Title
Unilateral hyperkeratotic plaques along blaschko lines

Journal
Dermatology Online Journal, 21(7)

Authors
Kumar, Piyush
Debbarman, Panchami
RK, Shilpa

Publication Date
2015

DOI
10.5070/D3217028137

Volume 21 Number 7
July 2015

Photo vignette

Unilateral hyperkeratotic plaques along blaschko lines

Piyush Kumar MD[1], Panchami Debbarman MD[2], Shilpa RK MBBS DVD[2]

Dermatology Online Journal 21 (7): 16

[1]Katihar Medical College and Hospital, India

[2]Consultant Dermatologist, India

Correspondence:

Piyush Kumar
Asst. Prof, Dermatology
Katihar Medical College and Hospital
Karim Bagh, Katihar
Bihar- 854105
Phone: +91 8987280259
docpiyush99@gmail.com
docpiyush099@gmail.com

Abstract

Epidermolytic ichthyosis (or epidermolytic hyperkeratosis) classically presents with erythroderma and increased fragility (blistering) at birth or soon thereafter. In later life, erythroderma and blistering improve gradually and the clinical picture is dominated by hyperkeratotic plaques in flexures and around joints. Linear epidermolytic hyperkeratosis is a unique, uncommon clinical variant and the absence of erythroderma and blistering are its hallmark. Linear lesions may be localized or generalized andunilateral or bilateral. Herein we report a 6-year-old girl with unilateral epidermolytic ichthyosis.

Keywords: Epidermolytic ichthyosis, Bullous congenital ichthyosiform erythroderma, linear Epidermolytic hyperkeratosis

Case synopsis

A 6-year-old girl, born of a non-consanguineous marriage, presented with mildly itchy widespread hyperkeratotic plaques in a linear distribution on the right side of the body since 3 months of age. There was no history of widespread erythema or development of bullous lesions at birth or thereafter. The plaques had been increasing in thickness and pigmentation with time. The girl was otherwise healthy and no other family members were affected by a similar skin condition. On examination, multiple linear hyperkeratotic plaques in a Blaschkoid distribution were noted on the right side of the body, with accentuation of lesions in the flexures and around joints. The surface of these lesions was notable for hyperkeratotic ridges arranged in a parallel fashion. Scalp, face, palms, soles, and mucosae were spared (Figures 1a and 1b). The sample for histopathology was collected from lesions on the abdomen and knee. The findings from both samples were similar and showed compact orthohyperkeratosis, coarse keratohyaline granules, perinuclear vacuolization of cells of granular layers, and a mild mononuclear infiltration in the upper dermis. However, the findings fromthe abdominal sample (Figures 2a and 2b) were more subtle than those from the knee (Figures 3a and 3b). Considering clinical presentation and histopathological findings, the case was diagnosed as unilateral epidermolytic ichthyosis. The parents were examined for verrucous epidermal nevus like lesions, which were notably absent.

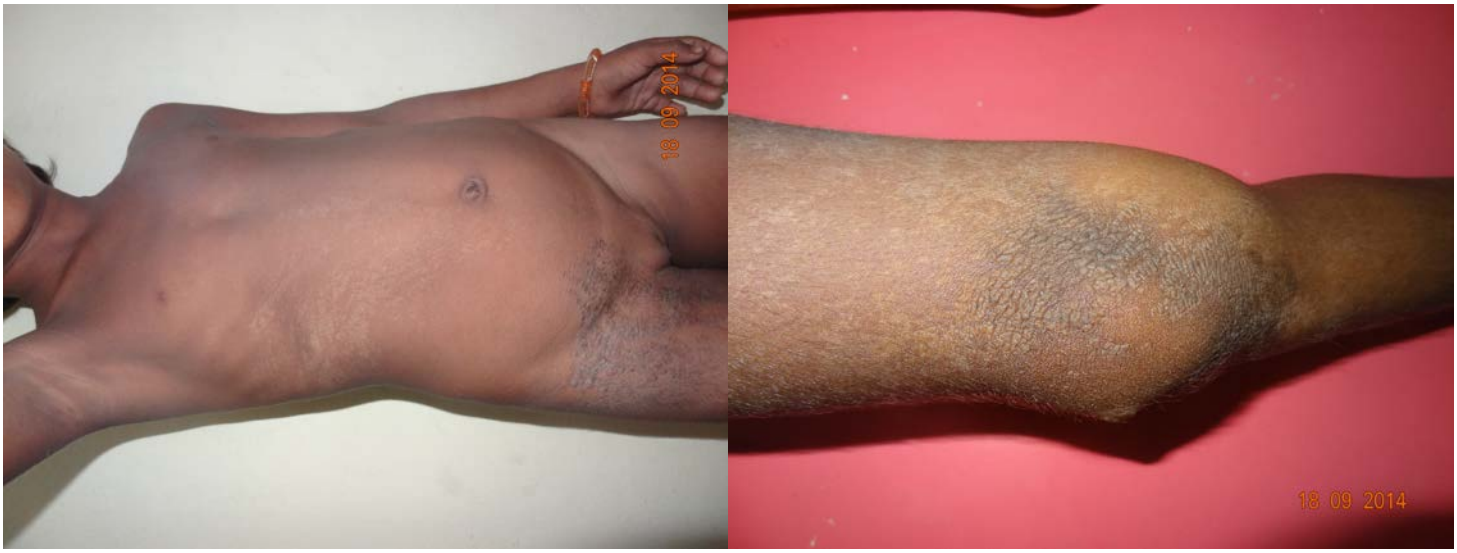

Figure 1. Hyperkeratotic plaques in Blaschkoid pattern on right side of body with accentuation of lesions in flexures (1a). Close up of lesions on knee (1b)

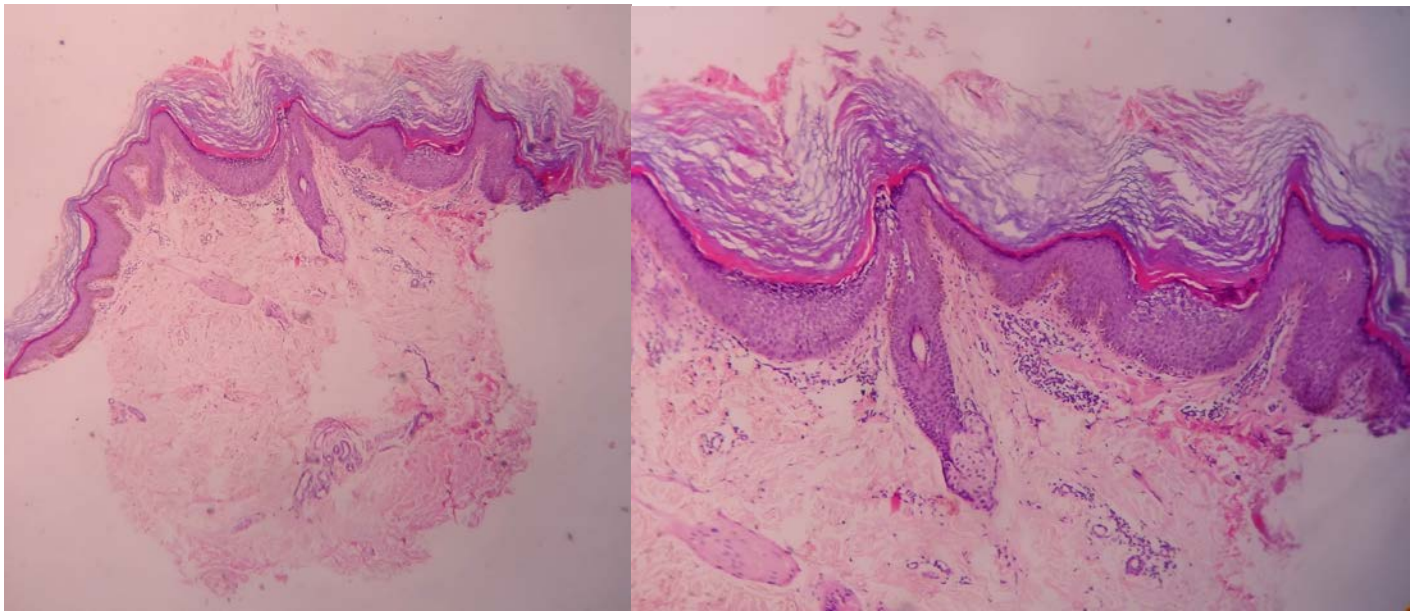

Figure 2. Histopathology from abdominal lesion showed compact hyperkeratosis, granular degeneration and mild mononuclear cell infiltration in upper dermis. (a) (H&E x 40). Higher magnification (b) (H&E x 100)

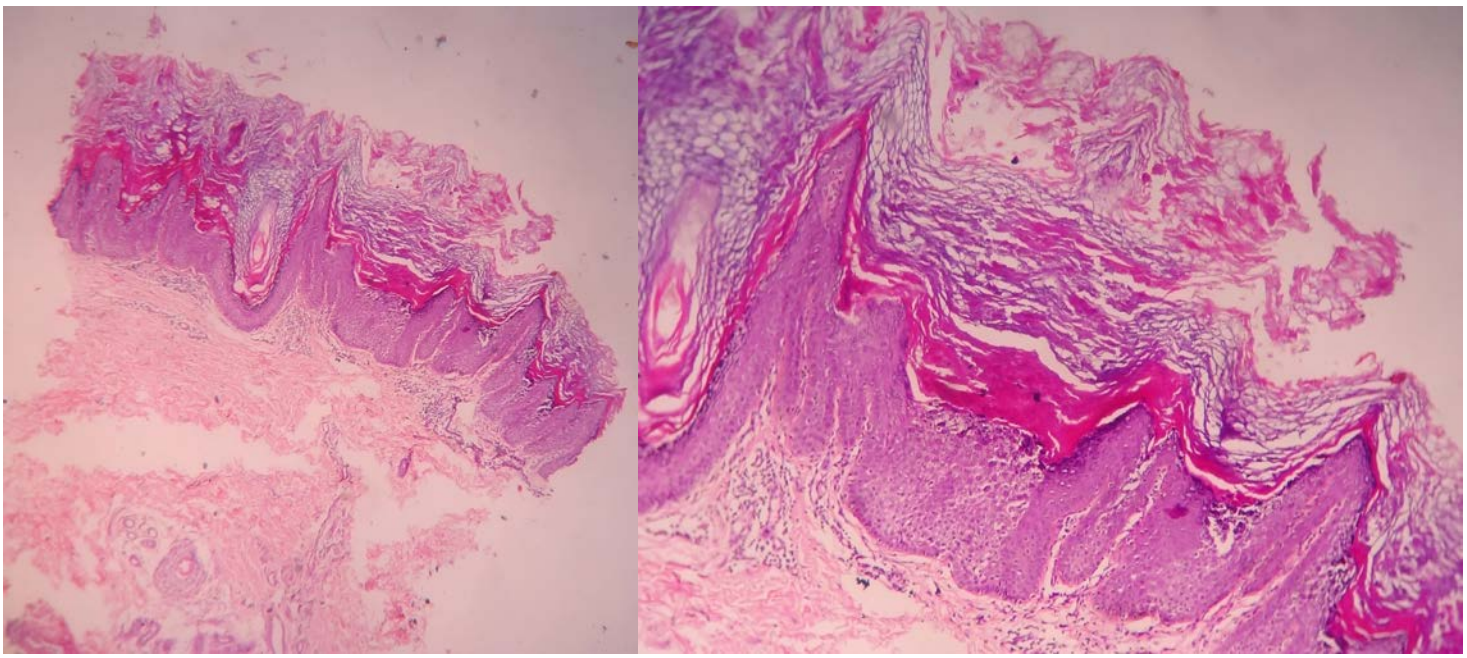

Figure 3. Histopathology from knee lesions showed pronounced hyperkeratosis, coarse keratohyaline granules, and perinuclear vacuolization in upper epidermal layers. (a) (H&E x 40). Higher magnification (b) (H&E x 100)

Discussion

Epidermolytic ichthyosis, also known as bullous congenital ichthyosiform erythroderma of Brocq (BCIE), is a rare disorder of keratinization and results from mutations affecting keratin genes KRT1 or KRT10. The term “epidermolytic hyperkeratosis” is often used synonymously for this condition, unless stated otherwise [1, 2, 3]. It may be sporadic or may be transmitted in an autosomal dominant manner. Classically, it presents with erythroderma and fragile skin (manifesting as flaccid blisters, peeling, and erosions) at birth. As the baby grows older, erythroderma and blistering improve gradually and verrucous plaques appear in flexures and around joints. The surface of these lesions is characterized by parallel hyperkeratotic ridges, giving an appearance of corrugated cardboard [4]. Two clinical subtypes have been described- one involving the palms and soles (PS type) and the other sparing palms and soles (NPS type). It is well documented that patients with K1 mutations are more likely to have PS type of BCIE and palm/sole involvement manifests as painful fissures and contractures [1, 2].

Annular epidermolytic ichthyosis and linear BCIE are uncommon clinical variants. Linear BCIE probably results from a postzygotic mutation and thus represents genetic mosaicism [2, 3]. It presents as linear verrucous plaques along Blaschko lines and may be unilateral or bilateral, and localized or generalized in distribution. Of note, linear BCIE variants are characterized by the absence of erythroderma and blistering, unlike classical BCIE, as evident in our case [5, 6]. Clinically, it is indistinguishable from linear verrucous epidermal nevus (VEN). However, accentuation of lesions in flexures and around joints with a “corrugated cardboard appearance” is a very important clinical clue for differentiating linear BCIE from linear VEN [3, 4]. In doubtful cases, histopathology is of diagnostic importance as BCIE is characterized by coarse keratohyaline granules and perinuclear vacuolization of keratinocytes of granular layers.

References

1. Kurosawa M1, Takagi A, Tamakoshi A et al. Epidemiology and clinical characteristics of bullous congenital ichthyosiformerythroderma (keratinolyticichthyosis) in Japan: results from a nationwide survey. J Am AcadDermatol 2013;68(2):278-83. [PMID: 23182068]
2. Mendes MS, Kouzak SS, Aquino TA et al. Mosaic epidermolyticichthyosis- case report. An Bras Dermatol 2013;88(6 Suppl 1):116-9. [PMID: 24346896]
3. Kumar P, Kumar R, Mandal RK et al. Systematized linear epidermolytic hyperkeratosis. Dermatol Online J 2014;20(1):21248. [PMID: 24456951]
4. Mondal AK, Kumar P, Mondal A. Bullous Congenital IchthyosiformErythroderma. Indian Pediatr 2011;48: 968. [PMID: 22253153]
5. Chassaing N, Kanitakis J, Sportich S et al. Generalized epidermolytic hyperkeratosis in two unrelated children from parents with localized linear form, and prenatal diagnosis. J Invest Dermatol. 2006;126(12):2715-7. [PMID: 16990804]
6. Bilgili SG, Karadag AS, Calka O, Bulut G, Cecen I. Widespread hyperpigmented and hyperkeratotic plaques following lines of Blaschko. Indian J DermatolVenereolLeprol. 2012;78:403-5. [PMID: 22565454]