Lamellar Ichthyosis with Bilateral Ectropion in a Nigerian Infant: A Case Report

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ABSTRACT

Lamellar ichthyosis is a rare genetic dermatosis, mostly inherited in an autosomal recessive fashion. Though not generally life-threatening, its distinctive features are not only distressing to the newborn but pose a challenge to the Nigerian mother due to the poor knowledge and understanding of genodermatoses. Several case reports have documented its occurrence amongst Nigerian children but its exact prevalence remains unknown.

We describe an eight-month-old male infant who presented at the Paediatric emergency unit of the University of Uyo Teaching Hospital (UUTH), where he was first diagnosed and managed for lamellar ichthyosis with bilateral ectropion complicated by sepsis and malnutrition. This case underscores the peculiar challenges with the medical management of lamellar ichthyosis, including late presentation, in limited-resource settings.

Keywords: Lamellar ichthyosis, Uyo, social stigma, collodion baby

Ichtyose Lamellaire avec Bctropion Bilatéral chez un Nourrisson Nigérian : Rapport de Cas

ABSTRAIT

L'ichtyose lamellaire est une dermatose génétique rare, principalement héritée d'un mode autosomique récessif. Bien qu'elles ne mettent généralement pas la vie en danger, ses caractéristiques distinctives sont non seulement pénibles pour le nouveau-né, mais posent un défi à la mère nigériane en raison de la mauvaise connaissance et compréhension des génodermatoses. Plusieurs rapports de cas ont documenté son apparition chez les enfants nigérians, mais sa prévalence exacte reste inconnue.

Nous décrivons un nourrisson de sexe masculin de huit mois qui s'est présenté au service des urgences pour enfants de l'hôpital universitaire de l'Université d'Uyo (UUTH), où il a été diagnostiqué et pris en charge pour la première fois pour une ichtyose lamellaire avec ectropion bilatéral compliqué de septicémie et de malnutrition. Ce cas souligne les défis particuliers de la prise en charge médicale de l'ichtyose lamellaire, y compris la présentation tardive, dans les milieux à ressources limitées.

Mots clés: Ichtyose lamellaire, Uyo, Stigmatisation sociale, Bébé au collodion

Introduction

Lamellar ichthyosis belongs to a group of non-syndromic congenital ichthyosis, referred to as autosomal recessive congenital ichthyoses (ARCI).[1] The hallmark of the disease arises from abnormal differentiation in the epidermal metabolism which is apparent from birth and throughout life. Mode of inheritance is predominantly in an autosomal recessive fashion except the rarely reported autosomal dominant lamellar ichthyosis (ADLI),[2,3] that is phenotypically indistinguishable from its autosomal recessive counterpart. The rarity of this condition, with a global prevalence of 1 in 200,000-3,000,000 live births,[4] contributes to the lack of population studies in most parts of the developing world with only pockets of case reports existing in Nigerian literature regarding this condition.[5,6,7]

As a common similarity amongst ARCI, the earliest sign of this condition is the birth of a newborn completely sheathed in a collodion, or a translucent parchment-like membrane.[8] This has also been described as having a "baked apple" appearance.[9] Shedding off this membrane occurs within 10-14 days

and the child develops dark, plate-like or lamellar scales on the skin.[10] Although the manifestations of this condition are often limited to the skin and not life-threatening in most cases,[11] it causes severe disfigurement and brings psychological stress to affected patients and their families.[12]

Case Description

Following the initial presentation at the Paediatric emergency unit of the University of Uyo Teaching Hospital, Uyo, Nigeria, the dermatology unit was called to review an 8-month-old male infant of the Ibibio tribe (accompanied by his mother) with a history of dark dry scaly skin and purulent discharge from both eyes, which was noticed from birth.

The birth mode of the infant was via spontaneous vertex delivery at term, done at home by a traditional birth attendant (TBA). After birth, the mother noticed the baby was covered with a 'thin shell' which later peeled off within weeks. The infant was said to have cried immediately at birth, but the birth weight and APGAR scores could not be ascertained in a traditional birth setting. The index pregnancy was not booked for antenatal care. However, the antenatal period was essentially uneventful and the pregnancy was carried to term. Owing to the appearance of a 'strange' skin condition noticed since birth, the mother had applied several herbal mixtures all over the baby's skin with no resolution of the rash.

The infant's mother was a 35-year-old widow and trader who had primary level of education and sold palm kernel oil. There was no indication of a consanguineous marriage. The infant was the only child of the mother who was unaware of a similar presentation in children born to any member of her family.

Examination revealed an ill-looking infant with fluffy hair and a weight of 4 kilograms (47% of expected), length was 6.1 centimetres (weight for length z-score of < -4 standard deviation, the expected value of -2 standard deviation to +2 standard deviation). The occipito-frontal circumference was 40.5 centimetres (expected range of 47 – 48 cm) and the mid-arm circumference was 9.5 cm (expected normal value of >14 cm). There were no signs of dehydration but the infant was febrile (38^0C). He had generalized thick

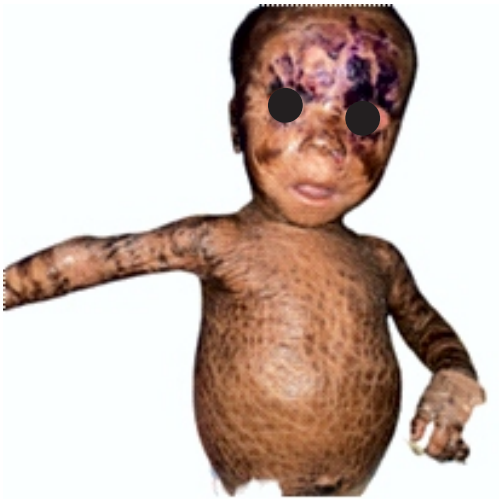

Figure 1: showing generalized brown scales on extremities, trunk and face, with bilateral ectropion (concealed with of the eye patches)

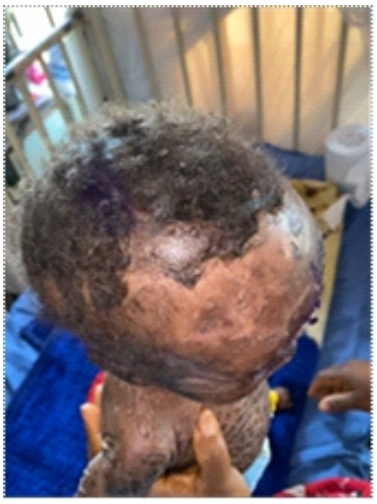

Figure 2: showing cicatricial/scarring alopecia

hyperpigmented 'fish-like' scales on the skin with marked xerosis and scarring alopecia involving the frontal aspect of the scalp extending to the vertex and hyperkeratosis of the palms and soles, (Figures 1 and 2). There was the presence of bilateral ectropion involving the upper and lower eyelids with non-copious yellowish-brown discharge. He was noticed to have a cough of a week's duration and crepitations and transmitted sounds in both middle and lower lung zones on chest auscultation. Other systems were essentially normal. A diagnosis of lamellar ichthyosis with bilateral ectropion complicated by sepsis and malnutrition was made.

The treatment was mainly supportive and the mother was educated on the condition, proper clothing of the child and adequate nutrition with complementary feeds. Generous application of petrolatum jelly to the

skin with concomitant use of a topical retinoid (0.05% tretinoin) was prescribed. Application of mupirocin ointment over fissures and non-intact skin was advised in addition to systemic antibiotics (ceftriaxone, azithromycin), oral zinc, folic acid and multivitamins suspension which were prescribed by the reviewing Paediatric unit. An ophthalmology consult was sent on account of bilateral ectropion and the use of lubricant eye drops was advised. However, there was no record of the mother procuring the tretinoin ointment before the child's discharge and the infant was booked for a follow-up visit at the paediatric clinic in two weeks. Unfortunately, both mother and infant were lost to follow-up despite efforts to trace them to assess the level of home care and support.

Discussion

Lamellar ichthyosis is a rare, chronic and refractory genodermatosis characterised by hyperkeratinisation and cornification of the skin which has been subdivided into types 1-5, depending on the gene mutation involved. The most common is type 1 LI involving a mutation in the gene for transglutaminase 1 (TGM1);[10] type 2 LI occurs with a mutation in the ABCA12 gene.[13] Since these types are clinically indistinguishable, the exact type for the index case cannot be identified as genetic testing is not available in general Nigerian dermatology practice. Thus, as with most cases diagnosed in Nigeria,[5] including the first diagnosed case of lamellar ichthyosis in the South-South region of Nigeria documented by Benson and Madubuko,[7] the diagnosis of LI relies solely on physical or clinical characteristics.

The condition has equal sex predilection and occurs with a similar phenotype in all races. Familial clustering has been identified especially in the presence of consanguinity.[14] Nevertheless, LI in the absence of consanguineous marriage has been reported,[5,15] which may in part be attributed to ignorance amongst families about this condition. Social factors like stigmatization and abandonment may contribute to concealing incidences of LI among family members; hence the mother of the index infant may be unaware of such occurrence in their family tree.

The accelerated epidermal turnover with proliferative hyperkeratosis of LI is demonstrated clinically at birth as the baby being wrapped in a collodion-like membrane that is subsequently shed during the first weeks of life with scaling of the skin involving the whole body.[16] This classical presentation was evident in this case. The defects in the skin barrier[17] give rise to dysregulated body temperature and a low threshold to developing cutaneous and systemic infections[18,19] explaining the fever and pneumonia present in the index case. The debilitating nature of the disease can be attributed to coexisting ocular abnormalities such as bilateral ectropion of the cicatricial type seen in one-third of cases,[20,21] which is one of the rare complications of the dermatosis. It can affect both eyelids but is more common with the lower eyelids, and it is often bilateral. The involvement of both eyelids reported here was unexpected as it is rarely documented in Nigerian cases.[22,23] This form of ocular deformity adds to the obvious physical anomalies of affected children which is the bane of stigmatization in our locality. As a culturally rooted community, some beliefs have labelled them as snake children,[12] that is to say snakes grown into humans limiting their social integration into schools and society as a whole.

Diagnosis of lamellar ichthyosis can be made on clinical grounds, as was done in the index case. The presenting clinical features such as onset at birth, presence of collodion membrane, ectropion and thick dark cutaneous scales are typical of lamellar ichthyosis as described by the 2009 Ichthyosis Consensus Conference.[24] Even with the myriad of identified gene mutations in LI, no gross phenotypical variation exists at the clinical level[25]; hence, the need for pre- and post-natal genetic diagnosis. However, this mode of investigation is unavailable in our facility and could not be carried out for the index case. The value of prenatal testing does not only assist in recognizing families at risk but offers the option of termination of the pregnancy if desired by the mother, as such concerns were expressed by an affected mother in Sub-Saharan Africa as documented by Kouadio et al.[12] The proffers the opportunity to minimize the economic and social burden families incurred with management after birth. However, the law remains frigid for such concerns in Nigeria as abortion is still illegal except in

instances where there is a threat to the life of the mother.[26] Considering the financial constraints of the mother and our facility's lack of capacity to carry out postnatal genetic testing, additional specialized tests that aid diagnosis of LI such as skin biopsy and detection of TGM1 expression/activity (TGM1 being the predominant mutation in LI)[10,14,27] were not requested and they may not significantly alter our outlined treatment.

The management is multidisciplinary and centred on counselling and supportive care. Infections are treated with antibiotics and the mother of an affected child is advised to apply emollients such as petrolatum jelly to enhance desquamation and re-epithelization of the skin. Other supportive care practices include regular baths to optimize hydration and stringent thermoregulation with proper clothing. The dramatic response to the administration of retinoids has been well documented[22] and is attributed to the keratolytic effect which helps in the elimination of scales and prevention of hyperkeratosis of the skin.[16] As a chronic condition, routine follow-up to assess home care and periodic counselling of parents and families is extremely vital to discouraging stigmatization and social isolation. Unfortunately, in this report, we could not reproduce the benefits of retinoids here as the mother did not procure tretinoin ointment as prescribed before hospital discharge. Coupled with the child's loss to outpatient follow-up, we were unable to have a long-term assessment to address the challenges to proper management, which in this case may be due to socioeconomic circumstances.

Conclusion

This report aims to highlight the late identification of the condition and presentation with concomitant ocular and systemic complications that could have been minimized if the mother received the necessary counselling and treatment right from the day of birth which is lacking in home deliveries with untrained traditional birth attendants as is prevalent in many Nigerian communities. These complications that may be encountered with this rare, life-long congenital skin defect can be an overwhelming burden for a widowed single mother. Given that this is the first documented report of lamellar ichthyosis in Uyo, it is imperative to heighten awareness within the community to encourage early and prompt diagnosis of this genodermatosis for appropriate and timely dermatological intervention.

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Conflict of interest: None

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