Junctional Epidermolysis Bullosa in a Young Patient

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INTRODUCTION

Epidermolysis bullosa is a cluster of hereditary disorders characterized by blister formation of the skin or mucosa either spontaneously or as a result of minor trauma. Major groups include epidermolysis bullosa simplex, junctional epidermolysis bullosa, autosomal dominant dystrophic epidermolysis bullosa, and autosomal recessive dystrophic epidermolysis bullosa.

Junctional epidermolysis bullosa, the rarest type of epidermolysis bullosa, is an autosomal recessive disorder with further classification into lethal and nonlethal forms. Patients with nonlethal junctional epidermolysis bullosa suffer from ocular and systemic complications. There is limited information in the literature on the long-term ocular prognosis of this rare condition, especially in children.

We describe the ocular findings in an 8-year-old boy who was diagnosed as having junctional epidermolysis bullosa at the age of 2 years.

CASE REPORT

A male born at term presented with skin lesions and respiratory distress at 5 months of age. His family history was remarkable for his older brother, who had previously been diagnosed as having epidermolysis bullosa.

A physical examination revealed hemorrhagic bullous lesions confined to the genital area. On further evaluation, a laryngeal web was detected and tracheostomy was performed. At 2 years of age, diffuse hemorrhagic bullous eruptions appeared on his extremities, forehead, and malar regions extending to his lower eyelids (Fig. 1). His fingernails were remarkably dystrophic. Cicatricial ectropion and exposure keratitis were noticeable inferiorly on both eyes. Findings from the rest of the ocular examination were normal.

A skin biopsy performed on the left thigh demonstrated a cleft located precisely at the lamina lucida of the basement membrane along the epidermo-dermal junction consistent with junctional epidermolysis bullosa.

The number of hemidesmosomes attached to the plasma membranes of the keratocytes was not remarkably decreased, unlike findings in typical junctional epidermolysis bullosa; however, some appeared to be smaller than others (Fig. 2).

During the 6-year period of follow-up, the patient presented with recurrent corneal epithelial erosions on several occasions; some occurred spontaneously as others were induced by minor trauma. These lesions responded well to treatment with lubricants and antibiotic ointment. Visual acuity remained stable at 20/30 in the right eye and 20/40 in the left eye. The patient underwent ectropion repair bilaterally and facial grafting several times for chronic wounds. On his last ocular examination, his eyelids were in the normal position. The left lower eyelid margin...
appeared atrophic with scar formation and the eyelashes were scarce in the medial region of the lower lids. Lower eyelid massage caused a small amount of meibomian secretion. Tear break-up time was reduced in both eyes. The conjunctiva was normal without any sign of symblepharon formation. The limbal tissue looked healthy and there was no corneal pannus other than superficial corneal scars located inferiorly on both eyes. No corneal staining was noticeable with fluorescein. A dilated funduscopic examination revealed hypertensive retinopathy. Systemically, renal failure and hypertension caused by obstructive uropathy were the main chronic problems and abdominal surgeries were performed to alleviate the obstruction.

**DISCUSSION**

The etiology of epidermolysis bullosa is not clearly understood. Increased collagenase activity within the bullous cavity was suggested to cause a breakdown of type VII collagen, a major component of anchoring fibrils at the lamina lucida.\(^1\) Mutation reducing adhesion between epidermis and dermis has been most frequently located within the LAMB3 gene.\(^2\)

In general, junctional epidermolysis bullosa is known to have absent or rudimentary hemidesmosomes between the basal cell layer and the basement membrane, but some subtypes have been shown by morphometric analysis to have a normal number of hemidesmosomes, similar to our findings.\(^3\) Irregularities of the basement membrane and discontinuity between the basement membrane and adjacent hemidesmosomes have been shown in a corneal specimen of a patient with epidermolysis bullosa simplex.\(^4\)

Ocular findings are less common and severe in junctional epidermolysis bullosa than in dystrophic epidermolysis bullosa. However, ectropion and recurrent corneal abrasions are frequently encountered. Ocular involvement has been reported in as young as a 1-month-old infant.\(^5\) Lin et al. described 20 patients with ocular findings (9 with corneal abrasion, 5 with corneal scar, 5 with eyelid scar or ectropion, and 1 with conjunctival blister) among 36 patients diagnosed as having junctional epidermolysis bullosa.\(^6\) Conjunctiva is relatively well spared in junctional epidermolysis bullosa. Our case and previous studies support this finding.\(^6,7\) Corneal lesions were identified in 40% (6 of 15) of patients with junctional epidermolysis bullosa in the study performed by Tong et al.\(^8\)

Lubricant is the mainstay treatment of corneal abrasions in junctional epidermolysis bullosa. Bandage contact lenses and lamellar keratectomy are reserved for more severe cases.\(^6\) Tarsorrhaphy and ectropion surgery are performed for exposure keratitis.

Early diagnosis and treatment of corneal problems prevent more serious ocular complications in junctional epidermolysis bullosa. However, clinicians and parents should be prepared for chronic multi-system morbidity associated with impaired wound healing.

Promising studies of gene therapy targeting defective genes previously identified are under way and prenatal diagnosis of the disease is now available.\(^9\)

**REFERENCES**