Inheritest®
CARRIER SCREEN

# Sjögren-Larsson syndrome

## What is Sjögren-Larsson syndrome?

Sjögren-Larsson syndrome is an inherited disease. Individuals with Sjögren-Larsson syndrome a have a deficiency in the enzyme fatty aldehyde dehydrogenase, which breaks down fatty aldehydes and fatty alcohols. Symptoms associated with Sjögren-Larsson syndrome are attributed to the accumulation of fatty aldehydes and fatty alcohols in various tissues of the body.[1] Sjögren-Larsson syndrome is also known as fatty aldehyde dehydrogenase deficiency.

## What are the symptoms of Sjögren-Larsson syndrome and what treatment is available?

Sjögren-Larsson syndrome causes skin problems that are usually seen at birth. Neurological symptoms typically appear during the first or second year of life. Symptoms of Sjögren-Larsson syndrome may include:[1,2]

- Ichthyosis (dry, scaly or thickened skin)
- Intellectual disability, varying from mild to profound, with delays in motor and cognitive milestones
- Spastic diplegia or tetraplegia (a form of muscle stiffness), which delays walking in most affected individuals
- Seizures
- Abnormalities in the retina of the eye and photophobia (eye discomfort in bright light)
- Leg contractures (shortening of muscles, tendons, and/or ligaments that prevents normal movement)
- Pruritis (itching)
- Preterm birth

There is no cure for Sjögren-Larsson syndrome. Survival to adulthood is expected. Available treatments include the use of medications to address specific symptoms such as Ichthyosis.[2]

## How is Sjögren-Larsson syndrome inherited?

Sjögren-Larsson syndrome is an autosomal recessive disease caused by mutations in the *ALDH3A2* gene. An individual who inherits one copy of an *ALDH3A2* gene mutation is a carrier and is not expected to have related health problems.[3] An individual who inherits two *ALDH3A2* mutations, one from each parent, is expected to be affected with Sjögren-Larsson syndrome.

If both members of a couple are carriers of a mutation in the same gene, the risk of having an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

## Who is at risk for Sjögren-Larsson syndrome?

Sjögren-Larsson syndrome can occur in individuals of all races and ethnicities, and is most common in individuals of Swedish ancestry.[4] In Sweden, the annual incidence of Sjögren-Larsson syndrome is approximately 1 in 167,000, and the carrier frequency is approximately 1 in 200.[5]

Having a relative who is a carrier or who is affected can increase an individual's risk to be a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

## What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

**What does a negative test result mean?**

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

**Where can I get more information?**

FIRST (Foundation for Ichthyosis and Related Skin Types): www.firstskinfoundation.org

Genetic and Rare Diseases (GARD) Information Center: https://rarediseases.info.nih.gov/gard/7654/sjogren-larsson-syndrome/resources/1

**References**

1. Rizzo, WB et *al.* The Molecular Basis of Sjögren-Larsson Syndrome: Mutation Analysis of the Fatty Aldehyde Dehydrogenase Gene. *Am J. Hum Genet*. 1999; 65: 1547-1560.
2. Ganemo, A *et al.*. Sjögren-Larsson Syndrome: A Study of Clinical Symptoms and Dermatological Treatment in 34 Swedish Patients. *Acta Derm Venereol*. 2009; 89:68-73.
3. Rizzo, WB *et al*. *Sjögren-Larsson Syndrome: Diversity of Mutations and Polymorphisms in the Fatty Aldehyde Dehydrogenase Gene (ALDH3A2)*. Human Mutation 2005; 26(1): 1-10.
4. Sillen, A *et al*. A missense mutation in the FALDH gene identified in Sjogren-Larsson syndrome patients originating from the northern part of Sweden. *Hum. Genet.* 1997; 100: 201-203
5. Jagell S *et al*. Sjögren-Larsson syndrome in Sweden. A clinical, genetic and epidemiological study. *Clinical Genetics*. 1981; 19: 233-256.