

24Genetics



Jane, this is your sport report



FORMI



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1. Introduction

1. Introduction

This report is a fundamental tool for athletic activity, both amateur and professional. For example you will see your genetic propensity to, for example, suffer injuries, have a lower heart rate, and your capacity for muscle regeneration.

Your genes determine your sport profile, the metabolic and even the overall benefit of the sport for your body. Thanks to the sequencing of your DNA done by 24genetics, and its subsequent analysis, you can optimize your workouts, discovering what types of exercise your body is best prepared for and for which ones you have to take special care if you want to avoid certain injuries.

Here are the sections you will find:

- Sports Performance

In this section, we tell you about the sporting potential stored in your genetics. Your capabilities in terms of your sporting efficiency range from your power to your muscular response to strength training, including training your aerobic capacity.

- Physical Exercise and Health

Every day, when you decide to start or resume your sports routine, you subject your muscles to different impacts. That's why knowing your genetic risk for injuries such as bone fracture becomes so essential.

- Metabolic profile

The food you eat daily is vital, but knowing how your body transforms those nutrients into energy, too. In this section of the report, we tell you, among many other things, how genetics influences your body's ability to control your glucose or cholesterol levels with exercise.

- Cardiac characteristics:

Your heart is the engine of your body; it moves you and allows you to train. Given its importance, our multidisciplinary scientific team decided to dedicate a complete section to our most important organ, where you will find parameters as necessary as your genetic tendencies in terms of heart rate or the morphology of your aortic root.

Special section: Sudden Death

We dedicate a special section to this type of natural death, given its implications at a sporting/genetic level. We typically define sudden death as natural and unexpected death from cardiac arrest within one hour of the onset of collapsing symptoms. Broader definitions of premature death are also used but do not generally apply to the sporting situation.

Most causes are related to cardiovascular disease, congenital or acquired, with no symptoms observed before the fatal event. It is important to note that 24Genetics tests do not sequence the entire genome but analyze 700,000 of the total 3.2 billion genetic links. Therefore, not finding any mutation does not mean that we are not carriers since the mutations may be found in the rest of our genome, which we are not analyzing.

Likewise, we do not analyze all the genetic information related to each disease. Specifically, in this section, we study on average slightly less than half of the pathogenic markers reported for pathologies or syndromes associated with sudden death in the databases consulted, so we could have mutations in the other half and not see them in this report.

The mutations we are looking for are reported in some of the most critical genetic databases worldwide, mainly OMIM and ClinVar. It is important to note that if you need a diagnosis of a particular disease, genetic tests analyze the entire gene or genes involved in that disease and are valid for clinical use. Therefore, if you have a family history, we recommend that you consult your doctor or geneticist to study the test's needs.

As usual in our reports, in the first pages you will find an iconographic summary of each of the analyzed values, which we develop more broadly in later pages. We remind you that any changes you want to make in your diet or your health treatments should be guided by health professionals.

Any doubts you have about any genetic test should be checked against health personnel who are experts in Genetic Diagnosis or Specialized Physicians.

1.1. Frequently Asked Questions

Do my genes determine everything?

Your genes are very important and determine the working predisposition of your body, but there are many other factors that influence it: lifestyle, exercise, diet... In any case, knowing yourself well, helps you to treat your body in the most appropriate way. That is what genetics gives you: information and, consequently, knowledge.

Can there be more features than those in my report?

Every day, new research is published worldwide that allows us to expand the knowledge we have in the field of genetics. This research is constantly analysed by 24Genetics to incorporate it into our algorithm and improve it, with the aim of obtaining more relevant information from your DNA data. This means that your report may evolve and offer more data than you currently have, in which case we will contact you to notify you.

Are all sports genetic tests the same?

Not all sports genetic testings are the same. There are not too many biotech companies with the capacity to perform these complex analyses and most of them give very poor conclusions in terms of the number of results. Thanks to our test, with some 700,000 genetic markers and our complex algorithms, we can offer what we believe is to date, the most comprehensive sports

genetic study on the market. The genetic information provided by 24Genetics is valid for research, information and educational purposes. Under no circumstances is it valid for clinical use.

What are 24genetics tests based on?

Our tests are developed based on countless genetic studies with the highest recognition and acceptance by the international scientific community. The scientific studies are published, through prestigious institutions and organisations in certain databases, as long as there is a certain level of consensus. These validated studies are used to create and update our algorithm, which is applied to our clients' genetic data.

If I have a low predisposition to injury, am I sure I won't get injured?

Absolutely not, not having a greater genetic predisposition is not a guarantee of not suffering injuries. These studies are normally carried out on the basis of statistical data obtained from a sufficiently large sample of people, in which the genetic differences between people who have a certain pathology and those who do not are studied. The type of conclusion that is usually reached is that people who have a genetic alteration have a greater predisposition to suffer from a certain pathology. However, this does not mean that 100% of people with that alteration will have that pathology. Equally, it does not mean that 100% of people who do not have this alteration cannot suffer from this pathology.

Example of some studies that support our sports genetics test

- Collins M et al; The COL1A1 gene and acute soft tissue ruptures; Br J Sports Med; 2010 Jun 11.
- Posthumus M et al; Components of the transforming growth factor-beta family and the pathogenesis of human Achilles tendon pathology—a genetic association study; Rheumatology; 2010 Apr 1.
- Posthumus M et al; The COL5A1 gene is associated with increased risk of anterior cruciate ligament ruptures in female participants; Am J Sports Med; 2009 Nov;37(11):2234-40.
- Raleigh SM et al; Variants within the MMP3 gene are associated with Achilles tendinopathy: possible interaction with the COL5A1 gene; Br J Sports Med; 2009 Jul;43(7):514-20.
- September AV et al; Variants within the COL5A1 gene are associated with Achilles tendinopathy in two populations; Br J Sports Med; 2009 May;43(5):357-65.

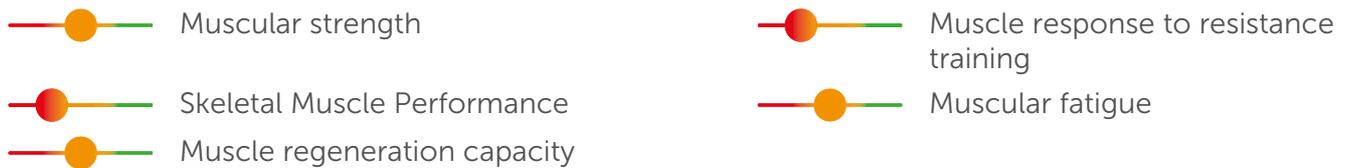
The information provided in this report is valid only for research, information and educational uses. It is not valid for clinical or diagnostic use.

2. Summary

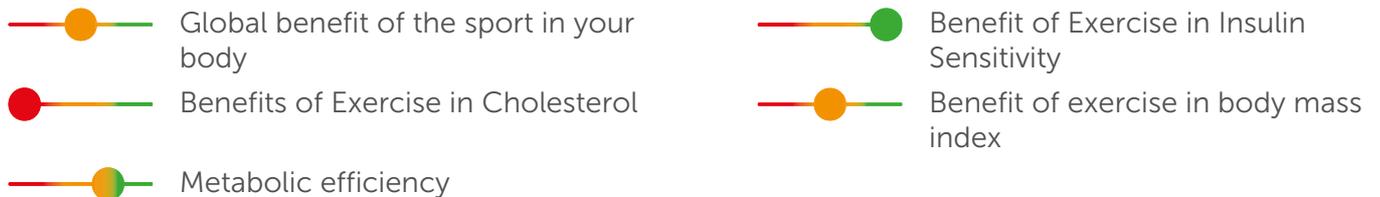
Sport profile



Muscle Profile



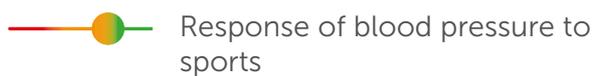
Metabolic Profile



Injury Risk



Cardiovascular profile



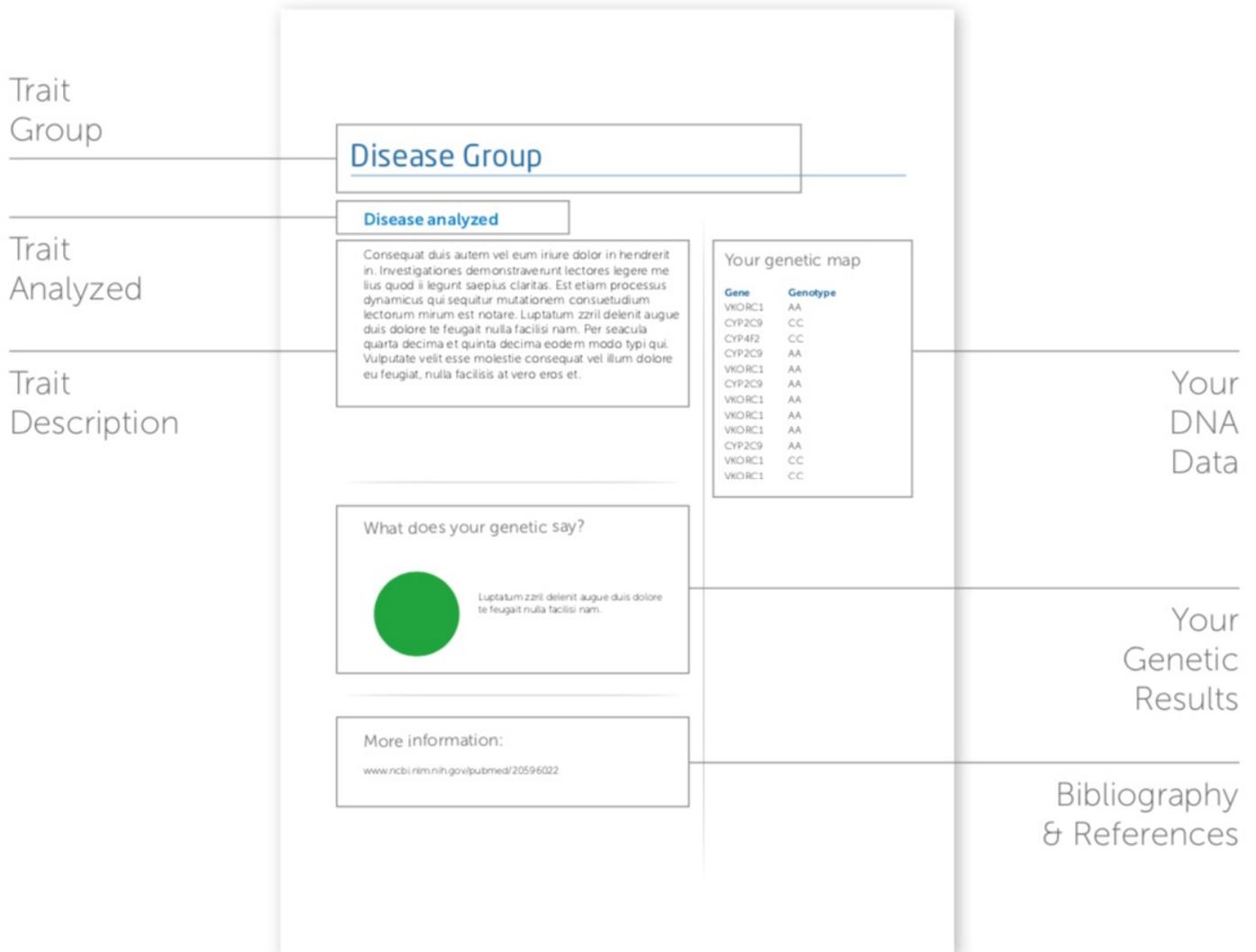
Caption:

- 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

Sport profile

Strength

Muscle strength measures the maximum amount of force that can be exerted over a limited period of time. Rapid shrinkage fibres generate a relatively high amount of force over a short period of time. They are characterised by great strength, power and speed, but they fatigue faster. They are less able to obtain aerobic energy, lower oxygen levels, and higher levels of glycogen, so at first they get energy from glycolysis (anaerobic respiration) for muscle contraction.

This process is very fast, but it is also quite inefficient at producing energy; and it produces lactic acid, which favours muscular fatigue. This explains why fast-twitch fibres tire faster.

It is estimated that power is 80% inherited, depending on the specific type of muscle (isometric strength of the knee, hand strength, elbow flexion). To assess the power predisposition profile, genetic markers have been used that have been associated with power sports.

Your genetic map

Gene	Genotype
ACE	AA
IGF2BP2	TT
NOS3	AG
PPARG	CC
AGT	AG
PPARA	GG
VEGFA	GG
VDR	GG
PPARGC1A	TC
HIF1A	CC

What do your genetics tell us?

Your genetic predisposition to stand out in strength-related sports is low.

More information:

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2658665/>

Sport profile

Resistance

Resistance training is defined as a low-intensity activity performed over a long period of time. Muscle endurance measures your ability to sustain an activity for a time period without feeling tired.

If your muscular structure favours endurance, you have the potential to thrive at sports that depend on this ability. The intrinsic ability to perform resistance exercise is influenced by several factors. First, resistance depends on the proportion of slow-twitch fibres of skeletal muscle. These are also known as red fibres because they contain more myoglobin, a protein that stores oxygen, and they have their own energy source, so they can maintain their strength longer. Second, it has been observed that the top athletes usually have what have been called "marathon genes".

Studies have identified genetic variants associated with a high proportion of these fibres and a high supply of oxygen to muscle tissue.

Your genetic map

Gene	Genotype
PPARGC1A	TC
ACE	AA
NFIA AS2	GG
HIF1A	CC

What do your genetics tell us?

Your genetic profile indicates that you are predisposed to perform endurance sports.

More information:

<https://www.ncbi.nlm.nih.gov/pubmed/15705733>



Sport profile

Aerobic capacity

Maximum aerobic capacity (or maximum volume of oxygen, VO₂max) is the maximum volume of oxygen a athlete's muscles can use for one minute to produce maximum physical effort. This measure reflects the person's aerobic physical condition and determines their power during prolonged exercise. The benefits of having good aerobic fitness are low pressure, low cholesterol, and reduced risk of obesity, type-2 diabetes and cardiovascular disease. VO₂max is measured in L/min, but is more commonly expressed in mL of O₂/kg/min in order to equitably compare athletes whose body masses are different. Absolute VO₂max figures are usually 40-60% higher in men than in women.

Beginning at age 30 lung capacity begins to decline, and at age 50 may be half of what it was. This decrease means that less oxygen enters our cells, leading to reduced respiration and endurance, and increased susceptibility to respiratory diseases with age. Numerous genetic variants have been associated with aerobic capacity.

Your genetic map

Gene	Genotype
NFIA AS2	GG
RGS18	AG
ACSL1	AA

What do your genetics tell us?

Your genotype does not tend to endow you with an enhanced lung capacity, but you can improve it, so your muscles can convert oxygen into energy more efficiently. Perform intense exercise, increasing your heart rate to 70-85%.

More information:

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4314597/>

Sport profile

Response to Strength training

With regards to physical condition, we will define strength as the capacity to overcome resistance through the contraction produced by muscles; that is, their capacity to perform a physical task.

The quality of strength is determined by the muscular structure itself: it depends on the orientation and types of muscle fibres, the length of the muscle, and temperature: muscle contraction is more rapid and potent when the internal temperature is slightly higher than normal; and by the osteoarticular system: strength depends on the type of lever that makes the movement; and, finally, age and gender. Training is another important factor because it improves the factors that impact muscular strength: metabolism and fuel deposits that increase muscle fibre thickness, the number of myofibrils, and a delay in the appearance of muscle fatigue.

In addition, genetic factors have been associated with greater benefits in the form of increased strength after training.

Your genetic map

Gene	Genotype
INSIG2	GG

What do your genetics tell us?

Strength training is less beneficial for people with your genotype, as you are likely to gain fat mass. Moderate training is recommended.

More information:

<https://www.ncbi.nlm.nih.gov/pubmed/19105843>

Sport profile

Resilience

Prolonged exercise involves muscle lengthening and may result in structural muscle disruption, deterioration of the excitation-contraction process, inflammation, and the breakdown of muscle proteins.

This process is known as exercise-induced muscle damage, and although a certain amount of muscle damage is required for adaptation to occur, excessive damage or inadequate recovery from muscle damage may increase the risk of injury.

After performing physical exercise, some people recover quickly and are ready to make a physical effort again after a brief rest. Other people do not recover as quickly, and need more rest time. Research has shown that certain genetic variants are associated with slower recovery after hard exercise. People with these markers should take special care with their training plan.

Your genetic map

Gene	Genotype
IL6	GC
CRP	TC
SOD2	AA

What do your genetics tell us?

Recovery time after exercise, in your case, is intermediate.

More information:

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4983298/>

Muscle Profile

Muscular strength

Muscle strength is the amount of force a muscle can exert in a single contraction. Muscles have two types of fibres: fast and slow. Fast-twitch fibres provide explosive energy; to lift weights, or run fast, for example. Slow-twitch fibres are for more endurance-related exercises.

Developing fast-twitch fibres requires aerobic activity, whereas for the development of slow-twitch fibres anaerobic exercise is required, to gain strength. Muscle strength is determined by fast fibres, which provide rapid explosions of energy.

Studies with relatives have shown that up to 90% of the variation in muscle mass, and up to 60% of variation in muscle strength, are heritable. Genetic variations have been associated with muscle strength.

Your genetic map

Gene	Genotype
HFE	CG
IGF1	GG
HIF1A	CC
GDF8	TT
IGF1	TT
SLC30A8	CC
CCL2	AA

What do your genetics tell us?

Your strength and muscle mass are intermediate, the same as those of the general population.

More information:

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4696732/>

Muscle Profile

Muscle response to resistance training

Workouts featuring resistance training are recommended at least two days a week. Systematic, long-term resistance training increases skeletal muscle size and strength in men and women of different ages, as well as the aerobic capacity of fast-twitch fibres.

There are studies that show that almost all people benefit from endurance exercises, although gains in muscle size and strength are highly variable amongst individuals, and depend on gender, age, general health, nutrition and personal genetics.

Several studies have reported an association between certain genetic variations and muscle size and strength. Some people gain more strength and muscle size in response to the same training as others.

Your genetic map

Gene	Genotype
BMP2	AA
IL15RA	CC
INSIG2	GG

What do your genetics tell us?

You do not have a propensity to easily increase your muscle strength through exercise.

More information:

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4147943/>

Muscle Profile

Skeletal Muscle Performance

Muscles such as biceps, pectorals and quadriceps are skeletal muscles that are attached to the skeleton to generate movement. Skeletal muscle is composed of elongated, thin cells, which include all the organelles necessary for cellular functions. More than 90% of the total volume of skeletal muscle cells is composed of muscle proteins, including actin and myosin contractile proteins.

When a muscle cell is activated by a nerve impulse, the interaction between actin and myosin generates a contraction. The total force depends on the sum of all the contractions that occur simultaneously in a muscle cell. Skeletal muscle is one of the three main types of muscles, the others being the heart and smooth muscle. The UCP2 and UCP3 proteins can negatively regulate mitochondrial ATP synthesis (energy that muscles use), thereby influencing physical performance. One study has found that genetic variants in these genes are associated with improved skeletal muscle performance through training.

Your genetic map

Gene	Genotype
UCP2	CC

What do your genetics tell us?

Your genotype does not favour enhanced muscle performance through exercise.

More information:

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3330832/>

Muscle Profile

Muscular fatigue

Muscle fatigue occurs when muscles cannot exert normal strength, or when it takes more effort than normal to achieve a desired level of strength. Late-onset muscle pain describes a phenomenon of muscle pain or stiffness that is felt 12-48 h after exercise, particularly when starting a new training program, after a change in sports activity, or after a considerable increase in the duration or intensity of exercise.

The proteins of an injured muscle are released into the blood. A higher concentration of these proteins means greater damage to muscle fibres and a greater likelihood of muscle fatigue.

In addition to exercise, genetic condition is another cause of muscle fatigue. There are studies that relate certain genetic variants with enhanced resistance to muscular fatigue.

Your genetic map

Gene	Genotype
HNF4A	GG
NAT2	AA

What do your genetics tell us?

The likelihood of your muscles suffering damage and fatigue is average.

More information:

<https://www.ncbi.nlm.nih.gov/pubmed/19406499>

Muscle Profile

Muscle regeneration capacity

Muscles are important for exercise and, after it, need between 24 and 48 hours to repair and rebuild. Making them work again too soon simply leads to tissue breakdown.

Are you one of those people who needs a lot of time to recover after muscle damage? Prolonged and tiring exercise, such as high-intensity training, activates inflammatory factors. Genetic variations in several genes improve the inflammatory response that allows for the slow repair of muscle damage after exercise.

A person with a high predisposition to inflammation will benefit from less frequent exercise and longer recovery periods. If the body is not fully recovered, it can suffer damage due to overexertion and excessive training. This is particularly important for high-intensity athletes and bodybuilders.

Your genetic map

Gene	Genotype
IL1B	GG
IL1B	AG

What do your genetics tell us?

Your muscular regeneration capacity is average, similar to that of the rest of the population.

More information:

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1665272/>



Metabolic Profile

Global benefit of the sport in your body

The benefits of exercise and regular physical activity are well known, and all people, regardless of age, sex or physical ability can notice its benefits. Exercise can prevent weight gain and help prevent many health problems, such as heart attacks, metabolic syndrome, type-2 diabetes, depression, various cancers, and arthritis.

Exercise releases oxygen and nutrients to tissues and helps the cardiovascular system work more efficiently. When the heart and lungs are healthier, the body has more energy.

Some people experience the benefits of exercise more quickly than others, but may also require dietary changes. People with certain genetic variants experience rapid results, such as lower cholesterol, triglycerides and blood pressure.

Your genetic map

Gene	Genotype
CETP	CC
BDNF	CC

What do your genetics tell us?

The benefits from exercise you will experience are those noted by the general population.

More information:

<https://www.ncbi.nlm.nih.gov/pubmed/21252145>



Metabolic Profile

Benefit of Exercise in Insulin Sensitivity

Insulin helps control changes in glucose levels (commonly known as sugar) in the body. Insulin sensitivity refers to the body's ability to respond to these changes.

Having a greater sensitivity to insulin means that the body is better able to process glucose. Insulin resistance, on the other hand, is an alteration that impedes the proper regulation of glucose, and is associated with obesity and type-2 diabetes. Many people can benefit from exercise to increase insulin sensitivity.

According to one study, people with the beneficial genotype in a marker of the LIPC gene benefit more in the form of increased insulin sensitivity.

Your genetic map

Gene	Genotype
LIPC	TC

What do your genetics tell us?

You enjoy increased benefits from exercise in the form of better insulin sensitivity. This is especially important if you are diabetic, are overweight, or have a metabolic syndrome.

More information:

<https://www.ncbi.nlm.nih.gov/pubmed/15983229>



Metabolic Profile

Benefits of Exercise in Cholesterol

One of the benefits of exercise is improved cholesterol levels. HDL cholesterol is known as good cholesterol, and having high levels of HDL is beneficial. Many people can improve their HDL levels through exercise.

Research has shown that exercise stimulates enzymes that help move bad cholesterol from the blood to the liver, allowing it to be excreted with bile. It has also been stipulated that exercise increases the size of protein particles that carry cholesterol through the blood, reducing the possibility that small particles clog arteries.

Individuals with certain genetic variants will do well to increase their good cholesterol levels while exercising, while carriers of other genetic variants are less likely to lower their bad cholesterol levels through exercise alone.

Your genetic map

Gene	Genotype
CETP	CC
PPARD	TT

What do your genetics tell us?

Your genotype is not associated with a greater capacity to regulate your cholesterol levels through exercise.

More information:

<https://www.ncbi.nlm.nih.gov/pubmed/21252145>



Metabolic Profile

Benefit of exercise in body mass index

Exercise is part of weight loss plans, and is a crucial tool for maintaining a healthy weight. Physical activity is beneficial for all people, regardless of their genetics, but exercise is especially recommended for people at increased risk of being overweight.

People with a certain variant in the genetic marker of the FTO gene are more likely to be overweight, have an increased Body Mass Index, and waist circumference. However, a large-scale study has shown that genetic susceptibility to obesity-induced variants in the FTO gene can be changed by adopting an active lifestyle.

In fact, people who are more susceptible to obesity experience greater weight loss by exercising at moderate intensities.

Your genetic map

Gene	Genotype
FTO	AA
FTO	CC

What do your genetics tell us?

Your capacity to lose weight through exercise and to enjoy its benefits is normal.

More information:

<https://www.ncbi.nlm.nih.gov/pubmed/19553294>

Metabolic Profile

Metabolic efficiency

Metabolism refers to the chemical processes that the body undergoes to convert food into energy, and a concept related to the way in which each body uses nutrients.

Physical fitness is a very complex phenotype, influenced by numerous genetic and environmental factors that contribute to inter-individual variation. Sports genomics studies the genetic components that determine sports performance.

Variations in various genes play an important role in how bodies respond to different types of physical activity, as these genes have a physiological impact on sports performance. Some genes analysed are involved in the metabolism of fatty acids whose expression can improve the oxidative capacity of skeletal muscle during exercise; i.e. different variants result in a more or less efficient acquisition of energy from fatty acids and other nutrients.

Your genetic map

Gene	Genotype
AMPD1	GG
PPARA	GG
ADRB2	AA
PPARD	TT
PPARGC1A	TC

What do your genetics tell us?

Your genotype predisposes you to a highly efficient metabolism, associated with enhanced athletic performance.

More information:

<https://www.ncbi.nlm.nih.gov/pubmed/20044476>



Injury Risk

Risk of injury to joints

Many sports-related injuries involve joint damage. The most common are wrist or ankle sprains, excessive elbow extension, and damage to the knee ligaments.

When exercising you are at higher risk of injury from the excessive use of joints, but you can strengthen and avoid injury by doing the exercise correctly, and stretching. Risk of joint damage is associated with an increased genetic predisposition to osteoarthritis.

Knowing personal risk is important to adjust the duration and intensity of one's training sessions. Sports and high-impact activities can lead to cartilage injuries and damage to the joints. Your risk of injury is calculated based on genetic variations that are associated with joint problems.

Your genetic map

Gene	Genotype
GNL3	GG
FTO	CC
SUPT3H	AG
IL1A	AG

What do your genetics tell us? 

Your risk of joint damage is greater than average.

More information:

<https://www.ncbi.nlm.nih.gov/pubmed/22763110>

Injury Risk

Risk of overload fracture

Overload fractures are small cracks in the bone caused by repetitive force or repetitive movements, such as running long distances or jumping repeatedly. They can also be caused by normal use of a weakened bone.

Anyone can suffer an overload fracture, but some people have a greater predisposition, which is associated with lower bone density. Overload fractures are a common injury in athletes, and affect up to 20% of athletes, particularly women. The main factor affecting one's risk to overload fractures is bone density, which has a genetic component (up to 85% of the variability is explained by genetic variations).

Using information from various genetic variants, the risk of overload fractures is estimated. Some variations increase risk while playing a protective role.

Your genetic map

Gene	Genotype
FUBP3	GG
RIN3	CC
C17ORF53	AC
MEPE	GG
ZBTB40	GG

What do your genetics tell us?

You are at a high risk of stress fractures.

More information:

<https://www.ncbi.nlm.nih.gov/pubmed/24945404>



Cardiovascular profile

Response of blood pressure to sports

High blood pressure, known as hypertension, is a common health issue. It is estimated that most people will have hypertension at some point in their lives.

Exercise has been shown to lower blood pressure. In fact, aerobic training is generally recommended as a therapy to prevent, treat, and control hypertension. An hour and a half of low-intensity aerobic exercise per week helps to lower blood pressure. There is great variability in the inter-individual response to the antihypertensive effects of exercise, and much of this variation is explained by genetic predisposition.

People more prone to controlling their hypertension see their blood pressure drop more quickly than the average person. For these people the benefits of 30 minutes of exercise a day are more noticeable than for the general population.

Your genetic map

Gene	Genotype
EDN1	TG
NOS3	GG
GNAS	TC
ADD1	TG

What do your genetics tell us?

Your blood pressure decreases faster through exercise than in the rest of the population.

More information:

<https://www.ncbi.nlm.nih.gov/pubmed/17938376>

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