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.

Erin, you **do not have the variants** we tested.

You could still have a variant not covered by this test.

0 variants detected in the HEXA gene

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**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

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**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.

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You are likely not a carrier.

This result is relevant for you because you have **Ashkenazi Jewish** ancestry.

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We ruled out the most common variants for Tay-Sachs disease in people of Ashkenazi Jewish descent.

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

You may still have up to a **1 in 2,700 chance** of carrying a variant not covered by this test.

See Scientific Details
About Tay-Sachs Disease

Also known as: Hexosaminidase A Deficiency

When symptoms develop
Symptoms typically develop during infancy.

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

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.

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

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

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

Chromosome 15

Gene: HEXA
You have no variants detected by this test.

### Variants Detected

<table>
<thead>
<tr>
<th>Marker Tested</th>
<th>Your Genotype*</th>
<th>Additional Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>G2695</td>
<td>C</td>
<td>Biological explanation</td>
</tr>
<tr>
<td>Gene: HEXA</td>
<td>Typical copy from one of your parents</td>
<td>%</td>
</tr>
<tr>
<td>Marker: i4000436</td>
<td>C</td>
<td>Typical copy from your other parent</td>
</tr>
<tr>
<td>1278InsTATC</td>
<td>(-)</td>
<td>Biological explanation</td>
</tr>
<tr>
<td>Gene: HEXA</td>
<td>Typical copy from one of your parents</td>
<td></td>
</tr>
<tr>
<td>Marker: i4000391</td>
<td>(-)</td>
<td>Typical copy from your other parent</td>
</tr>
<tr>
<td>IVS12+1G&gt;C</td>
<td>C</td>
<td>Biological explanation</td>
</tr>
<tr>
<td>Gene: HEXA</td>
<td>Typical copy from one of your parents</td>
<td></td>
</tr>
<tr>
<td>Marker: i4000393</td>
<td>C</td>
<td>Typical copy from your other parent</td>
</tr>
<tr>
<td>IVS9+1G&gt;A</td>
<td>C</td>
<td>Biological explanation</td>
</tr>
<tr>
<td>Gene: HEXA</td>
<td>Typical copy from one of your parents</td>
<td></td>
</tr>
<tr>
<td>Marker: i4000438</td>
<td>C</td>
<td>Typical copy from your other parent</td>
</tr>
</tbody>
</table>

*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.

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### Test Interpretation

#### 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**

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Carrier Risk</th>
<th>Risk(h)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ashkenazi Jewish</td>
<td>1 in 2,700</td>
<td>[4]</td>
</tr>
<tr>
<td>Cajun</td>
<td>1 in 29,000,000</td>
<td>[1]</td>
</tr>
</tbody>
</table>
Indications for Use

The 23andMe PGS Carrier 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 for people of Ashkenazi Jewish descent considering having children. This test includes the three 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.

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Rate</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ashkenazi Jewish</td>
<td>99%</td>
</tr>
<tr>
<td>Cajun</td>
<td>&gt;99%</td>
</tr>
<tr>
<td>French Canadian</td>
<td>[ 4 ]</td>
</tr>
<tr>
<td></td>
<td>[ 9 ]</td>
</tr>
<tr>
<td></td>
<td>[ 5 ]</td>
</tr>
</tbody>
</table>

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for 199 samples with known variant status. 199 out of 199 genotype results were correct. About 1 in 62,000 samples may receive a Not Determined result for one or more variants included in this test. 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


