ID: 00000

OMGENO

Genetics for people

Genetic Testing
Sarcopenia

My Prevention

SARCOPENIA

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1- Information about Sarcopenia

Sarcopenia is a generalised and progressive musculoskeletal disorder that leads **to accelerated loss of muscle mass and function**. It is strongly associated with functional impairment, falls, frailty (increased risk of bone fractures) and mortality. It commonly occurs as an age-related process and is influenced by both genetic and lifestyle factors that occur throughout life. It can also occur in middle-aged people in association with a variety of conditions.

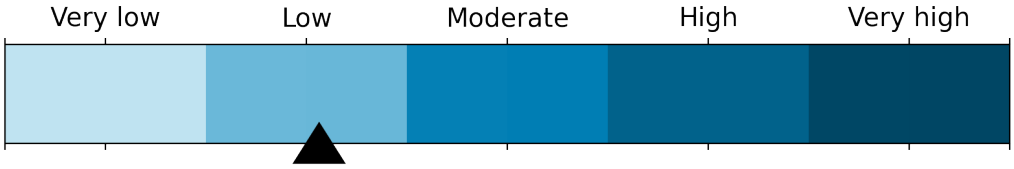
2- Genetic result

This section shows the distribution of your genetic profile as analysed for the *MySarcopenia* test. As a general rule, variants that do not influence the disease or cause undesirable effects, respectively, are classified as benign and malignant. As for heterozygotes, it should be emphasised that their effect is more complex to determine, since depending on the type of inheritance followed by the mutated allele, the effect may be different.

In the case of dominant inheritance, where a single mutated allele may be sufficient to manifest the disease, the effect of the mutated allele in the heterozygote may be evident. On the other hand, in recessive inheritance, where two copies of the mutated allele are needed to manifest the disease, the effect of the mutated allele in the heterozygote may be less obvious, but may still have an impact on the health of the individual. It is important to note that the results of this genetic analysis are a tool to help in the prevention and management of sarcopenia, but should always be interpreted in conjunction with other risk factors and under the supervision of a healthcare professional.

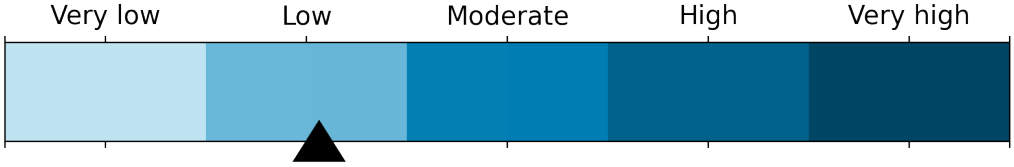
3- Your genetic predisposition

Below, we indicate your genetic predisposition to develop Sarcopenia:



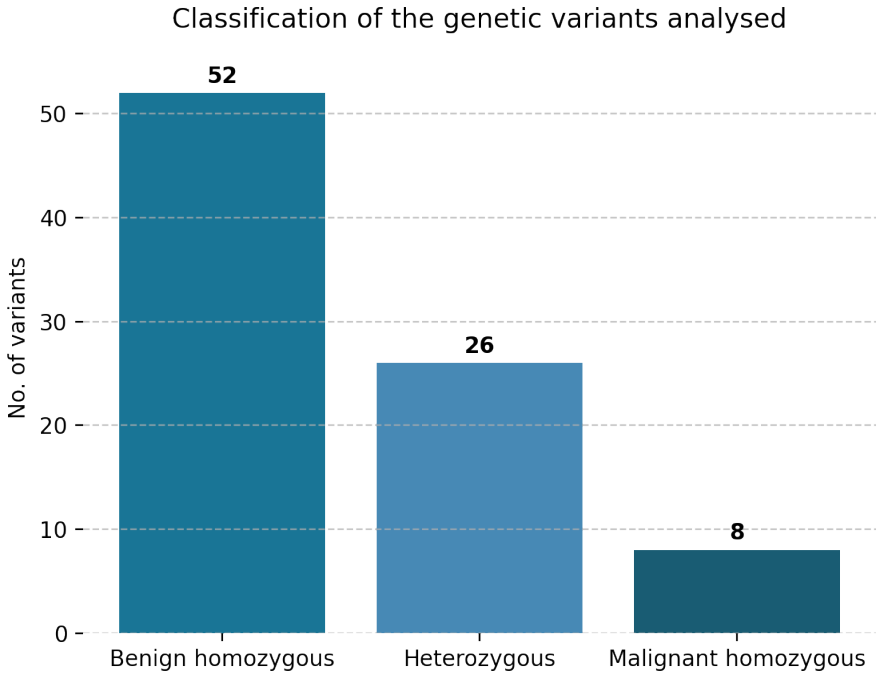
4- Your overall predisposition to develop sarcopenia considering the multiscale factor

Below, we indicate your overall predisposition to develop Sarcopenia based on your genetic predisposition and body mass index:



5- Your markers implicated in the risk of developing sarcopenia

Next, the impact of your genetic profile on your predisposition to develop the disease is shown.



6- Your conclusion

You have a **very low genetic predisposition** to develop sarcopenia, based on the genetic variants analysed. Therefore, according to your genetic results, it is **very unlikely that you will develop** sarcopenia.

However, the origin of sarcopenia is multifactorial and is closely associated with the natural ageing process. If you have any of the associated symptoms described in Annex I, it is recommended that you contact your specialist for more information and further tests to confirm the diagnosis, as well as to obtain the correct treatment guidelines.

Here are the **main factors that accelerate the loss of muscle mass** and the measures you can take to prevent it:

- Sedentary lifestyle: **regular physical activity** can reduce the chances of developing sarcopenia. This is one of the best prevention strategies. Getting at least half an hour of moderate exercise every day, such as walking, jogging or running, helps to keep the body active and has numerous health benefits.
- Unbalanced diet: a **balanced diet** with sufficient calories and protein prevents weight loss and the resulting reduction in muscle mass. To help prevent sarcopenia, experts recommend consuming 25-30 g of protein and omega-3 fatty acids. On the other hand, in the case of vitamin D deficiency, supplementation can increase muscle strength.

- Inflammatory state: sattention should be paid to **inflammatory processes** caused by chronic or long-term injury or illness. Studies link elevated blood levels of C-reactive protein (an indicator of inflammation) to sarcopenia.
- Stress: sarcopenia is more common when conditions that increase stress occur, such as when chronic diseases occur.

ANNEX 1: INFORMATION ABOUT SARCOPENIA

1- Common symptoms

The symptoms associated with sarcopenia are very diverse. However, most of them are related to loss of muscle mass and weakness. The most common symptoms are listed below:

SYMPTOMATOLOGY		
Muscular weakness	Difficulty in daily activities (walking, climbing stairs, etc.)	Fatigue Resistance loss
Lack of balance	Muscle size decrease	Loss of tissue elasticity

2- Links of interest

- National Institute of Health: nih.gov
- Spanish Federation of Rheumatology: inforeuma.com
  - Information Dossier on Sarcopenia: inforeuma.com
- Spanish Society of Rheumatology: ser.es
- Healthline Media (Symptoms, Causes, Treatment and Prevention): healthline.com
- Healthline Media (How to combat sarcopenia?): healthline.com

TECHNOLOGY

DNA Microarray technology consists of a solid surface with microscopic reactions (micro-reactions) or DNA chip, on which molecular probes are attached to detect the presence of target DNA molecules. Probe-target hybridization is usually detected and quantified by measuring the intensity of a specific fluorescence provided by the molecular probe in the samples. This type of technology allows the detection of thousands of specific DNA fragments present in a DNA sample. On the other hand, the specificity in terms of DNA sequence recognition is very high, since single nucleotide exchange (single base resolution) can be detected using short oligonucleotide probes (20-25 nucleotides). As a result, DNA Microarray technology has also evolved to be applied as a DNA sequencing technique to genotype several hundred thousand single nucleotide variants (SNVs) in target genes located throughout the genome (Whole Genome DNA Microarray).

QUALITY

The analytical laboratory has standard and effective procedures to protect against technical and operational problems. However, results can be altered due to problems with sample collection (contamination) and labeling (identification), delay in receiving the sample in the laboratory (integrity), among other problems. This could lead to invalidation of the test results. In such cases, the patient would be asked to repeat the entire testing process. As with all clinical screening tests, there is a small chance that the laboratory may report inaccurate information. If there is a suspicion of an error in the genotype detected, a verification test may be requested.

RISKS AND LIMITATIONS

The results presented in this report are limited to the scientific knowledge available at the time the test was developed. This test only detects the specified genetic variants, it does not detect other minority variants even if they are related to the pathologies. The recommendations described throughout this report of results are for guidance only, Overgenes cannot be held responsible for any possible misinterpretation of the results provided.

MySarcopenia is not a medical report. These results should **NOT** be interpreted as a diagnostic tool, nor do they indicate whether a person suffers from sarcopenia, they only inform about the genetic predisposition of each individual to develop it.

GLOSSARY

• **DNA:** abbreviation for deoxyribonucleic acid. A molecule present in our cells that contains the genetic information necessary for the development and proper functioning of living organisms.

• **Allele:** each of the alternative forms of a gene, which may have differences in their sequence.

• **Cell:** basic structural and functional unit of life.

• **Gene:** DNA segment representing the unit of hereditary information.

• **Heterozygous:** when the two alleles of the same gene are different.

• **Homozygous:** when the two alleles of the same gene are the same.

• **Phenotype:** the set of observable characteristics of an organism.

• **Genotype:** combination of the variants of a gene in an individual.

• **Haplotype:** a set of DNA variations, or polymorphisms, that tend to be inherited together.

• **Metabolism:** a set of chemical processes occurring within a cell or organism that serve to produce energy or use it as fuel.

• **Mutation:** nucleotide sequence variation in genes affecting less than 1% of the population.

• **Polymorphism:** nucleotide sequence variation in the genes affecting ≥1% the population.

• **SNP:** single nucleotide genetic polymorphism.

• **Genetic predisposition:** also known as genetic susceptibility. It is the increased likelihood of developing a certain condition or pathology due to the presence of one or more genetic variations.

Genetics for people

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