Epidermolysis Bullosa Pruriginosa with Nail Dystrophy: A Rare Case Report

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Epidermolysis bullosa (DEB) pruriginosa is a type of dystrophic DEB wherein there is a mutation in gene COL VII A1, which encodes anchoring fibril protein Type VII collagen. Clinically, it is characterized by intensely pruritic linear lichenified or nodular prurigo like lesions over extremities with milia, nail dystrophy, and in some cases albopapuloid lesions over trunk. Here we report a case of an adult onset DEB pruriginosa with typical clinical features which was confirmed by histopathology. In any severely itchy skin lesion over pretibial region, DEB pruriginosa should be kept in mind, and DEB pruriginosa can occur for the first time in adulthood also.

Keywords: Adult, Epidermolysis bullosa dystrophica, Lichenified eruption

INTRODUCTION

Epidermolysis bullosa (DEB) is a group of genetically transmitted disorders characterized by skin fragility and blistering, in response to mild mechanical trauma.¹ On the basis of the level of cleavage, DEB has been divided into three broad categories. (a) DEB simplex - in which blisters develop intra epidermally above the basement membrane, (b) junctional DEB - in which blisters develop within basement membrane, (c) dystrophic DEB - where blisters form beneath basement membrane. DEB pruriginosa is a type of dystrophic DEB wherein there is a mutation in gene COL VII A1 which encodes anchoring fibril protein Type VII collagen.² Most cases are sporadic, but both autosomal recessive and dominant inheritance is recognized.³

CASE REPORT

A 45-year-old female born of nonconsanguineous marriage presented to our outpatient department with complaints of blistering over both shins on mild mechanical trauma of 5 years duration. The lesions were intensely pruritic. The lesions started as papules and vesicles. The vesicles ruptured to form erosions which healed with scarring and atrophy. The papules coalesced to form lichenified plaques. All the toenails were dystrophic (Figure 1). Similar lesions were noted in her father and two brothers. The differential diagnosis entertained was DEB pruriginosa, pretibial DEB, lichen amyloid, prurigo nodularis, hypertrophic lichen planus, and pretibial bullous pemphigoid.

All routine investigations were within normal limits. Tzanck smear did not show any acantholytic cells or inflammatory cells. Biopsy revealed a subepidermal bulla with lymphocytic infiltrate in the dermis and perivascular region (Figures 2 and 3) which was consistent with the diagnosis of DEB pruriginosa. Electron microscopy and immunofluorescence were not done due to lack of facility.

Figure 1: Clinical photograph showing few vesicles with areas of atrophy over left leg with nail dystrophy

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DISCUSSION

DEB pruriginosa is a rare type of dystrophic DEB. Most cases are sporadic, but both autosomal recessive and dominant inheritance is recognized. Clinically, it is characterized by intensely pruritic lichenified nodular lesions over extremities with atrophy, milia, and nail dystrophy. It usually presents at birth or during infancy/childhood, but it can also occur for the first time in adulthood. The reason for the delayed presentation is not known. Histologically hyperkeratosis, mild acanthosis, blister at dermoeipidermal junction and mild to moderate dermal lymphohistiocytic infiltrate are common. Recent molecular analysis studies have shown glycine substitution within the triple helical collagen domain of Type VII collagen molecule is exclusively associated with DEB pruriginosa. Clinical management of DEB pruriginosa is difficult.

Modalities of treatment tried include topical tacrolimus cryotherapy, dermabrasion and systemic cyclosporine, thalidomide, and etretinate. Prenatal diagnosis is possible and genetic counseling and gene therapy remain the most promising approaches.

CONCLUSION

In any severely itchy skin lesion over pretibial region, DEB pruriginosa should be kept in mind and DEB pruriginosa can occur for the first time in adulthood also. This case is presented for its rarity and also for the adult onset of lesions.

REFERENCES


How to cite this article: Mathan R, Ramasamy PP, Mahadevan K, Eswaramoorthy B, Madhavan R. Epidermolysis Bullosa Pruriginosa with Nail Dystrophy: A Rare Case Report. IJSS Case Reports & Reviews 2015;1(10):43-44.

Source of Support: Nil, Conflict of Interest: None declared.