CASE REPORT

Bilateral Severe Ectropion and Mature Cataract in Lamellar Ichthyosis

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Abstract

Lamellar ichthyosis is a rare, autosomal recessive, genetically heterogeneous skin disorder caused by mutations in the transglutaminase-1 gene. Eye abnormalities include bilateral ectropion of lower lids, chronic blepharitis and rarely cataract. A case of lamellar ichthyosis with bilateral lower lid ectropion and bilateral mature cataract is hereby presented for its rarity.

Key words

Lamellar ichthyosis, Ectropion, Cataract.

Introduction

Lamellar ichthyosis is a rare type of ichthyosis manifesting at birth often encased in collodion membrane and large thick scales all over the body, except mucous membrane and lips. Bilateral ectropion is the main ocular involvement in lamellar ichthyosis. Though nuclear cataract has been reported earlier in lamellar ichthyosis, we report a case of lamellar ichthyosis with bilateral lower lid ectropion and bilateral mature cataract.

Case report

A 3 year old male boy presented in the eye OPD with scaling of the skin, out-turning of lower eyelids and inability to see with both eyes. As per the parents history, at birth, the patient's skin was covered with a shiny membrane all over the body, which desquamated and lead to peeling with scales. One week after birth, ectropion of lower eyelids was observed. Loss of vision was observed at about 1 year of age with the presence of a white pupillary reflex.

On general examination, the patient was malnourished with a weight of 9 Kg, covered with tough dry membrane with desquamation and scales. The child followed light and responded to sound stimuli briskly. Mild pyoderma of the scalp was present. Ocular examination revealed absence of the eyebrows, presence of scales over lid skin and eyelashes and grade III ectropion in lower eyelids. Conjunctiva was congested and dry. There was a mature cataract in both eyes. On the basis of history and examination, diagnosis of collodion baby developing into lamellar ichthyosis was made. The diagnosis was confirmed by skin biopsy. The patient was put on intensive anti-ichthyosis therapy in the form of frequent massage with vaseline, glycerine, emollients and keratolytics. Cephalexin, vitamin B complex and vitamin A was given in appropriate dosages. Ocular treatment included hourly instillation of ciprofloxacin 0.3% and methylcellulose 0.5% eyedrops and ciprofloxacin ointment at bed time. The conjunctival congestion subsided and the general condition improved.

The patient was operated upon for cataract and lens aspiration with posterior chamber intraocular lens

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implantation in right eye was done under general anesthesia. There was no intra-operative complication. Post-operatively, patient was put on topical antibiotics, cycloplegics and tear substitutes. Post-operative follow-up for a period of 1 year was uneventful.

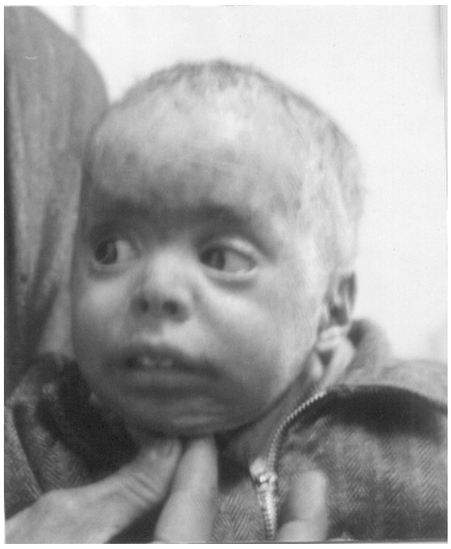

Fig. 1. Photograph showing lamellar ichthyosis with bilateral lower lid ectropion & mature cataract.

Discussion

Collodion baby is a term given to a baby covered with a shiny membrane all over the body except mucous membrane and lips, which gradually desquamates with large thick scales and mostly develops into lamellar ichthyosis. Ichthyosis is a skin disorder characterized by excessive dryness of skin and increased formation of epidermal scales. The four main types of ichthyosis are ichthyosis vulgaris, sex linked recessive, lamellar ichthyosis and epdermolytic hyperkeratosis. Lamellar ichthyosis is the rarest form with an incidence of less than 1 in 3 lacs (1). It has autosomal recessive inheritance and there is a defect on chromosome 14q11 causing transglutaminase-1 (TG) defect (2). TG mutations might adversely affect the formation of cross links essential to formation of cornified cell envelops and normal stratum corneum layer of the skin (2,3). Ocular manifestations of ichthyosis vary according to the type of ichthyosis (4). Scales on eyelashes and eyelids may be seen in all varieties. However, the tight collodion membrane covering the newborn and producing ectropion of lids is characteristically found in lamellar ichthyosis.

The ectropion may respond to the conservative management in the early periods of life in about half of the cases, while in rest of the cases, excessive ichthyosis may lead to severe cicatricial ectropion as in our case. Few cases of congenital ichthyosis especially the X-linked type have been reported as having congenital cataracts. However, in our case, the patient presented with bilateral mature cataract with accurate perception and projection of light .

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