

24Genetics



Jane, this is your
skin report





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1. Introduction

In this report, after a thorough analysis of your DNA, you will receive detailed information about the relationship between your genes and your skin.

Thanks to your DNA sequencing, you will learn about your skin's response to different factors, such as oxidation, premature ageing, redness, freckles, varicose veins, cellulite, and more. The report you have in your hands will help you, for example, to use the most suitable creams, according to your skin type, thereby optimising the results of your dermatological treatments.

As is common in our reports, on the first pages you will find a summary, featuring icons, of each of the values analysed, which we cover in more detail in later pages.

We remind you that any changes you make to your health or skin treatments should be reviewed and approved by health professionals. Any questions you have about any genetic test should be answered by medical personnel who are experts in genetic diagnosis, or by dermatologists. On our website we feature the services of these types of professionals.

1.1. Frequently Asked Questions

Should I make drastic changes in light of these test results?

No. Any changes you make regarding your health and skin care should be reviewed and approved by health professionals, such as geneticists or doctors. Any questions you have about any genetic test should be answered by experts in genetic diagnosis, or a dermatologist.

Does it all depend on my genes?

No. The body responds to many factors. Our genes are certainly an important parameter, but lifestyle, exercise, diet, and many other circumstances also affect the body. Knowing yourself well will help you to treat your body in the most appropriate way. And this is what these tests are all about: more knowledge.

Are all the genes analysed listed in the sections?

We include only a sample of the genes we analyse. Some of the sections are defined by the analysis of genes that we do not show in the report. Our algorithms combine all your genotypes from the markers analysed.



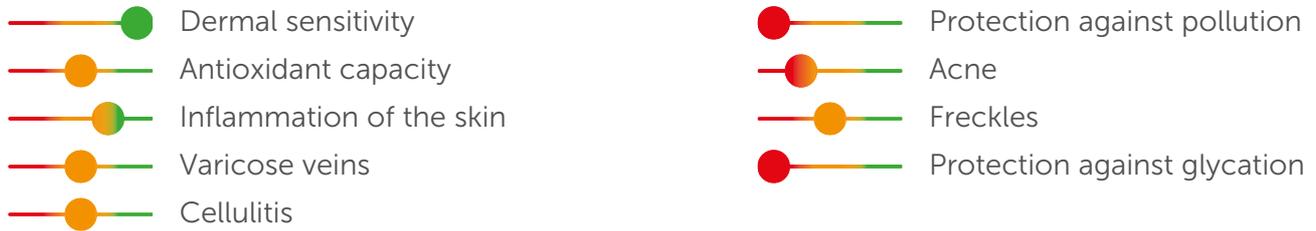
What is this report based on?

This test is based on different genetic studies that have been internationally verified and accepted by the scientific community. There are some databases where studies are published only when there exists a certain level of consensus. Our genetic tests are carried out by applying these studies to our clients' genotypes. In each section you will see some of the studies on which they are based. There are sections where more studies are used than those listed.

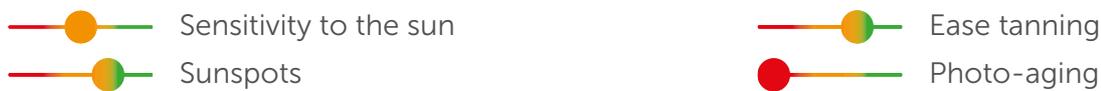
The information provided in this report is valid only for research, information and educational uses. It is not valid for clinical or diagnostic use.

2. Summary

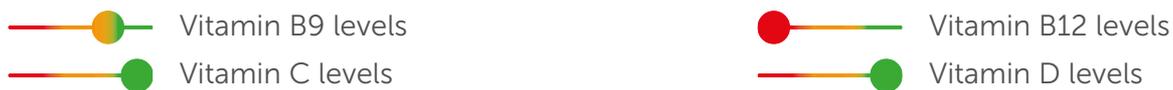
Skin care



The sun and your skin



Feed your skin



Caption:

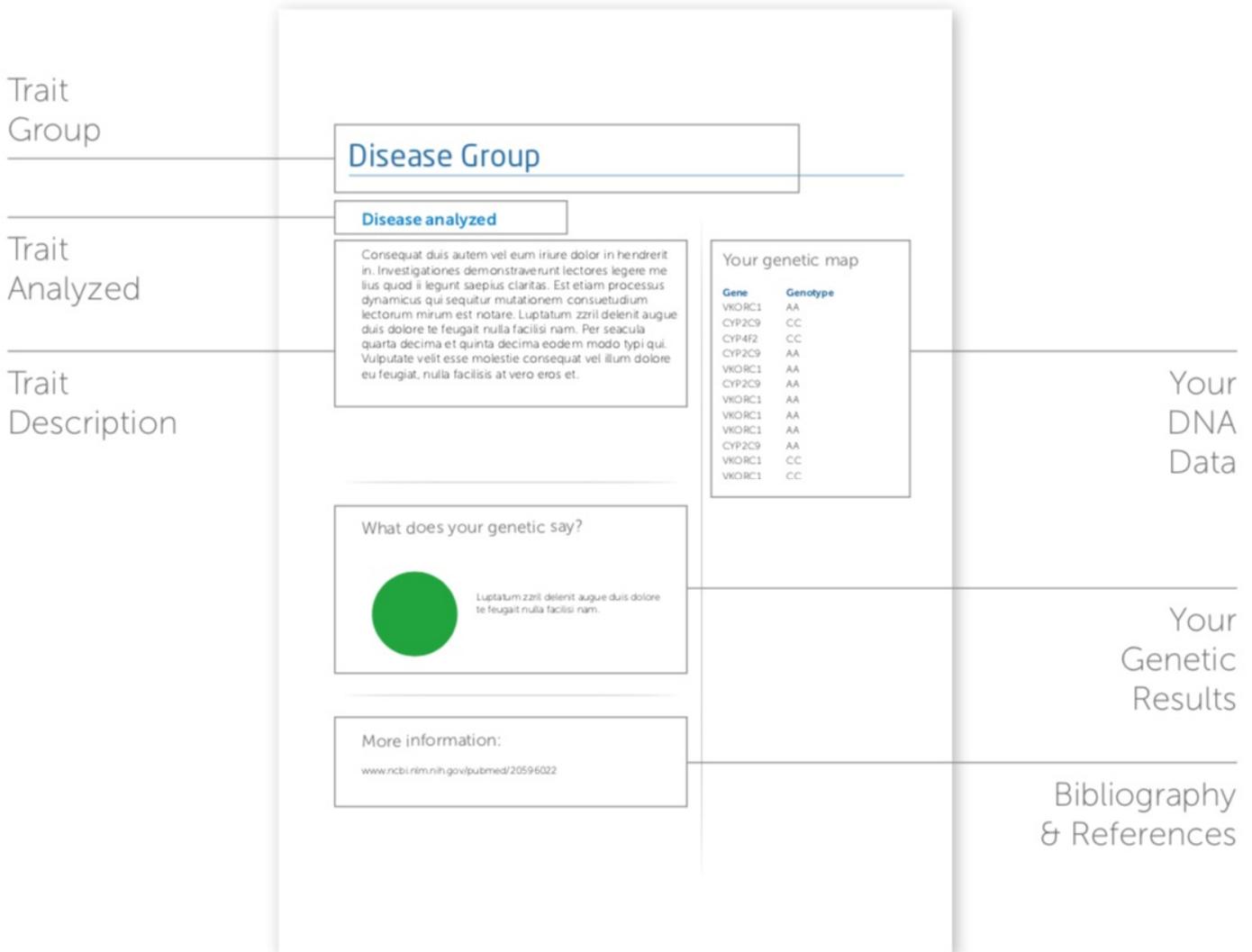
- Your analyzed genotype is favorable.
- Your analyzed genotype is a little favorable.
- Your analyzed genotype doesn't particularly affect you.
- Your analyzed genotype is a little unfavorable.
- Your analyzed genotype is unfavorable.





3. Genetic Results

3.1. How to understand your report?



3.2. Your genetic results



Skin care

Dermal sensitivity

The skin functions as a permeable barrier that blocks the penetration of harmful pathogens and toxins. A hyperreactive immune response to allergens and deficiencies in protection against environmental toxins contribute to the overall risk of dermal sensitivity. In some cases dermal sensitivity results in atopic dermatitis, or eczema, which is the most common skin involvement, with a prevalence of up to 20% in children and 3% in adults in developed countries. People living in cities and in dry climates are more susceptible to this disease. Atopic dermatitis is characterised by very dry skin and inflammatory lesions, which are frequently infected by bacteria and viruses. It is important to see a dermatologist if you have these symptoms.

Genetic and environmental factors appear to be the cause of increased dermal sensitivity. Your overall risk is calculated using the results of a large-scale study in which a number of genetic variants associated with increased risk have been identified.

Your genetic map

| Gene | Genotype |
|-------|----------|
| IL18 | CC |
| ADAD1 | GG |
| EPHX1 | TT |

What do your genetics tell us?

You have genetic variants associated with normal dermal sensitivity.

More information:

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4805319/>

Skin care

Protection against pollution

Environmental pollution causes signs of ageing on the skin, dark spots and inflammation. Two important enzymes (EPHX1 and NQO1) protect the skin and body from highly reactive external chemicals (epoxides and quinones).

The EPHX1 enzyme prevents the absorption of epoxides by converting them into less reactive, water-soluble forms. The NQO1 enzyme converts the coenzyme Q10 (ubiquinone) to its reduced form, ubiquinol, which captures free radicals in the mitochondria and in the lipid membrane of the skin. This enzyme detoxifies quinones by converting them into reduced forms that can be excreted. On the skin, both enzymes play an important role in preventing the outer layer (the epidermis) from absorbing toxins.

Genetic variations in the EPHX1 gene may cause deficiencies in its function and, in the NQO1 gene, decrease the production of ubiquinol. People with reduced levels of these two enzymes suffer from significantly reduced skin protection against environmental toxins.

Your genetic map

| Gene | Genotype |
|-------|----------|
| EPHX1 | TT |
| NQO1 | AG |

What do your genetics tell us?

You are at a greater risk of not properly eliminating the external agents that can damage your skin. Consider taking Coenzyme Q10 supplements and antioxidants, like Astaxanthin. Use products containing antioxidants and coenzyme Q10, and a high sun protection factor. Reduce your exposure to contaminants. Cleanse your skin at night.

More information:

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4868095/>



Skin care

Antioxidant capacity

A balance between free radicals and antioxidants is necessary for proper physiological functioning, as well as for healthy and youthful looking skin. In the skin, the increase in free radicals (called oxidative stress) causes a breakdown of collagen - a structural support of the skin - and alters the cycle of cell regeneration, causing premature ageing (dull complexion with spots and non-uniform texture), proteins and lipids. Free radicals can affect all layers of the skin (hypodermis, dermis and epidermis, being particularly vulnerable).

The antioxidant machinery present in the skin is started when there is oxidative stress, turning harmful free radicals into less harmful products. Antioxidants are the body's natural defence to minimise the damage caused by free radicals, and can drastically reduce some signs of ageing, decreasing wrinkles and preserving the skin's natural shine. Genetic variations encoding antioxidant enzymes (SOD2, EPHX1, CAT and NQO1) have been associated with an increased risk of oxidative stress and a reduction in antioxidant activity, which accelerates the ageing of the skin.

What do your genetics tell us?

The overall antioxidant capacity of your skin is average; some genetic variants are beneficial, while others decrease your skin's antioxidant power.

More information:

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4085290/>

Your genetic map

| Gene | Genotype |
|-------|----------|
| CAT | TC |
| NQO1 | AG |
| SOD2 | AA |
| EPHX1 | TT |
| CAT | TT |

Skin care

Acne

Acne is the most common skin disease, very common among adolescents and young people, although it can also occur in adults. It affects the sebaceous glands, connected to the pores of the skin through a channel called the follicle. These glands produce a fatty substance called sebum, which transports dead cells and sebum to the surface of the skin through the follicle. When a follicle is clogged a pimple is generated, and the bacteria inside the follicle cause swelling.

The treatment focuses on healing the pimples, preventing new ones from forming, and averting scarring. There are anti-acne medications that are applied directly to the skin, and also pills.

In addition to hormonal changes, stress, certain medications, and the use of greasy makeup, there are hereditary factors that contribute to the onset of acne. Variations in different genes contribute to this skin disorder.

Your genetic map

| Gene | Genotype |
|------------|----------|
| NQO1 | AG |
| SELL | GG |
| TGFB2 | AG |
| Intergenic | GG |

What do your genetics tell us?

Your genetic results predispose you to acne.

More information:

<https://www.ncbi.nlm.nih.gov/pubmed/25061327>

Skin care

Inflammation of the skin

Skin inflammation occurs when skin cells have a hyperreactive response to allergens or toxins. Acute inflammation is a natural reaction to repair the skin after being exposed to environmental infections or toxins, and usually lasts a few days. While it is a useful short-term response, if inflammation continues it can play a negative role. When the inflammation is chronic it begins to be destructive and damages the skin.

There are numerous stimuli that induce chronic inflammation: UV rays, stress, toxins, tobacco, alcohol, pathogen infections, excess free radicals. While inflammation is the skin's first line of defence, excessive inflammatory response causes premature ageing of the skin.

Signs include dermal tenderness, redness, and irritation. Genetic variations in various proinflammatory and antiinflammatory genes are associated with an increased risk of chronic skin inflammation.

Your genetic map

| Gene | Genotype |
|-------|----------|
| IL18 | CC |
| IL6 | AG |
| IFNG | AG |
| ADAD1 | GG |
| IL10 | GG |
| IL6 | GC |

What do your genetics tell us?

Your genetics predispose you to a lower risk of excessive inflammatory responses on your skin.

More information:

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4805319/>



Skin care

Freckles

Freckles, also known as ephelides, are hyperpigmented spots that often appear on the face, neck, thorax, and arms. They are the result of an increase in the production of melanin in the skin. They usually appear in childhood, but decrease with age, and darken with sun exposure.

Freckles are common among the Caucasian population and more frequent in light-skinned and red-haired people, who tend not to tan, are more likely to suffer sunburn and sun spots, and are at an increased risk for malignant melanoma and skin cancers.

Freckles are associated with genetic variations in the IRF4 and MC1R genes. The MC1R gene contributes the most to red hair and fair skin. The number of freckles depends on the number of variants of the MC1R gene.

Your genetic map

| Gene | Genotype |
|------------|----------|
| Intergenic | TT |
| Intergenic | GG |
| IRF4 | TC |
| TYR | CC |
| TYR | AG |
| MC1R | CC |

What do your genetics tell us?



Your risk of having freckles is moderate.

More information:

<https://www.ncbi.nlm.nih.gov/pubmed/17952075>



Skin care

Varicose veins

Varicose veins are small veins that have a strong purple-blue colour and extend into the skin like roots (also known as spider veins). They affect more than one third of the world's population and can cause pain, itching, ulceration and venous thrombosis.

There are very simple measures to prevent the formation of varicose veins. The Mayo Clinic recommends exercising regularly, maintaining a healthy diet, avoiding standing or sitting for prolonged periods, and raising the legs. Women should also avoid wearing high-heeled shoes and crossing their legs.

People who have a family history of varicose veins are more likely to have them. Genetic variations in the MTHFR gene have been associated with an increased risk of developing varicose veins. Other non-genetic risk factors are obesity, age, sitting or standing for a long time, and hormonal changes.

Your genetic map

| Gene | Genotype |
|-------|----------|
| MTHFR | TT |
| MTHFR | AG |

What do your genetics tell us?

Based on your genotype, the likelihood of you suffering from varicose veins is moderate.

More information:

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2674155/>



Skin care

Protection against glycation

Our body uses glucose as its main source of energy, but if it is not properly metabolised it can bind to collagen and elastin fibres and modify them, both structurally and functionally. The resulting products are known as Advanced Glycation End Products (AGEs).

This process, called glycation, is involved in the ageing of the skin and impairs its ability to regenerate and repair itself. Collagen fibres that have undergone glycation become rigid, less elastic and have a decreased capacity for regeneration, causing wrinkles, dryness, thickening of the skin, and a loss of firmness. AGEs increase with age, and are more harmful in combination with exposure to UV rays.

Glycation can be reduced by controlling blood glucose, LDL cholesterol and triglyceride levels through a proper diet. Variations in the genes that determine how our body processes sugar can alter the normal functioning of energy metabolism and glucose levels. Scientists have identified variations in genes like GLO1 and AGER associated with an excess of AGE.

Your genetic map

| Gene | Genotype |
|------|----------|
| AGER | AA |
| AGER | AG |
| GLO1 | AG |

What do your genetics tell us?



Your genotype predisposes you to a high risk of glycation in your skin's components. Niacinamide, carnosine and green tea reduce advanced glycation products on the skin. The use of creams with carnosine, niacinamide, silibinin and α -lipoic acid decreases the effects of advanced glycation products.

More information:

<https://www.ncbi.nlm.nih.gov/pubmed/23721855>

Skin care

Cellulitis

Cellulitis is a common cosmetic problem for many women (80-90%), characterised by alterations in the surface of the skin that give it an uneven, unattractive appearance due to irregular fibrous tissue and the accumulation of subcutaneous fat. It mainly affects the buttocks, hips and thighs, although it can also affect other parts of the body, such as the abdomen.

Caucasian women are more likely to have cellulitis than Asian women, partly because of differences in diet. The causes are complex and involve hormonal factors, the circulatory system, the extracellular matrix, inflammation, and substances produced by adipocytes, genetic predisposition and weight changes.

To minimise cellulitis it is recommended to maintain a healthy weight, stay active, follow a healthy diet, and stay hydrated. Certain anti-cellulitis creams, massages and medical-aesthetic treatments are beneficial in treating this disorder. Variations in the HIF1A gene, among others, have been associated with the risk of developing cellulitis.

Your genetic map

| Gene | Genotype |
|-------|----------|
| HIF1A | CC |

What do your genetics tell us?

You do not have the protective genotype, so your predisposition to cellulite is average. Caffeine is a very common component in anti-cellulite creams. Other ingredients are tetrahydroxypropyl ethylenediamide (THPE), retinol and/or red algae and glaucine. There are numerous formulations. See your beauty consultant.

More information:

<https://www.ncbi.nlm.nih.gov/pubmed/20059631>

The sun and your skin

Sensitivity to the sun

The skin can be sensitive to the sun for a variety of reasons: it is underdeveloped (childhood), or inflamed (atopic dermatitis or acne), or due to photosensitivity induced by drugs or dermatological treatments. In these cases it is vital to use protection with a Sun Protection Factor (SPF) suitable for each type of skin.

Sensitivity to the harmful effects of ultraviolet radiation is inheritable. Numerous large-scale studies have identified genetic variations that enhance sensitivity to the sun and the tendency we have to suffer from sunburn (erythema).

The genes related to skin pigmentation (ASIP, TYR, MC1R, and OCA2) and a low tanning capability are those that most influence the skin's sensitivity to the sun. In addition, there is a strong association between DNA repair genes and the tendency to suffer from sunburn. These genes have no relation to tendency to tan, so there is an underlying mechanism to burns that is independent of pigmentation.

Your genetic map

| Gene | Genotype |
|-------------|----------|
| NTM | GG |
| TYR | AG |
| ASIP | TC |
| LOC10537487 | CC |

What do your genetics tell us?

Your propensity towards sun-sensitive skin is intermediate.

More information:

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3690971/>

The sun and your skin

Ease tanning

Tanning is the physiological response stimulated by ultraviolet (UV) radiation from the sun's rays. Exposure to UV rays increases the production of eumelanin, a type of melanin pigment that darkens the skin to protect it from damage. Different individuals' tanning capacities vary, and can have positive and negative effects on the health of the skin.

People with less capacity are more prone to burns and sun spots, wrinkles, folate loss and melanoma, while people who tan easily are at risk for Vitamin D deficiency, because they can produce less Vitamin D in response to solar exposure.

The skin's tanning capability is variable and is genetically determined. People with certain variants in genes related to pigmentation usually have light-coloured eyes and skin, and a reduced tanning capacity. Variations in the MC1R gene (melanin receptor) are the most determinant, and are associated with red hair, freckles, increased sensitivity to the sun and less tanning.

Your genetic map

| Gene | Genotype |
|-------------|----------|
| LOC10537406 | TC |
| LOC10537487 | CC |
| HERC2 | GG |
| ASIP | TC |
| ASIP | GG |
| IRF4 | TC |
| MC1R | CC |
| TYR | CC |
| TYR | AG |
| MC1R | TC |

What do your genetics tell us?

Your skin is very likely to tan easily.

More information:

<https://www.ncbi.nlm.nih.gov/pubmed/23223146>

The sun and your skin

Sunspots

Facial sunspots (sun lentigos, or lentigines) are oval or round, pigmented spots that measure 2 to 20 mm, are brown, uniform, and located in areas frequently exposed to the sun, like the face, arms and back of the hands. They are larger than freckles/ephelides, do not disappear in the winter, and are common in ageing skin.

Solar lentigines are the result of the local growth of melanin-producing cells in response to ultraviolet radiation. These spots are more frequent among the Caucasian and Asian populations, and in women, especially after age 50. Although they are benign lesions that do not need medical treatment, they indicate that sun exposure has been excessive. For aesthetic reasons they can be eliminated by different treatments, although the best form of prevention is the use of sunscreens and limiting sun exposure.

Variations in MC1R and IRF4 genes have been associated with an increased risk of sunspots. There are numerous risk alleles in the MC1R (melanin receptor) gene.

Your genetic map

| Gene | Genotype |
|------|----------|
| IRF4 | TC |
| MC1R | GG |
| MC1R | CC |

What do your genetics tell us?

You are at a low risk of sun spots on your skin.

More information:

<https://www.ncbi.nlm.nih.gov/pubmed/25705849>



The sun and your skin

Photo-aging

Photoageing is defined as the premature ageing of the skin due to sun exposure. It is one of the harmful effects of prolonged and daily exposure to UV radiation, which causes DNA damage, oxidative stress and alterations to the normal architecture of the skin's connective tissue, impairing its function.

Most premature ageing is caused by exposure to the sun. Photoageing is responsible for uneven pigmentation, fine wrinkles, sagging skin, freckles, age spots, spider veins on the face, and rough skin. It is, therefore, crucial to protect the face and body against the negative effects of UVA and UVB rays.

Numerous studies suggest that photoageing has a genetic basis. Variations in the genes STXBP5L and FBXO40 have been associated with an overall photoageing score that combines factors such as irregularities in pigmentation, wrinkles and sagging skin.

Your genetic map

| Gene | Genotype |
|---------|----------|
| FBXO40 | AA |
| STXBP5L | GG |
| STXBP5L | TC |

What do your genetics tell us?



Your genotype indicates an increased risk of severe photoaging. Consuming fruits and vegetables rich in antioxidants is a fundamental step to prevent ageing. Creams with arbutin, kojic acid, L-ascorbic acid, licorice extract, retinol and Vitamin B3 can help you avert the signs of premature photoaging.

More information:

<https://www.ncbi.nlm.nih.gov/pubmed/23223146>



Feed your skin

Vitamin B9 levels

Vitamin B9 (folate) is a water-soluble vitamin that is essential in processes such as DNA synthesis, cell repair, protein metabolism and proper brain function. It is naturally present in foods such as leafy green vegetables, peas, lentils, fruits, cereals and other foods. Folic acid is an artificial (synthetic) folate contained in supplements and added to fortified foods. Vitamin B9 deficiencies are associated with anaemia, high homocysteine levels, increased risk of heart disease, complications during pregnancy, increased risk of cancer, and cognitive dysfunction in old age.

Genetic studies have shown that the MTHFR gene is associated with low levels of vitamin B9 in the blood and an increase in homocysteine, a substance that, at high levels, is linked to cardiovascular disease.

Your genetic map

| Gene | Genotype |
|-------|----------|
| MTHFR | AG |
| MTHFR | TT |

What do your genetics tell us?

Based on your genotype, you are not predisposed to have a vitamin B9 deficiency. Other genetic and clinical factors may be relevant.

More information:

<https://www.ncbi.nlm.nih.gov/pubmed/17115185>



Feed your skin

Vitamin B12 levels

Vitamin B12 (cobalamin) is a water-soluble vitamin that plays an important role in the functioning of the brain, the nervous system and the digestive system, and it is an essential component in the synthesis and regulation of DNA and for the metabolism of fatty acids and amino acids. It is produced by bacteria and is found naturally in foods of animal origin: meat, fish, eggs and dairy products. Excess vitamin B12 can cause blurred vision, vomiting, diarrhoea and gastric disorders, blood clots, and damage to the liver and kidneys. Various genetic studies have identified that the MTHFR gene is associated with excess blood concentrations of vitamin B12 in women.

Your genetic map

| Gene | Genotype |
|-------|----------|
| FUT2 | AG |
| MTHFR | TT |

What do your genetics tell us?

Based on your genotype, you are predisposed to have excess levels of vitamin B12. Other genetic and clinical factors may be relevant.

More information:

<https://www.ncbi.nlm.nih.gov/pubmed/18776911?dopt=Abstract>



Feed your skin

Vitamin C levels

Vitamin C (ascorbic acid) is a water-soluble vitamin that is critical for life processes, such as the functioning of the immune system, the production of red blood cells, and the maintenance of connective tissue, blood vessels, bones, teeth and gums. It is a powerful antioxidant and is involved in iron absorption. A severe deficiency can cause scurvy, which leads to anaemia, bleeding gums, bruising and poor wound healing. Some foods rich in vitamin C include kiwis, lemons, oranges, red pepper, watermelon, strawberries, broccoli and other vegetables.

A recent study in about 15,000 people found that a variant of the SLC23A1 gene is associated with low levels of vitamin C in the blood.

Your genetic map

| Gene | Genotype |
|---------|----------|
| SLC23A1 | CC |

What do your genetics tell us?

Based on your genotype, you are not predisposed to have low levels of vitamin C. Other genetic and clinical factors may be relevant. The best way to maintain adequate levels of vitamin C is to eat a balanced diet that contains a variety of foods. Smokers and pregnant or breastfeeding women may need to increase their daily intake of vitamin C.

More information:

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3605792/>



Feed your skin

Vitamin D levels

Vitamin D (calcidiol or calcifediol) is a fat-soluble vitamin that is important in the absorption and use of calcium, for maintaining good bone and muscle health, and for the proper functioning of the immune, endocrine and cardiovascular systems. It is synthesised in the skin after exposure to sunlight, which transforms it to its active form. Recently, an increase in cases of vitamin D deficiency has been identified in developed countries mainly due to lifestyle, the use of sunscreens and environmental conditions (pollution, geographic location).

Numerous studies have identified variations in the GC gene related to vitamin D deficiency.

Your genetic map

| Gene | Genotype |
|------|----------|
| GC | TC |
| GC | TT |

What do your genetics tell us?

Based on your genotype, you are not predisposed to vitamin D deficiency. Other genetic and clinical factors may be relevant. Exposure to sunlight is crucial to a person's vitamin D levels because there are few dietary sources of this vitamin.

More information:

<https://www.ncbi.nlm.nih.gov/pubmedhealth/PMH0065716/>

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