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Unveiling the Spectrum of Congenital Ichthyosis: A Case Series

Keyur Sabnis, Jayesh Panot, Vandana Kumavat, Mohit Rojekar*,

Department of Biochemistry, Rajiv Gandhi Medical College, Thane, India

CASE STUDY

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Corresponding Author:

Mohit Rojekar,
Department of Biochemistry,
Rajiv Gandhi Medical College,
Thane, India
drmohi44@gmail.com

ABSTRACT

Introduction

Congenital ichthyosis refers to a group of genetic disorders that affect the process of skin cell maturation, often leading to abnormal skin symptoms[1]. These conditions can involve the production of scales with unusual characteristics or quantities, an abnormal thickening of the outermost layer of the skin (stratum corneum), or irregularities in the turnover of keratinocytes, which are the predominant cells in the epidermis. In some cases, these disorders may also cause skin inflammation. Rare forms of ichthyosis can be accompanied by additional symptoms such as itching (pruritus), delicate skin that is prone to injury (skin fragility), turning out of the eyelids (ectropion) or lips (eclabium), and decreased sweating (anhydroses).

"Ichthyosis" is a term used to describe various keratinising disorders that have different causes but share common characteristics of generalised epidermal hyperkeratosis (thickening of the outermost layer of the skin) and dry, scaly skin. The severity can range from having a plastic wrapped appearance as seen in case of collodion babies, to thick hard plates that cover the skin all over the body as seen in case of Harlequin Ichthyosis.

Case Series

We present two cases of Congenital Ichthyosis (CI), one being the milder form while the other being the most severe form CI. The patients were seen at Rajiv Gandhi Medical College in Kalwa, Thane.

Case 1

The first case was a neonate born to a 20-years-old woman, of birth order 3, and third degree consanguinity, at the first day of life presented with ambiguous genitalia and features of white parchment like membrane covering whole of the body. The baby also had claw-like appearance of hands and feet (Figure 1). Ectropion was also reported. The baby was born with 1.8 kg birth weight at 34.2 weeks gestation through a preterm vaginal delivery. The first child of the mother was female, that died at the time of delivery, and had similar features.

On the first day, the baby was admitted in the NICU and umbilical line was secured. Prophylactic antibiotics were started. Paraffin gauze dressing was prescribed by the dermatologist. Gatifloxacin eye drops and lubricating eye gel was prescribed by the ophthalmologist.

In the next few days, USG was performed which confirmed the baby to be a male child. Distal fingers showed necrosis on the fourth day. Blood examination revealed a macrocyctic hypochromic anemia. Inspite of strict aseptic measures undertaken, the baby became septic. Blood and pus culture revealed Acinetobacter Spp. and Klebsiella pneumonia infection. The baby died due to septicaemia on the ninth day.

Case 2

The second case was a female neonate born to a 25-years-old woman, of similar birth order and degree of consanguinity as the previous case. She had a plastic wrapped appearance (Figure 2) on the entire body with ectropionandeclabium. Oedematous hand and feet, nasal and aural cartilage hypoplasia and an increased muscle tone was also reported. The baby was born with a 2.4 kg birth weight at 37.2 weeks of gestation through a cesarean section. No family history of ichthyosis was reported.

First day management was conducted in a similar manner as the previous case. A nasal blockage was noticed on the fifth day. On the sixth day, elevated C - Reactive Protein (CRP) and blood culture revealed Non- Albicans Candida infection. On the seventh day, she went into respiratory distress,

although X-Ray showed no significant findings. An episode of diarrhoea was reported on the ninth day for which probiotics were initiated. Declining SpO2 along with septic shock led to respiratory failure. The baby succumbed to septicaemia on the eleventh day.

A pedigree chart of the two cases has been shows in Figure 3 and 4.

Discussion

Ichthyoses refer to a collection of genetic disorders that involve abnormalities in the process of skin cell maturation, resulting in the clinical manifestation of skin scaling. These conditions often present with noticeable inflammation of the skin. In many cases, individuals affected by these disorders experience a considerable decline in their quality of life[2].

Autosomal Recessive Congenital Ichthyoses (ARCI) are lifelong skin conditions characterised by widespread scaling and varying degrees of redness that typically become apparent at birth or during early infancy. ARCI includes different types of non syndromic ichthyosis, which exhibit significant variations in their clinical presentation and severity. These types encompass a spectrum ranging from the most severe and potentially life- threatening form called harlequin ichthyosis to Lamellar Ichthyosis (LI), (Nonbullous) Congenital Ichthyosiform Erythroderma (CIE), and intermediate phenotypes displaying varying levels of redness, as well as variations in the size and texture of the scales.

The first to give the term Collodion Baby (CB) were Hallopeau and Watelet. Children with autosomal recessive congenital ichthyosis (ARCI) are frequently born prematurely, and they are often covered by a tight, shiny, and transparent membrane called a collodion membrane. This membrane, which resembles plastic wrap, is formed by thickened layers of the skin. These infants are prone to significant transepidermal water loss, leading to dehydration and high levels of sodium in the blood (hypernatremia). During the neonatal period, they face an increased risk of skin infections and sepsis[3]. The tautness of the collodion membrane commonly causes complications such as ectropion (outward turning of the eyelids), eclabium (eversion of the lips), and underdeveloped cartilage in the nose and ears. Impaired ability to suck and ventilate their lungs can result in dehydration, malnutrition, oxygen deprivation (hypoxia), and pneumonia.

A scoring system has been created to evaluate the prognosis of neonates with a collodion membrane by considering multiple factors[4]. Previous case reports have shown a connection between collodion baby and drug intake, such as infliximab[5]. Biopsy results have indicated a thickening of the outermost layer of the skin, known as the stratum corneum.

In the most uncommon form of Autosomal Recessive Congenital Ichthyosis (ARCI) known as harlequin ichthyosis, infants are born prematurely and have a covering of thick, hard, and armor-like plates of skin that is separated by deep cracks. This tight skin causes facial deformities, microcephaly (small head size), absence of eyelashes and eyebrows, and sometimes hair loss. Bands of hardened skin can constrict blood vessels, leading to restricted blood flow, tissue damage, and swelling in the hands and feet resembling mittens. A comparison between the two cases has been presented in Table 1.

Newborns with harlequin ichthyosis are susceptible to life-threatening complications during both the neonatal and postnatal periods. These complications include respiratory insufficiency, dehydration, imbalances in electrolytes, difficulty maintaining body temperature, problems with feeding, bacterial infections, and sepsis, which can potentially result in fatal outcomes.

Reported survival rates for harlequin ichthyosis have been around 56 Per cent[6]. However, with advancements in neonatal intensive care and treatment options such as early application of topical and/or systemic retinoids, it is anticipated that the survival rate will continue to improve.

While abnormalities in lipid transport, protein phosphatase activity, and cellular differentiation have been identified, the genetic cause behind the clinical and cellular characteristics of Harlequin ichthyosis (HI) remains unknown. However, the presence of a defect in the ABCA12 gene in individuals with HI may provide an explanation for the observed abnormalities in lamellar granules, which could be linked to a significantly impaired skin - Barrier function. This phenomenon is similar to the storage of surfactant in lamellar bodies within alveolar type II cells in the lungs[7].

The ectropion can attract opportunistic microorganisms. Preservation of visual function is crucial in survivors. Persistent ectropion is a frequent complication, and in a 45-cases study, 20% of the cases underwent surgical correction [8].

The treatment is mainly supportive like the use of incubators, IV fluids and tube feeding. Additionally, use of emollients and retinoids therapy ensures better prognosis[9].

Conclusion

Although very essential but we could not make a definitive diagnosis to determine the phenotype of the ichthyosis, since it needs genetic evaluation of both parents and the neonate. This facility is currently unavailable at our hospital, hence we had to rely on clinical presentations to make the diagnosis.

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Figures

Figure 1: Thick scales all over the body with claw-like hands and feet; ectropion and eclabium are clearly visible. Baby of case 1.

Figure 2: Plastic wrapped appearance all over the body with visible ectropion and eclabium. Baby of case 2.

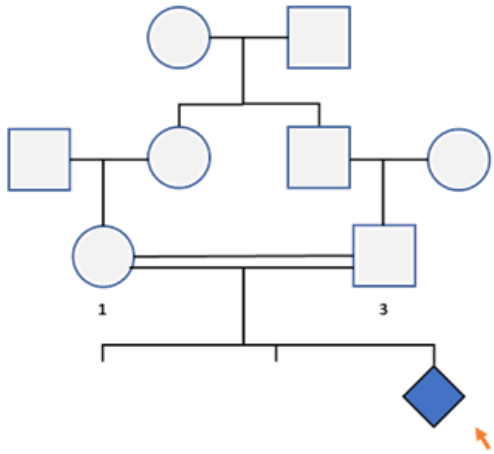

Figure 3: A pedigree chart of the first baby born with Harlequin ichthyosis.

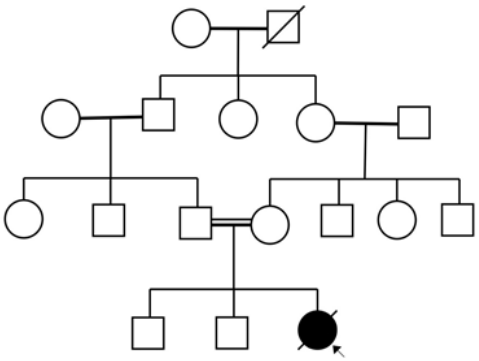

Figure 4: A pedigree chart of the second baby born with collodion membrane.

Table 1: Comparison between the two cases.

Patient	Sex	Family history of Ichthyosis	Perinatological History	Ectropion	Eclabium	Scales
1	Male	Yes	Preterm and Underweight	Severe	Severe	Present
2	Female	No	Full term baby	Severe	Mild	Absent

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