Limb-Girdle Muscular Dystrophy Type 2D

LGMD2D is a rare genetic disorder. It is characterized by muscle weakness that worsens over time as well as heart and lung problems. A person must have two variants in the SGCA gene in order to have this condition.

Erin, you **do not have the variant** we tested.

You could still have a variant not covered by this test.

How To Use This Test

This test does not diagnose any health conditions.

Please talk to a healthcare professional if this condition runs in your family, you think you might have this condition, or you have any concerns about your results.

Review the Carrier Status tutorial

See Scientific Details

Intended Uses

- To test for the R77C variant in the SGCA gene.
- To identify carrier status for LGMD2D.

Limitations

- Does **not test** for all possible variants for the condition.
- Does **not report** if someone has two copies of a tested variant.
- Does **not cover** other types of LGMD.

Important Ethnicities

- This test is expected to identify the majority of carriers of **Finnish** descent.

You are likely not a carrier.

We ruled out the tested variant for LGMD2D.

You still have a chance of being a carrier for LGMD2D.

You may still have up to a **1 in 5,500 chance** of carrying a variant not covered by this test.

See Scientific Details

About Limb-Girdle Muscular Dystrophy Type 2D

Also known as: Alpha-Sarcoglycanopathy

When symptoms develop

Symptoms typically develop between early childhood and adolescence.

How it's treated

There is currently no known cure. Therapy focuses on maintaining muscle function, preventing skeletal problems, and monitoring heart and lung function.

Typical signs and symptoms

- Wasting of arm and leg muscles closest to the torso
- Large calf muscles
- Curvature of the spine
- Heart and lung problems
- Shortened lifespan

Ethnicities most affected

This condition is rare in all ethnicities.

Read more at

Genetics Home Reference

GeneReviews

Consider talking to a healthcare professional if you are concerned about your results.

If you're starting a family, a genetic counselor can help you and your partner understand if additional testing might be appropriate.

Connect with a GC

Share your results with a healthcare professional.

Print report

Learn more about this condition and connect with support groups.

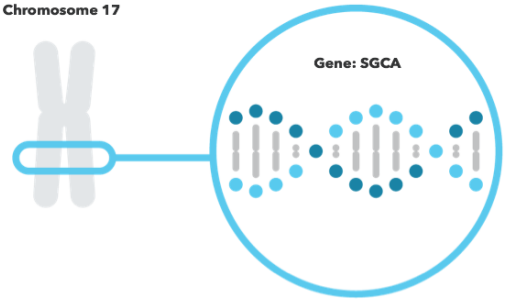
Learn more

LGMD2D is caused by variants in the SGCA gene.

SGCA

The SGCA gene contains instructions for making one part of a group of proteins. These proteins, called the sarcoglycan protein complex, are found in muscle tissue where they help strengthen and protect muscle fibers. Certain variants in the SGCA gene prevent the protein complex from working properly.

Read more at Genetics Home Reference



You have no variants detected by this test.

Variants Detected	View All Tested Markers
0	1

Marker Tested	Your Genotype*		Additional Information
R77C Gene: SGCA Marker: rs28933693	**C** Typical copy from one of your parents	**C** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [1 , 2 , 3 , 4] | ClinVar

*This test cannot distinguish which copy you received from which parent. This test also cannot determine whether multiple variants, if detected, were inherited from only one parent or from both parents. This may impact how these variants are passed down.

23andMe always reports genotypes based on the 'positive' strand of the human genome reference sequence (build 37). Other sources sometimes report genotypes using the opposite strand.

Test Interpretation

This report provides an estimate of the chances of still being a carrier for people who do not have the variant(s) tested. This is known as the **post-test carrier risk** .

Post-test carrier risk is based on the average chance of being a carrier for a given ethnicity and the carrier detection rate of the test for a given ethnicity.

View technical article on estimating post-test carrier risk.

Post-Test Carrier Risk

This report provides an estimate of the post-test carrier risk for people of Finnish descent only.

- For people of partial Finnish descent, post-test carrier risk is less than that for those who are fully Finnish. The exact post-test risk depends on how much Finnish ancestry a person has.
- Post-test risk for other ethnicities cannot be provided because sufficient data is not available.

Post-test carrier risk for relevant ethnicities

Finnish	1 in 5,500	[4]

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Limb-Girdle Muscular Dystrophy Type 2D is indicated for the detection of the R77C variant in the SGCA gene. This test is intended to be used to determine carrier status for LGMD2D in adults, but cannot determine if a person has two copies of a tested variant.

Special Considerations

- Symptoms can vary greatly in people with this condition, and can be mild in some cases.
- There are currently no professional guidelines in the U.S. for carrier testing for this condition.

Test Performance Summary

Carrier Detection Rate & Relevant Ethnicities

The "carrier detection rate" is an estimate of the percentage of carriers for this condition that would be identified by this test. Carrier detection rate differs by ethnicity and is provided only where sufficient data is available.

Finnish	95%	[4]

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for 49 samples with known variant status. 49 out of 49 genotype results were correct. About 1 in 3,300 samples may receive a **Not Determined** result. This can be caused by random test error or unexpected DNA sequences that interfere with the test. It can also be caused by having two copies of a variant tested.

Warnings and Limitations

- This test does not cover all variants that could cause this condition.*
- This test does not diagnose any health conditions.
- Positive results in individuals whose ethnicities are not commonly associated with this condition may be incorrect. Individuals in this situation should consider genetic counseling and follow-up testing.
- Share results with your healthcare professional for any medical purposes.
- If you are concerned about your results, consult with a healthcare professional.

See the Package Insert for more details on use and performance of this test.

* Variants not included in this test may be very rare, may not be available on our genotyping platform, or may not pass our testing standards.

References

1. Bartoli M et al. (2008). "Mannosidase I inhibition rescues the human alpha-sarcoglycan R77C recurrent mutation." Hum Mol Genet. 17(9):1214-21.
2. Carrié A et al. (1997). "Mutational diversity and hot spots in the alpha-sarcoglycan gene in autosomal recessive muscular dystrophy (LGMD2D)." J Med Genet. 34(6):470-5.
3. Draviam RA et al. (2006). "Alpha-sarcoglycan is recycled from the plasma membrane in the absence of sarcoglycan complex assembly." Traffic. 7(7):793-810.
4. Hackman P et al. (2005). "Enrichment of the R77C alpha-sarcoglycan gene mutation in Finnish LGMD2D patients." Muscle Nerve. 31(2):199-204.
5. Pegoraro E et al. (1993). "Limb-Girdle Muscular Dystrophy Overview"