Lamellar Ichthyosis in a Nigerian Child: A Case Report

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ABSTRACT

Lamellar ichthyosis is a form of congenital ichthyosis that is transmitted in an autosomal recessive fashion. Globally, it is a rare condition with estimated prevalence of 1 in 300000 in the United States. Its exact prevalence in Nigeria is not known though there has been an earlier reported case of two sisters presenting with lamellar ichthyosis. We therefore present this case for its rarity, late presentation and stigma associated with it in our environment.

Keywords: Lamellar ichthyosis, ectropion, stigma

INTRODUCTION

Lamellar ichthyosis (LI) is an autosomal recessive disorder that is apparent at birth and is present throughout life. The newborn is born encased in a collodion membrane that sheds within 10-14 days. The shedding of the membrane reveals generalized scaling with variable redness of the skin. The scaling may be fine or platelike, resembling fish skin.

Although the disorder is not life threatening, it is quite disfiguring and causes considerable psychological stress including social stigma in affected patients.

Patients with lamellar ichthyosis have accelerated epidermal turnover with proliferative hyperkeratosis. This involves a mutation in the gene for transglutaminase 1 (TGM1). The transglutaminase 1 enzyme is involved in the formation of the cornified cell envelope that is an essential scaffold upon which normal intercellular lipid layer formation in the stratum corneum occurs. How much a defective cornified cell envelope alone contributes to the barrier abnormality in ichthyoses however remains unclear

In the United States, prevalence is 1 in 300 000. The exact prevalence in our environment is however unknown though a case of two sisters with lamellar ichthyosis in Nigeria has been previously documented.

The case is being reported for its rarity, late presentation and associated social stigma.

CASE REPORT

A 2 year-old female child presented (with her mother) at the dermatology clinic of Obafemi Awolowo University teaching hospital, Ile-Ife, Nigeria with history of generalized skin lesions since birth.

Mother delivered her at a mission hospital and gave history of the child being in an enclosed membrane that shed over two weeks. Subsequently, baby developed generalised scaliness of the skin. There was associated history of itchiness but there was no history of weeping, oozing or crusting nor redness of the body. The mother gave history of incomplete closure of eyelids while sleeping. She has had no previous hospital admissions or surgeries and no history of blood transfusion. She was the last of four children. Mother was a 36 year old trader and father a 40 year old driver. She completed her immunization and attained developmental milestones at the appropriate time. However child was not enrolled in school due to her clinical condition and stigma she experiences in the community. No known family history of similar skin lesions and no history of consanguinity.

Physical examination showed generalised large,

brownish scales with some being firmly adherent. There were patchy areas of hair loss on the scalp especially at the periphery with adherent crusts. Also, ectropion was observed with plantar fissures and flexion contractures of the feet. No background erythema was observed. Systemic examination yielded no additional information.

A diagnosis of Lamellar icthyosis was made and this was confirmed by histological findings.

Hematological profile done were within reference range and blood film appearance had no abnormal cells detected. Electrolyte, urea and creatinine were also within reference ranges. The skin biopsy done showed marked hyperkeratosis, focal parakeratosis, acanthosis and a thickened granular cell layer.

She was started on multivitamins, Vaseline, urea lotion and 2% salicylic acid and she was to procure acitretin.

At one-month follow up, the child's clinical state had improved as evidenced by reduced scaliness and pruritus. Patient was however lost to follow-up subsequently.

DISCUSSION

Lamellar ichthyosis (LI) is an autosomal recessive disorder characterized by onset of ichthyosis at birth, involving the entire body surface.[1]Non-bullous congenital icthyosiform erythroderma (NCIE) is the other non-syndromic form of autosomal recessive icthyosis. [1]Clinically, LI can be distinguished from NCIE by the characteristics of the scales, which are large, adherent, dark and pigmented, and by the absence of erythema.[2,3]

It is a rare condition with a prevalence of 1: 300 000 though its exact prevalence in our enviroment is not known.[3]It occurs in both sexes equally and affects all population.[3] An earlier reported case in Nigeria documented two sisters with the condition thus showing its familial tendency[4]

The newborn with lamellar icthyosis, is usually born encased in a membrane like sheath, the phenotype known as collodion baby which later on sheds within 10-14 days as was the case being reported, with persistence of scales and variable redness of skin.[1]

The scales may be fine and white to thick, dark and plate like as in our case. The scales are arranged in a mosaic pattern resembling fish skin.[2]

During childhood and into adulthood, the degree of erythema may vary, but the severe presentation of classic LI usually has minimal to no erythroderma.[1]

Involvement of the palms and soles in LI is variable and ranges from minimal hyperlinearity to severe keratoderma.[3] The index case had mild hyperlinearity.

Nail abnormalities, which include subungual hyperkeratosis and nail dystrophy, scarring alopecia, ectropion, eclabium, bilateral conjunctivitis, small and deformed ear and inflexible digits due to taut skin are other manifestations of lamellar ichthyosis.[4] They could also present with fissures as was the case in our patient.

Lamellar ichthyosis is a disorder of cornification that leads to an abnormal quality and quantity of scale, the barrier function of the stratum corneum is compromised, and there may be alterations in the kinetics of epidermal cell proliferation. [3,5]

The pathogenesis is hypothesized to be due to mutation of gene located on chromosome 14q11 encoding Transglutaminase 1 enzyme, an enzyme that catalyzes the cross-linking of proteins (of lucrine and involucrine) and attachment of ceramides during the formation of corneocytes.[3,6]

Lucrine and involucrine serve as the scaffolding in the formation of cell envelope which acts as a physical barrier for epidermis.To date, 6 genes for lamellar ichthyosis have been localized and 5 of them identified, as follows :TGM1 (14q11), ABCA12 (2q34), 19p12-q12, 19p13, ALOXE3-ALOX12B (17p13) and ichthyin (5q33) and novel mutations are also been discovered.[7]

The histopathological findings include compact orthohyperkeratosis with variable mild focal parakeratosis, and stratum corneum thickness.There may be mild papillomatosis, extension and blunting of rete ridges, and dilatation of dermal capillaries reminiscent of psoriasis.[8]These were the findings in the index patient.

One of the difficulties individuals in sub-Saharan Africa as a whole encounter is social stigma as was experienced in this patient.[4,9,10] Ibekwe et al noted that social stigmatization of patients with lamellar

Figure 1 showing generalized, large brownish scales (anterior view)

Figure 2 showing generalized, large brownish scales (posterior view)

Figure 3 showing feet contractures

Figure 4 showing marked scaliness and crusts on the scalp and ectropion

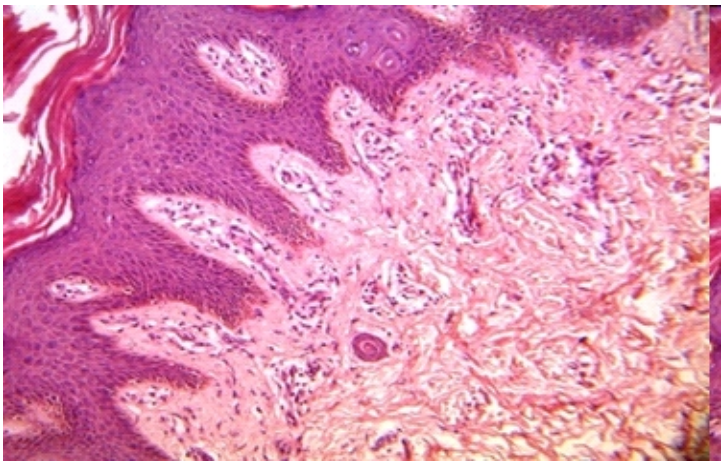

Figure 5: histology showing marked hyperkeratosis, acanthosis and thickened granular cell layer

ichthyosis hindered their access to good education, adequate healthcare and employment opportunities.[4]

As regards management, this is usually supportive and focuses on hydration, lubrication, and keratolysis.[1] Emollients include petrolatum, coconut oil and alpha hydroxy acetic acid.Keratolytic agents are used to enhance corneocyte disadhesion and thereby remove scale and thin hyperkeratotic stratum corneum.[2] They include urea, salicylic acid and propylene glycol.Topical and oral retinoids decrease the cohesiveness of follicular epithelial cells and thickening of scales and stimulate mitotic activity resulting in an increase in the turnover of follicular cells.[3]

The corrective gene therapy may be a modality of treatment in near future for lamellar ichthyosis.[3]Adequate nutrition is also important because of large turnover scales that can lead to malnutrition.[2]The education of patients for potential heat stress disorder and psychosocial and emotional support is required for the patient and family.[3]

CONCLUSION

Lamellar ichthyosis though rare do occur among Nigerian children. There is the need to look out for it and address social stigma that might be associated with the disorder in our environment.

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