MAK HILL Publications Research Journal of Medical Sciences

Case Report
doi: 10.36478/makrjms.2024.9.238.240

OPEN ACCESS

Key Words
Abnormal, features, significant, reaspiratory

Corresponding Author
Shreedevi Kori,
Department of Obstetrics and Gynecology, Blde(DU) Shri B.M. Patil Medical College, Hospital and Research Centre, Vijayapura, Karnataka, India

Author Designation
[1] Junior Resident
[2,3]Associate Professor
[4,5]Professor

Received: 15 July 2024
Accepted: 15 August 2024
Published: 18 August 2024

Citation: G.V. Vinithira Sri, Shreedevi Kori, Dayanand Biradar, Rajasri Yaliwal and Subhashchandra Mudanur, 2024. Harlequin Ichthyosis: Insights from a Neonatal Case Report. Res. J. Med. Sci., 18: 238-240, doi: 10.36478/makrjms. 2024.9.238.240

Harlequin Ichthyosis: Insights from a Neonatal Case Report

[1]G.V. Vinithira Sri, [2]Shreedevi Kori, [3]Dayanand Biradar, [4]Rajasri Yaliwal and [5]Subhashchandra Mudanur

[1-5]Department of Obstetrics and Gynecology, BLDE(DU) Shri B.M. Patil Medical College, Hospital and Research Centre, Vijayapura, Karnataka, India

ABSTRACT
Harlequin ichthyosis (HI) is a rare and severe genetic disorder marked by extreme skin thickening and abnormal shedding. This case report describes a neonate born to a first-time mother, presenting classic HI features, with no significant family history of the condition. Despite receiving intensive care, the infant unfortunately succumbed to sepsis and respiratory failure. This report emphasizes the need for early diagnosis, genetic testing, and thorough prenatal screening to enhance the management and prognosis of HI in newborns.

INTRODUCTION

Harlequin ichthyosis (HI) is a rare and life-threatening form of hereditary ichthyosis caused by the deletion of the keratinocyte lipid transporter and truncated proteins[1].It is a rare condition that affects one in every 3,000,000 live infants, with only 200 cases reported globally[2-3]. It is an autosomal recessive disorder caused by mutations in the ABCA12 gene, which encodes the ATP-binding cassette (ABC) transporter protein found in the lamellar granules of upper epidermal keratinocytes[4]. It is distinguished by aberrant desquamation and widespread epidermal hyper-keratinization. Harlequin ichthyosis can be quickly identified in a newborn by its characteristic appearance, which includes thick, plate-like scales separated by deep fissures, bilateral ectropion (where the eyelids turn outward), eclabium, flattened ears and nose, and constricting bands around the limbs. Neonates are particularly vulnerable to sepsis, dehydration and poor thermoregulation because of the catastrophic degradation of the epidermal barrier[4]. In this instance, we report a low-resource case of fatal Harlequin ichthyosis without a family history of any inherited skin condition.

Clinical presentation: A primigravid woman in her mid-twenties presented with a history of severe oligohydramnios at 35 weeks of gestation. She had been attending regular antenatal check-ups, and all previous scans, including the anomaly scan, had shown no abnormalities. Aside from a history of a second-degree consanguineous marriage, there was no other significant past or family history. The oligohydramnios was first noted at 33 weeks of gestation and was managed conservatively with intravenous hydration and corticosteroids. However, due to the persistence of severe oligohydramnios, an emergency cesarean section was performed at 35 weeks. A live female infant, weighing 2.45 kg, was delivered by cesarean section. The baby exhibited a weak cry and had APGAR scores of 7 at one minute and 8 at five minutes. Examination revealed hyperkeratotic, plate-like scales across the body, notably on the face. This led to bilateral ectropion (eversion of both upper and lower eyelids), eclabium (outward turning of the lips), and a flattened nasal bridge. These clinical features were consistent with Harlequin Ichthyosis. The neonate was transferred to the neonatal intensive care unit (NICU) for advanced care. The baby was initiated on broad-spectrum intravenous antibiotics, provided with sufficient nutrition via a nasogastric tube and had their body temperature closely monitored and maintained. Although the plan was to initiate oral retinoids on the third day of life, the baby's condition worsened, characterized by increased scaling and respiratory distress that necessitated ventilatory support. Despite these interventions, the neonate succumbed to sepsis and respiratory failure on the fifth day of life. The parents were advised to have the neonate undergo an autopsy and skin biopsy for further confirmation. Additionally, they were encouraged to undergo testing for plausible genetic mutations. However, they declined for both, due to cultural concerns and financial limitations.

RESULTS AND DISCUSSIONS

Harlequin Ichthyosis also known as ichthyosis fetalis, has a high mortality rate of up to 50% even with the recent advancements. The etiology and manifestation of harlequin ichthyosis could be more intricate than a simple keratinization condition. Research indicates that the functions of ABCA12 may be broader than previously thought, encompassing immunological control, antimicrobial peptide production, and respiratory homeostasis. Infants diagnosed with HI are more prone to infections and respiratory problems[5].Amniocentesis or chorionic villus sampling (CVS) can be used to get a fetal DNA sample and search for ABCA12 gene alterations to diagnose it. For prenatal diagnosis, ultrasound is also essential, particularly in situations where there is no family history. Sonographic diagnosis is difficult to make early; most instances are found in the third trimester. Atypical facial features, a large open mouth, flat nose skin, minimal fetal movement with stiff limbs and no associated visceral anomalies are prenatal ultrasonography findings indicative of a harlequin fetus. To diagnose HI and interpret 2D images, 3D imaging is necessary. Because distinctive traits frequently arise later, multiple scans are required, even if the second-trimester anatomy scan is normal.[6] The ultrasound may also show hyperechogenic amniotic fluid, intrauterine growth restriction, oligo, or polyhydramnios[6]. In the Neonatal Intensive Care Unit (NICU), initial management calls for monitoring and supportive therapy to preserve quality of life. Humidified incubators are used to monitor temperature regulation. Infants must receive nutritional support via tube feedings until their eclabium subsides and they can begin feeding. Amputation may result from gangrene and tissue

Fig 1: Neonate showing characteristic features of Harlequin Ichthyosis

necrosis caused by limb contracture. Therefore, it is important to debride the hyperkeratotic constrictive bands to avoid ischemia of digits. Physiotherapy, analgesics for deep fissure pain and adequate infection control are other management methods. Frequent evaluations by ophthalmologists and the use of artificial tears are advised methods of eye care. Skin softening and desquamation are facilitated by the use of saline compression dressings and mild ointments[7]. Early administration of systemic retinoids can accelerate this change in clinical presentation and potentially reduce mortality because they promote keratolysis, keratinocyte differentiation and the shedding of the thick hyperkeratotic encasing. The survival rate has increased with the use of retinoids and ranges from 10 months to 25 years.

CONCLUSION

Genetic testing for the ABCA12 gene is indicated in cases of consanguinity marriage and positive family history of HI. The likelihood of a recurrence in subsequent pregnancies is 25%. Due to the late manifestation of these traits, it is essential to perform a thorough scan at both the anomaly and following growth scans. Since there is no cure and the majority of care is supportive, early diagnosis is crucial.

REFERENCES

1. Harvey, H.B., M.G. Shaw and D.S. Morrell, 2010. Perinatal management of harlequin ichthyosis: A case report and literature review. J. Perinatology, 30: 66-72.
2. González, R.A., A.V. Ojeda, N.A. Nova and R.J. Prasca, 2024. Harlequin ichthyosis, prenatal diagnosis: The ultrasound recognition. BMJ Case Rep., Vol. 17, No. 1 .10.1136/bcr-2023-256859.
3. Baby, C., 2016. Harlequin ichthyosis: A disease chronic rather than fatal. wor Jo Phar PharScie., 14: 1957-1966.
4. Rajpopat, S., C. Moss, J. Mellerio, A. Vahlquist and A. Gånemo et al., 2011. Harlequin ichthyosis: a review of clinical and molecular findings in 45 cases. Arch. Dermatol., 147: 681-686.
5. Parikh, K., K. Brar, J.B. Glick, A. Flamm and S.A. Glick, 2016. A case report of fatal harlequin ichthyosis: Insights into infectious and respiratory complications. JAAD Case Rep., 2: 301-303.
6. Vijayakumari, M., D.K. Reddy, M. Routhu, M. Vuchuru and N.S. Reddy, 2020. Prenatal diagnosis of harlequin ichthyosis: A case report. Obstet. amp Gynecol. Sci., 63: 94-97.
7. Tsivilika, M., D. Kavvadas, S. Karachrysafi, A. Sioga and T. Papamitsou, 2022. Management of harlequin ichthyosis: A brief review of the recent literature. Children, Vol. 9, No. 6 .10.3390/ children9060893.