A SPORADIC CASE OF ERYTHROKERATODERMIA VARIABILIS IN AN ADULT

Dermatology

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ABSTRACT

Erythrokeratoderma are diverse group of genodermatosis affecting keratinization under which, Erythrokeratoderma Variabilis (EKV) was first described in 1925 by Mendes da Costa. Our patient is a 48 year old male presenting clinically with EKV Mendes Da Costa, hyperkeratotic subtype with the histopathology supportive of the diagnosis. We are reporting for its rarity and due to its late onset in our patient and also for the sporadic nature of this particular case of EKV.

KEYWORDS

Erythrokeratoderma Variabilis, Hyperkeratotic Subtype, Adult Onset, Sporadic

INTRODUCTION

Erythrokeratoderma are diverse group of rare genodermatosis affecting keratinization characterized by well demarcated erythematous, hyperkeratotic plaques either migratory or stationary.^[1] A recent proposal to name it as Erythrokeratoderma variabilis et progressiva (EKVP) as there is overlapping of clinical features along with underlying same genetic mutations of Erythrokeratoderma variabilis (EKV) with Progressive symmetric erythrokeratoderma (PSEK).^[2]

Case Report

A 48 year old male presented with complaints of skin lesions over the elbows & knees, the thigh and the feet of 8 years duration. These scaly lesions are asymptomatic & began first over both elbows then involved the knees, thigh and feet. Initially lesions were reddish. Over the years they have become brown, rough and thicker. No history of any transient lesions which changes shape or sizes or position. Patient took over the counter medications with the lesions regressing but not fully resolved. No history of aggravation on exposure to cold, heat, stress or sun. No history of similar complaint in the family. No significant past history of any other systemic or chronic illness. On general and systemic examination, no significant findings were seen. On dermatological examination, multiple scaly brown figurate hyperkeratotic plaques which are well demarcated with geographical pattern along with accentuation of skin creases, showing nearly symmetrical distribution over both the extensors of knees, elbows and dorsum of feet noted. Palms & Soles were normal. [Figure 1: (a,b,c,d)]

b)

c)

d)

a)

Figure 1: (a, b, c and d) Multiple well demarcated scaly brown figurate hyperkeratotic plaques with geographical pattern along with accentuation of skin creases, showing nearly symmetrical distribution over both the extensors of elbows, knees and dorsum of feet.

Histopathology examination from the skin lesion over the left knee was supportive with nonspecific findings. Epidermis showed hyperkeratosis, moderate acanthosis & papillomatosis. [Figure 2]

Figure 2: Epidermis showed hyperkeratosis, moderate acanthosis & papillomatosis (H and E, x10)

The final diagnosis reached based upon clinical features and histopathological features was erythrokeratoderma variabilis, hyperkeratotic subtype.

DISCUSSION

Classical Erythrokeratoderma Variabilis (EKV) /Mendes da Costa syndrome first described in 1925 by Mendes da Costa is usually inherited as autosomal dominant disorder with few cases of autosomal recessive pattern inheritance. The very few sporadic cases reported could be due to Denovo or new gene mutations where there is no history of family members being affected. [3] EKV has been reported worldwide. Both genders are affected equally and there is no racial predilection. Onset is usually at birth or early infancy. But it may be seen in childhood or manifest even later in adults. [4,5] Our patient had no positive family history and onset was in adulthood.

In about 2/3rd of EKV patients, mutations in GJB3 encoding connexin 31 or GJB4 encoding connexin 30.3 have been found. [1,3] In the skin, Cx31 and Cx30.3 are expressed in stratum granulosum. Connexins form gap junctions which are aqueous intercellular channels which are important for normal epidermal differentiation. Rest 1/3rd of cases have no known mutations in the connexin genes suggesting genetic heterogeneity. [3,6] Clinical features of EKV is characterized by two distinct skin lesions a) relatively fixed well demarcated hyperkeratotic plaques often bizarrely shaped showing predilection for extensor surfaces of extremities, lateral trunk & buttocks. b) Transient erythematous, polycyclic or comma shaped patches occurring at any site which may change its distribution and last days or weeks. [7,8] One feature may predominate or sometimes can be absent or missing. [6] This was noted in our case which is of the EKV hyperkeratotic type. Atypical variants are en cocardes type, reticulate type & erythema gyratum repens-like type. [8] Palms and soles may show diffuse hyperkeratosis. Palmoplantar keratoderma and lesions involving face are seen more in PSEK than in EKV. [9] Our patient had neither features.

Differential diagnoses are Progressive symmetric erythrokeratoderma (PSEK), Keratitis-ichthyosis-deafness (KID) syndrome, Netherton Syndrome, Schnyder syndrome, Greither syndrome, Giroux-barbeau syndrome. Others are plaques of psoriasis, urticaria in early stages. Atypical variants have to be differentiated from Erythema annulare centrifugum, erythema multiforme, subacute lupus erythematosus. [8]

Besides histopathological examination, if available genetic studies can be done to detect connexin genes mutation. EKV has a chronic course with remissions & exacerbations. Treatment is mainly directed at diminishing the hyperkeratosis to minimize discomfort. Current line of treatments are topical keratolytics, topical retinoids, tazarotene, topical corticosteroids and systemic retinoids acitretin, etretinate and isotretinoin. Our patient was started on oral Acitretin 25mg, emollients & keratolytics and is under follow up. Some studies have tried Psoralen plus ultraviolet A (PUVA). [10] Avoidance of mechanical irritation, exposure to extreme temperature changes are advised as general measures. Recent interest has been using drugs affecting connexions hemichannels including antimalarials- mefloquine, aminoglycoside-gentamycin and streptomycin, glycyrrhetic acid-carbenoxolone disodium and benzopyran-tonabersat in the management of connexin implicated conditions. [6] Further studies need to be done for the evaluation of their efficacy in EKV.

CONCLUSION

As the name suggests, Erythrokeratoderma variabilis can have variable clinical presentations as noted in our patient. This case has been reported for its rarity and due to its late onset in our patient and also for the sporadic nature of this particular case of EKV.

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