Tay-Sachs Disease

Tay-Sachs disease is a rare genetic disorder. It is characterized by a loss of strength and coordination over time as well as developmental disability, seizures, and early death. A person must have two variants in the HEXA 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 HEXA gene.
- To identify carrier status for Tay-Sachs disease.

Limitations

- Does **not test** for all possible variants for the condition.
- Does **not report** if someone has two copies of a tested variant.

Important Ethnicities

- This test is most relevant for people of **Ashkenazi Jewish** and **Cajun** descent.
- This test does **not** include the most common variant found in people of French Canadian descent with Tay-Sachs disease.

You are likely not a carrier.

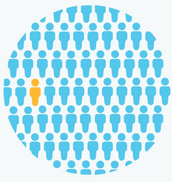
This result may be less relevant for you because the variants that cause Tay-Sachs disease are rarely found in people of your ethnicity.

We ruled out the tested variants for Tay-Sachs disease.

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

You still have a chance of being a carrier for Tay-Sachs disease.

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



About Tay-Sachs Disease

Also known as: Hexosaminidase A Deficiency

When symptoms develop

Symptoms typically develop during infancy.

Typical signs and symptoms

- Loss of strength and coordination that worsens over time
- Severe developmental disability
- Vision loss
- Seizures
- Death in early childhood in severe cases

Ethnicities most affected

This condition is most common in people of Ashkenazi Jewish, Cajun, and French Canadian descent.

How it's treated

There is currently no known cure. Treatment focuses on managing symptoms, providing nutritional support, and using seizure medications as needed.

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.

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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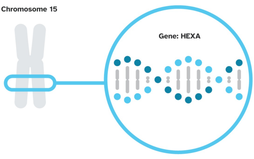
Overview | Scientific Details

Tay-Sachs disease is caused by variants in the HEXA gene.

HEXA

The HEXA gene contains instructions for making one part of an enzyme called hexosaminidase A. This enzyme helps break down harmful substances within compartments of nerve cells called lysosomes. Certain variants in HEXA disrupt this function, causing a buildup of these harmful substances inside nerve cells of the brain and spinal cord.

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
G269S **Gene:** HEXA **Marker:** i4000436	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 [5] | ClinVar
1278insTATC **Gene:** HEXA **Marker:** i4000391	(—) 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 [2, 5] | ClinVar
IVS12+1G>C **Gene:** HEXA **Marker:** i4000393	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 [5] | ClinVar
IVS9+1G>A **Gene:** HEXA **Marker:** i4000438	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 [1, 8, 10, 11, 12] | 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 and Cajun descent only.

- For people with partial ethnicity from one or more groups mentioned above, post-test carrier risk depends on the exact mixture in the person's background.
- 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 2,769	[5]
Cajun	1 in 29,999,999	[3]

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Tay-Sachs Disease is indicated for the detection of four variants in the HEXA gene. This test is intended to be used to determine carrier status for Tay-Sachs disease 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 and Cajun descent.

Special Considerations

- Symptoms of this disorder vary in severity depending on which variants are causing the condition.
- Carrier testing for Tay-Sachs disease is recommended by ACMG and ACOG for people of Ashkenazi Jewish descent considering having children. This test includes the three variants recommended for testing by ACMG. In addition, ACOG recommends offering carrier testing for Tay-Sachs disease to individuals of Cajun and French Canadian descent who are considering having children.
- When carrier testing for Tay-Sachs disease is indicated in people who are not of Ashkenazi Jewish descent, ACMG recommends biochemical carrier screening as a first step. Genetic testing can then be used to confirm carrier status in people with a positive result.

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	99%	[5]
Cajun	>99%	[19]
French Canadian	<19%	[6]

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. **Akli S et al. (1993). "A null allele frequent in non-Jewish Tay-Sachs patients." Hum Genet. 91(6):614-20.**
2. **Boles DJ et al. (1995). "The molecular basis of HEXA mRNA deficiency caused by the most common Tay-Sachs disease mutation." Am J Hum Genet. 56(3):716-24.**
3. **Committee on Genetics. (2017). "Committee Opinion No. 690: Carrier Screening in the Age of Genomic Medicine." Obstet Gynecol. 129(3):e35-e40.**
4. **Committee on Genetics. (2017). "Committee Opinion No. 691: Carrier Screening for Genetic Conditions." Obstet Gynecol. 129(3):e41-e55.**
5. **Gross SJ et al. (2008). "Carrier screening in individuals of Ashkenazi Jewish descent." Genet Med. 10(1):54-6.**
6. **Hechtman P et al. (1990). "More than one mutant allele causes infantile Tay-Sachs disease in French-Canadians." Am J Hum Genet. 47(5):815-22.**
7. **Kaback MM et al. (1999). "Hexosaminidase A Deficiency." [Accessed Nov 1, 2016].**
8. **Landels EC et al. (1992). "Beta-hexosaminidase splice site mutation has a high frequency among non-Jewish Tay-Sachs disease carriers from the British Isles." J Med Genet. 29(8):563-7.**
9. **Martin DC et al. (2007). "Evaluation of the risk for Tay-Sachs disease in individuals of French Canadian ancestry living in new England." Clin Chem. 53(3):392-8.**
10. **McDowell GA et al. (1992). "The presence of two different infantile Tay-Sachs disease mutations in a Cajun population." Am J Hum Genet. 51(5):1071-7.**

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
Feb. 18, 2016	Due to improvements in data analysis, some customers who previously received a "Not Determined" result for one or more of the following genetic markers may see a genotype at these markers: i4999393, i4999436. This may also update the overall report result for these customers.
Oct. 21, 2015	Tay-Sachs Disease report created.