Glycogen Storage Disease Type Ib

GSDIb is a rare genetic disorder. It is characterized by low blood sugar, liver and kidney problems, and frequent infections. A person must have two variants in the SLC37A4 gene in order to have this condition.

Overview Scientific Details

Jamie, you **do not have the variants** 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

- Tests for **multiple variants** in the SLC37A4 gene.
- To identify carrier status for GSDIb.

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 does **not** include the majority of SLC37A4 variants that cause GSDIb in any ethnicity.

You are likely not a carrier.

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

We ruled out the tested variants for GSDIb.

These variants are very rare in all ethnicities.

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

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

About Glycogen Storage Disease Type Ib

Also known as: von Gierke Disease

When symptoms develop

Symptoms typically develop during infancy.

Ethnicities most affected

This condition is rare in all ethnicities.

Typical signs and symptoms

- Low blood sugar
- Liver enlargement
- Kidney and liver problems
- Frequent infections
- Very short height
- Inflammatory bowel disease
- Oral problems, including gum disease and cavities

How it's treated

There is currently no known cure. Treatment focuses on managing diet in order to control blood sugar levels and prevent problems with metabolism. Medication can help prevent infections.

Read more at: MedlinePlus 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

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Overview | Scientific Details

GSDIb is caused by variants in the SLC37A4 gene.

SLC37A4

The SLC37A4 gene contains instructions for making part of a protein called the glucose-6-phosphate transporter. This protein helps control the level of certain sugars, called glycogen and glucose, in the body. Certain variants in SLC37A4 disrupt this protein's function, leading to a buildup of glycogen in cells and low glucose levels.

Read more at MedlinePlus

You have no variants detected by this test.

Variants Detected | View All Tested Markers

Marker Tested	Your Genotype*		Additional Information
1042_1043delCT Gene: SLC37A4 Marker: i5012880	**AG** Typical copy from one of your parents	**AG** Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [3, 7, 8, 10, 11, 13, 14] | ClinVar
W118R Gene: SLC37A4 Marker: i5012878	**A** Typical copy from one of your parents	**A** Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [1, 3, 6, 9] | 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 GSDIb is the chance of still being a carrier for the condition if you do not have the variants tested. This chance depends on how common it is to be a carrier for GSDIb and whether the variants we tested tend to be found in people of your ethnicity.

Because you do not have the variants 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 Glycogen Storage Disease Type Ib is indicated for the detection of two variants in the SLC37A4 gene. This test is intended to be used to determine carrier status for GSDIb in adults, but cannot determine if a person has two copies of a tested variant.

Special Considerations

- This test does not include the majority of SLC37A4 variants that cause GSDIb in any ethnicity.
- There are currently no professional guidelines in the U.S. for carrier testing for this condition. However, ACOG notes that testing for glycogen storage disease type I may be considered for people of Ashkenazi Jewish descent who are considering having children.

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.

Japanese	42%	[4]
Serbian	39%	[12]
European	31%	[4]

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. Avramova Z et al. (1988). "Metabolic behaviour of a stable DNA-protein complex." Int J Biochem. 20(1):61-5.
2. Bali DS et al. (2006). "Glycogen Storage Disease Type I." [Accessed Aug 12, 2021].
3. Chen LY et al. (2000). "Structural requirements for the stability and microsomal transport activity of the human glucose 6-phosphate transporter." J Biol Chem. 275(44):34280-6.
4. Chou JY et al. (2002). "Type I glycogen storage diseases: disorders of the glucose-6-phosphatase complex." Curr Mol Med. 2(2):121-43.
5. Committee on Genetics. (2017). "Committee Opinion No. 691: Carrier Screening for Genetic Conditions." Obstet Gynecol. 129(3):e41-e55.
6. Hou DC et al. (1999). "Glycogen storage disease type Ib: structural and mutational analysis of the microsomal glucose-6-phosphate transporter gene." Am J Med Genet. 86(3):253-7.
7. Janecke AR et al. (2000). "Mutation analysis in glycogen storage disease type 1 non-a." Hum Genet. 107(3):285-9.
8. Kishnani PS et al. (2014). "Diagnosis and management of glycogen storage disease type I: a practice guideline of the American College of Medical Genetics and Genomics." Genet Med. 16(11):e1.
9. Kure S et al. (1998). "Molecular analysis of glycogen storage disease type Ib: identification of a prevalent mutation among Japanese patients and assignment of a putative glucose-6-phosphate translocase gene to chromosome 11." Biochem Biophys Res Commun. 248(2):426-31.
10. Melis D et al. (2005). "Genotype/phenotype correlation in glycogen storage disease type 1b: a multicentre study and review of the literature." Eur J Pediatr. 164(8):501-8.

See all references

Change Log

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

Date	Change
Dec. 9, 2019	Information specific to people of Serbian descent was added. Customers may now see carrier detection rate information specific to that ancestry.
Feb. 18, 2016	Due to improvements in data analysis, some customers who previously received a "Not Determined" result for i5012889 may see a genotype at this marker. This may also update the overall report result for these customers.
Oct. 21, 2015	Glycogen Storage Disease Type Ib report created.

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