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.

J, you do not have the variant we tested.

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

0 variants detected
in the SLC17A5 gene

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.

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 is relevant for you because you have Finnish ancestry.

We ruled out the most common variant for Salla disease in people of Finnish descent.

You still have a chance of being a carrier for Salla disease.
You may still have up to a 1 in 2,200 chance of carrying a variant not covered by this test.

See Scientific Details

About Salla Disease

Also known as: Free Sialic Acid Storage Disease

When symptoms develop
Symptoms typically develop during infancy or childhood.

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.

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.

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

Salla Disease

**Scientific Details**

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.

Salla disease is caused by variants in the SLC17A5 gene.

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](https://ghr.nlm.nih.gov/gene/slc17a5)
You have no variants detected by this test.

<table>
<thead>
<tr>
<th>Marker Tested</th>
<th>Your Genotype*</th>
<th>Additional Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>R39C</td>
<td>G</td>
<td>- Biological explanation</td>
</tr>
<tr>
<td>Gene: SLC17A5</td>
<td>Typical copy from one of your parents</td>
<td></td>
</tr>
<tr>
<td>Marker: i5012634</td>
<td>G Typical copy from your other parent</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>- Typical vs. variant DNA sequence(s)</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- Percent of 23andMe customers with variant</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- References [2, 3, 5, 6, 7, 8, 9]</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.

Test Interpretation

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

<table>
<thead>
<tr>
<th>Ethnicity</th>
<th>Risk</th>
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<tbody>
<tr>
<td>Finnish</td>
<td>1 in 2,200</td>
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</table>

View technical article on estimating post-test carrier risk.
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 Ethnicties

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>
<th>Notes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Finnish</td>
<td>91%</td>
<td>[2]</td>
</tr>
<tr>
<td>Swedish</td>
<td>85%</td>
<td>[2, 4]</td>
</tr>
</tbody>
</table>

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for 47 samples with known variant status. 47 out of 47 genotype results were correct. 1 in 42,000 samples may receive a Not Determined result. 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.

References

## Change Log

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

<table>
<thead>
<tr>
<th>Date</th>
<th>Change</th>
</tr>
</thead>
<tbody>
<tr>
<td>Aug. 10, 2016</td>
<td>Salla Disease report created.</td>
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