ARSACS

ARSACS is a genetic disorder characterized by loss of sensation and muscle control, as well as muscle stiffness that worsens over time. A person may have two variants in the SACS gene in order to have this condition.

play+80658ac81, you have the variant we tested.

You could pass this variant on to your children.

1 variant detected
in the SACS 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 the condition, or you have any concerns about your results.

**Intended Uses**
- To test for the 80658ac81 variant in the SACS gene.
- To identify carrier status for ARSACS.

**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 French Canadian descent.

You are a carrier. You could pass the variant on to your children.

We detected one variant for ARSACS. People with only one variant are not expected to have ARSACS.

Your results may be relevant for you if you’re thinking about starting a family:
- If you and your partner are both carriers, each child may have a 25% chance of having this condition. Your relatives may also wish to consider testing if they plan to have children.

**About ARSACS**
Also known as: Autosomal recessive spastic ataxia of Charlevoix Saguenay

When symptoms develop:
Symptoms typically develop during early childhood.

**Typical signs and symptoms**
- Muscle stiffness that worsens over time
- Loss of sensation in hands and feet that worsens over time
- Impaired movement and balance that worsens over time

**Ethiobiotics most affected**
This condition is most common in people of French Canadian descent, specifically from the Charlevoix and Saguenay-Lac-Saint-Jean regions of Quebec.

**How it’s tested**
There is currently no cure for this disorder. Treatment focuses on managing symptoms and providing supportive care through speech, physical, and occupational therapy.

Read more at:
- Genomics Home Reference™
- GenomicsHomes
- March of Dimes Canada™

Consider talking to a healthcare professional if you are thinking about having children.

If you’re thinking about a family, a genetic counselor can help you and your partner understand additional testing might be appropriate.

Connect with a Genetic Counselor.

If you have other concerns about your results, consult with a healthcare professional.

Post Support.

Learn more about this condition and connect with support groups.
ARSACS is a variant in the SACS gene.

**Chromosome 15**

Gene: SACS

![Gene Diagram]

You have one variant detected by this test.

<table>
<thead>
<tr>
<th>Variant Detected</th>
<th>Genotype*</th>
<th>Additional Information</th>
</tr>
</thead>
<tbody>
<tr>
<td>c.540G&gt;A</td>
<td>(−)</td>
<td>isten copy from one of your parents.</td>
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<tr>
<td></td>
<td></td>
<td>A Typical copy from your other parent.</td>
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<td></td>
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<td><strong>Biallelic explanation</strong></td>
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<tr>
<td></td>
<td></td>
<td>- Typical vs. variant (DNA sequencing)</td>
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<tr>
<td></td>
<td></td>
<td>- Presence of 23andMe customers with variant</td>
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</tbody>
</table>
|                  |           | **References** [1] | [3d]

*For some variants, the Y-axis is shown from bottom to top. This does not affect the interpretation of the variant. The Y-axis orientation is arbitrary and does not affect the interpretation of the variant.

**Test Details**

**Indications for Use**

The 23andMe DNA Test for ARSACS is indicated for the detection of the c.540G>A variant in the SACS gene. This test is intended to be used to determine carrier status for ARSACS in adults, but cannot determine if a person has two copies of a tested variant. This test is most relevant for people of Persian or Jewish ancestry.

**Special Considerations**

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

**Test Performance Summary**

Carrier Detection Rate & Relevant Relatives

The “carrier detection rate” is an estimate of the percentage of carriers for this condition that would be identified by the test. Carrier detection rate differs by ethnicity and is provided only where sufficient data is available.

French-Canadian, particularly from the Charlevoix and Saguenay-Lac Saint-Jean regions of Quebec: 0.0% (6/5464)

**Analytical Performance**

Accuracy was determined by comparing results from this test with results from sequencing. Greater than 99.9% 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 ancestry are not commonly associated with this condition may be incorrect. Individuals in this situation should consult 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 Test Results for more details on the interpretation of this test.

**References**


**Change Log**

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

<table>
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<th>Change</th>
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<td>2013-05-10</td>
<td>ARSACS report created.</td>
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</table>

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