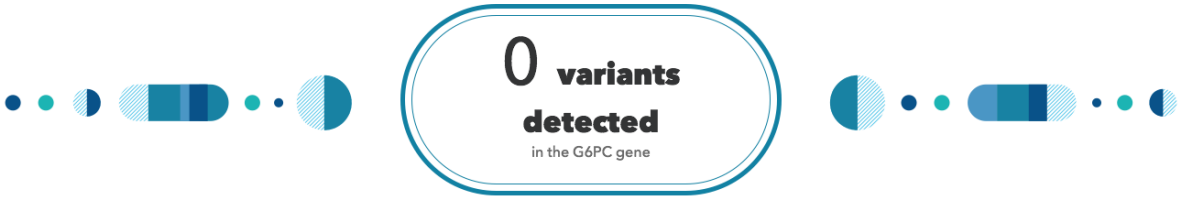
Glycogen Storage Disease Type Ia

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.



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.

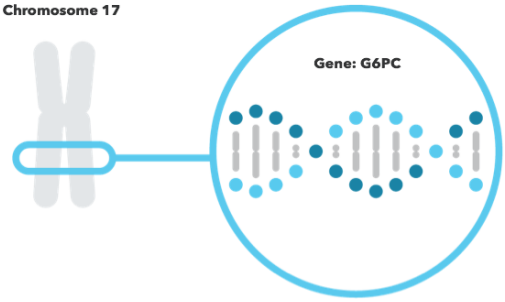
Learn more

GSDIa is caused by variants in the G6PC gene.

G6PC

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.

Variants Detected	View All Tested Markers
0	1

Marker Tested	Your Genotype*		Additional Information
R83C Gene: G6PC Marker: i3002486	**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** [2 , 3 , 4 , 5 , 6 , 7] | 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 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

Ashkenazi Jewish	1 in 3,200	[3]

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Glycogen Storage Disease Type Ia 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 GSDIa 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.

Ashkenazi Jewish	98%	[2]

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. Bali DS et al. (1993). "Glycogen Storage Disease Type I"
2. Chou JY et al. (2008). "Mutations in the glucose-6-phosphatase-alpha (G6PC) gene that cause type Ia glycogen storage disease." Hum Mutat. 29(7):921-30.
3. Ekstein J et al. (2004). "Mutation frequencies for glycogen storage disease Ia in the Ashkenazi Jewish population." Am J Med Genet A. 129A(2):162-4.
4. Lei KJ et al. (1993). "Mutations in the glucose-6-phosphatase gene that cause glycogen storage disease type 1a." Science. 262(5133):580-3.
5. Lei KJ et al. (1995). "Genetic basis of glycogen storage disease type 1a: prevalent mutations at the glucose-6-phosphatase locus." Am J Hum Genet. 57(4):766-71.
6. Seydewitz HH et al. (2000). "Molecular genetic analysis of 40 patients with glycogen storage disease type Ia: 100% mutation detection rate and 5 novel mutations." Hum Mutat. 15(1):115-6.
7. Stroppiano M et al. (1999). "Mutations in the glucose-6-phosphatase gene of 53 Italian patients with glycogen storage disease type Ia." J Inherit Metab Dis. 22(1):43-9.