

Name

Erythrokeratoderma Variabilis

Alternative Names

EKV

Erythrokeratoderma Figurata, Congenital Familial, in Plaques
Erythrokeratoderma Variabilis with Erythema Gydatum Repens
Greither Disease
Keratosi Palmoplantari Transgrediens et Progrediens
Mendes da Costa syndrome

Record Category

Disease phenotype

WHO International Classification of Diseases

Diseases of the skin and subcutaneous tissue > Other disorders of the skin and subcutaneous tissue

OMIM Number

133200

Mode of Inheritance

Autosomal dominant

Gene Map Locus

1p35.1, 1p35.1

Description

Erythrokeratoderma variabilis (EKV) is a rare genetic disorder that is inherited by an autosomal dominant gene with variable expressivity. The condition usually starts after birth between 3 months and 3 years of age or, rarely, at birth or in early adult life. EKV is

characterized by two distinct morphologic features: erythematous patches and hyperkeratotic plaques. These manifestations are extremely variable in a single family and within an individual patient. The erythematous patches usually consist of bright red to brownish well demarcated lesions that frequently vary in shape, size, and severity and change their location or involutes completely within hours or days. The hyperkeratotic plaques that are usually fixed in location are yellow-brown with greasy scales and are often polycyclical in shape. The distribution is symmetrical with a predilection of extensor surfaces of the limbs, axilla, buttocks, groin, and face. Palmoplantar keratoderma may be seen in some patients. Mucous membranes, hair, and nails are spared.

Exacerbations of the disease have been seen during pregnancy or with use of oral contraceptive pills. Usually there will be an improvement with age, particularly after menopause.

Molecular Genetics

The inheritance pattern of EKV is autosomal dominant with considerable variability of expression among family members. The gene is mapped to chromosome 1p34-p35, a region that has been found to house a cluster of connexin genes encoding gap junction proteins. Four connexins have been identified: GJB3 (connexin 31, Cx31), GJA4 (connexin 37, Cx37), GJB5 (connexin 31.1, Cx31.1), and GJB4 (connexin 30.3, Cx30.3). Mutations associated with EKV were subsequently found in both connexin 31 and connexin 30.3. Recently, a possible autosomal recessive variant of EKV has been reported.

Epidemiology in the Arab World

United Arab Emirates

Galadarai and Galadari (2004) described a 4-year-old female with a history of erythematous skin lesions on her face, extremities, forearms, and joints that started a few months after birth and the condition progressed over a period of time. Some lesions showed variable exacerbations and remissions. Her parents were healthy cousins. There was no family history of a similar problem, although her older brother showed marginal hair loss without any skin lesions. The hair showed normal appearance, but there was no hair growth on the margins of the scalp. History and physical examination revealed erythematous, hyperkeratotic patches with sharply demarcated borders, and a diagnosis of erythrokeratoderma variabilis was made. As the patient was only 4 years old, the treatment provided involved emollients only with not much improvement.

References

Galadari I, Galadari H. Case study: erythrokeratoderma variabilis. *Skinmed.* 2004; 3(4):231-2. PMID: 15249788

Related CTGA Records

N/A

Links

http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=GB&Expert=317

Contributors

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