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Epidermolytic palmoplantar keratoderma of Vorner: Case report

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Epidermolytic palmoplantar keratoderma (EPPK), Vorner type (EPPK, OMIM 144200), rare autosomal dominant genodermatosis, first described by Vorner in 1901, is the most common form of diffuse keratoderma characterized clinically by total area of hyperkeratosis on palms and soles. It is usually due to a dominant mutation in the keratin 9 gene (KRT9), located in chromosome 17, which is specific for the palmoplantar skin. A two-year-old boy presenting hyperkeratosis of palms and soles since the first month of birth, were noticed diffuse hypertrophic plaques with yellowish keratosis and fissures limited to palms and soles and distinct erythematous margin at the border of normal-appearing skin. A punch biopsy of the palms showed epidermolysis in spinous and granular layers, hyperkeratosis and irregular clumped keratohyalin granules. EPPK integrates a complex group of uncommon keratodermas, mainly based on the morphologic and topographical characteristics and associated systemic alteration. It is the most frequent form of such diseases and usually appears during the first weeks or months after birth, characterized by diffuse thickening of the skin of palms and soles with well-circumscribed erythematous borders. The disease may be followed by other ones, above all neoplasias, probably arises due to segregation of oncogenes. Histopathologically, EPPK presents the characteristic features of epidermolytic hyperkeratosis and vacuolar cytolysis in the upper spinous and granular layers, large irregularly shaped keratohyalin granules and hyperkeratosis and electron microscopy shows vacuolization of keratinocytes of the granular layer and clumping of keratin filaments. It is differentiated from other palmoplantar keratodermas by lack of other cutaneous findings and by the presence of histologic features of epidermolytic hyperkeratosis. Currently, there are few therapeutic resources, which are mostly symptomatic. RNA interference strategies are being investigated as an approach to allele-specific gene silencing for dominant-negative keratin diseases.

Biography

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