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Case Report

From blisters to nodules: clinical clues to epidermolysis bullosa pruriginosa

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ABSTRACT

Epidermolysis bullosa pruriginosa (EBP) is an uncommon variant of dystrophic epidermolysis bullosa characterised by severe pruritus, prurigo-like nodules, milia, nail dystrophy, and scarring. Delayed diagnosis is common because blistering may become inconspicuous with age. The case report demonstrates a 44-year-old woman with a 10–12-year history of intensely pruritic papules and nodules involving the scalp, trunk, and extremities. Detailed history revealed recurrent trauma-induced blistering since childhood, delayed wound healing, recurrent great toenail shedding, and similar illness in her sister. Examination showed multiple excoriated hyperpigmented papules and nodules, milia, atrophic scarring, and dystrophic toenails. Dermoscopy demonstrated milia-like white clods and scar-like white areas, while histopathology showed acanthosis, compact hyperkeratosis, papillary dermal fibroplasia, and infundibular cyst formation. Clinicopathological correlation established the diagnosis of epidermolysis bullosa pruriginosa. Recognition of childhood blistering, nail dystrophy, milia, scarring, and family history is essential for timely diagnosis and appropriate management.

Keywords: Epidermolysis bullosa pruriginosa, Dystrophic epidermolysis bullosa, Dermoscopy, Nail dystrophy, Milia, COL7A1

INTRODUCTION

Epidermolysis bullosa pruriginosa (EBP) is a rare clinical variant of dystrophic epidermolysis bullosa caused by mutations in the COL7A1 gene encoding type VII collagen, resulting in defective anchoring fibrils at the dermoepidermal junction.[1,2]

Unlike classical dystrophic epidermolysis bullosa, EBP is characterised predominantly by severe pruritus, excoriated papules, nodules, and lichenified plaques, whereas blistering may be mild or limited to childhood.[3] Consequently, patients are frequently misdiagnosed as having prurigonodularis or other chronic pruritic dermatoses, leading to delayed diagnosis.[4]

Recognition of childhood trauma-induced blistering, nail dystrophy, milia, scarring, delayed wound healing, and a positive family history is crucial for diagnosis, particularly where advanced diagnostic facilities are unavailable.[5] We report a patient in whom careful clinical evaluation, dermoscopy, histopathology, and clinicopathological correlation established the diagnosis of EBP after years of symptomatic treatment.

CASE REPORT

44-year-old woman presented with intensely pruritic raised lesions over the scalp, trunk, and extremities for 10–12 years. The lesions initially appeared on the scalp and gradually progressed to involve the trunk and limbs.

Pruritus was severe and associated with repeated scratching, resulting in excoriations and sleep disturbance. She had previously been treated with topical corticosteroids, antihistamines, oral prednisolone (10 mg/day), cyclosporine (50 mg/day), and thalidomide (50 mg twice daily) without sustained improvement.

On detailed questioning, she reported recurrent trauma-induced blistering since early childhood. The blisters healed with scarring and milia formation and were associated with delayed wound healing. She also described recurrent shedding of both great toenails since childhood with dystrophic regrowth. Her elder sister had similar pruritic lesions with nail abnormalities, suggesting an inherited disorder.

Routine laboratory investigations were within normal limits. Tuberculosis screening by interferon-gamma release assay was negative before initiation of immunosuppressive therapy.

Cutaneous examination revealed multiple discrete to coalescent hyperpigmented excoriated papules, nodules, and hyperkeratotic plaques distributed symmetrically over the scalp, trunk, upper limbs, and lower limbs with post-inflammatory hyperpigmentation and atrophic scarring (Figure 1).



Figure 1: Multiple excoriated hyperpigmented papules, nodules, plaques, and post-inflammatory scarring over both lower limbs.

Multiple milia were present over healed lesions. Both great toenails showed dystrophy with partial nail loss (Figure 2). Oral mucosa, palms, and soles were unaffected.

Dermoscopy using a DermLite DL5 (polarized mode, ×10 magnification) demonstrated multiple milia-like white clods, scar-like white structureless areas, focal erosions, and peripheral brown pigmentation, findings consistent with dermal fibrosis and keratin-filled cysts (Figure 3).



Figure 2: Dystrophy and partial loss of both great toenails with periungual scarring.

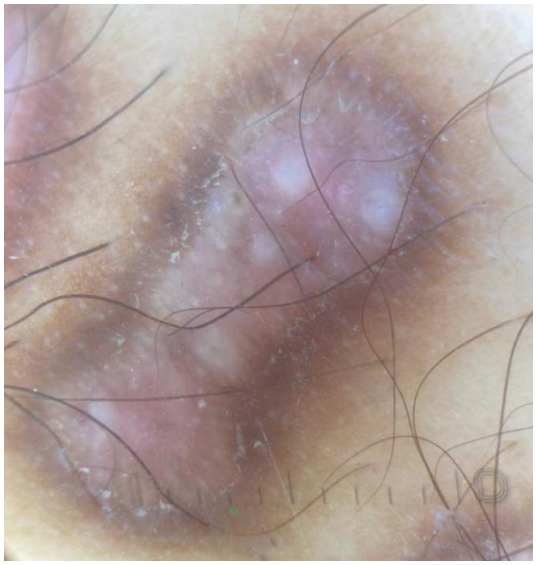


Figure 3: Dermoscopy (DermLite DL5, polarized mode, ×10) showing milia-like white clods, scar-like white areas, focal erosions, and peripheral brown pigmentation.

Histopathological examination revealed compact hyperkeratosis with focal parakeratosis, irregular acanthosis, focal epidermal atrophy, papillary dermal fibroplasia, increased capillary proliferation, superficial lymphoplasmacytic infiltrate with occasional eosinophils, and an infundibular keratin cyst (Figure 4). These findings supported the diagnosis in the appropriate clinical setting.

Based on the history of childhood trauma-induced blistering, delayed wound healing, recurrent nail dystrophy, positive family history, characteristic dermoscopic findings, and supportive histopathology, a

diagnosis of epidermolysis bullosa pruriginosa, a subtype of dystrophic epidermolysis bullosa, was established. Direct immunofluorescence antigen mapping and COL7A1 mutation analysis could not be performed because of financial constraints. The patient was counselled regarding the inherited nature of the disease and initiated on oral tofacitinib 5 mg twice daily, cyclosporine 100 mg/day, topical corticosteroids, intralesional triamcinolone for selected nodules, emollients, and oral doxepin with hydroxyzine for symptomatic relief. At follow-up, she reported reduction in pruritus, improved sleep, flattening of older nodules, and no new blister formation.

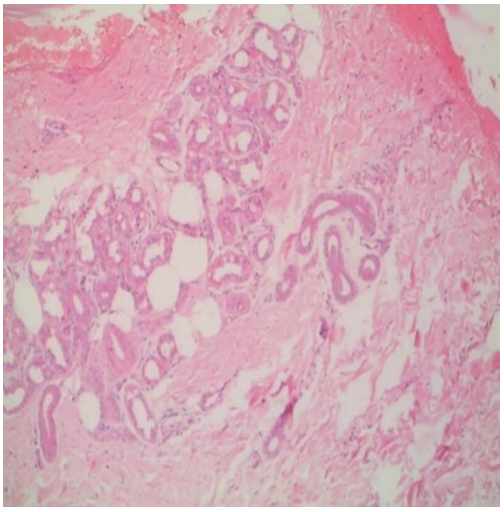


Figure 4: Histopathological examination showing compact hyperkeratosis, irregular acanthosis, papillary dermal fibroplasia, increased capillaries, and infundibular cyst formation (hematoxylin and eosin stain, ×40).

DISCUSSION

Epidermolysis bullosa pruriginosa (EBP) is a rare phenotypic variant of dystrophic epidermolysis bullosa caused by mutations in the COL7A1 gene encoding type VII collagen, resulting in defective anchoring fibrils and increased skin fragility.[1,2] The disorder is characterised by severe pruritus, prurigo-like papules and nodules, lichenification, milia, nail dystrophy, and variable blistering. Because blistering may become less frequent with age, many patients initially present with chronic pruritic nodules rather than active blisters, resulting in delayed diagnosis.[3]

Our patient had several characteristic clinical clues including recurrent trauma-induced blistering since childhood, delayed wound healing, recurrent shedding of both the great toenails, milia formation, atrophic scarring, and a positive family history. These findings strongly suggested an inherited mechanobullous disorder and helped distinguish EBP from prurigonodularis, hypertrophic lichen planus, lichen simplex chronicus, and other chronic pruritic dermatoses.[4]

Dermoscopy was a useful non-invasive adjunct in our patient. The presence of milia-like white clods, scar-like white structureless areas, focal erosions, and peripheral brown pigmentation corresponded to keratin-filled cysts, dermal fibrosis, and chronic excoriation, findings previously described in EBP.[5] Histopathological examination showed compact hyperkeratosis, irregular acanthosis, papillary dermal fibroplasia, superficial inflammatory infiltrate, and infundibular cyst formation. Although these findings are not pathognomonic, they support the diagnosis when interpreted together with the clinical features.[6]

Definitive diagnosis is established by immunofluorescence antigen mapping or molecular demonstration of COL7A1 mutations. However, these investigations are often unavailable or unaffordable in resource-limited settings. In such circumstances, careful clinicopathological correlation remains the cornerstone of diagnosis.[7,8]

Treatment of EBP is challenging and primarily aims to control pruritus, minimise trauma, and improve quality of life. Variable responses have been reported with topical corticosteroids, topical calcineurin inhibitors, cyclosporine, thalidomide, and phototherapy.[9] Increasing evidence suggests that Janus kinase inhibitors may provide benefit in refractory inflammatory and pruritic dermatoses by interrupting cytokine-mediated itch pathways.[10] The patient showed early improvement in pruritus and flattening of nodules following treatment with oral tofacitinib, cyclosporine, topical corticosteroids, intralesional triamcinolone, and supportive skin care.

This case highlights the importance of obtaining a detailed childhood history and carefully examining for nail dystrophy, milia, scarring, and family history in adults presenting with chronic pruritic nodules. Standard dermatology references also emphasise these clinical features as important diagnostic clues in EBP.[11,12]

CONCLUSION

This case highlights the diagnostic challenge posed by epidermolysis bullosa pruriginosa when blistering is subtle and chronic pruritic nodules dominate the clinical presentation. Careful elicitation of childhood trauma-induced blistering, delayed wound healing, nail dystrophy, milia, scarring, and a positive family history, together with dermoscopic and histopathological correlation, enabled the correct diagnosis after years of symptomatic treatment. This report reinforces the importance of maintaining a high index of suspicion for inherited mechanobullous disorders in adults presenting with longstanding pruritic nodules and demonstrates that meticulous clinicopathological evaluation can facilitate accurate diagnosis even when advanced investigations such as immunofluorescence antigen mapping and molecular testing are unavailable. Increased awareness of these distinguishing clinical clues may reduce diagnostic

delay, prevent inappropriate treatment, and improve patient counselling and management.

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