GENETICS AND GENODERMATOSES

HEREDITARY PUNCTATE PALMOPLANTAR KERATODERMA TYPE IA - MUTATION IN THE AAGAB GENE

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Background: Punctate palmoplantar keratoderma type IA (PPKP1A; OMIM 148600), also called keratosis punctate palmoplantaris type Buschke-Fischer-Brauer, is a very rare autosomal dominant hereditary skin disease characterized by multiple hyperkeratotic centrally indented papules that develop in early adolescence or later and are irregularly distributed on the palms and soles. In mechanically irritated areas, confluent plaques can be found. PPKP1A is caused by heterozygous mutation in the AAGAB (Alpha- And Gamma-Adaptin-Binding protein P34) gene on chromosome 15q23. Interfamilial and intrafamilial severity shows broad variation. There have been reports of an association between PPKP and the development of early- and late-onset malignancies, including squamous cell carcinoma, Hodgkin disease, and breast, pancreatic cancers.

Observation: Our patient is a 62-year-old man with punctate hyperkeratotic lesions on his palms and soles, which have been present from birth. The same skin changes were observed in his father and offspring. Dermatological examination showed multiple punctate keratoderma, with a yellowish, irregular surface and several fissures. Such alterations were limited to the palms and soles, without compromising other topographies. The diagnosis of the condition was confirmed histopatologically and genetically (heterozygous mutation in the AAGAB gen (NM_024666) :c.195_198delCAAA p.Lys66Phefs*43, Motol University Hospital, 03/2018). Our patient was treated topically with keratolytics, such as urea and corticosteroids under occlusion several nights per week in combination with systemic retinoids, which led to significant regression of the keratoses.

Key message: Hereditary PPKs are not curable, but symptoms can be controlled. The aim of treatment is to reduce the thickness of the skin and to soften the skin. Treatment options include regular use of emollients, keratolytics such as salicylic acid or urea, antifungal creams or tablets if indicated, topical retinoids/calcipotriol, and systemic retinoids. In many cases, treatment only results in short-term improvement. The long-term outlook may differ depending on each person's individual symptoms and pain tolerance.