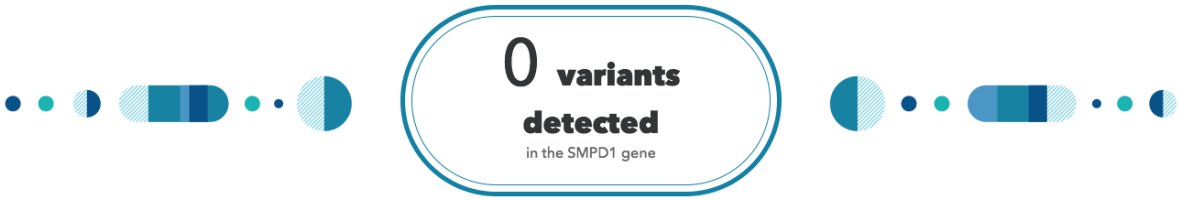
Niemann-Pick Disease Type A

Niemann-Pick disease type A is a rare genetic disorder. It is characterized by an enlarged liver and spleen, developmental disability, recurring lung infections, and early death. A person must have two variants in the SMPD1 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.



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 SMPD1 gene.
- To identify carrier status for Niemann-Pick disease type A.

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 Niemann-Pick disease type A are rarely found in people of your ethnicity.

We ruled out the tested variants for Niemann-Pick disease type A.

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

You still have a chance of being a carrier for Niemann-Pick disease type A.

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

About Niemann-Pick Disease Type A

Also known as: Acid Sphingomyelinase Deficiency

When symptoms develop

Symptoms typically develop during infancy.

How it's treated

There is currently no known cure. Treatment focuses on managing symptoms and preventing complications through physical and occupational therapy.

Typical signs and symptoms

- Enlarged liver and spleen
- Severe developmental disability
- Recurring lung infections
- Poor weight gain
- Death in early childhood

Ethnicities most affected

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

Read more at

Genetics Home Reference

GeneReviews

National Institute of Neurological Disorders and Stroke

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.

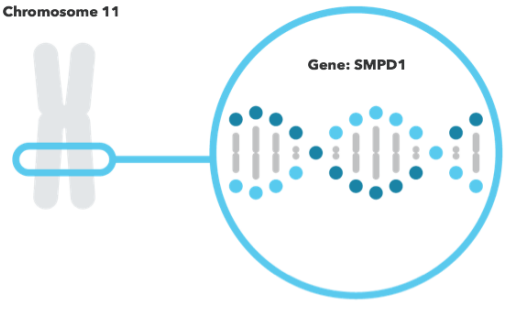
Learn more

Niemann-Pick disease type A is caused by variants in the SMPD1 gene.

SMPD1

The SMPD1 gene contains instructions for making an enzyme called acid sphingomyelinase. This enzyme plays a role in converting a fat called sphingomyelin into another fat called ceramide. Certain variants in SMPD1 disrupt this function, causing a harmful buildup of sphingomyelin.

Read more at Genetics Home Reference



You have no variants detected by this test.

Variants Detected	View All Tested Markers
0	3

Marker Tested	Your Genotype*		Additional Information
L302P Gene: SMPD1 Marker: i4000381	**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 [1]** | ClinVar
fsP330 Gene: SMPD1 Marker: i4000383	**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]** | ClinVar
R496L Gene: SMPD1 Marker: i4000430	**G** Typical copy from one of your parents	**G** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References [1]** | 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 3,000	[1]

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Niemann-Pick Disease Type A is indicated for the detection of three variants in the SMPD1 gene. This test is intended to be used to determine carrier status for Niemann-Pick disease type A 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 Niemann-Pick disease type A 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.

Ashkenazi Jewish	97%	[1]

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for 146 samples with known variant status. 146 out of 146 genotype results were correct. About 1 in 6,000 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

1. Gross SJ et al. (2008). "Carrier screening in individuals of Ashkenazi Jewish descent." Genet Med. 10(1):54-6.
2. Wasserstein MP et al. (1993). "Acid Sphingomyelinase Deficiency"