

Case report

Lamellar ichthyosis presenting as bilateral spontaneous corneal perforation

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Abstract

Introduction: Lamellar Ichthyosis is an autosomal recessive, inherited skin disorder characterized by thickening, fissuring and scaling of the skin. **Objective:** To report a case of lamellar ichthyosis and highlight the importance of monitoring corneal health in these patients. **Case:** We report a rare case of bilateral spontaneous corneal perforation in a patient with lamellar ichthyosis. The patient presented with complaints of diminution of vision, foreign body sensation, watering and discharge in both of his eyes for the last three months followed by pain and redness for one week. Visual acuities were light perception in both the eyes. Cicatricial ectropion was seen in both the lower eyelids. Corneal perforation was seen in both the eyes. Lamellar ichthyosis was diagnosed on the basis of scaling and excessive dryness of the entire body skin and was confirmed by skin biopsy. Penetrating keratoplasty of both eyes was done with cataract surgery of the right. The systemic anti-ichthyosis therapy was started. Ectropion of the right eye was corrected, and on follow-up at three months, the patient had a visual acuity of 4 /60 and CFCF in the right and the left eye respectively. **Conclusion:** In patients with cicatricial ectropion and dry eye secondary to ichthyosis, corneal health should be closely monitored because of the perforation risk.

Key-words: lamellar ichthyosis, cornea, spontaneous perforation, ectropion

Introduction

Ichthyosis refers to a relatively uncommon group of skin disorders characterized by presence of excessive amounts of dry surface scales. It is regarded as a disorder of keratinization or cornification, and it is due to abnormal epidermal differentiation or metabolism.

Lamellar ichthyosis is a rare, autosomal recessive, genetically heterogeneous skin disease caused by mutations involving multiple genetic loci. Lamellar ichthyosis manifests at birth often with collodion membrane and large thick scales all over the body except on the mucous membranes and the lips

(Burak et al, 2009). Cicatricial ectropion is the most common eyelid abnormality, which is assumed to lead to corneal exposure and ulceration (Cruz et al, 2000). Here we report a rare case of bilateral spontaneous corneal perforation and lower lid ectropion in a patient with lamellar ichthyosis.

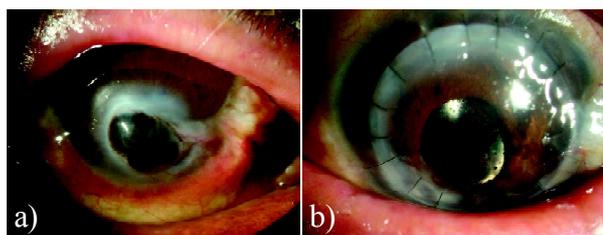
Case description

A 54-year-old male presented to us with pain, redness, foreign body sensation, watering, discharge and diminution of vision for the last one week. Visual acuities in both the eyes were perception of light. Scales, mucoid discharge over eyelid skin, cicatricial ectropion in the lower eyelids, conjunctival dryness, and congestion was noticed in both the eyes. The value of Schirmer's test II was 0 mm in both the

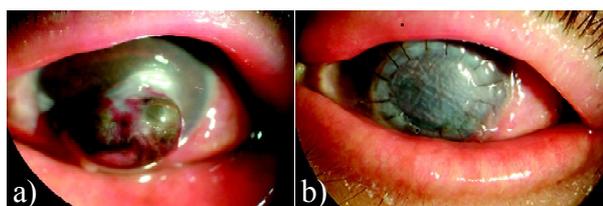
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eyes. In the center of the right cornea, a 3.0 mm x 3.5mm perforation and in the left eye a 4.0 mm x 4.5 mm corneal perforation with iris prolapse was noticed. Exudative membrane and vascularization was seen. Iris adhesion and prolapse led to irregular anterior chamber depth and the Siedel's test was found to be negative. The patient did not report any ocular trauma. On general examination, scaling and excessive dryness of the entire body skin was noticed, which according to the patient, had been present since childhood. No other family members had a significant medical history. On the basis of the family history and examination findings, a diagnosis of lamellar ichthyosis was suspected and confirmed by skin biopsy. The skin biopsy revealed orthokeratotic hyperkeratosis and epidermal thickening consistent with lamellar ichthyosis. Penetrating keratoplasty using a fresh cornea stored in MK media was used for the right eye and glycerin preserved cornea was used for left eye. Cataract surgery with PCIOL implantation was done in right eye. Ectropion correction was done in the right eye and systemic anti-ichthyosis therapy including vitamin A was started. The patient, on a follow-up visit after three months had a visual acuity of 4/60 in the right eye and is awaiting an optical keratoplasty in the left eye.

Photograph of patient showing scaling of skin and ectropion of both lower lids.



Right eye: a) Pre-operative picture (corneal perforation) b) Post-operative picture: penetrating keratoplasty



Left eye: a) Pre-operative picture (corneal perforation with iris prolapse) b) Post-operative picture: penetrating keratoplasty

Discussion

Ichthyosis is an inherited disorder characterized by thickening, fissuring, scaling and excessive cutaneous dryness of the skin. There are four different forms of ichthyosis, namely, ichthyosis vulgaris-autosomal dominant; X-linked ichthyosis; lamellar ichthyosis-autosomal recessive; and congenital ichthyosiform erythroderm (Sharma et al, 1993).

Lamellar ichthyosis is the rarest form with an incidence of 1 in 300,000. It has autosomal recessive inheritance (Ahmad et al, 2004), and is due to a defect on chromosome 14q11 encoding transglutaminase-1 (TG) (Esposito et al, 2007). In classic lamellar ichthyosis, children with the disease are referred to as collodion babies and are covered at birth by a thickened membrane that subsequently is shed. The scaling of the skin involves the whole body with no sparing of the flexural creases. Lamellar ichthyosis on histopathological examination displays massive, compact orthohyperkeratosis with variable degrees of parakeratosis and a markedly thick stratum corneum.

Different manifestations on some ocular and adnexal structures may accompany ichthyosis. Ocular manifestations include exposure keratitis secondary to ectropion, unilateral megalocornea, enlarged



corneal nerves, blepharitis, absence of the meibomian gland, trichiasis, madarosis, and absence of lacrimal puncta. Ectropion of both the upper and lower lids have been documented (Kenneth et al, 2007). Secondary corneal ulceration may occur due to exposure. Late presentation can lead to severe sight-threatening complications like descemetocele and corneal perforation.

Approximately one third of children affected with this disorder develop bilateral ectropion of the cicatricial type that appears to result from excessive dryness of the skin and subsequent contracture. The association between ichthyosis and cicatricial ectropion, which is the most common eyelid malposition, was first reported in 1834 (Jay et al, 1968), secondary to chronic exposure, corneal ulceration and perforation may occur in these patients (Eltutar et al, 1988). Cruz and colleagues postulated that corneal damage is not directly linked to lower eyelid ectropion. Apart from ectropion, the other factors causing corneal damage are madarosis; conjunctivitis, eyelash retraction, and lagophthalmus (Cruz et al, 2000). In patients with chronic corneal involvement, preservative-free artificial tears, petroleum ointment and bandage contact lenses diminish symptoms and ease epithelial healing. In the therapy of persistent corneal epithelial defect, amniotic membrane transplantation and tarsorrhaphy may promote epithelial wound healing. 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. Full-thickness skin grafting may be used to repair cicatricial ectropion (Cruz et al, 2000). Our patient underwent a full thickness keratoplasty for the corneal perforation and ectropion correction.

To our knowledge from an internet search, there is only one case that reports corneal perforation in lamellar ichthyosis (Burak et al, 2009). Our case is the first bilateral corneal perforation that was reported in a patient with lamellar ichthyosis. This is also the first case of lamellar ichthyosis ever reported from Nepal. Cicatricial ectropion, a poor

prognosis indicator for corneal health, can result in corneal perforation in ichthyosis.

Conclusion

In patients with cicatricial ectropion secondary to ichthyosis, corneal health should be closely monitored because of the perforation risk.

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