

Case report

Lamellar ichthyosis – a rare case report

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Abstract:

Lamellar Ichthyosis (LI) is an autosomal recessive disorder with an incidence of less than 1 in 3 lakhs. Diagnosis of lamellar ichthyosis is based on the history of collodion membrane at birth and the characteristic appearance of scales especially on shins.

Key words: Lamellar Ichthyosis, Collodion membrane

Introduction :

Ichthyoses form a clinically and etiologically heterogeneous group of cornification disorders. Clinical hallmarks of the diseases are visible scaling and a thickening of the cornified layer and very often an accompanying inflammation of the skin presenting as erythroderma.¹ Lamellar Ichthyosis (LI) is the rarest form with an incidence of less than 1 in 3 lakhs. It has autosomal recessive inheritance and there is a defect on chromosome 14q11 causing transglutaminase-1 (TG) defect. It is an autosomal recessive ichthyosis with hypotrichosis, mutation resulting in a Glycine to Arginine substitution in residue 827 of the matriptase protein resulting in thickened scaling, and grayish skin and curly, sparse, fragile, brittle, dry, lustreless, and slow growing hair.² Diagnosis of lamellar ichthyosis is based on the history of collodion membrane at birth and the characteristic appearance of scales especially on shins.

Case report:

A 10 year old girl presented with complaints of generalized scaling since birth. At birth her mother reported that her body was completely enclosed in a shiny taut membrane. The membrane shed over a period of one month followed by gradual development of generalized scaling of the skin and there was no history of mucosal and nail involvement and no history of similar complaints in the parents. History of parental consanguinity was present. Examination revealed generalized dryness and scaling all over the body. The scales were dark brown in colour, polygonal in shape, large in size, thick and adherent to skin prominently over shins. The hyperkeratosis was present on palms and soles. Mucosae, teeth and hair were normal. Ocular examination, routine investigations of the blood and urine were normal. On the basis of these findings, a diagnosis of lamellar ichthyosis was made and the child was treated with emollients and keratolytics.

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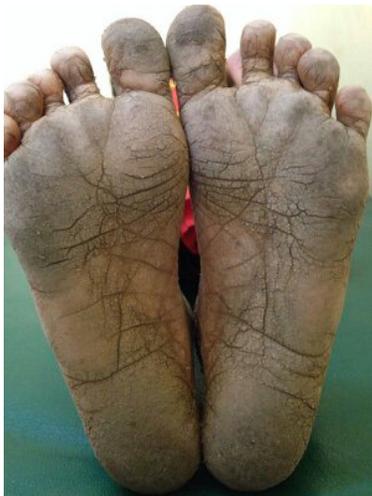
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Generalized scaling of back



Generalized scaling of trunk



Hyperkeratosis of soles



Large dark brown scales on shins

Discussion:

The term ichthyosis is derived from the Greek word 'ichthys' meaning "fish" and refers to the similarity in appearance of the skin to fish scales.³ Lamellar ichthyosis is an autosomal recessive disorder that is apparent at birth and persists throughout life.⁴ There is minimal erythema while the scales are large, thick, dark-brown in colour and firmly adherent. Sweating is also impaired.^{4,5} Lamellar ichthyosis and Congenital Ichthyosiform Erythroderma (CIE) are two different disorders of cornification based on disparities in clinical

presentation, lipid abnormalities, enzymatic studies and cellular kinetics. The skin in LI is covered with large, thick, dark scales that have a plate-like appearance.⁶ In contrast, CIE is milder than LI; scales on the body are finer and whiter in colour, but erythroderma is more prominent.⁷ The management is aimed at decreasing symptoms and include emollients (petrolatum, coconut oil, alpha hydroxyl acetic acid), keratolytics containing salicylates with propylene glycol and local and systemic retinoids.⁸

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