Case Report

Erythrokeratoderma variabilis

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ABSTRACT

Erythrokeratoderma variabilis is a rare autosomal dominant genodermatosis of variable expressivity. In this report, we describe the clinical features and microscopic findings in one of our patients born to unaffected parents. We also briefly review the literature on this disorder.

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Erythrokeratoderma variabilis (EKV) is an autosomal dominant genodermatosis of variable expressivity but sporadic cases are not infrequently reported. The term EKV was first used by Mendes da Costa[1] and almost 150 cases were reported in the literature mostly from Europe.[2] The condition usually manifests in the first year of life and clinically consists of 2 main components: erythematous patches and hyperkeratotic plaques with particular propensity to affect extensor sites. The hyperkeratosis is a constant feature and tend to be confluent and well demarcated while the erythema tend to be transient persisting for hours to few days and related to environmental changes and emotional upsets.[3]

Case Report. A 17-year-old girl originally from the Northern part of Kingdom of Saudi Arabia presented to our dermatology clinic with the chief complaint of a thick dry skin involving the whole body. According to her mother, the condition started one week after birth when she noticed redness of the skin of her daughter over both hands, which soon after involved the diaper area. Few days later, the skin became thickened involving the whole body with predilection to extensor surfaces, elbow, knees, groins and face with on and off appearance of red patches at different areas of the body. This was associated with marked thickening of the palms and sole. The patient continued to develop these lesions until puberty when it became stable by this presentation except for the red patches, which changed during summer months. There was no history of nail or hair changes and none of her parents or siblings had similar condition. There was no history of consanguinity.

On examination, widespread irregularly shaped hyperkeratotic plaques distributed over the whole body mainly on the extensor surfaces of the upper limbs, lower limbs, elbows and knees (**Figure 1**). Besides, there were sharply demarcated brightly erythematous scaly geographic patches, which vary in size from few to many centimeters that ran independent of the hyperkeratotic plaques. There was also marked palmoplantar keratoderma (**Figure 2**). The hair, nails and teeth were entirely normal. Skin punch biopsy was taken from the hyperkeratotic lesion, which showed marked orthohyperkeratosis, prominent stratum granulosum, acanthosis and perivascular mononuclear cell infiltrate (**Figure 3**). The diagnosis of EKV was established. Since the patient was only concerned with her asymptomatic dry skin, she chose to use emollients and keratolytics and not to take oral retinoids.

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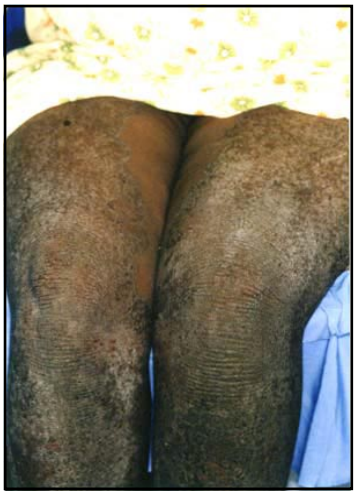

Figure 1 - Hyperkeratotic plaques with geographic outline involving the lower limbs.

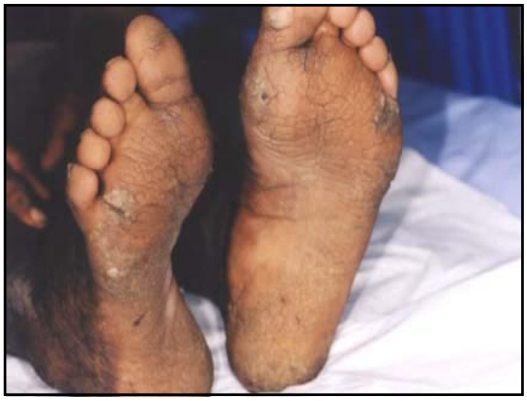

Figure 2 - Plantar keratoderma.

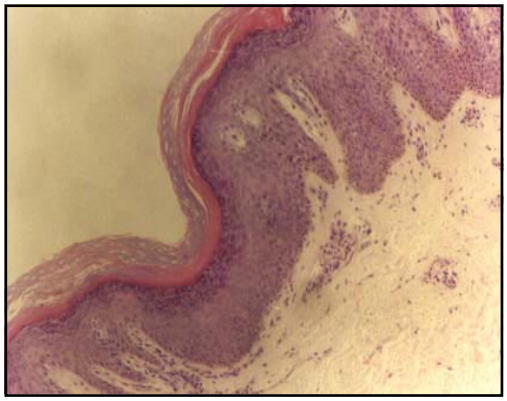

Figure 3 - Histopathology of hyperkeratotic lesion showing orthokeratosis, prominent stratum granulosum and acanthosis.

Discussion. Erythrokeratoderma variabilis is a rare genetic disorder, which has 2 characteristic distinctive morphological features: erythema and hyperkeratosis.[3] Its pathogenesis is unclear but many theories suggested that it represents a systemic ectodermal vascular dysplasia, which leads to disturbance in the keratinization process.[4,5] While Cram[6] believed that, since the 2 components are separate, a unique genetic disturbance of both cutaneous vasculature and epidermis is operative. Most authors consider EKV as a retention-type hyperkeratosis supported by the lack of evidence of increased epidermal proliferation.[7] The condition mode of inheritance is basically an autosomal dominant with variable expressivity but autosomal recessive cases have been rarely reported.[8] Clinically, the figurate sharply defined erythematous patches change their shape and distribution within minutes, hours or days. The hyperkeratotic plaques have a striking geographic outlines and arise independently on normal skin or on areas of persistent erythema. Affected sites include the face, buttocks and extensors of the limbs in symmetrical manner. Cold, wind, sunlight and emotional upsets can aggravate the erythema.[9] There may be palmoplantar keratoderma but mucus membranes, hair, nails and teeth are normal. Besides, congenital defects are absent.[8] The histologic findings of the hyperkeratotic lesion include orthokeratosis, irregular acanthosis, papillomatosis and suprapapillary thinning. In the papillary, dermis there may be few perivascular lymphohistiocytic infiltrates with capillary dilatation.[10,11] Electron microscopic findings have shown marked reduction in the number of keratinosomes in cells of the stratum spinosum and granulosum a feature found in the hyperkeratotic lesions but not the erythematous patches. In contrast, unmyelinated nerve axons and Schwann cells were seen in the superficial dermis of the erythematous lesions which could account to the sensitivity of these lesions to temperature changes, wind and emotional stress.[3] Erythrokeratoderma variabilis is a chronic condition that tends to increase in severity until puberty, which then becomes stable. Hormonal changes may play a role in its severity as it was reported to become severe during pregnancy, oral contraceptive pills use and resolve at menopause.[12] Erythrokeratoderma variabilis condition is a retinoid responsive dermatosis given topically or orally besides the use of emollients and topical keratolytics but recurrence is the rule upon discontinuation of the retinoids.

In summary, we report a patient with the diagnosis of EKV in whom detailed family history showed no evidence of similar condition which in turn suggest the condition could be due to either spontaneous mutation or autosomal recessive gene.[8] Oral retinoids are the treatment of choice with the consideration of the patient age, sex and clinical severity.

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