Corneal ulcer secondary to ectropion in lamellar Ichthyosis: A rare congenital disorder

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Abstract:

We present a rare case of Lamellar Ichthyosis with bilateral ectropion with left sided corneal ulcer with descemetocoele in a four-month-old female child, the youngest ever reported. Ichthyosis is a group of skin disorders characterized by the presence of fish-like scales all over the body (Mushriff, 2016; Turgut *et al.*, 2009). Lamellar Ichthyosis is a rare congenital condition with incidence of 1 in 100,000 (Conditions), affecting males and females equally. Inheritance is Autosomal Recessive (Conditions). It involves generalized body as a collodion baby at birth; once the membrane sheds, patient develops large, thick, brown, pasted scales associated with ectropion, eclabium, scarring alopecia, plantar and palmar hyperkeratosis (Mushriff, 2016). Mild cases are managed with hydration, lubrication and keratolytic-agents (Mushriff, 2016). Severe cases are treated with Oral Retinoid. Proper lubrication and patching of eyes with timely management of ectropion to prevent the exposure keraopathy and related complications are needed in such cases. Secondary infections can lead to vision threatening complications, so child with ichthyosis should be under Ophthalmic observation for early needed interventions.

Keywords:

Corneal ulcer, descemetocoele, ectropion, lamellar ichthyosis

INTRODUCTION

Ichthyosis is a group of skin disorders characterized by the presence of fish-like scales all over the body^[1]. Lamellar Ichthyosis is the rarest form with incidence of 1 in 1 lac, with autosomal recessive inheritance and is found equally in both sexes^[2,3]. It is commonly associated with cicatricial ectropion^[4] which can lead to exposure keratopathy, secondary infections, corneal ulceration and perforation^[2]. We present a case of Lamellar Ichthyosis with bilateral ectropion with left sided corneal ulcer with descemetocoele in a four-month-old female child, the youngest ever reported [Figure 1].

CASE REPORT

A 4-month-old female child was referred for the Ophthalmology consultation, with complaint of persistent watering in both the eyes since birth,

worsening in left eye with mucoid discharge and white lesion observed by parents for two months of age. As per mother, baby was born out of full term normal vaginal delivery with birth weight of 2.2 kg and was under neonatal intensive care for 5 days in view of respiratory distress, after delivery. She started developing rashes all over the body, from face to trunk and then limbs, after 4–5 weeks of birth. At 8 weeks of age, ectropion of lower eyelids was observed by mother. The marriage of parents was consanguineous in nature.

On general examination, the patient had desquamation all over the body. Ocular examination revealed absence of eyebrows, presence of scales over lid skin and eye lashes and grade III ectropion in upper and lower eyelids of both the eyes. Conjunctiva and Cornea was normal in the Right eye and there was no evidence of xerosis or exposure keratopathy. Left eye was congested with a central corneal ulcer and a descemetocoele.

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Figure 1: Lamellar Ichthyosis with bilateral ectropion with left sided corneal ulcer with descemetocoele in a four-month-old female child

Conjunctival swab taken under sterile precautions revealed presence of gram positive cocci. Culture and Antibiotic sensitivity test reported Methicillin Resistant *Staphylococcus aureus* sensitive to Fluoroquinolones and Vancomycin. KOH mount and culture growth was negative for fungi. The baby was started with intensive anti-ichthyosis therapy in the form of frequent massage with Vaseline, Rose Water Ointment and oral Retinoid plus Vitamin D drops, by Pediatrician. Topical lubricant eye drops Carboxymethylcellulose Sodium (0.25 percent) + Hypromellose (0.3%) 4 times a day with Hypromellose 2% eye ointment and proper eye patching at night for Right eye was prescribed to the patient for better lubrication of ocular surface and protection of cornea from exposure keratitis. Moxifloxacin 0.5% eyedrop 4 times a day with Tobramycin eye drops 0.3% 4 times a day was prescribed for the Left eye. As the corneal ulcer was responding well to the topical management, immediate surgical interventions^[5] like glue, bandage contact lens, membrane grafting, or ectropion correction were withheld. Corneal lesion healed with residual leucomatous corneal opacity involving visual axis. Once the acute phase was settled, surgical intervention for correction of bilateral ectropion with donor skin from mother was performed. Baby is due for the management of corneal opacity.

DISCUSSION

Ichthyosis is a group of skin disorders characterized by the presence of fish-like scales all over the body. Ichthyosis is classified into^[1]:

- Congenital Ichthyosis:
 - Ichthyosis Vulgaris
 - X-linked Ichthyosis

- Lamellar Ichthyosis
- Non-bullous Ichthyosis-form Erythroderma
- Epidermolysis hyperkeratosis
- Acquired Ichthyosis
 - Infections (leprosy)
 - Drugs (clofazimine)
 - Malignancies (lymphoma)
 - Nutritional deficiencies
 - Metabolic disorders (hypothyroidism)
 - Systemic Diseases (sarcoidosis)
- Lamellar Ichthyosis

It is an extremely rare condition with incidence of 1 in 1 lac, with autosomal recessive inheritance and is found equally in both sexes^[1,3,6]. Molecular defect involves abnormality of gene present on chromosome 14q11, which encodes for transglutaminase^[7]. It usually presents at birth with generalized body involvement which begins as a collodion baby at birth. Once the membrane sheds, patient develops large thick, brown, pasted (fish-like) scales associated with eclabium, scarring alopecia over scalp and eyebrows, plantar and palmar hyperkeratosis, cicatricial ectropion, chronic blepharitis, rarely cataract^[6].

The association between ichthyosis and cicatricial ectropion, which is the most common eyelid malposition, was first reported in 1834^[2]. Secondary to chronic exposure and infections, corneal ulceration and perforation may occur in these patients. Apart from ectropion, the other factors causing corneal damage are madarosis, conjunctivitis, eyelash retraction, and lagophthalmus^[2].

Mild cases of Lamellar Ichthyosis are managed with hydration, lubrication and keratolytic-agents^[1]. Severe cases are treated with Oral Retinoid. Proper lubrication and patching of eyes with timely management of ectropion to prevent the exposure keraopathy and related complications are needed in such cases. To promote the epithelial wound healing, bandage contact lenses, amniotic membrane transplantation and tarsorrhaphy can be attempted^[5]. Full-thickness skin grafting may be used to repair cicatricial ectropion. In cases with difficulty to procure healthy skin due to scales all over, donor skin (from mother in our case) can be used^[8]. For diffuse limbal stem cell deficiency, combined keratoplasty and limbal stem cell transplantation and long-term systemic immunosuppression may be necessary, although the success rate has been poor.^[2]

In our case of Lamellar Ichthyosis, bilateral Ectropion was an associated feature giving rise to left sided exposure keratopathy with corneal ulcer and descemetocoele. If the ulcer is responding well to the medical management, immediate surgical interventions like glue, bandage contact lens, membrane grafting or tarsorrhaphy can be withheld. The correction of bilateral ectropion to prevent further corneal complications and to promote the outcome of visual rehabilitation management can be performed once the acute phase settles. On review of literature, we found very few similar cases reported with the age of presentation between

6 months to 67 years. Ours is the youngest case reported at the age of 4 months. We insist that all cases diagnosed with Lamellar Ichthyosis should be cross referred to Ophthalmology to prevent the secondary infections and vision threatening complications.

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Conflicts of interest

There are no conflicts of interest.

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