

Limb-Girdle Muscular Dystrophy Type 2E

LGMD2E 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 SGCB gene in order to have this condition.

Overview Scientific Details

Jamie, 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 T151R variant in the SGCB gene.
- To identify carrier status for LGMD2E.

- 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 most relevant for people of **Southern Indiana Amish** descent.

You are likely not a carrier.

This result may be less relevant for you because the variants that cause LGMD2E are rarely found in people of your ethnicity.

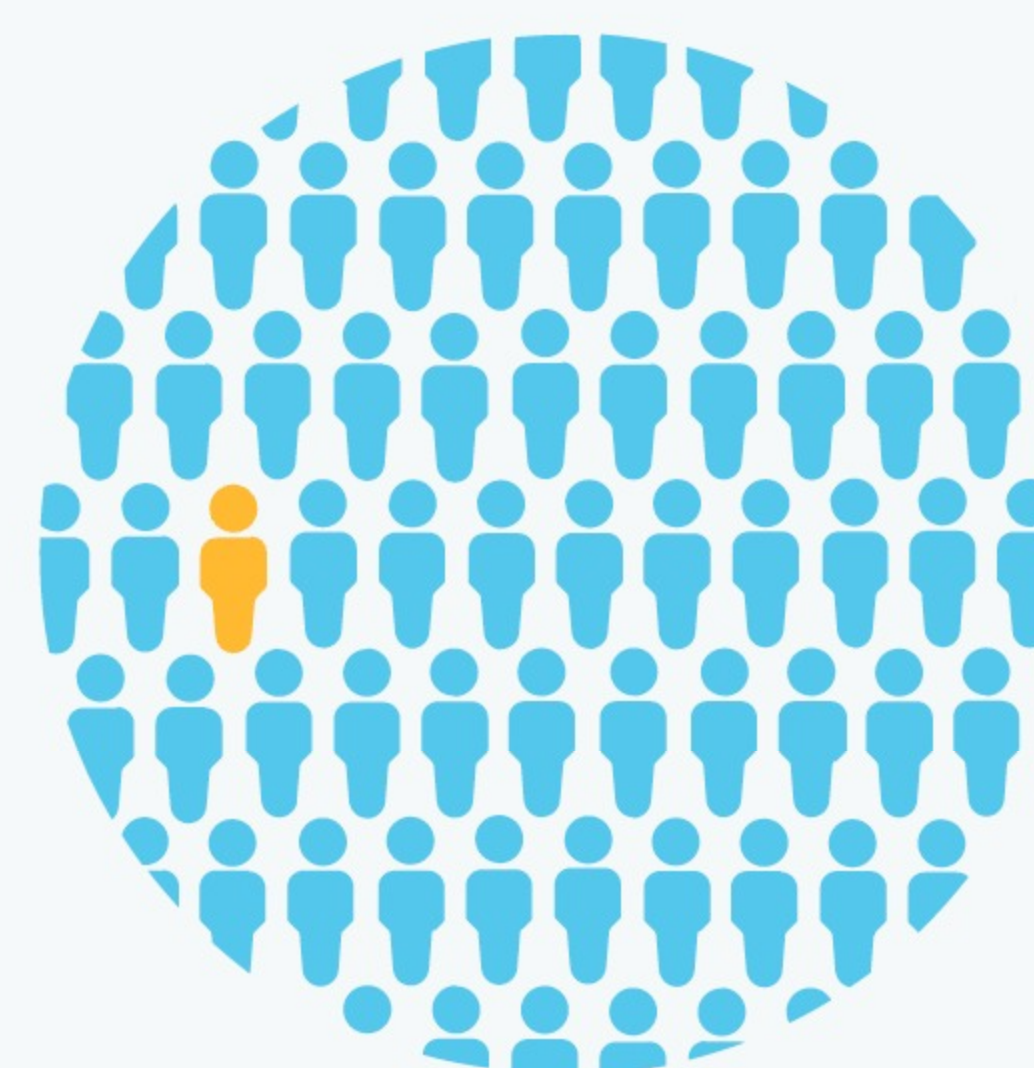


We ruled out the tested variant for LGMD2E.

This variant is most common in people of **Amish** descent.

You still have a chance of being a carrier for LGMD2E.

We cannot estimate your chances because this condition is rare and not well studied in your ethnicity.



About Limb-Girdle Muscular Dystrophy Type 2E

Also known as: Beta-Sarcoglycanopathy

📅 When symptoms develop

Symptoms typically develop between early childhood and adolescence.

🏠 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

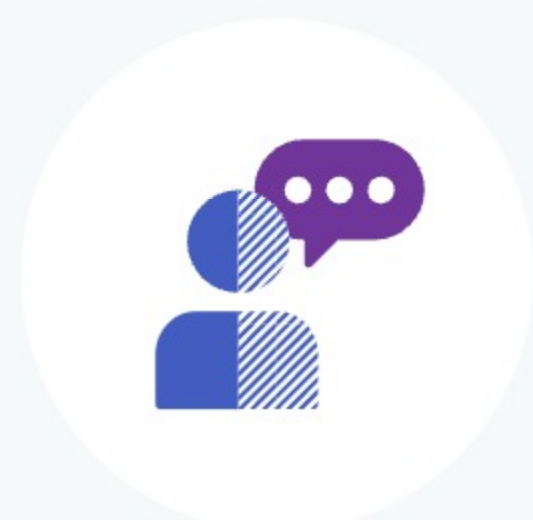
This condition is most common in people of Southern Indiana **Amish** descent.

🏥 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.

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](#)



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Limb-Girdle Muscular Dystrophy Type 2E

LGMD2E 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 SGCB gene in order to have this condition.

Overview **Scientific Details**

LGMD2E is caused by variants in the SGCB gene.

SGCB


The SGCB 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 SGCB gene prevent the protein complex from working properly.

Read more at [Genetics Home Reference](#)

Chromosome 4



You have no variants detected by this test.

Variants Detected		View All Tested Markers	
Marker Tested	Your Genotype*	Additional Information	
T151R Gene: SGCB Marker: rs28936383	G Typical copy from one of your parents	 G Typical copy from your other parent	<ul style="list-style-type: none"> ▼ Biological explanation ▼ Typical vs. variant DNA sequence(s) ▼ Percent of 23andMe customers with variant ▼ References [1, 2, 3] 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

Post-test carrier risk for LGMD2E is the chance of still being a carrier for the condition if you do not have the variant tested. This chance depends on how common it is to be a carrier for LGMD2E and whether the variants we tested tend to be found in people of your ethnicity.

Because you do not have the variant we tested, your chances of still being a carrier are lower than for someone who has not been tested. However, we cannot provide an exact estimate because the information needed to calculate post-test carrier risk is not available for your ethnicity.

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Limb-Girdle Muscular Dystrophy Type 2E is indicated for the detection of the T151R variant in the SGCB gene. This test is intended to be used to determine carrier status for LGMD2E in adults, but cannot determine if a person has two copies of a tested variant. The test is most relevant for people of Amish descent.

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.

Amish from southern Indiana	>99%	[3]
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Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing. Greater than 99% of test results were correct. While unlikely, this test may provide false positive or false negative results. For more details on the analytical performance of this test, refer to the package insert.

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. [Cottrell CE et al. \(2012\). "Maternal uniparental disomy of chromosome 4 in a patient with limb-girdle muscular dystrophy 2E confirmed by SNP array technology." Clin Genet. 81\(6\):578-83.](#)
2. [Duclos F et al. \(1998\). "Beta-sarcoglycan: genomic analysis and identification of a novel missense mutation in the LGMD2E Amish isolate." Neuromuscul Disord. 8\(1\):30-8.](#)
3. [Lim LE et al. \(1995\). "Beta-sarcoglycan: characterization and role in limb-girdle muscular dystrophy linked to 4q12." Nat Genet. 11\(3\):257-65.](#)
4. [Pegoraro E et al. \(2000\). "Limb-Girdle Muscular Dystrophy Overview." \[Updated 2012 Aug 30\].](#)

Change Log

Your report may occasionally be updated based on new information. This Change Log describes updates and revisions to this report.

Date	Change
Oct. 21, 2015	Limb-Girdle Muscular Dystrophy Type 2E report created.



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