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Successful Treatment of Dystrophic Epidermolysis Bullosa With the JAK1 Inhibitor Abrocitinib

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ABSTRACT

Introduction: JAK inhibitors are increasingly being utilized for the treatment of diseases beyond their nonclassical indications, and the highly selective JAK1 inhibitor abrocitinib may represent a promising therapeutic option for dystrophic epidermolysis bullosa (DEB).
Case Presentation: The first case was a 39-year-old male with DEB in whom conventional therapy was ineffective. After one month of abrocitinib treatment, marked improvement in both pruritus and skin lesions was observed, with no adverse drug reactions during six months of therapy. The second case involved a 46-year-old female DEB patient in whom conventional therapy resulted in mild improvement of skin lesions but no significant relief of pruritus.
Conclusion: The present two cases provide comparative evidence suggesting the superior efficacy of abrocitinib relative to conventional therapies for DEB.

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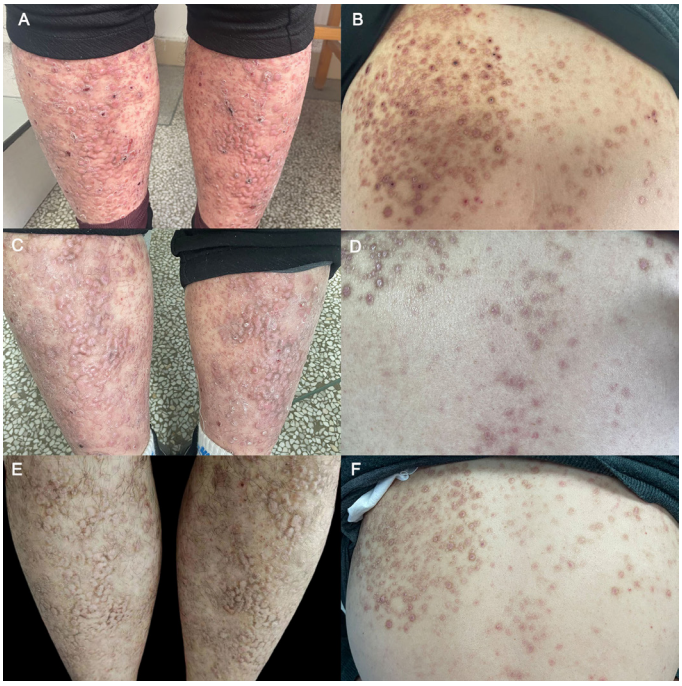
INTRODUCTION

Dystrophic epidermolysis bullosa (DEB), caused by mutations in *COL7A1* is characterized by the development of blisters, bullae, and erosions following minor trauma to the skin and mucous membranes, with healing typically resulting in atrophic scarring and associated nail dystrophy.[1,2] However, no cure for DEB is available; therefore, the clinical care and quality of life of patients need considerable improvement. Currently, JAK inhibitors are being increasingly utilized in the management of noncanonical indications, demonstrating promising clinical efficacy across diverse disease contexts. Here, we report that JAK1 inhibitor abrocitinib may be an effective therapeutic strategy for DEB, supported by case evidence.

CASE PRESENTATION

The first case was a 39-year-old male proband with DEB who initially presented with localized erythema and blisters during adolescence. Subsequently, the rash spread to the trunk and extremities with obvious itching. The proband's mother, grandmother, and aunt manifested similar clinical manifestations. Clinical examination revealed diffuse erythema with papules and nodules distributed across the trunk and extremities, accompanied by scattered hemorrhagic crusts, scales, and hypertrophic scar tissue formation (Figure 1A, B). Genetic testing of the proband and his mother revealed a heterozygous missense variant in *COL7A1*: c.5099G>T (p.Gly1700Val). The patient had previously undergone treatment with oral antihistamines (eg, ebastine, cetirizine, and loratadine), corticosteroids, and cyclosporine, yet the clinical efficacy was unsatisfactory. Due to significant pruritus, the patient was initiated on oral abrocitinib (Pfizer, New York, USA) at a dosage of 100 mg daily. One week later, the symptoms exhibited no significant improvement. Based on prescribing recommendations, the dosage of abrocitinib was increased to 200 mg orally per day. After only one month of treatment, a significant reduction in pruritus and inflammation was observed; the rash improved, and blood crusts sloughed off (Figure 1C, D). The Visual Analog Scale score for pruritus decreased from 8 to 1. The patient reported significant improvements in pruritus and sleep quality. After 3 months, complete resolution of diffuse erythema was achieved with flattening of the rash, although some dark-brown hyperpigmentation and scars remained (Figure 1E, F). During the six-month follow-up period, routine examinations of blood and urine, liver and kidney function, electrolytes, and coagulation parameters were performed periodically, with no abnormalities detected and no adverse drug reactions reported.

FIGURE 1. Clinical course in Case 1 during abrocitinib treatment. The patient at the baseline (A, B), after treatment for 1 month (C, D), and 3 months (E, F).

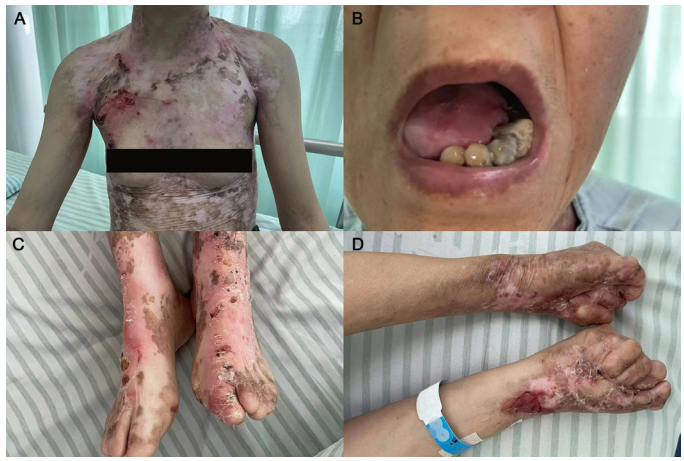


In the second case, a 46-year-old female with DEB had blisters on her right ankle at birth that recurred over many years, accompanied by ulceration, crusting, and oral mucosal involvement. Progressive dysphagia emerged later in her clinical trajectory. The patient had a family history. Clinical examination revealed large, broken red vesicular areas on the head and neck, trunk, and extremities; some were dry and crusted; extensive tooth loss with dental caries; and contracture deformity of the fingers of both hands (Figure 2A-D). Genetic testing revealed one pathogenic variant, c.6187C>T (p.Arg2063Trp), and one suspected pathogenic variant, c.4965C>T (p.(Gly1655=)) in the *COL7A1* gene. The patient exhibited prominent symptoms, including intense pruritus, for which the administration of abrocitinib was recommended as a therapeutic intervention. The patient declined this treatment option due to financial considerations, and was treated with loratadine, ebastine, sulperazone, aggressive debridement and dressing changes, red and blue light irradiation, albumin, and blood transfusion support. The patient exhibited mild regression of cutaneous lesions; however, pruritus remained persistent.

DISCUSSION

Refractory itching is the most distressing disease-related symptom in patients with DEB, resulting in an itch-scratch-blister cycle.[3] Research has shown that patients with DEB exhibit

FIGURE 2. Clinical appearance in Case 2. (A-D) shows widespread skin peeling, blisters, erosions, crusting, with caries and finger deformities throughout the patient.



immune dysregulation affecting both the skin and systemic levels, characterized by increased production of inflammatory cytokines, including IL-1β, IL-2, IL-6, IL-10, interferon-γ, and TNF-α.[4] The JAK-STAT signaling pathway is involved in the signal transduction of multiple cytokines and their receptors, playing a broad role in various biological processes, including immune regulation, inflammatory responses, cell proliferation, and differentiation.[5] Therefore, targeting the JAK-STAT pathway may be a promising approach for the treatment of patients with DEB. In addition to atopic dermatitis, abrocitinib, a highly selective JAK1 inhibitor, has been shown to be effective in dozens of other inflammatory diseases, such as lichen sclerosus, alopecia areata, prurigo nodularis, and hidradenitis suppurativa.[6]

Recent studies have underscored the significant efficacy of JAK inhibitors in alleviating pruritus associated with DEB. To date, a total of 30 DEB patients, aged 12 to 71 years, have received JAK inhibitor therapy, including 2 treated with tofacitinib, 7 with baricitinib, 15 with upadacitinib, and 6 with abrocitinib.[7-17] The majority of these patients exhibited substantial clinical improvement in both cutaneous lesions and pruritus, with only one patient displaying a lack of therapeutic response. Regarding safety, no serious adverse reactions were reported in any of the patients. Therefore, the treatment of a DEB patient without atopic dermatitis using 200 mg of abrocitinib was effective, affirming the drug's effectiveness and feasibility as a therapeutic option. However, the study's limitations include a small sample size and the absence of a comparator treatment group.

CONCLUSION

DEB is a persistent, relapsing disease that severely affects the quality of life. The findings of this study suggest that abrocitinib may represent a viable therapeutic option for DEB, demonstrating pronounced efficacy in pruritus relief compared

to conventional treatments. However, larger-scale studies are warranted to fully elucidate the efficacy and safety profile of JAK inhibitors in DEB.

DISCLOSURES

The authors declare that the research was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

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