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Original Article

Ocular manifestation of Ichthyosis

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Abstract

Purpose: Ichthyosis is a rare dermato-ocular disease. This study evaluates the presenting ocular signs, symptoms, complications and prognosis of ichthyosis in a case series from Saudi Arabia.
Methods: A retrospective chart review was performed for 11 patients with ichthyosis who presented to King Khaled Eye Specialist Hospital, Riyadh, Saudi Arabia, over the last 20 years.
Results: The most common presenting ocular diagnosis was ectropion of both the lids. Two patients developed corneal perforation with poor prognosis. Most of the patients underwent skin grafting to repair eyelid ectropion. The visual prognosis was excellent because timely surgical interventions were performed. Hence the rate of corneal complications such as perforation was low.
Conclusion: The most ocular presentation of ichthyosis is ectropion of both the upper and lower lids. Despite good visual prognosis, there were some devastating corneal complications such as perforation with unpredictable outcomes.

Keywords: Ichthyosis, Ectropion, Congenital, Skin graft, Collodion

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Introduction

Ichthyosis is an inherited group of skin disorders characterized by the skin thickening and scale formation. Some of these disorders include, ichthyosis vulgaris and epidermolytic hyperkeratosis (EHK) which are autosomal dominant and lamellar type which is autosomal recessive and X-linked ichthyosis.[1] Harlequin ichthyosis is an inherited autosomal recessive disorder with mutations in the abca12 gene. It is fatal and is characterized by deeply fissured skin and deformities of the hands and feet. Harlequin ichthyosis may be accompanied with lipid dysfunctions of the epidermal layer of the skin, starting prenatally, and has many names such as alligator baby and malignant keratosis.[2] KID syndrome stands for keratitis, ichthyosis and sensorineural deafness. It manifests itself as alopecia, dental disorders, susceptibility to bacterial and mycotic infections and squamous cell carcinoma.[3,4] Ichthyosis with confetti is a very rare type of ichthyosis characterized by dermatological features of collodion baby mixed with patches of confetti-like healthy skin.[5]

The most common ocular manifestation of ichthyosis is cicatricial ectropion. We present the ocular signs, symptoms, complications and prognosis of ichthyosis in a case series from Saudi Arabia.

Patients and methods

The medical records were reviewed for all patients diagnosed with ichthyosis who presented to King Khaled Eye Specialist Hospital, Riyadh, Saudi Arabia. This hospital is referral based eye care hospital that treats patients nationwide and from the Gulf region. A retrospective chart review was performed for cases of ichthyosis from 1994–2014. Data were collected on patient demographics, age at presentation, type

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of ichthyosis, the type of referral to the hospital and if the patient was treated by a local dermatologist. Other data were collected on skin and ocular presenting signs including lid ectropion and corneal complications, the medical and surgical treatment as well as the type of the surgical procedure and the incidence of repeat lid correcting procedures such as skin grafting.

Patients were excluded if they underwent surgical repair of the lids and/or cornea that was not due to ichthyosis or if the presenting or follow-up data were insufficient.

Patients (pediatric or adults) were included if they had documented or suspected ichthyosis and documented or suspected cases of congenital lid ectropion, corneal stem cell deficiency and/or skin manifestations of ichthyosis and any cases of ichthyosis with documented histopathology studies.

The study was registered with the institutional review board and approval was obtained from the ethics committee.

Results

Eleven out of 16 patients met the inclusion criteria. Four patients were excluded because there were no physical medical records located to retrieve the data, and 1 patient had a diagnosis of orbital inflammatory syndrome not ichthyosis.

All 11 patients were referred to the hospital with a straightforward diagnosis of lid ectropion related to ichthyosis (Fig. 1A and B) and exposure keratopathy (Fig. 2A and B, Table 1). Eight (72%) patients had documented ichthyosis by a dermatologist, a neonatologist or an ophthalmologist. Mean age of the 11 cases was 17.4 years (range, 1.6–38 years). Mean age at presentation was 8.9 years (range, 1.6–32 years). Follow-up ranged from 3 months to 9 years.

At presentation, best corrected visual acuity (BCVA) ranged from 20/20 to 20/25 in 6 out of 11 patients (54%) (Table 1). Two (18%) patients had vision ranging from 20/200 to count fingers secondary to corneal scarring. Three (27%) patients (all children) had fix and follow vision.

Lamellar ichthyosis was the most common type in 4 patients (36%) cases (Fig. 3, Table 1). Four (36%) patients had a positive family history for siblings with ichthyosis.

All cases (100%) presented with ectropion (Table 1, Fig. 1). All patients were treated for dry eye and exposure keratopathy with different topical lubricating drops and ointment. The cornea perforated in one patient that warranted tectonic penetrating keratoplasty. The same patient developed microbial keratitis on the graft surface. Eight (72%) eyes underwent skin grafting for lid ectropion. Of the patients who did not undergo skin grafting, 1 patient had no healthy skin and was referred to a dermatologist, 1 patient was referred to a dermatologist for further skin treatment and we could not determine why the patient did not undergo skin grafting.

Three of 8 patients were recommended for skin re-grafting but only 1 underwent the procedure. One patient was sick and was lost to follow-up and the other patient's family elected not to consent to any further surgical intervention.

The histopathology studies were performed on excised lid skin samples during skin grafting. The results were generally consistent with the classic dermatological changes confirming the disease. Histopathologic features were, hyperkeratosis, parakeratosis and/or keratotic plugging and in some cases, psoriatic epidermal hyperplasia and chronic inflammatory reaction of the dermis (Fig. 4A and B).

Discussion

In this case series of ichthyosis, we found lamellar ichthyosis was the most common, with ectropion of both the upper and lower eyelids and no conjunctival involvement. We found the ocular complications from ichthyosis were not severe in the majority of cases. This observation concurs with previous reports of low ophthalmic morbidity due to ichthyosis.[4] However, severe complications such as corneal perforation did occur in our case series. For example, there was one patient

A B

Figure 1. Ectropion of the upper and the lower eyelids.