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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.

play+b3811710e9, you have **two of the variants** we tested.

You are at risk for developing symptoms of Gaucher disease type 1.
Your result may also be relevant if you're considering having children.

2 variants detected
in the GBA gene

This test **does not diagnose** Gaucher disease type 1. If this result is unexpected, please discuss this report with a healthcare professional.

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
See Frequently Asked Questions

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 at risk for developing symptoms of Gaucher disease type 1.

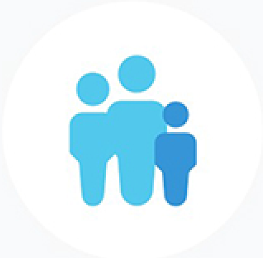
Your result may also be relevant if you're considering having children.

Talk to a healthcare professional.

People with your result are at risk for developing symptoms of Gaucher disease type 1, and early detection is important. A healthcare professional can answer questions you may have about your results. See Frequently Asked Questions for more information.

Your result may be relevant for your family.

Because you have two variants, you will most likely pass a variant on to each of your children. If your partner is a carrier for Gaucher disease, each child most likely has a 50% chance of having this condition. Your relatives may also wish to consider testing if they plan to have children.



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

It is important to talk to a healthcare professional if you are concerned about your result.

You are at risk for developing symptoms of Gaucher disease type 1. It is important to consult with a healthcare professional about your result.

Print report

If you're starting a family, a genetic counselor can help you and your partner understand if additional testing might be appropriate.

Learn more

See our Frequently Asked Questions for more information.

FAQs

Learn more about this condition and connect with support groups.

Learn more

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Gaucher Disease Type 1

Scientific Details

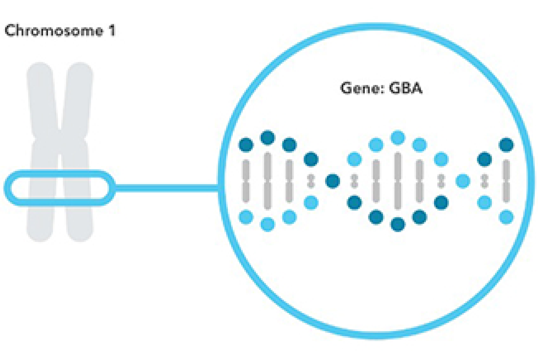
Gaucher disease type 1 is a rare genetic disorder that can affect many organs. It often leads to an enlarged liver and spleen, as well as bone abnormalities. A person must have two variants in the GBA gene, or two copies of a variant, in order to have Gaucher disease type 1.

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 two variants detected by this test.

Variants Detected | View All Tested Markers

Marker Tested	Your Genotype*		Additional Information
N370S Gene: GBA Marker: i4000415	C — Variant 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	C — Variant 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 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 for 438 samples with known variant status. 437 out of 438 genotype results were correct. 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. (1993). "Gaucher Disease"
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.

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Gaucher Disease Type 1

Frequently Asked Questions

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.

Gaucher Disease Type 1

What does this test do?

This test looks for three genetic variants in the GBA gene that are associated with Gaucher disease type 1.

People with one variant can pass the variant on to their children.

People with two variants, or two copies of a variant, are at risk for developing symptoms of Gaucher disease type 1, and they will most likely pass a variant on to each of their children.

This test does not include all possible genetic variants associated with Gaucher disease type 1.

Is this answer helpful? Yes No

What does this test not do?

This test does not diagnose Gaucher disease type 1 or any other health conditions. Only a healthcare professional can do that.

This test does not include all possible genetic variants associated with Gaucher disease type 1.

Is this answer helpful? Yes No

My report says two variants were detected. What does this mean?

This means you have two of the genetic variants we tested. You most likely inherited one variant from each of your parents.

People with the two variants we detected are at risk for developing symptoms of Gaucher disease type 1. However, the severity of symptoms, and when they develop, can vary greatly between individuals. It's important to talk with a healthcare professional if you have concerns about your results.

This result also means you will most likely pass a variant on to each of your children.

Is this answer helpful? Yes No

What does at risk for developing symptoms of Gaucher disease type 1 mean?

Symptoms of Gaucher disease type 1 include an enlarged liver and spleen, bone weakness and pain, growth impairment, anemia, and a low platelet count.

People with your genetic result are at risk for developing these symptoms. However, the severity of symptoms, and when they develop, can vary greatly.

Is this answer helpful? Yes No

The report says the test is most relevant for people of Ashkenazi Jewish descent. What if I'm not of Ashkenazi Jewish descent?

The genetic variants we tested are most common in people of Ashkenazi Jewish descent, but they are also found in people of other ethnicities. Because you have two variants, you are still at risk for developing symptoms of Gaucher disease type 1. In addition, you will still most likely pass a variant on to each of your children.

Is this answer helpful? Yes No

My report says I have two variants associated with Gaucher disease type 1. What are some things I could do?

Based on your genetic result, you are at risk for developing symptoms of Gaucher disease type 1. Early detection is important, so talk to a healthcare professional about your result.

If you're starting a family, a genetic counselor can help you and your partner understand if additional testing might be appropriate.

You may also want to share your results with your family. Because we share DNA with our relatives, your genetic result could be relevant for your family members.

Is this answer helpful? Yes No

How could my result affect my children?

Because you have two variants, you will most likely pass a variant on to each of your children. If your partner is a carrier for Gaucher disease, each of your children most likely has a 50% chance of having this condition. Depending on your partner's genotype, your children could inherit a combination of variants that may put them at risk for the more severe, type 2 or type 3 form of Gaucher disease. A genetic counselor can help you and your partner understand if additional testing might be appropriate. Learn more about genetic counseling.

Is this answer helpful? Yes No

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