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CASE REPORT

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Collodion baby, lamellar ichthyosis; A report of two cases and literature review

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Abstract: Ichthyosis are disorders of cornification characterized clinically by a pattern of scaling and histologically by hyperkeratosis. Babies with these conditions present as collodion babies that are covered in thick, taut, cellophane like membranes with associated bilateral ectropion and eclabium. The membrane sheds over a few days to weeks after birth to reveal the true nature of the underlying condition which may either be a lamellar ichthyosis, a recessive x-linked ichthyosis, congenital ichthyosiform erythroderma, harlequin ichthyosis or a normal skin variant otherwise known as the Self-healing Collodion baby. We present two cases of collodion babies that were seen in our hospital within a space of two months. The first case was considerably successfully managed in the neonatal period but social stigmatization prevented follow up while the second case was discharged against medical advice by the parents after 48 hours of admission.

Keywords: Ichthyosis, Collodion baby, Social Stigmatization

Introduction

Ichthyoses are disorders of keratinization or cornification in which abnormal differentiation and desquamation of the epidermis result in a defective cutaneous barrier[1]. The word ichthyosis comes from the Greek word ichthys, meaning fish, referring to the cutaneous scaling that is characteristic of these disorders, which is said to resemble the scales of a fish[1-3]. Scaling can be localized or generalized and can be associated with a variety of additional cutaneous and/or systemic manifestations[2, 3]. Babies with these conditions present as collodion babies that are covered in thick taut, cellophane like membranes with associated bilateral ectropion and eclabium[1-3]. The membrane sheds over a few days to weeks after birth to reveal the true nature of the underlying condition which may either be lamellar ichthyosis, recessive x-linked ichthyosis, congenitalivhthyosiform erythroderma, harlequin ichthyosis or a normal variant otherwise known as self-healing Collodiun baby. They are usually distinguishable on the basis of inheritance patterns, clinical features, associated defects, and histopathological changes[1, 3]. Some of these conditions cause disfigurement and considerable psychosocial trauma, so early diagnosis is helpful to predict probable course, prognosis and to provide supportive management for patients, parents and caregivers[1, 3]. We present two cases of collodion babies that were seen in our hospital within a space of two months.

Case 1

Baby A, was a female neonate who was brought to our hospital at the age of 40 hours. She was delivered at a peripheral hospital to a 37year old para 2, (2 alive) mother via spontaneous vaginal delivery at term. Baby was said to have cried well immediately after birth. The Apgar score was not stated. Birth weight was 2.6kg. She was referred on account of abnormal features with a thick shiny membranous covering all over the body, noticed at birth. Mother attended antenatal clinic regularly at the hospital of referral and regularly received heamatinics. The ultrasound scan done in pregnancy did not show any abnormality. Pregnancy was uneventful, there was no history of illicit drug use or exposure to radiation. Father and mother had tertiary level of education and were civil servants. There was no history of consanguinity and no family history of skin disorders or congenital abnormalities. Mother was devastated, refused to touch baby and would not express breast milk to feed the baby. Examination revealed a term female neonate with bilateral ectropion, flat nose with small nasal opening, deformed low set ears, widely opened mouth with small maxilla and generalised membranous sheet covering of the body. Systemic examination was normal.

An assessment of a collodion baby was made. Full Blood Count showed, PCV-38%, WBC-8,500/mm^3, Platelet- 234,000/mm^3, Neutrophil- 56%, Eosinophil-10%, Lymphocytes-3%, Monocytes- 04%, Target cells-++. She was nursed in an incubator for humidification and temperature control, commenced on oral vitamin A, antibiotics and Breast milk substitute feeding via orogastric tube. Saline compressed and moisturisers were used. She was co-managed with the Ophthalmology, Dermatology and ENT, teams. She was commenced on

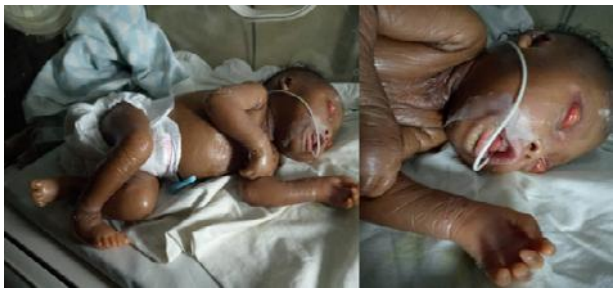
chloramphenicol and moxifloxacin eye drops with hypertonic saline dabs by the Ophthalmologists and six hourly use of Emollients (Vaseline petroleum jelly+5% urea) by the dermatologists. Mother was referred to the psychiatrists where she received psychotherapy and was better disposed to the baby before discharge, however she was still sceptical. Baby started shedding the skin membrane from the 3rd week of admission leaving a scally and reddish underlying skin. Hence the final diagnosis of lamellar ichthyosis.
She made significant clinical improvement and was discharged home to her parents after three weeks. She was worked up for biopsy however parents declined. At discharge they requested that the baby be followed up at the teaching hospital in the neighbouring city because of the social stigma. They however visited the hospital only once after so much persuasion on phone at the age of six months with features of malnutrition and did not go back. Further enquiry about the child on phone, revealed that she died at home at the age of eight months.

Case 2

Baby B was a 42day old male infant that presented to our hospital on account of abnormal facies and abnormal skin. Baby was delivered at home with abnormal fasciae and thick shiny membranous covering of the body. Both parents had no formal education, father is a herdsman and mother did not receive any form of antenatal care. Despite verbal referral to our hospital at the hospital where baby was taken to after delivery, parents were influenced by the family elders to take the baby back home because he was abnormal. No immunisation was given to the baby due to the skin condition. Herbal concoctions were given to the baby and also used to bath the baby however parent reluctantly brought the baby on advice of an acquaintance when no obvious changes were seen. There was no family history of similar skin disorders. Examination finding includes; widely opened mouth with thick membranous covering over the body, bilateral ectropion, flat nostrils with almost blocked nasal openings, flattened ears with small auditory canal, thick constricting bands over the joints and limbs.

He was admitted and commenced on oral vitamin A, moxifloxacin eye drops with hypertonic saline padding of both eyes, six hourly applications of emollients (Vaseline petroleum jelly+5% urea). The parents were counselled about the child`s condition and the prognosis. They refused to do any investigations and eventually discharged against medical advice (DAMA) after 48 hours of admission.

A – Case 1 at 40hours of age



B – Case 1at 6months of age

C – Case 2 at presentation

Discussion

Ichthyoses are rare genodermatosis that was first described in history by Rev. Oliver Hart in America 1750[4], while the term collodion baby was first used to describe a newborn in 1884 by Hallopeau and Watelet[2, 5, 6], since then about 270 cases have been reported in literatures worldwide with incidence ranging between 1 in 300,000 to 600,000[2, 7]. In Nigeria the first case reported was in 1985 by Okoro et al[8] with pockets of cases reported[9-15] since then. Cases have also been reported in other parts of Africa[6, 16, 17]. Congenital Ichthyosis constitute a wide phenotype of disorders of cornification and keratinization with varying outcome and severity. These different phenotypes can be differentiated based on their clinical manifestations, genetic presentation, and histologic findings[3]. They are mostly inherited in autosomal recessive mode example, Lamellar Ichthyosis (LI), Congenital Icthyosiform Erythroderma (CIE) and Harelquin Icthyosis (HI). Other modes of inheritance are x-linked and autosomal dominant[1, 3].

Lamellar ichthyosis (LI) is an autosomal recessive genetically heterogeneous skin disease caused by mutations involving multiple genetic loci. It involves mutation in the gene for transglutaminase 1(TGM1) on band

14q11[1, 2, 5], which is involved in the formation of the cornified cell envelope. The cornified cell envelope is the structure on which intercellular lipid layer forms in the stratum corneum, hence the mutation of this gene leads to a defective barrier function of the stratum corneum. Type 1 maps to band 14q11.2 and is caused by mutations in the gene for keratinocyte transglutaminase 1, an enzyme responsible for the assembly of the keratinized envelope. Type 2, which is clinically indistinguishable from type 1, maps to band 2q33-q35[18]. Loss of the fillagrin gene has also been identified in patients with ichthyosis, other genes identified in lamaller ichthyosis include ABCA12 (2q34), 19p12-q12, 19p13, ALOXE3-ALOX12B (17p13), Ichthyin (5q33)[2, 18].

Lamellar ichthyosis, though rare affects all populations with cases reported in different parts of the world including Africa. Global prevalence is less than 1 case per 300,000 individuals with no sexual predilections[5, 6]. Patients with lamellar ichthyosis have accelerated epidermal turn over with proliferative hyperkeratosis[19]. The disease is apparent at birth and continues through out life. The newborn is born encased in a collodion membrane, hence the name "collodion baby" [1-3, 5]. Collodion babies are often born prematurely and present at birth encased within a shiny, taut, cellophane-like membrane[1-3, 5]. They have flat noses with small openings and small deformed ears. The tight skin around the eyes and mouth often leads to ectropion (outturning of the eyelids) and eclabium (eversion of the lips), respectively[1, 2]. The tight shiny membrane is shed after about 2-4 weeks revealing a generalised scaling with variable redness of the skin. The scales are arranged in a mosaic pattern resembling fish skin, involve the entire body and is increased in the flexural surfaces causing constrictures/ fissures[2, 7].

The scalp may develop thick adherent scales that may lead to scarring alopecia[1] as seen in our first case when she presented at the neighbouring teaching hospital at the age of 6months. The baby was also malnourished, this is another complication[14] that can arise. They are prone to excessive dryness of the skin leading to problems with temperature regulation, water loss, secondary infections, and systemic infection[1, 20]. Keratitis with secondary corneal ulceration may occur as a result of long-term exposure[20]. There can be nail dystrophy with nail fold inflammation, subungual hyperkeratosis and longitudinal/ transverse stippling[20]. Ultimately, there is significant disfiguring of the body which may cause some psychological stress to affected patients and their parents, as observed in the cases presented[1, 3].
The exact cause of lamellar ichthyosis is not known but as with most diseases inherited in autosomal recessive inheritance pattern and they are very rare and associated with consanguinity[1, 5]. Consanguinity was observed in some reported cases[2, 3] including those reported in Nigeria among those of Fulani descent[2, 10], however in our cases there was no consanguinity even in the second case who was also of Fulani descent. Diagnosis is made through clinical features, histologic findings, genetic mutation analysis and hormonal assays[11]. Both cases presented were diagnosed clinically not for lack of availability but the parents did not consent to the diagnostic investigations.

The management of ichthyosis requires the collaborative efforts of the Paediatricians, Dermatologists, Ophthalmologists, Genetic counsellors and Physiotherapist[2, 3, 5, 7]. First steps in the management of this condition are extensive counselling of care givers on the nature of the disease and the management process[1, 3, 6]. There might be need for a psychologists/ psychiatrist intervention as in the first case presented. This is a very important step in the care of the baby which cannot be overemphasized as it determines the willingness to receive care as we discovered in both cases presented. Other cases reported[9, 21] have also shown that the fear of social stigma and misconceptions about the conditions have hampered care or prevented presentation at the hospital for care. The next important step in the management is to nurse in humidified incubator to prevent heat and water loss[2, 3, 5, 7]. Then the frequent use of Emollients to keep the skin lubricated example of emollients available are Vaseline (petrolatum jelly) water in oil preparations e.g., Eucerin, Urea creams, SSA, 5% lactic acid[1, 2, 5, 7]. Oral retinoids as keratolytic example is oral Isotretinoin which is structurally related to vitamin A, decreases sebaceous gland size and sebum production. Topical retinoids- inhibits microcomedone formation, makes keratinocytes in the sebaceous follicles less adherent and easier to remove[1-3, 5, 7]. The collodion membrane should not be peeled-off, however bands of tight skin constricting the chest, digits, hands or feet may occasionally require surgical division[2].

Eye care can be conservative or surgical[5]. The non-surgical treatment includes application of 5% hypertonic saline, eye lubrication with antibiotic ointment like chloramphenicol eye ointment, injection of hyaluronic acid and eye patching. While surgical treatment includes temporal tarsorrhaphy, subconjunctival injection of hyaluronic acid, fornix sutures and full thickness skin graft of the upper lid. The use of antibiotics to prevent or treat infection is paramount as they are highly predisposed to infection[2, 5, 7]. Close attention should also be paid to the fluid, electrolyte and nutrition of the baby[2, 5, 7]. Our first case survived the neonatal period and was discharged as meticulous attention was paid to the above line of management but the fear of stigmatization prevents the parents from attending follow up leading to the eventual demise of the baby.

Conclusion

Lamellar ichthyosis, though rare, is a condition that requires significant attention in the neonatal period. Successful management of ichthyosis in the newborn can be achieved through thoughtful, directed interdisciplinary approach.

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