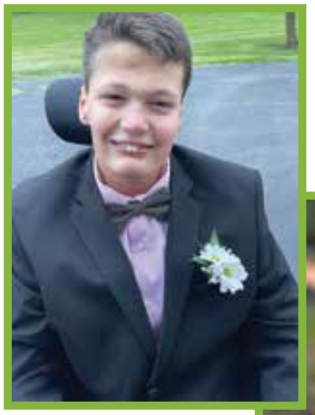

TREATMENT

There is currently no cure for SLS, but there are clinical trials underway. The skin can respond to the usual topical lotions and keratolytic agents, but the neurologic symptoms have no specific therapy.

UNMC SLS Research Fund is used for clinical and lab research. If you'd like to donate to the University of Nebraska Foundation all money goes to research and is tax deductible.
Scan QR code For More Info:

HELP

This document has been created by parents of those affected with SLS and is meant for informational purposes only. It is not designed to diagnose or treat SLS.

This information and much more can be found at:

www.sjogrenlarsson.com
www.firstskinfoundation.org
www.rarediseases.org

Although SLS is rare you are never alone. If you would like to get in touch with other SLS families, contact:
sjogrenlarsson@gmail.com
or Facebook@SLSfamilynetwork

For specialized doctor care in US or more information on upcoming trials and diagnosing, contact:

Dr. William Rizzo
University Of Nebraska
College Of Medicine
402-559-4000
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SJÖGREN-LARSSON SYNDROME FOUNDATION

2025

SJÖGREN-LARSSON SYNDROME FOUNDATION

www.sjogrenlarsson.com

Sjögren-Larsson syndrome (SLS) is a rare genetic disease affecting the skin and brain. Ichthyosis (dry scaly skin) is usually present at birth or shortly afterwards. Developmental delay and spasticity develop by two years of age. Brain white matter changes (leukodystrophy) appear by 5 years. SLS is caused by mutations in the *ALDH3A2* gene, which leads to abnormal metabolism of fatty alcohol and fatty aldehyde lipids. Symptoms of SLS vary widely in affected children and adults.

DIAGNOSIS

Genetic testing for SLS can be done with blood or cheek cell samples. SLS is only expressed when someone inherits two defective *ALDH3A2* genes.

SIGNS & SYMPTOMS

BIRTH

- Preterm birth is common

ICHTHYOSIS

- The outer surface of skin builds up
- Dry and rough scaly skin
- Extremely itchy
- Wax build up in ears

SPASTICITY

- Abnormal function of nerves causes muscle tightness
- Clonus
- Spastic Diplegia
- Apparent typically by age 2

SEIZURES/EPILEPSY

- Seen in many SLS patients
- May begin at various ages
- Likely to respond to anticonvulsant medications

VISION

- Tiny glistening white deposits that appear in retina
- Decreased vision treated with corrective lenses
- Photophobia

SPEECH

- Expressive speech delays
- Dysarthria

INTELLECTUAL

- Developmental delays from mild to severe
- Usually apparent during first few years of life

SYMPTOM MANAGEMENT

Management of SLS is multidisciplinary. Individuals are typically followed by a variety of specialists, including:

- Geneticist
- Neurologist
- Epileptologist
- Orthopedist
- Opthalmologist
- Dermatologist
- Otolaryngologist (ENT)
- Physiatrist
- Therapists such as occupational, speech and physical