

# Case Study on Rare Disease: Sjogren-Larsson Syndrome

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**Abstract:** I report a case of Sjögren-Larsson syndrome (SLS) which is a rare autosomal recessive disorder with clinical outline (spastic diplegia, ichthyosis, mental retardation). So, basically this disorder characterized by triad of congenital ichthyosis, spastic paresis, and mental retardation. It is an inherited fault of lipid metabolism triggered by absence of fatty aldehyde dehydrogenase enzyme.

**Key words:** Sjogren-Larsson syndrome, Congenital ichthyosis, mental retardation, spastic paresis

## 1. Introduction

Sjögren-Larsson syndrome (SLS) is a recessively inherited disease with congenital ichthyosis, spastic diplegia or tetraplegia, and mental retardation, caused by a deficiency of fatty aldehyde dehydrogenase. [1] SLS is an inborn error of lipid metabolism caused by a deficiency of the microsomal enzyme fatty aldehyde dehydrogenase (FALDH), a component of fatty alcohol: NAD-oxidoreductase enzyme complex. FALDH deficiency may lead to an accumulation of long-chain fatty alcohols with structural consequences for cell membrane integrity which disrupt the barrier function of skin and the white matter of the brain. [2]

## 2. Case Report

A 2 year old male child born of non-consanguineous marriage presented to paediatrics opd with complain of inability to speak, inability to walk since one year of age. Skin lesion present all over body since one year of age which started gradually, dry, non – itching and scaly. No other complaints at present.

On detailed history the child did not sitting without support, not standing with and without support and not walking and running. In addition patient has attained neck holding at six months which was gradually decreased by nine months of age and patient was having no mental retardation. On fine motor pincer grasp not achieved. On CNS examination tone of both upper limb was decreased, deep tendon reflex was brisk and ankle clonus +++. Other skeletal, eyes, ear examination was normal. Scaly skin lesion present over front and backside of body suggestive of lamellar ichthyosis.

All routine investigations (blood) was within normal limits, chest x-ray, usg abdomen was s/o normal study. **MRI BRAIN** done which s/o near symmetric non enhancing altered signal areas in bilateral periventricular white matter of fronto-parietal lobes and corpus callosum, **MR spectroscopy** shows lipid peak at 1.3 ppm and small peak at

0.8 ppm. This imaging findings along with clinical features are diagnostic of Sjogren-larsson syndrome.

## 3. Discussion

In 1957, Sjogren and Larsson described a rare neurocutaneous syndrome featured with skin changes like ichthyosis and neurological changes like spastic diplegia or tetraplegia, aphasia and dementia.

SLS is an inherited neurocutaneous disorder caused by mutations in the ALDH3A2 gene that encodes fatty aldehyde dehydrogenase (FALDH). More than 70 mutations in ALDH3A2 have been discovered in SLS patients including amino acid substitutions, deletions, insertions, and splicing errors. [3] FALDH catalyses the oxidation of long chain aldehyde to fatty acids. Due to deficiency of this enzyme, there is an accumulation of aldehyde-modified lipids or fatty alcohol in the skin and in the myelin. [4]

In Sjogren-larsson syndrome there is usually spastic diplegia occasionally tetraplegia with mental retardation, epilepsy, speech defects, dental, dermatological and retinal changes. Skin changes are in form of ichthyosis which is a generalized hyperkeratosis of the trunk, joints, and the dorsal aspects of the hands and the feet. Most patients have erythema at birth with worsening of cutaneous symptoms during the first year of life. Pruritus is a prominent feature that is not found in other types of ichthyotic skin disorders. [4] Photophobia, macular dystrophy and decreased visual acuity are the most prominent ophthalmologic abnormalities and may be caused by accumulation of long-chain fatty alcohols or fatty aldehydes. [4] These features are seen in one third of cases but not seen in our case.

MRI shows diffuse symmetrical white matter hyperintensities on T2W sequence especially in periventricular frontal, parietal lobes, corpus callosum, and corona radiata. Typically, subcortical white matter U fibers are spared. [2] In our case, Near symmetrical altered signal areas are seen in bilateral in white matter of fronto-parietal

lobes as well as corpus callosum (more in genu), this shows hyperintense signal seen on the T2W, FLAIR images and hypointense signal on T1W images.

In conclusion, the diagnosis of SLS should be considered in a patient with congenital ichthyosis and emerging

neurological features. One should look for ocular features and pruritus to make the diagnosis. Cerebral MRI reveals arrested myelination or demyelination in white matter and lipid peak on spectroscopy help in making the diagnosis.



Figure 1: Showing skin lesion (lamellar ichthyosis) ]

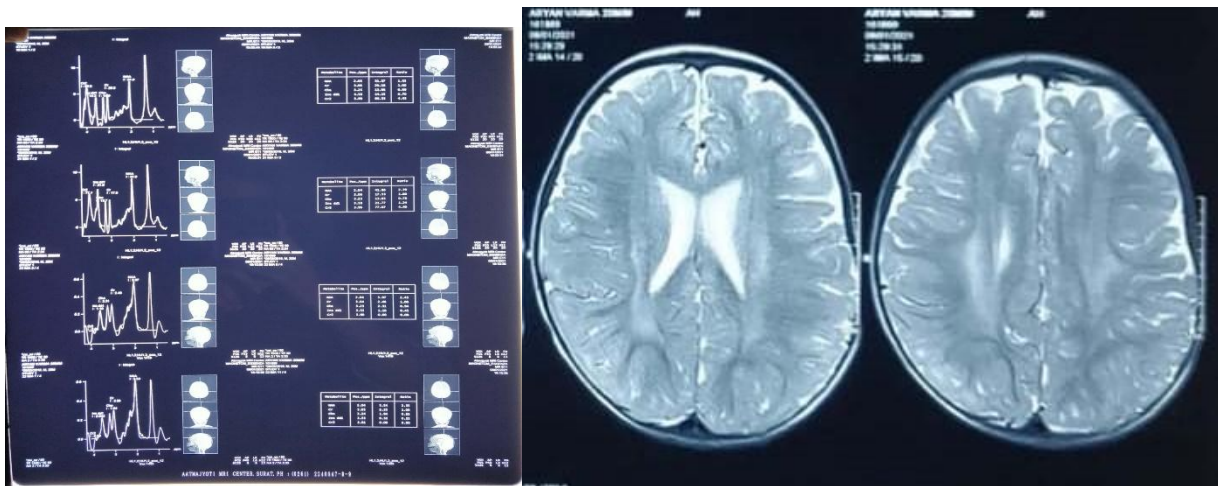


Figure 2: Showing MRI findings

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