**ARSACS**

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

Erin, you **do not have the variant** we tested.

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

0 **variants detected** in the SACS gene

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**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

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**Intended Uses**

- To test for the 6594delT 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.

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You are likely not a carrier.

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

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**We ruled out the tested variant for ARSACS.**

This variant is most common in people of **French Canadian** descent.

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You still have a chance of being a carrier for ARSACS.

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

Also known as: Autosomal Recessive Spastic Ataxia of Charlevoix-Segueney

When symptoms develop
Symptoms typically develop during early childhood.

How it’s treated
There is currently no known cure. Treatment focuses on managing symptoms and providing supportive care through speech, physical, and occupational therapy.

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

Ethnicities most affected
This condition is most common in people of French Canadian descent, particularly from the Charlevoix and Segueney-Lac Saint-Jean regions of Quebec.

Read more at
- Genetics Home Reference
- GeneReviews
- Muscular Dystrophy Canada

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

ARSACS is caused by variants in the SACS gene.

The SACS gene contains instructions for making a protein called saccin, a protein of unknown function that is mainly present in the brain, skin cells, and skeletal muscles. Certain variants in SACS lead to a shortened protein that cannot function properly.

Read more at Genetics Home Reference
You have no variants detected by this test.

Test Interpretation

Post-Test Carrier Risk

This report provides an estimate of the post-test carrier risk for people of French Canadian descent only.

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

French Canadian, particularly from the Charlevoix and Saguenay-Lac-Saint-Jean regions of Quebec

1 in 340

[1.2]

Test Details

Indications for Use

The 23andMe PGx Carrier Status Test for ARSACS is indicated for the detection of the 6594delT 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. The test is most relevant for people of French Canadian 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.

French Canadian, particularly from the Charlevoix and Saguenay-Lac-Saint-Jean regions of Quebec

94%

[4]

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for samples with known variant status. Out of genotype results were correct. About 1 in 5,200 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.

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.

*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.
References


