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Case Report

Epidermolysis bullosa: a tale of two sisters

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ABSTRACT

Epidermolysis bullosa (EB) is a clinically and genetically heterogenous, inherited, mechanobullous disorder characterised by spontaneous or trauma induced blisters over skin and mucous membranes. Four major forms are: EB Simplex, EB Junctional, EB dystrophic and mixed. A 11-year-old and a 9 year old female child, both sisters, accompanied by their father, presented to dermatology OPD, with chief complaints of multiple raw areas over elbows and knees since infancy. There was history of fluid filled lesions which were induced by trauma or friction during handling or while playing. These lesions ruptured on their own or on trauma to form raw areas with crusting which further healed with scarring and small white raised lesions. Also, he gave history of loss of nails following blistering on toes in infancy. No associated systemic complaints were noted. On examination, both of them presented with multiple erosions and ulcers with oozing of blood, few bullae and crusted lesions over elbows, knees and feet. There were multiple atrophic scars and skin coloured to whitish papules [milia] over feet, ankles, elbows and knees. There were loss of nails of toes of both feet. On biopsy, histopathological examination revealed bullae at dermoepidermal junction. Diagnosis of EB junctional was made. EB is a rare inherited disorder. Its management includes prevention and healing of blisters and infection. Psychological support to patients and their families is of prime importance.

Keywords: Epidermolysis, Blisters, Mechanobullous, Dermoepidermal

INTRODUCTION

Epidermolysis bullosa (EB) is a clinically and genetically heterogeneous, inherited skin fragility disorder characterized by structural anomalies that cause disruption at the dermoepidermal junction or in the basal layer of the epidermis, resulting in increased cutaneous vulnerability to mechanical stress.[1]

EB is due to a mutation in at least one of 16 different genes. Several genes encoding structural proteins that make up the intraepidermal adhesion and dermoepidermal anchoring complexes in the basement membrane zone of the skin and mucosae, focal adhesions, desmosome cell junctions, and hemidesmosome attachment complexes are the cause of EB.[2-5]

Based upon the level of skin cleavage, EB is classified into four major groups:[6] EB simplex (EBS)-Intraepidermal cleavage plane within the basal layer of keratinocytes (basal EBS). Junctional EB (JEB)-Cleavage plane within the lamina lucida of the dermoepidermal junction.

Dystrophic EB (DEB)-Cleavage plane below the lamina densa, within the upper papillary dermis at level of anchoring fibrils. Kindler EB (KEB)- Multiple cleavage planes (intraepidermal, intralamina lucida, or sublamina densa).

EB can be diagnosed either by a skin (punch) biopsy at the edge of a wound with immunofluorescent mapping, or via blood sample and genetic testing

CASE REPORT

Course in dermatology OPD

Our patients, two sisters, 11-year-old and 9 year old, both females presented to the dermatological OPD accompanied by their father.

Chief complaints of multiple raw erosions over bilateral elbows and knees. These erosions were preceded by multiple fluid filled lesions containing clear fluid. Erosions healed with scarring and small whitish raised atrophic lesions. Both these daughters developed such multiple lesions on trivial trauma or friction since infancy, just after few days of birth. On further questioning, the father also complained of loss of nails of bilateral hands and feet since infancy, also preceded by fluid filled lesions.

First case (11 year old)

Single ulcer with oozing of fluid and crust/scab present over left elbow (Figure 3).

Multiple atrophic hypopigmented scars with few areas of perifollicular hyperpigmentation over bilateral knees and right elbows (Figure 2).

Multiple skin colored to whitish papules to plaques over dorsum of bilateral feet, ankles and hands.

Loss of nail plate and few thickened dystrophic nails over bilateral feet and hands (Figure 1).

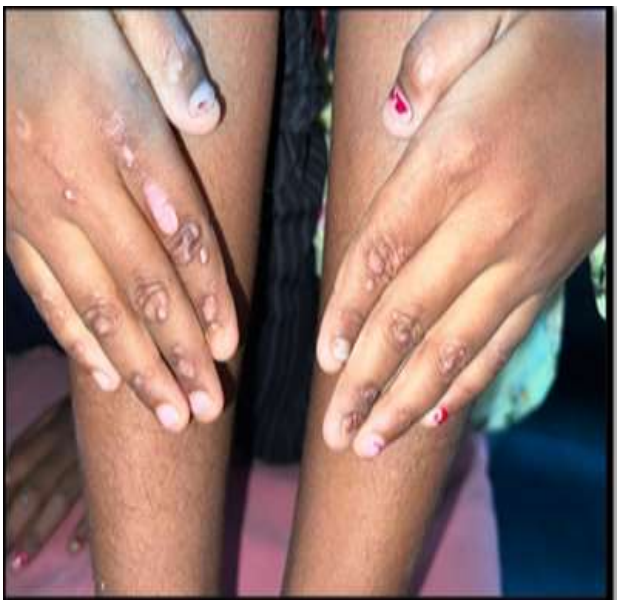

Figure 1: Loss of nail plate.

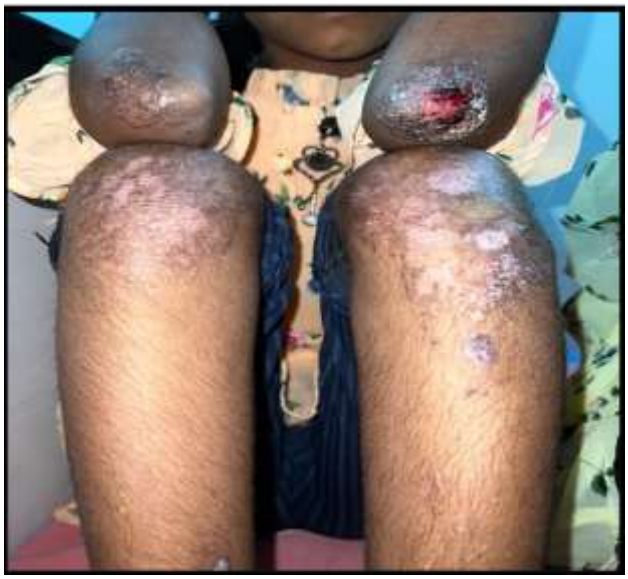

Figure 2: Atrophic scars.

Figure 3: Single crusted erosion.

Second case (9 year old)

Single erosion with crusting present over scalp, fourth web space of right foot, left knee each (Figure 5).

Multiple atrophic hypopigmented scars with few areas of perifollicular hyperpigmentation over bilateral knees, right elbow (Figure 4).

Multiple skin colored to whitish papules to plaques over dorsum of bilateral feet and hands. Loss of nail plate and few thickened dystrophic nails over bilateral feet and hands.

Figure 4: Atrophic scars.

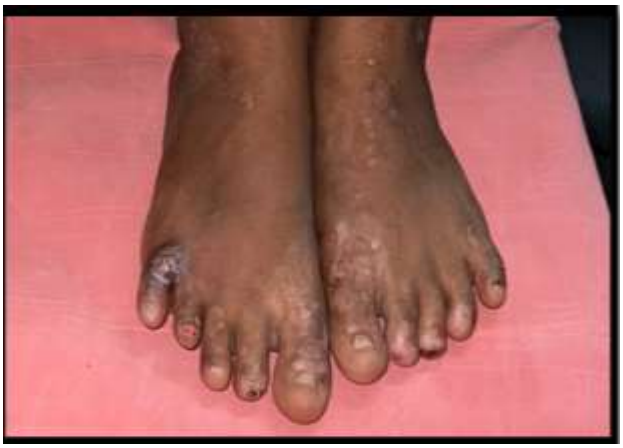

Figure 5: Single crusting erosion.

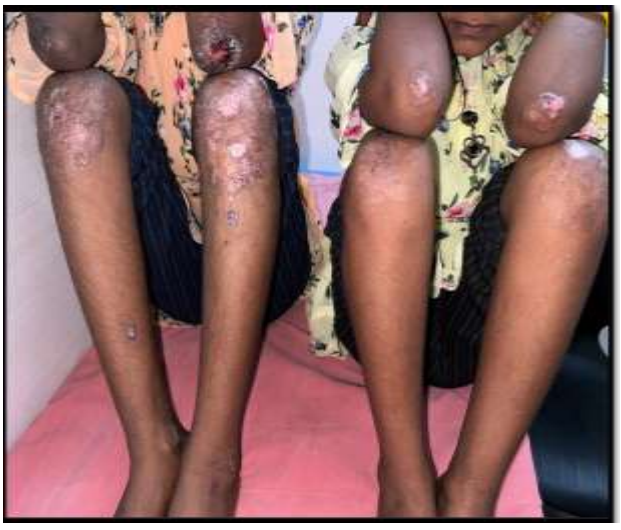

Figure 6: Multiple atrophic hypo/hyperpigmented scars.

Diagnosis

Skin punch biopsy was sent from vesicle over right foot from both the sisters.

Section was studied up to deep dermis. The epidermis was thinned out which showed hyperkeratosis and parakeratosis.

Bullae were seen at dermo-epidermal junction.

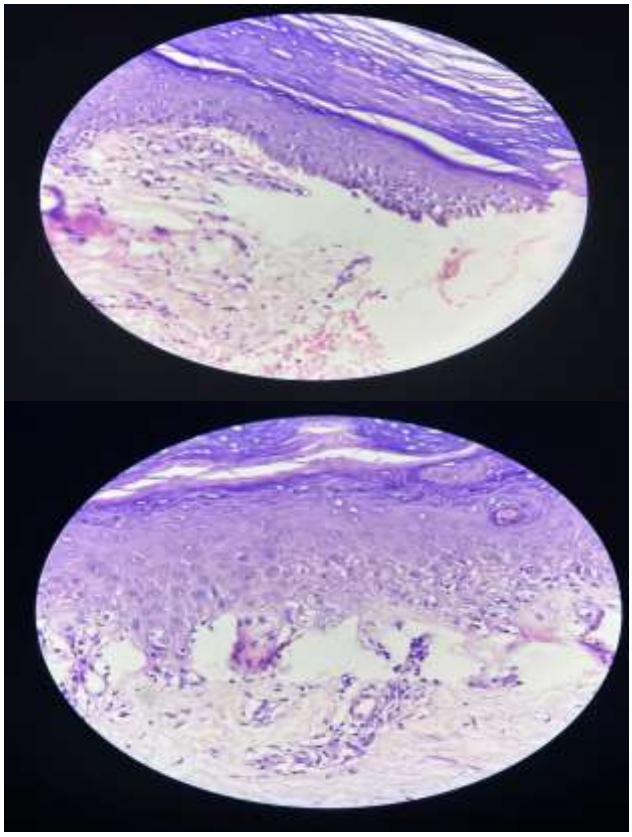

Figure 7: Skin punch biopsy diagnosis: JEB.

Based on clinical and histopathological results, final diagnosis was generalized atrophic benign EB-subtype of JEB.

Treatment

With respect to the treatment of lesions, blister puncturing to prevent dissemination and use of sterile dressings was done. Lesions were cleaned with solutions of low toxicity, such as saline solution and water. The skin was left in place, functioning as a biological dressing and preventing bacterial colonization. Firm and easily torn crusts require debridement. Analgesics and antibiotics to prevent infection were prescribed. Meticulous skin care as to avoid trauma and thereby blistering was advised. Patient's parents were counselled about the condition.

DISCUSSION

JEB encompasses a group of autosomal recessive disorders affecting 2.7 per million live births characterized by blistering of the skin and mucosae that heal with scarring.

Primary cause of JEB is autosomal recessive mutations in laminin-332 gene which result in a structural defect of the anchoring filaments located in the lamina lucida and superior lamina densa of the basal membrane zone.[7]

In atrophic benign JEB, Repeated blistering and healing causes pigmentary abnormalities, skin atrophy with a poikilodermatous look, and faint, stellate scars.[8-10]

Hence, wound care, pain control, nutritional support form the mainstay of treatment. Nonstick or nonadherent silicone dressings and foam dressings that absorb exudates are ideal as primary dressings for EB wounds.[11] For non-healing ulcers, PRF can be done for faster recovery. Punch grafting has been successfully used in a few patients with laminin 322-deficicent JEB for the treatment of chronic, deep ulcers on the extremities. The serum levels of micronutrients such as iron, zinc, selenium, vitamin B12, vitamin A, and folate should be checked at regular intervals (3-6 months for iron, 12 months for vitamins and trace elements) and necessary supplementation should be done.[12]

CONCLUSION

Most patients with junctional epidermolysis bullosa pose a considerable risk of infant mortality. Sepsis failure to thrive and respiratory failure are the major causes of death. Patients who live to adulthood are more likely to acquire squamous cell carcinoma thus, genetic counselling plays a major role in pre natal and pre implantation genetic diagnosis. Affected families should be offered genetic counselling so that they can better understand their reproductive risks and options.

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