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Case Report

Erythrokeratoderma variabilis: a case report

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ABSTRACT

Erythrokeratodermia variabilis (EKV) was first described by Mendes da Costa. It is a rare heterogeneous group of inherited cornification disorders characterized by two distinct types of skin lesions: fixed hyperkeratotic plaques and sharply marginated, pruritic, migratory erythematous lesions. We report a case of EKV in a 44-year-old male patient.

Keywords: EKV, Genodermatosis, Hyperkeratotic plaque, Migratory erythema

INTRODUCTION

Erythrokeratodermas (EK) are a rare, heterogenous group of inherited disorders of cornification.[1] They are classified into two types, EKV and progressive symmetric erythrokeratoderma (PSEK).[2] EKV is a rare subtype of EK. Clinically it has two types of presentation. One with discrete, erythematous, well defined patches of bizarre geographical configuration (polycyclic) that change size and shape in a short period of time and other type with erythematous, well defined, hyperkeratotic plaques that are fixed in location.[3] Here we report a case of a 44-year-old man with classical EKV, who was treated with Isotretinoin with good clinical improvement.

CASE REPORT

A 44-year-old man presented with itchy and scaly skin lesions over trunk. These lesions used to occur on and off since birth. Lesions would initially start as diffuse erythema followed by appearance of scaly plaques over the face and trunk. The plaques would be large and assume a map like configuration. Patient was receiving treatment for the same from local physicians. Lesions used to resolve with treatment but recur subsequently. Cutaneous examination revealed diffuse erythema over face, trunk, and limbs (Figure 1). Well-defined to ill-defined scaly plaques were present on the medial aspect of arm extending to the axilla, lateral aspect of chest and loin (Figure 2). Multiple such plaques of varying sizes were present over bilateral scapular area, lower back and cubital fossa (Figure 3 and 4). Plaques had assumed a map like configuration. Similar complaints were present in his elder sister who expired due to unknown cause 10 years back. His parents and other siblings were unaffected. Patient had undergone angioplasty 8 years back. He is not a known case of hypertension or diabetes mellitus. Skin biopsy was advised, but the patient refused. Based on history and clinical examination a diagnosis of EVK was made. The patient was treated with oral Isotretinoin 20 mg once a day, topical moistures lotion, clobetasol lotion and antihistamines.

Figure 1: Diffuse erythema over the face.

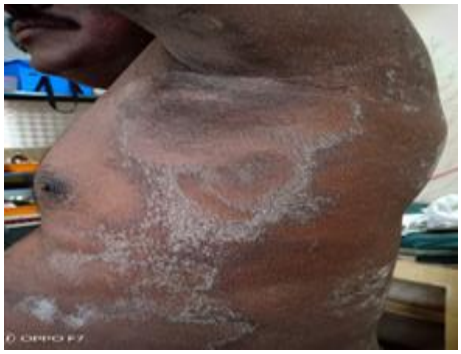

Figure 2: Scaly plaques over inner arm, extending to axilla, chest and loin map like configuration.

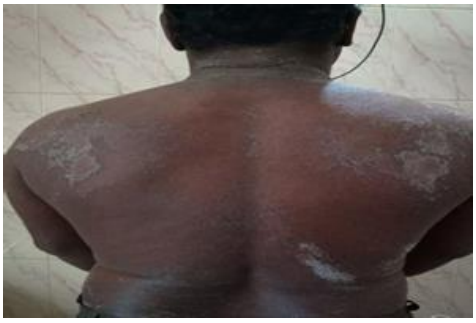

Figure 3: Scaly plaques on bilateral on scapular areas in a background of diffuse erythema.

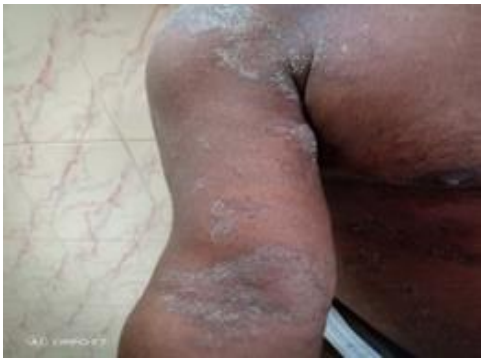

Figure 4: Scaly plaques on cubital fossa.

DISCUSSION

EKV was first described by Mendes Da Costa in 1925. It is a rare subtype of a heterogenous group of inherited cornification disorders called the erythrokeratodermi.[1,2] There are two major subtypes of EKV including Erythrokeratoderma variabilis (Mendes da Costa) and Erythrokeratoderma progressiva symmetrica (Gottron). Few rare variants of EKV have been described. These include erythroderma en cocardes, also known as Degos' disease, reticulate erythrokeratoderma and EKV with erythema gyratum repens-like lesions.[2]

Gap junctions are intercellular channels that allow the passage of water, ions, and small molecules. They are involved in quick, short-range messaging between cells and are found in skin, nervous tissue, heart, and muscle. Mutations in one of the genes coding for the constituent proteins of gap junctions in keratinocytes, known as connexins is responsible for EKV. Autosomal dominant EKV was initially linked to chromosome 1p34-1p35 where there is a cluster of connexin genes encoding the gap junction proteins connexin 30-Cx 30.3 (GJB4), Cx 31.1 (GJB5) and Cx 37 (GJA4). Later, pathogenic GJB3 mutations were identified in four families with EKV, demonstrating a link between defects in Cx 31-associated gap junctions and aberrant epidermal differentiation and function. All reported mutations in EKV, are non-conservative amino-acid substitution mutations, resulting in impaired epidermal differentiation.[4,5] Hence it is hypothesized that there is a systemic ectodermal vascular dysplasia and abnormal vascular dilatation that may lead to a disturbance in keratinization.[2]

EKV usually presents at birth or during infancy but may occasionally develop much later in life.[1,2] EKV Mendes da Costa has two types of morphological skin lesions: Migratory erythematous type and fixed hyperkeratotic plaques. Initially, they present as erythema, and later they become hyperkeratotic. Migratory lesions can occur all over the body, persist for hours to days, and disappear spontaneously to reappear again. They have irregular borders with fine scaling. Variations in the lesions can be influenced by emotions and physical factors like temperature, friction, pressure, or hormones. Fixed hyperkeratotic plaques are erythematous, well-demarcated and distributed on face, buttocks and extremities. They may sometimes be associated with palmoplantar keratoderma.[5] Atypical variants of EKV include Cram-Mevorah which presents as erythema gyratum repens-like skin lesions and erythrokeratoderma en cocardes or Degos' syndrome characterized by annular lesions with central scaling and surrounding erythema, giving the appearance of targetoid, or "en cocardes" distributed on the extremities.[6]

In our case, the patient presented with diffuse erythema followed by appearance of multiple polycyclic scaly plaques of varying sizes in a geographical or map-like configuration. These lesions were fixed in location. Few lesions were seen on flexures, unlike the typical extensor distribution.

Differentiation between the various EK is difficult and along with clinical findings, histopathology and genetic analysis is required to make a diagnosis.[2] Histopathology findings of skin biopsy taken from hyperkeratotic plaques, and erythematous patches is usually non-specific and findings include hyperkeratosis, papillomatosis, epidermal hyperplasia and normal granular layer.[5] Dilated blood vessels and a mononuclear perivascular infiltrate seen in the dermis. Electron microscopy studies show grain like cells at the junction of stratum granulosum and corneum, containing large amounts of clumped perinuclear tonofilaments.[2] Closest differential diagnosis of EKV is PSEK. Lesions of PSEK typically spare the

trunk and are nonmigratory well-demarcated, polycyclic, hyperkeratotic and distributed symmetrically over the elbows, knees, dorsal aspect of hands, feet and buttocks. Other differentials include congenital ichthyosis, and Netherton syndrome.[5] Atypical variants of EKV should be differentiated from subacute lupus erythematosus, erythema annulare centrifugum, and erythema multiforme.[6]

There is no specific therapy for EKV. Various topical therapies that have been used include emollients, retinoic acid, keratolytic, alpha hydroxyl acid, and topical corticosteroids. Systemic agents including oral retinoids like acitretin, etretinate and isotretinoin have been found to be effective. Lesions reappear once the treatment is stopped. Bath PUVA has also been reported to be effective in some cases. Parents have to be counselled regarding the chronic nature of the condition and its prognosis. The need for regular follow-up should also be emphasised.[5,6] Our patient showed good response to oral Isotretinoin.

CONCLUSION

We have presented this case study for its rarity. Clinicians have to be aware of this entity so that diagnosis is not missed. Parents should be counseled about prognosis and emphasized about the need for regular follow-up.

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REFERENCES

1. Brahma D, Jain VK, Aggarwal K. Erythrokeratoderma variabilis. Indian J Dermatol Venereol Leprol. 2003;69(S1):5-6.
2. Karadag AS, Bilgili SG, Calka O, Bayram I. Erythrokeratodermia variabilis: Two case reports. Indian Dermatol Online J. 2013;4:340-3.
3. Mehta TY, Bhuptani NV, Sheth BS. Disorders of keratinisation. In: Sacchidanand S, Oberai C, Inamadar AC. IADVL textbook of dermatology. 4th ed. Mumbai, IN: Bhalani publishing house. 2015;382-3.
4. Common JEA, O'Toole EA, Leigh IM, Thomas A, Griffiths WAD, Venning V et al. Clinical and Genetic Heterogeneity of Erythrokeratoderma Variabilis. J Invest Dermatol. 2005;125(5):920-7.
5. Srinivas SM, Dhar S. Erythrokeratodermia variabilis and erythrokeratoderma en cocardes: Case series with review of literature. Indian J Paediatr Dermatol. 2016;17:202-5.
6. Tiwary AK, Dixit A. Erythrokeratoderma Variabilis in a 6-Year-Old Child: An Uncommon Genodermatosis with Typical Presentation. J Pigmentary Dis. 2017;4(1):e108.

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