What is Sjögren-Larsson syndrome?
Sjögren-Larsson syndrome (SLS) is an inherited disease characterized by an inability of the body to break down certain molecules called fatty aldehydes and fatty alcohols due to a deficiency in the activity of an enzyme called fatty aldehyde dehydrogenase (FALDH). Symptoms associated with SLS are attributed to the accumulation of fatty aldehydes and fatty alcohols in various tissues in the body\(^1\). SLS 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. The symptoms of SLS may include\(^1,2\):

- Ichthyosis (dry, scaly or thickened skin)
- Mental retardation, varying from mild to profound, with delays in motor and cognitive milestones
- Spastic diplegia or tetraplegia (a form of muscle stiffness), which delays the development of walking in most affected individuals
- Seizures
- Abnormalities in the retina (the tissue at the back of the eye) and photophobia (eye discomfort in bright light)
- Leg contractures (or tightening of the muscles or tendons that prevents the leg from moving normally)
- Pruritis (itching)
- Preterm birth

There is no cure for SLS, but survival to adulthood is expected. Available treatments include the use of oral retinoids and topical ointments to address specific symptoms such as ichthyosis\(^2\).

How is Sjögren-Larsson syndrome inherited?
SLS 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 mutations in this gene, one from each parent, is expected to be affected with SLS.

If both members of a couple are carriers, the risk for an affected child is 25% in each pregnancy. Therefore, it is important that the reproductive partner of a carrier be offered testing.

Who is at risk for Sjögren-Larsson syndrome?
SLS can occur in individuals of all races and ethnicities, but is most common in individuals of Swedish ancestry\(^5\). In Sweden, the annual incidence of SLS is approximately 1 in 167,000, and the carrier frequency is approximately 1 in 200\(^4\).

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
- NIH Office of Rare Diseases Research - Genetic and Rare Diseases (GARD) Information Center www.rarediseases.info.nih.gov/GARD/

References