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Epidermolytic Ichthyosis With Palmoplantar Sparing in a Child: A Case Report of a Rare Keratinization Disorder

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Abstract

Epidermolytic ichthyosis (EI) is a rare, inherited keratinopathic disorder characterized by marked skin peeling shortly after birth that is followed by pronounced hyperkeratosis and the persistence of skin fragility. Here, we report a case of epidermolytic ichthyosis in a four-year-old female child who presented with generalized hyperkeratosis of the skin and skin fragility with sparing of the palms and soles. Histopathological examination revealed orthohyperkeratosis, hypergranulosis with large keratohyaline granules, and vacuolated keratinocytes, supporting the diagnosis. However, molecular testing was unavailable. This case highlights the importance of recognizing the characteristic clinical features of epidermolytic ichthyosis and correlating them with histopathological findings, particularly when genetic testing is unavailable.

Categories: Dermatology
Keywords: congenital ichthyosis, epidermolytic ichthyosis, keratin, keratin genes, rare genetic disorder

Introduction

Epidermolytic ichthyosis (EI), also termed epidermolytic hyperkeratosis (EHK) and previously termed bullous congenital ichthyosiform erythroderma (BCIE), is a rare autosomal dominant disorder resulting from mutations in genes encoding keratins 1 and 10 [1,2]. It has an estimated worldwide prevalence of one in 200000, with both sexes being affected equally. At least half of all cases arise sporadically due to new mutations [3]. Clinically, EI presents with generalized erythroderma at birth. The fragility of the skin results in blisters, peeling, and widespread areas of denuded skin. Over time, there is a decrease in blistering and the development of marked hyperkeratosis. Occasionally, skin fragility may persist, and patients shed large plates of the superficial epidermis periodically [4]. Palmoplantar involvement is associated with mutation in keratin 1, whereas keratin 10 mutation is associated with the sparing of the palmoplantar region [5].

The diagnosis is based on clinical and histopathological findings, while genetic analysis is regarded as the gold standard for the diagnosis of the condition [1]. Essential components of treatment include supportive care, the maintenance of hydration, and the prevention of infection [4,6]. Retinoids are used to prevent hyperkeratinization and superinfection in severe cases, being especially effective in patients with *KRT10* mutations [7,8]. The authors report a case of EI with palmoplantar sparing in a four-year-old child with characteristic clinical and histopathological phenotype. This case is presented to emphasize the importance of recognizing the typical clinical evolution and correlating it with histopathological findings in a resource-limited setting, particularly when molecular diagnostic testing is unavailable.

Case Presentation

A four-year-old female child was brought to the dermatology outpatient department by her grandfather regarding complaints of brown-to-gray, dry, hard scales over the entire body for four years. It was associated with mild-to-moderate itching and an occasional foul smell from her body. Additionally, she experienced occasional blisters, especially during summers, that ruptured spontaneously, leaving behind dry scales.

The patient was born at term via normal vaginal delivery and presented with redness and peeling of her skin on the first day of life. She was taken to a government hospital in Nepalgunj, where she was admitted for a week and discharged with some topical medicaments that her guardians are unable to recall now. By her seventh day of life, the peeling and redness had improved. Over the subsequent weeks, she developed brown-to-gray, dry, scaly lesions on her knees and elbows, which gradually progressed to involve the entire body. However, the child showed no evidence of intellectual disability, and no similar lesions were observed in other family members. Furthermore, there was no history of consanguinity within the immediate family.

Cutaneous examination revealed multiple brown-to-gray, scaly plaques, with a dry, rough, hyperkeratotic surface, of variable sizes over the entire body (Figure *1A*, *1B*), with a rippled or corrugated appearance over the extensor surfaces, particularly the bilateral knees and elbows (Figure *2*). There were multiple foci of

erythema that were discretely distributed surrounding some of the lesions. The scalp exhibited dry, grayish scaling. The palms and soles were spared. No abnormalities were observed in the nails or in the conjunctival, oral, or genital mucosa. No significant abnormalities were detected on systemic examination.



FIGURE 1: (A) Generalized brown, dry, hyperkeratotic scales on the face, trunk, and bilateral upper limbs. (B) Generalized brown, dry scales with areas of erythema on the nape of the neck and the back of the trunk



FIGURE 2: Hyperkeratotic scales on bilateral lower limbs with rippled appearance on the knees

Baseline investigations, including complete blood count, liver function tests, renal function tests, and serological screening for HIV, hepatitis B surface antigen (HBsAg), and hepatitis C virus (anti-HCV), were performed prior to skin biopsy. All results were within normal limits.

A punch biopsy was obtained from the left lower leg, adjacent to the knee. Histopathological examination revealed orthohyperkeratosis and the expansion of the granular layer with large keratohyaline granules. Stratum spinosum showed vacuolated keratinocytes, accompanied by sparse, mild intercellular spongiosis. Upper dermis showed mild-to-moderate perivascular and interstitial lymphocytic infiltration. Lower dermis and subcutaneous tissues revealed no remarkable findings (Figure *3A*, *3B*). The histopathological findings were thus compatible with the diagnosis of epidermolytic ichthyosis. Genetic testing could not be performed due to the unavailability of necessary testing facilities.

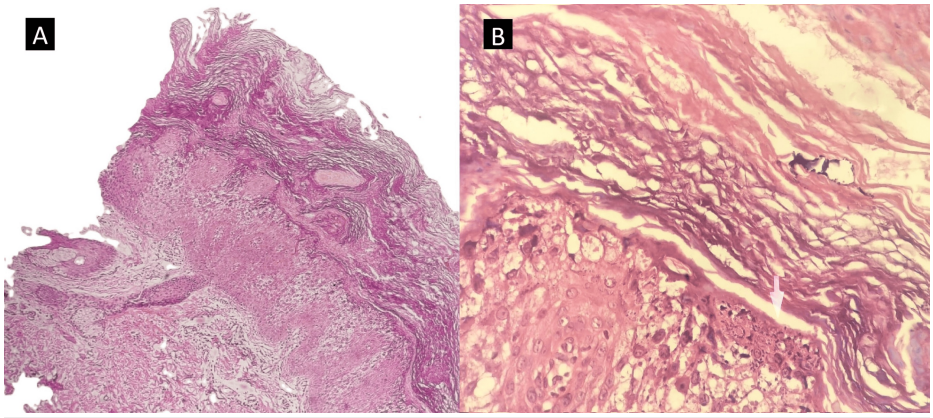


FIGURE 3: (A) Photomicrograph (10×) showing orthohyperkeratosis, hypergranulosis, stratum spinosum with vacuolated keratinocytes and spongiosis, and mild-to-moderate perivascular and interstitial lymphocytic infiltration in the upper dermis. (B) Photomicrograph (high-power view) showing hyperkeratosis, hypergranulosis with prominent keratohyaline granules (arrow), and spongiosis

The guardians of the patient were counseled regarding the nature of the condition and its clinical course. They were also advised regarding the appropriate care and measures required in this condition, including the liberal use of emollients and moisturizers. The patient was started on topical tazarotene 0.1% and a topical keratolytic preparation with a combination of urea and lactic acid, both applied twice a day, along with oral vitamin A supplementation 50000 international units twice weekly for two weeks. Careful monitoring and follow-up were planned to assess treatment response.

When the patient came for follow-up a month later, she was clinically evaluated for treatment response and tolerance. A slight improvement in the lesions was noted. The scaling and erythema had reduced; however, prominent scales still persisted over the extensor surfaces of the knees and elbows (Figure *4A*, *4B*). The patient was tolerating the treatments well, and no adverse effects were reported or observed. She was continued on the same topical treatment because of the persistent lesion, with further follow-up advised after one month.

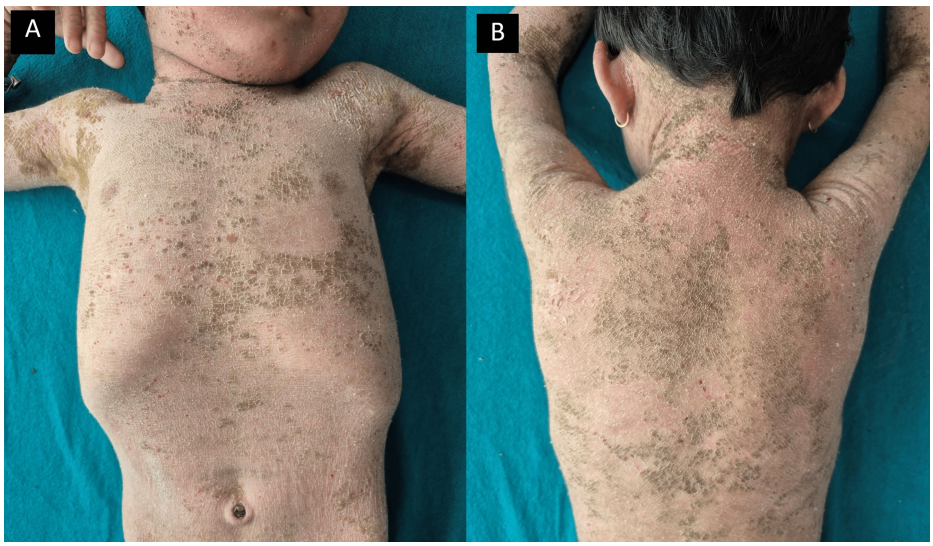


FIGURE 4: (A) Slightly reduced brown, dry, hyperkeratotic scales on the lower face, trunk, and bilateral axillae. (B) Slightly reduced brown, dry, hyperkeratotic scales on the nape of the neck and the back of the trunk

Written informed consent was obtained from the patient's grandfather, who served as the child's guardian,

for the publication of this case report and the accompanying clinical photographs.

Discussion

Epidermolytic ichthyosis (EI) is a rare autosomal dominant keratinopathic disorder caused by mutations in *KRT1* or *KRT10*, which encode keratins expressed in the suprabasal layers of the epidermis. The disorder has an estimated global prevalence of one in 200000 live births and affects both sexes equally [1,2]. Approximately 50% of the cases occur sporadically due to de novo mutations, which might explain the absence of family history in our patient [3].

This disorder typically manifests shortly after birth with generalized erythroderma, blistering, and skin fragility. Over time, blistering subsides, and hyperkeratosis becomes the dominant feature, particularly over flexural and extensor surfaces. Our patient demonstrated this classic evolution, with neonatal erythema and peeling that gradually progressed to generalized hyperkeratotic, scaly plaques with a rippled appearance over the elbows and knees. Her palms and soles were spared, a phenotype that has been described in association with *KRT10*-related disease.

Complications include the increased risk of sepsis, dehydration, and death in neonates. There can be significant disfigurement and pungent body odors, which can lead to psychological and social distress [4]. Our patient also had a history of occasional foul body odor, but no significant psychological concerns were reported.

Differential diagnoses vary according to the age of presentation. In early infancy, it can include disorders with erythroderma, bulla formation, or exfoliation such as epidermolysis bullosa, Netherton syndrome, collodion baby, and other forms of congenital ichthyosis. Meticulous clinical evaluation and histopathology are essential to distinguish EI from these conditions.

Histopathological features of EI include compact orthokeratotic hyperkeratosis, markedly thickened granular layer with coarse keratohyaline granules, and variously sized intracellular vacuolization in the upper stratum spinosum and in the stratum granulosum [9]. These findings correspond well with the histopathological findings in our patient. The gold standard diagnostic technique is genetic mutation analysis for keratin defects with multigene-panel screening [1]. The unavailability of tests for genetic analysis remains a limitation of this study.

As there is no cure, the management of EI remains largely supportive. The regular use of emollients and moisturizers is essential to maintain skin hydration and reduce scaling. Keratolytic agents and topical retinoids, such as tazarotene, may help to reduce hyperkeratosis in localized areas [10]. In severe cases, oral retinoids such as acitretin may dramatically improve hyperkeratosis but can paradoxically increase skin fragility and exacerbate blistering. Therefore, it is recommended to use low initial doses and then gradually increase with careful monitoring [11]. This patient was started on topical tazarotene 0.1%, topical emollients, a urea- and lactic acid-containing keratinolytic, and oral vitamin A supplementation, with slight clinical improvement on follow-up after one month. Topical retinoids, particularly tazarotene, have been used in the management of congenital ichthyoses to reduce hyperkeratosis and scaling. The pediatric consensus recommendations by Zaenglein et al. recognize the role of topical retinoids, including tazarotene, in selected disorders of cornification, including epidermolytic ichthyosis [12]. Although the use of tazarotene in children below the approved age is off-label, its use in this patient was considered on the basis of the available clinical literature and the extent of hyperkeratosis. As acitretin was not available in this region of Nepal, the patient was started on oral vitamin A supplementation instead. The dosing regimen was guided by a previously published case report of epidermolytic ichthyosis by Shah and Padhiar [13]. However, evidence supporting this approach remains limited to case-level observations, and its efficacy and safety cannot be established from these reports alone.

The main limitation of our study is the absence of molecular genetic testing, which limited the confirmation of the underlying *KRT1* or *KRT10* mutation, and the lack of documentation of the patient's weight at treatment initiation. Consequently, a retrospective calculation of the weight-based oral vitamin A dose was not possible. Nevertheless, the typical clinical and histopathological findings allowed a clinicopathological diagnosis of EI. This highlights the practical importance of the recognition of these features in settings where molecular diagnostic facilities may not be available. To the knowledge of the authors, the published documentation of epidermolytic ichthyosis from Nepal appears to be limited. This case, therefore, contributes to the limited published clinical documentation of this condition from Nepal.

Conclusions

Epidermolytic ichthyosis is a rare inherited keratinopathic disorder that is characterized by neonatal skin fragility and peeling that progresses to generalized hyperkeratosis and scaling. Genetic testing remains the gold standard; however, the recognition of its clinical evolution and histopathological findings can aid in the diagnosis when molecular testing facilities are unavailable.

This case demonstrates the characteristic clinical evolution and histopathological features of EI with

palmoplantar sparing in a child and contributes to the limited clinical documentation of the disorder from Nepal. A careful evaluation of clinical and histopathological findings and a close correlation between them allowed an appropriate clinical diagnosis and guided supportive management in the absence of molecular testing.

Additional Information

Author Contributions

All authors have reviewed the final version to be published and agreed to be accountable for all aspects of the work.

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Supervision: Alisha Aryal

Disclosures

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