Glycogen Storage Disease Type Ia

GSDIa is a rare genetic disorder. It is characterized by low blood sugar, liver and kidney problems, and poor growth. A person must have two variants in the G6PC 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 G6PC 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.

Review the Carrier Status tutorial
See Scientific Details

Intended Uses

• To test for the R83C variant in the G6PC gene.
• To identify carrier status for GSDIa.

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 subtypes of glycogen storage disease.

Important Ethnicities

• This test is most relevant for people of Ashkenazi Jewish descent.

You are likely not a carrier.

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

We ruled out the tested variant for GSDIa.

This variant is most common in people of Ashkenazi Jewish descent.

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

We cannot estimate your chances because this condition is rare and not well studied in your ethnicity.
About Glycogen Storage Disease Type Ia

Also known as: von Gierke Disease

When symptoms develop
Symptoms typically develop during infancy.

How it’s treated
There is currently no known cure. Treatment focuses on managing diet to control blood sugar levels and prevent problems with metabolism.

Typical signs and symptoms
- Low blood sugar
- Liver enlargement
- Very short height
- Kidney and liver problems
- Anemia

Ethnicities most affected
This condition is most common in people of Ashkenazi Jewish descent.

Read more at
Genetics Home Reference
GeneReviews
National Organization for Rare Disorders

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

GSD Ia is caused by variants in the G6PC gene.

The G6PC gene contains instructions for making part of a protein called glucose-6-phosphatase, also known as G6Pase. This protein helps control sugar levels in the body. Certain variants in G6PC disrupt this protein’s function.

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>R83C</td>
<td>C</td>
<td>Biological explanation</td>
</tr>
<tr>
<td>Gene: G6PC</td>
<td>Typical copy from one of your parents</td>
<td>Typical vs. variant DNA sequence(s)</td>
</tr>
<tr>
<td>Marker: 13002466</td>
<td>C</td>
<td>Percent of 23andMe customers with variant</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 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 resources sometimes report genotypes using the opposite strand.

Test Interpretation

Post-Test Carrier Risk

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

- For people of partial Ashkenazi Jewish descent, post-test carrier risk is less than that for those who are fully Ashkenazi Jewish. The exact post-test risk depends on how much Ashkenazi Jewish 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>
</tr>
</thead>
<tbody>
<tr>
<td>Ashkenazi Jewish</td>
<td>1 in 3,200</td>
</tr>
</tbody>
</table>

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Glycogen Storage Disease Type 1a is indicated for the detection of the R83C variant in the G6PC gene. This test is intended to be used to determine carrier status for GSD1a in adults, but cannot determine if a person has two copies of a tested variant. The test is most relevant for people of Ashkenazi Jewish descent.

Special Considerations

- 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>Ashkenazi Jewish</td>
<td>98%</td>
</tr>
</tbody>
</table>

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. Fewer than 1 in 100,000 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.

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. Bell DS et al. (1993). "Glycogen Storage Disease Type I." 