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Erythrokeratoderma variabilis in two cases with localized and generalized lesions

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Abstract

Erythrokeratoderma variabilis is an autosomal dominant genodermatosis characterized by fixed, brownish hyperkeratotic plaques and migratuar erythematous patches. Here we report a 44-year-old female patient with a common itching and rash over a 4-month period and a 21-year-old male with stable, asymptomatic plaques in both antecubital regions for 3 years due to the rare diagnosis of EKV. The aim of this study is to draw attention to the fact that the disease may be encountered in different clinical manifestations and may be severe itchy or asymptomatic. The absence of extracutaneous involvement and recurrence after discontinuation of treatment indicate a benign and chronic course.

Key words: genodermatosis, erythrokeratoderma variabilis, localized, generalized

Introduction

Erythrokeratoderma variabilis (EKV) is considered as the subtype of a group of skin diseases called Erythrokeratoderma [1]. EKV is an autosomal dominant inherited genodermatosis and is characterized by fixed, brownish hyperkeratotic plaques and figurative erythematous patches [2]. Progressive symmetric erythrokeratoderma and ECV are accepted as two main subtypes of erythrokeratodermas. Since these ichthyosiform dermatoses have similar characteristics, it has been suggested to call progressive symmetric erythrokeratoderma as "EKV progressive" [3]. Two cases with rare ECV are presented here to emphasize that the disease may follow different clinical presentations.

Case-presentation

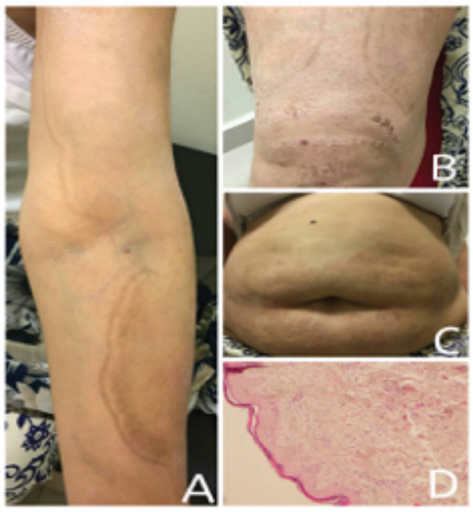
Case 1

A 44-year-old female patient was admitted to the dermatology clinic with a 4-month history of erythematous rash on the trunk, arms and legs. The patient's history did not reveal any drug, infection, emotional stress or other condition that could be associated with rash. She had no known dermatological or systemic disease in her medical history. Similarly, she did not describe a psoriasis or similar skin disease in her family history. Dermatological examination revealed widespread erythematous hyperpigmented sharply circumscribed annular patterned plaques on the trunk and extremities, and fine scales on these plaques (Figure 1). Nails, scalp and oral mucosa were normal. There was no palmoplantar hyperkeratosis. Direct fungal examination of the lesions was negative. As a result of biochemical analysis of blood tests, low density lipoprotein level was determined as 165 mg/dl. Punch biopsy revealed hyperkeratosis in the epidermis, thinning in the granular layer, spongiosis and acanthosis, and moderate mononuclear inflammatory cell infiltration in the upper and middle dermis (Figure 1D). Systemic isotretinoin 20 mg/day, topical moisturizer and corticosteroid mixture was started with the diagnosis of progressive symmetric erythrokeratoderma. After 1 month of regular treatment, the patient stopped treatment because of regression of complaints and did not come to control. After 4 months, his lesions reappeared.

Case 2

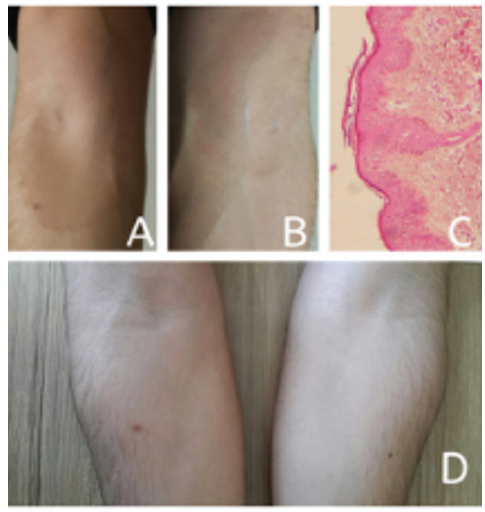
A 21-year-old male patient presented to the dermatology clinic with an asymptomatic, brown rash on the inner face of the arms which had been growing very slowly over the years. Similar to case 1, there was no specific feature in the medical history and family history of case 2. In the dermatological examination, there were two sharply circumscribed, brown plaques in both antecubital regions (Figure 2). Body, scalp, nails, oral mucosa, palms and soles were normal. Body, scalp, nails, oral mucosa, palms and soles were dermatologically normal. Direct fungal examination of the lesions was negative, too. Biochemistry tests including hemogram,

Figure 1 - Specific characteristics of Case 1



A: Geographically shaped hyperkeratotic plaque on the inner side of the left arm
B: Hyperkeratotic plaque and excoriated papules on the anterior aspect of the right leg
C: Erythematous squamous plaque on the anterior abdomen
D: Hyperkeratosis in the epidermis, thinning in the granular layer, spongiosis and acanthosis, and moderate mononuclear inflammatory cell infiltration in the upper and middle dermis (HE, x100)

Figure 2 - Specific characteristics of Case 2



A: Sharply circumscribed, brown, hyperkeratotic plaque on the right antecubital region
B: Lesion on the left antecubital region
C: Hyperkeratosis and acanthosis in the epidermis and perivascular lymphocytic infiltrate in the papillary dermis (HE, x100)
D: View after treatment

liver and kidney function tests and lipid profile were determined within normal limits. Punch biopsy showed hyperkeratosis and acanthosis in the epidermis and perivascular lymphocytic infiltrate in the papillary dermis (Figure 2C). The patient was started on topical potency corticosteroid because of the limited number of lesions. Following a 1-month intermittent treatment period, the complaints were completely regressed.

Discussion

The most important feature that differentiates PSEK from EKV is the lack of migration characteristics of the lesions. Moreover palmoplantar keratoderma is more specific to PSEK [4]. Progressive symmetric erythrokeratoderma is inherited as autosomal dominant in 50% of the cases, while in others there are spontaneous mutations or autosomal recessive transitions [5]. In this study, family history was not detected in both cases.

Two different types of skin lesions are seen in the EKV: fixed hyperkeratotic plaques and migratory erythematous areas. Both types of skin lesions are sharply demarcated and geographic. As in Case 1, hyperkeratotic plaques are located on the extensor faces of the trunk, hips and extremities. However, as in Case 2, where the antecubital region is affected, folds like axilla and groin have been reported to be affected by the disease [6]. In this study, lesions in Case 1 have been generalized since the onset of the complaints and have been localized in Case 2 for 3 years.

Palmoplantar keratoderma is seen in 50% of ECV cases. Emotional stress, extreme temperatures, friction and rarely sun exposure have been reported to worsen the disease. It was accepted that the disease was limited to the skin as in the cases in this study [7]. In both cases in this study a condition that causes the exacerbation of the disease, extracutaneous involvement or palmoplantar keratoderma were not detected. The fact that genetic studies are not always available in the routine clinical practice and there is no specific histopathological finding makes the diagnosis difficult. In this study, hyperkeratosis, acanthosis, papillomatosis, lymphocytic infiltration in the dermis and perivascular areas were found similar to the literature in the histopathology of both cases [8]. Therefore, the diagnosis was made by clinicopathological correlation in both cases.

Systemic retinoids, topical moisturizers, keratolytic ointments, coal tar, topical retinoids, tacrolimus, topical corticosteroids and vitamin D analogs have been effective in the treatment of the disease. However, due to the chronic course of the disease, recurrence is common with discontinuation of treatment [9]. In this study, Case 1 discontinued treatment due to a decrease in complaints after taking isotretinoin treatment for a month. 4 months after the discontinuation of the treatment, the lesions were returned to their original state. Isotretinoin treatment was started at low dose to case 1 because of LDL value was greater than 130 mg/dl.

In this study, it was aimed to emphasize that ECV may occur with different clinical findings and may be severe itchy or asymptomatic. Absence of extracutaneous involvement indicates a benign pathology, and recurrence after discontinuation of therapy indicates a chronic course.

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