ORIGINAL RESEARCH PAPER

Volume-7 | Issue-11 | November-2018 | PRINT ISSN No 2277 - 8179

INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

EPIDERMOLYSISBULLOSA PRURIGINOSA -A CASE REPORT

Dermatology

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ABSTRACT

INTRODUCTION: Epidermolysisbullosa refers to group of inherited disorders involving formation of blisters following trivial trauma. Epidermolysis bullosa pruriginosa is a type of dystrophic epidermolysis bullosa caused by type VII collagen gene COL7A1 mutation, characterized by pruritus, nodular prurigo like lichenified lesions in early phase and violaceous linear scarring, trauma induced blistering, excoriation, milia, nail dystrophy in late healing phase, in some cases albopapuloid lesions can be seen on the trunk.

CASE SUMMARY: 27 years old male had an itchy, discrete, multiple papular and lichenified nodular lesions of varying sizes from 0.5-1.5cm, present over legs since the age of 12. No history of blistering after trauma, nail or mucosal involvement. No history of consanguineous marriage. His father and mother had similar lesions over the extremities. Histo-pathological examination of skin biopsy showed sub-epidermal blister with thickened stratum corneum in roof of blister. Within the blister, fibrin, plasma and few neutrophils were seen. Overlying epidermis showed mild acanthosis, focal thinning and lamellated compact orthohyperkeratosis. Papillary dermis thickened & showed abundant fibroplasia with increased number of thick walled capillaries & moderately dense mixed inflammatory infiltrate. Clinical features & biopsy findings suggestive of epidermolysisbullosa pruriginosa. Treatment with topical super-potent steroids and systemic antihistamine showed no response after 1 month.

CONCLUSION: Epedermolysis bullosa pruriginosa is a type of dystrophic Epidermolysis bullosa caused by type VII collagen gene COL7A1 mutation. It can be complicated by infection, deformities, squamous cell carcinoma which may metastasize eventually leading to death. Multidisciplinary approach with avoidance of trauma is required. Genetic counselling of parents is advisable.

KEYWORDS

INTRODUCTION:

Epidermolysis bullosa (EB) refers to a group of inherited disorders that involve the formation of blisters following trivial trauma.[1] Epidermolysis bullosa pruriginosa is a recently described variant, caused by Type VII collagen gene mutation, with distinctive clinicopathological features characterized by pruritus, nodular prurigo like lichenified lesions in early phase and Violaceous linear scarring, trauma induced blistering, excoriation, milia, nail dystrophy in late healing phase, in some cases albopapuloid lesions can be seen on the trunk.[2] Most cases are sporadic,[3] but a few show autosomal dominant or autosomal recessive pattern of inheritance.[2] Microscopic studies of EB pruriginosa show typical findings of dystrophic EB,[4] and it has been postulated that itching lesions of EB pruriginosa could represent an abnormal dermal reactivity of some subjects to their inherited bullous disorder. The study of the molecular basis of dominant dystrophic EB (classical) and EB pruriginosa shows that both diseases are caused by a missense glycine substitution mutation by different amino acids in the same codon of COL 7A (G2028R and G2028A).[5]

CASE REPORT:

A 27 year old male presented with an itchy, discrete, multiple papular and lichenified nodular lesions of varying sizes from 0.5-1.5cm, present over legs since 12 years of age. There was no history of blistering after trauma, nail or mucosal involvement and no history of consanguineous marriage. Similar lesions were present over the extremities of his father and mother. Histo-pathological examination of skin biopsy showed sub-epidermal blister with thickened stratum corneum in roof of blister. Within the blister, fibrin, plasma and few neutrophils were seen. Overlying epidermis showed mild acanthosis, focal thinning and lamellated compact orthohyperkeratosis. Papillary dermis thickened & showed abundant fibroplasia with increased number of thick walled capillaries & moderately dense mixed inflammatory infiltrate. Clinical features & histo-pathological examination of biopsy findings suggest the probable diagnosis of epidermolysis bullosa pruriginosa. Topical super-potent steroids for local application twice a day and systemic antihistamine were given for 1 month, but treatment showed no response.

DISCUSSION:

EB pruriginosa is a newly characterized variant of dystrophic EB.[2] Patients had multiple papular and nodular prurigo-like lesions, mostly on the shins, and also on other parts of the legs, forearms, elbows, dorsal aspect of the hands, shoulders, and lower back, developing since birth in some cases, and between 6 months to 10 years of age in others. The face and flexures were always spared, and nail dystrophy, albopapuloid lesions, blisters, and milia were other common but not invariable features.

Histopathology showed hyperkeratosis, mild acanthosis, dermal lymphohistiocytic infiltrate, and also subepidermal bullae in some areas. Ultrastructural studies showed a reduction of anchoring fibrils in lesional, perilesional, and non-lesional skin, similar to dominant dystrophic or localized recessive EB, and morphometry of anchoring fibrils alone could not distinguish between different subtypes of dominant dystrophic or localized recessive EB and EB pruriginosa.[2]

In our patient, pruritus was the most oppressive symptom. Clinical presentation and histopathology were consistent with the diagnosis of EB pruriginosa, however, there were no albopapuloid lesions or milia. The occurrence of the condition in father points strongly towards a genetic origin, but the exact mode of inheritance is hard to specify.

Epedermolysisbullosa pruriginosa is a type of dystrophic Epidermolysisbullosa caused by type VII collagen gene COL7A1 mutation. It can be complicated by infection, deformities, squamous cell carcinoma which metastasized to death. Multidisciplinary approach with avoidance of trauma required. Genetic counselling of parents is advisable.

Figure I: lower extremities of patient

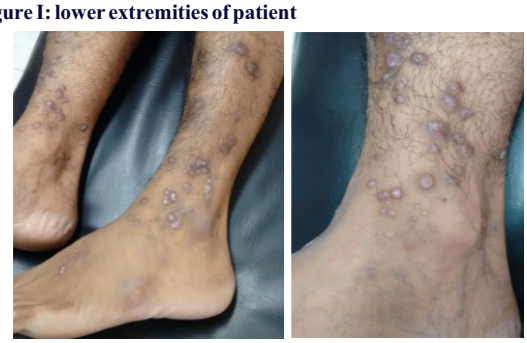


Figure II: Lower extremities of patient's father

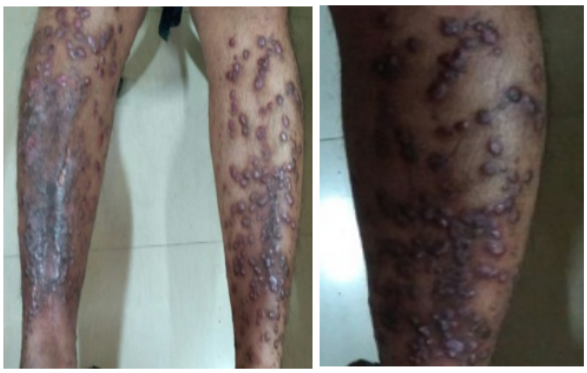
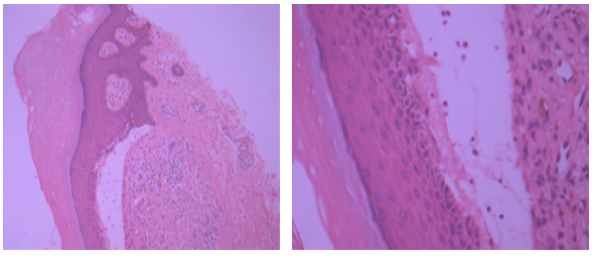


Figure III: histopathology of patient's punch biopsy



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