

Original Research Article

Ophthalmological manifestations in ichthyosis: from eyelid involvement to corneal complications

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Abstract

Ichthyosis is a rare disorder of keratinization with frequent but underrecognized ocular involvement. Eyelid abnormalities and ocular-surface disease may lead to severe corneal complications, especially in congenital forms. Objective: To describe ophthalmological manifestations in patients with ichthyosis and to identify factors associated with corneal complications. Methods: Retrospective observational analytical study of eight patients with clinically confirmed ichthyosis seen in an ophthalmology consultation. Data included demographics, ichthyosis type, eyelid examination, slit-lamp findings, dry-eye assessment, treatments, and outcomes. Results are presented as counts and percentages; associations between eyelid abnormalities and corneal involvement were explored descriptively. Results: Mean age was 12.5 years (range 6 months–32 years); five patients (62.5%) were male. Severe congenital forms represented 62.5% of cases. Eyelid involvement occurred in 6/8 patients (75%): lower eyelid ectropion in 5 (62.5%), bilateral in 4 (50%); lagophthalmos in 3 (37.5%); chronic blepharitis in 4 (50%). Dry eye affected all patients (100%): mild–moderate in 5 (62.5%), severe in 3 (37.5%), with severe dry eye consistently associated with ectropion. Ocular-surface disease was present in 5 patients (62.5%): chronic conjunctival hyperemia (50%), superficial punctate keratopathy (50%), exposure keratitis (37.5%), and corneal ulceration (12.5%). Corneal complications were more frequent in patients with lagophthalmos (66.7%) than in those without eyelid abnormalities (20%). Visual acuity decrease due to corneal damage occurred in 2 patients (25%); one child was at risk for amblyopia. Six patients (75%) received medical treatment alone; two (25%) underwent surgical correction of ectropion. Intensive lubrication was prescribed for all. Clinical improvement was observed in 7/8 patients (87.5%). Conclusions: Ophthalmic manifestations in ichthyosis are common and can be severe. Eyelid malposition, especially ectropion and lagophthalmos, is the main risk factor for corneal complications. Early ophthalmic assessment, aggressive ocular-surface management, and timely surgical repair of eyelid malposition are essential to preserve vision, particularly in congenital forms.

Keywords: *ichthyosis, ectropion, lagophthalmos, corneal ulceration, dry eye, ocular surface.*

Introduction

Ichthyosis is a rare keratinization disorder. Ophthalmological manifestations, often underestimated, are mainly related to eyelid malposition and ocular-surface involvement and can lead to serious corneal complications, particularly in congenital forms. This study aimed to analyze ophthalmological manifestations in patients with ichthyosis and to identify factors associated with corneal complications.

Materials and methods

We performed a retrospective observational analytical study of patients with clinically confirmed ichthyosis seen in ophthalmology consultation. Eight patients were included. Collected data: age, sex, ichthyosis type (congenital vs. acquired/severity), eyelid static and dynamic assessment, slit-lamp examination, dry-eye evaluation, treatments, and outcomes. Data are presented as counts and percentages; comparative description assessed associations between eyelid abnormalities and corneal involvement.

Results

Demographics: mean age 12.5 years (6 months–32 years); 5 males (62.5%). Severe congenital forms: 62.5%. Eyelid lesions Eyelid involvement: 6 patients (75%).

- Lower eyelid ectropion: 5 patients (62.5%), bilateral in 4 (50%).
- Lagophthalmos: 3 patients (37.5%).
- Chronic blepharitis: 4 patients (50%). Eyelid abnormalities were more frequent in severe congenital forms (83.3%).

Dry eye All patients (100%) had dry eye: mild–moderate in 5 (62.5%); severe in 3 (37.5%). Severe dry eye was consistently associated with ectropion.

Conjunctival and corneal disorders Ocular-surface involvement in 5 patients (62.5%):

- Chronic conjunctival hyperemia: 50%
- Superficial punctate keratopathy: 50%
- Exposure keratitis: 37.5%
- Corneal ulceration: 12.5% Corneal complications were more frequent with lagophthalmos (66.7%) than without eyelid abnormalities (20%).

Functional impact and treatment Visual acuity decreased due to corneal damage in 2 patients (25%); one child at risk of amblyopia. Conservative treatment alone was used in 6 patients (75%); 2 patients (25%) underwent surgical ectropion repair. Intensive lubrication prescribed for all; clinical improvement in 87.5%

Discussion

This series confirms frequent ocular involvement in ichthyosis, dominated by eyelid malposition and dry eye. Ectropion and lagophthalmos are major risk factors for exposure-related corneal disease. Severe congenital forms appear at higher risk, highlighting the need for early ophthalmological follow-up in children. Limitations include small sample size and retrospective design, but the rarity of the disease gives clinical value to the findings.

Conclusions

Ocular manifestations in ichthyosis are common and potentially sight-threatening. Early detection, aggressive ocular-surface therapy, and timely surgical correction of eyelid malposition are crucial to prevent corneal complications and preserve vision.

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