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Congenital Ichthyosis in a Nigerian preterm neonate: A case report and review of the literature

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Abstract: Congenital ichthyoses are relatively uncommon skin disorders with worldwide occurrence. The ichthyoses are heterogenous disorders of keratinisation characterised by scaling of the skin of varying severity. This report describes a case of congenital ichthyosis in a preterm, male Hausa infant which happened to be the first case managed at the OOUTH, Sagamu. The infant was managed using stringent thermoregulation, optimal hydration, use of topical emollient and antibiotics.

Although, the skin disorder resolved within fifteen days of treatment, the infant thereafter developed inguinoscrotal hernia and hydrocephalus necessitating surgical consult. The case is reported to highlight the good outcome of the cutaneous disorder following conservative management in a low-resource setting.

Key words: Collodion baby, Congenital ichthyosis, hydrocephalus, newborn, Sagamu

Introduction

Congenital Ichthyoses are a rare heterogenous group of skin disorders characterized by excessive skin scaling and dryness.¹ They represent a disorder of keratinisation or cornification arising from abnormal differentiation in the epidermal metabolism resulting in thick skin with poor elasticity, formation of blisters or bullae, trans-epidermal loss of water and weakened physical skin barrier to infections.² The mode of inheritance varies: there are autosomal recessive, autosomal dominant and X-linked forms.

Most autosomal recessive cases of Congenital Ichthyoses (lamellar ichthyosis, non-bullous ichthyosiform erythroderma) present in shiny (collodion) membranes which are taut and limit movements. Others present with thick excessive skin scaling or even blisters present at birth. Disfigurements are present, including but not limited to ectropion, corneal /retinal abnormalities, persistently open O-shaped mouth, eclabium, small flattened ears and nose, fixed flexion of extremities / autoamputation.²⁻⁴

The rarity of the condition is attested to by the fact that there are no population studies on congenital ichthyosis in most parts of the developing world, Nigeria inclusive. Only pockets of case reports are known in the literature in this part of the world. This report is meant to add to the literature on this rare condition in Nigeria, highlight the diagnostic challenges as well as showcase the relatively good outcome of the cutaneous disorder in a

low-resource setting.

Case Description

One hour old male neonate of the Hausa tribe, was referred from a private hospital on account of prematurity following spontaneous vertex delivery at 28 weeks + 5 days gestation.

The pregnancy was registered at the referral centre. There was no significant maternal illness during pregnancy and the mother received two doses of the anti-tetanus vaccine. The only major illness the mother had in pregnancy was prolonged drainage of the amniotic fluid from about 20 weeks of gestation necessitating hospitalization for bed rest. She also bled per vaginam for three days prior to delivery. Dexamethazone was not administered in pregnancy.

The baby cried immediately after birth but the birth weight and APGAR scores were not stated. The father was a 48-year old truck driver with Primary School Leaving Certificate married to two wives. The mother was a 27-year old Para1+0 cloth-dealer with a BSc degree and the second of the two wives. The step-mother had three children: 10yrs (F), 7yrs (M) and 4yrs (M). There was no indication of consanguineous marriage.

On examination, he had generalized taut, thick (leathery) and shiny wrinkled skin (Figure 1). He was plethoric, anicteric, acrocyanosed, cold (35.9°C) but not

dehydrated. No dysmorphism was observed. He weighed 1kg with length and head circumference of 29cm and 25cm respectively (all less than 5th percentile). The gestational age was 31 weeks as estimated by the modified Ballard Score. He was conscious and fairly active with intact primitive reflexes. Musculoskeletal examination revealed linear skin breaks over the ankle joints with mild bleeding, skin tethering over the joints and partially flexed left knee joint from skin tautness. He had a persistently open O-shaped mouth. No erythroderma, ectropion or corneal opacities were observed. The right testis was palpable in the upper part of the hemiscrotum while the left testis was not palpable in the scrotum or inguinal region. Examination of the cardiovascular, respiratory systems and the abdomen were essentially normal.

Fig 1: Preterm male neonate with Congenital Ichthyosis showing shiny wrinkled skin and open mouth.



Diagnosis of Congenital Ichthyosis-lamellar type (in a preterm, Very Low Birth Weight baby) was made. Skin biopsy for histologic and genetic studies could not be done as the facilities were not available. The Random Blood Glucose at presentation was 31mg/dl. It was corrected with 4ml/kg 10% D/W and subsequent monitoring showed values within normal limits. Initial laboratory screening revealed the following: PCV - 31%, WBC - $8.28 \times 10^9/L$, Platelet - $54 \times 10^9/L$, normal electrolytes, urea and creatinine, negative blood culture, normal G6PD status, Baby's blood group - A Rhesus+, Mother's blood group - O Rhesus+ and negative Direct Coomb's Test. Parenteral antibiotics (Cefuroxime and Gentamicin), IM Vitamin K, IV Aminophylline and 10% D/W infusion were instituted. He was nursed in an incubator and kept nil per os. Prophylactic phototherapy and generous bland Blue Seal Vaseline[®] application to the skin were also commenced.

Temperature instability was documented from the first day of admission. Jaundice was first noticed at 9hrs of life with total serum bilirubin rising to a peak of 11.1mg/dl on 5th day of life. Due to the poor clinical condition, double volume exchange transfusion could not be done but intensive phototherapy was commenced along with serial whole blood transfusion for three days to increase serum albumin concentration. This practice aimed to reduce the concentration of free bilirubin and by extension, the risk of bilirubin encephalopathy. Antibiotics were changed to Ceftriaxone and Metronidazole on account of the poorer clinical state, particularly with abdominal distension. Jaundice thereafter resolved and the post transfusion PCV rose to 45% (from 31%).

A course of oral quinine was commenced when fever persisted and blood film demonstrated asexual forms of *Plasmodium falciparum*. Fever and abdominal distension resolved and the taut skin membrane and desquamation was totally replaced with normal skin on the 15th day of life. Severe anaemia was documented on the 24th day of life and he was serially transfused with sedimented cells at the rate of 15ml/kg/session and the post-transfusion PCV was 48%.

On day 27, reducible left inguinal hernia was noticed but immediate surgery was deferred due to the baby's clinical state. Examination on the 43rd day of life revealed craniofacial disproportion with full anterior fontanelle, visible scalp veins and head circumference of 29.5cm. Transfontanelle ultrasound scan showed communicating hydrocephalus with moderately dilated lateral, third and fourth ventricles with thinning of cerebral mantle. The brain stem and cerebellum were normal. Thereafter, the baby was referred for neurosurgical intervention with the weight of 1.2kg. Surgical release of the left knee fixed flexion was not necessary as the membrane spontaneously broke with restoration of normal skin and mobility. The parents were counselled on the nature of underlying skin disorder.

Discussion

Congenital ichthyoses are a rare heterogenous primary group of inherited skin disorder characterized by excessive skin dryness and scaling. They can be classified into two broad groups- syndromic and non- syndromic forms. The latter can be further classified into different types based on the clinical features, genetic analysis and histology findings.⁵

This group of skin condition has been reported in different parts of the world with incidence of 1/6,000 in the X-Linked Ichthyosis and 1/20,000-30,000 in the other inheritable non- syndromic types.^{5,6} Many case reports of the non- syndromic type have been made in Nigeria.^{7,8,9} This case adds to the list of this relatively uncommon disorder in the country.

Although all races may be affected, this case was similar to three of the four cases reported at Enugu, southeast Nigeria by Obu *et al* who were of the Hausa/Fulani descent.⁷ All the three cases however had a history of consanguinity in contrast to this case.⁷ Orogade⁸ also reported a case of a collodion baby in Kaduna. The father was Hausa/Fulani but without history of consanguinity which was similar to the situation of our patient.⁸ Only about 25% of the cases of lamellar ichthyosis are born prematurely.¹⁰ Conversely, most are delivered at term or post-term. Similar to this case however, one of the two cases reported by Joh-Jong *et al* was born prematurely.¹¹

Similar to most cases diagnosed in Nigeria, the index case was diagnosed using only physical characteristics as the facilities for genetic analysis were not

available.^{7,8,9} Prenatal diagnosis is available via fetal ultrasound scan for physical features, amniotic fluid analysis for genetic mutations and hormonal assays and fetal skin biopsy. Post-natal diagnosis is also made through genetic mutation analysis, clinical features, histologic findings and hormonal assays. Unfortunately, both arms of the diagnostic possibilities could not be explored in this case. Indeed, histology could not be done before referral due to logistic reasons. The generalized collodion membrane, as is the case in 80% of cases was totally shed in the index infant by the 15th day of life, in tandem with the cases cited by literature.^{5,7,11} Thereafter, the skin remained normal although long term follow up would be needed to monitor for recurrence. This experience was not different from that of Obu *et al*⁷, Orogade⁸ and Okoro *et al*⁹. Ten percent of cases shed the collodion membrane and retain normal skin for life (self-healing collodion baby) while about 75% will develop lamellar ichthyosis or non-bullous ichthyosiform erythroderma later in life.⁸ Fifty percent or less of the lamellar type of ichthyosis has ectropion hence it was not unusual for the index baby not to have ectropion.^{5,11}

This cutaneous disorder was managed in the index case using lubrication with bland petrolatum as a universally accepted mode of treatment. This hydrates and lubricates the skin thereby enhancing desquamation and re-epithelialization.¹² Other aspects of care such as hydration and use of antibiotics were essentially the same as reported by other researchers.

Inguinoscrotal hernia and hydrocephalus found in the index baby are not known complications associated with congenital ichthyoses. While they may be some of the

usual morbidities associated with prematurity, it is attractive to consider them as possibly linked with the ichthyosiform disorder. This observation has previously been made by Small *et al*.¹³ Therefore, it would have been helpful to have extensive genetic analysis of the index case in an attempt to establish any genotypic or phenotypic link between congenital ichthyosis and inguinoscrotal hernia or hydrocephalus in the child. In spite of this shortcoming, this report highlights the good outcome in lamellar congenital ichthyosis in a low-resource setting.

Conclusion

This report of a case of lamellar congenital ichthyosis in a preterm infant adds to the existing literature database on genetic conditions and raises important diagnostic challenges. Health facilities should be better equipped to aid definitive diagnosis for better outcome and improvement of the quality of data.

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