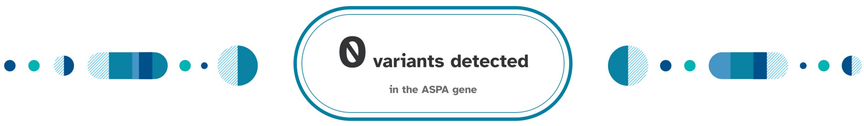
Canavan Disease

Canavan disease is a rare genetic disorder characterized by a loss of nerve cell function in the brain that worsens over time. A person must have two variants in the ASPA 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 ASPA gene.
- To identify carrier status for Canavan 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** descent.

You are likely not a carrier.

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

We ruled out the tested variants for Canavan disease.

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

You still have a chance of being a carrier for Canavan disease.

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

About Canavan Disease

Also known as: ASPA Deficiency

When symptoms develop

Symptoms typically develop during infancy.

Typical signs and symptoms

- Developmental disability
- Gradual loss of muscle tone
- Seizures
- Difficulty swallowing

Ethnicities most affected

This condition is most common in people of Ashkenazi Jewish descent.

How it's treated

There is currently no known cure. Treatment focuses on preventing complications by monitoring diet, treating infectious diseases, and managing seizures.

Read more at: Genetics Home Reference 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

Canavan Disease

Canavan disease is a rare genetic disorder characterized by a loss of nerve cell function in the brain that worsens over time. A person must have two variants in the ASPA gene in order to have this condition.

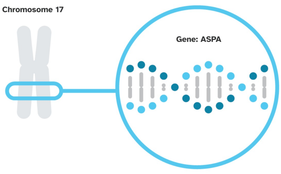
Overview | Scientific Details

Canavan disease is caused by variants in the ASPA gene.

ASPA

The ASPA gene contains instructions for making an enzyme called aspartoacylase. This enzyme breaks down a compound in the brain called N-acetyl-L-aspartic acid (NAA). Certain variants in the ASPA gene disrupt this breakdown and result in the harmful buildup of NAA in the brain. This buildup leads to a loss of myelin, a protective nerve covering that is important for nerve function.

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
Y231X **Gene:** ASPA **Marker:** rs12948217	**C** 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 [2] | ClinVar
E285A **Gene:** ASPA **Marker:** rs28940279	**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 [2] | ClinVar
A305E **Gene:** ASPA **Marker:** rs28940574	**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 [8, 9, 10] | 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 2,999	[7]

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Canavan Disease is indicated for the detection of three variants in the ASPA gene. This test is intended to be used to determine carrier status for Canavan 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 descent.

Special Considerations

- Carrier testing for Canavan disease is recommended by ACMG for people of Ashkenazi Jewish descent considering having children. This test includes the two 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	98%	[4, 7]
European	53%	[4, 5, 8, 9, 10]

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. **Feigenbaum A et al. (2004). "Canavan disease: carrier-frequency determination in the Ashkenazi Jewish population and development of a novel molecular diagnostic assay." Am J Med Genet A. 124A(2):142-7.**
2. **Gross SJ et al. (2008). "Carrier screening in individuals of Ashkenazi Jewish descent." Genet Med. 10(1):54-6.**
3. **Hoshino H et al. (2014). "Canavan disease: clinical features and recent advances in research." Pediatr Int. 56(4):477-83.**
4. **Kaul R et al. (1994). "Canavan disease: mutations among Jewish and non-Jewish patients." Am J Hum Genet. 55(1):34-41.**
5. **Kaul R et al. (1996). "Identification and expression of eight novel mutations among non-Jewish patients with Canavan disease." Am J Hum Genet. 59(1):95-102.**
6. **Matalon R et al. (1999). "Canavan Disease." [Updated 2011 Aug 11].**
7. **Monaghan KG et al. (2008). "Technical standards and guidelines for reproductive screening in the Ashkenazi Jewish population." Genet Med. 10(1):57-72.**
8. **Shaag A et al. (1995). "The molecular basis of canavan (aspartoacylase deficiency) disease in European non-Jewish patients." Am J Hum Genet. 57(3):572-80.**
9. **Sistermans EA et al. (2000). "Mutation detection in the aspartoacylase gene in 17 patients with Canavan disease: four new mutations in the non-Jewish population." Eur J Hum Genet. 8(7):557-60.**
10. **Zeng BJ et al. (2002). "Identification and characterization of novel mutations of the aspartoacylase gene in non-Jewish patients with Canavan disease." J Inherit Metab Dis. 25(7):557-70.**

Change Log

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

Date	Change
Sept. 13, 2018	Information specific to people of European descent was added. Customers who self-report having European ancestry may now see carrier detection rate information specific to that ancestry.
Oct. 12, 2016	Canavan Disease report created.