CASE REPORT

Lamellar ichthyosis with severe bilateral ectropion and self-healing collodion membrane

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Abstract  Congenital ichthyosis is a type of generalized hyperkeratinization of the skin at birth. Three forms of congenital ichthyosis have been defined based on clinical aspects and severity: (1) congenital ichthyosis mitis; (2) congenital ichthyosis tarda; and (3) congenital ichthyosis gravis. Desquamation of the parchment-like hyperkeratinized skin begins shortly after birth and may require several weeks to complete. Skin alterations in the eyelid cause shortening of the anterior lamella, subsequently resulting in ectropion. This affects the upper eyelid more often than the lower eyelid and can lead to complications such as chronic palpebral or bulbar conjunctivitis and keratinization or exposure keratopathy. In this paper, we present two case reports illustrating the course of ichthyosis congenita mitis and ichthyosis congenita tarda.

Introduction  The word “ichthyosis” is derived from the Greek word  ikthus, meaning “fish.” Ichthyosiform dermatoses are a group of hereditary disorders characterized by dryness and roughness of the skin with excessive accumulation of
epidermal scales. Lamellar ichthyosis is a rare congenital ichthyosiform dermatosis.1 The characteristic feature of the disease is a thin, dry, shining, brown-yellow, parchment-like membrane that completely envelops a newborn. This gives a collodion (i.e., "backed apple") appearance to the newborn; such children are called "collodion babies." This condition was first described by Seelingman in 1841. Ballantyne was the first author to make an extensive report on 33 cases.2 Since the Ballantyne study, approximately 300 cases have been described.3 We report two cases of lamellar ichthyosis with bilateral ectropion of the eyelids and generalized involvement of the body surface by a membrane.

Case report

Patient 1 (ichthyosis congenita mitis)

A male baby, born at 39² weeks of gestation, was delivered by vaginal delivery. His entire body was covered by a parchment-like hyperkeratinized skin. Both eyes showed ectropion of the upper and the lower eyelids, which was more obvious with enforced lid closure. Frequent application of external ointment and spontaneous desquamation led to the resolution of the ectropion.

Patient 1 could suck and cry. While being observed, the membrane cracked at the neck and groin within a few hours of birth. Treatment was initiated with frequent light massage of the entire body surface with white soft paraffin. Methylcellulose was frequently instilled in each eye to prevent exposure keratitis. A wet saline gauze was lightly placed over the face, thereby covering the eyes totally. Patient 1 was fed through a nasogastric tube. Approximately 2 days after birth, Patient 1 could move his mouth sufficiently and was able to suck and cry. He was able to close his eyes adequately after 2 days. The ectropion totally disappeared within 1 week (Fig. 3). In Patient 1, a biopsy of skin from the left groin (obtained approximately 24 hours after birth) showed thickening of the stratum corneum and stratum granulosum containing perivascular lymphocytic infiltration and the orifices of hair follicles by keratic precipitation. History revealed a non-consanguineous marriage of the parents.

Patient 2 (ichthyosis congenita tarda)

A male baby, born prematurely at 35³ weeks of gestation, was delivered by cesarean section. At birth the child showed signs of being a collodion baby with ectropion of all four eyelids, the characteristic "fish mouth," and rudimentary external ears. The child survived by fluid supplementation administered intravenously and by the topical application of an external ointment. Spontaneous desquamation led to resolution of the ectropion.

Both babies were covered all over their bodies with a brown, dry, shining membrane. Because of the tautness of the membrane, the baby's eyes remained open with marked ectropion of the lids of both eyes and the mouth was fixed in an open position (Figs. 1 and 2).

Patient 2 was unable to suck or cry. While being observed, the membrane cracked at the neck and groin within a few hours of birth. Treatment was initiated with frequent light massage of the entire body surface with white soft paraffin. Methylcellulose was frequently instilled in each eye to prevent exposure keratitis. A wet saline gauze was lightly placed over the face, thereby covering the eyes totally. Patient 2 was fed through a nasogastric tube. Approximately 5 days after birth, Patient 2 could move his mouth sufficiently and was able to suck and cry. He was able to close his eyes adequately after 1 week and ectropion totally disappeared in 2 weeks. History revealed
a nonconsanguineous marriage of the parents. Massage with white soft paraffin continued to be applied. After approximately 3 months, the membrane disappeared from his body surface, except over the forehead where several large dry scales persisted.

Discussion

Lamellar ichthyosis is an inherited autosomal recessive disorder. Eight percent of these patients have a history of consanguinity between the parents, although this was not a feature in our infants. The disease is reportedly two times more common in males; our patients were male infants. Twenty-five percent of these children are born prematurely and 51% of the children have similarly affected siblings. Patient 1 was born at full term, but Patient 2 was born prematurely. Some patients have the disease limited to the trunk or to one or two extremities, although 80% of patients have a generalized involvement. The flexor aspects of the body are the most severely affected. The other reported associations of the disease are bilateral ectropion (33%), diminished or absent sweating (10%), nail dystrophies (<5%) and seasonal recurrence of the dermatosis in summer (15%). These children are extra-susceptible to systemic infection. Alternate formation and shedding of scales on the skin has been described from time to time. The histopathology of the disease has been discussed. Other findings are marked hyperkeratosis, occasionally a normal to thickened granular layer on prominent rete ridges, follicular orifices that are filled with keratin, and smaller-than-normal hair follicles and sebaceous glands.

There is universal agreement regarding the management of these patients by using plain ointments, keratolytic preparations, and (in severe cases) systemic and topical corticosteroids. In mild forms of congenital ichthyosis, surgical treatment of the eyelid ectropion is not required. In more severe cases, a skin graft may be necessary. Various but limited sources of graft material, which are discussed, can be considered.

The present cases illustrate the severity of ectropion of the eyelids and highlight the importance of proper treatment to prevent exposure keratopathy. Skin biopsies from our patients revealed only hyperkeratosis. Both patients had to be managed with topical emollients because the family could not afford oral retinoids.

At present, there is no curative therapy for ichthyosis and some scientists may argue that current therapies are moderately effective and involve significant risks of side effects. Cutaneous gene therapy based on retransplanted cells is a complex procedure if large areas of skin need to be treated.

Lamellar ichthyosis is a genetically heterogeneous group of keratinization disorders that is inherited in an autosomal recessive fashion. Lamellar ichthyosis has been studied as a prototype for therapeutic cutaneous gene delivery. Mutations in the transglutaminase-1 (TGM1) gene on chromosome 14q11 account for approximately one-half of the cases of lamellar ichthyosis and a few cases of nonbullous congenital ichthyosis-forms erythroderma. The TGM1 mutations are heterogeneous (including point mutations, deletions, truncations, and splice-site mutations); however, consistent genotype-phenotype correlations have not been observed. A lack of transglutaminase-1 activity impairs the cross-linking of proteins and lipids in the cornified cell envelope of the upper epidermis and leads to defective cornification and desquamation. Restoration of transglutaminase activity in keratinocytes from lamellar ichthyosis patients has normalized protein cross-linking and cornification in vitro and in a human skin—immunodeficient mouse xenograft model.

References