Limb-Girdle Muscular Dystrophy Type 2I

LGMD2I 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 FKRP 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.

0 variants detected
in the FKRP gene

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.

Intended Uses

- To test for the L276I variant in the FKRP gene.
- To identify carrier status for LGMD2I.

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 European descent.

You are likely not a carrier.

We ruled out the tested variant for LGMD2I.

You still have a chance of being a carrier for LGMD2I.

You may still have up to a **1 in 520 chance** of carrying a variant not covered by this test.

See Scientific Details
About Limb-Girdle Muscular Dystrophy Type 2I

Also known as: Dystroglycanopathy, Muscular Dystrophy-Dystroglycanopathy Type C5

When symptoms develop
Symptoms typically develop between early childhood and early adulthood.

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
- Heart and lung problems
- Large calf muscles
- Curvature of the spine
- Shortened lifespan

Ethnicities most affected
This condition is rare in all ethnicities.

Read more at
Genetics Home Reference
GenetReviews

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.

Learn more about this condition and connect with support groups.

LGMD2I is caused by variants in the FKRP gene.

The FKRP gene contains instructions for making a protein that is found at especially high levels in muscle tissue. Its function is not fully understood, but it is believed to be involved in stabilizing and protecting muscle fibers.

Read more at Genetics Home Reference
You have no variants detected by this test.

<table>
<thead>
<tr>
<th>Marker Tested</th>
<th>Your Genotype*</th>
<th>Additional Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>L2761</td>
<td>C</td>
<td>Biological explanation</td>
</tr>
<tr>
<td>Genotype: FKRP</td>
<td>Typical copy from one of your parents</td>
<td>Typical vs. variant DNA sequence(s)</td>
</tr>
<tr>
<td>Marker: rs28937900</td>
<td>C</td>
<td>Percent of 23andMe customers with variant</td>
</tr>
<tr>
<td></td>
<td></td>
<td>References [1, 2, 3, 4, 5, 6, 7, 8, 9]</td>
</tr>
</tbody>
</table>

*This test cannot distinguish which copy you received from which parent. This test also cannot determine whether multiple variants, if detected, were inherited from 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.

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Test Interpretation

Post-Test Carrier Risk

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

- For people of partial European descent, post-test carrier risk is less than that for those who are fully European. The exact post-test risk depends on how much European 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

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Risk</th>
<th>95% CI</th>
</tr>
</thead>
<tbody>
<tr>
<td>European</td>
<td>1/520</td>
<td>0.3-3</td>
</tr>
</tbody>
</table>

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Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Limb-Girdle Muscular Dystrophy Type 2I is indicated for the detection of the L2761 variant in the FKRP gene. This test is intended to be used to determine carrier status for LGMD2I 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.

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Rate</th>
</tr>
</thead>
<tbody>
<tr>
<td>European</td>
<td>62%</td>
</tr>
</tbody>
</table>

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for 51 samples with known variant status. 51 out of 51 genotype results were correct. About 1 in 4,000 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.

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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


