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Acrokeratoelastoidosis: case report

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KEYWORDS

acrokeratoelastoidosis
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ABSTRACT

Acrokeratoelastoidosis (AKE) is a rare, benign keratoderma that presents as keratotic papules on the borders of palms and soles and frequently involves the dorsum of hands and feet. In most cases it is inherited in an autosomal dominant fashion but also sporadic forms have been reported. Its pathogenesis remains unclear. Histopathology shows the presence of a reduced number of thick and fragmented elastic fibers in the dermis (elastorhexis). Various forms of treatments have been attempted with modest or poor results. We present an inherited case of AKE in an 14-year-old girl with papular elements simmetrically spread to the knuckles of the fingers from the age of eight.

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1 Introduction

Acrokeratoelastoidosis(AKE)ofOswaldoCostaisarare, benign autosomal-dominant disease, only sometimes sporadic (1). It is part of the hereditary palmar-plantar keratoderma (PPK), characterized by thickening of the stratum corneum in the palmar-plantar area. The mode of genetic transmission (linkage chromosome 2) consists of a fibroblast secretion defect of elastic fibers (2). The confirmation genetic test in association with the morphological criteria is useful to differentiate this from other similar PPKs. It begins before the age of twenty, with a progressive evolution. Numerous asymptomatic yellowish-white papules, sometimes umbilicated or warty, that can converge in plaques, are typically located in the areas between the volar and dorsal sides of the hands and feet and on the dorsum of hands and feet. Sometimes they take on a "cobbled" appearance. Histological peculiarity is the presence of elastorhexis, a reduced number of thick and fragmented elastic fibers in the dermis (3). Therapy with oral keratolytics or oral retinoids have been tried with little help or without success. No associated systemic diseases are reported in the literature, and apart the frequent cosmetic discomfort the prognosis is generally good (3).

2 Case Description

We describe the case of 14-year-old girl with asymptomatic papular elements spread to the knuckles of the fingers from the age of eight (Fig. 1). The mother presented the same picture, at the same age and still presents it; including "transition" areas between the volar face and the back of the hands. A biopsy was performed on the left hand fifth finger papule. Histological examination showed marked orthokeratotic hyperkeratosis of the epidermis, mild hypergranulosis, irregular acanthosis and minimal lymphocytic infiltrate of the superficial dermis (Fig. 2).
Weigert-Van Gieson histochemical staining showed the presence of scattered fragmented elastic fibers in the dermis (Fig. 3). Findings consistent with the clinical diagnosis of autosomal dominant hereditary acrokeratoelastoidosis. The girl also presented on the fourth finger of the right hand a knuckle pad, that is very rarely described in association with acrokeratoelastoidosis.

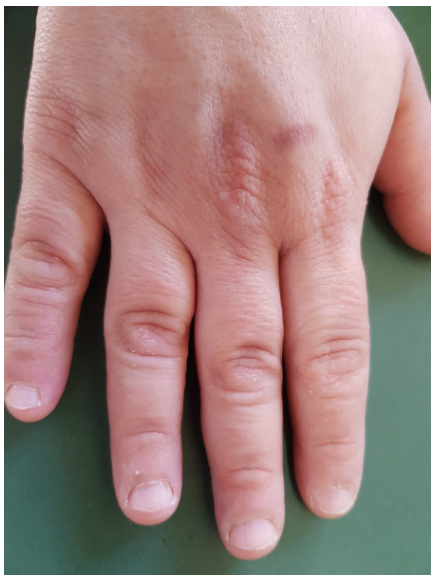


Fig. 1. *Papular elements spread to the knuckles of the fingers.*

Fig. 2. *Histological examination of a papular lesion shows marked orthokeratotic hyperkeratosis of the epidermis, mild hypergranulosis and irregular acanthosis and minimal superficial dermal lymphocytic infiltrate.*

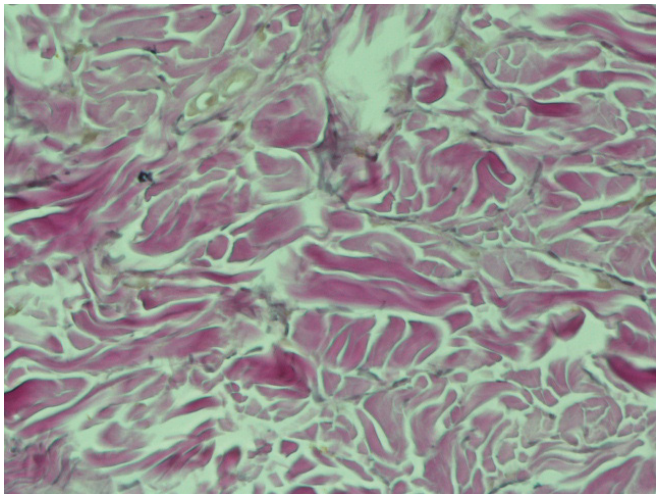
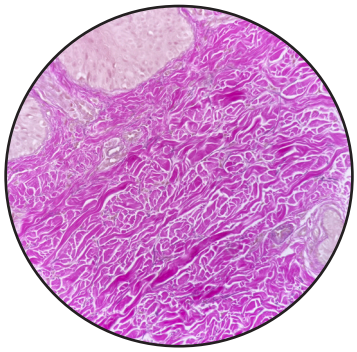


Fig. 3. *Weigert-Van Gieson histochemical staining performed which showed the presence of scattered fragmented elastic fibers in the dermis. Findings consistent with the clinical diagnosis of autosomal dominant hereditary acrokeratoelastoidosis.*



3 Discussion

Acrokeratoelastoidosis is a benign autosomal dominant genodermatoses, considered a type of inherited punctate palmoplantar keratoderma (PPKP), specifically Type 3 PPKP (4), where preliminary linkage suggests a possible locus on 2p25-p12. (5). The onset of the disease is frequently seen in infancy or adolescence. The clinical characteristic of AKE is multiple, yellowish papules located symmetrically on the lateral surfaces of the palms and soles and sometimes over the knuckles of the hands (2). Our case is characterized by the localization of the lesions on the dorsal surfaces of the hands and the unusual association with Knuckle pad.

Histological findings of AKE comprise hyperkeratosis with hypergranulosis, acanthosis, and epidermal hyperplasia. Van Gieson stain shows reduced number of elastic fibers in the dermis with a high proportion of fragmented fibers (2).

Usually, no treatment is required because the disease is asymptomatic without systemic involvement.

However, in cases where significant aesthetic discomfort occurs, various treatments have been attempted (topical and systemic medications, laser, surgery) with limited clinical improvement (2, 6). Our patient was so young that no treatment was necessary.

4 Conclusion

It is important for a modern dermatologist to recognize this condition and to differentiate it from other PPKs that can be associate with predisposition to malignant tumors. It is also important recognize this disorder to distinguish it from warts, which can show similar pictures, in order to avoid incongruous, useless and sometimes painfull treatments.

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DISCLOSURE

All authors report no conflict of interest.

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