Gaucher Disease Type 1

Gaucher disease type 1 is a rare genetic disorder that can affect many organs. It often leads to an enlarged liver and spleen, as well as bone abnormalities. A person must have two variants in the GBA gene, or two copies of a variant, in order to have Gaucher disease type 1.

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 Gaucher disease type 1 or any other 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 **three variants** in the GBA gene associated with Gaucher disease type 1. These variants are called N370S, 84GG, and V394L.
- To identify carrier status for Gaucher disease type 1.
- Informs individuals with two variants in the GBA gene, or two copies of the N370S variant, that they are at risk for developing symptoms of Gaucher disease type 1.

Limitations

- Does **not test** for all possible variants for the condition.
- Does **not report** if someone has two copies of the 84GG or V394L variant or has one copy of each of those variants.

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 Gaucher disease type 1 are rarely found in people of your ethnicity.

We ruled out the tested variants for Gaucher disease type 1.

These variants are most common in people of **Ashkenazi Jewish** descent.

You still have a chance of being a carrier for Gaucher disease type 1.

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

About Gaucher Disease Type 1

When symptoms develop

Symptoms can develop anytime from childhood to adulthood and can vary from mild to severe. Some people may never develop symptoms.

Typical signs and symptoms

- Enlargement of the liver and spleen
- Bone weakness and pain
- Growth impairment
- Anemia and low platelet count

Ethnicities most affected

This condition is most common in people of Ashkenazi Jewish descent but also occurs in people of other ethnicities.

How it's treated

There is currently no known cure. Treatment varies depending on the severity of symptoms, but often includes enzyme replacement therapy.

Read more at: Genetics Home Reference GeneReviews Mayo Clinic

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

Share your results with a healthcare professional.

Print report

Learn more about this condition and connect with support groups.

Learn more

©2022 23andMe, Inc. Terms of Service Membership Terms Privacy Statement Cookie Policy Help Resources

Gaucher Disease Type 1

Gaucher disease type 1 is a rare genetic disorder that can affect many organs. It often leads to an enlarged liver and spleen, as well as bone abnormalities. A person must have two variants in the GBA gene, or two copies of a variant, in order to have Gaucher disease type 1.

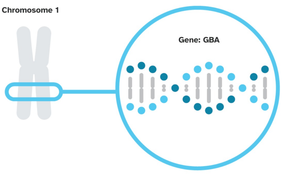
Overview | Scientific Details

Gaucher disease type 1 is caused by variants in the GBA gene.

GBA

The GBA gene contains instructions for making an enzyme that breaks down a large, fatty molecule called glucocerebroside into simpler sugars and fats. Certain variants in the GBA gene prevent the enzyme from functioning. This can cause glucocerebroside to build up to harmful levels in various organs.

Read more at Genetics Home Reference



You have no variants detected by this test.

Variants Detected | View All Tested Markers

Marker Tested	Your Genotype*			Additional Information
N370S **Gene:** GBA **Marker:** i4000415	**T** Typical copy from one of your parents		**T** Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [5, 6] | ClinVar
84GG **Gene:** GBA **Marker:** i4000417	**(—)** Typical copy from one of your parents		**(—)** Typical copy from your other parent	Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [1, 5] | ClinVar
V394L **Gene:** GBA **Marker:** i4000419	**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] | 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 209	[5]

Test Details

Indications for Use

The 23andMe PGS Carrier Status Report for Gaucher Disease Type 1 is indicated for reporting of the N370S, 84GG, and V394L variants in the GBA gene. This report describes carrier status for Gaucher disease type 1 in adults. This report also describes if a result is associated with personal risk for developing symptoms of Gaucher disease type 1, but it does not describe a person's overall risk of developing symptoms. This test is most relevant for people of Ashkenazi Jewish descent.

Special Considerations

- The severity of symptoms, and when they develop, can vary greatly in people with Gaucher disease type 1. Some people may never develop symptoms.
- The 84GG and V394L variants can occasionally be found in people with the more severe, type 2 or type 3 forms of Gaucher disease. People with two copies of the N370S variant, or one copy of N370S and one copy of another variant, typically have the less severe, type 1 form of the disease.
- Carrier testing for Gaucher disease type 1 is recommended by ACMG for people of Ashkenazi Jewish descent considering having children. This test includes two of four variants recommended for testing by ACMG.

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	92%	[5]

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. The performance of this test may be affected by the presence of rare mutations, such as c.1265_1319del55.

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. **Beutler E et al. (1991). "Identification of the second common Jewish Gaucher disease mutation makes possible population-based screening for the heterozygous state." Proc Natl Acad Sci U S A. 88(23):10544-7.**
2. **Beutler E et al. (1992). "Mutations in Jewish patients with Gaucher disease." Blood. 79(7):1662-6.**
3. **Elstein D et al. (2005). "Phenotypic heterogeneity in patients with Gaucher disease and the N370S/V394L genotype." Genet Test. 9(1):26-9.**
4. **Grace ME et al. (1994). "Analysis of human acid beta-glucosidase by site-directed mutagenesis and heterologous expression." J Biol Chem. 269(3):2283-91.**
5. **Gross SJ et al. (2008). "Carrier screening in individuals of Ashkenazi Jewish descent." Genet Med. 10(1):54-6.**
6. **Montfort M et al. (2004). "Functional analysis of 13 GBA mutant alleles identified in Gaucher disease patients: Pathogenic changes and "modifier" polymorphisms." Hum Mutat. 23(6):567-75.**
7. **Pastores GM et al. (2000). "Gaucher Disease." [Updated 2015 Feb 26].**
8. **Torralba MA et al. (2002). "High prevalence of the 55-bp deletion (c.1263del55) in exon 9 of the glucocerebrosidase gene causing misdiagnosis (for homozygous N370S (c.1226A > G) mutation) in Spanish Gaucher disease patients." Blood Cells Mol Dis. 29(1):35-40.**

Change Log

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

Date	Change
April 17, 2017	Gaucher Disease Type 1 report created.

©2022 23andMe, Inc. Terms of Service Membership Terms Privacy Statement Cookie Policy Help Resources