

## FACT SHEET: SJOGREN-LARSSON SYNDROME

### What causes SLS?

SLS is caused by mutations in a gene called fatty aldehyde dehydrogenase, or FADH. The protein that is made by this gene is responsible for breaking down certain molecules called medium- and long-chain fatty aldehydes. If FADH is not functioning properly, these and related molecules build up in the body, leading to SLS.

### How is SLS diagnosed?

SLS can be diagnosed by a biochemical blood test that determines if FADH activity is normal. In addition, because mutations in FADH are known to cause SLS, the gene can be sequenced in order to determine if any mutations are present. This also provides the option of genetic and prenatal testing, which can allow parents to make informed decisions about having children.

### What are the symptoms of SLS?

SLS causes symptoms within the first two years of life. A primary feature of SLS is dry, scaly skin, which is called ichthyosis. In addition to these and can develop some or all of the following symptoms:

- Developmental delay
- Mental retardation
- Speech difficulties
- Seizures
- Spastic diplegia/tetraplegia: paralysis (diplegia is paralysis of both legs, tetraplegia is paralysis of all four limbs)
- Spasticity in the legs: leg spasms, which can impair motor abilities and waking
- Glistening white dots in the retina of the eye

- Pruritis: itching
- Preterm birth

### What is the treatment for SLS?

Some recent clinical studies have found that a drug called zileuton can be beneficial in the treatment of SLS. In these studies, the drug successfully reduced the severity of the pruritis (itching), and improved the behavior of the treated child. While this drug does not cure SLS, it has the potential to greatly improve the quality of life of children with SLS.

### How is scientific research on SLS progressing towards improved treatment or diagnosis?

The identification of mutations in FADH as the cause of SLS has allowed scientists to identify potential treatments for SLS, including zileuton, which was described above. In addition, this discovery is allowing scientists to develop improved methods of diagnosing SLS.

### Other Clinical Names for Sjogren-Larsson Syndrome

Other clinical names of Sjogren-Larsson Syndrome include:

- SLS
- Ichthyosis, Spastic Neurological Disorder, and Oligophrenia
- Fatty Alcohol: NAD<sup>+</sup> Oxidoreductase Deficiency
- FAO deficiency
- Fatty aldehyde dehydrogenase deficiency
- FALDH deficiency