CASE REPORT

Lamellar Ichthyosis: A Rare Mucocutaneous Disease

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Abstract:

Lamellar Ichthyosis is a rare genodermatotic condition, which occurs due to the mutation in the trans-glutaminase-1gene. It is a rare disease with a global prevalence of 1 in 300000 live births with no specific gender predilection. It is a rare genetic disorder with a characteristic fish scale appearance of the skin. Here we report one such rare case of Lamellar Ichthyosis in a 7-year-old girl reported to us with difficulty in chewing.

Keywords: Fish Scale, Genodermatosis, Lamellar Ichthyosis

Introduction:

Lamellar Ichthyosis (LI) is an autosomal recessive condition primarily affecting the skin. This disorder may inherit or acquired, which may be limited to the skin or appear as a multisystem disorder. This disorder occurs due to the mutation in the TGM1 gene, which codes for trans-glutaminase 1. It is a rare disease with a global prevalence of 1 in 300000 live births with no specific gender predilection [1]. It is seen more commonly in some communities in Norway and in children born out of consanguineous marriages. This disorder is extremely rare in the Indian population. It is a severe disorder of skin present at birth and continues throughout life with periods of remission and exacerbations. In this disorder, the epidermal layers of skin become cornified and lead to defect in keratinization. The skin is covered with dark, large, polygonal, thick plate-like scales giving the appearance of typical "fish scales". Other structures, which can be involved, are eye, ear, hair, nails, and teeth. Our case was classified as nonsyndromic Autosomal Recessive Congenital Ichthyosis (ARCI) based on the worldwide first Ichthyosis Consensus Classification (2009) [2]. Here we are reporting a case of LI in a 7-year-old girl with characteristic fish scale skin lesions with oral findings.

Case Report:

A 7-year-old female child, born of a non-consanguineous marriage reported to the Department of Oral Medicine and Radiology, Manipal with the chief complaints of pain in lower left back tooth for one week and difficulty in chewing due to multiple missing teeth. Her mother revealed that she had a complete set of deciduous dentition but reported early exfoliation leading to difficulty in chewing. A detailed medical history revealed that she was a diagnosed case of LI. There was no history of a similar medical condition in her family members. The dermatological examination revealed that she had hyperhidrosis and often had heat intolerance. Ophthalmologic examination revealed the presence of abnormal eversion in her eyelid resulting in lacrimation and photo-sensitivity. She had normal IQ and was home-schooled. Extraoral examination revealed that her

entire body was covered with typical thick brown fish scales (Figs.1 and 2). Her scalp was dry and covered with the plate-like scales, and hair was sparse. No nail abnormalities detected. The mouth opening was restricted due to multiple angular fissuring in the lower lip.

Intraoral examination revealed grossly decayed 55, 36, and 46 with multiple missing teeth in relation to 53,54,64,65,74,75,85. Retained root stumps were present about 61 and 84. No soft tissue abnormalities noted. The oral mucosa appeared less hydrated with no anomalies in texture suggestive of mild xerostomia. A panoramic radiograph was advised which confirmed deep caries approximating pulp with respect to 55, 36 and 46 (Fig. 3) with evidence of root resorption about 63,73. The dental treatment was instituted with the preventive approach as the patient was at high risk of caries. Full mouth oral prophylaxis was done followed by fluoride application. The patient was advised pulpectomy concerning 36, 46, and fabrication of space maintainers for edentulous regions 54, 64, 65, 74, 75, 85. She was referred to a dermatologist for her skin lesions and treated with moisturizers to lubricate the skin.

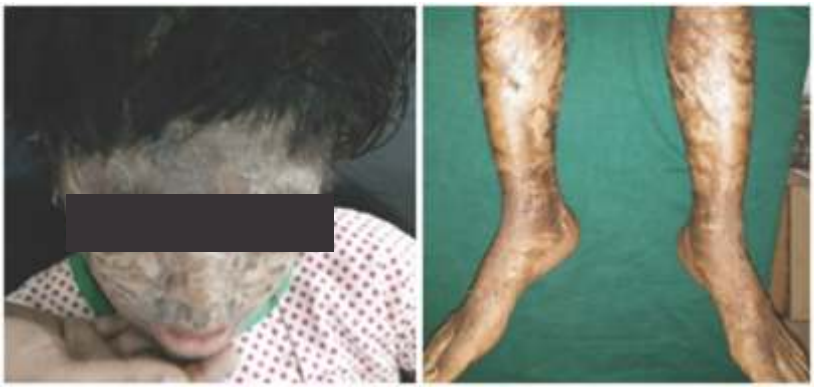

Fig. 1 and Fig. 2: Typical Fish Scale Appearance of Skin

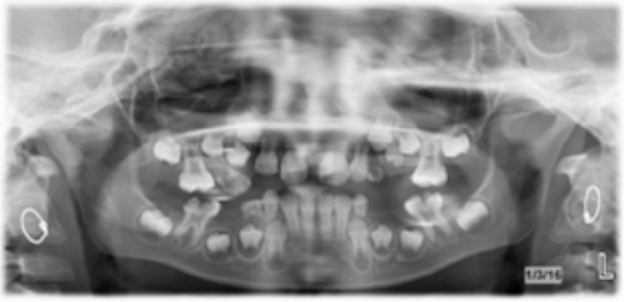

Fig. 3: Panoramic Radiograph showing Mixed Dentition Status

Discussion:

LI is an autosomal recessive disease, which is present since birth. The term has been derived from the Latin word "Icthus," meaning fish [3]. According to literature, seven causative genes for autosomal ARCI have been identified [4]. The child at birth, are usually born with an anomaly known as the collodion membrane, which ruptures off to reveal scaly skin with polygonal patches. Our case had a similar typical fish scales all over her body. Ichthyosis is a genodermatotic condition, which is characterized by exaggerated hyper-keratosis and desquamation of the skin. It is a multifaceted disease involving different structures of ectodermal origin viz. hair, skin, nails, eyes, and ears. Clinical presentation remains heterogeneous and may vary from the milder, dry skin type to a full-blown disorder with severe ocular manifestations [5]. Our case reported with bilateral ectropion in the lower eyelid, dry scalp along with the scarcity of hair. Bilateral ectropion have been reported in only thirty-three percentage of cases of LI. Various disciplines have focused on the abnormalities of LI associated with a majority of ectodermal structures, but few studies have emphasized on its association with teeth.

There is scarce information regarding the oral manifestations of these disorders. In some patients, teeth are usually developed, but in others, they are defective and likely to develop caries. The dental findings reported in persons with ichthyosis have included gingivitis, periodontitis, enamel hypoplasia, high caries incidence, delayed primary and secondary eruption, bruxism, bifid teeth, an irregular morphology of teeth, and hyperkeratotic plaques on the tongue [6]. Our case presented with early childhood caries type 2, premature exfoliation of primary teeth, and a significant amount of plaque-induced gingivitis. Our treatment included restorations for carious teeth, pulpotomies with the fabrication of stainless steel crowns for pulpal involved teeth, extraction of anterior root stumps, and fabrication of space maintainers for edentulous areas relating to unerupted premolars. The dermatological treatment of ichthyosis consists of substantial administration of emollients and keratolytic agents such as 0.1% cream of retinoic acid, lactic acid, and urea, a mixture of lactic acid and propylene glycol, and phototherapy [7].

Conclusion:

The present case signifies the need for the dental clinician to be well verse with the general manifestations of LI. Since this condition is extremely rare and involves multiple ectodermal structures like scalp, eye, nail, skin and teeth and requires multidisciplinary approach to manage this disorder. Additionally, we should look to build up a social rapport with the patient to promote oral health care.

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