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Genetics of Sjögren Larsson Syndrome and a Case Report from India

Arvind Rup Singh[1], Jai Rup Singh[2], Harshinder Kaur[3], Gurpal Singh Sachdeva[4], Anupam Kaur[2] and Amrita Darshan Singh[2]

1. A-7, Guru Nanak Dev University Campus, Amritsar, India
2. Centre for Genetic Disorders, Guru Nanak Dev University, Amritsar, India
3. Department of Paediatrics, Govt Medical College, Patiala, India
4. Department of Medicine, Govt Medical College, Patiala, India

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ABSTRACT Sjögren Larsson syndrome (SLS) is a rare autosomal disorder that is characterised by congenital ichthyosis, spastic diplegia or qudriplegia and mental retardation. It is caused by the deficiency of the enzyme, fatty aldehyde dehydrogenase (FALDH) that is required for the oxidation of fatty alcohol to fatty acid. The metabolism of leukotrine B4 (LTB4) has also been reported to be defective in SLS patients. The gene, ADLH3A2, encoding for FALDH has been localised at 17p11.2 and mutations in it cause SLS. The worldwide frequency of SLS is reported to be less than 1: 100,000 births but rarely a case has been reported from India. This article reviews the genetic factors in SLS and reports a case of SLS from India, with two similarly affected sibs. The management of SLS including genetic counselling and prenatal diagnostic possibilities are also discussed.

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