

## Karl Gustaf Torsten Sjögren and Sjögren-Larsson syndrome

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Karl Gustaf Torsten Sjögren (1896-1974), (Figure 1), a Swedish psychiatrist and geneticist, was a pioneer of modern Swedish psychiatry.<sup>1,4</sup> Sjögren studied medicine at the University of Uppsala.<sup>1</sup> From 1932 to 1935, he was Head Physician and Director of Lillhagen Hospital in Gothenburg, and between 1935-1945, he was physician-in-chief at the psychiatric department of Sahlgrenska Hospital in Gothenburg. Sjögren was professor of psychiatry at Karolinska Institutet from 1945 to 1961 and was elected to the Royal Swedish Academy of Sciences in 1951.<sup>3</sup>

Sjögren was an expert of psychiatry for the World Health Organization.<sup>3</sup> Among his many contributions to medicine, he is credited for describing several medical conditions,<sup>5-8</sup> which were later named after him, including Graefe-Sjögren syndrome, Marinesco-Sjögren syndrome, and Sjögren-Larsson syndrome (SLS).

During his work on juvenile amaurotic idiocy, Sjögren forged a collaboration with Tage K.L. Larsson, a statistics lecturer at the University of Lund.<sup>3</sup> Their study on the combination of oligophrenia, congenital ichthyosis, and spastic disorders in 1957 established the clinical and genetic profile of a new disease entity, later known as Sjogren-Larsson syndrome (SLS).<sup>3</sup>

### Sjögren-Larsson syndrome

Sjögren-Larsson syndrome – also known in OMIM as ichthyosis, spastic neurologic disorder, and oligophrenia – is a rare autosomal recessive condition comprising congenital ichthyotic hyperkeratosis, spastic diplegia, mild to moderate mental retardation, and retinopathy.<sup>9-12</sup> Hair, nails, and the ability to sweat are generally normal. SLS was originally described in Swedish patients, but over 200 cases have been reported worldwide in all races. The incidence in Sweden is 1 in 100,000, rising to 1 in 10,000 in the northwest region of Vasterbotten.<sup>10</sup>

SLS presents at birth as ichthyosis that ranges from fine scaling to generalized hyperkeratosis. Preterm birth is common and is been attributed to abnormal leukotriene B4 inactivation.<sup>10</sup> Velvety orange or brown lichenification, often with verrucous hyperkeratosis, develops noticeably in and around the flexures, neck, and midabdomen. The lichenification progresses slowly and is embarrassing to older



**Figure 1.** Karl Gustaf Torsten Sjögren (1896-1974). Image is provided by the Center for History of Science, the Royal Swedish Academy of Sciences. Permission for republication is granted by Norstedts, Sweden.

children, in whom it can restrict the mobility of already hypertonic limbs.<sup>10</sup>

In the first 2 to 3 years, SLS patients can develop neurological manifestations, such as spastic diplegia or tetraplegia and mental retardation, and can be accompanied by defects in speech. Glistening white dots develop in the macula of the retina after 1 year of age but are not present in all patients. Most SLS patients never walk<sup>9</sup> and tend to have short stature. About half of SLS patients experience seizures. The histological findings of hyperkeratosis, papillomatosis, acanthosis, and a mildly thickened granular layer are nonspecific.<sup>11</sup>

SLS is caused by a deficiency in fatty aldehyde dehydrogenase (FALDH), which maps to 17p11.2. Mutations occur in the FALDH gene (ALDH10, recently renamed ALDH3A2).<sup>11</sup> Clinical improvements occurs with fat restriction and supplementation with medium-chain triglycerides.<sup>9</sup>

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