
24Genetics
Olivia, this is your
health report

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1. Introduction

On the following pages, we offer you the health report obtained from the analysis of your DNA. In it, you will find information about your genetic predispositions to health.

Here are some basic things to keep in mind before reading this report.

The process with which we obtain your personalized report

The process we have followed to prepare your health report consists of the following:

1. Extract DNA from the saliva sample you sent us.
2. Transform the biological data contained in DNA into bioinformatics data. This process is called sequencing. In case you already had your DNA sequencing, these first two steps were not necessary, and we went directly to step 3 with the raw data of your genetic map (RAW DATA file).
3. Please apply the algorithms developed exclusively by 24Genetics to this computer data, allowing us to obtain your personalized report.

As you can see, we combine purely biological processes with computer processes so that, without losing an iota of scientific rigor, we can process vast amounts of information and offer you such detailed reports.

How is our algorithm?

The 24Genetics algorithm is based on the **analysis and study of thousands of publications** (called " papers " in the scientific environment), contrasted, validated, and recognized by the international scientific community, adding value to our reports.

Thanks to the reliability of our ancestry test, the first step in our genetic analysis is to **identify the sex and ancestry** of each individual. From there, **we exclusively apply the appropriate studies for each profile** whenever it is possible to do so. To obtain the genetic report of a European woman, we do not usually use, for example, studies whose analyzed population has been exclusively male or Asian. At this point, we could apply a single analysis, but **we combine a multitude of validated publications**, refining the process with artificial intelligence. Thus, we could use all available scientific knowledge to calculate genetic predispositions.

With this, we gain **accuracy and reliability in our results.**

Methodology

Our genetic reports are obtained based on three types of analysis methodology:

- **GWAS** (Genome-Wide Association Study). This is a type of study in which DNA markers in the whole genome (a person's complete genetic material) of people with a disease or trait are compared with those of people who do not have that disease or feature. It is a study based on statistics, which considers many genes associated with a predisposition in a not-so-direct way but whose sum offers a relevant conclusion.

- **Multivariate analysis**. In this case, our algorithm analyzes several genetic variants or mutations of one or several genes, which correlate more directly with the predisposition.

- **Monovariate analysis**. In this type of methodology, it is a single variant of a single gene that determines the predisposition due to its strong correlation with the genotype.

Each of the traits discussed in this report is based on one of these three types of methodology.

The data and conclusions in this report, like the progress of scientific research in genetics, may evolve. New mutations are continually being discovered, and the ones we analyze today are getting to know better. At 24Genetics, we make a great effort to apply newly established scientific discoveries to our reports.

What information do we offer you?

The information provided by our reports speaks of **predispositions**. And what do we mean by this?

In the case of this health report, we have two main types of diseases: complex and hereditary.

- **Complex diseases** have two factors of influence: genetics and environmental factors, or environment and habits. Depending on each pathology, both types of characteristics have a greater or lesser weight.

Complex diseases are analyzed using the three studies mentioned in the previous section: GWAS, multivariate analysis, and monovarietal analysis.

Let's give an example. The possibility of suffering from diabetes is influenced by the two types of factors that we have just mentioned: **genetic and environmental.** Genetic factors indicate the natural propensity we have to suffer from diabetes. On the other hand, the so-called ecological factors include elements that also affect, such as diet, habits, stress level, place where we live, climate, age, etc. Whether or not we eventually develop diabetes depends on the combination of both factors. And, even if we have a genetic predisposition to suffer from it, if we maintain a healthy weight, control glucose consumption, have stress under control, play sports, etc, we may never develop it. Or vice versa.

Conversely, **hereditary diseases** are only influenced by genetics and are analyzed based exclusively on mutations (monovarietal or multivariate analysis). In this case, only a particular modification or transformation determines the propensity to suffer from the disease or be a carrier without developing it. In this case, environmental factors do not play a role.

However, even though environmental factors do not play a role, each pathogenic mutation associated with a possible disease may or may not cause the development of said disease and may do so at different levels. In this sense, we can talk about two concepts:

o Penetrance is the percentage of people who develop the disease out of all those with the pathogenic mutation. In some cases, this figure is 100% since mutations necessarily cause the disease.

o Expressivity consists of the range of clinical manifestations associated with the disease being suffered. With the same condition, one person can have very few symptoms, and another, all that can entail.

In addition to complex and hereditary diseases, our report includes other types of pathologies or indicators, such as intolerances, biomarkers, and others, which you can see described later in the "Structure of this report" section.

In this report you could see some pathologies that cannot develop in your biological sex, such as ovarian cancer, which for obvious reasons cannot occur in a biological male. We did not want to remove that information from your report, because you may be a carrier of a mutation or mutations associated with that disease and pass it on to offspring, who could develop the disease, so the information is equally valuable.

In any of the cases, our reports tell you are always genetic predispositions, either because environmental factors play a role or because our tests do not analyze the entire genome and are not considered diagnostic tests.

What does this genetic report give you?

In this report, you have a large amount of **scientifically validated information** about your predispositions, and this allows you to know **how your body works** naturally and what aspects you should possibly pay attention to

At 24Genetics, we recommend that you always consult a doctor, who will act with all his knowledge and experience, be able to clarify your doubts, complement this report with your health history and available family history, supervise the follow-up of your possible pathology, or prescribe additional diagnostic tests, if he deems it necessary to confirm the risk of one or more specific predispositions.

A fundamental concept: the genetic variant.

Regarding genetic concepts, we want to share a basic one, which appears in all the traits in our reports and is essential for you to understand at least briefly, such as genetic variants (also called variation or mutation). The variant is a permanent change in the DNA sequence that forms a gene and is what marks an individual predisposition. Therefore, in each trait in this report, you will see information about the gene or genes affected in that trait. One or more variants in that gene or genes determine the different predispositions of some people compared to others.

For example, in the case of thyroid cancer, the rs77316810 and rs79781594 variants of the RET gene can mark the predisposition to suffer from this disease.

1.1. Structure of this report

This report is organized into the following categories:

1. Complex diseases: GWAS

Complex diseases are defined as pathologies whose development is influenced by multiple factors. Genetics is only one part, and other environmental factors, such as lifestyle, diet, where we live, our daily stress level, age, etc., can be as essential or more significant than our genes.

This section will exclusively include complex diseases analyzed using the GWAS (Genome-Wide Association) methodology. Studies), that is, biostatistical analysis, to which we have already referred in the "Methodology" section.

In these pathologies, the information we will obtain is based on a comparison with the population's mean. Therefore, your result will indicate whether you have a higher, equal, or lower predisposition than the population average. Usually, we will tell you that you have a higher genetic predisposition than the average if you are in the 10% of the population with the highest propensity to that disease and less if you are in the 10% of the people with the most negligible bias. We remind you, as we have already indicated in this report, that having a penchant or not does not mean that you are going to suffer from a disease or that you are free of it since many other factors influence it. In addition, it is common to have a greater predisposition than the average in between 10 and 20% of the pathologies analyzed.

To facilitate the understanding of the information, we have classified these diseases by medical specialties or areas of the body.

1.1. Neurology
1.2. Circulatory system
1.3. Digestive system
1.4. musculoskeletal system
1.5. Endocrinology
1.6. urogenital system
1.7. Dermatology
1.8. other

2. Complex diseases: oncogenic mutations

In this section we continue to analyze complex diseases, i.e. multifactorial diseases, which are influenced by both genetic and environmental factors, but the difference with the previous section is that we rely on the detection of mutations in one or more markers of one or more genes (monovariate or multivariate analysis, as described in the "Methodology" section). These mutations by themselves already mark the genetic predisposition to suffer from that disease, without any comparison with the population. Therefore, in the results of these diseases, we tell you whether or not we have found mutations likely to be pathogenic, and we do not make any comparison with the population. For this section, we consider pathogenic the mutations included in the ClinVar database.

3. Complex diseases: others

In this section, we include complex diseases analyzed by detecting mutations in one or more markers of one or more genes (monovarietal or multivariate analysis) unrelated to oncological processes. They share the methodology with the previous section, but they are not cancer-related diseases. As in the earlier cases, these are complex diseases and, as such, multifactorial.

4. Viruses, bacteria and fungi

Genetics are essential in the relationship between viruses, bacteria, and fungi and the diseases they can cause. Your genes may indicate greater susceptibility or resistance to a viral, bacterial, or fungal infection. Using all our types of methodologies (GWAS, multivariate or monovarietal), in this section, we will inform you of your genetic predisposition to multiple infectious diseases, such as tuberculosis, Covid, pneumonia, bronchitis or herpes, among others, and even the risk of aggravation of some of them.

5. Allergies and intolerances

In this section, we analyze a series of intolerances and allergies in the food, dermatological and respiratory fields, and we tell you if you have a genetic predisposition to suffer from them. Thus, with the help of a health professional, you can take the appropriate measures to try to avoid them or modulate their symptoms and improve your well-being. We use our three methodologies in the allergies and intolerances section, so the result of each of your analyzed traits will depend on the specific methods we have used.

6. Biomarkers and others

Some physiological parameters, such as cholesterol or triglyceride levels, bone density, or the number of white blood cells, platelets, or neutrophils, among many others, are influenced by your DNA, which determines your possible tendency to have abnormal indicators.

In this section, we exclusively use the GWAS methodology, so the results will indicate whether you are more, equal, or less predisposed than the population average to having abnormal levels of each parameter.

7. Pharmacogenetics

The same drug can work differently in different people; part of that possible effect depends on DNA. That is, your genetics can influence the response to varying types of drugs in terms of level of toxicity, effectiveness, metabolism, or necessary dose.

In this section, through monovarietal and multivariate analysis, we study your genetic predisposition for your body to respond in one way or another to certain medications.

8. Hereditary diseases: genetics

Hereditary diseases, unlike complex ones, are not influenced by environmental factors. DNA is the only influence factor to suffer from them or not. In this section, for each of the diseases that we analyze, we look for pathogenic mutations, or mutations likely to be, reported in the most critical genetic databases worldwide, mainly OMIM and ClinVar, and that have been associated with said pathologies.

Most of the diseases listed in this section can be classified into the so-called "rare diseases," and, as we have commented, lifestyle or other external factors do not affect the possibility of suffering from these ailments, only DNA influences. Additionally, we remind you that the mutations associated with a disease can cause its development or not and, in case of developing it, do so with different intensity, according to the concepts of penetrance and expressivity described earlier in this introduction.

As their name suggests, hereditary diseases are likely to be transmitted to your descendants. In this sense, it should be noted that having a pathogenic mutation that predisposes to a condition does not always imply suffering from it, and there can be 2 cases:

1. Being a carrier and also developing the disease.

2. You are a carrier of the disease (which happens whenever you have the pathogenic mutation) but not developing it. In this case, although the condition does not create, the pathogenic mutation can be transmitted to the offspring and, therefore, the predisposition to the disease. The greater or lesser probability of inheriting the pathogenic mutation by the offspring also depends on the genetics of the other parent. Therefore, this information is precious.

These types of diseases are mostly monogenetic, so one or more mutations of a single gene mark the predisposition to suffer a specific pathology.

It is important to note that this test does not sequence the complete genome. Still, we analyze just over 700,000 of the 3.2 million genetic markers that mark variability in the human genome, so there may be other mutations **in areas of the genome that we are not analyzing.**

*** The information provided in this report is for research, information, and educational uses only. In no case is it valid for clinical or diagnostic use.**

1.2. Frequently Asked Questions

Does it all depend on my genes?

No. The body responds to a whole series of conditions. Our genes are certainly an important parameter, but lifestyle, such as exercise and diet, influence our body. Undoubtedly, knowing yourself well helps to treat the body in the most appropriate way, and this is what you can get from genetics. Thanks to a genetic test for disease prevention, you obtain more knowledge for yourself and for the professionals who care for your health.

If my report says that I have a high genetic predisposition to suffer from a certain disease, does that mean that I will suffer from it?

We are our genes and our experiences.

Apart from your genes, there are many other environmental and internal factors that influence the development or not of a disease, so you can be genetically prone to a pathology and never develop it due to environmental reasons, health habits, lifestyle... But you can also not have a predisposition and suffer from a certain disease at some point in your life.

In addition, depending on the pathology, genetics may have a greater or lesser influence on the appearance or development of a disease.

Knowledge of our genetics through a disease DNA test allows health professionals to carry out their work with much more information. In addition, it allows designing prevention plans that can make a difference.

Do I have to make drastic changes in my health treatments on my own as a result of the results of this health and disease DNA test?

Our reports show data on your body's genetic predispositions, but there are many other external, environmental or habit factors that influence it. For this reason, we consider our reports as preventive, not diagnostic. Our recommendation is to always consult with medical professionals in case of any doubt that may arise from your health DNA test. Therefore, the answer is no, you should not make major changes without the validation of a professional.

If my report says that I am not prone to a certain disease, does that mean that I am not at risk?

Most diseases do not depend only on our genes, they also depend on countless internal and external factors that can cause them. In addition, our health DNA test has partial information about your genome.

We are not sequencing the complete genome, but only a part, so it does not exclude the possibility that you may carry other mutations associated with said pathology in other gene regions that we are not analyzing or that are not currently known.

There are genetic tests for clinical or diagnostic use, which analyze all the genes involved in a certain pathology or disease and which a medical service can prescribe if deemed appropriate. And, of course, one must always take into account multiple environmental factors, as these can also have a high degree of influence on the possibility of disease development.

Our genetic health and disease tests are not valid for clinical or diagnostic use. Therefore, when in doubt, we always recommend consulting your doctors so that they are the ones who prescribe the appropriate clinical genetic tests.

Does my genetic predisposition to suffer from certain pathologies mean that my relatives also have it?

The genetics of each person is unique, so we always recommend that you consult with your reference clinical service the decisions to be made in terms of health. However, in genetics, many of the patterns that are expressed are often related to those of close relatives, so it would be normal for the reports to be quite similar. However, keep in mind that multiple external factors also influence the development or not of a disease, so that the probability of suffering from it will be very different among family members with different lifestyles, health habits, place of residence, etc.

Some of the studies on which our DNA test for health is based.

The 24Genetics genetic health test is based on thousands of genetic investigations agreed upon by the international scientific community. Our system selects the research that is applicable to you (depending on your gender and ancestry) and our algorithm combines it to provide you with the most useful information for your health and well-being. Here are some examples of genetic research used:

- Ahmed S et al; Newly discovered breast cancer susceptibility loci on 3p24 and 17q23; Nat Genet; 2009 May;41(5):585-90.

- Cox A et al; A common coding variant in CASP8 disassociated with breast cancer risk; Nat Genet; 2007 Mar;39(3):352-8.

- Dickson C et al; Tyrosine kinase signalling in breast cancer: fibroblast growth factors and their receptors; Breast Cancer Res; 2000;2(3):191-6.

- Easton DF et al; Genome-wide association study identifies novel breast cancer susceptibility loci; Nature; 2007 Jun28;447(7148):1087-93.

- Hunter DJ et al; A genome-wide association study identifies alleles in FGFR2 associated with risk of sporadic postmenopausal breast cancer; Nat Genet; 2007 Jul;39(7):870-4.

- Chang YK et al; Association of BANK1 and TNFSF4 with systemic lupus erythematosus in Hong Kong Chinese; Genes Immun.; 2009; 10(5):414-20.

2. Summary

GWAS Complex Diseases: Neurology

- Parkinson's disease
- Motion sickness
- Multiple sclerosis
- Neuroblastoma
- Glioma
- Intracranial aneurysm
- Alzheimer's disease (late onset)
- Schizophrenia
- Conduct disorder

Caption:

- According to this study, you have a predisposition similar to most of the population.
- According to this study, you are less likely to suffer from this disease than the majority of the population.
- According to this study, you are more likely to suffer from this disease than most of the population.

GWAS Complex Diseases: Circulatory System

- Primary biliary cirrhosis
- Myocardial infarction (early onset)
- Hodgkin's lymphoma
- Follicular lymphoma
- Coronary heart disease
- Chronic lymphocytic leukemia
- Diffuse large B cell lymphoma
- Wilms tumor

Caption:

- According to this study, you have a predisposition similar to most of the population.
- According to this study, you are less likely to suffer from this disease than the majority of the population.
- According to this study, you are more likely to suffer from this disease than most of the population.

GWAS Complex Diseases: Respiratory System

- Upper aerodigestive tract cancers
- Asthma
- Chronic bronchitis and chronic obstructive pulmonary disease

Caption:

- According to this study, you have a predisposition similar to most of the population.
- According to this study, you are less likely to suffer from this disease than the majority of the population.
- According to this study, you are more likely to suffer from this disease than most of the population.

GWAS Complex Diseases: Musculoskeletal System

- Systemic sclerosis
- Rheumatoid arthritis
- Myasthenia gravis
- Osteosarcoma
- Multiple myeloma

Caption:

- According to this study, you have a predisposition similar to most of the population.
- According to this study, you are less likely to suffer from this disease than the majority of the population.
- According to this study, you are more likely to suffer from this disease than most of the population.

GWAS Complex Diseases: Endocrinology

- Type 1 diabetes
- Type 1 diabetes nephropathy

- Type 2 diabetes
- Hypothyroidism

Caption:

- According to this study, you have a predisposition similar to most of the population.
- According to this study, you are less likely to suffer from this disease than the majority of the population.
- According to this study, you are more likely to suffer from this disease than most of the population.

GWAS Complex Diseases: Urogenital System

- Endometriosis
- Bladder cancer

Caption:

- According to this study, you have a predisposition similar to most of the population.
- According to this study, you are less likely to suffer from this disease than the majority of the population.
- According to this study, you are more likely to suffer from this disease than most of the population.

GWAS Complex Diseases: Dermatology

- Basal cell carcinoma
- Psoriasis
- Vitiligo

Caption:

- According to this study, you have a predisposition similar to most of the population.
- According to this study, you are less likely to suffer from this disease than the majority of the population.
- According to this study, you are more likely to suffer from this disease than most of the population.

GWAS Complex Diseases: Others

- Celiac disease
- Age-related macular degeneration

Caption:

- According to this study, you have a predisposition similar to most of the population.
- According to this study, you are less likely to suffer from this disease than the majority of the population.
- According to this study, you are more likely to suffer from this disease than most of the population.

Complex Diseases: Oncogenic Mutations

- APC: colorrectal and pancreatic cancer
- ATM: breast cancer
- BARD1: breast cancer
- BLM: colorrectal cancer
- BMPR1A: colorrectal, gastric and pancreatic cancer
- BRCA1: breast and ovarian cancer
- BRCA2: breast and ovarian cancer
- BRIP1: breast cancer
- CDH1: breast and gastric cancer
- CDK4: Familial melanoma
- CDKN2A: pancreatic cancer
- CHEK2: breast and colorrectal cancer
- DICER1: ovarian cancer
- EPCAM: Lynch syndrome, breast, ovarian, uterine, colorrectal, gastric and pancreatic cancer
- FH: Hereditary leiomyomatosis and renal cell cancer
- FLCN: Kidney cancer
- MEN1: multiple endocrine neoplasia type 1
- MET: Lung and gastric cancer
- MITF: MITF-related melanoma and renal cell carcinoma predisposition syndrome
- MLH1: Lynch syndrome
- MSH2: Lynch syndrome and colorrectal cancer
- MSH6: Lynch syndrome and colorrectal cancer

- MUTYH: colorrectal cancer
- NBN: breast, ovarian, colorrectal and gastric cancer
- NF1: type 1 neurofibromatosis
- NF2: Familial multiple meningioma
- NTHL1: Attenuated familial adenomatous polyposis
- RAD50: breast and pancreatic cancer
- PMS2: Lynch syndrome and colorrectal cancer
- POLD1: breast, ovarian, uterine and colorrectal cancer
- POLE: ovarian, uterine, colorrectal andpancreatic cancer
- MSH3-related attenuated familial adenomatous polyposis
- POT1: Familial melanoma
- PTCH1: Basal cell carcinoma
- PTEN: breast, uterine and colorrectal cancer
- RAD50: breast and ovarian cancer
- RAD51C: ovarian cancer
- RB1: Lynch syndrome and retinoblastoma
- RECQL4: Stomach and colon cancer
- RET: thyroid carcinoma
- SDHA: gastric cancer
- SDHAF2: Hereditary pheochromocytoma-paraganglioma
- SDHB: gastric cancer
- SDHC: gastric cancer
- SDHD: breast, uterine and gastric cancer
- BAP1-related tumor predisposition syndrome
- SMAD4: juvenile polyposis syndrome and colorrectal cancer
- SMARCA4: ovarian cancer
- SMARCB1: Familial rhabdoid tumor
- SMARCE1: Familial multiple meningioma
- STK11: breast, ovarian, uterine, colorrectal, gastric and pancreatic cancer
- TERT: Familial melanoma
- TP53: Li-Fraumeni syndrome, breast cancer and more
- VHL: Von Hippel-Lindau syndrome
- WT1: Nephroblastoma
- Familial adenomatous polyposis
- Kenny-Caffey syndrome

Caption:

- We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions.
- We have detected at least one mutation that could be pathogenic.

Complex Diseases: Multivariate Analysis

- Septic shock
- TSC1: tuberous sclerosis complex 1
- TSC2: tuberous sclerosis complex 2

Caption:

- We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions.
- We have detected at least one mutation that could be pathogenic.

Viruses, Bacteria and Fungi

- The severity of COVID-19 infection
- Severe Acute Respiratory Syndrome (SARS)
- HIV Transmission
- Genital herpes
- Cirrhosis due to Hepatitis B
- Community-acquired pneumonia
- Severe hospital pneumonia
- Bronchitis

Caption:

- According to this study, you have a predisposition similar to most of the population.
- According to this study, you are less likely to suffer from this disease than the majority of the population.
- According to this study, you are more likely to suffer from this disease than most of the population.

Allergies and Intolerances

- Lactose intolerance
- Shellfish allergy
- Allergic rhinitis
- DAO deficiency and migraines
- Mercury Accumulation
- Allergy to grass pollen

Caption:

- According to this study, you have a predisposition similar to most of the population.
- According to this study, you are less likely to suffer from this disease than the majority of the population.
- According to this study, you are more likely to suffer from this disease than most of the population.

Biomarkers and Others

- Calcium levels
- Magnesium levels
- Beta-2 microglubulin plasma levels
- Serum total protein level
- Glycerophospholipid levels
- Phospholipid levels (plasma)
- Heart rate
- Thyroid hormone levels
- Neutrophil levels
- Platelet levels
- Monocyte levels
- Menopause (age at onset)
- Lung volume
- Phosphorus levels
- Plasma omega-6 polyunsaturated fatty acid levels (dihomo-gamma-linolenic acid)
- Glycated hemoglobin levels
- GGT levels
- Serum albumin level
- Aortic root size
- Bilirubin levels
- Eosinophil levels
- Interleukin 6 and Inflammation
- White blood cell count
- Uric acid levels
- Bone mineral density
- Longevity

Caption:

- According to this study, you have a similar predisposition to the majority of the population to have normal levels.
- According to this study, you have a better predisposition than the majority of the population to have normal levels.
- According to this study, you have a greater predisposition than most of the population to suffer abnormal levels.

Pharmacogenetics

- Warfarin
- Pentazocine
- Aspirin
- Bupropion
- Methotrexate
- Vincristine
- Peginterferon Alpha-2b
- Meperidine
- Morphine
- Simvastatin
- Pravastatin
- Fluorouracil, capecitabine, pyrimidine analogues, tegafur and Neoplasms
- Tacrolimus
- Ribavirin

Caption:

- We have not found anything in your genetics that indicates a predisposition to an abnormal effect of this drug. Other non-analyzed and non-genetic genetic factors may play a role.
- According to your genotype you have a greater predisposition for the drug to have an abnormal effect on you. Other non-analyzed and non-genetic genetic factors may play a role.
- According to your genotype you have a greater predisposition for the drug to have a harmful effect on you. Other non-analyzed and non-genetic genetic factors may play a role.
- According to your genotype, you have a greater predisposition to respond positively to this drug. Other non-analyzed and non-genetic genetic factors may play a role.

Hereditary Diseases (genetics)

- Isovaleric acidemia
- Combined malonic and methylmalonic acidemia
- Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency
- Vitamin B12-unresponsive methylmalonic acidemia
- Vitamin B12-responsive methylmalonic acidemia
- Propionic acidemia
- Congenital lactic acidosis, Saguenay-Lac-Saint-Jean type
- Distal renal tubular acidosis
- 3-methylglutaconic aciduria type 1
- 3-methylglutaconic aciduria type 7
- 3-methylglutaconic aciduria type 9
- Argininosuccinic aciduria
- D-2-hydroxyglutaric aciduria
- Formiminoglutamic aciduria
- Fumaric aciduria
- Mevalonic aciduria
- Achondroplasia
- Achromatopsia
- Gastric adenocarcinoma and proximal polyposis of the stomach
- X-linked adrenoleukodystrophy
- Neurological conditions associated with aminoacylase 1 deficiency
- X-linked agammaglobulinemia
- Oculocutaneous albinism type 1
- Oculocutaneous albinism type 2
- Oculocutaneous albinism type 3
- Oculocutaneous albinism type 4
- Alkaptonuria
- Alpha-thalassemia
- Alpha-mannosidosis
- ALG1-CDG
- ALG6-CDG
- ALG8-CDG
- ATTRV30M amyloidosis
- Familial primary localized cutaneous amyloidosis
- Multiple myeloma
- Congenital dyserythropoietic anemia type I
- Congenital dyserythropoietic anemia type II
- Sickle cell anemia
- Hemolytic anemia due to glucophosphate isomerase deficiency
- Hemolytic anemia due to pyrimidine 5' nucleotidase deficiency
- Hemolytic anemia due to red cell pyruvate kinase deficiency
- X-linked sideroblastic anemia
- X-linked sideroblastic anemia and spinocerebellar ataxia
- Enteric anendocrinosis
- Hereditary angioedema
- Distal anoctaminopathy
- Peters anomaly
- Rieger anomaly
- Uhl anomaly

- 46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency
- Aplasia of lacrimal and salivary glands
- Systemic-onset juvenile idiopathic arthritis
- Distal arthrogryposis type 5D
- VACTERL/VATER association
- Autosomal recessive ataxia due to ubiquinone deficiency
- Adult-onset autosomal recessive cerebellar ataxia
- Non-progressive cerebellar ataxia with intellectual disability
- Autosomal dominant spastic ataxia type 1
- Spinocerebellar ataxia with axonal neuropathy type 1
- Infantile-onset spinocerebellar ataxia
- Spinocerebellar ataxia type 19/22
- Spinocerebellar ataxia type 28
- Multiple intestinal atresia
- Autosomal dominant congenital benign spinal muscular atrophy
- Scapuloperoneal spinal muscular atrophy
- Congenital bilateral absence of vas deferens
- Beta-mannosidosis
- Bradyopsia
- Nasopharyngeal carcinoma
- Cystinuria
- Keratosis follicularis spinulosa decalvans
- COG5-CDG
- Neonatal intrahepatic cholestasis due to citrin deficiency
- Metaphyseal chondrodysplasia, Spahr type
- Infantile convulsions and choreoathetosis
- Cranio-osteoarthropathy
- Autosomal recessive cutis laxa type 1
- Isolated congenital anonychia
- Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy
- Distal arthrogryposis type 1
- Progressive pseudorheumatoid arthropathy of childhood
- Aspartylglucosaminuria
- Autosomal recessive ataxia, Beauce type
- Autosomal recessive cerebellar ataxia due to CWF19L1 deficiency
- X-linked progressive cerebellar ataxia
- Spinocerebellar ataxia with epilepsy
- Spinocerebellar ataxia with axonal neuropathy type 2
- Spinocerebellar ataxia type 13
- Spinocerebellar ataxia type 21
- Ataxia-oculomotor apraxia type 1
- Gyrate atrophy of choroid and retina
- Spinal muscular atrophy with respiratory distress type 1
- Autosomal dominant childhood-onset proximal spinal muscular atrophy
- Autosomal recessive bestrophinopathy
- Beta-thalassemia
- Autosomal dominant brachyolmia
- Familial papillary or follicular thyroid carcinoma
- Citrullinemia type I
- COG4-CDG
- Progressive familial intrahepatic cholestasis
- Tuberous sclerosis complex
- X-linked dominant chondrodysplasia punctata
- Paroxysmal dystonic choreathetosis with episodic ataxia and spasticity
- Hereditary cryohydrocytosis with reduced stomatin
- Autosomal recessive cutis laxa type 2A

- Autosomal recessive cutis laxa type 2B
- Congenital bile acid synthesis defect type 1
- Isolated cytochrome C oxidase deficiency
- Isolated complex III deficiency
- Combined oxidative phosphorylation defect type 15
- Combined oxidative phosphorylation defect type 8
- Congenital fibrinogen deficiency
- Congenital factor V deficiency
- Congenital factor XIII deficiency
- 3-hydroxy-3-methylglutaryl-CoA synthase deficiency
- Acyl-CoA dehydrogenase 9 deficiency
- Medium chain acyl-CoA dehydrogenase deficiency
- Adenylosuccinate lyase deficiency
- Aromatase deficiency
- Beta-ureidopropionase deficiency
- Butyrylcholinesterase deficiency
- Carnitine palmitoyl transferase 1A deficiency
- Carnitine-acylcarnitine translocase deficiency
- Fatal infantile cytochrome C oxidase deficiency
- Dimethylglycine dehydrogenase deficiency
- Fructose-1,6-bisphosphatase deficiency
- Glutaryl-CoA dehydrogenase deficiency
- Guanidinoacetate methyltransferase deficiency
- LCAT deficiency
- Lipoyl transferase 1 deficiency
- Myeloperoxidase deficiency
- Alpha-N-acetylgalactosaminidase deficiency
- Pyruvate carboxylase deficiency, benign type
- Prolidase deficiency
- DDOST-CDG
- Congenital bile acid synthesis defect type 4
- Isolated complex I deficiency
- Non-acquired isolated growth hormone deficiency
- Combined oxidative phosphorylation defect type 20
- Congenital intrinsic factor deficiency
- Congenital sucrase-isomaltase deficiency
- Congenital factor XI deficiency
- 3-phosphoglycerate dehydrogenase deficiency, infantile/juvenile form
- Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency
- Short chain acyl-CoA dehydrogenase deficiency
- Very long chain acyl-CoA dehydrogenase deficiency
- Alpha-1-antitrypsin deficiency
- Beta-ketothiolase deficiency
- Biotinidase deficiency
- Carbamoyl-phosphate synthetase 1 deficiency
- Carnitine palmitoyltransferase II deficiency
- Cernunnos-XLF deficiency
- Dihydropyrimidine dehydrogenase deficiency
- Dopamine beta-hydroxylase deficiency
- Class I glucose-6-phosphate dehydrogenase deficiency
- Glutathione synthetase deficiency
- Holocarboxylase synthetase deficiency
- Lysosomal acid lipase deficiency
- Homocystinuria without methylmalonic aciduria
- Monoamine oxidase A deficiency
- Ornithine transcarbamylase deficiency
- Pyruvate dehydrogenase deficiency

- Mitochondrial trifunctional protein deficiency
- Purine nucleoside phosphorylase deficiency
- Succinyl-CoA:3-oxoacid CoA transferase deficiency
- Multiple acyl-CoA dehydrogenase deficiency
- Combined pituitary hormone deficiencies, genetic forms
- Brain demyelination due to methionine adenosyltransferase deficiency
- Desmosterolosis
- Nephrogenic diabetes insipidus
- Congenital sodium diarrhea
- Dihydropyrimidinuria
- Severe intellectual disability and progressive spastic paraplegia
- X-linked intellectual disability, Cabezas type
- X-linked intellectual disability, Najm type
- Intellectual disability, Birk-Barel type
- Paroxysmal exertion-induced dyskinesia
- Cortical dysgenesis with pontocerebellar hypoplasia due to TUBB3 mutation
- Postaxial acrofacial dysostosis
- Cerebrofaciothoracic dysplasia
- Craniofrontonasal dysplasia
- Singleton-Merten dysplasia
- Hidrotic ectodermal dysplasia
- Multiple epiphyseal dysplasia, Beighton type
- Spondyloepimetaphyseal dysplasia, PAPSS2 type
- Spondyloepimetaphyseal dysplasia with multiple dislocations
- Acromelic frontonasal dysplasia
- Schimke immuno-osseous dysplasia
- Otospondylomegaepiphyseal dysplasia
- FLNA-related X-linked myxomatous valvular dysplasia
- Dopa-responsive dystonia due to sepiapterin reductase deficiency
- Adult-onset dystonia-parkinsonism
- Pterin-4 alpha-carbinolamine dehydratase deficiency
- S-adenosylhomocysteine hydrolase deficiency
- Familial glucocorticoid deficiency
- Systemic primary carnitine deficiency
- Infantile cerebellar-retinal degeneration
- Desminopathy
- Maternally-inherited diabetes and deafness
- Congenital chloride diarrhea
- Syndromic diarrhea
- Familial dysautonomia
- Syndromic X-linked intellectual disability due to JARID1C mutation
- X-linked intellectual disability, Snyder type
- 2q23.1 microdeletion syndrome
- Familial dyskinesia and facial myokymia
- Familial aortic dissection
- X-linked complicated corpus callosum dysgenesis
- Acromicric dysplasia
- FGFR2-related bent bone dysplasia
- Non-epidermolytic palmoplantar keratoderma
- Diastrophic dysplasia
- Hypohidrotic ectodermal dysplasia
- Spondyloepiphyseal dysplasia congenita
- Spondyloepiphyseal dysplasia, Stanescu type
- Spondyloepimetaphyseal dysplasia congenita, Strudwick type
- Gnathodiaphyseal dysplasia
- Odonto-onycho-dermal dysplasia
- Thanatophoric dysplasia
- Familial isolated arrhythmogenic right ventricular dysplasia
- Early-onset generalized limb-onset dystonia
- Reis Bücklers corneal dystrophy

- Granular corneal dystrophy type II
- Lattice corneal dystrophy type I
- Congenital hereditary endothelial dystrophy type II
- Congenital muscular dystrophy with cerebellar involvement
- Congenital muscular dystrophy, Ullrich type
- Becker muscular dystrophy
- DNAJB6-related limb-girdle muscular dystrophy D1
- Titin-related limb-girdle muscular dystrophy R10
- Anoctamin-5-related limb-girdle muscular dystrophy R12
- GMPPB-related limb-girdle muscular dystrophy R19
- Alpha-sarcoglycan-related limb-girdle muscular dystrophy R3
- Gamma-sarcoglycan-related limb-girdle muscular dystrophy R5
- FKRP-related limb-girdle muscular dystrophy R9
- Tibial muscular dystrophy
- Infantile neuroaxonal dystrophy
- Progressive cone dystrophy
- Best vitelliform macular dystrophy
- Isolated ectopia lentis
- Mitochondrial neurogastrointestinal encephalomyopathy
- Early infantile epileptic encephalopathy
- Severe neonatal-onset encephalopathy with microcephaly
- Glycine encephalopathy
- Central core disease
- Addison disease
- Glycogen storage disease due to glycogen debranching enzyme deficiency
- Glycogen storage disease due to muscle phosphofructokinase deficiency
- Glycogen storage disease due to liver phosphorylase kinase deficiency
- Glycogen storage disease due to liver glycogen phosphorylase deficiency
- Granular corneal dystrophy type I
- Bietti crystalline dystrophy
- Benign concentric annular macular dystrophy
- Congenital muscular dystrophy with integrin alpha-7 deficiency
- Congenital muscular dystrophy due to LMNA mutation
- Autosomal dominant limb-girdle muscular dystrophy type 1A
- Calpain-3-related limb-girdle muscular dystrophy R1
- POMT1-related limb-girdle muscular dystrophy R11
- POMT2-related limb-girdle muscular dystrophy R14
- Dysferlin-related limb-girdle muscular dystrophy R2
- Beta-sarcoglycan-related limb-girdle muscular dystrophy R4
- Telethonin-related limb-girdle muscular dystrophy R7
- Duchenne muscular dystrophy
- Muscular dystrophy, Selcen type
- Butterfly-shaped pigment dystrophy
- Bothnia retinal dystrophy
- DPM1-CDG
- Microcephalic osteodysplastic primordial dwarfism type II
- KCNQ2-related epileptic encephalopathy
- Ethylmalonic encephalopathy
- Encephalopathy due to sulfite oxidase deficiency
- STAT3-related early-onset multisystem autoimmune disease
- Juvenile neuronal ceroid lipofuscinosis
- Alexander disease
- Glycogen storage disease due to glycogen branching enzyme deficiency
- Glycogen storage disease due to phosphoglycerate mutase deficiency
- Glycogen storage disease due to liver and muscle phosphorylase kinase deficiency
- Glycogen storage disease due to muscle glycogen phosphorylase deficiency

- Glycogen storage disease due to hepatic glycogen synthase deficiency
- Canavan disease
- Autosomal dominant Charcot-Marie-Tooth disease type 2D
- X-linked Charcot-Marie-Tooth disease type 5
- Charcot-Marie-Tooth disease type 1D
- Autosomal dominant Charcot-Marie-Tooth disease type 2N
- SURF1-related Charcot-Marie-Tooth disease type 4
- Charcot-Marie-Tooth disease type 4C
- Charcot-Marie-Tooth disease type 4J
- Sporadic Creutzfeldt-Jakob disease
- Dent disease
- Fabry disease
- Hirschsprung disease
- Lafora disease
- Menkes disease
- Niemann-Pick disease type A
- Niemann-Pick disease type C
- Oguchi disease
- Refsum disease
- Sandhoff disease
- Tangier disease
- Thomsen and Becker disease
- Von Willebrand disease type 1
- Von Willebrand disease type 3
- Fatal mitochondrial disease due to combined oxidative phosphorylation defect type 3
- Muscle-eye-brain disease
- Glycogen storage disease due to LAMP-2 deficiency
- Glycogen storage disease due to acid maltase deficiency
- Autosomal dominant generalized dystrophic epidermolysis bullosa
- Dystrophic epidermolysis bullosa pruriginosa
- Intermediate epidermolysis bullosa simplex with cardiomyopathy
- Caffey disease
- Autosomal dominant Charcot-Marie-Tooth disease type 2A2
- X-linked Charcot-Marie-Tooth disease type 1
- Charcot-Marie-Tooth disease type 1B
- Charcot-Marie-Tooth disease type 2B5
- Charcot-Marie-Tooth disease type 2T
- Charcot-Marie-Tooth disease type 4A
- Charcot-Marie-Tooth disease type 4F
- Coats disease
- Crouzon disease
- Free sialic acid storage disease
- Gaucher disease
- Krabbe disease
- Leber plus disease
- Naxos disease
- Niemann-Pick disease type B
- Norrie disease
- Pelizaeus-Merzbacher disease
- Chylomicron retention disease
- Stargardt disease
- Tay-Sachs disease
- Von Hippel-Lindau disease
- Von Willebrand disease type 2A
- Wilson disease
- Rippling muscle disease
- Åland Islands eye disease
- Glycogen storage disease due to glucose -6-phosphatase deficiency
- Autosomal recessive polycystic kidney disease
- Recessive dystrophic epidermolysis bullosa inversa
- Junctional epidermolysis bullosa with pyloric atresia
- Autosomal dominant generalized epidermolysis bullosa simplex, severe form

- Autosomal dominant generalized epidermolysis bullosa simplex, intermediate form
- Autosomal dominant epilepsy with auditory features
- Juvenile myoclonic epilepsy
- Progressive myoclonic epilepsy type 6
- Benign familial neonatal epilepsy
- Multiple self-healing squamous epithelioma
- Chuvash erythrocytosis
- Supravalvular aortic stenosis
- Dehydrated hereditary stomatocytosis
- Phenylketonuria
- Familial atrial fibrillation
- Idiopathic ventricular fibrillation, non Brugada type
- Congenital fibrosis of extraocular muscles
- Cystic fibrosis
- Phocomelia, Schinzel type
- Symptomatic form of hemochromatosis type 1
- Fucosidosis
- Fundus albipunctatus
- GM1 gangliosidosis
- MOGS-CDG
- Juvenile glaucoma
- Hawkinsinuria
- Hemochromatosis type 2
- Mild hemophilia A
- Mild hemophilia B
- Paroxysmal nocturnal hemoglobinuria
- Hepatoblastoma
- Hepatoencephalopathy due to combined oxidative phosphorylation defect type 1
- Hydrocephalus with stenosis of the aqueduct of Sylvius
- Hb Bart's hydrops fetalis
- Phosphoribosylpyrophosphate synthetase superactivity
- Familial hyperaldosteronism type I
- Transient familial neonatal hyperbilirubinemia
- Hyperimmunoglobulinemia D with periodic fever
- Autosomal dominant hyperinsulinism due to SUR1 deficiency
- Hyperinsulinism due to INSR deficiency
- Endosteal hyperostosis, Worth type
- Primary hyperoxaluria
- Familial isolated hyperparathyroidism
- Heritable pulmonary arterial hypertension
- Malignant hyperthermia of anesthesia
- Familial hypoaldosteronism
- Hypochondroplasia
- Hypophosphatasia
- X-linked hypophosphatemia
- Primary hypomagnesemia with secondary hypocalcemia
- Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement
- Focal dermal hypoplasia
- Pontocerebellar hypoplasia type 10
- Pontocerebellar hypoplasia type 2
- Pontocerebellar hypoplasia type 6
- Pontocerebellar hypoplasia type 8
- X-linked adrenal hypoplasia congenita
- Isolated optic nerve hypoplasia/aplasia
- Hypothyroidism due to TSH receptor mutations
- Hypotonia with lactic acidemia and hyperammonemia
- Hereditary renal hypouricemia
- Classic homocystinuria
- Homocystinuria due to methylene tetrahydrofolate reductase deficiency
- Harlequin ichthyosis

- Autosomal dominant epidermolytic ichthyosis
- Exfoliative ichthyosis
- Lamellar ichthyosis
- Recessive X-linked ichthyosis
- Incontinentia pigmenti
- Male infertility due to large-headed multiflagellar polyploid spermatozoa
- Combined immunodeficiency with granulomatosis
- Severe combined immunodeficiency due to adenosine deaminase deficiency
- Severe combined immunodeficiency due to DCLRE1C deficiency
- T-B+ severe combined immunodeficiency due to gamma chain deficiency
- Combined immunodeficiency due to partial RAG1 deficiency
- Immunodeficiency due to a late component of complement deficiency
- Immunodeficiency by defective expression of MHC class I
- Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins
- Isolated cleft lip
- Leprechaunism
- Acute lymphoblastic leukemia
- B-cell chronic lymphocytic leukemia
- Acute myeloid leukemia
- Juvenile myelomonocytic leukemia
- RARS-related autosomal recessive hypomyelinating leukodystrophy
- Hereditary diffuse leukoencephalopathy with axonal spheroids and pigmented glia
- Lymphangioleiomyomatosis
- Familial partial lipodystrophy, Dunnigan type
- Late infantile neuronal ceroid lipofuscinosis
- ATP13A2-related juvenile neuronal ceroid lipofuscinosis
- X-linked lissencephaly with abnormal genitalia
- Lissencephaly due to LIS1 mutation
- Lissencephaly due to TUBA1A mutation
- Lissencephaly type 1 due to doublecortin gene mutation
- Lysinuric protein intolerance
- Malaria
- MELAS
- Metachondromatosis
- Microlissencephaly
- Infantile hypertrophic cardiomyopathy due to MRPL44 deficiency
- Mitochondrial hypertrophic cardiomyopathy with lactic acidosis due to MTO1 deficiency
- Familial isolated restrictive cardiomyopathy
- Infantile myofibromatosis
- Autosomal dominant centronuclear myopathy
- X-linked centronuclear myopathy
- X-linked myopathy with excessive autophagy
- Polyglucosan body myopathy type 2
- Reducing body myopathy
- Congenital fiber-type disproportion myopathy
- Bethlem myopathy
- Miyoshi myopathy
- Distal myopathy with anterior tibial onset
- Laing early-onset distal myopathy
- Progressive scapulohumeroperoneal distal myopathy
- GNE myopathy
- Hereditary myopathy with early respiratory failure

- Mitochondrial myopathy with reversible cytochrome C oxidase deficiency
- Severe congenital nemaline myopathy
- Potassium-aggravated myotonia
- Complete hydatidiform mole
- Mucolipidosis type III
- Mucopolysaccharidosis type 2
- Mucopolysaccharidosis type 4
- Mucopolysaccharidosis type 7
- Mitochondrial membrane protein-associated neurodegeneration
- Neurofibromatosis-Noonan syndrome
- Autosomal recessive axonal neuropathy with neuromyotonia
- Autosomal recessive severe congenital neutropenia due to CSF3R deficiency
- Woolly hair nevus
- Obesity due to melanocortin 4 receptor deficiency
- Hypertrichotic osteochondrodysplasia, Cantu type
- Osteopetrosis with renal tubular acidosis
- Osteosarcoma
- Non-acquired panhypopituitarism
- Pachyonychia congenita
- Paramyotonia congenita of Von Eulenburg
- Autosomal dominant spastic paraplegia type 17
- Autosomal dominant spastic paraplegia type 8
- Autosomal recessive spastic paraplegia type 35
- Autosomal recessive spastic paraplegia type 56
- Spastic paraplegia type 2
- Pycnodysostosis
- PMM2-CDG
- Polymicrogyria due to TUBB2B mutation
- Syndactyly type 2
- Acute intermittent porphyria
- Multiminicore myopathy
- Inclusion body myopathy with Paget disease of bone and frontotemporal dementia
- MODY
- MPI-CDG
- Mucopolysaccharidosis type 1
- Mucopolysaccharidosis type 3
- Mucopolysaccharidosis type 6
- Multiple endocrine neoplasia type 2
- Neurofibromatosis type 6
- Navajo neurohepatopathy
- Leber hereditary optic neuropathy
- Autosomal recessive severe congenital neutropenia due to JAGN1 deficiency
- Obesity due to leptin receptor gene deficiency
- Autosomal recessive progressive external ophthalmoplegia
- Multiple osteochondromas
- Albers-Schönberg osteopetrosis
- Hereditary chronic pancreatitis
- Pachydermoperiostosis
- Hypokalemic periodic paralysis
- Autosomal dominant spastic paraplegia type 10
- Autosomal dominant spastic paraplegia type 31
- Autosomal recessive spastic paraplegia type 15
- Autosomal recessive spastic paraplegia type 54
- Autosomal recessive spastic paraplegia type 5A
- Spastic paraplegia type 7
- Familial clubfoot with or without associated lower limb anomalies
- Bilateral polymicrogyria
- Autosomal recessive spastic ataxia of Charlevoix-Saguenay
- Porencephaly
- Hepatoerythropoietic porphyria

- Congenital erythropoietic porphyria
- Autosomal erythropoietic protoporphyria
- Pseudopseudohypoparathyroidism
- Thrombotic thrombocytopenic purpura
- Autosomal dominant focal non-epidermolytic palmoplantar keratoderma with plantar blistering
- Palmoplantar keratoderma, Nagashima type
- Keratoderma hereditarium mutilans
- Autosomal dominant hypophosphatemic rickets
- Resistance to thyroid hormone due to a mutation in thyroid hormone receptor beta
- X-linked retinoschisis
- 3M syndrome
- ADNP syndrome
- Auriculocondylar syndrome
- BOR syndrome
- Branchiootic syndrome
- Cardiofaciocutaneous syndrome
- CHILD syndrome
- Congenital vertebral-cardiac-renal anomalies syndrome
- Heart-hand syndrome, Slovenian type
- Adams-Oliver syndrome
- Aicardi-Goutières syndrome
- Alazami syndrome
- Alpers-Huttenlocher syndrome
- Thiamine-responsive megaloblastic anemia syndrome
- Angelman syndrome
- Palatal anomalies-widely spaced teeth-facial dysmorphism-developmental delay syndrome
- Apert syndrome
- Progeroid and marfanoid aspect-lipodystrophy syndrome
- Lipoid proteinosis
- Pseudohypoparathyroidism type 1C
- Familial male-limited precocious puberty
- Striate palmoplantar keratoderma
- Isolated focal non-epidermolytic palmoplantar keratoderma
- Transgrediens et progrediens palmoplantar keratoderma
- Hypocalcemic vitamin D-dependent rickets
- Hereditary hypophosphatemic rickets with hypercalciuria
- Retinoblastoma
- Sebocystomatosis
- Acrocallosal syndrome
- ADULT syndrome
- Autosomal dominant intellectual disability-craniofacial anomalies-cardiac defects syndrome
- Branchio-oculo-facial syndrome
- CACH syndrome
- CHARGE syndrome
- Classic glucose transporter type 1 deficiency syndrome
- Constitutional mismatch repair deficiency syndrome
- Aarskog-Scott syndrome
- Corpus callosum agenesis-neuronopathy syndrome
- Alagille syndrome
- Allan-Herndon-Dudley syndrome
- Andersen-Tawil syndrome
- Aneurysm-osteoarthritis syndrome
- Anophthalmia/microphthalmia-esophageal atresia syndrome
- Antley-Bixler syndrome
- Pyogenic arthritis-pyoderma gangrenosum-acne syndrome
- Cerebellar ataxia-areflexia-pes cavus-optic atrophy-sensorineural hearing loss syndrome

- Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to WWOX deficiency
- Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome
- Ataxia-intellectual disability-oculomotor apraxia-cerebellar cysts syndrome
- Spinal muscular atrophy-progressive myoclonic epilepsy syndrome
- Autosomal dominant optic atrophy plus syndrome
- Optic atrophy-intellectual disability syndrome
- Barth syndrome
- Bartter syndrome
- Beta-thalassemia-X-linked thrombocytopenia syndrome
- Björnstad syndrome
- Blau syndrome
- Bohring-Opitz syndrome
- Borjeson-Forssman-Lehmann syndrome
- Bosley-Salih-Alorainy syndrome
- Bruck syndrome
- Brugada syndrome
- Carney-Stratakis syndrome
- Carvajal syndrome
- Congenital cataract-progressive muscular hypotonia-hearing loss-developmental delay syndrome
- Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome
- Chédiak-Higashi syndrome
- Christianson syndrome
- Chudley-McCullough syndrome
- Cockayne syndrome
- Coffin-Lowry syndrome
- Atrial septal defect-atrioventricular conduction defects syndrome
- Lethal congenital contracture syndrome type 1
- Autosomal recessive chorioretinopathy-microcephaly syndrome
- Cornelia de Lange syndrome
- Costello syndrome
- Recurrent metabolic encephalomyopathic crises-rhabdomyolysis-cardiac arrhythmia-intellectual disability syndrome
- Crouzon syndrome-acanthosis nigricans syndrome
- De Barsy syndrome
- DEND syndrome
- Denys-Drash syndrome
- Mitochondrial DNA depletion syndrome, encephalomyopathic form
- Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency
- Acral peeling skin syndrome
- Dysequilibrium syndrome
- Cognitive impairment-coarse facies-heart defects-obesity-pulmonary involvement-short stature-skeletal dysplasia syndrome
- TBCK-related intellectual disability syndrome
- Severe intellectual disability-progressive spastic diplegia syndrome
- X-linked intellectual disability-cerebellar hypoplasia syndrome
- X-linked intellectual disability-hypotonia-movement disorder syndrome
- X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome
- X-linked intellectual disability-psychosis-macroorchidism syndrome
- Intellectual disability-expressive aphasia-facial dysmorphism syndrome
- Intellectual disability-cataracts-calcified pinnae-myopathy syndrome
- Intellectual disability-seizures-hypophosphatasia-ophthalmic-skeletal anomalies syndrome
- Intellectual disability-macrocephaly-hypotonia-behavioral abnormalities syndrome

- Intellectual disability-severe speech delay-mild dysmorphism syndrome
- CNTNAP2-related developmental and epileptic encephalopathy
- Spondylometaphyseal dysplasia-cone-rod dystrophy syndrome
- Corneal dystrophy-perceptive deafness syndrome
- Dravet syndrome
- Dyggve-Melchior-Clausen disease
- Hypermobile Ehlers-Danlos syndrome
- Periodontal Ehlers-Danlos syndrome
- Neonatal encephalomyopathy-cardiomyopathy-respiratory distress syndrome
- Progressive epilepsy-intellectual disability syndrome, Finnish type
- Gingival fibromatosis-hypertrichosis syndrome
- Bloom's Syndrome
- Gerstmann-Straussler-Scheinker syndrome
- Hermansky-Pudlak syndrome due to BLOC-3 deficiency
- Hydrops-lactic acidosis-sideroblastic anemia-multisystemic failure syndrome
- Autosomal dominant hyper-IgE syndrome
- Hyperinsulinism-hyperammonemia syndrome
- Hypoplastic pancreas-intestinal atresia-hypoplastic gallbladder syndrome
- Hypotonia-speech impairment-severe cognitive delay syndrome
- Hutchinson-Gilford progeria syndrome
- Ichthyosis-prematurity syndrome
- Early-onset seizures-distal limb anomalies-facial dysmorphism-global developmental delay syndrome
- Partial androgen insensitivity syndrome
- Jackson-Weiss syndrome
- Johanson-Blizzard syndrome
- Joubert syndrome with ocular defect
- Multiple mitochondrial dysfunctions syndrome type 4
- Spondyloperipheral dysplasia-short ulna syndrome
- Corneal intraepithelial dyskeratosis-palmoplantar hyperkeratosis-laryngeal dyskeratosis syndrome
- Donnai-Barrow syndrome
- Dubin-Johnson syndrome
- Cardiac-valvular Ehlers-Danlos syndrome
- Musculocontractural Ehlers-Danlos syndrome
- Vascular Ehlers-Danlos syndrome
- Interstitial lung disease-nephrotic syndrome-epidermolysis bullosa syndrome
- Female restricted epilepsy with intellectual disability
- Floating-Harbor syndrome
- Frasier syndrome
- Gitelman syndrome
- Hermansky-Pudlak syndrome due to BLOC-2 deficiency
- Hyper-IgM syndrome with susceptibility to opportunistic infections
- Hyperphosphatasia-intellectual disability syndrome
- Hypohidrosis-enamel hypoplasia-palmoplantar keratoderma-intellectual disability syndrome
- Pancreatic hypoplasia-diabetes-congenital heart disease syndrome
- Holt-Oram syndrome
- Ichthyosis follicularis-alopecia-photophobia syndrome
- Imerslund-Gräsbeck syndrome
- Complete androgen insensitivity syndrome
- Acute infantile liver failure-multisystemic involvement syndrome
- Jeune syndrome
- Joubert syndrome with hepatic defect
- Joubert syndrome with oculorenal defect

- Kabuki syndrome
- Hypoxanthine guanine phosphoribosyltransferase partial deficiency
- Stiff skin syndrome
- Leigh syndrome
- Leigh syndrome with nephrotic syndrome
- Lesch-Nyhan syndrome
- Leukoencephalopathy with brain stem and spinal cord involvement-high lactate syndrome
- Leukoencephalopathy-thalamus and brainstem anomalies-high lactate syndrome
- Leukoencephalopathy-dystonia-motor neuropathy syndrome
- Lissencephaly syndrome, Norman-Roberts type
- Loeys-Dietz syndrome
- Macrocephaly-intellectual disability-autism syndrome
- Macrocephaly-intellectual disability-left ventricular non compaction syndrome
- Macrothrombocytopenia-lymphedema-developmental delay-facial dysmorphism-camptodactyly syndrome
- Lethal fetal brain malformation-duodenal atresia-bilateral renal hypoplasia syndrome
- 3MC syndrome
- Marfan syndrome
- Marinesco Sjogren syndrome
- Marshall syndrome
- McCune-Albright syndrome
- McKusick-Kaufman syndrome
- Meacham syndrome
- Goldberg-Shprintzen megacolon syndrome
- Megalencephaly-severe kyphoscoliosis-overgrowth syndrome
- Megalencephaly-capillary malformation-polymicrogyria syndrome
- Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome
- Familial atypical multiple mole melanoma syndrome
- Congenital microcephaly-severe encephalopathy-progressive cerebral atrophy syndrome
- Postnatal microcephaly-infantile hypotonia-spastic diplegia-dysarthria-intellectual disability syndrome
- Macrocephaly-intellectual disability-neurodevelopmental disorder-small thorax syndrome
- Microcephaly-corpus callosum hypoplasia-intellectual disability-facial dysmorphism syndrome
- Microcephaly-lymphedema-chorioretinopathy syndrome
- Microcephaly-capillary malformation syndrome
- 5q14.3 microdeletion syndrome
- Colobomatous microphthalmia-rhizomelic dysplasia syndrome
- Action myoclonus-renal failure syndrome
- Early-onset myopathy-areflexia-respiratory distress-dysphagia syndrome
- Mohr-Tranebjaerg syndrome
- Mowat-Wilson syndrome
- Muckle-Wells syndrome
- Muir-Torre syndrome
- Mulibrey nanism
- Myhre syndrome
- Nager syndrome
- Nance-Horan syndrome
- Netherton syndrome
- Peripheral neuropathy-myopathy-hoarseness-hearing loss syndrome
- Noonan syndrome with multiple lentigines
- Omenn syndrome
- Opitz GBBB syndrome

- Ear-patella-short stature syndrome
- Osteoporosis-pseudoglioma syndrome
- Early-onset parkinsonism-intellectual disability syndrome
- Perry syndrome
- Peutz-Jeghers syndrome
- Pierson syndrome
- Short rib-polydactyly syndrome, Majewski type
- Autosomal recessive multiple pterygium syndrome
- Familial short QT syndrome
- Resistance to thyrotropin-releasing hormone syndrome
- Retinitis pigmentosa-juvenile cataract-short stature-intellectual disability syndrome
- Global developmental delay-neuro-ophthalmological abnormalities-seizures-intellectual disability syndrome
- Autosomal dominant Robinow syndrome
- Rotor syndrome
- Schinzel-Giedion syndrome
- Senior-Boichis syndrome
- Shprintzen-Goldberg syndrome
- Simpson-Golabi-Behmel syndrome
- Smith-Lemli-Opitz syndrome
- Stickler syndrome
- Short stature-pituitary and cerebellar defects-small sella turcica syndrome
- Spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome
- Arterial tortuosity syndrome
- Noonan syndrome-like disorder with loose anagen hair
- Vici syndrome
- Wiedemann-Steiner syndrome
- Wolcott-Rallison syndrome
- Carney complex-trismus-pseudocamptodactyly syndrome
- Osteopathia striata-cranial sclerosis syndrome
- Pancytopenia-developmental delay syndrome
- Pendred syndrome
- Peters plus syndrome
- Pfeiffer syndrome
- Pitt-Hopkins syndrome
- Serrated polyposis syndrome
- Autosomal dominant popliteal pterygium syndrome
- Palmoplantar keratoderma-deafness syndrome
- Insulin-resistance syndrome type A
- Growth and developmental delay-hypotonia-vision impairment-lactic acidosis syndrome
- Rett syndrome
- Rothmund-Thomson syndrome
- Rubinstein-Taybi syndrome
- Scott syndrome
- Sheldon-Hall syndrome
- Shwachman-Diamond syndrome
- Sjögren Larsson syndrome
- Steel syndrome
- Short stature-brachydactyly-obesity-global developmental delay syndrome
- Tatton-Brown-Rahman syndrome
- Toriello-Lacassie-Droste syndrome
- Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-skeletal anomalies syndrome
- Renal tubulopathy-encephalopathy-liver failure syndrome
- Wiedemann-Rautenstrauch syndrome
- Wiskott-Aldrich syndrome
- Wolfram syndrome
- Isolated cloverleaf skull syndrome

- Occipital horn syndrome
- Linear nevus sebaceus syndrome
- Neurogenic scapuloperoneal syndrome, Kaeser type
- Familial hyperphosphatemic tumoral calcinosis/Hyperphosphatemic hyperostosis syndrome
- Atypical hemolytic uremic syndrome
- KID syndrome
- MASA syndrome
- Micro syndrome
- Nephrogenic syndrome of inappropriate antidiuresis
- PRUNE1-related neurological syndrome
- Oculocerebrorenal syndrome of Lowe
- Orofaciodigital syndrome type 4
- Otopalatodigital syndrome type 2
- RAPADILINO syndrome
- Congenital intrauterine infection-like syndrome
- Wolfram-like syndrome
- Triple A syndrome
- Sitosterolemia
- Short stature due to GHSR deficiency
- Catecholaminergic polymorphic ventricular tachycardia
- Tyrosinemia type 1
- TELO2-related intellectual disability-neurodevelopmental disorder
- ITPA-related lethal infantile neurological disorder with cataract and cardiac involvement
- Noonan syndrome-like disorder with juvenile myelomonocytic leukemia
- Carney triad
- Glanzmann thrombasthenia
- Paris-Trousseau thrombocytopenia
- Lateral meningocele syndrome
- EEC syndrome
- Enamel-renal syndrome
- H syndrome
- Hydrolethalus
- Lacrimoauriculodentodigital syndrome
- MEGDEL syndrome
- Multisystemic smooth muscle dysfunction syndrome
- Congenital nephrotic syndrome, Finnish type
- Oculocerebrofacial syndrome, Kaufman type
- Orofaciodigital syndrome type 14
- Orofaciodigital syndrome type 5
- Tumor necrosis factor receptor 1 associated periodic syndrome
- SHORT syndrome
- NPHP3-related Meckel-like syndrome
- Larsen-like syndrome, B3GAT3 type
- Spondylocarpotarsal synostosis
- Deafness with labyrinthine aplasia, microtia, and microdontia
- Microcephalic cortical malformations-short stature due to RTTN deficiency
- Hereditary hemorrhagic telangiectasia
- 46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency
- Lethal acantholytic erosive disorder
- Familial progressive cardiac conduction defect
- Nijmegen breakage syndrome-like disorder
- Severe primary trimethylaminuria
- Congenital amegakaryocytic thrombocytopenia
- Severe hereditary thrombophilia due to congenital protein C deficiency

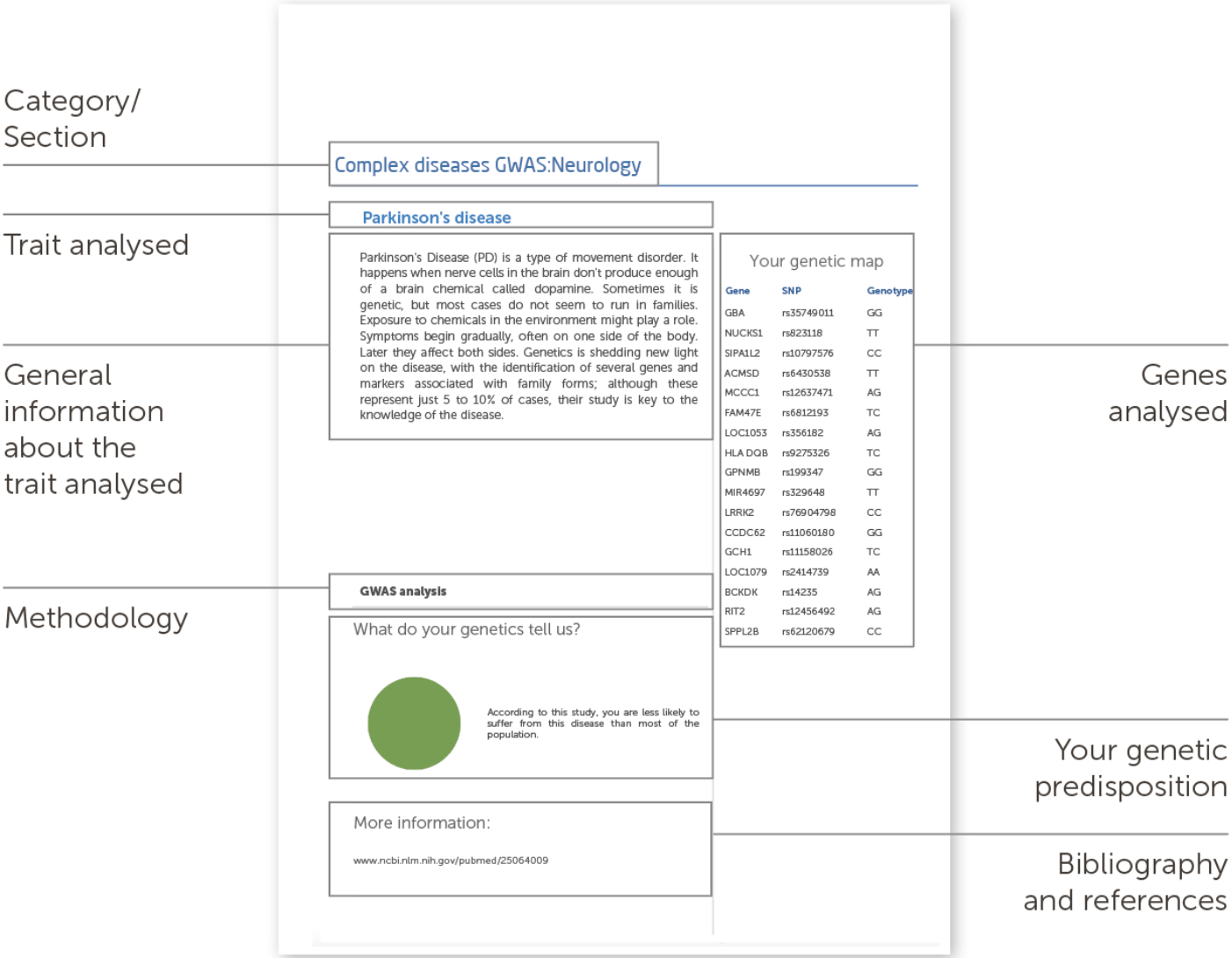
- Hereditary thrombophilia due to congenital antithrombin deficiency
- Familial cold urticaria
- STING-associated vasculopathy with onset in infancy
- Cerebrotendinous xanthomatosis
- Desmoid tumor
- Vasculitis due to ADA2 deficiency
- Hereditary xanthinuria
- Xeroderma pigmentosum

Caption:

- We have not detected any pathogenic mutation but, since we only analyze a part of the gene, you could have some pathogenic mutation in the non-analyzed genetic regions.
- We have detected at least one mutation that could be pathogenic.

3. Genetic Results

3.1. How to understand your report?



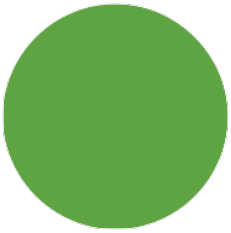
GWAS Complex Diseases: Neurology

Parkinson's disease

Parkinson's Disease (PD) is a type of movement disorder. It happens when nerve cells in the brain don't produce enough of a brain chemical called dopamine. Sometimes it is genetic, but most cases do not seem to run in families. Exposure to chemicals in the environment might play a role. Symptoms begin gradually, often on one side of the body. Later they affect both sides. Genetics is shedding new light on the disease, with the identification of several genes and markers associated with family forms; although these represent just 5 to 10% of cases, their study is key to the knowledge of the disease.

GWAS analysis

What do your genetics tell us?



According to this study, you are less likely to suffer from this disease than most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/25064009

Your genetic map

Gene	SNP	Genotype
GBA	rs35749011	GG
NUCKS1	rs823118	CC
SIPA1L2	rs10797576	CC
ACMSD	rs6430538	TC
MCCC1	rs12637471	AG
FAM47E	rs6812193	CC
LOC1053	rs356182	AA
HLA DQB	rs9275326	CC
GPNMB	rs199347	AG
MIR4697	rs329648	TC
LRRK2	rs76904798	CC
CCDC62	rs11060180	AG
GCH1	rs11158026	CC
LOC1079	rs2414739	AG
BCKDK	rs14235	GG
RIT2	rs12456492	AA
SPPL2B	rs62120679	TC

GWAS Complex Diseases: Neurology

Intracranial aneurysm

A brain aneurysm is an abnormal bulge or "ballooning" in the wall of an artery in the brain. They are sometimes called "berry aneurysms" because they are often the size of a small berry. Most brain aneurysms produce no symptoms until they become large, begin to leak blood, or burst.

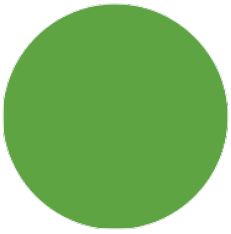
If a brain aneurysm presses on nerves in your brain, it can cause signs and symptoms.

Your genetic map

Gene	SNP	Genotype
RP1	rs9298506	AA
CDKN2B	rs1333040	CC
CNNM2	rs12413409	GG
STARD13	rs9315204	CC
RBBP8	rs11661542	AA

GWAS analysis

What do your genetics tell us?



According to this study, you are less likely to suffer from this disease than most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/20364137

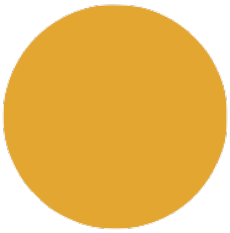
GWAS Complex Diseases: Neurology

Motion sickness

Motion sickness is a common problem in people traveling by car, train, airplanes and boats, especially. Anyone can suffer it, but it is more common in children, pregnant women, and people taking certain medicines. Motion sickness can start suddenly, causing a queasy feeling and cold sweats. It can then lead to dizziness, nausea and vomiting. Your brain senses movement by getting signals from your inner ears, eyes, muscles, and joints. When it receives signals that do not match, you can suffer from motion sickness. For example, if you are reading on your phone while riding a bus, your eyes are focused on something that is not moving, but your inner ear senses motion. Despite its high heritability, no associated genetic factors have been discovered. This section is based on a genome association study on motion sickness in 80,494 individuals who were surveyed about this pathology.

GWAS analysis

What do your genetics tell us?



According to this study, you are more likely to suffer from this disease than most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/25628336

Your genetic map

Gene	SNP	Genotype
PVRL3	rs66800491	AG
GPD2	rs56051278	AG
LINC0124	rs10970305	AC
AUTS2	rs1195218	GG
LINC026	rs705145	AA
CBLN4	rs6069325	TT
MUTED	rs2153535	GC
LINGO2	rs2150864	AG
CPNE4	rs9834560	AA
LOC1019	rs1858111	AG
PRDM16	rs61759167	TT
NLGN1	rs11713169	AC
HOXD3	rs2551802	GG
COPS8	rs2318131	AC
TLE4	rs149951341	AA
HOXB3	rs9906289	CC
ST18	rs2360806	AA
SDK1	rs4343996	AG
LINC009	rs7170668	TC
CELF2	rs10752212	AG
PDZRN4	rs7957589	AA
MCTP2	rs62018380	CC
ARAP2	rs6833641	CC
AUTS2	rs6946969	AG
RGS5	rs4076764	TT
MAP2K5	rs997295	TT
AGA	rs1378552	CC
POU6F2	rs60464047	AT
LINC0124	rs1782032	AG
GXYLT2	rs1847202	TT
SDK1	rs34912216	AG

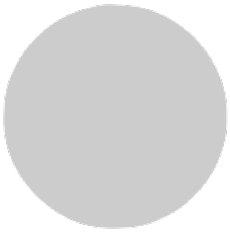
GWAS Complex Diseases: Neurology

Alzheimer's disease (late onset)

Alzheimer's Disease (AD) is the most common form of dementia among older people. Dementia is a brain disorder that seriously affects a person's ability to carry out daily activities. AD begins slowly. It first involves the parts of the brain that control thought, memory and language. People with AD may have trouble remembering things that happened recently, or names of people they know. A related problem, Mild Cognitive Impairment (MCI), causes more memory problems than normal for people of the same age. Many, but not all, people with MCI will develop AD. This section analyses the predisposition to Late-Onset Alzheimer's.

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/24162737

Your genetic map

Gene	SNP	Genotype
CR1	rs6656401	GG
LOC1053	rs6733839	TC
CD2AP	rs10948363	AG
EPHA1	rs11771145	AG
CLU	rs9331896	TT
MS4A6A	rs983392	AA
PICALM	rs10792832	GG
INPP5D	rs35349669	TC
MEF2C	rs190982	AG
NME8	rs2718058	AG
ZCWPW1	rs1476679	TT
CELF1	rs10838725	TC
FERMT2	rs17125944	TT
CASS4	rs7274581	TT
HLA	rs9271192	AC
PTK2B	rs28834970	TC
SORL1	rs11218343	TT
SLC24A4	rs10498633	TT
SQSTM1	rs72807343	CC
LOC1079	rs9381040	CC
CD33	rs3865444	AC

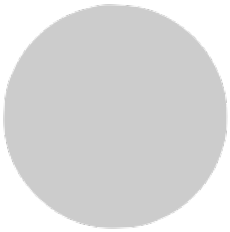
GWAS Complex Diseases: Neurology

Multiple sclerosis

Multiple Sclerosis (MS) is a nervous system disease that affects your brain and spinal cord. It damages the myelin sheath, the material that surrounds and protects your nerve cells. This damage slows down or blocks messages between your brain and your body, leading to the symptoms of MS. These can include: visual disturbances, muscle weakness, trouble with coordination and balance, sensations such as numbness, prickling, "pins and needles", and thinking and memory problems. No one knows what causes MS. It may be an autoimmune disease, which happens when your immune system attacks healthy cells in your body by mistake. Multiple Sclerosis affects women more than men. It often begins between the ages of 20 and 40. Epidemiological studies show that genetic factors are responsible for its occurrence, which explains the higher frequency of the disease in the relatives of affected people.

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/21833088

Your genetic map

Gene	SNP	Genotype
AGAP2	rs12368653	AG
AHI1	rs11154801	CC
BACH2	rs12212193	AG
BATF	rs2300603	TC
INAVA	rs7522462	AA
TIMMDC1	rs2293370	AA
LOC1053	rs650258	TC
CD58	rs1335532	AA
CD86	rs9282641	GG
CHST12	rs6952809	TT
CLECL1P	rs10466829	GG
CXCR5	rs630923	CC
CYP24A1	rs2248359	TT
DDAH1	rs233100	GG
DKKL1	rs2303759	TG
DLEU1	rs806321	TC
EOMES	rs11129295	TC
EVI5	rs11810217	TT
VCAM1	rs12048904	TT
FCRL3	rs3761959	CC
LINC0114	rs2119704	CC
HHEX	rs7923837	GG
IL12A	rs2243123	TT
LOC2856	rs2546890	AA
IL22RA2	rs17066096	AG
IL7R	rs6897932	CC
IRF8	rs13333054	CC
MALT1	rs7238078	TT
MAMSTR	rs281380	CC
MAPK1	rs2283792	TT
MERTK	rs17174870	CC

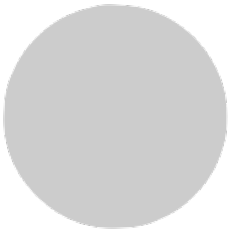
GWAS Complex Diseases: Neurology

Schizophrenia

Schizophrenia is a serious brain illness. People who have it may hear voices that aren't there. They may think other people are trying to hurt them. Sometimes they don't make sense when they talk. The disorder makes it hard for them to keep a job or take care of themselves. Symptoms of schizophrenia usually start between ages 16 and 30. Men often develop symptoms at a younger age than women. People usually do not develop schizophrenia after age 45.

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/25056061

Your genetic map

Gene	SNP	Genotype
PLCH2	rs4648845	CC
KDM4A	rs11210892	AA
LOC1053	rs12129573	CC
MIR137H	rs1702294	CC
FAM5B	rs6670165	CC
MIR29B2	rs7523273	AA
AKT3	rs77149735	GG
FANCL	rs11682175	TC
CYP26B1	rs3768644	GG
PCGEM1	rs59979824	CC
SATB2	rs6704641	AA
GIGYF2	rs6704768	AA
CNTN4	rs17194490	GG
TRANK1	rs75968099	CC
THOC7	rs832187	TT
STAG1	rs7432375	GG
CLCN3	rs10520163	TT
GPM6A	rs1106568	AA
HCN1	rs1501357	TC
LINC020	rs4391122	AA
MEF2C	rs16867576	AG
MAN2A1	rs4388249	CC
ETF1	rs3849046	TC
GALNT10	rs11740474	TT
RIMS1	rs1339227	CC
FUT9	rs117074560	CC
GRM3	rs12704290	GG
SRPK2	rs6466055	AA
IMMP2L	rs13240464	TC
PODXL	rs7801375	GG
DGKI	rs3735025	TC

GWAS Complex Diseases: Neurology

Neuroblastoma

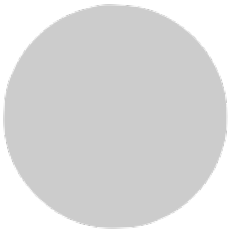
Neuroblastoma is a cancer that forms in your nerve tissue. It usually begins in the adrenal glands, located above your kidneys. It may also begin in the neck, chest or spinal cord. The cancer often begins in early childhood. Sometimes it begins before a child is born. By the time doctors find the cancer, it has usually spread to other parts of the body.

Your genetic map

Gene	SNP	Genotype
HACE1	rs4336470	TC
LIN28B	rs17065417	AA
BARD1	rs7587476	CC
CASC15	rs9295536	AC
LMO1	rs110419	AG
HSD17B1	rs11037575	TT

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/22941191

GWAS Complex Diseases: Neurology

Conduct disorder

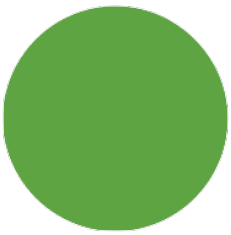
Behavioural disorder is one of the most prevalent psychiatric disorders in children. The related symptoms have an important genetic component, whose heritability is estimated at 50%, and include aggression, rule-breaking, the harassment of other children, robberies, violence, etc. This disorder is a risk factor for future addictive behaviour. Different genetic variants have been associated with the risk of onset of this disorder.

Your genetic map

Gene	SNP	Genotype
C1QTNF7	rs16891867	AA
PDE10A	rs7762160	TC
TOX2	rs6031252	CC
ERCC4	rs3136202	AG
LOC1053	rs4434872	CC
ARHGAP2	rs10776612	CC
Intergeni	rs7950811	CC
LINC003	rs11838918	TT
Intergeni	rs1256531	AA
LOC1079	rs4792394	AC
Intergeni	rs13398848	AA
Intergeni	rs2184898	GG
RNF150	rs1550057	AA
CC2D2A	rs1861050	CC

GWAS analysis

What do your genetics tell us?



According to this study, you are less likely to suffer from this disease than most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/20585324

GWAS Complex Diseases: Neurology

Glioma

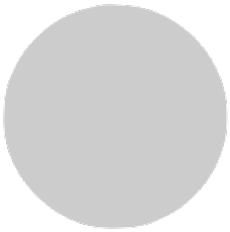
Glioma is a type of neoplasm that occurs in the brain or spinal cord. It is called glioma because it arises from glial cells. Its most frequent location is the brain.

Your genetic map

Gene	SNP	Genotype
TERT	rs2736100	AC
TERT	rs2853676	CC
CCDC26	rs891835	TG
CCDC26	rs4295627	TT
CDKN2B	rs4977756	AG
PHLDB1	rs498872	GG
RTEL1	rs6010620	GG

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/19578367

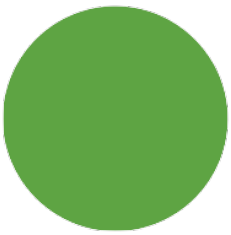
GWAS Complex Diseases: Circulatory System

Primary biliary cirrhosis

The bile ducts are tubes that move bile from the liver to the small intestine. Bile is a substance that facilitates digestion. All of the bile ducts together are called the biliary tract. When the bile ducts become swollen or inflamed, it blocks the flow of bile. The buildup of bile damages the liver cells and leads to scarring of the liver, called cirrhosis. This is called biliary cirrhosis.
Genetic susceptibility has been suggested, as well as the influence of environmental factors (infections, smoking, exposure to chemicals).

GWAS analysis

What do your genetics tell us?



According to this study, you are less likely to suffer from this disease than most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/21399635

Your genetic map

Gene	SNP	Genotype
DENND1	rs12134279	CC
NAB1	rs10931468	CC
TIMMDC1	rs2293370	AA
NFKB1	rs7665090	AG
IL7R	rs860413	AA
ELMO1	rs6974491	GG
CXCR5	rs6421571	CC
TNFRSF1	rs1800693	TT
RAD51B	rs911263	TC
CLEC16A	rs12924729	GG
Intergeni	rs11117432	AG
MAP3K7I	rs968451	GG
LINC0110	rs485499	TC
MHC	rs7774434	TC
TNPO3	rs12531711	AA
FBXL20	rs7208487	TG
SPIB	rs3745516	GG
PLCL2	rs1372072	AG
RPS6KA4	rs538147	GG
EXOC3L4	rs8017161	AG

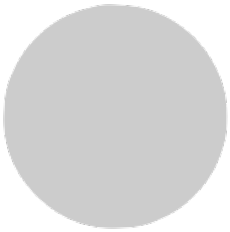
GWAS Complex Diseases: Circulatory System

Coronary heart disease

Coronary Heart Disease is a narrowing of the small blood vessels that supply blood and oxygen to the heart. Coronary Heart Disease (CHD) is also called coronary artery disease. CHD is the leading cause of death in the United States for men and women. CHD is caused by the buildup of plaque in the arteries to your heart. This may also be called "hardening of the arteries". Fatty material and other substances form a plaque buildup on the walls of your coronary arteries. The coronary arteries carry blood and oxygen to your heart.
This buildup causes the arteries to narrow. As a result, blood flow to the heart can slow down or stop.

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/21378990

Your genetic map

Gene	SNP	Genotype
PCSK9	rs11206510	TC
CXCL12	rs1746048	CC
PLPP3	rs17114036	AA
ANKS1A	rs17609940	GG
ZC3HC1	rs11556924	TT
ABO	rs579459	TC
CNNM2	rs12413409	GG
ZPR1	rs964184	GC
COL4A1	rs4773144	AA
HHIPL1	rs2895811	TC
ADAMTS7	rs3825807	AG
SMG6	rs216172	GG
RASD1	rs12936587	AG
UBE2Z	rs46522	TT
MIA3	rs17465637	AC
WDR12	rs6725887	TT
MRAS	rs2306374	TC
LPA	rs3798220	TT
CDKN2B	rs4977574	AG
SH2B3	rs3184504	CC
SMARCA	rs1122608	GG
SLC5A3	rs9982601	CC
INPP5D	rs10933436	AC
BTD	rs7651039	TC
ASZ1	rs7808424	TT
SMG6	rs1231206	AG

GWAS Complex Diseases: Circulatory System

Myocardial infarction (early onset)

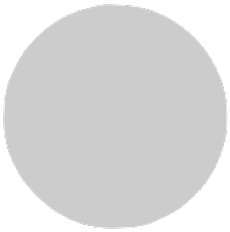
Myocardial infarction has a hereditary component and is among the leading causes of death and disability worldwide. While most cases occur in individuals older than 65, 5-10% occur in younger patients (men under 50 and women under 60). These cases are associated with a substantially greater heritability, so it is important to identify the genes responsible. A large-scale association study has found several genetic variants that increase the risk of early onset myocardial infarction.

Your genetic map

Gene	SNP	Genotype
CDKN2B	rs4977574	AG
CELSR2	rs646776	TT
MIA3	rs17465637	AC
CXCL12	rs1746048	CC
SLC5A3	rs9982601	CC
WDR12	rs6725887	TT
SMARCA	rs1122608	GG
PCSK9	rs11206510	TC

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/19198609

GWAS Complex Diseases: Circulatory System

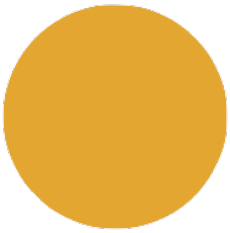
Chronic lymphocytic leukemia

Leucemia is cancer of the white blood cells. White blood cells help your body fight infection. Your blood cells form in your bone marrow. In leucemia, the bone marrow produces abnormal white blood cells. These cells crowd out the healthy blood cells, making it hard for blood to do its work. In Chronic Lymphocytic Leucemia (CLL), there are too many lymphocytes, a type of white blood cell.

CLL is the second most common type of leucemia in adults. It often occurs during or after middle age, and is rare in children.

GWAS analysis

What do your genetics tell us?



According to this study, you are more likely to suffer from this disease than most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/23770605

Your genetic map

Gene	SNP	Genotype
ACOXL	rs17483466	AG
SP110	rs13397985	TG
FARP2	rs757978	CC
IRF4	rs872071	AG
HLA	rs9273363	AA
BAK1	rs210142	CC
CASC19	rs2466035	TT
GRAMD1	rs735665	GG
LOC1053	rs11636802	AA
RPLP1	rs7176508	AA
IRF8	rs391023	TC
BCL2	rs4987852	TT
FAS	rs4406737	GG
BCL2	rs4987855	CC
TSPAN32	rs7944004	TG
LEF1	rs898518	AA
CASP8	rs3769825	AG
AS1	rs1679013	TC
PMAIP1	rs4368253	TC
ACOXL	rs13401811	AG
ODF1	rs2511714	GG

GWAS Complex Diseases: Circulatory System

Hodgkin's lymphoma

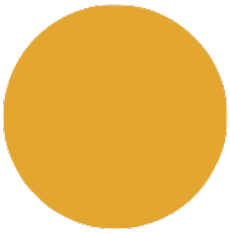
Hodgkin Lymphoma is a cancer of the lymphatic system produced by the germ cells of the B lymphocytes (defensive cells of the immune system). The incidence in our country is 30 new cases per million inhabitants per year. It features a bimodal distribution, affecting either the young, ages 15 to 35, or those well over 55. 60-70% of patients are asymptomatic, and cases are usually detected due to an increase in the volume of the lymph nodes. 45-60% of cases are associated with an Epstein-Barr virus infection.

Your genetic map

Gene	SNP	Genotype
EOMES	rs3806624	GG
HBS1L	rs7745098	TT
NR	rs1432295	GG
GATA3	rs501764	TG
PVT1	rs2019960	TT
NR	rs6903608	TT

GWAS analysis

What do your genetics tell us?



According to this study, you are more likely to suffer from this disease than most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/24149102

GWAS Complex Diseases: Circulatory System

Diffuse large B cell lymphoma

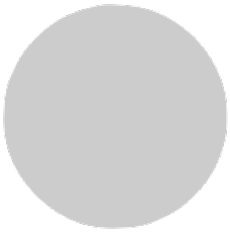
Diffuse Large B-cell Lymphoma (DLBCL) is a clinically aggressive B-cell (immune system) cancer and is the most common non-Hodgkin lymphoma. In some European countries the incidence of non-Hodgkin lymphoma is estimated at 12.3 cases per 100,000/year in men, whereas in women it is 10.8 cases. It is a disease of the elderly, with an average diagnosis age of around 70. Diagnosis in the early stages may improve prognosis. Family history is a risk factor.

Your genetic map

Gene	SNP	Genotype
NCOA1	rs79480871	CC
HLA B	rs2523607	TT
PVT1	rs13255292	TC
MYC	rs4733601	AA

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/25261932

GWAS Complex Diseases: Circulatory System

Follicular lymphoma

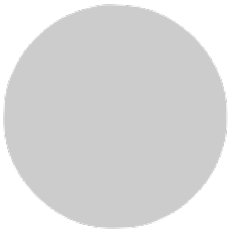
Follicular lymphoma is a form of non-Hodgkin lymphoma that is characterised by a proliferation of B cells with the nodular structure of the follicular architecture being preserved. The prevalence of follicular lymphoma is estimated at about 1/3,000. The average diagnosis age is 60 -65. The disease is extremely rare in children. Follicular lymphoma is found mainly in lymph nodes, but can also affect the spleen, bone marrow, peripheral blood and Waldeyer's ring. In exceptional cases the skin and central nervous system are affected.

Your genetic map

Gene	SNP	Genotype
HLA	rs12195582	CC
CXCR5	rs4938573	TT
LOC1053	rs4937362	TC
LPP	rs6444305	AG
BCL2	rs17749561	GG
PVT1	rs13254990	CC
SLC14A2	rs11082438	GG

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/25279986

GWAS Complex Diseases: Circulatory System

Wilms tumor

Wilms Tumour is a rare type of kidney cancer. It causes a tumor on one or both kidneys. It usually affects children, but can occur in adults. Having certain genetic conditions, or birth defects, can increase the risk of contracting it. Children that are at risk should be screened for Wilms tumor every three months until they turn eight.

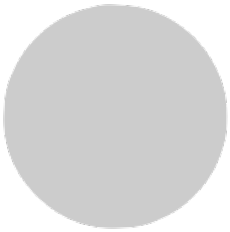
Symptoms include a lump in the abdomen, blood in the urine, and a fever for no reason. Tests that examine the kidney and blood are used to find the tumor.

Your genetic map

Gene	SNP	Genotype
DDX1	rs3755132	TT
LOC1053	rs1027643	TC
DLG2	rs790356	AG
TCN2	rs2283873	GG
NHS	rs5955543	AA
MYCN	rs807624	TG

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/22544364

GWAS Complex Diseases: Respiratory System

Upper aerodigestive tract cancers

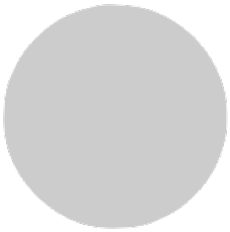
Cancer of the upper aerodigestive tract includes tumours of the oral cavity, pharynx, larynx, nasal cavity and paranasal sinuses, ear and salivary glands. Head and neck carcinoma is the most common among them, and has a high mortality rate (in Spain it is 37%). Alcohol and tobacco use are the main risk factors, although the human papilloma virus infection and family history also play an important role. A large-scale genetic association study has found genetic variants that increase risk of the disease.

Your genetic map

Gene	SNP	Genotype
ADH1B	rs1229984	CC
ADH7	rs971074	CC
HELQ	rs1494961	TC
NAA25	rs4767364	GG

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/21437268

GWAS Complex Diseases: Respiratory System

Chronic bronchitis and chronic obstructive pulmonary disease

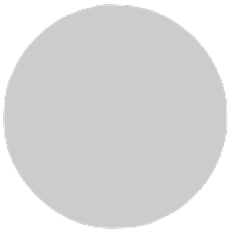
Chronic Obstructive Pulmonary Disease (COPD) is a common lung disease. Having COPD makes it hard to breathe.
There are two main forms of COPD: Chronic bronchitis, which involves a long-term cough with mucus; and Emphysema, which involves damage to the lungs over timeMost people with COPD have a combination of both conditions. Smoking is the main cause of COPD. The more a person smokes, the more likely it is that he will develop COPD. However, some people smoke for years and never get COPD. In rare cases, non-smokers who lack a protein called alpha-1 antitrypsin can develop emphysema.

Your genetic map

Gene	SNP	Genotype
FAM13A	rs2869966	TC
IREB2	rs8042238	TC
FAM13A	rs2869967	TT
CD151	rs34391416	GG
HHIP AS1	rs13141641	TC
CHRNA3	rs12914385	TC
FAM13A	rs4416442	TT
CYS1	rs12692398	AA

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/25241909

GWAS Complex Diseases: Respiratory System

Asthma

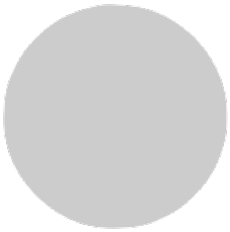
Asthma is a chronic inflammatory disease that affects the lungs, causing reversible airflow obstruction, bronchospasms, and other recurring and variable symptoms, including wheezing, coughing, chest tightness, and shortness of breath. Symptoms can occur several times a day or week and are often worse at night, first thing in the morning, or with exercise. We could say that the disease is always there when you have asthma, but you only have crises when something affects your lungs. Environmental factors, such as exposure to allergens and pollutants, significantly influence asthma, but genetics also play a crucial role in its development. Specific variants in genes, such as TSBP1-AS1 and LOC105369781, are associated with a greater genetic predisposition to suffer from asthma.

Your genetic map

Gene	SNP	Genotype
HLA	rs7775228	TT
GAB1	rs3805236	GG
LOC1053	rs1701704	TG
NOTCH4	rs404860	TT
PBX2	rs204993	AA
TSBP1	rs3117098	AG
TSBP1	rs3129943	AG
Unknown	rs9500927	AG
Unknown	rs9275698	AA
Unknown	rs7686660	TG
Unknown	rs3129890	CC
Unknown	rs1837253	CC
Unknown	rs10508372	GG

GWAS analysis

What do your genetics tell us?



According to this study, you have a predisposition similar to most of the population. Other genetic and clinical factors may influence.

More information:

https://pubmed.ncbi.nlm.nih.gov/21804548/

GWAS Complex Diseases: Musculoskeletal System

Systemic sclerosis

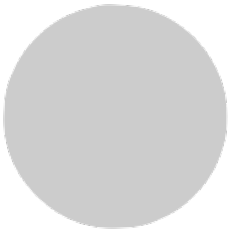
Systemic Sclerosis is a chronic autoimmune disease that causes an alteration of the collagen (protein of the connective tissue) and, as a consequence, the skin sclerosis; that is, it hardens. It can also affect other organs of the body such as the lungs, heart, kidneys, etc. although the part most often affected is the skin. The prognosis is highly variable from person to person. Exposure to certain toxic products (such as tobacco), excessive stress, exposure to cold, and some drugs can worsen the symptoms. It affects one in 50,000 people and is more common in middle-aged women. It is a rare disease of unknown, severely disabling origin. A large-scale study has found that different genetic variants are associated with the pathogenesis of the disease.

Your genetic map

Gene	SNP	Genotype
PSORS1C	rs3130573	GG
HLA	rs6457617	CC
LOC1079	rs13021401	CC
TNIP1	rs2233287	GG
CD247	rs2056626	TG
STAT4	rs7574865	GG
TNPO3	rs10488631	TC

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/21750679

GWAS Complex Diseases: Musculoskeletal System

Osteosarcoma

Osteosarcoma is a very rare type of cancerous bone tumour that usually develops in teenagers. It often occurs when a teen is growing rapidly. Osteosarcoma is the most common bone cancer in children. The average age at diagnosis is 15. Boys and girls are just as likely to develop this tumour, until the late teens, when it occurs more often in boys. Osteosarcoma is also common in people over age 60.

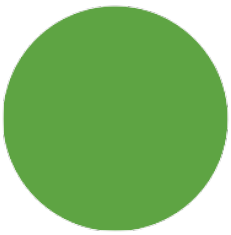
The cause is not known. In some cases, osteosarcoma runs in families. At least one gene has been linked to an increased risk. This gene is also associated with familial retinoblastoma. This is a cancer of the eye that occurs in children.

Your genetic map

Gene	SNP	Genotype
GRM4	rs1906953	CC
AJ412031	rs573666	CC
Intergeni	rs7591996	AA
ADAMTS6	rs17206779	TT

GWAS analysis

What do your genetics tell us?



According to this study, you are less likely to suffer from this disease than most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/23727862

GWAS Complex Diseases: Musculoskeletal System

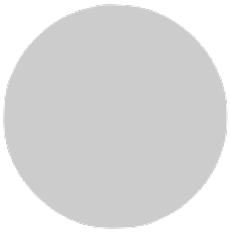
Rheumatoid arthritis

Rheumatoid Arthritis (RA) is a form of arthritis that causes pain, swelling, stiffness and a loss of function in your joints. It can affect any joint, but is common in the wrist and fingers.

More women than men suffer from rheumatoid arthritis. It often starts in middle age, and is most common in older people. You might have the disease for only a short time, or symptoms might come and go. The severe form can last a lifetime.

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/24390342

Your genetic map

Gene	SNP	Genotype
ACOXL	rs6732565	AG
LINC0110	rs9653442	TT
ANKRD55	rs7731626	AA
ARID5B	rs71508903	CC
ATG5	rs9372120	TG
BLK	rs2736337	TT
RABEP1	rs72634030	CC
C4orf52	rs11933540	TC
MACIR	rs2561477	GG
CCL21	rs11574914	AG
CD2	rs624988	CC
CD226	rs2469434	TT
CD28	rs1980422	TC
CD40	rs4239702	TC
CDK6	rs4272	AA
TYR	rs4409785	CC
FLACC1	rs6715284	CC
CLNK	rs13142500	TT
CTLA4	rs3087243	AA
RPP14	rs73081554	CC
EOMES	rs3806624	GG
ETS1	rs73013527	TC
FADS2	rs968567	CC
GRHL2	rs678347	AA
HLA	rs9268839	AG
STAG1	rs9826828	GG
CSF2 IL3	rs657075	GG
MECP2	rs5987194	GC
IRF8	rs13330176	TT
JAZF1	rs67250450	TC
LBH	rs10175798	GG

GWAS Complex Diseases: Musculoskeletal System

Multiple myeloma

Multiple myeloma is a cancer that begins in plasma cells, a type of white blood cell. These cells are part of your immune system, which helps protect the body from germs and other harmful substances. Over time myeloma cells collect in the bone marrow and in the solid parts of bones.

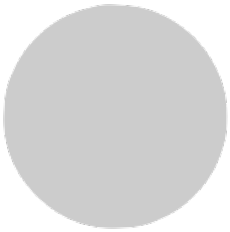
No one knows the exact causes of multiple myeloma, but it is more common in older people and African Americans. It can run in families.

Your genetic map

Gene	SNP	Genotype
MYNN	rs10936599	CC
PSORS1C	rs2285803	CC
MXI1	rs11195062	CC
TNFRSF1	rs4273077	AA
CBX7	rs877529	AG

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/23955597

GWAS Complex Diseases: Musculoskeletal System

Myasthenia gravis

Myasthenia gravis is a disease that causes weakness in the voluntary muscles. These are the muscles that you control. For example, you may suffer weakness in the muscles used for eye movement, facial expressions, and swallowing. You can also have weakness in other muscles. This weakness gets worse with activity, and better with rest.

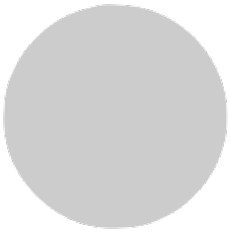
Myasthenia gravis is an autoimmune disease. Your body's immune system produces antibodies that block or alter some of the nerve signals to your muscles. This makes your muscles weaker.

Your genetic map

Gene	SNP	Genotype
PTPN22	rs2476601	GG
TNIP1	rs4958881	TC
LINC0112	rs6719884	AC
NR	rs3130544	CC

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/23055271

GWAS Complex Diseases: Endocrinology

Type 1 diabetes

Diabetes means your blood glucose, or blood sugar, levels are too high. With type-1 diabetes, your pancreas does not make insulin. Insulin is a hormone that helps your cells get energy from glucose. Without insulin, too much glucose remains in your blood. Over time, high blood glucose can lead to serious problems with your heart, eyes, kidneys, nerves, and gums and teeth.

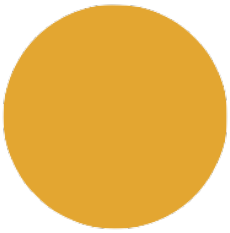
Type-1 diabetes happens most often in children and young adults, but can appear at any age.

Your genetic map

Gene	SNP	Genotype
BACH2	rs11755527	GG
LINC026	rs947474	AA
CTSH	rs3825932	TC
C1QTNF6	rs229541	AA
PHTF1	rs6679677	CC
CTLA4	rs3087243	AA
IL2RA	rs12251307	CC
NAA25	rs17696736	AA
ERBB3	rs2292239	GG
CLEC16A	rs12708716	AA
PTPN2	rs2542151	TT

GWAS analysis

What do your genetics tell us?



According to this study, you are more likely to suffer from this disease than most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/18978792

GWAS Complex Diseases: Endocrinology

Type 1 diabetes nephropathy

Type-1 Diabetes Mellitus (DM1) is an autoimmune and metabolic disease in which the pancreas does not produce insulin, resulting in elevated blood glucose levels. Type-1 diabetes occurs most frequently in children and young adults, and accounts for 13% of all cases of diabetes in countries like Spain, where the number of cases for children under 15 is 11.5-27.6 cases/100,000 inhabitants. Susceptibility to Type-1 diabetes mellitus appears to be associated with multiple genetic factors, although interaction with certain environmental factors (infections, diet ...) is required for the development of the disease.

Your genetic map

Gene	SNP	Genotype
LOC1079	rs12437854	TT
AFF3	rs7583877	TT
Intergeni	rs878889	GG
LINC0115	rs4871297	AA
RNF10	rs614226	CC
EFCAB8	rs13045180	TC

GWAS analysis

What do your genetics tell us?

According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/23028342

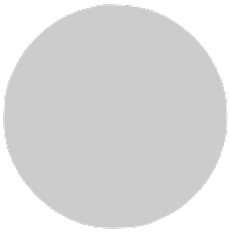
GWAS Complex Diseases: Endocrinology

Type 2 diabetes

Diabetes means your blood glucose, or blood sugar, levels are too high. With type-2 diabetes, the more common type, your body does not make or use insulin well. Insulin is a hormone that helps your cells get energy from glucose. Without insulin, too much glucose remains in your blood. Over time, high blood glucose can lead to serious problems with your heart, eyes, kidneys, nerves, and gums and teeth. You have a higher risk of type 2 diabetes if you are older, obese, have a family history of diabetes, or do not exercise. Having pre-diabetes also increases your risk. Prediabetes means that your blood sugar is higher than normal, but not high enough to be called diabetes.

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/24509480

Your genetic map

Gene	SNP	Genotype
RREB1	rs9502570	TT
FAF1	rs17106184	GG
POU5F1	rs3132524	CC
LOC1079	rs6808574	TC
ARL15	rs702634	AA
MPHOSP	rs1727313	GG
PLEKHA1	rs10510110	TC
LINC008	rs1561927	TC
LOC1079	rs9472138	CC
ETV1	rs7795991	AG
C6orf173	rs4273712	AA
TCF7L2	rs7903146	TT
CDKAL1	rs7756992	AG
GRB14	rs3923113	AA
TLE4	rs17791513	AA
CDC123	rs11257655	TC
ARAP1	rs1552224	AC
KCNQ1	rs163184	GG
JAZF1	rs849135	AG
KCNJ11	rs5215	TT
ST6GAL1	rs16861329	TC
MTNR1B	rs10830963	CC
HNF4A	rs4812829	AG
RPSAP52	rs2261181	CC
LOC1053	rs1359790	AG
AP3S2	rs2028299	AC
FTO	rs9936385	TT
GLIS3	rs7041847	GG
IGF2BP2	rs4402960	TT
PPARG	rs1801282	CC
HNF1B	rs4430796	AG

GWAS Complex Diseases: Endocrinology

Hypothyroidism

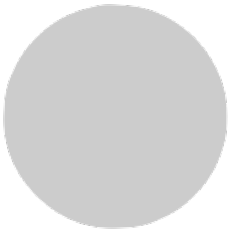
Your thyroid is a butterfly-shaped gland in your neck, just above your collarbone. It is one of your endocrine glands, which produce hormones. Thyroid hormones control the rate of many activities in your body. These include how fast you burn calories and how fast your heart beats. All of these activities comprise your body's metabolism. If your thyroid gland is not active enough, it does not produce enough thyroid hormone to meet your body's needs. This condition is known as hypothyroidism. Hypothyroidism is more common in women, people with other thyroid problems, and those over age 60. Hashimoto's Disease, an autoimmune disorder, is the most common cause. Other causes include thyroid nodules, thyroiditis, congenital hypothyroidism, surgical removal of part or all of the thyroid, radiation treatment of the thyroid, and some medicines.

Your genetic map

Gene	SNP	Genotype
INSR	rs4804416	TG
TRNAH	rs10961534	AA
TNFRSF1	rs10162002	GG
HLA C	rs2517532	AG
MTF1	rs3748682	TT
PDE8B	rs4704397	AG
ZBTB10	rs1051920	TC
ZNF804B	rs10248351	TT
KRT18P13	rs925489	TT
VAV3	rs4915077	TT
SH2B3	rs3184504	CC
PTPN22	rs6679677	CC
HLA	rs3129720	CC

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/22493691

GWAS Complex Diseases: Urogenital System

Endometriosis

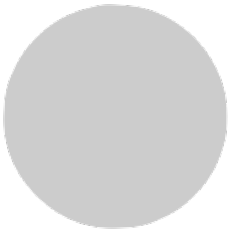
The uterus, or womb, is the place where a baby grows when a woman is pregnant. Endometriosis is a disease in which the kind of tissue that normally grows inside the uterus grows outside it. It can grow on the ovaries, fallopian tubes, bowels, or bladder. Rarely, it grows in other parts of the body.

Your genetic map

Gene	SNP	Genotype
GREB1	rs13394619	AA
LNC	rs7739264	TC
Intergeni	rs12700667	GG
CDKN2B	rs1537377	CC
VEZT	rs10859871	AC

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/23104006

GWAS Complex Diseases: Urogenital System

Bladder cancer

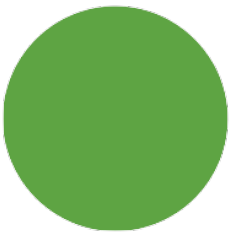
Bladder cancer is the fourth most frequently diagnosed in men. It is much more frequent in men than women, the ratio being 7-to-1. The incidence (new cases diagnosed in one year) in our country is the highest in the world: 11% of tumours in men, and 2.4% in women. 70-75% of the cases are attributed to tobacco consumption. Another risk factor is urinary tract infection. People with affected relatives are at increased risk of developing this type of tumour, suggesting that there is an underlying genetic factor. In fact, large-scale association studies have found genes predisposing one to the disease.

Your genetic map

Gene	SNP	Genotype
MYNN	rs10936599	CC
LSP1	rs907611	AG
LINC028	rs6104690	GG
MCF2L	rs4907479	AA
UGT1A10	rs11892031	AC
TP63	rs710521	TT
TACC3	rs798766	CC
CLPTM1L	rs401681	CC
NAT2	rs1495741	AG
PSCA	rs2204008	TT
CASC11	rs9642880	GG
SLC14A1	rs10775480	TC
CCNE1	rs8102137	TT
CBX6	rs1014971	CC

GWAS analysis

What do your genetics tell us?



According to this study, you are less likely to suffer from this disease than most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/24163127

GWAS Complex Diseases: Dermatology

Basal cell carcinoma

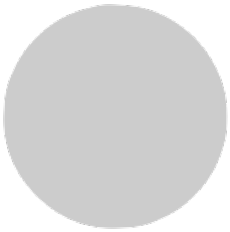
Non-melanoma type tumours occur on the outermost layer of the epidermis, and account for some 95% of the cancers that appear on the skin. About 20% are squamous carcinomas, which come from the malignization of the skin's squamous cells. It is among the most common cancers among people of European descent. The main cause of occurrence is DNA damage caused by ultraviolet exposure, although large-scale genetic studies have described genetic variants predisposing one to the disease.

Your genetic map

Gene	SNP	Genotype
MYCN	rs57244888	TT
FLACC1	rs13014235	GG
LOC1079	rs28727938	CC
GATA3	rs73635312	GG
PADI6	rs7538876	GG
RHOU	rs801114	TT
CLPTM1L	rs401681	CC
KRT5	rs11170164	CC
CDKN2B	rs2151280	AG
LINC	rs157935	TG
TP53	rs78378222	TT
TGM3	rs214782	AG
RGS22	rs7006527	AA

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/25855136

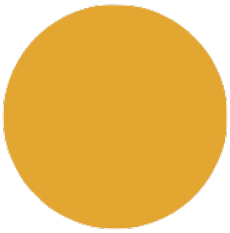
GWAS Complex Diseases: Dermatology

Psoriasis

Psoriasis is a skin disease that causes itchy or sore patches of thick, red skin with silvery scales. Patients usually get the patches on their elbows, knees, scalp, back, face, palms and feet, but they can show up on other parts of the body. Some people who have psoriasis also get a form of arthritis called psoriatic arthritis. A problem with your immune system causes psoriasis. In a process called cell turnover, skin cells that grow deep in your skin rise to the surface. This normally takes a month. In cases of psoriasis this happens in just days, because one's cells rise too fast. The disease is not hereditary, but there is a genetic predisposition to it, and a third of those affected have direct relatives with psoriasis.

GWAS analysis

What do your genetics tell us?



According to this study, you are more likely to suffer from this disease than most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/25903422

Your genetic map

Gene	SNP	Genotype
TP63	rs28512356	AC
COG6	rs34394770	TC
LOC1448	rs9533962	TC
RUNX1	rs8128234	CC
CLIC6	rs9305556	GG
LOC1079	rs11922372	TC
LOC2856	rs7709212	TT
TNIP	rs17728338	GG
IL12B	rs4921493	TC
IFIH1	rs3747517	TT
LCE	rs4845459	AA
TNFAIP3	rs643177	TC
REL DT	rs842625	AG
IL12B	rs2853694	GG
IFIH1	rs1990760	TT
PSMA6	rs8016947	TG
NOS2	rs4795067	AG
IL13	rs20541	GG
RIGI	rs11795343	TC
IL28RA	rs10794648	CC
QTRT1	rs892085	AG
IL23R	rs12564022	TT
STAT2	rs2066807	GC
REV3L	rs240993	CC
ETS1	rs6590334	TC
TRAF3IP2	rs7769061	AA

GWAS Complex Diseases: Dermatology

Vitiligo

Vitiligo causes white patches on your skin. It can also affect your eyes, mouth, and nose. It occurs when the cells that give your skin its color are destroyed. No one knows what destroys them. It is more common in people with autoimmune diseases, and it might run in families. It usually starts before age 40.

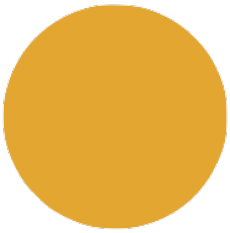
The white patches are more common where your skin is exposed to the sun. In some cases, the patches spread. Vitiligo can cause your hair to grey prematurely. If you have dark skin, you may lose colour inside your mouth.

Your genetic map

Gene	SNP	Genotype
IFIH1	rs2111485	GG
CD80	rs59374417	AC
CLNK	rs16872571	TC
BACH2	rs3757247	CC
CASP7	rs3814231	CC
SLC1A2	rs10768122	AG
TYR	rs4409785	CC
IKZF4	rs2456973	AC
ATXN2	rs4766578	TA
HERC2	rs1129038	TC
FANCA	rs9926296	AG
TICAM1	rs6510827	TT
TOB2	rs4822024	AG

GWAS analysis

What do your genetics tell us?



According to this study, you are more likely to suffer from this disease than most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/22561518

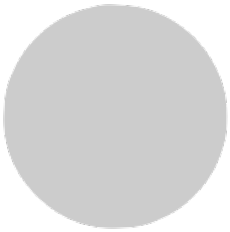
GWAS Complex Diseases: Others

Celiac disease

Celiac disease is an immune disease in which people cannot eat gluten because it damages their small intestine. If you have celiac disease and eat foods with gluten, your immune system responds by damaging the small intestine. Gluten is a protein found in wheat, rye, and barley. It may also be found in other products, like vitamins and supplements, hair and skin products, toothpastes, and lip balm. Celiac disease affects each person differently. Symptoms may occur in the digestive system, or in other parts of the body. One person might have diarrhea and abdominal pain, while another may be irritable or depressed. Irritability is one of the most common symptoms in children. Some people have no symptoms.

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/20190752

Your genetic map

Gene	SNP	Genotype
LOC1053	rs2816316	AA
PUS10	rs13003464	AG
IL18R1	rs917997	CC
LINC0193	rs13010713	GG
ICOS	rs4675374	TC
CCRL2	rs13098911	CC
IL12A AS1	rs17810546	AA
LPP	rs1464510	AC
BLTP1	rs13151961	AA
HLA	rs2187668	TT
TNFAIP3	rs2327832	AG
ATXN2	rs653178	CC
PTPN2	rs1893217	AA
MMEL1	rs3748816	AG
RUNX3	rs10903122	AG
MROH3P	rs296547	TC
PLEK	rs17035378	TC
ARHGAP3	rs11712165	TG
BACH2	rs10806425	AC
THEMIS	rs802734	AA
Intergeni	rs9792269	AA
ZMIZ1	rs1250552	AG
ETS1	rs11221332	TC
LOC1053	rs12928822	CC
ICOSLG	rs4819388	TT
CD247	rs864537	AA
TNFSF18	rs859637	CC
FRMD4B	rs6806528	CC
MYNN	rs10936599	CC
ELMO1	rs6974491	GG
DLEU1	rs2762051	CC

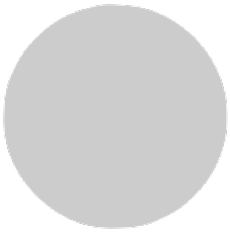
GWAS Complex Diseases: Others

Age-related macular degeneration

Macular degeneration, or age-related macular degeneration (AMD), is a leading cause of vision loss in Americans 60 and older. It is a disease that destroys your sharp, central vision. You need central vision to see objects clearly and to perform tasks such as reading and driving. AMD affects the macula, the part of the eye that allows you to perceive details. It does not hurt, but it causes cells in the macula to die. There are two types: wet and dry. Wet AMD happens when abnormal blood vessels grow under the macula. These new blood vessels often leak blood and fluid. Wet AMD damages the macula quickly. Blurred vision is a common early symptom. Dry AMD happens when the light-sensitive cells in the macula slowly break down. You gradually lose your central vision. A common early symptom is that straight lines appear crooked.

GWAS analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/23455636

Your genetic map

Gene	SNP	Genotype
ARMS2	rs10490924	GG
SKIC2	rs429608	AG
C3	rs2230199	CG
APOC1	rs4420638	AA
CETP	rs1864163	GG
LOC1079	rs943080	CC
TNFRSF1	rs13278062	TG
LOC1019	rs920915	CC
MCUB	rs4698775	TT
COL10A1	rs3812111	AT
COL8A1	rs13081855	GG
LOC1079	rs3130783	AA
SLC16A8	rs8135665	TC
TGFBR1	rs334353	TT
RAD51B	rs8017304	AG
ADAMTS9	rs6795735	TT
B3GLCT	rs9542236	CC

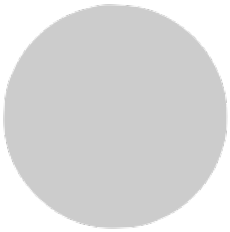
Complex Diseases: Oncogenic Mutations

APC: colorrectal and pancreatic cancer

APC gene mutations may be related to diseases such colorrectal and pancreatic cancer. Some publications associate it, in some cases, with gastric cancer.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=733

Your genetic map

Gene	SNP	Genotype
APC	rs137854571	CC
APC	rs387906230	TT
APC	rs137854568	CC
APC	rs137854569	CC
APC	rs137854570	CC
APC	rs121913327	CC
APC	rs137854572	CC
APC	rs137854573	CC
APC	rs137854574	CC
APC	rs137854577	CC
APC	rs137854580	CC
APC	rs137854582	TT
APC	rs397515734	CC
APC	rs398123116	GG
APC	rs398123117	CC
APC	rs398123121	CC
APC	rs587779780	CC
APC	rs587779783	CC
APC	rs587779786	AA
APC	rs62619935	CC
APC	rs587781392	CC
APC	rs587781809	TT
APC	rs587782518	CC
APC	rs587783029	CC
APC	rs587783035	AA
APC	rs376213437	TT
APC	rs730881240	CC
APC	rs145945630	CC
APC	rs786201291	AA
APC	rs786201856	CC
APC	rs775126020	CC

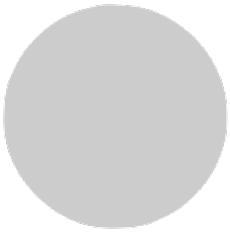
Complex Diseases: Oncogenic Mutations

ATM: breast cancer

Mutations of the ATM gene may be related to diseases like breast cancer. Some publications have associated this gene, to a lesser extent, with other cancers, such as ovarian.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene?Db=gene&Cmd=DetailsSearch&Term=472

Your genetic map

Gene	SNP	Genotype
ATM	rs587781545	CC
ATM	rs747855862	GG
ATM	rs55861249	CC
ATM	rs587776551	GG
ATM	rs1137887	GG
ATM	rs796051858	GG
ATM	rs587779813	GG
ATM	rs587779815	CC
ATM	rs587779818	GG
ATM	rs587779826	TT
ATM	rs587779833	CC
ATM	rs587779836	GG
ATM	rs587778080	CC
ATM	rs587781511	AA
ATM	rs587781558	GG
ATM	rs587781698	CC
ATM	rs200196781	GG
ATM	rs587781911	GG
ATM	rs587781927	TT
ATM	rs587781950	AA
ATM	rs587782103	GG
ATM	rs587782124	TT
ATM	rs587782192	TT
ATM	rs587782276	AA
ATM	rs587782280	GG
ATM	rs373226793	TT
ATM	rs376170600	CC
ATM	rs730881359	AA
ATM	rs730881333	CC
ATM	rs730881336	CC
ATM	rs730881347	GG

Complex Diseases: Oncogenic Mutations

BARD1: breast cancer

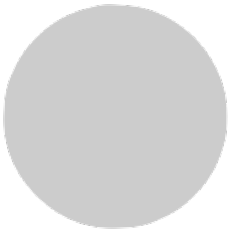
BARD1 gene mutations may be related to diseases like breast cancer. Some publications have associated this gene, to a minor extent, with ovarian cancer.

Your genetic map

Gene	SNP	Genotype
BARD1	rs587781430	GG
BARD1	rs587781707	GG
BARD1	rs587781948	GG
BARD1	rs587782681	GG
BARD1	rs730881422	GG
BARD1	rs730881415	CC
BARD1	rs730881411	GG
BARD1	rs786202559	GG
BARD1	rs786202500	GG
BARD1	rs758972589	GG
BARD1	rs786201912	GG
BARD1	rs864622239	TT

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.orpha.net/consor/cgi-bin/Disease_Search.php?lng=EN&data_id=3384&Disease_Disease_Search_diseaseGroup=BARD1&Disease_Disease_Search_diseaseType=Gen&Disease(s)/group%20of%20diseases=Hereditary-breast-and-ovarian-cancer-syndrome&title=Hereditary%20breast%20and%20ovarian%20cancer%20syndrome&search=Disease_Search_Simple

Complex Diseases: Oncogenic Mutations

BLM: colorrectal cancer

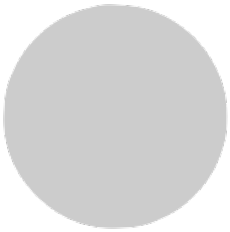
BLM gene mutations may be related to diseases such bloom syndrome and colorrectal cancer.

Your genetic map

Gene	SNP	Genotype
BLM	rs367543036	GG
BLM	rs367543029	GG
BLM	rs367543017	CC
BLM	rs200389141	CC
BLM	rs587779884	CC
BLM	rs587783037	CC
BLM	rs730881428	TT
BLM	rs1057516964	GG
BLM	rs1356090839	GG

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/641

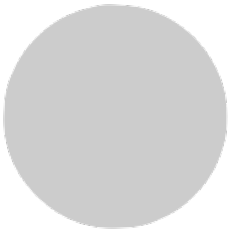
Complex Diseases: Oncogenic Mutations

BMPR1A: colorrectal, gastric and pancreatic cancer

BMPR1A gene mutations may be related to diseases such juvenile polyposis syndrome, colorrectal, gastric and pancreatic cancer.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2929

Your genetic map

Gene	SNP	Genotype
BMPR1A	rs199476085	GG
BMPR1A	rs199476086	CC
BMPR1A	rs199476087	TT
BMPR1A	rs587782388	GG
BMPR1A	rs587782400	CC
BMPR1A	rs587782682	CC
BMPR1A	rs786203157	AA
BMPR1A	rs764466442	CC
BMPR1A	rs786201040	CC
BMPR1A	rs878854672	GG
BMPR1A	rs878854664	GG
BMPR1A	rs759363072	CC
BMPR1A	rs1131691178	CC
BMPR1A	rs1131691185	CC
BMPR1A	rs1230919713	CC
BMPR1A	rs1404557708	CC
BMPR1A	rs1392086533	CC

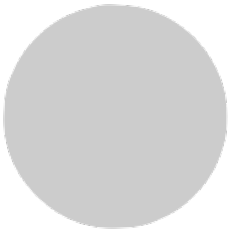
Complex Diseases: Oncogenic Mutations

BRCA1: breast and ovarian cancer

Mutations of the BRCA1 gene may be related to diseases such as breast and ovarian cancer. There are some studies that associated this gene, to a lesser extent, with other cancers, such as colon and pancreatic.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=145

Your genetic map

Gene	SNP	Genotype
BRCA1	rs80357064	AA
BRCA1	rs28897672	AA
BRCA1	rs62625308	GG
BRCA1	rs28897686	CC
BRCA1	rs41293455	GG
BRCA1	rs62625306	CC
BRCA1	rs80357382	TT
BRCA1	rs41293463	AA
BRCA1	rs80357498	CC
BRCA1	rs80357253	TT
BRCA1	rs80358038	CC
BRCA1	rs80358158	CC
BRCA1	rs80357010	GG
BRCA1	rs80356898	GG
BRCA1	rs80357005	CC
BRCA1	rs80357355	TT
BRCA1	rs80358061	AA
BRCA1	rs80358163	TT
BRCA1	rs80357233	GG
BRCA1	rs80357147	TT
BRCA1	rs80356875	CC
BRCA1	rs80357131	GG
BRCA1	rs80356925	GG
BRCA1	rs80357251	CC
BRCA1	rs80357170	TT
BRCA1	rs80357035	CC
BRCA1	rs80357115	AA
BRCA1	rs397507206	GG
BRCA1	rs80358051	GG
BRCA1	rs80357161	CC
BRCA1	rs397507215	GG

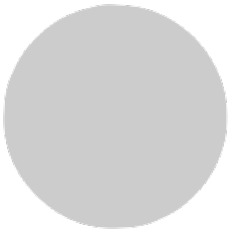
Complex Diseases: Oncogenic Mutations

BRCA2: breast and ovarian cancer

Mutations of the BRCA2 gene may be related to diseases such as breast and ovarian cancer. Some studies have related this gene, to a lesser extent, with other cancers, such as pancreatic.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/675

Your genetic map

Gene	SNP	Genotype
BRCA2	rs80359062	CC
BRCA2	rs80359070	TT
BRCA2	rs80358695	GG
BRCA2	rs80358979	TT
BRCA2	rs80358785	CC
BRCA2	rs80359180	CC
BRCA2	rs81002897	GG
BRCA2	rs81002899	TT
BRCA2	rs397507266	CC
BRCA2	rs397507275	AA
BRCA2	rs80358464	TT
BRCA2	rs80358474	CC
BRCA2	rs397507278	CC
BRCA2	rs397507279	TT
BRCA2	rs397507282	CC
BRCA2	rs80358504	TT
BRCA2	rs397507285	TT
BRCA2	rs80358529	CC
BRCA2	rs80358532	CC
BRCA2	rs397507296	CC
BRCA2	rs80358544	GG
BRCA2	rs80358550	AA
BRCA2	rs80358557	CC
BRCA2	rs41293477	TT
BRCA2	rs397507303	GG
BRCA2	rs397507305	TT
BRCA2	rs80358638	GG
BRCA2	rs397507320	TT
BRCA2	rs80358650	GG
BRCA2	rs397507325	TT
BRCA2	rs80358663	CC

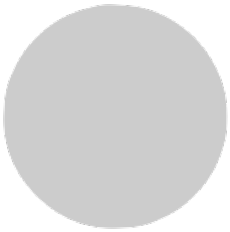
Complex Diseases: Oncogenic Mutations

BRIP1: breast cancer

Mutations in the BRIP1 gene may be related to diseases like breast cancer. There are some studies that associated this gene, on a smaller scale, with ovarian cancer.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=227535

Your genetic map

Gene	SNP	Genotype
BRIP1	rs587780226	GG
BRIP1	rs587780228	CC
BRIP1	rs587780833	CC
BRIP1	rs587780875	AA
BRIP1	rs587781292	CC
BRIP1	rs587781321	GG
BRIP1	rs587781655	CC
BRIP1	rs368796923	GG
BRIP1	rs587781786	GG
BRIP1	rs587782047	CC
BRIP1	rs574552037	GG
BRIP1	rs587782410	AA
BRIP1	rs587782539	CC
BRIP1	rs587782574	GG
BRIP1	rs730881635	TT
BRIP1	rs730881633	GG
BRIP1	rs786202927	TT
BRIP1	rs786203451	CC
BRIP1	rs747604569	GG
BRIP1	rs775171520	CC
BRIP1	rs864622277	CC
BRIP1	rs575595017	GG

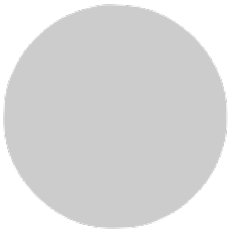
Complex Diseases: Oncogenic Mutations

CDH1: breast and gastric cancer

Mutations of the CDH1 gene may be associated with diseases such as breast and gastric cancer. There are some studies linking this gene, to a lesser extent, with ovarian and colon cancer.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/999

Your genetic map

Gene	SNP	Genotype
CDH1	rs121964877	CC
CDH1	rs587780113	GG
CDH1	rs149127230	GG
CDH1	rs587780537	GG
CDH1	rs587780784	CC
CDH1	rs587780787	GG
CDH1	rs587782750	CC
CDH1	rs587782798	CC
CDH1	rs587783047	CC
CDH1	rs587783050	GG
CDH1	rs730881663	CC
CDH1	rs786202817	TT
CDH1	rs786202290	GG
CDH1	rs786202785	GG
CDH1	rs876660771	GG

Complex Diseases: Oncogenic Mutations

CDK4: Familial melanoma

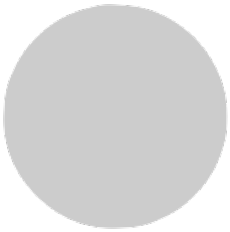
Mutations of the CDK4 gene may be related to diseases such as familial melanoma.

Your genetic map

Gene	SNP	Genotype
CDK4	rs11547328	GG

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=618

Complex Diseases: Oncogenic Mutations

CDKN2A: pancreatic cancer

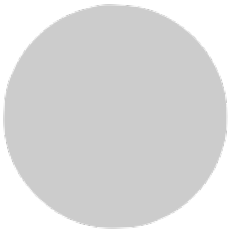
CDKN2A gene mutations may be related to diseases such as pancreatic cancer.

Your genetic map

Gene	SNP	Genotype
CDKN2A	rs104894095	CC
CDKN2A	rs104894097	CC
CDKN2A	rs104894098	AA
CDKN2A	rs104894099	AA
CDKN2A	rs587778189	TT
CDKN2A	rs1800586	CC
CDKN2A	rs45476696	CC
CDKN2A	rs749714198	GG
CDKN2A	rs199907548	AA
CDKN2A	rs730881677	CC

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/1029

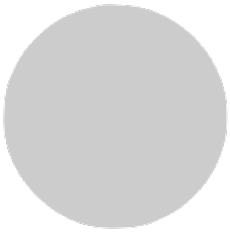
Complex Diseases: Oncogenic Mutations

CHEK2: breast and colorrectal cancer

CHEK2 gene mutations may be related to diseases such as breast and colorrectal cancer.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=227535

Your genetic map

Gene	SNP	Genotype
CHEK2	rs137853007	GG
CHEK2	rs121908702	CC
CHEK2	rs121908698	CC
CHEK2	rs200432447	GG
CHEK2	rs28909982	TT
CHEK2	rs536907995	GG
CHEK2	rs587781269	GG
CHEK2	rs587781592	GG
CHEK2	rs587781699	CC
CHEK2	rs587781705	AA
CHEK2	rs587782070	CC
CHEK2	rs587782401	AA
CHEK2	rs587782575	TT
CHEK2	rs587782830	CC
CHEK2	rs730881702	CC
CHEK2	rs730881687	CC
CHEK2	rs730881701	GG
CHEK2	rs786201906	CC
CHEK2	rs760502479	GG
CHEK2	rs786203650	CC
CHEK2	rs786203229	CC
CHEK2	rs786203889	CC
CHEK2	rs761494650	GG
CHEK2	rs864622149	CC
CHEK2	rs545982789	AA
CHEK2	rs864622613	CC
CHEK2	rs371418985	CC
CHEK2	rs768384031	GG
CHEK2	rs756250205	GG
CHEK2	rs778989252	GG
CHEK2	rs768172525	CC

Complex Diseases: Oncogenic Mutations

DICER1: ovarian cancer

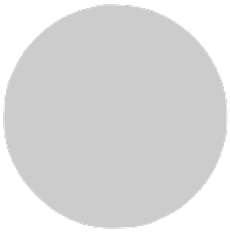
DICER1 gene mutations may be related to diseases such as ovarian cancer or DICER1 syndrome related to various types of tumors.

Your genetic map

Gene	SNP	Genotype
DICER1	rs137852976	AA
DICER1	rs137852977	CC
DICER1	rs137852978	GG
DICER1	rs137852979	GG

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=284343

Complex Diseases: Oncogenic Mutations

EPCAM: Lynch syndrome, breast, ovarian, uterine, colorrectal, gastric and pancreatic cancer

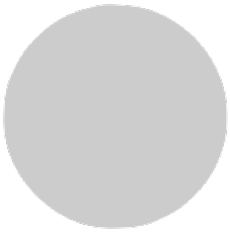
EPCAM gene mutations may be related to diseases such as Lynch syndrome, breast, ovarian, uterine, colorrectal, gastric and pancreatic cancer.

Your genetic map

Gene	SNP	Genotype
EPCAM	rs606231203	GG
EPCAM	rs376155665	AA

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/4072

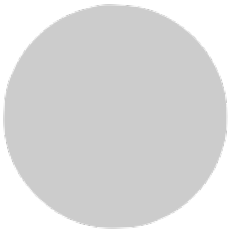
Complex Diseases: Oncogenic Mutations

FH: Hereditary leiomyomatosis and renal cell cancer

Mutations of the FH gene may be related to hereditary leiomyomatosis and renal cell cancer (HLRCC).

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/2271

Your genetic map

Gene	SNP	Genotype
FH	rs121913120	GG
FH	rs121913122	GG
FH	rs121913123	CC
FH	rs75086406	CC
FH	rs398123160	GG
FH	rs121913121	TT
FH	rs398123168	GG
FH	rs727503927	AA
FH	rs863224010	TT
FH	rs863224007	CC
FH	rs863223966	TT
FH	rs863223968	GG
FH	rs863223980	GG
FH	rs886039368	CC
FH	rs398123159	AA
FH	rs398123166	GG
FH	rs587781682	GG
FH	rs587782618	CC
FH	rs372505976	TT
FH	rs863223978	CC
FH	rs863224008	TT
FH	rs863224004	CC
FH	rs863223973	AA
FH	rs863224002	GG
FH	rs863224000	AA
FH	rs863223967	TT
FH	rs863223965	AA
FH	rs863224015	TT
FH	rs863223983	TT
FH	rs863223982	CC

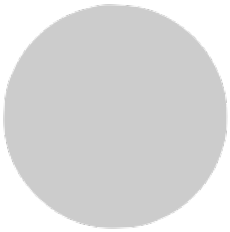
Complex Diseases: Oncogenic Mutations

FLCN: Kidney cancer

Mutations of the FLCN gene may be related to diseases such as kidney cancer. In addition, some studies associated this gene, to a lesser extent, with other tumors of the skin and lungs.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=122

Your genetic map

Gene	SNP	Genotype
FLCN	rs137852929	GG
FLCN	rs398124524	GG
FLCN	rs398124528	TT
FLCN	rs398124530	CC
FLCN	rs398124533	TT
FLCN	rs398124536	GG
FLCN	rs587782069	GG
FLCN	rs786202081	CC
FLCN	rs758175953	CC
FLCN	rs876658409	CC
FLCN	rs878855218	CC
FLCN	rs879255683	GG
FLCN	rs755959303	CC
FLCN	rs879255678	GG
FLCN	rs879255668	AA
FLCN	rs879255667	GG

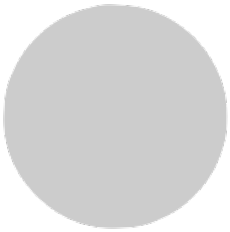
Complex Diseases: Oncogenic Mutations

MEN1: multiple endocrine neoplasia type 1

MEN1 gene mutations may be related to diseases such as multiple endocrine neoplasia type 1.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=652

Your genetic map

Gene	SNP	Genotype
MEN1	rs104894256	AA
MEN1	rs28931612	CC
MEN1	rs104894263	GG
MEN1	rs1060499976	CC
MEN1	rs386134250	TT
MEN1	rs386134254	GG
MEN1	rs386134256	AA
MEN1	rs386134260	GG
MEN1	rs794728622	CC
MEN1	rs398124437	CC
MEN1	rs786204242	CC
MEN1	rs794728627	GG
MEN1	rs794728625	CC
MEN1	rs794728652	CC
MEN1	rs794728624	CC
MEN1	rs104894257	CC
MEN1	rs794728650	CC
MEN1	rs376872829	CC
MEN1	rs794728647	GG
MEN1	rs794728616	GG
MEN1	rs794728614	GG
MEN1	rs878855192	TT
MEN1	rs886039416	GG
MEN1	rs886039415	AA
MEN1	rs886039414	CC
MEN1	rs886039553	GG
MEN1	rs886039413	GG
MEN1	rs886042035	TT
MEN1	rs1057518572	CC
MEN1	rs794728648	CC
MEN1	rs1057520733	GG

Complex Diseases: Oncogenic Mutations

MET: Lung and gastric cancer

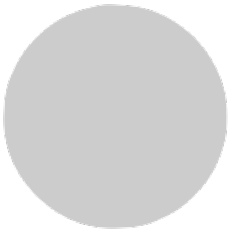
Mutations of the MET gene may be related to lung and gastric cancer. Some studies associated this gene, to a lesser extent, with other cancers, such as cell ovarian and colorectal.

Your genetic map

Gene	SNP	Genotype
MET	rs121913670	GG
MET	rs121913243	AA
MET	rs794728016	TT
MET	rs786202724	GG

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/4233

Complex Diseases: Oncogenic Mutations

MITF: MITF-related melanoma and renal cell carcinoma predisposition syndrome

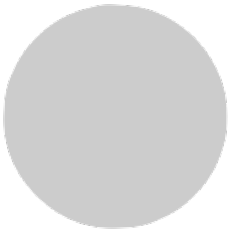
Mutations of the MITF gene may be related to diseases such as melanoma and renal cell carcinoma predisposition syndrome. In addition, some studies associated this gene, to a lesser extent, with other cancers, such as breast cancer.

Your genetic map

Gene	SNP	Genotype
MITF	rs104893746	CC
MITF	rs149617956	GG

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/4286

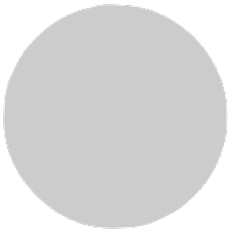
Complex Diseases: Oncogenic Mutations

MLH1: Lynch syndrome

MLH1 gene mutations may be related to diseases such as Lynch Syndrome.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=144

Your genetic map

Gene	SNP	Genotype
MLH1	rs63750198	CC
MLH1	rs63751109	CC
MLH1	rs63750710	AA
MLH1	rs63751615	CC
MLH1	rs63750206	GG
MLH1	rs63750899	CC
MLH1	rs63750691	CC
MLH1	rs63750217	GG
MLH1	rs63749939	GG
MLH1	rs63751194	CC
MLH1	rs63750540	AA
MLH1	rs63751221	CC
MLH1	rs63751715	GG
MLH1	rs587778894	CC
MLH1	rs267607823	AA
MLH1	rs63750443	GG
MLH1	rs63749795	CC
MLH1	rs267607836	AA
MLH1	rs267607853	GG
MLH1	rs63751657	GG
MLH1	rs267607867	GG
MLH1	rs63751632	GG
MLH1	rs267607871	AA
MLH1	rs63750726	CC
MLH1	rs63751275	CC
MLH1	rs267607720	CC
MLH1	rs267607894	TT
MLH1	rs63750437	GG
MLH1	rs63750005	CC
MLH1	rs267607735	GG
MLH1	rs267607750	GG

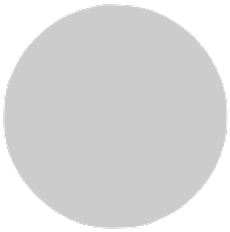
Complex Diseases: Oncogenic Mutations

MSH2: Lynch syndrome and colorrectal cancer

MSH2 gene mutations may be related to diseases such as Lynch Syndrome and colorrectal cancer.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=144

Your genetic map

Gene	SNP	Genotype
MSH2	rs267607939	CC
MSH2	rs267607940	GG
MSH2	rs63750224	CC
MSH2	rs267607972	GG
MSH2	rs267607970	GG
MSH2	rs63751411	GG
MSH2	rs63750636	CC
MSH2	rs63750843	CC
MSH2	rs587779190	GG
MSH2	rs28929483	CC
MSH2	rs63751108	CC
MSH2	rs28929484	CC
MSH2	rs63750047	CC
MSH2	rs63751207	GG
MSH2	rs63750875	GG
MSH2	rs63749932	CC
MSH2	rs193922376	AA
MSH2	rs63750396	GG
MSH2	rs587779067	CC
MSH2	rs267607943	AA
MSH2	rs63750558	CC
MSH2	rs63749849	CC
MSH2	rs587779075	CC
MSH2	rs63751412	CC
MSH2	rs267607950	GG
MSH2	rs63751693	CC
MSH2	rs63751646	AA
MSH2	rs267607957	GG
MSH2	rs587779087	TT
MSH2	rs63750615	GG
MSH2	rs267607969	GG

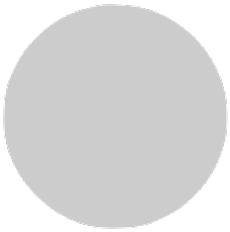
Complex Diseases: Oncogenic Mutations

MSH6: Lynch syndrome and colorrectal cancer

MSH6 gene mutations may be related to diseases such as Lynch Syndrome and colorrectal cancer.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/2956

Your genetic map

Gene	SNP	Genotype
MSH6	rs1800937	CC
MSH6	rs63751405	TT
MSH6	rs63750741	TT
MSH6	rs63750909	CC
MSH6	rs587779227	GG
MSH6	rs267608068	TT
MSH6	rs63751419	CC
MSH6	rs63750138	CC
MSH6	rs63751017	CC
MSH6	rs587779246	CC
MSH6	rs63750563	CC
MSH6	rs587779252	GG
MSH6	rs267608098	AA
MSH6	rs587779279	GG
MSH6	rs267608066	CC
MSH6	rs267608048	CC
MSH6	rs200492211	CC
MSH6	rs587781462	CC
MSH6	rs786201042	CC
MSH6	rs786201049	GG
MSH6	rs864622153	CC
MSH6	rs876660943	GG
MSH6	rs1064795256	CC
MSH6	rs587779204	TT
MSH6	rs587779215	CC
MSH6	rs63751127	CC
MSH6	rs63751321	CC
MSH6	rs63750111	CC
MSH6	rs63750258	GG
MSH6	rs63749999	CC
MSH6	rs587779255	GG

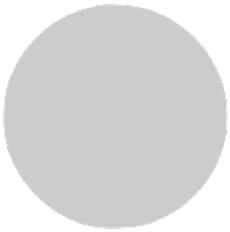
Complex Diseases: Oncogenic Mutations

MUTYH: colorrectal cancer

MUTYH gene mutations may be related to diseases such as MYH-associated polyposis and colorrectal cancer.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=247798

Your genetic map

Gene	SNP	Genotype
MUTYH	rs34612342	TT
MUTYH	rs121908380	GG
MUTYH	rs121908381	CC
MUTYH	rs200495564	GG
MUTYH	rs587780082	GG
MUTYH	rs587780088	GG
MUTYH	rs587781295	CC
MUTYH	rs587781337	CC
MUTYH	rs587781338	GG
MUTYH	rs140342925	CC
MUTYH	rs587781628	TT
MUTYH	rs587782228	CC
MUTYH	rs529008617	GG
MUTYH	rs587782730	AA
MUTYH	rs587782885	GG
MUTYH	rs587783057	GG
MUTYH	rs558173961	GG
MUTYH	rs730881833	CC
MUTYH	rs143353451	CC
MUTYH	rs730881832	AA
MUTYH	rs376790729	CC
MUTYH	rs374950566	GG
MUTYH	rs786203115	GG
MUTYH	rs34126013	GG
MUTYH	rs747993448	GG
MUTYH	rs786203161	TT
MUTYH	rs372267274	CC
MUTYH	rs765123255	GG
MUTYH	rs863224502	TT
MUTYH	rs863224452	TT
MUTYH	rs876659420	CC

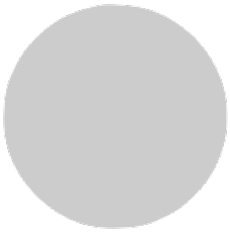
Complex Diseases: Oncogenic Mutations

NBN: breast, ovarian, colorrectal and gastric cancer

NBN gene mutations may be related to diseases such as breast, ovarian, colorrectal and gastric cancer.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/4683

Your genetic map

Gene	SNP	Genotype
NBN	rs121908973	GG
NBN	rs121908974	GG
NBN	rs587782130	GG
NBN	rs587782545	TT
NBN	rs730881857	GG
NBN	rs730881850	AA
NBN	rs142301194	AA
NBN	rs786201965	CC
NBN	rs786203223	AA
NBN	rs786201745	CC
NBN	rs574673404	CC
NBN	rs786204181	CC
NBN	rs767215758	GG
NBN	rs786205135	AA
NBN	rs864622090	TT
NBN	rs876659521	TT
NBN	rs1057517262	CC
NBN	rs756363734	CC

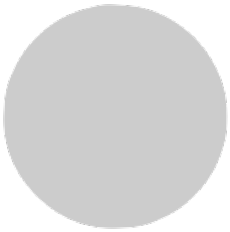
Complex Diseases: Oncogenic Mutations

NF1: type 1 neurofibromatosis

NF1 gene mutations may be related to diseases such as type 1 neurofibromatosis.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/4763

Your genetic map

Gene	SNP	Genotype
NF1	rs137854550	AA
NF1	rs137854552	CC
NF1	rs137854560	CC
NF1	rs267606599	GG
NF1	rs137854556	GG
NF1	rs267606603	GG
NF1	rs137854559	CC
NF1	rs267606604	AA
NF1	rs137854562	CC
NF1	rs137854563	TT
NF1	rs397514641	CC
NF1	rs199474737	TT
NF1	rs199474760	AA
NF1	rs199474762	TT
NF1	rs199474746	CC
NF1	rs199474747	TT
NF1	rs199474786	TT
NF1	rs199474742	CC
NF1	rs199474790	AA
NF1	rs587781517	GG
NF1	rs587781577	GG
NF1	rs587782088	GG
NF1	rs786203448	CC
NF1	rs786203390	GG
NF1	rs786202112	GG
NF1	rs772295894	CC
NF1	rs786202457	CC
NF1	rs786201367	CC
NF1	rs768638173	CC
NF1	rs786204211	TT
NF1	rs786204253	TT

Complex Diseases: Oncogenic Mutations

NF2: Familial multiple meningioma

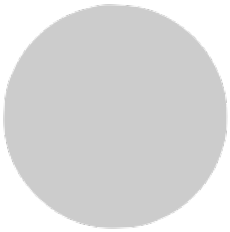
Mutations of the NF2 gene may be related to diseases such as multiple familial meningiomas.

Your genetic map

Gene	SNP	Genotype
NF2	rs121434259	CC
NF2	rs74315496	CC
NF2	rs74315499	CC
NF2	rs74315503	GG
NF2	rs74315504	CC
NF2	rs74315505	GG
NF2	rs794728682	GG
NF2	rs878853925	AA
NF2	rs1060503667	CC
NF2	rs1060503670	AA
NF2	rs1060503666	AA
NF2	rs1064796632	GG
NF2	rs917257652	CC
NF2	rs587776562	GG

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=637

Complex Diseases: Oncogenic Mutations

NTHL1: Attenuated familial adenomatous polyposis

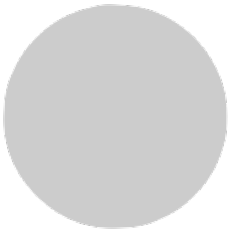
Mutations of the NTHL1 gene may be related to diseases such as familial adenomatous polyposis and colorectal cancer. In addition, some studies associated this gene, to a lesser extent, with other cancers, such as breast cancer.

Your genetic map

Gene	SNP	Genotype
NTHL1	rs146347092	GG
NTHL1	rs779757251	CC

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/4913

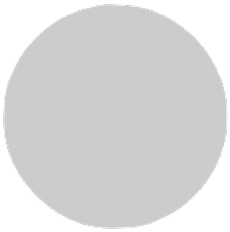
Complex Diseases: Oncogenic Mutations

RAD50: breast and pancreatic cancer

RAD50 gene mutations may be related to diseases such as breast and pancreatic cancer

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/79728

Your genetic map

Gene	SNP	Genotype
PALB2	rs118203997	AA
PALB2	rs118203998	GG
PALB2	rs118203999	GG
PALB2	rs180177097	GG
PALB2	rs180177103	CC
PALB2	rs180177083	GG
PALB2	rs180177111	GG
PALB2	rs180177112	CC
PALB2	rs587776417	CC
PALB2	rs180177122	CC
PALB2	rs515726099	CC
PALB2	rs180177132	CC
PALB2	rs515726111	CC
PALB2	rs587776527	GG
PALB2	rs180177091	GG
PALB2	rs180177100	GG
PALB2	rs587778587	CC
PALB2	rs587776423	CC
PALB2	rs375699023	GG
PALB2	rs587782005	TT
PALB2	rs180177110	GG
PALB2	rs587782446	GG
PALB2	rs587776411	GG
PALB2	rs587776413	GG
PALB2	rs587776419	CC
PALB2	rs587776407	GG
PALB2	rs730881888	AA
PALB2	rs730881876	CC
PALB2	rs730881905	CC
PALB2	rs730881879	TT
PALB2	rs730881897	TT

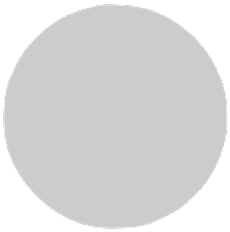
Complex Diseases: Oncogenic Mutations

PMS2: Lynch syndrome and colorrectal cancer

PMS2 gene mutations may be related to diseases such as Lynch Syndrome and colorrectal cancer.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=144

Your genetic map

Gene	SNP	Genotype
PMS2	rs63751466	GG
PMS2	rs63750451	GG
PMS2	rs121434629	CC
PMS2	rs267608158	AA
PMS2	rs587778617	GG
PMS2	rs63750490	TT
PMS2	rs63751422	GG
PMS2	rs201451115	TT
PMS2	rs267608172	CC
PMS2	rs587779338	GG
PMS2	rs587779343	GG
PMS2	rs267608153	CC
PMS2	rs200640585	GG
PMS2	rs587780062	GG
PMS2	rs587780064	CC
PMS2	rs587778618	GG
PMS2	rs587780724	GG
PMS2	rs587781339	TT
PMS2	rs730881919	CC
PMS2	rs863224450	CC
PMS2	rs876659736	TT
PMS2	rs1064794577	CC
PMS2	rs1064794083	AA
PMS2	rs988423880	CC
PMS2	rs1458321358	GG
PMS2	rs63750871	GG
PMS2	rs267608161	CC
PMS2	rs63750261	GG
PMS2	rs587779347	TT
PMS2	rs141577476	GG
PMS2	rs786201047	GG

Complex Diseases: Oncogenic Mutations

POLD1: breast, ovarian, uterine and colorrectal cancer

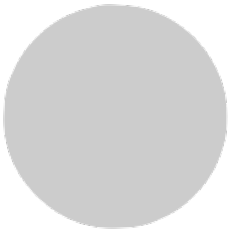
POLD1 gene mutations may be related to diseases such breast, ovarian, uterine and colorrectal cancer.

Your genetic map

Gene	SNP	Genotype
POLD1	rs587777627	TT

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/5424

Complex Diseases: Oncogenic Mutations

POLE: ovarian, uterine, colorrectal andpancreatic cancer

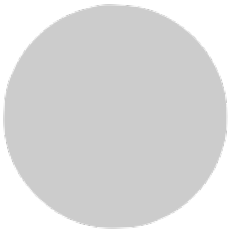
POLE gene mutations may be related to diseases such ovarian, uterine, colorrectal andpancreatic cancer.

Your genetic map

Gene	SNP	Genotype
POLE	rs483352909	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=220460

Complex Diseases: Oncogenic Mutations

MSH3-related attenuated familial adenomatous polyposis

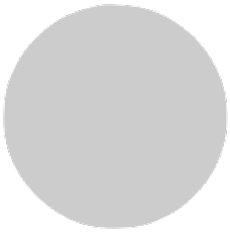
Mutations of the MSH3 gene may be related to diseases such as familial adenomatous polyposis and colorectal and stomach cancer.

Your genetic map

Gene	SNP	Genotype
MSH3	rs539295465	CC

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/4437

Complex Diseases: Oncogenic Mutations

POT1: Familial melanoma

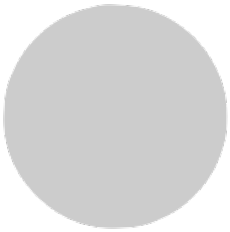
Mutations of the POT1 gene may be related to diseases such as familial melanoma. In addition, some studies associated this gene, to a lesser extent, with gliomas.

Your genetic map

Gene	SNP	Genotype
POT1	rs756198077	GG
POT1	rs531061783	GG

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=618

Complex Diseases: Oncogenic Mutations

PTCH1: Basal cell carcinoma

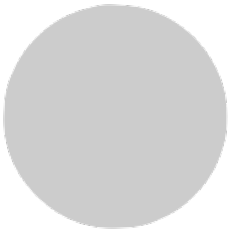
Mutations of the PTCH1 gene may be related to diseases such as basal cell carcinoma and skin cancer.

Your genetic map

Gene	SNP	Genotype
PTCH1	rs786204056	AA
PTCH1	rs863224443	TT
PTCH1	rs863224444	CC
PTCH1	rs863224487	AA
PTCH1	rs863224486	GG
PTCH1	rs863225054	TT
PTCH1	rs864622293	CC
PTCH1	rs779388970	GG

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=377

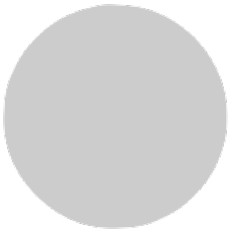
Complex Diseases: Oncogenic Mutations

PTEN: breast, uterine and colorrectal cancer

PTEN gene mutations may be related to diseases such as breast, uterine and colorrectal cancer.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/5728

Your genetic map

Gene	SNP	Genotype
PTEN	rs121909224	CC
PTEN	rs121909238	AA
PTEN	rs121909239	AA
PTEN	rs121909240	TT
PTEN	rs397514559	CC
PTEN	rs397514560	CC
PTEN	rs587782360	AA
PTEN	rs786204863	GG
PTEN	rs786204865	AA
PTEN	rs121913293	CC
PTEN	rs863224909	CC
PTEN	rs1057519368	TT
PTEN	rs876660507	GG
PTEN	rs1057517809	GG
PTEN	rs370795352	TT
PTEN	rs1114167667	TT
PTEN	rs121909218	GG
PTEN	rs121909219	CC
PTEN	rs121909221	TT
PTEN	rs121909222	AA
PTEN	rs121909223	TT
PTEN	rs587776667	GG
PTEN	rs121909225	TT
PTEN	rs121909226	TT
PTEN	rs121909227	CC
PTEN	rs121909228	GG
PTEN	rs121909229	GG
PTEN	rs121909231	CC
PTEN	rs121909232	CC
PTEN	rs121909241	GG
PTEN	rs398123321	TT

Complex Diseases: Oncogenic Mutations

RAD50: breast and ovarian cancer

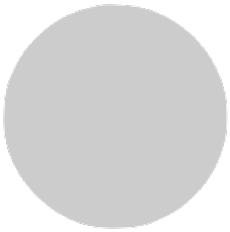
RAD50 gene mutations may be related to diseases such as breast and ovarian cancer.

Your genetic map

Gene	SNP	Genotype
RAD50	rs373428259	CC
RAD50	rs587780150	CC
RAD50	rs377260382	GG
RAD50	rs587781904	CC
RAD50	rs587782078	GG
RAD50	rs587782090	GG
RAD50	rs149201802	CC
RAD50	rs587781742	GG

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/10111

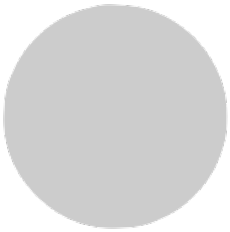
Complex Diseases: Oncogenic Mutations

RAD51C: ovarian cancer

RAD51C gene mutations may be related to diseases such as ovarian cancer.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/5889

Your genetic map

Gene	SNP	Genotype
RAD51C	rs267606997	GG
RAD51C	rs267606999	GG
RAD51C	rs387907159	CC
RAD51C	rs587780259	AA
RAD51C	rs200293302	CC
RAD51C	rs587781490	AA
RAD51C	rs587782036	GG
RAD51C	rs587782702	GG
RAD51C	rs587782818	CC
RAD51C	rs730881931	TT
RAD51C	rs786201909	TT
RAD51C	rs770637624	CC
RAD51C	rs757128712	GG
RAD51C	rs767796996	GG
RAD51C	rs760235677	GG
RAD51C	rs876659874	GG

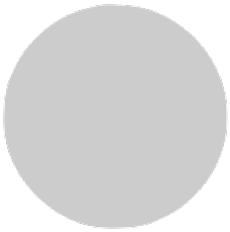
Complex Diseases: Oncogenic Mutations

RB1: Lynch syndrome and retinoblastoma

Mutations of the RB1 gene may be related to a rare inherited cancer-predisposing syndrome characterized by a predisposition to a wide variety of cancers, including neoplasms of the digestive tract, urinary tract, kidney, endometrium, ovary, brain, and prostate, as well as sebaceous skin tumors. In addition, some studies associated this gene, to a lesser extent, with other cancers, such as retinoblastoma.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=790

Your genetic map

Gene	SNP	Genotype
RB1	rs3092891	CC
RB1	rs137853293	CC
RB1	rs121913301	AA
RB1	rs137853294	CC
RB1	rs121913304	CC
RB1	rs137853296	TT
RB1	rs137853297	TT
RB1	rs483352690	GG
RB1	rs587778864	CC
RB1	rs121913305	CC
RB1	rs587778871	GG
RB1	rs587778850	GG
RB1	rs587778839	TT
RB1	rs121913296	GG
RB1	rs587778870	CC
RB1	rs587778842	CC
RB1	rs121913300	CC
RB1	rs587776783	GG
RB1	rs587778831	GG
RB1	rs587778846	GG
RB1	rs121913302	CC
RB1	rs121913303	CC
RB1	rs794727199	GG
RB1	rs794727481	GG
RB1	rs878853947	TT
RB1	rs878853949	CC
RB1	rs886043247	CC
RB1	rs1060503088	TT
RB1	rs1060503067	GG
RB1	rs1060503079	CC
RB1	rs1060503074	TT

Complex Diseases: Oncogenic Mutations

RECQL4: Stomach and colon cancer

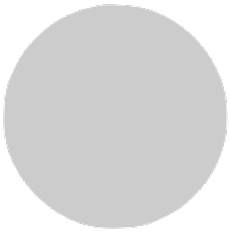
Mutations of the RECQL4 gene may be related to diseases such as stomach and colon cancer. In addition, some studies associated this gene with other cancers, such as endometrial cancer, to a lesser extent.

Your genetic map

Gene	SNP	Genotype
RECQL4	rs137853229	GG
RECQL4	rs117642173	CC
RECQL4	rs386833844	GG
RECQL4	rs386833851	GG
RECQL4	rs398124117	CC
RECQL4	rs373130543	CC

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/9401

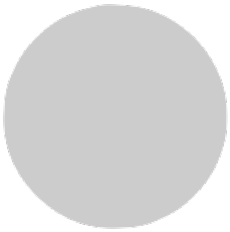
Complex Diseases: Oncogenic Mutations

RET: thyroid carcinoma

RET gene mutations may be related to diseases such thyroid carcinoma.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/5979

Your genetic map

Gene	SNP	Genotype
RET	rs76262710	TT
RET	rs75076352	TT
RET	rs75996173	GG
RET	rs79781594	GG
RET	rs77316810	TT
RET	rs77503355	GG
RET	rs77709286	CC
RET	rs77939446	GG
RET	rs77558292	TT
RET	rs75873440	GG
RET	rs75234356	TT
RET	rs377767404	TT
RET	rs267607011	CC
RET	rs74799832	TT
RET	rs78014899	GG
RET	rs377767391	TT
RET	rs377767412	GG
RET	rs143795581	AA
RET	rs193922699	AA

Complex Diseases: Oncogenic Mutations

SDHA: gastric cancer

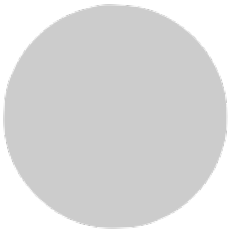
SDHA gene mutations may be related to diseases such gastric cancer.

Your genetic map

Gene	SNP	Genotype
SDHA	rs137852768	GG
SDHA	rs142441643	CC
SDHA	rs781764920	CC
SDHA	rs151170408	CC
SDHA	rs766667009	GG
SDHA	rs748089700	CC

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=29072

Complex Diseases: Oncogenic Mutations

SDHAF2: Hereditary pheochromocytoma-paraganglioma

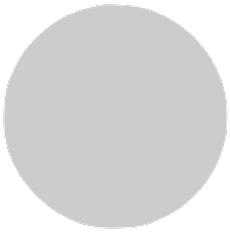
Mutations of the SDHAF2 gene may be related to diseases such as pheochromocytoma/paraganglioma tumors.

Your genetic map

Gene	SNP	Genotype
SDHAF2	rs113560320	GG

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/54949

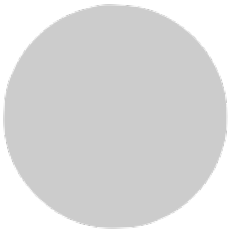
Complex Diseases: Oncogenic Mutations

SDHB: gastric cancer

SDHB gene mutations may be related to diseases such as gastric cancer.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/6390

Your genetic map

Gene	SNP	Genotype
SDHB	rs74315366	GG
SDHB	rs74315367	GG
SDHB	rs74315368	CC
SDHB	rs74315369	GG
SDHB	rs74315370	GG
SDHB	rs74315372	TT
SDHB	rs398122805	CC
SDHB	rs267607032	CC
SDHB	rs202101384	TT
SDHB	rs397516833	CC
SDHB	rs397516835	CC
SDHB	rs397516836	CC
SDHB	rs587781270	AA
SDHB	rs587782243	CC
SDHB	rs587782604	CC
SDHB	rs727504457	AA
SDHB	rs786201085	CC
SDHB	rs786203251	GG
SDHB	rs138996609	GG
SDHB	rs200245469	GG
SDHB	rs786202732	AA
SDHB	rs786201161	TT
SDHB	rs786203506	GG
SDHB	rs786201063	CC
SDHB	rs772551056	CC
SDHB	rs786203800	AA
SDHB	rs751000085	GG
SDHB	rs864321636	CC
SDHB	rs876658461	GG
SDHB	rs876658367	CC
SDHB	rs876658451	GG

Complex Diseases: Oncogenic Mutations

SDHC: gastric cancer

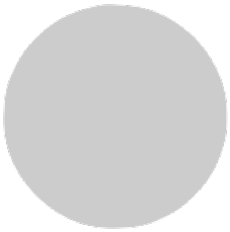
SDHC gene mutations may be related to diseases such gastric cancer.

Your genetic map

Gene	SNP	Genotype
SDHC	rs587776652	GG
SDHC	rs201286421	CC
SDHC	rs786203457	AA
SDHC	rs764575966	CC
SDHC	rs1057517818	GG
SDHC	rs755235380	AA
SDHC	rs981049067	GG
SDHC	rs1131691062	AA
SDHC	rs898854295	AA
SDHC	rs587776653	GG

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/6391

Complex Diseases: Oncogenic Mutations

SDHD: breast, uterine and gastric cancer

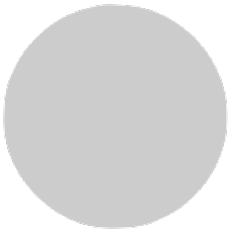
SDHD gene mutations may be related to diseases such breast, uterine and gastric cancer.

Your genetic map

Gene	SNP	Genotype
SDHD	rs80338844	CC
SDHD	rs80338845	GG
SDHD	rs104894302	AA
SDHD	rs104894304	AA
SDHD	rs878854594	CC
SDHD	rs1060503769	GG
SDHD	rs786202403	CC
SDHD	rs786203932	GG
SDHD	rs1060503770	CC
SDHD	rs1050032491	TT

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/6392

Complex Diseases: Oncogenic Mutations

BAP1-related tumor predisposition syndrome

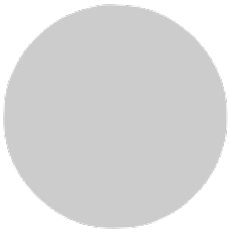
Mutations of the BAP1 gene may be related to diseases such as renal cell carcinoma and breast cancer. In addition, some studies associated this gene, to a lesser extent, with meningioma and ovarian and kidney cancer.

Your genetic map

Gene	SNP	Genotype
BAP1	rs387906848	GG
BAP1	rs864622592	GG
BAP1	rs200156887	AA

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=289539

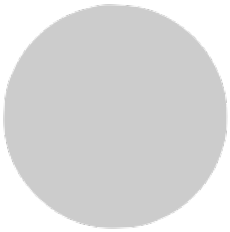
Complex Diseases: Oncogenic Mutations

SMAD4: juvenile polyposis syndrome and colorrectal cancer

SMAD4 gene mutations may be related to diseases such as Juvenile Polyposis Syndrome and colorrectal cancer. Some studies have associated this gene, to a lesser extent, with pancreatic cancer.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/4089

Your genetic map

Gene	SNP	Genotype
SMAD4	rs80338963	CC
SMAD4	rs80338964	CC
SMAD4	rs377767326	CC
SMAD4	rs377767331	CC
SMAD4	rs121912581	GG
SMAD4	rs377767347	GG
SMAD4	rs377767350	TT
SMAD4	rs377767360	CC
SMAD4	rs377767382	TT
SMAD4	rs377767353	GG
SMAD4	rs587781359	CC
SMAD4	rs587781618	GG
SMAD4	rs863224507	TT
SMAD4	rs876660079	GG
SMAD4	rs876660556	GG
SMAD4	rs878854769	GG
SMAD4	rs1060500738	TT
SMAD4	rs1060500733	CC
SMAD4	rs1060500740	TT
SMAD4	rs1316902116	CC
SMAD4	rs377767371	GG
SMAD4	rs281875321	TT
SMAD4	rs281875322	AA
SMAD4	rs397518413	CC
SMAD4	rs730881954	CC
SMAD4	rs876658694	CC
SMAD4	rs1057519739	GG
SMAD4	rs863224400	CC

Complex Diseases: Oncogenic Mutations

SMARCA4: ovarian cancer

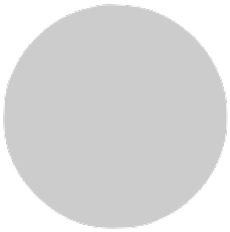
SMARCA4 gene mutations may be related to diseases such ovarian cancer.

Your genetic map

Gene	SNP	Genotype
SMARCA	rs281875227	CC
SMARCA	rs587779750	CC

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/6597

Complex Diseases: Oncogenic Mutations

SMARCB1: Familial rhabdoid tumor

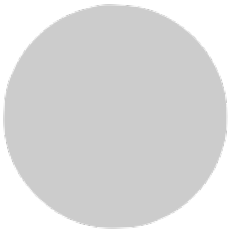
Mutations of the SMARCB1 gene may be related to diseases such as schwannomatosis.

Your genetic map

Gene	SNP	Genotype
SMARCB1	rs797045989	AA

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/6598

Complex Diseases: Oncogenic Mutations

SMARCE1: Familial multiple meningioma

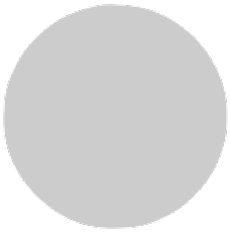
Mutations of the SMARCE1 gene may be related to diseases such as multiple familial meningiomas.

Your genetic map

Gene	SNP	Genotype
SMARCE1	rs387906857	TT

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=263662

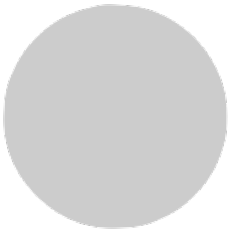
Complex Diseases: Oncogenic Mutations

STK11: breast, ovarian, uterine, colorrectal, gastric and pancreatic cancer

STK11 gene mutations may be related to diseases such breast, ovarian, uterine, colorrectal, gastric and pancreatic cancer.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/6794

Your genetic map

Gene	SNP	Genotype
STK11	rs730881975	GG
STK11	rs137853076	AA
STK11	rs137854584	GG
STK11	rs121913315	GG
STK11	rs137853082	GG
STK11	rs137853083	CC
STK11	rs587782018	GG
STK11	rs730881971	GG
STK11	rs730881979	GG
STK11	rs730881984	GG
STK11	rs786202134	CC
STK11	rs786201090	CC
STK11	rs730881976	CC
STK11	rs863224448	GG
STK11	rs876658584	AA
STK11	rs886037926	AA
STK11	rs886037859	AA
STK11	rs886039554	GG
STK11	rs398123406	GG
STK11	rs1057517830	GG
STK11	rs121913324	CC
STK11	rs775595174	GG
STK11	rs786201213	CC
STK11	rs1131690950	GG
STK11	rs1131690925	CC
STK11	rs1131690951	AA
STK11	rs730881973	CC
STK11	rs1131690923	CC
STK11	rs1131690940	CC
STK11	rs1131690921	GG
STK11	rs1131690945	CC

Complex Diseases: Oncogenic Mutations

TERT: Familial melanoma

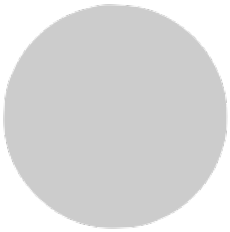
Mutations of the TERT gene may be related to diseases such as familial melanoma.

Your genetic map

Gene	SNP	Genotype
TERT	rs121918666	CC
TERT	rs770066110	GG
TERT	rs797046041	GG

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/7015

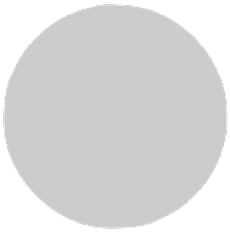
Complex Diseases: Oncogenic Mutations

TP53: Li-Fraumeni syndrome, breast cancer and more

TP53 gene mutations may be related to diseases such Li-Fraumeni Syndrome; and breast, ovarian, uterine, colorrectal and pancreatic cancer. There are some studies that have associated this gene, to a lesser extent, with gastric cancer.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=145

Your genetic map

Gene	SNP	Genotype
TP53	rs121912652	CC
TP53	rs28934575	CC
TP53	rs121912655	CC
TP53	rs11540652	CC
TP53	rs28934873	AA
TP53	rs121912657	CC
TP53	rs28934574	GG
TP53	rs28934578	CC
TP53	rs121912662	AA
TP53	rs28934875	CC
TP53	rs121912664	CC
TP53	rs121912666	TT
TP53	rs121912667	TT
TP53	rs397514495	CC
TP53	rs397516434	GG
TP53	rs397516435	GG
TP53	rs397516439	TT
TP53	rs267605076	CC
TP53	rs201744589	CC
TP53	rs483352695	TT
TP53	rs11540654	CC
TP53	rs587780068	GG
TP53	rs587780071	GG
TP53	rs587780073	TT
TP53	rs587780074	AA
TP53	rs587778720	CC
TP53	rs587781589	AA
TP53	rs587781664	TT
TP53	rs587781702	CC
TP53	rs55832599	GG
TP53	rs17882252	CC

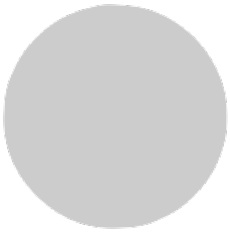
Complex Diseases: Oncogenic Mutations

VHL: Von Hippel-Lindau syndrome

VHL gene mutations may be related to diseases such Von Hippel-Lindau Syndrome.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=892

Your genetic map

Gene	SNP	Genotype
VHL	rs5030823	CC
VHL	rs104893826	GG
VHL	rs5030818	CC
VHL	rs5030820	CC
VHL	rs104893824	TT
VHL	rs5030809	TT
VHL	rs104893825	GG
VHL	rs104893830	GG
VHL	rs28940297	TT
VHL	rs28940301	CC
VHL	rs5030827	GG
VHL	rs5030808	GG
VHL	rs267607170	AA
VHL	rs193922608	CC
VHL	rs193922609	GG
VHL	rs193922610	CC
VHL	rs193922613	AA
VHL	rs5030826	CC
VHL	rs5030802	GG
VHL	rs397516440	CC
VHL	rs397516441	AA
VHL	rs5030817	GG
VHL	rs397516444	GG
VHL	rs397516445	TT
VHL	rs5030804	AA
VHL	rs398123481	CC
VHL	rs587780077	GG
VHL	rs727504215	GG
VHL	rs730882034	CC
VHL	rs119103277	GG
VHL	rs730882032	GG

Complex Diseases: Oncogenic Mutations

WT1: Nephroblastoma

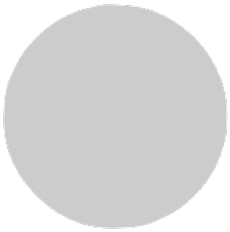
Mutations of the WT1 gene may be related to diseases such as rare malignant renal and Wilms tumors.

Your genetic map

Gene	SNP	Genotype
WT1	rs121907909	GG
WT1	rs28942089	GG
WT1	rs121907900	GG
WT1	rs121907901	CC
WT1	rs121907902	TT
WT1	rs28941778	CC
WT1	rs587776576	CC
WT1	rs121907906	GG
WT1	rs587776577	GG
WT1	rs121907910	GG
WT1	rs1423753702	GG

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/7490

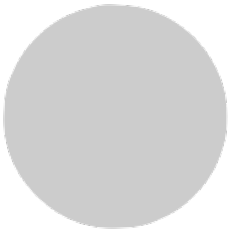
Complex Diseases: Oncogenic Mutations

Familial adenomatous polyposis

Familial adenomatous polyposis (FAP) is characterized by the development of hundreds to thousands of adenomas in the rectum and colon during the second decade of life.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=733

Your genetic map

Gene	SNP	Genotype
APC	rs387906230	TT
APC	rs137854568	CC
APC	rs137854569	CC
APC	rs137854570	CC
APC	rs121913327	CC
APC	rs137854572	CC
APC	rs137854573	CC
APC	rs137854574	CC
APC	rs137854577	CC
APC	rs137854580	CC
APC	rs137854582	TT
APC	rs199740875	GG
APC	rs141576417	CC
APC	rs397515734	CC
APC	rs74953290	CC
APC	rs77056664	CC
APC	rs398123116	GG
APC	rs398123117	CC
APC	rs398123121	CC
APC	rs587779780	CC
APC	rs587779783	CC
APC	rs587779786	AA
APC	rs587779798	GG
APC	rs587781392	CC
APC	rs587781809	TT
APC	rs587782518	CC
APC	rs587783029	CC
APC	rs587783035	AA
APC	rs376213437	TT
APC	rs730881240	CC
APC	rs145945630	CC

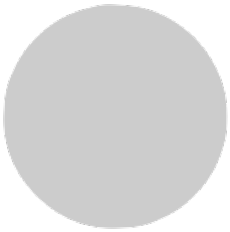
Complex Diseases: Oncogenic Mutations

Kenny-Caffey syndrome

A rare inherited cancer-predisposing syndrome characterized by predisposition to a wide variety of cancers, including neoplasms of the digestive tract, urinary tract, kidney, endometrium, ovary, brain, and prostate, as well as sebaceous skin tumors, depending on the gene involved. Tumors may occur at any age but often arise in young people. Factors influencing individual tumor risk include sex, age, affected gene, and personal history of cancer.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=144

Your genetic map

Gene	SNP	Genotype
AIMP2	rs587779333	TT
EPM2AIP1	rs63750580	AA
EPM2AIP1	rs267607706	CC
EPM2AIP1	rs587778967	AA
EPM2AIP1	rs111052004	TT
EPM2AIP1	rs72481822	GG
EPM2AIP1	rs63750648	AA
EPM2AIP1	rs63750706	CC
MLH1	rs63750781	CC
MLH1	rs193922370	GG
MLH1	rs267607816	GG
MLH1	rs267607713	GG
MLH1	rs267607832	GG
MLH1	rs267607837	GG
MLH1	rs63750193	TT
MLH1	rs63751596	GG
MLH1	rs63751460	CC
MLH1	rs63749792	CC
MLH1	rs63750610	CC
MLH1	rs63751202	TT
MLH1	rs63751662	GG
MLH1	rs267607884	GG
MLH1	rs63750603	GG
MLH1	rs63750561	GG
MLH1	rs63751022	GG
MLH1	rs63749859	TT
MLH1	rs63749990	TT
MLH1	rs587778998	AA
MLH1	rs63750266	GG
MLH1	rs267607727	GG
MLH1	rs63750453	GG

Complex Diseases: Multivariate Analysis

Septic shock

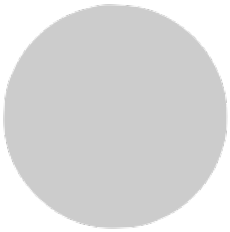
Septic shock is a highly serious condition in the development of sepsis. Its symptoms generally match those of this condition, but usually also include dangerously low blood pressure, a decrease in the amount of urine produced, and changes in mental status. These profound circulatory, cellular, and metabolic abnormalities, specific to septic shock, are associated with a higher risk of mortality than in sepsis, making it a critical condition. DNA also plays an essential role in this condition, as the SFTPB and TNFAIP3 genes have been linked to genetic susceptibility to septic shock.

Your genetic map

Gene	SNP	Genotype
SFTPB	rs1130866	AG
TNFAIP3	rs6920220	GG

Multivariate analysis

What do your genetics tell us?



Based on your genotype, you are not particularly predisposed to septic shock. Other genetic and clinical factors may play a role.

More information:

https://dx.doi.org/10.1097/01.ccm.0000124872.55243.5a

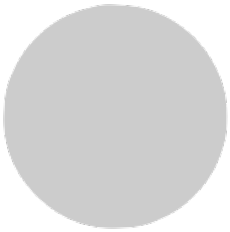
Complex Diseases: Multivariate Analysis

TSC1: tuberous sclerosis complex 1

TSC1 gene mutations may be related to diseases such as tuberous sclerosis complex 1.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/7248

Your genetic map

Gene	SNP	Genotype
TSC1	rs118203419	CC
TSC1	rs118203447	AA
TSC1	rs118203426	AA
TSC1	rs118203537	GG
TSC1	rs118203542	GG
TSC1	rs118203549	GG
TSC1	rs118203345	AA
TSC1	rs118203606	GG
TSC1	rs118203610	CC
TSC1	rs118203614	CC
TSC1	rs118203631	GG
TSC1	rs118203353	CC
TSC1	rs118203352	TT
TSC1	rs118203647	GG
TSC1	rs118203661	GG
TSC1	rs118203668	GG
TSC1	rs118203680	GG
TSC1	rs118203682	GG
TSC1	rs118203687	CC
TSC1	rs118203727	GG
TSC1	rs118203728	GG
TSC1	rs118203732	GG
TSC1	rs118203384	GG
TSC1	rs118203387	CC
TSC1	rs118203402	CC
TSC1	rs118203403	AA
TSC1	rs118203423	CC
TSC1	rs118203427	GG
TSC1	rs118203434	GG
TSC1	rs118203438	CC
TSC1	rs118203440	TT

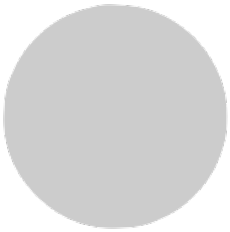
Complex Diseases: Multivariate Analysis

TSC2: tuberous sclerosis complex 2

TSC2 gene mutations may be related to diseases such as tuberous sclerosis complex 2

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

https://www.ncbi.nlm.nih.gov/gene/7249

Your genetic map

Gene	SNP	Genotype
TSC2	rs45483392	CC
TSC2	rs45517179	CC
TSC2	rs28934872	GG
TSC2	rs121964862	CC
TSC2	rs45516293	AA
TSC2	rs45517259	GG
TSC2	rs45517258	CC
TSC2	rs45515894	GG
TSC2	rs45517218	GG
TSC2	rs137854380	AA
TSC2	rs397514994	GG
TSC2	rs397515169	AA
TSC2	rs794727602	AA
TSC2	rs794727906	GG
TSC2	rs796053484	GG
TSC2	rs796053492	GG
TSC2	rs796053509	GG
TSC2	rs773920155	GG
TSC2	rs886041772	CC
TSC2	rs368710573	CC
TSC2	rs886041919	CC
TSC2	rs1057518230	GG
TSC2	rs1057523509	TT
TSC2	rs1057521562	GG
TSC2	rs1060499676	CC
TSC2	rs1060500931	CC
TSC2	rs1060500972	GG
TSC2	rs1060500924	GG
TSC2	rs1064796970	GG
TSC2	rs1085307853	GG
TSC2	rs1131691794	GG

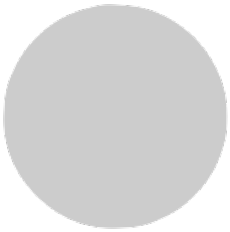
Viruses, Bacteria and Fungi

The severity of COVID-19 infection

Coronavirus (COVID-19) is an infectious disease caused by the SARS-CoV-2 virus, which caused a global pandemic in 2020. Severe disease status occurs in 5% of patients overall and 22 % of patients hospitalized, and it may assume that those affected require mechanical ventilation due to respiratory failure; who suffer from other organ failures such as coagulopathy, acute myocardial or renal lesions; and in the worst case, death. Preventing the progression toward the critical state of the disease is essential to reduce the mortality rate. A 2020 study, in which hundreds of international institutions and companies collaborated (24Genetics among them) demonstrated the interrelationship of genetics and Covid-19 since it was possible to verify that the TYK2 gene was related to the genetic predisposition to evolve towards the severe condition of Covid-19.

GWAS analysis

What do your genetics tell us?



According to this study, you are predisposed to evolve to a severe state of this disease, similar to most of the population. Other genetic and clinical factors may play a role.

More information:

https://www.nature.com/articles/s41586-021-03767-x

Your genetic map

Gene	SNP	Genotype
APCDD1L	rs117463534	CC
DPP9	rs2109069	GG
FYCO1	rs13079478	GG
IFNAR2	rs1131964	TC
KAT7	rs3785928	GG
LAMB1	rs2237698	CC
LOC1053	rs676314	AA
LOC1053	rs79708423	CC
LOC1053	rs4076440	AG
NLN	rs114969787	CC
OAS3	rs10735079	AG
THBS3	rs35154152	TT
TNFSF15	rs6478109	GG
TYK2	rs2304256	AC
Unknown	rs1264701	GG

Viruses, Bacteria and Fungi

Severe Acute Respiratory Syndrome (SARS)

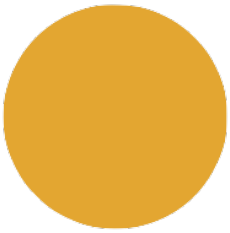
Severe acute respiratory syndrome (SARS) is a highly infectious disease caused by the SARS-CoV virus, which can cause severe lung infections in humans. Initial symptoms often include fever, headache, and muscle pain, followed by respiratory symptoms such as cough, shortness of breath, and pneumonia. In addition, SARS patients often show a decrease in the number of lymphocytes in the blood, which usually affects the severity of the disease. Personal genetics play an important role in predisposing to SARS-CoV infection. Specifically, specific variants in genes such as MBL2, IFNG, and CCL2 have been associated with a greater predisposition to suffer from SARS. Therefore, understanding the genetics of SARS may provide valuable information for developing new treatments and preventive measures for the disease.

Your genetic map

Gene	SNP	Genotype
IFNG	rs2430561	TA
CCL2	rs1024611	AA
MBL2	rs1800450	CC

Multivariate analysis

What do your genetics tell us?



Depending on your genotype, you are predisposed to SARS. Other genetic and clinical factors may play a role.

More information:

https://www.journalofinfection.com/article/S0163-4453(15)00090-0/pdf

Viruses, Bacteria and Fungi

HIV Transmission

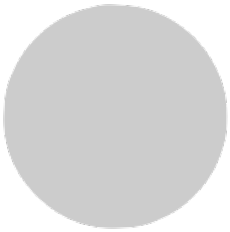
HIV-1 (Human Immunodeficiency Virus type 1) is a virus that usually weakens the immune system of infected people and evolves towards Acquired Immune Deficiency Syndrome (AIDS), which facilitates the appearance of opportunistic infections and cancer, whose treatment is more complicated due to the patient's immunosuppressed situation. Transmission occurs through exposure to the infected person's blood and other body fluids, so sexual contact is one of the main routes of infection. In the genetic field, it has been found that the TLR8-AS1 gene has been linked to HIV infection in women.

Your genetic map

Gene	SNP	Genotype
TLR7	rs179012	GG
IL4	rs2243250	CC
TLR8 AS1	rs3764880	AG
TLR2	rs3804099	TC

Multivariate analysis

What do your genetics tell us?



Based on your genotype, you are not particularly predisposed to HIV-1 infection. Other genetic and clinical factors may play a role.

More information:

https://pubmed.ncbi.nlm.nih.gov/18605904/

Viruses, Bacteria and Fungi

Genital herpes

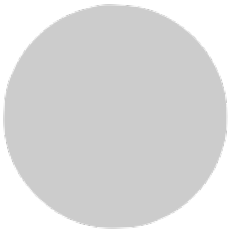
Genital herpes, or herpes simplex virus type 2 (HSV-2), is a common viral infection that causes blisters and sores on the genital area of infected people. It is a highly contagious disease that is spread through sexual contact. The infected person can transmit the virus from the time it begins to incubate until a week after the appearance of the skin lesions. There is no cure for genital herpes, and antivirals only mitigate the frequency of outbreaks. Additionally, other specific medications can be taken to treat the symptoms. Genetics plays a vital role in predisposing to genital herpes virus infection. It has been verified that specific genetic variants in the TLR3 gene are linked to a lower predisposition to contracting the herpes simplex virus type 2.

Your genetic map

Gene	SNP	Genotype
TLR3	rs13126816	GG
TLR3	rs3775291	CC

Multivariate analysis

What do your genetics tell us?



Depending on your genotype, you do not have a particular predisposition to herpes simplex virus type 2 infection. Other genetic and clinical factors may play a role.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/22552940

Viruses, Bacteria and Fungi

Cirrhosis due to Hepatitis B

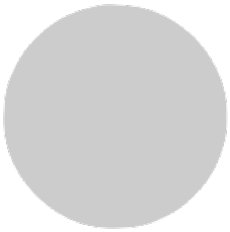
Hepatitis B is a severe liver infection caused by the Hepatitis B Virus (HBV). It is usually a brief infection, but sometimes it becomes chronic, increasing the risk of developing liver failure, liver cancer, or cirrhosis. Cirrhosis is a liver disease that causes lesions in the form of fibrosis when the hepatitis B virus attacks the liver, resulting in severe damage with a consequent increased risk of liver cancer. Symptoms of hepatitis B infection are usually non-existent until cirrhosis develops. Genetics is vital in predisposing to hepatitis B-related liver cirrhosis. Mutations in genes such as STAT4 and NOD2 are related to the predisposition to suffer from these pathologies.

Your genetic map

Gene	SNP	Genotype
ESR1	rs2234693	TC
LOC1053	rs2227982	GG
NOD2	rs2066845	GG
NOD2	rs2066844	CC
STAT4	rs7574865	GG
TLR3	rs3775290	TC

Multivariate analysis

What do your genetics tell us?



Based on your genotype, you are not particularly predisposed to hepatitis B-related cirrhosis. Other genetic and clinical factors may play a role.

More information:

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6616055/

Viruses, Bacteria and Fungi

Community-acquired pneumonia

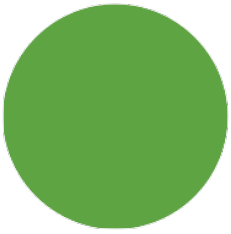
The so-called Community-Acquired Pneumonia (CAP), or community-acquired pneumonia, refers to pneumonia, in any of its variants, contracted by a person outside the health system, that is, in daily life. CAP is a lung infection that can be caused by multiple microorganisms (bacteria, viruses, and fungi), affects people of all ages, and occurs as a result of the oxygen-absorbing areas of the lung (alveoli) filling up. Consequently, the lung inhibits its function, causing symptoms such as dyspnea, fever, chest pain, and cough. The treatment for this pathology usually depends on the microorganism that has generated it. Genetics plays an essential role in the development of this disease, as variants in the IL6-AS1 gene have been linked to developing community-acquired pneumonia.

Your genetic map

Gene	SNP	Genotype
CYP1A1	rs2606345	AC
IL6 AS1	rs1800795	GC
TNFRSF1	rs1061622	TG

Multivariate analysis

What do your genetics tell us?



According to your genotype, you have a low predisposition to suffer from community-acquired pneumonia. Other genetic and clinical factors may play a role.

More information:

https://pubmed.ncbi.nlm.nih.gov/19900796/

Viruses, Bacteria and Fungi

Severe hospital pneumonia

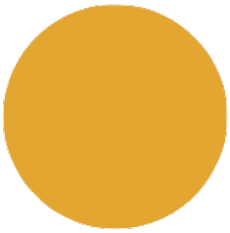
Hospital Acquired Pneumonia (HAP), or nosocomial pneumonia, is a hospital-acquired lung infection that usually presents in patients 48-72 hours after admission. Bacteria mainly cause this disease, although viruses and fungi can also cause it, and it is the second most common nosocomial infection (15-20% of the total) after urinary tract infections. In general, nosocomial pneumonia is a severe and life-threatening disease, and genetics may play an important role in susceptibility to developing a severe stage of pneumonia. It has been verified that people with specific variants in the ABCB1 and AGTR1 genes have a greater predisposition to hospital pneumonia leading to a more extended hospital stay.

Your genetic map

Gene	SNP	Genotype
ABCB1	rs1045642	AA
AGTR1	rs5186	AA

Multivariate analysis

What do your genetics tell us?



Based on your genotype, you have a high predisposition to severe hospital-acquired pneumonia. Other genetic and clinical factors may play a role.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/24127120

Viruses, Bacteria and Fungi

Bronchitis

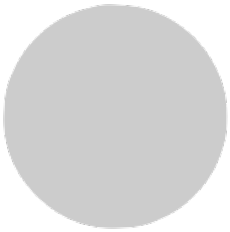
Bronchitis is a respiratory disease caused by inflammation of the bronchi, which causes coughing, wheezing, shortness of breath, and chest pain. Although environmental factors, such as exposure to tobacco smoke and air pollutants, can influence the development of the disease, genetics also play a role. Specifically, the LOC100287329 gene has been related to a genetic predisposition to bronchitis. This gene produces a protein called alpha-lymphoid tumor necrosis factor, which is involved in our body's inflammatory response. Research has shown that specific genetic variants of the LOC100287329 gene may increase the susceptibility to bronchitis. Therefore, understanding the role of the LOC100287329 gene in the development of bronchitis could help to develop new therapeutic approaches for the disease.

Your genetic map

Gene	SNP	Genotype
LOC1002	rs909253	AG
LOC1002	rs1041981	AC

Multivariate analysis

What do your genetics tell us?



According to your genotype, you do not have a particular predisposition to suffer from bronchitis. Other genetic and clinical factors may play a role.

More information:

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5524954/pdf/41598_2017_Article_6791.pdf

Allergies and Intolerances

Lactose intolerance

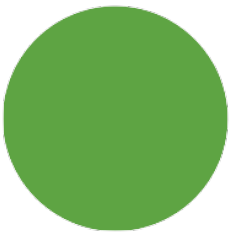
Lactose is the main naturally-occurring sugar in milk and dairy products. It consists of a glucose molecule and a galactose molecule, two simple sugars that the body uses to produce energy. The enzyme lactase is essential for breaking down lactose into glucose and galactose, a key step in certain immune and neuronal processes. Some people cannot produce enough lactase; as a result, they do not digest lactose, which ferments in the intestine, generating gas, digestive distress, abdominal distension, and/or diarrhoea.
There are genetic factors that play an important role in lactose absorption, such as the MCM6 gene, which is directly related to this process.

Your genetic map

Gene	SNP	Genotype
MCM6	rs4988235	AA

Monovariant analysis

What do your genetics tell us?



Based on your genotype, you are predisposed to metabolise lactose easily. Other genetic and clinical factors may be relevant.

More information:

https://onlinelibrary.wiley.com/doi/full/10.1002/jbmr.83

Allergies and Intolerances

DAO deficiency and migraines

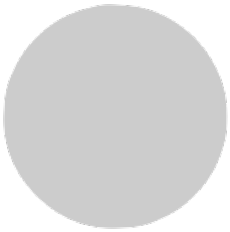
Diamine oxidase (DAO) is the enzyme responsible for reducing histamine, which is a molecule the body uses to respond to substances it considers harmful. With a DAO deficiency, histamine builds up, causing allergies and bothersome symptoms, which can be worsened by eating foods that contain high levels of histamine, such as tomatoes, fish preserves, processed sauces, dairy products and other foods. One of the best-known consequences of DAO deficiency is migraines, but dizziness, irritable bowel syndrome, Crohn's disease, stomach pain, nausea and/or vomiting, abnormal blood pressure and arrhythmias can also occur.
The AOC1 gene is responsible for producing the DAO enzyme, and several studies confirm that mutations in this gene create a propensity for this process to malfunction, with the consequent generation of reduced levels of DAO.

Your genetic map

Gene	SNP	Genotype
AOC1	rs2052129	GG
AOC1	rs1049793	CC

Multivariate analysis

What do your genetics tell us?



Based on your genotype, your predisposition to have reduced DAO enzyme activity is average. Other genetic and clinical factors may be relevant.

More information:

https://pubmed.ncbi.nlm.nih.gov/21488903/

Allergies and Intolerances

Shellfish allergy

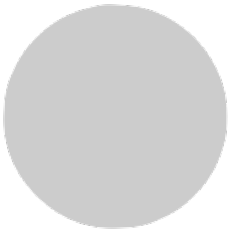
Shellfish allergy is a critical immune system reaction to proteins present mainly in crustaceans. Shrimp and other shellfish are one of the most common sources of food allergies. The symptoms are multiple and can vary from slight irritation in the area in contact with food (lips, tongue, mouth) or inflammation in the throat area, which can make breathing difficult or even impossible, to a life-threatening reaction called anaphylaxis. At the genetic level, mutations in the TH2LCRR gene have been associated with an increased risk of developing an allergy to shrimp and, by analogy, to other crustaceans.

Your genetic map

Gene	SNP	Genotype
IL13	rs20541	GG
TH2LCRR	rs1800925	CC
Unknown	rs9275596	TC

Multivariate analysis

What do your genetics tell us?



Based on your genotype, your predisposition to shellfish allergy is standard. Other genetic and clinical factors may play a role.

More information:

https://pubmed.ncbi.nlm.nih.gov/33175217/

Allergies and Intolerances

Mercury Accumulation

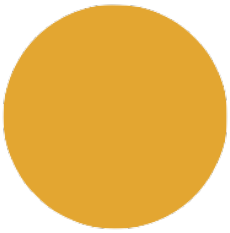
Mercury is a heavy metal, which reaches the body of people mainly through the ingestion of fish, is absorbed by the intestinal tract, transported through the blood, and accumulated in different body organs. Elevated levels of this heavy metal can cause damage to the gastrointestinal tract, nervous system, and kidneys, especially in infants, children, and pregnant women. At the genetic level, it has been proven that some individuals may have an easier time accumulating mercury in their blood due to their genetics. Specifically, the GCLC and GSTP1 genes code for an enzyme that helps detoxify the body of toxic compounds such as mercury and reduce cell damage.

Your genetic map

Gene	SNP	Genotype
GSTP1	rs1138272	TC
GCLC	rs17883901	GG

Multivariate analysis

What do your genetics tell us?



You are predisposed to accumulate mercury in your blood depending on your genotype. Other genetic and clinical factors may play a role.

More information:

https://pubmed.ncbi.nlm.nih.gov/16599007/

Allergies and Intolerances

Allergic rhinitis

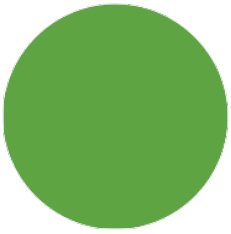
Allergic rhinitis is inflammation of the nasal mucosa, the symptoms of which are similar to those of a cold: nasal itching, sneezing, runny nose and nasal congestion, red and watery eyes, coughing, and itchy palate. Sometimes it can cause asthma or eczema. Its cause is exposure to specific allergens, mainly pollen, dust mites, fungi, or animal epithelia. Symptoms usually appear shortly after contact with the allergen. Specific immunotherapy is sometimes used for its treatment, which consists of the controlled administration of an extract of the substance that the patient is allergic to until their symptoms decrease. The condition may or may not be heritable. Still, at the genetic level, the correlation of the GLI3 gene with allergic rhinitis has been verified, which suggests an essential role in the predisposition to suffer from this pathology.

Your genetic map

Gene	SNP	Genotype
GLI3	rs4724100	TT
Unknown	rs6898653	GG
Unknown	rs216518	CC
Unknown	rs2155219	TG
Unknown	rs17513503	CC

GWAS analysis

What do your genetics tell us?



According to this study, you are less predisposed to suffering from this disease than most of the population. Other genetic and clinical factors may play a role.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/23817571

Allergies and Intolerances

Allergy to grass pollen

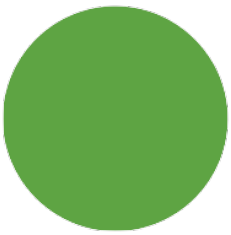
Grasses are monocotyledonous herbaceous plants with more than 800 genera and 12,000 known species, including wheat, canary grass, oats, rice, sugarcane, grass, and weeds. Their pollen is known to cause allergies in many people, manifesting in symptoms such as nasal congestion, watery eyes, hives, and even anaphylactic shock in extreme cases. Genetics plays an important role in this type of allergy, as demonstrated by variants in the DNAH5 gene, among others, which are correlated with a higher or lower predisposition to grass allergies.

Your genetic map

Gene	SNP	Genotype
HLA	rs7775228	TT
LOC1019	rs631208	AG
DNAH5	rs6554809	TC
Unknown	rs7617456	AG
Unknown	rs2155219	TG
Unknown	rs17513503	CC

GWAS analysis

What do your genetics tell us?



According to this study, you are less predisposed to suffering from this disease than most of the population. Other genetic and clinical factors may play a role.

More information:

https://pubmed.ncbi.nlm.nih.gov/23817571/

Biomarkers and Others

Calcium levels

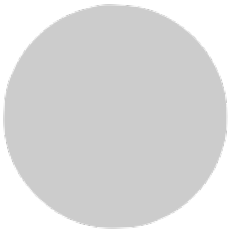
Calcium is vital to the normal functioning of multiple organ systems, and its serum concentration is tightly regulated.

Your genetic map

Gene	SNP	Genotype
CASR	rs1801725	GG
DGKD	rs1550532	GG
GCKR	rs780094	TC
LINC007	rs10491003	TC
CARS1	rs7481584	AG
LOC1053	rs7336933	AG
CYP24A1	rs1570669	AA
WDR81	rs12150338	CC

GWAS analysis

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/24068962

Biomarkers and Others

Phosphorus levels

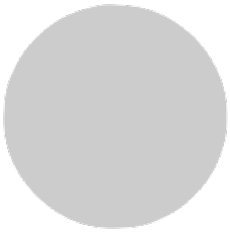
Phosphorus is an essential mineral that sustains cellular energy and mineralizes the skeleton. Because the complex actions of ion transporters and regulatory hormones regulate serum phosphorus concentrations, genetic variation may determine inter-individual variations in phosphorus metabolism.

Your genetic map

Gene	SNP	Genotype
NBPF3	rs1697421	TT
CSTA	rs17265703	AA
IP6K3	rs9469578	CC
PDE7B	rs947583	TT
FERRY3	rs2970818	TT

GWAS analysis

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/20558539

Biomarkers and Others

Magnesium levels

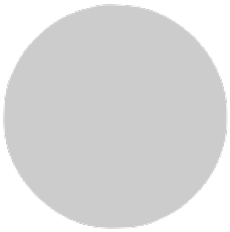
Magnesium, potassium, and sodium, cations commonly measured in serum, are involved in many physiological processes, including energy metabolism, nerve and muscle function, signal transduction, and fluid and blood pressure regulation.

Your genetic map

Gene	SNP	Genotype
MUC1	rs4072037	TC
SHROOM	rs13146355	GG
LOC1079	rs7965584	AA
LOC1019	rs3925584	TT
LOC1001	rs2592394	GG
MECOM	rs448378	AG

GWAS analysis

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/20700443

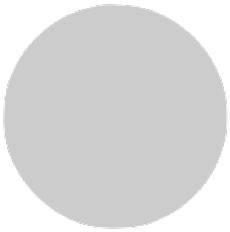
Biomarkers and Others

Plasma omega-6 polyunsaturated fatty acid levels (dihomo-gamma-linolenic acid)

Omega6 (n6) Polyunsaturated Fatty Acids (PUFAs) and their metabolites are involved in cell signaling, inflammation, clot formation, and other crucial biological processes. Genetic components, such as variants of Fatty Acid Desaturase (FADS) genes, determine the composition of n6 PUFAs.

GWAS analysis

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/24823311

Your genetic map

Gene	SNP	Genotype
PDXDC1	rs2280018	AA
TMEM25	rs102275	TC
IL23R	rs7517847	TT
C10orf12	rs17009617	GG
FADS1	rs174550	TC
FADS2	rs2727270	CC
PDXDC1	rs1136001	GG
FTLP19	rs2069036	CC
FADS1	rs174547	TT
PDXDC1	rs4985155	AG
TMEM39	rs16829840	CC
PDXDC1	rs1741	GC
ELOVL2	rs2236212	CC
FADS1	rs174555	TC

Biomarkers and Others

Beta-2 microglubulin plasma levels

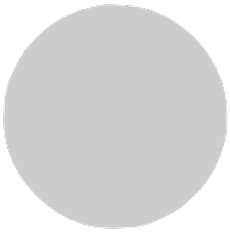
Beta-2-microglobulin (B2M) is a protein found on the surface of many cells, including those that make up the immune system and can therefore be considered a marker of immune defense system activity. If found in high levels, it may indicate an overactive defense system or the presence of disease, although it is not diagnostic of a specific illness. On the other hand, low levels of this protein may indicate a compromised immune system or the presence of kidney or cardiovascular disease. Genetics plays a vital role in regulating this protein, and, in particular, variants in the TRIM31 gene, among others, can influence circulating beta -2-microglobulin levels.

Your genetic map

Gene	SNP	Genotype
TRIM31	rs2023472	GG
HLA B	rs2523608	AG
MICA AS1	rs16899524	CC
SH2B3	rs3184504	CC

GWAS analysis

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/23417110

Biomarkers and Others

Glycated hemoglobin levels

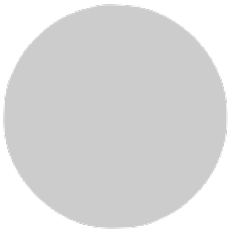
Glycosylated hemoglobin, or HbA1c, measures the average blood sugar level over the past two to three months. It is a crucial indicator as a measure of glycaemic control and also as a diagnostic criterion for diabetes, as it helps to determine how well the disease is being controlled. A high level may indicate inadequate control and an increased risk of diabetes complications, while a good level suggests that diabetes is either not present or under control. Scientific studies have confirmed that variants in the FADS2 gene, among others, can influence glycosylated hemoglobin levels, thus ensuring the influence of genetics on this marker.

Your genetic map

Gene	SNP	Genotype
SMG5	rs6684514	GG
LOC1079	rs9399137	TC
FADS2	rs174570	CC
PIEZO1	rs9933309	CC
MYO9B	rs11667918	CC
ANK1	rs4737009	GG
FN3KRP	rs1046875	GG
ABCB11	rs3755157	CC
CDKAL1	rs7772603	TT
GCK	rs1799884	CC
SLC30A8	rs13266634	CC

GWAS analysis

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/24647736

Biomarkers and Others

Serum total protein level

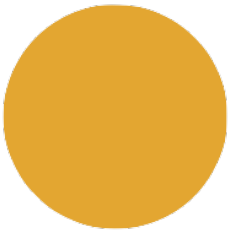
We could say that serum is the liquid part of blood that remains after blood cells (such as red blood cells and white blood cells) and platelets have been removed, and contains elements such as water, salts, sugars, proteins, and other compounds necessary for the functioning of your body. The proteins present in blood serum play a crucial role in modulating and monitoring multiple biological processes in our body and are not only a reflection of our general health and nutritional status but can also be affected by diseases, infections, and nutritional imbalances, such as malnutrition, cancer, and cardiovascular, renal and inflammatory diseases. At the genetic level, variants in the RPS11 gene, among others, have been confirmed to have the ability to influence predisposition to abnormal serum protein levels.

Your genetic map

Gene	SNP	Genotype
TNFRSF1	rs4561508	CC
intergeni	rs204999	AG
TNFRSF1	rs4561508	CC
GCKR	rs1260326	TC
ARID5B	rs2675609	CC
RPS11	rs2280401	AA
TNFRSF1	rs4561508	CC
intergeni	rs204999	AG
ELL2	rs3777200	CC
GCKR	rs1260326	TC
RPS11	rs2280401	AA

GWAS analysis

What do your genetics tell us?



According to this study, you are more prone than the average person to suffering abnormal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/23022100

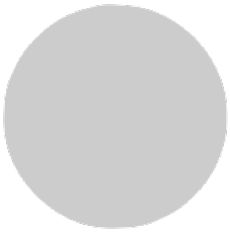
Biomarkers and Others

GGT levels

GGT (Gamma Glutamyl Transferase) is a type of liver enzyme essential in the metabolic process of amino acids, which stands out for its ability to diagnose potential liver disorders. Low GGT, in many cases, is not due to a disease but simply to an unbalanced diet with specific nutrient and vitamin deficiencies. However, elevated blood levels may indicate liver disease or damage to the bile ducts, the tubes through which bile enters and exits the liver. Environmental factors, such as alcohol intake, certain medications, and some diseases, can directly affect these levels, but we also find a determining influence in our genetic inheritance. Specifically, specific gene variants, such as PNPLA3, can influence GGT levels in the blood.

GWAS analysis

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/22001757

Your genetic map

Gene	SNP	Genotype
PNPLA3	rs738409	CC
RNU6	rs6984305	TT
LOC1053	rs10819937	GG
ABO	rs579459	TC
JMJD1C	rs7923609	GG
FADS2	rs174601	TC
ST3GAL4	rs2236653	TT
ASGR1	rs314253	TT
ABHD12	rs7267979	GG
LOC1019	rs1497406	AG
CEPT1	rs1335645	AA
EFHD1	rs2140773	AA
SLC2A2	rs10513686	GG
HPRT1P2	rs6888304	AA
MLXIPL	rs17145750	TC
DLG5	rs754466	AA
EXOC3L4	rs944002	AG
RORA	rs339969	AC
CD276	rs8038465	CC
LOC1027	rs4581712	AA
SOX9 AS1	rs9913711	CC
FUT2	rs516246	TC
MICAL3	rs1076540	TC
GGT1	rs2073398	CC

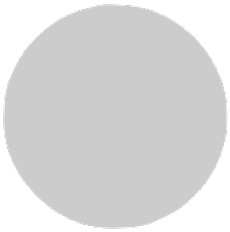
Biomarkers and Others

Glycerophospholipid levels

Phosphoglycerides or glycerophospholipids are lipid molecules of the phospholipid group that form the cell membrane and, therefore, play a vital role in the structure and function of membranes, especially in cell signaling and recognisability. An adequate proportion of these lipids is essential for the proper functioning and structure of cells, and changes in their levels can affect membrane fluidity and functionality. Genetics is an influential factor, and scientifically validated studies have identified variants in the MYRF gene, among others, as a determining factor in regulating glycerophospholipids.

GWAS analysis

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/26068415

Your genetic map

Gene	SNP	Genotype
PKD2L1	rs603424	GG
MYRF	rs174536	AA
MYRF	rs174537	GG
TMEM25	rs102275	TC
TMEM25	rs102275	TC
TMEM25	rs102275	TC
TMEM25	rs102275	TC
TMEM25	rs102275	TC
TMEM25	rs102275	TC
TMEM25	rs102275	TC
TMEM25	rs102275	TC
TMEM25	rs102275	TC
FADS1	rs174546	TC
FADS1	rs174546	TC
FADS1	rs174547	TT
FADS1	rs174547	TT
FADS1	rs174547	TT
FADS1	rs174547	TT
FADS1	rs174547	TT
FADS1	rs174547	TT
FADS1	rs174547	TT
FADS1	rs174550	TC
FADS1	rs174555	TC
FADS2	rs968567	CC
FADS2	rs1535	AG
FADS2	rs1535	AG
FADS2	rs174576	CC
FADS2	rs174578	TA
FADS2	rs174578	TA
SYNE2	rs7157785	GG
GPHN	rs1077989	AC
GPHN	rs1077989	AC

Biomarkers and Others

Serum albumin level

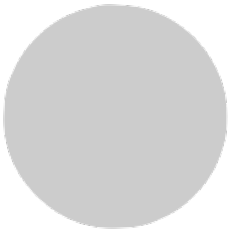
Albumin is a protein produced by the liver that stands out as the most prevalent protein in blood serum. It is vital for regulating osmotic balance, the relationship between the fluids inside the cell (intracellular) and its external environment (extracellular), and for transporting various molecules. A decreased albumin level can be a warning sign of possible kidney or liver disease; low albumin levels usually indicate dehydration. In any case, either too high or too low, abnormal levels are not necessarily associated with a health problem. It has been shown that certain medications can have an impact on albumin levels, and genetics is also an important influencing factor. Specifically, variants in genes, such as FRMD5, have been identified that influence serum albumin concentration.

Your genetic map

Gene	SNP	Genotype
MIR22HG	rs11078597	TT
ACTBP9	rs694419	CC
RPS11	rs2280401	AA
FRMD5	rs16948098	GG
TNFRSF1	rs4561508	CC
FKBPL	rs204999	AG
LOC1079	rs2675609	CC
HPN AS1	rs11671010	TC
CHRNA3	rs12914385	TC
ELL2	rs3777200	CC

GWAS analysis

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/23022100

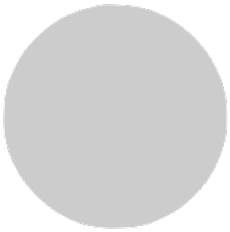
Biomarkers and Others

Phospholipid levels (plasma)

Phospholipids are a source of essential fatty acids and act as critical components in the formation and function of cell membranes, making them vital to ensure optimal cellular health, as well as functioning as a biological vehicle for the absorption of fat-soluble vitamins, such as A, D, E, and K. Stored lipids represent the body's energy pantry and are a source of energy during exercise. Alterations in the balance of these lipids can be a precursor to metabolic dysfunction and cardiovascular problems, among other pathologies. Diet and the individual's metabolism are determining factors in the concentration of these lipids, but scientific studies have shown the influence of genetics in this process. In particular, it has been highlighted that variants in genes such as LCT influence the predisposition to have abnormal levels of phospholipids.

GWAS analysis

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/21829377

Your genetic map

Gene	SNP	Genotype
TMEM25	rs102275	TC
MYRF	rs174536	AA
RPLP0P2	rs1692120	AG
FADS1	rs174547	TT
FADS2	rs1535	AG
FADS2	rs174448	AG
FEN1	rs4246215	GG
LCT	rs16832011	AA
TMEM25	rs174538	AG
MYRF	rs174535	TC
FADS1	rs174550	TC
FADS2	rs174574	AC
ELOVL2	rs3798713	GC
BEST1	rs1109748	AC
LOC1019	rs1514178	TT
ELOVL2	rs3734398	CC
SYCP2L	rs4713103	TT
RAB3IL1	rs2521572	GG
DAGLA	rs198426	TT
GCKR	rs780094	TC
LOC1053	rs9586179	TT
RPS2P37	rs4963452	TT
STIM2	rs6844153	TC
ELOVL2	rs2236212	CC
ELOVL2	rs4711171	CC

Biomarkers and Others

Aortic root size

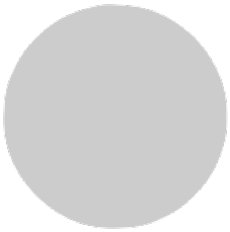
Echocardiographic measures of Left Ventricular (LV) structure and function are heritable phenotypes of cardiovascular disease.

Your genetic map

Gene	SNP	Genotype
SLC35F1	rs89107	GG
TMEM23	rs17132261	CC
SMG6	rs10852932	TG
PRDM6	rs17470137	AG
HMGA2	rs4026608	TT
LINC023	rs10770612	AA
LOXL1	rs893817	AG

GWAS analysis

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/19584346

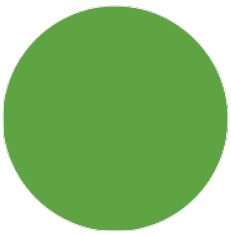
Biomarkers and Others

Heart rate

An elevated resting heart rate is associated with a greater risk of cardiovascular disease.

GWAS analysis

What do your genetics tell us?



According to this study, you are more prone than the average person to having normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/23583979

Your genetic map

Gene	SNP	Genotype
TFPI	rs4140885	GG
LOC1053	rs180242	AA
RNU3P3	rs17796783	TC
SYT10	rs7980799	CC
LOC1053	rs17287293	AG
CD46	rs11118555	TT
MYH6	rs365990	AA
LOC1053	rs1015451	TT
ACHE	rs13245899	AA
FADS1	rs174549	GG
SLC35F1	rs11153730	TC
KIAA1755	rs6127471	TC
CCDC141	rs17362588	GG
GNB4	rs7612445	GG
CHRM2	rs2350782	TT
NKX2 5	rs6882776	GG
LOC1053	rs13030174	AC
FNDC3B	rs9647379	CG
RFX4	rs2067615	AT
CPNE8	rs826838	TT
RBFOX1	rs11645781	GG
SLC10A7	rs10213084	GG
RNU4	rs11154027	TC
LOC1079	rs11578508	AA
HMGN2P	rs17083533	GG
LOC1019	rs7722600	AA

Biomarkers and Others

Bilirubin levels

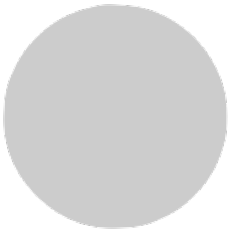
Bilirubin is a yellowish pigment produced during the breakdown of red blood cells, passes through the liver, and is eventually excreted from the body. Lower than average levels are not a concern, but abnormally high levels may indicate that the liver is not eliminating bilirubin properly, which may indicate liver disease or damage. It is, therefore, considered an essential indicator for detecting certain conditions. While liver disease is a common factor influencing these levels, genetics also plays a role. Variations in specific genes, such as UGT1A10, play a role in determining bilirubin levels.

Your genetic map

Gene	SNP	Genotype
UGT1A10	rs6742078	GG
HIST1H1T	rs12206204	CC
ARHGEF7	rs4773330	GG
SLCO1B1	rs4149056	TT

GWAS analysis

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/19414484

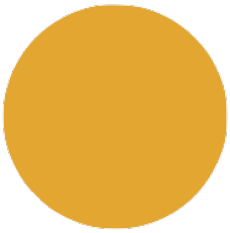
Biomarkers and Others

Thyroid hormone levels

Thyroid hormones, produced by the thyroid gland, play a crucial role in regulating metabolism and growth. Abnormal levels of thyroid hormones affect about 10% of people in their lifetime. Low levels of thyroid hormones lead to hypothyroidism, the consequences of which include tiredness, intolerance to colds, apathy and indifference, depression, reduced memory and mental concentration, dry skin, dry and brittle hair, brittle nails, pale skin, weight gain, persistent constipation, and excessive sleepiness. Conversely, high levels (hyperthyroidism) can cause weight loss or irregular or rapid heartbeat. Variants in the PDE10A gene, among others, have been identified as influencing thyroid hormone levels, supporting the importance of genetics in thyroid hormone levels.

GWAS analysis

What do your genetics tell us?



According to this study, you are more prone than the average person to suffering abnormal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/23408906

Your genetic map

Gene	SNP	Genotype
PDE8B	rs6885099	AG
PDE10A	rs753760	GC
LOC1053	rs10799824	GG
LOC1053	rs3813582	TT
LOC1079	rs9472138	CC
LINC0151	rs11755845	CC
LOC1079	rs10032216	TT
IGFBP	rs13015993	AA
SOX9	rs9915657	TT
NFIA	rs334699	GG
FGF7	rs10519227	TT
PRDM11	rs17723470	TC
DET1	rs17776563	GG
INSR	rs4804416	TG
	rs657152	AC
ITPK1	rs11624776	AA
NRG1	rs7825175	GG
LINC006	rs1537424	TC
SASH1	rs9497965	CC
GLIS3	rs1571583	GG
DIO1	rs2235544	AC
LHX3	rs7860634	AA
PTCSC2	rs7045138	TC
LOC1053	rs11726248	GG
LPCAT2	rs6499766	AA
LOC1005	rs7240777	GG

Biomarkers and Others

Eosinophil levels

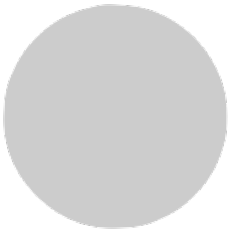
Eosinophils, a variety of white blood cells, are essential for responding to allergies and infections. These cells play a crucial role in the immune response, especially when dealing with parasites and allergic reactions. Diagnostically, high eosinophils may indicate a parasitic infection or an ongoing allergic reaction. On the other hand, low levels may indicate a weakened immune system, stress, or the presence of certain medications, such as beta-blockers or corticosteroids. Genetics has also been shown to be an influential factor, and specifically, variants in the IL1RL1 gene, among others, can correlate with the number of eosinophils in the blood. This relationship gives physicians a clearer picture of an individual's immune health and tailor treatments accordingly.

Your genetic map

Gene	SNP	Genotype
IL1RL1	rs1420101	TC
LOC1027	rs12619285	AG
TMED10P	rs4857855	CC
SH2B3	rs3184504	CC
IRF1 IL5	rs4143832	GG
WDR36	rs2416257	TC
TNXB	rs2269426	AA

GWAS analysis

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/19198610

Biomarkers and Others

Neutrophil levels

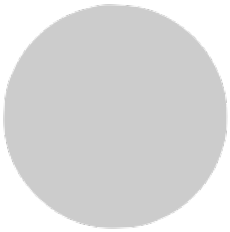
Neutrophils are granulocyte-type leukocytes (white blood cells), also called polymorphonuclears. They are the most common type of white blood cell and play a crucial role in the body's defense against infection. They respond rapidly to the presence of foreign bodies and are essential in the initial phase of the immune response. Low levels of neutrophils (neutropenia) make it difficult for the body to fight infection, making the person more likely to become ill. Increased levels result in a condition known as neutrophilic leukocytosis, which is a normal immune response to infection, injury, inflammation, or certain medications, among other causes. Variations in the CDK6 gene, among others, have been shown to correlate with neutrophil levels in the blood, confirming that genetics is an important influencing factor.

Your genetic map

Gene	SNP	Genotype
CDK6	rs445	CC
Intergeni	rs8078723	TC
Intergeni	rs8078723	TC
PSMD3	rs4794822	CC
PSMD3	rs4794822	CC
AK12388	rs6936204	TC

GWAS analysis

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/25096241

Biomarkers and Others

Interleukin 6 and Inflammation

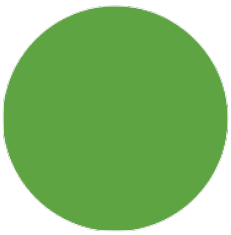
Interleukin 6 (IL-6) is a proinflammatory cytokine contributing to host defense against infection and tissue injury. However, the exaggerated and excessive synthesis of IL-6 while fighting environmental stress leads to a severe and acute systemic inflammatory response known as a "cytokine storm" since high levels of IL-6 can activate the pathway of IL-6. Coagulation and vascular endothelial cells inhibit myocardial function. As previously shown in the literature, increased circulating levels of proinflammatory cytokines are associated with lung inflammation and extensive lung involvement in SARS patients. Genetics also plays a key role, as the IL6R gene has been linked to genetic susceptibility to such inflammation.

Your genetic map

Gene	SNP	Genotype
IL6 AS1	rs1800796	GC
IL6R	rs4537545	CC

Multivariate analysis

What do your genetics tell us?



Depending on your genotype, you are predisposed to maintaining standard levels of interleukin 6. Other genetic and clinical factors may play a role.

More information:

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2668154/pdf/nihms45547.pdf

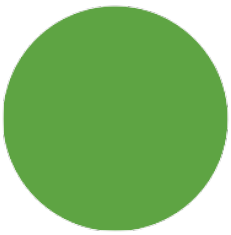
Biomarkers and Others

Platelet levels

Platelets, also known as thrombocytes, are fragments of blood cells produced by the bone marrow that are essential for blood clotting. They contribute to the repair of damaged blood vessels and prevent excessive bleeding. If your blood is low in platelets, it is called thrombocytopenia, and you may be at risk of moderate to severe bleeding. Abnormally high levels of platelets increase the risk of forming blood clots (thrombi) that can block blood flow in the body. If a thrombus moves from the site where it started, it is called an embolism. The thrombus or embolism blocks the supply of oxygen and blood flow to surrounding tissues and can cause significant damage. At the genetic level, studies show that variants in the MFN2 gene, among others, have been identified as influencing blood platelet levels.

GWAS analysis

What do your genetics tell us?



According to this study, you are more prone than the average person to having normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/22139419

Your genetic map

Gene	SNP	Genotype
MFN2	rs2336384	TT
DNM3	rs10914144	TC
TMCC2	rs1668871	TT
GCSAML	rs7550918	TT
TRIM58	rs3811444	TT
EHD3	rs625132	AG
THADA	rs17030845	TT
LOC3398	rs7641175	AA
ARHGEF3	rs1354034	TC
PDIA5	rs3792366	AG
KLHL8	rs7694379	GG
F2R	rs17568628	TT
MEF2C	rs700585	TC
IRF1	rs2070729	AC
CARMIL1	rs441460	AA
HLA B	rs3819299	TT
HLA DOA	rs399604	TT
BAK1	rs210134	GG
LOC1079	rs9399137	TC
CTB	rs342275	TC
HYAL4	rs4731120	AA
PLEC	rs6995402	TC
AK3	rs409801	TC
RCL1	rs13300663	GG
CDKN2A	rs3731211	TA
PSMD13	rs505404	TT
FEN1	rs4246215	GG
CBL	rs4938642	GG
LOC1053	rs7342306	GG
BAZ2A	rs941207	CC
SH2B3	rs3184504	CC

Biomarkers and Others

White blood cell count

White blood cells are a type of blood cell that is produced in the bone marrow and found in blood and lymphatic tissues. White blood cells are part of the body's immune system. These help the body fight infections and other diseases. The types of white blood cells are granulocytes (neutrophils, eosinophils, and basophils), monocytes, and lymphocytes (T cells and B cells).

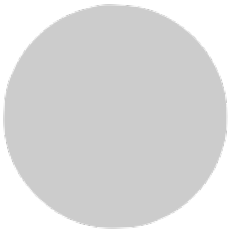
White blood cell count is a common clinical measurement of whole blood count tests, and varies widely among healthy individuals.

Your genetic map

Gene	SNP	Genotype
LINC0156	rs4328821	AA
EPS15L1	rs10411936	AG
LOC1019	rs1449263	TC
LINC0156	rs9880192	GC
CCDC26	rs10098310	AG
LOC1053	rs10980800	TT
PSMD3	rs8078723	TC
HCG22	rs2517510	TG
PSMD3	rs4794822	CC

GWAS analysis

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/21738480

Biomarkers and Others

Monocyte levels

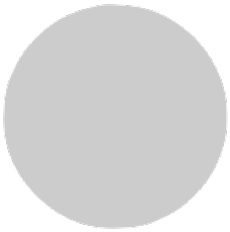
Monocytes belong to the agranulocytes, a type of white blood cell or leukocyte. They are part of the immune system, responsible for fighting particular infections, and are also involved in the inflammatory response. Having altered levels of monocytes may mean that the immune system is weakened or fighting a disease. In both cases, this is usually related to infectious processes, such as a virulent flu, a blood infection, viral infections, infectious mononucleosis, mumps, measles, or parasitic infections. Occasionally, specific medical treatments can also influence monocyte levels. In addition to all these diseases, genetics can play a role, and variants in the ITGA4 gene, among others, have been shown to influence blood monocyte levels.

Your genetic map

Gene	SNP	Genotype
ITGA4	rs2124440	AG
RPN1	rs2712381	AC
ACKR2	rs2228467	TT
PTGR1	rs2273788	CC
IRF8	rs424971	TT

GWAS analysis

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/23314186

Biomarkers and Others

Uric acid levels

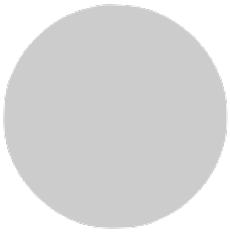
Purines are essential organic compounds that our body uses to build DNA and are obtained from certain foods and drinks. When we have too many of them, or the body cannot handle them properly, they can turn into uric acid, a waste product produced by the breakdown of purines and usually excreted by the kidneys and urine. It sometimes builds up, which can form needle-like crystals in the joints, called gout, a painful form of arthritis. On the flip side, having low uric acid levels in the blood is rare and usually does not cause health problems. At the genetic level, variants in the GCKR gene, among others, correlate with a predisposition to abnormal uric acid levels.

Your genetic map

Gene	SNP	Genotype
PDZK1	rs12129861	AG
GCKR	rs780094	TC
SLC2A9	rs734553	TT
ABCG2	rs2231142	GG
CARMIL1	rs742132	AG
SLC17A1	rs1183201	AT
SLC16A9	rs12356193	AA
SLC22A11	rs17300741	AA
SLC22A1	rs505802	TT

GWAS analysis

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/19503597

Biomarkers and Others

Menopause (age at onset)

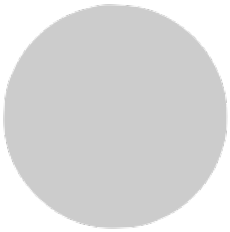
Menopause is the natural stage in a woman's life when menstruation ceases and fertility ends. This stage of life is associated with one of the significant hormonal changes in women, characterized by a decrease in the secretion of estrogen, progesterone, and testosterone to a lesser extent. It influences a woman's well-being and can be associated with cardiovascular disease, breast cancer, osteoarthritis, and osteoporosis. Some women experience menopause at an earlier age than expected, known as premature menopause, leading to earlier infertility and the possibility of the diseases above. Genetics is an influencing factor, as variants in the EXO1 gene, among others, have been found to correlate with the likelihood of early onset of menopause.

Your genetic map

Gene	SNP	Genotype
EXO1	rs1635501	TC
FNDC4	rs2303369	TC
TLK1	rs10183486	TC
UIMC1	rs365132	TG
SYCP2L	rs2153157	AG
ASH2L	rs2517388	TT
LOC1027	rs12294104	CC
PRIM1	rs2277339	TT
TDRD3	rs4886238	GG
POLG	rs2307449	TG
GSPT1	rs10852344	TT
TMEM150	rs11668344	AA
NLRP11	rs12461110	GG
MCM8	rs16991615	GG

Monovariant analysis

What do your genetics tell us?



According to this study, you have a propensity similar to that of most of the population.

More information:

www.ncbi.nlm.nih.gov/pubmed/22267201

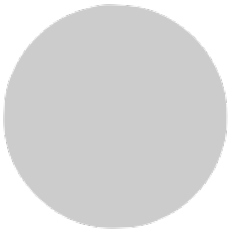
Biomarkers and Others

Bone mineral density

Bone Mineral Density (BMD) is the most widely used predictor of fracture risk.

GWAS analysis

What do your genetics tell us?



According to this study, your propensity is to have normal levels, in line with the average person.

More information:

www.ncbi.nlm.nih.gov/pubmed/22504420

Your genetic map

Gene	SNP	Genotype
FABP3P2	rs9533090	CC
Intergeni	rs7932354	TC
AXIN1	rs9921222	TC
TMEM26	rs1053051	TC
RPS3AP2	rs13336428	AG
HROB	rs227584	AC
FAM210A	rs4796995	AG
CCDC170	rs4869742	TC
CPED1	rs13245690	AA
CBR1 AS1	rs4817775	CC
CPN1	rs7084921	CC
LOC1053	rs430727	TC
LOC1079	rs1564981	AG
DCDC1	rs163879	TC
RHEBL1	rs12821008	CC
DNM3	rs479336	GG
LOC1079	rs2887571	AA
FOXL1	rs10048146	AA
FUBP3	rs7851693	CC
CSRNP3	rs1346004	GG
GPATCH1	rs10416218	TC
Intergeni	rs736825	CG
IDUA	rs3755955	AG
LOC1053	rs1878526	GG
JAG1	rs3790160	CC
KCNMA1	rs7071206	TT
USF3	rs1026364	TG
LOC1053	rs7953528	TT
LEKR1	rs344081	TT
RPL37AP	rs10835187	TC
LRP5	rs3736228	CC

Biomarkers and Others

Lung volume

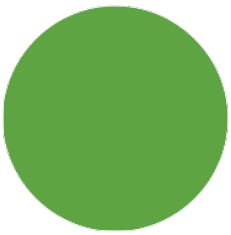
Lung volume is an essential factor influencing our respiratory function. It is measured by forced vital capacity (FVC), which indicates the maximum volume of air exhaled at maximum possible effort, starting from a maximal inspiration. It is expressed as volume (in ml). Low levels of this indicator may indicate lung obstruction. The analysis tool used is spirometry, which is used to diagnose and monitor respiratory diseases such as asthma and COPD (chronic obstructive pulmonary disease), among others. Environmental factors such as smoking and pollution exposure can influence the results, but genetics also plays a significant role. It has been found that specific variants in genes, such as BMP6, can affect a person's forced vital capacity.

Your genetic map

Gene	SNP	Genotype
EFEMP1	rs1430193	TT
BMP6	rs6923462	CC
MIR129 2	rs4237643	TT
PRDM11	rs2863171	AA
WWOX	rs1079572	AG

GWAS analysis

What do your genetics tell us?



According to this study, you are more prone than the average person to having normal levels.

More information:

www.ncbi.nlm.nih.gov/pubmed/24929828

Biomarkers and Others

Longevity

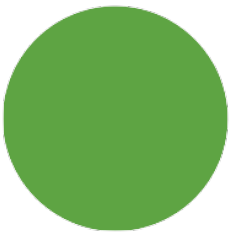
Longevity is described as a person's lifespan and is a multifactorial phenomenon involving environmental factors, mainly diet, sport, stress, lifestyle, and genetics. Research on the genetic component in human longevity has focused on stress response signaling pathways, DNA repair, and nutrient storage and utilization. These processes are mediated by a wide variety of genes, some of which have been identified as possible determinants of longevity. Therefore, although longevity is a complex and multifactorial phenomenon, evidence indicates that genetics plays a vital role in its determination, and a particular variant of the TAS2R16 gene is related to the natural propensity for longevity in women.

Your genetic map

Gene	SNP	Genotype
TAS2R16	rs978739	TT

Monovariant analysis

What do your genetics tell us?



According to your genotype, you have a propensity to be a long-lived person. Other genetic and clinical factors may play a role

More information:

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3487725/pdf/pone.0045232.pdf

Pharmacogenetics

Warfarin

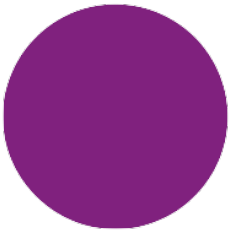
Warfarin is an anticoagulant drug normally used to prevent blood clot formation, as well as migration. Although originally marketed as a pesticide (d-Con, Rodex, among others), Warfarin has since become the most frequently prescribed oral anticoagulant in North America. Warfarin has several properties that should be noted when used medicinally, including its ability to cross the placental barrier during pregnancy, which can result in fetal bleeding, spontaneous abortion, preterm birth, stillbirth, and neonatal death. Additional adverse effects, such as necrosis, purple toe syndrome, osteoporosis, valve and artery calcification, and drug interactions, have also been documented with warfarin use. Warfarin does not actually affect blood viscosity. Rather, it inhibits Vitamin-k dependent synthesis of biologically active forms of various clotting factors, in addition to several regulatory factors.

Your genetic map

Gene	SNP	Genotype
VKORC1	rs9923231	CC

Monovariant analysis

What do your genetics tell us?



Patients with the CC genotype may require an increased dose of warfarin as compared to patients with the TC or TT genotype. Other genetic and clinical factors may also influence a patient's warfarin dose requirement.

More information:

https://www.ncbi.nlm.nih.gov/gtr/conditions/CN078029

Pharmacogenetics

Meperidine

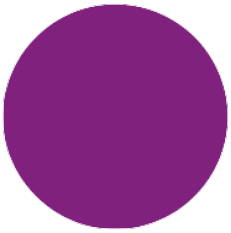
A narcotic analgesic that can be used for the relief of most types of moderate to severe pain, including postoperative pain and the pain of labour. Prolonged use may lead to dependence on the morphine type; withdrawal symptoms appear more rapidly than with morphine and are of shorter duration.

Your genetic map

Gene	SNP	Genotype
CREB1	rs2952768	TC

Monovariant analysis

What do your genetics tell us?



Patients with the TC genotype may have decreased opioid analgesic requirements after surgery. Other genetic and clinical factors may also have an effect.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/23183491

Pharmacogenetics

Pentazocine

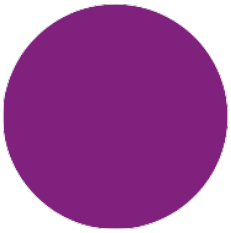
The first mixed agonist-antagonist analgesic to be marketed. It is an agonist at the kappa and sigma opioid receptors, and has a weak antagonist action at the mu receptor

Your genetic map

Gene	SNP	Genotype
CREB1	rs2952768	TC

Monovariant analysis

What do your genetics tell us?



Patients with the TC genotype may have decreased opioid analgesic requirements after surgery as compared to patients with the CC genotype. Other genetic and clinical factors may influence a patient's opioid dose requirement.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/23183491

Pharmacogenetics

Morphine

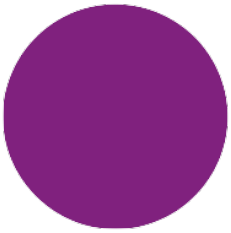
The principal alkaloid in opium and the prototype opiate analgesic and narcotic. Morphine has widespread effects in the central nervous system and on smooth muscle. In January, 2017, morphine was approved for the treatment of chronic pain.

Your genetic map

Gene	SNP	Genotype
CREB1	rs2952768	TC

Monovariant analysis

What do your genetics tell us?



Patients with the TC genotype may have decreased opioid analgesic requirements after surgery as compared to patients with the CC genotype. Other genetic and clinical factors may affect a patient's opioid dose requirement.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/23183491

Pharmacogenetics

Aspirin

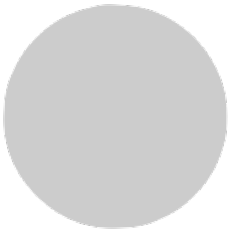
Aspirin, also known as acetylsalicylic acid (ASA), is a medication used to treat pain, fever, and inflammation. Specific inflammatory conditions for which aspirin is used include Kawasaki disease, pericarditis, and rheumatic fever. Aspirin is a non-steroidal anti-inflammatory drug (NSAID) and works similar to other NSAIDs, but also suppresses the normal functioning of platelets.

Your genetic map

Gene	SNP	Genotype
PTGS1	rs10306114	AA

Monovariant analysis

What do your genetics tell us?



Patients with the AA genotype who are treated with aspirin may be at a decreased, though not absent, risk for non-response to aspirin as compared to patients with the AG or GG genotype. Other genetic and clinical factors may also influence a patient's response to aspirin.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/16493486

Pharmacogenetics

Simvastatin

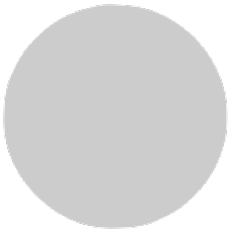
Simvastatin is a lipid-lowering agent that is derived synthetically from the fermentation of Aspergillus terreus. It is a potent, competitive inhibitor of 3-hydroxy-3-methylglutaryl coenzyme A reductase (hydroxymethylglutaryl COA reductases), which is the rate-limiting enzyme in cholesterol biosynthesis. It may also interfere with steroid hormone production. Due to the induction of hepatic LDL receptors, it increases the breakdown of LDL cholesterol.

Your genetic map

Gene	SNP	Genotype
SLCO1B1	rs4149056	TT

Monovariant analysis

What do your genetics tell us?



Patients with the TT genotype may be at a lower risk of simvastatin-related myopathy as compared to patients with the CT or CC genotype. Other genetic and clinical factors may also affect a patient's risk for toxicity.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/28482130

Pharmacogenetics

Bupropion

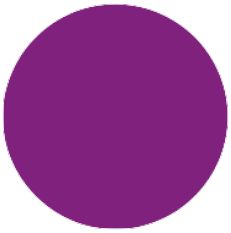
A unicyclic, aminoketone antidepressant. The mechanism of its therapeutic actions is not well understood, but it does appear to block dopamine uptake. Hydrochloride is available as an aid to smoking cessation treatments.

Your genetic map

Gene	SNP	Genotype
ANKK1	rs1800497	AA

Monovariant analysis

What do your genetics tell us?



Patients with the AA genotype who are treated with bupropion may be less likely to quit smoking as compared to patients with the GG genotype, although this has been contradicted in one study. Other genetic and clinical factors may also influence a patient's capacity to quit smoking.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/15492764

Pharmacogenetics

Pravastatin

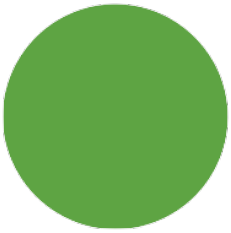
Pravastatin is a cholesterol-lowering agent that belongs to a class of medications known as statins. It was derived from microbial transformation of mevastatin, the first statin discovered. It is a ring-opened dihydroxyacid with a 6'-hydroxyl group that does not require in vivo activation. Pravastatin is one of the lower potency statins. However, its increased hydrophilicity is thought to confer advantages, such as minimal penetration through lipophilic membranes of peripheral cells, increased selectivity for hepatic tissues, and a reduction in side effects compared with lovastatin and simvastatin.

Your genetic map

Gene	SNP	Genotype
HMGCR	rs17244841	AA

Multivariate analysis

What do your genetics tell us?



Patients with the AA genotype who are treated with statins may be more likely to respond as compared to patients with the AT or TT genotype. Other genetic and clinical factors may also influence a patient's response when treated with statins.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/15199031

Pharmacogenetics

Methotrexate

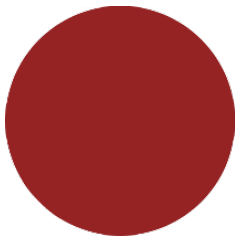
An antineoplastic antimetabolite with immunosuppressive properties. It is an inhibitor of tetrahydrofolate dehydrogenase and prevents the formation of tetrahydrofolate, necessary for synthesis of thymidylate, an essential component of DNA.

Your genetic map

Gene	SNP	Genotype
MTHFR	rs1801133	AG

Monovariant analysis

What do your genetics tell us?



Patients with AG genotype and leucemia or lymphoma who are treated with methotrexate: 1) may have a poorer response 2) may be at an increased risk of toxicity 3) may require a lower dose of methotrexate, and 4) may be at a greater risk of folate deficiency as compared to patients with GG genotype. When comparing with AA genotype, the opposite is true. This association has been contradicted in other studies. Other factors may also have an effect.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/22143415

Pharmacogenetics

Fluorouracil, capecitabine, pyrimidine analogues, tegafur and Neoplasms

Fluorouracil (5-FU), sold under the brand name Adrucil, among others, is a medication used to treat cancer. By injection into a vein, it is used for colon cancer, esophageal cancer, stomach cancer, pancreatic cancer, breast cancer, and cervical cancer. As a cream it is used for actinic keratosis and basal cell carcinoma. It is a potent antimetabolite used in the treatment of cancer. It is a drug that blocks the methylation reaction of deoxyuridic acid, converting it into thymidylic acid by inhibiting an enzyme that is important for the synthesis of thymidine, which, being part of the DNA molecule, stops its formation. The drug is specific to the S phase of the cell phase cycle. 5-Fluorouracil is involved in the synthesis of DNA and inhibits, to a small degree, the formation of RNA. The two actions combine to promote a metabolic imbalance that results in cell death. The inhibitory activity of the drug, by its analogy with uracil, has an effect on the rapid growth of the neoplastic cells, which, preferentially, take advantage of the uracil molecule for nucleic acid biosynthesis.

Your genetic map

Gene	SNP	Genotype
DPYD	rs67376798	TT

Monovariant analysis

What do your genetics tell us?

TT-genotype patients treated with fluoropyrimidine-based chemotherapy may exhibit 1) increased clearance of the drug and 2) decreased, but not absent, risk and reduced severity of drug toxicity as compared to patients with the AT genotype. The combination (FOLFOX, FOLFIRI or FEC) and delivery of the drug may influence risk for toxicity. Other genetic and clinical factors may also have an influence.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/17700593

Pharmacogenetics

Vincristine

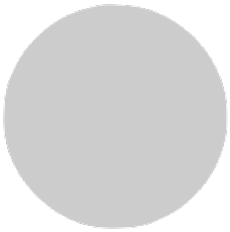
Vincristine is an anti-tumour vinca alkaloid isolated from Vinca Rosea. It is marketed under several brand names, many of which have different formulations, such as Marqibo (liposomal injection) and Vincasar. Vincristine is indicated for the treatment of acute leucemia, malignant lymphoma, Hodgkin's disease, acute erythraemia, and acute panmyelosis. Vincristine sulfate is often chosen as part of polychemotherapy because of its lack of significant bone–marrow suppression (at recommended doses) and unique clinical toxicity (neuropathy).

Your genetic map

Gene	SNP	Genotype
CEP72	rs924607	TC

Monovariant analysis

What do your genetics tell us?



Patients with the TC genotype may have decreased, but not absent, risk of peripheral nervous system diseases when treated with vincristine as compared to patients with the TT genotype. Other genetic and clinical factors may also affect a patient's response to vincristine.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/25710658

Pharmacogenetics

Tacrolimus

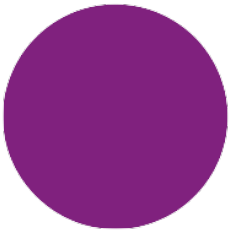
Tacrolimus (also FK-506 or Fujimycin) is an immunosuppressive drug mainly used after an organ transplant, to reduce the activity of the patient's immune system and, thereby, the risk of organ rejection. It is also used in a topical preparation for the treatment of severe atopic dermatitis, severe refractory uveitis, after bone marrow transplants; and the skin condition vitiligo. It was discovered in 1984 from the fermentation broth of a Japanese soil sample containing the bacteria Streptomyces tsukubaensis. Tacrolimus is chemically known as a macrolide. It reduces peptidyl-prolyl isomerase activity by binding to the immunophilin FKBP-12 (FK506 binding protein), creating a new complex. This FKBP12-FK506 complex interacts with and inhibits calcineurin, thus inhibiting both T-lymphocyte signal transduction and IL-2 transcription.

Your genetic map

Gene	SNP	Genotype
CYP3A4	rs2740574	TT

Monovariant analysis

What do your genetics tell us?



Transplant recipients with the TT (CYP3A4) genotype may require a decreased dose of tacrolimus as compared to patients with the TC or CC genotype. Other genetic and clinical factors, such as CYP3A5 (rs776746), may also influence a patient's dose requirements.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/23778326

Pharmacogenetics

Peginterferon Alpha-2b

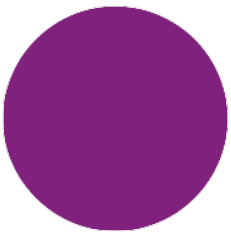
Peginterferon alfa-2b is a form of recombinant interferon used as part of combination therapy to treat chronic Hepatitis C, an infectious liver disease caused by infection with the Hepatitis C Virus (HCV). HCV is a single-stranded RNA virus that is categorised into nine distinct genotypes, with genotype 1 being the most common in the United States, and affecting 72% of all chronic HCV patients. Treatment options for chronic Hepatitis C have advanced significantly since 2011, with the development of Direct Acting Antivirals (DAAs) resulting in less use of Peginterferon alfa-2b. Peginterferon alfa-2b is derived from the alfa-2b moiety of recombinant human interferon, and acts by binding to human type-1 interferon receptors. The activation and dimerization of this receptor induces the body's innate antiviral response by activating the Janus kinase/signal transducer and activator of transcription (JAK/STAT) pathway.

Your genetic map

Gene	SNP	Genotype
IFNL4	rs12979860	TC

Monovariant analysis

What do your genetics tell us?



Patients with the TC genotype and Hepatitis C genotype 1 may exhibit a decreased response (sustained virological response, SVR) when administered peg interferon alpha (2a, 2b) and ribavirin as compared to patients with the CC genotype. Patients with the TC genotype may also have lower spontaneous clearance in acute HCV infections than patients with the CC genotype. Other genetic and clinical factors may also affect a patient's response to peg interferon and ribavirin.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/21145807

Pharmacogenetics

Ribavirin

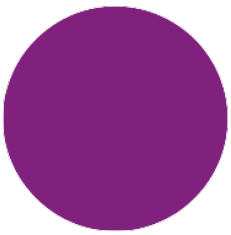
Producing broad-spectrum activity against several RNA and DNA viruses, Ribavirin is a synthetic guanosine nucleoside and antiviral agent that interferes with the synthesis of viral mRNA. It is primarily indicated for use in treating hepatitis C and viral hemorrhagic fevers. HCV is a single-stranded RNA virus that is categorised into nine distinct genotypes, with genotype 1 being the most common in the United States, and affecting 72% of all chronic HCV patients. It is reported that ribavirin might be effective only in the early stages of viral hemorrhagic fevers, including Lasser fever, Crimean-Congo hemorrhagic fever, Venezuelan hemorrhagic fever, and Hantavirus infection. Ribavirin is a prodrug that is metabolised into nucleoside analogs, blocking viral RNA synthesis and viral mRNA capping. Before the development of newer drugs, ribavirin and dual therapy was considered the first-generation and standard antiviral treatment. Newer drugs developed as hepatitis C viral infection treatments can be used to reduce or eliminate the use of ribavirin, which is associated with serious adverse effects.

Your genetic map

Gene	SNP	Genotype
IFNL4	rs12979860	TC

Monovariant analysis

What do your genetics tell us?



Patients with the TC genotype and Hepatitis C genotype 1 may exhibit a decreased response (sustained virological response, SVR) when administered peg interferon alpha (2a, 2b) and ribavirin. They may also exhibit lower spontaneous clearance in acute HCV infections than patients with the CC genotype. Other genetic and clinical factors may also affect a patient's response to peg interferon and ribavirin.

More information:

https://www.ncbi.nlm.nih.gov/pubmed/21145807

Hereditary Diseases (genetics)

Isovaleric acidemia

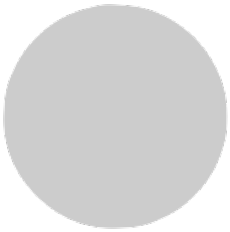
A rare, autosomal recessive, organic aciduria that is characterized by variable clinical presentation ranging from acute neonatal onset of metabolic decompensation to later onset of chronic, non-specific manifestations including failure to thrive and/or developmental delay. All patients are prone to intermittent, acute metabolic decompensation. During metabolic episodes, urine analysis demonstrates elevated isovaleric acid derivatives.

Your genetic map

Gene	SNP	Genotype
IVD	rs121434285	GG
IVD	rs34695403	CC
IVD	rs28940889	CC
IVD	rs398123683	TT
IVD	rs142761835	GG
IVD	rs748026507	TT
IVD	rs796051983	CC
IVD	rs765815516	CC

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=33

Hereditary Diseases (genetics)

Combined malonic and methylmalonic acidemia

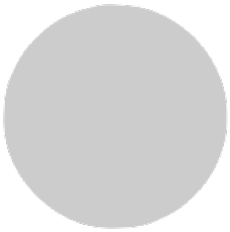
Combined malonic and methylmalonic acidemia is a rare inborn error of metabolism characterized by elevation of malonic acid (MA) and methylmalonic acid (MMA) in body fluids, with higher levels of MMA than MA. CMAMMA presents in childhood with metabolic acidosis, developmental delay, dystonia and failure to thrive or in adulthood with seizures, memory loss and cognitive decline.

Your genetic map

Gene	SNP	Genotype
ACSF3	rs387907119	GG
ACSF3	rs370382601	AA
ACSF3	rs757905943	GG
ACSF3	rs752338222	GG
ACSF3	rs145583876	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=289504

Hereditary Diseases (genetics)

Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency

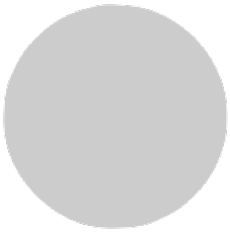
Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency is a rare inborn error of metabolism disease characterized by mild to moderate, persistent elevation of methylmalonic acid in plasma, urine and cerebrospinal fluid. Clinical presentation may include acute metabolic decompensation with metabolic acidosis (presenting with vomiting, dehydration, confusion, hallucinations), nonspecific neurological symptoms, or may also be asymptomatic.

Your genetic map

Gene	SNP	Genotype
MCEE	rs111033538	GG

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=308425

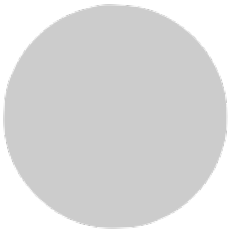
Hereditary Diseases (genetics)

Vitamin B12-unresponsive methylmalonic acidemia

Vitamin B12-unresponsive methylmalonic acidemia is an inborn error of vitamin B12 (cobalamin) metabolism characterized by recurrent ketoacidotic crises or transient vomiting, dehydration, hypotonia and intellectual deficit, which does not respond to administration of vitamin B12. There are two types of vitamin B12-unresponsive methylmalonic acidemia: mut0 and mut- (see these terms).

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=27

Your genetic map

Gene	SNP	Genotype
MMUT	rs121918249	AA
MMUT	rs121918251	CC
MMUT	rs121918252	CC
MMUT	rs121918253	CC
MMUT	rs121918254	CC
MMUT	rs121918256	TT
MMUT	rs121918257	GG
MMUT	rs398123276	TT
MMUT	rs398123278	GG
MMUT	rs727504020	GG
MMUT	rs727504022	CC
MMUT	rs753564352	CC
MMUT	rs200019422	CC
MMUT	rs564069299	CC
MMUT	rs777031588	TT
MMUT	rs796052002	GG
MMUT	rs796052007	AA
MMUT	rs796052006	AA
MMUT	rs760782399	GG
MMUT	rs796052005	TT
MMUT	rs779990936	GG
MMUT	rs777758903	GG
MMUT	rs753288303	CC
MMUT	rs772552898	GG
MMUT	rs778702777	CC
MMUT	rs879253852	GG
MMUT	rs774159791	GG

Hereditary Diseases (genetics)

Vitamin B12-responsive methylmalonic acidemia

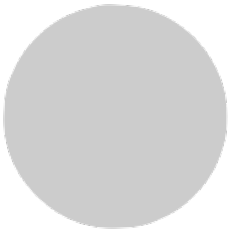
An inborn error of vitamin B12 (cobalamin) metabolism characterized by recurrent ketoacidotic comas or transient vomiting, dehydration, hypotonia and intellectual deficit, which responds to vitamin B12. There are three types: cblA, cblB and cblD-variant 2 (cblDv2).

Your genetic map

Gene	SNP	Genotype
MMAA	rs104893846	CC
MMAA	rs104893851	CC
MMAA	rs796051992	CC
MMAA	rs571038432	CC
MMAA	rs757548934	CC

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=28

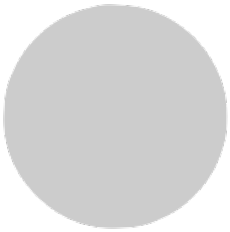
Hereditary Diseases (genetics)

Propionic acidemia

Propionic acidemia (PA) is an organic aciduria caused by the deficient activity of the propionyl Coenzyme A carboxylase and is characterized by life threatening episodes of metabolic decompensation, neurological dysfunction and that may be complicated by cardiomyopathy.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=35

Your genetic map

Gene	SNP	Genotype
PCCA	rs121964958	TT
PCCA	rs138149179	CC
PCCA	rs776496862	GG
PCCA	rs796052019	GG
PCCA	rs796052018	GG
PCCA	rs776281864	AA
PCCB	rs121964959	CC
PCCB	rs121964960	GG
PCCB	rs111033542	CC
PCCB	rs121964961	AA
PCCB	rs186710233	CC
PCCB	rs202247822	TT
PCCB	rs202247823	AA
PCCB	rs398123464	GG
PCCB	rs374722096	CC
PCCB	rs572246667	CC
PCCB	rs879253815	CC
PCCB	rs186031457	CC

Hereditary Diseases (genetics)

Congenital lactic acidosis, Saguenay-Lac-Saint-Jean type

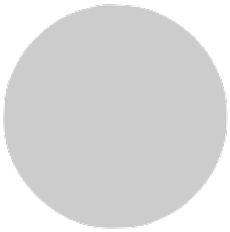
Saguenay-Lac-St. Jean (SLSJ) type congenital lactic acidosis, a French Canadian form of Leigh syndrome (see this term), is a mitochondrial disease characterized by chronic metabolic acidosis, hypotonia, facial dysmorphism and delayed development.

Your genetic map

Gene	SNP	Genotype
LRPPRC	rs119466000	GG
LRPPRC	rs863224052	GG

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=70472

Hereditary Diseases (genetics)

Distal renal tubular acidosis

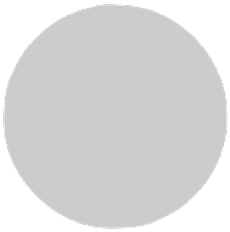
Distal renal tubular acidosis (dRTA) is a disorder of impaired net acid secretion by the distal tubule characterized by hyperchloremic metabolic acidosis. The classic form is often associated with hypokalemia whereas other forms of acquired dRTA may be associated with hypokalemia, hyperkalemia or normokalemia.

Your genetic map

Gene	SNP	Genotype
SLC4A1	rs121912744	GG
SLC4A1	rs121912751	GG

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=18

Hereditary Diseases (genetics)

3-methylglutaconic aciduria type 1

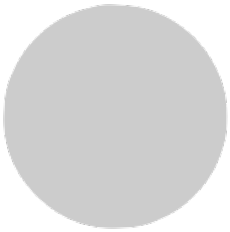
3-methylglutaconic aciduria (3-MGA) type I is an inborn error of leucine metabolism with a variable clinical phenotype ranging from mildly delayed speech to psychomotor retardation, coma, failure to thrive, metabolic acidosis and dystonia.

Your genetic map

Gene	SNP	Genotype
AUH	rs387906755	CC

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=67046

Hereditary Diseases (genetics)

3-methylglutaconic aciduria type 7

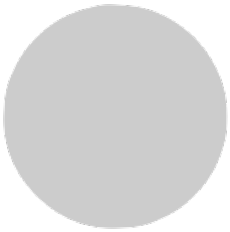
A rare organic aciduria characterized by increased urinary excretion of 3-methylglutaconic acid, variably associated with neutropenia (sometimes causing recurrent severe infections and potentially resulting in leukemia) and progressive neurologic manifestations, such as global developmental delay, intellectual disability, hypotonia, movement disorder, and seizures. Microcephaly, cataract, facial dysmorphism, growth retardation, endocrine abnormalities, and cardiomyopathy have also been reported. Brain imaging may show cerebral or cerebellar atrophy, or abnormalities of the basal ganglia.

Your genetic map

Gene	SNP	Genotype
CLPB	rs144078282	TT
CLPB	rs200203460	GG
CLPB	rs374473067	CC
CLPB	rs185461628	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=445038

Hereditary Diseases (genetics)

3-methylglutaconic aciduria type 9

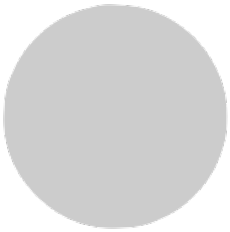
A rare organic aciduria characterized by early onset of global developmental delay with severe intellectual disability, seizures, and 3-methylglutaconic aciduria. Additional features are hypotonia, hyperactivity and aggressive behavior, optic atrophy, or spasticity. Brain imaging may show generalized cerebral atrophy and white matter abnormalities.

Your genetic map

Gene	SNP	Genotype
TIMM50	rs797044891	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=505216

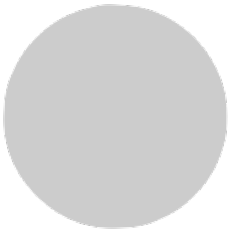
Hereditary Diseases (genetics)

Argininosuccinic aciduria

A rare, genetic disorder of urea cycle metabolism typically characterized by either a severe, neonatal-onset form that manifests with hyperammonemia accompanied with vomiting, hypothermia, lethargy and poor feeding in the first few days of life, or late-onset forms that manifest with stress- or infection-induced episodic hyperammonemia or, in some, behavioral abnormalities and/or learning disabilities, or chronic liver disease. Patients often manifest liver dysfunction.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=23

Your genetic map

Gene	SNP	Genotype
ASL	rs28941472	AA
ASL	rs28940286	CC
ASL	rs28941473	GG
ASL	rs28940287	CC
ASL	rs367543005	CC
ASL	rs374304304	CC
ASL	rs145138923	GG
ASL	rs142637046	GG
ASL	rs398123126	CC
ASL	rs201523601	GG
ASL	rs199938613	CC
ASL	rs751590073	GG
ASL	rs369879957	CC
ASL	rs770167670	CC

Hereditary Diseases (genetics)

D-2-hydroxyglutaric aciduria

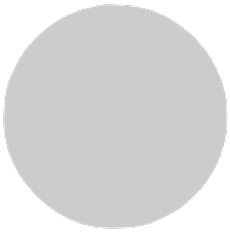
D-2-hydroxyglutaric aciduria (D-2-HGA) is a rare clinically variable neurological form of 2-hydroxyglutaric aciduria characterized biochemically by elevated D-2-hydroxyglutaric acid (D-2-HG) in the urine, plasma and cerebrospinal fluid.

Your genetic map

Gene	SNP	Genotype
D2HGDH	rs753528947	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79315

Hereditary Diseases (genetics)

Formiminoglutamic aciduria

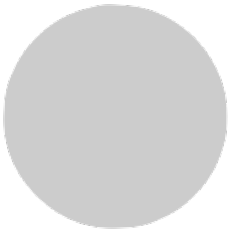
A rare disorder of folate metabolism and transport characterized, biochemically, by elevated formiminoglutamate in urine and plasma due to glutamate formiminotransferase deficiency, associated with a highly variable clinical phenotype, ranging from developmental delay, intellectual disability and anemia to normal development without anemia. Increased hydantoin-5-propionic acid and/or folate in plasma may also be associated.

Your genetic map

Gene	SNP	Genotype
FTCD AS1	rs140217223	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=51208

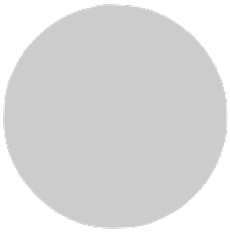
Hereditary Diseases (genetics)

Fumaric aciduria

Fumaric aciduria (FA), an autosomal recessive metabolic disorder, is most often characterized by early onset but non-specific clinical signs: hypotonia, severe psychomotor impairment, convulsions, respiratory distress, feeding difficulties and frequent cerebral malformations, along with a distinctive facies. Some patients present with only moderate intellectual impairment.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=24

Your genetic map

Gene	SNP	Genotype
FH	rs398123159	AA
FH	rs398123166	GG
FH	rs587781682	GG
FH	rs587782618	CC
FH	rs372505976	TT
FH	rs863223978	CC
FH	rs863224008	TT
FH	rs863224004	CC
FH	rs863223973	AA
FH	rs863224002	GG
FH	rs863224000	AA
FH	rs863223967	TT
FH	rs863223965	AA
FH	rs863224015	TT
FH	rs863223983	TT
FH	rs863223982	CC

Hereditary Diseases (genetics)

Mevalonic aciduria

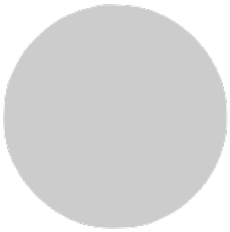
A rare, severe form of mevalonate kinase deficiency (MKD) characterized by dysmorphic features, failure to thrive, psychomotor delay, ocular involvement, hypotonia, progressive ataxia, myopathy, and recurrent inflammatory episodes.

Your genetic map

Gene	SNP	Genotype
MVK	rs104895319	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=29

Hereditary Diseases (genetics)

Achondroplasia

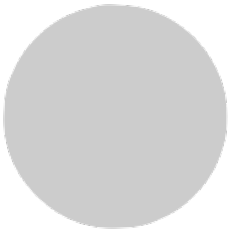
A primary bone dysplasia with micromelia characterized by rhizomelia, exaggerated lumbar lordosis, brachydactyly, and macrocephaly with frontal bossing and midface hypoplasia.

Your genetic map

Gene	SNP	Genotype
FGFR3	rs28931614	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=15

Hereditary Diseases (genetics)

Achromatopsia

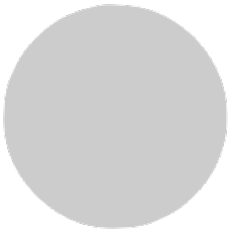
A rare autosomal recessive retinal disorder characterized by color blindness, nystagmus, photophobia, and severely reduced visual acuity due to the absence or impairment of cone function.

Your genetic map

Gene	SNP	Genotype
CNGA3	rs104893613	CC
CNGA3	rs104893614	GG
CNGA3	rs104893617	CC
CNGA3	rs137852608	CC
CNGA3	rs104893619	GG
CNGA3	rs104893620	CC
CNGA3	rs753625117	TT
CNGA3	rs141386891	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=49382

Hereditary Diseases (genetics)

Gastric adenocarcinoma and proximal polyposis of the stomach

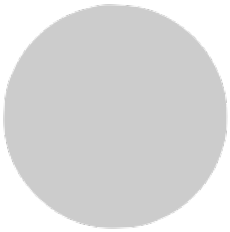
Gastric adenocarcinoma and proximal polyposis of the stomach (GAPPS) is a rare hereditary gastric cancer characterized by proximal gastric polyposis and increased risk of early-onset, intestinal-type adenocarcinoma of the gastric body, with no duodenal or colorectal polyposis.

Your genetic map

Gene	SNP	Genotype
APC	rs879253784	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=314022

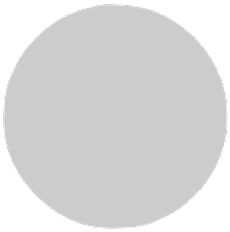
Hereditary Diseases (genetics)

X-linked adrenoleukodystrophy

A rare progressive peroxisomal disorder characterized by endocrine dysfunction (adrenal failure and sometimes testicular insufficiency), progressive myelopathy, peripheral neuropathy and, variably, progressive leukodystrophy.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=43

Your genetic map

Gene	SNP	Genotype
ABCD1	rs128624215	CC
ABCD1	rs128624219	GG
ABCD1	rs128624220	CC
ABCD1	rs128624221	CC
ABCD1	rs128624224	CC
ABCD1	rs4010613	CC
ABCD1	rs193922094	TT
ABCD1	rs398123100	CC
ABCD1	rs398123102	GG
ABCD1	rs398123105	CC
ABCD1	rs398123106	CC
ABCD1	rs398123108	GG
ABCD1	rs727503786	CC
ABCD1	rs797044726	CC
BCAP31	rs128624216	AA
BCAP31	rs128624218	GG
BCAP31	rs193922097	GG
BCAP31	rs193922098	CC
BCAP31	rs398123110	GG
BCAP31	rs398123113	CC
BCAP31	rs797044610	AA

Hereditary Diseases (genetics)

Neurological conditions associated with aminoacylase 1 deficiency

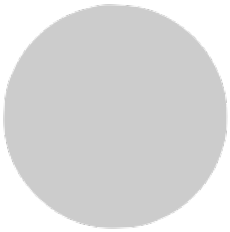
An inborn error of metabolism marked by a characteristic pattern of urinary N-acetyl amino acid excretion and neurologic symptoms.

Your genetic map

Gene	SNP	Genotype
ABHD14A	rs121912699	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=137754

Hereditary Diseases (genetics)

X-linked agammaglobulinemia

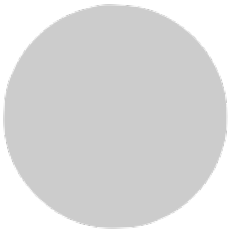
A clinically variable form of isolated agammaglobulinemia, an inherited immunodeficiency disorder, characterized in affected males by recurrent bacterial infections during infancy.

Your genetic map

Gene	SNP	Genotype
BTK	rs128620183	CC
BTK	rs128620187	GG
BTK	rs128620185	CC
BTK	rs128621201	GG
BTK	rs128621204	GG
BTK	rs128621210	AA
BTK	rs104894770	CC
BTK	rs193922124	GG
BTK	rs193922125	TT
BTK	rs193922131	CC
BTK	rs193922132	TT
BTK	rs193922133	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=47

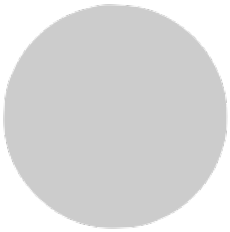
Hereditary Diseases (genetics)

Oculocutaneous albinism type 1

A form of oculocutaneous albinism (OCA) characterized by a spectrum of hypopigmentation of skin hair and eyes, ranging from little or no pigmentation to localized pigementation. Nystagmus, photophobia and reduced visual acuity are frequently present. The subtypes include OCA1A, OCA1B, type 1 minimal pigment oculocutaneous albinism (OCA1-MP) and type 1 temperature sensitive oculocutaneous albinism (OCA1-TS).

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=352731

Your genetic map

Gene	SNP	Genotype
LOC1079	rs28940876	CC
LOC1079	rs61754388	CC
LOC1079	rs121908011	GG
LOC1079	rs61753185	GG
LOC1079	rs28940880	GG
LOC1079	rs61754392	GG
LOC1079	rs61753178	CC
LOC1079	rs61753180	GG
LOC1079	rs61754387	AA
LOC1079	rs104894316	GG
LOC1079	rs104894317	GG
LOC1079	rs104894318	GG
LOC1079	rs61754381	TT
LOC1079	rs61754386	AA
LOC1079	rs62645917	CC
LOC1079	rs61754362	CC
LOC1079	rs61754365	GG
LOC1079	rs61754371	CC
LOC1079	rs62645904	CC
LOC1079	rs61754380	GG
LOC1079	rs797046082	AA
LOC1079	rs797046083	CC
LOC1079	rs758115945	GG

Hereditary Diseases (genetics)

Oculocutaneous albinism type 2

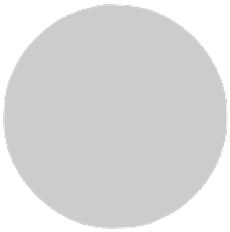
A form of oculocutaneous albinism characterized by variable hypopigmentation of the skin and hair, numerous characteristic ocular changes and misrouting of the optic nerves at the chiasm.

Your genetic map

Gene	SNP	Genotype
OCA2	rs121918167	GG
OCA2	rs121918170	TT
OCA2	rs797045839	CC
OCA2	rs797045838	TT
OCA2	rs368124046	CC
OCA2	rs763819379	TT
OCA2	rs371963034	CC
OCA2	rs142988897	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79432

Hereditary Diseases (genetics)

Oculocutaneous albinism type 3

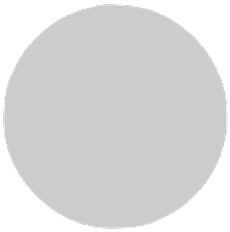
A form of oculocutaneous albinism (OCA) characterized by rufous or brown albinism and occurring mainly in the African population.

Your genetic map

Gene	SNP	Genotype
LURAP1L	rs281865424	GG
LURAP1L	rs776174514	TT
TYRP1	rs104894130	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79433

Hereditary Diseases (genetics)

Oculocutaneous albinism type 4

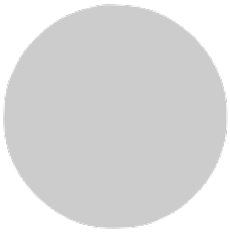
A form of oculocutaneous albinism characterized by varying degrees of skin and hair hypopigmentation, numerous ocular changes and misrouting of the optic nerves at the chiasm.

Your genetic map

Gene	SNP	Genotype
SLC45A2	rs797045970	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79435

Hereditary Diseases (genetics)

Alkaptonuria

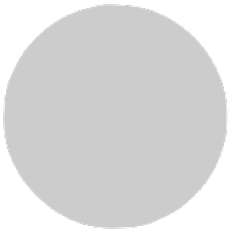
A rare disorder of phenylalanine and tyrosine metabolism characterized by the accumulation of homogentisic acid (HGA) and its oxidized product, benzoquinone acetic acid (BQA), in various tissues (e.g. cartilage, connective tissue) and body fluids (urine, sweat), causing urine to darken when exposed to air as well as grey-blue coloration of the sclera and ear helix (ochronosis), and a disabling joint disease involving both the axial and peripheral joints (ochronotic arthropathy).

Your genetic map

Gene	SNP	Genotype
HGD	rs28942100	GG
HGD	rs120074170	AA
HGD	rs28941783	CC
HGD	rs397515347	CC
HGD	rs120074173	TT
HGD	rs120074174	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=56

Hereditary Diseases (genetics)

Alpha-thalassemia

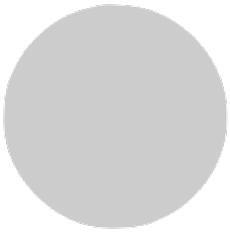
A rare inherited hemoglobinopathy characterized by impaired synthesis of two to all four alpha-globin chains leading to a variable clinical picture depending on the number of affected alleles.

Your genetic map

Gene	SNP	Genotype
HBA2	rs41464951	TT
HBA2	rs41397847	TT
HBA2	rs41417548	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=846

Hereditary Diseases (genetics)

Alpha-mannosidosis

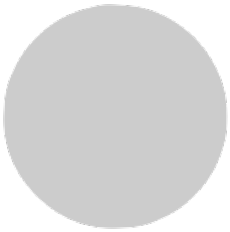
An inherited lysosomal storage disorder characterized by immune deficiency, facial and skeletal abnormalities, hearing impairment, and intellectual deficit.

Your genetic map

Gene	SNP	Genotype
MAN2B1	rs121434331	GG
MAN2B1	rs80338680	GG
MAN2B1	rs80338677	CC
MAN2B1	rs398123455	CC
MAN2B1	rs398123456	CC
MAN2B1	rs398123457	AA
MAN2B1	rs775200333	GG
MAN2B1	rs561991886	CC
MAN2B1	rs768734132	CC
MAN2B1	rs779769525	GG
WDR83	rs370803545	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=61

Hereditary Diseases (genetics)

ALG1-CDG

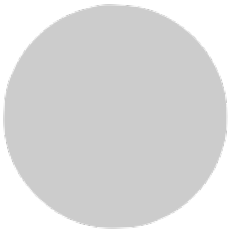
A severe form of congenital disorders of N-linked glycosylation characterized by severe developmental and psychomotor delay, muscular hypotonia, intractable early-onset seizures, and microcephaly. Additional features include altered blood coagulation with a high probability of hemorrhages or thromboses, nephrotic syndrome, ascites, hepatomegaly, cardiomyopathy, ocular manifestations (strabismus, nystagmus), and immunodeficiency. The disease is caused by loss-of-function mutations in the gene ALG1 (16p13.3).

Your genetic map

Gene	SNP	Genotype
ALG1	rs28939378	CC
ALG1	rs121908340	CC
ALG1	rs151173406	CC
ALG1	rs374928784	GG
ALG1	rs369160589	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79327

Hereditary Diseases (genetics)

ALG6-CDG

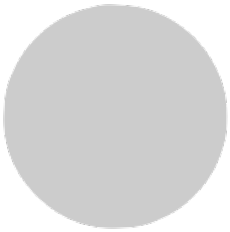
A form of congenital disorders of N-linked glycosylation characterized by feeding problems, mild-to-moderate neurologic involvement with hypotonia, poor head control, developmental delay, ataxia, strabismus, and seizures, ranging from febrile convulsions to epilepsy. Retinal degeneration has also been reported. A minority of patients show other manifestations, particularly intestinal (such as protein-losing enteropathy) and liver involvement. The disease is caused by loss of function mutations of the gene ALG6 (1p31.3).

Your genetic map

Gene	SNP	Genotype
ALG6	rs199682486	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79320

Hereditary Diseases (genetics)

ALG8-CDG

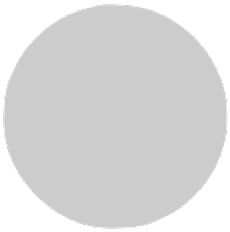
A form of congenital disorders of N-linked glycosylation that is characterized by gastrointestinal symptoms (diarrhea, vomiting, feeding problems with failure to thrive, protein-losing enteropathy), edema and ascites (including hydrops fetalis), hepatomegaly, renal tubulopathy, coagulation anomalies due to thrombocytopenia, brain involvement (psychomotor delay, seizures, ataxia), facial dysmorphism (low-set ears and retrognathia), pes equinovarus, and muscular hypotonia. Cataracts may also be observed. Prognosis is usually poor. The disease is caused by loss-of-function mutations in the gene ALG8 (11q14.1), resulting in a block in the initial step of protein glycosylation.

Your genetic map

Gene	SNP	Genotype
ALG8	rs121908293	TT
ALG8	rs200888240	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79325

Hereditary Diseases (genetics)

ATTRV30M amyloidosis

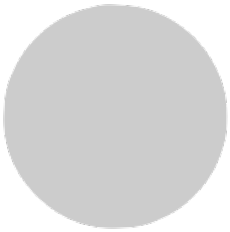
Familial amyloid polyneuropathy (FAP) or transthyretin (TTR) amyloid polyneuropathy is a progressive sensorimotor and autonomic neuropathy of adulthood onset. Weight loss and cardiac involvement are frequent; ocular or renal complications may also occur.

Your genetic map

Gene	SNP	Genotype
TTR	rs28933979	GG
TTR	rs121918069	TT
TTR	rs121918070	AA
TTR	rs76992529	GG
TTR	rs121918076	TT
TTR	rs121918082	GG
TTR	rs121918091	TT
TTR	rs121918093	GG
TTR	rs121918098	AA
TTR	rs267607161	GG
TTR	rs386134269	AA
TTR	rs11541790	CC
TTR	rs730881169	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=85447

Hereditary Diseases (genetics)

Familial primary localized cutaneous amyloidosis

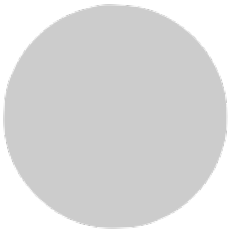
A rare primary cutaneous amyloidosis characterized by familial occurrence of lichen and/or macular amyloidosis due to fibrillary degeneration and apoptosis of basal keratinocytes, followed by conversion of filamentous masses into amyloid material in the papillary dermis. Patients typically present with a pruritic eruption of grouped hyperkeratotic papules, which may coalesce to form hyperkeratotic plaques, with a predilection for the lower limbs (lichen amyloidosis), or with hyperpigmented macules, sometimes with a reticulate pattern, most commonly arising on the back, chest or interscapular areas (macular amyloidosis).

Your genetic map

Gene	SNP	Genotype
OSMR	rs387906822	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=353220

Hereditary Diseases (genetics)

Multiple myeloma

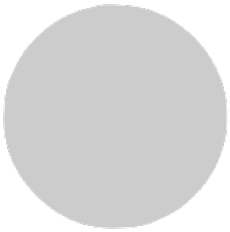
Multiple myeloma (MM) is a malignant tumor of plasma cell characterized by overproduction of abnormal plasma cells in the bone marrow and skeletal destruction. The clinical features are bone pain, renal impairment, immunodeficiency, anemia and presence of abnormal immunoglobulins (Ig).

Your genetic map

Gene	SNP	Genotype
BRAF	rs121913355	CC
FGFR3	rs78311289	AA
KRAS	rs121913527	CC
KRAS	rs121913240	TT
NRAS	rs121913250	CC
TP53	rs28934576	CC
TP53	rs28934874	GG
TP53	rs587781288	CC
TP53	rs730882005	CC
TP53	rs876660333	AA
TP53	rs17849781	GG
TP53	rs764146326	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=314701

Hereditary Diseases (genetics)

Congenital dyserythropoietic anemia type I

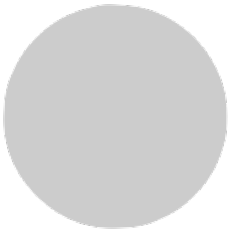
Congenital dyserythropoietic anemiatype I (CDA I) is a hematologic disorder of erythropoiesis characterized by moderate to severe macrocytic anemia occasionally associated with limb or nail deformities and scoliosis.

Your genetic map

Gene	SNP	Genotype
CDAN1	rs80338694	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=98869

Hereditary Diseases (genetics)

Congenital dyserythropoietic anemia type II

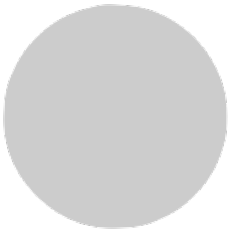
Congenital dyserythropoietic anemia type II (CDA II) is the most common form of CDA characterized by anemia, jaundice and splenomegaly and often leading to liver iron overload and gallstones.

Your genetic map

Gene	SNP	Genotype
SEC23B	rs121918221	GG
SEC23B	rs121918222	CC
SEC23B	rs398124225	CC
SEC23B	rs199939108	CC
SEC23B	rs727504145	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=98873

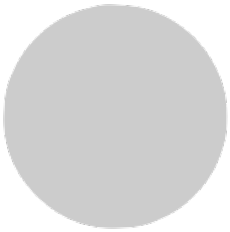
Hereditary Diseases (genetics)

Sickle cell anemia

A severe form of sickle cell disease (SCD) characterized by homozygosity for the sickle hemoglobin (HbS) gene and which acutely manifests with severe anemia, susceptibility to severe bacterial infections, and ischemic vasoocclusive accidents (VOA). It is a red cell disease of genetic origin which manifests with hemolytic disease and loss of red cell deformability leading to other occlusive events.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=232

Your genetic map

Gene	SNP	Genotype
HBB	rs33946267	CC
HBB	rs33950507	CC
HBB	rs33960103	CC
HBB	rs35424040	CC
HBB	rs35256489	AA
HBB	rs33986703	TT
HBB	rs11549407	GG
HBB	rs63750783	CC
HBB	rs33971440	CC
HBB	rs33945777	CC
HBB	rs33915217	CC
HBB	rs35004220	CC
HBB	rs34690599	GG
HBB	rs34451549	GG
HBB	rs33951465	AA
HBB	rs33941377	GG
HBB	rs33931746	TT
HBB	rs33978907	AA
HBB	rs33914668	TT
HBB	rs33941849	AA

Hereditary Diseases (genetics)

Hemolytic anemia due to glucophosphate isomerase deficiency

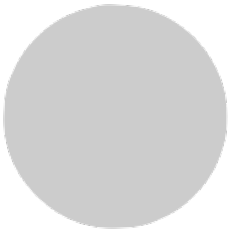
Glucosephosphate isomerase (GPI) deficiency is an erythroenzymopathy characterized by chronic nonspherocytic hemolytic anemia.

Your genetic map

Gene	SNP	Genotype
GPI	rs137853583	GG
GPI	rs61754634	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=712

Hereditary Diseases (genetics)

Hemolytic anemia due to pyrimidine 5' nucleotidase deficiency

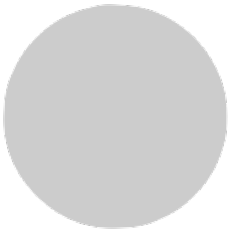
Hemolytic anemia due to pyrimidine 5' nucleotidase deficiency is a rare, hereditary, hemolytic anemia due to an erythrocyte nucleotide metabolism disorder characterized by mild to moderate hemolytic anemia associated with basophilic stippling and the accumulation of high concentrations of pyrimidine nucleotides within the erythrocyte. Patients present with variable features of jaundice, splenomegaly, hepatomegaly, gallstones, and sometimes require transfusions. Rare cases of mild development delay and learning difficulties are reported.

Your genetic map

Gene	SNP	Genotype
NT5C3A	rs104894025	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=35120

Hereditary Diseases (genetics)

Hemolytic anemia due to red cell pyruvate kinase deficiency

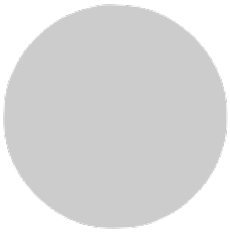
A rare, genetic metabolic disorder due to pyruvate kinase deficiency characterized by a variable degree of chronic nonspherocytic hemolytic anemia resulting in a variable clinical manifestations ranging from fatal anemia at birth to a to a fully compensated hemolysis without apparent anemia.

Your genetic map

Gene	SNP	Genotype
PKLR	rs118204085	CC
PKLR	rs113403872	CC
PKLR	rs201953584	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=766

Hereditary Diseases (genetics)

X-linked sideroblastic anemia

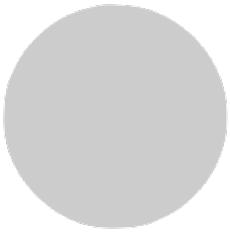
X-linked sideroblastic anemia is a constitutional microcytic, hypochromic anemia of varying severity that is clinically characterized by manifestations of anemia and iron overload and that may respond to treatment with pyridoxine and folic acid.

Your genetic map

Gene	SNP	Genotype
ALAS2	rs137852304	CC
ALAS2	rs137852311	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=75563

Hereditary Diseases (genetics)

X-linked sideroblastic anemia and spinocerebellar ataxia

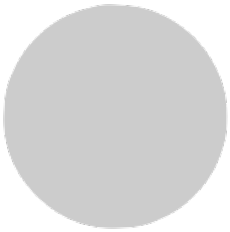
A rare syndromic, inherited form of sideroblastic anemia characterized by mild to moderate anemia (with hypochromia and microcytosis) and early-onset, non- or slowly progressive spinocerebellar ataxia.

Your genetic map

Gene	SNP	Genotype
ABCB7	rs72554634	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2802

Hereditary Diseases (genetics)

Enteric anendocrinosis

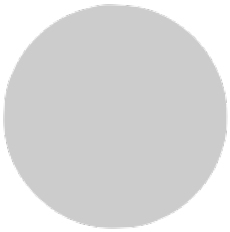
A very rare genetic gastroenterological disease characterized by severe malabsorptive diarrhea (requiring parenteral nutrition and disappearing at fasting) due to a lack of intestinal enteroendocrine cells. It is associated with early-onset (within the first weeks of life) dehydration, metabolic acidosis and diabetes mellitus (that can develop until late childhood). Patient may display various degrees of pancreatic insufficiency that does not explain diarrhea, as it is not reduced with pancreatic enzyme supplementation. Central hypogonadism (developing in the second decade), as well as an association with celiac disease have been reported.

Your genetic map

Gene	SNP	Genotype
LOC1019	rs121917837	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=83620

Hereditary Diseases (genetics)

Hereditary angioedema

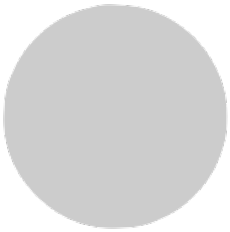
Hereditary angioedema (HAE) is a genetic disease characterized by the occurrence of transitory and recurrent subcutaneous and/or submucosal edemas resulting in swelling and/or abdominal pain.

Your genetic map

Gene	SNP	Genotype
SERPING	rs121907948	GG
SERPING	rs28940870	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=91378

Hereditary Diseases (genetics)

Distal anoctaminopathy

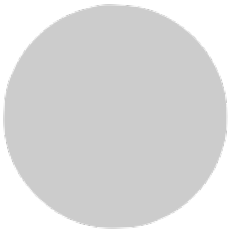
Distal anoctaminopathy is a rare, autosomal recessive distal myopathy characterized by early adult-onset, slowly progressive, often asymmetrical, lower limb muscle weakness initially affecting the calves (with relative anterior muscle sparing) and later proximal muscle involvement, as well as highly elevated creatine kinase (CK) serum levels.

Your genetic map

Gene	SNP	Genotype
ANO5	rs137854529	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=399096

Hereditary Diseases (genetics)

Peters anomaly

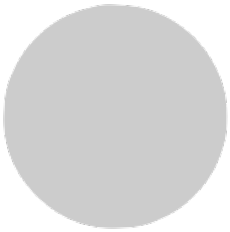
Peters anomaly (PA) is a congenital corneal opacity disorder characterized by a central corneal leukoma that obstructs the pupil leading to visual loss as well as absence of the posterior corneal stroma and Descemet membrane.

Your genetic map

Gene	SNP	Genotype
CYP1B1	rs72549387	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=708

Hereditary Diseases (genetics)

Rieger anomaly

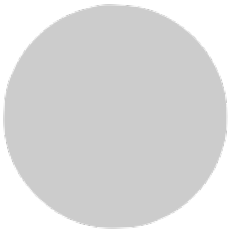
Rieger's anomaly is a congenital ocular defect caused by anterior segment dysgenesis and is characterized by severe anterior chamber deformity with prominent strands and marked atrophy of the iris stroma, with hole or pseudo-hole formation and corectopia. The term covers the association of these iris and pupil anomalies with the features of Axenfeldís anomaly (see this term).

Your genetic map

Gene	SNP	Genotype
PITX2	rs104893861	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=91483

Hereditary Diseases (genetics)

Uhl anomaly

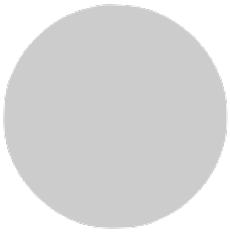
Uhl anomaly is characterized by an almost complete absence of the myocardium in the right ventricle resulting in a thin walled nonfunctional right ventricle manifesting with cardiac arrhythmias and right ventricular failure. Cases of partial absence of right ventricular myocardium which remains asymptomatic or mildly symptomatic until adulthood have also been reported. Patients presenting with complete Uhl anomaly should be considered for cardiac transplantation.

Your genetic map

Gene	SNP	Genotype
DSP	rs730880082	CC
PKP2	rs878898365	CC
SCN5A	rs1060499941	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3403

Hereditary Diseases (genetics)

46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency

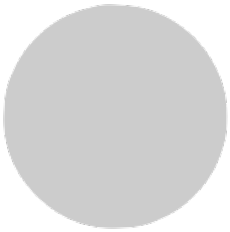
46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency is a rare, genetic, developmental defect during embryogenesis disorder characterized by severe, early-onset, salt-wasting adrenal insufficiency and ambiguous/female external genitalia (irrespective of chromosomal sex) due to mutations in the CYP11A1 gene. Milder cases may present delayed onset of adrenal gland dysfunction and genitalia phenotype may range from normal male to female in individuals with 46,XY karyotype. Imaging studies reveal hypoplastic/absent adrenal glands and biochemical findings include low serum cortisol, mineralocorticoids, androgens, and sodium, with elevated potassium levels.

Your genetic map

Gene	SNP	Genotype
CYP11A1	rs72547508	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=168558

Hereditary Diseases (genetics)

Isolated congenital anonychia

Isolated congenital anonychia is characterized by nail abnormalities ranging from onychodystrophy (dystrophic nails) to anonychia (absence of nails). Onychodystrophy-anonychia has been described in at least four generations of a family with male-to-male transmission, suggesting autosomal dominant transmission. Anonychia has been described in approximately less than 20 cases; it is likely to be transmitted as an autosomal recessive trait. Total anonychia congenita, in which all the fingernails and toenails are absent, may have an autosomal dominant inheritance pattern.

Your genetic map

Gene	SNP	Genotype
COL7A1	rs780261665	GG

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79143

Hereditary Diseases (genetics)

Aplasia of lacrimal and salivary glands

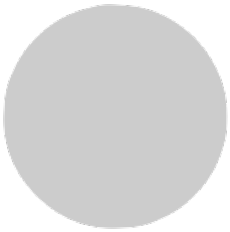
A rare autosomal dominant disorder characterized by aplasia, atresia or hypoplasia of the lacrimal and salivary glands leading to varying features since infancy such as recurrent eye infections, irritable eyes, epiphora, xerostomia, dental caries, dental erosion and oral inflammation.

Your genetic map

Gene	SNP	Genotype
FGF10	rs104893884	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=86815

Hereditary Diseases (genetics)

Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy

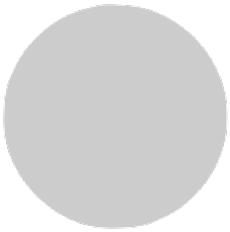
CADASIL (Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy) is a hereditary cerebrovascular disorder characterized by mid-adult onset of recurrent subcortical ischemic stroke and cognitive impairment progressing to dementia in addition to migraines with aura and mood disturbances seen in about a third of patients.

Your genetic map

Gene	SNP	Genotype
NOTCH3	rs137852642	GG
NOTCH3	rs201118034	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=136

Hereditary Diseases (genetics)

Systemic-onset juvenile idiopathic arthritis

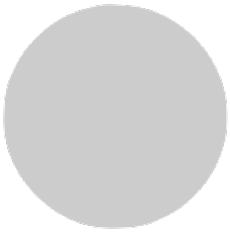
A rare pediatric rheumatological disease characterized by the variable occurrence of chronic arthritis, intermittently high spiking fever, maculopapular rash during fever episodes, hepatomegaly and/or splenomegaly, lymphadenopathy, and serositis.

Your genetic map

Gene	SNP	Genotype
LACC1	rs730880295	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=85414

Hereditary Diseases (genetics)

Distal arthrogryposis type 1

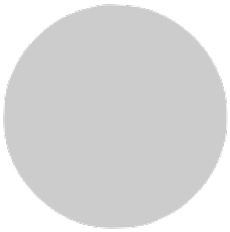
A form of arthrogryposis characterized by contractures of the distal regions of the hands and feet in the absence of a primary neurological and/or muscle disease affecting limb function. Facial involvement is limited to a small mouth and impaired mouth opening. No additional anomalies are reported.

Your genetic map

Gene	SNP	Genotype
TNNT3	rs199474721	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1146

Hereditary Diseases (genetics)

Distal arthrogryposis type 5D

Distal arthrogryposis type 5D is a rare subtype of distal arthrogryposis syndrome characterized by arthrogryposis multiplex congenita affecting the hands, feet, ankle, shoulders and/or neck, with camptodactyly of the fingers and limited knee and hip extension, associated with asymmetric ptosis and, less frequently, other ocular manifestations (e.g. ophthalmoplegia, strabismus). Affected individuals frequently have a bulbous nose, furrowed tongue, micro/retrognathia, a short neck, congenital hip dislocation, club feet, scoliosis and short stature.

Your genetic map

Gene	SNP	Genotype
ECEL1	rs532757890	GG
ECEL1	rs370167241	GG

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=329457

Hereditary Diseases (genetics)

Progressive pseudorheumatoid arthropathy of childhood

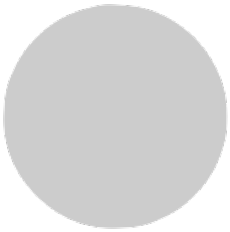
Progressive pseudorheumatoid arthropathy (dysplasia) of childhood (PPAC; PPD) presents as spondyloepiphyseal dysplasia (SED) tarda with progressive arthropathy and is described as a specific autosomal recessive subtype of SED.

Your genetic map

Gene	SNP	Genotype
CCN6	rs121908901	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1159

Hereditary Diseases (genetics)

VACTERL/VATER association

VACTERL/VATER is an association of congenital malformations typically characterized by the presence of at least three of the following: vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies, and limb abnormalities.

Your genetic map

Gene	SNP	Genotype
FOXF1	rs752504125	GG

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=887

Hereditary Diseases (genetics)

Aspartylglucosaminuria

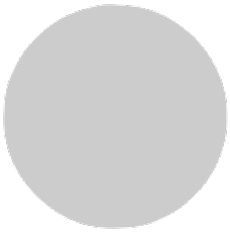
An autosomal recessive lysosomal storage disease belonging to the oligosaccharidosis group (also called glycoproteinosis).

Your genetic map

Gene	SNP	Genotype
AGA	rs121964904	CC
AGA	rs121964908	GG
AGA	rs386833437	CC
AGA	rs121964909	AA
AGA	rs386833431	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=93

Hereditary Diseases (genetics)

Autosomal recessive ataxia due to ubiquinone deficiency

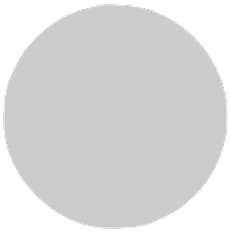
This syndrome is characterised by childhood-onset progressive ataxia and cerebellar atrophy.

Your genetic map

Gene	SNP	Genotype
COQ8A	rs119468004	GG
COQ8A	rs578189699	CC
COQ8A	rs771578775	CC
COQ8A	rs201908721	CC
COQ8A	rs752130338	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=139485

Hereditary Diseases (genetics)

Autosomal recessive ataxia, Beauce type

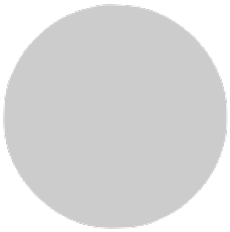
A rare disorder characterised by a slowly progressive pure cerebellar ataxia associated with dysarthria. It has been described in 53 individuals from 26 families of Canadian origin. The mode of transmission is autosomal recessive. Positional cloning has led to the identification of several SYNE1 gene mutations.

Your genetic map

Gene	SNP	Genotype
SYNE1	rs606231134	TT
SYNE1	rs797046025	GG
SYNE1	rs797046024	GG
SYNE1	rs375077588	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=88644

Hereditary Diseases (genetics)

Adult-onset autosomal recessive cerebellar ataxia

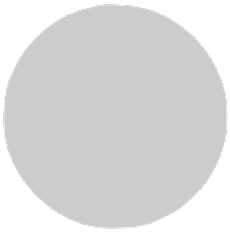
A rare, genetic, autosomal recessive cerebellar ataxia disease characterized by adulthood-onset of slowly progressive spinocerebellar ataxia, manifesting with gait and appendicular ataxia, dysarthria, ocular movement anomalies (e.g. horizontal, vertical, and/or downbeat nystagmus, hypermetric saccades), increased deep tendon reflexes and progressive cognitive decline. Additional variable features may include proximal leg muscle wasting and fasciculations, pes cavus, inspiratory stridor, epilepsy, retinal degeneration and cataracts. Brain imaging reveals marked cerebellar atrophy and electromyography shows evidence of lower motor neuron involvement.

Your genetic map

Gene	SNP	Genotype
ANO10	rs797045240	TT
ANO10	rs765592794	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=284289

Hereditary Diseases (genetics)

Autosomal recessive cerebellar ataxia due to CWF19L1 deficiency

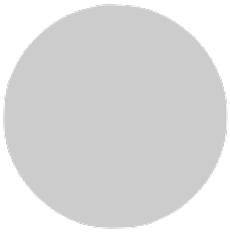
A rare autosomal recessive cerebellar ataxia characterized by early onset of slowly progressive cerebellar atrophy, clinically manifesting with extremity and truncal ataxia, global developmental delay, intellectual impairment, nystagmus, dysarthria, intention tremor, and pyramidal signs, among others.

Your genetic map

Gene	SNP	Genotype
CWF19L1	rs587780326	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=453521

Hereditary Diseases (genetics)

Non-progressive cerebellar ataxia with intellectual disability

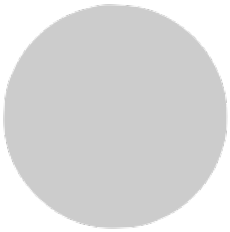
Non-progressive cerebellar ataxia with intellectual deficit is a rare subtype of autosomal dominant cerebellar ataxia type 1 (ADCA type 1; see this term) characterized by the onset in infancy of cerebellar ataxia, neonatal hypotonia (in some), mild developmental delay and, in later life, intellectual disability. Less common features include dysarthria, dysmetria and dysmorphic facial features (long face, bulbous nose long philtrum, thick lower lip and pointed chin).

Your genetic map

Gene	SNP	Genotype
CAMTA1	rs863224853	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=314647

Hereditary Diseases (genetics)

X-linked progressive cerebellar ataxia

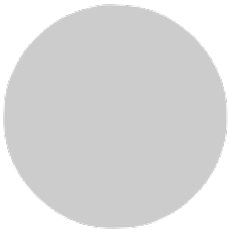
A rare X-linked cerebellar ataxia, characterized by a combination of upper and lower motor neuron signs, with an age of onset in the first or second decade, slow progression, and normal intelligence. Typical features of cerebellar dysfunction include gait and limb ataxia, intention tremor, dysmetria, dysdiadochokinesia, dysarthria, nystagmus, and hyperreflexia. Further phenotypic features are pes cavus, scoliosis, muscle atrophy, and peripheral sensory and motor nerve abnormalities.

Your genetic map

Gene	SNP	Genotype
ATP2B3	rs397514619	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1175

Hereditary Diseases (genetics)

Autosomal dominant spastic ataxia type 1

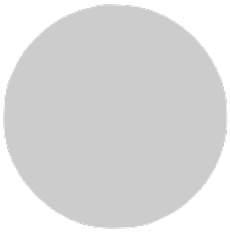
A rare, genetic, autosomal dominant spastic ataxia disorder characterized by lower-limb spasticity and ataxia in the form of head jerks, ocular movement abnormalities, dysarthria, dysphagia and gait disturbances.

Your genetic map

Gene	SNP	Genotype
TAPBPL	rs878854975	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=251282

Hereditary Diseases (genetics)

Spinocerebellar ataxia with epilepsy

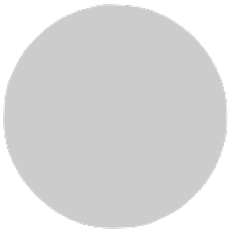
Spinocerebellar ataxia with epilepsy is a rare, mitochondrial DNA maintenance syndrome characterized by cerebellar ataxia, sensory peripheral neuropathy, myoclonus, epilepsy, progressive cognitive impairment, late-onset ptosis and external ophthalmoplegia. Liver failure may also occur, most often in association with the use of antiepileptic drug sodium valproate.

Your genetic map

Gene	SNP	Genotype
FANCI	rs139562274	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=254881

Hereditary Diseases (genetics)

Spinocerebellar ataxia with axonal neuropathy type 1

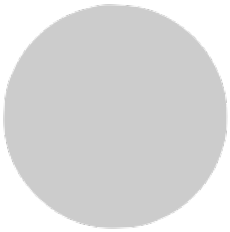
Spinocerebellar ataxia with axonal neuropathy type 1 is a rare, genetic neurological disorder characterized by a late childhood onset of slowly progressive cerebellar ataxia. Initial manifestations include weakness and atrophy of distal limb muscles, areflexia and loss of pain, vibration and touch sensations in upper and lower extremities. Gaze nystagmus, cerebellar dysarthria, peripheral neuropathy, stepagge gait and pes cavus develop as disease progresses. Cerebellar atrophy (especially of the vermis) is present in all affected individuals. Additional reported manifestations include seizures, mild brain atrophy, mild hypercholesterolemia and borderline hypoalbuminemia.

Your genetic map

Gene	SNP	Genotype
TDP1	rs370121773	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=94124

Hereditary Diseases (genetics)

Spinocerebellar ataxia with axonal neuropathy type 2

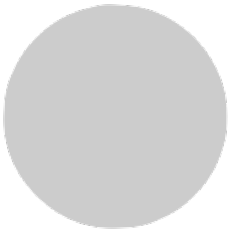
A rare autosomal recessive cerebellar ataxia (ARCA), characterized by progressive cerebellar ataxia associated with frequent oculomotor apraxia, severe neuropathy and an elevated serum alpha-fetoprotein (AFP) level.

Your genetic map

Gene	SNP	Genotype
SETX	rs29001665	GG
SETX	rs121434379	AA
SETX	rs797045068	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=64753

Hereditary Diseases (genetics)

Infantile-onset spinocerebellar ataxia

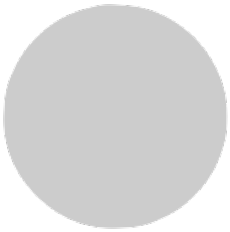
Infantile-onset spinocerebellar ataxia (IOSCA) is a hereditary neurological disorder with early and severe involvement of both the peripheral and central nervous systems. It has only been described in Finnish families.

Your genetic map

Gene	SNP	Genotype
TWNK	rs80356540	AA
TWNK	rs386834146	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1186

Hereditary Diseases (genetics)

Spinocerebellar ataxia type 13

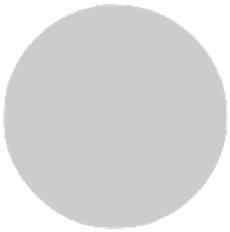
Spinocerebellar ataxia type 13 (SCA13) is a very rare subtype of type I autosomal dominant cerebellar ataxia (ADCA type I; see this term). It is characterized by onset in childhood marked by delayed motor and cognitive development followed by mild progression of cerebellar ataxia.

Your genetic map

Gene	SNP	Genotype
KCNC3	rs797044872	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=98768

Hereditary Diseases (genetics)

Spinocerebellar ataxia type 19/22

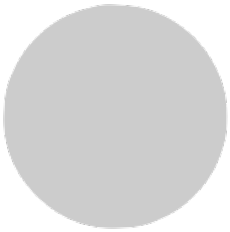
Spinocerebellar ataxia type 19 (SCA19) is a very rare subtype of type I autosomal dominant cerebellar ataxia (ADCA type I; see this term). It is characterized by mild cerebellar ataxia, cognitive impairment, low scores on the Wisconsin Card Sorting Test measuring executive function, myoclonus, and postural tremor.

Your genetic map

Gene	SNP	Genotype
KCND3	rs797045634	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=98772

Hereditary Diseases (genetics)

Spinocerebellar ataxia type 21

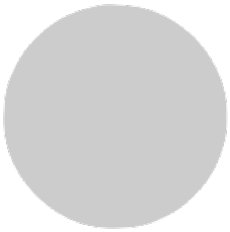
Spinocerebellar ataxia type 21 (SCA21) is a very rare subtype of type I autosomal dominant cerebellar ataxia (ADCA type I; see this term). It is characterized by slowly progressive cerebellar ataxia, mild cognitive impairment, postural and/or resting tremor, bradykinesia, and rigidity.

Your genetic map

Gene	SNP	Genotype
TMEM24	rs606231451	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=98773

Hereditary Diseases (genetics)

Spinocerebellar ataxia type 28

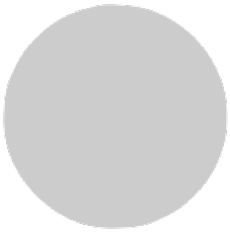
Spinocerebellar ataxia type 28 (SCA28) is a very rare subtype of type I autosomal dominant cerebellar ataxia (ADCA type I; see this term). It is characterized by juvenile onset, slowly progressive cerebellar ataxia due to Purkinje cell degeneration.

Your genetic map

Gene	SNP	Genotype
LOC1079	rs151344523	CC
LOC1079	rs151344514	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=101109

Hereditary Diseases (genetics)

Ataxia-oculomotor apraxia type 1

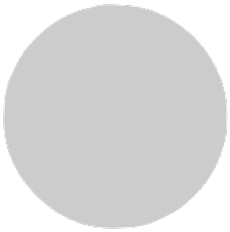
A rare autosomal recessive cerebellar ataxia, characterized by progressive cerebellar ataxia associated with oculomotor apraxia, severe neuropathy, and hypoalbuminemia.

Your genetic map

Gene	SNP	Genotype
APTX	rs104894103	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1168

Hereditary Diseases (genetics)

Multiple intestinal atresia

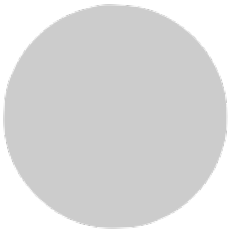
Multiple intestinal atresia is a rare form of intestinal atresia characterized by the presence of numerous atresic segments in the small bowel (duodenum) or large bowel and leading to symptoms of intestinal obstruction: vomiting, abdominal bloating and inability to pass meconium in newborns.

Your genetic map

Gene	SNP	Genotype
TTC7A	rs886042805	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2300

Hereditary Diseases (genetics)

Gyrate atrophy of choroid and retina

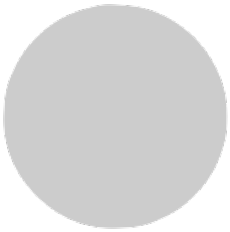
Gyrate atrophy of the choroid and retina (GACR) is a very rare, inherited retinal dystrophy, characterized by progressive chorioretinal atrophy, myopia and early cataract.

Your genetic map

Gene	SNP	Genotype
OAT	rs121965040	CC
OAT	rs121965043	AA
OAT	rs121965053	CC
OAT	rs386833598	AA
OAT	rs386833618	GG
OAT	rs386833621	CC
OAT	rs200068769	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=414

Hereditary Diseases (genetics)

Autosomal dominant congenital benign spinal muscular atrophy

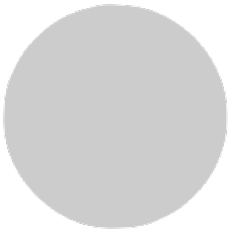
A rare distal hereditary motor neuropathy, with a variable clinical phenotype, typically characterized by congenital, non-progressive, predominantly distal, lower limb muscle weakness and atrophy and congenital (or early-onset) flexion contractures of the hip, knee and ankle joints. Reduced or absent lower limb deep tendon reflexes, skeletal anomalies (bilateral talipes equinovarus, scoliosis, kyphoscoliosis, lumbar hyperlordisis), late ambulation, waddling gait, joint hyperlaxity and/or bladder and bowel dysfuntion are usually also associated.

Your genetic map

Gene	SNP	Genotype
TRPV4	rs267607144	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1216

Hereditary Diseases (genetics)

Spinal muscular atrophy with respiratory distress type 1

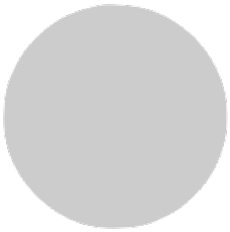
Spinal muscular atrophy with respiratory distress type 1 is a rare genetic motor neuron disease characterized by severe respiratory distress/respiratory failure in association with diaphragmatic eventration and palsy, as well as progressive, symmetrical, distal-to-proximal muscle weakness and atrophy (in lower limbs especially). Patients typically have a history of intrauterine growth retardation, low birth weight, feeble cry, weak suck and failure to thrive and present with inspiratory stridor, recurrent episodes of dyspnea or apnea, cyanosis and absent deep tendon reflexes. Kyphosis/scoliosis, foot deformities and joint contractures are frequently associated features.

Your genetic map

Gene	SNP	Genotype
IGHMBP2	rs137852665	GG
IGHMBP2	rs137852667	GG
IGHMBP2	rs200089714	CC
IGHMBP2	rs35193202	CC
IGHMBP2	rs145226920	CC
IGHMBP2	rs797044802	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=98920

Hereditary Diseases (genetics)

Scapuloperoneal spinal muscular atrophy

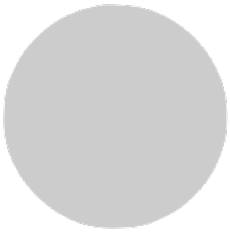
A rare, genetic motor neuron disease characterized by predominantly motor axonal peripheral neuropathy manifesting with progressive scapuloperoneal muscular atrophy and weakness, laryngeal palsy, congenital absence of muscles, and, in some, skeletal abnormalities.

Your genetic map

Gene	SNP	Genotype
TRPV4	rs267607143	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=431255

Hereditary Diseases (genetics)

Autosomal dominant childhood-onset proximal spinal muscular atrophy

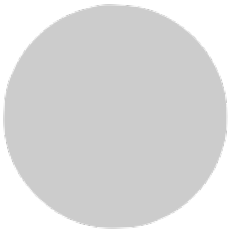
A rare genetic neuromuscular disease characterized by early onset muscular weakness with predominant proximal lower limb involvement. The disorder is static or only mildly progressive. The severity of manifestations ranges from lethal, congenital muscular atrophy with arthrogryposis to asymptomatic with subclinical features.

Your genetic map

Gene	SNP	Genotype
DYNC1H1	rs587780564	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=209341

Hereditary Diseases (genetics)

Congenital bilateral absence of vas deferens

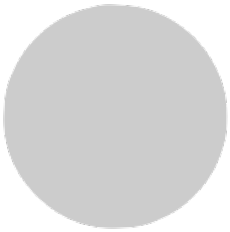
Congenital bilateral absence of the vas deferens (CBAVD) is a condition leading to male infertility.

Your genetic map

Gene	SNP	Genotype
CFTR	rs78655421	GG
CFTR AS1	rs121908805	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=48

Hereditary Diseases (genetics)

Autosomal recessive bestrophinopathy

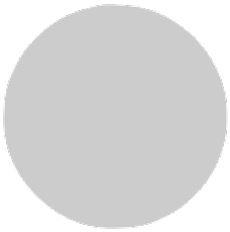
A rare retinal dystrophy, characterized by central visual loss in the first 2 decades of life, associated with an absent electrooculogram (EOG) light rise and a reduced electroretinogram (ERG).

Your genetic map

Gene	SNP	Genotype
LOC1079	rs200277476	CC
LOC1079	rs281865238	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=139455

Hereditary Diseases (genetics)

Beta-mannosidosis

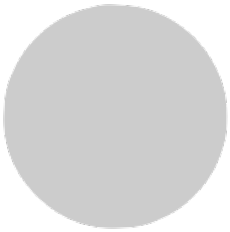
Beta-mannosidosis is a very rare lysosomal storage disease characterized by developmental delay of varying severity and hearing loss, but that can manifest a wide phenotypic heterogeneity.

Your genetic map

Gene	SNP	Genotype
MANBA	rs374545788	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=118

Hereditary Diseases (genetics)

Beta-thalassemia

Beta-thalassemia (BT) is characterized by deficiency (Beta+) or absence (Beta0) of synthesis of the beta globin chains of hemoglobin (Hb).

Your genetic map

Gene	SNP	Genotype
HBB	rs33941849	AA
HBB	rs34999973	GG

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=848

Hereditary Diseases (genetics)

Bradyopsia

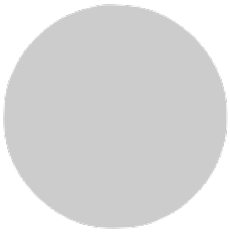
Bradyopsia is characterised by prolonged electroretinal response suppression leading to difficulties adjusting to changes in luminance, normal to subnormal acuity and photophobia.

Your genetic map

Gene	SNP	Genotype
RGS9	rs121908449	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=75374

Hereditary Diseases (genetics)

Autosomal dominant brachyolmia

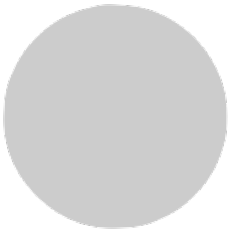
A relatively severe form of brachyolmia, a group of rare genetic skeletal disorders, characterized by short-trunked short stature, platyspondyly and kyphoscoliosis. Degenerative joint disease (osteoarthropathy) in the spine, large joints and interphalangeal joints becomes manifest in adulthood.

Your genetic map

Gene	SNP	Genotype
TRPV4	rs121912633	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=93304

Hereditary Diseases (genetics)

Nasopharyngeal carcinoma

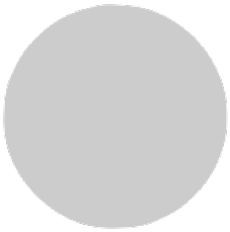
Nasopharyngeal carcinoma (NPC) is a tumor arising from the epithelial cells that cover the surface and line the nasopharynx.

Your genetic map

Gene	SNP	Genotype
TP53	rs121912660	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=150

Hereditary Diseases (genetics)

Familial papillary or follicular thyroid carcinoma

Familial papillary or follicular thyroid carcinoma is a rare, hereditary nonmedullary thyroid carcinoma characterized by the presence of differentiated thyroid cancer of follicular cell origin in two or more first-degree relatives, in the absence of other familial tumor syndromes or radiation exposure. Frequent capsular invasion is observed. Biopsy reveals multicentric tumors with multiple adenomatous nodules with or without oxyphilia and follicular or papillary carcinoma histology.

Your genetic map

Gene	SNP	Genotype
BRAF	rs121913364	TT
NRAS	rs11554290	TT

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=319487

Hereditary Diseases (genetics)

Cystinuria

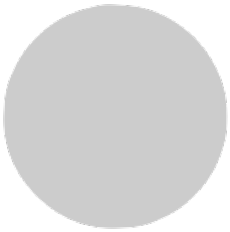
A rare disorder of renal tubular amino acid transport characterized by recurrent formation of kidney cystine stones.

Your genetic map

Gene	SNP	Genotype
SLC3A1	rs200483989	CC
SLC7A9	rs121908480	CC
SLC7A9	rs121908484	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=214

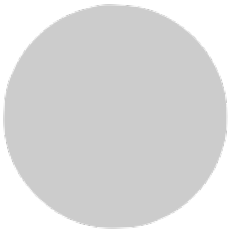
Hereditary Diseases (genetics)

Citrullinemia type I

Citrullinemia type I is a rare autosomal recessive urea cycle defect characterized biologically by hyperammonemia and clinically by progressive lethargy, poor feeding and vomiting in the neonatal form (Acute neonatal citrullinemia type I, see this term) and by variable hyperammonemia in the later-onset form (Adult-onset citrullinemia type I, see this term).

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=247525

Your genetic map

Gene	SNP	Genotype
ASS1	rs121908638	GG
ASS1	rs121908639	GG
ASS1	rs121908645	CC
ASS1	rs121908646	TT
ASS1	rs398123130	AA
ASS1	rs192838388	GG
ASS1	rs148918985	CC
ASS1	rs398123131	GG
ASS1	rs371265106	GG
ASS1	rs751930594	AA
ASS1	rs183276875	CC
LOC1053	rs121908640	CC
LOC1053	rs121908641	GG
LOC1053	rs121908647	GG
LOC1053	rs727503814	GG
LOC1053	rs771937610	GG

Hereditary Diseases (genetics)

Keratosis follicularis spinulosa decalvans

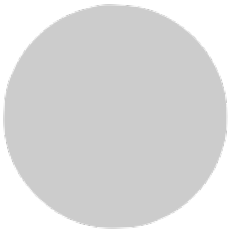
A severe subtype of citrin deficiency characterized clinically by adult onset (20 and 50 years of age), recurrent episodes of hyperammonemia and associated neuropsychiatric symptoms such as nocturnal delirium, confusion, restlessness, disorientation, drowsiness, memory loss, abnormal behavior (aggression, irritability, and hyperactivity), seizures, and coma.

Your genetic map

Gene	SNP	Genotype
SLC25A13	rs80338721	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=247585

Hereditary Diseases (genetics)

COG4-CDG

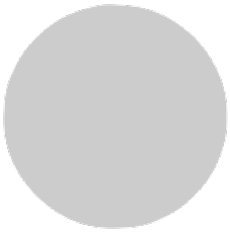
COG4-CDG is an extremely rare form of CDG syndrome characterized clinically in the single reported case to date by seizures, some dysmorphic features, axial hyponia, slight peripheral hypertonia and hyperreflexia.

Your genetic map

Gene	SNP	Genotype
COG4	rs267606740	GG
COG4	rs376663459	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=263501

Hereditary Diseases (genetics)

COG5-CDG

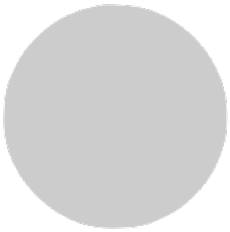
COG5-CDG is an extremely rare form of CDG syndrome characterized clinically in the single reported case to date by moderate mental retardation with slow and inarticulate speech, truncal ataxia, and mild hypotonia.

Your genetic map

Gene	SNP	Genotype
COG5	rs548774836	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=263487

Hereditary Diseases (genetics)

Progressive familial intrahepatic cholestasis

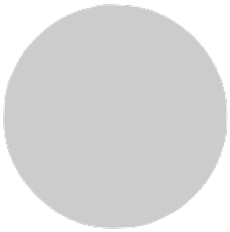
Progressive familial intrahepatic cholestasis (PFIC) refers to a heterogeneous group of autosomal recessive disorders of childhood that disrupt bile formation and present with cholestasis of hepatocellular origin.

Your genetic map

Gene	SNP	Genotype
ABCB4	rs863225298	GG
ABCB4	rs377160065	GG
NR1H4	rs113090017	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=172

Hereditary Diseases (genetics)

Neonatal intrahepatic cholestasis due to citrin deficiency

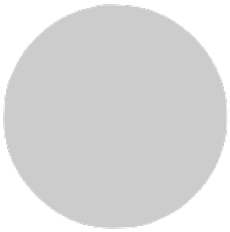
A mild subtype of citrin deficiency characterized clinically by low birth weight, failure to thrive, transient intrahepatic cholestasis, multiple aminoacidemia, galactosemia, hypoproteinemia, hepatomegaly, decreased coagulation factors, hemolytic anemia, variable but mostly mild liver dysfunction, and hypoglycemia.

Your genetic map

Gene	SNP	Genotype
SLC25A13	rs80338722	CC
SLC25A13	rs80338729	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=247598

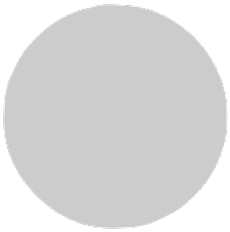
Hereditary Diseases (genetics)

Tuberous sclerosis complex

A rare neurocutaneous disorder characterized by multisystem hamartomas, most commonly involving the skin, brain, kidneys, lungs, eye, and heart, and associated with neuropsychiatric disorders.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=805

Your genetic map

Gene	SNP	Genotype
TSC1	rs118203447	AA
TSC1	rs118203426	AA
TSC1	rs118203504	GG
TSC1	rs118203537	GG
TSC1	rs118203542	GG
TSC1	rs118203549	GG
TSC1	rs118203345	AA
TSC1	rs118203606	GG
TSC1	rs118203610	CC
TSC1	rs118203614	CC
TSC1	rs118203631	GG
TSC1	rs118203353	CC
TSC1	rs118203352	TT
TSC1	rs118203647	GG
TSC1	rs118203661	GG
TSC1	rs118203668	GG
TSC1	rs118203680	GG
TSC1	rs118203682	GG
TSC1	rs118203687	CC
TSC1	rs118203727	GG
TSC1	rs118203728	GG
TSC1	rs118203732	GG
TSC1	rs118203384	GG
TSC1	rs118203402	CC
TSC1	rs118203403	AA
TSC1	rs118203423	CC
TSC1	rs118203427	GG
TSC1	rs118203434	GG
TSC1	rs118203438	CC
TSC1	rs118203440	TT
TSC1	rs118203450	CC

Hereditary Diseases (genetics)

Metaphyseal chondrodysplasia, Spahr type

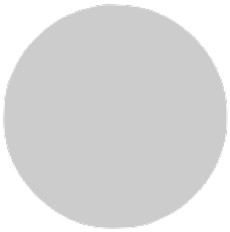
A rare, genetic, primary bone dysplasia disease characterized by usually moderate, postnatal short stature, progressive genu vara deformity, a waddling gait, and radiological signs of metaphyseal dysplasia (i.e. irregular, sclerotic and widened metaphyses), in the absence of biochemical abnormalities suggestive of rickets disease. Intermittent knee pain, lordosis, and delayed motor development may also occasionally be associated.

Your genetic map

Gene	SNP	Genotype
MMP13	rs140059558	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2501

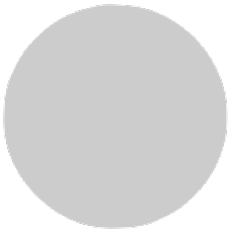
Hereditary Diseases (genetics)

X-linked dominant chondrodysplasia punctata

A rare genodermatosis disease with great phenotypic variation and characterized most commonly by ichthyosis following the lines of Blaschko, chondrodysplasia punctata (CDP), asymmetric shortening of the limbs, cataracts and short stature.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=35173

Your genetic map

Gene	SNP	Genotype
EBP	rs104894799	CC
EBP	rs104894800	GG
EBP	rs587783599	GG
EBP	rs587783601	GG
EBP	rs587783602	TT
EBP	rs587783603	GG
EBP	rs587783605	TT
EBP	rs587783607	GG
EBP	rs587783608	AA
EBP	rs587783609	TT
EBP	rs587783610	AA
EBP	rs587783611	CC
EBP	rs587783612	GG
EBP	rs587783613	CC
EBP	rs587783614	TT
EBP	rs587783616	TT
EBP	rs587783617	GG
EBP	rs587783619	TT

Hereditary Diseases (genetics)

Infantile convulsions and choreoathetosis

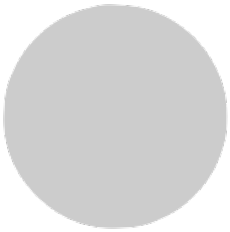
Infantile Convulsions and paroxysmal ChoreoAthetosis (ICCA) syndrome is a neurological condition characterized by the occurrence of seizures during the first year of life (Benign familial infantile epilepsy ; see this term) and choreoathetotic dyskinetic attacks during childhood or adolescence.

Your genetic map

Gene	SNP	Genotype
PRRT2	rs387907126	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=31709

Hereditary Diseases (genetics)

Paroxysmal dystonic choreathetosis with episodic ataxia and spasticity

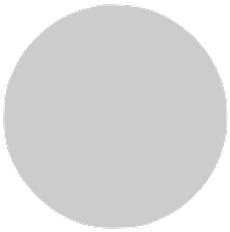
A rare, genetic, paroxysmal dystonia disorder characterized by childhood to adolescent-onset of episodic paroxysmal choreoathetosis, triggered mainly by sudden movements, prolonged exercise, anxiety and emotional stress, in association with progressive spastic paraparesis (onest in adulthood), gait ataxia, mild to moderate cognitive impairment, and/or epileptic seizures. Episodes typically last from a few minutes to hours, have a variable frequency (daily to yearly), and are relieved by rest. Frequency of episodes tends to decrease with age.

Your genetic map

Gene	SNP	Genotype
SLC2A1	rs387907312	GG
SLC2A1	rs796053254	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=53583

Hereditary Diseases (genetics)

Cranio-osteoarthropathy

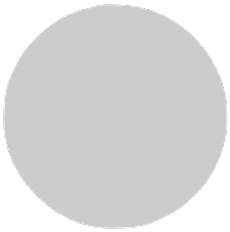
Cranio-osteoarthropathy (COA) is a form of primary hypertrophic osteoarthropathy characterized by delayed closure of the cranial sutures and fontanels, digital clubbing, arthropathy, and periostosis.

Your genetic map

Gene	SNP	Genotype
HPGD	rs121434480	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1525

Hereditary Diseases (genetics)

Hereditary cryohydrocytosis with reduced stomatin

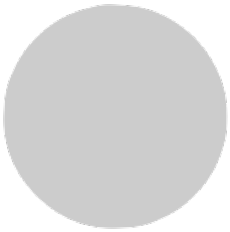
Hereditary cryohydrocytosis with reduced stomatin is a rare hemolytic anemia characterized by combination of neurologic features, such as psychomotor delay, seizures, variable movement disorders, and hemolytic anemia with stomatocytosis, resulting in cation-leaky erythrocytes, pseudohyperkalemia, hemolytic crises and hepatosplenomegaly. Cataracts are also a presenting feature.

Your genetic map

Gene	SNP	Genotype
SLC2A1	rs796053272	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=168577

Hereditary Diseases (genetics)

Autosomal recessive cutis laxa type 1

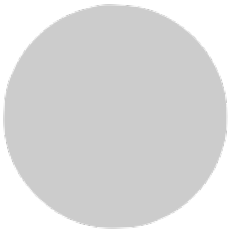
A generalized connective tissue disorder characterized by the association of wrinkled, redundant and sagging inelastic skin with severe systemic manifestations (lung atelectesias and emphysema, vascular anomalies, and gastrointestinal and genitourinary tract diverticuli).

Your genetic map

Gene	SNP	Genotype
EFEMP2	rs193302867	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=90349

Hereditary Diseases (genetics)

Autosomal recessive cutis laxa type 2A

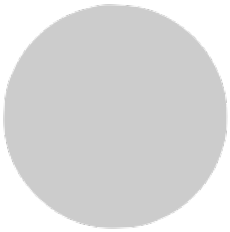
A rare, genetic, dermis elastic tissue disease characterized by redundant, overfolded skin of variable severity, ranging from wrinkly skin to cutis laxa associated with pre- and post-natal growth retardation, hypotonia, mild to moderate developmental delay, late closure of anterior fontanelle, and craniofacial dysmorphism (including microcephaly, hypertelorism, downslanting palpebral fissures, large, prominent nasal root with funnel nose, small, low-set ears, long philtrum, drooping facial skin). Additional manifestations may include seizures, intellectual disability, congenital hip dislocation, inguinal hernia, and cortical and cerebellar malformations. Pretibial pseudo-ecchymotic skin lesions have occasionally been associated.

Your genetic map

Gene	SNP	Genotype
ATP6V0A	rs374480381	GG
LOC1053	rs80356750	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=357074

Hereditary Diseases (genetics)

Autosomal recessive cutis laxa type 2B

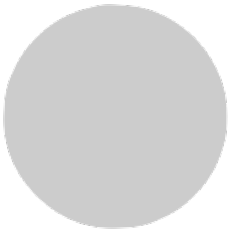
A rare, hereditary, developmental defect with connective tissue involvement characterized by cutis laxa of variable severity, in utero growth restriction, congenital hip dislocation and joint hyperlaxity, wrinkling of the skin, in particular the dorsum of hands and feet, and progeroid facial features. Hypotonia, developmental delay, and intellectual disability are common. In addition, cataracts, corneal clouding, wormian bones, lipodystrophy and osteopenia have been reported.

Your genetic map

Gene	SNP	Genotype
PYCR1	rs121918377	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=357064

Hereditary Diseases (genetics)

DDOST-CDG

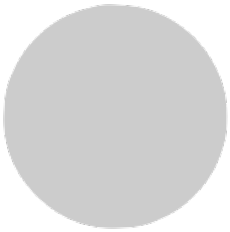
DDOST-CDG is a form of congenital disorders of N-linked glycosylation characterized by failure to thrive, developmental delay, hypotonia, strabismus and hepatic dysfunction. The disease is caused by mutations in the gene DDOST (1p36.1).

Your genetic map

Gene	SNP	Genotype
DDOST	rs387906831	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=300536

Hereditary Diseases (genetics)

Congenital bile acid synthesis defect type 1

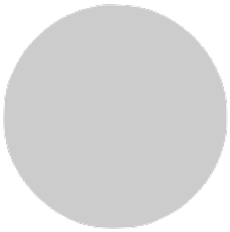
Congenital bile acid synthesis defect type 1 (BAS defect type 1) is the most common anomaly of bile acid synthesis characterized by variable manifestations of progressive cholestatic liver disease, and fat malabsorption.

Your genetic map

Gene	SNP	Genotype
HSD3B7	rs104894518	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79301

Hereditary Diseases (genetics)

Congenital bile acid synthesis defect type 4

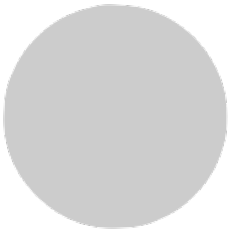
Congenital bile acid synthesis defect type 4 (BAS defect type 4) is an anomaly of bile acid synthesis characterized by mild cholestatic liver disease, fat malabsorption and/or neurological disease.

Your genetic map

Gene	SNP	Genotype
C1QTNF3	rs121917814	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79095

Hereditary Diseases (genetics)

Isolated cytochrome C oxidase deficiency

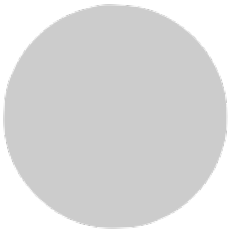
A rare mitochondrial oxidative phosphorylation disorder characterized by a highly variable clinical phenotype, including a benign infantile mitochondrial type affecting mainly the skeletal muscle, a lethal infantile mitochondrial myopathy linked to severe metabolic acidosis and mitochondrial dysfunction in skeletal muscle and often also in heart, Leigh syndrome, which causes severe, early-onset, progressive, and fatal encephalopathy, and French-Canadian type Leigh syndrome, which affects mostly the skeletal muscle, but also brain and liver.

Your genetic map

Gene	SNP	Genotype
MT TN	rs199476130	GG
PET100	rs587777839	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=254905

Hereditary Diseases (genetics)

Isolated complex I deficiency

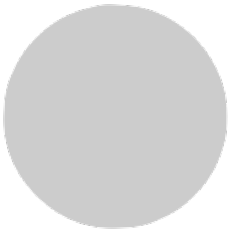
Isolated complex I deficiency is a rare inborn error of metabolism due to mutations in nuclear or mitochondrial genes encoding subunits or assembly factors of the human mitochondrial complex I (NADH: ubiquinone oxidoreductase) and is characterized by a wide range of manifestations including marked and often fatal lactic acidosis, cardiomyopathy, leukoencephalopathy, pure myopathy and hepatopathy with tubulopathy. Among the numerous clinical phenotypes observed are Leigh syndrome, Leber hereditary optic neuropathy and MELAS syndrome (see these terms).

Your genetic map

Gene	SNP	Genotype
NDUFS3	rs28939714	CC
NDUFS3	rs104894270	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2609

Hereditary Diseases (genetics)

Isolated complex III deficiency

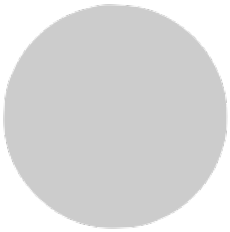
Isolated complex III deficiency is a rare, genetic, mitochondrial oxidative phosphorylation disorder characterized by a wide spectrum of clinical manifestations ranging from isolated myopathy or transient hepatopathy to severe multisystem disorder (that may include hypotonia, failure to thrive, psychomotor delay, cardiomyopathy, encephalopathy, renal tubulopathy, hearing impairment, lactic acidosis, hypoglycemia and other signs and symptoms).

Your genetic map

Gene	SNP	Genotype
TTC19	rs747166010	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1460

Hereditary Diseases (genetics)

Non-acquired isolated growth hormone deficiency

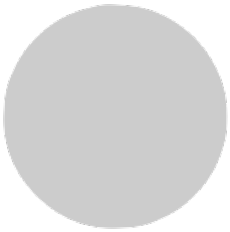
A rare non-acquired pituitary hormone deficiency characterized by growth deficiency, delayed bone age, and short stature of variable severity and age of onset, and with variable response to treatment with recombinant human growth hormone, depending on the respective subtype of the disease. Hormone deficiency may be quantitative or qualitative in nature.

Your genetic map

Gene	SNP	Genotype
GH1	rs71640277	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=631

Hereditary Diseases (genetics)

Combined oxidative phosphorylation defect type 15

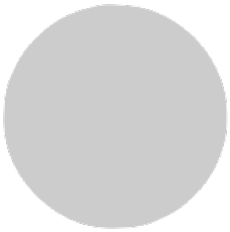
Combined oxidative phosphorylation defect type 15 is a rare mitochondrial disease due to a defect in mitochondrial protein synthesis characterized by onset in infancy or early childhood of muscular hypotonia, gait ataxia, mild bilateral pyramidal tract signs, developmental delay (affecting mostly speech and coordination) and subsequent intellectual disability. Short stature, obesity, microcephaly, strabismus, nystagmus, reduced visual acuity, lactic acidosis, and a brain neuropathology consistent with Leigh syndrome are also reported.

Your genetic map

Gene	SNP	Genotype
MTFMT	rs201431517	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=319524

Hereditary Diseases (genetics)

Combined oxidative phosphorylation defect type 20

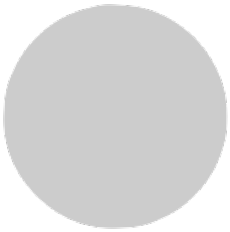
Combined oxidative phosphorylation defect type 20 is a rare mitochondrial oxidative phosphorylation disorder characterized by variable combination of psychomotor delay, hypotonia, muscle weakness, seizures, microcephaly, cardiomyopathy and mild dysmorphic facial features. Variable types of structural brain anomalies have also been reported. Biochemical studies typically show decreased activity of mitochondrial complexes (mainly complex I).

Your genetic map

Gene	SNP	Genotype
VARS2	rs143821815	GG
VARS2	rs769768815	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=420728

Hereditary Diseases (genetics)

Combined oxidative phosphorylation defect type 8

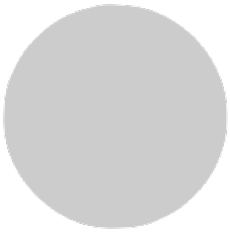
Combined oxidative phosphorylation defect type 8 is a mitochondrial disease due to a defect in mitochondrial protein synthesis resulting in deficiency of respiratory chain complexes I, III and IV in the cardiac and skeletal muscle and brain characterized by severe hypertrophic cardiomyopathy, pulmonary hypoplasia, generalized muscle weakness and neurological involvement.

Your genetic map

Gene	SNP	Genotype
AARS2	rs138119149	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=319504

Hereditary Diseases (genetics)

Congenital intrinsic factor deficiency

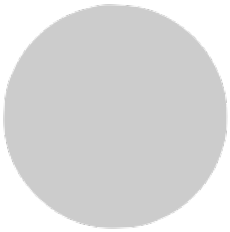
Congenital intrinsic factor deficiency (IFD) is a rare disorder of vitamin B12 (cobalamin) absorption that is characterized by megaloblastic anemia and neurological abnormalities.

Your genetic map

Gene	SNP	Genotype
CBLIF	rs147785187	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=332

Hereditary Diseases (genetics)

Congenital fibrinogen deficiency

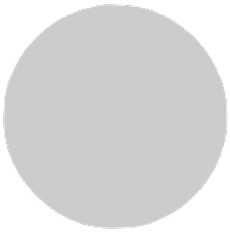
Congenital deficiencies of fibrinogen are coagulation disorders characterized by bleeding symptoms ranging from mild to severe resulting from reduced quantity and/or quality of circulating fibrinogen. Afibrinogenemia (complete absence of fibrinogen) and hypofibrinogenemia (reduced plasma fibrinogen concentration) (see these terms) correspond to quantitative anomalies of fibrinogen while dysfibrinogenemia corresponds to a functional anomaly of fibrinogen. Hypo- and dysfibrinogenemia may be frequently combined (hypodysfibrinogenemia).

Your genetic map

Gene	SNP	Genotype
FGA	rs146387238	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=335

Hereditary Diseases (genetics)

Congenital sucrase-isomaltase deficiency

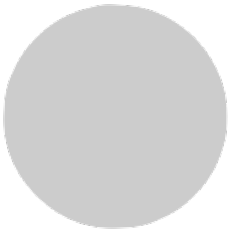
A rare, genetic, congenital carbohydrate intolerance disorder characterized by lack of endogenous sucrase activity, marked reduction in isomaltase activity, and moderate decrease in maltase activity, and clinically manifesting with diarrhea, abdominal pain and bloating, failure to thrive.

Your genetic map

Gene	SNP	Genotype
SI	rs200451408	GG
SI	rs200328403	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=35122

Hereditary Diseases (genetics)

Congenital factor V deficiency

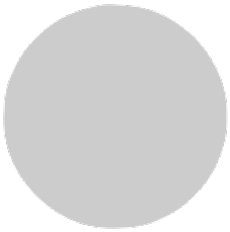
Congenital factor V deficiency is an inherited bleeding disorder due to reduced plasma levels of factor V (FV) and characterized by mild to severe bleeding symptoms.

Your genetic map

Gene	SNP	Genotype
F5	rs118203907	TT
F5	rs118203910	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=326

Hereditary Diseases (genetics)

Congenital factor XI deficiency

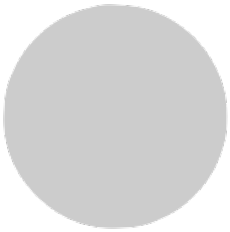
A rare inherited bleeding disorder characterized by reduced levels and/or activity of factor XI (FXI) resulting in moderate bleeding symptoms, usually occurring after trauma or surgery.

Your genetic map

Gene	SNP	Genotype
F11	rs121965063	GG
F11	rs121965064	TT
F11	rs28934608	CC
F11	rs121965069	TT
F11	rs121965071	GG
F11	rs770505620	CC
F11 AS1	rs281875250	CC
F11 AS1	rs201007090	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=329

Hereditary Diseases (genetics)

Congenital factor XIII deficiency

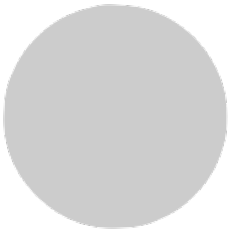
Congenital factor XIII deficiency is an inherited bleeding disorder due to reduced levels and activity of factor XIII (FXIII) and characterized by hemorrhagic diathesis frequently associated with spontaneous abortions and defective wound healing. Factor XIII deficiency is one of the most rare coagulation factor deficiencies.

Your genetic map

Gene	SNP	Genotype
F13A1	rs372296352	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=331

Hereditary Diseases (genetics)

3-phosphoglycerate dehydrogenase deficiency, infantile/juvenile form

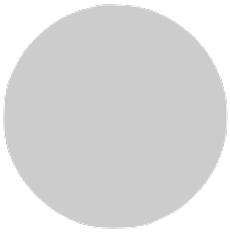
3-Phosphoglycerate dehydrogenase deficiency (3-PGDH deficiency) is an autosomal recessive form of serine deficiency syndrome characterized clinically in the few reported cases by congenital microcephaly, psychomotor retardation and intractable seizures in the infantile form and by absence seizures, moderate developmental delay and behavioral disorders in the juvenile form

Your genetic map

Gene	SNP	Genotype
PHGDH	rs121907987	GG
PHGDH	rs886041874	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79351

Hereditary Diseases (genetics)

3-hydroxy-3-methylglutaryl-CoA synthase deficiency

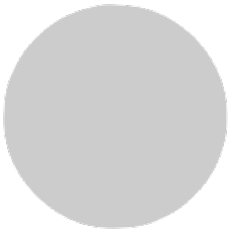
3-hydroxy-3-methylglutaryl-CoA synthase deficiency (HMG-CoA synthase deficiency) is a rare autosomal recessively inherited disorder of ketone body metabolism (see this term), reported in less than 20 patients to date, characterized clinically by episodes of decompensation (often associated with gastroenteritis or fasting) that present with vomiting, lethargy, hepatomegaly, non ketotic hypoglycemia and, in rare cases, coma. Patients are mostly asymptomatic between acute epidodes. HMG-CoA synthase deficiency requires an early diagnosis in order to avoid hypoglycemic crises that can lead to permanent brain damage or death.

Your genetic map

Gene	SNP	Genotype
HMGCS2	rs137852638	CC
HMGCS2	rs142637231	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=35701

Hereditary Diseases (genetics)

Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency

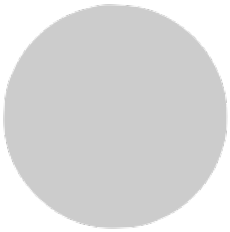
A mitochondrial disorder of long chain fatty acid oxidation characterized in most patients by onset in infancy/ early childhood of hypoketotic hypoglycemia, metabolic acidosis, liver disease, hypotonia and, frequently, cardiac involvement with arrhythmias and/or cardiomyopathy.

Your genetic map

Gene	SNP	Genotype
GAREM2	rs794727219	CC
HADHA	rs786204607	GG
LOC1079	rs1057516217	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=5

Hereditary Diseases (genetics)

Acyl-CoA dehydrogenase 9 deficiency

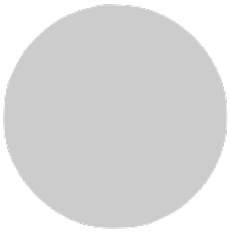
A rare disorder characterized by neurological dysfunction, hepatic failure and cardiomyopathy due to a deficiency of complex I of the respiratory chain.

Your genetic map

Gene	SNP	Genotype
ACAD9	rs368949613	CC
ACAD9	rs387907042	GG
ACAD9	rs753711253	CC
ACAD9	rs773586510	GG
ACAD9	rs149753643	GG
ACAD9	rs150283105	CC
CFAP92	rs377022708	CC
CFAP92	rs863224845	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=99901

Hereditary Diseases (genetics)

Short chain acyl-CoA dehydrogenase deficiency

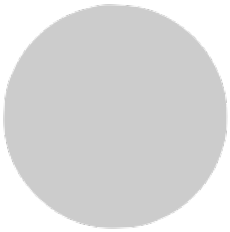
Short-chain acyl-CoA dehydrogenase (SCAD) deficiency is a very rare inborn error of mitochondrial fatty acid oxidation characterized by variable manifestations ranging from asymptomatic individuals (in most cases) to those with failure to thrive, hypotonia, seizures, developmental delay and progressive myopathy.

Your genetic map

Gene	SNP	Genotype
ACADS	rs121908003	CC
ACADS	rs57443665	TT
ACADS	rs28940872	CC
ACADS	rs121908006	CC
ACADS	rs28941773	CC
ACADS	rs387906950	AA
ACADS	rs140853839	CC
ACADS	rs796051905	GG
ACADS	rs749491616	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=26792

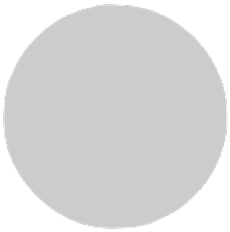
Hereditary Diseases (genetics)

Medium chain acyl-CoA dehydrogenase deficiency

Medium chain acyl-CoA dehydrogenase (MCAD) deficiency (MCADD) is an inborn error of mitochondrial fatty acid oxidation characterized by a rapidly progressive metabolic crisis, often presenting as hypoketotic hypoglycemia, lethargy, vomiting, seizures and coma, which can be fatal in the absence of emergency medical intervention.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=42

Your genetic map

Gene	SNP	Genotype
ACADM	rs77931234	AA
ACADM	rs121434274	GG
ACADM	rs121434277	GG
ACADM	rs121434278	GG
ACADM	rs121434280	TT
ACADM	rs121434281	CC
ACADM	rs398123072	CC
ACADM	rs398123073	TT
ACADM	rs398123074	TT
ACADM	rs148207467	CC
ACADM	rs778906552	GG
ACADM	rs762114560	CC
ACADM	rs745844469	AA
ACADM	rs866388216	GG
ACADM	rs779759347	GG
ACADM	rs150310121	GG
DLSTP1	rs373715782	CC
DLSTP1	rs200724875	GG

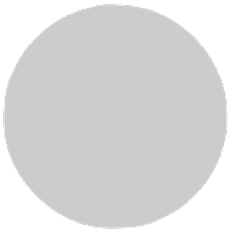
Hereditary Diseases (genetics)

Very long chain acyl-CoA dehydrogenase deficiency

Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency (VLCADD) is an inherited disorder of mitochondrial long-chain fatty acid oxidation with a variable presentation including: cardiomyopathy, hypoketotic hypoglycemia, liver disease, exercise intolerance and rhabdomyolysis.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=26793

Your genetic map

Gene	SNP	Genotype
ACADVL	rs113994167	TT
ACADVL	rs398123092	AA
ACADVL	rs751995154	GG
DLG4	rs369560930	GG
DLG4	rs398123091	GG
DLG4	rs794727773	GG
DLG4	rs545215807	GG
MIR324	rs113690956	GG
MIR324	rs118204014	CC
MIR324	rs118204018	GG
MIR324	rs118204016	GG
MIR324	rs2309689	GG
MIR324	rs113994171	GG
MIR324	rs398123083	GG
MIR324	rs794727113	CC
MIR324	rs112406105	GG
MIR324	rs766742117	CC

Hereditary Diseases (genetics)

Adenylosuccinate lyase deficiency

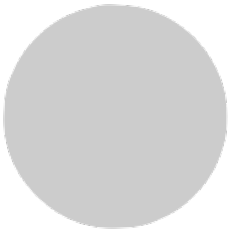
A disorder of purine metabolism characterized by intellectual disability, psychomotor delay and/or regression, seizures, and autistic features.

Your genetic map

Gene	SNP	Genotype
ADSL	rs119450941	GG
ADSL	rs374259530	TT
ADSL	rs756210458	CC
ADSL	rs750614500	CC
ADSL	rs761493155	CC
ADSL	rs763542069	GG
ADSL	rs796052248	CC
ADSL	rs372895468	CC
ADSL	rs776496275	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=46

Hereditary Diseases (genetics)

Alpha-1-antitrypsin deficiency

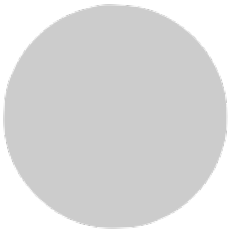
A rare hereditary, metabolic disease characterized by serum levels of alpha-1-antitrypsin (AAT) that are well below the normal range. In the most severe form, the disease can clinically manifest with chronic liver disorders (cirrhosis, fibrosis), respiratory disorders (emphysema, bronchiectasis), and rarely panniculitis or vasculitis.

Your genetic map

Gene	SNP	Genotype
SERPINA1	rs199422209	GG
SERPINA1	rs121912714	TT
SERPINA1	rs199422211	TT
SERPINA1	rs55819880	GG
SERPINA1	rs864622051	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=60

Hereditary Diseases (genetics)

Aromatase deficiency

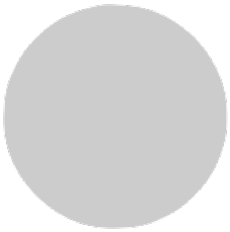
A rare disorder that disrupts the synthesis of estradiol, resulting in hirsutism of mothers during gestation of an affected child; pseudohermaphroditism and virilization in women; and tall stature, osteoporosis and obesity in men.

Your genetic map

Gene	SNP	Genotype
MIR4713	rs121434534	GG
MIR4713	rs121434538	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=91

Hereditary Diseases (genetics)

Beta-ketothiolase deficiency

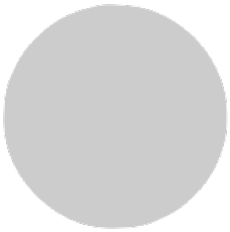
A rare, genetic organic aciduria affecting ketone body metabolism and the catabolism of isoleucine and characterized by intermittent ketoacidotic episodes associated with vomiting, dyspnea, tachypnoea, hypotonia, lethargy and coma, with an onset during infancy and usually ceasing by adolescence.

Your genetic map

Gene	SNP	Genotype
ACAT1	rs120074141	GG
ACAT1	rs145229472	AA
ACAT1	rs120074144	CC
ACAT1	rs120074146	TT
ACAT1	rs148639841	AA
ACAT1	rs398123096	TT
ACAT1	rs727503796	GG
ACAT1	rs762991875	GG
ACAT1	rs199524907	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=134

Hereditary Diseases (genetics)

Beta-ureidopropionase deficiency

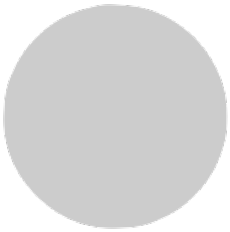
Beta-ureidopropionase deficiency is a very rare pyrimidine metabolism disorder described in fewer than 10 patients to date with an extremely wide clinical picture ranging from asymptomatic cases to neurological (epilepsy, autism) and developmental disorders (urogenital, colorectal).

Your genetic map

Gene	SNP	Genotype
UPB1	rs747539101	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=65287

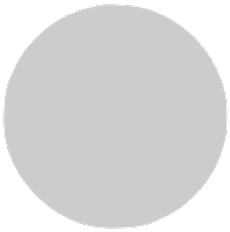
Hereditary Diseases (genetics)

Biotinidase deficiency

A late-onset form of multiple carboxylase deficiency, an inborn error of biotin metabolism that, if untreated, is characterized by seizures, breathing difficulties, hypotonia, skin rash, alopecia, hearing loss and delayed development.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79241

Your genetic map

Gene	SNP	Genotype
BTD	rs104893688	CC
BTD	rs80338686	CC
BTD	rs28934601	AA
BTD	rs80338685	AA
BTD	rs104893686	TT
BTD	rs104893687	CC
BTD	rs397514360	GG
BTD	rs397514363	CC
BTD	rs397514367	GG
BTD	rs397514369	GG
BTD	rs190386869	CC
BTD	rs138818907	CC
BTD	rs146136265	CC
BTD	rs397507174	AA
BTD	rs397507175	GG
BTD	rs146015592	GG
BTD	rs397507170	GG
BTD	rs398123139	GG
BTD	rs587783005	CC

Hereditary Diseases (genetics)

Butyrylcholinesterase deficiency

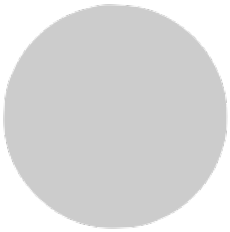
Butyrylcholinesterase (BChE) deficiency is a metabolic disorder characterised by prolonged apnoea after the use of certain anaesthetic drugs, including the muscle relaxants succinylcholine or mivacurium and other ester local anaesthetics. The duration of the prolonged apnoea varies significantly depending on the extent of the enzyme deficiency.

Your genetic map

Gene	SNP	Genotype
BCHE	rs104893684	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=132

Hereditary Diseases (genetics)

Carbamoyl-phosphate synthetase 1 deficiency

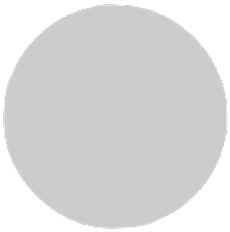
A rare, severe disorder of urea cycle metabolism typically characterized by either a neonatal-onset of severe hyperammonemia that occurs few days after birth and manifests with lethargy, vomiting, hypothermia, seizures, coma and death or a presentation outside the newborn period at any age with (sometimes) milder symptoms of hyperammonemia.

Your genetic map

Gene	SNP	Genotype
CPS1	rs121912592	CC
CPS1	rs121912595	GG
CPS1	rs201716417	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=147

Hereditary Diseases (genetics)

Carnitine palmitoyl transferase 1A deficiency

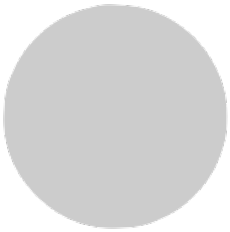
Carnitine palmitoyltransferase 1A (CPT-1A) deficiency is an inborn error of metabolism that affects mitochondrial oxidation of long chain fatty acids (LCFA) in the liver and kidneys, and is characterized by recurrent attacks of fasting-induced hypoketotic hypoglycemia and risk of liver failure.

Your genetic map

Gene	SNP	Genotype
CPT1A	rs80356774	GG
CPT1A	rs80356798	CC
CPT1A	rs80356780	CC
CPT1A	rs80356779	GG
CPT1A	rs398123654	GG
CPT1A	rs191107774	CC
CPT1A	rs189174414	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=156

Hereditary Diseases (genetics)

Carnitine palmitoyltransferase II deficiency

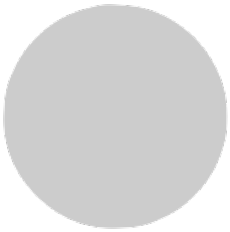
Carnitine palmitoyltransferase II (CPT II) deficiency is an inherited metabolic disorder that affects mitochondrial oxidation of long chain fatty acids (LCFA). Three forms of CPT II deficiency have been described: a myopathic form, a severe infantile form and a neonatal form (see these terms).

Your genetic map

Gene	SNP	Genotype
CPT2	rs28936375	CC
CPT2	rs74315295	TT
CPT2	rs74315296	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=157

Hereditary Diseases (genetics)

Carnitine-acylcarnitine translocase deficiency

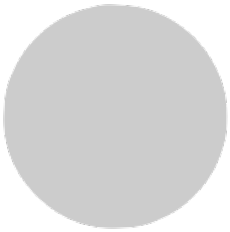
Carnitine-acylcarnitine translocase (CACT) deficiency is a life-threatening, inherited disorder of fatty acid oxidation which usually presents in the neonatal period with severe hypoketotic hypoglycemia, hyperammonemia, cardiomyopathy and/or arrhythmia, hepatic dysfunction, skeletal muscle weakness, and encephalopathy.

Your genetic map

Gene	SNP	Genotype
SLC25A2	rs541208710	AA
SLC25A2	rs756998699	GG
SLC25A2	rs147540030	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=159

Hereditary Diseases (genetics)

Cernunnos-XLF deficiency

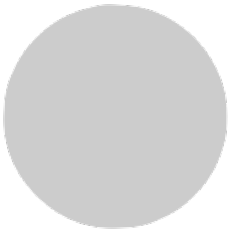
Cernunnos-XLF deficiency is a rare form of combined immunodeficiency characterized by microcephaly, growth retardation, and T and B cell lymphopenia.

Your genetic map

Gene	SNP	Genotype
NHEJ1	rs118204453	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=169079

Hereditary Diseases (genetics)

Fatal infantile cytochrome C oxidase deficiency

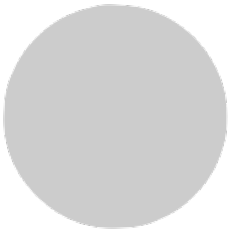
Fatal infantile cytochrome C oxidase deficiency is a very rare mitochondrial disease characterized clinically by cardioencephalomyopathy resulting in death in infancy.

Your genetic map

Gene	SNP	Genotype
COX15	rs28939711	GG
COX15	rs397514662	AA
COX15	rs778412019	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1561

Hereditary Diseases (genetics)

Dihydropyrimidine dehydrogenase deficiency

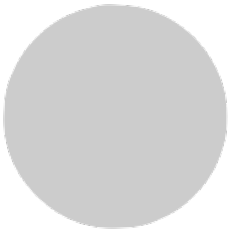
A rare disorder of pyrimidine metabolism characterized by a variable phenotype ranging from absence of symptoms to severe neurological involvement with developmental delay, intellectual disability, and seizures. Additional signs and symptoms may include hypotonia, microcephaly, ocular abnormalities (such as microphthalmia, nystagmus, and strabismus), and autistic behavior, among others. Analysis of urine typically shows high levels of uracil and thymine. Patients are at risk of suffering from severe toxicity after the administration of the anti-neoplastic agent 5-fluorouracil.

Your genetic map

Gene	SNP	Genotype
DPYD	rs72549310	GG
DPYD	rs146170505	CC
DPYD	rs568132506	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1675

Hereditary Diseases (genetics)

Dimethylglycine dehydrogenase deficiency

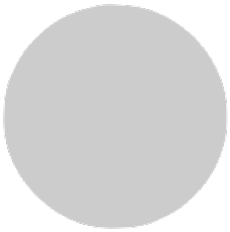
Dimethylglycine dehydrogenase deficiency is an extremely rare autosomal recessive glycine metabolism disorder characterized clinically in the single reported case to date by muscle fatigue and a fish-like odor.

Your genetic map

Gene	SNP	Genotype
DMGDH	rs121908331	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=243343

Hereditary Diseases (genetics)

Dopamine beta-hydroxylase deficiency

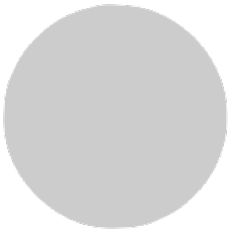
A very rare primary monoamine neurotransmitter synthesis disorder with norepinephrine and adrenaline deficiency that leads to young-onset severe orthostatic hypotension and eyelid ptosis.

Your genetic map

Gene	SNP	Genotype
DBH	rs74853476	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=230

Hereditary Diseases (genetics)

Fructose-1,6-bisphosphatase deficiency

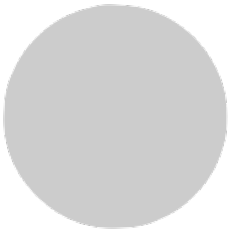
Fructose-1,6-biphosphatase (FBP) deficiency is a disorder of fructose metabolism characterized by recurrent episodes of fasting hypoglycemia with lactic acidosis, that may be life-threatening in neonates and infants.

Your genetic map

Gene	SNP	Genotype
FBP1	rs121918188	CC
FBP1	rs758609113	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=348

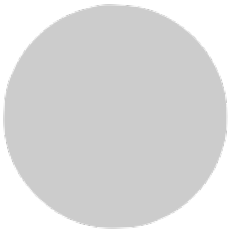
Hereditary Diseases (genetics)

Class I glucose-6-phosphate dehydrogenase deficiency

A rare constitutional hemolytic anemia characterized in symptomatic forms by mild to severe chronic hemolysis, which is further exacerbated by oxidative stress and may lead to chronic non-shperocytic hemolytic anemia of variable severity. Variation in glucose-6-phosphate dehydrogenase levels accounts for differences in sensitivity to oxidants, with chronic hemolysis occurring in association with very low enzyme levels, while the majority of affected individuals remain asymptomatic. The most common clinical manifestations are neonatal jaundice and signs and symptoms of acute hemolysis (such as fatigue, back pain, anemia, and jaundice).

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=466026

Your genetic map

Gene	SNP	Genotype
CASK	rs398122844	TT
G6PD	rs5030869	CC
G6PD	rs137852314	CC
G6PD	rs5030868	GG
G6PD	rs137852315	CC
G6PD	rs137852316	CC
G6PD	rs137852317	CC
G6PD	rs137852319	AA
G6PD	rs137852320	TT
G6PD	rs137852321	CC
G6PD	rs137852322	AA
G6PD	rs137852323	CC
G6PD	rs137852324	CC
G6PD	rs72554665	CC
G6PD	rs137852333	GG
G6PD	rs137852327	CC
G6PD	rs137852329	GG
G6PD	rs137852330	GG
G6PD	rs137852331	TT
G6PD	rs137852332	CC
G6PD	rs137852334	GG
G6PD	rs137852335	CC
G6PD	rs137852336	CC
G6PD	rs137852337	CC
G6PD	rs137852339	CC
G6PD	rs76645461	AA
G6PD	rs78478128	GG
G6PD	rs137852343	AA
G6PD	rs137852344	GG
G6PD	rs137852345	GG
G6PD	rs137852346	CC

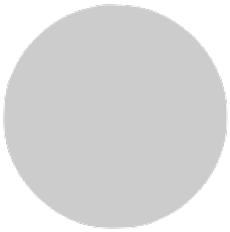
Hereditary Diseases (genetics)

Glutaryl-CoA dehydrogenase deficiency

Glutaryl-CoA dehydrogenase (GCDH) deficiency (GDD) is an autosomal recessive neurometabolic disorder clinically characterized by encephalopathic crises resulting in striatal injury and a severe dystonic dyskinetic movement disorder.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=25

Your genetic map

Gene	SNP	Genotype
GCDH	rs121434366	TT
GCDH	rs121434370	GG
GCDH	rs121434373	GG
GCDH	rs398123195	GG
GCDH	rs142967670	CC
GCDH	rs777201305	GG
GCDH	rs766518430	CC
GCDH	rs786205862	GG
GCDH	rs786205861	CC
GCDH	rs794726972	CC
GCDH	rs149120354	TT
SYCE2	rs121434367	CC
SYCE2	rs121434369	CC
SYCE2	rs121434372	GG
SYCE2	rs147611168	GG
SYCE2	rs141437721	AA
SYCE2	rs372983141	GG
SYCE2	rs199999619	AA

Hereditary Diseases (genetics)

Glutathione synthetase deficiency

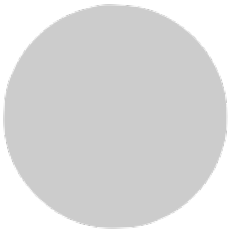
A rare disorder characterised by hemolytic anemia, associated with metabolic acidosis and 5-oxoprolinuria in moderate forms, and with progressive neurological symptoms and recurrent bacterial infections in the most severe forms.

Your genetic map

Gene	SNP	Genotype
GSS	rs28938472	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=32

Hereditary Diseases (genetics)

Guanidinoacetate methyltransferase deficiency

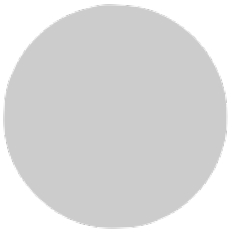
Guanidinoacetate methyltransferase (GAMT) deficiency is a creatine deficiency syndrome characterized by global developmental delay/intellectual disability (DD/ID), prominent speech delay, autistic/hyperactive behavioral disorders, seizures, and various types of pyramidal and/or extra-pyramidal manifestations.

Your genetic map

Gene	SNP	Genotype
GAMT	rs80338735	CC
GAMT	rs370421531	CC
GAMT	rs753198836	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=382

Hereditary Diseases (genetics)

Holocarboxylase synthetase deficiency

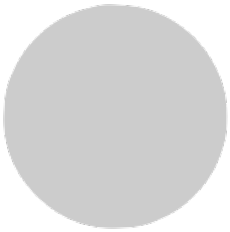
A rare, early-onset and life-threatening, multiple carboxylase deficiency that when left untreated, is characterized by vomiting, tachypnea, irritability, lethargy, exfoliative dermatitis, and seizures that can worsen to coma and death.

Your genetic map

Gene	SNP	Genotype
HLCS	rs119103227	AA
HLCS	rs119103229	GG
HLCS	rs119103230	CC
HLCS	rs119103231	CC
HLCS	rs753887925	CC
HLCS	rs146448211	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79242

Hereditary Diseases (genetics)

LCAT deficiency

LCAT (lecithin-cholesterol acyltransferase) deficiency is a rare lipoprotein metabolism disorder characterized clinically by corneal opacities, and sometimes renal failure and hemolytic anemia, and biochemically by severely reduced HDL cholesterol.

Your genetic map

Gene	SNP	Genotype
LCAT	rs121908050	GG

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=650

Hereditary Diseases (genetics)

Lysosomal acid lipase deficiency

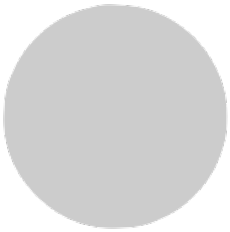
A rare, progressive metabolic liver disease due to marked to complete lysosomal acid lipase deficiency and characterized by dyslipidemia and massive lipid accumulation leading to hepatomegaly and liver dysfunction, splenomegaly, accelerated atherosclerosis.

Your genetic map

Gene	SNP	Genotype
LIPA	rs121965086	AA
LIPA	rs116928232	CC
LIPA	rs797045094	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=275761

Hereditary Diseases (genetics)

Lipoyl transferase 1 deficiency

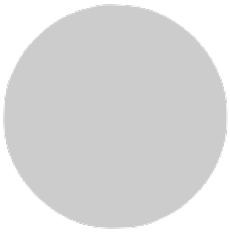
Lipoyl transferase 1 deficiency is a very rare inborn error of metabolism disorder, with a highly variable phenotype, typically characterized by neonatal to infancy-onset of seizures, psychomotor delay, and abnormal muscle tone that may include hypo- and/or hypertonia, resulting in generalized weakness, dystonic movements, and/or progressive respiratory distress, associated with severe lactic acidosis and elevated lactate, ketoglutarate and 2-oxoacids in urine. Additional manifestations may include dehydration, vomiting, signs of liver dysfunction, extrapyramidal signs, spastic tetraparesis, brisk deep tendon reflexes, speech impairment, swallowing difficulties, and pulmonary hypertension.

Your genetic map

Gene	SNP	Genotype
MITD1	rs137891647	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=401862

Hereditary Diseases (genetics)

Homocystinuria without methylmalonic aciduria

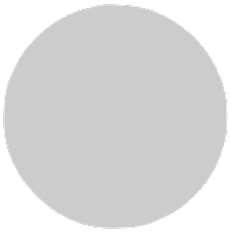
Homocystinuria without methylmalonic aciduria is an inborn error of vitamin B12 (cobalamin) metabolism characterized by megaloblastic anemia, encephalopathy and, sometimes, developmental delay, and associated with homocystinuria and hyperhomocysteinemia. There are three types of homocystinuria without methylmalonic aciduria; cblE, cblG and cblD-variant 1 (cblDv1).

Your genetic map

Gene	SNP	Genotype
MTR	rs121913578	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2170

Hereditary Diseases (genetics)

Myeloperoxidase deficiency

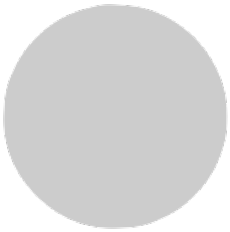
A rare primary immunodeficiency due to a defect in innate immunity characterized by a marked decrease or absence of myeloperoxidase activity in neutrophils and monocytes. Clinically, most patients are asymptomatic. Occasionally, severe infectious complications may occur, particularly recurrent candida infections, being especially severe in the setting of comorbid diabetes mellitus.

Your genetic map

Gene	SNP	Genotype
MPO	rs119468010	GG
MPO	rs778013714	CC
MPO	rs762526880	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2587

Hereditary Diseases (genetics)

Monoamine oxidase A deficiency

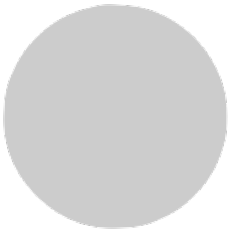
Monoamine oxidase-A deficiency is a very rare recessive X-linked biogenic amine metabolism disorder characterized clinically by mild intellectual deficit, impulsive aggressiveness, and sometimes violent behavior and presenting from childhood.

Your genetic map

Gene	SNP	Genotype
MAOA	rs796065312	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3057

Hereditary Diseases (genetics)

Alpha-N-acetylgalactosaminidase deficiency

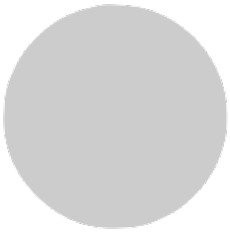
A very rare lysosomal storage disease that is clinically and pathologically heterogeneous and is characterized by deficient NAGA activity.

Your genetic map

Gene	SNP	Genotype
LOC1079	rs779423223	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3137

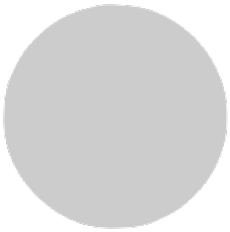
Hereditary Diseases (genetics)

Ornithine transcarbamylase deficiency

A rare, genetic disorder of urea cycle metabolism and ammonia detoxification characterized by either a severe, neonatal-onset disease found mainly in males, or later-onset (partial) forms of the disease. Both present with episodes of hyperammonemia that can be fatal and which can lead to neurological sequelae.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=664

Your genetic map

Gene	SNP	Genotype
OTC	rs68026851	GG
OTC	rs67960011	CC
OTC	rs72556267	GG
OTC	rs68031618	GG
OTC	rs72558454	CC
OTC	rs67120076	CC
OTC	rs66626662	GG
OTC	rs72558465	GG
OTC	rs66656800	GG
OTC	rs72558412	TT
OTC	rs72554307	CC
OTC	rs72554308	GG
OTC	rs72558495	TT
OTC	rs74518351	AA
OTC	rs72554310	CC
OTC	rs66521141	GG
OTC	rs66677059	TT
OTC	rs72554326	CC
OTC	rs67418243	CC
OTC	rs66550389	GG
OTC	rs68058881	GG
OTC	rs72552295	TT
OTC	rs72556257	AA
OTC	rs72556260	GG
OTC	rs72556271	AA
OTC	rs72556274	CC
OTC	rs72556275	GG
OTC	rs66867430	AA
OTC	rs72556277	CC
OTC	rs72556278	CC
OTC	rs72556284	CC

Hereditary Diseases (genetics)

Pyruvate carboxylase deficiency, benign type

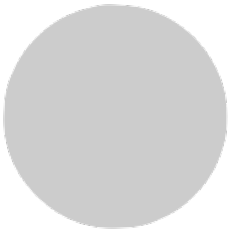
Benign pyruvate carboxylase (PC) deficiency (Type C) is a rare, very mild form of PC deficiency characterized by episodic metabolic acidosis and normal or mildly delayed neurological development.

Your genetic map

Gene	SNP	Genotype
PC	rs796052029	CC
PC	rs113994142	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=353320

Hereditary Diseases (genetics)

Pyruvate dehydrogenase deficiency

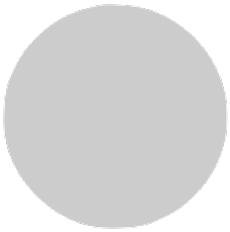
Pyruvate dehydrogenase deficiency (PDHD) is a rare neurometabolic disorder characterized by a wide range of clinical signs with metabolic and neurological components of varying severity. Manifestations range from often fatal, severe, neonatal lactic acidosis to later-onset neurological disorders. Six subtypes related to the affected subunit of the PDH complex have been recognized with significant clinical overlap: PDHD due to E1-alpha, E1-beta, E2 and E3 deficiency, PDHD due to E3-binding protein deficiency, and PDH phosphatase deficiency (see these terms).

Your genetic map

Gene	SNP	Genotype
DLAT	rs797044957	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=765

Hereditary Diseases (genetics)

Prolidase deficiency

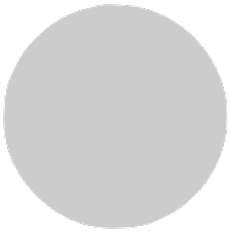
Prolidase deficiency is an inherited disorder of peptide metabolism characterized by severe skin lesions, recurrent infections (involving mainly the skin and respiratory system), dysmorphic facial features, variable cognitive impairment, and splenomegaly.

Your genetic map

Gene	SNP	Genotype
PEPD	rs121917723	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=742

Hereditary Diseases (genetics)

Mitochondrial trifunctional protein deficiency

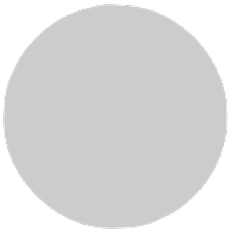
A rare disorder of fatty acid oxidation characterized by a wide clinical spectrum ranging from severe neonatal manifestations including cardiomyopathy, hypoglycemia, metabolic acidosis, skeletal myopathy and neuropathy, liver disease and death to a mild phenotype with peripheral polyneuropathy, episodic rhabdomyolysis and pigmentary retinopathy.

Your genetic map

Gene	SNP	Genotype
HADHA	rs137852770	GG
HADHA	rs781222705	TT
HADHA	rs137852774	AA
HADHA	rs147103714	GG
HADHB	rs121913132	GG
HADHB	rs121913133	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=746

Hereditary Diseases (genetics)

Pterin-4 alpha-carbinolamine dehydratase deficiency

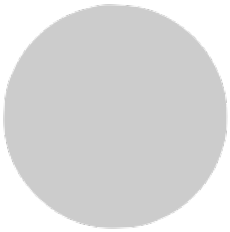
Dehydratase deficiency or pterin-4 alpha-carbinolamine dehydratase (PCD) is considered a transient and benign form of hyperphenylalaninemia due to tetrahydrobiopterin deficiency (see this term), characterized by muscular hypotonia, irritability (detected by EEG), slow acquisition of psychomotor skills, age-dependent movement disorders, including dystonia and an accompanying excretion of 7-substituted pterins. Neurological developement is normal with dietary control of blood phenyalanine. PCD is inherited in an autosomal recessive manner.

Your genetic map

Gene	SNP	Genotype
PCBD1	rs104894172	CC
PCBD1	rs121913015	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1578

Hereditary Diseases (genetics)

Purine nucleoside phosphorylase deficiency

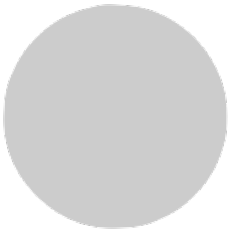
A rare immune disease characterized by progressive immunodeficiency leading to recurrent and opportunistic infections, autoimmunity and malignancy as well as neurologic manifestations.

Your genetic map

Gene	SNP	Genotype
PNP	rs104894451	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=760

Hereditary Diseases (genetics)

S-adenosylhomocysteine hydrolase deficiency

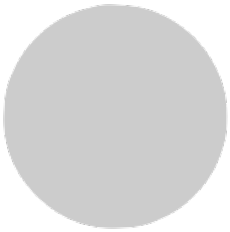
A rare, multisystemic inherited metabolic diseases characterized clinically, by a variable spectrum of severity, primarily comprised of psychomotor delay, myopathy and liver dysfunction. Most patients present in infancy, but the onset can be already in utero or in adult age. Hypermethioninemia is frequent, but often absent in infancy. Creatine kinase is elevated in most patients.

Your genetic map

Gene	SNP	Genotype
AHCY	rs121918608	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=88618

Hereditary Diseases (genetics)

Succinyl-CoA:3-oxoacid CoA transferase deficiency

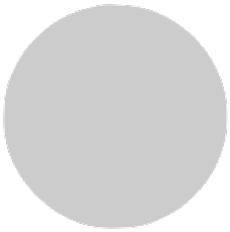
A rare, genetic disorder in ketone body utilization characterized by severe, potentially fatal intermittent episodes of ketoacidosis.

Your genetic map

Gene	SNP	Genotype
OXCT1	rs121909301	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=832

Hereditary Diseases (genetics)

Familial glucocorticoid deficiency

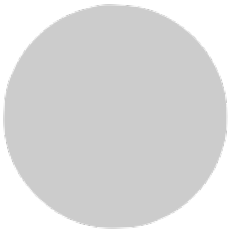
Familial glucocorticoid deficiency (FGD) is a group of primary adrenal insufficiencies characterized clinically by neonatal hyperpigmentation, hypoglycemia, failure to thrive, and recurrent infections, and biochemically by glucocorticoid deficiency without mineralocorticoid deficiency.

Your genetic map

Gene	SNP	Genotype
MC2R	rs104894658	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=361

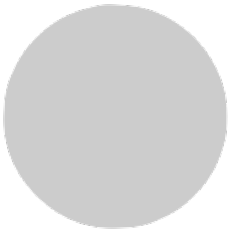
Hereditary Diseases (genetics)

Multiple acyl-CoA dehydrogenase deficiency

Multiple acyl-CoA dehydrogenation deficiency (MADD) is a disorder of fatty acid and amino acid oxidation and is a clinically heterogeneous disorder ranging from a severe neonatal presentation with metabolic acidosis, cardiomyopathy and liver disease, to a mild childhood/adult disease with episodic metabolic decompensation, muscle weakness, and respiratory failure.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=394532

Your genetic map

Gene	SNP	Genotype
ETFA	rs119458969	AA
ETFA	rs199763682	GG
ETFDH	rs121964954	GG
ETFDH	rs121964955	GG
ETFDH	rs387907170	TT
ETFDH	rs377656387	CC
ETFDH	rs398124151	GG
ETFDH	rs398124152	CC
ETFDH	rs377686388	TT
ETFDH	rs796051965	AA
ETFDH	rs796051959	GG
ETFDH	rs558005496	GG
ETFDH	rs863224869	TT
ETFDH	rs200920510	CC
FLAD1	rs771466122	CC
FLAD1	rs199979286	CC

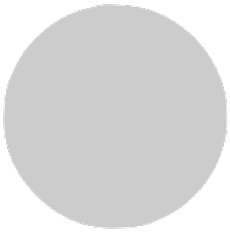
Hereditary Diseases (genetics)

Systemic primary carnitine deficiency

A disorder of carnitine cycle and carnitine transport that is characterized classically by early childhood onset cardiomyopathy often with weakness and hypotonia, failure to thrive and recurrent hypoglycemic hypoketotic seizures and/or coma.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=158

Your genetic map

Gene	SNP	Genotype
MIR3936	rs267607052	GG
MIR3936	rs11568520	CC
MIR3936	rs72552725	AA
MIR3936	rs202088921	CC
SLC22A5	rs72552727	GG
SLC22A5	rs121908886	CC
SLC22A5	rs121908888	AA
SLC22A5	rs121908889	GG
SLC22A5	rs121908890	CC
SLC22A5	rs267607054	CC
SLC22A5	rs151231558	GG
SLC22A5	rs114269482	CC
SLC22A5	rs386134208	CC
SLC22A5	rs386134210	GG
SLC22A5	rs386134212	CC
SLC22A5	rs144547521	CC
SLC22A5	rs72552732	CC
SLC22A5	rs60376624	CC
SLC22A5	rs386134223	GG
SLC22A5	rs377724489	AA
SLC22A5	rs796052039	GG
SLC22A5	rs777004046	AA
SLC22A5	rs185551386	GG

Hereditary Diseases (genetics)

Combined pituitary hormone deficiencies, genetic forms

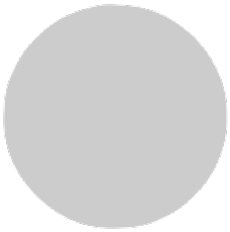
Congenital hypopituitarism is characterized by multiple pituitary hormone deficiency, including somatotroph, thyrotroph, lactotroph, corticotroph or gonadotroph deficiencies, due to mutations of pituitary transcription factors involved in pituitary ontogenesis. Congenital hypopituitarism is rare compared with the high incidence of hypopituitarism induced by pituitary adenomas, transsphenoidal surgery or radiotherapy.

Your genetic map

Gene	SNP	Genotype
POU1F1	rs104893764	CC
POU1F1	rs104893765	CC
PROP1	rs140016178	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=95494

Hereditary Diseases (genetics)

Infantile cerebellar-retinal degeneration

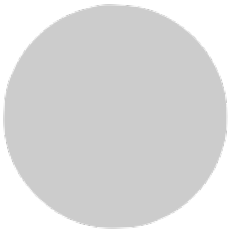
Infantile cerebellar-retinal degeneration is a rare, neurodegenerative disorder characterized by an early onset of truncal hypotonia, variable forms of seizures, athetosis, severe global developmental delay, intellectual disability and various ophthalmologic abnormalities, including strabismus, nystagmus, optic atrophy and retinal degeneration.

Your genetic map

Gene	SNP	Genotype
POLR3H	rs375761361	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=313850

Hereditary Diseases (genetics)

Brain demyelination due to methionine adenosyltransferase deficiency

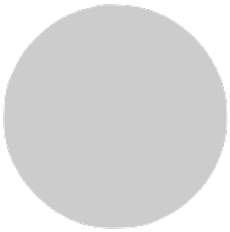
Hypermethioninemia due to methionine adenosyltransferase deficiency is a very rare metabolic disorder resulting in isolated hepatic hypermethioninemia that is usually benign due to partial inactivation of enzyme activity. Rarely patients have been found to have an odd odor or neurological disorders such as brain demyelination.

Your genetic map

Gene	SNP	Genotype
MAT1A	rs118204001	AA
MAT1A	rs118204003	GG
MAT1A	rs116659053	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=168598

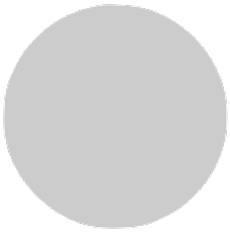
Hereditary Diseases (genetics)

Desminopathy

A rare genetic skeletal muscle disease characterized by abnormal chimeric aggregates of desmin and other cytoskeletal proteins and granulofilamentous material at the ultrastructural level in muscle biopsies and variable clinical myopathological features, age of disease onset and rate of disease progression. Patients present with bilateral skeletal muscle weakness that starts in distal leg muscles and spreads proximally, sometimes involving trunk, neck flexors and facial muscles and often cardiomyopathy manifested by conduction blocks, arrhythmias, chronic heart failure, and sometimes tachyarrhythmia. Weakness eventually leads to wheelchair dependence. Respiratory insufficiency can be a major cause of disability and death, beginning with nocturnal hypoventilation with oxygen desaturation and progressing to daytime respiratory failure.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=98909

Your genetic map

Gene	SNP	Genotype
DES	rs57639980	TT
DES	rs121913003	CC
DES	rs121913005	CC
DES	rs62635763	CC
DES	rs397516698	GG
DES	rs267607482	AA
DES	rs59308628	TT
DES	rs57694264	GG
DES	rs61726467	GG
DES	rs267607485	AA
DES	rs267607499	AA
DES	rs267607495	CC
DES	rs267607483	AA
DES	rs150974575	CC
DES	rs781590560	CC

Hereditary Diseases (genetics)

Desmosterolosis

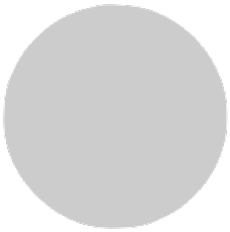
Desmosterolosis is a very rare sterol biosynthesis disorder characterized by multiple congenital anomalies, failure to thrive, and intellectual disability, with elevated levels of desmosterol.

Your genetic map

Gene	SNP	Genotype
DHCR24	rs119475041	CC
DHCR24	rs387906939	CC
DHCR24	rs387906940	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=35107

Hereditary Diseases (genetics)

Maternally-inherited diabetes and deafness

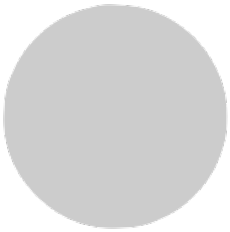
Maternally inherited diabetes and deafness (MIDD) is a mitochondrial disorder characterized by maternally transmitted diabetes and sensorineural deafness.

Your genetic map

Gene	SNP	Genotype
MT TE	rs121434453	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=225

Hereditary Diseases (genetics)

Nephrogenic diabetes insipidus

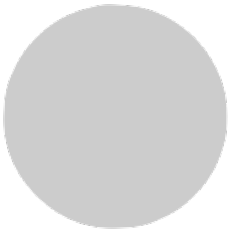
A rare, genetic renal tubular disease that is characterized by polyuria with polydipsia, recurrent bouts of fever, constipation, and acute hypernatremic dehydration after birth that may cause neurological sequelae.

Your genetic map

Gene	SNP	Genotype
AQP2	rs28931580	AA
AQP5 AS1	rs104894328	CC
AQP5 AS1	rs104894326	GG
AQP5 AS1	rs104894334	GG
AQP5 AS1	rs104894338	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=223

Hereditary Diseases (genetics)

Congenital chloride diarrhea

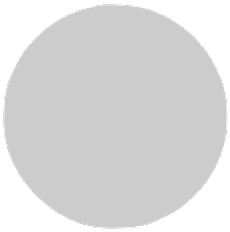
A rare genetic intestinal disease characterized by persistent, potentially life-threatening, watery diarrhea with excessive levels of chloride in stools, hypochloremia, hyponatremia, hypokalemia, and metabolic alkalosis, resulting in chronic dehydration and failure to thrive. Antenatal ultrasound typically reveals polyhydramnios and significant dilatation of the fetal intestinal loops.

Your genetic map

Gene	SNP	Genotype
SLC26A3	rs386833471	CC
SLC26A3	rs386833479	CC
SLC26A3	rs386833480	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=53689

Hereditary Diseases (genetics)

Congenital sodium diarrhea

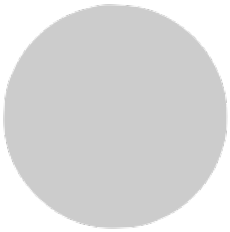
A rare, genetic, non-syndromic intestinal transport defect characterized by congenital onset of severe watery diarrhea containing high concentrations of sodium, hyponatremia and metabolic acidosis.

Your genetic map

Gene	SNP	Genotype
SPINT2	rs121908403	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=103908

Hereditary Diseases (genetics)

Syndromic diarrhea

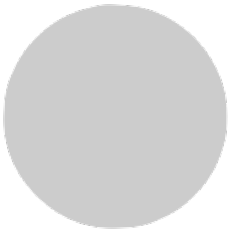
A rare gastroenterologic disease manifesting as intractable diarrhea in the first month of life with failure to thrive and associated with facial dysmorphism, hair abnormalities, and, in some cases, immune disorders and intrauterine growth restriction.

Your genetic map

Gene	SNP	Genotype
SKIC3	rs534237033	CC
SKIC3	rs140800288	GG
SKIC3	rs200085753	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=84064

Hereditary Diseases (genetics)

Dihydropyrimidinuria

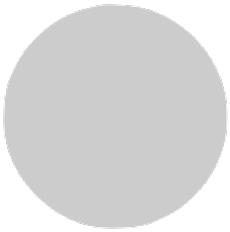
Dihydropyrimidinase (DPD) deficiency is a very rare pyrimidine metabolism disorder with a variable clinical presentation including gastrointestinal manifestations (feeding problems, cyclic vomiting, gastroesophageal reflux, malabsorption with villous atrophy), hypotonia, intellectual deficit, seizures, and less frequently growth retardation, failure to thrive, microcephaly and autism. Asymptomatic cases are also reported. DPD deficiency increases the risk of 5-FU toxicity.

Your genetic map

Gene	SNP	Genotype
DPYS	rs61758444	GG
DPYS	rs201280871	GG
DPYS	rs142574766	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=38874

Hereditary Diseases (genetics)

Familial dysautonomia

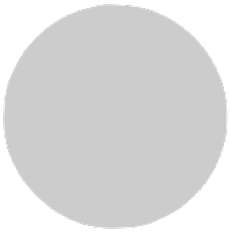
A rare hereditary sensory and autonomic neuropathy characterized by decreased pain and temperature perception, absent deep tendon reflexes, proprioceptive ataxia, afferent baroreflex failure and progressive optic neuropathy.

Your genetic map

Gene	SNP	Genotype
ELP1	rs111033171	AA
ELP1	rs137853022	CC
ELP1	rs28939712	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1764

Hereditary Diseases (genetics)

Severe intellectual disability and progressive spastic paraplegia

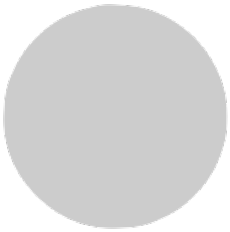
Severe intellectual disability and progressive spastic paraplegia is a rare complex spastic paraplegia characterized by an early onset hypotonia that progresses to spasticity, global developmental delay, severe intellectual disability and speech impairment, microcephaly, short stature and dysmorphic features. Patients often become non-ambulatory, and some develop seizures and stereotypic laughter.

Your genetic map

Gene	SNP	Genotype
AP4S1	rs200440467	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=280763

Hereditary Diseases (genetics)

Syndromic X-linked intellectual disability due to JARID1C mutation

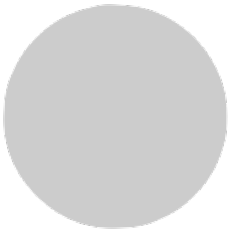
Syndromic X-linked intellectual disability due to JARID1C mutation is characterised by mild to severe intellectual deficit associated with variable clinical manifestations including spasticity, cryptorchidism, maxillary hypoplasia, alopecia areata, epilepsy, short stature, impaired speech and behavioural problems. To date, it has been described in less than 15 families. Transmission is X-linked recessive and the syndrome is caused by mutations in the JARID1C (SMCX) gene encoding a JmjC-domain protein with histone demethylase activity.

Your genetic map

Gene	SNP	Genotype
KDM5C	rs199422235	CC
KDM5C	rs587780372	GG
MIR6895	rs782246658	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=85279

Hereditary Diseases (genetics)

X-linked intellectual disability, Cabezas type

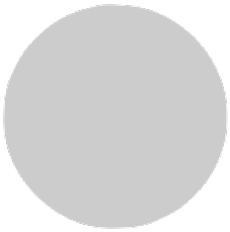
An X-linked syndromic intellectual disability characterized by developmental delay, intellectual disability (ID) with severe speech impairment, and short stature. Variable additional clinical features have been associated, including behavioral disturbances, gait abnormalities, tremor, seizures, hypogonadism, truncal obesity, unspecific facial dysmorphism, and small hands and feet.

Your genetic map

Gene	SNP	Genotype
CUL4B	rs121434616	GG
CUL4B	rs797044862	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=85293

Hereditary Diseases (genetics)

X-linked intellectual disability, Snyder type

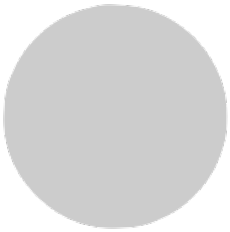
X-linked intellectual disability, Snyder type is a rare X-linked intellectual disability syndrome characterized by hypotonia, asthenic build with diminished muscle mass, severe generalized psychomotor delay, unsteady gait and moderate to severe intellectual disability, as well as a long, thin, asymmetrical face with prominent lower lip, long fingers and toes and nasal, dysarthric or absent speech. Bone abnormalities (e.g., osteoporosis, kyphoscoliosis, fractures, joint contractures) are also characteristic. Myoclonic, or myoclonic-like, seizures and renal abnormalities have been associated in some patients.

Your genetic map

Gene	SNP	Genotype
SMS	rs121434610	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3063

Hereditary Diseases (genetics)

X-linked intellectual disability, Najm type

Najm type X-linked intellectual deficit is a rare cerebellar dysgenesis syndrome characterized by variable clinical manifestations ranging from mild intellectual deficit with or without congenital nystagmus, to severe cognitive impairment associated with cerebellar and pontine hypoplasia/atrophy and abnormalities of cortical development.

Your genetic map

Gene	SNP	Genotype
CASK	rs137852815	GG
CASK	rs387906705	GG
CASK	rs587783360	GG
CASK	rs587783361	GG
CASK	rs587783364	GG
CASK	rs587783366	TT
CASK	rs587783368	CC
CASK	rs587783369	CC
CASK	rs587783371	GG
CASK	rs794727270	GG
CASK	rs749742837	GG
CASK	rs863224854	TT

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=163937

Hereditary Diseases (genetics)

2q23.1 microdeletion syndrome

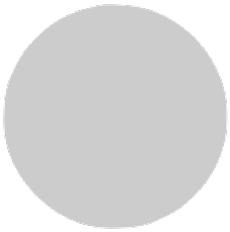
The newly described 2q23.1 microdeletion syndrome includes severe intellectual deficit with pronounced speech delay, behavioral abnormalities including hyperactivity and inappropriate laughter, short stature and seizures.

Your genetic map

Gene	SNP	Genotype
MBD5	rs886041003	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=178469

Hereditary Diseases (genetics)

Intellectual disability, Birk-Barel type

Intellectual disability, Birk-Barel type is a rare, genetic, syndromic intellectual disability characterized by congenital central hypotonia, developmental delay, moderate to severe intellectual disability and subtle dysmorphic features which evolve over time (dolichocephaly, myopathic facies, ptosis, short and broad philtrum, tented upper lip vermillion, palatal anomalies, mild micro- and/or retrognathia). Patients present reduced facial movements, lethargy, weak cry, transient neonatal hypoglycemia, severe feeding difficulties and failure to thrive. Dysphagia, particularly of solid food, asthenic body build, joint contractures and scoliosis are additional features.

Your genetic map

Gene	SNP	Genotype
KCNK9	rs121908332	CC

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=166108

Hereditary Diseases (genetics)

Familial dyskinesia and facial myokymia

Familial dyskinesia and facial myokymia is a rare paroxysmal movement disorder, with childhood or adolescent onset, characterized by paroxysmal choreiform, dystonic, and myoclonic movements involving the limbs (mostly distal upper limbs), neck and/or face, which can progressively increase in both frequency and severity until they become nearly constant. Patients may also present with delayed motor milestones, perioral and periorbital dyskinesias, dysarthria, hypotonia, and weakness.

Your genetic map

Gene	SNP	Genotype
ADCY5	rs796065306	CC

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=324588

Hereditary Diseases (genetics)

Paroxysmal exertion-induced dyskinesia

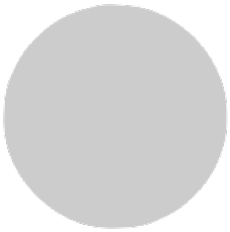
Paroxysmal exertion-induced dyskinesia (PED) is a form of paroxysmal dyskinesia (see this term), characterized by painless attacks of dystonia of the extremities triggered by prolonged physical activities.

Your genetic map

Gene	SNP	Genotype
SLC2A1	rs121909739	CC
SLC2A1	rs121909740	CC
SLC2A1	rs267607061	GG
SLC2A1	rs202060209	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=98811

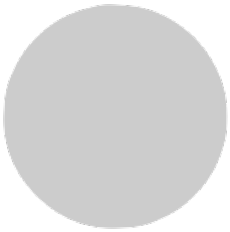
Hereditary Diseases (genetics)

Familial aortic dissection

Familial aortic dissection is the term used to describe rupture of the aortic wall at the level of the media, resulting in the formation of a false channel and deviation of part of the aortic flux. Familial predisposition to thoracic aortic aneurysms and type A dissections (concerning the ascending aorta and/or the aortic arch) has been demonstrated in around 19% of patients presenting with thoracic aortic dissections and several loci have been identified so far (16p12.2-p13.13, 3p24-25). This predisposition is transmitted in an autosomal dominant manner.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=229

Your genetic map

Gene	SNP	Genotype
ACTA2	rs869025352	AA
ACTA2	rs397516685	CC
COL3A1	rs869312034	GG
COL3A1	rs587779433	GG
COL3A1	rs587779458	GG
COL3A1	rs587779685	GG
COL3A1	rs794728057	CC
COL3A1	rs1393544920	CC
FBN1	rs137854460	CC
FBN1	rs137854477	CC
FBN1	rs267606800	CC
FBN1	rs193922183	AA
FBN1	rs112660651	CC
FBN1	rs193922209	CC
FBN1	rs149062442	CC
FBN1	rs193922243	CC
FBN1	rs397515764	CC
FBN1	rs397515773	AA
FBN1	rs397515775	CC
FBN1	rs397515784	GG
FBN1	rs397515789	CC
FBN1	rs397515827	CC
FBN1	rs397515828	CC
FBN1	rs587782947	CC
FBN1	rs794728283	GG
FBN1	rs794728281	CC
FBN1	rs112118237	CC
FBN1	rs794728256	CC
FBN1	rs794728253	AA
FBN1	rs794728247	CC
FBN1	rs794728241	CC

Hereditary Diseases (genetics)

Cortical dysgenesis with pontocerebellar hypoplasia due to TUBB3 mutation

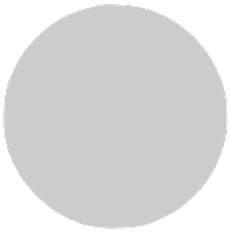
A rare, genetic, non-syndromic cerebral malformation due to abnormal neuronal migration disease characterized by the association of cortical dysplasia and pontocerebellar hypoplasia, manifesting with global developmental delay, mild to severe intellectual disability, axial hypotonia, strabismus, nystagmus and, occasionally, optic nerve hypoplasia. Brain imaging reveals variable malformations, including frontally predominant microgyria, gyral disorganization and simplification, dysmorphic and hypertrophic basal ganglia, cerebellar vermis dysplasia, brainstem/corpus callosum hypoplasia, and/or olfactory bulbs agenesis.

Your genetic map

Gene	SNP	Genotype
TUBB3	rs747480526	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=300570

Hereditary Diseases (genetics)

X-linked complicated corpus callosum dysgenesis

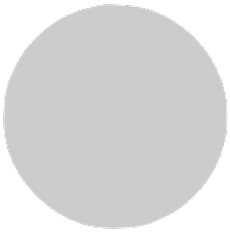
A congenital, X-linked, clinical subtype of L1 syndrome, characterized by variable spastic paraplegia, mild to moderate intellectual disability, and dysplasia, hypoplasia or aplasia of the corpus callosum. In this subtype hydrocephalus, adducted thumbs, or absent speech are not observed.

Your genetic map

Gene	SNP	Genotype
L1CAM	rs797045673	GG
L1CAM	rs367665974	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1497

Hereditary Diseases (genetics)

Postaxial acrofacial dysostosis

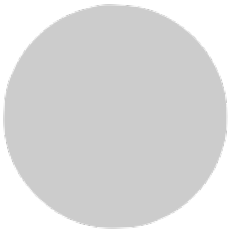
A rare acrofacial dysostosis that is characterized by mandibular and malar hypoplasia, small and cup-shaped ears, lower lid ectropion, and symmetrical postaxial limb deficiencies with absence of the fifth digital rays and ulnar hypoplasia.

Your genetic map

Gene	SNP	Genotype
DHODH	rs201947120	CC
DHODH	rs201230446	CC

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=246

Hereditary Diseases (genetics)

Acromicric dysplasia

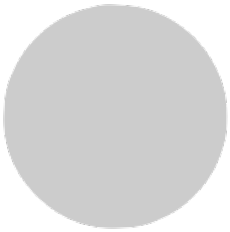
A rare bone dysplasia characterized by short stature, short hands and feet, mild facial dysmorphism, and characteristic X-ray abnormalities of the hands.

Your genetic map

Gene	SNP	Genotype
FBN1	rs1131692052	AA
FBN1	rs387906626	TT
FBN1	rs1064797059	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=969

Hereditary Diseases (genetics)

Cerebrofaciothoracic dysplasia

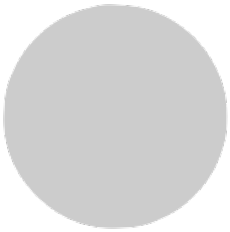
Cerebro-facio-thoracic dysplasia or Pascual-Castroviejo syndrome type 1 is a rare syndrome characterized by facial dysmorphism, intellectual deficit and costovertebral abnormalities.

Your genetic map

Gene	SNP	Genotype
TMCO1	rs372701032	CC
TMCO1	rs765824628	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1394

Hereditary Diseases (genetics)

FGFR2-related bent bone dysplasia

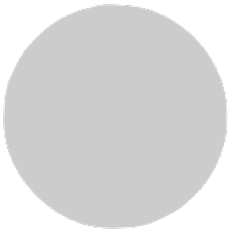
FGFR2-related bent bone dysplasia is a rare, genetic, lethal, primary bone dysplasia characterized by dysmorphic craniofacial features (low-set, posteriorly rotated ears, hypertelorism, megalophtalmos, flattened and hypoplastic midface, micrognathia), hypomineralization of the calvarium, craniosynostosis, hypoplastic clavicles and pubis, and bent long bones (particularly involving the femora), caused by germline mutations in the FGFR2 gene. Prematurely erupted fetal teeth, osteopenia, hirsutism, clitoromegaly, gingival hyperplasia, and hepatosplenomegaly with extramedullary hematopoesis may also be associated.

Your genetic map

Gene	SNP	Genotype
FGFR2	rs387906678	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=313855

Hereditary Diseases (genetics)

Craniofrontonasal dysplasia

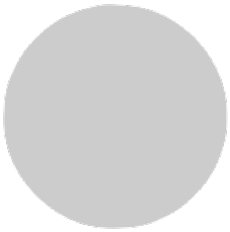
A rare X-linked malformation syndrome characterized by craniofacial abnormalities, grooved nails, intellectual disability and various skeletal and soft tissue abnormalities.

Your genetic map

Gene	SNP	Genotype
EFNB1	rs104894801	CC
EFNB1	rs104894804	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1520

Hereditary Diseases (genetics)

Non-epidermolytic palmoplantar keratoderma

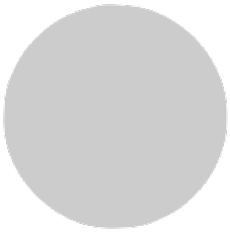
Kniest dysplasia is a severe type II collagenopathy characterized by a short trunk and limbs, prominent joints and midface hypoplasia (round face with a flat nasal root).

Your genetic map

Gene	SNP	Genotype
COL2A1	rs121912877	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=485

Hereditary Diseases (genetics)

Singleton-Merten dysplasia

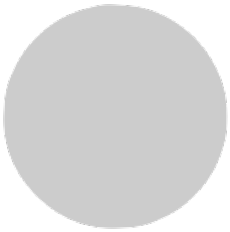
Singleton-Merten dysplasia is characterized by dental dysplasia, progressive calcification of the thoracic aorta with stenosis, osteoporosis and expansion of the marrow cavities in hand bones. Additional features included generalized muscle weakness and atrophy, and chronic psoriasiform skin eruptions. It has been reported in four unrelated patients (male and female) and in a family with multiple affected members (male).

Your genetic map

Gene	SNP	Genotype
IFIH1	rs376048533	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=85191

Hereditary Diseases (genetics)

Diastrophic dysplasia

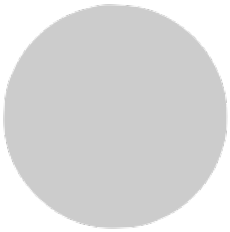
A rare disorder marked by short stature with short extremities (final adult height is 120cm +/- 10cm), and joint malformations leading to multiple joint contractures (principally involving the shoulders, elbows, interphalangeal joints and hips).

Your genetic map

Gene	SNP	Genotype
SLC26A2	rs104893919	CC
SLC26A2	rs386833492	TT
SLC26A2	rs386833493	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=628

Hereditary Diseases (genetics)

Hidrotic ectodermal dysplasia

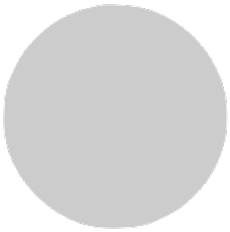
Clouston syndrome (or hidrotic ectodermal dysplasia) is characterised by the clinical triad of nail dystrophy, alopecia, and palmoplantar hyperkeratosis.

Your genetic map

Gene	SNP	Genotype
GJB6	rs104894415	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=189

Hereditary Diseases (genetics)

Hypohidrotic ectodermal dysplasia

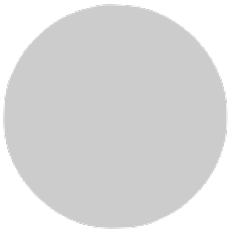
A rare genetic ectodermal dysplasia syndrome characterized by sparse hair, abnormal or missing teeth, decrease or absent sudation and typical facial features.

Your genetic map

Gene	SNP	Genotype
EDAR	rs121908452	GG
EDAR	rs121908453	CC
EDAR	rs747806672	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=238468

Hereditary Diseases (genetics)

Multiple epiphyseal dysplasia, Beighton type

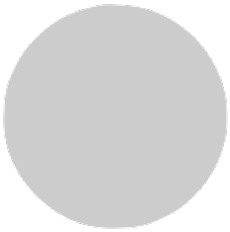
A rare primary bone dysplasia characterized by the association of multiple epiphyseal dysplasia, visual impairment (with early-onset progressive myopia, retinal thinning, and cataracts), and conductive hearing loss. Patients are of short stature and present brachydactyly, genu valgus deformity, and joint pain.

Your genetic map

Gene	SNP	Genotype
LOC1053	rs121912882	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=166011

Hereditary Diseases (genetics)

Spondyloepiphyseal dysplasia congenita

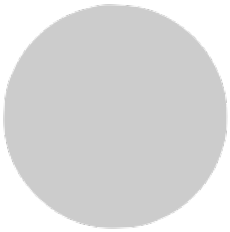
Spondyloepiphyseal dysplasia congenita (SEDC) is a chondrodysplasia characterized by disproportionate short stature, abnormal epiphyses and flattened vertebral bodies.

Your genetic map

Gene	SNP	Genotype
COL2A1	rs121912870	CC
COL2A1	rs121912874	GG
COL2A1	rs864621973	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=94068

Hereditary Diseases (genetics)

Spondyloepimetaphyseal dysplasia, PAPSS2 type

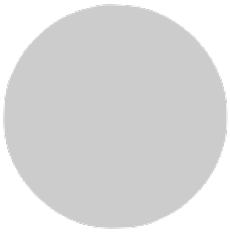
Spondyloepimetaphyseal dysplasia (SEMD), Pakistani type is characterized by short stature, short and bowed lower limbs, mild brachydactyly, kyphoscoliosis, abnormal gait, enlarged knee joints, precocious osteoarthropathy, and normal intelligence.

Your genetic map

Gene	SNP	Genotype
PAPSS2	rs121908952	CC
PAPSS2	rs201203612	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=93282

Hereditary Diseases (genetics)

Spondyloepiphyseal dysplasia, Stanescu type

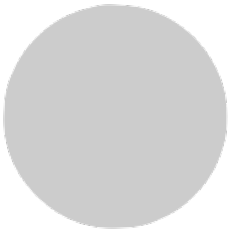
A rare spondyloepiphyseal dysplasia characterized by progressive joint contractures with premature degenerative joint disease, particularly in the knee, hip, and finger joints. Patients are of normal height and present with gait problems, joint pain, and enlarged joints with joint restriction and contractures. Radiological features include generalized platyspondyly, hypoplastic ilia, epiphyseal flattening with metaphyseal splaying of the tubular bones, and broad, elongated femoral necks with marked coxa valga. Histopathologic examination of cartilage shows PAS-positive cytoplasmic inclusion bodies in chondrocytes.

Your genetic map

Gene	SNP	Genotype
COL2A1	rs869312907	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=459051

Hereditary Diseases (genetics)

Spondyloepimetaphyseal dysplasia with multiple dislocations

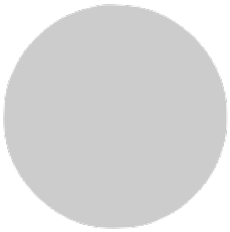
Spondyloepimetaphyseal dysplasia with multiple dislocations is a rare genetic primary bone dysplasia disorder characterized by midface hypoplasia, short stature, generalized joint laxity, multiple joint dislocations (most frequently of knees and hips), limb malalignment (genu valgum/varum) and progressive spinal deformity (e.g. kyphosis/scoliosis). Radiography reveals distinctive slender metacarpals and metatarsals, as well as small, irregular epiphyses, metaphyseal irregularities with vertical striations, constricted femoral necks and mild platyspondyly, among others.

Your genetic map

Gene	SNP	Genotype
KIF22	rs193922921	CC
KIF22	rs193922922	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=93360

Hereditary Diseases (genetics)

Spondyloepimetaphyseal dysplasia congenita, Strudwick type

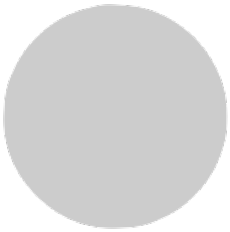
Spondyloepimetaphyseal dysplasia congenita, Strudwick type is characterized by disproportionate short stature from birth (with a very short trunk and shortened limbs) and skeletal abnormalities (lordosis, scoliosis, flattened vertebrae, pectus carinatum, coxa vara, clubfoot, and abnormal epiphyses or metaphyses).

Your genetic map

Gene	SNP	Genotype
COL2A1	rs121912880	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=93346

Hereditary Diseases (genetics)

Acromelic frontonasal dysplasia

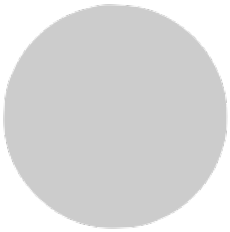
A rare frontonasal dysplasia characterized by distinct craniofacial (large fontanelle, hypertelorism, bifid nasal tip, nasal clefting, brachycephaly, median cleft face, carp-shaped mouth), brain (interhemispheric lipoma, agenesis of the corpus callosum), and limb (tibial hypoplasia/aplasia, club foot, symmetric preaxial polydactyly of the feet and bilateral clubbed and thickened nails of halluces) malformations as well as intellectual disability. Other manifestations sometimes reported include absent olfactory bulbs, hypopituitarism and cryptorchidism.

Your genetic map

Gene	SNP	Genotype
ZSWIM6	rs587777695	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1827

Hereditary Diseases (genetics)

Gnathodiaphyseal dysplasia

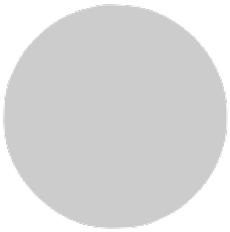
Gnathodiaphyseal dysplasia (GDD) is a bone dysplasia characterized by bone fragility, frequent bone fractures at a young age, cemento-osseous lesions of the jaw bones, bowing of tubular bones (tibia and fibula) and diaphyseal sclerosis of long bones associated with generalized osteopenia. GD follows an autosomal dominant mode of transmission.

Your genetic map

Gene	SNP	Genotype
ANO5	rs142027093	GG
ANO5	rs749645231	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=53697

Hereditary Diseases (genetics)

Schimke immuno-osseous dysplasia

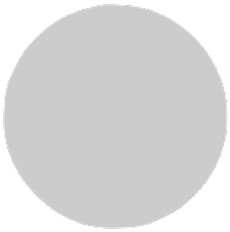
A rare a multisystem disorder characterized by spondyloepiphyseal dysplasia and disproportionate short stature, facial dysmorphism, T-cell immunodeficiency, and progressive, proteinuric steroid-resistant nephropathy.

Your genetic map

Gene	SNP	Genotype
SMARCAL	rs119473033	GG
SMARCAL	rs119473037	CC
SMARCAL	rs119473038	CC
SMARCAL	rs267607071	GG
SMARCAL	rs864309531	GG
SMARCAL	rs761546902	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1830

Hereditary Diseases (genetics)

Odonto-onycho-dermal dysplasia

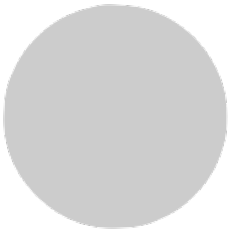
A rare, genetic, ectodermal dysplasia syndrome characterized by dental abnormalities (primarily agenesis of the permanent and deciduous teeth with cone-shaped incisors and canines), onychodysplasia, palmoplantar hyperkeratosis, dry skin and, more variably, hypotrichosis, and sweat gland dysfunction (hyper- or hypohidrosis).

Your genetic map

Gene	SNP	Genotype
WNT10A	rs121908118	GG
WNT10A	rs121908121	GG
WNT10A	rs762739726	CC
WNT10A	rs377416834	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2721

Hereditary Diseases (genetics)

Otospondylomegaepiphyseal dysplasia

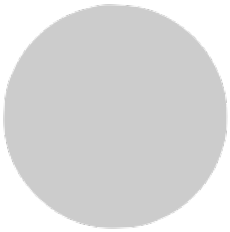
Otospondylomegaepiphyseal dysplasia (OSMED) is an inborn error of cartilage collagen formation characterized by sensorineural hearing loss, enlarged epiphyses, skeletal dysplasia with disproportionately short limbs, vertebral body anomalies and a characteristic facies.

Your genetic map

Gene	SNP	Genotype
COL11A2	rs121912945	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1427

Hereditary Diseases (genetics)

Thanatophoric dysplasia

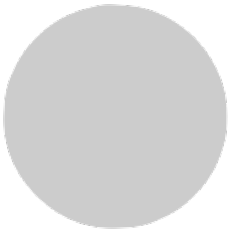
A primary bone dysplasia with micromelia characterized by micromelia, macrocephaly, narrow thorax, and distinctive facial features. It includes TD, type 1 (TD1) and TD, type 2 (TD2), that can be differentiated from each other by femur and skull shape.

Your genetic map

Gene	SNP	Genotype
FGFR3	rs121913479	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2655

Hereditary Diseases (genetics)

FLNA-related X-linked myxomatous valvular dysplasia

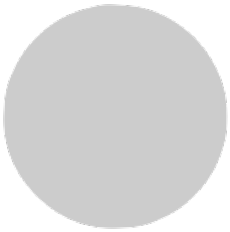
A rare genetic cardiac malformation characterized by progressive myxomatous degeneration predominantly of the mitral valve (but not uncommonly with multivalvular involvement), presenting as valve thickening and dysfunction with variable stenosis, prolapse, and/or regurgitation, and potentially resulting in lethal heart failure. Hyperextensible skin and joint hypermobility have been reported in some patients. Hemizygous males display a more severe phenotype than heterozygous females.

Your genetic map

Gene	SNP	Genotype
FLNA	rs267606815	GG
FLNA	rs797045044	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=555877

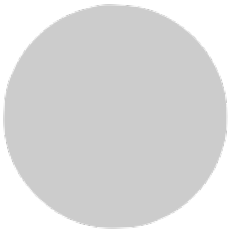
Hereditary Diseases (genetics)

Familial isolated arrhythmogenic right ventricular dysplasia

Familial isolated arrhythmogenic right ventricular dysplasia (ARVC) is the familial autosomal dominant form of ARVC (see this term), a heart muscle disease characterized by life-threatening ventricular arrhythmias with left bundle branch block configuration that may manifest with palpitations, ventricular tachycardia, syncope and sudden fatal attacks, and that is due to dystrophy and fibro-fatty replacement of the right ventricular myocardium that may lead to right ventricular aneurysms.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=217656

Your genetic map

Gene	SNP	Genotype
DSP	rs397516915	CC
DSP	rs397516940	CC
DSP	rs397516943	CC
DSP	rs397516955	GG
DSP	rs727504443	GG
DSP	rs767643821	CC
DSP	rs770873593	CC
DSP	rs794728124	CC
DSP	rs141026028	CC
DSP	rs886039178	CC
DSP	rs886039343	CC
DSP	rs1060500618	CC
DSP	rs746877365	CC
DSP	rs1060500609	GG

Hereditary Diseases (genetics)

Dopa-responsive dystonia due to sepiapterin reductase deficiency

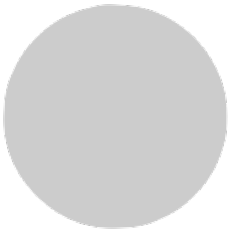
Dopa-responsive dystonia (DRD) due to sepiapterin reductase deficiency (SRD) is a very rare neurometabolic disorder characterized by dystonia with diurnal fluctuations, axial hypotonia, oculogyric crises, and delays in motor and cognitive development.

Your genetic map

Gene	SNP	Genotype
SPR	rs104893665	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=70594

Hereditary Diseases (genetics)

Early-onset generalized limb-onset dystonia

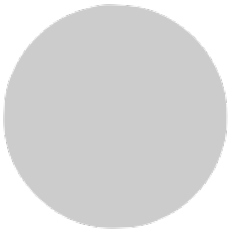
A rare movement disorder characterized by involuntary, repetitive, sustained muscle contractions or postures that typically begins in a single limb and, in most individuals, followed by progressive involvement of other limbs and the trunk, typically sparing the cranial and cervical region.

Your genetic map

Gene	SNP	Genotype
TOR1A	rs760768475	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=256

Hereditary Diseases (genetics)

Adult-onset dystonia-parkinsonism

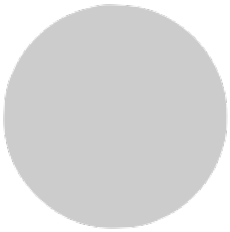
A rare neurodegenerative disease usually presenting before the age of 30 and which is characterized by dystonia, L-dopa-responsive parkinsonism, pyramidal signs and rapid cognitive decline.

Your genetic map

Gene	SNP	Genotype
BAIAP2L2	rs121908686	CC
BAIAP2L2	rs121908687	GG
PLA2G6	rs199935023	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=199351

Hereditary Diseases (genetics)

Reis Bücklers corneal dystrophy

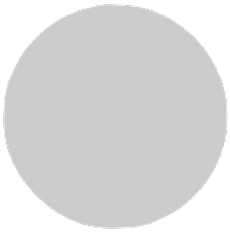
Reis Bücklers corneal dystrophy (RBCD), also known as granular corneal dystrophy type III, is a rare form of superficial corneal dystrophy characterized by bilateral symmetrical reticular opacities in the superficial central cornea, with progressive visual impairment.

Your genetic map

Gene	SNP	Genotype
TGFBI	rs121909211	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=98961

Hereditary Diseases (genetics)

Granular corneal dystrophy type II

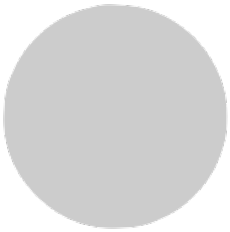
Type II granular corneal dystrophy (GCDII) is a rare form of stromal corneal dystrophy characterized by irregular-shaped well-demarcated granular deposits in the superficial central corneal stroma, and progressive visual impairment.

Your genetic map

Gene	SNP	Genotype
TGFBI	rs121909211	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=98963

Hereditary Diseases (genetics)

Granular corneal dystrophy type I

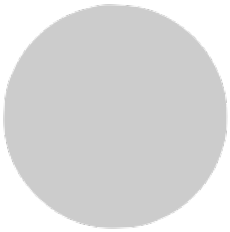
Type I granular corneal dystrophy (GCDI) is a rare form of stromal corneal dystrophy characterized by multiple small deposits in the superficial central corneal stroma, and progressive visual impairment, which may sometimes be severe.

Your genetic map

Gene	SNP	Genotype
TGFBI	rs121909210	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=98962

Hereditary Diseases (genetics)

Lattice corneal dystrophy type I

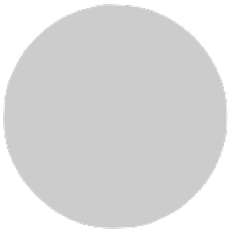
Type I lattice corneal dystrophy (LCDI) is a frequent form of stromal corneal dystrophy characterized by a network of delicate interdigitating branching filamentous opacities within the cornea with progressive visual impairment and no systemic manifestations.

Your genetic map

Gene	SNP	Genotype
TGFBI	rs121909210	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=98964

Hereditary Diseases (genetics)

Bietti crystalline dystrophy

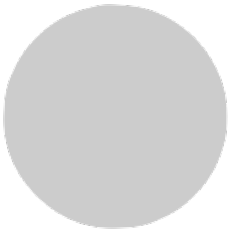
Bietti's crystalline dystrophy (BCD) is a rare progressive autosomal recessive tapetoretinal degeneration disease, occurring in the third decade of life, characterized by small sparkling crystalline deposits in the posterior retina and corneal limbus in addition to sclerosis of the choroidal vessels and manifesting as nightblindness, decreased vision, paracentral scotoma, and, in the end stages of the disease, legal blindness.

Your genetic map

Gene	SNP	Genotype
CYP4V2	rs119103283	TT
CYP4V2	rs199476183	AA
CYP4V2	rs199476203	GG
CYP4V2	rs199476204	CC
CYP4V2	rs199476189	GG
CYP4V2	rs199476197	AA
CYP4V2	rs369063468	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=41751

Hereditary Diseases (genetics)

Congenital hereditary endothelial dystrophy type II

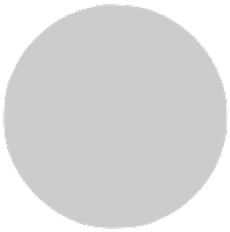
Congenital hereditary endothelial dystrophy II (CHED II) is a rare subtype of posterior corneal dystrophy characterized by a diffuse ground-glass appearance of the corneas and marked corneal thickening from birth with nystagmus, and blurred vision.

Your genetic map

Gene	SNP	Genotype
SLC4A11	rs121909388	GG
SLC4A11	rs121909392	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=293603

Hereditary Diseases (genetics)

Benign concentric annular macular dystrophy

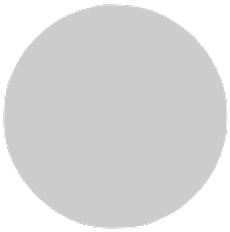
Benign concentric annular macular dystrophy (BCAMD) is a progressive autosomal dominant macular dystrophy characterized by parafoveal hypopigmentation followed by a retinitis pigmentosa-like phenotype (nyctalopia and peripheral vision loss) with a bullís eye configuration.

Your genetic map

Gene	SNP	Genotype
ABCA4	rs61749423	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=251287

Hereditary Diseases (genetics)

Congenital muscular dystrophy with cerebellar involvement

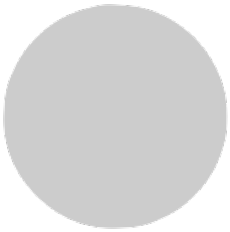
Congenital muscular dystrophy with cerebellar involvement is a rare, congenital muscular dystrophy due to dystroglycanopathy characterized by proximal muscle weakness with a tendency for muscle hypertrophy and pseudohypertrophy, variable cognitive impairment, microcephaly, cerebellar hypoplasia with or without cysts, and other structural brain anomalies.

Your genetic map

Gene	SNP	Genotype
FKRP	rs104894681	CC
FKRP	rs28937903	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=370959

Hereditary Diseases (genetics)

Congenital muscular dystrophy with integrin alpha-7 deficiency

Congenital muscular dystrophy with integrin alpha-7 deficiency is a rare, genetic, congenital muscular dystrophy due to extracellular matrix protein anomaly characterized by early motor development delay and muscle weakness with mild elevation of serum creatine kinase, that may be followed by progressive disease course with predominantly proximal muscle weakness and atrophy, motor development regress, scoliosis and respiratory insufficiency.

Your genetic map

Gene	SNP	Genotype
ITGA7	rs17854600	GG

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=34520

Hereditary Diseases (genetics)

Congenital muscular dystrophy, Ullrich type

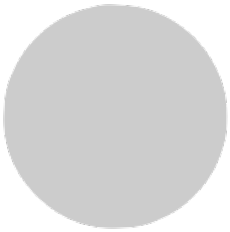
Ullrich congenital muscular dystrophy (UCMD) is characterized by early-onset, generalized and slowly progressive muscle weakness, multiple proximal joint contractures, marked hypermobility of the distal joints and normal intelligence.

Your genetic map

Gene	SNP	Genotype
COL6A3	rs398124119	GG
COL6A3	rs398124126	CC
COL6A3	rs398124128	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=75840

Hereditary Diseases (genetics)

Congenital muscular dystrophy due to LMNA mutation

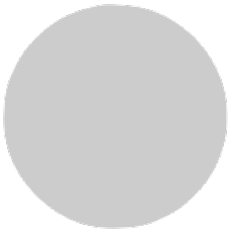
A rare congenital muscular dystrophy characterized by prominent axial hypotonia, predominantly proximal muscle weakness in upper limbs and distal in lower limbs, joint contractures (initially distal, later proximal), spinal rigidity, and progressive respiratory insufficiency, in the presence of moderately elevated serum creatine kinase. Cardiac arrhythmias and sudden death have also been reported.

Your genetic map

Gene	SNP	Genotype
LMNA	rs121912496	CC
LMNA	rs60458016	GG
LMNA	rs267607632	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=157973

Hereditary Diseases (genetics)

Becker muscular dystrophy

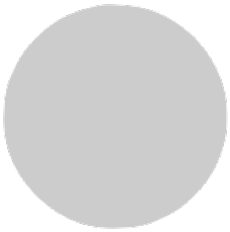
A rare, genetic muscular dystrophy characterized by progressive muscle wasting and weakness due to degeneration of skeletal, smooth and cardiac muscle.

Your genetic map

Gene	SNP	Genotype
DMD	rs5030730	GG
DMD	rs398122853	CC
DMD	rs398123935	GG
DMD	rs398124002	AA
DMD	rs373286166	CC
DMD	rs794727666	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=98895

Hereditary Diseases (genetics)

Autosomal dominant limb-girdle muscular dystrophy type 1A

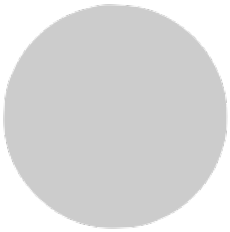
A rare subtype of autosomal dominant limb girdle muscular dystrophy characterized by an adult onset of proximal shoulder and hip girdle weakness (that later progresses to include distal weakness), nasal speech and dysarthria. Other frequent findings include tightened heel cords, reduced deep-tendon reflexes and elevated creatine kinase serum levels. Respiratory failure, as well as mild facial weakness and dysphagia, may also be observed.

Your genetic map

Gene	SNP	Genotype
MYOT	rs121908457	CC
PKD2L2	rs28937597	CC
PKD2L2	rs121908458	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=266

Hereditary Diseases (genetics)

DNAJB6-related limb-girdle muscular dystrophy D1

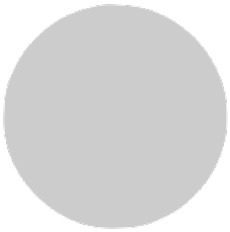
A subtype of autosomal dominant limb-girdle muscular dystrophy characterized by an adult-onset of slowly progressive, proximal pelvic girdle weakness, with none, or only minimal, shoulder girdle involvement, and absence of cardiac and respiratory symptoms. Mild to moderate elevated creatine kinase serum levels and gait abnormalities are frequently observed.

Your genetic map

Gene	SNP	Genotype
DNAJB6	rs149278319	CC
DNAJB6	rs387907150	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=34516

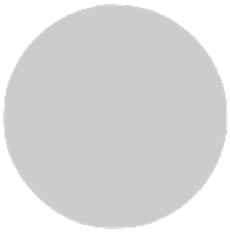
Hereditary Diseases (genetics)

Calpain-3-related limb-girdle muscular dystrophy R1

A subtype of autosomal recessive limb girdle muscular dystrophy characterized by a variable age of onset of progressive, typically symmetrical and selective weakness and atrophy of proximal shoulder- and pelvic-girdle muscles (gluteus maximus, thigh adductors, and muscles of the posterior compartment of the limbs are most commonly affected) without cardiac or facial involvement. Clinical manifestations include exercise intolerance, a waddling gait, scapular winging and calf pseudo-hypertrophy.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=267

Your genetic map

Gene	SNP	Genotype
CAPN3	rs80338802	GG
CAPN3	rs121434544	GG
CAPN3	rs121434547	CC
CAPN3	rs121434548	GG
CAPN3	rs201736037	AA
CAPN3	rs149095128	CC
CAPN3	rs587780290	GG
CAPN3	rs727503839	GG
CAPN3	rs141656719	CC
CAPN3	rs794726871	CC
CAPN3	rs557164942	CC
CAPN3	rs147774793	CC
CAPN3	rs778768583	GG
CAPN3	rs774048743	GG
CAPN3	rs374665929	AA
CAPN3	rs863224956	GG
CAPN3	rs863224957	CC
CAPN3	rs863224959	CC
CAPN3	rs863224960	GG
CAPN3	rs863224961	GG
CAPN3	rs863224962	AA
CAPN3	rs761211705	GG
CAPN3	rs776043976	CC
CAPN3	rs149914792	GG
CAPN3	rs369552114	GG
CAPN3	rs199806879	CC
CAPN3	rs200379491	AA
DYSF	rs727503915	GG
LOC1053	rs863224964	GG
LOC1053	rs878854364	CC

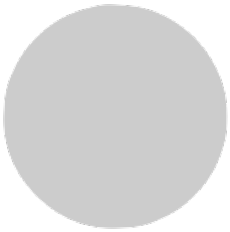
Hereditary Diseases (genetics)

Titin-related limb-girdle muscular dystrophy R10

A form of limb-girdle muscular dystrophy that usually has a childhood onset (but can range from the first to third decade of life) of severe progressive proximal weakness, eventually involving the distal muscles. Some patients may remain ambulatory but most are wheelchair dependant 20 years after onset.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=140922

Your genetic map

Gene	SNP	Genotype
TTN	rs397517481	CC
TTN	rs397517689	GG
TTN	rs751746401	GG
TTN AS1	rs397517589	GG
TTN AS1	rs397517601	CC
TTN AS1	rs397517624	CC
TTN AS1	rs72646831	GG
TTN AS1	rs72646846	GG
TTN AS1	rs397517735	AA
TTN AS1	rs727503586	AA
TTN AS1	rs557312035	GG
TTN AS1	rs574660186	GG
TTN AS1	rs794727539	GG
TTN AS1	rs112188483	CC
TTN AS1	rs781540455	GG
TTN AS1	rs794729278	GG
TTN AS1	rs72646837	CC
TTN AS1	rs761807131	CC
TTN AS1	rs751502842	GG
TTN AS1	rs565675340	GG
TTN AS1	rs543860009	GG
TTN AS1	rs72677247	AA
TTN AS1	rs886038916	GG
TTN AS1	rs886042331	GG

Hereditary Diseases (genetics)

POMT1-related limb-girdle muscular dystrophy R11

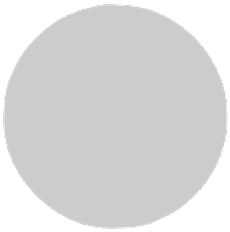
A form of limb-girdle muscular dystrophy characterized by the onset of slowly progressive proximal muscle weakness during childhood (with fatigue and difficulty running and climbing stairs) and developmental delay. Mild intellectual deficit and microcephaly, without any obvious structural brain abnormality, are found in all patients. Mild pseudohypertrophy and joint contractures of the ankles have also been reported.

Your genetic map

Gene	SNP	Genotype
POMT1	rs119462982	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=86812

Hereditary Diseases (genetics)

Anoctamin-5-related limb-girdle muscular dystrophy R12

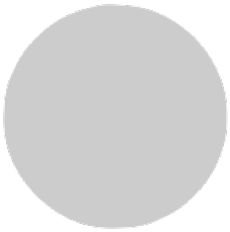
A form of limb-girdle muscular dystrophy most often characterized by an adult onset (but ranging from 11 to 51 years) of mainly proximal lower limb weakness, with difficulties standing on tiptoes being one of the initial signs. Proximal upper limb and distal lower limb weakness is also common, as well as atrophy of the quadriceps (most commonly), biceps brachii, and lower leg muscles. Calf hypertrophy has also been reported in some cases. LGMD2L progresses slowly, with most patients remaining ambulatory until late adulthood.

Your genetic map

Gene	SNP	Genotype
ANO5	rs137854524	CC
ANO5	rs398124625	GG
ANO5	rs137854526	TT
ANO5	rs372221490	GG
ANO5	rs566415362	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=206549

Hereditary Diseases (genetics)

POMT2-related limb-girdle muscular dystrophy R14

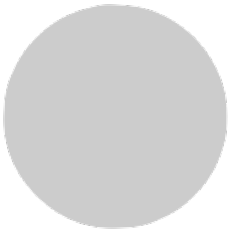
A form of limb-girdle muscular dystrophy characterized by proximal weakness (manifesting as slowness in running) presenting in infancy, along with calf hypertrophy, mild lordosis, scapular winging and normal intelligence (or mild intellectual disability).

Your genetic map

Gene	SNP	Genotype
POMT2	rs587780423	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=206559

Hereditary Diseases (genetics)

GMPPB-related limb-girdle muscular dystrophy R19

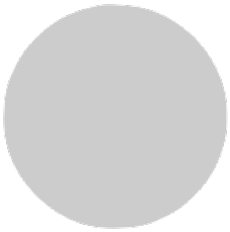
A form of limb-girdle muscular dystrophy, that can present from birth to early childhood, characterized by hypotonia, microcephaly, mild proximal muscle weakness (leading to delayed walking and difficulty climbing stairs), mild intellectual disability and epilepsy. Additional manifestations reported in some patients include cataracts, nystagmus, cardiomyopathy, and respiratory insufficiency.

Your genetic map

Gene	SNP	Genotype
GMPPB	rs142336618	CC
RNF123	rs199922550	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=363623

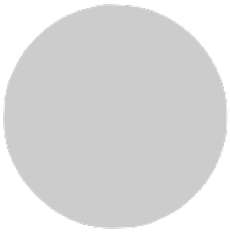
Hereditary Diseases (genetics)

Dysferlin-related limb-girdle muscular dystrophy R2

A subtype of autosomal recessive limb-girdle muscular dystrophy characterized by an onset in late adolescence or early adulthood of slowly progressive, proximal weakness and atrophy of shoulder and pelvic girdle muscles. Cardiac and respiratory muscles are not involved. Hypertrophy of the calf muscles and highly elevated serum creatine kinase levels are frequently observed.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=268

Your genetic map

Gene	SNP	Genotype
DYSF	rs121908955	CC
DYSF	rs121908956	CC
DYSF	rs121908963	GG
DYSF	rs786205084	GG
DYSF	rs398123763	GG
DYSF	rs398123765	TT
DYSF	rs202044973	CC
DYSF	rs398123768	GG
DYSF	rs377735262	CC
DYSF	rs140108514	GG
DYSF	rs398123787	GG
DYSF	rs398123789	CC
DYSF	rs398123794	GG
DYSF	rs373585652	CC
DYSF	rs398123800	GG
DYSF	rs201869739	GG
DYSF	rs727503911	CC
DYSF	rs201049092	GG
DYSF	rs756328339	AA
DYSF	rs370874727	AA
DYSF	rs794727636	CC
DYSF	rs766016391	GG
DYSF	rs794727851	GG
DYSF	rs141497053	GG
DYSF	rs746873768	CC
DYSF	rs369607332	CC
DYSF	rs863225021	CC
DYSF	rs150877497	GG
DYSF	rs746315830	CC
DYSF	rs199543257	CC

Hereditary Diseases (genetics)

Alpha-sarcoglycan-related limb-girdle muscular dystrophy R3

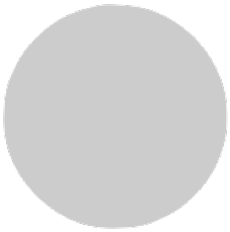
A subtype of autosomal recessive limb-girdle muscular dystrophy characterized by childhood onset of progressive proximal weakness of the shoulder and pelvic girdle muscles, resulting in difficulty walking, scapular winging, calf hypertrophy and contractures of the Achilles tendon, which lead to a tiptoe gait pattern. Cardiac and respiratory involvement is rare.

Your genetic map

Gene	SNP	Genotype
LOC1053	rs137852621	GG
LOC1053	rs28933693	CC
LOC1053	rs371675217	GG
LOC1053	rs768814872	TT
LOC1053	rs758647756	CC
LOC1053	rs138945081	CC
SGCA	rs137852623	CC
SGCA	rs143570936	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=62

Hereditary Diseases (genetics)

Beta-sarcoglycan-related limb-girdle muscular dystrophy R4

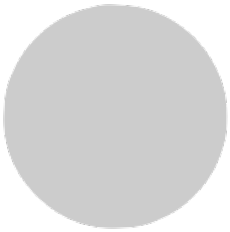
A subtype of autosomal recessive limb girdle muscular dystrophy characterized by a childhood to adolescent onset of progressive pelvic- and shoulder-girdle muscle weakness, particularly affecting the pelvic girdle (adductors and flexors of hip). Usually the knees are the earliest and most affected muscles. In advanced stages, involvement of the shoulder girdle (resulting in scapular winging) and the distal muscle groups are observed. Calf hypertrophy, cardiomyopathy, respiratory impairment, tendon contractures, scoliosis, and exercise-induced myoglobinuria may be observed.

Your genetic map

Gene	SNP	Genotype
SGCB	rs28936383	GG
SGCB	rs104893868	AA
SGCB	rs104893869	CC
SGCB	rs150518260	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=119

Hereditary Diseases (genetics)

Gamma-sarcoglycan-related limb-girdle muscular dystrophy R5

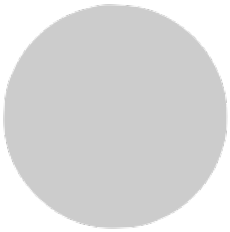
A subtype of autosomal recessive limb-girdle muscular dystrophy characterized by a childhood onset of progressive shoulder and pelvic girdle muscle weakness and atrophy frequently associated with calf hypertrophy, diaphragmatic weakness, and/or variable cardiac abnormalities. Mild to moderate elevated serum creatine kinase levels and positive Gowers sign are reported.

Your genetic map

Gene	SNP	Genotype
LOC1079	rs104894422	GG
LOC1079	rs104894423	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=353

Hereditary Diseases (genetics)

Telethonin-related limb-girdle muscular dystrophy R7

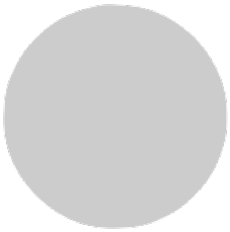
A mild subtype of autosomal recessive limb-girdle muscular dystrophy characterized by a variable onset (ranging from infancy to adolescence) of progressive proximal upper and lower limb muscle weakness and atrophy. Mild scapular winging, calf hypertrophy, and lack of respiratory and cardiac involvement are also observed.

Your genetic map

Gene	SNP	Genotype
TCAP	rs104894655	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=34514

Hereditary Diseases (genetics)

FKRP-related limb-girdle muscular dystrophy R9

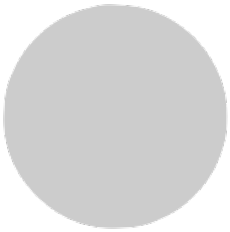
A form of autosomal recessive limb-girdle muscular dystrophy that presents a highly variable age of onset and phenotypic spectrum typically characterized by slowly progressive proximal weakness of the pelvic and shoulder girdle musculature (predominantly affecting the lower limbs), frequently associated with waddling gait, scapular winging, calf and tongue hypertrophy, exercise-induced myalgia, abdominal muscle weakness, cardiomyopathy, respiratory muscle involvement, and myoglobinuria and/or elevated creatine kinase serum levels.

Your genetic map

Gene	SNP	Genotype
FKRP	rs28937900	CC
FKRP	rs104894682	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=34515

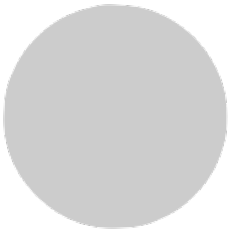
Hereditary Diseases (genetics)

Duchenne muscular dystrophy

A rare, genetic, muscular dystrophy characterized by rapidly progressive muscle weakness and wasting due to degeneration of skeletal, smooth and cardiac muscle.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=98896

Your genetic map

Gene	SNP	Genotype
DMD	rs128625228	GG
DMD	rs128625229	GG
DMD	rs104894787	GG
DMD	rs201366610	GG
DMD	rs128626235	GG
DMD	rs146071084	AA
DMD	rs128626232	GG
DMD	rs128626242	CC
DMD	rs128626246	CC
DMD	rs128626249	GG
DMD	rs128626250	GG
DMD	rs128626251	GG
DMD	rs104894790	GG
DMD	rs104894797	GG
DMD	rs128627256	GG
DMD	rs398123827	GG
DMD	rs398123828	CC
DMD	rs398123832	GG
DMD	rs398123833	GG
DMD	rs398123834	CC
DMD	rs398123852	GG
DMD	rs398123853	GG
DMD	rs398123862	CC
DMD	rs398123865	GG
DMD	rs398123870	GG
DMD	rs398123883	GG
DMD	rs398123884	CC
DMD	rs398123888	GG
DMD	rs398123889	CC
DMD	rs398123901	CC
DMD	rs398123903	GG

Hereditary Diseases (genetics)

Tibial muscular dystrophy

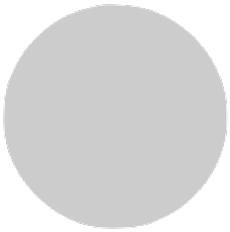
Tibial muscular dystrophy (TMD) is a distal myopathy characterized by weakness of the muscles of the anterior compartment of lower limbs, appearing in the fourth to seventh decade of life.

Your genetic map

Gene	SNP	Genotype
TTN AS1	rs587780495	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=609

Hereditary Diseases (genetics)

Muscular dystrophy, Selcen type

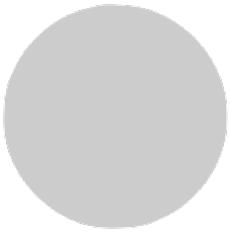
Selcen type muscular dystrophy is characterized by progressive limb and axial muscle weakness associated with cardiomyopathy and severe respiratory insufficiency during adolescence. The disease manifests during childhood and progresses rapidly.

Your genetic map

Gene	SNP	Genotype
BAG3	rs121918312	CC
BAG3	rs397516881	GG
BAG3	rs117749531	GG
BAG3	rs869248137	CC
BAG3	rs1057517945	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=199340

Hereditary Diseases (genetics)

Infantile neuroaxonal dystrophy

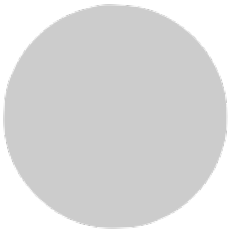
Infantile neuroaxonal dystrophy/atypical neuroaxonal dystrophy (INAD/atypical NAD) is a type of neurodegeneration with brain iron accumulation (NBIA; see this term) characterized by psychomotor delay and regression, increasing neurological involvement with symmetrical pyramidal tract signs and spastic tetraplegia. INAD may be classic or atypical and patients present with symptoms anywhere along a continuum between the two.

Your genetic map

Gene	SNP	Genotype
PLA2G6	rs200075782	GG
PLA2G6	rs587784347	GG
PLA2G6	rs587784339	GG
PLA2G6	rs587784327	CC
PLA2G6	rs587784363	CC
PLA2G6	rs587784359	GG
PLA2G6	rs794729212	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=35069

Hereditary Diseases (genetics)

Butterfly-shaped pigment dystrophy

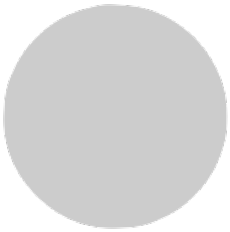
A rare patterned dystrophy of the retinal pigment epithelium characterized by abnormal accumulation of lipofuscin in a butterfly-shaped distribution at the retinal pigment epithelium level. Patients manifest with a slowly progressive loss of vision that often only becomes apparent in old age.

Your genetic map

Gene	SNP	Genotype
PRPH2	rs121918563	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=99001

Hereditary Diseases (genetics)

Progressive cone dystrophy

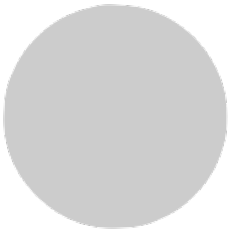
A rare retinal dystrophy characterized by photophobia, progressive loss of visual acuity, nystagmus, visual field abnormalities, abnormal color vision, and psychophysical and electrophysiological evidence of abnormal cone function. Progressive cone dystrophy usually presents in childhood or early adult life, and patients tend to develop rod photoreceptor dysfunction in later life.

Your genetic map

Gene	SNP	Genotype
PDE6C	rs762426409	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1871

Hereditary Diseases (genetics)

Bothnia retinal dystrophy

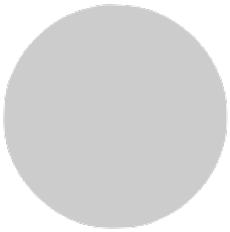
Bothnia retinal dystrophy is a rare form of retinal dystrophy, seen mostly in Northern Sweden, presenting in early childhood with night blindness and progressive maculopathy with a decrease in visual acuity, eventually leading to blindness by adulthood. Retinal degeneration, without obvious bone spicule formation, accompanied by affected visual fields and the typical presence of retinitis punctata albescens in the posterior pole are also noted.

Your genetic map

Gene	SNP	Genotype
RLBP1	rs28933990	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=85128

Hereditary Diseases (genetics)

Best vitelliform macular dystrophy

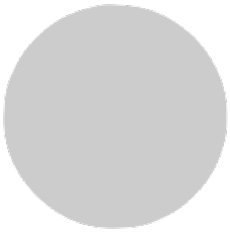
Best vitelliform macular dystrophy (BVMD) is a genetic macular dystrophy characterized by loss of central visual acuity, metamorphopsia and a decrease in the Arden ratio secondary to an egg yolk-like lesion located in the foveal or parafoveal region.

Your genetic map

Gene	SNP	Genotype
LOC1079	rs28940570	CC
LOC1079	rs267606677	AA
LOC1079	rs281865238	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1243

Hereditary Diseases (genetics)

DPM1-CDG

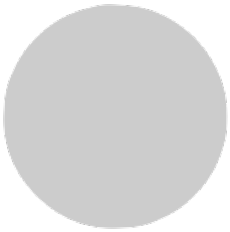
The CDG (Congenital Disorders of Glycosylation) syndromes are a group of autosomal recessive disorders affecting glycoprotein synthesis. CDG syndrome type Ie is characterised by psychomotor delay, seizures, hypotonia, facial dysmorphism and microcephaly. Ocular anomalies are also very common.

Your genetic map

Gene	SNP	Genotype
MOCS3	rs139624629	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79322

Hereditary Diseases (genetics)

Isolated ectopia lentis

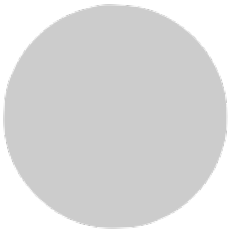
Isolated ectopia lentis (IEL) is a rare, clinically variable, eye disorder characterized by dislocation of the lens, often causing significant reduction in visual acuity.

Your genetic map

Gene	SNP	Genotype
FBN1	rs137854464	CC
FBN1	rs137854480	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1885

Hereditary Diseases (genetics)

Microcephalic osteodysplastic primordial dwarfism type II

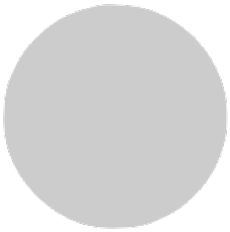
A rare bone disease and a form of microcephalic primordial dwarfism characterized by severe pre- and postnatal growth retardation, with marked microcephaly in proportion to body size, skeletal dysplasia, abnormal dentition, insulin resistance, and increased risk for cerebrovascular disease.

Your genetic map

Gene	SNP	Genotype
PCNT	rs119479063	GG
PCNT	rs181690344	CC
PCNT	rs587784308	GG
PCNT	rs369195346	GG
PCNT	rs587784321	CC
PCNT	rs151020551	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2637

Hereditary Diseases (genetics)

Mitochondrial neurogastrointestinal encephalomyopathy

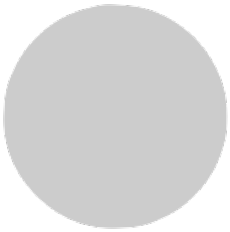
Mitochondrial NeuroGastroIntestinal Encephalomyopathy (MNGIE) syndrome is characterized by the association of gastrointestinal dysmotility, peripheral neuropathy, chronic progressive external ophthalmoplegia and leukoencephalopathy.

Your genetic map

Gene	SNP	Genotype
SCO2	rs121913039	CC
TYMP	rs863224255	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=298

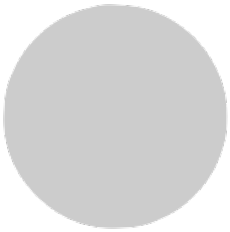
Hereditary Diseases (genetics)

KCNQ2-related epileptic encephalopathy

KCNQ2-related epileptic encephalopathy is a severe form of neonatal epilepsy that usually manifests in newborns during the first week of life with seizures (that affect alternatively both sides of the body), often accompanied by clonic jerking or more complex motor behavior, as well as signs of encephalopathy such as diffuse hypotonia, limb spasticity, lack of visual fixation and tracking and mild to moderate intellectual deficiency. The severity can range from controlled to intractable seizures and mild/moderate to severe intellectual disability.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=439218

Your genetic map

Gene	SNP	Genotype
KCNQ2	rs74315392	GG
KCNQ2	rs587777219	GG
KCNQ2	rs727503974	GG
KCNQ2	rs794727740	CC
KCNQ2	rs794727813	CC
KCNQ2	rs796052643	GG
KCNQ2	rs796052626	GG
KCNQ2	rs796052621	CC
KCNQ2	rs796052620	AA
KCNQ2	rs864321707	GG
KCNQ2	rs886041262	CC
KCNQ2	rs1057516095	GG
KCNQ2	rs1057516094	GG
KCNQ2	rs796052618	CC
LOC1053	rs796052645	CC
LOC1053	rs796052655	CC
LOC1053	rs796052653	CC
LOC1053	rs796052652	GG
LOC1053	rs118192234	CC

Hereditary Diseases (genetics)

Early infantile epileptic encephalopathy

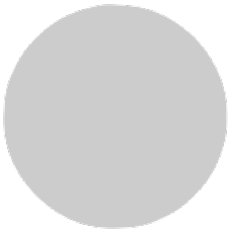
A severe form of age-related epileptic encephalopathies characterized by the onset of tonic spasms within the first 3 months of life that can be generalized or lateralized, independent of the sleep cycle, and that can occur hundreds of times per day, leading to psychomotor impairment and death.

Your genetic map

Gene	SNP	Genotype
GNAO1	rs797044878	GG
GNAO1	rs797044951	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1934

Hereditary Diseases (genetics)

Ethylmalonic encephalopathy

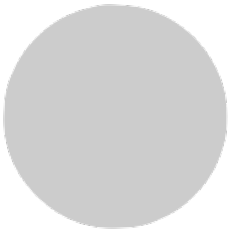
Ethylmalonic acid encephalopathy (EE) is defined by elevated excretion of ethylmalonic acid (EMA) with recurrent petechiae, orthostatic acrocyanosis and chronic diarrhoea associated with neurodevelopmental delay, psychomotor regression and hypotonia with brain magnetic resonance imaging (MRI) abnormalities.

Your genetic map

Gene	SNP	Genotype
ETHE1	rs28940289	GG
ETHE1	rs863223954	TT
ETHE1	rs745656120	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=51188

Hereditary Diseases (genetics)

Severe neonatal-onset encephalopathy with microcephaly

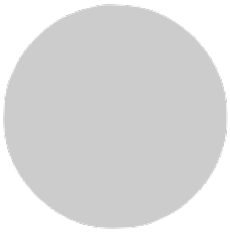
Severe neonatal-onset encephalopathy with microcephaly is a rare monogenic disease with epilepsy characterized by neonatal-onset encephalopathy, microcephaly, severe developmental delay or absent development, breathing abnormalities (including central hypoventilation and/or respiratory insufficiency), intractable seizures, abnormal muscle tone and involuntary movements. Early death is usual.

Your genetic map

Gene	SNP	Genotype
MECP2	rs61754437	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=209370

Hereditary Diseases (genetics)

Encephalopathy due to sulfite oxidase deficiency

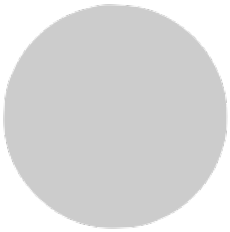
Encephalopathy due to sulfite oxidase deficiency is a rare neurometabolic disorder characterized by seizures, progressive encephalopathy and lens dislocation.

Your genetic map

Gene	SNP	Genotype
MOCS1	rs104893969	CC
MOCS1	rs104893970	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=833

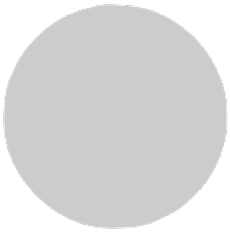
Hereditary Diseases (genetics)

Glycine encephalopathy

Glycine encephalopathy (GE) is an inborn error of glycine metabolism characterized by accumulation of glycine in body fluids and tissues, including the brain, resulting in neurometabolic symptoms of variable severity.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=407

Your genetic map

Gene	SNP	Genotype
AMT	rs121964984	CC
AMT	rs121964985	CC
AMT	rs386833690	CC
AMT	rs797045082	CC
GLDC	rs121964974	CC
GLDC	rs386833549	CC
GLDC	rs121964979	GG
GLDC	rs121964980	CC
GLDC	rs386833517	GG
GLDC	rs386833536	TT
GLDC	rs386833555	TT
GLDC	rs386833560	GG
GLDC	rs386833576	GG
GLDC	rs386833585	GG
GLDC	rs386833587	GG
GLDC	rs772871471	GG
GLDC	rs149070244	CC
GLDC	rs191905539	CC
GLDC	rs188269735	AA
NICN1	rs386833679	GG
PCDH19	rs796052815	GG

Hereditary Diseases (genetics)

STAT3-related early-onset multisystem autoimmune disease

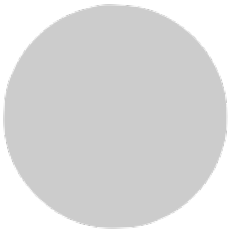
A rare, genetic, lymphoproliferative syndrome characterized by early onset recurrent infections, lymphadenopathy with hepatosplenomegaly and variable autoimmune disorders, including hemolytic anemia, thrombocytopenia, neutropenia, enteropathy, type I diabetes, scleroderma, arthritis, atopic dermatitis, and inflammatory lung disease. Patients commonly have failure to thrive. Variable immunologic findings include decreased regulatory T-cells, hypogammaglobulinemia, and reduction in memory B cells.

Your genetic map

Gene	SNP	Genotype
STAT3	rs869312892	GG
STAT3	rs869312894	CC
STAT3	rs869312889	GG
STAT3	rs869312887	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=438159

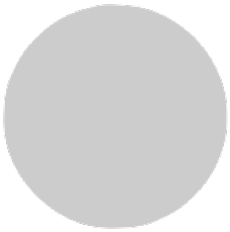
Hereditary Diseases (genetics)

Central core disease

Central core disease (CCD) is an inherited neuromuscular disorder characterised by central cores on muscle biopsy and clinical features of a congenital myopathy.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=597

Your genetic map

Gene	SNP	Genotype
RYR1	rs28933396	GG
RYR1	rs118192166	AA
RYR1	rs118192143	CC
RYR1	rs118192133	GG
RYR1	rs118192156	TT
RYR1	rs118192136	GG
RYR1	rs118192183	GG
RYR1	rs118192139	AA
RYR1	rs118192147	CC
RYR1	rs118192123	TT
RYR1	rs118192131	TT
RYR1	rs118192138	TT
RYR1	rs118192124	CC
RYR1	rs118192122	GG
RYR1	rs118192178	CC
RYR1	rs118192125	GG
RYR1	rs118192180	CC
RYR1	rs118192150	CC
RYR1	rs118192184	AA
RYR1	rs118192154	GG
RYR1	rs118192134	CC
RYR1	rs193922884	CC
RYR1	rs113928116	GG
RYR1	rs113460156	GG
RYR1	rs1456276440	CC

Hereditary Diseases (genetics)

Juvenile neuronal ceroid lipofuscinosis

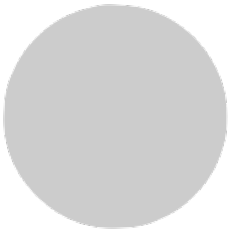
Juvenile neuronal ceroid lipofuscinoses (JNCLs) are a genetically heterogeneous group of neuronal ceroid lipofuscinoses (NCLs; see this term) typically characterized by onset at early school age with vision loss due to retinopathy, seizures and the decline of mental and motor capacities.

Your genetic map

Gene	SNP	Genotype
CLN3	rs386833694	GG
CLN3	rs386833695	CC
CLN3	rs386833744	CC
CLN3	rs796052335	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=228346

Hereditary Diseases (genetics)

Addison disease

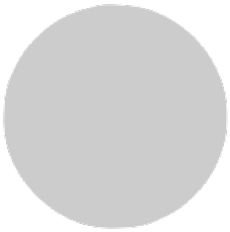
A chronic and rare endocrine disorder due to autoimmune destruction of the adrenal cortex and resulting in a glucocorticoid and mineralocorticoid deficiency. Properly speaking, it designates autoimmune adrenalitis, but it is a term commonly used to describe any form of chronic primary adrenal insufficiency (CPAI).

Your genetic map

Gene	SNP	Genotype
ABCD1	rs128624225	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=85138

Hereditary Diseases (genetics)

Alexander disease

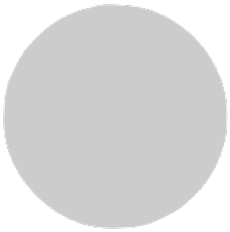
A rare neurodegenerative disorder of the astrocytes comprised of two clinical forms: Alexander disease (AxD) type I and type II manifesting with various degrees of macrocephaly, spasticity, ataxia and seizures and leading to psychomotor regression and death.

Your genetic map

Gene	SNP	Genotype
GFAP	rs58064122	GG
GFAP	rs59565950	CC
GFAP	rs59793293	GG
GFAP	rs61622935	GG
GFAP	rs58075601	CC
GFAP	rs797044590	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=58

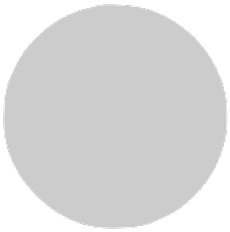
Hereditary Diseases (genetics)

Glycogen storage disease due to glycogen debranching enzyme deficiency

Glycogen debranching enzyme (GDE) deficiency, or glycogen storage disease type 3 (GSD 3), is a form of glycogen storage disease characterized by severe muscle weakness and hepatopathy.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=366

Your genetic map

Gene	SNP	Genotype
AGL	rs113994126	CC
AGL	rs113994129	GG
AGL	rs369973784	AA
AGL	rs199922945	GG
AGL	rs113994128	CC
AGL	rs267606640	GG
AGL	rs113994130	CC
AGL	rs113994131	CC
AGL	rs771961377	CC
AGL	rs370792293	AA
AGL	rs193186112	CC
AGL	rs794729208	TT
AGL	rs201201443	GG
AGL	rs775498547	CC

Hereditary Diseases (genetics)

Glycogen storage disease due to glycogen branching enzyme deficiency

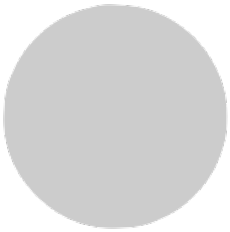
Glycogen branching enzyme (GBE) deficiency (Andersen's disease or amylopectinosis), or glycogen storage disease type 4 (GSD4), is a rare and severe form of glycogen storage disease which accounts for approximately 3% of all the glycogen storage diseases (see these terms).

Your genetic map

Gene	SNP	Genotype
GBE1	rs80338671	TT
GBE1	rs80338672	GG
GBE1	rs137852887	AA
GBE1	rs80338673	CC
GBE1	rs201958741	CC
GBE1	rs192044702	AA
GBE1	rs766935302	GG
GBE1	rs781198373	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=367

Hereditary Diseases (genetics)

Glycogen storage disease due to muscle phosphofructokinase deficiency

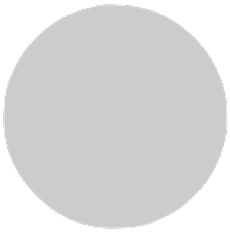
Muscle phosphofructokinase (PFK) deficiency (Tarui's disease), or glycogen storage disease type 7 (GSD7), is a rare form of glycogen storage disease characterized by exertional fatigue and muscular exercise intolerance. It occurs in childhood.

Your genetic map

Gene	SNP	Genotype
MIR6505	rs202143236	GG
MIR6505	rs138893744	CC
PFKM	rs121918193	GG
PFKM	rs746348793	GG
PFKM	rs770066278	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=371

Hereditary Diseases (genetics)

Glycogen storage disease due to phosphoglycerate mutase deficiency

Muscle phosphoglycerate mutase deficiency (PGAMD) is a metabolic myopathy characterised by exercise-induced cramp, myoglobinuria, and presence of tubular aggregates in the muscle biopsy. Serum creatine kinase (CK) levels are increased between episodes of myoglobinuria. Less than 50 cases have been described so far. The disease is due to an anomaly in one of the last steps of glycolysis. The enzymatic defect in PGAMD is caused by mutations in the cDNA coding for the M-isoform of PGAM. Residual PGAM activity in the muscles of patients (2%-6%) is due to activity of the B-isoform. Transmission is autosomal recessive. Differential diagnosis includes muscle phosphorylase deficiency (McArdle disease) and phosphofructokinase deficiency (PFKD) (see these terms).

Your genetic map

Gene	SNP	Genotype
PGAM2	rs10250779	CC
PGAM2	rs104894030	TT

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=97234

Hereditary Diseases (genetics)

Glycogen storage disease due to liver phosphorylase kinase deficiency

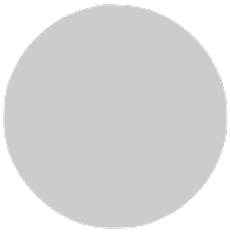
Glycogen storage disease (GSD) due to liver phosphorylase kinase (PhK) deficiency is a benign inborn error of glycogen metabolism characterized by hepatomegaly, growth retardation, and mild delay in motor development during childhood.

Your genetic map

Gene	SNP	Genotype
PHKA2	rs137852290	CC
PHKA2	rs137852291	TT
PHKA2	rs137852292	GG
PHKA2	rs137852294	GG
PHKA2	rs797044877	CC
PHKA2	rs137852293	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=264580

Hereditary Diseases (genetics)

Glycogen storage disease due to liver and muscle phosphorylase kinase deficiency

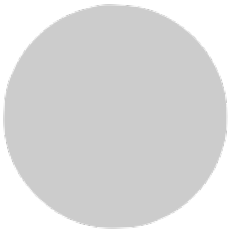
A benign inborn error of glycogen metabolism. It is the mildest form of GSD due to PhK deficiency.

Your genetic map

Gene	SNP	Genotype
PHKB	rs371296953	GG
PHKB	rs535749057	AA

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79240

Hereditary Diseases (genetics)

Glycogen storage disease due to liver glycogen phosphorylase deficiency

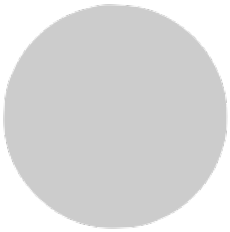
Liver phosphorylase deficiency, or glycogen storage disease type 6b (Hers' disease, GSD 6b) is a benign and rare form of glycogen storage disease.

Your genetic map

Gene	SNP	Genotype
PYGL	rs113993982	CC
PYGL	rs113993981	CC
PYGL	rs113993973	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=369

Hereditary Diseases (genetics)

Glycogen storage disease due to muscle glycogen phosphorylase deficiency

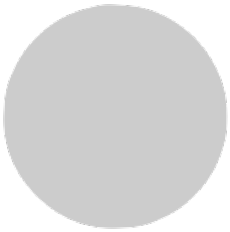
Myophosphorylase deficiency (McArdle's disease), or glycogen storage disease type 5 (GSD5) , is a severe form of glycogen storage disease characterized by exercise intolerance.

Your genetic map

Gene	SNP	Genotype
PYGM	rs119103251	CC
PYGM	rs119103252	TT
PYGM	rs144081869	CC
PYGM	rs267606993	TT
PYGM	rs119103259	CC
PYGM	rs398124208	CC
PYGM	rs398124209	GG
PYGM	rs527236146	GG
PYGM	rs771427957	CC
RASGRP2	rs119103258	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=368

Hereditary Diseases (genetics)

Glycogen storage disease due to hepatic glycogen synthase deficiency

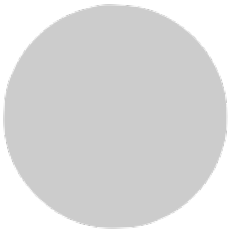
A genetically inherited anomaly of glycogen metabolism and a form of glycogen storage disease (GSD) characterized by fasting hypoglycemia. This is not a glycogenosis, strictly speaking, as the enzyme deficiency decreases glycogen reserves.

Your genetic map

Gene	SNP	Genotype
GYS2	rs121918419	GG
GYS2	rs121918421	CC
GYS2	rs150382575	GG
GYS2	rs201157731	GG
GYS2	rs146195866	GG
GYS2	rs372079212	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2089

Hereditary Diseases (genetics)

Caffey disease

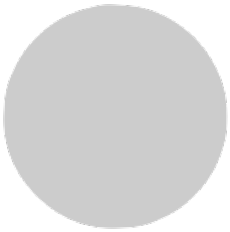
Caffey disease is an osteosclerotic dysplasia characterized by acute inflammation with massive subperiosteal new bone formation usually involving the diaphyses of the long bones, as well as the ribs, mandible, scapulae, and clavicles. The disease is associated with fever, irritability pain and soft tissue swelling, with onset around the age of 2 months and resolving spontaneously by the age of 2 years. However, prenatal disease onset has also been described.

Your genetic map

Gene	SNP	Genotype
COL1A1	rs72653170	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1310

Hereditary Diseases (genetics)

Canavan disease

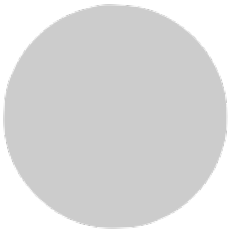
Canavan disease (CD) is a neurodegenerative disorder; its spectrum varies between severe forms with leukodystrophy, macrocephaly and severe developmental delay, and a very rare mild/juvenile form characterized by mild developmental delay.

Your genetic map

Gene	SNP	Genotype
SPATA22	rs28940279	AA
SPATA22	rs28940574	CC
SPATA22	rs104894552	AA
SPATA22	rs104894553	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=141

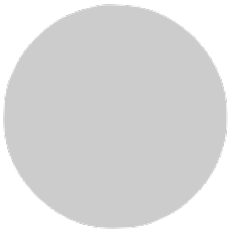
Hereditary Diseases (genetics)

Autosomal dominant Charcot-Marie-Tooth disease type 2A2

A subtype of Autosomal dominant Charcot-Marie-Tooth disease type 2 characterized by the childhood onset of distal weakness and areflexia (with earlier and more severe involvement of the lower extremities), reduced sensory modalities (primarily pain and temperature sensation), foot deformities, postural tremor, scoliosis and contractures. Optic atrophy, vocal cord palsy with dysphonia, sensorineural hearing loss, spinal cord abnormalities and hydrocephalus have also been reported.

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=99947

Your genetic map

Gene	SNP	Genotype
MFN2	rs28940291	GG
MFN2	rs28940292	GG
MFN2	rs28940293	TT
MFN2	rs28940294	GG
MFN2	rs119103263	CC
MFN2	rs119103265	CC
MFN2	rs119103268	CC
MFN2	rs387906991	CC
MFN2	rs587777875	CC
MFN2	rs794729198	CC
MFN2	rs863224069	CC
MFN2	rs863224969	CC
MFN2	rs863224970	AA
MFN2	rs863224967	AA
MFN2	rs863224968	CC

Hereditary Diseases (genetics)

Autosomal dominant Charcot-Marie-Tooth disease type 2D

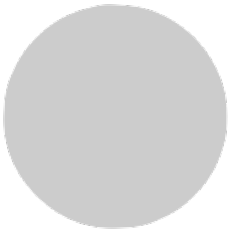
A form of axonal Charcot-Marie-Tooth disease, a peripheral sensorimotor neuropathy, characterized by distal weakness primarily and predominantly occurring in the upper limbs and tendon reflexes absent or reduced in the arms and decreased in the legs. Progression is slow.

Your genetic map

Gene	SNP	Genotype
GARS1	rs137852643	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=99938

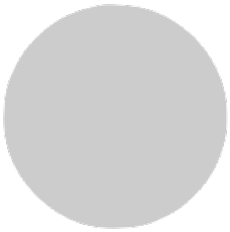
Hereditary Diseases (genetics)

X-linked Charcot-Marie-Tooth disease type 1

X-linked Charcot-Marie-Tooth disease type 1 is a rare, genetic, peripheral sensorimotor neuropathy characterized by an X-linked dominant inheritance pattern and the childhood-onset (within the first decade in males) of progressive, distal, moderate to severe muscle weakness and atrophy in lower extremities and intrinsic hand muscles, pes cavus, bilateral foot drop, reduced or absent tendon reflexes, as well as mild to moderate sensory impairment in lower extremities. Females tend to have milder manifestations or may be asymptomatic. Sensorineural deafness and central nervous system involvement have also been reported.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=101075

Your genetic map

Gene	SNP	Genotype
GJB1	rs104894810	CC
GJB1	rs104894811	CC
GJB1	rs104894812	GG
GJB1	rs104894814	CC
GJB1	rs104894819	AA
GJB1	rs104894821	GG
GJB1	rs104894822	AA
GJB1	rs104894824	CC
GJB1	rs116840818	GG
GJB1	rs116840819	CC
GJB1	rs116840815	CC
GJB1	rs116840822	GG
GJB1	rs756928158	GG
GJB1	rs863224471	CC
GJB1	rs863224971	CC
GJB1	rs863224972	GG
GJB1	rs863224973	CC
GJB1	rs139643362	CC
GJB1	rs864622215	GG
GJB1	rs879254047	GG

Hereditary Diseases (genetics)

X-linked Charcot-Marie-Tooth disease type 5

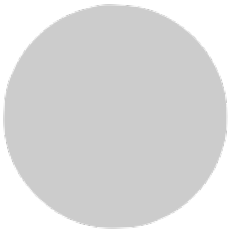
A rare form of X-linked Charcot-Marie-Tooth disease, a peripheral sensorimotor neuropathy, characterized by infancy- to childhood-onset of: 1) progressive distal muscle weakness and atrophy (first appearing and more prominent in the lower extremities than the upper) which usually manifests with foot drop and gait disturbance, 2) bilateral, profound, prelingual sensorineural hearing loss and 3) progressive optic neuropathy.

Your genetic map

Gene	SNP	Genotype
PRPS1	rs80338732	TT
PRPS1	rs587781262	AA
PRPS1	rs587781263	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=99014

Hereditary Diseases (genetics)

Charcot-Marie-Tooth disease type 1B

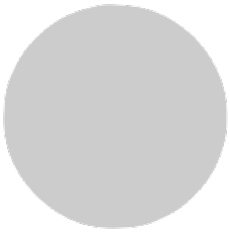
Charcot-Marie-Tooth disease type 1B (CMT1B) is a form of CMT1 (see this term), caused by mutations in the MPZ gene (1q22), that presents with the manifestations of peripheral neuropathy (distal muscle weakness and atrophy, foot deformities and sensory loss). The phenotype is variable depending on the particular mutation. Two distinct presentations have been described: (1) an early infantile onset severe phenotype with delayed walking and motor nerve conduction velocities (MNCV) <10 m/s, often referred to as Dejerine-Sottas syndrome (see this term), or (2) a much later onset phenotype (>age 40), with normal or mildly slowed MNCV and more frequent hearing loss and pupillary abnormalities. CMT1B can also cause the classical CMT phenotype in about 15% of total CMT1B cases.

Your genetic map

Gene	SNP	Genotype
MPZ	rs121913584	GG
MPZ	rs121913585	GG
MPZ	rs121913586	CC
MPZ	rs121913587	AA
MPZ	rs121913588	CC
MPZ	rs121913589	CC
MPZ	rs121913590	GG
MPZ	rs121913594	TT
MPZ	rs121913601	GG
MPZ	rs121913603	TT
MPZ	rs281865128	CC
MPZ	rs863225025	CC

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=101082

Hereditary Diseases (genetics)

Charcot-Marie-Tooth disease type 1D

Charcot-Marie-Tooth disease type 1D (CMT1D) is a form of CMT1 (see this term), caused by mutations in the EGR2 gene (10q21.1), with a variable severity and age of onset (from infancy to adulthood), that usually presents with gait abnormalities, progressive wasting and weakness of distal limb muscles, with possible later involvement of proximal muscles, foot deformity and severe reduction in nerve conduction velocity. Additional features may include scoliosis, cranial nerve deficits such as diplopia, and bilateral vocal cord paresis.

Your genetic map

Gene	SNP	Genotype
EGR2	rs104894161	GG

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=101084

Hereditary Diseases (genetics)

Charcot-Marie-Tooth disease type 2B5

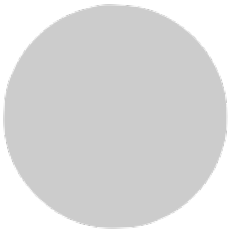
A rare axonal hereditary motor and sensory neuropathy characterized by infantile onset of slowly progressive distal motor weakness and atrophy (more severe in legs and moderate in arms) with mildly delayed motor development, hypotonia, and distal sensory impairment of all sensory modalities.

Your genetic map

Gene	SNP	Genotype
NEFL	rs58982919	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=228374

Hereditary Diseases (genetics)

Autosomal dominant Charcot-Marie-Tooth disease type 2N

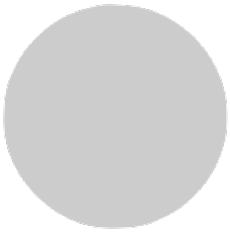
A mild form of axonal Charcot-Marie-Tooth disease, a peripheral sensorimotor neuropathy, characterized by distal legs sensory loss and weakness that can be asymmetric. Tendon reflexes are reduced in the knees and absent in ankles. Progression is slow.

Your genetic map

Gene	SNP	Genotype
AARS1	rs267606621	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=228174

Hereditary Diseases (genetics)

Charcot-Marie-Tooth disease type 2T

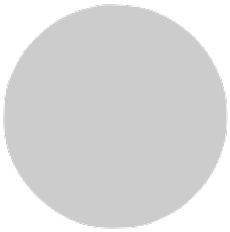
A rare autosomal recessive axonal hereditary motor and sensory neuropathy characterized by adult onset of slowly progressive distal muscle weakness and atrophy, sensory impairment, and decreased or absent deep tendon reflexes predominantly in the lower extremities. Patients present gait disturbances but remain ambulatory. Mild involvement of the upper limbs may be seen.

Your genetic map

Gene	SNP	Genotype
DNAJB2	rs797045039	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=495274

Hereditary Diseases (genetics)

SURF1-related Charcot-Marie-Tooth disease type 4

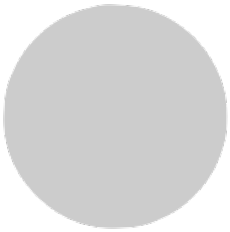
A subtype of Charcot-Marie-Tooth disease type 4 characterized by childhood onset of severe, progressive, demyelinating sensorimotor neuropathy manifesting with distal muscle weakness and atrophy of hands and feet, distal sensory impairment (vibration and pinprick) of lower limbs, lactic acidosis, areflexia and severely reduced motor nerve conduction velocities (25 m/s or less). Patients may also present kyphoscoliosis, nystagmus, hearing loss, cerebellar ataxia and/or brain MRI abnormalities (putaminal and periaqueductal lesions).

Your genetic map

Gene	SNP	Genotype
SURF1	rs782190413	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=391351

Hereditary Diseases (genetics)

Charcot-Marie-Tooth disease type 4A

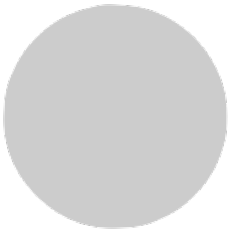
Charcot-Marie-Tooth disease type 4A (CMT4A) is a subtype of Charcot-Marie-Tooth disease type 4 characterized by early-onset (infancy to early childhood) of severe, rapidly progressing demyelinating, axonal, or intermediate sensorimotor neuropathy usually affecting first, and more severely, the distal lower extremities and later the proximal muscles and upper extremities. Nerve conduction velocities range from very slow to normal. Apart from the typical CMT phenotype (distal muscle weakness and atrophy, sensory loss, frequent pes cavus foot deformity), patients commonly present delayed motor development, vocal cord paresis, mild sensory loss, abolished deep tendon reflexes, and skeletal deformities.

Your genetic map

Gene	SNP	Genotype
GDAP1	rs104894075	CC
GDAP1	rs745663149	CC
GDAP1	rs864622501	GG

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=99948

Hereditary Diseases (genetics)

Charcot-Marie-Tooth disease type 4C

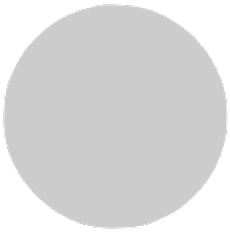
Charcot-Marie-Tooth disease type 4C (CMT4C) is a subtype of Charcot-Marie-Tooth type 4 characterized by childhood or adolescent-onset of a relatively mild, demyelinating sensorimotor neuropathy that contrasts with a severe, rapidly progressing, early-onset scoliosis, and the typical CMT phenotype (i.e. distal muscle weakness and atrophy, sensory loss, and often foot deformity). A wide spectrum of nerve conduction velocities are observed and cranial nerve involvement and kyphoscoliosis have also been reported.

Your genetic map

Gene	SNP	Genotype
MIR584	rs864309709	TT
SH3TC2	rs80338933	GG
SH3TC2	rs80338934	GG
SH3TC2	rs80338925	CC
SH3TC2	rs80338926	GG
SH3TC2	rs80338931	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=99949

Hereditary Diseases (genetics)

Charcot-Marie-Tooth disease type 4F

Charcot-Marie-Tooth disease type 4F (CMT4F) is a severe, demyelinating subtype of Charcot-Marie-Tooth disease type 4 characterized by the childhood onset of a slowly-progressing typical CMT phenotype (i.e. distal muscle weakness and atrophy, as well as pes cavus) that presents severe sensory loss (frequently with sensory ataxia), moderately to severely reduced motor nerve conduction velocities and almost invariable absence of sensory nerve action potentials, and delayed motor milestones.

Your genetic map

Gene	SNP	Genotype
PRX	rs104894714	GG
PRX	rs104894707	AA

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=99952

Hereditary Diseases (genetics)

Charcot-Marie-Tooth disease type 4J

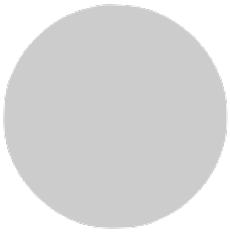
Charcot-Marie-Tooth disease type 4J is a subtype of Charcot-Marie-Tooth disease type 4 characterized by childhood- to adulthood-onset of variably severe, rapidly progressive, axonal and demyelinating sensorimotor neuropathy typically manifesting with delayed motor development, proximal and distal asymmetric muscle weakness and atrophy of the lower and upper extremities, severe motor dysfunction with mildly reduced sensory impairment, and areflexia. Nerve conduction velocities range from very mildly to severely reduced.

Your genetic map

Gene	SNP	Genotype
FIG4	rs377357931	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=139515

Hereditary Diseases (genetics)

Coats disease

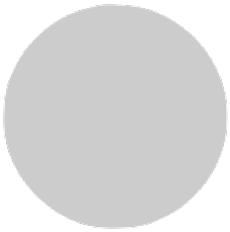
Coats disease (CD) is an idiopathic disorder characterized by retinal telangiectasia with deposition of intraretinal or subretinal exudates, potentially leading to retinal detachment and unilateral blindness. CD is classically an isolated and unilateral condition affecting otherwise healthy young children.

Your genetic map

Gene	SNP	Genotype
PRSS23	rs80358284	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=190

Hereditary Diseases (genetics)

Sporadic Creutzfeldt-Jakob disease

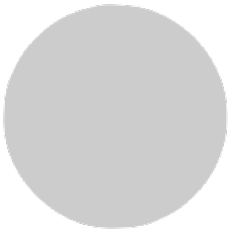
A rare sporadic human prion disease characterized by rapidly progressive cognitive impairment in combination with variable neurologic signs and symptoms including myoclonus, visual or cerebellar problems, pyramidal or extrapyramidal features, or akinetic mutism. Brain imaging may show high signal intensity in caudate, putamen, and/or cortical regions, and a typical EEG pattern consisting of generalized periodic sharp wave complexes is observed in many cases. The disease is invariably fatal within less than two years. Neuropathologic examination reveals deposition of abnormal prion protein in brain tissue, as well as spongiform change and massive neuronal loss and gliosis.

Your genetic map

Gene	SNP	Genotype
PRNP	rs74315408	GG
PRNP	rs74315412	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=204

Hereditary Diseases (genetics)

Crouzon disease

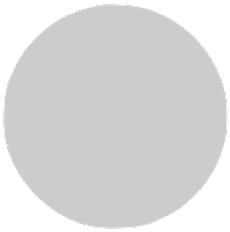
Crouzon disease is characterized by craniosynostosis and facial hypoplasia.

Your genetic map

Gene	SNP	Genotype
FGFR2	rs121918487	CC
FGFR2	rs121918489	AA
FGFR2	rs121918490	GG
FGFR2	rs121918491	CC
FGFR2	rs121918493	TT
FGFR2	rs121918494	GG
FGFR2	rs121918497	TT
FGFR2	rs121918501	AA
FGFR2	rs121918488	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=207

Hereditary Diseases (genetics)

Dent disease

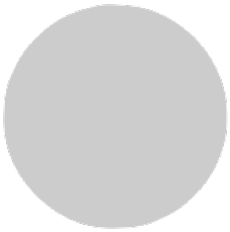
Dent disease is a rare genetic renal tubular disease characterized by manifestations of proximal tubule dysfunction.

Your genetic map

Gene	SNP	Genotype
CLCN5	rs151340621	CC
CLCN5	rs797044810	CC
CLCN5	rs797044813	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1652

Hereditary Diseases (genetics)

Free sialic acid storage disease

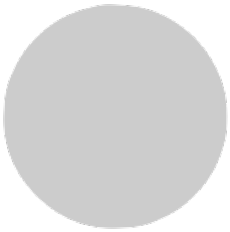
Free sialic acid storage disease (free SASD), is a group of lysosomal storage diseases characterized by a spectrum of clinical manifestations including neurological and developmental disorders with severity ranging from the milder phenotype, Salla disease (SD), to the most severe phenotype, infantile free sialic acid storage disease (ISSD).

Your genetic map

Gene	SNP	Genotype
SLC17A5	rs201284672	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=834

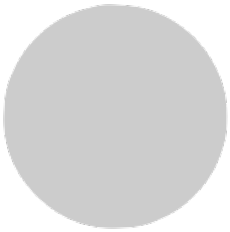
Hereditary Diseases (genetics)

Fabry disease

Fabry disease (FD) is a progressive, inherited, multisystemic lysosomal storage disease characterized by specific neurological, cutaneous, renal, cardiovascular, cochleo-vestibular and cerebrovascular manifestations.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=324

Your genetic map

Gene	SNP	Genotype
GLA	rs797044747	GG
GLA	rs869312142	AA
RPL36A	rs104894827	GG
RPL36A	rs104894828	CC
RPL36A	rs104894830	TT
RPL36A	rs104894831	GG
RPL36A	rs104894832	CC
RPL36A	rs104894835	TT
RPL36A	rs28935196	AA
RPL36A	rs28935197	TT
RPL36A	rs104894840	CC
RPL36A	rs104894841	GG
RPL36A	rs28935486	TT
RPL36A	rs28935487	TT
RPL36A	rs28935492	CC
RPL36A	rs104894842	CC
RPL36A	rs28935493	CC
RPL36A	rs104894843	GG
RPL36A	rs104894844	CC
RPL36A	rs104894845	CC
RPL36A	rs104894851	GG
RPL36A	rs104894852	TT
RPL36A	rs28935495	TT
RPL36A	rs104894834	GG
RPL36A	rs397515870	GG
RPL36A	rs398123199	GG
RPL36A	rs398123201	AA
RPL36A	rs398123206	CC
RPL36A	rs398123207	CC
RPL36A	rs398123208	CC
RPL36A	rs113173389	CC

Hereditary Diseases (genetics)

Gaucher disease

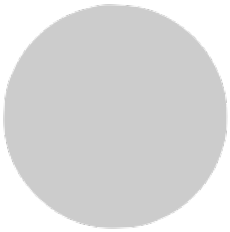
Gaucher disease (GD) is a lysosomal storage disorder encompassing three main forms (types 1, 2 and 3), a fetal form and a variant with cardiac involvement (Gaucher disease - ophthalmoplegia - cardiovascular calcification or Gaucher-like disease).

Your genetic map

Gene	SNP	Genotype
GBA1	rs76763715	TT
GBA1	rs80356769	CC
GBA1	rs80356771	GG
GBA1	rs76539814	GG
GBA1	rs75822236	CC
GBA1	rs364897	TT
GBA1	rs121908312	CC
GBA1	rs80356772	CC
GBA1	rs398123527	CC
GBA1	rs398123528	CC
GBA1	rs409652	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=355

Hereditary Diseases (genetics)

Hirschsprung disease

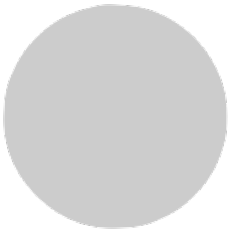
A rare congenital intestinal motility disorder that is characterized by signs of intestinal obstruction due to the presence of an aganglionic segment of variable extent in the terminal part of the colon.

Your genetic map

Gene	SNP	Genotype
RET	rs377767391	TT
RET	rs377767412	GG
RET	rs143795581	AA
RET	rs193922699	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=388

Hereditary Diseases (genetics)

Krabbe disease

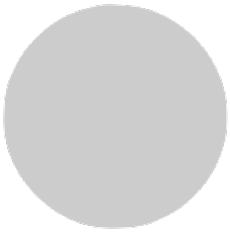
A rare lysosomal disorder that affects the white matter of the central and peripheral nervous systems characterized by neurodegeneration with severity depending on the age of onset (infantile, late-infantile, juvenile, adolescent and adulthood).

Your genetic map

Gene	SNP	Genotype
GALC	rs199847983	CC
GALC	rs200378205	CC
GALC	rs752537626	TT
GALC	rs771111145	GG
GALC	rs756690487	CC
GALC	rs756352952	GG
GALC	rs1057516453	AA
GALC	rs200960659	GG
GALC	rs200532368	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=487

Hereditary Diseases (genetics)

Lafora disease

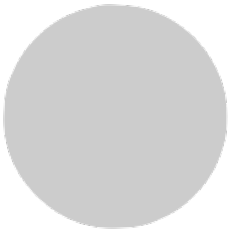
A rare, inherited, severe, progressive myoclonic epilepsy characterized by myoclonus and/or generalized seizures, visual hallucinations (partial occipital seizures), and progressive neurological decline.

Your genetic map

Gene	SNP	Genotype
EPM2A	rs104893950	GG
NHLRC1	rs28940575	AA
NHLRC1	rs28940576	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=501

Hereditary Diseases (genetics)

Leber plus disease

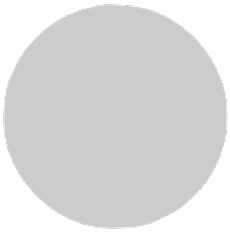
A rare inherited mitochondrial disease characterized by the clinical features of Leber hereditary optic neuropathy in combination with other systemic or neurological abnormalities. These abnormalities include: postural tremor, motor disorder, multiple sclerosis-like syndrome, spinal cord disease, skeletal changes, Parkinsonism with dystonia, anarthria, dystonia, motor and sensory peripheral neuropathy, spasticity, mild encephalopathy, and cardiac arrhythmias.

Your genetic map

Gene	SNP	Genotype
ND1	rs199476122	GG
ND6	rs199476105	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=99718

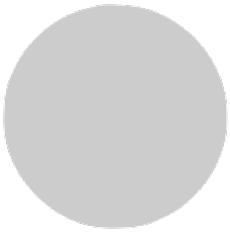
Hereditary Diseases (genetics)

Menkes disease

A rare congenital disorder of copper metabolism with severe multisystemic manifestations that are primarily characterized by progressive neurodegeneration and marked connective tissue anomalies. A pathognomonic feature is the typical sparse, abnormal steely hair.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=565

Your genetic map

Gene	SNP	Genotype
ATP7A	rs72554649	CC
ATP7A	rs72554652	GG
ATP7A	rs151340633	CC
ATP7A	rs797045399	CC
ATP7A	rs797045325	GG
ATP7A	rs72554636	CC
ATP7A	rs797045330	CC
ATP7A	rs797045332	CC
ATP7A	rs797045337	GG
ATP7A	rs797045338	GG
ATP7A	rs797045339	TT
ATP7A	rs72554639	GG
ATP7A	rs72554640	CC
ATP7A	rs797045340	GG
ATP7A	rs797045341	GG
ATP7A	rs797045342	GG
ATP7A	rs797045346	TT
ATP7A	rs797045348	GG
ATP7A	rs797045347	GG
ATP7A	rs797045349	AA
ATP7A	rs72554644	GG
ATP7A	rs797045351	GG
ATP7A	rs797045354	TT
ATP7A	rs72554645	CC
ATP7A	rs797045357	TT
ATP7A	rs797045359	GG
ATP7A	rs797045360	CC
ATP7A	rs797045363	GG
ATP7A	rs72554650	CC
ATP7A	rs797045367	GG
ATP7A	rs797045370	TT

Hereditary Diseases (genetics)

Naxos disease

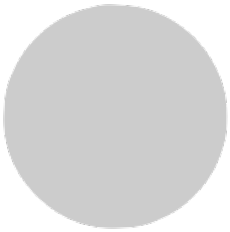
A recessively inherited condition with arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVD/C) and a cutaneous phenotype, characterised by peculiar woolly hair and palmoplantar keratoderma.

Your genetic map

Gene	SNP	Genotype
JUP	rs373761090	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=34217

Hereditary Diseases (genetics)

Niemann-Pick disease type A

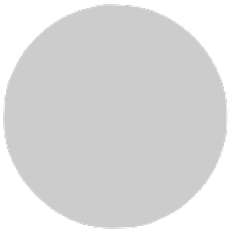
A rare, autosomal recessive, acid sphingomyelinase deficiency characterized clinically by onset in infancy or early childhood with failure to thrive, hepatosplenomegaly, interstitial lung disease and rapidly progressive neurodegenerative disorders.

Your genetic map

Gene	SNP	Genotype
SMPD1	rs120074117	GG
SMPD1	rs120074119	GG
SMPD1	rs120074122	GG
SMPD1	rs120074124	TT
SMPD1	rs120074125	TT
SMPD1	rs398123474	GG
SMPD1	rs398123475	TT
SMPD1	rs182812968	CC
SMPD1	rs398123478	CC
SMPD1	rs398123479	GG
SMPD1	rs727504166	TT
SMPD1	rs769904764	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=77292

Hereditary Diseases (genetics)

Niemann-Pick disease type B

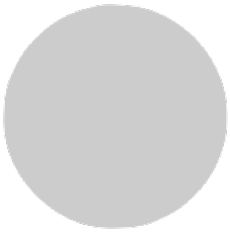
A rare autosomal recessive, chronic, acid sphingomyelinase deficiency characterized clinically by onset in childhood with hepatosplenomegaly, growth retardation, interstitial lung disease and absence of neurodegenerative disorders.

Your genetic map

Gene	SNP	Genotype
SMPD1	rs120074126	CC
SMPD1	rs120074127	CC
SMPD1	rs120074128	CC
SMPD1	rs120074117	GG
SMPD1	rs182812968	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=77293

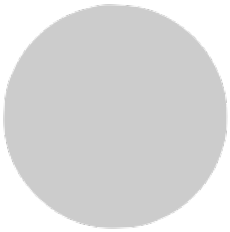
Hereditary Diseases (genetics)

Niemann-Pick disease type C

A rare lysosomal lipid storage disease characterized by variable clinical signs, depending on the age of onset, such as prolonged unexplained neonatal jaundice or cholestasis, isolated unexplained splenomegaly, and progressive, often severe neurological symptoms such as cognitive decline, cerebellar ataxia, vertical supranuclear gaze palsy (VSPG), dysarthria, dysphagia, dystonia, seizures, gelastic cataplexy, and psychiatric disorders.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=646

Your genetic map

Gene	SNP	Genotype
NPC1	rs28942105	TT
NPC1	rs80358254	CC
NPC1	rs80358259	AA
NPC1	rs120074135	CC
NPC1	rs28942107	GG
NPC1	rs786200877	CC
NPC1	rs80358252	CC
NPC1	rs28942108	GG
NPC1	rs80358253	TT
NPC1	rs483352886	CC
NPC1	rs543206298	GG
NPC1	rs369368181	GG
NPC1	rs758902805	GG
NPC1	rs200444084	CC
NPC1	rs786204455	GG
NPC1	rs139751448	CC
NPC1	rs372030650	TT
NPC1	rs794727897	CC
NPC1	rs777286835	GG
NPC1	rs886042268	TT
NPC1	rs759826138	GG

Hereditary Diseases (genetics)

Norrie disease

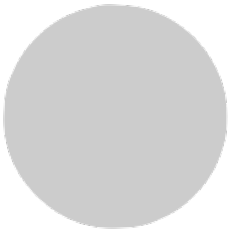
A rare developmental defect during embryogenesis characterized by abnormal retinal development with congenital blindness. Common associated manifestations include sensorineural hearing loss and developmental delay, intellectual disability and/or behavioral disorders.

Your genetic map

Gene	SNP	Genotype
NDP AS1	rs398123283	GG
NDP AS1	rs727504031	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=649

Hereditary Diseases (genetics)

Oguchi disease

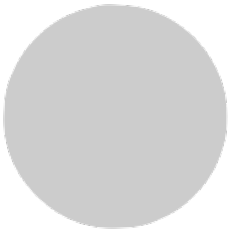
Oguchi disease is an autosomal recessive retinal disorder characterized by congenital stationary night blindness and the Mizuo-Nakamura phenomenon.

Your genetic map

Gene	SNP	Genotype
SAG	rs397514681	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=75382

Hereditary Diseases (genetics)

Pelizaeus-Merzbacher disease

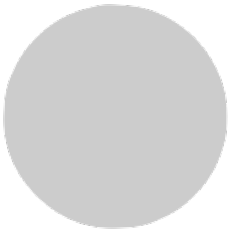
Pelizaeus-Merzbacher disease (PMD) is an X-linked leukodystrophy characterized by developmental delay, nystagmus, hypotonia, spasticity, and variable intellectual deficit. It is classified into three sub-forms based on the age of onset and severity: connatal, transitional, and classic PMD (see these terms).

Your genetic map

Gene	SNP	Genotype
RAB9B	rs132630278	CC
RAB9B	rs132630279	TT
RAB9B	rs11543022	CC
RAB9B	rs797045064	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=702

Hereditary Diseases (genetics)

Refsum disease

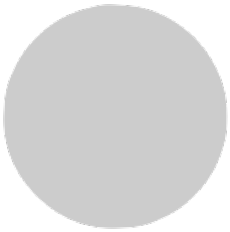
A metabolic disease characterized by anosmia, cataract, early-onset retinitis pigmentosa and possible neurological manifestations, including peripheral neuropathy and cerebellar ataxia. Other features can be deafness, ichthyosis, skeletal abnormalities, and cardiac arrhythmia. It is characterized biochemically by accumulation of phytanic acid in plasma and tissues.

Your genetic map

Gene	SNP	Genotype
PHYH	rs201578674	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=773

Hereditary Diseases (genetics)

Chylomicron retention disease

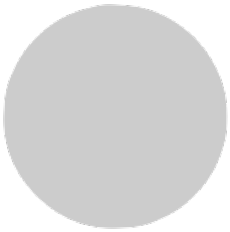
Chylomicron retention disease (CRD) is a type of familial hypocholesterolemia characterized by malnutrition, failure to thrive, growth failure, vitamin E deficiency and hepatic, neurologic and ophthalmologic complications.

Your genetic map

Gene	SNP	Genotype
SAR1B	rs28942109	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=71

Hereditary Diseases (genetics)

Sandhoff disease

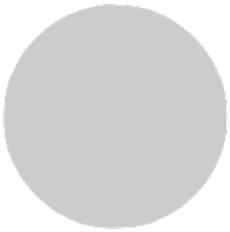
Sandhoff disease is a lysosomal storage disorder from the GM2 gangliosidosis family and is characterised by central nervous system degeneration.

Your genetic map

Gene	SNP	Genotype
HEXB	rs28942073	CC
HEXB	rs121907983	GG
HEXB	rs121907985	CC
HEXB	rs121907986	CC
HEXB	rs398123446	AA
HEXB	rs761197472	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=796

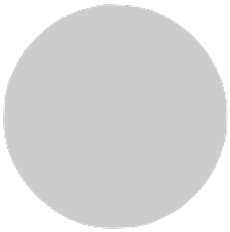
Hereditary Diseases (genetics)

Stargardt disease

A rare ophthalmic disorder that is usually characterized by a progressive loss of central vision associated with irregular macular and perimacular yellow-white fundus flecks, and a so-called "beaten bronze" atrophic central macular lesion.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=827

Your genetic map

Gene	SNP	Genotype
ABCA4	rs61751408	GG
ABCA4	rs61750200	GG
ABCA4	rs121909205	GG
ABCA4	rs61750130	GG
ABCA4	rs61751383	GG
ABCA4	rs61753033	AA
ABCA4	rs61751399	CC
ABCA4	rs61750120	GG
ABCA4	rs398123339	TT
ABCA4	rs61752390	AA
ABCA4	rs61748550	GG
ABCA4	rs61751410	CC
ABCA4	rs61748556	GG
ABCA4	rs150774447	CC
ABCA4	rs55732384	GG
ABCA4	rs61749414	GG
ABCA4	rs61752401	CC
ABCA4	rs62654395	CC
ABCA4	rs61749420	GG
ABCA4	rs201738997	TT
ABCA4	rs62654397	GG
ABCA4	rs61749428	CC
ABCA4	rs61750202	CC
ABCA4	rs61752406	CC
ABCA4	rs61749459	CC
ABCA4	rs61751397	GG
ABCA4	rs61752416	TT
ABCA4	rs61750121	CC
ABCA4	rs61752425	CC
ABCA4	rs61752427	GG
ABCA4	rs62642573	CC

Hereditary Diseases (genetics)

Tangier disease

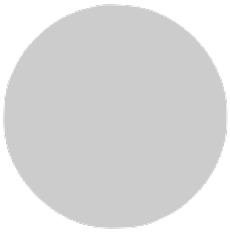
A rare, genetic neurometabolic disease characterized biochemically by an almost complete absence of plasma high-density lipoproteins (HDL), and clinically by liver, spleen, lymph node and tonsil enlargement along with multifocal peripheral neuropathy, corneal, skin and nail and, occasionally, cardiovascular disease.

Your genetic map

Gene	SNP	Genotype
ABCA1	rs28937313	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=31150

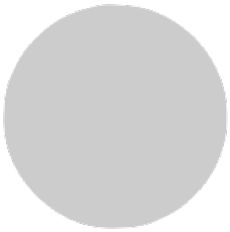
Hereditary Diseases (genetics)

Tay-Sachs disease

A rare disorder characterized by accumulation of G2 gangliosides due to hexosaminidase A deficiency.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=845

Your genetic map

Gene	SNP	Genotype
HEXA	rs147324677	CC
HEXA	rs121907952	CC
HEXA	rs797044432	CC
HEXA	rs121907955	CC
HEXA	rs28941770	CC
HEXA	rs121907953	GG
HEXA	rs121907956	CC
HEXA	rs121907957	CC
HEXA	rs121907958	CC
HEXA	rs121907959	CC
HEXA	rs28942071	GG
HEXA	rs121907966	GG
HEXA	rs76173977	CC
HEXA	rs121907972	GG
HEXA	rs387906311	CC
HEXA	rs121907980	CC
HEXA	rs587779406	GG
HEXA	rs786204585	GG
HEXA	rs370266293	CC
HEXA	rs772180415	CC
HEXA	rs762374961	CC
HEXA	rs767041069	CC
HEXA	rs150675340	GG
HEXA	rs762060470	CC
HEXA	rs185429231	CC
HEXA AS1	rs786204721	AA

Hereditary Diseases (genetics)

Thomsen and Becker disease

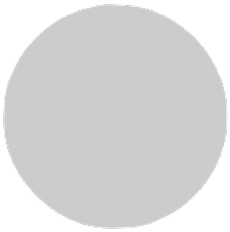
A rare, genetic, skeletal muscle channelopathy characterized by slow muscle relaxation after contraction (myotonia).

Your genetic map

Gene	SNP	Genotype
CLCN1	rs80356700	GG
CLCN1	rs80356703	GG
CLCN1	rs80356697	TT
CLCN1	rs80356685	CC
CLCN1	rs80356687	CC
CLCN1	rs80356692	GG
CLCN1	rs375596425	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=614

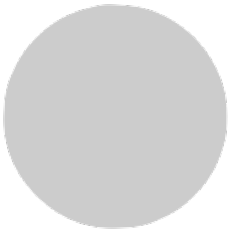
Hereditary Diseases (genetics)

Von Hippel-Lindau disease

Von Hippel-Lindau disease (VHL) is a familial cancer predisposition syndrome associated with a variety of malignant and benign neoplasms, most frequently retinal, cerebellar, and spinal hemangioblastoma, renal cell carcinoma (RCC), and pheochromocytoma.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=892

Your genetic map

Gene	SNP	Genotype
VHL	rs5030821	GG
VHL	rs5030818	CC
VHL	rs5030820	CC
VHL	rs119103277	GG
VHL	rs104893824	TT
VHL	rs5030809	TT
VHL	rs104893825	GG
VHL	rs28940297	TT
VHL	rs5030827	GG
VHL	rs5030808	GG
VHL	rs267607170	AA
VHL	rs193922608	CC
VHL	rs193922609	GG
VHL	rs193922610	CC
VHL	rs193922613	AA
VHL	rs143985153	AA
VHL	rs5030826	CC
VHL	rs5030802	GG
VHL	rs397516440	CC
VHL	rs397516441	AA
VHL	rs5030817	GG
VHL	rs397516444	GG
VHL	rs397516445	TT
VHL	rs5030804	AA
VHL	rs398123481	CC
VHL	rs587780077	GG
VHL	rs5030829	GG
VHL	rs727504215	GG
VHL	rs730882034	CC
VHL	rs730882032	GG
VHL	rs121913346	TT

Hereditary Diseases (genetics)

Von Willebrand disease type 1

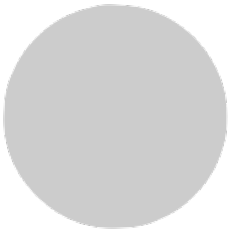
A form of von Willebrand disease (VWD) characterized by a bleeding disorder associated with a partial, quantitative plasmatic deficiency of an otherwise structurally and functionally normal von Willebrand factor (VWF).

Your genetic map

Gene	SNP	Genotype
VWF	rs41276738	CC
VWF	rs61751286	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=166078

Hereditary Diseases (genetics)

Von Willebrand disease type 2A

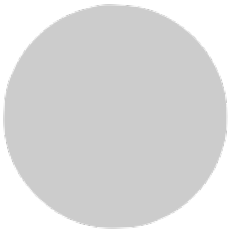
A subtype of type 2 von Willebrand disease characterized by a bleeding disorder associated with a decrease in the affinity of the Willebrand factor (VWF) for platelets and the subendothelium caused by a deficiency of high molecular weight VWF multimers. The disease manifests as mucocutaneous bleeding (menorrhagia, epistaxis, gastrointestinal hemorrhage, etc.).

Your genetic map

Gene	SNP	Genotype
VWF	rs61749397	CC
VWF	rs61750074	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=166084

Hereditary Diseases (genetics)

Von Willebrand disease type 3

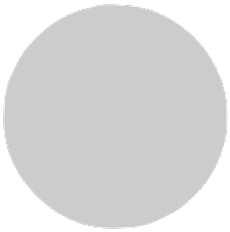
A form of von Willebrand disease (VWD) characterized by a bleeding disorder associated with a total or near-total absence of Willebrand factor (VWF) in the plasma and cellular compartments, also leading to a profound deficiency of plasmatic factor VIII (FVIII). It is the most severe form of VWD.

Your genetic map

Gene	SNP	Genotype
VWF	rs61751296	GG
VWF	rs2363337	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=166096

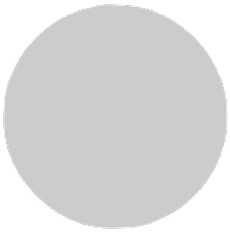
Hereditary Diseases (genetics)

Wilson disease

A rare genetic disorder of copper metabolism presenting with non-specific hepatic, neurologic, psychiatric or ophthalmologic manifestations due to impaired biliary copper excretion and consecutive excessive copper deposition in the body.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=905

Your genetic map

Gene	SNP	Genotype
ALG11	rs369488210	TT
ATP7B	rs76151636	GG
ATP7B	rs121907992	CC
ATP7B	rs28942074	CC
ATP7B	rs28942075	CC
ATP7B	rs28942076	CC
ATP7B	rs121907993	GG
ATP7B	rs121907990	TT
ATP7B	rs121907996	CC
ATP7B	rs121907997	GG
ATP7B	rs60431989	AA
ATP7B	rs121907999	GG
ATP7B	rs121908000	AA
ATP7B	rs121908001	CC
ATP7B	rs137853279	CC
ATP7B	rs193922102	AA
ATP7B	rs193922103	TT
ATP7B	rs72552255	GG
ATP7B	rs193922107	GG
ATP7B	rs193922109	GG
ATP7B	rs193922110	CC
ATP7B	rs398123137	AA
ATP7B	rs137853285	CC
ATP7B	rs137853284	GG
ATP7B	rs137853283	CC
ATP7B	rs201738967	TT
ATP7B	rs587783306	CC
ATP7B	rs587783307	TT
ATP7B	rs587783317	CC
ATP7B	rs776848753	GG
ATP7B	rs786204578	GG

Hereditary Diseases (genetics)

Fatal mitochondrial disease due to combined oxidative phosphorylation defect type 3

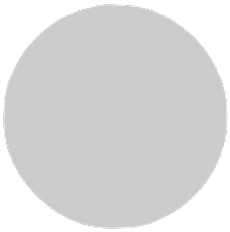
Combined oxidative phosphorylation deficiency type 3 is an extremely rare clinically heterogenous disorder described in about 5 patients to date. Clinical signs included hypotonia, lactic acidosis, and hepatic insufficiency, with progressive encephalomyopathy or hypertrophic cardiomyopathy.

Your genetic map

Gene	SNP	Genotype
TSFM	rs121909485	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=168566

Hereditary Diseases (genetics)

Rippling muscle disease

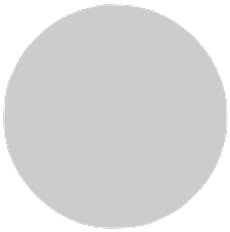
Rippling muscle disease is a rare, genetic, neuromuscular disorder characterized by muscle hyperirritability triggered by stretch, percussion or movement. Patients present wave-like, electrically-silent muscle contractions (rippling), muscle mounding, painful muscle stiffness and muscle hypertrophy, usually with elevated serum creatine kinase.

Your genetic map

Gene	SNP	Genotype
SSUH2	rs116840773	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=97238

Hereditary Diseases (genetics)

Muscle-eye-brain disease

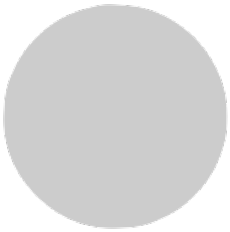
A rare, congenital muscular dystrophy due to dystroglycanopathy characterized by early onset muscular dystrophy, severe muscular hypotonia, severe mental retardation and typical brain and eye malformations, including pachygyria, polymicrogyria, agyria, brainstem and cerebellar structural anomalies, severe myopia, glaucoma, optic nerve and retinal hypoplasia. Patients may present with seizures, macrocephaly or microcephaly, microphthalmia, and congenital contractures. Depending on the severity, limited motor function is acquired. Less severe cases have been reported.

Your genetic map

Gene	SNP	Genotype
FKRP	rs104894680	CC
FKRP	rs121908110	AA
FKTN	rs377417974	CC
POMT1	rs119462985	CC
POMT1	rs119462987	GG
POMT1	rs794727208	CC
POMT1	rs149682171	CC
POMT1	rs138902646	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=588

Hereditary Diseases (genetics)

Åland Islands eye disease

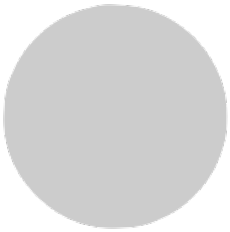
An X-linked recessive retinal disease characterized by fundus hypopigmentation, decrased visual acuity, nystagmus, astigmatism, progressive axial myopia, defective dark adaptation and protanopia.

Your genetic map

Gene	SNP	Genotype
CACNA1F	rs797044676	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=178333

Hereditary Diseases (genetics)

Glycogen storage disease due to LAMP-2 deficiency

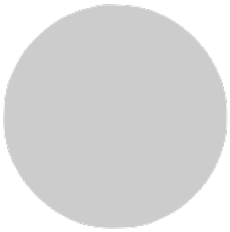
Glycogen storage disease due to LAMP-2 (Lysosomal-Associated Membrane Protein 2) deficiency is a lysosomal glycogen storage disease characterised by severe cardiomyopathy and variable degrees of muscle weakness, frequently associated with intellectual deficit.

Your genetic map

Gene	SNP	Genotype
LAMP2	rs104894858	CC
LAMP2	rs397516740	CC
LAMP2	rs397516743	TT
LAMP2	rs727504742	CC
LAMP2	rs727503118	GG
LAMP2	rs727503120	CC
LAMP2	rs727503119	CC
LAMP2	rs730880485	AA
LAMP2	rs730880483	GG
LAMP2	rs730880496	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=34587

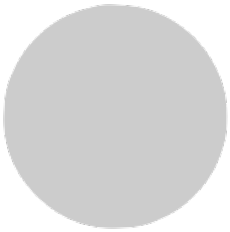
Hereditary Diseases (genetics)

Glycogen storage disease due to glucose-6-phosphatase deficiency

Glycogenosis due to glucose-6-phosphatase (G6P) deficiency or glycogen storage disease, (GSD), type 1, is a group of inherited metabolic diseases, including types a and b (see these terms), and characterized by poor tolerance to fasting, growth retardation and hepatomegaly resulting from accumulation of glycogen and fat in the liver.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=364

Your genetic map

Gene	SNP	Genotype
G6PC1	rs1801175	CC
G6PC1	rs104894563	CC
G6PC1	rs80356487	CC
G6PC1	rs104894566	TT
G6PC1	rs80356484	GG
G6PC1	rs104894565	AA
G6PC1	rs104894567	GG
G6PC1	rs80356482	GG
G6PC1	rs1801176	GG
G6PC1	rs80356485	CC
G6PC1	rs80356483	GG
G6PC1	rs387906505	TT
G6PC1	rs780226142	CC
G6PC1	rs863224023	GG

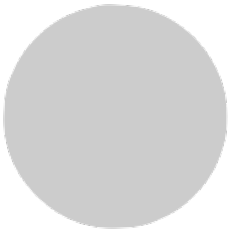
Hereditary Diseases (genetics)

Glycogen storage disease due to acid maltase deficiency

A rare lysosomal storage disease characterized by lysosomal accumulation of glycogen particularly in skeletal, cardiac, and respiratory muscles, as well as the liver and nervous system, due to acid maltase deficiency. The clinical spectrum comprises infantile-onset disease with severe hypertrophic cardiomyopathy, generalized muscle weakness, poor feeding and failure to thrive, and respiratory insufficiency, and late-onset disease manifesting before or after twelve months of age without cardiomyopathy, with proximal muscle weakness and respiratory insufficiency.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=365

Your genetic map

Gene	SNP	Genotype
GAA	rs121907937	GG
GAA	rs28937909	GG
GAA	rs121907938	CC
GAA	rs28940868	CC
GAA	rs121907942	CC
GAA	rs121907943	CC
GAA	rs398123169	GG
GAA	rs369532274	CC
GAA	rs398123174	TT
GAA	rs370950728	GG
GAA	rs140826989	GG
GAA	rs374143224	GG
GAA	rs1800312	GG
GAA	rs779556619	TT
GAA	rs142752477	GG

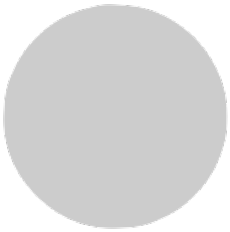
Hereditary Diseases (genetics)

Autosomal recessive polycystic kidney disease

A rare, genetic hepatorenal fibrocystic syndrome characterized by cystic dilatation and ectasia of renal collecting tubules, and a ductal plate malformation of the liver resulting in congenital hepatic fibrosis. Clinical presentation, whilst typically in utero or at birth, is variable and in the most severe cases includes Potter-sequence, oligohydramnios, pulmonary hypoplasia, and massively enlarged echogenic kidneys.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=731

Your genetic map

Gene	SNP	Genotype
LOC1053	rs148617572	GG
LOC1053	rs201082169	GG
PKHD1	rs398124476	CC
PKHD1	rs398124478	GG
PKHD1	rs398124480	GG
PKHD1	rs398124503	GG
PKHD1	rs146649803	CC
PKHD1	rs727504089	GG
PKHD1	rs786204688	GG
PKHD1	rs773136605	CC
PKHD1	rs794727566	AA
PKHD1	rs748365248	CC
PKHD1	rs180675584	CC
PKHD1	rs369925690	TT
PKHD1	rs759851475	CC
PKHD1	rs1240212722	TT

Hereditary Diseases (genetics)

Autosomal dominant generalized dystrophic epidermolysis bullosa

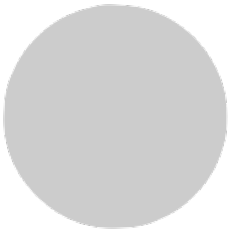
A rare dystrophic epidermolysis bullosa (DEB) characterized by generalized blistering, milia formation, atrophic scarring, and dystrophic nails.

Your genetic map

Gene	SNP	Genotype
COL7A1	rs121912836	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=231568

Hereditary Diseases (genetics)

Recessive dystrophic epidermolysis bullosa inversa

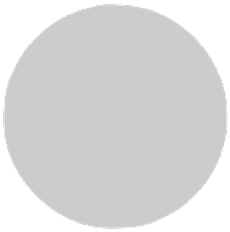
A rare subtype of dystrophic epidermolysis bullosa (DEB) characterized by blisters and erosions which from adolescence or early adulthood are primarily confined to flexural skin sites.

Your genetic map

Gene	SNP	Genotype
COL7A1	rs121912839	CC
COL7A1	rs121912847	GG
COL7A1	rs121912849	GG
COL7A1	rs121912852	GG
COL7A1	rs121912854	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79409

Hereditary Diseases (genetics)

Dystrophic epidermolysis bullosa pruriginosa

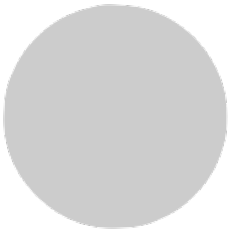
A rare dystrophic epidermolysis bullosa (DEB) characterized by generalized or localized skin lesions associated with severe, if not intractable, pruritus.

Your genetic map

Gene	SNP	Genotype
COL7A1	rs121912855	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=89843

Hereditary Diseases (genetics)

Junctional epidermolysis bullosa with pyloric atresia

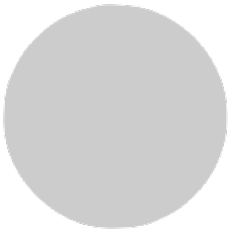
A severe form of junctional epidermolysis bullosa (JEB) characterized by generalized blistering at birth and congenital atresia of the pylorus and rarely of other portions of the gastrointestinal tract.

Your genetic map

Gene	SNP	Genotype
ITGB4	rs80338755	GG
ITGB4	rs147222357	GG
ITGB4	rs121912467	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79403

Hereditary Diseases (genetics)

Intermediate epidermolysis bullosa simplex with cardiomyopathy

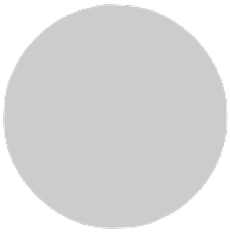
A rare, inherited, epidermolysis bullosa characterized by aplasia cutis congenita on the extremities, leaving behind hypopigmentation and atrophy in a whirled pattern. Generalized blistering persists during childhood and heals with cutaneous and follicular atrophy, linear and stellate scars, and hypopigmentation. Skin fragility decreases with adulthood. Adult patients exhibit dyspigmentation and atrophy of the skin, scars, follicular atrophoderma, sparse body hair, progressive diffuse alopecia of the scalp, diffuse palmoplantar keratoderma, and nail changes. Dilative cardiomyopathy with heart failure complicates the disease course in young adulthood or later and may have lethal outcome. Ultra-structurally, intraepidermal splitting appears at the level of the basal keratinocytes, above the hemidesmosomes.

Your genetic map

Gene	SNP	Genotype
KLHL24	rs886037957	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=508529

Hereditary Diseases (genetics)

Autosomal dominant generalized epidermolysis bullosa simplex, severe form

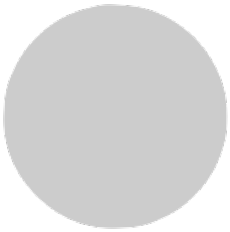
Epidermolysis bullosa simplex, Dowling-Meara type (EBS-DM) is a basal subtype of epidermolysis bullosa simplex (EBS, see this term) characterized by the presence of generalized vesicles and small blisters in grouped or arcuate configuration.

Your genetic map

Gene	SNP	Genotype
KRT14	rs60399023	GG
KRT14	rs58330629	CC
KRT14	rs60171927	TT
KRT14	rs61027685	CC
KRT5	rs57599352	AA
KRT5	rs59115483	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79396

Hereditary Diseases (genetics)

Autosomal dominant generalized epidermolysis bullosa simplex, intermediate form

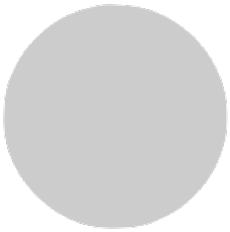
Non-Dowling-Meara generalized epidermolysis bullosa simplex, formerly known as epidermolysis bullosa simplex, Koebner type (EBS-K) is a generalized basal subtype of epidermolysis bullosa simplex (EBS, see this term) characterized by non-herpetiform blisters and erosions arising in particular at sites of friction.

Your genetic map

Gene	SNP	Genotype
KLHL24	rs886037957	GG
KLHL24	rs886037956	AA
KRT14	rs58380626	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79399

Hereditary Diseases (genetics)

Autosomal dominant epilepsy with auditory features

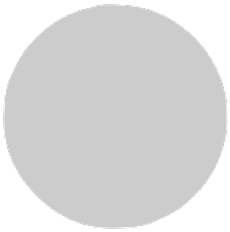
A rare, genetic, familial partial epilepsy disease characterized by focal seizures associated with prominent ictal auditory symptoms, and/or receptive aphasia, presenting in two or more family members and having a relatively benign evolution.

Your genetic map

Gene	SNP	Genotype
LOC1053	rs119488099	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=101046

Hereditary Diseases (genetics)

Juvenile myoclonic epilepsy

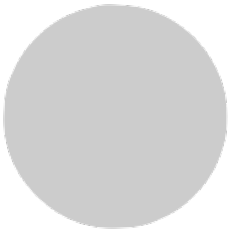
Juvenile myoclonic epilepsy is the most common hereditary idiopathic generalized epilepsy syndrome and is characterized by myoclonic jerks of the upper limbs on awakening, generalized tonic-clonic seizures manifesting during adolescence and triggered by sleep deprivation, alcohol intake, and cognitive activities, and typical absence seizures (30% of cases).

Your genetic map

Gene	SNP	Genotype
EFHC1	rs796052414	CC
GABRA1	rs796052488	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=307

Hereditary Diseases (genetics)

Progressive myoclonic epilepsy type 6

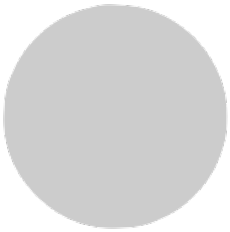
A rare, genetic, neurological disorder characterized by early-onset, progressive ataxia associated with myoclonic seizures (frequently associated with other seizure types such as generalized tonic-clonic, absence and drop attacks), scoliosis of variable severity, areflexia, elevated creatine kinase serum levels, and relative preservation of cognitive function until late in the disease course.

Your genetic map

Gene	SNP	Genotype
GOSR2	rs387906881	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=280620

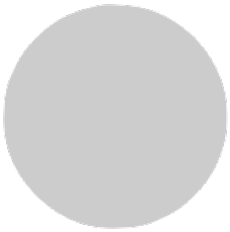
Hereditary Diseases (genetics)

Benign familial neonatal epilepsy

Benign familial neonatal epilepsy (BFNE) is a rare genetic epilepsy syndrome characterized by the occurrence of afebrile seizures in otherwise healthy newborns with onset in the first few days of life.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1949

Your genetic map

Gene	SNP	Genotype
KCNQ2	rs118192226	GG
KCNQ2	rs118192208	CC
KCNQ2	rs118192216	CC
KCNQ2	rs796052619	GG
KCNQ2	rs1057516121	CC
KCNQ2	rs118192194	GG
KCNQ2	rs796052615	TT
KCNQ2	rs864321712	GG
KCNQ3	rs796052678	GG
KCNQ3	rs796052675	GG
LOC1053	rs118192234	CC
LOC1053	rs118192235	CC
LOC1053	rs759584387	GG
LOC1053	rs796052650	GG
LOC1053	rs1057516123	TT

Hereditary Diseases (genetics)

Multiple self-healing squamous epithelioma

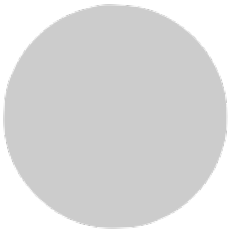
Multiple self-healing squamous epithelioma (also known as Ferguson-Smith disease (FSD)) is a rare inherited skin cancer syndrome characterized by the development of multiple locally invasive skin tumors resembling keratoacanthomas of the face and limbs which usually heal spontaneously after several months leaving pitted scars.

Your genetic map

Gene	SNP	Genotype
TGFBR1	rs387906697	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=65748

Hereditary Diseases (genetics)

Chuvash erythrocytosis

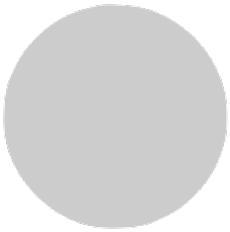
Chuvash erythrocytosis is a rare, genetic, congenital secondary polycythemia disorder characterized by increased hemoglobin, hematocrit and erythropoietin serum levels and normal oxygen affinity, which usually manifests with headache, dizziness, dyspnea and/or plethora. Patients present an increased risk of hemorrhage, thrombosis and early death.

Your genetic map

Gene	SNP	Genotype
VHL	rs104893830	GG
VHL	rs28940301	CC
VHL	rs869025636	GG
VHL	rs5030812	AA
VHL	rs786202787	AA
VHL	rs28940297	TT
VHL	rs5030821	GG
VHL	rs5030818	CC
VHL	rs1352275281	GG
VHL	rs869025622	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=238557

Hereditary Diseases (genetics)

Supravalvular aortic stenosis

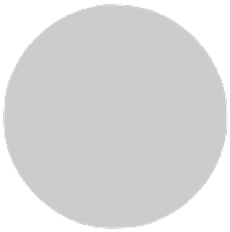
A rare aortic malformation characterized by the narrowing of the aorta lumen (close to its origin) associated or not with stenosis of other arteries (branch pulmonary arteries, coronary arteries). This narrowing of the aorta or pulmonary branches may impede blood flow, resulting in heart murmur and ventricular hypertrophy (left ventricle in case of aorta involvement, right ventricle in case of pulmonary artery involvement).

Your genetic map

Gene	SNP	Genotype
ELN	rs137854452	CC
ELN	rs397516433	CC
ELN	rs727503027	AA
ELN	rs727503029	GG
ELN	rs863223518	TT
ELN	rs200862792	GG
ELN AS1	rs137854453	CC
ELN AS1	rs727503033	TT
ELN AS1	rs727503035	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3193

Hereditary Diseases (genetics)

Dehydrated hereditary stomatocytosis

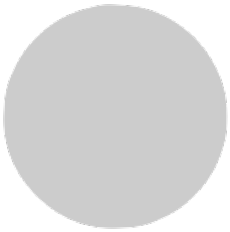
Dehydrated hereditary stomatocytosis (DHS) is a rare hemolytic anemia characterized by a decreased red cell osmotic fragility due to a defect in cation permeability, resulting in red cell dehydration and mild to moderate compensated hemolysis. Pseudohyperkalemia (loss of potassium ions from red cells on storage at room temperature) is sometimes observed.

Your genetic map

Gene	SNP	Genotype
PIEZO1	rs587776989	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3202

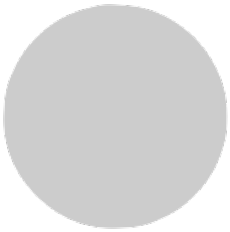
Hereditary Diseases (genetics)

Phenylketonuria

A rare inborn error of amino acid metabolism characterized by elevated blood phenylalanine and low levels or absence of phenylalanine hydroxylase enzyme. If not detected early or left untreated, the disorder manifests with mild to severe mental disability.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=716

Your genetic map

Gene	SNP	Genotype
PAH	rs5030861	CC
PAH	rs5030858	GG
PAH	rs62642936	AA
PAH	rs62508698	CC
PAH	rs76296470	GG
PAH	rs5030849	CC
PAH	rs62516151	GG
PAH	rs5030847	GG
PAH	rs62514891	TT
PAH	rs5030843	CC
PAH	rs5030846	GG
PAH	rs5030851	GG
PAH	rs62514927	TT
PAH	rs62508588	CC
PAH	rs79931499	CC
PAH	rs5030860	TT
PAH	rs62514907	CC
PAH	rs62516095	GG
PAH	rs62514952	CC
PAH	rs62514953	GG
PAH	rs5030852	CC
PAH	rs118203921	GG
PAH	rs78655458	AA
PAH	rs62642926	GG
PAH	rs5030855	CC
PAH	rs5030841	AA
PAH	rs62514934	TT
PAH	rs5030850	GG
PAH	rs5030859	CC
PAH	rs62642933	AA
PAH	rs62508646	AA

Hereditary Diseases (genetics)

Familial atrial fibrillation

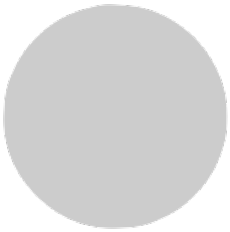
Familial atrial fibrillation is a rare, genetically heterogenous cardiac disease characterized by erratic activation of the atria with an irregular ventricular response, in various members of a single family. It may be asymptomatic or associated with palpitations, dyspnea and light-headedness. Concomitant rhythm disorders and cardiomyopathies are frequently reported.

Your genetic map

Gene	SNP	Genotype
KCNQ1	rs199472705	TT

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=334

Hereditary Diseases (genetics)

Idiopathic ventricular fibrillation, non Brugada type

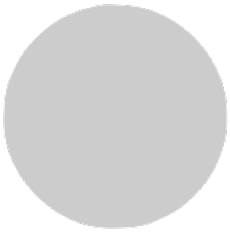
A rare, genetic, cardiac rhythm disease characterized by ventricular fibrillation in the absence of any structural or functional heart disease, or known repolarization abnormalities. The presence of J waves is associated with a higher risk of nocturnal ventricular fibrillation events and a higher risk of recurrence.

Your genetic map

Gene	SNP	Genotype
CACNA1	rs587782933	GG
SCN5A	rs137854604	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=228140

Hereditary Diseases (genetics)

Congenital fibrosis of extraocular muscles

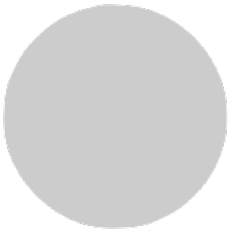
A rare syndromic disorder with strabismus characterized by congenital non-progressive ophthalmoplegia affecting the oculomotor and/or trochlear nucleus/nerve and their innervated muscles. Patients present with abnormal resting position of the eyes (in most cases infraducted and exotropic), limitation of vertical and horizontal gaze, impaired binocular vision, amblyopia, unilateral or bilateral blepharoptosis, and compensatory abnormal head posture. Extraocular manifestations include intellectual disability, peripheral neuropathy, and skeletal abnormalities, among others.

Your genetic map

Gene	SNP	Genotype
KIF21A	rs121912585	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=45358

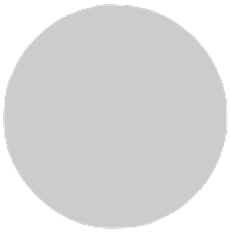
Hereditary Diseases (genetics)

Cystic fibrosis

A rare, genetic pulmonary disorder characterized by sweat, thick mucus secretions causing multisystem disease, chronic infections of the lungs, bulky diarrhea and short stature.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=586

Your genetic map

Gene	SNP	Genotype
CFTR	rs113993958	GG
CFTR	rs78655421	GG
CFTR	rs77932196	GG
CFTR	rs76713772	GG
CFTR	rs80055610	GG
CFTR	rs121908755	GG
CFTR	rs121909005	TT
CFTR	rs121908758	CC
CFTR	rs75527207	GG
CFTR	rs74597325	CC
CFTR	rs75549581	GG
CFTR	rs76649725	CC
CFTR	rs267606722	GG
CFTR	rs77010898	GG
CFTR	rs80034486	CC
CFTR	rs74767530	CC
CFTR	rs387906362	AA
CFTR	rs121909011	CC
CFTR	rs121909012	CC
CFTR	rs121909013	GG
CFTR	rs75961395	GG
CFTR	rs79850223	CC
CFTR	rs121909019	GG
CFTR	rs143570767	GG
CFTR	rs78194216	CC
CFTR	rs75039782	CC
CFTR	rs77902683	GG
CFTR	rs121908748	GG
CFTR	rs141158996	GG
CFTR	rs121908766	CC
CFTR	rs387906369	GG

Hereditary Diseases (genetics)

Phocomelia, Schinzel type

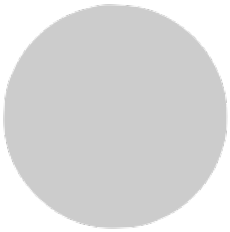
Schinzel phocomelia syndrome, also called limb/pelvis hypoplasia/aplasia syndrome, is characterized by skeletal malformations affecting the ulnae, pelvic bones, fibulae and femora. As the phenotype is similar to that described in the malformation syndrome known as Al-Awadi/Raas-Rothschild syndrome, they are thought to be the same disorder.

Your genetic map

Gene	SNP	Genotype
WNT7A	rs387907231	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2879

Hereditary Diseases (genetics)

Symptomatic form of hemochromatosis type 1

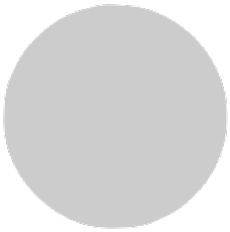
Symptomatic form of hemochromatosis type 1 is a rare, hereditary hemochromatosis characterized by inappropriately regulated intestinal iron absorption which leads to excessive iron storage in various organs and manifests with a wide range of signs and symptoms, including abdominal pain, weakness, lethargy, weight loss, elevated serum aminotransferase levels, increase in skin pigmentation, and/or arthropathy in the metacarpophalangeal joints. Other commonly associated manifestations include hepatomegaly, cirrhosis, liver fibrosis, hepatocellular carcinoma, restrictive cardiomyopathy and/or diabetes mellitus.

Your genetic map

Gene	SNP	Genotype
HFE AS1	rs146519482	GG
TFR2	rs786204108	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=465508

Hereditary Diseases (genetics)

Fucosidosis

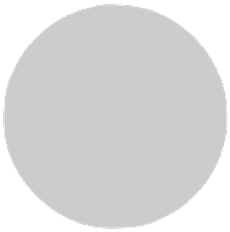
Fucosidosis is an extremely rare lysosomal storage disorder characterized by a highly variable phenotype with common manifestations including neurologic deterioration, coarse facial features, growth retardation, and recurrent sinopulmonary infections, as well as seizures, visceromegaly, angiokeratoma and dysostosis.

Your genetic map

Gene	SNP	Genotype
FUCA1	rs794727774	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=349

Hereditary Diseases (genetics)

Fundus albipunctatus

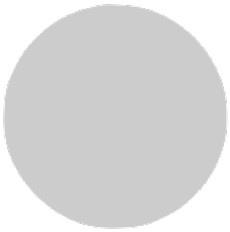
Fundus albipunctatus is a rare, genetic retinal dystrophy disorder characterized by the presence of numerous small, round, yellowish-white retinal lesions that are distributed throughout the retina but spare the fovea. Patients present in childhood with non-progressive night blindness with prolonged cone and rod adaptation times. The macula may or may not be involved, which may result in a decrease of central visual acuity with age.

Your genetic map

Gene	SNP	Genotype
BLOC1S1	rs62638191	GG
BLOC1S1	rs62638193	GG
BLOC1S1	rs774122562	GG
RLBP1	rs137853290	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=227796

Hereditary Diseases (genetics)

GM1 gangliosidosis

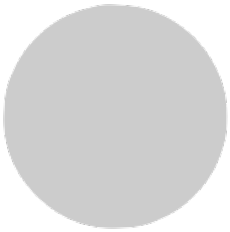
GM1 gangliosidosis is a rare lysosomal storage disorder characterized biochemically by deficient beta-galactosidase activity and clinically by a wide range of variable neurovisceral, ophthalmological and dysmorphic features.

Your genetic map

Gene	SNP	Genotype
GLB1	rs28934274	CC
GLB1	rs72555366	GG
GLB1	rs72555392	CC
GLB1	rs192732174	GG
GLB1	rs794727165	GG
LOC1079	rs72555391	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=354

Hereditary Diseases (genetics)

MOGS-CDG

MOGS-CDG is a form of congenital disorders of N-linked glycosylation characterized by generalized hypotonia, craniofacial dysmorphism (prominent occiput, short palpebral fissures, long eyelashes, broad nose, high arched palate , retrognathia), hypoplastic genitalia, seizures, feeding difficulties, hypoventilation, severe hypogammaglobulinemia with generalized edema, and increased resistance to particular viral infections (particularly to enveloped viruses). The disease is caused by loss-of-function mutations in the gene MOGS (2p13.1).

Your genetic map

Gene	SNP	Genotype
MOGS	rs587777323	GG

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79330

Hereditary Diseases (genetics)

Juvenile glaucoma

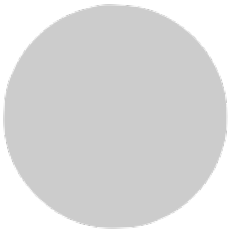
A primary early-onset glaucoma that is characterized by early onset, severe elevation of intra ocular pressure of rapid progression, leading to optic nerve excavation and, when untreated, substantial visual impairment.

Your genetic map

Gene	SNP	Genotype
MYOC	rs74315330	GG
MYOC	rs74315329	GG
MYOC	rs74315334	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=98977

Hereditary Diseases (genetics)

Hawkinsinuria

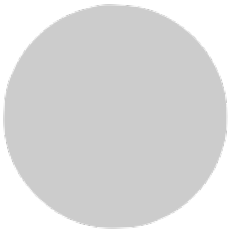
Hawkinsinuria is an inborn error of tyrosine metabolism characterized by failure to thrive, persistent metabolic acidosis, fine and sparse hair, and excretion of the unusual cyclic amino acid metabolite, hawkinsin ((2-l-cystein-S-yl, 4-dihydroxycyclohex-5-en-1-yl)acetic acid), in the urine.

Your genetic map

Gene	SNP	Genotype
TIALD	rs367674632	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2118

Hereditary Diseases (genetics)

Hemochromatosis type 2

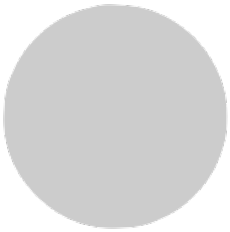
Hemochromatosis type 2 (juvenile) is the early-onset and most severe form of rare hereditary hemochromatosis (HH; see this term), a group of diseases characterized by excessive tissue iron deposition of genetic origin.

Your genetic map

Gene	SNP	Genotype
HJV	rs74315323	GG
HJV	rs28940586	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79230

Hereditary Diseases (genetics)

Mild hemophilia A

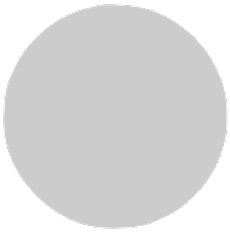
Mild hemophilia A is a form of hemophilia A characterized by a small deficiency of factor VIII leading to abnormal bleeding as a result of minor injuries, or following surgery or tooth extraction.

Your genetic map

Gene	SNP	Genotype
F8	rs137852355	GG
F8	rs28935499	CC
F8	rs137852382	AA
F8	rs137852403	CC
F8	rs137852428	GG
F8	rs137852439	GG
F8	rs137852459	TT
F8	rs137852464	GG
F9	rs137852253	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=169808

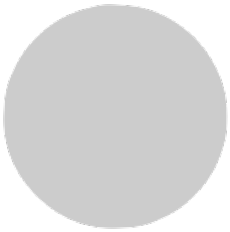
Hereditary Diseases (genetics)

Mild hemophilia B

Mild hemophilia B is a form of hemophilia B characterized by a small deficiency of factor IX leading to abnormal bleeding as a result of minor injuries, or following surgery or tooth extraction.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=169799

Your genetic map

Gene	SNP	Genotype
F8	rs139526001	TT
F9	rs137852227	CC
F9	rs137852228	GG
F9	rs137852232	CC
F9	rs137852233	GG
F9	rs137852237	CC
F9	rs137852238	GG
F9	rs137852248	CC
F9	rs137852240	CC
F9	rs137852241	GG
F9	rs137852249	GG
F9	rs137852250	CC
F9	rs137852254	CC
F9	rs137852257	GG
F9	rs137852258	CC
F9	rs137852259	GG
F9	rs137852261	CC
F9	rs137852268	TT
F9	rs137852271	GG
F9	rs137852272	CC
F9	rs137852275	GG
F9	rs387906481	TT
F9	rs137852247	GG

Hereditary Diseases (genetics)

Paroxysmal nocturnal hemoglobinuria

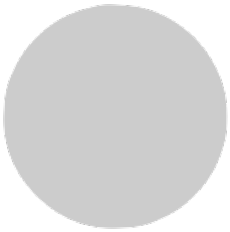
Paroxysmal nocturnal hemoglobinuria (PNH) is an acquired clonal hematopoietic stem cell disorder characterized by corpuscular hemolytic anemia, bone marrow failure and frequent thrombotic events.

Your genetic map

Gene	SNP	Genotype
PIGA	rs199422232	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=447

Hereditary Diseases (genetics)

Hepatoblastoma

A malignant hepatic tumor, typically affecting the pediatric population, arising mostly in an otherwise healthy liver. The most common signs are abdominal distension and abdominal mass. Sometimes patients present with anorexia, weight loss, fatigue. Most HBLs are sporadic, but some cases are associated with genetic factors, especially overgrowth syndromes, such as Beckwith-Wiedemann syndrome (BWS) or hemihypertrophy, and familial adenomatous polyposis (FAP).

Your genetic map

Gene	SNP	Genotype
TP53	rs121912656	CC
TP53	rs397516436	GG
TP53	rs148924904	TT
TP53	rs587782177	CC
TP53	rs876660754	CC
TP53	rs876658468	GG
TP53	rs138729528	GG
TP53	rs1057519975	AA
TP53	rs1057519983	AA
TP53	rs1057520007	TT
TP53	rs530941076	AA
TP53	rs28934874	GG
TP53	rs1057519747	AA

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=449

Hereditary Diseases (genetics)

Hepatoencephalopathy due to combined oxidative phosphorylation defect type 1

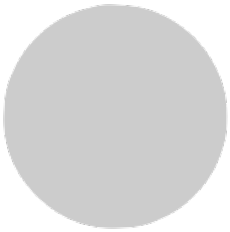
Hepatoencephalopathy due to combined oxidative phosphorylation deficiency type 1 is a rare, inherited mitochondrial disorder due to a defect in mitochondrial protein synthesis characterized by intrauterine growth retardation, metabolic decompensation with recurrent vomiting, persistent severe lactic acidosis, encephalopathy, seizures, failure to thrive, severe global developmental delay, poor eye contact, severe muscular hypotonia or axial hypotonia with limb hypertonia, hepatomegaly and/or liver dysfunction and/or liver failure, leading to fatal outcome in severe cases. Neuroimaging abnormalities may include corpus callosum thinning, leukodystrophy, delayed myelination and basal ganglia involvement.

Your genetic map

Gene	SNP	Genotype
GFM1	rs119470018	AA
GFM1	rs119470019	CC
GFM1	rs139430866	CC
GFM1	rs863224030	GG
GFM1	rs863224032	CC
GFM1	rs201408725	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=137681

Hereditary Diseases (genetics)

Hydrocephalus with stenosis of the aqueduct of Sylvius

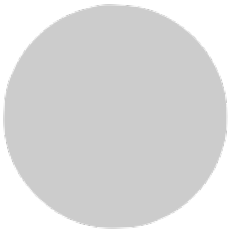
A congenital, X-linked, clinical subtype of L1 syndrome characterized by severe hydrocephalus often of prenatal onset, adducted thumbs, spasticity (mostly evidenced by brisk tendon reflexes and extensor plantar responses) and moderate to severe intellectual disability. This subtype represents the severe end of the L1 syndrome spectrum and is associated with poor prognosis.

Your genetic map

Gene	SNP	Genotype
L1CAM	rs137852520	CC
L1CAM	rs137852522	GG
L1CAM	rs797044787	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2182

Hereditary Diseases (genetics)

Hb Bart's hydrops fetalis

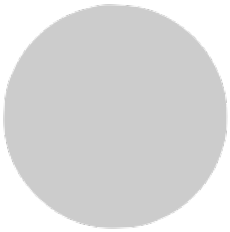
A severe form of alpha-thalassemia that is mostly lethal, and associated with severe long-term outcome and lifelong transfusions in survivors. It is characterized by fetal onset of generalized edema, pleural and pericardial effusions, and severe hypochromic anemia.

Your genetic map

Gene	SNP	Genotype
GUSB	rs786205674	TT
GUSB	rs786205671	CC
GUSB	rs786205673	GG
LOC1027	rs786205667	AA
NEB	rs769345284	GG
THSD1	rs9536062	GG
THSD1	rs786205669	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=163596

Hereditary Diseases (genetics)

Phosphoribosylpyrophosphate synthetase superactivity

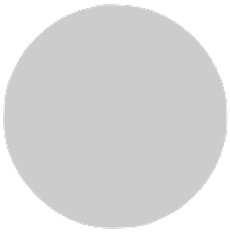
A rare X-linked disorder of purine metabolism associated with hyperuricemia and hyperuricosuria, and comprised of two forms: an early-onset severe form characterized by gout, urolithiasis, and neurodevelopmental anomalies and a mild late-onset form with no neurologic involvement.

Your genetic map

Gene	SNP	Genotype
PRPS1	rs137852540	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3222

Hereditary Diseases (genetics)

Familial hyperaldosteronism type I

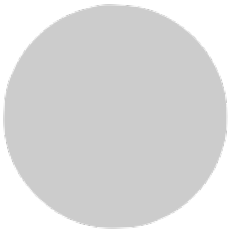
A rare heritable, glucocorticoid remediable form of primary aldosteronism (PA) characterized by early-onset hypertension, hyperaldosteronism, variable hypokalemia, low plasma renin activity (PRA), and abnormal production of 18-oxocortisol and 18-hydroxycortisol.

Your genetic map

Gene	SNP	Genotype
CYP11B1	rs193922538	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=403

Hereditary Diseases (genetics)

Transient familial neonatal hyperbilirubinemia

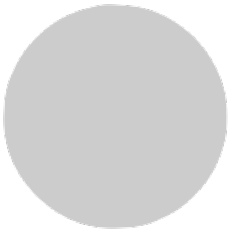
A rare genetic hepatic disease characterized by very high serum bilirubin levels in a newborn, clinically presenting as jaundice during the first few days of life. The condition is usually self-resolving, although in some cases it can lead to kernicterus with corresponding symptoms (including lethargy, high-pitched crying, hypotonia, missing reflexes, vomiting, or seizures, among others), which may result in chronic disability and even death.

Your genetic map

Gene	SNP	Genotype
MROH2A	rs34993780	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2312

Hereditary Diseases (genetics)

Hyperimmunoglobulinemia D with periodic fever

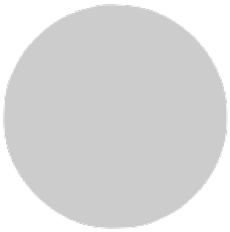
A rare autoinflammatory disease, and form of mevalonate kinase deficiency (MKD), characterized by periodic attacks of fever and a systemic inflammatory reaction (cervical lymphadenopathy, abdominal pain, vomiting, diarrhea, arthralgia and skin manifestations.

Your genetic map

Gene	SNP	Genotype
MVK	rs104895304	TT
MVK	rs104895300	CC
MVK	rs104895360	CC
MVK	rs104895382	TT
MVK	rs104895298	GG
MVK	rs104895311	GG
MVK	rs104895332	TT
MVK	rs104895366	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=343

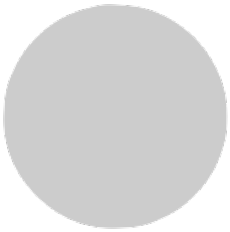
Hereditary Diseases (genetics)

Autosomal dominant hyperinsulinism due to SUR1 deficiency

A form of diazoxide-sensitive diffuse hyperinsulinism (DHI) characterized by hypoglycemic episodes that are usually mild, escaping detection during infancy, and usually present a good clinical response to diazoxide. Autosomal dominant hyperinsulinism due to SUR1 deficiency usually has a milder phenotype when compared to that resulting from recessive K-ATP mutations (recessive forms of Diazoxide-resistant hyperinsulinism).

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=276575

Your genetic map

Gene	SNP	Genotype
ABCC8	rs28936370	CC
ABCC8	rs28938469	GG
ABCC8	rs137852671	CC
ABCC8	rs137852672	AA
ABCC8	rs193922402	GG
ABCC8	rs193922405	CC
ABCC8	rs541269678	GG
ABCC8	rs570388861	GG
ABCC8	rs797045211	CC
ABCC8	rs797045207	CC
ABCC8	rs797045206	AA
ABCC8	rs797045213	TT
ABCC8	rs761749884	CC
ABCC8	rs797045208	AA
ABCC8	rs773306994	CC
ABCC8	rs139328569	GG

Hereditary Diseases (genetics)

Hyperinsulinism due to INSR deficiency

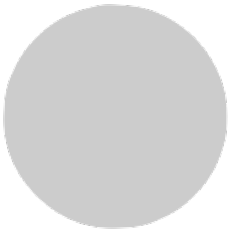
Hyperinsulinemic hypoglycemia due to INSR deficiency is a very rare autosomal dominant form of familial hyperinsulinism characterized clinically in the single reported family by postprandial hypoglycemia, fasting hyperinsulinemia, and an elevated serum insulin-to-C peptide ratio, and a variable age of onset.

Your genetic map

Gene	SNP	Genotype
INSR	rs797045624	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=263458

Hereditary Diseases (genetics)

Endosteal hyperostosis, Worth type

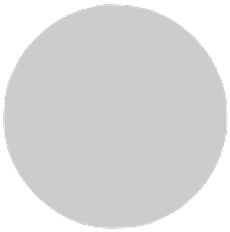
Worth type autosomal dominant osteosclerosis is a sclerozing bone disorder characterized by generalized skeletal densification, particularly of the cranial vault and tubular long bones, which is not associated to an increased risk of fracture.

Your genetic map

Gene	SNP	Genotype
LRP5	rs121908670	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2790

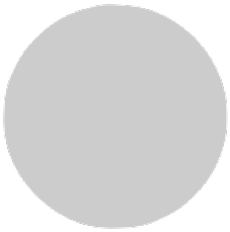
Hereditary Diseases (genetics)

Primary hyperoxaluria

A disorder of glyoxylate metabolism characterized by an excess of oxalate resulting in kidney stones, nephrocalcinosis and ultimately renal failure and systemic oxalosis. There are 3 types of PH, types 1-3, all caused by liver-specific enzyme defects.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=416

Your genetic map

Gene	SNP	Genotype
AGXT	rs121908520	TT
AGXT	rs121908521	CC
AGXT	rs121908522	GG
AGXT	rs121908523	GG
AGXT	rs121908524	TT
AGXT	rs121908525	TT
AGXT	rs121908526	CC
AGXT	rs121908527	GG
AGXT	rs121908530	GG
AGXT	rs121908529	GG
AGXT	rs180177238	CC
AGXT	rs180177157	CC
AGXT	rs180177168	GG
AGXT	rs180177195	TT
AGXT	rs180177197	TT
AGXT	rs180177207	GG
AGXT	rs180177227	GG
AGXT	rs180177253	CC
AGXT	rs180177259	GG
AGXT	rs180177267	GG
AGXT	rs180177156	GG
AGXT	rs180177225	CC
AGXT	rs180177298	GG
AGXT	rs796052064	GG

Hereditary Diseases (genetics)

Familial isolated hyperparathyroidism

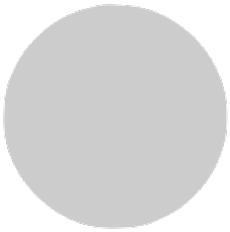
A rare, hereditary, familial primary hyperparathyroidism disease characterized by primary hyperparathyroidism due to single or multiple parathyroid tumors in at least two first-degree relatives in the absence of evidence of other endocrine disorders, tumors and/or systemic manifestations.

Your genetic map

Gene	SNP	Genotype
GCM2	rs104893960	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=99879

Hereditary Diseases (genetics)

Heritable pulmonary arterial hypertension

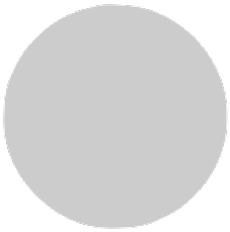
Heritable pulmonary arterial hypertension (HPAH) is a form of pulmonary arterial hypertension (PAH, see this term), occurring due to mutations in PAH predisposing genes or in a familial context. HPAH is characterized by elevated pulmonary arterial resistance leading to right heart failure. HPAH is progressive and potentially fatal.

Your genetic map

Gene	SNP	Genotype
SMAD9	rs397514716	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=275777

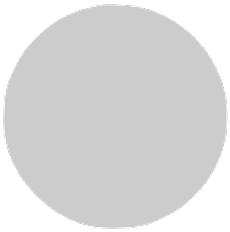
Hereditary Diseases (genetics)

Malignant hyperthermia of anesthesia

Malignant hyperthermia (MH) is a pharmacogenetic disorder of skeletal muscle that presents as a hypermetabolic response to potent volatile anesthetic gases such as halothane, sevoflurane, desflurane and the depolarizing muscle relaxant succinylcholine, and rarely, to stresses such as vigorous exercise and heat.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=423

Your genetic map

Gene	SNP	Genotype
RYR1	rs1801086	GG
RYR1	rs118192161	CC
RYR1	rs121918592	GG
RYR1	rs28933397	CC
RYR1	rs121918594	GG
RYR1	rs118192175	CC
RYR1	rs118192163	GG
RYR1	rs118192177	CC
RYR1	rs121918595	CC
RYR1	rs118192162	AA
RYR1	rs193922747	TT
RYR1	rs193922839	GG
RYR1	rs148399313	GG
RYR1	rs193922843	GG
RYR1	rs193922766	GG
RYR1	rs193922876	CC
RYR1	rs193922878	CC
RYR1	rs193922768	CC
RYR1	rs193922770	CC
RYR1	rs193922772	GG
RYR1	rs193922753	GG
RYR1	rs193922781	CC
RYR1	rs193922757	CC
RYR1	rs112563513	GG
RYR1	rs193922801	AA
RYR1	rs193922802	GG
RYR1	rs193922807	GG
RYR1	rs193922810	GG
RYR1	rs193922816	CC
RYR1	rs193922818	GG
RYR1	rs193922832	GG

Hereditary Diseases (genetics)

Familial hypoaldosteronism

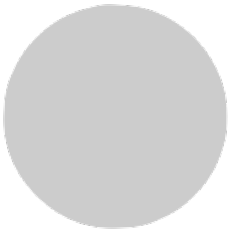
A rare genetic hypoaldosteronism that typically presents in infancy (earl-onset familial hypoaldosternism) as a life-threatening electrolyte imbalance (failure to thrive, recurrent vomiting, and severe dehydration). A history of fever, diarrhoea, lethargy, poor weight gain, poor feeding since birth may also be present. Older subjects (late-onset familial hypoaldosteronism) are less severely affected or asymptomatic.

Your genetic map

Gene	SNP	Genotype
CYP11B2	rs104894072	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=427

Hereditary Diseases (genetics)

Hypochondroplasia

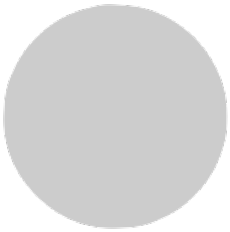
A primary bone dysplasia with micromelia characterized by disproportionate short stature, mild lumbar lordosis and limited extension of the elbow joints.

Your genetic map

Gene	SNP	Genotype
FGFR3	rs77722678	AA
FGFR3	rs121913115	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=429

Hereditary Diseases (genetics)

Hypophosphatasia

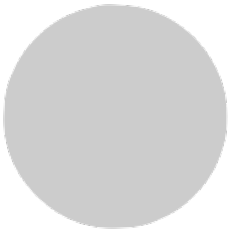
A rare, genetic metabolic disorder characterized by reduced activity of unfractionated serum alkaline phosphatase (ALP) and various symptoms from life-threatening, severely impaired mineralization at birth to musculo-skeletal pain in adulthood.

Your genetic map

Gene	SNP	Genotype
ALPL	rs121918007	GG
ALPL	rs121918008	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=436

Hereditary Diseases (genetics)

X-linked hypophosphatemia

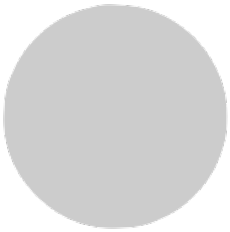
X-linked hypophosphatemia (XLH) is a hereditary renal phosphate-wasting disorder characterized by hypophosphatemia, rickets and/or osteomalacia, and diminished growth.

Your genetic map

Gene	SNP	Genotype
PHEX	rs193922454	TT
PHEX	rs193922455	GG
PHEX	rs193922458	GG
PHEX	rs193922459	GG
PTCHD1	rs193922457	GG
PTCHD1	rs875989883	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=89936

Hereditary Diseases (genetics)

Primary hypomagnesemia with secondary hypocalcemia

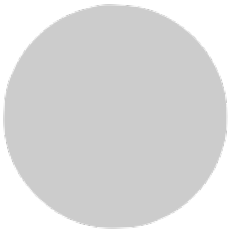
Primary hypomagnesemia with secondary hypocalcemia (PHSH) is a form of familial primary hypomagnesemia (FPH, see this term), characterized by severe hypomagnesemia and secondary hypocalcemia associated with neurological symptoms, including generalized seizures, tetany and muscle spasms. PHSH may be fatal or may result in chronic irreversible neurological complications.

Your genetic map

Gene	SNP	Genotype
TRPM6	rs869025214	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=30924

Hereditary Diseases (genetics)

Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement

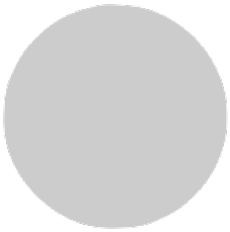
Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement (FHHNCOI) is a form of familial primary hypomagnesemia (FPH, see this term), characterized by excessive magnesium and calcium renal wasting, bilateral nephrocalcinosis, progressive renal failure and severe ocular abnormalities.

Your genetic map

Gene	SNP	Genotype
CLDN19	rs118203979	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2196

Hereditary Diseases (genetics)

Focal dermal hypoplasia

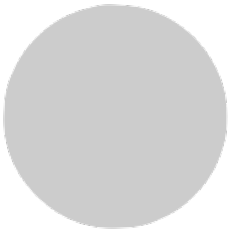
A rare multiple congenital anomalies/dysmorphic syndrome characterized by abnormalities in ectodermal- and mesodermal-derived tissues, classically manifesting with skin abnormalities, limb defects, ocular malformations, and mild facial dysmorphism.

Your genetic map

Gene	SNP	Genotype
PORCN	rs267606973	GG
PORCN	rs137852218	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2092

Hereditary Diseases (genetics)

Pontocerebellar hypoplasia type 10

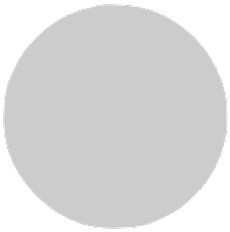
Pontocerebellar hypoplasia type 10 is a rare, genetic, pontocerebellar hypoplasia subtype characterized by severe psychomotor developmental delay, progressive microcephaly, progressive spasticity, seizures, and brain abnormalities consisting of mild atrophy of the cerebellum, pons and corpus callosum and cortical atrophy with delayed myelination. Patients may present dysmorphic facial features (high arched eyebrows, prominent eyes, long palpebral fissures and eyelashes, broad nasal root, and hypoplastic alae nasi) and an axonal sensorimotor neuropathy.

Your genetic map

Gene	SNP	Genotype
CLP1	rs587777616	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=411493

Hereditary Diseases (genetics)

Pontocerebellar hypoplasia type 2

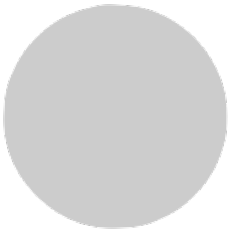
A rare, genetic form of pontocerebellar hypoplasia characterized by pontocerebellar hypoplasia and progressive neocortical atrophy that manifests clinically with uncoordinated sucking and swallowing, and generalized clonus in the neonate. In early childhood, spasticity, chorea/dyskinesia, seizures and progressive microcephaly develop. Voluntary motor development is lacking.

Your genetic map

Gene	SNP	Genotype
TSEN54	rs113994152	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2524

Hereditary Diseases (genetics)

Pontocerebellar hypoplasia type 6

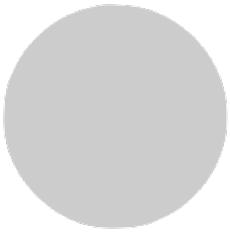
A rare, genetic form of pontocerebellar hypoplasia (PCH) characterized by neocortical and severe cerebral cortical atrophy associated with pontocerebellar hypoplasia with the pons and cerebellum equally affected. Clinically the disorder manifests at birth with hypotonia, clonus, epilepsy impaired swallowing and from infancy by progressive microcephaly, spasticity and lactic acidosis.

Your genetic map

Gene	SNP	Genotype
RARS2	rs199835443	GG
RARS2	rs772887102	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=166073

Hereditary Diseases (genetics)

Pontocerebellar hypoplasia type 8

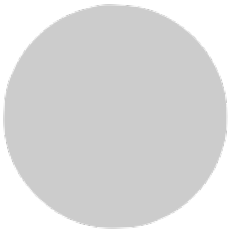
Pontocerebellar hypoplasia type 8 (PCH8) is a novel very rare form of pontocerebellar hypoplasia characterized clinically by progressive microencephaly, feeding difficulties, severe developmental delay, although walking may be achieved, hypotonia often associated with increased muscle tone of lower extremities and deep tendon reflexes, joint deformities in the lower extremities, and occasionally complex seizures. PCH8 is caused by a loss-of-function mutation in the CHMP1A gene. MRI demonstrates a pontocerebellar hypoplasia with vermis and hemispheres equally affected and mild to severely reduced cerebral white matter volume with a fully formed very thin corpus callosum.

Your genetic map

Gene	SNP	Genotype
CHMP1A	rs397515426	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=324569

Hereditary Diseases (genetics)

X-linked adrenal hypoplasia congenita

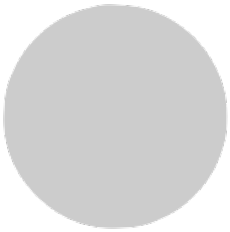
A rare genetic adrenal disease characterized by primary adrenal insufficiency (AI) and/or hypogonadotropic hypogonadism (HH). Male patients typically present with AI with acute onset in infancy or insidious onset in childhood. Clinical features of AI include hyperpigmentation, vomiting, poor feeding, failure to thrive, seizures, vascular collapse, and sometimes sudden death. HH manifests later as delayed or arrested puberty. In rare cases, patients become symptomatic in early adulthood with delayed-onset AI, partial HH, and/or infertility. Histologically, the adrenal glands lack the permanent adult cortical zone. The remaining cells are larger than fetal adrenal cells (''cytomegalic'') and contain characteristic nuclear inclusions.

Your genetic map

Gene	SNP	Genotype
NR0B1	rs104894892	GG
NR0B1	rs104894894	GG
NR0B1	rs132630327	CC
NR0B1	rs386134262	AA
NR0B1	rs386134263	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=95702

Hereditary Diseases (genetics)

Isolated optic nerve hypoplasia/aplasia

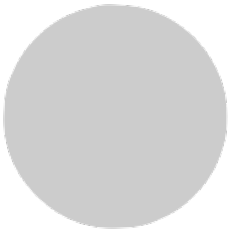
A rare genetic optic nerve disorder characterized by visual impairment or blindness resulting from varying degrees of underdevelopment of the optic nerve or even complete absence of the optic nerve, ganglion cells, and central retinal vessels. It may be unilateral, typically with otherwise normal brain development, or bilateral with accompanying severe and widespread congenital malformations of the central nervous system.

Your genetic map

Gene	SNP	Genotype
PAX6	rs121907924	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=137902

Hereditary Diseases (genetics)

Hypothyroidism due to TSH receptor mutations

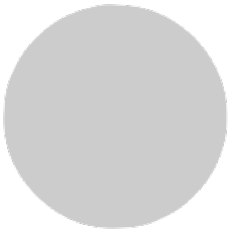
A type of primary congenital hypothyroidism, a permanent thyroid hormone deficiency that is present from birth due to thyroid resistance to TSH.

Your genetic map

Gene	SNP	Genotype
CEP128	rs121908869	GG
LOC1019	rs121908871	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=90673

Hereditary Diseases (genetics)

Hypotonia with lactic acidemia and hyperammonemia

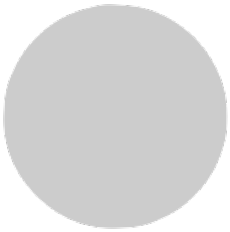
This syndrome is characterised by severe hypotonia, lactic academia and congenital hyperammonaemia.

Your genetic map

Gene	SNP	Genotype
MRPS22	rs119478059	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=137908

Hereditary Diseases (genetics)

Hereditary renal hypouricemia

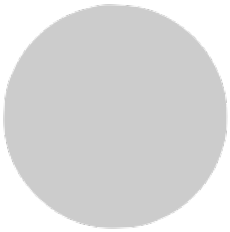
A genetic renal tubular disorder characterized by urinary urate wasting that typically leads to asymptomatic hypouricemia and predisposes to urolithiasis and exercise-induced acute renal failure (EIARF).

Your genetic map

Gene	SNP	Genotype
SLC22A1	rs121907892	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=94088

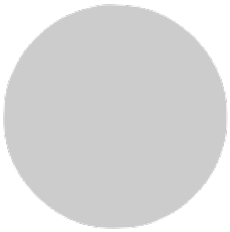
Hereditary Diseases (genetics)

Classic homocystinuria

Classical homocystinuria due to cystathionine beta-synthase (CbS) deficiency is characterized by the multiple involvement of the eye, skeleton, central nervous system, and vascular system.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=394

Your genetic map

Gene	SNP	Genotype
CBS	rs121964962	CC
CBS	rs121964964	GG
CBS	rs121964969	CC
CBS	rs28934891	CC
CBS	rs375846341	TT
CBS	rs398123151	GG
CBS	rs763036586	CC
CBS	rs771298943	CC
CBS	rs770095972	CC
CBS	rs781444670	CC
CBS	rs149119723	GG
CBS	rs863223433	CC
CBS	rs372010465	CC
CBS	rs863223432	CC
CBS	rs775992753	GG
CBS	rs778220779	AA
CBS	rs863223435	CC
CBS	rs148865119	GG
CBS	rs781567152	AA
CBS	rs762065361	CC

Hereditary Diseases (genetics)

Homocystinuria due to methylene tetrahydrofolate reductase deficiency

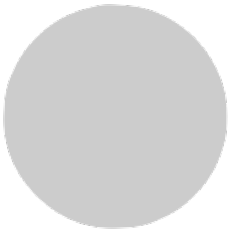
Homocystinuria due to methylene tetrahydrofolate reductase (MTHFR) deficiency is a metabolic disorder characterised by neurological manifestations.

Your genetic map

Gene	SNP	Genotype
MTHFR	rs121434295	CC
MTHFR	rs200137991	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=395

Hereditary Diseases (genetics)

Harlequin ichthyosis

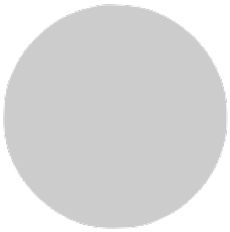
Harlequin ichthyosis (HI) is the most severe variant of autosomal recessive congenital ichthyosis (ARCI; see this term). It is characterized at birth by the presence of large, thick, plate-like scales over the whole body associated with severe ectropion, eclabium, and flattened ears, that later develops into a severe scaling erythroderma.

Your genetic map

Gene	SNP	Genotype
SNHG31	rs137853289	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=457

Hereditary Diseases (genetics)

Autosomal dominant epidermolytic ichthyosis

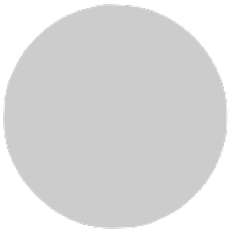
Epidermolytic ichthyosis (EI) is a rare keratinopathic ichthyosis (KPI; see this term), that is characterized by a blistering phenotype at birth which progressively becomes hyperkeratotic.

Your genetic map

Gene	SNP	Genotype
KRT10	rs58075662	CC
KRT10	rs58852768	GG
KRT10	rs58901407	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=312

Hereditary Diseases (genetics)

Exfoliative ichthyosis

Exfoliative ichthyosis is an inherited, non-syndromic, congenital ichthyosis disorder characterized by the infancy-onset of palmoplantar peeling of the skin (aggravated by exposure to water and by occlusion) associated with dry, scaly skin over most of the body. Pruritus and hypohidrosis may also be associated. Well-demarcated areas of denuded skin appear in moist and traumatized regions and skin biopsies reveal reduced cell-cell adhesion in the basal and suprabasal layers, prominent intercellular edema, numerous aggregates of keratin filaments in basal keratinocytes, attenuated cornified cell envelopes, and epidermal barrier impairment.

Your genetic map

Gene	SNP	Genotype
CSTA	rs149474339	CC

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=289586

Hereditary Diseases (genetics)

Lamellar ichthyosis

Lamellar ichthyosis (LI) is a keratinization disorder characterized by the presence of large scales all over the body without significant erythroderma.

Your genetic map

Gene	SNP	Genotype
TGM1	rs121918716	GG
TGM1	rs121918717	CC
TGM1	rs121918718	CC
TGM1	rs121918721	CC
TGM1	rs121918723	CC
TGM1	rs121918725	CC
TGM1	rs121918727	CC
TGM1	rs121918731	GG
TGM1	rs121918732	CC
TGM1	rs143473912	CC
TGM1	rs142634031	TT
TGM1	rs139208806	TT
TGM1	rs140000324	GG

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=313

Hereditary Diseases (genetics)

Recessive X-linked ichthyosis

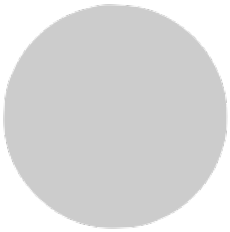
Recessive X-linked ichthyosis (RXLI) is a genodermatosis belonging to the Mendelian Disorders of Cornification (MeDOC) and characterized by generalized hyperkeratosis and scaling of the skin.

Your genetic map

Gene	SNP	Genotype
STS	rs137853167	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=461

Hereditary Diseases (genetics)

Incontinentia pigmenti

An X-linked syndromic muti-systemic ectodermal dysplasia presenting neonatally in females with a bullous rash along Blaschko's lines (BL) followed by verrucous plaques and hyperpigmented swirling patterns. It is further characterized by teeth abnormalities, alopecia, nail dystrophy and can affect the retinal and the central nervous system (CNS) microvasculature. It may have other aspects of ectodermal dysplasia such as sweat gland abnormalities. Germline pathogenic variants in males result in embryonic lethality.

Your genetic map

Gene	SNP	Genotype
IKBKG	rs137853323	CC

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=464

Hereditary Diseases (genetics)

Male infertility due to large-headed multiflagellar polyploid spermatozoa

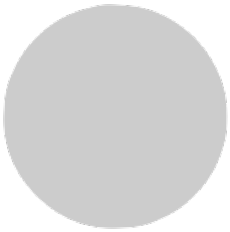
Male infertility due to large-headed multiflagellar polypoid spermatozoa is a male infertility due to sperm disorder characterized by the presence, in sperm, of a very high percentage of spermatozoa with enlarged head, irregular head shape, multiple flagella, and abnormal midpiece and acrosome. It is generally associated with severe oligoasthenozoospermia and a high rate of sperm chromosomal abnormalities (polyploidy, aneuploidy).

Your genetic map

Gene	SNP	Genotype
AURKC	rs55658999	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=137893

Hereditary Diseases (genetics)

Combined immunodeficiency with granulomatosis

A rare, genetic, non-severe combined immunodeficiency disease characterized by immunodeficiency (manifested by recurrent and/or severe bacterial and viral infections), destructive noninfectious granulomas involving skin, mucosa and internal organs, and various autoimmune manifestations (including cytopenias, vitiligo, psoriasis, myasthenia gravis, enteropathy). Immunophenotypically, T-cell and B-cell lymphopenia, hypogammaglobulinemia, abnormal specific antibody production and impaired T-cell function are observed.

Your genetic map

Gene	SNP	Genotype
IFTAP	rs121917894	CC
IFTAP	rs193922574	GG
RAG1	rs121918569	GG
RAG1	rs121918570	CC
RAG1	rs193922461	GG
RAG1	rs193922464	CC

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=157949

Hereditary Diseases (genetics)

Severe combined immunodeficiency due to adenosine deaminase deficiency

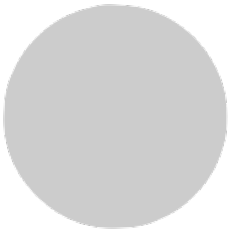
Severe combined immunodeficiency (SCID) due to adenosine deaminase (ADA) deficiency is a form of SCID characterized by profound lymphopenia and very low immunoglobulin levels of all isotypes resulting in severe and recurrent opportunistic infections.

Your genetic map

Gene	SNP	Genotype
ADA	rs121908716	CC
ADA	rs199422327	AA
ADA	rs121908715	GG
ADA	rs121908739	AA
ADA	rs121908723	CC
ADA	rs121908735	GG
ADA	rs121908725	GG
ADA	rs749484894	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=277

Hereditary Diseases (genetics)

Severe combined immunodeficiency due to DCLRE1C deficiency

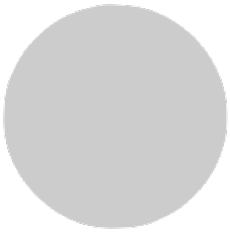
Severe combined immunodeficiency (SCID) due to DCLRE1C deficiency is a type of SCID characterized by severe and recurrent infections, diarrhea, failure to thrive, and cell sensitivity to ionizing radiation.

Your genetic map

Gene	SNP	Genotype
DCLRE1C	rs121908156	GG
DCLRE1C	rs121908157	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=275

Hereditary Diseases (genetics)

T-B+ severe combined immunodeficiency due to gamma chain deficiency

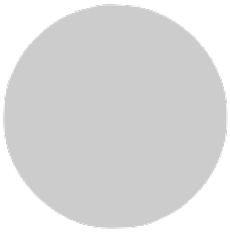
Severe combined immunodeficiency (SCID) due to gamma chain deficiency, also called SCID-X1, is a form of SCID characterized by severe and recurrent infections, associated with diarrhea and failure to thrive.

Your genetic map

Gene	SNP	Genotype
CXorf65	rs111033617	CC
CXorf65	rs137852508	GG
IL2RG	rs193922346	CC
IL2RG	rs193922347	TT
IL2RG	rs193922348	AA
IL2RG	rs193922350	CC
IL2RG	rs869320660	CC
IL2RG	rs869320659	GG
IL2RG	rs869320658	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=276

Hereditary Diseases (genetics)

Combined immunodeficiency due to partial RAG1 deficiency

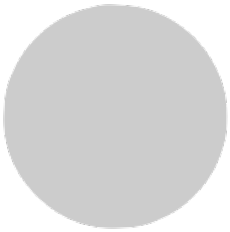
Combined immunodeficiency due to partial RAG1 deficiency is a form of combined T and B cell immunodeficiency (CID; see this term) characterized by severe and persistent cytomegalovirus (CMV) infection and autoimmune cytopenia.

Your genetic map

Gene	SNP	Genotype
RAG1	rs104894287	CC
RAG1	rs141524540	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=231154

Hereditary Diseases (genetics)

Immunodeficiency due to a late component of complement deficiency

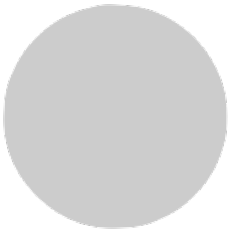
Immunodeficiency due to a late component of complement deficiency is a primary immunodeficiency due to an anomaly in either complement components C5, C6, C7, C8 or C9 and is typically characterized by meningitis due to often recurrent meningococcal infections. The prognosis is generally favorable.

Your genetic map

Gene	SNP	Genotype
C7	rs121964921	GG
C7	rs531103546	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=169150

Hereditary Diseases (genetics)

Immunodeficiency by defective expression of MHC class I

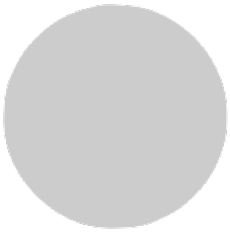
A rare autosomal recessive primary immunodeficiency characterized by severe reduction in the cell surface expression of HLA class I molecules, typically resulting in childhood-onset of chronic bacterial infections of the respiratory tract evolving to widespread bronchiectasis and respiratory insufficiency. Sterile necrotizing granulomatous skin lesions mainly involving the extremities and the mid-face may be observed in some patients. Severe viral infections do not occur as part of the condition. Atypical variants without respiratory or cutaneous manifestations, as well as asymptomatic individuals have been reported.

Your genetic map

Gene	SNP	Genotype
TAP1	rs143800384	GG
TAP2	rs765335850	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=34592

Hereditary Diseases (genetics)

Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins

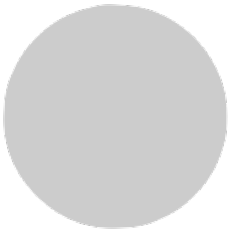
A very rare mitochondrial respiratory chain deficiency characterized clinically by transient but life-threatening liver failure with elevated liver enzymes, jaundice, vomiting, coagulopathy, hyperbilirubinemia, and lactic acidemia.

Your genetic map

Gene	SNP	Genotype
TRMU	rs387907022	GG
TRMU	rs367683258	CC
TRMU	rs766314948	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=217371

Hereditary Diseases (genetics)

Isolated cleft lip

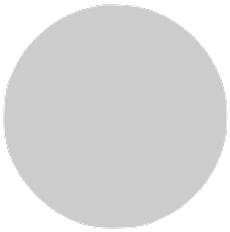
Isolated cleft lip is a fissure type embryopathy extending from the upper lip to the nasal base.

Your genetic map

Gene	SNP	Genotype
TP63	rs121908840	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=199302

Hereditary Diseases (genetics)

Leprechaunism

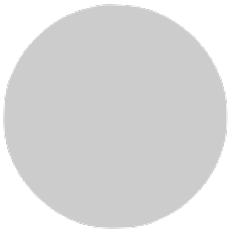
Leprechaunism is a congenital form of extreme insulin resistance (a group of syndromes that also includes Rabson-Mensenhall syndrome, type A insulin-resistance syndrome, and acquired type B insulin-resistance syndrome; see these terms) characterized by intrauterine and mainly postnatal severe growth retardation.

Your genetic map

Gene	SNP	Genotype
INSR	rs121913145	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=508

Hereditary Diseases (genetics)

Acute lymphoblastic leukemia

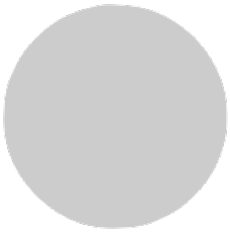
A rare disease characterized by malignant proliferation of lymphoid cells blocked at an early stage of differentiation and accounts for 75% of all cases of childhood leukaemia.

Your genetic map

Gene	SNP	Genotype
JAK1	rs869312953	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=513

Hereditary Diseases (genetics)

B-cell chronic lymphocytic leukemia

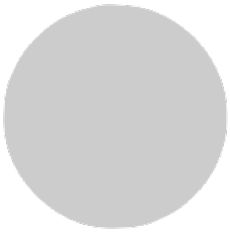
B-cell chronic lymphocytic leukemia (B-CLL) is a type of B-cell non-Hodgkin lymphoma (see this term), and the most common form of leukemia in Western countries, affecting elderly adults (mean age of 67 and 72 years) with a slight male predominance (1.7:1), and characterized by a highly variable clinical presentation that can include asymptomatic disease or non-specific B-symptoms such as unintentional weight loss, severe fatigue, fever (without evidence of infection), and night sweats as well as cervical lymphadenopathy, splenomegaly and frequent infections. Some patients can also develop autoimmune complications such as autoimmune hemolytic anemia or immune thrombocytopenia (see these terms). The clinical course is extremely heterogeneous with survival ranging from a few months to several decades.

Your genetic map

Gene	SNP	Genotype
BRAF	rs121913348	CC
LRRC56	rs104894226	CC
PTPN11	rs121918453	GG
TP53	rs121912651	GG
TP53	rs121913343	GG
TP53	rs587781525	TT
TP53	rs786201838	TT
TP53	rs1057519981	AA
TP53	rs764146326	CC
TP53	rs1057519990	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=67038

Hereditary Diseases (genetics)

Acute myeloid leukemia

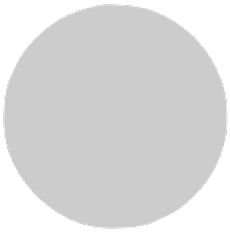
A group of neoplasms arising from precursor cells committed to the myeloid cell-line differentiation. All of them are characterized by clonal expansion of myeloid blasts. They manifest by fever, pallor, anemia, hemorrhages and recurrent infections.

Your genetic map

Gene	SNP	Genotype
NRAS	rs121913250	CC
TERT	rs797046041	GG
TP53	rs587780070	GG
TP53	rs587782082	TT
TP53	rs876660821	AA
TP53	rs1057519747	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=167714

Hereditary Diseases (genetics)

Juvenile myelomonocytic leukemia

A rare myelodysplastic/myeloproliferative neoplasm characterized by a proliferation primarily of granulocytic and monocytic lineages with infiltration of the liver and spleen, among other organs. Blasts and promonocytes account for less than 20% of white blood cells in peripheral blood and bone marrow. Erythroid and megakaryocytic abnormalities are often present. BCR-ABL1 fusion is absent, while somatic mutations in genes of the RAS pathway or monosomy 7 may be found. The condition may also occur in the context of neurofibromatosis type 1 or Noonan syndrome-like disorder. Children of less than three years are predominantly affected, with a clear male preponderance. Most patients present with constitutional symptoms, signs of infection, and hepatosplenomegaly.

Your genetic map

Gene	SNP	Genotype
NRAS	rs121434596	CC
PTPN11	rs121918458	TT
PTPN11	rs121918465	AA
PTPN11	rs397507520	GG
PTPN11	rs397507550	GG
PTPN11	rs397507510	GG

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=86834

Hereditary Diseases (genetics)

RARS-related autosomal recessive hypomyelinating leukodystrophy

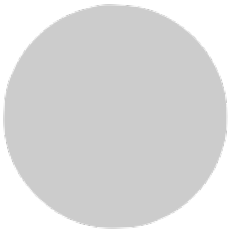
A rare, genetic leukodystrophy characterized by developmental delay, increased muscle tone leading later to spasticity, mild ataxia, nystagmus, dysarthria, intentional tremor, and mild intellectual disability. Brain imaging reveals supratentorial and infratentorial hypomyelination.

Your genetic map

Gene	SNP	Genotype
RARS1	rs672601375	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=438114

Hereditary Diseases (genetics)

Hereditary diffuse leukoencephalopathy with axonal spheroids and pigmented glia

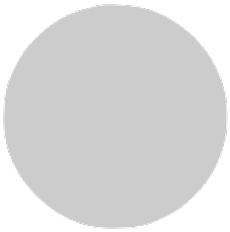
Hereditary diffuse leukoencephalopathy with axonal spheroids and pigmented glia is a rare autosomal dominant disease characterized by a complex phenotype including progressive dementia, apraxia, apathy, impaired balance, parkinsonism, spasticity and epilepsy.

Your genetic map

Gene	SNP	Genotype
CSF1R	rs281860274	AA
CSF1R	rs587777247	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=313808

Hereditary Diseases (genetics)

Lymphangioleiomyomatosis

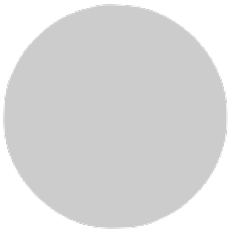
Lymphangioleiomyomatosis (LAM) is a multiple cystic lung disease characterized by progressive cystic destruction of the lung and lymphatic abnormalities, frequently associated with renal angiomyolipomas (AMLs). LAM occurs either sporadically or as a manifestation of tuberous sclerosis complex (TSC).

Your genetic map

Gene	SNP	Genotype
TSC1	rs118203387	CC
TSC2	rs45517403	AA
TSC2	rs1131691965	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=538

Hereditary Diseases (genetics)

Familial partial lipodystrophy, Dunnigan type

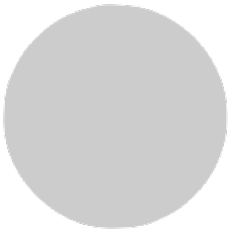
A rare, genetic lipodystrophy characterized by a loss of subcutaneous adipose tissue from the trunk, buttocks and limbs; fat accumulation in the neck, face, axillary and pelvic regions; muscular hypertrophy; and usually associated with metabolic complications such as insulin resistance, diabetes mellitus, dyslipidemia and liver steatosis.

Your genetic map

Gene	SNP	Genotype
LMNA	rs60864230	GG
LMNA	rs57920071	CC
LMNA	rs267607555	CC
LMNA	rs59981161	GG
LMNA	rs267607543	GG
LMNA	rs57629361	CC
LMNA	rs56793579	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2348

Hereditary Diseases (genetics)

Late infantile neuronal ceroid lipofuscinosis

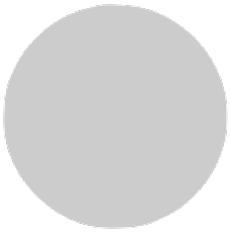
Late infantile neuronal ceroid lipofuscinoses (LINCLs) are a genetically heterogeneous group of neuronal ceroid lipofuscinoses (NCLs; see this term) typically characterized by onset during infancy or early childhood with decline of mental and motor capacities, epilepsy, and vision loss through retinal degeneration.

Your genetic map

Gene	SNP	Genotype
FBXL3	rs121908292	GG
FBXL3	rs386833980	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=168491

Hereditary Diseases (genetics)

ATP13A2-related juvenile neuronal ceroid lipofuscinosis

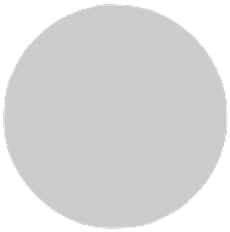
A rare neuronal ceroid lipofiscinosis disorder characterized by juvenile-onset of progressive spinocerebellar ataxia, bulbar syndrome (manifesting with dysarthria, dysphagia and dysphonia), pyramidal and extrapyramidal involvement (including myoclonus, amyotrophy, unsteady gait, akinesia, rigidity, dysarthric speech) and intellectual deterioration. Muscle biopsy displays autofluorescent bodies and lipofuscin deposits in brain and, occasionally the retina, upon post mortem.

Your genetic map

Gene	SNP	Genotype
ATP13A2	rs150519745	CC
ATP13A2	rs758014228	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=314632

Hereditary Diseases (genetics)

X-linked lissencephaly with abnormal genitalia

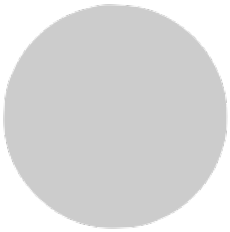
X-linked lissencephaly with abnormal genitalia (XLAG) is a rare, genetic, central nervous system malformation disorder characterized, in males, by lissencephaly (with posterior predominance and moderately thickened cortex), complete absence of corpus callosum, neonatal-onset (mainly perinatal) intractable seizures, postnatal microcephaly, severe hypotonia, poor responsiveness and hypogonadism (micropenis, hypospadias, cryptorchidism, small scrotal sac). Defective temperature regulation and chronic diarrhea may be additionally observed.

Your genetic map

Gene	SNP	Genotype
ARX	rs587783183	AA
ARX	rs587783184	GG
ARX	rs587783189	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=452

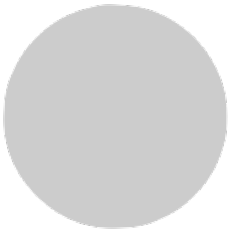
Hereditary Diseases (genetics)

Lissencephaly due to LIS1 mutation

Lissencephaly due to LIS1 mutation is a cerebral malformation with epilepsy characterized predominantly by posterior isolated lissencephaly with developmental delay, intellectual disability and epilepsy that usually evolves from West syndrome to Lennox-Gastaut syndrome. Additional features include muscular hypotonia, acquired microcephaly, failure to thrive and poor control of airways leading to aspiration pneumonia.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=95232

Your genetic map

Gene	SNP	Genotype
DCX	rs104894784	CC
DCX	rs587783592	GG
PAFAH1B	rs121434483	CC
PAFAH1B	rs121434487	GG
PAFAH1B	rs113994203	GG
PAFAH1B	rs113994202	TT
PAFAH1B	rs587784265	GG
PAFAH1B	rs587784260	CC
PAFAH1B	rs587784262	CC
PAFAH1B	rs587784272	TT
PAFAH1B	rs587784250	GG
PAFAH1B	rs587784257	GG
PAFAH1B	rs587784258	CC
PAFAH1B	rs587784261	TT
PAFAH1B	rs587784263	AA
PAFAH1B	rs587784267	CC
PAFAH1B	rs587784269	CC
PAFAH1B	rs587784273	CC
PAFAH1B	rs587784276	GG
PAFAH1B	rs587784281	GG
PAFAH1B	rs587784280	GG
PAFAH1B	rs587784282	CC
PAFAH1B	rs587784286	CC
PAFAH1B	rs587784287	AA
PAFAH1B	rs587784288	TT
PAFAH1B	rs587784291	GG
PAFAH1B	rs587784290	GG
PAFAH1B	rs587784293	CC
PAFAH1B	rs587784294	TT
PAFAH1B	rs587784235	GG
PAFAH1B	rs587784239	GG

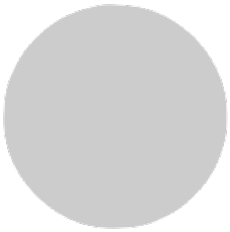
Hereditary Diseases (genetics)

Lissencephaly due to TUBA1A mutation

Lissencephaly (LIS) due to TUBA1A mutation is a congenital cortical development anomaly due to abnormal neuronal migration involving neocortical and hippocampal lamination, corpus callosum, cerebellum and brainstem. A large clinical spectrum can be observed, from children with severe epilepsy and intellectual and motor deficit to cases with severe cerebral dysgenesis in the antenatal period leading to pregnancy termination due to the severity of the prognosis.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=171680

Your genetic map

Gene	SNP	Genotype
TUBA1A	rs137853043	GG
TUBA1A	rs137853044	CC
TUBA1A	rs137853049	GG
TUBA1A	rs137853050	CC
TUBA1A	rs587784483	GG
TUBA1A	rs587784482	GG
TUBA1A	rs587784481	TT
TUBA1A	rs587784497	AA
TUBA1A	rs587784495	TT
TUBA1A	rs587784494	CC
TUBA1A	rs587784492	TT
TUBA1A	rs587784488	AA
TUBA1A	rs587784485	GG
TUBA1A	rs587784491	CC
TUBA1A	rs797046071	CC
TUBA1A	rs797046073	CC
TUBA1A	rs797046072	TT
TUBA1A	rs863224938	CC
TUBA1A	rs1057517843	CC

Hereditary Diseases (genetics)

Lissencephaly type 1 due to doublecortin gene mutation

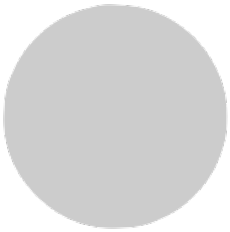
Type 1 lissencephaly due to doublecortin (DCX) gene mutations is a semi-dominant X-linked disease characterised by intellectual deficiency and seizures that are more severe in male patients.

Your genetic map

Gene	SNP	Genotype
DCX	rs104894780	GG
DCX	rs104894782	GG
DCX	rs56030372	CC
DCX	rs587783590	GG
DCX	rs587783589	CC
DCX	rs587783568	GG
DCX	rs587783534	GG
DCX	rs797045512	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2148

Hereditary Diseases (genetics)

Lysinuric protein intolerance

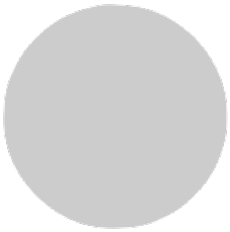
Lysinuric protein intolerance (LPI) is a very rare inherited multisystem condition caused by distrubance in amino acid metabolism.

Your genetic map

Gene	SNP	Genotype
SLC7A7	rs121908678	GG
SLC7A7	rs121908679	CC
SLC7A7	rs386833823	GG
SLC7A7	rs146582474	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=470

Hereditary Diseases (genetics)

Malaria

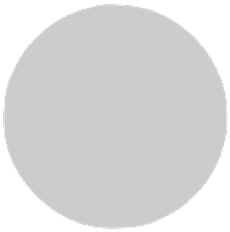
A life-threatening parasitic disease caused by Plasmodium (P.) parasites that are transmitted by Anophles mosquito bites to humans and is typically clinically characterized by attacks of fever, headache, chills and vomiting.

Your genetic map

Gene	SNP	Genotype
G6PD	rs72554664	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=673

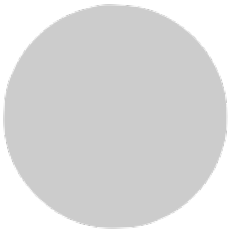
Hereditary Diseases (genetics)

MELAS

A rare neurometabolic genetic disorder which is progressive and multisystemic due to mitochondrial dysfunction and that is characterized by encephalomyopathy, lactic acidosis, and stroke-like episodes.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=550

Your genetic map

Gene	SNP	Genotype
MT TA	rs121434458	GG
MT TF	rs118203885	GG
MT TG	rs121434475	TT
MT TH	rs121434474	GG
MT TL1	rs199474657	AA
MT TL1	rs199474658	TT
MT TL1	rs199474660	CC
MT TL1	rs199474661	AA
MT TL1	rs199474662	AA
MT TL1	rs199474663	AA
MT TL2	rs121434462	GG
MT TP	rs199474701	GG
MT TS2	rs118203889	GG
MT TW	rs199474673	GG
MT TW	rs199474674	GG
ND1	rs199476123	GG
ND5	rs267606897	GG
ND5	rs267606898	GG
ND6	rs199476107	GG
NDUFS1	rs786205666	AA

Hereditary Diseases (genetics)

Metachondromatosis

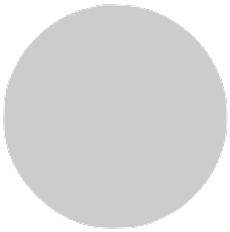
Metachondromatosis (MC) is a rare disorder characterized by the presence of both multiple enchondromas and osteochondroma-like lesions.

Your genetic map

Gene	SNP	Genotype
PTPN11	rs267606989	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2499

Hereditary Diseases (genetics)

Microlissencephaly

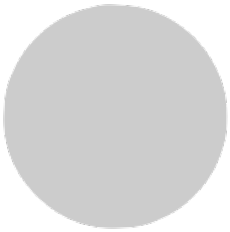
Microlissencephaly describes a heterogenous group of a rare cortical malformations characterized by lissencephaly in combination with severe congenital microcephaly, presenting with spasticity, severe developmental delay, and seizures and with survival varying from days to years.

Your genetic map

Gene	SNP	Genotype
NDE1	rs576928842	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1083

Hereditary Diseases (genetics)

Infantile hypertrophic cardiomyopathy due to MRPL44 deficiency

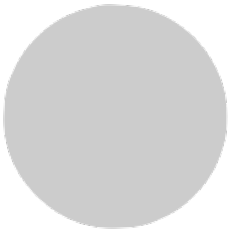
A rare mitochondrial oxidative phosphorylation disorder with complex I and IV deficiency characterized by hypertrophic cardiomyopathy, hepatic steatosis with elevated liver transaminases, exercise intolerance and muscle weakness. Neuro-opthalmological features (hemiplegic migraine, Leigh-like lesions on brain MRI, pigmentary retinopathy) have been reported later in life.

Your genetic map

Gene	SNP	Genotype
MRPL44	rs143697995	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=352563

Hereditary Diseases (genetics)

Mitochondrial hypertrophic cardiomyopathy with lactic acidosis due to MTO1 deficiency

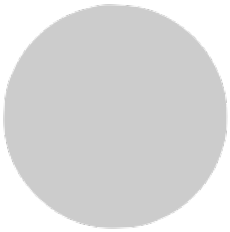
A rare mitochondrial oxidative phosphorylation disorder with complex I and IV deficiency characterized by lactic acidosis, hypotonia, hypertrophic cardiomyopathy and global developmental delay. Other clinical features include feeding difficulties, failure to thrive, seizures, optic atrophy and ataxia.

Your genetic map

Gene	SNP	Genotype
MTO1	rs201544686	GG
MTO1	rs200583827	CC
MTO1	rs775623164	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=314637

Hereditary Diseases (genetics)

Familial isolated restrictive cardiomyopathy

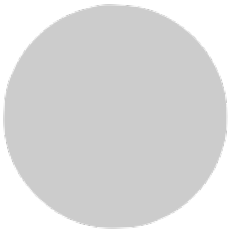
A rare genetic cardiac disease characterized by restrictive ventricular filling due to high ventricular stiffness that results in severe diastolic dysfunction in the absence of dilated or hypertrophied ventricles.

Your genetic map

Gene	SNP	Genotype
TNNI3	rs104894724	GG
TNNI3	rs104894729	CC
TNNI3	rs104894730	TT
TNNI3	rs727503504	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=75249

Hereditary Diseases (genetics)

Infantile myofibromatosis

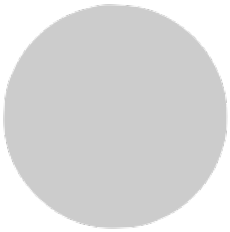
A rare benign soft tissue tumor characterized by the development of nodules in the skin, striated muscles, bones, and in exceptional cases, visceral organs, leading to a broad spectrum of clinical symptoms. It contains myofibroblasts.

Your genetic map

Gene	SNP	Genotype
PDGFRB	rs367543286	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2591

Hereditary Diseases (genetics)

Autosomal dominant centronuclear myopathy

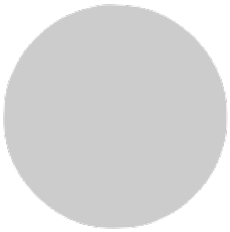
A rare, autosomal dominant congenital myopathy characterized by numerous centrally placed nuclei on muscle biopsy and clinical features of a congenital myopathy (hypotonia, distal/proximal muscle weakness, rib cage deformities (sometimes associated with respiratory insufficiency), ptosis, ophthalmoparesis and weakness of the muscles of facial expression with dysmorphic facial features.

Your genetic map

Gene	SNP	Genotype
DNM2	rs121909089	GG
DNM2	rs121909090	CC
DNM2	rs121909091	CC
DNM2	rs121909092	GG
DNM2	rs587783594	TT
DNM2	rs587783595	GG
DNM2	rs587783597	TT
DNM2	rs587783598	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=169189

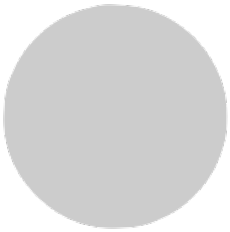
Hereditary Diseases (genetics)

X-linked centronuclear myopathy

A rare X-linked congenital myopathy characterized by numerous centrally placed nuclei on muscle biopsy and that presents at birth with marked weakness, hypotonia and respiratory failure.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=596

Your genetic map

Gene	SNP	Genotype
DNM2	rs121909095	CC
MTM1	rs132630302	AA
MTM1	rs132630304	CC
MTM1	rs397518445	AA
MTM1	rs132630305	CC
MTM1	rs132630306	CC
MTM1	rs398123272	GG
MTM1	rs398123275	CC
MTM1	rs587783817	TT
MTM1	rs587783823	GG
MTM1	rs587783843	GG
MTM1	rs587783844	AA
MTM1	rs587783846	GG
MTM1	rs587783857	CC
MTM1	rs587783753	CC
MTM1	rs587783796	GG
MTM1	rs587783809	CC
MTM1	rs587783810	GG
MTM1	rs587783814	CC
MTM1	rs587783813	AA
MTM1	rs587783812	GG
MTM1	rs587783816	TT
MTM1	rs587783820	AA
MTM1	rs587783825	CC
MTM1	rs587783828	GG
MTM1	rs587783830	GG
MTM1	rs587783831	AA
MTM1	rs587783832	CC
MTM1	rs587783834	GG
MTM1	rs587783835	AA
MTM1	rs587783836	CC

Hereditary Diseases (genetics)

X-linked myopathy with excessive autophagy

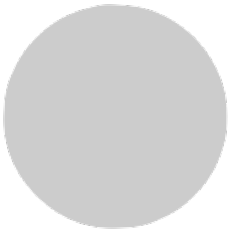
X-linked myopathy with excessive autophagy is a childhood-onset X-linked myopathy characterised by slow progression of muscle weakness and unique histopathological findings.

Your genetic map

Gene	SNP	Genotype
VMA21	rs797044909	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=25980

Hereditary Diseases (genetics)

Polyglucosan body myopathy type 2

A rare glycogen storage disease characterized by slowly progressive myopathy with storage of polyglucosan in muscle fibers. Age of onset ranges from childhood to late adulthood. Patients present proximal or proximodistal weakness predominantly of limb-girdle muscles. Variable features include exercise intolerance or myalgia. Serum creatine kinase is normal or mildly elevated. There is usually no overt cardiac involvement.

Your genetic map

Gene	SNP	Genotype
GYG1	rs370652040	GG

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=456369

Hereditary Diseases (genetics)

Reducing body myopathy

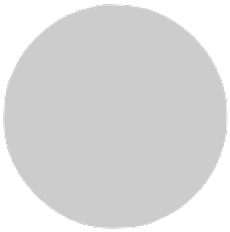
Reducing body myopathy (RBM) is a rare muscle disorder marked by progressive muscle weakness and the presence of characteristic inclusion bodies in affected muscle fibres.

Your genetic map

Gene	SNP	Genotype
FHL1	rs122459146	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=97239

Hereditary Diseases (genetics)

Congenital fiber-type disproportion myopathy

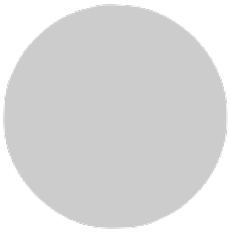
A rare genetic, congenital, non-dystrophic myopathy characterized by neonatal or infantile-onset hypotonia and mild to severe generalized muscle weakness.

Your genetic map

Gene	SNP	Genotype
MYH7	rs1060505018	CC
RYR1	rs772494345	GG
RYR1	rs1057518940	GG
RYR1	rs142929172	GG
RYR1	rs193922810	GG
TPM3	rs121964854	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2020

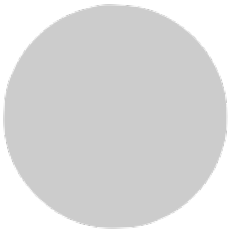
Hereditary Diseases (genetics)

Bethlem myopathy

Bethlem myopathy is a benign autosomal dominant form of slowly progressive muscular dystrophy.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=610

Your genetic map

Gene	SNP	Genotype
COL6A1	rs121912936	AA
COL6A1	rs398123631	GG
COL6A1	rs121912938	GG
COL6A1	rs121912939	GG
COL6A1	rs398123639	AA
COL6A1	rs398123640	GG
COL6A1	rs398123643	GG
COL6A1	rs398123644	GG
COL6A1	rs794727060	TT
COL6A1	rs797045477	AA
COL6A2	rs267606750	GG
COL6A2	rs387906609	CC
COL6A2	rs397515333	GG
COL6A2	rs727502827	GG
COL6A2	rs727502828	GG
COL6A2	rs138948335	GG
COL6A2	rs794727715	GG
COL6A2	rs794727788	GG
COL6A2	rs794727855	GG
COL6A2	rs770842374	TT
COL6A3	rs121434553	CC
COL6A3	rs794727188	CC
COL6A3	rs886043737	CC

Hereditary Diseases (genetics)

Miyoshi myopathy

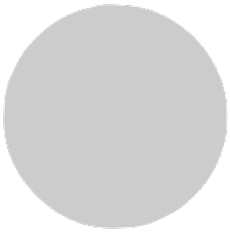
A recessive distal myopathy characterized by weakness in the distal lower extremity posterior compartment (gastrocnemius and soleus muscles) and associated with difficulties in standing on tip toes.

Your genetic map

Gene	SNP	Genotype
DYSF	rs121908953	CC
DYSF	rs121908958	GG
DYSF	rs398123792	AA
DYSF	rs758180890	CC
DYSF	rs121908963	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=45448

Hereditary Diseases (genetics)

Distal myopathy with anterior tibial onset

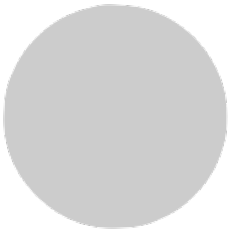
Distal myopathy with anterior tibial onset is a rare, genetic neuromuscular disease characterized by a progressive muscle weakness starting in the anterior tibial muscles, later involving lower and upper limb muscles, associated with an increased serum creatine kinase levels and absence of dysferlin on muscle biopsy. Patients become wheelchair dependent.

Your genetic map

Gene	SNP	Genotype
DYSF	rs121908959	CC
DYSF	rs398123773	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=178400

Hereditary Diseases (genetics)

Laing early-onset distal myopathy

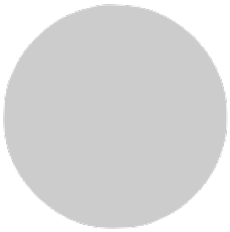
Laing distal myopathy, also called myopathy distal, type 1 (MPD1), is characterized by early-onset selective weakness of the great toe and ankle dorsiflexors, and a very slowly progressive course.

Your genetic map

Gene	SNP	Genotype
MHRT	rs121913647	CC
MHRT	rs397516248	CC
MHRT	rs397516254	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=59135

Hereditary Diseases (genetics)

Progressive scapulohumeroperoneal distal myopathy

A rare genetic muscular dystrophy characterized by progressive muscle weakness in a scapulo-humero-peroneal and distal distribution, featuring wrist extensor weakness, finger and foot drop, scapular winging, mild facial weakness, contractures of the Achilles tendon, elbow, and shoulder, and diminished or absent deep tendon reflexes. A predilection for the upper extremities has been reported in some patients. Respiratory muscles are spared until late in the disease course. Age of onset, progression, and severity of the disease vary significantly between individuals. Muscle biopsy shows groups of atrophic type I fibers and increased internal nuclei.

Your genetic map

Gene	SNP	Genotype
ACTA1	rs869312739	CC

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=447977

Hereditary Diseases (genetics)

GNE myopathy

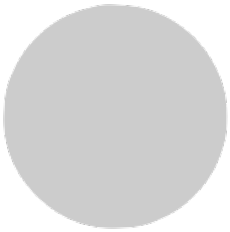
GNE myopathy is a rare autosomal recessive distal myopathy characterized by early adult-onset, slowly to moderately progressive distal muscle weakness that preferentially affects the tibialis anterior muscle and that usually spares the quadriceps femoris. Muscle biopsy reveals presence of rimmed vacuoles.

Your genetic map

Gene	SNP	Genotype
GNE	rs28937594	AA
GNE	rs121908629	CC
GNE	rs121908632	CC
GNE	rs62541771	GG
GNE	rs139425890	TT
GNE	rs748949603	AA
GNE	rs773729410	GG
GNE	rs779694939	AA
GNE	rs745517517	GG
GNE	rs1209266607	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=602

Hereditary Diseases (genetics)

Hereditary myopathy with early respiratory failure

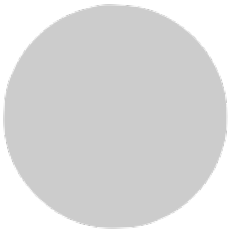
A rare genetic neuromuscular disease characterized by adult onset of slowly progressive distal and/or proximal muscle weakness in the upper and lower extremities, and early involvement of respiratory muscles leading to respiratory failure. Additional features are neck flexor weakness, foot extensor weakness, and, in rare cases, mildly impaired cardiac function. Muscle biopsy shows eosinophilic myofibrillar inclusions referred to as cytoplasmic bodies, as well as fiber size variation, increased internal nuclei and connective tissue, fiber splitting, and rimmed vacuoles.

Your genetic map

Gene	SNP	Genotype
TTN AS1	rs869320740	AA
TTN AS1	rs753334568	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=178464

Hereditary Diseases (genetics)

Mitochondrial myopathy with reversible cytochrome C oxidase deficiency

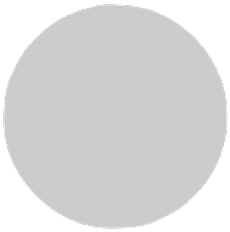
A rare, genetic, mitochondrial oxidative phosphorylation disorder characterized by a potentially life-threatening, severe myopathy manifesting in the neonatal to early infantile period, followed by marked, spontaneous improvement of muscular function by early childhood. Associated biochemical findings include lactic acidosis and a transient, marked decrease in respiratory chain activity.

Your genetic map

Gene	SNP	Genotype
CYTB	rs207459997	GG
CYTB	rs207459998	GG
CYTB	rs207460002	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=254864

Hereditary Diseases (genetics)

Multiminicore myopathy

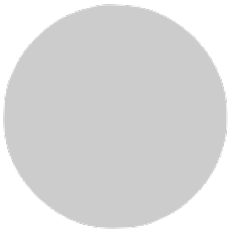
A rare hereditary neuromuscular disorder characterized by multiple cores on muscle biopsy and clinical features of a congenital myopathy.

Your genetic map

Gene	SNP	Genotype
RYR1	rs118192173	CC
RYR1	rs118192174	TT
RYR1	rs193922803	CC
RYR1	rs193922809	GG
RYR1	rs200563280	CC
RYR1	rs878854365	CC
RYR1	rs111436401	GG
RYR1	rs1057524858	GG
RYR1	rs1432807966	CC
RYR1	rs1346257891	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=598

Hereditary Diseases (genetics)

Severe congenital nemaline myopathy

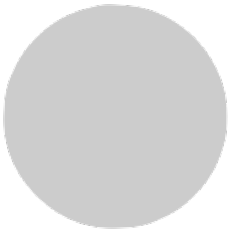
Severe congenital nemaline myopathy is a severe form of nemaline myopathy (NM; see this term) characterized by severe hypotonia with little spontaneous movement in neonates.

Your genetic map

Gene	SNP	Genotype
KLHL40	rs397509419	GG
KLHL40	rs367579275	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=171430

Hereditary Diseases (genetics)

Inclusion body myopathy with Paget disease of bone and frontotemporal dementia

Inclusion body myopathy with Paget disease of bone and frontotemporal dementia (IBMPFD) is a multisystem degenerative genetic disorder characterized by adult-onset proximal and distal muscle weakness (clinically resembling limb-girdle muscular dystrophy; see this term); early-onset Paget disease of bone (see this term), manifesting with bone pain, deformity and enlargement of the long-bones; and premature frontotemporal dementia (see this term), manifesting first with dysnomia, dyscalculia and comprehension deficits followed by progressive aphasia, alexia, and agraphia. As the disease progresses, muscle weakness begins to affect the other limbs and respiratory muscles, ultimately resulting in respiratory or cardiac failure.

Your genetic map

Gene	SNP	Genotype
VCP	rs121909330	GG
VCP	rs121909335	CC

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=52430

Hereditary Diseases (genetics)

Potassium-aggravated myotonia

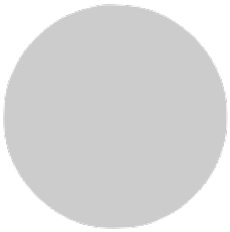
A muscular channelopathy presenting with a pure myotonia dramatically aggravated by potassium ingestion, with variable cold sensitivity and no episodic weakness. This group includes three forms: myotonia fluctuans, myotonia permanens, and acetazolamide-responsive myotonia.

Your genetic map

Gene	SNP	Genotype
LOC1053	rs121908552	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=612

Hereditary Diseases (genetics)

MODY

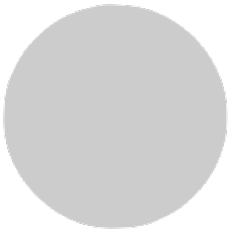
MODY (maturity-onset diabetes of the young) is a rare, familial, clinically and genetically heterogeneous form of diabetes characterized by young age of onset (generally 10 -45 years of age) with maintenance of endogenous insulin production, lack of pancreatic beta-cell autoimmunity, absence of obesity and insulin resistance and extra-pancreatic manifestations in some subtypes.

Your genetic map

Gene	SNP	Genotype
HNF4A	rs193922470	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=552

Hereditary Diseases (genetics)

Complete hydatidiform mole

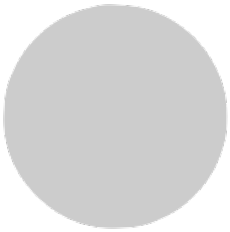
Complete hydatidiform mole is a type of hydatiform mole characterized by abnormal hyperplastic trophoblasts and hydropic villi due to fertilization of an enucleated ovocyte by one or two haploid spermatozoa that can manifest with vaginal bleeding accompanied by nausea and frequent vomiting, hyperemesis gravidarum, risk of spontaneous miscarriage, hyperthyroidism, and has the potential of developing into choriocarcinoma (see this term).

Your genetic map

Gene	SNP	Genotype
NLRP7	rs104895506	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=254688

Hereditary Diseases (genetics)

MPI-CDG

MPI-CDG is a form of congenital disorders of N-linked glycosylation, characterized by cyclic vomiting, profound hypoglycemia, failure to thrive, liver fibrosis, gastrointestinal complications (protein-losing enteropathy with hypoalbuminaemia, life-threatening intestinal bleeding of diffuse origin), and thrombotic events (protein C and S deficiency, low anti-thrombine III levels), whereas neurological development and cognitive capacity is usually normal. The clinical course is variable even within families. The disease is caused by loss of function of the gene MPI (15q24.1).

Your genetic map

Gene	SNP	Genotype
MPI	rs104894489	GG
MPI	rs28928906	GG
MPI	rs863225086	AA
MPI	rs863225087	GG

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79319

Hereditary Diseases (genetics)

Mucolipidosis type III

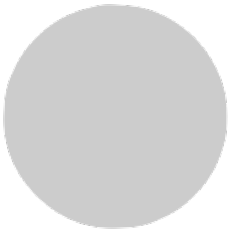
A rare lysosomal disease characterized by dysmorphic features and skeletal changes, restricted joint mobility, short stature, and hand deformities (such as claw hands, stiffness of hands, carpal tunnel syndrome, inability to make fists). Most patients have normal intellectual capacity and the clinical progression is less rapid than that of mucolipidosis type II (MLII).

Your genetic map

Gene	SNP	Genotype
GNPTAB	rs137852897	GG
GNPTAB	rs281864969	GG
GNPTAB	rs281864980	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=577

Hereditary Diseases (genetics)

Mucopolysaccharidosis type 1

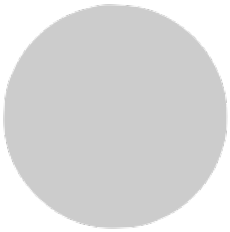
Mucopolysaccharidosis type 1 (MPS 1) is a rare lysosomal storage disease belonging to the group of mucopolysaccharidoses. There are three variants, differing widely in their severity, with Hurler syndrome being the most severe, Scheie syndrome the mildest and Hurler-Scheie syndrome giving an intermediate phenotype.

Your genetic map

Gene	SNP	Genotype
IDUA	rs121965021	CC
IDUA	rs398123256	GG
IDUA	rs199801029	GG
IDUA	rs794727701	GG
IDUA	rs777295041	AA
SLC26A1	rs121965020	CC
SLC26A1	rs398123259	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=579

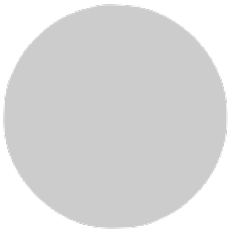
Hereditary Diseases (genetics)

Mucopolysaccharidosis type 2

A lysosomal storage disease with multisystemic involvement leading to a massive accumulation of glycosaminoglycans and a wide variety of symptoms including distinctive coarse facial features, short stature, cardio-respiratory involvement and skeletal abnormalities. It manifests as a continuum varying from a severe form with neurodegeneration to an attenuated form without neuronal involvement.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=580

Your genetic map

Gene	SNP	Genotype
IDS	rs199422227	GG
IDS	rs104894853	GG
IDS	rs113993948	GG
IDS	rs199422231	GG
IDS	rs113993946	CC
IDS	rs864622773	TT
IDS	rs864622777	CC
IDS	rs864622771	AA
IDS	rs193302912	CC
IDS	rs113993953	TT
IDS	rs193302907	CC
IDS	rs864622778	CC
IDS	rs113993945	GG
IDS	rs864622779	CC
IDS	rs193302904	CC
IDS	rs113993947	CC
IDS	rs193302908	GG
IDS	rs193302910	CC
IDS	rs781997631	AA
IDS	rs113993955	AA

Hereditary Diseases (genetics)

Mucopolysaccharidosis type 3

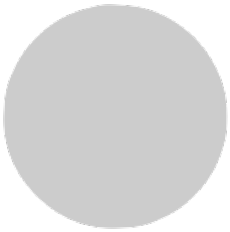
Mucopolysaccharidosis type III (MPS III) is a lysosomal storage disease belonging to the group of mucopolysaccharidoses and characterised by severe and rapid intellectual deterioration.

Your genetic map

Gene	SNP	Genotype
SGSH	rs104894635	CC
SGSH	rs104894636	GG
SGSH	rs104894641	CC
SGSH	rs104894637	GG
SGSH	rs104894638	CC
SGSH	rs104894639	CC
SGSH	rs104894640	CC
SGSH	rs138504221	AA
SGSH	rs143947056	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=581

Hereditary Diseases (genetics)

Mucopolysaccharidosis type 4

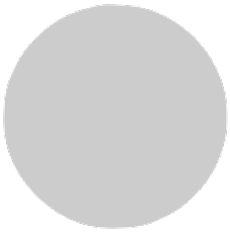
A rare lysosomal storage disease characterized by mild to severe spondylo-epiphyso-metaphyseal dysplasia, manifesting with disproportionate short stature (short neck and trunk), joint laxity, pectus carinatum, genum valgum, abnormal gait, tracheal narrowing, spinal abnormalities (kyphosis and scoliosis), respiratory impairment and valvular heart disease.

Your genetic map

Gene	SNP	Genotype
GALNS	rs118204438	TT
GALNS	rs118204443	CC
GALNS	rs118204444	GG
GALNS	rs372893383	CC
GALNS	rs398123438	CC
GALNS	rs398123440	GG
GALNS	rs746756997	AA
LOC1079	rs118204437	GG
LOC1079	rs398123429	TT
LOC1079	rs398123430	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=582

Hereditary Diseases (genetics)

Mucopolysaccharidosis type 6

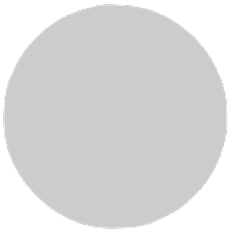
Mucopolysaccharidosis type 6 (MPS 6) is a lysosomal storage disease with progressive multisystem involvement, associated with a deficiency of arylsulfatase B (ASB) leading to the accumulation of dermatan sulfate.

Your genetic map

Gene	SNP	Genotype
ARSB	rs118203943	TT
ARSB	rs431905495	CC
ARSB	rs398123125	CC
ARSB	rs727503809	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=583

Hereditary Diseases (genetics)

Mucopolysaccharidosis type 7

A rare, genetic lysosomal storage disease characterized by accumulation of glycosaminoglycans in connective tissue which results in progressive multisystem involvement with severity ranging from mild to severe. The most consistent features include musculoskeletal involvement (particularly dysostosis multiplex, joint restriction, thorax abnormalities, and short stature), limited vocabulary, intellectual disability, coarse facies with a short neck, pulmonary involvement (predominantly decreased pulmonary function), corneal clouding, and cardiac valve disease.

Your genetic map

Gene	SNP	Genotype
GUSB	rs121918172	GG
GUSB	rs121918173	GG
GUSB	rs121918181	GG
GUSB	rs121918185	GG

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=584

Hereditary Diseases (genetics)

Multiple endocrine neoplasia type 2

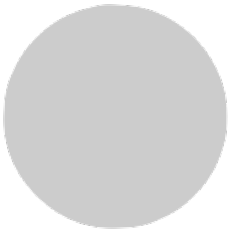
A rare multiple endocrine neoplasia (MEN) syndrome that is principally characterized by the association of medullary thyroid carcinoma (MTC) with other endocrine tumors. The variant MEN 2A is defined by MTC associated with pheochromocytoma and/or primary hyperparathyroidism (MEN2A); the variant MEN 2B is defined as an aggressive form of MTC in association with pheochromocytoma but without primary hyperparathyroidism.

Your genetic map

Gene	SNP	Genotype
RET	rs74799832	TT
RET	rs78014899	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=653

Hereditary Diseases (genetics)

Mitochondrial membrane protein-associated neurodegeneration

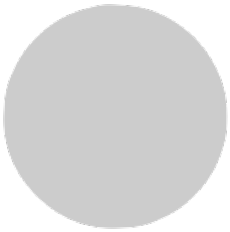
A rare neurodegenerative disorder characterized by iron accumulation in specific regions of the brain, usually the basal ganglia, and associated with slowly progressive pyramidal (spasticity) and extrapyramidal (dystonia) signs, motor axonal neuropathy, optic atrophy, cognitive decline, and neuropsychiatric abnormalities.

Your genetic map

Gene	SNP	Genotype
C19orf12	rs397514477	GG
C19orf12	rs515726205	CC
C19orf12	rs752450983	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=289560

Hereditary Diseases (genetics)

Neurofibromatosis type 6

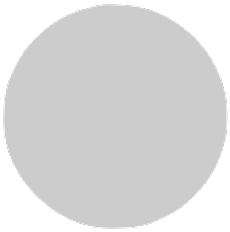
Neurofibromatosis type 6 (NF6), also referred as cafe-au-lait spots syndrome, is a cutaneous disorder characterized by the presence of several cafe-au-lait (CAL) macules without any other manifestations of neurofibromatosis or any other systemic disorder.

Your genetic map

Gene	SNP	Genotype
NF1	rs1057518904	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2678

Hereditary Diseases (genetics)

Neurofibromatosis-Noonan syndrome

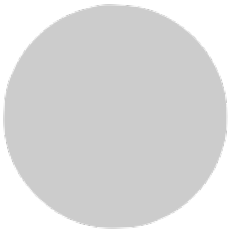
Neurofibromatosis-Noonan syndrome (NFNS) is a RASopathy and a variant of neurofibromatosis type 1 (NF1) characterized by the combination of features of NF1, such as cafe-au-lait spots, iris Lisch nodules, axillary and inguinal freckling, optic nerve glioma and multiple neurofibromas, and Noonan syndrome (NS), such as short stature, typical facial features (hypertelorism, ptosis, downslanting palpebral fissures, low-set posteriorly rotated ears with a thickened helix, and a broad forehead), congenital heart defects and unusual pectus deformity. As these three entities have significant phenotypic overlap, molecular genetic testing is often necessary for a correct diagnosis (such as when cafe-au-lait spots are present in patients diagnosed with NS).

Your genetic map

Gene	SNP	Genotype
NF1	rs199474789	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=638

Hereditary Diseases (genetics)

Navajo neurohepatopathy

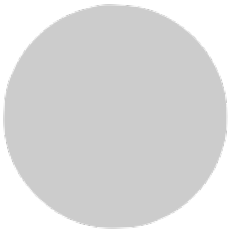
A rare, life-threatening, mitochondrial DNA depletion syndrome disease characterized by severe, progressive sensorimotor neuropathy associated with corneal ulceration, scarring or anesthesia, acral mutilation, metabolic and immunologic derangement, and hepatopathy (which can manifest with fulminant hepatic failure, a Reye-like syndrome or indolent progression to liver cirrhosis, depending on clinical form involved), present in the Navajo Native American population. Clinical presentation includes failure to thrive, distal limb weakness with reduced sensation, limb contractures with loss of funtion, areflexia, recurrent metabolic acidosis with intercurrent illness, immunologic anomalies manifesting with severe systemic infections, and sexual infantilism.

Your genetic map

Gene	SNP	Genotype
MPV17	rs121909721	CC
MPV17	rs121909723	GG
MPV17	rs267607258	GG

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=255229

Hereditary Diseases (genetics)

Autosomal recessive axonal neuropathy with neuromyotonia

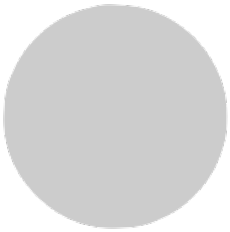
A rare peripheral neuropathy characterized by slowly progressive axonal, motor greater than sensory, polyneuropathy combined with neuromytonia (including spontaneous muscular activity at rest (myokymia), impaired muscle relaxation (pseudomyotonia), and contractures of hands and feet) and neuromyotonic or myokymic discharges on needle EMG. It presents with distal lower limb weakness with gait impairment, muscle stiffness, fasciculations and cramps in hands and legs worsened by cold, decreased to absent tendon reflexes, intrinsic hand muscle atrophy and, variably, mild distal sensory impairment.

Your genetic map

Gene	SNP	Genotype
HINT1	rs149782619	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=324442

Hereditary Diseases (genetics)

Leber hereditary optic neuropathy

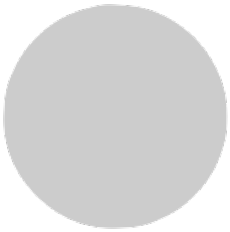
A rare hereditary optic neuropathy characterized by sudden onset, painless central vision loss, loss of retinal ganglion cells and optic atrophy.

Your genetic map

Gene	SNP	Genotype
ND1	rs397515507	GG
ND6	rs199476104	TT
ND6	rs199476106	AA
ND6	rs397515506	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=104

Hereditary Diseases (genetics)

Autosomal recessive severe congenital neutropenia due to CSF3R deficiency

Autosomal recessive severe congenital neutropenia due to CSF3R deficiency is a rare, genetic, primary immunodeficiency disorder characterized by predisposition to recurrent, life-threatening bacterial infections associated with decreased peripheral neutrophil granulocytes (absolute neutrophil count less than 500 cells/microliter), resulting from recessively inherited loss-of-function mutations in the CSF3R gene. Full maturation of all three lineages in the bone marrow and refractoriness to in vivo rhG-CSF treatment are associated.

Your genetic map

Gene	SNP	Genotype
CSF3R	rs138156467	CC

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=420702

Hereditary Diseases (genetics)

Autosomal recessive severe congenital neutropenia due to JAGN1 deficiency

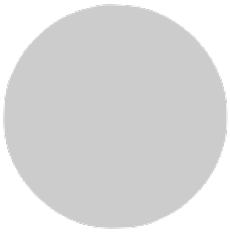
Autosomal recessive severe congenital neutropenia due to JAGN1 deficiency is a rare, genetic, primary immunodeficiency disorder characterized by early-onset, recurrent, severe bacterial infections, granulopoiesis maturation arrest at the promyelocyte/myelocyte stage and markedly reduced absolute neutrophil counts, resulting from recessively inherited mutations in the JAGN1 gene. Mild facial dysmorphism (i.e. triangular face), short stature, failure to thrive, hypothyroidism, developmental delay, pancreatic insufficiency and coractation of aorta, as well as bone and urogenital abnormalities, may also be associated.

Your genetic map

Gene	SNP	Genotype
JAGN1	rs587777728	CC
JAGN1	rs587777730	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=423384

Hereditary Diseases (genetics)

Woolly hair nevus

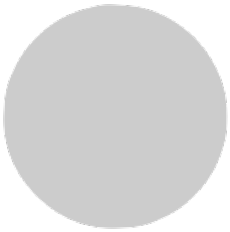
Woolly hair nevus (WHN) is a rare non-familial hair anomaly characterized by kinky, tightly coiled, and hypopigmented fine hair with an average diameter of 0.5 cm, noted, since birth or during the first two years of life, in a localized circumscribed distribution on the scalp. Occassionally, WHN grows in areas observed to be alopecic in the neonatal period. WHN can be associated with features like ocular defects (persistent pupillary membrane, retinal defects), precocious puberty, and epidermal nevi.

Your genetic map

Gene	SNP	Genotype
NRAS	rs121913237	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79414

Hereditary Diseases (genetics)

Obesity due to leptin receptor gene deficiency

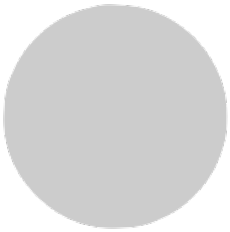
A rare, genetic, non-syndromic, obesity disease characterized by severe, early-onset obesity, associated with major hyperphagia and endocrine abnormalities, resulting from leptin receptor deficiency.

Your genetic map

Gene	SNP	Genotype
LEPR	rs144159890	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=179494

Hereditary Diseases (genetics)

Obesity due to melanocortin 4 receptor deficiency

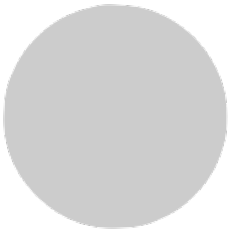
Melanocortin 4 receptor (MC4R) deficiency is the commonest form of monogenic obesity identified so far. MC4R deficiency is characterised by severe obesity, an increase in lean body mass and bone mineral density, increased linear growth in early childhood, hyperphagia beginning in the first year of life and severe hyperinsulinaemia, in the presence of preserved reproductive function.

Your genetic map

Gene	SNP	Genotype
MC4R	rs121913564	AA
MC4R	rs52804924	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=71529

Hereditary Diseases (genetics)

Autosomal recessive progressive external ophthalmoplegia

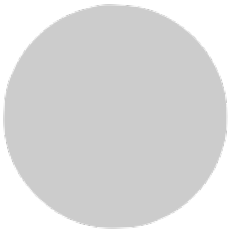
A rare genetic, neuro-ophthalmological disease characterized by progressive weakness of the external eye muscles, resulting in bilateral ptosis and diffuse, symmetric ophthalmoparesis. Additional signs may include generalized skeletal muscle weakness, muscle atrophy, sensory axonal neuropathy, ataxia, cardiomyopathy, and psychiatric symptoms. It is usually more severe than autosomal dominant form.

Your genetic map

Gene	SNP	Genotype
MIR6766	rs113994095	CC
POLG	rs113994098	CC
POLG	rs121918054	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=254886

Hereditary Diseases (genetics)

Hypertrichotic osteochondrodysplasia, Cantu type

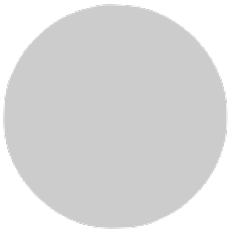
Cantu syndrome is a rare disorder characterized by congenital hypertrichosis, osteochondrodysplasia, cardiomegaly, and dysmorphism.

Your genetic map

Gene	SNP	Genotype
ABCC9	rs387907208	GG
ABCC9	rs387907209	CC
ABCC9	rs387907227	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1517

Hereditary Diseases (genetics)

Multiple osteochondromas

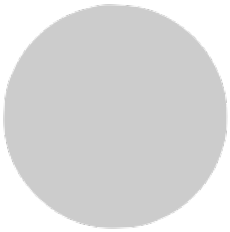
A primary bone disorder characterized by development of two or more cartilage capped bony outgrowths (osteochondromas) at the surface of the bones.

Your genetic map

Gene	SNP	Genotype
EXT1	rs119103287	CC
EXT1	rs119103290	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=321

Hereditary Diseases (genetics)

Osteopetrosis with renal tubular acidosis

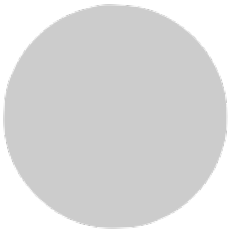
Osteopetrosis with renal tubular acidosis is a rare disorder characterized by osteopetrosis (see this term), renal tubular acidosis (RTA), and neurological disorders related to cerebral calcifications.

Your genetic map

Gene	SNP	Genotype
CA3 AS1	rs573750741	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2785

Hereditary Diseases (genetics)

Albers-Schönberg osteopetrosis

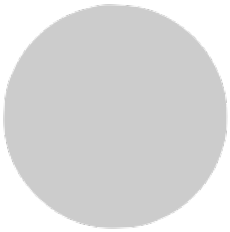
A sclerosing disorder of the skeleton characterized by increased bone density that classically displays the radiographic sign of "sandwich vertebrae" (dense bands of sclerosis parallel to the vertebral endplates).

Your genetic map

Gene	SNP	Genotype
CLCN7	rs387907576	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=53

Hereditary Diseases (genetics)

Osteosarcoma

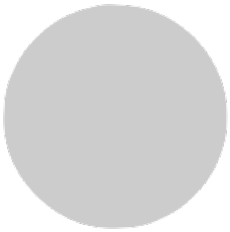
Osteosarcoma is a primary malignant tumour of the skeleton characterised by the direct formation of immature bone or osteoid tissue by the tumour cells.

Your genetic map

Gene	SNP	Genotype
TP53	rs28934573	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=668

Hereditary Diseases (genetics)

Hereditary chronic pancreatitis

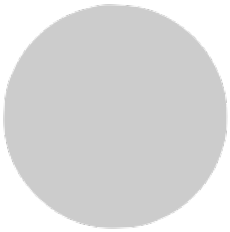
A rare gastroenterologic disease characterized by recurrent acute pancreatitis and/or chronic pancreatitis in at least 2 first-degree relatives, or 3 or more second-degree relatives in 2 or more generations, for which no predisposing factors are identified. This rare inherited form of pancreatitis leads to irreversible damage to both exocrine and endocrine components of the pancreas.

Your genetic map

Gene	SNP	Genotype
CTRC	rs121909294	GG
PRSS1	rs111033565	GG
PRSS1	rs111033567	AA
PRSS1	rs111033568	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=676

Hereditary Diseases (genetics)

Non-acquired panhypopituitarism

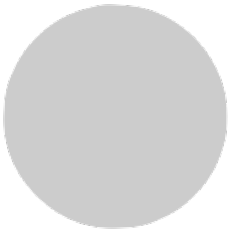
A rare genetic pituitary disease characterized by variable deficiency of all hormones produced in the anterior lobe of the pituitary gland. Clinical manifestations include hypothyroidism, hypogonadism, growth retardation and short stature, and secondary adrenal insufficiency. Age of onset is variable. Signs and symptoms usually develop gradually, and loss of the different hormones is often sequential.

Your genetic map

Gene	SNP	Genotype
PROP1	rs121917839	GG
PROP1	rs121917840	AA
PROP1	rs121917843	GG
PROP1	rs121917845	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=90695

Hereditary Diseases (genetics)

Pachydermoperiostosis

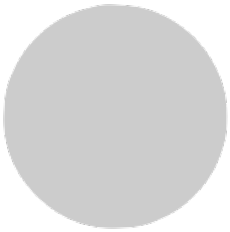
Pachydermoperiostosis (PDP) is a form of primary hypertrophic osteoarthropathy (see this term), a rare hereditary disorder, and is characterized by digital clubbing, pachydermia and subperiosteal new bone formation associated with pain, polyarthritis, cutis verticis gyrata, seborrhea and hyperhidrosis. Three forms have been described: a complete form with pachydermia and periostitis, an incomplete form with evidence of bone abnormalities but lacking pachydermia, and a forme frusta with prominent pachydermia and minimal-to-absent skeletal changes.

Your genetic map

Gene	SNP	Genotype
SLCO2A1	rs776813259	GG
SLCO2A1	rs765249238	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2796

Hereditary Diseases (genetics)

Pachyonychia congenita

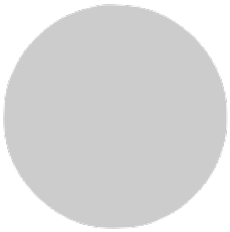
Pachyonychia congenita (PC) is a rare genodermatosis predominantly featuring painful palmoplantar keratoderma, thickened nails, cysts and whitish oral mucosa.

Your genetic map

Gene	SNP	Genotype
KRT16	rs60944949	AA
KRT16	rs58293603	AA
KRT16	rs59328451	TT
KRT16	rs28928894	AA
KRT16	rs58608173	TT
KRT16	rs59856285	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2309

Hereditary Diseases (genetics)

Hypokalemic periodic paralysis

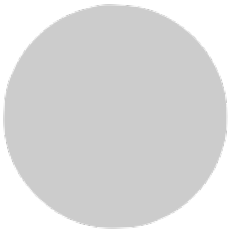
A rare genetic, muscle channelopathy characterized by recurrent episodic attacks of generalized muscle weakness associated with a decrease in blood potassium levels.

Your genetic map

Gene	SNP	Genotype
CACNA1S	rs28930068	CC
CACNA1S	rs28930069	GG
CACNA1S	rs80338777	CC
CACNA1S	rs267606698	AA
CACNA1S	rs797045031	TT
CACNA1S	rs770073633	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=681

Hereditary Diseases (genetics)

Paramyotonia congenita of Von Eulenburg

Paramyotonia congenita of Von Eulenburg is characterised by exercise- or cold-induced myotonia and muscle weakness. Prevalence is unknown. The syndrome is nonprogressive and is transmitted as an autosomal dominant trait. It is caused by mutations in the gene encoding the alpha subunit of the type IV voltage-gated sodium channel (SCN4A; 17q23.3).

Your genetic map

Gene	SNP	Genotype
LOC1053	rs80338956	AA
SCN4A	rs121908544	GG
SCN4A	rs121908547	GG

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=684

Hereditary Diseases (genetics)

Autosomal dominant spastic paraplegia type 10

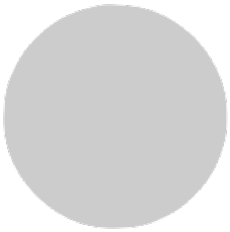
A rare, hereditary spastic paraplegia that can present as either a pure or complex phenotype. The pure form is characterized by lower limb spasticity, hyperreflexia and extensor plantar responses, presenting in childhood or adolescence. The complex form is characterized by the association with additional manifestations including peripheral neuropathy with upper limb muscle atrophy, moderate intellectual disability and parkinsonism. Deafness and retinitis pigmentosa have also been reported.

Your genetic map

Gene	SNP	Genotype
KIF5A	rs387907285	GG
KIF5A	rs387907287	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=100991

Hereditary Diseases (genetics)

Autosomal dominant spastic paraplegia type 17

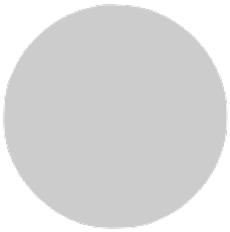
A complex hereditary spastic paraplegia characterized by progressive spastic paraplegia, upper and lower limb muscle atrophy, hyperreflexia, extensor plantar responses, pes cavus and occasionally impaired vibration sense. Association with hand muscles amyotrophy typical.

Your genetic map

Gene	SNP	Genotype
HNRNPU	rs137852973	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=100998

Hereditary Diseases (genetics)

Autosomal dominant spastic paraplegia type 31

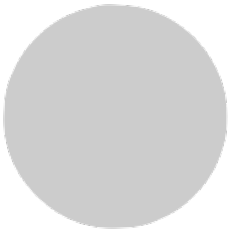
A rare type of hereditary spastic paraplegia usually characterized by a pure phenotype of proximal weakness of the lower extremities with spastic gait and brisk reflexes, with a bimodal age of onset of either childhood or adulthood (>30 years). In some cases, it can present as a complex phenotype with additional associated manifestations including peripheral neuropathy, bulbar palsy (with dysarthria and dysphagia), distal amyotrophy, and impaired distal vibration sense.

Your genetic map

Gene	SNP	Genotype
REEP1	rs121918262	GG
REEP1	rs786204081	TT

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=101011

Hereditary Diseases (genetics)

Autosomal dominant spastic paraplegia type 8

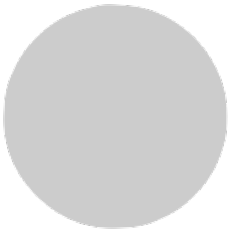
A rare, pure or complex form of hereditary spastic paraplegia characterized by early adulthood onset of slowly progressive lower limb spasticity resulting in gait disturbances, hyperreflexia and extensor plantar responses, urinary urgency and/or incontinence, muscle weakness, decreased vibration sense and mild muscular atrophy in lower extremities. It may be associated with complicating signs, such as sensory neuropathy, ataxia (i.e. mild dysmetria, uncoordinated eye movement) and mild dysphagia.

Your genetic map

Gene	SNP	Genotype
WASHC5	rs80338867	CC
WASHC5	rs80338866	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=100989

Hereditary Diseases (genetics)

Autosomal recessive spastic paraplegia type 15

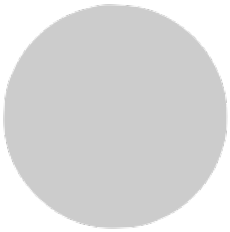
Autosomal recessive spastic paraplegia type 15 is a complex form of hereditary spastic paraplegia characterized by a childhood to adulthood onset of slowly progressive lower limb spasticity (resulting in gait disturbance, extensor plantar responses and decreased vibration sense) associated with mild intellectual disability, mild cerebellar ataxia, peripheral neuropathy (with distal upper limb amyotrophy) and retinal degeneration. Thin corpus callosum is a common imaging finding.

Your genetic map

Gene	SNP	Genotype
ZFYVE26	rs118204049	GG
ZFYVE26	rs370828455	CC
ZFYVE26	rs769329153	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=100996

Hereditary Diseases (genetics)

Autosomal recessive spastic paraplegia type 35

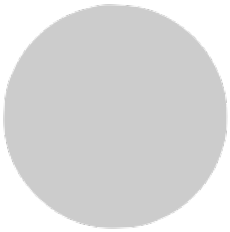
Autosomal recessive spastic paraplegia type 35 is a rare form of hereditary spastic paraplegia characterized by childhood (exceptionally adolescent) onset of a complex phenotype presenting with lower limb (followed by upper limb) spasticity with hyperreflexia and extensor plantar responses, with additional manifestations including progressive dysarthria, dystonia, mild cognitive decline, extrapyramidal features, optic atrophy and seizures. White matter abnormalities and brain iron accumulation have also been observed on brain magnetic resonance imaging.

Your genetic map

Gene	SNP	Genotype
FA2H	rs863224870	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=171629

Hereditary Diseases (genetics)

Autosomal recessive spastic paraplegia type 54

Autosomal recessive spastic paraplegia type 54 (SPG54) is a rare, complex form of hereditary spastic paraplegia characterized by the onset in early childhood of progressive spastic paraplegia associated with cerebellar signs, short stature, delayed psychomotor development, intellectual disability and, less commonly, foot contractures, dysarthria, dysphagia, strabismus and optic hypoplasia. SPG54 is caused by mutations in the DDHD2 gene (8p11.23) encoding phospholipase DDHD2.

Your genetic map

Gene	SNP	Genotype
DDHD2	rs375168720	GG
DDHD2	rs755267771	CC

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=320380

Hereditary Diseases (genetics)

Autosomal recessive spastic paraplegia type 56

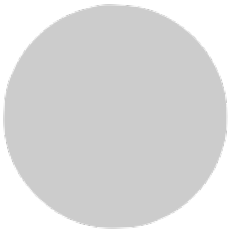
A rare form of hereditary spastic paraplegia characterized by delayed walking, toe walking, unsteady and spastic gait, hyperreflexia of the lower limbs, and extensor plantar responses. Upper limbs spasticity and dystonia, subclinical axonal neuropathy, cognitive impairment and intellectual disability have also been associated.

Your genetic map

Gene	SNP	Genotype
LOC1079	rs397514513	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=320411

Hereditary Diseases (genetics)

Autosomal recessive spastic paraplegia type 5A

Autosomal recessive spastic paraplegia type 5A is a form of hereditary spastic paraplegia characterized by either a pure phenotype of slowly progressive spastic paraplegia of the lower extremities with bladder dysfunction and pes cavus or a complex presentation with additional manifestations including cerebellar signs, nystagmus, distal or generalized muscle atrophy and cognitive impairment. Age of onset is highly variable, ranging from early childhood to adulthood. White matter hyperintensity and cerebellar and spinal cord atrophy may be noted, on brain magnetic resonance imaging, in some patients.

Your genetic map

Gene	SNP	Genotype
CYP7B1	rs121908611	CC
CYP7B1	rs121908613	AA
CYP7B1	rs116171274	GG
CYP7B1	rs587777222	TT

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=100986

Hereditary Diseases (genetics)

Spastic paraplegia type 2

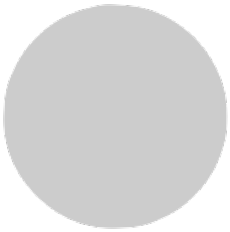
A rare, X-linked leukodystrophy characterized primarily by spastic gait and autonomic dysfunction. When additional central nervous system (CNS) signs, such as intellectual deficit, ataxia, or extrapyramidal signs, are present, the syndrome is referred to as complicated SPG.

Your genetic map

Gene	SNP	Genotype
RAB9B	rs132630292	GG
RAB9B	rs132630294	CC
RAB9B	rs398123467	GG
RAB9B	rs864622194	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=99015

Hereditary Diseases (genetics)

Spastic paraplegia type 7

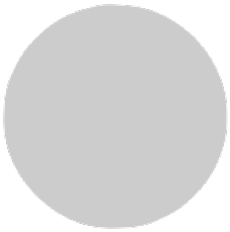
A form of hereditary spastic paraplegia characterized by an onset usually in adulthood (but ranging from 10-72 years) of progressive bilateral lower limb weakness and spasticity, sphincter dysfunction, decreased vibratory sense at the ankles and with additional manifestations including optical neuropathy, nystagmus, strabismus, decreased hearing, scoliosis, pes cavus, motor and sensory neuropathy, amyotrophy, blepharoptosis and ophthalmoplegia.

Your genetic map

Gene	SNP	Genotype
SPG7	rs121918358	TT
SPG7	rs369227537	AA
SPG7	rs752623413	TT
SPG7	rs748555510	CC
SPG7	rs748309520	GG
SPG7	rs72547551	CC
SPG7	rs864622094	TT
SPG7	rs141644720	GG
SPG7	rs779055639	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=99013

Hereditary Diseases (genetics)

Pycnodysostosis

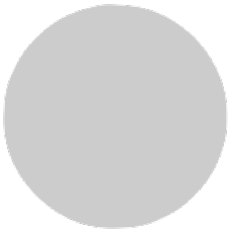
Pycnodysostosis is a genetic lysosomal disease characterized by osteosclerosis of the skeleton, short stature and brittle bones.

Your genetic map

Gene	SNP	Genotype
CTSK	rs74315303	GG
CTSK	rs74315304	GG
CTSK	rs29001685	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=763

Hereditary Diseases (genetics)

Familial clubfoot with or without associated lower limb anomalies

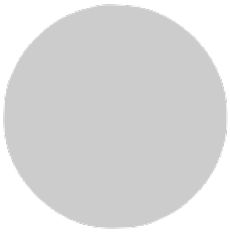
Familial clubfoot with or without associated lower limb anomalies is a rare congenital limb malformation syndrome characterized by malalignment of the bones and joints of the foot and ankle, with presence of forefoot and midfoot adductus, hindfoot varus, and ankle equinus, presenting as rigid inward turning of the foot towards the midline, in various members of a single family. Hypoplasia of lower leg muscles is a frequently associated finding. Patients may present with other low-limb malformations, such as patellar hypoplasia, oblique talus, tibial hemimelia, and polydactyly.

Your genetic map

Gene	SNP	Genotype
BLTP1	rs775292946	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=199315

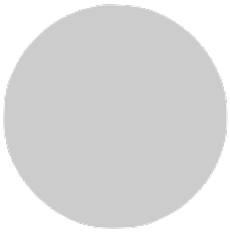
Hereditary Diseases (genetics)

PMM2-CDG

PMM2-CDG is the most frequent form of congenital disorder of N-glycosylation and is characterized by cerebellar dysfunction, abnormal fat distribution, inverted nipples, strabismus and hypotonia. 3 forms of PMM2-CDG can be distinguished: the infantile multisystem type, late-infantile and childhood ataxia-intellectual disability type (3 -10 yrs old), and the adult stable disability type. Infants usually develop ataxia, psychomotor delay and extraneurological manifestations including failure to thrive, enteropathy, hepatic dysfunction, coagulation abnormalities and cardiac and renal involvement. The phenotype is however highly variable and ranges from infants who die in the first year of life to mildly involved adults.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79318

Your genetic map

Gene	SNP	Genotype
LOC1001	rs78290141	AA
LOC1001	rs80338708	CC
LOC1001	rs80338709	GG
LOC1001	rs80338707	GG
PMM2	rs104894526	CC
PMM2	rs80338701	CC
PMM2	rs80338704	AA
PMM2	rs80338702	TT
PMM2	rs80338700	CC
PMM2	rs104894534	TT
PMM2	rs80338703	GG
PMM2	rs200503569	CC
PMM2	rs398123309	GG
PMM2	rs190521996	TT
PMM2	rs150719105	TT
PMM2	rs148032587	GG
PMM2	rs139716296	TT
PMM2	rs764353860	CC
TMEM186	rs104894532	GG

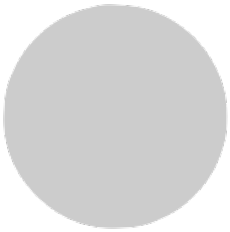
Hereditary Diseases (genetics)

Bilateral polymicrogyria

Bilateral polymicrogyria is a rare cerebral malformation due to abnormal neuronal migration defined as a cerebral cortex with many excessively small convolutions. It presents with developmental delay, intellectual disability, seizures and various neurological impairments and may be isolated or comprise a clinical feature of many genetic syndromes. It may also be associated with perinatal cytomegalovirus infection.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=268940

Your genetic map

Gene	SNP	Genotype
ADGRG1	rs587776623	GG
ADGRG1	rs121908462	CC
ADGRG1	rs121908464	CC
ADGRG1	rs121908465	GG
ADGRG1	rs587783658	CC
ADGRG1	rs146278035	CC
ADGRG1	rs587783660	GG
ADGRG1	rs532188689	GG
ADGRG1	rs587783652	CC
ADGRG1	rs587783654	TT
ADGRG1	rs587783655	TT
ADGRG1	rs587783656	GG
ADGRG1	rs587783657	GG
ADGRG1	rs786204777	CC

Hereditary Diseases (genetics)

Polymicrogyria due to TUBB2B mutation

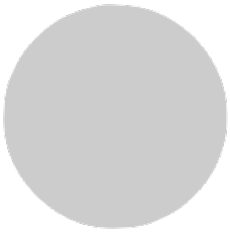
A rare, genetic, complex cerebral cortical malformation characterized by generalized or focal dysgyria (also named polymicrogryia-like cortical dysplasia) or alternatively by microlissencephaly with dysmorphic basal ganglia and dysgenesis of the corpus callosum. Clinical manifestations are variable and include microcephaly, seizures, hypotonia, developmental delay, severe psychomotor delay, ataxia, spastic diplegia or tetraplegia, and ocular abnormalities (strabismus, ptosis or optic atrophy).

Your genetic map

Gene	SNP	Genotype
TUBB2B	rs397514569	AA
TUBB2B	rs587784498	CC
TUBB2B	rs587784502	GG
TUBB2B	rs797046075	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=300573

Hereditary Diseases (genetics)

Autosomal recessive spastic ataxia of Charlevoix-Saguenay

Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) is a neurodegenerative disorder characterised by early-onset cerebellar ataxia with spasticity, a pyramidal syndrome and peripheral neuropathy.

Your genetic map

Gene	SNP	Genotype
SACS	rs281865118	GG
SACS	rs281865120	GG
SACS	rs780247476	GG
SACS	rs752059006	GG
SACS	rs202199411	GG
SACS	rs145766983	GG

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=98

Hereditary Diseases (genetics)

Syndactyly type 2

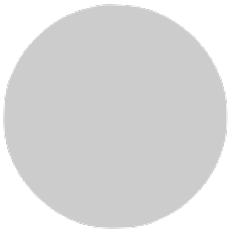
A rare non-syndromic syndactyly characterized by a distinctive combination of syndactyly and polydactyly, generally affecting the 3rd and 4th fingers and the 4th and 5th toes, bilaterally, with partial or complete reduplication of a digital ray within the syndactylous web. Additional features include 5th finger clinodactyly, camptodactyly and/or brachydactyly.

Your genetic map

Gene	SNP	Genotype
HOXD13	rs200750564	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=295195

Hereditary Diseases (genetics)

Porencephaly

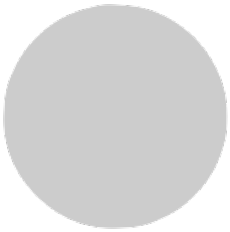
A rare, genetic or acquired, cerebral malformation characterized by an intracerebral fluid-filled cyst or cavity with or without communication between the ventricle and subarachnoid space. Clinical manifestations depend on location and severity and may include hemiparesis, seizures, intellectual disability, and dystonia.

Your genetic map

Gene	SNP	Genotype
COL4A1	rs587780588	CC
COL4A1	rs797044867	CC
COL4A1	rs797045034	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2940

Hereditary Diseases (genetics)

Acute intermittent porphyria

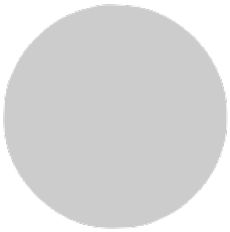
A rare, severe form of the acute hepatic porphyrias characterized by the occurrence of neuro-visceral attacks without cutaneous manifestations.

Your genetic map

Gene	SNP	Genotype
HMBS	rs118204095	GG
HMBS	rs118204101	CC
HMBS	rs118204109	CC
HMBS	rs118204120	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79276

Hereditary Diseases (genetics)

Hepatoerythropoietic porphyria

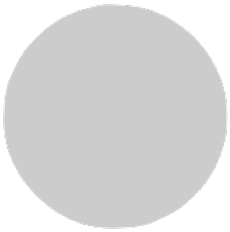
Hepatoerythropioetic porphyria (HEP) is a very rare form of chronic hepatic porphyria characterized by bullous photodermatitis.

Your genetic map

Gene	SNP	Genotype
UROD	rs121918065	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=443062

Hereditary Diseases (genetics)

Congenital erythropoietic porphyria

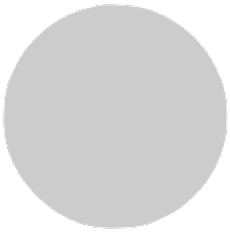
Congenital erythropoietic porphyria, or Gunther disease, is a form of erythropoietic porphyria characterized by very severe and mutilating photodermatosis.

Your genetic map

Gene	SNP	Genotype
UROS	rs121908012	AA
UROS	rs121908014	GG
UROS	rs121908015	GG
UROS	rs121908020	CC
UROS	rs373864821	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79277

Hereditary Diseases (genetics)

Lipoid proteinosis

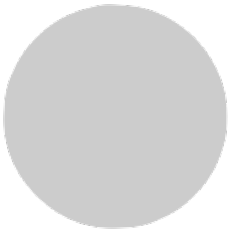
Lipoid proteinosis (LP) is a rare genodermatosis characterized clinically by mucocutaneous lesions, hoarseness developing in early childhood and, at times, neurological complications.

Your genetic map

Gene	SNP	Genotype
ECM1	rs121909115	CC
ECM1	rs121909116	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=530

Hereditary Diseases (genetics)

Autosomal erythropoietic protoporphyria

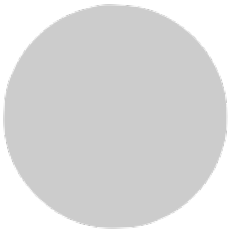
Erythropoietic protoporphyria (EPP) is an inherited disorder of the heme metabolic pathway characterized by accumulation of protoporphyrin in blood, erythrocytes and tissues, and cutaneous manifestations of photosensitivity.

Your genetic map

Gene	SNP	Genotype
FECH	rs150146721	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79278

Hereditary Diseases (genetics)

Pseudohypoparathyroidism type 1C

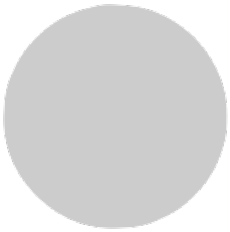
Pseudohypoparathyroidism type 1c (PHP1c) is a rare type of pseudohypoparathyroidism (PHP; see this term) characterized by resistance to parathyroid hormone (PTH) and other hormones, which manifests with hypocalcemia, hyperphosphatemia and elevated PTH levels, a constellation of clinical features collectively termed Albright's hereditary osteodystrophy (AHO; see this term), but normal activity of the stimulatory protein G (Gs alpha).

Your genetic map

Gene	SNP	Genotype
GNAS	rs397514456	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79444

Hereditary Diseases (genetics)

Pseudopseudohypoparathyroidism

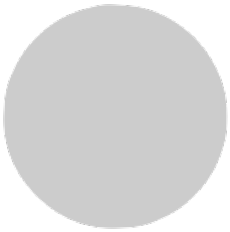
Pseudopseudohypoparathyroidism (pseudo-PHP) is a disease characterized by a constellation of clinical features collectively termed Albright hereditary osteodystrophy (AHO; see this term) but no evidence of resistance to parathyroid hormone (PTH), which is seen in other forms of pseudohypoparathyroidism (PHP; see this term).

Your genetic map

Gene	SNP	Genotype
GNAS	rs797045046	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79445

Hereditary Diseases (genetics)

Familial male-limited precocious puberty

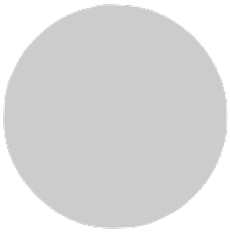
Familial male limited precocious puberty (FMPP) is a gonadotropin-independent familial form of male-limited precocious puberty, generally presenting between 2-5 years of age as accelerated growth, early development of secondary sexual characteristics and reduced adult height.

Your genetic map

Gene	SNP	Genotype
STON1	rs121912532	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3000

Hereditary Diseases (genetics)

Thrombotic thrombocytopenic purpura

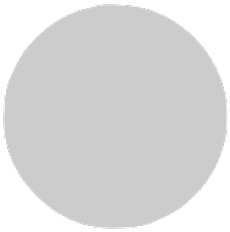
An aggressive and life-threatening form of thrombotic microangiopathy (TMA) characterized by profound peripheral thrombocytopenia, microangiopathic hemolytic anemia (MAHA) and organ failure of variable severity and is comprised of a congenital (cTTP) and acquired, immune-mediated (iTTP) form.

Your genetic map

Gene	SNP	Genotype
ADAMTS1	rs121908470	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=54057

Hereditary Diseases (genetics)

Striate palmoplantar keratoderma

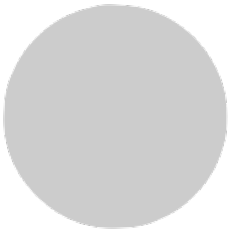
Striate palmoplantar keratoderma is an isolated, focal, hereditary palmoplantar keratoderma characterized by linear hyperkeratosis along the flexor aspect of the fingers and on palms, as well as focal hyperkeratosis of the plantar skin. Patients present with painful thickening of the skin on palms and soles, with occasional fissuring, blistering and hyperhidrosis. Rarely, hyperkeratosis on other areas may be seen (knees, dorsal aspects of the digits). Histopatologically, widened intercellular spaces between keratinocytes are observed.

Your genetic map

Gene	SNP	Genotype
DSP	rs121912991	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=50942

Hereditary Diseases (genetics)

Autosomal dominant focal non-epidermolytic palmoplantar keratoderma with plantar blistering

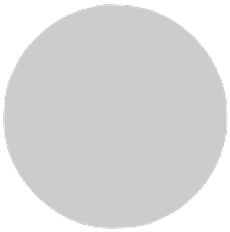
A rare, genetic, isolated, focal palmoplantar keratoderma disease characterized by focal thickening of the skin of the soles, and often of the palms, associated with minimal or no nail involvement. Patients frequently present non-epidermolytic painful plantar blistering and, occasionally, subtle oral leukokeratosis or plantar hyperhidrosis.

Your genetic map

Gene	SNP	Genotype
KRT6C	rs587777292	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=402003

Hereditary Diseases (genetics)

Isolated focal non-epidermolytic palmoplantar keratoderma

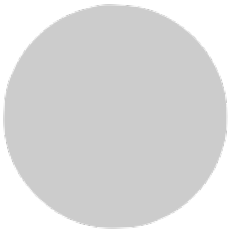
A rare hereditary palmoplantar keratoderma characterized by focal hyperkeratotic lesions on the palms and soles. Histopathologic examination reveals prominent hyperkeratosis, thickened stratum spinosum with reduced stratum granulosum, disadhesion of cells in the suprabasal layers, elongation of rete ridges, and sparse lymphocyte infiltration in the dermis.

Your genetic map

Gene	SNP	Genotype
KRT16	rs59856285	GG
KRT16	rs60723330	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=448264

Hereditary Diseases (genetics)

Palmoplantar keratoderma, Nagashima type

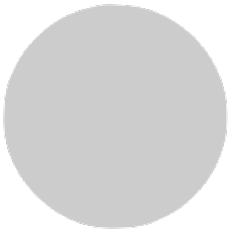
A rare autosomal recessive, isolated diffuse palmoplantar keratoderma charactized by transgressive and nonprogressive palmoplantar keratoderma resembling a mild form of mal de Meleda.

Your genetic map

Gene	SNP	Genotype
SERPINB7	rs142859678	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=140966

Hereditary Diseases (genetics)

Transgrediens et progrediens palmoplantar keratoderma

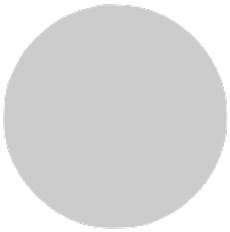
A rare, isolated, diffuse palmoplantar keratoderma disorder characterized by red-yellow, moderate to severe hyperkeratosis of the palms and soles, extending to the dorsal aspects of the hands, feet and/or wrists and involving the skin over the Achilles' tendon (transgrediens), gradually worsening with age (progrediens) to include patchy hyperkeratosis over the shins, knees, elbows and, sometimes, skin flexures. Hyperhidrosis is usually associated. Histologically, either epidermolytic or nonepidermolytic changes may be seen.

Your genetic map

Gene	SNP	Genotype
LOC1053	rs148182439	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=495

Hereditary Diseases (genetics)

Keratoderma hereditarium mutilans

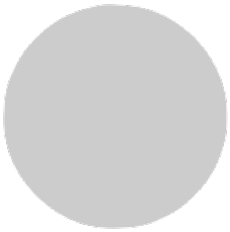
Keratosis follicularis spinulosa decalvans is a rare genodermatosis occurring during infancy or childhood, predominantly affecting males, and characterized by diffuse follicular hyperkeratosis associated with progressive cicatricial alopecia of the scalp, eyebrows and eyelashes. Additional findings can include photophobia, corneal dystrophy, facial erythema, and/or palmoplantar keratoderma.

Your genetic map

Gene	SNP	Genotype
MBTPS2	rs587776867	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2340

Hereditary Diseases (genetics)

Hypocalcemic vitamin D-dependent rickets

An early-onset hereditary vitamin D metabolism disorder characterized by severe hypocalcemia leading to osteomalacia and rachitic bone deformations, and moderate hypophosphatemia.

Your genetic map

Gene	SNP	Genotype
CYP27B1	rs28934604	CC
CYP27B1	rs118204008	GG
CYP27B1	rs118204009	CC

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=289157

Hereditary Diseases (genetics)

Autosomal dominant hypophosphatemic rickets

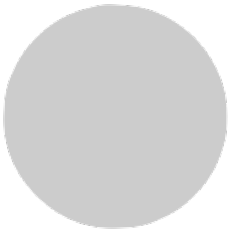
A rare hereditary renal phosphate-wasting disorder characterized by hypophosphatemia, rickets and/or osteomalacia.

Your genetic map

Gene	SNP	Genotype
FGF23	rs28937882	GG
FGF23	rs193922701	CC
FGF23	rs193922702	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=89937

Hereditary Diseases (genetics)

Hereditary hypophosphatemic rickets with hypercalciuria

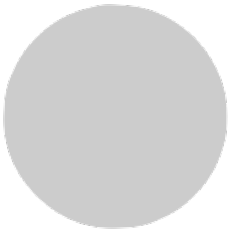
Hereditary hypophosphatemic rickets with hypercalciuria (HHRH) is a hereditary renal phosphate-wasting disorder characterized by hypophosphatemia and hypercalciuria associated with rickets and/or osteomalacia.

Your genetic map

Gene	SNP	Genotype
SLC34A3	rs201293634	TT
SLC34A3	rs150841256	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=157215

Hereditary Diseases (genetics)

Resistance to thyroid hormone due to a mutation in thyroid hormone receptor beta

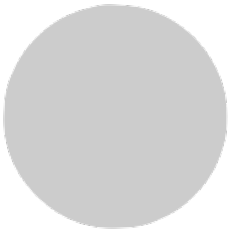
A rare genetic hyperthyroidism characterized by elevated levels of circulating free thyroid hormones, normal or elevated thyroid-stimulating hormone, decreased peripheral tissue responses to iodothyronine action, and a highly variable clinical phenotype which most commonly includes goiter, resting tachycardia, osteoporosis, short stature, and attention deficit disorder. Some patients may be entirely asymptomatic.

Your genetic map

Gene	SNP	Genotype
THRB	rs121918695	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=165994

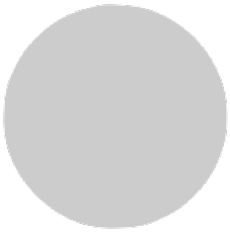
Hereditary Diseases (genetics)

Retinoblastoma

A rare eye tumor disease representing the most common intraocular malignancy in children. It is a life threatening neoplasia but is potentially curable and it can be hereditary or non hereditary, unilateral or bilateral.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=790

Your genetic map

Gene	SNP	Genotype
RB1	rs587776780	TT
RB1	rs3092891	CC
RB1	rs137853293	CC
RB1	rs121913301	AA
RB1	rs137853294	CC
RB1	rs121913304	CC
RB1	rs137853296	TT
RB1	rs137853297	TT
RB1	rs483352690	GG
RB1	rs587778864	CC
RB1	rs121913305	CC
RB1	rs587778871	GG
RB1	rs587778850	GG
RB1	rs587778839	TT
RB1	rs121913296	GG
RB1	rs587778870	CC
RB1	rs587778842	CC
RB1	rs121913300	CC
RB1	rs587776783	GG
RB1	rs587778831	GG
RB1	rs587778846	GG
RB1	rs121913302	CC
RB1	rs121913303	CC
RB1	rs794727199	GG
RB1	rs794727481	GG
RB1	rs878853947	TT
RB1	rs878853949	CC
RB1	rs886043247	CC
RB1	rs1060503088	TT
RB1	rs1060503067	GG
RB1	rs1060503079	CC

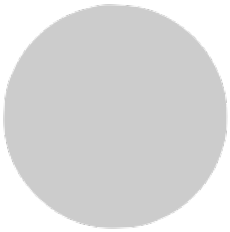
Hereditary Diseases (genetics)

X-linked retinoschisis

A rare disorder involving multiple structure of the eye characterized by reduced visual acuity in males due to juvenile macular degeneration. Clinical features such as vitreous hemorrhage, retinal detachment, and neovascular glaucoma can be observed in advanced stages.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=792

Your genetic map

Gene	SNP	Genotype
CDKL5	rs61752063	AA
CDKL5	rs61752067	GG
CDKL5	rs104894928	CC
CDKL5	rs104894933	CC
CDKL5	rs104894934	CC
CDKL5	rs104894929	AA
CDKL5	rs104894930	GG
CDKL5	rs61752068	CC
CDKL5	rs61752060	TT
CDKL5	rs61752147	CC
CDKL5	rs61752159	CC
CDKL5	rs281865348	CC
CDKL5	rs61753174	GG
CDKL5	rs281865357	GG
CDKL5	rs281865365	GG

Hereditary Diseases (genetics)

Sebocystomatosis

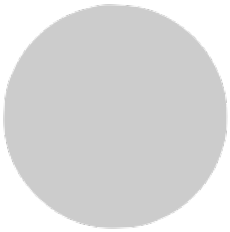
Sebocystomatosis is characterized by multiple (100 to 2000) asymptomatic dermal cysts that usually occur on the sternal region, upper back, axillae and proximal parts of the extremities.

Your genetic map

Gene	SNP	Genotype
KRT17	rs58730926	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=841

Hereditary Diseases (genetics)

3M syndrome

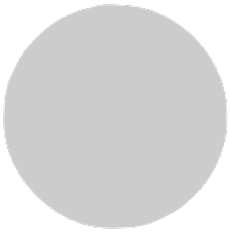
A rare primordial growth disorder characterized by low birth weight, reduced birth length, severe postnatal growth restriction, large head size, a spectrum of minor anomalies (including facial dysmorphism) and normal intelligence.

Your genetic map

Gene	SNP	Genotype
CUL7	rs121918229	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2616

Hereditary Diseases (genetics)

Acrocallosal syndrome

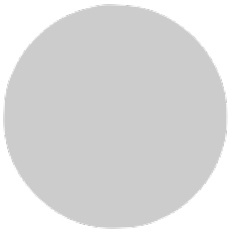
A rare polymalformative syndrome characterized by agenesis of corpus callosum (CC), distal anomalies of limbs, minor craniofacial anomalies and intellectual disability.

Your genetic map

Gene	SNP	Genotype
KIF7	rs794727316	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=36

Hereditary Diseases (genetics)

ADNP syndrome

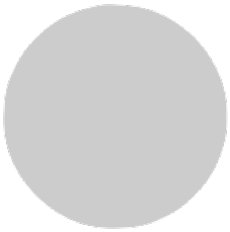
A rare syndromic intellectual disability characterized by global developmental delay, gastrointestinal problems, hypotonia, delayed speech, behavioral and sleep problems, pain insensitivity, seizures, structural brain anomalies, dysmorphic features, visual problems, early tooth eruption and autistic features.

Your genetic map

Gene	SNP	Genotype
ADNP	rs587777526	GG
ADNP	rs886041116	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=404448

Hereditary Diseases (genetics)

ADULT syndrome

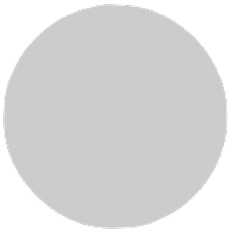
A rare ectodermal dysplasia syndrome characterized by ectrodactyly, syndactyly, mammary hypoplasia, and excessive freckling as well as other typical ectodermal defects such as hypodontia, lacrimal duct anomalies, hypotrichosis, and onychodysplasia.

Your genetic map

Gene	SNP	Genotype
TP63	rs113993967	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=978

Hereditary Diseases (genetics)

Auriculocondylar syndrome

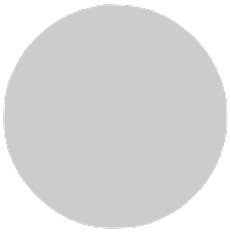
A rare, genetic dysostosis with predominant craniofacial involvement characterized by bilateral external ear malformations, mandibular condyle hypoplasia, microstomia, micrognathia, microglossia and facial asymmetry. Additional manifestations include hypotonia, ptosis, cleft palate, full cheeks, developmental delay, hearing impairment and respiratory distress. Significant intra- and interfamilial phenotypic variation has been reported.

Your genetic map

Gene	SNP	Genotype
GNAI3	rs387907178	GG
PLCB4	rs387907179	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=137888

Hereditary Diseases (genetics)

Autosomal dominant intellectual disability-craniofacial anomalies-cardiac defects syndrome

A rare genetic neurodevelopmental disorder characterized by global developmental delay (DD) and variable degrees of intellectual disability (ID) with delayed or limited/absent speech development associated with neonatal hypotonia, feeding difficulties, cardiac anomalies and dysmorphic facial features, predominantly broad nasal tip and thin, tented upper lip. Microcephaly, frequent infections, gastrointestinal and/or ocular anomalies have also been described.

Your genetic map

Gene	SNP	Genotype
KAT6A	rs786200960	GG

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=457193

Hereditary Diseases (genetics)

BOR syndrome

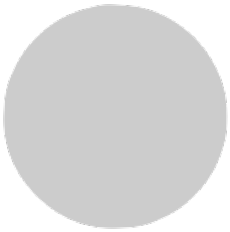
Branchiootorenal (BOR) syndrome is characterized by branchial arch anomalies (branchial clefts, fistulae, cysts), hearing impairment (malformations of the auricle with pre-auricular pits, conductive or sensorineural hearing impairment), and renal malformations (urinary tree malformation, renal hypoplasia or agenesis, renal dysplasia, renal cysts).

Your genetic map

Gene	SNP	Genotype
EYA1	rs121909195	GG
EYA1	rs121909196	CC
EYA1	rs606231357	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=107

Hereditary Diseases (genetics)

Branchio-oculo-facial syndrome

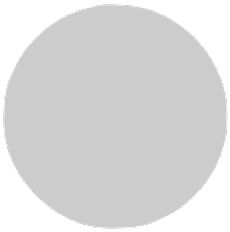
A rare, dominantly inherited multiple congenital anomalies syndrome characterized by highly variable clinical phenotype involving the three main affected systems: branchial (cutaneous) defects, ophthalmic malformations and facial anomalies. Additional features can be present.

Your genetic map

Gene	SNP	Genotype
TFAP2A	rs793888540	GG
TFAP2A	rs793888541	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1297

Hereditary Diseases (genetics)

Branchiootic syndrome

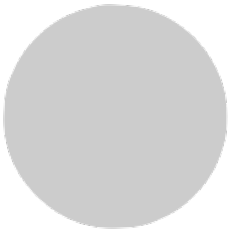
Branchiootic syndrome is a rare, genetic multiple congenital anomalies syndrome characterized by second branchial arch anomalies (branchial cysts and fistulae), malformations of the outer, middle and inner ear associated with sensorineural, mixed or conductive hearing loss, and the absence of renal abnormalities. Typical ear findings consist of malformed auricles (e.g. lop or cupped ears), preauricular pits and/or tags, and middle and/or inner ear dysplasias (inculding cochlear, vestibular and semicircular channel hypoplasia, malformation of the ossicles and of middle ear space).

Your genetic map

Gene	SNP	Genotype
EYA1	rs397517917	CC
LOC1053	rs397517920	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=52429

Hereditary Diseases (genetics)

CACH syndrome

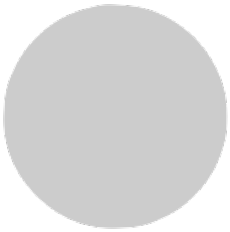
A new leukoencephalopathy, the CACH syndrome (Childhood Ataxia with Central nervous system Hypomyelination) or VWM (Vanishing White Matter) was identified on clinical and MRI criteria. Classically, this disease is characterized by (1) an onset between 2 and 5 years of age, with a cerebello-spastic syndrome exacerbated by episodes of fever or head trauma leading to death after 5 to 10 years of disease evolution, (2) a diffuse involvement of the white matter on cerebral MRI with a CSF-like signal intensity (cavitation), (3) a recessive autosomal mode of inheritance, (4) neuropathologic findings consistent with a cavitating orthochromatic leukodystrophy with increased number of oligodendrocytes with sometimes ``foamy'' aspect.

Your genetic map

Gene	SNP	Genotype
EIF2B2	rs104894425	AA
EIF2B2	rs104894426	TT
EIF2B2	rs113994012	GG
EIF2B5	rs113994049	GG
EIF2B5	rs113994054	GG
EIF2B5	rs113994053	CC
EIF2B5	rs113994048	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=157716

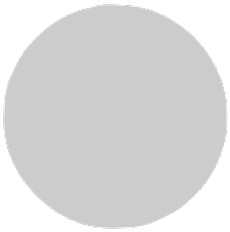
Hereditary Diseases (genetics)

Cardiofaciocutaneous syndrome

A rare, multiple congenital anomalies syndrome characterized by craniofacial dysmorphology, congenital heart disease, dermatological abnormalities (most commonly hyperkeratotic skin and sparse, curly hair), neurological manifestations (hypotonia, seizures), failure to thrive and intellectual disability.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1340

Your genetic map

Gene	SNP	Genotype
BRAF	rs113488022	AA
BRAF	rs121913348	CC
BRAF	rs180177034	CC
BRAF	rs121913364	TT
BRAF	rs121913355	CC
BRAF	rs180177035	TT
BRAF	rs180177036	CC
BRAF	rs180177037	TT
BRAF	rs180177038	CC
BRAF	rs180177039	TT
BRAF	rs180177040	TT
BRAF	rs180177042	AA
BRAF	rs387906661	TT
BRAF	rs397507465	TT
BRAF	rs397507466	TT
BRAF	rs397507469	GG
BRAF	rs397507473	AA
BRAF	rs397507474	TT
BRAF	rs397507475	AA
BRAF	rs397507476	TT
BRAF	rs397507479	CC
BRAF	rs397507480	AA
BRAF	rs397507481	GG
BRAF	rs397507483	CC
BRAF	rs121913375	GG
BRAF	rs397507484	TT
BRAF	rs397516892	GG
BRAF	rs397516893	AA
BRAF	rs397516894	GG
BRAF	rs397516895	AA
BRAF	rs397516903	AA

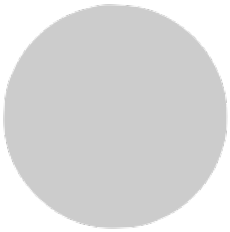
Hereditary Diseases (genetics)

CHARGE syndrome

CHARGE syndrome is a multiple congenital anomaly syndrome characterized by the variable combination of multiple anomalies, mainly Coloboma; Choanal atresia/stenosis; Cranial nerve dysfunction; Characteristic ear anomalies (known as the major 4 C's).

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=138

Your genetic map

Gene	SNP	Genotype
CHD7	rs121434338	AA
CHD7	rs267606724	CC
CHD7	rs398124321	GG
CHD7	rs587783428	GG
CHD7	rs587783429	CC
CHD7	rs587783432	GG
CHD7	rs587783433	TT
CHD7	rs587783434	GG
CHD7	rs587783440	CC
CHD7	rs587783441	AA
CHD7	rs587783442	CC
CHD7	rs587783445	TT
CHD7	rs587783446	CC
CHD7	rs587783447	GG
CHD7	rs587783448	AA
CHD7	rs587783450	CC
CHD7	rs587783451	AA
CHD7	rs587783454	CC
CHD7	rs587783457	CC
CHD7	rs587783458	CC
CHD7	rs587783459	GG
CHD7	rs794727293	CC
CHD7	rs794727423	GG
CHD7	rs794727569	GG
CHD7	rs797045467	CC
CHD7	rs864622523	AA
CHD7	rs886040983	CC
CHD7	rs768184220	AA
CHD7	rs886040991	CC
CHD7	rs757160222	CC
CHD7	rs886040995	CC

Hereditary Diseases (genetics)

CHILD syndrome

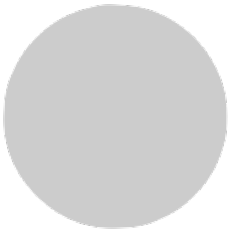
CHILD syndrome (Congenital Hemidysplasia with Ichthyosiform nevus and Limb Defects, CS) is an X-linked dominant genodermatosis characterized by unilateral inflammatory and scaling skin lesions with ipsilateral visceral and limb anomalies.

Your genetic map

Gene	SNP	Genotype
NSDHL	rs141571609	CC
NSDHL	rs587784226	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=139

Hereditary Diseases (genetics)

Classic glucose transporter type 1 deficiency syndrome

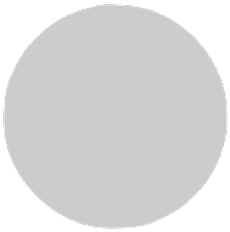
Glucose transporter type 1 (GLUT1) deficiency syndrome is characterized by an encephalopathy marked by childhood epilepsy that is refractory to treatment, deceleration of cranial growth leading to microcephaly, psychomotor retardation, spasticity, ataxia, dysarthria and other paroxysmal neurological phenomena often occurring before meals. Symptoms appear between the age of 1 and 4 months, following a normal birth and gestation.

Your genetic map

Gene	SNP	Genotype
SLC2A1	rs80359816	CC
SLC2A1	rs80359818	GG
SLC2A1	rs587784397	GG
SLC2A1	rs587784396	GG
SLC2A1	rs587784390	TT
SLC2A1	rs794727642	CC
SLC2A1	rs80359825	GG
SLC2A1	rs794729221	GG
SLC2A1	rs796053253	GG
SLC2A1	rs80359823	GG
SLC2A1	rs80359819	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=71277

Hereditary Diseases (genetics)

Congenital vertebral-cardiac-renal anomalies syndrome

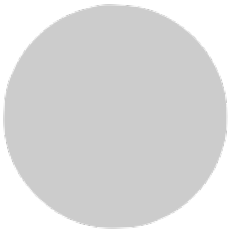
A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by vertebral segmentation defects associated with cardiac (patent ductus arteriosus, atrial septal defect, hypoplastic left heart) and renal (hypoplastic kidneys, chronic kidney disease) anomalies. Additional reported features include limb defects, short stature, global developmental delay, intellectual disability, and sensorineural hearing loss, among others.

Your genetic map

Gene	SNP	Genotype
NADSYN1	rs368115694	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=521438

Hereditary Diseases (genetics)

Constitutional mismatch repair deficiency syndrome

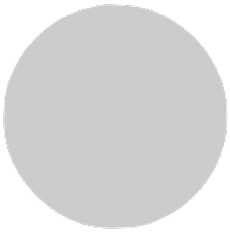
Constitutional mismatch repair deficiency syndrome is a rare, inherited cancer-predisposing syndrome characterized by the development of a broad spectrum of malignancies during childhood, including mainly brain, hematological and gastrointestinal cancers, although embryonic and other tumors have also been occasionally reported. Non-neoplastic features, in particular manifestations reminiscent of neurofibromatosis type 1 (e.g., cafe-au-lait spots, freckling, neurofibromas), as well as premalignant and non-malignant lesions (such as adenomas/polpyps) are frequently present before malignancy development.

Your genetic map

Gene	SNP	Genotype
PMS2	rs63750871	GG
PMS2	rs587779347	TT
PMS2	rs758304323	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=252202

Hereditary Diseases (genetics)

Heart-hand syndrome, Slovenian type

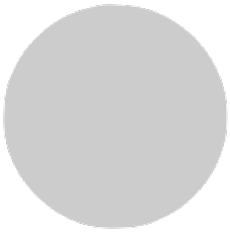
Heart-hand syndrome of Slovenian type is a rare autosomal dominant form of heart-hand syndrome (see this term), first described in members of a Slovenian family, that is characterized by adult onset, progressive cardiac conduction disease, tachyarrhythmias that can lead to sudden death, dilated cardiomyopathy and brachydactyly, with the hands less severely affected than the feet. Muscle weakness and/or myopathic electromyographic findings have been observed in some cases.

Your genetic map

Gene	SNP	Genotype
LMNA	rs386134243	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=168796

Hereditary Diseases (genetics)

Aarskog-Scott syndrome

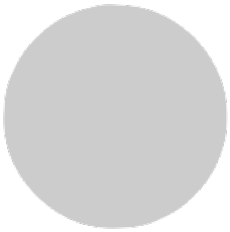
A rare developmental disorder characterized by facial, limbs and genital features, and a disproportionate acromelic short stature.

Your genetic map

Gene	SNP	Genotype
FGD1	rs28935497	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=915

Hereditary Diseases (genetics)

Adams-Oliver syndrome

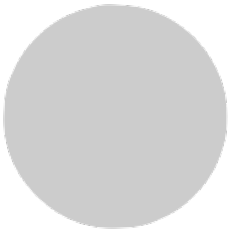
A rare disorder characterized by the combination of congenital limb abnormalities and scalp defects, often accompanied by skull ossification defects.

Your genetic map

Gene	SNP	Genotype
DLL4	rs796065350	GG
DLL4	rs796065348	CC
DLL4	rs796065347	TT
DLL4	rs796065346	GG
DLL4	rs796065345	CC
DLL4	rs61750844	CC
DOCK6	rs372751467	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=974

Hereditary Diseases (genetics)

Corpus callosum agenesis-neuronopathy syndrome

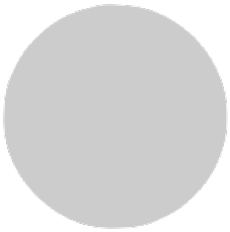
Corpus callosum agenesis-neuronopathy syndrome is a neurodegenerative disorder characterized by severe progressive sensorimotor neuropathy beginning in infancy with resulting hypotonia, areflexia, amyotrophy and variable degrees of dysgenesis of the corpus callosum. Additional features include mild-to-severe intellectual and developmental delays, and psychiatric manifestations that include paranoid delusions, depression, hallucinations, and 'autistic-like' features. Affected individuals are usually wheelchair restricted in the second decade of life and die in the third decade of life. The disease is inherited as an autosomal recessive trait.

Your genetic map

Gene	SNP	Genotype
SLC12A6	rs121908427	GG
SLC12A6	rs121908429	GG
SLC12A6	rs199747285	CC
SLC12A6	rs751184319	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1496

Hereditary Diseases (genetics)

Aicardi-Goutières syndrome

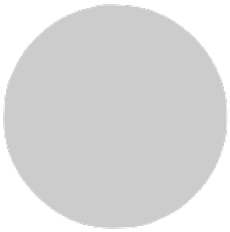
An inherited, subacute encephalopathy characterised by the association of basal ganglia calcification, leukodystrophy and cerebrospinal fluid (CSF) lymphocytosis.

Your genetic map

Gene	SNP	Genotype
TREX1	rs78218009	CC
TREX1	rs121908117	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=51

Hereditary Diseases (genetics)

Alagille syndrome

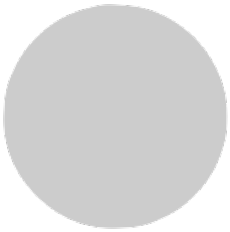
A rare syndrome variably characterized by chronic cholestasis due to paucity of intrahepatic bile ducts, peripheral pulmonary artery stenosis, vertebrae segmentation anomalies, characteristic facies, posterior embryotoxon/anterior segment abnormalities, pigmentary retinopathy, and dysplastic kidneys.

Your genetic map

Gene	SNP	Genotype
JAG1	rs121918351	CC
JAG1	rs863223655	GG
JAG1	rs863223649	GG
JAG1	rs863223648	CC
JAG1	rs876660980	GG
JAG1	rs886043603	GG
MIR6870	rs863223650	CC

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=52

Hereditary Diseases (genetics)

Alazami syndrome

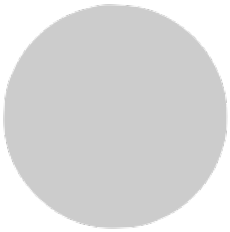
A rare form of primordial dwarfism, often microcephalic, characterized by short stature, global developmental delay, variable intellectual disability and recognizable dysmorphic facial features (triangular face, prominent forehead, deeply set eyes, low-set ears, wide nose, malar hypoplasia, wide mouth, thick lips, and widely spaced teeth).

Your genetic map

Gene	SNP	Genotype
MIR302C	rs775430086	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=319671

Hereditary Diseases (genetics)

Allan-Herndon-Dudley syndrome

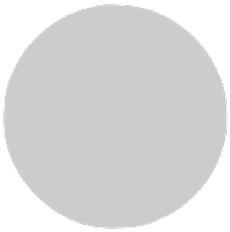
An X-linked intellectual disability syndrome with neuromuscular involvement characterized by infantile hypotonia, muscular hypoplasia, spastic paraparesis with dystonic/athetoic movements, and severe cognitive deficiency.

Your genetic map

Gene	SNP	Genotype
LOC1053	rs587784386	CC
SLC16A2	rs104894936	CC
SLC16A2	rs122455132	TT
SLC16A2	rs587784382	CC
SLC16A2	rs587784383	GG
SLC16A2	rs587784384	CC
SLC16A2	rs766773277	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=59

Hereditary Diseases (genetics)

Alpers-Huttenlocher syndrome

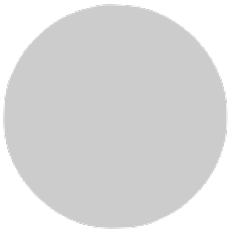
A cerebrohepatopathy and a rare and severe form of mitochondrial DNA (mtDNA) depletion syndrome characterized by the triad of progressive developmental regression, intractable seizures, and hepatic failure.

Your genetic map

Gene	SNP	Genotype
FANCI	rs139562274	GG
POLG	rs121918049	CC
POLG	rs548076633	TT
POLG	rs56047213	CC
POLG	rs201732356	GG
POLG	rs796052888	CC
POLG	rs796052887	CC
POLG	rs796052906	GG
POLG	rs769410130	GG
POLG	rs753160398	GG
POLG	rs139590686	TT
POLG	rs142347031	AA
POLG	rs140079523	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=726

Hereditary Diseases (genetics)

Andersen-Tawil syndrome

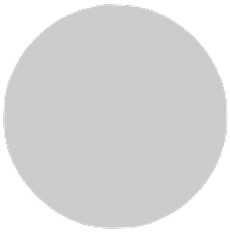
A rare disorder characterized by periodic muscle paralysis, prolongation of the QT interval with a variety of ventricular arrhythmias (leading to predisposition to sudden cardiac death) and characteristic physical features: short stature, scoliosis, low-set ears, hypertelorism, broad nasal root, micrognathia, clinodactyly, brachydactyly and syndactyly.

Your genetic map

Gene	SNP	Genotype
KCNJ2	rs104894578	CC
KCNJ2	rs104894579	GG
KCNJ2	rs104894580	CC
KCNJ2	rs104894585	CC
KCNJ2	rs199473373	CC
KCNJ2	rs199473381	GG
KCNJ2	rs199473384	GG
KCNJ2	rs786205817	AA
KCNJ2	rs786205820	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=37553

Hereditary Diseases (genetics)

Thiamine-responsive megaloblastic anemia syndrome

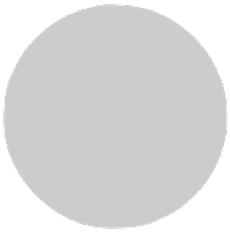
Thiamine-responsive megaloblastic anemia (TRMA) is characterized by a triad of megaloblastic anemia, non-type I diabetes mellitus, and sensorineural deafness.

Your genetic map

Gene	SNP	Genotype
SLC19A2	rs28937595	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=49827

Hereditary Diseases (genetics)

Aneurysm-osteoarthritis syndrome

A rare, genetic, systemic disease characterized by the presence of arterial aneurysms, tortuosity and dissection throughout the arterial tree, associated with early-onset osteoarthritis (predominantly affecting the spine, hands and/or wrists, and knees) and mild craniofacial dysmorphism (incl. long face, high forehead, flat supraorbital ridges, hypertelorism, malar hypoplasia and, a raphe, broad or bifid uvula), as well as mild skeletal and cutaneous anomalies. Joint abnormalities, such as osteochondritis dissecans and intervertebral disc degeneration, are frequently associated. Additional cardiovascular anomalies may include mitral valve defects, congenital heart malformations, ventricular hypertrophy and atrial fibrillation.

Your genetic map

Gene	SNP	Genotype
SMAD3	rs387906850	CC
SMAD3	rs387906853	GG

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=284984

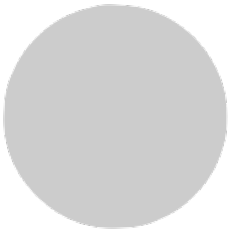
Hereditary Diseases (genetics)

Angelman syndrome

A neurogenetic disorder characterized by severe intellectual deficit and distinct facial dysmorphic features.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=72

Your genetic map

Gene	SNP	Genotype
MECP2	rs61748396	GG
MECP2	rs61754453	GG
SNHG14	rs111033595	CC
SNHG14	rs587780577	AA
SNHG14	rs587781208	CC
SNHG14	rs587781220	CC
SNHG14	rs587781241	GG
SNHG14	rs587782919	TT
SNHG14	rs587783097	GG
SNHG14	rs587784526	AA
SNHG14	rs587784518	TT
SNHG14	rs587784516	CC
SNHG14	rs587784515	AA
SNHG14	rs587784514	CC
SNHG14	rs587784508	CC
SNHG14	rs587784533	CC

Hereditary Diseases (genetics)

Anophthalmia/microphthalmia-esophageal atresia syndrome

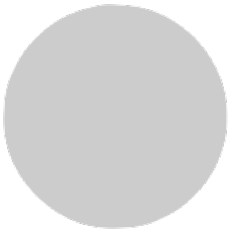
A syndrome that belongs to the group of syndromic microphthalmias and is characterized by the association of uni- or bilateral anophthalmia or microphthalmia, and esophageal atresia with or without trachoesophageal fistula.

Your genetic map

Gene	SNP	Genotype
SOX2 OT	rs55683010	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=77298

Hereditary Diseases (genetics)

Palatal anomalies-widely spaced teeth-facial dysmorphism-developmental delay syndrome

Palatal anomalies-widely spaced teeth-facial dysmorphism-developmental delay syndrome is a rare, genetic multiple congenital anomalies/dysmorphic syndrome characterized by global developmental delay, axial hypotonia, palate abnormalities (including cleft palate and/or high and narrow palate), dysmorphic facial features (including prominent forehead, hypertelorism, downslanting palpebral fissures, wide nasal bridge, thin lips and widely spaced teeth), and short stature. Additional manifestations may include digital anomalies (such as brachydactyly, clinodactyly, and hypoplastic toenails), a single palmar crease, lower limb hypertonia, joint hypermobility, as well as ocular and urogenital anomalies.

Your genetic map

Gene	SNP	Genotype
KDM1A	rs864309715	GG
KDM1A	rs864309716	AA

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=477993

Hereditary Diseases (genetics)

Antley-Bixler syndrome

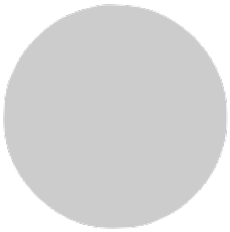
A rare syndromic craniosynostosis characterized by craniosynostosis with midface hypoplasia, radiohumeral synostosis, femoral bowing and joint contractures.

Your genetic map

Gene	SNP	Genotype
FGFR2	rs121918502	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=83

Hereditary Diseases (genetics)

Apert syndrome

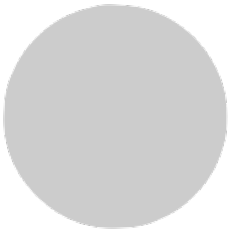
A frequent form of acrocephalosyndactyly, a group of inherited congenital malformation disorders, characterized by craniosynostosis, midface hypoplasia, and finger and toe anomalies and/or syndactyly.

Your genetic map

Gene	SNP	Genotype
FGFR2	rs79184941	GG
FGFR2	rs77543610	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=87

Hereditary Diseases (genetics)

Pyogenic arthritis-pyoderma gangrenosum-acne syndrome

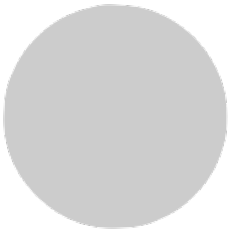
Pyogenic arthritis-pyoderma gangrenosum-acne syndrome is a rare pleiotropic autoinflammatory disorder of childhood, primarily affecting the joints and skin.

Your genetic map

Gene	SNP	Genotype
PSTPIP1	rs28939089	GG
PSTPIP1	rs121908130	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=69126

Hereditary Diseases (genetics)

Progeroid and marfanoid aspect-lipodystrophy syndrome

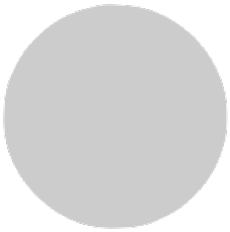
Progeroid and marfanoid aspect-lipodystrophy syndrome is a rare systemic disease characterized by a neonatal progeroid appearance (not associated with other manifestations of premature aging) associated with facial dysmorphism (e.g. macrocephaly or arrested hydrocephaly, proptosis, downslanting palpebral fissures, retrognathia), generalized, extreme, congenital lack of subcutaneous fat tissue (except in the breast and iliac region) and incomplete signs of Marfan syndrome (mainly severe myopia, joint hyperextensibility and arachnodactyly). Metabolic disturbances are not associated.

Your genetic map

Gene	SNP	Genotype
FBN1	rs398122833	CC
FBN1	rs794728325	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=300382

Hereditary Diseases (genetics)

Cerebellar ataxia-areflexia-pes cavus-optic atrophy-sensorineural hearing loss syndrome

Cerebellar ataxia - areflexia - pes cavus - optic atrophy - sensorineural hearing loss (CAPOS syndrome) is a rare autosomal dominant neurological disorder characterized by early onset cerebellar ataxia, associated with areflexia, progressive optic atrophy, sensorineural deafness, a pes cavus deformity, and abnormal eye movements.

Your genetic map

Gene	SNP	Genotype
ATP1A3	rs587777771	CC
ATP1A3	rs863224847	CC

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1171

Hereditary Diseases (genetics)

Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to WWOX deficiency

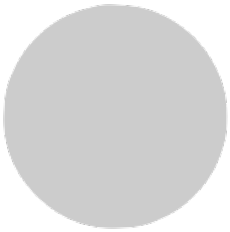
A rare autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome characterized by early-childhood onset of cerebellar ataxia associated with generalized tonic-clonic epilepsy and psychomotor development delay, dysarthria, gaze-evoked nystagmus and learning disability. Other features in some patients include upper motor neuron signs with leg spasticity and extensor plantar responses, and mild cerebellar atrophy on brain MRI.

Your genetic map

Gene	SNP	Genotype
WWOX	rs756762196	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=284282

Hereditary Diseases (genetics)

Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome

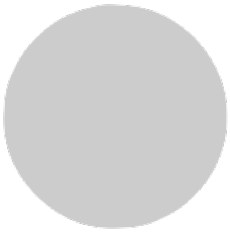
Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome is a rare hereditary spastic ataxia disorder characterized by childhood onset of slowly progressive lower limb spastic paraparesis and cerebellar ataxia (with dysarthria, swallowing difficulties, motor degeneration), associated with sensorimotor neuropathy (including muscle weakness and distal amyotrophy in lower extremities) and progressive myoclonic epilepsy. Ocular signs (ptosis, oculomotor apraxia), dysmetria, dysdiadochokinesia, dystonic movements and myoclonus may also be associated.

Your genetic map

Gene	SNP	Genotype
LOC1079	rs387906889	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=313772

Hereditary Diseases (genetics)

Ataxia-intellectual disability-oculomotor apraxia-cerebellar cysts syndrome

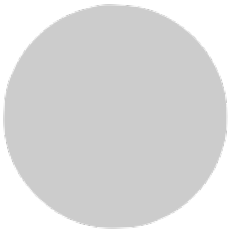
A rare neuro-ophthalmological disease characterized by nonprogressive cerebellar ataxia, delayed motor and language development and intellectual disability, in addition to ophthalmological abnormalities (e.g. oculomotor apraxia, strabismus, amblyopia, retinal dystrophy and myopia). Cerebellar cysts, cerebellar dysplasia and cerebellar vermis hypoplasia, seen on magnetic resonance imaging, are also characteristic of the disease.

Your genetic map

Gene	SNP	Genotype
LAMA1	rs587777677	AA
LAMA1	rs587777681	AA
LAMA1	rs797045184	CC
LAMA1	rs141914419	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=370022

Hereditary Diseases (genetics)

Spinal muscular atrophy-progressive myoclonic epilepsy syndrome

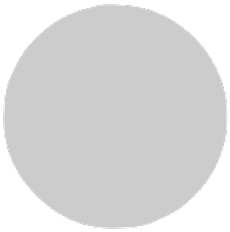
Spinal muscular atrophy-progressive myoclonic epilepsy syndrome is characterized by hereditary myoclonus and progressive distal muscular atrophy. Less than 10 cases have been reported. Treatment with clonazepam results in complete and lasting improvement of the myoclonus.

Your genetic map

Gene	SNP	Genotype
ASAH1	rs145873635	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2590

Hereditary Diseases (genetics)

Autosomal dominant optic atrophy plus syndrome

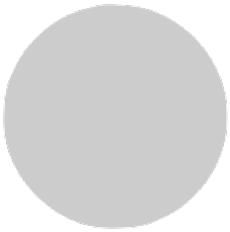
A rare neuro-ophthalmological disease associating the typical optic atrophy with other extra-ocular manifestations such as sensorineural deafness, myopathy, chronic progressive external ophthalmoplegia, ataxia and peripheral neuropathy. More rarely, other manifestations have been associated with this condition, such as spastic paraplegia or multiple-sclerosis like illness.

Your genetic map

Gene	SNP	Genotype
LOC1027	rs398124298	CC
OPA1	rs80356529	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1215

Hereditary Diseases (genetics)

Optic atrophy-intellectual disability syndrome

Optic atrophy-intellectual disability syndrome is a rare, hereditary, syndromic intellectual disability characterized by developmental delay, intellectual disability, and significant visual impairment due to optic nerve atrophy, optic nerve hypoplasia or cerebral visual impairment. Other common clinical signs and symptoms are hypotonia, oromotor dysfunction, seizures, autism spectrum disorder, and repetitive behaviors. Dysmorphic facial features are variable and nonspecific.

Your genetic map

Gene	SNP	Genotype
NR2F1	rs587777274	GG
NR2F1	rs587777275	CC
NR2F1	rs587777276	TT
NR2F1	rs587777277	GG
NR2F1	rs863224903	TT

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=401777

Hereditary Diseases (genetics)

Barth syndrome

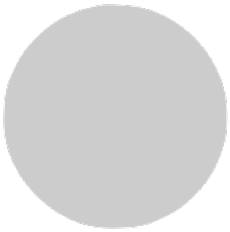
Barth syndrome (BTHS) is an inborn error of phospholipid metabolism characterized by dilated cardiomyopathy (DCM), skeletal myopathy, neutropenia, growth delay and organic aciduria.

Your genetic map

Gene	SNP	Genotype
TAFAZZIN	rs387907218	GG
TAFAZZIN	rs397515738	CC
TAFAZZIN	rs397515739	TT
TAFAZZIN	rs397515740	TT
TAFAZZIN	rs397515741	TT
TAFAZZIN	rs397515746	GG
TAFAZZIN	rs397515747	GG
TAFAZZIN	rs727504327	GG
TAFAZZIN	rs727504431	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=111

Hereditary Diseases (genetics)

Bartter syndrome

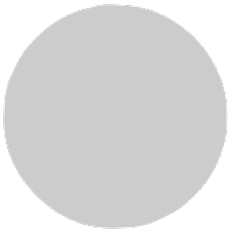
Bartter syndrome is a group of rare renal tubular disease characterized by impaired salt reabsorption in the thick ascending limb of Henle's loop and clinically by the association of hypokalemic alkalosis, hypercalciuria/nephrocalcinosis, increased levels of plasma renin and aldosterone, low blood pressure and vascular resistance to angiotensin II.

Your genetic map

Gene	SNP	Genotype
KCNJ1	rs377205432	GG
KCNJ1	rs746509804	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=112

Hereditary Diseases (genetics)

Beta-thalassemia-X-linked thrombocytopenia syndrome

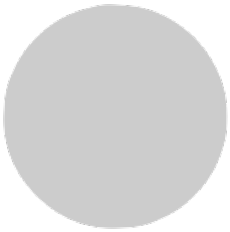
Beta-thalassemia - X-linked thrombocytopenia is a form of beta-thalassemia characterized by splenomegaly and petechiae, moderate thrombocytopenia, prolonged bleeding time due to platelet dysfunction, reticulocytosis and mild beta-thalassemia.

Your genetic map

Gene	SNP	Genotype
GATA1	rs104894809	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=231393

Hereditary Diseases (genetics)

Björnstad syndrome

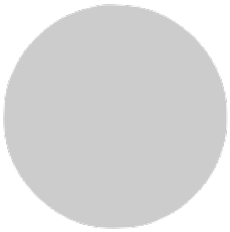
Björnstad syndrome is characterized by congenital sensorineural hearing loss and pili torti.

Your genetic map

Gene	SNP	Genotype
BCS1L	rs121908577	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=123

Hereditary Diseases (genetics)

Blau syndrome

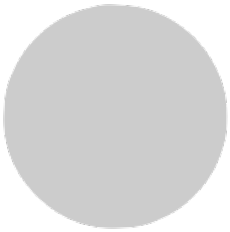
Blau syndrome (BS) is a rare systemic inflammatory disease characterized by early onset granulomatous arthritis, uveitis and skin rash. BS now refers to both the familial and sporadic (formerly early-onset sarcoidosis) form of the same disease. The proposed term pediatric granulomatous arthritis is currently questioned since it fails to represent the systemic nature of the disease.

Your genetic map

Gene	SNP	Genotype
NOD2	rs104895461	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=90340

Hereditary Diseases (genetics)

Bohring-Opitz syndrome

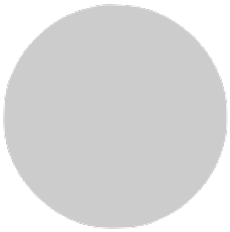
A rare multiple congenital anomalies syndrome characterized by intrauterine growth retardation (IUGR), postnatal failure to thrive, severe feeding difficulties, microcephaly/trigonocephaly, facial dysmorphism, a recognizable upper limb posture and severe developmental delay. The upper limb posture consists of internal rotation of the shoulders, flexion of the elbows, ulnar deviation of wrists and/or metacarpophalangeal joints.

Your genetic map

Gene	SNP	Genotype
ASXL1	rs373145711	CC
ASXL1	rs397515401	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=97297

Hereditary Diseases (genetics)

Borjeson-Forssman-Lehmann syndrome

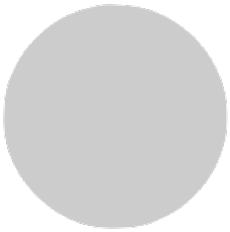
Borjeson-Forssman-Lehmann syndrome (BFLS) is a rare X-linked obesity syndrome characterized by intellectual deficit, truncal obesity, characteristic facial features, hypogonadism, tapered fingers and short toes.

Your genetic map

Gene	SNP	Genotype
PHF6	rs132630297	CC
PHF6	rs864309532	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=127

Hereditary Diseases (genetics)

Bosley-Salih-Alorainy syndrome

Bosley-Salih-Alorainy syndrome (BSAS) is characterized by variable horizontal gaze dysfunction, profound and bilateral sensorineural deafness associated commonly with severe inner ear maldevelopment, cerebrovascular anomalies (ranging from unilateral internal carotid artery hypoplasia to bilateral agenesis), cardiac malformation, developmental delay and occasionally autism. The syndrome is caused by homozygous mutations in the HOXA1 gene (7p15.2) and is transmitted in an autosomal recessive manner. The syndrome overlaps clinically and genetically with Athabaskan brain dysfunction syndrome (ABDS,). However unlike ABDS, BSAS does not manifest central hypoventilation.

Your genetic map

Gene	SNP	Genotype
HOTAIRM	rs104894018	GG

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=69737

Hereditary Diseases (genetics)

Bruck syndrome

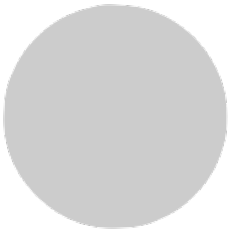
Bruck syndrome is characterised by the association of osteogenesis imperfecta and congenital joint contractures.

Your genetic map

Gene	SNP	Genotype
COL1A2	rs794727669	GG
FKBP10	rs387906960	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2771

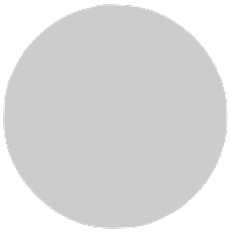
Hereditary Diseases (genetics)

Brugada syndrome

A cardiac disorder characterized on electrocardiogram (ECG) by ST segment elevation with a coved aspect on the right precordial leads, and a clinical susceptibility to ventricular tachyarrhythmias and sudden death occurring in the absence of overt myocardial abnormalities.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=130

Your genetic map

Gene	SNP	Genotype
FBN1 DT	rs886039072	CC
SCN5A	rs28937318	CC
SCN5A	rs137854611	GG
SCN5A	rs199473282	GG
SCN5A	rs199473579	CC
SCN5A	rs199473565	CC
SCN5A	rs199473153	CC
SCN5A	rs199473161	GG
SCN5A	rs199473168	GG
SCN5A	rs199473172	CC
SCN5A	rs199473055	GG
SCN5A	rs199473554	CC
SCN5A	rs199473556	GG
SCN5A	rs199473058	CC
SCN5A	rs199473598	CC
SCN5A	rs199473220	CC
SCN5A	rs199473225	GG
SCN5A	rs199473249	CC
SCN5A	rs199473613	TT
SCN5A	rs199473305	CC
SCN5A	rs199473083	CC
SCN5A	rs483353016	CC
SCN5A	rs786204839	AA
SCN5A	rs794728880	AA
SCN5A	rs794728879	CC
SCN5A	rs794728865	GG
SCN5A	rs794728849	GG
SCN5A	rs794728843	CC
SCN5A	rs794728846	CC
SCN5A	rs863224532	GG
SCN5A	rs863225273	CC

Hereditary Diseases (genetics)

Carney-Stratakis syndrome

Carney-Stratakis syndrome is a recently described familial syndrome characterized by gastrointestinal stromal tumors (GIST) and paragangliomas, often at multiple sites.

Your genetic map

Gene	SNP	Genotype
SDHB	rs587782703	CC
SDHC	rs587776653	GG
SDHD	rs786202403	CC
SDHD	rs786203932	GG
SDHD	rs1060503770	CC
SDHD	rs1050032491	TT
TIMM8B	rs587776644	TT
TIMM8B	rs80338842	GG
TIMM8B	rs587782210	CC

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=97286

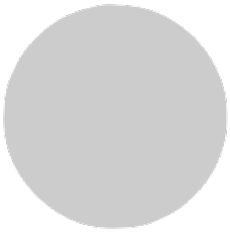
Hereditary Diseases (genetics)

Carvajal syndrome

A syndrome that is characterized by woolly hair, palmoplantar keratoderma and dilated cardiomyopathy principally affecting the left ventricle.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=65282

Your genetic map

Gene	SNP	Genotype
DSP	rs121912997	CC
DSP	rs397516946	CC
DSP	rs140474226	CC
DSP	rs730880081	GG
DSP	rs794728106	GG
DSP	rs149701627	CC
DSP	rs794728118	CC
DSP	rs777573018	CC
DSP	rs869025395	CC
DSP	rs876657638	CC
DSP	rs1057517903	GG
DSP	rs774514264	TT
DSP	rs778178956	CC
DSP	rs1304410089	GG
DSP	rs1236464864	TT
DSP	rs1267435790	CC
DSP	rs113726158	AA
DSP	rs1194358112	GG

Hereditary Diseases (genetics)

Congenital cataract-progressive muscular hypotonia-hearing loss-developmental delay syndrome

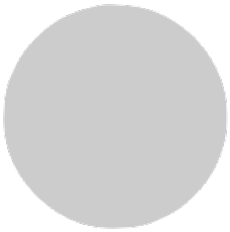
Congenital cataract-progressive muscular hypotonia-hearing loss-developmental delay syndrome is a rare, genetic, mitochondrial myopathy disorder characterized by congenital cataract, progressive muscular hypotonia that particularly affects the lower limbs, reduced deep tendon reflexes, sensorineural hearing loss, global development delay and lactic acidosis. Muscle biopsy reveals reduced complex I, II and IV respiratory chain activity.

Your genetic map

Gene	SNP	Genotype
GFER	rs121908192	GG
GFER	rs771809901	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=330054

Hereditary Diseases (genetics)

Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome

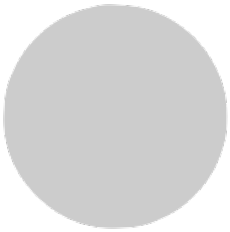
Congenital cataract - hypertrophic cardiomyopathy - mitochrondrial myopathy (CCM) is a mitochondrial disease characterized by cataracts, hypertrophic cardiomyopathy, muscle weakness and lactic acidosis after exercise.

Your genetic map

Gene	SNP	Genotype
AGK	rs387907025	CC
AGK	rs746709222	CC
AGK	rs863223895	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1369

Hereditary Diseases (genetics)

Chédiak-Higashi syndrome

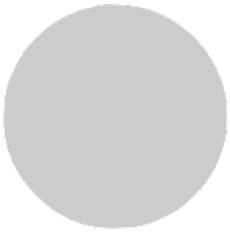
Chédiak-Higashi syndrome (CHS) is a rare severe genetic disorder generally characterized by partial oculocutaneous albinism (OCA, see this term), severe immunodeficiency, mild bleeding, neurological dysfunction and lymphoproliferative disorder. A classic, early-onset form and an attenuated, later-onset form (Atypical CHS; see this term) have been described.

Your genetic map

Gene	SNP	Genotype
LYST	rs80338652	GG
LYST	rs80338651	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=167

Hereditary Diseases (genetics)

Christianson syndrome

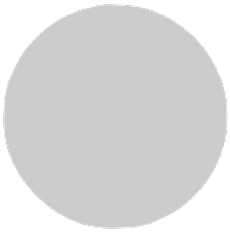
A rare developmental defect during embryogenesis characterized by intellectual deficit, ataxia, postnatal microcephaly, and hyperkinesis.

Your genetic map

Gene	SNP	Genotype
SLC9A6	rs122461162	CC
SLC9A6	rs398124224	CC
SLC9A6	rs587784399	TT
SLC9A6	rs797044508	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=85278

Hereditary Diseases (genetics)

Chudley-McCullough syndrome

Chudley-McCullough syndrome is a rare, genetic, syndromic deafness characterized by severe to profound, bilateral, sensorineural hearing loss (congenital or rapidly progressive in infancy) associated with a complex brain malformation including hydrocephalus, varying degrees of partial corpus callosum agenesis, colpocephaly, cerebral and cerebellar cortical dysplasia (bilateral medial frontal polymicrogyria, bilateral frontal subcortical heteropia) and, in some, arachnoid cysts. Major physical abnormalities or psychomotor delay are usually not associated.

Your genetic map

Gene	SNP	Genotype
GPSM2	rs145191476	CC
GPSM2	rs370907055	CC

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=314597

Hereditary Diseases (genetics)

Cockayne syndrome

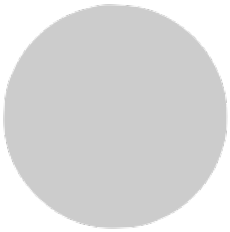
Cockayne syndrome (CS) is a multisystem condition characterized by short stature, a characteristic facial appearance, premature aging, photosensitivity, progressive neurological dysfunction, and intellectual deficit.

Your genetic map

Gene	SNP	Genotype
ERCC6	rs786205174	GG
ERCC6	rs373227647	TT
ERCC6	rs151242354	GG
ERCC6	rs202080674	GG
ERCC6	rs371739894	CC
ERCC6	rs368728467	AA
ERCC6	rs751838040	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=191

Hereditary Diseases (genetics)

Coffin-Lowry syndrome

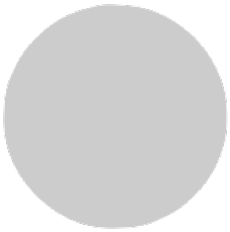
A rare X-linked syndromic intellectual disability characterized by global development delay, postnatal growth retardation leading to short stature, facial dysmorphism, short hands with tapering fingers and progressive skeletal abnormalities including kyphoscoliosis and pectus carinatum/excavatum. Intellectual disability ranges from mild to severe.

Your genetic map

Gene	SNP	Genotype
RPS6KA3	rs28935171	CC
RPS6KA3	rs398124177	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=192

Hereditary Diseases (genetics)

Atrial septal defect-atrioventricular conduction defects syndrome

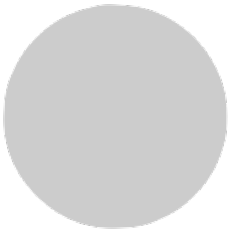
An extremely rare genetic congenital heart disease characterized by the presence of atrial septal defect, mostly of the ostium secundum type, associated with conduction anomalies like atrioventricular block, atrial fibrillation or right bundle branch block.

Your genetic map

Gene	SNP	Genotype
NKX2 5	rs104893901	GG
NKX2 5	rs72554028	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1479

Hereditary Diseases (genetics)

Lethal congenital contracture syndrome type 1

Lethal congenital contracture syndrome type 1 is a rare, genetic arthrogryposis syndrome characterized by total fetal akinesia (detectable since the 13th week of gestation) accompanied by hydrops, micrognathia, pulmonary hypoplasia, pterygia and multiple joint contractures (usually flexion contractures in the elbows and extension in the knees), leading invariably to death before the 32nd week of gestation. Lack of anterior horn motoneurons, severe atrophy of the ventral spinal cord and severe skeletal muscle hypoplasia are characteristic neuropathological findings, with no evidence of other organ structural anomalies.

Your genetic map

Gene	SNP	Genotype
LOC1019	rs121434407	GG

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1486

Hereditary Diseases (genetics)

Autosomal recessive chorioretinopathy-microcephaly syndrome

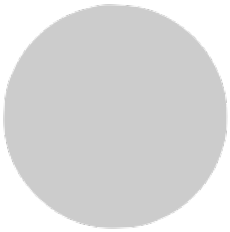
A rare neuro-opthalmological disease characterized by severe microcephaly of prenatal onset (with diminutive anterior fontanelle and sutural ridging), growth retardation, global developmental delay and intellectual disability (ranging from mild to profound), dysmorphic features (sloping forehead, micro/retrognathia, prominent ears) and visual impairments (including microphthalmia to anophtalmia, generalized retinopathy or multiple punched-out retinal lesions, retinal folds with retinal detachment, optic nerve hypoplasia, strabismus, nystagmus). Brain MRI may show reduced cortical size, cerebral hemispheres, corpus callosum, pachygyria, symplified gyral folding or normal pattern. Other associated features include epilepsy and neurological deficits.

Your genetic map

Gene	SNP	Genotype
TUBGCP	rs192919234	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2518

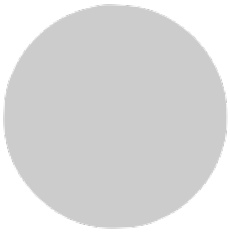
Hereditary Diseases (genetics)

Cornelia de Lange syndrome

A rare multiple congenital anomalies syndrome characterized by facial dysmorphism, hypertrichosis, mild to profound intellectual disability, intrauterine growth restriction (IUGR) and/or postnatal growth restriction, feeding difficulties, abnormalities of the hands and feet (ranging from severe reductional limb abnormalities, oligodactyly, to brachymetacarpia of the first metacarpus). Variable visceral malformations may be present.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=199

Your genetic map

Gene	SNP	Genotype
CPLANE1	rs398124474	CC
CPLANE1	rs587784053	GG
HDAC8	rs886041936	GG
NIPBL	rs121918267	CC
NIPBL	rs121918269	CC
NIPBL	rs398124466	CC
NIPBL	rs80358362	CC
NIPBL	rs398124471	CC
NIPBL	rs80358367	CC
NIPBL	rs80358369	TT
NIPBL	rs80358366	GG
NIPBL	rs80358373	AA
NIPBL	rs80358360	CC
NIPBL	rs80358363	GG
NIPBL	rs80358376	CC
NIPBL	rs80358384	AA
NIPBL	rs80358370	CC
NIPBL	rs587783937	GG
NIPBL	rs587784009	GG
NIPBL	rs587784012	AA
NIPBL	rs587783886	GG
NIPBL	rs587783895	TT
NIPBL	rs587783922	AA
NIPBL	rs587783927	GG
NIPBL	rs587783928	GG
NIPBL	rs587783988	CC
NIPBL	rs587783993	GG
NIPBL	rs587784042	AA
NIPBL	rs587784048	GG
NIPBL	rs587784049	GG
NIPBL	rs587784059	GG

Hereditary Diseases (genetics)

Costello syndrome

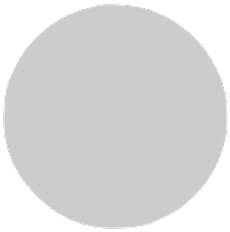
A rare syndrome with intellectual disability, characterized by failure to thrive, short stature, joint laxity, soft skin, and distinctive facial features. Cardiac and neurological involvement is common and there is an increased lifetime risk of certain tumors. Costello syndrome belongs to the RASopathies, a group of conditions resulting from germline derived point mutations affecting the RAS-mitogen activated protein kinase pathway.

Your genetic map

Gene	SNP	Genotype
LRRC56	rs104894230	CC
LRRC56	rs104894229	CC
LRRC56	rs104894226	CC
LRRC56	rs104894227	TT
LRRC56	rs104894228	CC
LRRC56	rs121917756	CC
LRRC56	rs121917757	GG
LRRC56	rs121917758	GG
LRRC56	rs121917759	GG
LRRC56	rs727503093	CC
LRRC56	rs730880460	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3071

Hereditary Diseases (genetics)

Recurrent metabolic encephalomyopathic crises-rhabdomyolysis-cardiac arrhythmia-intellectual disability

Recurrent metabolic encephalomyopathic crises-rhabdomyolysis-cardiac arrhythmia-intellectual disability syndrome is a rare, genetic, neurodegenerative disease characterized by episodic metabolic encephalomyopathic crises (of variable frequency and severity which are frequently precipitated by an acute illness) which manifest with profound muscle weakness, ataxia, seizures, cardiac arrhythmias, rhabdomyolysis with myoglobinuria, elevated plasma creatine kinase, hypoglycemia, lactic acidosis, increased acylcarnitines and a disorientated or comatose state. Global developmental delay, intellectual disability and cortical, pyramidal and cerebellar signs develop with subsequent progressive neurodegeneration causing loss of expressive language and varying degrees of cerebral atrophy.

Your genetic map

Gene	SNP	Genotype
TANGO2	rs372949028	GG
TANGO2	rs199801224	CC

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=480864

Hereditary Diseases (genetics)

Crouzon syndrome-acanthosis nigricans syndrome

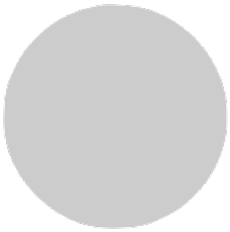
Crouzon syndrome with acanthosis nigricans (CAN) is a very rare, clinically heterogeneous form of faciocraniostenosis with Crouzon-like features and premature synostosis of cranial sutures (Crouzon disease, see this term), associated with acanthosis nigricans (AN; see this term).

Your genetic map

Gene	SNP	Genotype
FGFR3	rs28931615	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=93262

Hereditary Diseases (genetics)

De Barsy syndrome

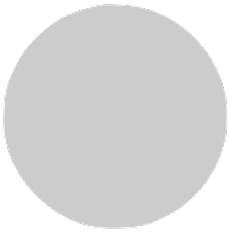
De Barsy syndrome (DBS) is characterized by facial dysmorphism (down-slanting palpebral fissures, a broad flat nasal bridge and a small mouth) with a progeroid appearance, large and late-closing fontanel, cutis laxa (CL), joint hyperlaxity, athetoid movements and hyperreflexia, pre- and postnatal growth retardation, intellectual deficit and developmental delay, and corneal clouding and cataract.

Your genetic map

Gene	SNP	Genotype
ALDH18A	rs556267618	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2962

Hereditary Diseases (genetics)

DEND syndrome

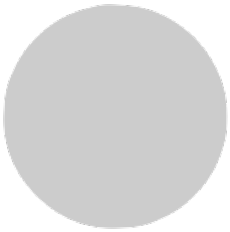
DEND syndrome is a very rare, generally severe form of neonatal diabetes mellitus (NDM, see this term) characterized by a triad of developmental delay, epilepsy, and neonatal diabetes.

Your genetic map

Gene	SNP	Genotype
ABCC8	rs1048095	AA
INS IGF2	rs80356663	GG
INS IGF2	rs80356669	GG
INS IGF2	rs80356664	CC
INS IGF2	rs797045623	CC
KCNJ11	rs80356611	CC
KCNJ11	rs193929356	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79134

Hereditary Diseases (genetics)

Denys-Drash syndrome

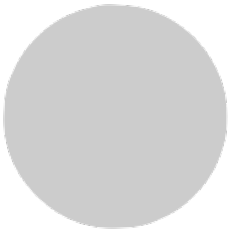
A rare genetic, syndromic glomerular disorder characterized by the association of nephropathy presenting as persistent proteinuria or overt nephrotic syndrome, Wilms tumor and genitourinary structural defects. In addition, disorders of testicular development are common in subjects with 46,XY karyotype.

Your genetic map

Gene	SNP	Genotype
WT1	rs121907900	GG
WT1	rs121907901	CC
WT1	rs121907902	TT
WT1	rs28941778	CC
WT1	rs587776576	CC
WT1	rs121907906	GG
WT1	rs1423753702	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=220

Hereditary Diseases (genetics)

Mitochondrial DNA depletion syndrome, encephalomyopathic form

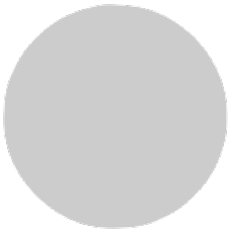
Mitochondrial DNA depletion syndrome, encephalomyopathic form is a group of mitochondrial DNA maintenance syndrome diseases characterized by predominantly neuromuscular manifestations with typically infantile onset of hypotonia, lactic acidosis, psychomotor delay, progressive hyperkinetic-dystonic movement disorders, external ophtalmoplegia, sensosineural hearing loss, generalized seizures and variable renal tubular dysfunction. It may be associated with a broad range of other clinical features.

Your genetic map

Gene	SNP	Genotype
RRM2B	rs515726196	AA
RRM2B	rs776184830	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=254803

Hereditary Diseases (genetics)

Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency

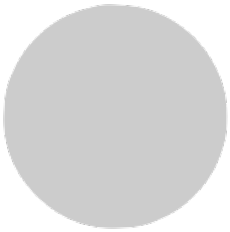
A rare immune disease characterized by severely reduced mitochondrial DNA content due to DGUOK deficiency typically manifesting with early-onset liver dysfunction, psychomotor delay, hypotonia, rotary nystagmus that develops into opsoclonus, lactic acidosis and hypoglycemia.

Your genetic map

Gene	SNP	Genotype
DGUOK	rs748597500	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=279934

Hereditary Diseases (genetics)

Acral peeling skin syndrome

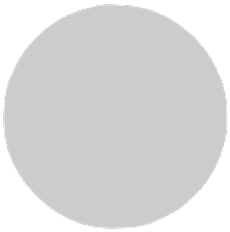
A rare peeling skin syndrome characterized by superficial peeling of the skin predominantly affecting the dorsa of the hands and feet.

Your genetic map

Gene	SNP	Genotype
TGM5	rs112292549	CC
TGM5	rs115677373	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=263534

Hereditary Diseases (genetics)

Dysequilibrium syndrome

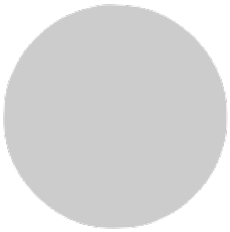
Dysequilibrium syndrome (DES) is a non-progressive cerebellar disorder characterized by ataxia associated with an intellectual disability, delayed ambulation and cerebellar hypoplasia.

Your genetic map

Gene	SNP	Genotype
VLDLR	rs770269674	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1766

Hereditary Diseases (genetics)

Cognitive impairment-coarse facies-heart defects-obesity-pulmonary involvement-short stature-skeletal dysplasia

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by global developmental delay, intellectual disability, short stature, skeletal abnormalities (such as brachydactyly and vertebral anomalies), obesity, cardiac, respiratory, and genitourinary anomalies, and dysmorphic facial features (including coarse facies, thick eyebrows, synophrys, hypertelorism, short, upturned nose, and long philtrum). Additional reported manifestations are microcephaly, hearing impairment, cataract, and gastroesophageal reflux.

Your genetic map

Gene	SNP	Genotype
AFF4	rs786205680	GG

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=444077

Hereditary Diseases (genetics)

TBCK-related intellectual disability syndrome

TBCK-related intellectual disability syndrome is a rare, genetic, syndromic intellectual disability characterized by usually profound intellectual disability with absent speech, severe infantile hypotonia with decreased or absent reflexes, markedly slow motor development (with no progress beyond the ability to sit independently), early-onset epilepsy, strabismus and post-natal onset of progressive brain atrophy (incl. loss of brain volume, ex vacuo ventriculomegaly, dysgenesis of corpus callosum, white matter abnormalities ranging from non-specific changes to leukodystrophy). Swallowing difficulties, respiratory insufficiency, osteoporosis and variable craniofacial dysmorphisms (incl. plagio/brachicephaly, bitemporal narrowing, high-arched eyebrows, high nasal bridge, anteverted nares, high palate, tented upper lip) may constitute additional clinical features.

Your genetic map

Gene	SNP	Genotype
TBCK	rs575822089	GG
TBCK	rs376699648	TT

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=488632

Hereditary Diseases (genetics)

Severe intellectual disability-progressive spastic diplegia syndrome

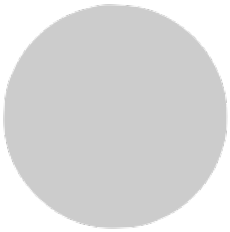
Severe intellectual disability-progressive spastic diplegia syndrome is a rare, genetic, syndromic intellectual disability disorder characterized by intellectual disability, significant motor delay, severe speech impairment, early-onset truncal hypotonia with progressive distal hypertonia/spasticity, microcephaly, and behavioral anomalies (autistic features, aggression or auto-aggressive behavior, sleep disturbances). Variable facial dysmorphism includes broad nasal tip with small alae nasi, long and/or flat philtrum, thin upper lip vermillion. Visual impairment (strabismus, hyperopia, myopia) is commonly associated.

Your genetic map

Gene	SNP	Genotype
CTNNB1	rs397514554	CC
CTNNB1	rs797044875	GG
CTNNB1	rs863224864	TT
CTNNB1	rs775104326	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=404473

Hereditary Diseases (genetics)

X-linked intellectual disability-cerebellar hypoplasia syndrome

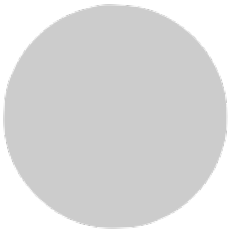
X-linked intellectual deficit-cerebellar hypoplasia, also known as OPHN1 syndrome, is a rare syndromic form of cerebellar dysgenesis characterized by moderate to severe intellectual deficit and cerebellar abnormalities.

Your genetic map

Gene	SNP	Genotype
OPHN1	rs587784234	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=137831

Hereditary Diseases (genetics)

X-linked intellectual disability-hypotonia-movement disorder syndrome

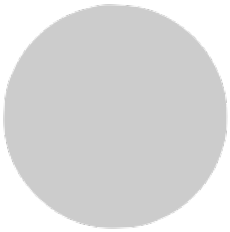
A rare, genetic, syndromic intellectual disability characterized by mild to severe intellectual disability associated with variable features, including hypotonia, dyskinesia, spasticity, wide-based gait, microcephaly, epilepsy and behavioral problems. MRI imaging may show a corpus callosum hypoplasia or ventricular enlargement. Other variable features, such as joint hyperlaxity, skin pigmentary abnormalities, and visual impairment, have also been reported.

Your genetic map

Gene	SNP	Genotype
DDX3X	rs796052231	CC
DDX3X	rs796052232	TT
DDX3X	rs796052235	GG
DDX3X	rs796052226	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=457260

Hereditary Diseases (genetics)

X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome

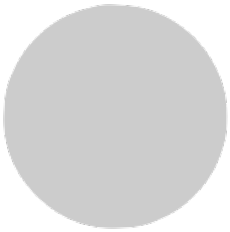
X-linked Dandy-Walker malformation with intellectual disability, basal ganglia disease and seizures (XDIBS), or Pettigrew syndrome is a central nervous system malformation characterized by severe intellectual deficit, early hypotonia with progression to spasticity and contractures, choreoathetosis, seizures, dysmorphic face (long face with prominent forehead), and brain imaging abnormalities such as Dandy-Walker malformation (see this term), and iron deposition.

Your genetic map

Gene	SNP	Genotype
AP1S2	rs587777542	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1568

Hereditary Diseases (genetics)

X-linked intellectual disability-psychosis-macroorchidism syndrome

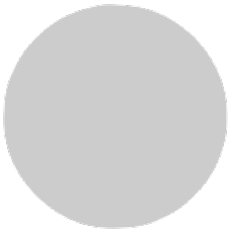
An X-linked syndromic intellectual disability characterized by developmental delay, variable degree of intellectual disability, speech delay or absent speech, pyramidal signs, tremor, macroorchidism and variable mood and behavior problems, including psychosis and autistic-like behavior. Males are predominantly affected, some females show lower cognitive abilities.

Your genetic map

Gene	SNP	Genotype
MECP2	rs63094662	CC
MECP2	rs28934908	GG
MECP2	rs61751444	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3077

Hereditary Diseases (genetics)

Intellectual disability-expressive aphasia-facial dysmorphism syndrome

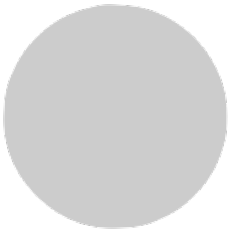
A rare genetic syndromic intellectual disability characterized by moderate to severe intellectual deficiency, language deficit (completely absent or significantly impaired speech), and distinctive facial dysmorphism (long face, straight eyebrows, and, less frequently, low-set ears and cafe-au-lait spots). Additional, variably observed features include motor delays, behavioral difficulties, and seizures.

Your genetic map

Gene	SNP	Genotype
SETBP1	rs606231272	CC
SETBP1	rs606231273	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=436151

Hereditary Diseases (genetics)

Intellectual disability-cataracts-calcified pinnae-myopathy syndrome

Intellectual disability-cataracts-calcified pinnae-myopathy syndrome is a rare, genetic intellectual disability syndrome characterized by macrocephaly, hypotonia, dysmorphic facial features (wide forehead, ptosis, downslanting palpebral fissures, enlarged and calcified external ears, large jaw), sparse body hair, tall stature, and intellectual disability. Hearing loss, insulin-resistant diabetes, and progressive distal muscle wasting (leading to joint contractures) have also been reported in adulthood. Rare manifestations include behavioral abnormalities (aggression and restlessness), hypothyroidism, cerebral calcification, ataxia, and peripheral neuropathy.

Your genetic map

Gene	SNP	Genotype
ZBTB20	rs483353069	TT

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3042

Hereditary Diseases (genetics)

Intellectual disability-seizures-hypophosphatasia-ophthalmic-skeletal anomalies syndrome

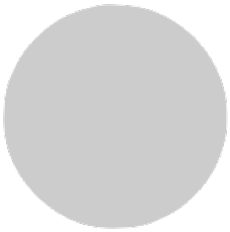
The syndrome of intellectual disability, seizures, hypotonia, ophthalmologic and skeletal anomalies is a rare congenital glycosylation disorder. It presents with neonatal hypotonia, developmental delays, and significant intellectual disability. Infants experience seizures, initially during fever, evolving to unprovoked seizures. Vision is affected with esotropia and nystagmus. Brain atrophy is progressive, alongside skeletal issues like brachycephaly, scoliosis, and osteopenia. Dysmorphic features include a distinct face with a high forehead, short nose, and facial hypotonia. Cardiac and urogenital abnormalities, as well as low alkaline phosphatase levels, can also occur.

Your genetic map

Gene	SNP	Genotype
LOC1079	rs200790673	AA
PIGT	rs201317502	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=369837

Hereditary Diseases (genetics)

Intellectual disability-macrocephaly-hypotonia-behavioral abnormalities syndrome

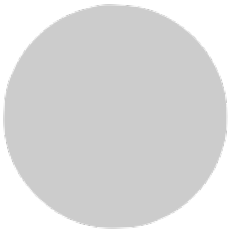
A rare, syndromic intellectual disability characterized by hypotonia, global developmental delay, limited or absent speech, intellectual disability, macrocephaly, mild dysmorphic features, seizures and autism spectrum disorder. Associated ophthalmologic, heart, skeletal and central nervous system anomalies have been reported.

Your genetic map

Gene	SNP	Genotype
PPP2R5D	rs863225079	GG
PPP2R5D	rs863225081	GG
PPP2R5D	rs863225080	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=457279

Hereditary Diseases (genetics)

Intellectual disability-severe speech delay-mild dysmorphism syndrome

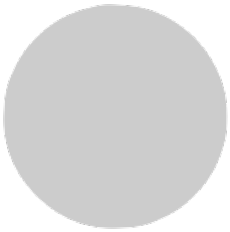
Intellectual disability-severe speech delay-mild dysmorphism syndrome is a rare, genetic, syndromic intellectual disability disorder, with highly variable phenotype, typically characterized by mild to severe global development delay, severe speech and language impairment, mild to severe intellectual disability, dysphagia, hypotonia, relative to true macrocephaly, and behavioral problems that may include autistic features, hyperactivity, and mood lability. Facial gestalt typically features a broad, prominent forehead, hypertelorism, downslanting palpebral fissures, ptosis, a short bulbous nose with broad tip, thick vermilion border, wide, and open mouth with downturned corners. Brain, cardiac, urogenital and ocular malformations may be associated.

Your genetic map

Gene	SNP	Genotype
FOXP1	rs794727155	GG
FOXP1	rs797045586	CC
FOXP1	rs797045584	GG
FOXP1	rs869025203	GG
FOXP1	rs869025202	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=391372

Hereditary Diseases (genetics)

Multiple mitochondrial dysfunctions syndrome type 4

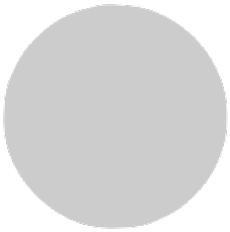
A rare, severe, genetic, neurometabolic disease characterized by infantile-onset of progressive neurodevelopmental regression, optic atrophy with nystagmus and diffuse white matter disease. Affected individuals usually have central hypotonia that progresses to limb spasticity and hyperreflexia, eventually resulting in a vegetative state. Recurrent chest infections are frequently associated and seizures (usually generalized tonic-clonic) may occasionally be observed. Brain magnetic resonance imaging shows diffuse bilateral symmetric abnormalities in the cerebral periventricular white matter, with variable lesions in other areas but sparing the basal ganglia.

Your genetic map

Gene	SNP	Genotype
ISCA2	rs730882246	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=457406

Hereditary Diseases (genetics)

CNTNAP2-related developmental and epileptic encephalopathy

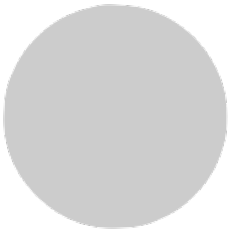
A rare, genetic, syndromic neurodevelopmental disorder characterized by moderate to mostly severe intellectual disability, speech impairment with normal or mildly delayed motor development and early-onset seizures often accompanied by developmental regression. Autistic behavior and stereotypic movements are common.

Your genetic map

Gene	SNP	Genotype
CNTNAP	rs730880276	GG
CNTNAP	rs398124268	GG
CNTNAP	rs752550849	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=163681

Hereditary Diseases (genetics)

Spondyloperipheral dysplasia-short ulna syndrome

Spondyloperipheral dysplasia-short ulna syndrome is a rare, genetic, primary bone dysplasia, with highly variable phenotype, typically characterized by platyspondyly, brachydactyly type E changes (short metacarpals and metatarsals, short distal phalanges in hands and feet), bilateral short ulnae and mild short stature. Other reported features include additional skeletal findings (e.g. midface hypoplasia, degenerative changes in proximal femora, limited elbow extension, bilateral sacralization of L5, clubfeet), as well as myopia, hearing loss, and intellectual disability.

Your genetic map

Gene	SNP	Genotype
COL2A1	rs121912880	CC

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1856

Hereditary Diseases (genetics)

Spondylometaphyseal dysplasia-cone-rod dystrophy syndrome

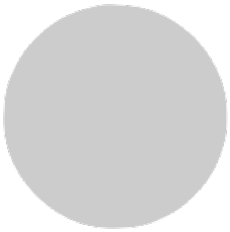
Spondylometaphyseal dysplasia-cone-rod dystrophy syndrome is characterised by the association of spondylometaphyseal dysplasia (marked by platyspondyly, shortening of the tubular bones and progressive metaphyseal irregularity and cupping), with postnatal growth retardation and progressive visual impairment due to cone-rod dystrophy. So far, it has been described in eight individuals. Transmission appears to be autosomal recessive.

Your genetic map

Gene	SNP	Genotype
PCYT1A	rs587777189	GG
PCYT1A	rs587777190	GG
PCYT1A	rs587777191	CC
PCYT1A	rs587777192	GG
PCYT1A	rs540053239	CC
PCYT1A	rs587777194	CC
PCYT1A	rs587777195	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=85167

Hereditary Diseases (genetics)

Corneal intraepithelial dyskeratosis-palmoplantar hyperkeratosis-laryngeal dyskeratosis syndrome

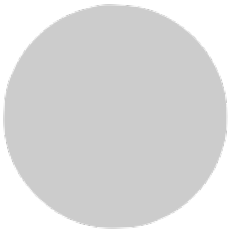
Corneal intraepithelial dyskeratosis-palmoplantar hyperkeratosis-laryngeal dyskeratosis syndrome is a rare, genetic, corneal dystrophy disorder characterized by corneal opacification and dyskeratosis (which may cause visual impairment), associated with systemic features including palmoplantar hyperkeratosis, laryngeal dyskeratosis, pruritic hyperkeratotic scars, chronic rhintis, dyshidrosis and/or nail thickening.

Your genetic map

Gene	SNP	Genotype
NLRP1	rs1057519493	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=352662

Hereditary Diseases (genetics)

Corneal dystrophy-perceptive deafness syndrome

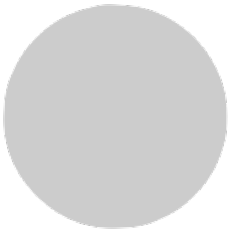
Corneal dystrophy-perceptive deafness (CDPD) or Harboyan syndrome is a degenerative corneal disorder characterized by the association of congenital hereditary endothelial dystrophy (CHED; see this term) with progressive, postlingual sensorineural hearing loss.

Your genetic map

Gene	SNP	Genotype
SLC4A11	rs121909394	AA
SLC4A11	rs759540763	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1490

Hereditary Diseases (genetics)

Donnai-Barrow syndrome

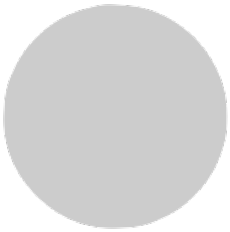
A multiple congenital malformation syndrome characterized by typical facial dysmorphism, myopia and other ocular findings, hearing loss, agenesis of the corpus callosum, low-molecular-weight proteinuria, and variable intellectual disability. Congenital diaphragmatic hernia (CDH) and/or omphalocele are common.

Your genetic map

Gene	SNP	Genotype
LRP2	rs80338747	AA
LRP2	rs752197557	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2143

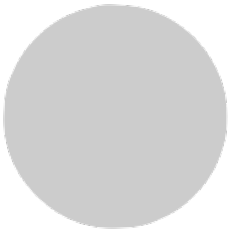
Hereditary Diseases (genetics)

Dravet syndrome

A rare, genetic, developmental and epileptic encephalopathy characterized by infantile onset of intractable seizures that are often febrile, and associated with cognitive and motor impairment.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=33069

Your genetic map

Gene	SNP	Genotype
LOC1027	rs121917912	CC
LOC1027	rs121917960	CC
LOC1027	rs121917986	CC
LOC1027	rs121917919	AA
LOC1027	rs121917915	CC
LOC1027	rs121917976	CC
LOC1027	rs121917922	GG
LOC1027	rs121917980	CC
LOC1027	rs121917921	GG
LOC1027	rs121917981	AA
LOC1027	rs121918738	GG
LOC1027	rs121918739	TT
LOC1027	rs121918740	AA
LOC1027	rs121918741	CC
LOC1027	rs121918742	CC
LOC1027	rs121918791	GG
LOC1027	rs121918763	GG
LOC1027	rs121918757	AA
LOC1027	rs121918751	AA
LOC1027	rs139300715	GG
LOC1027	rs727504136	GG
LOC1027	rs794726737	CC
LOC1027	rs794726739	GG
LOC1027	rs794726845	GG
LOC1027	rs779614747	GG
LOC1027	rs794726801	GG
LOC1027	rs794726769	CC
LOC1027	rs794726781	GG
LOC1027	rs794726780	CC
LOC1027	rs794726722	TT
LOC1027	rs794726763	CC

Hereditary Diseases (genetics)

Dubin-Johnson syndrome

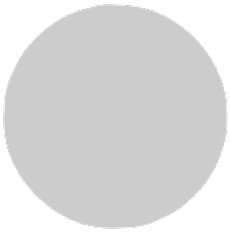
Dubin-Johnson syndrome (DJS) is a benign, inherited liver disorder characterized clinically by chronic, predominantly conjugated, hyperbilirubinemia and histopathologically by black-brown pigment deposition in parenchymal liver cells.

Your genetic map

Gene	SNP	Genotype
ABCC2	rs56199535	CC
ABCC2	rs72558199	CC
ABCC2	rs34937870	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=234

Hereditary Diseases (genetics)

Dyggve-Melchior-Clausen disease

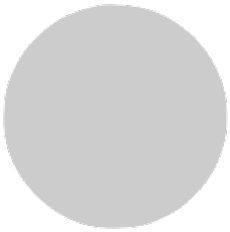
A rare, genetic primary bone dysplasia of the spondylo-epi-metaphyseal dysplasia (SEMD) group characterized by progressive short-trunked dwarfism, protruding sternum, microcephaly, intellectual disability and pathognomonic radiological findings (generalized platyspondyly with double-humped end plates, irregularly ossified femoral heads, a hypoplastic odontoid, and a lace-like appearance of iliac crests)

Your genetic map

Gene	SNP	Genotype
DYM	rs775414124	TT
DYM	rs768509996	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=239

Hereditary Diseases (genetics)

Cardiac-valvular Ehlers-Danlos syndrome

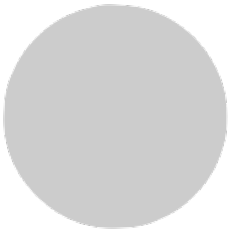
A rare form of Ehlers-Danlos syndrome (EDS) characterized by soft skin, skin hyperextensibility, easy bruisability, atrophic scar formation, joint hypermobility and severe, progressive cardiac valvular defects comprising mitral and/or aortic valve insufficiency.

Your genetic map

Gene	SNP	Genotype
COL1A2	rs67162110	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=230851

Hereditary Diseases (genetics)

Hypermobile Ehlers-Danlos syndrome

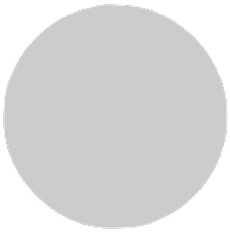
Ehlers-Danlos syndrome, hypermobility type (HT-EDS) is the most frequent form of EDS (see this term), a group of hereditary connective tissue diseases, and is characterized by joint hyperlaxity, mild skin hyperextensibility, tissue fragility and extra-musculoskeletal manifestations.

Your genetic map

Gene	SNP	Genotype
COL3A1	rs863224860	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=285

Hereditary Diseases (genetics)

Musculocontractural Ehlers-Danlos syndrome

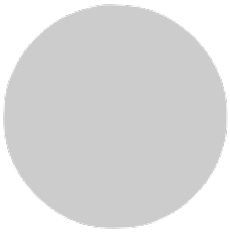
A rare systemic disease characterized by congenital multiple contractures, characteristic craniofacial features (like large fontanel, hypertelorism, downslanting palpebral fissures, blue sclerae, ear deformities, high palate) evident at birth or in early infancy, and characteristic cutaneous features like skin hyperextensibility, skin fragility with atrophic scars, easy bruising, and increased palmar wrinkling. Additional features include recurrent/chronic dislocations, chest and spinal deformities, peculiarly shaped fingers, colonic diverticula, pneumothorax, and urogenital and ophthalmological abnormalities, among others. Molecular testing is obligatory to confirm the diagnosis.

Your genetic map

Gene	SNP	Genotype
CHST14	rs121908257	GG
CHST14	rs121908258	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2953

Hereditary Diseases (genetics)

Periodontal Ehlers-Danlos syndrome

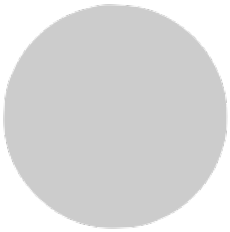
A rare type of Ehlers-Danlos syndrome characterized by childhood or adolescence onset of severe, intractable periodontitis, lack of attached gingiva, and presence of pretibial plaques. Additional manifestations are easy bruising, hypermobility mainly of the distal joints, skin hyperextensibility and fragility, abnormal scarring, recurrent infections, hernias, marfanoid facial features, acrogeria, and prominent vasculature.

Your genetic map

Gene	SNP	Genotype
C1S	rs886040975	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=75392

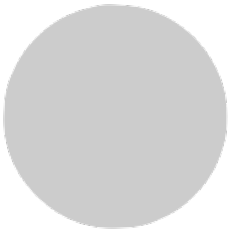
Hereditary Diseases (genetics)

Vascular Ehlers-Danlos syndrome

A rare genetic connective tissue disorder typically characterized by the association of unexpected organ fragility (arterial/bowel/gravid uterine rupture) with inconstant physical features as thin, translucent skin, easy bruising and acrogeric traits.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=286

Your genetic map

Gene	SNP	Genotype
COL3A1	rs113485686	GG
COL3A1	rs121912927	GG
COL3A1	rs397509372	GG
COL3A1	rs121912917	GG
COL3A1	rs121912918	GG
COL3A1	rs397509376	GG
COL3A1	rs121912921	GG
COL3A1	rs121912925	GG
COL3A1	rs121912926	GG
COL3A1	rs267599120	GG
COL3A1	rs587779424	GG
COL3A1	rs587779427	GG
COL3A1	rs587779429	TT
COL3A1	rs587779431	GG
COL3A1	rs587779437	GG
COL3A1	rs587779438	GG
COL3A1	rs587779442	GG
COL3A1	rs587779444	GG
COL3A1	rs587779450	GG
COL3A1	rs587779454	GG
COL3A1	rs587779461	GG
COL3A1	rs587779471	GG
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COL3A1	rs587779477	GG
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COL3A1	rs587779482	GG
COL3A1	rs587779483	GG
COL3A1	rs587779487	GG
COL3A1	rs587779492	GG

Hereditary Diseases (genetics)

Neonatal encephalomyopathy-cardiomyopathy-respiratory distress syndrome

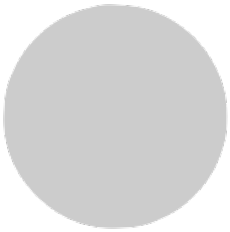
A rare mitochondrial disease characterized by neonatal onset of severe cardiac and/or neurologic signs and symptoms mostly associated with a fatal outcome in the neonatal period or in infancy, although a milder phenotype with later onset and slowly progressive neurologic deterioration has also been reported. Clinical manifestations are variable and include respiratory insufficiency, hypotonia, cardiomyopathy, and seizures. Serum lactate is elevated in most cases. Brain imaging may show cerebellar atrophy or hypoplasia.

Your genetic map

Gene	SNP	Genotype
COQ4	rs143441644	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=457185

Hereditary Diseases (genetics)

Interstitial lung disease-nephrotic syndrome-epidermolysis bullosa syndrome

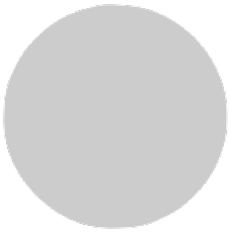
Congenital nephrotic syndrome-interstitial lung disease-epidermolysis bullosa syndrome is a life-threatening multiorgan disorder which develops in the first months of life, presenting with respiratory distress and proteinuria in the nephrotic range, and leading to severe interstitial lung disease and renal failure. Some patients additionally display cutaneous alterations, ranging from blistering and skin erosions to an epidermolysis bullosa-like phenotype, with toe nail dystrophy and sparse hair.

Your genetic map

Gene	SNP	Genotype
ITGA3	rs540704248	CC
ITGA3	rs797045048	GG

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=306504

Hereditary Diseases (genetics)

Progressive epilepsy-intellectual disability syndrome, Finnish type

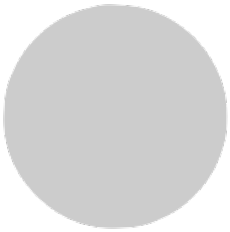
Progressive epilepsy-intellectual deficit, Finnish type (also known as Northern epilepsy) is a subtype of neuronal ceroid lipofuscinosis (NCL; see this term) characterized by seizures, progressive decline of intellectual capacities and variable loss of vision.

Your genetic map

Gene	SNP	Genotype
CLN8	rs104894064	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1947

Hereditary Diseases (genetics)

Female restricted epilepsy with intellectual disability

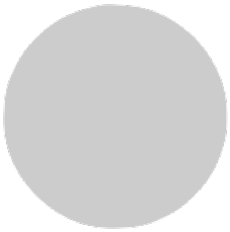
Female restricted epilepsy with intellectual disability is a rare X-linked epilepsy syndrome characterized by febrile or afebrile seizures (mainly tonic-clonic, but also absence, myoclonic, and atonic) starting in the first years of life and, in most cases, developmental delay and intellectual disability of variable severity. Behavioral disturbances (e.g. autistic features, hyperactivity, and aggressiveness) are also frequently associated. This disease affects exclusively females, with male carriers being unaffected, despite an X-linked inheritance.

Your genetic map

Gene	SNP	Genotype
PCDH19	rs267606933	GG
PCDH19	rs398123603	TT
PCDH19	rs587784299	TT
PCDH19	rs796052812	GG
PCDH19	rs796052839	TT
PCDH19	rs796052802	GG
PCDH19	rs796052837	GG
PCDH19	rs796052800	CC
PCDH19	rs796052799	GG
PCDH19	rs797045873	GG
PCDH19	rs1060502176	GG
PCDH19	rs796052813	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2076

Hereditary Diseases (genetics)

Gingival fibromatosis-hypertrichosis syndrome

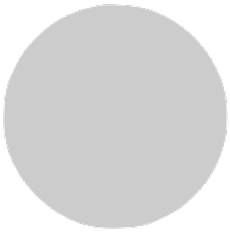
A rare autosomal dominant disorder characterized by a generalized enlargement of the gingiva occurring at birth or during childhood that is associated with generalized hypertrichosis developing at birth, during the first years of life, or at puberty and predominantly affecting the face, upper limbs, and midback.

Your genetic map

Gene	SNP	Genotype
ABCA5	rs199753304	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2026

Hereditary Diseases (genetics)

Floating-Harbor syndrome

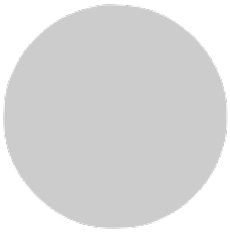
A multiple congenital anomalies/dysmorphic syndrome-intellectual disability that is characterized by facial dysmorphism, short stature with delayed bone age, and expressive language delay.

Your genetic map

Gene	SNP	Genotype
SRCAP	rs199469464	CC
SRCAP	rs199469465	CC
SRCAP	rs587784444	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2044

Hereditary Diseases (genetics)

Bloom's Syndrome

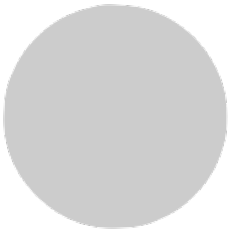
Bloom syndrome is a rare disorder associated with pre- and postnatal growth deficiency, a telangiectatic erythematous rash of the face and other sun-exposed areas, insulin resistance and predisposition to early onset and recurrent cancer of multiple organ systems.

Your genetic map

Gene	SNP	Genotype
BLM	rs367543036	GG
BLM	rs367543029	GG
BLM	rs367543017	CC
BLM	rs587779884	CC
BLM	rs587783037	CC
BLM	rs730881428	TT
BLM	rs1057516964	GG
BLM	rs1356090839	GG
BLM	rs200389141	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=125

Hereditary Diseases (genetics)

Frasier syndrome

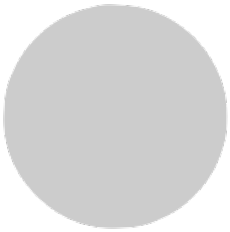
A rare genetic, syndromic glomerular disorder characterized by the association of progressive glomerular nephropathy and 46,XY complete gonadal dysgenesis with a high risk of developing gonadoblastoma.

Your genetic map

Gene	SNP	Genotype
WT1	rs587776577	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=347

Hereditary Diseases (genetics)

Gerstmann-Straussler-Scheinker syndrome

A rare inherited human prion disease characterized by adult onset of slowly progressive cerebellar ataxia, with dementia developing relatively late in the disease course (classic ataxic phenotype). Patients may present with gait disturbances and frequent falls, dysarthria, dysphagia, nystagmus, dysmetry, and eventually pancerebellar syndrome, myoclonus, spasticity, severe dementia, and mutism. The disease is invariably fatal after five years on average. Neuropathological hallmark is the presence of numerous multicentric prion protein plaques in the cerebral and cerebellar cortex.

Your genetic map

Gene	SNP	Genotype
PRNP	rs11538758	CC

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=356

Hereditary Diseases (genetics)

Gitelman syndrome

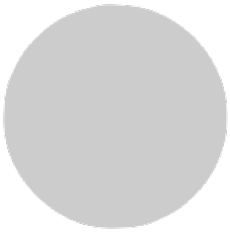
A rare syndrome characterized by hypokalemic metabolic alkalosis in combination with significant hypomagnesemia and low urinary calcium excretion.

Your genetic map

Gene	SNP	Genotype
MIR6863	rs199974259	GG
SLC12A3	rs121909382	CC
SLC12A3	rs267607050	CC
SLC12A3	rs568513106	TT
SLC12A3	rs374163823	GG
SLC12A3	rs140012781	CC
SLC12A3	rs749098014	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=358

Hereditary Diseases (genetics)

Hermansky-Pudlak syndrome due to BLOC-3 deficiency

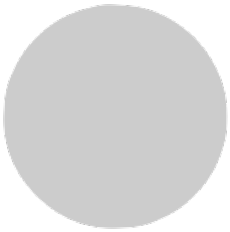
Hermansky-Pudlak syndrome with pulmonary fibrosis as a complication includes two types (HPS-1 and HPS-4) of Hermansky-Pudlak syndrome (HPS; see this term), a multi-system disorder characterized by oculocutaneous albinism, bleeding diathesis and, in some cases, pulmonary fibrosis or granulomatous colitis.

Your genetic map

Gene	SNP	Genotype
HPS1	rs121908385	CC

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=231500

Hereditary Diseases (genetics)

Hermansky-Pudlak syndrome due to BLOC-2 deficiency

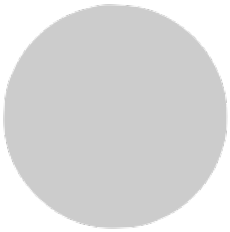
Hermansky-Pudlak syndrome without pulmonary fibrosis as a complication includes three relatively mild types (HPS-3, HPS-5 and HPS-6) of Hermansky-Pudlak syndrome (HPS; see this term), a multi-system disorder characterized by ocular or oculocutaneous albinism, bleeding diathesis and, in some cases, granulomatous colitis.

Your genetic map

Gene	SNP	Genotype
HPS3	rs201227603	GG
HPS3	rs121908316	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=231512

Hereditary Diseases (genetics)

Hydrops-lactic acidosis-sideroblastic anemia-multisystemic failure syndrome

A rare mitochondrial disease characterized by prenatal complications including oligohydramnios, fetal growth restriction, hydrops, and anemia, followed by severe lactic acidosis, hyaline membrane disease, pulmonary hypertension, cardiac anomalies, liver dysfunction, urogenital abnormalities and progressive renal disease, seizures, thrombocytopenia, and sideroblastic anemia resulting in multisystem organ failure and death shortly after birth. Less severely affected patients surviving the neonatal period and showing sensorineural hearing loss and developmental delay have been reported.

Your genetic map

Gene	SNP	Genotype
LARS2	rs786205560	AA

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=528091

Hereditary Diseases (genetics)

Hyper-IgM syndrome with susceptibility to opportunistic infections

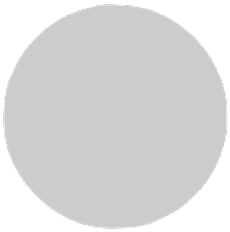
Hyper-IgM syndrome with susceptibility to opportunistic infections is a rare, genetic, non-severe combined immunodeficiency disorder characterized by normal or elevated IgM serum levels with low or absent IgG, IgA and IgE serum concentrations, which manifests with recurrent or severe bacterial infections and increased susceptibility to opportunistic infections (in particular, pneumonia due to P. jiroveci, but also chronic cryptosporidial, cryptococcal, cytomegalovirus and toxoplasma infections). Hematologic disorders (neutropenia, anemia, thrombocytopenia) are frequently associated. Immunologic findings reveal decreased numbers of CD27+ memory B cells and lack of germinal center formation.

Your genetic map

Gene	SNP	Genotype
CD40LG	rs104894769	TT
CD40LG	rs104894774	TT
CD40LG	rs104894777	TT
CD40LG	rs104894778	CC
CD40LG	rs193922135	CC
CD40LG	rs193922136	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=183663

Hereditary Diseases (genetics)

Autosomal dominant hyper-IgE syndrome

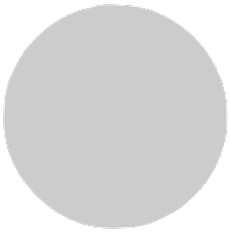
A very rare primary immunodeficiency disorder characterized by the clinical triad of high serum IgE (>2000 IU/ml), recurring staphylococcal skin abscesses, and recurrent pneumonia with formation of pneumatoceles.

Your genetic map

Gene	SNP	Genotype
STAT3	rs113994135	GG
STAT3	rs113994139	CC
STAT3	rs193922716	GG
STAT3	rs193922717	CC
STAT3	rs193922719	TT
STAT3	rs193922720	CC
STAT3	rs193922721	TT
STAT3	rs193922722	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2314

Hereditary Diseases (genetics)

Hyperphosphatasia-intellectual disability syndrome

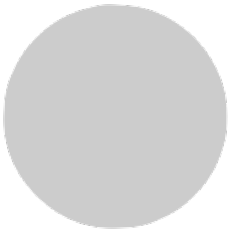
A rare, congenital disorder of glycosylation-related bone disorder characterized by hypotonia, severe developmental delay, intellectual disability, seizures, increased serum alkaline phosphatase, short distal phalanges with hypoplastic nails, and dysmorphic facial features. In some cases, cleft palate, megacolon, anorectal malformations, and congenital heart defects have been reported.

Your genetic map

Gene	SNP	Genotype
PIGV	rs139073416	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=247262

Hereditary Diseases (genetics)

Hyperinsulinism-hyperammonemia syndrome

Hyperinsulinism-hyperammonemia syndrome (HIHA) is a frequent form of diazoxide-sensitive diffuse hyperinsulinism (see this term), characterized by an excessive/ uncontrolled insulin secretion (inappropriate for the level of glycemia), asymptomatic hyperammonemia and recurrent episodes of profound hypoglycemia induced by fasting and protein rich meals, requiring rapid and intensive treatment to prevent neurological sequelae. Epilepsy and cognitive deficit that are unrelated to hypoglycemia may also occur.

Your genetic map

Gene	SNP	Genotype
GLUD1	rs121909731	GG
GLUD1	rs121909734	CC
GLUD1	rs797045597	CC

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=35878

Hereditary Diseases (genetics)

Hypohidrosis-enamel hypoplasia-palmoplantar keratoderma-intellectual disability syndrome

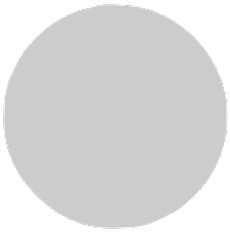
Hypohidrosis-enamel hypoplasia-palmoplantar keratoderma-intellectual disability syndrome is a rare, genetic, syndromic intellectual disability disorder characterized by severe intellectual disability with significant speech and language impairment, hypohydrosis (often resulting in hyperthermia) with normal sweat gland appearance, tooth enamel hypoplasia, palmoplantar hyperkeratosis and a high frequency of acquired microcephaly. Mild facial dysmorphism, including lateral flaring of the eyebrows, broad nasal tip, and thick vermilion border, may also be observed.

Your genetic map

Gene	SNP	Genotype
COG6	rs730882236	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=363523

Hereditary Diseases (genetics)

Hypoplastic pancreas-intestinal atresia-hypoplastic gallbladder syndrome

Hypoplastic pancreas-intestinal atresia-hypoplastic gallbladder syndrome is a rare, potentially fatal, genetic, visceral malformation syndrome characterized by neonatal diabetes, hypoplastic or annular pancreas, duodenal and jejunal atresia, as well as gallbladder aplasia or hypoplasia. Patients typically present intrauterine growth restriction, failure to thrive, malnutrition, intestinal malrotation, malabsorption, conjugated hyperbilirubinemia, acholia and infections. Cardiac anomalies may also be associated.

Your genetic map

Gene	SNP	Genotype
RFX6	rs587780440	CC

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=293864

Hereditary Diseases (genetics)

Pancreatic hypoplasia-diabetes-congenital heart disease syndrome

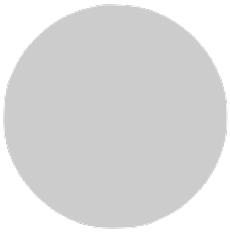
A rare, syndromic diabetes mellitus characterized by partial pancreatic agenesis, diabetes mellitus, and heart anomalies (including transposition of the great vessels, ventricular or atrial septal defects, pulmonary stenosis, or patent ductus arteriosis).

Your genetic map

Gene	SNP	Genotype
GATA6	rs387906818	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2255

Hereditary Diseases (genetics)

Hypotonia-speech impairment-severe cognitive delay syndrome

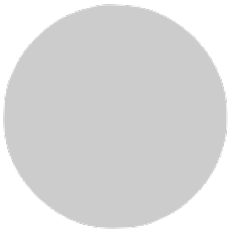
Hypotonia-speech impairment-severe cognitive delay syndrome is a rare, genetic neurodegenerative disorder characterized by severe, persistent hypotonia (presenting at birth or in early infancy), severe global developmental delay (with poor or absent speech, difficulty or inability to roll, sit or walk), profound intellectual disability, and failure to thrive. Additional manifestations include microcephaly, progressive peripheral spasticity, bilateral strabismus and nystagmus, constipation, and variable dysmorphic facial features (including plagiocephaly, broad forehead, small nose, low-set ears, micrognathia and open mouth with tented upper lip).

Your genetic map

Gene	SNP	Genotype
UNC80	rs864321623	GG
UNC80	rs200659479	CC
UNC80	rs864321622	CC
UNC80	rs869025317	GG
UNC80	rs869025319	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=371364

Hereditary Diseases (genetics)

Holt-Oram syndrome

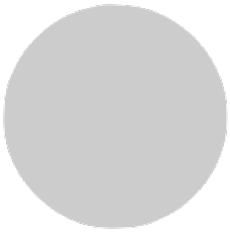
A genetic syndrome with limb reduction defects characterized by skeletal abnormalities of the upper limbs and mild-to-severe congenital cardiac defects.

Your genetic map

Gene	SNP	Genotype
TBX5	rs104894378	CC
TBX5	rs104894382	GG
TBX5	rs863223776	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=392

Hereditary Diseases (genetics)

Hutchinson-Gilford progeria syndrome

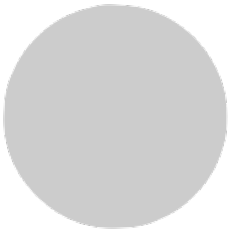
Hutchinson-Gilford progeria syndrome is a rare, fatal, autosomal dominant and premature aging disease, beginning in childhood and characterized by growth reduction, failure to thrive, a typical facial appearance (prominent forehead, protuberant eyes, thin nose with a beaked tip, thin lips, micrognathia and protruding ears) and distinct dermatologic features (generalized alopecia, aged-looking skin, sclerotic and dimpled skin over the abdomen and extremities, prominent cutaneous vasculature, dyspigmentation, nail hypoplasia and loss of subcutaneous fat).

Your genetic map

Gene	SNP	Genotype
LMNA	rs58596362	CC
LMNA	rs267607547	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=740

Hereditary Diseases (genetics)

Ichthyosis follicularis-alopecia-photophobia syndrome

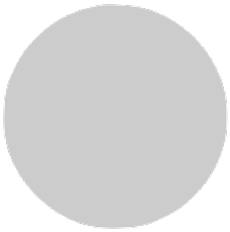
Ichthyosis follicularis - alopecia - photophobia (IFAP) is a rare genetic disorder characterized by the triad of ichthyosis follicularis, alopecia, and photophobia from birth.

Your genetic map

Gene	SNP	Genotype
MBTPS2	rs122468178	GG

Multivariate analysis

What do your genetics tell us?



We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2273

Hereditary Diseases (genetics)

Ichthyosis-prematurity syndrome

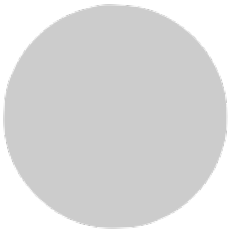
Ichthyosis prematurity syndrome is a rare, syndromic congenital ichthyosis characterized by premature birth (at gestational weeks 30-32, in general) in addition to thick, caseous and desquamating epidermis, neonatal respiratory asphyxia, and persistent eosinophilia. After the perinatal period, a spontaneous improvement in the health of affected patients is observed and skin features (vernix caseosa-like scale) evolve into a mild presentation of flat follicular hyperkeratosis with atopy.

Your genetic map

Gene	SNP	Genotype
SLC27A4	rs137853134	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=88621

Hereditary Diseases (genetics)

Imerslund-Gräsbeck syndrome

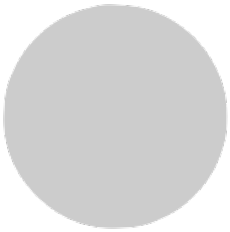
Imerslund-Grasbeck syndrome (IGS) or selective vitamin B12 (cobalamin) malabsorption with proteinuria is a rare autosomal recessive disorder characterized by vitamin B12 deficiency commonly resulting in megaloblastic anemia, which is responsive to parenteral vitamin B12 therapy and appears in childhood.

Your genetic map

Gene	SNP	Genotype
CUBN	rs386833778	GG
CUBN	rs143944436	GG
CUBN	rs374417889	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=35858

Hereditary Diseases (genetics)

Early-onset seizures-distal limb anomalies-facial dysmorphism-global developmental delay syndrome

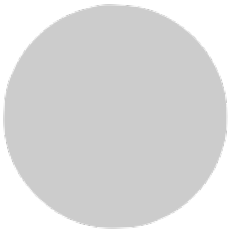
A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by variable developmental delay, intellectual disability, early-onset seizures, and facial dysmorphism (including arched eyebrows, long palpebral fissures, prominent nasal bridge, large ears, thin upper lip, and high arched palate). Other reported features are microcephaly, hypotonia, growth retardation, congenital heart defects, and malformations of the fingers and toes, as well as additional neurologic manifestations (such as ataxia or spastic quadriplegia). Brain imaging may show hypoplastic corpus callosum, white matter abnormalities, or cortical atrophy.

Your genetic map

Gene	SNP	Genotype
OTUD6B	rs368313959	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=505237

Hereditary Diseases (genetics)

Complete androgen insensitivity syndrome

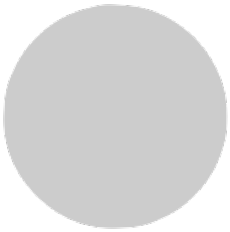
Complete androgen insensitivity syndrome (CAIS) is a form of androgen insensitivity syndrome (AIS; see this term), a disorder of sex development (DSD), characterized by the presence of female external genitalia in a 46,XY individual with normal testis development but undescended testes and unresponsiveness to age-appropriate levels of androgens.

Your genetic map

Gene	SNP	Genotype
AR	rs137852562	CC
AR	rs137852564	GG
AR	rs137852565	GG
AR	rs137852572	GG
AR	rs137852594	CC
AR	rs754201976	GG
AR	rs9332970	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=99429

Hereditary Diseases (genetics)

Partial androgen insensitivity syndrome

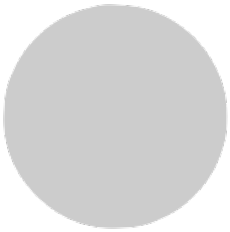
A disorder of sex development (DSD) distinct from complete AIS (CAIS) characterized by the presence of abnormal genital development in a 46,XY individual with normal testis development and partial responsiveness to age-appropriate levels of androgens.

Your genetic map

Gene	SNP	Genotype
AR	rs137852569	GG
AR	rs9332971	GG
AR	rs137852577	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=90797

Hereditary Diseases (genetics)

Acute infantile liver failure-multisystemic involvement syndrome

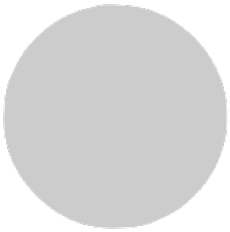
A rare, genetic, parenchymal hepatic disease characterized by acute liver failure, that occurs in the first year of life, which manifests with failure to thrive, hypotonia, moderate global developmental delay, seizures, abnormal liver function tests, microcytic anemia and elevated serum lactate. Other associated features include hepatosteatosis and fibrosis, abnormal brain morphology, and renal tubulopathy. Minor illness exacerbates deterioration of liver failure.

Your genetic map

Gene	SNP	Genotype
NBAS	rs761330483	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=370088

Hereditary Diseases (genetics)

Jackson-Weiss syndrome

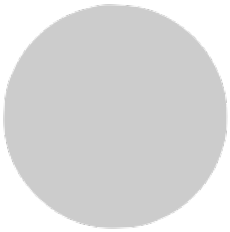
Jackson-Weiss syndrome (JWS) is a rare genetic disorder characterized by foot malformations (tarsal and metatarsal fusions; short, broad, medially deviated great toes) and in some patients craniosynostosis with facial anomalies. Hands are normal in affected patients.

Your genetic map

Gene	SNP	Genotype
FGFR1	rs121909627	GG
FGFR2	rs121918487	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1540

Hereditary Diseases (genetics)

Jeune syndrome

Jeune syndrome, also called asphyxiating thoracic dystrophy, is a short-rib dysplasia characterized by a narrow thorax, short limbs and radiological skeletal abnormalities including 'trident' aspect of the acetabula and metaphyseal changes.

Your genetic map

Gene	SNP	Genotype
DYNC2LI	rs769975073	GG
DYNC2LI	rs745930390	CC

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=474

Hereditary Diseases (genetics)

Johanson-Blizzard syndrome

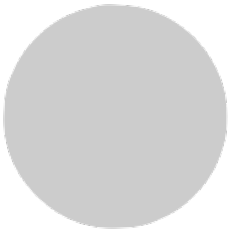
Johanson-Blizzard syndrome (JBS) is a multiple congenital anomaly characterized by exocrine pancreatic insufficiency, hypoplasia/aplasia of the nasal alae, hypodontia, sensorineural hearing loss, growth retardation, anal and urogenital malformations, and variable intellectual disability.

Your genetic map

Gene	SNP	Genotype
UBR1	rs797045112	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2315

Hereditary Diseases (genetics)

Joubert syndrome with hepatic defect

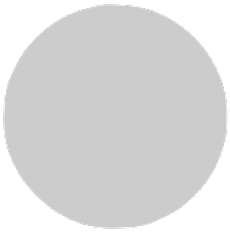
Joubert syndrome with hepatic defect is a very rare subtype of Joubert syndrome and related disorders (JSRD, see this term) characterized by the neurological features of JS associated with congenital hepatic fibrosis (CHF).

Your genetic map

Gene	SNP	Genotype
TMEM67	rs758948621	AA
TMEM67	rs267607119	TT
TMEM67	rs267607115	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1454

Hereditary Diseases (genetics)

Joubert syndrome with ocular defect

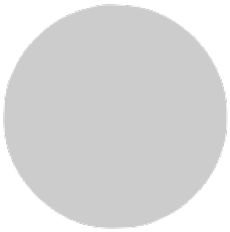
Joubert syndrome with ocular defect is, along with pure JS, the most frequent subtype of Joubert syndrome and related disorders (JSRD, see these terms) characterized by the neurological features of JS associated with retinal dystrophy.

Your genetic map

Gene	SNP	Genotype
AHI1	rs201391050	GG
AHI1	rs397514726	CC
AHI1	rs797045224	TT
AHI1	rs797045223	CC
AHI1	rs372659908	GG
AHI1	rs863225147	TT
AHI1	rs777668842	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=220493

Hereditary Diseases (genetics)

Joubert syndrome with oculorenal defect

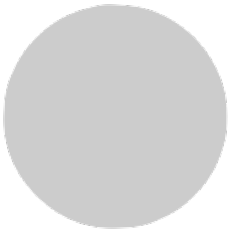
A rare subtype of Joubert syndrome (JS) and related disorders (JSRD) characterized by the neurological features of JS associated with both renal and ocular disease.

Your genetic map

Gene	SNP	Genotype
TMEM216	rs201108965	GG
TMEM216	rs755459875	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2318

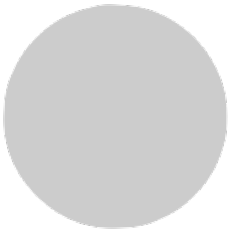
Hereditary Diseases (genetics)

Kabuki syndrome

A rare multiple congenital anomalies/neurodevelopmental disorder characterized by five major features: intellectual disability (typically mild to moderate), visceral malformations (frequently congenital heart defects), persistence of fetal fingertip pads, post-natal short stature, skeletal anomalies (brachymesophalangy, brachydactyly V, spinal column abnormalities and fifth digit clinodactyly) and specific facial features (arched and broad eyebrows, long palpebral fissures, eversion of the lower eyelid, large prominent, cupped ears, depressed nasal tip and short columella). Various additional features are frequently observed.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2322

Your genetic map

Gene	SNP	Genotype
KMT2D	rs267607237	CC
KMT2D	rs398123704	GG
KMT2D	rs398123708	GG
KMT2D	rs398123721	GG
KMT2D	rs398123729	CC
KMT2D	rs587783700	TT
KMT2D	rs587783699	GG
KMT2D	rs587783698	GG
KMT2D	rs587783697	CC
KMT2D	rs587783696	CC
KMT2D	rs587783695	GG
KMT2D	rs587783692	GG
KMT2D	rs587783690	GG
KMT2D	rs587783688	GG
KMT2D	rs587783685	GG
KMT2D	rs587783682	GG
KMT2D	rs587783681	GG
KMT2D	rs587783729	GG
KMT2D	rs587783727	GG
KMT2D	rs556669370	GG
KMT2D	rs587783712	GG
KMT2D	rs587783711	GG
KMT2D	rs587783705	CC
KMT2D	rs587783714	CC
KMT2D	rs587783708	CC
KMT2D	rs727503979	GG
KMT2D	rs727503987	GG
KMT2D	rs727503983	GG
KMT2D	rs794727420	GG
KMT2D	rs794727688	GG
KMT2D	rs797045659	GG

Hereditary Diseases (genetics)

Hypoxanthine guanine phosphoribosyltransferase partial deficiency

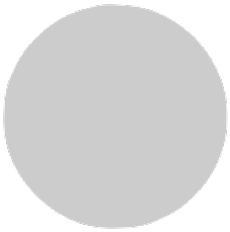
Kelley-Seegmiller syndrome (KSS) is the mildest form of hypoxanthine-guanine phosphoribosyltransferase (HPRT) deficiency (see this term), a hereditary disorder of purine metabolism, and is associated with uric acid overproduction (UAO) leading to urolithiasis, and early-onset gout.

Your genetic map

Gene	SNP	Genotype
HPRT1	rs137852484	GG
HPRT1	rs398123241	GG
HPRT1	rs369065223	CC
HPRT1	rs137852490	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=79233

Hereditary Diseases (genetics)

Stiff skin syndrome

Stiff skin syndrome is a rare, slowly progressive cutaneous disease characterized by rock-hard skin bound firmly to the underlying tissues (mainly on the shoulders, lower back, buttocks and thighs), mild hypertrichosis and hyperpigmentation overlying the affected areas of skin, as well as limited joint mobility (mainly of large joints) with flexion contractures. Cutaneous nodules, affecting mostly distal interphalangeal joints, as well as extracutaneous manifestations, including diffuse entrapment neuropathy, scoliosis, a tiptoe gait and a narrow thorax, may be associated. Restrictive pulmonary changes, muscle weakness, short stature and growth delay have also been reported. No vascular hyperreactivity, immunologic abnormalities nor visceral, muscular or bone involvement has been described.

Your genetic map

Gene	SNP	Genotype
FBN1	rs267606798	CC

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2833

Hereditary Diseases (genetics)

Leigh syndrome

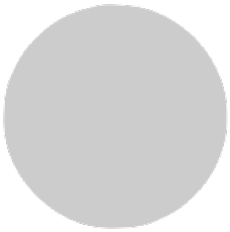
A progressive neurological disease defined by specific neuropathological features associating brainstem and basal ganglia lesions.

Your genetic map

Gene	SNP	Genotype
ATP6	rs199476138	TT
CYTB	rs207459999	GG
MIR3944	rs587776498	GG
MT TK	rs118192098	AA
MT TV	rs199476144	CC
ND6	rs199476109	TT
SURF1	rs782623477	GG
SURF1	rs781948238	CC
SURF1	rs782682492	TT
SURF2	rs863224926	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=506

Hereditary Diseases (genetics)

Leigh syndrome with nephrotic syndrome

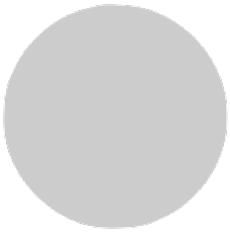
A rare, genetic neurometabolic disease characterized by encephalomyopathy (including developmental delay, nystagmus, progressive ataxia, dystonia, amyotrophy, visual loss, sensorineural deafness, seizures) and bilateral, symmetrical lesions in the basal ganglia or brainstem on imaging, associated with nephrotic syndrome.

Your genetic map

Gene	SNP	Genotype
COQ2	rs121918231	CC
COQ2	rs121918233	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=255249

Hereditary Diseases (genetics)

Lesch-Nyhan syndrome

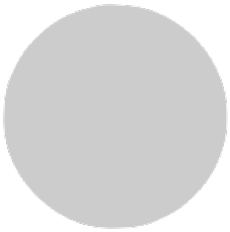
Lesch-Nyhan syndrome (LNS) is the most severe form of hypoxanthine-guanine phosphoribosyltransferase (HPRT) deficiency (see this term), a hereditary disorder of purine metabolism, and is associated with uric acid overproduction (UAO), neurological troubles, and behavioral problems.

Your genetic map

Gene	SNP	Genotype
HPRT1	rs137852487	GG
HPRT1	rs137852488	GG
HPRT1	rs137852489	CC
HPRT1	rs137852490	CC
HPRT1	rs387906725	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=510

Hereditary Diseases (genetics)

Leukoencephalopathy with brain stem and spinal cord involvement-high lactate syndrome

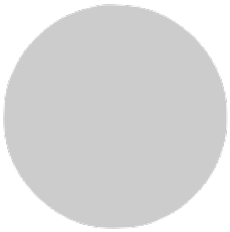
This disease is characterised by progressive cerebellar ataxia with pyramidal and spinal cord dysfunction, associated with distinctive MRI anomalies and increased lactate in the abnormal white matter.

Your genetic map

Gene	SNP	Genotype
DARS2	rs121918207	GG
DARS2	rs121918208	GG
DARS2	rs142433332	TT
DARS2	rs121918210	GG
DARS2	rs182811621	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=137898

Hereditary Diseases (genetics)

Leukoencephalopathy-thalamus and brainstem anomalies-high lactate syndrome

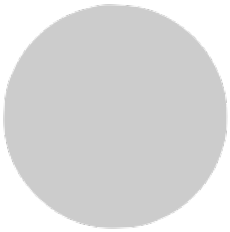
Leukoencephalopathy-thalamus and brainstem anomalies-high lactate (LTBL) syndrome is a rare, genetic neurological disorder defined by early-onset of neurologic symptoms, biphasic clinical course, unique MRI features (incl. extensive, symmetrical, deep white matter abnormalities), and increased lactate in body fluids. The severe form is characterized by delayed psychomotor development, seizures, early-onset hypotonia, and persistently increased lactate levels. The mild form usually presents with irritability, psychomotor regression after six months of age, and temporary high lactate levels, with overall clinical improvement from the second year onward.

Your genetic map

Gene	SNP	Genotype
EARS2	rs376103091	GG
EARS2	rs201842633	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=314051

Hereditary Diseases (genetics)

Leukoencephalopathy-dystonia-motor neuropathy syndrome

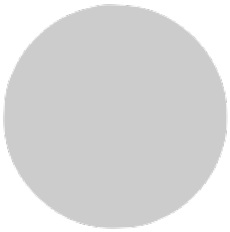
Leukoencephalopathy-dystonia-motor neuropathy syndrome is a peroxisomal neurodegenerative disorder characterized by spasmodic torticollis, dystonic head tremor, intention tremor, nystagmus, hyposmia, and hypergonadotrophic hypogonadism with azoospermia. Slight cerebellar signs (left-sided intention tremor, balance and gait impairment) are also noted. Magnetic resonance imaging (MRI) shows bilateral hyperintense signals in the thalamus, butterfly-like lesions in the pons, and lesions in the occipital region, whereas nerve conduction studies of the lower extremities shows a predominantly motor and slight sensory neuropathy.

Your genetic map

Gene	SNP	Genotype
SCP2	rs144132787	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=163684

Hereditary Diseases (genetics)

Lissencephaly syndrome, Norman-Roberts type

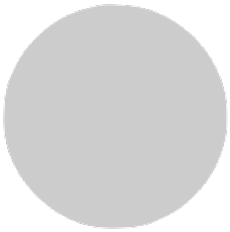
Lissencephaly syndrome, Norman-Roberts type is characterised by the association of lissencephaly type I with craniofacial anomalies (severe microcephaly, a low sloping forehead, a broad and prominent nasal bridge and widely set eyes) and postnatal growth retardation.

Your genetic map

Gene	SNP	Genotype
RELN	rs587780435	GG
RELN	rs587780437	CC
RELN	rs797045915	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=89844

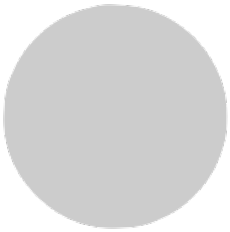
Hereditary Diseases (genetics)

Loeys-Dietz syndrome

Loeys-Dietz syndrome is a rare genetic connective tissue disorder characterized by a broad spectrum of craniofacial, vascular and skeletal manifestations with four genetic subtypes described forming a clinical continuum.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=60030

Your genetic map

Gene	SNP	Genotype
SMAD3	rs587782977	GG
SMAD3	rs730880214	GG
TGFBR1	rs113605875	GG
TGFBR1	rs111854391	CC
TGFBR1	rs111426349	CC
TGFBR1	rs727503470	GG
TGFBR1	rs760079636	GG
TGFBR1	rs886038919	AA
TGFBR2	rs193922664	TT
TGFBR2	rs397516840	GG
TGFBR2	rs587782979	GG
TGFBR2	rs727504292	GG
TGFBR2	rs727503475	GG
TGFBR2	rs727504421	GG
TGFBR2	rs869025537	GG
TGFBR2	rs886039551	GG

Hereditary Diseases (genetics)

Macrocephaly-intellectual disability-autism syndrome

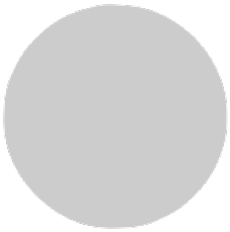
A rare, genetic, neurological disease characterized by association of macrocephaly, dysmorphic facial features and psychomotor delay leading to intellectual disability and autism spectrum disorder. Facial dysmorphism may include frontal bossing, hypertelorism, midface hypoplasia, depressed nasal bridge, short nose, and long philtrum.

Your genetic map

Gene	SNP	Genotype
LOC1079	rs387907053	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=210548

Hereditary Diseases (genetics)

Macrocephaly-intellectual disability-left ventricular non compaction syndrome

Macrocephaly-intellectual disability-left ventricular non compaction syndrome is a rare, genetic, syndromic intellectual disability characterized by motor and cognitive developmental delay with language impairment, macrocephaly, hypotonia, dysmorphic facial features (including long face, slanting palpebral fissures and prominent, flattened nose) and left ventricular noncompaction cardiomyopathy. Patients also present skeletal abnormalities (e.g. scoliosis, finger clinodactyly, pes planus), slender build and shy behavior. Strabismus and various neurological signs (including ataxia, tremor and hyperreflexia) may be associated, as well as epilepsy, autism and MRI findings showing a small cerebellum and abnormalities of the corpus callosum. A phenotypic variant with no cardiac involvement has been reported.

Your genetic map

Gene	SNP	Genotype
NONO	rs869025343	GG

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=466791

Hereditary Diseases (genetics)

Macrothrombocytopenia-lymphedema-developmental delay-facial dysmorphism-camptodactyly syndrome

A rare multiple congenital anomalies/dysmorphic syndrome with intellectual disability characterized by global developmental delay, intellectual disability, macrothrombocytopenia, lymphedema, and dysmorphic facial features (like synophrys, ptosis, eversion of the lateral portion of the lower eyelid, and thin upper lip, among others). Additional reported manifestations include cardiac and genitourinary anomalies, sensorineural hearing loss, ophthalmologic abnormalities, skeletal anomalies, and immunodeficiency. Brain imaging may show enlarged ventricles, cerebellar atrophy, or white matter changes.

Your genetic map

Gene	SNP	Genotype
CDC42	rs797044916	AA
CDC42	rs797044870	AA

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=487796

Hereditary Diseases (genetics)

Lethal fetal brain malformation-duodenal atresia-bilateral renal hypoplasia syndrome

A rare genetic lethal multiple congenital anomalies/dysmorphic syndrome characterized by mid-gestation lethality and features of a ciliopathy. Clinical manifestations include hydrocephalus, cerebellar vermis hypoplasia, corpus callosum agenesis, duodenal atresia, gastrointestinal malrotation, bilateral renal hypoplasia, and dysmorphic craniofacial features (such as microcephaly, hypertelorism, low-set ears, prominent nose, short columella, cleft palate, micrognathia, and wide mouth).

Your genetic map

Gene	SNP	Genotype
CENPF	rs779120472	GG
CENPF	rs375014198	CC

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=444069

Hereditary Diseases (genetics)

3MC syndrome

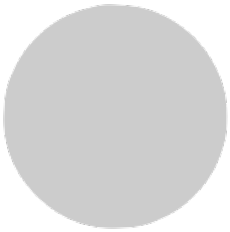
A rare multiple congenital anomalies syndrome characterized by a spectrum of developmental anomalies including cleft lip and/or palate, craniosynostosis, intellectual disability and/or learning disability, radioulnar synostosis, genital and vesicorenal anomalies. Observed facial dysmorphism includes hypertelorism, blepharophimosis, blepharoptosis, high arched eyebrows. Less common features reported include anterior chamber defects, cardiac anomalies (e.g. ventricular septal defect; see this term), caudal appendage, umbilical hernia/omphalocele and diastasis recti.

Your genetic map

Gene	SNP	Genotype
LOC1019	rs149010496	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2453

Hereditary Diseases (genetics)

Marfan syndrome

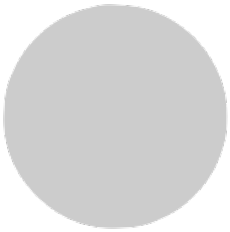
Marfan syndrome is a systemic disease of connective tissue characterized by a variable combination of cardiovascular, musculo-skeletal, ophthalmic and pulmonary manifestations.

Your genetic map

Gene	SNP	Genotype
TGFBR2	rs121918715	GG
TGFBR2	rs104893809	CC
TGFBR2	rs104893815	GG
TGFBR2	rs104893810	CC
TGFBR2	rs104893811	CC
TGFBR2	rs104893816	GG
TGFBR2	rs104893819	CC
TGFBR2	rs863224935	TT
TGFBR2	rs886038794	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=558

Hereditary Diseases (genetics)

Marinesco Sjogren syndrome

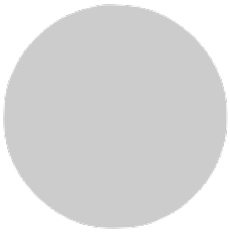
Marinesco Sjogren syndrome (MSS) belongs to the group of autosomal recessive cerebellar ataxias. Cardinal features of MSS are cerebellar ataxia, congenital cataract, and delayed psychomotor development.

Your genetic map

Gene	SNP	Genotype
SIL1	rs119456966	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=559

Hereditary Diseases (genetics)

Marshall syndrome

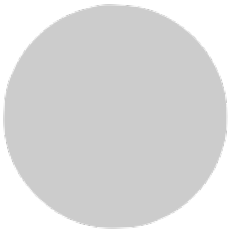
A malformation syndrome that is characterized by facial dysmorphism, severe hypoplasia of the nasal bones and frontal sinuses, ocular involvement, early-onset hearing loss, skeletal and anhidrotic ectodermal anomalies and short stature with spondyloepiphyseal dysplasia and early-onset osteoarthritis.

Your genetic map

Gene	SNP	Genotype
COL11A1	rs398122828	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=560

Hereditary Diseases (genetics)

McCune-Albright syndrome

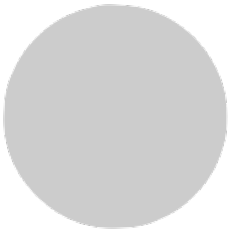
McCune-Albright syndrome (MAS) is classically defined by the clinical triad of fibrous dysplasia of bone (FD), cafe-au-lait skin spots, and precocious puberty (PP).

Your genetic map

Gene	SNP	Genotype
GNAS	rs121913495	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=562

Hereditary Diseases (genetics)

McKusick-Kaufman syndrome

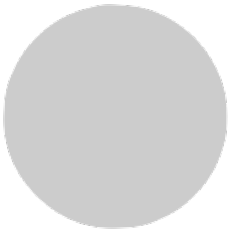
A rare, genetic multiple congenital anomalies syndrome characterized by genitourinary malformations (hydrometrocolpos in females and in males, glanular hypospadias and prominent scrotal raphe) , postaxial polydactyly that may affect only one or several limbs, and to a lesser extent cardiac defects. Hydrometrocolpos is due to either a congenital obstruction, imperforate hymen or vaginal atressia, and causes a palpable mass and possibly hydronephrosis. Other anomalies occasionally reported include choanal atresia, pituitary dysplasia, esophageal atresia and distal tracheoesophageal fistula, Hirschsprung disease, vertebral anomalies, and hydrops fetalis. The disorder is allelic with Bardet-Biedl, and as some phenotypic overlap has been observed, patients should be reevaluated in later childhood for retinistis pigmentosas and other signs of Bardet-Biedl syndrome.

Your genetic map

Gene	SNP	Genotype
MKKS	rs74315396	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2473

Hereditary Diseases (genetics)

Meacham syndrome

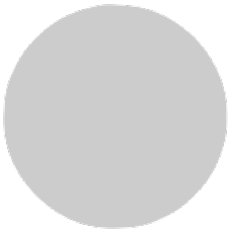
Meacham syndrome is a multiple malformation syndrome characterized by congenital diaphragmatic abnormalities, genital defects and cardiac malformations.

Your genetic map

Gene	SNP	Genotype
WT1	rs121907910	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3097

Hereditary Diseases (genetics)

Goldberg-Shprintzen megacolon syndrome

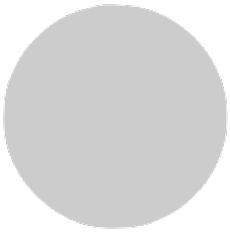
A rare multiple congenital anomalies/dysmorphic syndrome characterized by Hirschsprung disease, facial dysmorphism (sloping forehead, high arched eyebrows, long eyelashes, telecanthus/hypertelorism, ptosis, prominent ears, thick earlobes, prominent nasal bridge, thick philtrum, everted lower lip vermillion and pointed chin), global developmental delay, intellectual disability and variable cerebral abnormalities (focal or generalized polymicrogyria, or hypoplastic corpus callosum).

Your genetic map

Gene	SNP	Genotype
KIFBP	rs730882150	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=66629

Hereditary Diseases (genetics)

Megalencephaly-severe kyphoscoliosis-overgrowth syndrome

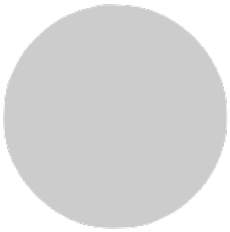
A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by overgrowth and macrocephaly with megalencephaly apparent at birth, global developmental delay, intellectual disability, and dysmorphic facial features (including frontal bossing, long face, sparse eyebrows, hypertelorism, downslanting palpebral fissures, and prognathism). Patients may exhibit tall stature with dolichostenomelia, arachnodactyly, kyphoscoliosis, and joint laxity, as well as neurologic manifestations, such as hypotonia, gait ataxia, or seizures. Brain imaging may show increased white matter volume, thick corpus callosum, or small cerebellum.

Your genetic map

Gene	SNP	Genotype
HERC1	rs753780877	GG
HERC1	rs797045141	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=457359

Hereditary Diseases (genetics)

Megalencephaly-capillary malformation-polymicrogyria syndrome

A rare developmental defect during embryogenesis that is characterized by growth dysregulation with overgrowth of the brain and multiple somatic tissues, with capillary skin malformations, megalencephaly (MEG) or hemimegalencephaly (HMEG), cortical brain abnormalities (in particular polymicrogyria), typical facial dysmorphisms, abnormalities of somatic growth with asymmetry of the body and brain, developmental delay and digital anomalies.

Your genetic map

Gene	SNP	Genotype
PIK3CA	rs587776932	GG

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=60040

Hereditary Diseases (genetics)

Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome

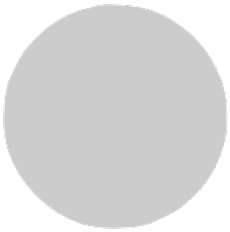
A rare syndrome with a central nervous system malformation as a major feature characterized by macrocephaly, megalencephaly, bilateral perisylvian polymicrogyria, variable degrees of ventriculomegaly/hydrocephalus, developmental delay and intellectual disability, oromotor dysfunction, hypotonia, seizures, and dysmorphic facial features (such as frontal bossing, low-set ears, a flat nasal bridge, and high-arched palate). Postaxial polydactyly of one or more extremities is also common.

Your genetic map

Gene	SNP	Genotype
CCND2	rs587777620	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=83473

Hereditary Diseases (genetics)

Familial atypical multiple mole melanoma syndrome

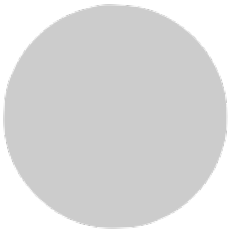
Familial atypical multiple mole melanoma (FAMMM) syndrome is an inherited genodermatosis characterized by the presence of multiple melanocytic nevi (often >50) and a family history of melanoma as well as, in a subset of patients, an increased risk of developing pancreatic cancer and other malignancies.

Your genetic map

Gene	SNP	Genotype
CDKN2A	rs199907548	AA
CDKN2A	rs730881677	CC
CDKN2A	rs1800586	CC
CDKN2A	rs45476696	CC
CDKN2A	rs749714198	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=404560

Hereditary Diseases (genetics)

Congenital microcephaly-severe encephalopathy-progressive cerebral atrophy syndrome

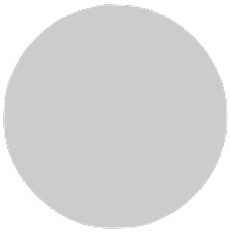
Congenital microcephaly-severe encephalopathy-progressive cerebral atrophy syndrome is a rare, genetic, neurometabolic disorder characterized by severe, progressive microcephaly, severe to profound global development delay, intellectual disability, seizures (typically tonic and/or myoclonic and frequently intractable), hyperekplexia, and axial hypotonia with appendicular spasticity, as well as hyperreflexia, dyskinetic quadriplegia, and abnormal brain morphology (cerebral atrophy with variable additional features including ventriculomeglay, pons and/or cerebellar hypoplasia, simplified gyral pattern and delayed myelination). Cortical blindness, feeding difficulties and respiratory insufficiency may also be associated.

Your genetic map

Gene	SNP	Genotype
CZ1P	rs398122974	GG
CZ1P	rs398122975	GG
CZ1P	rs754043007	GG
CZ1P	rs148111963	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=391376

Hereditary Diseases (genetics)

Postnatal microcephaly-infantile hypotonia-spastic diplegia-dysarthria-intellectual disability syndrome

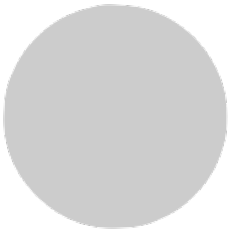
A rare genetic neurological disorder characterized by postnatal microcephaly, hypotonia during infancy followed in most cases by progressive spasticity mainly affecting the lower limbs, and spastic diplegia or paraplegia, intellectual disability, delayed or absent speech, and dysarthria. Seizures and mildly dysmorphic features have been described in some patients.

Your genetic map

Gene	SNP	Genotype
GPT2	rs115352435	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=477673

Hereditary Diseases (genetics)

Macrocephaly-intellectual disability-neurodevelopmental disorder-small thorax syndrome

A rare multiple congenital anomalies/dysmorphic syndrome with intellectual disability, characterized by macrocephaly, intellectual disability, seizures, dysmorphic facial features (including tall forehead, downslanting palpebral fissures, hypertelorism, depressed nasal bridge, and macrostomia), megalencephaly, and small thorax. Other reported features are umbilical hernia, muscular hypotonia, global developmental delay, autistic behavior, and cafe-au-lait spots, among others.

Your genetic map

Gene	SNP	Genotype
MTOR	rs786205165	CC
MTOR	rs863225264	CC
MTOR	rs878855328	CC

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=457485

Hereditary Diseases (genetics)

Microcephaly-corpus callosum hypoplasia-intellectual disability-facial dysmorphism syndrome

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by variable degrees of developmental delay and intellectual disability with poor or absent speech, hypotonia, hypoplastic or absent corpus callosum, and facial dysmorphism (such as long face, frontal bossing, hypertelorism, downslanting palpebral fissures, and tented upper lip). Additional reported features include microcephaly, seizures, gait ataxia, scoliosis, and syndactyly of fingers, among others.

Your genetic map

Gene	SNP	Genotype
PPP2R1A	rs786205227	CC
PPP2R1A	rs786205228	CC
PPP2R1A	rs863225094	GG

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=457284

Hereditary Diseases (genetics)

Microcephaly-lymphedema-chorioretinopathy syndrome

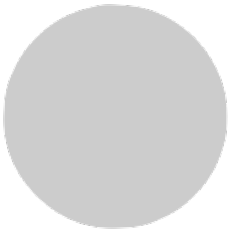
Microcephaly with or without chorioretinopathy, lymphedema or intellectual disability (MCLID) is a rare autosomal dominant condition characterized by variable expression of microcephaly, ocular disorders including chorioretinopathy, congenital lymphedema of the lower limbs, and mild to moderate intellectual disability.

Your genetic map

Gene	SNP	Genotype
KIF11	rs797045650	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2526

Hereditary Diseases (genetics)

Microcephaly-capillary malformation syndrome

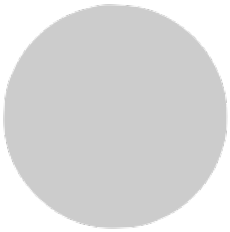
Microcephaly-capillary malformation syndrome is a rare, genetic vascular anomaly characterized by severe congenital microcephaly, poor somatic growth, diffuse multiple capillary malformations on the skin, intractable epilepsy, profound global developmental delay, spastic quadriparesis and hypoplastic distal phalanges.

Your genetic map

Gene	SNP	Genotype
LOC1053	rs143739249	CC
STAMBP	rs397509390	CC
STAMBP	rs797046015	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=294016

Hereditary Diseases (genetics)

5q14.3 microdeletion syndrome

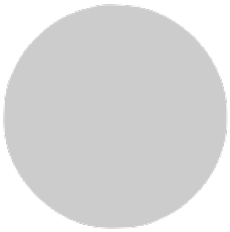
The newly described 5q14.3 microdeletion syndrome includes severe intellectual deficit with no speech, stereotypic movements and epilepsy.

Your genetic map

Gene	SNP	Genotype
MEF2C	rs587783747	GG
MEF2C	rs796052733	GG
MEF2C	rs797045053	TT
MEF2C	rs545185248	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=228384

Hereditary Diseases (genetics)

Colobomatous microphthalmia-rhizomelic dysplasia syndrome

Colobomatous microphthalmia-rhizomelic dysplasia syndrome is a rare, genetic developmental defect during embryogenesis characterized by a range of developmental eye anomalies (including anophthalmia, microphthalmia, colobomas, microcornea, corectopia, cataract) and symmetric limb rhizomelia with short stature and contractures of large joints. Intellectual disability with autistic features, macrocephaly, dysmorphic features, urogenital anomalies (hypospadia, cryptorchidism), cutaneous syndactyly and precocious puberty may also be present.

Your genetic map

Gene	SNP	Genotype
MAB21L2	rs587777511	GG
MAB21L2	rs587777512	CC
MAB21L2	rs587777513	GG
MAB21L2	rs587777514	GG

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=424099

Hereditary Diseases (genetics)

Action myoclonus-renal failure syndrome

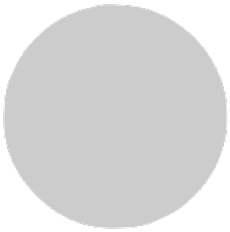
A rare epilepsy syndrome characterized by progressive myoclonus epilepsy in association with primary glomerular disease. Patients present with neurologic symptoms (including tremor, action myoclonus, tonic-clonic seizures, later ataxia and dysarthria) that may precede, occur simultaneously or be followed by renal manifestations including proteinuria that progresses to nephrotic syndrome and end-stage renal disease. In some patients, sensorimotor peripheral neuropathy, sensorineural hearing loss and dilated cardiomyopathy are associated symptoms.

Your genetic map

Gene	SNP	Genotype
SCARB2	rs200053119	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=163696

Hereditary Diseases (genetics)

Early-onset myopathy-areflexia-respiratory distress-dysphagia syndrome

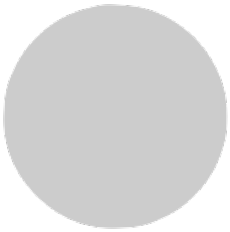
A rare congenital myopathy characterized by early onset of severe muscular weakness, respiratory distress due to diaphragmatic paralysis, dysphagia and areflexia, joint contractures, and scoliosis. Decreased fetal movements are seen in some individuals. Muscle biopsy may show a combination of dystrophic and myopathic features. The clinical course is variable, with some patients becoming ventilator-dependent and never achieving ambulation.

Your genetic map

Gene	SNP	Genotype
MEGF10	rs387907071	CC
MEGF10	rs387907072	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=439212

Hereditary Diseases (genetics)

Mohr-Tranebjaerg syndrome

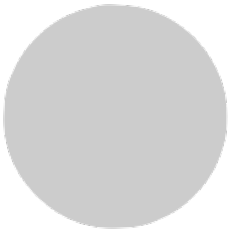
An X-linked syndromic intellectual disability characterized by clinical manifestations commencing with early childhood onset hearing loss, followed by adolescent onset progressive dystonia or ataxia, visual impairment from early adulthood onwards and dementia from the 4th decade onwards.

Your genetic map

Gene	SNP	Genotype
TIMM8A	rs80356560	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=52368

Hereditary Diseases (genetics)

Mowat-Wilson syndrome

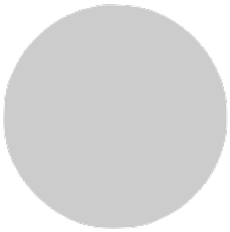
A rare multiple congenital anomaly syndrome characterized by a distinct facial phenotype, intellectual disability, epilepsy, Hirschsprung disease (HSCR) and variable congenital malformations.

Your genetic map

Gene	SNP	Genotype
ZEB2	rs137852981	GG
ZEB2	rs398124274	GG
ZEB2	rs398124282	AA
ZEB2	rs587784566	GG
ZEB2	rs587784571	GG
ZEB2	rs587784570	GG
ZEB2	rs727504224	CC
ZEB2	rs786204815	GG
ZEB2	rs797046121	GG
ZEB2	rs797046122	GG
ZEB2	rs886041338	GG
ZEB2	rs587784563	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2152

Hereditary Diseases (genetics)

Muckle-Wells syndrome

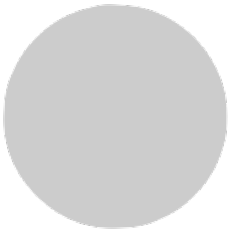
Muckle-Wells syndrome (MWS) is an intermediate form of cryopyrin-associated periodic syndrome (CAPS; see this term) and is characterized by recurrent fever (with malaise and chills), recurrent urticaria-like skin rash, sensorineural deafness, general signs of inflammation (eye redness, headaches, arthralgia/myalgia) and potentially life-threatening secondary amyloidosis (AA type).

Your genetic map

Gene	SNP	Genotype
NLRP3	rs121908149	CC
NLRP3	rs121908150	CC
NLRP3	rs121908153	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=575

Hereditary Diseases (genetics)

Muir-Torre syndrome

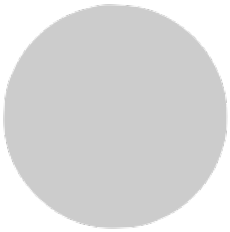
A form of hereditary nonpolyposis colon cancer characterized by the development of cutaneous sebaceous neoplasia and at least one visceral malignancy, most frequently gastrointestinal carcinoma. The malignancies are usually multiple, occur at an early age, but tend to be of low-grade and have a relatively low incidence of metastases. Sebaceous tumors are usually multiple, with sebaceous adenomas being the commonest. Multiple keratoacanthomas, usually located on the face or the trunk, have been reported as a feature. Cutaneous tumors may precede or follow the first presentation of internal malignancy, which usually involves the gastrointestinal tract, the breast or the genitourinary tract.

Your genetic map

Gene	SNP	Genotype
MLH1	rs587778913	CC
MLH1	rs63749900	GG
MLH1	rs587778983	AA
MLH1	rs267607745	GG
MLH1	rs267607795	GG
MSH2	rs63750047	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=587

Hereditary Diseases (genetics)

Mulibrey nanism

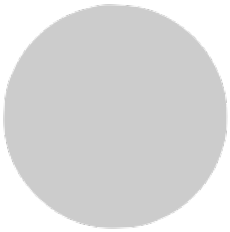
A rare developmental defect during embryogenesis characterized by growth delay and multiorgan manifestations.

Your genetic map

Gene	SNP	Genotype
TRIM37	rs386834008	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2576

Hereditary Diseases (genetics)

Myhre syndrome

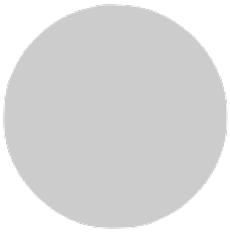
A rare multiple congenital anomalies syndrome characterized by short stature, distinctive facial dysmorphism, brachydactyly, stiff and thick skin, muscular pseudohypertrophy, restricted joint mobility, hearing loss, and variable intellectual disability. Cardiovascular and respiratory involvement are common.

Your genetic map

Gene	SNP	Genotype
SMAD4	rs281875321	TT
SMAD4	rs281875322	AA
SMAD4	rs397518413	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2588

Hereditary Diseases (genetics)

Nager syndrome

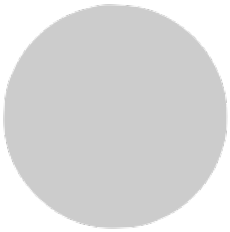
A congenital malformation syndrome characterized by mandibulofacial dystosis (malar hypoplasia, micrognathia, external ear malformations) and variable preaxial limb defects.

Your genetic map

Gene	SNP	Genotype
SF3B4	rs797045955	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=245

Hereditary Diseases (genetics)

Nance-Horan syndrome

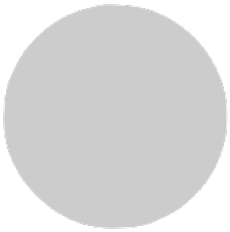
Nance-Horan syndrome (NHS) is characterized by the association in male patients of congenital cataracts with microcornea, dental anomalies and facial dysmorphism.

Your genetic map

Gene	SNP	Genotype
NHS	rs132630322	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=627

Hereditary Diseases (genetics)

Netherton syndrome

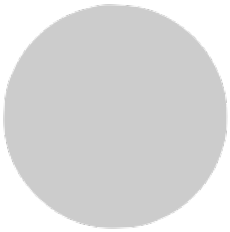
Netherton syndrome (NS) is a skin disorder characterized by congenital ichthyosiform erythroderma (CIE), a distinctive hair shaft defect (trichorrhexis invaginata; TI) and atopic manifestations.

Your genetic map

Gene	SNP	Genotype
SPINK5	rs199757347	CC
SPINK5	rs368134354	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=634

Hereditary Diseases (genetics)

Peripheral neuropathy-myopathy-hoarseness-hearing loss syndrome

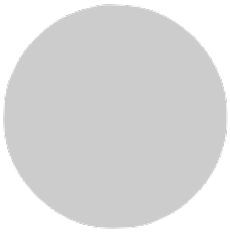
Peripheral neuropathy-myopathy-hoarseness-hearing loss syndrome is a rare, syndromic genetic deafness characterized by a combination of muscle weakness, chronic neuropathic and myopathic features, hoarseness and sensorineural hearing loss. A wide range of disease onset and severity has been reported even within the same family.

Your genetic map

Gene	SNP	Genotype
MYH14	rs113993956	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=397744

Hereditary Diseases (genetics)

Noonan syndrome with multiple lentigines

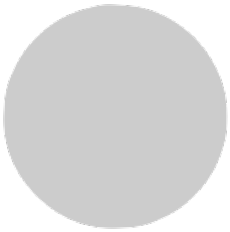
A rare multisystem genetic disorder characterized by cutaneous lentigines, hypertrophic cardiomyopathy, short stature, pectus deformity, and dysmorphic facial features.

Your genetic map

Gene	SNP	Genotype
PTPN11	rs121918456	AA
PTPN11	rs121918457	CC
PTPN11	rs121918468	GG
PTPN11	rs121918469	GG
PTPN11	rs121918470	AA
PTPN11	rs397507510	GG
PTPN11	rs397507529	AA
PTPN11	rs397507537	AA
PTPN11	rs397507541	CC
PTPN11	rs397507542	GG
PTPN11	rs397507548	AA
PTPN11	rs397507549	CC
RAF1	rs80338799	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=500

Hereditary Diseases (genetics)

Omenn syndrome

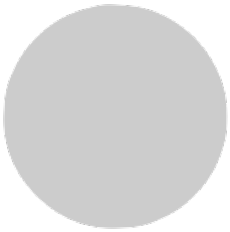
Omenn syndrome (OS) is an inflammatory condition characterized by erythroderma, desquamation, alopecia, chronic diarrhea, failure to thrive, lymphadenopathy, and hepatosplenomegaly, associated with severe combined immunodeficiency (SCID; see this term).

Your genetic map

Gene	SNP	Genotype
IFTAP	rs36001797	CC
RAG1	rs104894284	GG
RAG1	rs104894291	GG
RAG1	rs104894285	CC
RAG1	rs104894286	GG
RAG1	rs121918571	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=39041

Hereditary Diseases (genetics)

Opitz GBBB syndrome

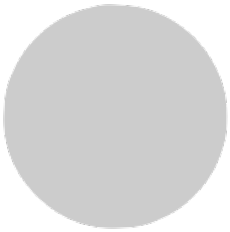
A rare X-linked congenital midline malformation syndrome characterized by hypertelorism, laryngo-tracheo-esophageal defects and hypospadias.

Your genetic map

Gene	SNP	Genotype
MID1	rs398123341	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2745

Hereditary Diseases (genetics)

Ear-patella-short stature syndrome

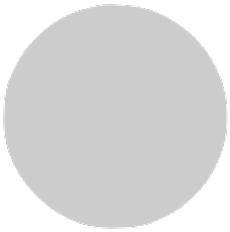
Ear-patella-short stature syndrome is an association of malformations including bilateral microtia (severe hypoplasia of ear pinnae), absent patellae, short stature, poor weight gain, and characteristic facial features such as high forehead, micrognathism with full lips and small mouth, and accentuated nasolabial folds (smile wrinkles linking the nostrils to the labial commissure).

Your genetic map

Gene	SNP	Genotype
GMNN	rs864309488	AA
ORC1	rs143141689	CC
ORC1	rs387906828	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2554

Hereditary Diseases (genetics)

Osteopathia striata-cranial sclerosis syndrome

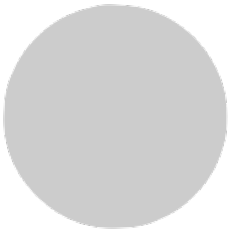
Osteopathia striata with cranial sclerosis (OS-CS) is a bone dysplasia characterized by longitudinal striations of the metaphyses of the long bones, sclerosis of the craniofacial bones, macrocephaly, cleft palate and hearing loss.

Your genetic map

Gene	SNP	Genotype
AMER1	rs137852217	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2780

Hereditary Diseases (genetics)

Osteoporosis-pseudoglioma syndrome

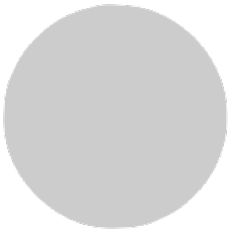
Osteoporosis pseudoglioma syndrome is a very rare autosomal recessive disorder characterized by congenital or infancy-onset blindness and severe juvenile-onset osteoporosis and spontaneous fractures.

Your genetic map

Gene	SNP	Genotype
LRP5	rs121908664	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2788

Hereditary Diseases (genetics)

Pancytopenia-developmental delay syndrome

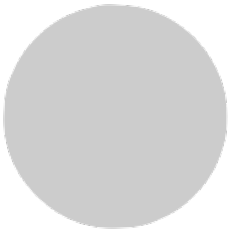
Pancytopenia-developmental delay syndrome is a rare, genetic, hematologic disorder characterized by progressive trilineage bone marrow failure (with hypocellularity), developmental delay with learning disabilities, and microcephaly. Mild facial dysmorphism and hypotonia have also been reported.

Your genetic map

Gene	SNP	Genotype
ERCC6L2	rs147948835	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=401764

Hereditary Diseases (genetics)

Early-onset parkinsonism-intellectual disability syndrome

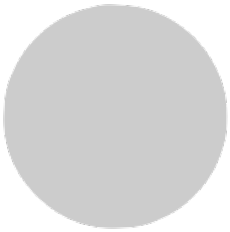
A rare X-linked syndromic intellectual disability characterized by infantile-onset non-progressive intellectual deficit (with psychomotor developmental delay, cognitive impairment and macrocephaly) and early-onset parkinsonism (before 45 years of age), in male patients.

Your genetic map

Gene	SNP	Genotype
RAB39B	rs587777874	GG
RAB39B	rs864309527	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2379

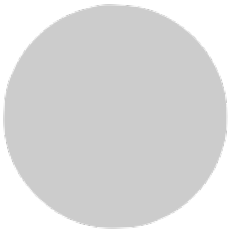
Hereditary Diseases (genetics)

Pendred syndrome

A syndromic genetic deafness clinically variable characterized by bilateral sensorineural hearing loss and euthyroid goiter.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=705

Your genetic map

Gene	SNP	Genotype
SLC26A4	rs80338848	TT
SLC26A4	rs80338849	GG
SLC26A4	rs111033244	AA
SLC26A4	rs121908363	CC
SLC26A4	rs111033307	TT
SLC26A4	rs111033348	CC
SLC26A4	rs111033199	GG
SLC26A4	rs111033254	TT
SLC26A4	rs397516414	GG
SLC26A4	rs111033305	GG
SLC26A4	rs111033311	GG
SLC26A4	rs397516416	CC
SLC26A4	rs397516418	TT
SLC26A4	rs111033316	AA
SLC26A4	rs111033312	GG
SLC26A4	rs111033257	GG
SLC26A4	rs397516424	AA
SLC26A4	rs111033318	TT
SLC26A4	rs111033256	TT
SLC26A4	rs397516432	TT
SLC26A4	rs111033454	GG
SLC26A4	rs111033245	GG
SLC26A4	rs727503430	GG
SLC26A4	rs727503431	CC
SLC26A4	rs542620119	GG
SLC26A4	rs146281367	GG
SLC26A4	rs876657722	GG
SLC26A4	rs147952620	CC
SLC26A4	rs111033200	CC
SLC26A4	rs111033302	TT
SLC26A4	rs397516430	CC

Hereditary Diseases (genetics)

Perry syndrome

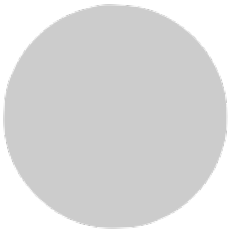
A rare inherited neurodegenerative disorder characterized by rapidly progressive early-onset parkinsonism, central hypoventilation, weight loss, insomnia and depression.

Your genetic map

Gene	SNP	Genotype
DCTN1	rs72466487	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=178509

Hereditary Diseases (genetics)

Peters plus syndrome

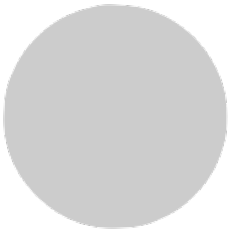
Peters plus syndrome is an autosomal recessively inherited syndromic developmental defect of the eye characterized by a variable phenotype including Peters anomaly and other anterior chamber eye anomalies, short limbs, limb abnormalities (i.e. rhizomelia and brachydactyly), characteristic facial features (upper lip with cupid bow, short palpebral fissures), cleft lip/palate, and mild to severe developmental delay/intellectual disability. Other associated abnormalities reported in some patients include congenital heart defects (i.e. hypoplastic left heart, absence of right pulmonary vein, bicuspid pulmonary valve), genitourinary anomalies (hydronephrosis, renal hypoplasia, renal and ureteral duplication, multicystic dysplastic kidneys, glomerulocystic kidneys) and congenital hypothyroidism.

Your genetic map

Gene	SNP	Genotype
B3GLCT	rs80338851	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=709

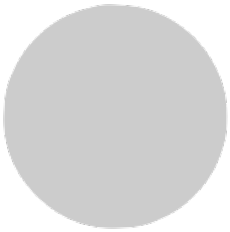
Hereditary Diseases (genetics)

Peutz-Jeghers syndrome

A genetic intestinal polyposis syndrome characterized by development of characteristic hamartomatous polyps throughout the gastrointestinal (GI) tract, and by mucocutaneous pigmentation. This disorder carries a considerably increased risk of GI and extra-GI malignancies.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2869

Your genetic map

Gene	SNP	Genotype
STK11	rs137853076	AA
STK11	rs137854584	GG
STK11	rs121913315	GG
STK11	rs137853082	GG
STK11	rs137853083	CC
STK11	rs398123406	GG
STK11	rs587782018	GG
STK11	rs730881971	GG
STK11	rs730881979	GG
STK11	rs730881976	CC
STK11	rs730881984	GG
STK11	rs786201349	CC
STK11	rs786202134	CC
STK11	rs786201213	CC
STK11	rs786201090	CC
STK11	rs863224448	GG
STK11	rs876658584	AA
STK11	rs886037926	AA
STK11	rs886037859	AA
STK11	rs886039554	GG
STK11	rs1057517830	GG
STK11	rs121913324	CC
STK11	rs1057520039	CC
STK11	rs775595174	GG
STK11	rs1131690950	GG
STK11	rs1131690925	CC
STK11	rs1131690951	AA
STK11	rs730881973	CC
STK11	rs1131690923	CC
STK11	rs1131690940	CC
STK11	rs1131690921	GG

Hereditary Diseases (genetics)

Pfeiffer syndrome

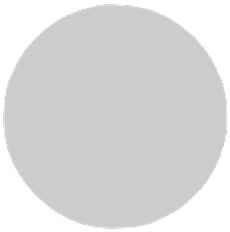
An acrocephalosyndactyly associated with craniosynostosis, midfacial hypoplasia, hand and foot malformation with a wide range of clinical expression and severity. Most of the affected patients show various other associated manifestations.

Your genetic map

Gene	SNP	Genotype
FGFR2	rs121918488	AA
FGFR2	rs121918495	TT
FGFR2	rs121918499	CC
FGFR2	rs121918505	AA
FGFR2	rs121918506	TT
FGFR2	rs121918510	TT
FGFR2	rs776587763	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=710

Hereditary Diseases (genetics)

Pierson syndrome

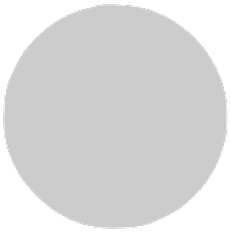
A rare primary glomerular disease characterized by the association of congenital nephrotic syndrome, early onset renal failure and ocular anomalies with microcoria and severe neurodevelopment deficits.

Your genetic map

Gene	SNP	Genotype
LAMB2	rs121912488	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2670

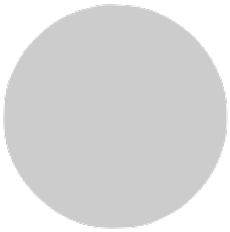
Hereditary Diseases (genetics)

Pitt-Hopkins syndrome

A rare multiple congenital anomalies syndrome characterized by the association of intellectual deficit, characteristic facial morphology and problems of abnormal and irregular breathing.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2896

Your genetic map

Gene	SNP	Genotype
TCF4	rs121909120	GG
TCF4	rs121909121	CC
TCF4	rs121909122	GG
TCF4	rs121909123	CC
TCF4	rs398123560	CC
TCF4	rs587784462	CC
TCF4	rs587784460	CC
TCF4	rs587784459	CC
TCF4	rs587784458	CC
TCF4	rs587784469	CC
TCF4	rs587784466	CC
TCF4	rs587784464	GG
TCF4	rs727504175	GG
TCF4	rs796053418	GG
TCF4	rs797045072	CC
TCF4	rs797046034	TT
TCF4	rs797046033	GG
TCF4	rs863224934	TT

Hereditary Diseases (genetics)

Short rib-polydactyly syndrome, Majewski type

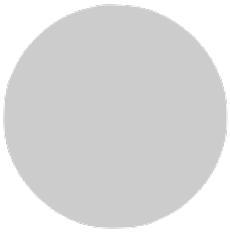
A rare ciliopathy with major skeletal involvement characterized by a hypoplastic thorax with short ribs and protuberant abdomen, micromelia with particularly short tibiae with ovoid configuration, pre- and postaxial polydactyly, brachydactyly, hypoplasia or aplasia of nails, and dysmorphic craniofacial features (such as prominent forehead, low-set and malformed ears, short and flat nose, lobulated tongue, micrognathia, and cleft lip/palate). Additional reported manifestations include urogenital, gastrointestinal, cardiovascular, and cerebral malformations, among others. The condition is fatal in the neonatal period.

Your genetic map

Gene	SNP	Genotype
EVC2	rs769864196	GG
NEK1	rs794727032	CC
NEK1	rs794727285	CC
NEK1	rs199947197	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=93269

Hereditary Diseases (genetics)

Serrated polyposis syndrome

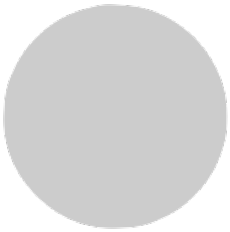
A rare, genetic intestinal disease characterized by the presence of multiple (usually large) hyperplastic/serrated colorectal polyps, usually with a pancolonic distribution. Histology reveals hyperplastic polyps, sessile serrated adenomas (most common), traditional serrated adenomas or mixed polyps. It is associated with an increased personal and familial (first-degree relatives) risk of colorectal cancer.

Your genetic map

Gene	SNP	Genotype
RNF43	rs786205215	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=157798

Hereditary Diseases (genetics)

Autosomal recessive multiple pterygium syndrome

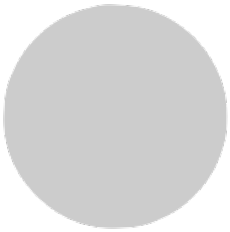
A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by congenital pterygia (webbing) mainly affecting the neck and large joints, arthrogryposis multiplex, short stature, and craniofacial dysmorphism (including ptosis, downslanting palpebral fissures, high-arched palate, and retrognathia). Additional manifestations are decreased movements, facial weakness, respiratory distress, vertebral anomalies, scoliosis, anomalies of the fingers, and cryptorchidism, among others. The disease is a non-lethal variant of multiple pterygium syndrome.

Your genetic map

Gene	SNP	Genotype
CHRNG	rs121912672	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2990

Hereditary Diseases (genetics)

Autosomal dominant popliteal pterygium syndrome

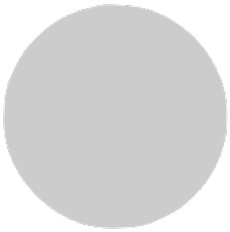
A rare genetic, multiple congenital anomalies syndrome characterized by cleft lip, with or without cleft palate, pits in the lower lip, contractures of the lower extremities, abnormal external genitalia, syndactyly of fingers and/or toes, and a pyramidal skin fold over the hallux nail.

Your genetic map

Gene	SNP	Genotype
IRF6	rs121434226	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1300

Hereditary Diseases (genetics)

Familial short QT syndrome

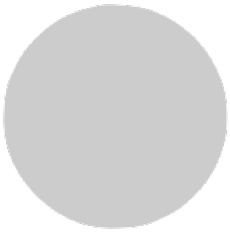
A rare, genetic cardiac rhythm disease characterized by a short QTc interval on the surface electrocardiogram (ECG) with a high risk of syncope or sudden death due to malignant ventricular arrhythmia.

Your genetic map

Gene	SNP	Genotype
KCNH2	rs794728382	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=51083

Hereditary Diseases (genetics)

Palmoplantar keratoderma-deafness syndrome

Palmoplantar keratoderma-deafness syndrome is a keratinization disorder characterized by focal or diffuse palmoplantar keratoderma. A patchy distribution is observed with accentuation on the thenars, hypothenars and the arches of the feet. The disease becomes apparent in infancy and is associated with sensorineural hearing loss that shows a variable age of onset. Due to genetic and clinical similarities, it has been proposed that palmoplantar keratoderma-deafness syndrome, knuckle pads-leukonychia-sensorineural deafness-palmoplantar hyperkeratosis syndrome and keratoderma hereditarium mutilans may represent variants of one broad disorder of syndromic deafness with heterogeneous phenotype. The disease is transmitted in an autosomal dominant manner with incomplete penetrance.

Your genetic map

Gene	SNP	Genotype
GJB2	rs28931593	CC

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2202

Hereditary Diseases (genetics)

Resistance to thyrotropin-releasing hormone syndrome

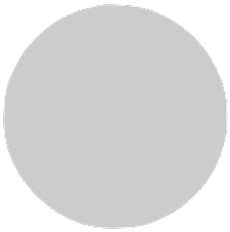
Resistance to thyrotropin-releasing hormone (TRH) syndrome is a type of central congenital hypothyroidism characterized by low levels of thyroid hormones due to insufficient release of thyroid-stimulating hormone (TSH) caused by pituitary resistance to TRH. It may or may not be observed from birth.

Your genetic map

Gene	SNP	Genotype
TRHR	rs121917847	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=99832

Hereditary Diseases (genetics)

Insulin-resistance syndrome type A

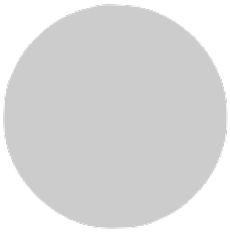
Type A insulin-resistance syndrome belongs to the group of extreme insulin-resistance syndromes (which includes leprechaunism, the lipodystrophies, Rabson-Mendenhall syndrome and type B insulin resistance syndrome; see these terms) and is characterized by the triad of hyperinsulinemia, acanthosis nigricans (skin lesions associated with insulin resistance), and signs of hyperandrogenism in females without lipodystrophy and who are not overweight.

Your genetic map

Gene	SNP	Genotype
INSR	rs121913148	CC
INSR	rs121913156	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2297

Hereditary Diseases (genetics)

Retinitis pigmentosa-juvenile cataract-short stature-intellectual disability syndrome

A rare, genetic, syndromic rod-cone dystrophy disorder characterized by psychomotor developmental delay from early childhood, intellectual disability, short stature, mild facial dysmorphism (e.g. upslanted palpebral fissures, hypoplastic alae nasi, malar hypoplasia, attached earlobes), excessive dental spacing and malocclusion, juvenile cataract and ophthalmologic findings of atypical retinitis pigmentosa (i.e. salt-and-pepper retinopathy, attenuated retinal arterioles, generalized rod-cone dysfunction, mottled macula, peripapillary sparing of retinal pigment epithelium).

Your genetic map

Gene	SNP	Genotype
RDH11	rs606231423	GG
RDH11	rs606231424	GG

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=436245

Hereditary Diseases (genetics)

Growth and developmental delay-hypotonia-vision impairment-lactic acidosis syndrome

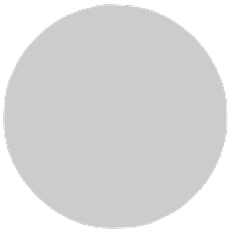
Growth and developmental delay-hypotonia-vision impairment-lactic acidosis syndrome is a rare, genetic, mitochondrial oxidative phosphorylation disorder characterized by intrauterine growth retardation, microcephaly, hypotonia, vision impairment, speech and language delay and lactic acidosis with reduced respiratory chain activity (typically complex I). Additonal features may include macrocytic anemia, tremor, muscular atrophy, dysmetria and mild intellectual disability.

Your genetic map

Gene	SNP	Genotype
SFXN4	rs756173225	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=391348

Hereditary Diseases (genetics)

Global developmental delay-neuro-ophthalmological abnormalities-seizures-intellectual disability syndrome

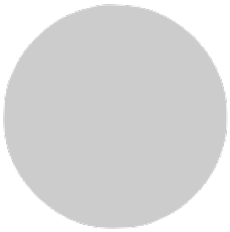
A rare genetic neurological disorder characterized by infantile to childhood onset of global developmental delay, hypotonia, seizures, growth delay, and intellectual disability. Additional variable features include strabismus, cortical visual impairment, nystagmus, movement disorder (such as dystonia, ataxia, or chorea), or mild dysmorphic features, among others.

Your genetic map

Gene	SNP	Genotype
GNB1	rs752746786	AA
GNB1	rs869312825	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=488613

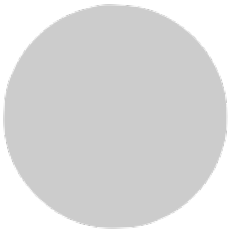
Hereditary Diseases (genetics)

Rett syndrome

A rare severe, X-linked, neurodevelopmental disorder characterized by rapid developmental regression in infancy, partial or complete loss of purposeful hand movements, loss of speech, gait abnormalities, and stereotypic hand movements, commonly associated with deceleration of head growth, severe intellectual disability, seizures, and breathing abnormalities. The disorder has a progressive clinical course and may associate various comorbidities including gastrointestinal diseases, scoliosis, and behavioral disorders.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=778

Your genetic map

Gene	SNP	Genotype
MECP2	rs28934904	GG
MECP2	rs28934906	GG
MECP2	rs28934907	GG
MECP2	rs61750240	GG
MECP2	rs61751362	GG
MECP2	rs28935468	GG
MECP2	rs61748421	GG
MECP2	rs61749721	GG
MECP2	rs28935168	GG
MECP2	rs61749715	GG
MECP2	rs179363901	GG
MECP2	rs193922679	TT
MECP2	rs61748408	GG
MECP2	rs61749724	GG
MECP2	rs61749747	GG
MECP2	rs61752372	GG
MECP2	rs61753965	GG
MECP2	rs61753979	GG
MECP2	rs61754432	GG
MECP2	rs61754437	GG
MECP2	rs61754452	GG
MECP2	rs61754455	CC
MECP2	rs61754457	CC
MECP2	rs267608455	CC
MECP2	rs61755763	CC
MECP2	rs61748389	CC
MECP2	rs61748390	GG
MECP2	rs61748391	TT
MECP2	rs61748395	TT
MECP2	rs61748404	GG
MECP2	rs61748407	TT

Hereditary Diseases (genetics)

Autosomal dominant Robinow syndrome

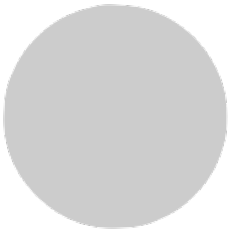
The more common type of Robinow syndrome (RS) characterized by mild to moderate limb shortening and abnormalities of the head, face and external genitalia.

Your genetic map

Gene	SNP	Genotype
DVL3	rs869025216	AA
DVL3	rs869025217	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3107

Hereditary Diseases (genetics)

Rothmund-Thomson syndrome

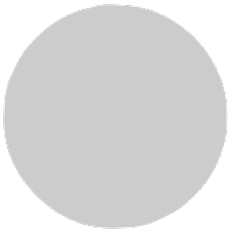
Rothmund-Thomson syndrome (RTS) is a genodermatosis presenting with a characteristic facial rash (poikiloderma) associated with short stature due to pre- and postnatal growth delay, sparse scalp hair, sparse or absent eyelashes and/or eyebrows, juvenile cataracts, skeletal abnormalities, radial ray defects, premature aging and a predisposition to certain cancers.

Your genetic map

Gene	SNP	Genotype
RECQL4	rs137853229	GG
RECQL4	rs117642173	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2909

Hereditary Diseases (genetics)

Rotor syndrome

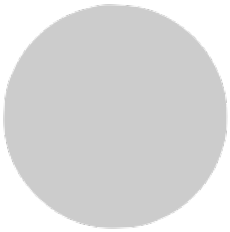
A benign, inherited liver disorder characterized by chronic, predominantly conjugated, nonhemolytic hyperbilirubinemia with normal liver histology.

Your genetic map

Gene	SNP	Genotype
SLCO1B1	rs183501729	CC
SLCO1B3	rs201833947	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3111

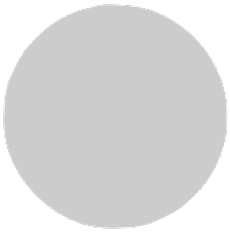
Hereditary Diseases (genetics)

Rubinstein-Taybi syndrome

A rare, genetic malformation syndrome characterized by congenital anomalies (microcephaly, specific facial characteristics, and broad thumbs and halluces), short stature, intellectual disability and behavioral characteristics.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=783

Your genetic map

Gene	SNP	Genotype
CREBBP	rs267606752	CC
CREBBP	rs398124145	CC
CREBBP	rs587783510	GG
CREBBP	rs587783505	GG
CREBBP	rs587783503	AA
CREBBP	rs587783497	TT
CREBBP	rs587783496	TT
CREBBP	rs147688139	AA
CREBBP	rs587783494	TT
CREBBP	rs587783493	GG
CREBBP	rs587783492	AA
CREBBP	rs587783491	CC
CREBBP	rs587783490	GG
CREBBP	rs587783489	GG
CREBBP	rs587783488	CC
CREBBP	rs587783486	TT
CREBBP	rs200782888	CC
CREBBP	rs587783483	CC
CREBBP	rs587783482	CC
CREBBP	rs587783480	CC
CREBBP	rs587783479	GG
CREBBP	rs587783475	GG
CREBBP	rs587783471	GG
CREBBP	rs587783464	GG
CREBBP	rs587783463	CC
CREBBP	rs587783461	GG
CREBBP	rs587783460	GG
CREBBP	rs587783516	GG
CREBBP	rs587783509	GG
CREBBP	rs587783478	GG
CREBBP	rs587783476	GG

Hereditary Diseases (genetics)

Schinzel-Giedion syndrome

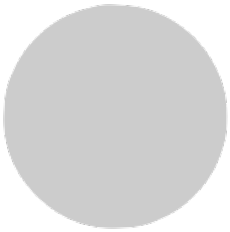
Schinzel-Giedion syndrome (SGS) is an ectodermal dysplasia syndrome chiefly characterized by a distinctive facial dysmorphism, hydronephrosis, severe developmental delay, typical skeletal malformations, and genital and cardiac anomalies.

Your genetic map

Gene	SNP	Genotype
SETBP1	rs267607042	GG
SETBP1	rs267607040	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=798

Hereditary Diseases (genetics)

Scott syndrome

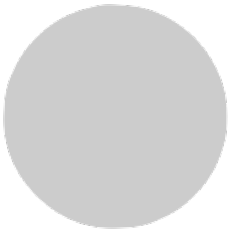
Scott syndrome is an extremely rare congenital hemorrhagic disorder characterized by hemorrhagic episodes due to impaired platelet coagulant activity.

Your genetic map

Gene	SNP	Genotype
LOC1053	rs374664255	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=806

Hereditary Diseases (genetics)

Senior-Boichis syndrome

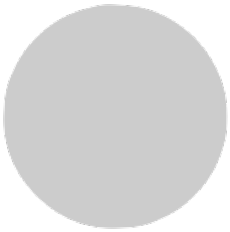
A rare ciliopathy characterized by the association of nephronophthisis and liver fibrosis. Renal manifestations include chronic renal failure, polyuria, polydipsia, anemia, as well as increased echogenicity on renal ultrasound and interstitial fibrosis and tubular dilation on biopsy. Hepatic involvement manifests as hepatosplenomegaly with extensive fibrosis, destruction of the bile ducts, and cholestasis. Mild psychomotor retardation and ocular symptoms, such as strabismus, nystagmus, retinal degeneration, and anisocoria, have been reported in some patients.

Your genetic map

Gene	SNP	Genotype
DCDC2	rs760040426	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=84081

Hereditary Diseases (genetics)

Sheldon-Hall syndrome

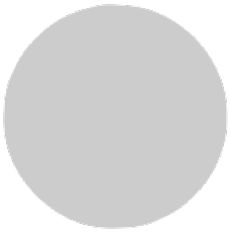
Sheldon-Hall syndrome (SHS) is a rare multiple congenital contracture syndrome characterized by contractures of the distal joints of the limbs, triangular face, downslanting palpebral fissures, small mouth, and high arched palate.

Your genetic map

Gene	SNP	Genotype
NALCN	rs786203988	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1147

Hereditary Diseases (genetics)

Shprintzen-Goldberg syndrome

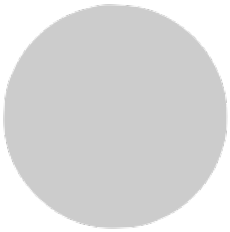
Shprintzen-Goldberg syndrome (SGS) is a very rare genetic disorder characterized by craniosynostosis, craniofacial and skeletal abnormalities, marfanoid habitus, cardiac anomalies, neurological abnormalities, and intellectual disability.

Your genetic map

Gene	SNP	Genotype
SKI	rs387907303	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2462

Hereditary Diseases (genetics)

Shwachman-Diamond syndrome

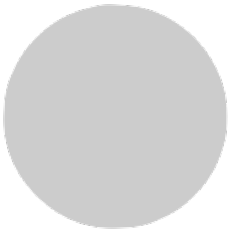
Shwachman-Diamond syndrome (SDS) is a rare multisystemic syndrome characterized by chronic and usually mild neutropenia, pancreatic exocrine insufficiency associated with steatorrhea and growth failure, skeletal dysplasia with short stature, and an increased risk of bone marrow aplasia or leukemic transformation.

Your genetic map

Gene	SNP	Genotype
SBDS	rs113993992	CC
TYW1	rs373730800	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=811

Hereditary Diseases (genetics)

Simpson-Golabi-Behmel syndrome

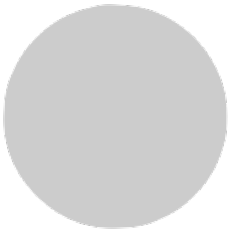
A rare X-linked multiple congenital anomalies syndrome characterized by pre- and postnatal overgrowth, distinctive craniofacial features, variable congenital malformations, organomegaly and an increased tumor risk.

Your genetic map

Gene	SNP	Genotype
GPC3	rs122453121	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=373

Hereditary Diseases (genetics)

Sjögren Larsson syndrome

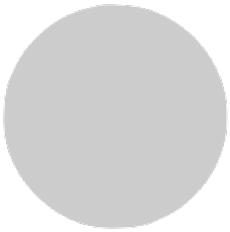
A rare neurocutaneous disorder caused by an inborn error of lipid metabolism and characterized by congenital ichthyosis, intellectual deficit, and spasticity.

Your genetic map

Gene	SNP	Genotype
ALDH3A2	rs72547571	CC
ALDH3A2	rs72547569	GG
ALDH3A2	rs72547575	AA
ALDH3A2	rs72547562	CC
ALDH3A2	rs72547561	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=816

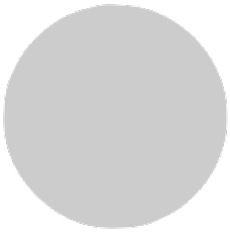
Hereditary Diseases (genetics)

Smith-Lemli-Opitz syndrome

Smith-Lemli-Opitz syndrome (SLOS) is characterized by multiple congenital anomalies, intellectual deficit, and behavioral problems.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=818

Your genetic map

Gene	SNP	Genotype
DHCR7	rs28938174	TT
DHCR7	rs121909764	CC
DHCR7	rs80338853	GG
DHCR7	rs80338860	GG
DHCR7	rs61757582	GG
DHCR7	rs121909765	GG
DHCR7	rs121909767	CC
DHCR7	rs80338864	CC
DHCR7	rs104886033	TT
DHCR7	rs121909768	CC
DHCR7	rs80338862	CC
DHCR7	rs11555217	CC
DHCR7	rs80338856	GG
DHCR7	rs80338857	CC
DHCR7	rs80338858	GG
DHCR7	rs104886035	GG
DHCR7	rs398123607	CC
DHCR7	rs143312232	GG
DHCR7	rs779709646	CC
DHCR7	rs104886039	GG
DHCR7	rs751604696	CC
DHCR7	rs753960624	AA
DHCR7	rs121912195	AA

Hereditary Diseases (genetics)

Steel syndrome

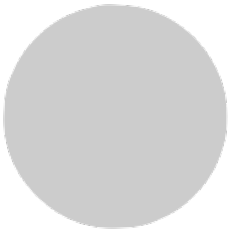
A rare genetic bone disease characterized by short stature, bilateral congenital hip dislocation, radial head dislocation, carpal coalition, scoliosis, pes cavus, and atlantoaxial subluxation. Dysmorphic facial features include broad forehead, broad nasal bridge, hypertelorism, and mild midface hypoplasia. Association with bilateral sensorineural hearing loss has also been described.

Your genetic map

Gene	SNP	Genotype
COL27A1	rs140950220	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=438117

Hereditary Diseases (genetics)

Stickler syndrome

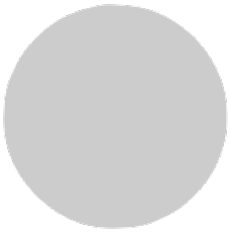
A rare group of genetic connective tissue disorders characterized by ophthalmic, auditory, orofacial and articular manifestations. The two main clinical forms are clinically distinguished by the vitreous phenotype; stickler type 1 by a vestigial vitreous gel in the immediate retrolental space, bordered by a distinct folded membrane, and Stickler type 2 by sparse and irregularly thickened bundles of 64257;bers throughout the vitreous cavity.

Your genetic map

Gene	SNP	Genotype
COL2A1	rs121912884	GG
COL2A1	rs121912893	GG
COL2A1	rs748459670	GG
LOC1053	rs121912866	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=828

Hereditary Diseases (genetics)

Short stature-brachydactyly-obesity-global developmental delay syndrome

A rare genetic, multiple congenital anomalies syndrome characterized by short stature, hand brachydactyly with hypoplastic distal phalanges, global development delay, intellectual disability, and more variably seizures, obesity, and craniofacial dysmorphism that includes microcephaly, high forehead, flat face, hypertelorism, deep set eyes, flat nasal bridge, averted nostrils, long philtrum, thin lip vermilion, and short neck.

Your genetic map

Gene	SNP	Genotype
PRMT7	rs201824659	GG

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=464288

Hereditary Diseases (genetics)

Short stature-pituitary and cerebellar defects-small sella turcica syndrome

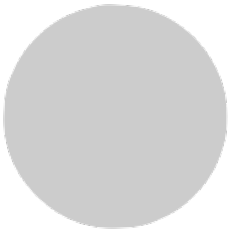
Short stature-pituitary and cerebellar defects-small sella turcica syndrome is characterised by short stature, anterior pituitary hormone deficiency, small sella turcica, and a hypoplastic anterior hypophysis associated with pointed cerebellar tonsils. It has been described in three generations of a large French kindred. Ectopia of the posterior hypophysis was observed in some patients. The syndrome is transmitted as a dominantly inherited trait and is caused by a germline mutation within the LIM-homeobox transcription factor LHX4 gene (1q25).

Your genetic map

Gene	SNP	Genotype
LHX4 AS1	rs786204780	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=85442

Hereditary Diseases (genetics)

Tatton-Brown-Rahman syndrome

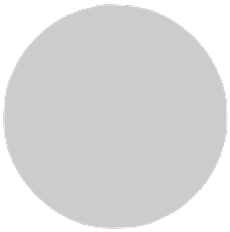
A rare multiple congenital anomalies syndrome characterized by greater hight, mild to moderate intellectual disability and distinctive facial appereance like round face, heavy, horizontal eyebrows and narrow palpebral fissures.

Your genetic map

Gene	SNP	Genotype
DNMT3A	rs779626155	GG
DNMT3A	rs778270132	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=404443

Hereditary Diseases (genetics)

Spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome

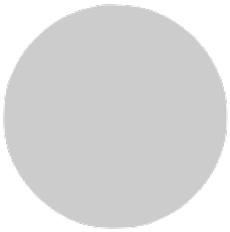
A rare neurometabolic disorder due to serine deficiency characterized by neonatal to infantile onset of global developmental delay, postnatal microcephaly and intellectual disability, which may be associated with slowly progressive spastic tetraplegia mainly affecting the lower extremities, seizures, and brain MRI findings including thin corpus callosum, delayed myelination and cerebral atrophy. Additional symptoms include brisk deep tendon reflexes, extensor plantar responses, behavioral abnormalities (such as irritability, hyperactivity, sleep disorder), abnormal hand movements and stereotypy.

Your genetic map

Gene	SNP	Genotype
LOC1053	rs201278558	GG
LOC1053	rs761533681	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=447997

Hereditary Diseases (genetics)

Toriello-Lacassie-Droste syndrome

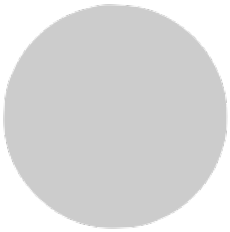
Oculo-ectodermal syndrome (OES) is characterized by the association of epibulbar dermoids and aplasia cutis congenital.

Your genetic map

Gene	SNP	Genotype
CLUAP1	rs751218423	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3339

Hereditary Diseases (genetics)

Arterial tortuosity syndrome

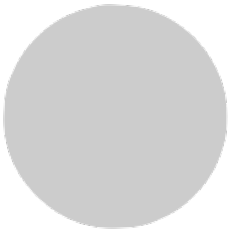
A rare autosomal recessive connective tissue disorder characterized by tortuosity and elongation of the large and medium-sized arteries and a propensity towards aneurysm formation, vascular dissection, and stenosis of the pulmonary arteries.

Your genetic map

Gene	SNP	Genotype
SLC2A10	rs121908172	GG
SLC2A10	rs756457861	CC
SLC2A10	rs761721442	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3342

Hereditary Diseases (genetics)

Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-skeletal anomalies syndrome

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by global developmental delay, intellectual disability, hypotonia, craniofacial dysmorphism (such as ridged metopic sutures, long palpebral fissures, broad nasal bridge, hypoplastic alae nasi, low-set, prominent ears, prominent midline tongue groove, and downturned mouth), congenital heart defects, and variable skeletal abnormalities including hip dysplasia, vertebral anomalies, and scoliosis. Additional reported manifestations include high pain tolerance and genitourinary anomalies. Brain imaging may show a thin corpus callosum or white matter abnormalities.

Your genetic map

Gene	SNP	Genotype
HNRNPK	rs863223403	CC

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=453499

Hereditary Diseases (genetics)

Noonan syndrome-like disorder with loose anagen hair

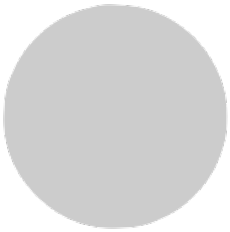
A Noonan-related syndrome, characterized by facial anomalies suggestive of Noonan syndrome, loose anagen hair, frequent congenital heart defects, distinctive skin features (darkly pigmented skin, keratosis pilaris, eczema or ichtyosis), and short stature that is often associated with a growth hormone deficiency. Psychomotor delay with attention deficit/hyperactivity disorder (ADHD) is frequently observed.

Your genetic map

Gene	SNP	Genotype
SHOC2	rs267607048	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2701

Hereditary Diseases (genetics)

Renal tubulopathy-encephalopathy-liver failure syndrome

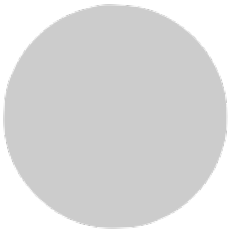
Renal tubulopathy - encephalopathy - liver failure describes a spectrum of phenotypes with manifestations similar but milder than those seen in GRACILE syndrome and that can be associated with encephalopathy and psychiatric disorders.

Your genetic map

Gene	SNP	Genotype
BCS1L	rs121908575	CC
ZNF142	rs121908576	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=254902

Hereditary Diseases (genetics)

Vici syndrome

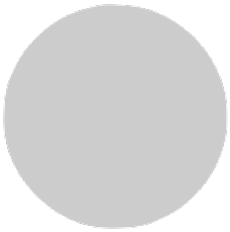
Vici syndrome is a very rare and severe congenital multisystem disorder characterized by the principal features of agenesis of the corpus callosum, cataracts, oculocutaneous hypopigmentation, cardiomyopathy and combined immunodeficiency.

Your genetic map

Gene	SNP	Genotype
EPG5	rs587776942	GG
EPG5	rs201757275	TT
EPG5	rs183478189	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1493

Hereditary Diseases (genetics)

Wiedemann-Rautenstrauch syndrome

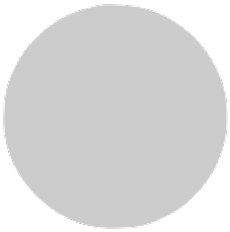
A rare multiple congenital anomalies/dysmorphic syndrome characterized by marked prenatal and postnatal growth retardation, decreased subcutaneous fat, hypotrichosis, relative macrocephaly and an unusual face. Mild to moderate intellectual disability is common.

Your genetic map

Gene	SNP	Genotype
POLR3A	rs148932047	GG
POLR3A	rs141659018	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3455

Hereditary Diseases (genetics)

Wiedemann-Steiner syndrome

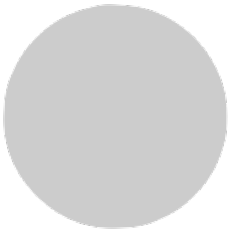
A rare, genetic multiple congenital anomalies/dysmorphic syndrome characterized by short stature, hypertrichosis (most commonly of the back or elbow regions), facial dysmorphism, behavioral problems, developmental delay and, most commonly, mild to moderate intellectual disability.

Your genetic map

Gene	SNP	Genotype
KMT2A	rs587783678	CC
KMT2A	rs587783679	GG
KMT2A	rs587783680	CC
KMT2A	rs797045051	CC
KMT2A	rs863224889	GG
KMT2A	rs886041856	CC
TTC36	rs782477344	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=319182

Hereditary Diseases (genetics)

Wiskott-Aldrich syndrome

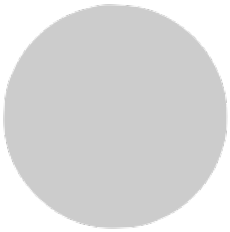
A primary immunodeficiency disease characterized by microthrombocytopenia, eczema, infections and an increased risk for autoimmune manifestations and malignancies.

Your genetic map

Gene	SNP	Genotype
WAS	rs132630268	GG
WAS	rs193922414	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=906

Hereditary Diseases (genetics)

Wolcott-Rallison syndrome

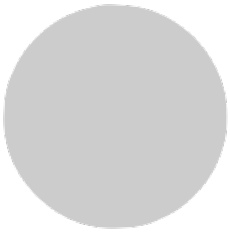
Wolcott-Rallison syndrome (WRS) is a very rare genetic disease, characterized by permanent neonatal diabetes mellitus (PNDM) with multiple epiphyseal dysplasia and other clinical manifestations, including recurrent episodes of acute liver failure.

Your genetic map

Gene	SNP	Genotype
EIF2AK3	rs864621972	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1667

Hereditary Diseases (genetics)

Wolfram syndrome

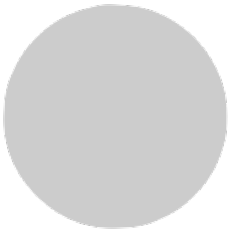
A rare, genetic, endocrine disorder characterized by type I diabetes mellitus (DM), diabetes insipidus (DI), sensorineural deafness (D), bilateral optical atrophy (OA) and neurological signs.

Your genetic map

Gene	SNP	Genotype
WFS1	rs28937892	CC
WFS1	rs387906930	CC
WFS1	rs71530923	CC
WFS1	rs797045075	TT
WFS1	rs777580652	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3463

Hereditary Diseases (genetics)

Carney complex-trismus-pseudocamptodactyly syndrome

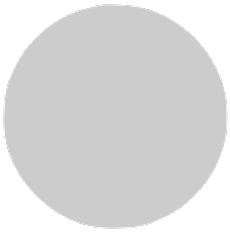
Carney complex-trismus-pseudocamptodactyly syndrome is a rare genetic heart-hand syndrome characterized by typical manifestations of the Carney complex (spotty pigmentation of the skin, familial cardiac and cutaneous myxomas and endocrinopathy) associated with trismus and distal arthrogryposis (presenting as involuntary contraction of distal and proximal interphalangeal joints of hands evident only on dorsiflexion of wrist and similar lower-limb contractures producing foot deformities).

Your genetic map

Gene	SNP	Genotype
MYHAS	rs121434590	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=319340

Hereditary Diseases (genetics)

Isolated cloverleaf skull syndrome

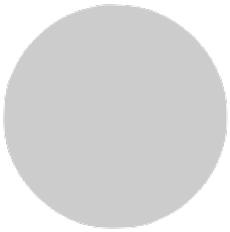
A form of craniosynostosis involving multiple sutures (coronal, lambdoidal, sagittal and metopic) characterized by a trilobular skull of varying severity (frontal towering and bossing, temporal bulging and a flat posterior skull), dysmorphic features (downslanting palpebral fissures, midface hypoplasia, and extreme proptosis) and that is complicated by hydrocephalus, cerebral venous hypertension, developmental delay/intellectual disability and hind brain herniation.

Your genetic map

Gene	SNP	Genotype
ERF	rs587777008	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2343

Hereditary Diseases (genetics)

Occipital horn syndrome

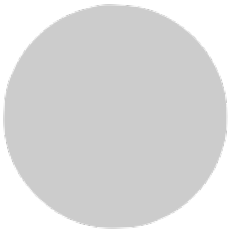
A rare congenital disorder of copper metabolism that is principally characterized by bony exostoses (including the pathognomonic occipital horns), and connective tissue manifestations with cutis laxa and bladder diverticula. Central nervous system involvement is variable.

Your genetic map

Gene	SNP	Genotype
ATP7A	rs151340631	CC
ATP7A	rs151340632	AA
ATP7A	rs797045340	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=198

Hereditary Diseases (genetics)

Lateral meningocele syndrome

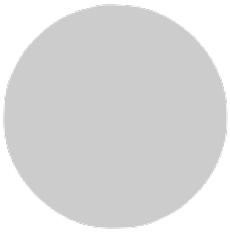
A rare genetic neurological disorder characterized by multiple lateral meningoceles, distinctive facial dysmorphism (including hypertelorism, downslanting palpebral fissures, posteriorly rotated ears, micrognathia, and high, narrow palate, among others), and skeletal abnormalities (e. g. vertebral anomalies, wormian bones, short stature, and scoliosis). Multiple additional features may present, such as conductive hearing impairment, hypotonia, and connective tissue and urogenital abnormalities. Cognition is usually normal.

Your genetic map

Gene	SNP	Genotype
NOTCH3	rs869312910	GG
NOTCH3	rs869312911	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2789

Hereditary Diseases (genetics)

Linear nevus sebaceus syndrome

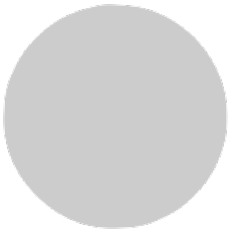
A rare nevus syndrome characterized by the association of an nevus sebaceous with a broad spectrum of abnormalities that affect many organ systems, most commonly the eye, skeletal and central nervous system.

Your genetic map

Gene	SNP	Genotype
LRRC56	rs121913233	TT
LRRC56	rs104894228	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2612

Hereditary Diseases (genetics)

EEC syndrome

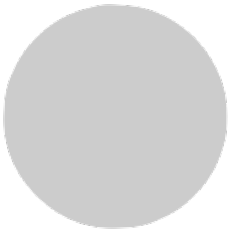
EEC syndrome is a genetic developmental disorder characterized by ectrodactyly, ectodermal dysplasia, and orofacial clefts (cleft lip/palate).

Your genetic map

Gene	SNP	Genotype
TP63	rs121908835	CC
TP63	rs121908841	GG
TP63	rs121908844	AA
TP63	rs121908849	GG
TP63	rs864621968	AA
TP63	rs797044484	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1896

Hereditary Diseases (genetics)

Neurogenic scapuloperoneal syndrome, Kaeser type

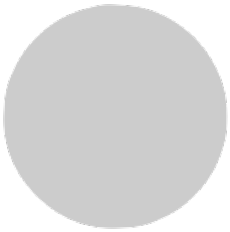
A rare, genetic, neuromuscular disease characterized by adult-onset muscle weakness and atrophy in a scapuloperoneal distribution, mild involvement of the facial muscles, dysphagia, and gynecomastia. Elevated serum CK levels and mixed myopathic and neurogenic abnormalities are associated clinical findings.

Your genetic map

Gene	SNP	Genotype
DES	rs57965306	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=85146

Hereditary Diseases (genetics)

Enamel-renal syndrome

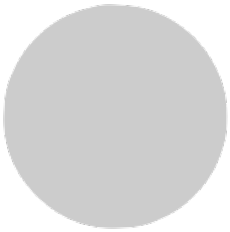
A extremely rare, genetic malformation syndrome characterized by hypoplastic amelogenesis imperfecta (hypoplastic dental enamel) and nephrocalcinosis (precipitation of calcium salts in renal tissue). Oral manifestations include yellow and misshaped teeth, delayed tooth eruption, and intrapulpal calcifications. Nephrocalcinosis is often asymptomatic but can progress during late childhood or early adulthood to impaired renal function, recurrent urinary infections, renal tubular acidosis, and rarely to end-stage renal failure.

Your genetic map

Gene	SNP	Genotype
FAM20A	rs144411158	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1031

Hereditary Diseases (genetics)

Familial hyperphosphatemic tumoral calcinosis/Hyperphosphatemic hyperostosis syndrome

Familial tumoral calcinosis (FTC) refers to a rare autosomal recessive disorder characterized by the occurrence of cutaneous and subcutaneous calcified masses, usually adjacent to large joints, such as hips, shoulders and elbows. FTC can occur in the setting of hyperphosphatemia or normophosphatemia, depending on the type of gene mutation involved.

Your genetic map

Gene	SNP	Genotype
GALNT3	rs137853086	GG

Multivariate analysis

What do your genetics tell us?

We have not detected any pathogenic mutations, but, since we only analyse a part of the gene, you could have a pathogenic mutation in non-analysed genetic regions.

More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=306661

Hereditary Diseases (genetics)

H syndrome

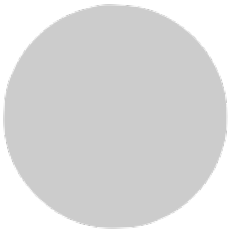
A rare cutaneous disease and a systemic inherited histiocytosis mainly characterized by hyperpigmentation, hypertrichosis, hepatosplenomegaly, heart anomalies, hearing loss, hypogonadism, low height, and occasionally, hyperglycemia/diabetes mellitus. Due to overlapping clinical features, it is now considered to include pigmented hypertrichosis with insulin dependent diabetes mellitus syndrome (PHID), Faisalabad histiocytosis (FHC) and familial sinus histiocytosis with massive lymphadenopathy (FSHML). Some cases of dysosteosclerosis may also represent the syndrome.

Your genetic map

Gene	SNP	Genotype
SLC29A3	rs121912583	GG
SLC29A3	rs267607056	GG
SLC29A3	rs121912584	GG
SLC29A3	rs387907066	GG
SLC29A3	rs387907067	CC
SLC29A3	rs587780462	CC
SLC29A3	rs587780463	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=168569

Hereditary Diseases (genetics)

Atypical hemolytic uremic syndrome

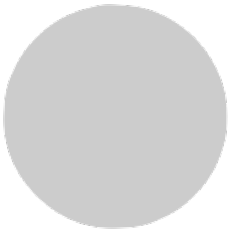
A rare, genetic thrombotic microangiopathy due to dysregulation of the alternative complement pathway and characterized by the triad of hemolytic anemia, thrombocytopenia, and acute renal dysfunction.

Your genetic map

Gene	SNP	Genotype
DGKE	rs138924661	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2134

Hereditary Diseases (genetics)

Hydrolethalus

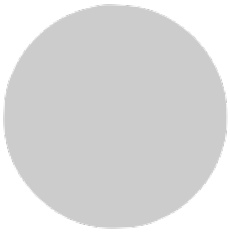
Hydrolethalus (HLS) is a severe fetal malformation syndrome characterized by craniofacial dysmorphic features, central nervous system, cardiac, respiratory tract and limb abnormalities.

Your genetic map

Gene	SNP	Genotype
HYLS1	rs104894232	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2189

Hereditary Diseases (genetics)

KID syndrome

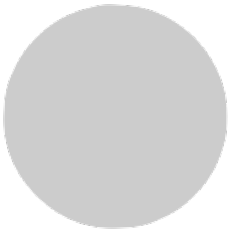
A rare congenital ectodermal disorder characterized by vascularizing keratitis, hyperkeratotic skin lesions and hearing loss.

Your genetic map

Gene	SNP	Genotype
GJB2	rs28931594	CC
GJB2	rs72561723	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=477

Hereditary Diseases (genetics)

Lacrimoauriculodentodigital syndrome

A rare, genetic, multiple congenital anomalies/dysmorphic syndrome characterized by hypoplasia, aplasia or atresia of the lacrimal system, anomalies of the ears with sensorineural or mixed hearing loss, hypoplasia, aplasia or atresia of the salivary glands, dental anomalies, and digital malformations. Patients present obstruction of the nasal lacrimal ducts that can lead to epiphora, and chronic conjunctivitis due to alacrimia. Aplasia or hypoplasia of the salivary glands lead to dry mouth and early onset of severe dental caries. Dental features include late tooth eruption, small and peg-shaped lateral maxillary incisors and mild enamel dysplasia. The digital features are variable and include fifth finger clinodactyly, duplication of the distal phalanx of the thumb, triphalangeal thumb, and/or syndactyly. Unilateral radial aplasia and radial-ulnar synostosis have also been reported in association.

Your genetic map

Gene	SNP	Genotype
FGFR2	rs121918509	CC

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2363

Hereditary Diseases (genetics)

MASA syndrome

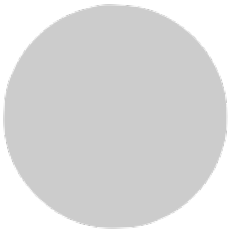
A X-linked, clinical subtype of L1 syndrome, characterized by mild to moderate intellectual disability, delayed development of speech, hypotonia progressing to spasticity or spastic paraplegia, adducted thumbs, and mild to moderate distension of the cerebral ventricles.

Your genetic map

Gene	SNP	Genotype
FA2H	rs765086319	GG
L1CAM	rs137852524	CC
SPG7	rs562890289	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2466

Hereditary Diseases (genetics)

MEGDEL syndrome

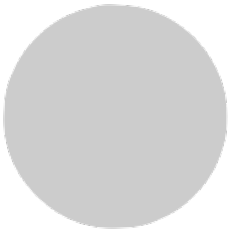
MEGDEL syndrome is a rare, genetic, neurometabolic disorder characterized by neonatal hypoglycemia, features of sepsis that are not linked to infection, development of feeding problems, failure to thrive, transient liver dysfunction, and truncal hypotonia followed by dystonia and spasticity which results in psychomotor development arrest and/or regression. Progressive sensorineural deafness, intellectual disability and absent speech are also associated. Laboratory tests demonstrate 3-methylglutaconic aciduria and temporary elevated serum lactate and transaminases.

Your genetic map

Gene	SNP	Genotype
SERAC1	rs387907236	GG
SERAC1	rs199632531	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=352328

Hereditary Diseases (genetics)

Micro syndrome

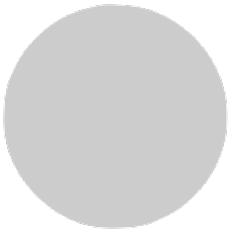
Micro syndrome is an autosomal recessive disorder caracterised by ocular and neurodevelopmental defects and by microgenitalia. It presents with severe intellectual disability, microcephaly, congenital cataract, microcornea, microphthalmia, agenesis/hypoplasia of the corpus callosum, and hypogenitalism.

Your genetic map

Gene	SNP	Genotype
RAB3GAP	rs532964185	CC
ZRANB3	rs797045905	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2510

Hereditary Diseases (genetics)

Multisystemic smooth muscle dysfunction syndrome

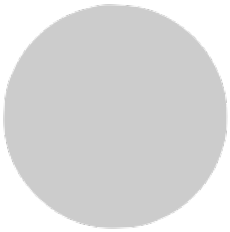
Multisystemic smooth muscle dysfunction syndrome is a rare, genetic, vascular disease characterized by congenital dysfunction of smooth muscle throughout the body, manifesting with cerebrovascular disease, aortic anomalies, intestinal hypoperistalsis, hypotonic bladder, and pulmonary hypertension. Congenital mid-dilated pupils non-reactive to light associated with a large, persistent patent ductus arteriosus are characteristic hallmarks of the disease.

Your genetic map

Gene	SNP	Genotype
ACTA2	rs387906592	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=404463

Hereditary Diseases (genetics)

Nephrogenic syndrome of inappropriate antidiuresis

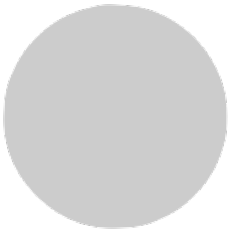
Nephrogenic syndrome of inappropriate antidiuresis (NSIAD) is a rare genetic disorder of water balance, closely resembling the far more frequent syndrome of inappropriate antidiuretic secretion (SIAD), and characterized by euvolemic hypotonic hyponatremia due to impaired free water excretion and undetectable or low plasma arginine vasopressin (AVP) levels.

Your genetic map

Gene	SNP	Genotype
AVPR2	rs104894761	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=93606

Hereditary Diseases (genetics)

Congenital nephrotic syndrome, Finnish type

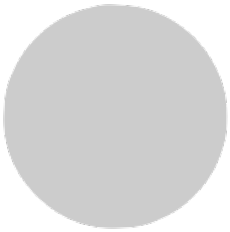
A rare congenital nephrotic syndrome characterized by massive protein loss and marked edema manifesting in utero or during the first 3 months of life.

Your genetic map

Gene	SNP	Genotype
KIRREL2	rs386833955	TT
NPHS1	rs137853042	GG
NPHS1	rs267606919	GG
NPHS1	rs386833865	GG
NPHS1	rs386833871	GG
NPHS1	rs386833874	GG
NPHS1	rs386833889	CC
NPHS1	rs386833895	CC
NPHS1	rs386833909	GG
NPHS1	rs386833915	GG
NPHS1	rs386833920	GG
NPHS1	rs749341977	GG
NPHS1	rs140018064	GG
NPHS1	rs142883811	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=839

Hereditary Diseases (genetics)

PRUNE1-related neurological syndrome

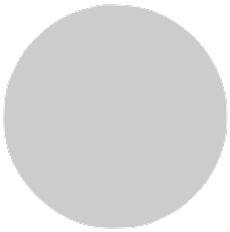
A rare genetic syndromic intellectual disability characterized by infantile onset of global developmental delay and profound intellectual disability in association with a heterogeneous spectrum of manifestations, such as features of lower motor neuron disease, hypotonia, spasticity, contractures, seizures, respiratory insufficiency, and optic atrophy, among others. Dysmorphic craniofacial features include microcephaly, tall forehead, bitemporal narrowing, flat nasal bridge, low-set ears, and high-arched palate. Brain imaging may show cerebral and cerebellar atrophy, delayed myelination, and thin corpus callosum.

Your genetic map

Gene	SNP	Genotype
PRUNE1	rs1057521927	GG
PRUNE1	rs767769359	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=544469

Hereditary Diseases (genetics)

Oculocerebrofacial syndrome, Kaufman type

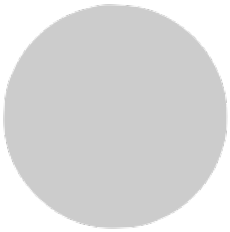
A rare, genetic, syndromic intellectual disability characterized by severe intellectual disability, distinctive craniofacial features and variable multiple congenital anomalies including ocular, brain, urogenital and skeletal abnormalities.

Your genetic map

Gene	SNP	Genotype
UBE3B	rs539407162	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2707

Hereditary Diseases (genetics)

Oculocerebrorenal syndrome of Lowe

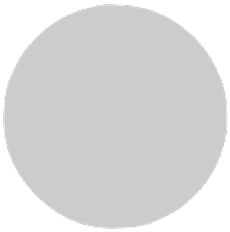
A rare multisystem disorder characterized by congenital cataracts, glaucoma, intellectual disabilities, seizures, postnatal growth retardation and renal tubular dysfunction with chronic renal failure.

Your genetic map

Gene	SNP	Genotype
OCRL	rs387906484	CC
OCRL	rs137853260	GG
OCRL	rs137853831	CC
OCRL	rs137853858	CC
OCRL	rs398123287	CC
OCRL	rs794727182	GG
OCRL	rs794727333	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=534

Hereditary Diseases (genetics)

Orofaciodigital syndrome type 14

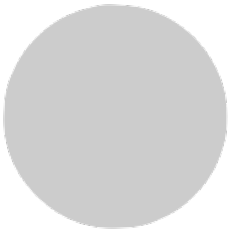
Orofaciodigital syndrome type 14 is a rare subtype of orofaciodigital syndrome, with autosomal recessive inheritance and C2CD3 mutations, characterized by severe microcephaly, trigonocephaly, severe intellectual disability and micropenis, in addition to oral, facial and digital malformations (gingival frenulae, lingual hamartomas, cleft/lobulated tongue, cleft palate, telecanthus, up-slanting palpebral fissures, microretrognathia, postaxial polydactyly of hands and duplication of hallux). Corpus callosum agenesis and vermis hypoplasia with molar tooth sign, on brain imaging, are also associated.

Your genetic map

Gene	SNP	Genotype
C2CD3	rs587777653	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=434179

Hereditary Diseases (genetics)

Orofaciodigital syndrome type 4

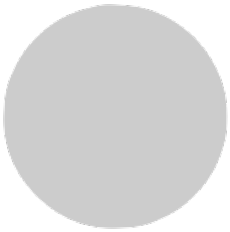
Oral-facial-digital syndrome, type 4 is characterized by lingual hamartoma, postaxial polysyndactyly of hands and feet, and mesomelic shortening of the legs with supinate equinovarus feet.

Your genetic map

Gene	SNP	Genotype
TCTN3	rs764091969	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2753

Hereditary Diseases (genetics)

Orofaciodigital syndrome type 5

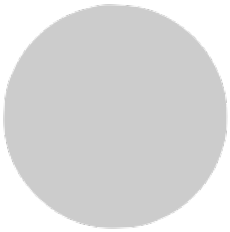
A rare orofaciodigital syndrome characterized by median cleft of the upper lip, postaxial polydactyly of hands and feet, and oral manifestations (duplicated frenulum).

Your genetic map

Gene	SNP	Genotype
DDX59	rs587777067	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2919

Hereditary Diseases (genetics)

Otopalatodigital syndrome type 2

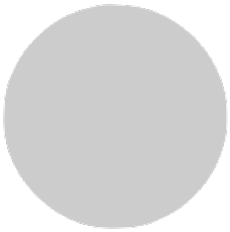
A severe form of otopalatodigital syndrome spectrum disorder, and is characterized by dysmorphic facies, severe skeletal dysplasia affecting the axial and appendicular skeleton, extraskeletal anomalies (including malformations of the brain, heart, genitourinary system, and intestine) and poor survival.

Your genetic map

Gene	SNP	Genotype
FLNA	rs28935470	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=90652

Hereditary Diseases (genetics)

Tumor necrosis factor receptor 1 associated periodic syndrome

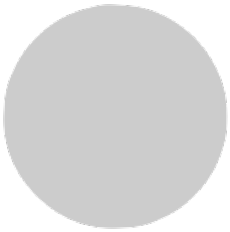
Tumor necrosis factor receptor 1 associated periodic syndrome (TRAPS) is a periodic fever syndrome, characterized by recurrent fever, arthralgia, myalgia and tender skin lesions lasting for 1 to 3 weeks, associated with skin, joint, ocular and serosal inflammation and complicated by secondary amyloidosis (see this term).

Your genetic map

Gene	SNP	Genotype
TNFRSF1	rs104895219	GG
TNFRSF1	rs104895217	AA
TNFRSF1	rs104895220	CC
TNFRSF1	rs104895223	CC
TNFRSF1	rs104895228	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=32960

Hereditary Diseases (genetics)

RAPADILINO syndrome

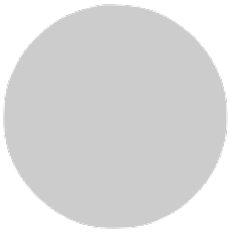
A rare syndrome for which the acronym indicates the principal signs: RA for radial ray defect, PA for both patellae hypoplasia or aplasia and cleft or highly arched palate, DI for diarrhea and dislocated joints, LI for little size and limb malformations, NO for long, slender nose and normal intelligence.

Your genetic map

Gene	SNP	Genotype
RECQL4	rs386833844	GG
RECQL4	rs386833851	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3021

Hereditary Diseases (genetics)

SHORT syndrome

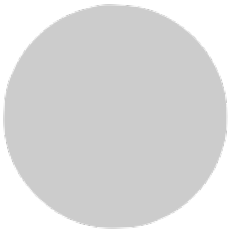
A rare disorder characterized by multiple congenital anomalies. The name is a mneumonic for the common features observed in SHORT syndrome that include; short stature, hyperextensibility of joints, ocular depression, Rieger anomaly and teething delay. Other common manifestations of SHORT syndrome are mild intrauterine growth restriction, partial lipodystrophy, delayed bone age, hernias and a recognizable facial gestalt.

Your genetic map

Gene	SNP	Genotype
PIK3R1	rs397515453	CC
PIK3R1	rs587784325	CC
PIK3R1	rs797045063	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3163

Hereditary Diseases (genetics)

Congenital intrauterine infection-like syndrome

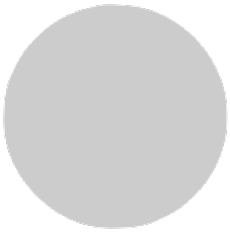
Congenital intrauterine infection-like syndrome is characterised by the presence of microcephaly and intracranial calcifications at birth accompanied by neurological delay, seizures and a clinical course similar to that seen in patients after intrauterine infection with Toxoplasma gondii, Rubella, Cytomegalovirus, Herpes simplex (so-called TORCH syndrome), or other agents, despite repeated tests revealing the absence of any known infectious agent.

Your genetic map

Gene	SNP	Genotype
OCLN	rs797045840	GG
OCLN	rs373915080	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1229

Hereditary Diseases (genetics)

NPHP3-related Meckel-like syndrome

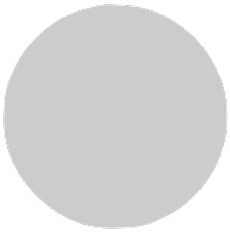
NPHP3-related Meckel-like syndrome is a rare, genetic, syndromic renal malformation characterized by cystic renal dysplasia with or without prenatal oligohydramnios, central nervous system abnormalities (commonly Dandy-Walker malformation), congenital hepatic fibrosis, and absence of polydactyly.

Your genetic map

Gene	SNP	Genotype
NPHP3	rs119456962	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3032

Hereditary Diseases (genetics)

Wolfram-like syndrome

Wolfram-like syndrome is a rare endocrine disease characterized by the triad of adult-onset diabetes mellitus, progressive hearing loss (usually presenting in the first decade of life and principally of low to moderate frequencies), and/or juvenile-onset optic atrophy. Psychiatric (i.e. anxiety, depression, hallucinations) and sleep disorders, the only neurologic abnormalities observed in this disease, have been reported in rare cases. Unlike Wolfram syndrome, patients with Wolfram-like syndrome do not report endocrine or cardiac findings.

Your genetic map

Gene	SNP	Genotype
LOC1079	rs74315205	GG
WFS1	rs201239579	GG
WFS1	rs71539673	GG

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=411590

Hereditary Diseases (genetics)

Larsen-like syndrome, B3GAT3 type

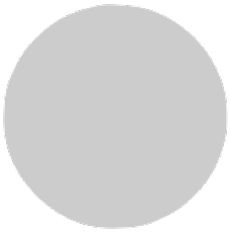
Larsen-like syndrome, B3GAT3 type is a rare, genetic, primary bone dysplasia characterized by laxity, dislocations and contractures of the joints, short stature, foot deformities (e.g. clubfeet), broad tips of fingers and toes, short neck, dysmorphic facial features (hypertelorism, downslanting palpebral fissures, upturned nose with anteverted nares, high arched palate) and various cardiac malformations. Severe disease is associated with multiple fractures, osteopenia, arachnodactyly and blue sclerae. A broad spectrum of additional features, including scoliosis, radio-ulnar synostosis, mild developmental delay, and various eye disorders (glaucoma, amblyopia, hyperopia, astigmatism, ptosis), are also reported.

Your genetic map

Gene	SNP	Genotype
B3GAT3	rs387906937	CC
B4GALT7	rs28937869	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=284139

Hereditary Diseases (genetics)

Triple A syndrome

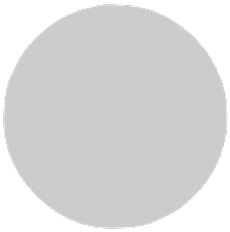
Triple A syndrome is a very rare multisystem disease characterized by adrenal insufficiency with isolated glucocorticoid deficiency, achalasia, alacrima, autonomic dysfunction and neurodegeneration.

Your genetic map

Gene	SNP	Genotype
AAAS	rs121918548	GG
AAAS	rs121918549	GG
AAAS	rs121918550	AA
AAAS	rs754637718	CC
AAAS	rs150511103	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=869

Hereditary Diseases (genetics)

Spondylocarpotarsal synostosis

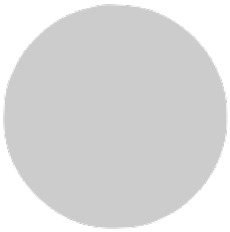
A spondylodysplasic dysplasia clinically characterized by postnatal progressive vertebral fusions frequently manifesting as block vertebrae, contributing to an shortened trunk and hence disproportionate short stature, scoliosis, lordosis, carpal and tarsal synostosis and infrequently, club feet.

Your genetic map

Gene	SNP	Genotype
FLNB	rs80356520	CC
FLNB	rs80356517	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3275

Hereditary Diseases (genetics)

Sitosterolemia

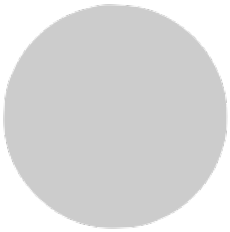
Sitosterolemia is a rare autosomal recessive sterol storage disease characterized by the accumulation of phytosterols in the blood and tissues. Clinical manifestations include xanthomas, arthralgia and premature atherosclerosis. Hematological manifestations include hemolytic anemia with stomatocytosis and macrothrombocytopenia. The disease is caused by homozygous or compound heterozygous mutations in ABCG5 (2p21) and ABCG8 (2p21) genes.

Your genetic map

Gene	SNP	Genotype
ABCG5	rs199689137	GG
ABCG8	rs137852987	GG
ABCG8	rs137852988	GG
ABCG8	rs137852991	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=2882

Hereditary Diseases (genetics)

Deafness with labyrinthine aplasia, microtia, and microdontia

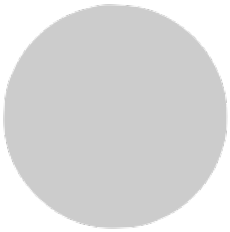
Deafness with labyrinthine aplasia, microtia, and microdontia (LAMM) is a genetic transmission deafness syndrome.

Your genetic map

Gene	SNP	Genotype
FGF3	rs281860303	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=90024

Hereditary Diseases (genetics)

Short stature due to GHSR deficiency

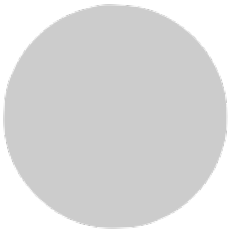
Short stature due to GHSR deficiency is a rare, genetic, endocrine growth disease, resulting from growth hormone secretagogue receptor (GHSR) deficiency, characterized by postnatal growth delay that results in short stature (less than -2 SD). The pituitary gland is typically without morphological changes, although anterior pituitary gland hypoplasia has been reported.

Your genetic map

Gene	SNP	Genotype
GHSR	rs121917883	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=314811

Hereditary Diseases (genetics)

Microcephalic cortical malformations-short stature due to RTTN deficiency

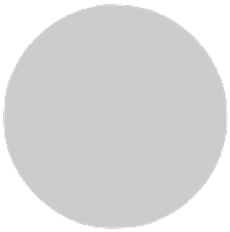
A rare, genetic, neurodevelopmental disorder with primordial microcephaly characterized by primary microcephaly, moderate to severe intellectual disability, and global developmental delay. Variable brain malformations are common ranging from simplified gyration, to cortical malformations such as pachygyria, polymicrogyria, reduced sulcation and midline defects. Craniofacial dysmorphism (e. g. sloping forehead, high and broad nasal bridge) are related to the primary microcephaly. Short stature is frequently observed, and may be severe.

Your genetic map

Gene	SNP	Genotype
RTTN	rs864321621	TT
RTTN	rs864321620	TT
RTTN	rs775277800	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=468631

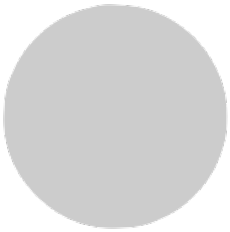
Hereditary Diseases (genetics)

Catecholaminergic polymorphic ventricular tachycardia

A rare, severe genetic arrhythmogenic disorder of the structurally normal heart characterized by catecholamine-induced ventricular tachycardia (VT) manifesting as syncope and sudden death in young individuals.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3286

Your genetic map

Gene	SNP	Genotype
CASQ2	rs139228801	GG
CASQ2	rs786205791	CC
RYR2	rs121918597	CC
RYR2	rs121918600	CC
RYR2	rs121918603	CC
RYR2	rs121918605	AA
RYR2	rs397516508	GG
RYR2	rs397516539	GG
RYR2	rs730880187	CC
RYR2	rs730880196	AA
RYR2	rs794728708	GG
RYR2	rs794728721	GG
RYR2	rs794728740	GG
RYR2	rs794728746	GG
RYR2	rs794728753	GG
RYR2	rs794728754	CC
RYR2	rs794728756	GG
RYR2	rs794728777	GG
RYR2	rs794728779	AA
RYR2	rs794728782	CC
RYR2	rs794728785	CC
RYR2	rs771994461	CC
RYR2	rs794728786	GG
RYR2	rs794728787	AA
RYR2	rs794728802	AA
RYR2	rs794728804	GG
RYR2	rs794728810	TT
RYR2	rs794728811	GG
RYR2	rs794728832	AA
RYR2	rs886037908	CC
RYR2	rs886037907	CC

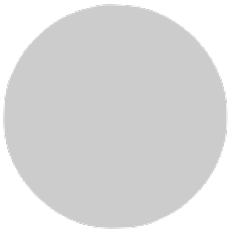
Hereditary Diseases (genetics)

Hereditary hemorrhagic telangiectasia

An inherited disorder of angiogenesis characterized by mucocutaneous telangiectases and visceral arteriovenous malformations.

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=774

Your genetic map

Gene	SNP	Genotype
ACVRL1	rs121909284	GG
ACVRL1	rs28936399	TT
ACVRL1	rs28936401	CC
ACVRL1	rs121909287	CC
ACVRL1	rs121909288	CC
ACVRL1	rs28936688	GG
ACVRL1	rs267606632	GG
ACVRL1	rs863223409	GG
ACVRL1	rs863223414	GG
ACVRL1	rs863223410	GG
ACVRL1	rs758683062	CC
ACVRL1	rs863223412	GG
ACVRL1	rs863223413	GG
ACVRL1	rs863223406	GG
ACVRL1	rs863223407	GG
ACVRL1	rs863223408	GG

Hereditary Diseases (genetics)

Tyrosinemia type 1

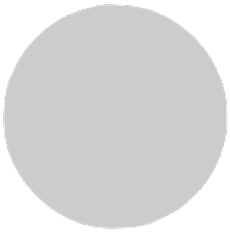
Tyrosinemia type 1 (HTI) is an inborn error of tyrosine catabolism caused by defective activity of fumarylacetoacetate hydrolase (FAH) and is characterized by progressive liver disease, renal tubular dysfunction, porphyria-like crises and a dramatic improvement in prognosis following treatment with nitisinone.

Your genetic map

Gene	SNP	Genotype
FAH	rs121965076	GG
FAH	rs80338900	GG
FAH	rs80338901	GG
FAH	rs121965075	GG
FAH	rs80338899	GG
FAH	rs80338895	GG
FAH	rs80338894	GG
FAH	rs80338898	CC
FAH	rs370686447	GG
FAH	rs149052294	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=882

Hereditary Diseases (genetics)

46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency

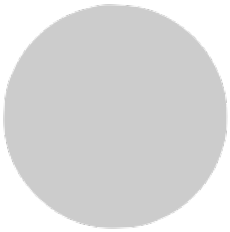
17-beta-hydroxysteroid dehydrogenase isozyme 3 (17betaHSD III) deficiency is a rare disorder leading to male pseudohermaphroditism (MPH), a condition characterized by incomplete differentiation of the male genitalia in 46X,Y males.

Your genetic map

Gene	SNP	Genotype
HSD17B3	rs119481077	GG
HSD17B3	rs119481079	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=752

Hereditary Diseases (genetics)

TELO2-related intellectual disability-neurodevelopmental disorder

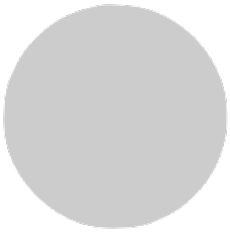
A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by global developmental delay and intellectual disability, infantile hypotonia, microcephaly, movement disorder, and impaired balance. More variable manifestations are hearing loss, cortical visual impairment, abnormalities of fingers and/or toes, congenital cardiac anomalies, kyphoscoliosis, dysmorphic facial features, abnormal sleep pattern, and seizures, among others.

Your genetic map

Gene	SNP	Genotype
TELO2	rs754162070	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=488642

Hereditary Diseases (genetics)

Lethal acantholytic erosive disorder

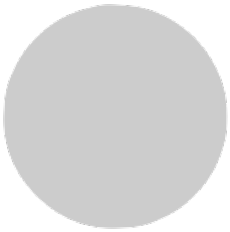
Lethal acantholytic epidermolysis bullosa is a suprabasal subtype of epidermolysis bullosa simplex (EBS, see this term) characterized by generalized oozing erosions, usually in the absence of blisters.

Your genetic map

Gene	SNP	Genotype
DSP	rs121912996	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=158687

Hereditary Diseases (genetics)

ITPA-related lethal infantile neurological disorder with cataract and cardiac involvement

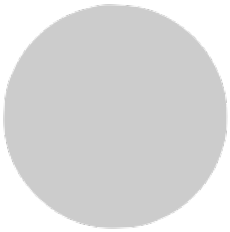
A rare, genetic, neurometabolic disease characterized by early onset encephalopathy with progressive microcephaly, severe global development delay, seizures, hypotonia, feeding difficulties, variable cardiac abnormalities, and cataracts. Brain MRI shows distinct pattern with high T2 signal and restricted diffusion in the posterior limb of the internal capsule in combination with delayed myelination and progressive cerebral atrophy. The disease is typically fatal.

Your genetic map

Gene	SNP	Genotype
ITPA	rs200086262	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=457375

Hereditary Diseases (genetics)

Familial progressive cardiac conduction defect

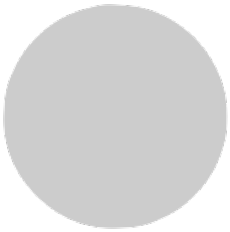
A genetic cardiac rhythm disease that may progress to complete atrioventricular (AV) block. The disease is either asymptomatic or manifests as dyspnea, dizziness, syncope, abdominal pain, heart failure or sudden death.

Your genetic map

Gene	SNP	Genotype
DSP	rs1135401735	AA
SCN5A	rs397514447	AA
SCN5A	rs137854607	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=871

Hereditary Diseases (genetics)

Noonan syndrome-like disorder with juvenile myelomonocytic leukemia

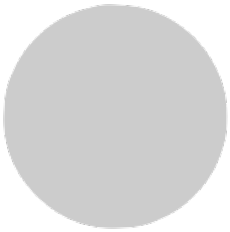
A rare, genetic, polymalformative syndrome characterized by a Noonan-like phenotype associated with increased risk of developing juvenile myelomonocytic leukemia (JMML). The Noonan-like (NS) phenotype includes dysmorphic facial features (i.e. high forehead, hypertelorism, downslanting palpebral fissures, ptosis, low-set ears, prominent philtrum and short neck with or without pterygium colli), developmental delay, hypotonia and small head circumference. It can be associated with congenital heart defects or cardiomyopathy, ectodermal anomalies, and short stature. The NS phenotype is subtle or even inapparent in a large proportion of subjects, but may occasionally be severe. Leukemia can be the only clinical manifestation of the syndrome.

Your genetic map

Gene	SNP	Genotype
CBL	rs267606706	TT
CBL	rs397507489	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=363972

Hereditary Diseases (genetics)

Nijmegen breakage syndrome-like disorder

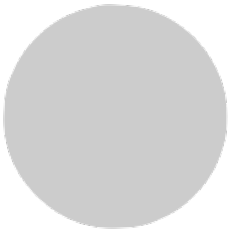
Nijmegen breakage syndrome-like disorder is a rare, genetic multiple congenital anomalies/dysmorphic syndrome characterized by growth retardation, short stature, developmental delay, intellectual disability, craniofacial dysmorphism (i.e. severe microcephaly, sloping forehead, prominent eyes, broad nasal ridge, hypoplastic nasal septum, epicanthal folds), spontaneous chromosomal instability, cellular hypersensitivity to ionizing radiation and radioresistant DNA synthesis, without severe infections, immunodeficiency or cancer predisposition. Additional reported features include mild spasticity, slight and nonprogressive ataxia, hyperopia, multiple pigmented nevi, widely spaced nipples, and clinodactyly.

Your genetic map

Gene	SNP	Genotype
RAD50	rs587780150	CC
RAD50	rs377260382	GG
RAD50	rs587781742	GG
RAD50	rs587781904	CC
RAD50	rs587782078	GG
RAD50	rs587782090	GG
RAD50	rs149201802	CC
TH2LCRR	rs750586158	CC
TH2LCRR	rs745797941	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=240760

Hereditary Diseases (genetics)

Carney triad

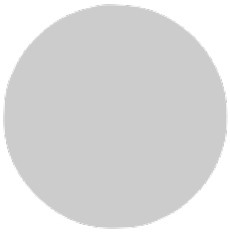
A rare non-hereditary condition characterized by gastrointestinal stromal tumors (GIST, intramural mesenchymal tumors of the gastrointestinal tract with neuronal or neural crest cell origin), pulmonary chondromas and extraadrenal paragangliomas.

Your genetic map

Gene	SNP	Genotype
SDHB	rs786201095	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=139411

Hereditary Diseases (genetics)

Severe primary trimethylaminuria

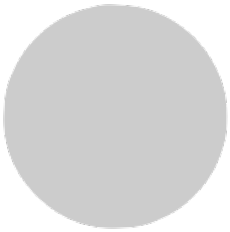
A rare inborn error of metabolism characterized by the presence of large amounts of trimethylamine in urine, sweat, and breath, resulting in a fishy body odor in affected individuals. While there are no additional signs and symptoms, the condition can have profound psychosocial consequences.

Your genetic map

Gene	SNP	Genotype
FMO3	rs61753344	GG
FMO3	rs72549326	CC
LOC1053	rs72549334	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=468726

Hereditary Diseases (genetics)

Glanzmann thrombasthenia

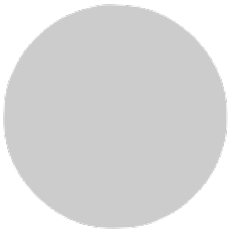
Glanzmann thrombasthenia (GT) is a bleeding syndrome characterized by spontaneous mucocutaneous bleeding and an exaggerated response to trauma due to a constitutional thrombocytopenia.

Your genetic map

Gene	SNP	Genotype
ITGB3	rs121918446	CC
ITGB3	rs121918452	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=849

Hereditary Diseases (genetics)

Congenital amegakaryocytic thrombocytopenia

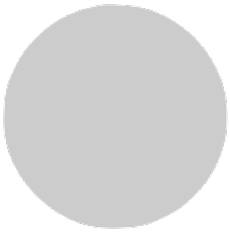
An isolated constitutional thrombocytopenia characterized by an isolated and severe decrease in the number of platelets and megakaryocytes during the first years of life that develops into bone marrow failure with pancytopenia later in childhood.

Your genetic map

Gene	SNP	Genotype
MPL	rs121913611	CC
MPL	rs28928907	GG
MPL	rs146249964	TT
MPL	rs148434485	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3319

Hereditary Diseases (genetics)

Paris-Trousseau thrombocytopenia

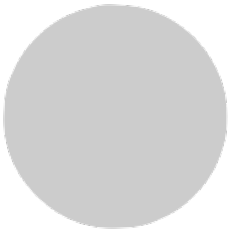
Paris-Trousseau thrombocytopenia (TCPT) is a contiguous gene syndrome characterized by mild bleeding tendency, variable thrombocytopenia (THC), dysmorphic facies, abnormal giant alpha-granules in platelets and dysmegakaryopoiesis.

Your genetic map

Gene	SNP	Genotype
FLI1	rs773148506	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=851

Hereditary Diseases (genetics)

Severe hereditary thrombophilia due to congenital protein C deficiency

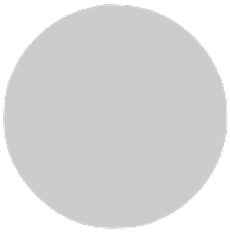
Congenital protein C deficiency is an inherited coagulation disorder characterized by deep venous thrombosis symptoms due to reduced synthesis and/or activity levels of protein C.

Your genetic map

Gene	SNP	Genotype
LOC1053	rs121918143	CC
LOC1053	rs121918150	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=745

Hereditary Diseases (genetics)

Hereditary thrombophilia due to congenital antithrombin deficiency

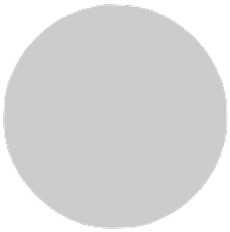
Hereditary thrombophilia due to congenital antithrombin deficiency is a rare, genetic, hematological disease characterized by decreased levels of antithrombin activity in plasma resulting in impaired inactivation of thrombin and factor Xa. Patients have an increased risk for venous thromboembolism, usually in the deep veins of the arms, legs and pulmonary system and, on occasion, in other venous territories (e.g. cerebral veins or sinus, mesenteric, portal, hepatic, renal and/or retinal veins).

Your genetic map

Gene	SNP	Genotype
SERPINC1	rs121909551	GG
SERPINC1	rs121909554	GG
SERPINC1	rs28929469	GG
SERPINC1	rs121909567	GG
SERPINC1	rs121909569	AA

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=82

Hereditary Diseases (genetics)

Desmoid tumor

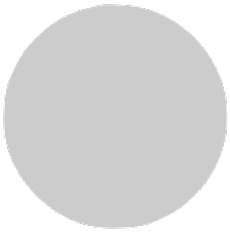
A desmoid tumor (DT) is a benign, locally invasive soft tissue tumor associated with a high recurrence rate but with no metastatic potential.

Your genetic map

Gene	SNP	Genotype
APC	rs62619935	CC
APC	rs876660765	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=873

Hereditary Diseases (genetics)

Familial cold urticaria

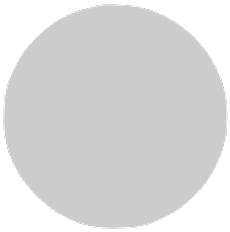
Familial cold urticaria (FCAS) is the mildest form of cryopyrin-associated periodic syndrome (CAPS; see this term) and is characterized by recurrent episodes of urticaria-like skin rash triggered by exposure to cold associated with low-grade fever, general malaise, eye redness and arthralgia/myalgia.

Your genetic map

Gene	SNP	Genotype
NLRP3	rs121908146	CC
NLRP3	rs121908148	AA
NLRP3	rs28937896	TT
NLRP3	rs151344629	CC
NLRP3	rs180177445	AA
NLRP3	rs180177452	AA
NLRP3	rs180177484	GG
NLRP3	rs180177431	TT

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=47045

Hereditary Diseases (genetics)

Vasculitis due to ADA2 deficiency

Vasculitis due to ADA2 deficiency is a rare, genetic, systemic and rheumatologic disease due to adenosine deaminase-2 inactivating mutations, combining variable features of autoinflammation, vasculitis, and a mild immunodeficiency. Variable clinical presentation includes chronic or recurrent systemic inflammation with fever, livedo reticularis or racemosa, early-onset ischemic or hemorrhagic strokes, peripheral neuropathy, abdominal pain, hepatosplenomegaly, portal hypertension, cutaneous polyarteritis nodosa, variable cytopenia and immunoglobulin deficiency.

Your genetic map

Gene	SNP	Genotype
ADA2	rs200930463	CC
ADA2	rs139750129	TT

Multivariate analysis

What do your genetics tell us?

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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=404553

Hereditary Diseases (genetics)

STING-associated vasculopathy with onset in infancy

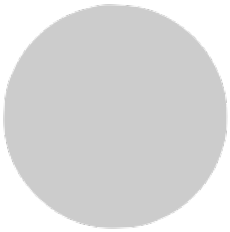
STING-associated vasculopathy with onset in infancy (SAVI) is a rare, genetic autoinflammatory disorder, type I interferonopathy due to constitutive STING (STimulator of INterferon Genes) activation, characterized by neonatal or infantile onset systemic inflammation and small vessel vasculopathy resulting in severe skin, pulmonary and joint lesions. Patients present with intermittent low-grade fever, recurrent cough and failure to thrive, in association with progressive interstitial lung disease, polyarthritis and violaceous scaling lesions on fingers, toes, nose, cheeks, and ears (which are exacerbated by cold exposure) that often progress to chronic acral ulceration, necrosis and autoamputation.

Your genetic map

Gene	SNP	Genotype
STING1	rs587777610	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=425120

Hereditary Diseases (genetics)

Hereditary xanthinuria

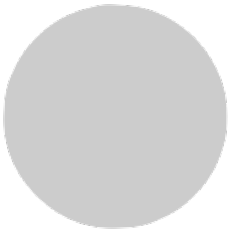
A rare purine metabolism disorder due to inherited deficiency of the xanthine dehydrogenase/oxidase enzyme and is characterized by very low (or undetectable) concentrations of uric acid in blood and urine and very high concentration of xanthine in urine, leading to urolithiasis.

Your genetic map

Gene	SNP	Genotype
XDH	rs119460972	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3467

Hereditary Diseases (genetics)

Cerebrotendinous xanthomatosis

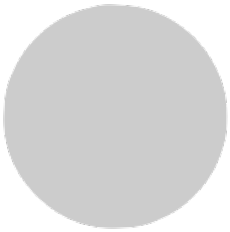
Cerebrotendinous xanthomatosis (CTX) is an anomaly of bile acid synthesis characterized by neonatal cholestasis, childhood-onset cataract, adolescent to young adult-onset tendon xanthomata, and brain xanthomata with adult-onset neurologic dysfunction.

Your genetic map

Gene	SNP	Genotype
CYP27A1	rs397515353	GG
CYP27A1	rs121908097	GG
CYP27A1	rs121908098	CC
CYP27A1	rs121908099	GG
CYP27A1	rs397515355	GG
CYP27A1	rs121908102	CC
CYP27A1	rs72551314	CC
CYP27A1	rs533885672	CC
CYP27A1	rs188850202	CC

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=909

Hereditary Diseases (genetics)

Xeroderma pigmentosum

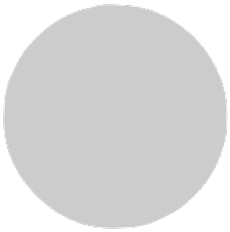
Xeroderma pigmentosum (XP) is a rare genodermatosis characterized by extreme sensitivity to ultraviolet (UV)-induced changes in the skin and eyes, and multiple skin cancers. It is subdivided into 8 complementation groups, according to the affected gene: classical XP (XPA to XPG) and XP variant (XPV) (see these terms).

Your genetic map

Gene	SNP	Genotype
XPA	rs104894132	GG

Multivariate analysis

What do your genetics tell us?



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More information:

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=910

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