Beyond retina in Sjogren–Larsson syndrome

Neelam Pawar, Meenakshi R, Devendra Maheshwari, Fathima A, Sabyasachi Chakrabarty, Mihir Paranjape

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Sjogren–Larsson syndrome (SLS) is a triad of generalized ichthyosis, mental retardation, and spastic paralysis caused by accumulation of fatty aldehydes and fatty alcohols in body tissues.[1] The pathognomonic ocular manifestations include crystalline juvenile macular dystrophy, cystoid foveal atrophy, and lack of macular pigment due the degeneration of Muller cells in the inner retina, retinal pigment epithelium (RPE) compromise from lipofuscin accumulation, leading to progressive RPE atrophy.[2–4] These changes generally start from the age of 2 and progress with the age.

The disease was first described 50 years ago by Sjogren from Northern Sweden, where maximum number of SLS patients have been reported.[1] The estimated prevalence in Sweden is about one in 250,000.[2] However, isolated cases have been described worldwide.[3–5] Diagnosis requires measurement of fatty aldehyde dehydrogenase (FALDH) activity in cultured fibroblasts or mutation analysis of the FALDH gene. Fatty aldehydes are harmful products to the cells when they exceed physiological levels. Various mechanisms have been postulated for this cytotoxicity.[2]

Our patient had characteristic features of SLS. He had erythematos and dry scaly lesions all over body with flexion contractures and often complained of pruritis [Fig. 1a and b]. He had visual acuity of 3/60 in both eyes with severe photophobia. He had bilateral posterior subcapsular cataracts [Fig. 2] for which he underwent uneventful phacoemulsification with hydrophobic acrylic with posterior capsulorrhexis and anterior vitrectomy under general anesthesia. Postoperatively, he had a vision of 6/12 vision in each eye. Fundus photograph showed crystalline deposits and pigmentary changes with atrophy in both eyes [Fig. 3a and b]. The autofluorescence images in both eyes showed lack of normal fundus autofluorescence (FAF) attenuation at the fovea [Fig. 3c and d].

Discussion

SLS is defined by the classical triad of ichthyosis, mental retardation, and spastic paraparesis. Additional findings include preterm birth, pruritis, macular dystrophy, leukoencephalopathy, and seizures.[1,2]

The ichthyosis in SLS is present since infancy. The skin at birth is erythematos and hyperkeratotic which over days appear as a dry scaly lesions over the entire body. The ichthyosis is mainly seen in flexural areas of the neck and lower abdomen.

SLS patients have delayed motor milestones, hypertonia, brisk deep tendon reflexes, and spasticity. Most of them have contractures which make it difficult for them to walk; majority becomes wheelchair bound. Isolated seizures occur in less than 50% of the patients. Magnetic resonance imaging of brain in these patients usually demonstrates hyperintense signal changes in the periventricular and deep white matter on FLAIR axial view. These patients show clinical improvement with a low-fat diet supplemented with medium chain fatty acids.

SLS patients demonstrate characteristic ophthalmic features ranging from retinopathy to cataracts.[6,5]

The main ocular features described in SLS patients are crystalline juvenile macular dystrophy, cystoid foveal atrophy, and lack of macular pigment.[6] Papathemeli et al.[7] reported high refractive errors and others have reported blepharo conjunctivitis, ichthyosis of lids, and photophobia in these patients. The occurrence of various eye abnormalities could
be due to direct toxic mechanisms of abnormal metabolic products or accumulation of normal metabolites by errors of synthetic pathways or by deficient energy metabolism. The exact pathophysiology of cataract in SLS patients is not clearly evident.

Ganemo A et al. found glistening dots around the foveola in 35 Swedish cases with SLS.\(^2\) Van der Veen et al.,\(^5\) Nanda et al.,\(^3\) and Willemsen described FAF changes in the SLS patient with lack of normal FAF attenuation at the fovea.\(^4\)

Cataract screening should be done in all patients with SLS. A multidisciplinary team of neurologist, dermatologist, ophthalmologist, and rehabilitation specialist is required in their management. SLS patients should have routine ophthalmologic examination as cataract may be underreported in this syndrome.

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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**Conflicts of interest**

There are no conflicts of interest.

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