CASE REPORT

Epidermolytic ichthyosis sparing the palms and soles: A rare case report

Majed Aldayhum,[1] Mousa Alshehri,[2] Norah Abu Mughaedh,[2] Areej Alasiri,[2] Noah Alghanemy,[2] Hamza Alshehri,[2] Fawziah Alfaifi,[2]

[1]MD, Armed Forces Hospital, Southern Region, KSA
[2]MD, Aseer Central Hospital, KSA

ABSTRACT

Background: Epidermolytic ichthyosis (EI), formerly known as epidermolytic hyperkeratosis (EHK) or bullous congenital ichthyosiform erythroderma (bullous CIE), is a form of congenital ichthyosis. It is inherited in an autosomal dominant fashion, with about 50% of cases representing spontaneous mutations. Epidermolytic ichthyosis presents at birth with erythroderma, blisters, and erosions and evolves over time into varying degrees of hyperkeratosis. We present a rare case of generalized epidermolytic icthyosis with sparing of the palms, and soles.

Case: This case report describes a 12-year-old Saudi boy who presented with generalized hyperkeratotic brownish linear plaques following Blaschko's lines, sparing the palms and soles. The patient had a history of skin lesions since birth, initially presenting as generalized body erythema and scattered blisters triggered by minor trauma. Over time, these symptoms decreased, but hyperkeratotic plaques began to develop, primarily affecting flexural and extensor areas and later spreading to involve the trunk and extremities. The patient exhibited normal hair, teeth, and nails, with no systemic symptoms or convulsions. A skin biopsy revealed characteristic histopathological findings. Extensive laboratory investigations yielded normal results. Treatment with acitretin and topical urea creams was initiated.

Conclusion: This case report highlights a unique presentation of epidermolytic ichthyosis in a 12-year-old boy, characterized by generalized hyperkeratotic plaques following Blaschko's lines, with sparing of the palms and soles. The absence of systemic involvement, coupled with normal laboratory findings, underscores the localized nature of this variant.

INTRODUCTION

Skin thickening, blistering, and hyperkeratotic plaques are symptoms of epidermolytic ichthyosis, a rare hereditary disorder.[1] Most often, mutations in the KRT1 or KRT10 genes, which code for epidermal structure-maintaining keratins, produce this genetic illness. Despite extensive hyperkeratosis, epidermolytic ichthyosis seldom spares the palms and soles.[2, 3] Known as "Epidermolytic Ichthyosis sparing the palms and soles," this unusual hereditary skin condition presents a unique treatment challenge and illuminates its complex biology.[4] This report describes the clinical symptoms, diagnostic workup, and management of a 12 year old Saudi boy with this uncommon symptomatology. This example illustrates the clinical diversity and complexity of epidermolytic ichthyosis, which must be carefully diagnosed and treated.

CASE REPORT

A 12-year-old Saudi boy presented with generalized hyperkeratotic brownish linear plaques involving the trunk, upper, and lower extremities,

Correspondence: *Dr. Majed Aldayhum, Armed Forces Hospital, Southern Region, KSA*

following Blaschko's lines (Fig. 1). Notably, the palms and soles were spared. Hair, teeth, and nails appeared normal. The skin lesions had been present since birth, initially manifesting as generalized body erythema with scattered blisters triggered by minor trauma. By the age of one, erythema and blister formation had diminished, replaced by hyperkeratotic plaques, mainly affecting both flexural and extensor areas. As the patient grew older, the hyperkeratotic plaques became more generalized and spread to involve the trunk and extremities. The patient exhibited good school performance and had no history of convulsions or systemic symptoms. There was no family history of similar skin lesions.

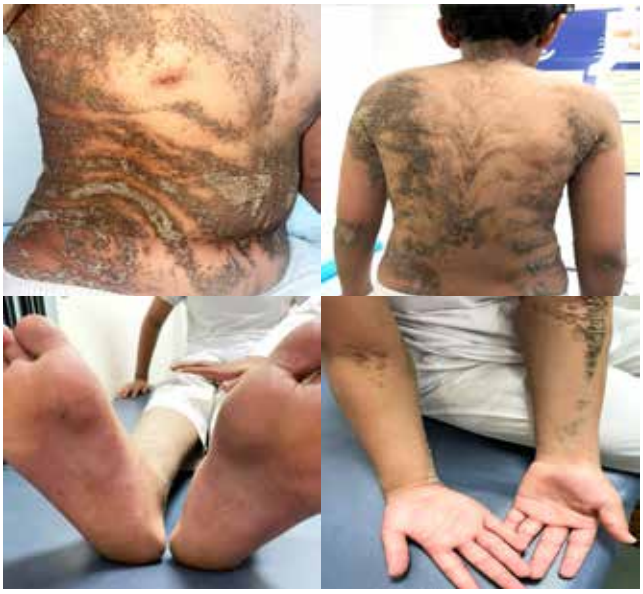

Fig. 1 Generalized hyperkerat otic brownish linear plaques involving the trunk, upper, and lower extremities, following Blaschko's lines

A skin biopsy was performed, revealing histopathological features consistent with the clinical presentation (Fig. 2). The biopsy demonstrated hyperkeratosis, acanthosis, granular degeneration in the superficial epidermis, vacuolar degeneration in the granular and spinous layers, and edematous keratinocytes. The vacuolated keratinocytes exhibited clear and pale cytoplasm with variable-sized nuclei.

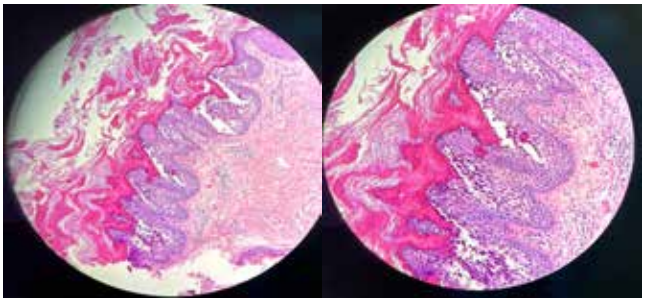

Fig. 2 Biopsy showing hyperkeratosis, acanthosis, granular degeneration in the superficial epidermis, vacuolar degeneration in the granular and spinous layers, and edematous keratinocytes.

Laboratory Investigations and Treatment

To assess any potential systemic involvement, a comprehensive set of laboratory tests were conducted, including liver function tests, renal function tests, lipid profile, and complete blood count. All results were within normal ranges, indicating no systemic abnormalities. The patient was started on acitretin tablets at a dose of 10 mg per day, in addition to topical urea creams, as part of the therapeutic management plan.

DISCUSSION

Epidermolytic ichthyosis (EI), also known as epidermolytic hyperkeratosis (EHK) or bullous congenital ichthyosiform erythroderma (bullous CIE), a rare genetic illness, with several symptoms. About half of this congenital condition is caused by random gene mutations. Heterozygous mutations in the KRT1 or KRT10 genes cause EI. These protein structural alterations cause epidermal keratins to misalign and fail to form intermediate filaments. Blisters form and spread when the skin's protective barrier breaks. Keratin overproduction results from the body's healing efforts.[1-3]

Palmoplantar keratoderma, which thickens the palms and soles, is typically connected to KRT1

mutations. KRT10 mutations may potentially cause palmoplantar keratoderma in rare circumstances. Only 1 in 200,000–300,000 people have epidermolytic ichthyosis. Race and gender do not seem to affect this disease.[4,5]

Epidermolytic ichthyosis in infants causes widespread surface blisters that rupture to reveal naked skin. In the first few months of life, hyperkeratosis becomes the norm and skin fragility lessens, with blistering and erosions becoming rare. Depending on the severity, hyperkeratosis may cause corrugated or cardboard-like skin.[6,7] Often, epidermolytic ichthyosis does not affect hair, nails, or teeth. Eyelid ectropion eyelids turning out is rare. These patients typically have odorous, superficial bacterial infections. Rarely, epidermolytic ichthyosis is associated with hypocalcemic rickets with or without vitamin D resistance, platelike osteoma cutis, and localized vulvar involvement.[8-10]

Epidermolytic ichthyosis is generally diagnosed without routine laboratory tests, unless it is required to monitor medication or investigate infections. Genetic tests may be done on buccal swabs or blood samples, and if a mutation is detected, families can get mutant-specific testing or prenatal diagnosis.[7] Epidermolytic ichthyosis exhibits several histological features. In hematoxylin and eosin staining of the top epidermis, hyperkeratosis, a thicker granular layer, coarse keratohyaline granules, and vacuolar degeneration are common Variable degrees of dyskeratosis are possible. When present across the horizontal epidermis, these distinct traits suggest mosaic epidermolytic ichthyosis rather than generalized disease.[8]

Electron microscopy shows large, round to oval, thick keratin tonofilaments in the basal epidermal layers, validating the diagnosis. Correct diagnosis is essential for treating epidermolytic ichthyosis and providing patients and families with relevant information and counseling. Prenatal diagnostics and genetic counseling help with this issue. Epidermolytic ichthyosis therapy reduces hyperkeratosis and relieves symptoms, but there is no cure.[9]

Newborns with denuded skin are at risk of infection, subsequent sepsis, and electrolyte problems from epidermolytic ichthyosis. They must be placed in the NICU for monitoring and care. These infants have sensitive skin, therefore caregivers should be careful.[10]

Topical emollients and keratolytics like lactic acid, alpha-hydroxy acid, or urea treat epidermolytic ichthyosis in older children and adults. Emollients based on glycerin are also utilized for dryness and discomfort. Topical and oral retinoids reduce hyperkeratosis and improve skin quality. Due to scale-induced bacterial overgrowth, mild antibacterial soaps or bleach baths may be used to treat associated illnesses.[1,7,10]

Our case had an unusual epidermolytic ichthyosis with hyperkeratotic linear plaques similar to Blaschko's lines but avoiding the palms and soles. This instance expands epidermolytic ichthyosis clinical variability research. The clinical history, physical traits, and histological results of this patient suggest an uncommon type, making the diagnosis challenging. Laboratory studies under normal settings show little systemic involvement, indicating the condition's restricted scope. Using acitretin and topical urea creams to decrease hyperkeratotic plaques may enhance quality of life. The treatment reaction of this variant will illuminate the condition. This example shows the necessity of recognizing and treating odd skin problems. More rare variant research

and clinical data are needed to improve patient treatment.

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