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Congenital Ectropion of the Upper Eyelids Associated with Ichthyosis: A Case Report

Sylla A[1]*, Barry MS[2], Touré A[3], Toure MS[3] and Cissé M[2]

[1]*Faculty of Health Sciences and Techniques, Guinea.*

[2]*Center for the Application of the Specialized Diploma in Ophthalmology (CADESSO), Donka University Hospital (CHU Donka), Guinea.*

[3]*Department of Dermatology, CHU Donka, Guinea.*

***Correspondence:**
Sylla A, Faculty of Health Sciences and Techniques, Guinea.

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Keywords
Congenital ectropion, upper eyelid, Congenital ichthyosis, Lamellar ichthyosis.

Introduction
Congenital ectropion of the upper eyelid is a rare condition [1].

It is defined as an outward turning (eversion) of the eyelid margin, resulting in loss of apposition between the eyelid and the ocular globe [2]. Clinically, this presentation may be associated with hereditary disorders such as Down syndrome and with dermatologic diseases such as ichthyosis [1].

The term **ichthyosis** derives from the Greek *ichthys* ("fish"), and refers to a heterogeneous group of congenital or acquired disorders characterized by **cutaneous xerosis** and **excessive desquamation** [3-5]. Clinicians distinguish ichthyoses limited to epidermal involvement from **syndromic ichthyoses**, which are associated with extracutaneous manifestations [6]. We report a case of ichthyosis associated with **bilateral ectropion of the upper eyelids**.

Case Report (Observation)
We report the case of a **4-day-old female neonate**, referred for an ocular malformation noted at birth.

History: The mother's history was notable for delivery by **cesarean section due to placental abruption**, with no need for neonatal resuscitation at birth. There was a family history of congenital malformations consistent with ichthyosis in two cousins. This was the mother's third full-term pregnancy. The father had no significant medical history.

Ophthalmologic examination: Inspection revealed **total bilateral eversion of the upper eyelids** (Figure 1). After instillation of topical anesthetic and placement of a lid speculum, the following were observed:

- Hyperemic conjunctiva, without keratinization
- Minimal yellowish purulent discharge
- Clear cornea
- Quiet, deep anterior chamber
- Round, centered pupil with a good photomotor response
- Transparent crystalline lens

Fundus examination was planned after adequate eyelid repositioning.

General examination: The neonate had generalized "fish-scale" skin involving the entire body, prompting dermatology consultation (Figure 1). The dermatology team concluded the diagnosis of **congenital ichthyosis**.

Figure 1: Bilateral ectropion of the upper eyelids with fish-scale skin.

Final diagnosis: Total bilateral congenital ectropion of the upper eyelids associated with congenital ichthyosis, based on (i)

complete eversion of the upper lids with conjunctival hyperemia and minimal purulent secretions and a normal anterior segment, and (ii) generalized ichthyosiform scaling on physical examination.

Management: Care was multidisciplinary and conservative, including:

- Ocular cleansing with **0.9% normal saline**
- **Occlusive moist compressive dressing** after manual eyelid repositioning, renewed every 24 hours for 48 hours
- Topical **rifamycin-based antibiotic ointment**
- Dermatologic care with **Neutraderm cleansing oil** and a **lipid-replenishing emollient balm** to restore skin barrier function and hydration

Outcome:

- **Day 3 post-treatment:** early eyelid repositioning was observed (Figure 2)
- **3 months post-treatment:** normal eyelid position was documented (Figure 3)
- **1-year post-treatment:** both ophthalmologic and dermatologic examinations were normal (Figure 4).

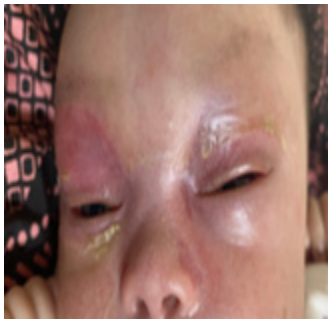

Figure 2: Post-therapeutic day 3, repositioning of the upper eyelids.

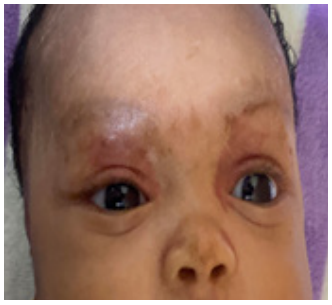

Figure 3: At 3 months, no recurrence of ectropion.

Figure 4: A 1 an, paupières normales et peau lisse.

Discussion

Ectropion is the eversion of the eyelid margin with loss of contact with the globe [2]. It can lead to significant ophthalmic complications - primarily from exposure keratopathy due to incomplete eyelid closure - potentially progressing to corneal ulceration, corneal perforation, subsequent phthisis bulbi, and poor visual prognosis [7].

Ectropion may be congenital or acquired [8]. Congenital ectropion can present at birth or within the first days of life. Proposed etiologies include infectious or inflammatory mechanisms and vertical deficiency of the anterior lamella (skin-orbicularis complex). It may occur in association with hereditary disorders (e.g., Down syndrome) and dermatologic diseases such as ichthyosis [9].

Ichthyoses represent a heterogeneous group of disorders characterized by abnormal stratum corneum formation, with hyperkeratotic cutaneous lesions that manifest as generalized scaling, with or without epidermal hyperproliferation or dermal inflammation [10]. Ichthyosis may be hereditary or acquired [11].

The four principal types commonly cited are:

- **Ichthyosis vulgaris**
- **X-linked recessive ichthyosis**
- **Lamellar ichthyosis**
- **Epidermolytic hyperkeratosis** (epidermolytic ichthyosis) [12].

Clinical severity ranges from mild forms (e.g., ichthyosis vulgaris) to severe forms (e.g., lamellar ichthyosis). Lamellar ichthyosis, historically described in the 19th century, is a rare congenital autosomal recessive disorder characterized by generalized hyperkeratinization. It is classically associated with a collodion membrane at birth that later desquamates, leaving thick, dark scales. Bilateral ectropion is frequently associated (reported in approximately 45–80% of cases), though unilateral cases have been described [7].

Management of ichthyosis requires a collaborative approach involving neonatology, dermatology, and ophthalmology [13].

In congenital lamellar ichthyosis, ectropion should be treated **very early in the neonatal period** to prevent eyelid fibrosis and to avoid complications related to meibomian gland dysfunction and corneal exposure [7].

In about half of cases, ectropion in lamellar ichthyosis may initially respond to conservative management [12]. During the neonatal stage, medical therapy typically includes:

- Frequent lubrication with artificial tears (e.g., methylcellulose or carboxymethylcellulose preparations)
- Gentle eyelid massage several times daily
- Ocular protection with moist compresses

Once cicatricial eyelid fibrosis is established, surgical correction

becomes necessary [7].

In our case, likely due to early intervention, no complications were observed. Treatment was conservative with artificial tears, an antibiotic–corticosteroid ointment, and occlusive moist compressive dressing, alongside dermatologic barrier-repair therapy (cleansing oil and lipid-replenishing emollients).

Conclusion

Early multidisciplinary collaboration and prompt conservative management are crucial to achieve rapid eyelid repositioning, prevent corneal exposure complications, and ensure effective skin hydration and barrier support, thereby improving overall quality of life.

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