ERYTHROKERATODERMIA VARIABILIS WITH RARE ASSOCIATIONS IN A CHILD A CASE REPORT

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ABSTRACT
Erythrokeratoderma variabilis (EKV) or Mendes da Costa disease is a genomic ailment transmitted as an autosomal dominant trait. EKV presents with two distinctive skin lesions which comprises well demarcated, fixed, figurate hyperkeratotic plaques and transient erythematous patches which typically presents at birth or initial infancy. We report a case on Erythrokeratoderma variabilis in a child who presented with grievances of erythematous raised lesions with scaling over the abdomen, back, extremities with left side clubfoot deformity.

INTRODUCTION
Erythrokeratoderma variabilis belongs to erythrokeratoderma group of sicknesses. In 1907, De Buy Wenninger described the first case of erythrokeratoderma variabilis in Netherland (De Buy Wenninger LM, 1907). Mendes da Costa described relatively fixed hyperkeratotic plaques and erythematous areas with geographical margins that changed by the hour in a mother and daughter in 1925 and coined the name "Erythro et keratodermia variabilis" (Mendes da Costa S, 1925). EKV presents with hyperkeratotic and well-marginated plaques which have a propensity to become confluent usually scattered over the extremities, buttocks and face and secondly, erythematous transient lesions which expresses disparity with stress and heat.

CASE REPORT
A 10-year-old boy born to non-consanguineous parents was carried to the skin OPD with complaints of erythematous raised lesions with scaling over the extremities, face, back and abdomen. History from the mother revealed that the child was delivered by full term, forceps delivery with birth weight of 2.82kg. Antenatal and perinatal period was monotonous. Social and motor milestones were delayed. There was no antiquity of hearing or visual disturbances or seizures were noted.

The child was diagnosed with bilateral congenital equino varus deformity at birth, for which surgery was performed at one year of age, but the child was not able to walk. At 4 years of age, mother noticed dryness of skin over the left forearm which later progressed to multiple pigmented raised lesions with scaling over the right forearm, back, both the legs. At 5 years of age, she noticed erythematous circular lesions which changed in shape once in every 15 days and was associated with itching. There was no history of atopy, seasonal exacerbation or thickening of palms and soles. There was no history of infections or trauma triggering the onset of skin lesions. The child was started on emollients, topical steroids and...
systemic antihistamines but showed no enhancement. There is no significant family history. Dermatological examination revealed multiple pigmented papules and scaly plaques of variable thickness and well defined borders present over both the thighs extending to the knees (Figure 1). Multiple erythematous circular patches with pigmented papules over the abdomen (Figure 2). Left side clubfoot was present (Figure 3). Hair, nails, teeth, palms, soles and mucosa were normal. Systemic examination was normal. Routine laboratory investigations such as complete haemogram, urine analysis, renal and liver function tests, serum electrolytes were within normal limits. MRI brain showed features suggestive of mild form of perinatal hypoxic-ischemic injury. Audiogram was done and proved to be normal.

A skin biopsy was taken from the pigmented papule and histopathology showed nonspecific features of hyperkeratosis with papillomatosis and acanthosis (Fig 4).

**DISCUSSION**

Erythrokeratodermia variabilis is an infrequent autosomal dominant disorder but autosomal recessive transmission has also been reported. More than 50% of patients frequently present at birth and 90% within the first year of life. Both sexes are affected similarly. The two types of skin lesions which occur in EKV are fixed, well demarcated, keratotic erythematous plaques, often bizarrely shaped which occurs mainly over the buttocks, extensor surfaces and lateral trunk and transient erythematous patches which may be topographical, targetoid or circinate in shape occur at any site. They extend and regress in area, thickness and degree of erythema. Triggering factors are irritation, trauma or temperature change and grow in shape over hours. After few days or weeks, lesions fade or migrate leaving a residual fine scale. The diagnosis of erythrokeratodermia variabilis is recognized on the basis of clinical features. The condition persists throughout life, might improve at
puberty or worsen during pregnancy or with oral contraceptives. EKV have been shown to be associated with mutations in GJB3 and GJB4 encoding connexin-30.3 and connexin-31 (Papadavid et al., 1998; Ishida-Yamamoto et al., 2000). Connexins are a family of proteins that form gap junctions, which are networks important for intercellular communication (Fuchs-Telem D et al., 2011). The main feature distinguishing EKV from PSEK is the presence of hyperkeratotic plaques on an erythematous base and possibly lack of facial lesions with features of palmoplantar keratoderma in most cases. EKV is associated with palmoplantar keratoderma, erythema annulare centrifugum like lesions, erythema gyratum repens like erythema, bilateral talipes equinovarus (clubfoot) deformity (Wei et al., 2011; Beare JM et al., 1972) and ataxia.

Acitretin is the drug of choice but isotretinoin, bath PUVA and non sedating antihistamines may also be useful. There is only one case reported in Ireland where EKV was associated with congenital equinovarus deformity and bilateral deafness. In our case, EKV was associated with bilateral talipes equinovarus deformity without sensorineural deafness. Hence this case is reported because of its rare associated features.

REFERENCES