Sjögren-Larsson Syndrome

Sjögren-Larsson syndrome is a rare genetic disorder. It is characterized by scaly dry skin, intellectual disability, and persistent muscle stiffness. A person must have two variants in the ALDH3A2 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 P315S variant in the ALDH3A2 gene.
- To identify carrier status for Sjögren-Larsson syndrome.

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 **Swedish** descent.

You are likely not a carrier.

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

We ruled out the tested variant for Sjögren-Larsson syndrome.

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

You still have a chance of being a carrier for Sjögren-Larsson syndrome.

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

About Sjögren-Larsson Syndrome

Also known as: FALDH Deficiency, Fatty Aldehyde Dehydrogenase Deficiency

When symptoms develop

Symptoms typically develop in infancy or early childhood.

How it's treated

There is currently no known cure. Treatment focuses on managing symptoms and providing supportive care through speech and physical therapy as well as skin care.

Typical signs and symptoms

- Dry scaly skin
- Persistent muscle stiffness
- Intellectual disability

Ethnicities most affected

This syndrome is most common in people of Swedish descent.

Read more at

Genetics Home Reference
GeneReviews
National Organization for Rare Disorders
Orphanet

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.

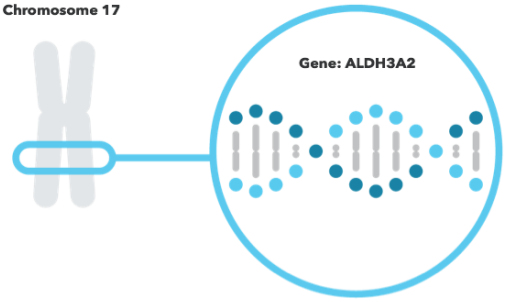
Learn more

Sjögren-Larsson syndrome is caused by variants in the ALDH3A2 gene.

ALDH3A2

The ALDH3A2 gene contains instructions for making a protein called fatty aldehyde dehydrogenase, also known as FALDH. This protein helps break down molecules called fatty acids to make energy. Certain variants in ALDH3A2 disrupt this protein's function, leading to a harmful buildup of fats inside of cells.

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
P315S Gene: ALDH3A2 Marker: rs72547571	**C** Typical copy from one of your parents	**C** Typical copy from your other parent	> **Biological explanation** > **Typical vs. variant DNA sequence(s)** > **Percent of 23andMe customers with variant** > **References** [1 , 2 , 4 , 6 , 7] | 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 Swedish descent only.

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

Swedish	1 in 1,200	[3]

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Sjögren-Larsson Syndrome is indicated for the detection of the P315S variant in the ALDH3A2 gene. This test is intended to be used to determine carrier status for Sjögren-Larsson syndrome in adults, but cannot determine if a person has two copies of a tested variant. The test is most relevant for people of 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.

Swedish	84%	[2]

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing for 79 samples with known variant status. 79 out of 79 genotype results were correct. About 1 in 62,000 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.

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 Laurenzi V et al. (1997). "Sjögren-Larsson syndrome is caused by a common mutation in northern European and Swedish patients." J Invest Dermatol. 109(1):79-83.
2. Gånemo A et al. (2009). "Sjögren-larsson syndrome: a study of clinical symptoms and dermatological treatment in 34 Swedish patients." Acta Derm Venereol. 89(1):68-73.
3. Jagell S et al. (1981). "Sjögren-Larsson syndrome in Sweden. A clinical, genetic and epidemiological study." Clin Genet. 19(4):233-56.
4. Keller MA et al. (2014). "A gatekeeper helix determines the substrate specificity of Sjögren-Larsson Syndrome enzyme fatty aldehyde dehydrogenase." Nat Commun. 5:4439.
5. Richard G et al. (1993). "Autosomal Recessive Congenital Ichthyosis"
6. Rizzo WB et al. (1999). "The molecular basis of Sjögren-Larsson syndrome: mutation analysis of the fatty aldehyde dehydrogenase gene." Am J Hum Genet. 65(6):1547-60.
7. Rizzo WB et al. (2008). "Abnormal fatty alcohol metabolism in cultured keratinocytes from patients with Sjögren-Larsson syndrome." J Lipid Res. 49(2):410-9.