Beyond retina in Sjogren–Larsson syndrome

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

Key words: Cataract, crystalline retinopathy, Sjogrens–Larsson

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 erythematous 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 erythematous and hyperkeratotic which over days appear as a dry scaly lesions over the entire body. The icthiyosis 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 wheel-chair 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.[4,5]

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

Access this article online

Quick Response Code:

Website: www.ijo.in

DOI: 10.4103/ijo.IJO_2994_21

Department of Pediatric Ophthalmology and Strabismus, Aravind Eye Hospital, Tirunelveli, Tamil Nadu, India

Correspondence to: Dr. Neelam Pawar, Aravind Eye Hospital, Tirunelveli - 627 001, Tamil Nadu, India. E-mail: drneels2pawar@yahoo.co.in

Received: 12-Dec-2021 Revision: 16-Jan-2022
Accepted: 29-Apr-2022 Published: 30-Jun-2022

Cite this article as: Pawar N, Meenakshi R, Maheshwari D, Fathima A, Chakrabarty S, Paranjape M. Beyond retina in Sjogren–Larsson syndrome. Indian J Ophthalmol 2022;70:2727-8.

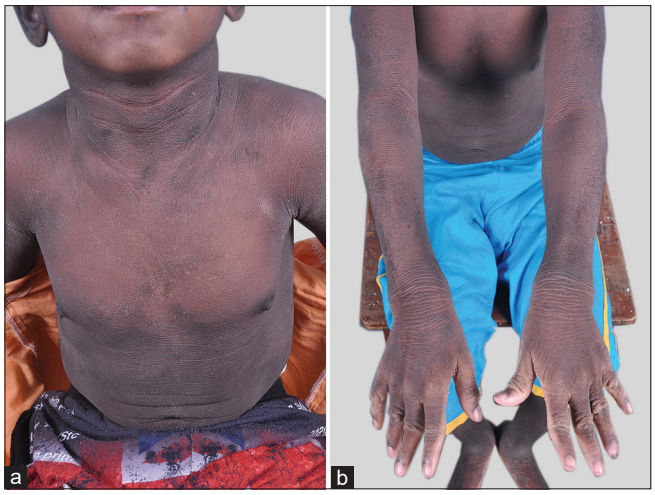

Figure 1: (a and b) The characteristic thickening and hyperpigmentation of axillae, flexion contractures in leg and abdomen, respectively

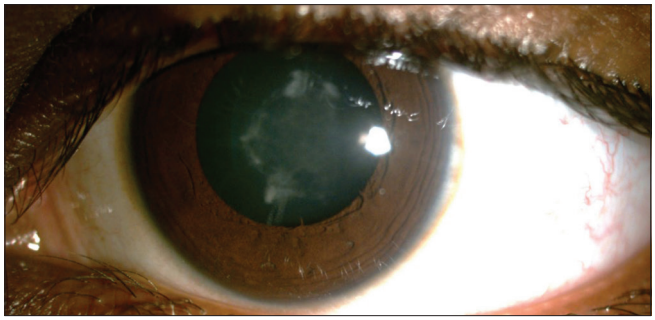

Figure 2: The clinical photo showing posterior capsular cataract in this patient

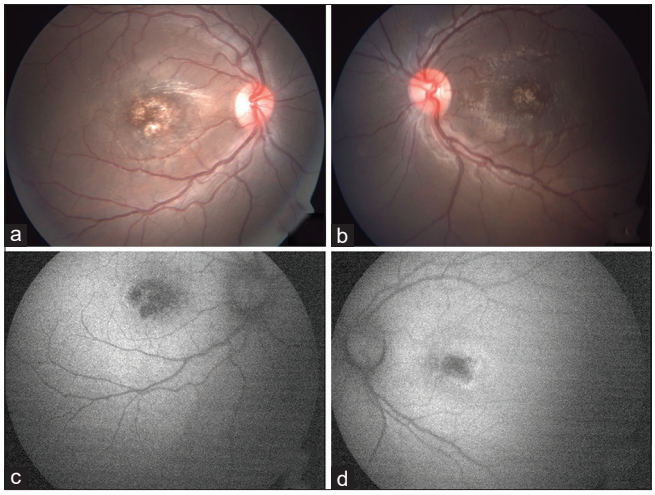

Figure 3: (a and b) Fundus photo exhibited intraretinal crystals in the macular region in both eyes along with retinal pigmentary changes and atrophy. (c and d) The autofluorescence images in both eyes showed lack of normal FAF attenuation at the fovea

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.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

References

1. Sjögren T, Larsson T. Oligophrenia in combination with congenital ichthyosis and spastic disorders. Acta Psychiatr Neurol Scand Suppl 1957;32(suppl113):1-13.
2. Ganemo A, Jagell S, Vahlquist A. Sjogren-Larsson syndrome and a study of clinical symptoms and dermatological treatment in 34 Swedish patients. Acta Derm Venerol 2009;89:68-73.
3. Nanda T, Kovach JL. Ophthalmic findings in late stage Sjogren-Larsson syndrome. Retin Cases Brief Rep 2019;13:251-4.
4. Willemsen MA, Cruysberg JR, Rotteveel JJ, Aandekerk AL, Van Domburg, PH, Deutmal AF. Juvenile macular dystrophy associated with deficient activity of fatty aldehyde dehydrogenase in Sjögren-Larsson syndrome. Am J Ophthalmol 2000;130:782-9.
5. Van der Veen RL, Fuijkschot J, Willemsen MA, Cruysberg JR, Berendschot TT, Theelen T. Patients with Sjögren-Larsson syndrome lack macular pigment. Ophthalmology 2010;117:966-71.
6. Fouzdar-Jain S, Suh DW, Rizzo WB. Sjögren-Larsson syndrome: A complex metabolic disease with a distinctive ocular phenotype. Ophthalmic Genet 2019;40:298-308.
7. Papathemeli D, Mataftsi A, Patsatsi A, Sotiriadis D, Samouilidou M, Chondromatidou S, *et al.* Atypical presentation of Sjögren-Larsson syndrome. Case Rep Pediatr 2017;2017:7981750.