What is Sjögren-Larsson syndrome?

Sjögren-Larsson syndrome is an inherited disease. Individuals with Sjögren-Larsson syndrome 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. 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:

- 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.

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. 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. In Sweden, the annual incidence of Sjögren-Larsson syndrome is approximately 1 in 167,000, and the carrier frequency is approximately 1 in 200.

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.
Sjögren-Larsson syndrome

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