"FISHY SKIN"- SJOGREN LARSSON SYNDROME : CASE REPORT WITH REVIEW OF CLINICAL FEATURES AND LITERATURE

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ABSTRACT

GOAL

To understand Sjögren-Larsson syndrome (SLS) and to better manage patients with this condition.

OBJECTIVES

1. Describe the clinical features of SLS.
2. Discuss the genetic component of SLS.
3. Identify potential therapeutic options for patients with SLS.

Sjogren-Larsson syndrome (SLS) is a rare autosomal recessive disorder characterized by triad of congenital ichthyosis, spastic paresis, and mental retardation. We report a case of sjogren-larsson syndrome, discussed along with clinical features & review of literature.

KEYWORDS: Ichthyosis, Onychonycosis, Spastic Paraplegia, Furfuraceous

Sjogren-Larsson syndrome, first described by Sjogren in 1956, and then jointly with Larsson in 1957, is known to occur in different populations throughout the world (Blumel, Watkins, and Eggers, 1958; Richards, 1960; Link and Roldan, 1958; Zaleski, 1962; Heijer and Reed, 1965;Selmanowitz and Porter, 1967)1. Shortly after that the autosomal recessive pattern of this disease in bigger cohorts was established.2,3 Thirty years later in its initial description, the deficiency of fatty aldehyde dehydrogenase (FALDH) was identified for the disease4,5 and accumulation of long-chain fatty alcohols and modificationof macromolecules by an excess of fatty aldehydeswere considered to be the pathophysiologicmechanisms causing the manifestations of SLS.6Prevalence of SLS is estimated to be 1 in every 1000mentally retarded cases and 1 in every 2500 dermatologiccases.7 The clinical features of SLS is developed during fetal and infantile period8. (Table 1)Skin findings are present at birth and include varying degrees of erythema and ich¬thyosis. The ichthyosis

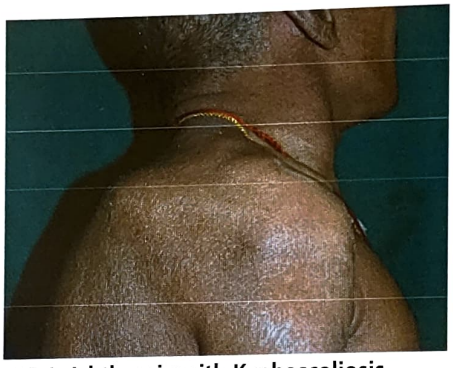
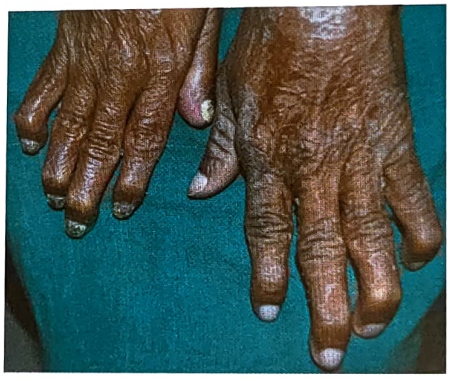


FIG 1: Ichthyosis with Kyphoscoliosis



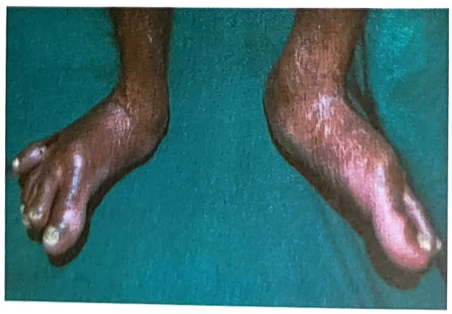


FIG 2: Deformity of bilateral upper and lower limb(spastic paraplegia), nail changes (onychonycosis)

ranges from a fine white scale to larger plaquelike scales or nonscaling hyperkeratosis with accentuation of skin markings.8

CASE REPORT

A 25 year old male patient visited the Department of Oral Medicine and Radiology with a chief complaint of multiple decayed and mobile teeth. Detail medical history revealed full term normal vaginal delivery(FTNVD) , with history of colloidal membrane at birth, diffuse erythema with fine scaling all over the body since birth (ichthyosis) (Figure 1), itchiness all over the body(pruritis), loss of hair(alopecia), nail changes(onychonycosis)(Figure 2), inability to walk and deformity of bilateral upper and lower limb(spastic paraplegia) (Figure 2), short stature (Figure 3), mental retardation, ocular changes(photophobia), inadequate speech, phonation , hearing defects, delayed milestones with delayed development of dentition.

On intra oral examination, generalized enamel hypoplasia, generalized mobility, loosening and shedding of teeth with gingivitis and poor hygiene control was observed. Orthopantomogram revealed decreased bone density, multiple missing teeth with history of self-exfoliation, microdontia of all the third molars, hypoplastic enamel and dentin and generalized loss of lamina dura.(Figure 4). Hand-wrist (Figure 5), PA-skull (Figure 6) & lateral spine radiographs (Figure 7) were also taken. Patient was on Tablet Terbinafine 250 mg once a day with topical emollients and keratolytics.

DISCUSSION

Sjögren-Larsson syndrome (SLS)sometimes called T9is arare neurocutaneousdisorder characterized by severe mental retardation, spastic di-or tetraplegia and congenital ichthyosis.10 SLS iscaused due to recessive mutations in the fatty aldehydedehydrogenase (FALDH) gene FALDH3A2 on the short arm ofchromosome 17p11.11 More than 72 mutations in FALDH3A2are known in SLS patients.2 This results in the accumulation of fatty alcohols in the skin and myelin of the central nervous system.12Very high prevalence of SLS has beenobserved in north east of Swe-

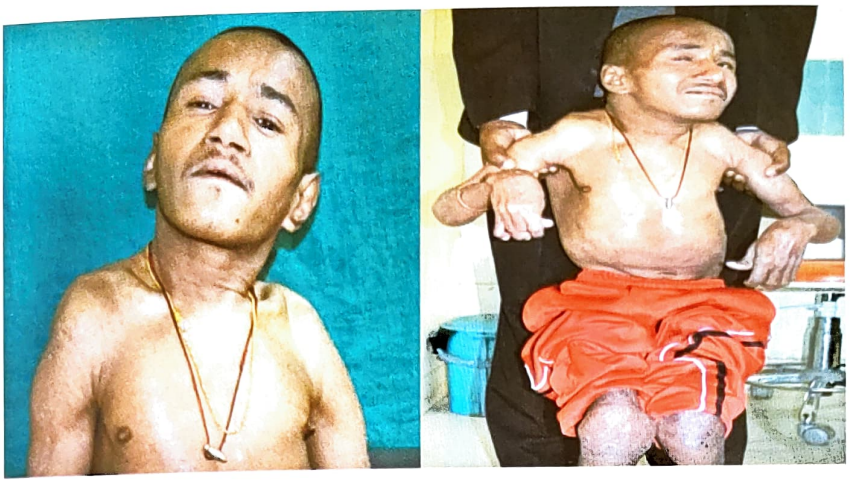


FIG 3: Short stature

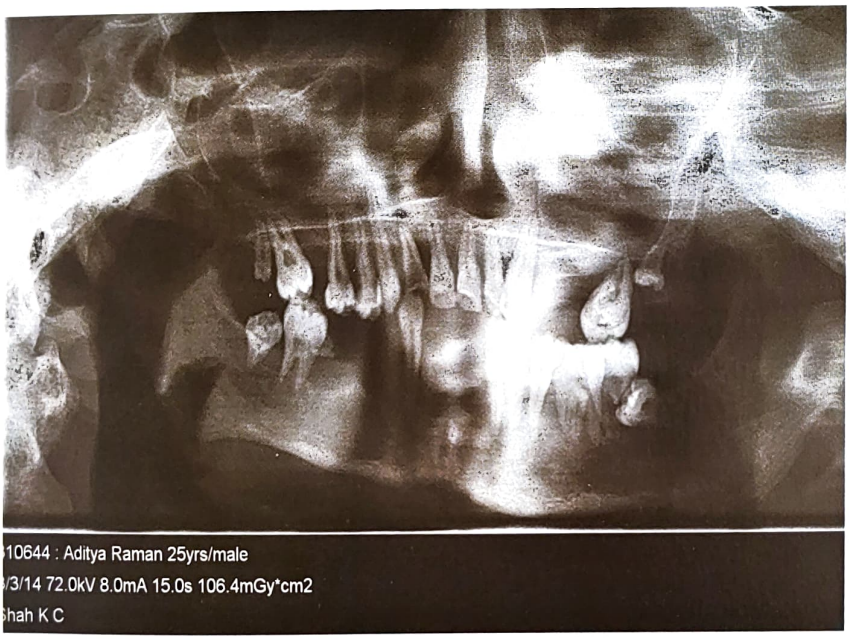


FIG 4: On orthopantomogram, decreased bone density, multiple missing teeth with self-exfoliation (history), microdontia of all the third molars, hypoplastic enamel and dentin and generalized loss of lamina dura.

den where an incidentof 8.3:100,000 births have been reported.13

The ichthyosis of SLS is characteristically of three types:furfuraceous, lamellar or non-scaly hyperkeratoticthickening of the stratum corneum. Lamellar ichthyosisis present on the legs. The non-scaly hyperkeratosisproduces a characteristic accentuation of the skinmarkings in the flexures. The hair, nails and teeth areunaffected.14

The occurrence of glistening dots on fundusexamination strongly suggests SLS.14Neurologic signs are nonspecific; however, by 1 to 2 years of age, severe motor and mental develop-mental delay usually is obvious. Spasticity may be apparent before 3 years of age and is more severe in the lower limbs than in other parts of the body.15Non-progressive mental retardation, associated with delayed or impaired speech, is an invariable feature of classical SLS and can range from mild to severe.

Diagnosis of SLS is suggestedwhen both neurologic and cutaneous symptoms arerecognized simultaneously.The histopathological features are nonspecific and includehyperkeratosis and acanthosis with a normal granular layer.The diagnosis of SLS can be confirmed by measurementof enzyme activity in cultured skin fibroblasts or leucocytes.Sequence analysis of FALDH3A2 gene causing mutations ishighly sensitive and also detects possible carrier.16

In SLS, MR imagingabnormalities of the brain are confined to the cerebralwhite matter and the corticospinal tracts. They consist ofthe accumulation of lipid substrates, delayed myelination,periventricular gliosis, and a permanent myelin deficit.17 In atypical cases, thedifferential diagnosis might encompass peroxisomal disorders(e.g. Refsum disease), and many

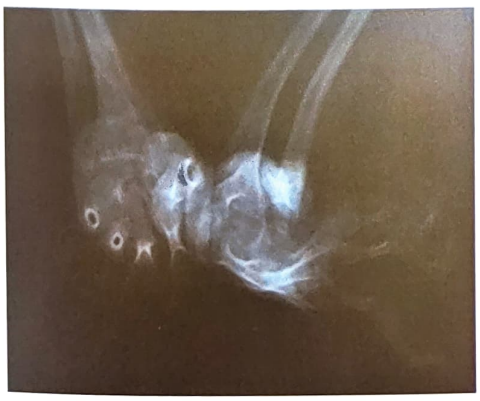


FIG 5: Hand – wrist Radiograph

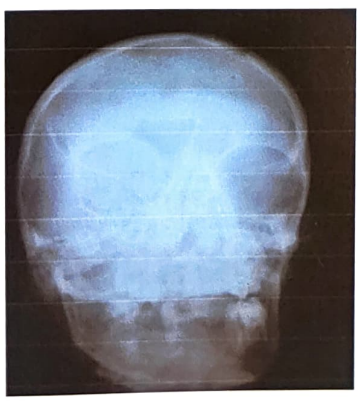


FIG 6: PA- Skull Radiograph

FIG 7: Lateral Spine Radiograph

TABLES

MAJOR FEATURES	MINOR FEATURES
• Ichthyosis • Glistening white dots on the retina • Mental retardation • Seizures • Short stature • Spastic diplegia or quadriplegia • Speech defects	• Enamel hypoplasia • Hypertelorism • Kyphoscoliosis • Macular degeneration • Metaphyseal dysplasia • Wide-spaced teeth

Table 1: Major & Minor Clinical Features inSjogren-Larsson Syndrome

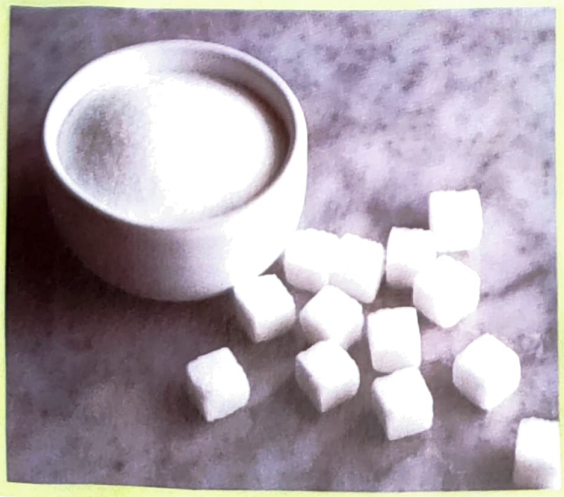
other raresyndromes which involve multiple organs.18 Management in SLS is supportive.19There is no permanent cure for SLS and no specific therapy.Multidisciplinary approach includes team of dermatologist,neurologist, ophthalmologist, orthopaedician, andphysiotherapist. Topical emollients, keratolytics, calcipotriol,and oral retinoids can be used to improve the cutaneoussymptoms.13

SUGAR'S IMPACT ON TOOTH DECAY STILL MAJOR CONCERN

Many people don't realize it but the adverse impact of sugar intake is just as bad for teeth as it is for the increased risk of obesity, type 2 diabetes and heart disease.The information came to the forefront again because of new plans soon to be underway in the United Kingdom. Food and all things that a person consumes are just as important to oneís oral health as other aspects. This issue is a pressing matter based on the amounts of sugary drinks children consume these days.

Toothy decay results from the acid produced when sugar and oral bacteria combine. A review of studies conducted by the World Health Organization supports the link involving the level of sugar consumed and the onset of cavities. The risk of tooth decay is reduced when the level of sugar intake is less then 10 percent of the caloric intake.

Even though fluoride is readily available, tooth decay is a major health concern. That's the reason these new plans are being put in place. There are also certain new policies looking to be recommended, such as not having added sugar contribute more than 5 percent of total energy intake. Also, one of the key goals is to reach an industry standard as far as reducing sugar in processed foods and drinks.



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