Section: Dermatology, Venereology and Leprosy

Case Report

A RARE CASE OF LAMELLAR ICHTHYOSIS: A CASE REPORT

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ABSTRACT

Lamellar ichthyosis (LI) is classified as one of the autosomal recessive congenital ichthyoses and is considered to be more severe than congenital ichthyosiform erythroderma (CIE). It is estimated that LI occurs in approximately 1 in 300,000 live births, and is primarily caused by a deficiency of transglutaminase-1 (TGM-1), inherited in an autosomal recessive fashion. Additionally, several mutations have been linked to LI, including missense mutations in the ATP-binding cassette (ABC) transporter gene (ABCA12). Usually, LI presents at birth with ectropion and a collodion-like membrane that begins to desquamate within the initial three weeks of life. This collodion-like membrane eventually gives way to large, thick scales that cover the entire body. Patients with TGM-1 mutations often experience scarring alopecia, and short stature may manifest in more severe cases of the condition. Individuals can expect a typical life expectancy; however, there is a heightened risk of mortality during the neonatal stage due to complications like secondary sepsis and hypernatremic dehydration. A case of 14 year old male presented with thick scaly lesions all over the body since birth. On Clinial Examination Multiple well defined dark browned, hyperkeratotic and plate – like scales present all over the body.

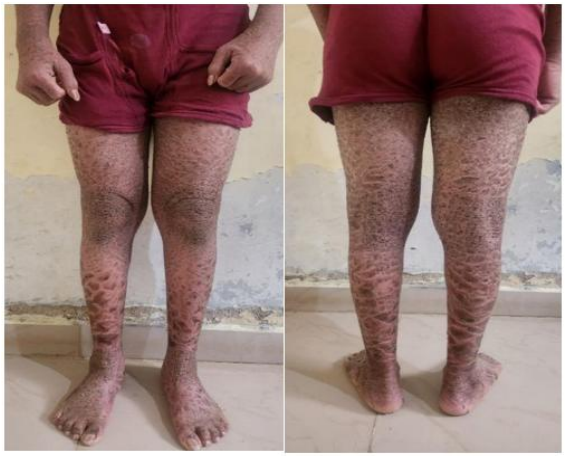
Keywords: Lamellar ichthyosis, scales, genetic.

INTRODUCTION

Lamellar ichthyosis is a genetic disorder inherited in an autosomal recessive pattern, and most affected individuals are born with a colloid membrane.[1] Mutations in the genes TGM1, ALOX12B, ALOXE3, and ABCA12 interfere with the normal mechanism that allows the skin to shed old cells and maintain moisture, resulting in the thick, plate-like scales characteristic of the condition. The global incidence of this disorder is 1/300000 live births.[2]

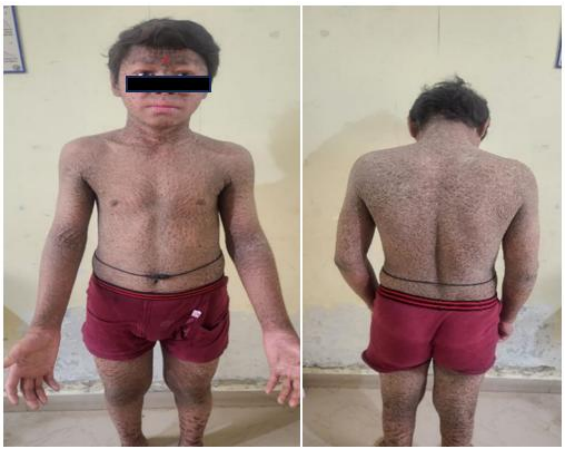
CASE REPORT

A 14 year old male presented with thick scaly lesions all over the body since birth. No h/o similar complaints in family. h/o 1st degree consanguinity among parents.



On examination: Multiple well defined dark browned, hyperkeratotic and plate – like scales

present all over the body. Madarosis present over the both sides. Nail pitting present.



DISCUSSION

The word ichthyosis comes from the Greek term 'ichthys,' which translates to "fish" and relates to the resemblance of the skin to fish scales. Lamellar ichthyosis is the least common type, occurring in fewer than one in 300,000 cases.[3] This condition follows an autosomal recessive pattern of inheritance, with a mutation located on chromosome 14q11 that leads to a deficiency in transglutaminase-1 (TG). Autosomal recessive ichthyosis with hypotrichosis (ARIH) is caused by a mutation that leads to a Glycine to Arginine change at residue 827 of the matriptase protein, resulting in thickened, scaly, grayish skin, along with curly, sparse, fragile, brittle, dry, and lacking luster hair that grows slowly. Individuals with LI frequently experience severe hypohydrosis and face an increased risk of hyperpyrexia in hot environments.[4]

Upon birth, neonates diagnosed with LI are typically covered by a collodion membrane, which results in a redness on their skin that is more pronounced than that of healthy neonates. Nevertheless, as indicated in various case reports, there have been instances where a collodion membrane was absent. Over time, substantial scales begin to emerge. These scales are generally quite large, with most of them concentrated on the lower limbs. Large scales that resemble plates can be found, often separated by shallow fissures that create the appearance of dry troughs. Aknesa's structure is distinguished by its thick scales. Patients with LI often exhibit symptoms of evaporative dry eye disease as well.

LI is often diagnosed in a clinical setting. Gene testing, however, could be used to confirm the diagnosis in situations that are unclear. Topical and oral pharmaceutical therapies, such as hydration aids, keratolytics, and keratinocyte differentiation regulators, are commonly used to treat ichthyosis.[5]

CONCLUSION

This is a rare case of Lamellar Ichthyosis. Knowledge of this condition helps in its early identification and diagnosis which in turn helps in its early management and prevention of complications.

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