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.

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 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.

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.

We ruled out the tested variant for ARSACS.

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

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-Saguenay

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 Saguenay-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.

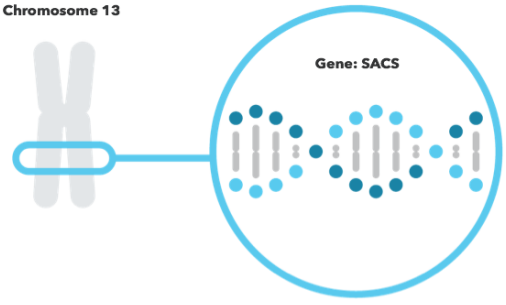
Learn more

ARSACS is caused by variants in the SACS gene.

SACS

The SACS gene contains instructions for making a protein called sacsin, 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.

Variants Detected	View All Tested Markers
0	1

Marker Tested	Your Genotype*		Additional Information
6594delT Gene: SACS Marker: i5012578	**A** Typical copy from one of your parents	**A** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [3 , 4 , 5] | 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 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 PGS 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.

References

1. De Braekeleer M et al. (1993). "Genetic epidemiology of autosomal recessive spastic ataxia of Charlevoix-Saguenay in northeastern Quebec." Genet Epidemiol. 10(1):17-25.
2. Dupré N et al. (2006). "Hereditary ataxia, spastic paraparesis and neuropathy in the French-Canadian population." Can J Neurol Sci. 33(2):149-57.
3. Engert JC et al. (2000). "ARSACS, a spastic ataxia common in northeastern Québec, is caused by mutations in a new gene encoding an 11.5-kb ORF." Nat Genet. 24(2):120-5.
4. Mercier J et al. (2001). "Rapid detection of the sacsin mutations causing autosomal recessive spastic ataxia of Charlevoix-Saguenay." Genet Test. 5(3):255-9.
5. Romano A et al. (2013). "Comparative analysis and functional mapping of SACS mutations reveal novel insights into sacsin repeated architecture." Hum Mutat. 34(3):525-37.
6. Vermeer S et al. (1993). "ARSACS"