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SHORT COMMUNICATION

Epidermolysis Bullosa Pruriginosa *Excoriée*: A Deceptive Pruritic Variant in Two Female Patients

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A skin condition that is scratched open, sometimes subtyped by the adjective "*excoriée*", is a feature of a number of skin diseases, such as acne and atopic dermatitis. The consequences of excoriation can be extreme in patients with fragile skin, such as epidermolysis bullosa (EB), in which scratching rapidly leads to wounds. Pruritus can manifest in all forms of EB secondary to wound healing. In a primary form it is seen in patients with dominant dystrophic EB (DDEB) due to glycine substitution mutations in the *COL7A1* gene (1), a subtype known as EB pruriginosa (EBP) (2). EBP occurs in young adults and starts with pruritus of EB lesions, most often in the pretibial area, with compulsive scratching leading to lichenoid, violaceous papules and plaques (2). In patients in whom bulla formation is not prominent, EBP can easily be misdiagnosed as hypertrophic lichen planus or psychogenic pruritus (3). We report here 2 young women affected by EBP and pruritus on the face and upper chest, a presentation not previously reported in the literature. What was remarkable in these 2 unrelated patients is that the itch did not start pretibially, nor did it lead to violaceous papules and plaques. The itch was not secondary to EB lesions, but was localized to intact non-inflamed skin of the face and upper chest. The excoriations, which resulted from compulsive scratching, strongly resembled neurotic artefacts.

CASE REPORTS

Patient 1, a 21-year-old woman with DDEB caused by a heterozygous glycine substitution c.6227G>T, p.Gly2076Val in *COL7A1*. Her EB lesions were localized acrally. The patient was seen for exacerbation of facial atopic dermatitis. Conventional therapy, including topical corticosteroids, calcineurin inhibitors and emollients, as well as cyclosporine, had little or no effect; her pruritus became unbearable, and she reported scratching in her sleep. To attempt to prevent her from scratching, we provided a physical barrier for the patient's face; a custom made perforated silicone mask worn at night. However, the mask worsened the pruritus due to condensation and heat accumulation. The patient was regularly seen with progressive seething wounds, crusts (Fig. 1) and scar tissue formation on her face (not shown). Clinically, the lesions resembled neurotic excoriations. Psychological examination revealed no psychopathological base consistent with self-inflicted compulsive disorders that the patient denied (dermatitis artefacta) or admitted (automutilation).

Patient 2, a 22-year-old woman with DDEB caused by a heterozygous glycine substitution c.6128G>A, p.Gly2043Glu in *COL7A1*, had had *acne excoriée des jeunes filles* in addition to EB since 11 years of age. After using oral contraceptives, her acne lesions (comedones and pustules) disappeared. However, the patient returned to our clinic with severe itch in her face and upper chest. Relief with topical calcineurin inhibitors and corticosteroids, and oral antibiotics was short-lived. Compulsive scratching of clinically healthy, non-inflamed skin after several weeks resulted in erythematous, excoriated papules, scabs and scars in the face (Fig. 1B). Persisting *acne excoriée*

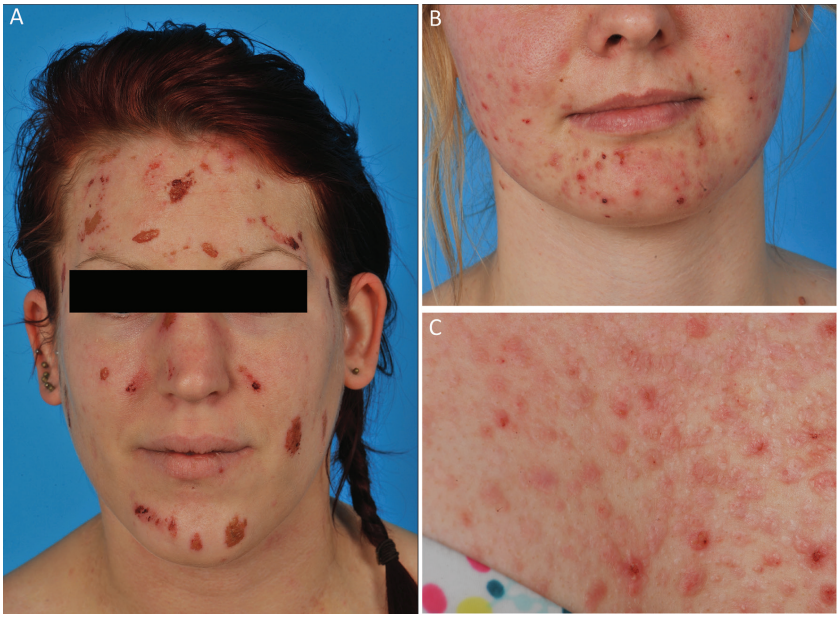

Fig. 1. Both patients seen at initial consultation. (A) Patient 1, with erythematous linear excoriations and scabs with secondary impetiginization localized on the face. (B) Patient 2, with erythematous macules, crusts and scabs localized on the face. (C) Albopapuloid papules and atrophic scars typical for epidermolysis bullosa pruriginosa (EBP) on the upper chest. The patients gave permission to publish the photographs.

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des jeunes filles was ruled out since comedones and pustules were absent. Consultation with a psychologist revealed no neurotic psychopathology. Scratching of her upper chest, but not her face, had evolved to shiny papules and plaques with scarring pathognomonic lesions of EBP (Fig. 1C).

DISCUSSION

Traditional EBP is seen clinically as linear hypertrophic scarring and lichenified plaques, occurring most commonly on the forearms and lower legs (2).

In both cases a diagnosis of atypical EBP with facial involvement, EBP-excoriée, was made. Prior to the development of itch on the face, both patients had had albopapuloid lesions, milia and skin blistering localized acrally on the chest, in the cubital and popliteal fossa, since childhood; all typical of DDEB. The pretibial region was spared. Due to the fragility of the skin, compulsive scratching of the face easily resulted in dramatic excoriations that mimicked neurotic artefacts (3). Atopic, acneiform and psychogenic causes were ruled out in both patients. Suggested therapeutic options for EBP, including the anti-inflammatory agents and immunomodulators already mentioned, were prescribed, but provided little or no relief. Persisting pruritus was observed during the 2-year follow-up. Both young women had periods of relief from itch, not associated with any of the treatments applied; however, the pruritus remains refractory. Lesions on the face have now healed with small atrophic scars that are not typical for EBP.

The aetiology of itch in EBP remains unknown. Cases continue to be reported of DEB patients with glycine substitution mutations in *COL7A1* (4). Internal diseases, mineral deficiencies and associations with atopy have all proven inconclusive. The presence of itch on clinically healthy, non-affected skin by our 2 patients with EBP-excoriée is difficult to explain. It is known that patients with chronic pruritic skin diseases, such as psoriasis and atopic dermatitis, experience feelings of helplessness and increased anxiety, which, in turn, can perpetuate perceived itch (5). One might speculate that such a socially stigmatizing disease as is EB, caused itch to be perceived on non-lesional skin in the face, therefore starting the vicious cycle of itching and scratching. As described by both young women, chronic lesions located on the body were also itchy; however, to a lesser extent than on the face. Further studies examining larger EBP cohorts must be performed in order to consider other aetiologies.

EBP has been classified by the International Forum for the Study of Itch (IFSI) into Group I, pruritus on primary diseased, inflamed skin (6). However, our cases demonstrate that violaceous hypertrophic papules and plaques, as typically seen on the shins in classic EBP, do not always form. We therefore conclude that EBP can manifest with pruritus on primary normal, non-inflamed skin, classified as IFSI Group II, in the form of EBP-excoriée. Consequently, it can evolve into IFSI Group III, pruritus with chronic secondary scratch lesions with typical EBP presentation. The atypical presentation of EBP will undoubtedly challenge physicians even more than classic EBP. EBP should be considered when encountering a patient with clinical characteristics of fragile skin with blistering, nail dystrophy and milia in patients (and family members), which are almost always present. Frequently reported sites of predilection for albopapuloid lesions and hypertrophic scarring of EBP are the shins (3); however, as seen in the excoriée variant, these features may be absent. Unexplained pruritus and excoriations mimicking artefacts should prompt physicians to consider EBP in the differential diagnosis, especially in younger patients. When encountering a patient with such clinical findings, immunofluorescence antigen mapping of type VII collagen may indicate the underlying genetic disorder; however, this may be inconclusive in patients with mild phenotypes. Diagnosis of EBP should always be confirmed by genetic sequencing of *COL7A1* (3).

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