

Corneal Perforation as a Rare Ocular Manifestation in Lamellar Ichthyosis: Case Report and Literature Review

Dear Editor,

Lamellar ichthyosis (LI), an autosomal recessive congenital oculocutaneous genodermatosis, is among the most severe forms of congenital ichthyosis.^[1,2] Its clinical findings include collodion baby in the neonatal period, scaly skin lesions, and ocular abnormalities.^[1,2]

Although LI is rare, it has numerous important complications. One of the most serious complications of LI is corneal perforation, which is a worrisome sight-threatening problem and is very rarely found in patients with LI.^[1-5] Herein, we report a case and review the literature on corneal perforation as an extremely rare ocular complication in patients with LI.

A 21-year-old male with LI was referred for decreased vision over a period of 10 days. He complained of pain, foreign body sensation, burning, and redness in both eyes. On cutaneous examination, mild to moderate scaling and lamellar desquamation with excessive dryness of the whole body including the periorbital skin [Figure 1], onychodystrophy [Figure 2a], chronic paronychia of all nails, and palmoplantar keratoderma [Figure 2b] were noticed. He was under treatment with topical emollient agents, topical keratolytics (lactic acid), and systemic acitretin 10 mg/day orally for his skin lesions.

On ophthalmologic examination, visual acuities were 20/40 in the right eye and 20/200 in the left eye, respectively. Mild degree cicatricial ectropion due to facial skin tightening in the lower eyelids, thick edematous lower eyelids, medial madarosis, and conjunctival dryness were seen in both eyes [Figure 1]. On slit-lamp biomicroscopy, there was a 2-mm thinning area in the paracentral area of the left cornea that was infiltrated. In the right eye, a corneal epithelial defect and a slight thinning area were seen [Figure 3]. Goldman tonometry revealed intraocular pressures of 14 mm Hg in each eye. His pupils were reactive to the light with no afferent pupillary defect. Full funduscopy following mydriasis was normal in both eyes.

Despite aggressive therapy with emollients, ocular lubricants, vancomycin 50 mg/mL and ceftazidime 50 mg/mL ophthalmic eye drops hourly, and blepharorrhaphy, the patient developed a perforation approximately 2 mm in size in the left cornea. There was no clinical evidence of iris prolapse.

Tectonic corneal patch graft and temporary tarsorrhaphy were performed emergently. Ophthalmic anti-microbial agents and acitretin were continued. After 3 months of follow-up, he had a visual acuity of 20/30 in the right eye and 20/60 in the left eye, respectively. His ophthalmic management is currently conservative, including lubricant drops and ointments.

Although ocular complications in ichthyosis are not severe in the majority of cases and there is low ophthalmic morbidity due to LI, extremely rare and severe ocular presentations such as corneal perforation can occur.^[1,3]

LI is associated with ophthalmologic involvement including cicatricial ectropion of both upper and lower lids (most common presentation), blepharitis, dry eyes, exposure keratopathy and secondary corneal ulceration, trichiasis, and madarosis.^[1-3]



Figure 1: Lamellar ichthyosis. Lamellar desquamation and excessive dryness of the whole face with edematous and erythematous eyelids, medial madarosis, and lower lid ectropion

The reason why few patients developed corneal perforation despite sufficient treatment is unclear. We hypothesized that other factors rather than ectropion may play a role in the development of corneal perforation. Thus, we suggest corneal perforation should be considered as a differential diagnosis of visual impairment in patients with LI. It

is therefore important to manage these patients in the context of a multidisciplinary team with involvement from neonatologists, dermatologists, and ophthalmologists. Early diagnosis and treatment of ectropion and the associated exposure keratitis in LI can prevent later major ophthalmic complications such as corneal perforation and visual loss.



Figure 2: Hand involvement in lamellar ichthyosis. Onychodystrophy, large dark brown adherent scales on the dorsum of the hand (a) and palmar keratoderma (b)

Six cases of corneal perforation have been reported in patients with congenital ichthyosis in the literature; five of these cases have been reported in LI^[1-5] and one of them is seen in congenital ichthyosiform erythroderma.^[6] Of the five cases of LI, two had bilateral corneal perforation.^[2,3] Ectropion was seen in all patients [Table 1].

In severe cases of ectropion and in patients who develop exposure keratopathy with/without corneal perforation, surgical intervention is necessary. In our case, similar to Al-Amry's study^[1], we chose a tectonic patch corneal graft as a promising surgical intervention for corneal perforation in LI.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s) has/have

Table 1: Oculocutaneous characteristics of reported corneal perforation in patients with lamellar ichthyosis

Case no	Author (s)/ years	Age (years)/ Sex	Cutaneous presentation	Ocular signs, symptoms/VA	Main management of corneal perforation
1	Turgut et al. 2009 ^[5]	67/F	Lamellar ichthyosis Scaling Excessive dryness	Cicatricial ectropion Conjunctival dryness Nuclear cataract Corneal perforation V/A: light perception	Amniotic membrane transplantation Temporary tarsorrhaphy
2	Nayak et al. 2011 ^[4]	26/F	Lamellar ichthyosis Scaling Excessive dryness	Cicatricial ectropion Tarsal eversion with fornix prolapse Lagophthalmos Exposure keratopathy Corneal perforation V/A: 20/200	Oral mucous membrane graft Temporary tarsorrhaphy
3	Chaudhary et al. 2013 ^[2]	54/M	Lamellar ichthyosis Scaling Excessive dryness	Cicatricial ectropion Conjunctival dryness Nuclear cataract Bilateral spontaneous corneal perforation Corneal vascularization Iris Prolapse V/A: light perception	PKP Eyelids construction
4	Cinar et al. 2014 ^[3]	8 months/F	Lamellar ichthyosis Scaling Excessive dryness	Cicatricial ectropion Bilateral inferior paracentral cornea, descemetocele with a small perforation V/A: NA	Double-layered amniotic membrane transplantation Upper and lower eyelids construction
5	Al-Amry et al. 2016 ^[1]	1/M	Lamellar ichthyosis Collodion baby	Bilateral ectropion Corneal perforation V/A: NA	1. Bilateral ectropion repair via skin graft 2. Tectonic PKP
6	Present case. 2022	21/M	Lamellar ichthyosis Scaling Excessive dryness	Bilateral ectropion Corneal perforation V/A: 20/200	1. Tectonic patch cornea graft 2. Temporary tarsorrhaphy

M=Male, F=Female, PKP=Penetrating keratoplasty, VA=Visual acuity, NA=Data not available

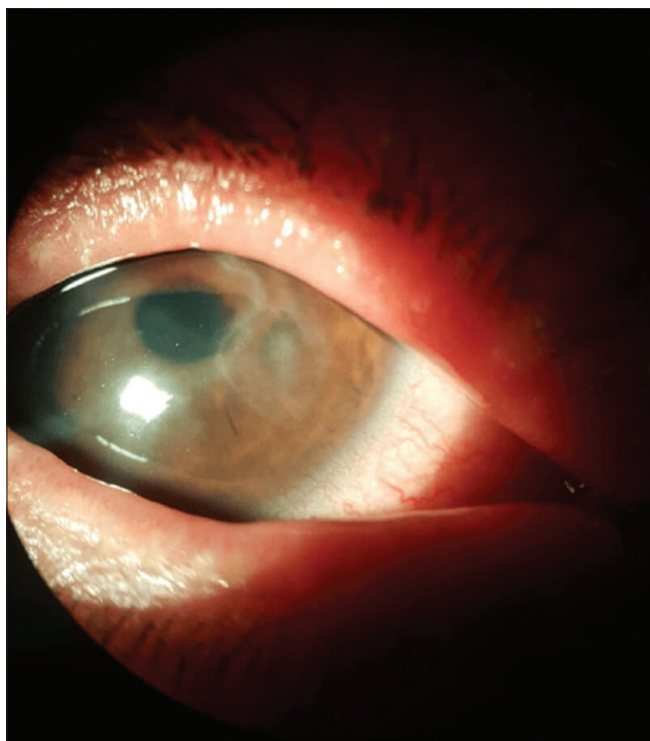


Figure 3: Corneal epithelial defect and slight thinning area in the patient's right eye

given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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