Severe Bilateral Ectropion in Lamellar Ichthyosis: A Case Report

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Patient: Female, newborn
Final Diagnosis: Ectropion in lamellar ichthyosis
Symptoms: Entropion • parchment-like scales • dry eyes
Medication: —
Clinical Procedure: Ophthalmological and dermatological follow-up
Specialty: Ophthalmology

Objective: Rare disease
Background: Lamellar ichthyosis is a rare type of hereditary ichthyosis disease that is responsible for generalized dry skin and severe scaling. Congenital ectropion and eclabium are often associated with this condition. The ectropion can cause many ophthalmologic complications, mostly due to corneal exposure caused by the lack of eyelid cleft closure. The corneal exposure can cause ulceration and even corneal perforation leading to phthisis bulbi.

Case Report: We report the case of a North African newborn baby diagnosed with congenital lamellar ichthyosis responsible for severe skin scaling, eclabium, and bilateral ectropion of both the upper and lower eyelids. The therapeutic care of the baby was ensured by a collaboration between ophthalmologist and dermatologist. The newborn was treated by oral acitretin and skin emollients for his dermatological conditions and on the ophthalmological level he was put on lubricating and moisturizing eye drops and gel, associated with daily repeated eyelid massage. The evolution after 6 months of treatment showed excellent results on both the dermatological and the ophthalmological level.

Conclusions: At the stage of cicatricial ectropion in lamellar ichthyosis, the management is mainly based on a chirurgical approach. However, if the ectropion is managed early enough, the treatment could be medical, which is much easier, produces very good results, and prevents the progression toward eyelid fibrosis and its associated complications that can make the management of the ectropion much harder.

Keywords: Collodion • Ectropion • Ichthyosis Exfoliativa • Lamellar Ichthyosis, Type 2

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Background

Hereditary ichthyosis is a group of heterogeneous genodermatoses clinically characterized by dry, thickened, scaly skin that is often associated with inflammation. Many types are described, in which the clinical presentation can vary from mild forms such as ichthyosis vulgaris to severe ones like lamellar ichthyosis. The lamellar form is characterized by a collodion-like membrane encasing the baby at birth, which desquamates afterwards, leaving thick dark scales all over the body; it is generally associated with an eclabium and a congenital ectropion (45% to 80% of cases) [1]. The latter is responsible for corneal exposure that can lead to a poor visual prognosis.

We report the case of a newborn baby treated for a congenital lamellar ichthyosis responsible for severe bilateral ectropion, which regressed very well following rigorous ophthalmological and dermatological medical management.

Case Report

This is a case of a 20-day old North African newborn baby from Morocco, premature at 7 months, vaginally delivered, descendant of a non-consanguineous marriage, and sibling of 2 sisters. The newborn was referred by his pediatrician for a dermo-ophthalmological check-up in front of a collodion baby phenotype.

The physical examination found a clinical aspect in favor of congenital lamellar ichthyosis; the new born was covered with parchment-like scales that had started to detach all over his body, giving an appearance of a “collodion baby”. In addition, the baby presented an eclabium and scaling of the scalp without alopecia. However, the palms, soles, and nails were not affected by the disease.

Ophthalmologic examination noted the presence of a major bilateral ectropion of the upper and lower eyelids with tarsal eversion, madarosis of the lower eyelids, and diffuse conjunctival hyperemia (Figure 1). The biomicroscopic examination with the slit lamp of the anterior and posterior segments was normal, with a clear cornea and a negative fluorescence test.

Faced with severe skin damage, the dermatologists started treatment based on oral acitretin (Neotigason [10 mg]: retinoid derived from vit A with keratolytic action) at a dose of 0.5 mg/kg/day and skin emollients (hydrating cream based on Niacinamide, Vitreoscilla Ferment, Colza oil, and butter shea) to hydrate and soften the skin.

On the ophthalmological level, the patient received topical treatment based on lubricating and moisturizing eye drops (1.8/1 ml mg of sodium hyaluronate, sodium chloride, potassium chloride, and calcium chloride) applied hourly during the daytime and carbomer eye gel (30 mg of carbomer in 10 g) applied at

Figure 1. Newborn on admission: Parchment-like scales that detach from all of his body with a “collodion baby” morphotype. Note the major bilateral ectropion.
night-time, associated with antiseptic eye drops (Hexamidine 0.1%) 3 times a day. The mother was advised to regularly and gently massage the eyelids to prevent fibrosis.

The control examination 2 months later noted a significant reduction in skin scales with persistence of the ectropion to a lesser extent (Figure 2). The presence of purulent secretions in both eyes without keratitis was also noted. At this time, topical antibiotic (Tobramycin 0.3%) combined with repeated eye washes throughout the day with physiological serum (sodium chloride solution 0.9%) were prescribed.

At 4-month follow-up, we noticed great dermatological and ophthalmological improvement, with significant decrease of scaling and ectropion (Figure 3). Treatment with skin emollients and eye lubricants, as well as eyelid massages, were advised to be maintained.

After 6 months of treatment, the ectropion had completely resolved, with the appearance of the upper eyelid crease (Figure 4). Overall, there was also great improvement on the dermatological level by the resolution of the eclairum and remarkable reduction of the desquamation.
Discussion

The word “Ichthyosis” is derived from the Greek word “Ikthus”, which means fish. Ichthyosiform dermatoses are rare skin conditions characterized by defective skin desquamation [2].

Lamellar ichthyosis, originally described by Seelingman in 1841, is a rare, autosomal, recessively inherited, congenital disorder characterized by generalized hyper-keratinization of the epidermis. Clinically, the newborn is encapsulated in a shiny membrane, called “collodion baby” [3,4]. Bilateral ectropion is often associated (in 45% to 80% of cases) [1]. Some cases of unilateral ectropion have been described [5]. The ectropion can be isolated or it can be associated with lagophthalmos or exposure keratitis. The risk of exposure keratitis increases if a meibomian gland dysfunction is present. The meibomian glands are ectodermal in origin, just like the skin, and are likely to be affected in lamellar ichthyosis [6].

Early, at the neonatal stage, the management of ectropion is medical. The treatment is based on an hourly instillation of lubricants (methyl cellulose, carboxymethylcellulose), associated with a gentle palpebral massage several times a day and protection of the eyes with moist compresses [7]. This was also the local treatment used for our patient. Gupta et al also describes the benefit of using a topical anti-inflammatory agent (clobetasol 0.05%) twice a day. This protocol allows resolution of ectropion in 2-3 months.

In addition to local treatments, Singh et al used oral retinoids (acitretin) under hematological monitoring associated with a high-calorie diet and daily baths. Clinical improvement was noted at the end of the second week [3]. Gicquel et al used a combination of oral acitretin with topical N-acetylcysteine, known for its anti-proliferative action, as a skin preparation (10% water-in-oil emulsion) applied twice a day [8]. In our patient, oral acitretin showed very good ophthalmological and dermatological results, with progressive improvement in the phenotype of the baby.

It should be noted that the use of oral acitretin in children with ichthyosis can be highly effective in reducing scaling and improving both function and appearance. However, the long-term use of systemic retinoids has known adverse effects involving the bones and eyes. Additionally, potential psychiatric and cardiovascular effects need to be considered [9].

At the cicatricial stage, ectropion management is surgical. The goal is to release the ectropion through an incision along the eyelid, followed by a skin graft that will cover the area of the incision and maintain eyelid relaxation. Several skin grafts harvesting sites have been described (groin fold, retro-auricular region); the rule is to find a flexible and above all healthy skin area within this dehydrated skin covered with scales [10,11].

Conclusions

The management of ectropion in congenital lamellar ichthyosis should be undertaken very early in the neonatal period, immediately after birth, in order to prevent palpebral fibrosis and avoid complications related to meibomian dysfunction and corneal exposure. At this stage, medical treatment with a gentle palpebral massage can be effective. However, later, when the palpebral fibrosis is established, surgery is necessary to treat the cicatricial ectropion.

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References: