Salla Disease

Salla disease is a rare genetic disorder. It is characterized by a gradual loss of muscle tone and coordination, as well as impaired growth, intellectual disability, and seizures. A person must have two variants in the SLC17A5 gene in order to have this condition.

Overview Scientific Details

Jamie, you **do not have the variant** 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

- To test for the R39C variant in the SLC17A5 gene.
- To identify carrier status for Salla 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 **Finnish** and **Swedish** descent.

You are likely not a carrier.

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

We ruled out the tested variant for Salla disease.

This variant is most common in people of **Finnish or Swedish** descent.

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

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

About Salla Disease

Also known as: Free Sialic Acid Storage Disease

When symptoms develop

Symptoms typically develop during infancy or childhood.

Typical signs and symptoms

- Intellectual disability
- Loss of muscle tone and coordination over time
- Seizures

Ethnicities most affected

This disease is most common in people of Finnish and Swedish descent.

How it's treated

There is currently no known cure. Treatment focuses on managing seizures and providing supportive care through speech, physical, and occupational therapy.

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

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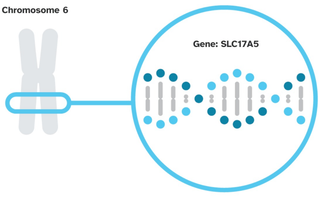
Overview | Scientific Details

Salla disease is caused by variants in the SLC17A5 gene.

SLC17A5

The SLC17A5 gene contains instructions for making a protein called sialin. Certain variants in SLC17A5 prevent the sialin protein from removing sialic acid from lysosomes (compartments within cells that break down and digest material). This results in a harmful buildup of sialic acid.

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
R39C **Gene:** SLC17A5 **Marker:** i5012634	**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 [2, 3, 5, 6, 7, 8, 9]** | **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 Finnish descent only.

- For people of partial Finnish descent, post-test carrier risk is less than that for those who are fully Finnish. The exact post-test risk depends on how much Finnish 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

Finnish	1 in 2,200	[2]

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Salla Disease is indicated for the detection of the R39C variant in the SLC17A5 gene. This test is intended to be used to determine carrier status for Salla disease in adults, but cannot determine if a person has two copies of a tested variant. The test is most relevant for people of Finnish and Swedish descent.

Special Considerations

- There are currently no professional guidelines in the U.S. for carrier testing for this condition.

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.

Finnish	91%	[2]
Swedish	85%	[2, 4]

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. Adams D et al. (2003). "Free Sialic Acid Storage Disorders." [Updated 2013 Jun 06].
2. Aula N et al. (2000). "The spectrum of SLC17A5-gene mutations resulting in free sialic acid-storage diseases indicates some genotype-phenotype correlation." Am J Hum Genet. 67(4):832-48.
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4. Erikson A et al. (2002). "Free sialic acid storage (Salla) disease in Sweden." Acta Paediatr. 91(12):1324-7.
5. Kleta R et al. (2003). "Biochemical and molecular analyses of infantile free sialic acid storage disease in North American children." Am J Med Genet A. 120A(1):28-33.
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7. Morin P et al. (2004). "Functional characterization of wild-type and mutant human sialin." EMBO J. 23(23):4560-70.
8. Strauss KA et al. (2005). "Genome-wide SNP arrays as a diagnostic tool: clinical description, genetic mapping, and molecular characterization of Salla disease in an Old Order Mennonite population." Am J Med Genet A. 138A(3):262-7.
9. Wreden CC et al. (2005). "Varied mechanisms underlie the free sialic acid storage disorders." J Biol Chem. 280(2):1408-16.

Change Log

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

Date	Change
Aug. 10, 2016	Salla Disease report created.