GENETICS AND GENODERMATOSES

ACROKERATOELASTOIDOSIS: REPORT OF A FAMILIAL CASE WITH ADDITIONAL BLASCHKO-LINEAR HYPOPIGMENTED LESIONS

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Background: Acrokeratoelastoidosis is a rare disorder characterized by small keratotic papules that primarily involve the margins of the hands and feet, appearing during adolescence or adult life. Both sporadic and familial cases following autosomal dominant inheritance have been reported. Histological findings reveal a degeneration of elastic fibers in the reticular dermis. The gene(s) responsible for acrokeratoelastoidosis remains unknown.

Observations: Two sisters, a 21-year-old and a 19-year-old, showed up at our Department of Dermatology with a history of asymptomatic lesions on their hands. On physical examination, several round, skin-colored, flat-topped, firm papules were observed on the lateral sides and dorsum of the hands. In addition, linear hypopigmented, slightly atrophic lesions were seen on both wrists extending to the arms. Hyperextensible joints were noted on the hands and swan neck deformity of the fingers were additional findings on both cases. Histological examination of a biopsy taken from one of the warty papules of both patients revealed marked hyperkeratosis, mild granulosis and acanthosis in the epidermis. Verhoeff–van Gieson stain showed a decrease in number of elastic fibres in the dermis. Transmission Electromicroscopy (TEM) showed decreased thickness of the elastic fibers, with surface indentations. Marked fragmentation of elastic fibers could also be observed as well as areas of granular degeneration. Collagen bundles showed normal aspect.

Key message: It is important to consider the diagnosis of acrokeratoelastoidosis in the presence of papular lesions on the dorsal and lateral sides of hands and feet. Familial cases should be searched as well as additional acral joint hypermobility. The presence of linear hypopigmented lesions is also a newly reported finding.