CASE REPORT

Epidermolysis Bullosa Puriginosa: Report of a Case

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Abstract

Epidermolysis Bullosa Puriginosa is a genetic mechanobullous disease characterized by pruritus, lichenified or nodular prurigo-like lesions, occasional trauma-induced blistering, excoriations, milia, nail dystrophy and albopapuloid lesions that appear at birth or later. Scarring and prurigo are most prominent on the shins. Herein, we report a case with a history of blisters since childhood followed by intensely pruritic lesions predominantly on the shins and dystrophy of the toenails, milia, excoriations and diffuse post-lesional hyper and hypopigmentation. *(Iran J Dermatol 2010;13: 20-23)*

Key words: dystrophic epidermolysis bullosa puriginosa, pruritis, dermal–epidermal junction

Case Report

A 15-year-old Iranian girl, the older of two daughters with no family history of skin diseases, presented with trauma-induced blistering, pruritus and nail dystrophy since she was 1.5 years old. The blisters were non trauma-induced since she was 6 years old. She had localized areas of repeated blistering, involving the same area of both shins and forearms and occasionally affecting other sites. Severe pruritus on the pretibial aspects of both legs worsened over time. Physical signs which were most apparent on the shins but could also seen on the forearms, trunk, back and neck consisted of intact blisters, erosions and scars (Figure1). Many of the scars were raised, taking the form of either nodules or plaques, often with a lichenified surface. There were diffuse post-lesional hyper and hypopigmentation patterns, particularly on the trunk and neck. Apart from the skin changes and dystrophy of her toenails (Figure 2), she was in good general condition with no significant problems in swallowing, bowel functions, nutrition, eyes or teeth.

There was no evidence for other causes of itching, such as thyroid dysfunction, anemia, eczema or atopy. The patient received different topical therapies and oral antihistamines.

Total serum IgE level was elevated more than 200 U/ml (normal <100). Other laboratory test results were within normal limits.

Skin biopsies of the shin and the back of the trunk showed dermo-epidermal separation with ulceration and crust formation. Dermis revealed mononuclear cellular infiltration with melanophages and extravasation of RBCs. A prominent scar tissue was present in the dermis (Figure 3).

We started topical clobetasol propionate 0.05% ointment twice daily with oral vitamin E 400 u/daily and oral antihistamines (loratadine 10 mg/daily, cetirizine 10 mg/night) with some symptomatic improvement after four months. She was followed up every four months.

Discussion

The term Epidermolysis Bullosa (EB) was first described in 1886. It was not until 1962, however, when the first sophisticated classification scheme was proposed by Pearson, based on the application of transmission electron microscopy to the study of inherited blistering diseases [1]. The prevalence is estimated to be one in every 20 000 live births [2]. Three major types are redefined: Epidermolytic (EB simplex [EBS]), Lucidolytic (Junctional EB [JEB]), and Dermolytic (dystrophic EB [DEB]) based on differences in the ultrastructural

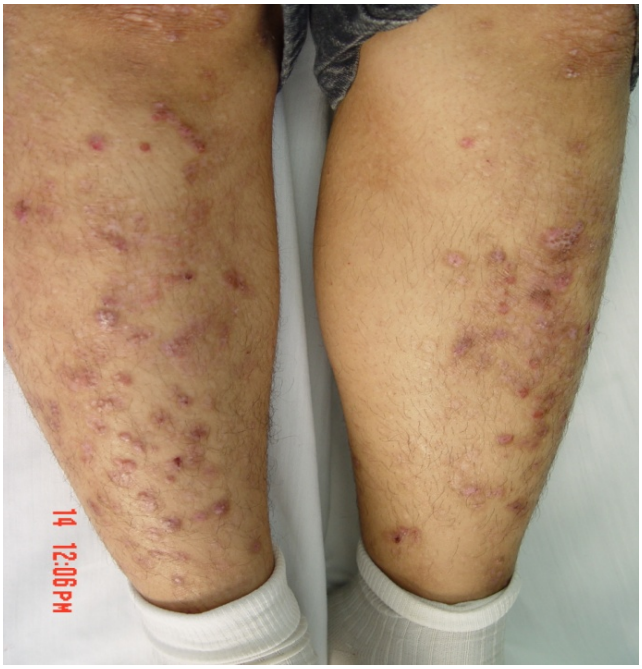

Figure 1. *Multiple nodules or plaques with a lichenified surface on the shin*

Figure 2. *Marked toenails dystrophy*

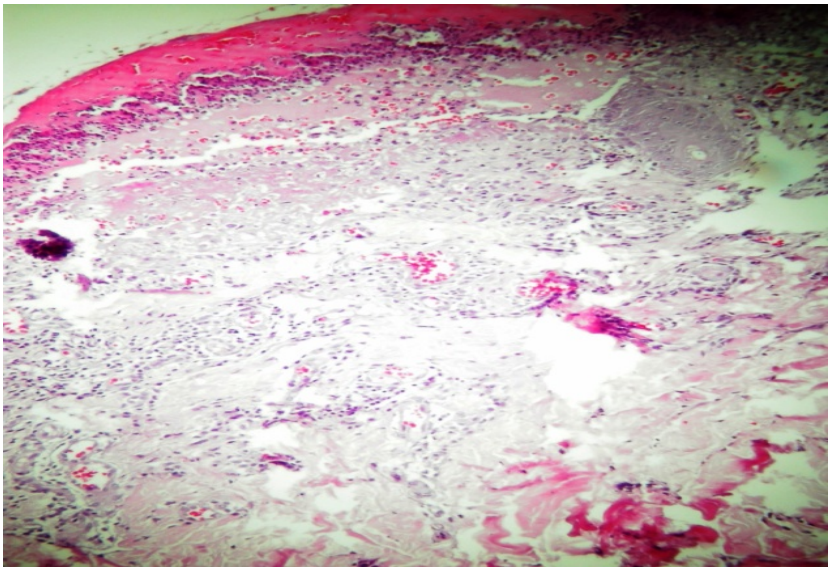

Figure 3. *Histopathology view: surface ulceration with crust and exudate collection at the dermo-epidermal dissociated area (H&E*!0).*

level within which blisters develop in EB skin, either spontaneously or following minor friction or trauma[1].

Dystrophic epidermolysis bullosa is characterized by skin fragility, blistering, scarring, nail changes and milia formation. Unlike the other types of EB, there are both major autosomal recessive and autosomal dominant subtypes [3]. Fewer than 40 patients with autosomal dominant or recessive inheritance or sporadic DEB-Pr have been described in the literature [4].

Blistering is due to abnormalities in anchoring fibrils (AF), microstructures mainly composed of type VII collagen (COLL VII), which contribute to the maintaining of dermal–epidermal adhesion [5].

Table 1. *Ultrastructural findings among major types and selected subtypes of EB* [1]

EB type or subtype	Ultrastructural site of skin findings	Other ultrastructural findings
EB simplex (EBS)		
EBS, localized	Basal layer	Split may spread to suprabasilar layer
EBS, DM	Basal layer in subnuclear cytoplasm	Dense, circumscribed clumps of keratin filaments (most commonly observed within lesional biopsy sites)
BS-MD	Predominantly in basal layer, above level of HD attachment plaque	Reduced integration of keratin filaments with HD
EBS-AR	Basal keratinocytes	Absent or reduced keratin filaments within basal keratinocytes
EBSS	Split usually at interface between granular and cornified cell layers	—
EBS, lethal acantholytic	Suprabasal cleavage and acantholysis	Perinuclear retraction of keratin filaments
EBS, plakophilin-1 deficiency	Mid-epidermal cell-cell separation	Diminutive suprabasal desmosomes; perinuclear retraction of keratin filaments
EBS-PA	Lower basal layer, above level of HD plaque	Reduced integration of keratin filaments with HD
Junctional EB (JEB)		
JEB-H	Lamina lucida	Markedly reduced or absent HD; absent SBDP
JEB-nH	Lamina lucida	HDs may be normal or reduced in size and Number
JEB-PA	Lamina lucida	Small HD plaques often with attenuated SBD
Dominant dystrophic EB (DDEB)		
DDEB, generalized	Subelamina densa	Normal or decreased numbers of AFs
DDEB-BDN	Subelamina densa	Electron-dense stellate bodies within basal layer; reduced AFs
Recessive dystrophic EB (RDEB)		
RDEB, severe generalized	Subelamina densa	Absent or rudimentary AFs
RDEB, generalized other	Subelamina densa	Reduced or rudimentary-appearing AFs
RDEB-BDN	Subelamina densa	Electron-dense stellate bodies within basal layer; reduced AFs

AF: Anchoring Fibril, AR: Autosomal Recessive, BDN: Bullous Dermolysis of the newborn, DM: Dowling-Meara, EBSS: EBS Superficialis, H: Herlitz, HD: Hemi Desmosome, MD: Muscular Dystrophy, nH: non-Herlitz, PA: Pyloric Atresia, SBDP: Sub-Basal Dense Plate.

Dystrophic epidermolysis bullosa puriginosa (DEB-Pr) is a distinctive clinical subtype of dystrophic EB [6]. In DEB-Pr patients, autosomal dominant and autosomal recessive inheritance and sporadic inheritance patterns have been recognized [5]. DEB-Pr presents either at birth with mild acral blistering and erosions, or during infancy or childhood [3]. It is clinically characterized by pruritus lichenified plaques or nodular prurigo-like lesions, violaceous linear scarring, occasional trauma-induced blistering, excoriations, milia, nail dystrophy and, in some cases, albopapuloid lesions on the trunk. The scarring is most evident on the limbs, particularly on the shins, with relative sparing elsewhere. Intact blisters are rarely seen. The diagnosis of EB in these patients may therefore be difficult, particularly as the condition may only manifest itself some years after birth. Scars frequently have a lichenoid appearance which may cause confusion with non-EB dermatoses, particularly hypertrophic lichen planus, lichen simplex, cutaneous amyloidosis and Nekam's disease [6].

Histologically, a split may be evident at the dermal-epidermal junction, although frank blisters are rarely seen. The cause of the severe pruritus is unknown; however, a number of patients have raised blood levels of immunoglobulin E (IgE), suggesting a possible atopic background. It shares several features with the pretibial form of dystrophic EB, but is clinically much more striking [3].

There is no specific treatment for any form of EB, and treatment is unsatisfactory [7]. The mainstay of clinical management is based on protection and avoidance of provoking factors. Long-term systemic corticosteroid treatment is not considered because of the high risk of complications. Phenytoin, which once appeared to control blistering in certain

patients in an open study, did not prove to be more effective than placebo in a controlled trial. Other systemic drugs that have been tried with variable results in small numbers of patients include vitamin E, minocycline, ciclosporin, and retinoic acid [3]. Some reports of helpful interventions in EBP have been published which include topical treatments (e.g. tacrolimus), systemic agents (e.g. ciclosporin or thalidomide), and cryotherapy [8]. Oral thalidomide therapy has been highly beneficial in patient with dominant Dystrophic EB Pruriginosa [7].

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