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IMAGES IN CLINICAL PRACTICE

HEREDITARY PALMOPLANTAR SKIN LESIONS

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A 2-year-old boy was referred to pediatric dermatology department with symmetrical orange/yellowish, diffuse, scaly palmoplantar hyperkeratotic lesions with erythematous borders involving dorsal surface of phalanges since his first year of life (Figures 1 and 2). There were no teeth or hair abnormalities nor history of cutaneous infections. His father, paternal aunt (Figure 3), and cousins had similar features.

Figure 1. Diffuse plantar keratoderma.

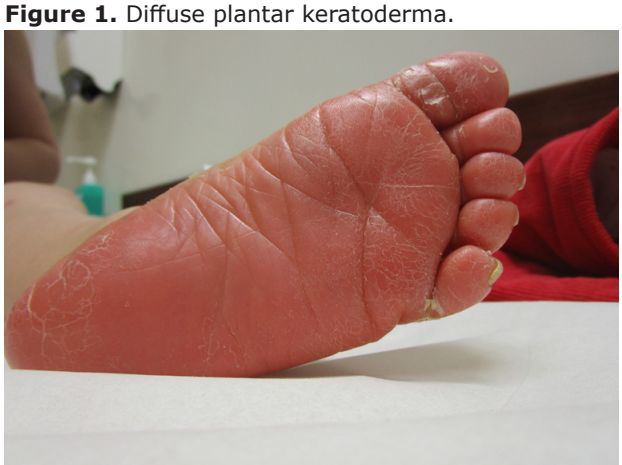

Figure 2. Palmar keratoderma.

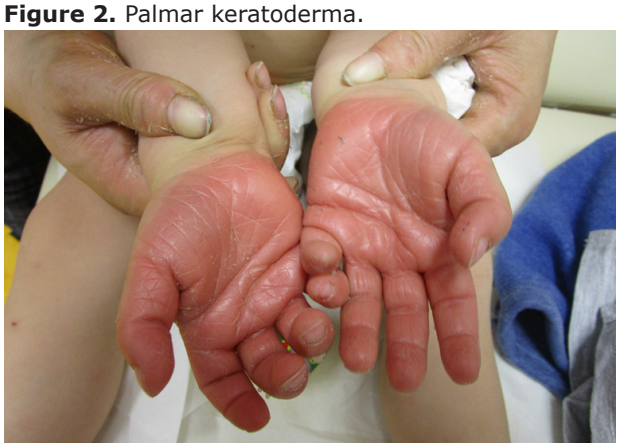

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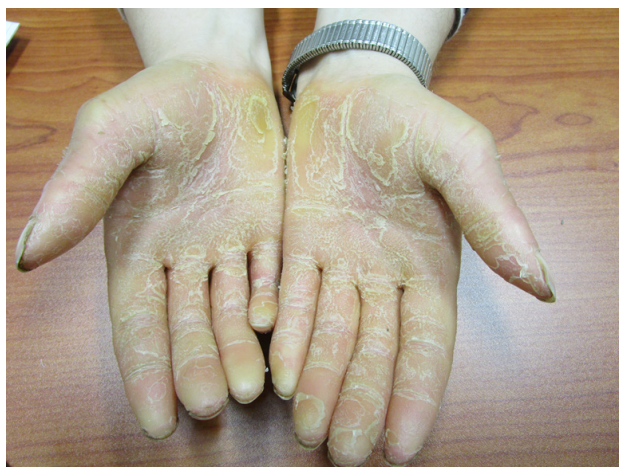
Figure 3. Aunt's hands showing palmar keratoderma, with limited extension of the fingers.



What is the diagnosis?

The diagnosis of *transgrediens* hereditary palmoplantar keratoderma (H-PPK), with probable autosomal dominant transmission was made and he was started on topical emollients with urea to which he improved. His aunt underwent a skin biopsy, that confirmed non-epidermolytic palmoplantar keratodermas (PPK), and keratin 1 and 9 gene testing was done, where no mutations were identified. No further study was performed based on his family decision. PPK are conditions characterized by palmoplantar hyperkeratotic epidermal thickening.[1,2,3] Sporadic and hereditary forms exist. H-PPK is characterized by an early onset of disease, positive family history and treatment resistance[3,4] and can present as isolated form or with associated features (ectodermal defects or multisystemic involvement).[1,2] H-PPK can be focal, striate, punctate or diffuse,[1,2,3] and the latter may be classified as: without *transgrediens* (limited to palmoplantar surfaces), or *transgrediens* and *progrediens*, with lesions on the dorsal surfaces of acral regions, Achilles tendon or nails.[1,2,3] Although the majority of PPK are not life-threatening, this is a chronic disease, with lifelong burden, both physically and psychologically.[2]

Diagnosis of PPK is made by clinical-based morphological classification and skin biopsy. If an inherited or syndromic disorder is suspected, genetic testing should be performed, for prognosis and for prenatal counseling

and diagnosis.[2] H-PPK are clinically and genetically heterogenous[4] and diffuse PPK can be inherited in an autosomal dominant pattern, involving mutations in various genes, such as KRT1 or KRT9, DSG1, AQP5; or in a recessive dominant pattern, involving SERPINB57 and SLURP1 genes.[1]

Although lacking specific treatment, regular baths, emollients, topical keratolytics (such as urea or salicylic acid-based ointments), topical retinoids and mechanical removal of hyperkeratotic areas can be performed.[2,3,4] Oral retinoids (acitretin, alitretinoin) are usually effective, but side effects must be taken into account.[2] Dermatophyte or bacterial infections are common and should be diagnosed and treated with antifungal and antibacterial agents.[2]

Compliance with ethical standards

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Conflict of Interest: None

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