Familial Epidermolysis Bullosa Pruriginosa

Dear Editor,

Epidermolysis bullosa (EB) refers to a group of inherited mechanobullous disorders. Epidermolysis bullosa pruriginosa (EBP) is a type of dystrophic EB which occurs due to mutation in type VII collagen gene.[1,2] It is characterized by intense pruritus, prurigo nodularis like lesions and occasionally tense blisters. It occurs mostly on shin but other body sites like knees, elbows and hands may also be involved. Scarring, milia formation and nail dystrophy are commonly seen. The lesions showed exacerbation in hot and humid climate. The mode of inheritance can be sporadic or autosomal dominant or recessive.[3] We report EBP occurring in a family across two generations affecting three members.

A 25-year-old male presented to dermatology outpatient department with intensely itchy skin lesions over lower limbs for 7–8 years [Figure 1]. Similar lesions were also reported in 20-year-old younger sister [Figure 2] and maternal uncle. There was no history of consanguinity in family [Figure 3]. On examination patient had multiple, excoriated nodules on bilateral lower limbs [Figure 1a] with a few tense clear fluid filled blisters. Few of the lesions healed with scarring and milia [Figure 1b]. All toe nails and right ring finger nail were dystrophic [Figure 1c]. Mucosal, hair and teeth examination was normal. The lesions showed exacerbation during hot and humid weather.

Based on clinical findings, a differential diagnosis of EBP, prurigo nodularis, hypertrophic lichen planus and lichen amyloidosis was made. Punch biopsy was taken from blister for histological examination and sample from perilesional skin was taken for direct immunofluorescence (DIF). Histological examination revealed subepidermal bulla with mild hyperkeratosis and acanthosis. The bullous cavity was filled with proteinaceous fluid with scattered inflammatory cells. Dermis showed mild perivascular lymphocytic infiltrate [Figure 1d]. DIF study was negative for any

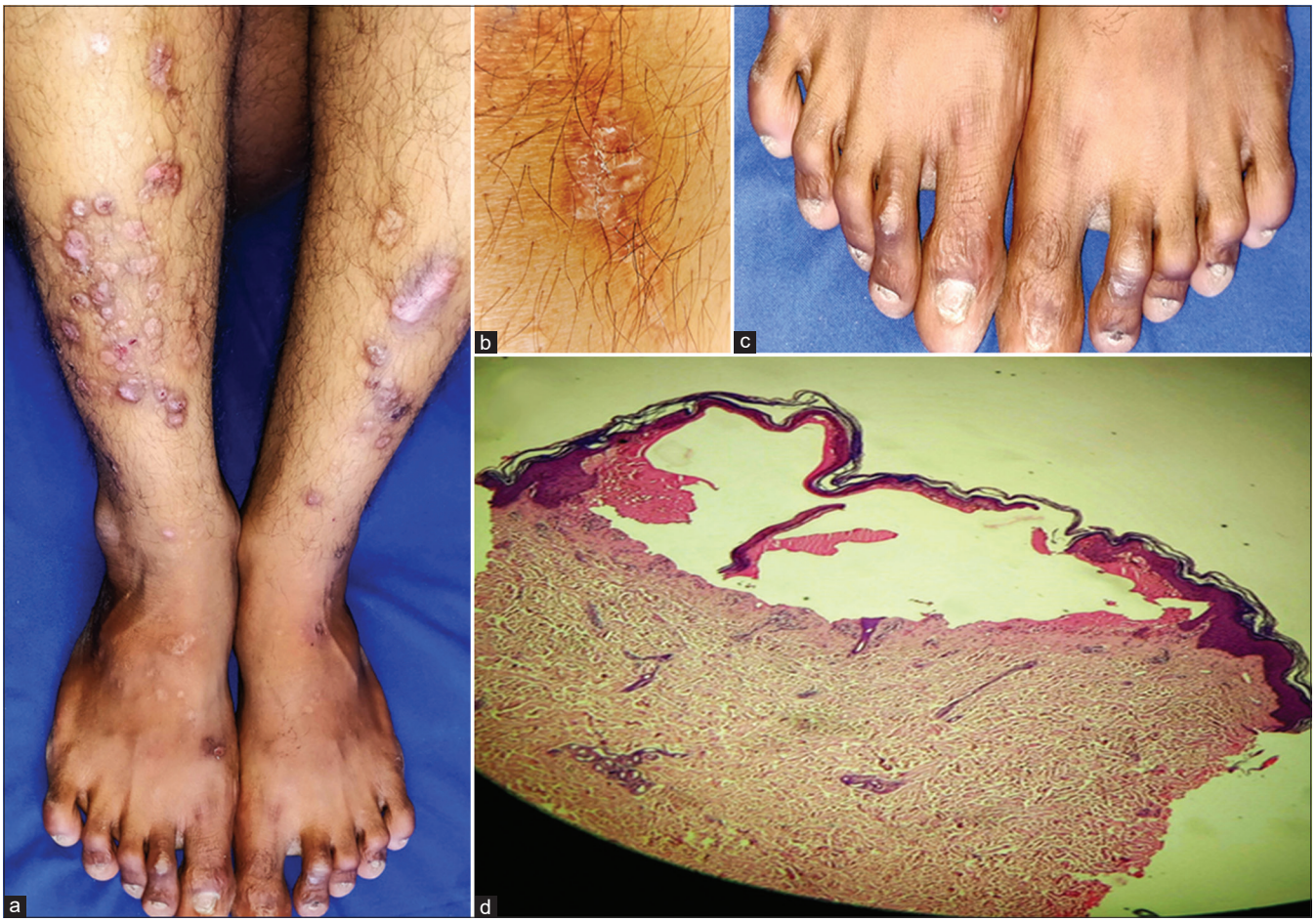

Figure 1: 25-year-old man with epidermolysis bullosa pruriginosa who presented with (a) multiple excoriated nodules on bilateral lower limbs. (b) Scarring and milia. (c) Dystrophic toe nails. (d) Subepidermal bulla with mild hyperkeratosis and acanthosis. Mild perivascular lymphocytic infiltration in dermis (Hematoxylin and eosin, x100)

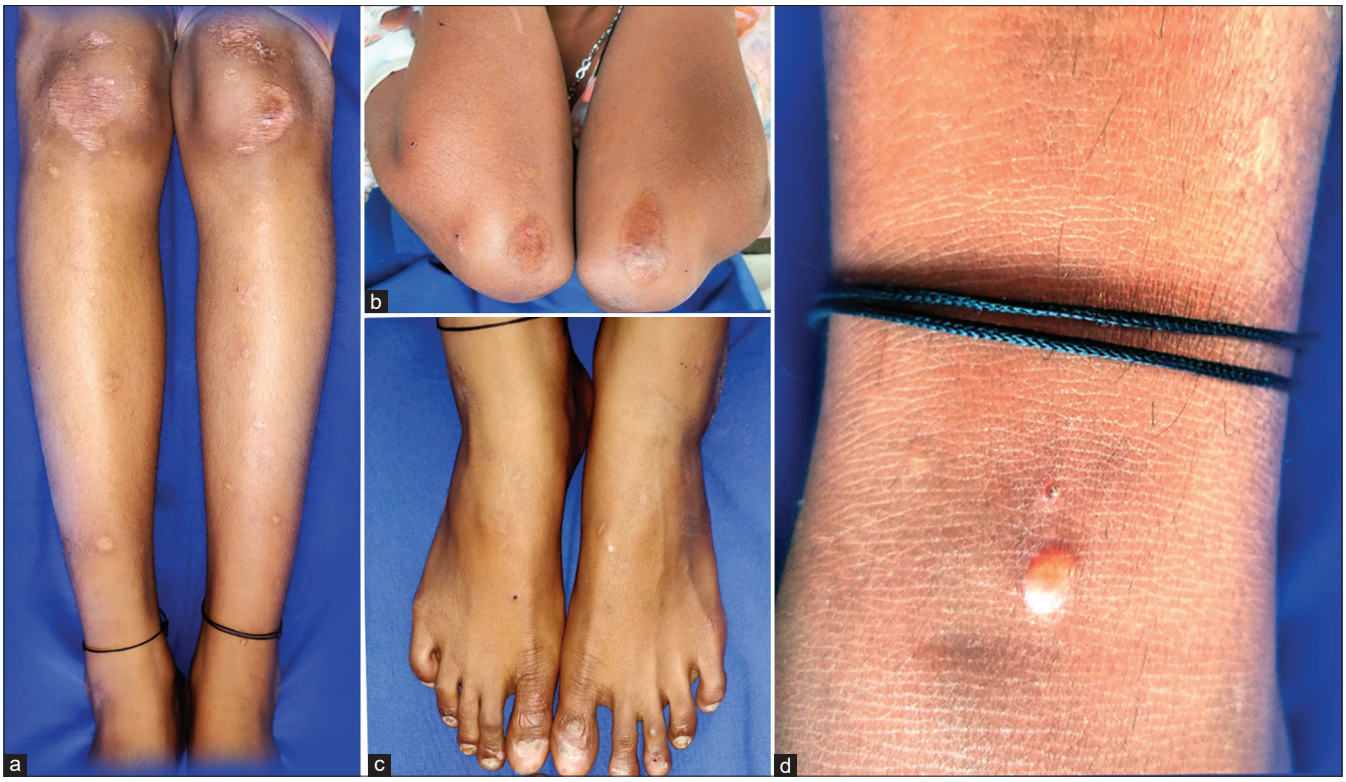

Figure 2: 20-year-old female (younger sister of patient) with (a) Scarring over bilateral knees and (b) elbows. (c) Dystrophic toe nails. (d) Tense clear fluid filled bulla over ankle

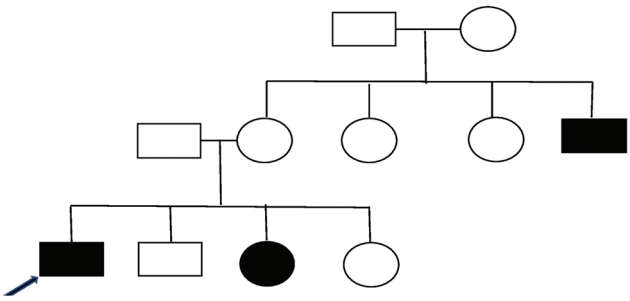

Figure 3: Pedigree chart showing disease across two generations affecting three family members

immunoreactants. Based on clinical and histopathological findings, a final diagnosis of EBP was made.

EB is broadly classified into three types on the basis of splitting in skin—EB simplex (intraepidermal), EB junctional (at the level of lamina lucida) and EB dystrophic (sublamina densa). EBP is a subtype of dystrophic EB.[1] EBP is caused by mutation in *COL7A1* gene. Glycine substitution in the triple helical domain of type VII collagen is implicated.[2] The term EBP was coined by McGrath JC in 1994.[4]

EBP is characterized by hypertrophic and lichenified excoriated papules and nodules with scarring and milia formation. Intact blister is rarely seen. Nail dystrophy is a consistent finding. Albopapuloid lesions can also be seen. Lesions typically occur on shin but elbows, forearms and dorsal aspect of hands can also be affected. Mucosa, flexures, face and hair are usually spared. It is associated with severe pruritus. The exact cause of pruritus in this disease is not known. It could be due to activation of kinin cascade by type VII collagen. Interaction of bradykinin with other mediators might be responsible for severe pruritus.[3] Age of onset is highly variable. It can present at birth or in adulthood. Mode of inheritance can be autosomal dominant or recessive or sporadic.[5]

There is no specific treatment. Management is mostly symptomatic. Pruritus is managed with oral antihistamines and topical or intralesional steroids. Various treatment modalities have been tried with variable success. These include tacrolimus, cyclosporine, naltrexone, thalidomide, etretinate, cryotherapy and surgical treatment in the form of dermabrasion or excision grafting. Genetic counselling and gene therapy are more promising approaches.[6,7]

After thorough literature search on PubMed and MEDLINE database with search terms "epidermolysis bullosa pruriginosa," we found only sixty eight cases have been reported till date and out of that, only eight cases have been documented from India. Although it is very rare condition, it mimics some common skin diseases like prurigo nodularis and hypertrophic lichen planus. Differentiation of EBP from these common diseases is extremely necessary because treatment, prognosis and counseling is quite different as far as EBP is concerned. Thus, a proper clinical examination keeping in mind about this rare entity will help

in early diagnosis, better management and counseling of patients.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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