# 24Genetics

# Mike, this is your sports test

CKC

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

Here are some essential aspects to take into account before reading this report.

#### The process by which we obtain your personalized report

The process we have followed to make your sports report consists of the following:

- **Extracting the DNA** from the saliva sample you sent us.

- Transforming the biological data contained in the DNA into computerized data. This process is called **sequencing.** 

- Applying **the algorithms** developed exclusively by 24Genetics to this computer data, which allows us to obtain your personalized report.

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

#### What is our algorithm like?

24Genetics algorithms are based on **the analysis and study of thousands of reports** (called "papers" in the scientific environment), contrasted, validated and recognized by the scientific community at an international level and that add value to our reports. In addition, we only apply to each individual the studies related to the population analyzed in them, i.e., we do not use a survey that has been carried out only with an Asian population for a European person, for example.

The most straightforward case, as a differential value of the 24Genetics reports compared to others, is that the first filter applied by our algorithm is sex. We classify genotypes into males and females. In this way, we only use the appropriate studies for each sex, avoiding applying a survey that only analyzed a male population sample to females.

In this way, 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). It is a study in which the DNA markers in the whole genome (the complete genetic material of a person) of people with a disease or trait are compared with those of people who do not have that disease or feature. It is a statistically based study, which considers a large number of genes associated with a predisposition in a less direct way but whose sum provides a relevant conclusion.

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

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

Each trait analyzed 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 we are learning more about the modifications we are analyzing today. At 24Genetics we make a great effort to apply new and consolidated scientific findings to our reports.

#### What information do we offer you?

The information provided by our reports speaks of **predispositions**. And what do we mean by that? Let's take an example. The possibility of suffering a bone fracture due to stress is influenced by multiple factors, which we could include in 2 groups: **genetic and environmental.** Genetic factors indicate our innate propensity to suffer a bone stress fracture.

On the other hand, environmental factors include elements that also have an effect, such as diet, habits, age, weight, etc. So, whether we ultimately suffer a fracture depends on the combination of both factors. And, even if we have a genetic predisposition, we may never suffer a stress fracture if we eat a diet rich in calcium and vitamin D, maintain a healthy weight, and practice sports with caution and without risks. Or vice versa.

Therefore, what our reports tell you are always genetic predispositions. By controlling environmental factors, we can help prevent these predispositions from developing.

#### What does this genetic test give me?

In this report, you have a lot of **scientifically validated information** about your **predispositions**. This lets you know **how your body works** naturally and what aspects you should pay attention to.

At 24Genetics, we recommend that you always consult a health or sports professional who can clarify your doubts, complement this report with your health history and available family history, supervise the follow-up of a personalized sports routine, or prescribe additional diagnostic tests if deemed necessary to confirm the risk of one or more specific predispositions.

#### A fundamental concept: the genetic variant.

In terms of genetic concepts, we want to share a basic one, which appears in all the features of our reports and is essential for you to understand at least briefly, that of genetic **genetic variant** (also called **variation**). The variant is a permanent change in the DNA sequence that forms a gene and is what marks an individual predisposition. Therefore, in each of the traits in this report, you will see information on the gene or genes affected in that trait. A variant in that gene or genes determines the different predispositions of some people versus others.

#### 1.1. Structure of this report

For ease of understanding, this report is organized into the following categories:

#### **Training and Abilities**

Training defines your athletic performance, and choosing a sport based on personal preferences is more than understandable and reasonable. Still, genetics tells you what your natural predispositions are, allowing you to develop training plans to help you exploit your capabilities or improve your deficiencies. In this category, you will be able to check whether you are more likely to perform better in power or endurance sports or what your overall innate strength is according to your DNA.

Practising sports correctly, safely and with caution will help you avoid physical damage. Still, you can also find out if you have a certain genetic predisposition to suffer specific injuries and thus be able to design sports routines to strengthen those parts of your anatomy. In this category, we tell you about it.

#### Biomarkers

Cholesterol levels, blood pressure or body mass index (BMI) and their relationship to the sport are influenced by your DNA, which determines your possible tendency to have additional advantages or risks than usual. Knowing yourself is the best tool to control your body. In this category, we inform you about your genetic predispositions so that you can use this information to your advantage.

#### Heart

The heart is the physiological centre of our organism and deserves special attention. Sport is intrinsically related to this organ, and preventing sudden death due to marks remains a medical challenge. Most causes are related to congenital or acquired cardiovascular diseases with no symptoms observed before the fatal event.

In this category, we include, among other data, information on your genetic predisposition to various cardiac pathologies that could lead to sudden death. However, this study does not analyze many other diseases and potential causes of premature death.

It should be borne in mind that the 24Genetics tests do not sequence the entire genome but analyze just over 700,000 markers of the 3.2 million genetic links that mark variability between individuals (99.9% of the genome is common to the human species). Therefore, not finding any mutation does not mean we are not carriers since the modification can be found in genetic areas not analyzed in our study. Likewise, we did not examine all the genetic information related to each disease studied. Specifically, we explored, on average, slightly less than 50% of the pathogenic markers reported for the pathologies or syndromes analyzed and associated with sudden death in the databases consulted so that mutations could exist in the other half and not be seen in this report. The modifications we are looking for are those reported in some of the most critical genetic databases worldwide, mainly OMIM and ClinVar.

It is essential to consider that, if you need to deepen the study of a specific disease, there are genetic tests that analyze the entire gene or genes involved in that disease and that have clinical validity. If you have a family history, we recommend you consult a physician or geneticist to study the need for such a test.

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

#### 1.2. Frequently Asked Questions

#### Do my genes determine everything?

Your genes are essential and determine the predisposition of your body's functioning, but many other factors influence it: lifestyle, exercise, and diet. In any case, knowing yourself well helps you to treat your body most appropriately. That is what genetics gives you: information and, consequently, knowledge.

#### Can there be more traits than those in my report?

Every day, new research is published worldwide, allowing us to expand our knowledge in the genetics field. 24Genetics constantly analyzes this research to incorporate it into our algorithm and improve it, to obtain 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 testing is the same. There are not too many biotechnology companies with the capacity to perform these complex analyses, and most give very poor conclusions regarding 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 uses. In no case is it suitable 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 organizations, in specific 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?

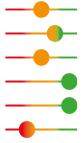
Not having a higher genetic predisposition is not a guarantee of not being injured. Usually, these studies are carried out based on statistical data from a sufficiently large sample of people. The genetic differences between people with a specific pathology and those without are observed. The type of conclusion typically reached is that people with a genetic alteration have a greater predisposition to suffer a particular pathology. However, this does not mean that 100% of people with that alteration will have that pathology. Likewise, it does not mean that 100% of the people who do not have this alteration cannot suffer from this pathology.

#### Examples of some of the 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.

# 2. Summary

#### Training and capabilities



- General strength
  Predisposition to power sports
  Aerobic capacity
  Skeletal muscle development
  Frequency of sport practice
  - Rowing

#### Risk of injury



- General risk of muscle injury due to sport Risk of a bone stress fracture
- Risk of Achilles tendon rupture Meniscus recovery after meniscus surgery
  - Concussion

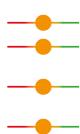
#### **Biomarkers**

- Influence of sport on body mass index (BMI)
  - Power of sport on glucose levels



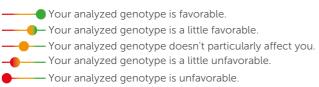
Sport and testosterone levels

#### Heart



- Resting heart rate
  Familial hypertrophic cardiomyopathy type I
  Familial isolated arrhythmogenic right ventricular dysplasia
  - Familial long QT syndrome

#### Caption:





Predisposition to endurance sports Muscle hypertrophy Personal motivation Endurance swimming



Risk of shoulder dislocation

Risk of anterior cruciate ligament rupture

Risk of meniscus tear

Sports-related skeletal muscle inflammation



- Impact levels
  - Impact of sport on cholesterol levels
  - Influence of resistance training on blood pressure
  - Oxidative stress



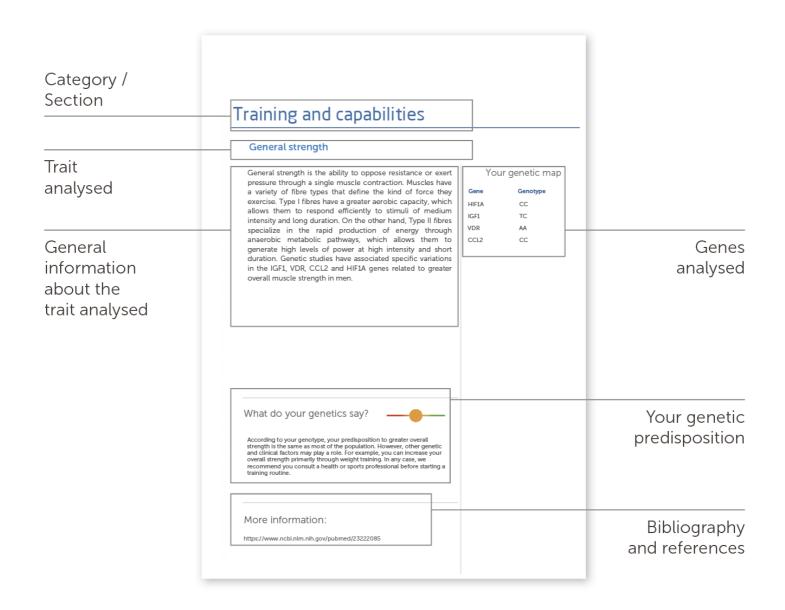
Alteration of cardiac structures

Familiar hypertrophic cardiomyopathy type II Brugada syndrome



# 3. Genetic Results

### 3.1. What do the results show?



### **General strength**

General strength is the ability to oppose resistance or exert pressure through a single muscle contraction. Muscles have a variety of fibre types that define the kind of force they exercise. Type I fibres have a greater aerobic capacity, which allows them to respond efficiently to stimuli of medium intensity and long duration. On the other hand, Type II fibres specialize in the rapid production of energy through anaerobic metabolic pathways, which allows them to generate high levels of power at high intensity and short duration. Genetic studies have associated specific variations in the IGF1, VDR, CCL2 and HIF1A genes related to greater overall muscle strength in men.

#### Your genetic map

Gene	Genotype
HIF1A	СС
IGF1	TC
VDR	AA
CCL2	СС

#### What do your genetics say?



According to your genotype, your predisposition to greater overall strength is the same as most of the population. However, other genetic and clinical factors may play a role. For example, you can increase your overall strength primarily through weight training. In any case, we recommend you consult a health or sports professional before starting a training routine.

More information:

### Flexibility

Flexibility is the ability of a muscle to passively lengthen through a range of motion and mobility. It could be defined as the ability to actively move a joint through its range of motion. Programs to improve joint mobility and flexibility are standard in clinical and physical sports settings. They generate multiple benefits, such as improved physical performance, reduced risk of injury or joint discomfort, improved posture and lower back pain. In addition, it helps to increase blood and nutrient flow to the tissues and, above all, improves muscle coordination and increases the range of mobility available in the joints.

At the genetic level, several studies show that people with mutations in the COL5A1 and ACTN3 genes tend to have less flexibility than individuals without those mutations.

#### Your genetic map

Gene	Genotype
COL5A1	ТТ
ACTN3	СС

#### What do your genetics say?



According to your genotype, your flexibility tends to be reduced. Other genetic and clinical factors may play a role. The flexibility program design should be personalized whenever possible, so we recommend that you seek the advice of a specialized personal trainer or physiotherapist.

More information:

https://pubmed.ncbi.nlm.nih.gov/21362053/

#### Predisposition to power sports

Power measures the speed at which force is applied. That is, it is an athlete's ability to exert force quickly, as opposed to endurance, which measures the ability to repeat an activity over a long period of time without feeling fatigue. Fast twitch fibers generate a high amount of force in a short time, but fatigue faster. They have a lower capacity to obtain aerobic energy and higher levels of glycogen, so, for muscle contraction, they get points through anaerobic respiration. Several studies have analyzed genetic markers associated with power sports and show that mutations in the AGTR2, MSTN, EPAS1, IGF2BP2AGT, IL6, AMPD1, ACE, VEGFA, ACVR1B and NOS3 genes, among others, correlate with a greater or lesser predisposition to achieve a good performance in power sports in men.

#### Your genetic map

Gene	Genotype
ACE	AG
ACVR1B	GG
AGT	GG
AGTR2	AA
AMPD1	GG
EPAS1	AG
IGF2BP2	TG
IL6	СС
MSTN	TT
NOS3	ТТ
VEGFA	GG

#### What do your genetics say?



You are highly predisposed to perform well in power sports, according to your genotype. Other genetic and clinical factors may play a role. Regardless of your genetic predisposition, power can be trained with different types of exercise. We recommend you consult a health or sports professional before starting a training routine.

#### More information:

#### Predisposition to endurance sports

Endurance measures the ability to repeat an activity without feeling fatigued. Therefore, endurance training is defined as a high or low-intensity activity performed for a prolonged period, as opposed to power training, in which the action is very high intensity for a short period. Endurance is greatly influenced by the proportion of slow twitch fibres in skeletal muscle, known as red fibres because they contain more myoglobin. This protein stores oxygen, obtaining its energy source and allowing it to maintain its strength for longer. In addition, studies have associated a specific variant in the PPARA gene with a predisposition to better performance in endurance sports in men.

#### Your genetic map

**Gene** PPARA

CG

Genotype

What do your genetics say?



According to your genotype, you are predisposed to perform well in endurance sports. Other genetic and clinical factors may play a role. Training such as slow long-distance running, cycling or swimming, yoga, sit-ups, squats or push-ups will improve your endurance. However, we recommend you consult a health or sports professional before starting a training routine.

More information:

https://pubmed.ncbi.nlm.nih.gov/22983821/

#### **Aerobic capacity**

Maximal aerobic capacity (or maximal volume of oxygen -VO2 max) is the maximum amount of oxygen that the body can absorb, transport and use per unit of time and, consequently, determines a person's aerobic fitness and power during prolonged exercise. Aerobic capacity can be improved through sports training to achieve a higher maximal oxygen volume, leading to low blood pressure, low cholesterol levels, and lower risk of obesity, type 2 diabetes and cardiovascular disease. Conversely, a low flow of oxygen to our cells explains possible shortness of breath, decreased endurance and increased susceptibility to respiratory infections with age. In addition, studies show that mutations in the ACSL1 gene correlate with poorer trainability of aerobic capacity.

#### Your genetic map

GG

Genotype

Gene ACSL1

What do your genetics say?



According to your genotype, your predisposition to improve your aerobic capacity through sport is normal. However, other genetic and clinical factors may play a role. Cross-training, combining several sports, is an excellent way to train aerobic capacity.

### More information:

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3098655/

### **Muscle hypertrophy**

Muscle hypertrophy is the scientific name given to the growth of muscle cell size, which implies an increase in fibre size and thus muscle development. Almost any type of sports training usually involves muscle development to a greater or lesser extent. Still, sometimes hypertrophy is a goal in itself for some athletes, in which case specific routines are designed for that purpose, based mainly on strength training. Some benefits of muscle hypertrophy are increased strength and endurance, joint strengthening, and injury prevention.

One study shows that individuals with mutations in the PPARG gene tend to obtain more significant muscle hypertrophy, i.e., increased muscle development, due to sports training.

#### Your genetic map

PPARG

Gene

СС

Genotype

#### What do your genetics say?



According to your genotype, your predisposition to obtain muscular hypertrophy due to the practice of sport is normal. However, other genetic and clinical factors may have an influence. For example, training is usually based on weight lifting to achieve muscle hypertrophy. The so-called "training to failure" system is standard among other routines, combining repetitions and weight until the last repetition cannot be performed entirely.

#### More information:

https://pubmed.ncbi.nlm.nih.gov/19526109/

#### Skeletal muscle development

There are three types of muscles: skeletal, cardiac and smooth. Skeletal muscles are striated muscles attached to the skeleton, formed by cells or elongated fibres. Their function is to facilitate movement and maintain the bonejoint union through their contraction. The biceps, pectorals or quadriceps are examples of skeletal muscles. Strength is influenced, among other factors, by muscle volume, which can be increased by training through functional exercises, repetitions with weight load and an adequate diet. At the genetic level, a study has found that a genetic variant in the BMP2 gene is associated with a better predisposition to skeletal muscle development due to endurance sports.

#### Your genetic map

Gene BMP2

CC

Genotype

What do your genetics say?



According to your genotype, you have a good predisposition to skeletal muscle development, thanks to endurance sports. However, other genetic and clinical factors may play a role. For example, a proper diet is essential in addition to sports to achieve muscle growth goals. Therefore, we recommend you share your genetic reports with health and sports professionals (doctor, nutritionist, personal trainer) to reach your maximum potential safely.

### More information:

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

#### **Personal motivation**

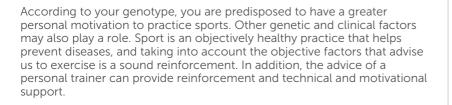
When we undertake any task or project, there can be two motivations. Rational motivation is the set of objective reasons why it is positive to carry out such a project. On the other hand, personal or intrinsic motivation is the set of psychological forces that lead us to undertake such action. It arises from within the individual and is usually a reliable indicator of our possibilities to carry it out. In sports, both types of motivations have an influence. For example, we can exercise because we are overweight or have high blood pressure, and we can do it because our body asks us to, without any apparent objective reasons.

In the case of personal motivation, genetics has a lot to say. For example, several studies show that variations in the BDNF and VDR genes can influence our motivation to practice sport.

#### Your genetic map

Gene	Genotype
BDNF	TC
VDR	AA

#### What do your genetics say?



#### More information:

https://pubmed.ncbi.nlm.nih.gov/24805993/

#### **Frequency of sport practice**

By training frequency, we mean the number of sports sessions performed, or the number of times a specific muscle group is exercised, during a given period of time, (usually, a whole week). But, leaving aside technical definitions, we can say that frequency in sport is the periodicity with which we carry out this activity and is directly related to the achievement of the objectives set, together with other factors, such as the type of training, the intensity, the duration of the sessions, etc.

Genetics can influence our predisposition to exercise more frequently. In this regard, a study shows that a mutation in the GCKR gene is directly related to a greater tendency to practice sports more assiduously.

#### Your genetic map

TC

Genotype

Gene GCKR

What do your genetics say?



According to your genotype, you are predisposed to practice sports frequently. However, other genetic and clinical factors may play a role. Regardless of your genetic predisposition, we recommend you consult a personal trainer, who can define your personalized routine with the optimal training, intensity, frequency and rest times according to your goals and physical condition.

### More information:

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6722860/pdf/genes-10 -00570.pdf

#### **Endurance swimming**

Swimming is a sport that mainly exercises the latissimus dorsi, pectoralis, deltoid, triceps, biceps, teres major and trapezius muscles, and, secondarily, the abdominals, gluteus, intercostals, and quadriceps. On a cardiorespiratory level, it improves oxygen consumption by up to 10%. In addition, it allows the heart to pump up to 18% more blood, resulting in better circulation and reducing the heart rate. It is also one of the sports that helps to improve coordination. There are four basic swimming styles (crawl, breaststroke, backstroke, and butterfly), each with its specific technique and level of difficulty, as well as its muscular and skeletal indications so that it is a sport that can be adapted to the particular needs of each person.

In genetics, a specific mutation in the NOS3 gene has been correlated with a predisposition to a better performance in endurance swimming.

#### Your genetic map

Gene NOS3

ТТ

Genotype

#### What do your genetics say?

Depending on your genotype, you are predisposed to perform well in endurance swimming. Other genetic and clinical factors may play a role. We can say that swimming is a sport recommended for almost any type of person, as it generates a little impact on bones and joints, improves flexibility and elasticity, and burns fat. It can also help to manage stress and anxiety.

#### More information:

https://pubmed.ncbi.nlm.nih.gov/30765915/

#### Rowing

Rowing is a sport discipline that consists of propelling a boat over the water using the muscular strength of one or more rowers, each using one or two oars as levers. At body level, the benefits of rowing are strengthening of the musculature, reduction of joint stiffness, elimination of body fat and improvement of cardiovascular endurance. But, at a mental level, it also helps release endorphins. The muscle groups most exercised with rowing are the cervical musculature, shoulder, back and arm, although indirectly also work the abdomen, buttocks and legs. In general, it helps build muscle and increases strength and endurance simultaneously.

At the genetic level, it has been shown that mutations in the ACTN3 gene are correlated with better rowing performance.

#### Your genetic map

ACTN3

Gene

CC

Genotype

#### What do your genetics say?



You are not predisposed to perform well in rowing, depending on your genotype. Other genetic and clinical factors may play a role.

#### More information:

https://pubmed.ncbi.nlm.nih.gov/25268288/

### Sprinting

Sport sprinting is the increase in exercise speed over a period of time, usually short. It is also known as doubling, because it is estimated to consist of doubling the energy level during that time interval. It should be noted that, like any form of exercise, it requires prior preparation in which the body becomes accustomed to these changes in intensity. Sprinting has many benefits: it improves reaction capacity and increases bone density and sports performance in general. Studies have associated specific variations in the AMPD1 and ACVR1B genes with the level of sprinting performance.

#### Your genetic map

Gene	Genotype
AMPD1	GG
ACVR1B	GG

#### What do your genetics say?



According to your genotype, your predisposition to correct sprint performance is average. However, other genetic and clinical factors may have an influence. Therefore, before you start sprinting, it is advisable to seek the advice of a doctor and a sports specialist to help you define your goals and burst times in a personalized and controlled manner.

More information:

#### General risk of muscle injury due to sport

Sport has numerous health benefits, but any physical activity carries a risk of injury from the activity itself and the possibility of performing the exercises incorrectly. To avoid the latter, having a sports or health professional supervise the correct development of the sports routine is essential. When a muscle is injured, proteins are released into the blood, the concentration of which is evidence of more significant damage to the muscle fibres and a greater likelihood of muscle fatigue. However, some people have a greater predisposition to injury than others, and this, in part, is due to their genetics. Scientific evidence has shown that specific variation in the IL6 gene correlates with the risk of injury.

#### Your genetic map

CC

Genotype

Gene

IL6

#### What do your genetics say?



According to your genotype, you have a high predisposition to suffer muscle injuries due to sports. Other genetic and clinical factors may also play a role. In addition to trying to avoid falls or using protective equipment, if you do certain activities, such as risky or contact sports, it is advisable to do physical exercise to strengthen and maintain the flexibility of your joints and muscles.

More information:

https://pubmed.ncbi.nlm.nih.gov/18758806/

### **Risk of shoulder dislocation**

Shoulder dislocation occurs when the head of the humerus is displaced out of the joint. It is usually caused by trauma due to accidents, falls or seizures, among other reasons. It is a relatively common injury; approximately 1.7% of the population suffers it at some point. Recent genetic studies link mutations in the COL1A1 gene with a lower risk of shoulder dislocation. Your genetic map

Genotype

Gene

What do your genetics say?

According to your genotype, your predisposition to shoulder dislocation is normal. However, other genetic and clinical factors may play a role. In addition to trying to avoid falls or using protective equipment, if you do certain activities, such as risky or contact sports, it is advisable to do physical exercise to strengthen and maintain the flexibility of your joints and muscles.

More information:

https://bjsm.bmj.com/content/44/14/1063.long





This report is not valid for clinical or diagnostic use

#### **Risk of a bone stress fracture**

Bone mineral density (BMD), also called bone density or bone mass, is the most widely used indicator to assess the risk of a stress fracture. Stress fractures are small cracks in the bone caused by repetitive application of force, repetitive motion or regular use of a weakened bone. Some people have a greater predisposition to stress fractures associated with lower bone density, with a genetic component of up to 85%. The most advanced case of low bone density is osteoporosis, a disease that affects more in older age and especially women after menopause. In addition, studies have associated specific variations in the FAM210A and C18orf19 genes, among many others, with the risk of bone stress fractures.

### What do your genetics say?



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	Genotype
I	FABP3P2	ТС
-	ZNF408;	ТС
/	AXIN1	ТТ
-	TMEM263	ТТ
ł	RPS3AP2	GG
ł	HROB	AA
I	FAM210A	GG
(	CCDC170	ТТ
(	CPED1	AG
(	CBR1 AS1	AC
(	CPN1	ТС
l	LOC10537704	СС
l	LOC10798396	GG
[	DCDC1	TT
I	RHEBL1 DHH	ТС
[	DNM3	TG
l	LOC10798450	AA
I	FOXL1	AG
I	FUBP3	СС
(	CSRNP3	AG
(	GPATCH1	СС
ł	HOXC6;	CG
I	DUA	AG
l	LOC10537357	GG
,	JAG1	TT
ł	KCNMA1	СС
l	USF3	GG
l	LOC10536970	TT
l	LEKR1	TT
ł	RPL37AP7	СС
l	LRP5	СС

#### Risk of anterior cruciate ligament rupture

The anterior cruciate ligament connects the posterior-lateral part of the femur with the anterior-middle part of the tibia, passing behind the patella. This junction prevents a forward displacement of the tibia with respect to the femur, while the posterior cruciate ligament prevents backward а displacement of the tibia with respect to the femur. The combination of the two provides rotational stability to the knee. Ruptures of the anterior cruciate ligament are frequent during physical activities. Recent genetic studies point to mutations in the COL1A1 gene as a protective factor against injuries to this ligament.

#### Your genetic map

СС

Genotype

COL1A1

Gene

What do your genetics say?



According to your genotype, your predisposition to anterior cruciate ligament rupture is normal. However, other genetic and clinical factors may play a role. In addition to avoiding falls or using protective equipment, if you do certain activities, such as risky or contact sports, it is advisable to do physical exercise to strengthen and maintain the flexibility of your joints and muscles.

More information:

https://bjsm.bmj.com/content/44/14/1063.long

### **Risk of Achilles tendon rupture**

The Achilles tendon is a strong band of fibrous tissue that connects the muscles in the back of your calf to the heel bone, and its function is plantar flexion of the foot. It allows the calf muscle to move the foot, making it essential for walking, running and jumping. Abnormalities of the tendon can include inflammation, degeneration and rupture. Recent genetic studies point to mutations in the COL1A1 gene as a protective factor against Achilles tendon injury.

#### Your genetic map

COL1A1

Gene

СС

Genotype

### What do your genetics say?



According to your genotype, your predisposition to Achilles tendon rupture is normal. However, other genetic and clinical factors may play a role. In addition to trying to avoid falls or using protective equipment, if you do certain activities, such as risky or contact sports, it is advisable to engage in physical activity to strengthen and maintain the flexibility of your joints and muscles.

More information:

https://bjsm.bmj.com/content/44/14/1063.long

#### **Risk of meniscus tear**

Cartilage is a strong but flexible tissue that cushions between the ends of bones in a joint. The meniscus is a C-shaped piece of cartilage, of which there are two in each knee. A torn meniscus usually causes medial or lateral pain depending on whether it is internal or external, although sometimes it can also cause pain in the back of the knee. In addition to pain, meniscal tears can cause a sensation of the knee giving way, inability to move the knee normally, and increased susceptibility to osteoarthritis in the injured joint. Studies have associated a particular variation in the GDF5 gene with the risk of meniscal tears in men.

#### 24Genetics Sports Genetic Test of Mike. 356548

#### Your genetic map

AA

Genotype

Gene	
GDF5	

What do your genetics say?



According to your genotype, you are predisposed to meniscus tears. Other genetic and clinical factors may play a role. In addition to trying to avoid falls or using protective equipment, if you do certain activities, such as risky or contact sports, it is advisable to do physical exercise to strengthen and maintain the flexibility of your joints and muscles.

More information:

#### Meniscus recovery after meniscus surgery

The treatment of a meniscal tear usually depends on the disability that the injury causes to the patient, among other factors. It is generally conservative in degenerative lesions of this age, but it is often necessary to go to the operating room. Meniscus surgery is usually performed by arthroscopy, using a camera inserted through a small incision in the joint, which allows the problem to be identified and solved from the inside. The most common techniques are meniscal suture and partial removal (partial meniscectomy). The postoperative period depends on many factors, such as the result of the surgery, the previous state of the joint, the patient's physical condition, age, etc. But genetics also has something to say since studies associate a specific variation in the GDF5 gene with better recovery and more excellent knee stability after meniscus surgery.

#### Your genetic map

AA

Genotype

Gene GDF5

What do your genetics say?



According to your genotype, you have a normal predisposition to make a good recovery after meniscus surgery. However, other genetic and clinical factors may play a role. Therefore, healing after trauma surgery should always be supervised by a healthcare professional, rehabilitation physician or physiotherapist.

More information:

#### Sports-related skeletal muscle inflammation

There are three types of muscles: skeletal, cardiac and smooth. Skeletal muscles are striated muscles attached to the skeleton, formed by elongated cells or fibres. Their function is to facilitate movement and maintain the bonejoint union through their contraction. Muscle inflammation related to sports can have different causes, such as overexertion, strains, blows, prolonged tension, and sports practice accidents, which are frequently associated with incorrect routines and physical performance. But genetics is also an influential factor, as demonstrated by a study that shows the correlation of a mutation in the IL1B gene with the risk of suffering skeletal muscle inflammation after endurance sports.

#### Your genetic map

AG

Genotype

Gene

IL1B

### What do your genetics say?



Based on your genotype, your predisposition to skeletal muscle inflammation from endurance sports is normal. However, other genetic and clinical factors may play a role. We recommend that you strengthen and maintain flexible joints and muscles. You can also see your genetic predisposition to endurance sports in another feature of this report.

#### More information:

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

#### Concussion

A concussion, also called a traumatic brain injury, can occur when the head hits an object or when a moving object strikes the head. This type of injury can cause headaches, changes in alertness, loss of consciousness (rare), or memory loss. It can also affect how the brain functions, and depending on the severity of the trauma, the extent of the injury and its duration, the consequences can be more or less significant. Sports activities are a common cause of concussion, which a physician should diagnose.

Several studies have shown that mutations in the IL6R and APOE genes are correlated with a greater or lesser likelihood of concussion.

#### Your genetic map

Gene	Genotype
IL6R	AC
APOE	TG

#### What do your genetics say?



According to your genotype, you have a low predisposition to concussion. Other genetic and clinical factors may play a role. Since a concussion is caused by trauma, it is a complex injury to prevent. Helmets, mouthguards and other safety items can reduce the risk of head injury, especially in high-risk sports where the trauma may be more severe.

#### More information:

https://bjsm.bmj.com/content/bjsports/52/3/192.full.pdf? ijkey=ZKH90hYIAcypOJa&keytype=ref

#### Cramps

Muscle cramps are sudden involuntary contractions or spasms in one or more muscles. They are relatively frequent and usually occur after exercise. The most typical case is muscle cramps in the legs at night. They can be very painful and last from a few seconds to several minutes. Dehydration; low levels of electrolytes, such as magnesium, potassium or calcium; muscles with poor blood supply; pregnancy; or certain medications are some of the causes, although the main reason is usually excessive tension of a specific muscle. If you have a cramp, it is essential to stop exercising, gently stretch the muscle, drink water, walk a little and relieve the pain with ice, if necessary.

On a genetic level, a study shows that people with a particular mutation in the COL5A1 gene are more prone to muscle cramps than people without the modification.

#### Your genetic map

COL5A1

Gene

TT

Genotype

What do your genetics say?



According to your genotype, you are not particularly predisposed to muscle cramps. However, other genetic and clinical factors may play a role. To avoid cramps, it is recommended to stretch your muscles before exercising and drink plenty of fluids to stay hydrated. If cramps occur at night, it is advisable to stretch before going to bed. If you have frequent cramps, we recommend you visit your doctor or physiotherapist.

More information:

https://pubmed.ncbi.nlm.nih.gov/22894972/

#### Influence of sport on body mass index (BMI)

Body mass index (BMI) is a number that is calculated based on a person's weight and height and is used to identify weight ranges that can lead to health problems. A healthy weight is considered between 18.5 and 24.9 BMI; overweight corresponds to the range between 25 and 29.9; and over 30 is considered obese, from moderate to severe. Physical activity is beneficial for all people, regardless of their genetics. Still, studies show that a specific variant of the FTO gene has a direct relationship with a more significant benefit of the practice of sport in reducing BMI.

#### Your genetic map

Gene	Genotype
FTO	GG
FTO	AC

#### What do your genetics say?



According to your genotype, your predisposition to reduce your BMI by practising sports is normal. However, other genetic and clinical factors may have an influence. In addition to a correct sports routine, a balanced diet can help you reduce your weight and improve your health and wellbeing, always under the supervision of a health professional. You can also see your predisposition to the greater or lesser effectiveness of different types of diet in our nutrigenetic report.

#### More information:

#### Impact of sport on cholesterol levels

Cholesterol is a waxy, fat-like substance found in every cell in our bodies. It is needed to produce hormones, vitamin D and substances that help digest food, but abnormally high levels can lead to an increased risk of heart disease, stroke and other problems. Research shows that one benefit of exercise is improving cholesterol levels by increasing the size of the protein particles that carry cholesterol through the blood, reducing the chance of smaller protein particles clogging arteries. It also stimulates enzymes that help move lousy cholesterol from the blood to the liver, allowing it to be excreted. In addition, specific variants in the LIPC gene have been correlated with better regulation of cholesterol levels by sports training in men.

#### Your genetic map

CC

Genotype

Gene

What do your genetics say?



According to your genotype, you are predisposed to better-regulating cholesterol levels through sport. However, other genetic and clinical factors may also play a role. In addition to practising sports, it is advisable to consume skimmed dairy products, replace butter with olive oil, fatty meats with lean meats, and avoid the consumption of cold meats, fried foods and industrial pastries. Baking, steaming, grilling or broiling are the most advisable.

#### More information:

#### Power of sport on glucose levels

Insulin is a hormone generated by the pancreas, which controls glucose levels in two ways: by using it as a source of energy, mainly by the muscle and by regulating its production by the liver. This function is "insulin sensitivity". When insulin does not perform this function well, it is called "insulin resistance", and glucose accumulates in the blood, producing hyperglycemia, which can lead to obesity and type II diabetes. Sport involves increased energy expenditure, which positively influences glucose levels. According to genetics, specific variants of the HNF4A and LIPC genes show a predisposition to obtain adequate insulin and glucose levels, thanks to the practice of sports regularly.

#### Your genetic map

Gene	Genotype
HNF4A	GG
LIPC	СС

#### What do your genetics say?



According to your genotype, you are predisposed to obtain adequate insulin and glucose levels through regular sports practice. Other genetic and clinical factors may play a role. A decrease in weight, under the supervision of a physician or nutritionist, with particular emphasis on reducing the consumption of sweets, bread, pasta and other carbohydrates, can help to achieve better insulin control.

More information:

#### Influence of resistance training on blood pressure

Blood pressure is the force exerted by the blood against the walls of the blood vessels throughout the circulatory system. Arterial hypertension is a chronic condition in which the blood vessels have persistently high blood pressure, which can damage them. It is estimated that most of the population will have hypertension at some point. The practice of endurance sports (running, swimming, cycling, prolonged sessions on the elliptical bike or rowing machine, among others) can help reduce blood pressure. Still, this influence depends very much on the individual, and genetics plays an important role. Several studies have associated specific variations in the NOS3 gene with an antihypertensive effect in particular genotypes.

#### Your genetic map

AG

Genotype

Gene

#### What do your genetics say?



According to your genotype, your predisposition to lower blood pressure through resistance training is typical. Other genetic and clinical factors may play a role. Besides sports, healthy lifestyle habits to prevent hypertension are well known to the population: maintaining an adequate BMI, reducing sodium in our diet, limiting alcohol, tobacco and caffeine consumption, and reducing stress levels.

#### More information:

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2714087/

#### Sport and testosterone levels

Testosterone is the primary sex hormone in men. During puberty, it causes body hair growth, muscle development, and deepening of the voice. In adult men, it controls sexual desire, helps produce sperm and maintains muscle mass. As for the relationship of testosterone with sport, it is worth noting that it has an essential role in regulating metabolism, whose proper functioning is vital for weight control, maintaining sufficient energy levels throughout the day, or having a good mood, among other functions. In particular, testosterone plays a significant role in making muscle fibres grow and become stronger. And since muscle is the most energy-consuming tissue in our body, metabolic activity improves when testosterone levels are adequate.

A study shows that mutations in the SHBG gene are correlated with the tendency to have low testosterone levels.

#### Your genetic map

Gene	Genotype
SHBG	GG
SHBG	СС

#### What do your genetics say?



According to this study, you have a predisposition similar to most of the population to have low levels. Other genetic and clinical factors may play a role. To maintain adequate testosterone levels, it is advisable to maintain a healthy weight; practice sports regularly; do not consume too much caffeine, tobacco or alcohol; sleep well; and avoid stress. A doctor can prescribe vitamin supplements or particular medication if deemed necessary.

#### More information:

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3188559/

#### **Oxidative stress**

Oxidative stress is the process by which our body's cells become oxidized, resulting in damage that affects their functions. It occurs due to an excess of free radicals and oxygen and a lack of antioxidants to counteract it. Environmental pollution, active and passive smoking, excessive sunbathing, and consuming drugs or too much alcohol are some leading causes. But during the practice of sport, free radicals and the consequent oxidative stress are also increased, which can be damaging in the case of a weakened antioxidant defence system. Its consequences are usually excessive muscle pain and fatigue after training. Genetics is an influential factor in this regard, and a study

shows that a particular variant of the SOD2 gene reduces the efficiency of the organism in its defence against oxidative stress caused by sport, which could be unfavourable in the practice of sports, especially high-intensity sports.

#### Your genetic map

Gene SOD2 **Genotype** GG

What do your genetics say?



According to your genotype, you are not particularly predisposed to suffer oxidative stress due to sport. However, other genetic and clinical factors may play a role. Broccoli, blueberries, carrots, tomatoes, cocoa, peppers, almonds, green tea, red wine in moderation, and some vegetable oils, such as olive oil, are antioxidant foods. Other micronutrients, such as selenium, beta-carotene and vitamins C and D, also have this effect.

### More information:

https://www.ncbi.nlm.nih.gov/pubmed/24865797?dopt=Abstract

#### **Resting heart rate**

The adult heart usually beats between 60 and 100 times per minute at rest. When the heart beats below 60 times per minute, it is bradycardia, which can be a severe problem if the heart rate is too slow and the heart cannot pump enough oxygen-rich blood to the body. A heart rate above 100 beats/min at rest is called tachycardia and is associated with an increased risk of cardiovascular disease. In terms of sports practice, cardiovascular or aerobic exercises such as running, swimming or cycling increase heart rate to a greater extent. In contrast, isometric activities, such as weight lifting, induce a minor increase in heart rate. Several studies have associated specific variations in the FADS1 and CD46 genes, among many others, with abnormal heart rates in particular genotypes.

### What do your genetics say?



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/23583979

#### Your genetic map

Gene	Genotype
TFPI	GG
LOC10537540	AA
RNU3P3	TT
SYT10	AC
LOC10536969	AA
CD46	TT
MYH6	AA
LOC10537797	СС
ACHE	AG
FADS1	AG
SLC35F1	TT
KIAA1755	TT
CCDC141	GG
GNB4	TG
CHRM2	TT
NKX2 5	AA
LOC10537392	AA
FNDC3B	GG
RFX4	ТТ
CPNE8	ТС
RBFOX1	GG
SLC10A7	GG
RNU4 35P	ТС
LOC10798525	AG
HMGN2P29	GG
LOC10192800	AA

#### Alteration of cardiac structures

The left ventricle's morphological structure and the aortic root's size are inherited traits that, if altered, can cause cardiovascular disease. The practice of endurance sports entails an overload of pressure and volume on all cardiac structures. During exercise, the left ventricle increases its contractile capacity in proportion to the demand for cardiac output, increasing cardiovascular risk in the case of altered structures. lt is, therefore, essential to take echocardiographic measurements into account. Possible complications may include heart failure, heart attack, stroke, or aneurysm. Studies show that mutations in the SMG6 and LOXL1 genes, among many others, correlate with a greater predisposition to suffer alterations in cardiac structures.

#### Your genetic map

Gene	Genotype
SLC35F1	GG
TMEM232	ТС
SMG6	GG
PRDM6	GG
HMGA2	TT
LINC02398	AA
LOXL1	GG

### What do your genetics say?



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

#### Familial hypertrophic cardiomyopathy type I

Familial hypertrophic cardiomyopathy type I is an inherited coronary artery disease characterized by a thickening of the heart muscle, which typically occurs in the anterior part of the wall separating the left and right ventricle (interventricular septum). This thickening is called left ventricular hypertrophy (LVH). The clinical manifestations of this disease range from asymptomatic LVH to sudden cardiac death, including arrhythmias or atrial fibrillation, and it develops mainly during adolescence or adulthood. However, it can also occur in childhood and senescence. Studies show that mutations in the MYH7 and MYBPC3 genes correlate with an increased predisposition to familial hypertrophic type I heart disease.

#### Your genetic map

Gene	Genotype
MYBPC3	DD
MYBPC3	СС
MYH7	СС
MYH7	GG
MYH7	II
MYH7	TT
MYH7	AA
MYBPC3	GG

### What do your genetics say?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

#### More information:

https://www.omim.org/entry/192600

#### Familiar hypertrophic cardiomyopathy type II

Type II familial hypertrophic heart disease is an inherited coronary artery disease characterized by a thickening of the heart muscle, which typically occurs in the wall that separates the left and right ventricle (interventricular septum). The difference with type I is morphological since type II affects the entire interventricular septum and not only the anterior part. This thickening is called left ventricular hypertrophy (LVH). The clinical manifestations of this disease range from asymptomatic LVH to sudden cardiac death, including arrhythmias or atrial fibrillation, and it develops mainly during adolescence or adulthood. However, it can also occur in childhood and senescence. Studies show that mutations in the TNNT2 gene correlate with an increased predisposition to type I familial hypertrophic heart disease.

#### Your genetic map

Gene	Genotype
TNNT2	GG
TNNT2	СС
TNNT2	

### What do your genetics say?



We have not detected any pathogenic mutations, but you might have some in non-analysed genetic regions.

#### More information:

https://www.omim.org/entry/115195

#### 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.

#### Your genetic map

Gene	Genotype
DSP	СС
DSP	GG

#### What do your genetics say?



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

#### More information:

http://www.orpha.net/consor/cgi-bin/OC\_Exp.php?lng=en&Expert=217656

### 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.

#### Your genetic map

Gene	Genotype
FBN1 DT	СС
SCN5A	СС
SCN5A	GG
SCN5A	TT
SCN5A	AA

### What do your genetics say?



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

### More information:

http://www.orpha.net/consor/cgi-bin/OC\_Exp.php?lng=en&Expert=130

### Familial long QT syndrome

A rare group of genetic, cardiac rhythm diseases characterized by a prolongation of the QT interval at basal electrocardiography (ECG) and by a high risk of lifethreatening arrhythmias.

#### Your genetic map

GG

Genotype

Gene CACNA1C

What do your genetics say?



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

### More information:

http://www.orpha.net/consor/cgi-bin/OC\_Exp.php?lng=en&Expert=768

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