Congenital Icthyosiform – A case report

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Summary

Congenital icthyosis is a rare group of disorders of keratinisation. A case of this condition is reported in order to highlight the clinical features and essential components of management: resuscitation, skincare, nutrition and counselling. This case is reported in order to appraise clinicians of its presence because there was delay in the diagnosis and management due to non-recognition.

Keywords: ***Ichthyosis, Congenital, Kaduna, Nigeria.***

Résumé

Les démengeaisons congénitales - ichthyosis sont des rares groupes de mutation de la peau-keratinisation. Un cas de cette maladie rare nous ai reporté afin d'éclaisir les differents apparences et leurs traitment: qui inclus la reanimation, les soins de la peau, la nutrition et un exposé aux parents. Ce cas entraine un delai de diagnostics et de traitments du a la non identification de la maladie. Donc, nous tenons a vous rappeler son existence.

Introduction

Congenital ichthyosis is a group of disorders of keratinisation which are generally determined: autosomal recessive, X-linked or could occur spontaneously as fresh dominant mutations.[1,2] Presentation ranges from mild cases with collodion membrane covering to the severe lethal form - Harlequin infant[3]. Reports of its local literature are scanty – probably a true reflection of the rarity of this condition or may be under-reporting from non-recognition. This communication highlights the clinical features of congenital ichthyosis in order to increase the awareness regarding the condition and therefore to enhance early management especially in the severe forms that have a high mortality.

Case report

A.H. a 3 day old female was admitted to the Special Care Baby Unit (S.C.B.U) of Ahmadu Bello University Teaching Hospital (ABUTH), Kaduna on the 4th January 2000 with a history of abnormal skin appearance and eye discharge noticed at birth. She was born after a full pregnancy to a 29 year old Para 5+0 (all alive) Hausa mother; she had generalised pruritic rash at the 4th month of pregnancy; there was no associated fever or jaundice. The rash spontaneously disappeared after 4 days. There was no history of vaginal discharge, per vagina bleeding or premature rupture of membranes. Delivery was attended to at a peripheral hospital and the child cried immediately after birth. The medical staff at the hospital noticed an abnormal skin appearance and A. H. and her mother were immediately discharged inspite of mother's plea for medical information and care with regards to her baby. On discharge, A. H. was taken home and then to 3 other private hospitals from where she was finally referred to ABUTH, on the 3rd day of life. The child is the last of 5 siblings in a monogamous home setting. Her father is a 37 year old Hausa-Fulani civil servant and there is no history of consanguinity. There is no known family history of such skin disorder. On presentation at ABUTH, physical examination revealed a full term baby covered with a thick, taut membrane (Collodion).

Face

There was wrinkling with ectropion, chemosis and bilateral conjunctivitis. The apposition of the upper and lower eyelids was not good. The ears and nose were flattened with partial membrane covering the nostrils. The lips were everted and fixed in an O-shape and there were fissures at the angles of the mouth.

Fig. 1 ***Facial features in Congenital icthyosiform***

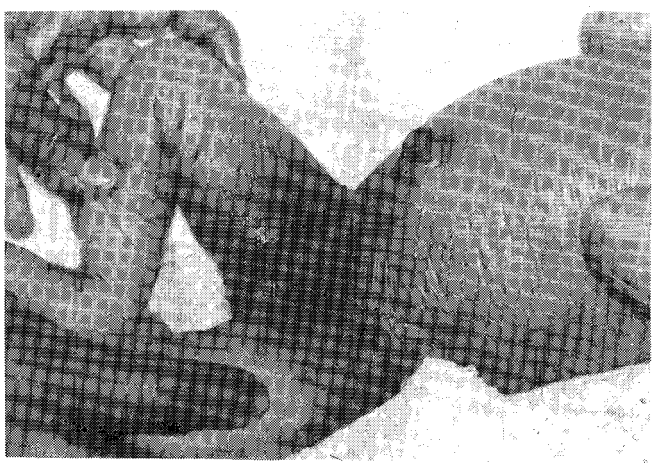

Fig. 2 ***Wrinkling and fissures in the trunk and groin flexures***

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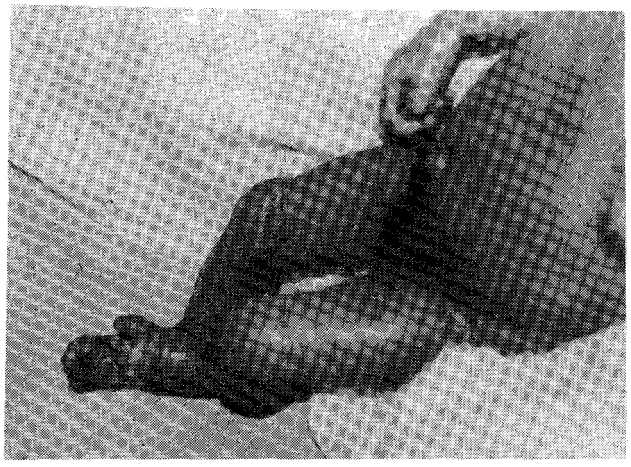

Fig. 3 ***Constriction in the digits***

The scalp was dry and moistureless with hair piercing through the membrane covering on it.

Trunk and Neck

The skin over these was rough and dry with erythematous patches over the axillary folds. There were fissures in the membrane over the chestwall and also at the groin flexures.

Limbs

There was constriction of the digits by the taut membrane with ischaemia, edema and restriction of movement of the fingers and toes. Crusted flexural membrane cracks were seen, while the palms and soles were hyperkeratotic. The nails were atrophic.

She was mildly jaundiced and dehydrated with an admission weight of 2.7kg. Gestational age assessment by the mother's dates was 39 weeks. Respiratory, cardiovascular and abdominal findings were normal. Based on these clinical findings, a diagnosis of congenital non-bullous ichthyosiform Erythroderma was made. Histopathologic diagnosis was not available. A. H. was managed for one week on admission with emphasis placed on resuscitation, skincare, nutrition and counselling. Skincare in the management of this baby involved emphasis on prolonged baths with bath oil to remove excessive scaling and mal-odours which resulted from infected, macerated cracks on the skin. Generous and frequent application of keratolytic agents – 2% Salicylate in Vaseline to prevent dryness and to lessen the scales.

Infection at the flexural surfaces due to fissuring were treated by systemic antibiotics and fresh ones prevented by skincare. The ophthalmologist treated for exposure keratitis and referred to the plastic surgeons for correction of the ectropion. The parents were counselled as to the nature of the disorder, and to help reduce the psychological stress associated with the disfigurement and enable them to understand how to manage the child well.

At the time of this report, A. H. has been seen twice on follow-up and is now 3 weeks old, gaining weight and the fissures are healing. Her skin is still very dry and coarse but her mother is well adjusted to the management of her child.

Discussion

The diagnosis of congenital ichthyosis is usually made from clinical features as well as histopathological changes from light and electron microscopy; the inheritance patterns will also differentiate the type. There is no doubt from the characteristic clinical features in this child that she presented with non-bullous ichthyosiform erythroderma which could have made the diagnosis easy. However, non-recognition of the condition led to delay in therapy. Congenital ichthyosis may present in the severest forms as the harlequin infant which is covered by markedly thickened ridged and cracked membrane that disfigures the facial features and constricts. This results in respiratory difficulty, poor suck and severe cutaneous infections which are all associated with a high mortality. The Collodion baby is also the presentation at birth of other forms of ichthyosis such as bullous congenital icthyosiform erythema, X-linked and ichthyosis vulgaris, which are differentiated by other clinical features. Other methods of diagnosis by histopathologic changes from light and electron microscopy also help to differentiate subtypes of varied forms.[4] Genetic factors suggested in X-linked congenital ichthyosis include partial or complete deletion of steroid sulfatase (STS) gene[1], mutation at an X chromosome site genetically linked to the STS gene.[5] Controversy abound concerning mutation of keratocyte transglutamate gene as causative in lamellar ichthyosis[6]. Counselling in ichthyosis is necessary as the clinical disfigurement leads to considerable psychological stress. Parents may also be offered the option of prenatal diagnosis where available. Low levels of maternal serum unconjugated oestriol in the 2nd trimester are indicative of congenital ichthyois. Follow-up of the children is done in view of associated disorders seen as the child grows older such as Sjogren syndrome (degenerative defect of retinal pigment, motor speech development delays, epilepsy and mental derangement), Netherton syndrome (failure to thrive, marked hypernatraemia, urticaria, angioedema and asthma), SLE and cutaneous carcinomas. Though histopathological means are not available, congenital ichthyosis should be diagnosed at least clinically to enhance saving the lives of these children.

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