Rathod KG et al. GAIMS J Med Sci 2024;4(2) (Jul-Dec):53-55
Online ISSN: 2583-1763

Case Report

Harlequin Ichthyosis: Navigating the Challenges of a Rare Case

Kamleshkumar G Rathod[1*], Parth Mali[2], Bharat Muliya[1]

Department of Paediatrics, [1]C.U. Shah Medical College and Hospital, Surendranagar, [2]Himalaya Medical College, Patna
* Correspondence: Dr Kamleshkumar G Rathod (kamleshrathod1711@gmail.com)

ABSTRACT

Ichthyosis, derived from the Greek word "ichthys" meaning fish, encompasses various skin disorders characterized by dry, scaly, and thickened skin, often inherited through autosomal or X-linked modes. This case report discusses a rare and severe form known as Harlequin ichthyosis, presenting a second gravida woman with consanguinity history who delivered a male baby with characteristic features including porcelain-like skin, ectropion, eclabion, and flexion deformities. Despite supportive measures, the baby died shortly after birth. Harlequin ichthyosis, with an incidence of 1 in 300,000 births, arises from mutations in the ABCA12 gene, affecting lipid exocytosis and desquamation. Antenatal diagnosis is feasible through ultrasound findings and genetic testing, with a grim prognosis and limited survival beyond infancy. This report underscores the challenges in managing such conditions and highlights the importance of ongoing research for better understanding and management.

Keywords: Harlequin ichthyosis, Congenital ichthyosiform erythroderma, Autosomal, ABCA12 gene, Antenatal diagnosis

INTRODUCTION

Ichthyosis is derived from a Greek word, ichthys, which means fish. It refers to a fish-scale- like skin's look. Skin conditions known as ichthyoses are characterized by thicker, dry, and scaly skin. Ichthyosis can be inherited or acquired, affecting not just the skin but also other organs. Autosomal or X-linked inheritance can be either recessive or dominant. The three primary types of autosomal recessive congenital ichthyosis are Harlequin ichthyosis, lamellar ichthyosis, and congenital ichthyosiform erythroderma. The least prevalent kind of harlequin ichthyosis is also the most severe. Hart recorded the first case in 1750 in South Carolina, USA.[1] In 1983, the first instance to be identified as prenatal was recorded.[2] Electron microscopy of fetal skin biopsies and DNA-based diagnosis using chorionic villus sample or amniocentesis can be used to confirm the antenatal diagnosis in suspected cases.[3] Since there is no known treatment for this sickness, supportive care is the only thing that can be done to prolong life.

CASE HISTORY

A 27-year-old primigravida who had registered with a private hospital went to the labor room at 35 weeks of gestation, exhibiting breech presentation and preterm premature rupture of membranes in latent labor. She had regular menstrual cycles with no significant medica and surgical history. After three years of marriage, a history of second-degree consanguinity was discovered. Polyhydramnios was found at a previous ultrasound. The scan did not reveal the other anomalies. Due to her breech presentation, she had an emergency cesarean section, and on August 3, 2021, a male baby weighing 2.1 kg was born.

As seen in Figure-1, the infant's body was covered in deep wrinkles and a white, porcelain-like skin that resembled armor. There was visible bleeding from the wrinkles. At birth, the baby let out a typical cry. Ectropion and eclabion were displayed by everted lips and eyelids, respectively. Nasal hypoplasia was seen. As shown in Figure-2, the lips were thick and the mouth was open. The pinna was closed and the ears were little. There was a permanent flexion deformity observed when the toes and fingers were pointed out. At birth, the newborn's Apgar scores were 7 and 9 at

one and five minutes, respectively. With a respiratory rate of 56 breaths per minute and a stable temperature of 37°C. The baby was Hemodynamically stable and oxygen saturation remained at 92% on room air. The head circumference was 35 cm. For additional care, the infant was admitted to the neonatal critical care unit. Afterwards, it was observed that the baby's skin was peeling off, creating erythematous cracks. The umbilical vein was used since a peripheral intravenous line could not be established. Conservative treatment included intravenous antibiotics, emollients and acitretin. There was a feeding tube used to administer meals. The patient was died after 20 hours of life. The parents refused to have the infant's skin sampled for a skin biopsy and an autopsy. Genetic counseling was summoned for the parents.

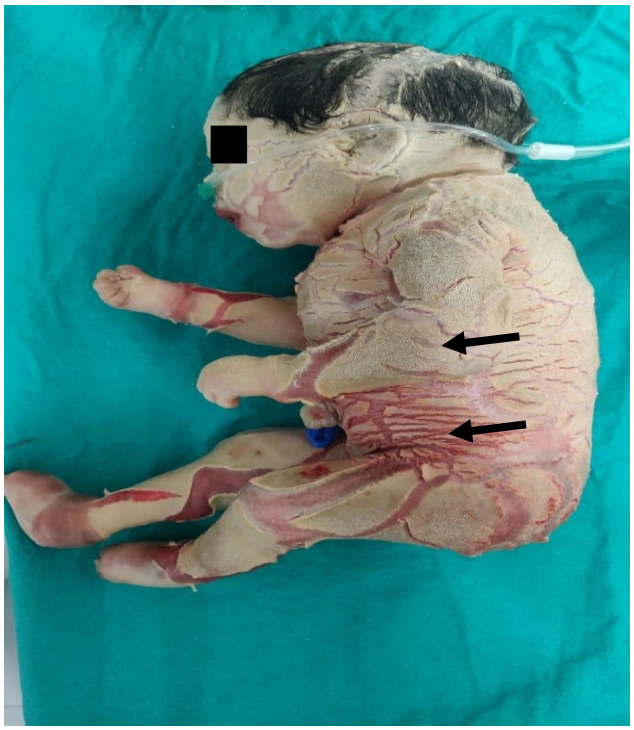

Figure-1: Deep wrinkles and a white, porcelain-like skin

DISCUSSION

Other names for Harlequin ichthyosis include ichthyosis congenita, harlequin baby syndrome, and harlequin fetus syndrome. A comedic servant character with a similar appearance is the source of the word "Harlequin." With a prevalence of 1 in 300,000 overall births, harlequin ichthyosis is an uncommon kind of congenital ichthyosis.[3] Globally, there have been about 200 cases documented.[4] A case from June 2016 in Nagpur, India was just described and the International Journal of Pharmacy published it.[5] It is inherited as a recessive autosomal.[3] Children, whether male and female, might be affected equally. It is brought on by a mutation in the adenosine triphosphate binding cassette A12 (ABCA12) gene. This gene is in charge of lipid-containing lamellar granule exocytosis, which regulates the desquamation process.[6] The ABCA12 gene locus is located on chromosome 2q35.[7] Null mutations and frameshift mutations are the main mutation types that cause this.[7] Utilizing the following characteristics, prenatal screening for this disorder can be carried out: flexion of the extremities, mottled, breeched skin on the face and limbs, hyperflexion of the fingers and toes, lack of finger opening movements, closed eyes with eversion of the lips and eyelids (ectropion and eclabion, respectively), restricted fetal growth, flat nose, open mouth, and malformed ears.[1] It is preferable to use a 3D scan to diagnose this illness. Respiratory insufficiency, supervening infections, and dehydration are among the potentially fatal consequences. The outcome is not good. Most affected babies die during the first week of their lives. The survival rate with supportive care has been reported to range from 10 months to 25 years, depending on the severity of the condition.[8,9] Pregnancy-related recurrence of this disease is 25%.[1] In these circumstances, genetic counseling ought to be initiated.

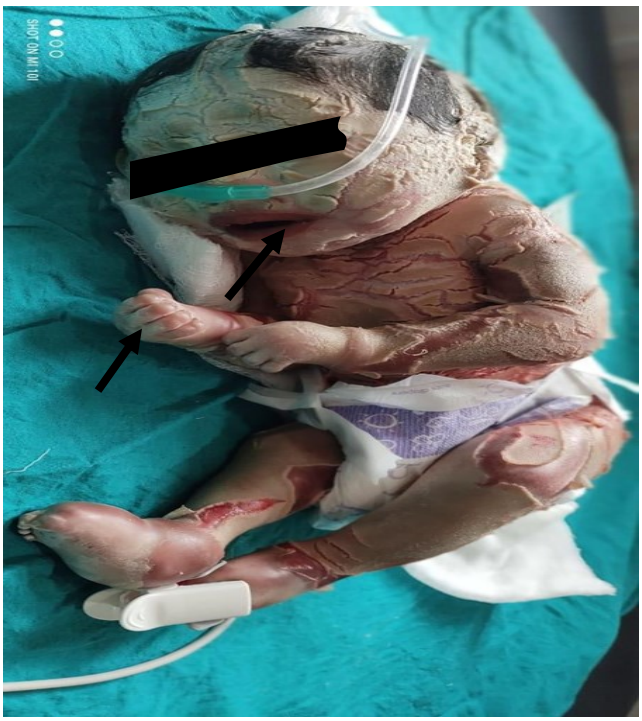

Figure-2: Thick lips and flexion deformities

CONCLUSIONS

This case report provides the information about intricacies and difficulties encountered in managing a rare and severe genetic disorder. The challenges faced in the diagnosis,

treatment and ongoing care of the patient emphasize the need for continued research and advancements in medical science to enhance our understanding of this rare condition. The report serves as a valuable resource for medical professionals, offering insights into the potential complications, therapeutic interventions and long-term considerations for individuals affected by this rare disorder. Continued awareness, research and collaborative efforts are essential to pave the way for better outcomes and support for individuals and families affected by Harlequin Ichthyosis.

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Authors' Contributions

Author 1: Contributed by Collection, analysis and interpretation of data related to the case and also drafting of the original manuscript, including writing and formatting.
Author 2: Conducted detailed investigation and literature review related to the case. Provided critical review and editing of the manuscript for intellectual content and accuracy.
Author 3: Provided oversight and guidance throughout the project as the senior author. Validated the accuracy and integrity of the data and analyses presented and gave final approval for the submission of the manuscript for publication.

Source of support: Nil

Conflict of interest: None declared

How to cite: Rathod KG, Mali P, Muliya B. Harlequin Ichthyosis: Navigating the Challenges of a Rare Case. GAIMS J Med Sci 2024;4(2):53-55.

https://doi.org/10.5281/zenodo.11575779