Epidermolysis bullosa pruriginosa

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Sri Lanka Journal of Dermatology, 2006, **10**, 49-50

Abstract

Epidermolysis bullosa pruriginosa (EBP) is a unique form of dystrophic epidermolysis bullosa with marked pruritus, intermittent blistering, violaceous linear plaques mainly on shins and forearms, milia formation and toe nail dystrophy. EBP may present either at birth, during infancy or childhood. Most cases are sporadic. However both autosomal recessive and autosomal doinant inheritance are recognized in some cases. Diagnosis is mainly clinical but can be confirmed with immunofluorescence and electrone microscopy. Treatment for this genetic blistering disease is unsatisfactory. We report a child with EBP and analysis of his pedigree revealed number of similarly affected people

An 11 year old boy a product of non consanguineous marriage presented with spontaneous blistering in hand and feet associated with marked pruritus. Examination revealed scarring, milia, and toe nail dystrophy. His mother also had similar features in her childhood and later developed linear lichenified plaques on shins, forearms and hands. Pedigree analysis revealed an autosomal dominant inheritance. The clinical diagnosis of EBP was made and child was managed conservatively.

Case report

An 11 year old boy a product of a non consaguinous marriage presented with blistering since birth. Blisters were both spontaneous and trauma induced. Blistering were more common on the hands and the feet. Pruritus which was troublesome occurred few years later. Itching was particularly severe on the legs. Examination of the child revealed acro located, fresh erosions, and pink lichenoid papules and milia (Figure 1). His toe nails were abnormal but the finger nails were normal.

His mother also has been suffering from a similar illness since childhood. Pedigree analysis revealed a number of affected members in all 4 generations indicating an autosomal dominant inheritance.

Examination of the mother's limbs revealed numerous excoriated and lichenified papules and plaques with associated scarring. (Figure 2 mother's hands) Many lesions had a curvi-linear configuration. The signs were most apparent on shins and backs of the hands. Lesions clinically resembled hypertrophic lichen planus. Nail dystrophy was noticed on the toe nails, and consisted of shedding of nail plates leaving vestigial nail spurs (Figure 3). Her face, neck and the major flexures were spared.

A diagnosis of Epidermolysis bullosa pruriginosa was made clinically. Histological confirmation of the diagnosis could not be entertained due to mother's refusal.

Treatment for this mecanobullous disorder was disappointing. Secondary bacterial infection was controlled with topical antibiotics – 2% Mupirocin ointment and a course of oral – Cephalexin. Pruritus responded to a course of oral – Chlorpheniramine.

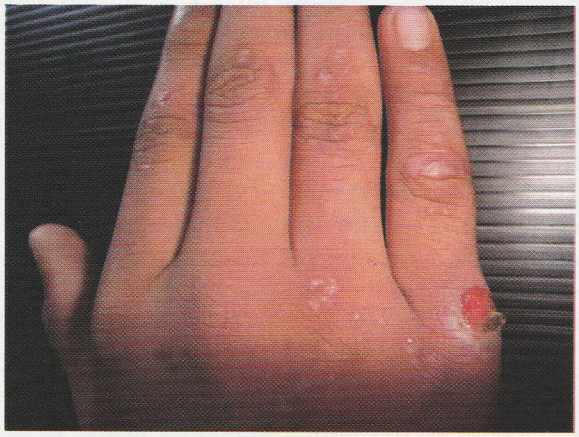

Figure 1.

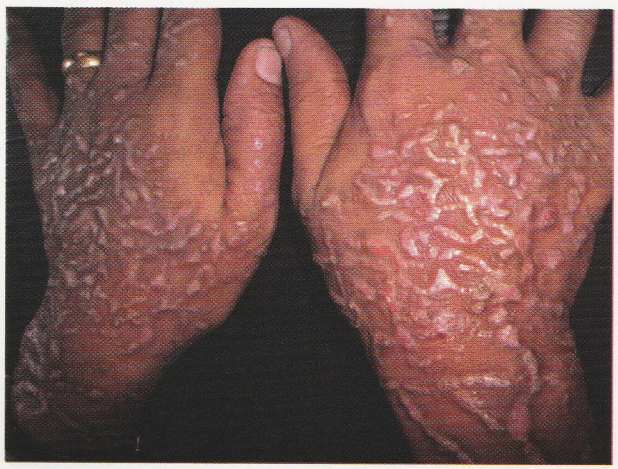

Figure 2. *Mother's hands.*

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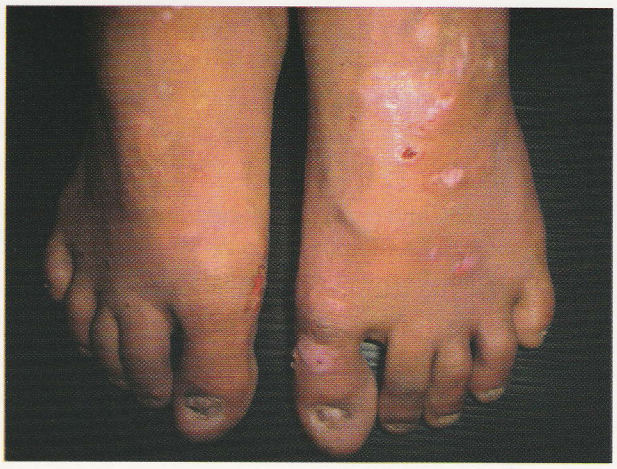

Figure 3.

Discussion

EBP was first described by McGrath in 1994. It's a unique form of DEB with mild acral blistering and erosions at birth, during infancy or childhood. Violaceous papular and nodular lesions, often in a linear arrangement, are mainly confined to the shins and forearms, although rare lesions may occur on the trunk. In adults, the lesions are chiefly lichenified plaques. Pruritus is invariable giving rise to excoriations. Occasionally trauma induced blisters may occur. Milia formation and toe nail dystrophy are associated features. In some cases albopapuloid lesions on the trunk can be seen. Intact blisters are rarely seen. When patients are seen with these features an erroneous diagnosis of lichen planus is often made.

Most cases of EBP are sporadic. However autosomal recessive and autosomal dominant inheritance is recognized. Squamous cell carcinoma can arise from a scar.

The diagnosis is mainly clinical but can be confirmed by light microscopy, immunofluorescence and electron microscopy. Light microscopic examination of lichenified plaque shows hyperkeratosis and mild acanthosis Sub epidermal blister formation with marked to moderate perivascular and interstitial lymphohistiocytic infiltration can be seen in upper and mid dermis. Fibrosis of papillary dermis and milia are also aiding the diagnosis. Direct immunofluorescence shows bright linear staining at dermo-epidermal junction. Electron microscopy can be used to demonstrate few or absent anchoring fibrils.

EBP should be considered in the differential diagnosis of pruritic lichenified papules and plaques occurring in adolescents and adults, particularly when it affects the legs. It may closely resemble hypertrophic lichen planus. A history of blistering in early childhood, autosomal dominant inheritance, presences of milia, immunofluorecence helps to distinguish the two conditions.

References

1. Fine JD, Bauer EA, Briggaman RA, et al. Revised clinical and laboratory criteria for subtypes of inherited epidermolysis bullosa. *J Am Acad Dermatol* 1991; **24**: 119-35.

2. Eady RAJ. Classification of epidermolysis bullosa. In: Epidermolysis Bullosa. A Comprehensive Review of Classification, Management and Laboratory Studies. 1990; 1-9.

3. Rook's Textbook of Dermatology Edition. Vol 2 .40.22.