External Ocular Manifestations in Autosomal Dominant Dystrophic Epidermolysis Bullosa; a Case Report

Manizheh Mahdavi, MD; Mohammad-Ali Javadi, MD

Shaheed Beheshti Medical University, Tehran, Iran

Purpose: To present a case of autosomal dominant dystrophic epidermolysis bullosa with symblepharon formation due to eye rubbing.

Case Report: A 10-year-old girl suffering from blistering and ulcerative lesions of the trunk and palms and dystrophic nails since childhood was referred to our clinic with a symblepharon connecting the medial portion of the right upper lid to the superonasal quadrant of the cornea. The central cornea in both eyes exhibited mild subepithelial opacification. She had history of eye rubbing due to foreign body sensation in the right eye, resulting in red eye and blister-like conjunctival lesions since three years ago. She had previously undergone surgical symblepharon removal leading to more severe recurrence of the condition.

Conclusion: Dominant dystrophic epidermolysis bullosa may be accompanied by external ocular manifestations. Protection of the eye from minor trauma such as rubbing may help prevent ocular complications.

J Ophthalmic Vis Res 2008; 3 (1): 70-73.

Correspondence to: Mohammad-Ali Javadi, MD. Professor of Ophthalmology; Ophthalmic Research Center, No. 5, Boostan 9 St., Amir Ebrahimi Ave., Pasdaran, Tehran 16666, Iran; Tel: +98 21 22585952, Fax: +98 21 22590607; e-mail: ma_ javadi@yahoo.com

INTRODUCTION

Epidermolysis bullosa, an autoimmune mucocutaneous disorder, is characterized by bullae formation in response to minor trauma to the skin and mucous membranes. This disorder presents in hereditary and acquired forms. The hereditary forms include epidermolysis bullosa simplex (autosomal dominant), junctional epidermolysis bullosa (autosomal dominant) and dystrophic epidermolysis bullosa (DEB, autosomal dominant or recessive). DEB is characterized by mucocutaneous blister formation following minor mechanical trauma, which results in dermal scarring and nail dystrophy. Presence of subepidermal blisters beneath the basal lamina is characteristic for the dystrophic subtype.[1]

Genetic studies have disclosed mutations in COL7A1 in DEB (both dominant and recessive types). This gene is responsible for synthesis of collagen type VII which is the coordinating agent for tissue collagen fibrils; therefore, in the presence of these mutations, collagen fibrils are disrupted.[2]

The dominant and recessive forms of DEB are thoroughly different in terms of clinical manifestations and severity. The dominant type of DEB (DDEB) usually presents at birth or during infancy or early childhood with skin blistering,

dermal scarring and milia formation. Dystrophic nails are a common presentation in the dominant form, which may exhibit as alterations in the shape of the nail or even absence of the nail altogether. However, the teeth are typically normal and the oral mucosa is rarely involved in this subtype. Recessive type of DEB (RDEB) commonly manifests at birth and comprises a broader range of clinical manifestations as compared to the dominant form. Extensive dermal blisters on the extremities, knees and elbows result in joint deformity and scarring around the fingers leading to polysyndactyly, can be observed in the recessive form. Furthermore, mucocutaneous scars in the recessive form are common places for developing squamous cell carcinoma. Oral mucosal involvement is frequently seen in this form resulting in microstomia and ankyloglossia in some cases, moreover, tooth decay is often noted. Gastrointestinal strictures cause problems in eating and swallowing.[3]

Ocular manifestations of epidermolysis bullosa include corneal ulceration, symblepharon formation, blepharitis, ectropion, nasolacrimal duct obstruction, disorders of vision and even blindness in some cases.[4] Herein we report a case of DDEB who developed a symblepharon following eye rubbing.

CASE REPORT

A 10-year-old girl was referred to Labbafinejad Medical Center with redness and bullous conjunctival lesions in her right eye. She had history of bullous and ulcerated itchy lesions on her trunk and palms since the age of two. The patient had also developed erythema and a bullous-like lesion in her right eye resulting in symblepharon formation and obliteration of the upper fornix following eye rubbing. She had previously twice undergone surgery to remove the symblepharon, however, each time it shortly relapsed with more severity.

On general examination, active and chronic bullous lesions were observed on the trunk and palms, which had led to scarring in some areas (figures 1 & 2). She had dystrophic nails and absence of nails in some fingers (Fig. 3). There were also a number of milia on the back of her hands and hypertrophic scars on the upper and lower extremities (Fig. 4). The oral mucosa was normal and the patient did not have any swallowing complaints or other gastrointestinal disorders.

On ophthalmic examination she had best-corrected visual acuity of 20/50 with +3.0-3.0 @140° in the right eye and uncorrected vision of 20/20 in the left. In the right eye, a symblepharon connected the medial portion of the upper lid to the upper nasal quadrant of the cornea resulting in limitation of eye movement especially downward and to the right (Fig. 5). Slitlamp biomicroscopy revealed mild subepithelial opacification in both corneas. The rest of the ophthalmic examination was unremarkable.

DISCUSSION

Dystrophic epidermolysis bullosa is a skin disorder resulting in dissociation of its superficial layers. This disorder is the most severe type of epidermolysis bullosa.[3] In a study of 204 patients with different types of epidermolysis bullosa, the most severe ocular manifestations were associated with RDEB such that out of 40 cases of RDEB, 17 suffered from corneal scarring and 6 had symblepharon formation.[5] Tong et al[6] reported that RDEB is associated with ocular manifestations including corneal abrasions, corneal pannus and scarring, exposure keratitis secondary to lower lid ectropion, bullous lesions of the lids and complications involving the conjunctiva. Adhesions of the eyelid to the cornea have been reported to severely reduce visual acuity to as low as hand motions.[7]

The patient presented herein was a case of DDEB who developed conjunctival lesions and symblepharon formation following foreign body sensation and eye rubbing in whom the lesion had recurred with more severity following symblepharon removal. Altan et al[8] reported symblepharon removal surgery and lamellar keratectomy in a case of RDEB. They used

amniotic membrane to cover the cornea and fornices and did not encounter any relapse up to 22 months. Abnormal structure of collagen type VII in patients suffering from DEB results in deficient tissue repair leading to scar and fibrous formation[2], this can explain the relapse of symblepharon of more severe nature following excision. Use of protective glasses and avoiding minor eye trauma such as rubbing can be beneficial in patients with DEB for preventing ocular complications such as symblepharon.

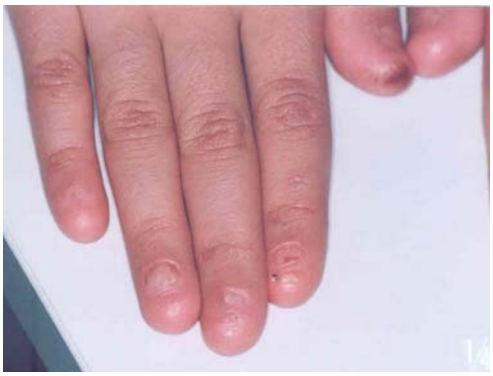

Figure 3 Nail dystrophy together with missing nails in some fingers.

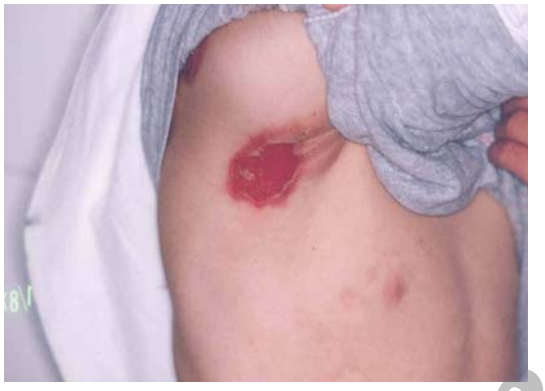

Figure 1 Bullous lesions on the trunk.

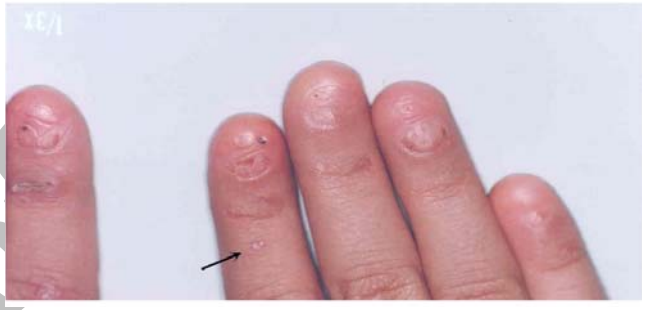

Figure 4 Milia on the dorsal aspect of fingers.

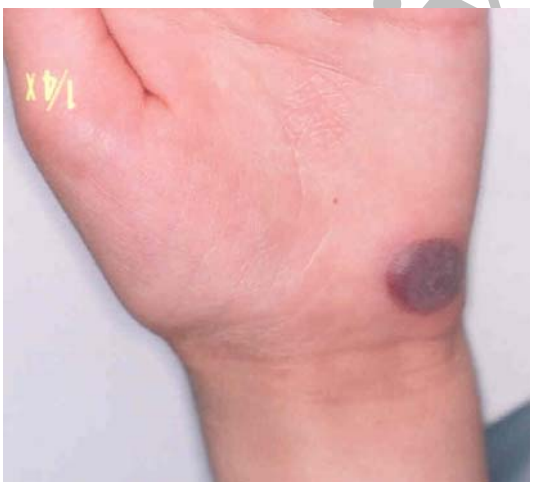

Figure 2 Dermal blister on the palm.

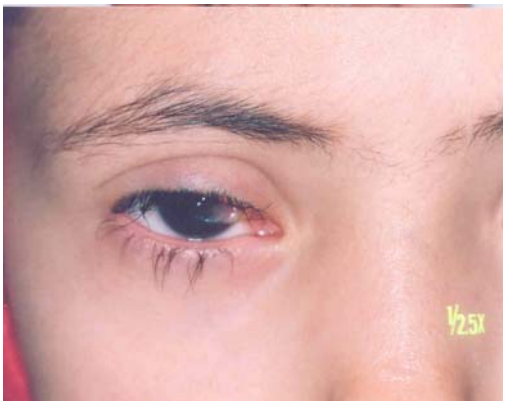

Figure 5 Symblepharon in the superonasal quadrant extending to the cornea.

REFERENCES

1. Fine JD, Johnson LB, Weiner M, Stein A, Cash S, Deleoz J, et al. Eye involvement in inherited epidermolysis bullosa: experience of the National Epidermolysis Bullosa Registry. *Am J Ophthalmol* 2004;138:254-262.
2. Sawamura D, Nizeki H, Miyagawa S, Shinkuma S, Shimizu H. Novel indel COL7A1 mutation 8068 del17ins GA causes dominant dystrophic epidemolysis bullosa. *Br J Dermatol* 2006;154:995-997.

3. Azrak B, Kaevel K, Hofmann L, Gleissner C, Willershausen B. Dystrophic epidermolysis bullosa: oral findings and problems. *Spec Care Dentist* 2006;26:111-115.
4. Matsumoto Y, Dogru M, Tsubota K. Ocular surface findings in Hallepau-Siemens subtype of dystrophic epidemolysis bullosa: report of a case and literature review. *Cornea* 2005;24:474-479.
5. Lin AN, Murphy F, Brodie SE, Carter DM. Review of ophthalmic findings in 204 patients with epidermolysis bullosa. *Am J Ophthalmol* 1994;118:384-390.
6. Tong L, Hodgkins PR, Denyer J, Brosnahan D, Harper J, Russel-Eggitt I, et al. The eye in epidermolysis bullosa. *Br J Ophthalmol* 1999;83:323-326.
7. Lazzaro DR, Kwon IS. Epidermolysis bullosa in a child. *CLAO J* 2002;28:75-76.
8. Altan-Xaxioglu R, Akova YA, Oto S. Amniotic membrane transplantation for treatment of symblepharon in a patient with recessive dystrophic epidermolysis bullosa. *Cornea* 2006;25:971-973.