ORIGINAL RESEARCH PAPER
Volume - 13 | Issue - 11 | November - 2024 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

BENEATH THE COLLODION MASK: OCULAR CHALLENGES IN HARLEQUIN ICHTHYOSIS – A CASE REPORT

Ophthalomology

Dr. Pranay Sah* Junior Resident, Department Of Ophthalmology, D Y Patil Medical College, Kadamwadi Kolhapur. *Corresponding Author

Dr. Gayatri Rajendra Gondhali Junior Resident, Department Of Ophthalmology, D Y Patil Medical College, Kadamwadi Kolhapur.

Dr Rahul Krishnamurthy Junior Resident Department Of Ophthalmology D Y Patil Medical College Kadamwadi Kolhapur

Dr Milind Sabnis Head Of Department Department Of Ophthalmology D Y Patil Medical College Kadamwadi Kolhapur

ABSTRACT

Harlequin ichthyosis is autosomal recessive disorder with mutation on ABCA 12 gene, characterized by thick, plate-like skin that causes significant ophthalmological signs including ectropion (outward turning of the eyelids), leading to exposure keratopathy. This condition results in dry eyes, corneal abrasions, ulceration, and increased risk of conjunctivitis. This case focusses on 20 days old neonate 2.40 kg who presented in neonatal intensive care unit with ectropion in both eyes. This report aims to highlight the clinical presentation, diagnosis, and management of Harlequin Ichthyosis in this patient.

KEYWORDS

Harlequin Ichthyosis, ectropion, exposure keratopathy, corneal abrasions, ulcerations, conjunctivitis

INTRODUCTION

Harlequin ichthyosis (HI) is a life-threatening genetic skin disorder that significantly impacts quality of life and requires multidisciplinary management. The disorder results from mutations in the ABCA12 gene, leading to severe abnormalities in the skin barrier function. This report presents a case of a 20-day-old infant diagnosed with harlequin ichthyosis, highlighting the ophthalmic complications associated with this condition.[6]

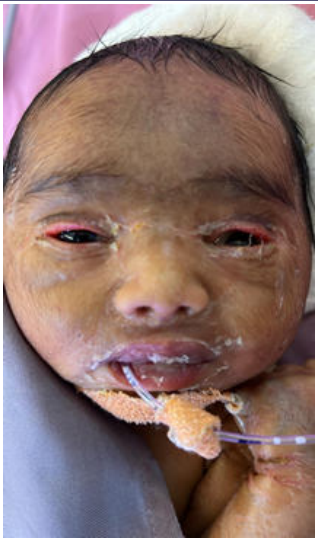
Case Report

The infant was born via full-term normal vaginal delivery at 34.3 weeks gestation, weighing 2.440 kg. He exhibited characteristic features of harlequin ichthyosis at birth, including fine scaly skin and facial deformities. At 20 days, he presented with ectropion affecting both eyes,

Ophthalmic Examination

Upon examination, the following findings were noted:

- **Visual Acuity**: Unable to assess due to age
- **Eyelid Examination**:
- Ectropion present bilaterally, exposing the conjunctiva and cornea
- **Incomplete blinking** and inability to fully close the eyelids
- **Conjunctival Examination**:
- Mild conjunctival hyperemia (redness), observed due to exposure
- No signs of conjunctivitis or discharge
- **Corneal Examination**:
- Superficial punctate keratitis noted
- No corneal ulcers or scarring
- **Pupil and Fundoscopy**:
- Pupil was round regular and reactive to light
- Fundoscopic examination was done to check red reflex



Systemic Evaluation

The infant was active and stable during the evaluation. No other significant systemic issues were noted. The following features were documented:

- **Skin**: Thick, scaly skin; evidence of collodion membrane remnants.
- **Facial Features**: Flat nasal bridge, hyper-telorism, and fish-mouth appearance.
- **Neurological Examination**: No abnormalities detected.
- **Cardiovascular And Respiratory**: Stable vitals; no signs of distress.

Diagnostic Workup

- **Genetic Testing**: Confirmed mutation in the **ABCA12** gene, consistent with harlequin ichthyosis.
- **Ocular Surface Staining**: Fluorescein staining demonstrated areas of dryness and superficial punctate keratitis.

Management and Follow up

The superficial punctate keratitis in both eyes was treated with intensive topical antibiotics. Frequent application of preservative-free artificial tears to prevent dryness and protect the corneal surface. Temporary eyelid taping was initiated to assist in closing the eyelids and reducing exposure.

Every weekly follow up was scheduled to closely monitor ocular health and corneal healing.

Collaboration with Dermatology department was done to manage the skin abnormalities, alongside Paediatrics department for ongoing evaluation and nutritional support to promote healthy growth and development.

DISCUSSION

Harlequin ichthyosis (HI) is a rare autosomal recessive disorder with a significant impact on multiple systems, including the skin, eyes, and respiratory tract. Ocular complications are a prominent concern in the management of infants with HI, with ectropion the outward turning of the eyelids being one of the most common and visually threatening manifestations. The unique pathophysiology of the disorder, characterized by a defect in lipid transport in the skin due to mutations in the ABCA12 gene, results in hyperkeratotic plaques that distort normal eyelid anatomy, leading to exposure-related ocular damage.[1,2]

In this 20-day-old infant with bilateral ectropion, the risk of significant exposure keratopathy was evident from the outset. Ectropion causes the eyelids to pull away from the globe, which prevents normal eyelid closure. This is a critical issue in neonates, as eyelid function is essential for distributing tears, maintaining corneal hydration, and protecting the ocular surface from environmental irritants and infections.[3]

Tear film deficiency is another concern in patients with ectropion. The role of the tear film is essential in maintaining corneal health by providing lubrication, nutrient delivery, and protection against infections. In this case, the infant's reduced tear production and impaired tear distribution due to incomplete eyelid closure predisposed the corneas to drying and epithelial damage, further increasing the risk of exposure keratopathy.[4]

The management of ectropion in Harlequin ichthyosis is multifaceted and focuses on protecting the cornea, preventing infections, and maintaining moisture on the ocular surface. The cornerstone of conservative management in this case was the regular application of lubricating eye drops and ointment. Frequent use of preservative-free artificial tears during the day helped to maintain adequate moisture on the corneal surface, while the use of thicker ointments at night provided prolonged protection during sleep. Neonates with ectropion are at a high risk of ocular infections, particularly bacterial conjunctivitis and keratitis. In this case, prophylactic topical were initiated to prevent bacterial colonization and infection. [5,6]

The long-term prognosis for infants with Harlequin ichthyosis and ectropion depends on the degree of corneal damage and the success of preventive measures. Early and effective management of ocular exposure is key to preventing permanent vision loss. In this case, the multidisciplinary approach—combining dermatologic care, neonatal support, and ophthalmic management—was critical to optimizing outcomes.[7]

CONCLUSION

In this 20-day-old infant with Harlequin ichthyosis and bilateral ectropion, early and proactive ophthalmic intervention was critical in preventing serious complications such as exposure keratopathy and corneal damage. Management with frequent lubrication, infection prevention, and mechanical eyelid support effectively protected the ocular surface in the short term. However, given the ongoing risk of exposure-related complications, long-term follow-up and potential surgical intervention may be necessary. This case underscores the importance of a multidisciplinary approach in managing Harlequin ichthyosis, particularly in addressing its ophthalmological challenges to preserve visual health and quality of life.

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