

Vohwinkel syndrome: ichthyosiform variant in a family*

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Abstract: Vohwinkel syndrome belongs to the group of hereditary palmoplantar keratoderma, having an autosomal dominant inheritance. In this report, the authors present a case of a four-year-old boy with diffuse scaling over his entire body and transgredient palmoplantar hyperkeratosis with some fissured areas. Family evaluation revealed that his mother and other family members were affected. Based on his clinical findings and on family history, the diagnosis of the ichthyotic Vohwinkel syndrome subtype, characterized by generalized ichthyosis and palmoplantar hyperkeratosis, was established.

Keywords: Genetic diseases, inborn; Ichthyosis; Keratinocytes; Keratoderma, palmoplantar; Skin diseases, genetic

INTRODUCTION

Vohwinkel syndrome (VS) belongs to the group of hereditary palmoplantar keratoderma and has an autosomal dominant inheritance.¹ The authors report a case of the ichthyosis subtype in a child (Camisa's syndrome).

CASE REPORT

A four-year-old boy presented with pruritus and diffuse scaling since birth. The lesions worsen during winter, and the use of moisturizers partially relieves the symptoms. Some relatives had similar experiences (Figure 1). On examination, the patient presented scaling over his entire body and transgredient palmoplantar hyperkeratosis with some fissured areas (Figures 2 and 3). His mother showed similar changes, with honeycomb scales on her

body, transgredient palmoplantar hyperkeratosis and digital constricting bands (pseudoainhum) (Figure 4). Based on the patient's clinical findings and his family history, the diagnosis of Vohwinkel syndrome (VS) was established.

DISCUSSION

Mutilating Palmoplantar Keratoderma, or VS, belongs to the group of the hereditary palmoplantar keratoderma characterized by focal or diffuse palmoplantar thickening.¹ It starts in childhood and has an autosomal dominant inheritance, occurring predominantly in females and in Caucasian individuals.¹ VS is characterized by diffuse transgredient palmoplantar hyperkeratosis with a honeycomb appearance, and also affects the extensor surfaces of the limbs. Digi-

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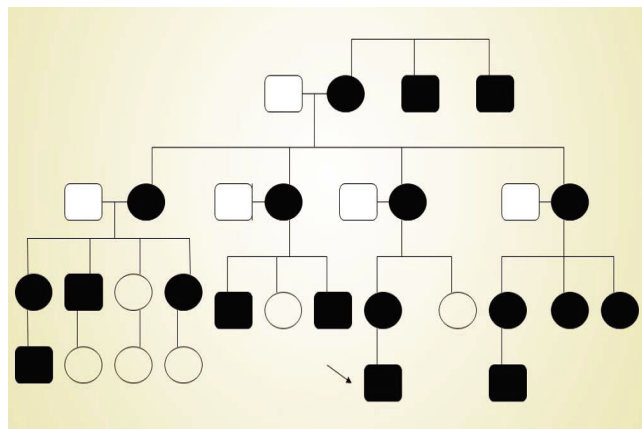


FIGURE 1: Heredogram: family members affected by the syndrome are represented by rectangles (men) or black circles (women). The patient is identified by the black arrow



FIGURE 2: Patient presenting fine scales throughout body surface (A, B)

tal constricting bands, composed by fibrous connective tissue (pseudoainhum), can lead to autoamputation. Distal starfish-shaped keratotic papules on the dorsum of the feet and hands, wrists, forearms, elbows and knees are also typical findings.¹⁻³

Two variants of the syndrome are described: one is associated with ichthyosis (Camisa's syndrome), and the other is accompanied by sensorineural hearing loss.¹ The ichthyotic variant, which is associated with a mutation in the *loricrin* gene and results in defective formation of the stratum corneum, is characterized by generalized ichthyosis and palmoplantar hyperkeratosis, as observed in our patient.^{2,4,5} Starfish-shaped keratotic papules and deafness are not observed in this variant.^{1,4} The variant with sensorineural hearing loss occurs due to a mutation in the *connexin 26* gene (*GJB2* gene) and presents with starfish-shaped keratotic papules and palmoplantar hyperkeratosis. Ichthyosis is not seen in this form of the disease.⁶

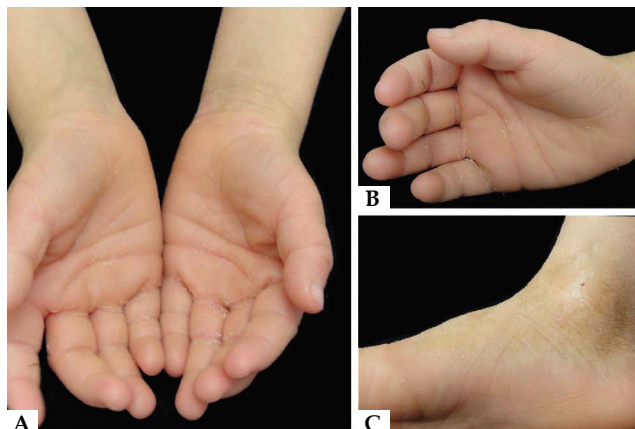


FIGURE 3: Patient with transgredient palmoplantar hyperkeratosis and some palmar fissures (A, B, C)

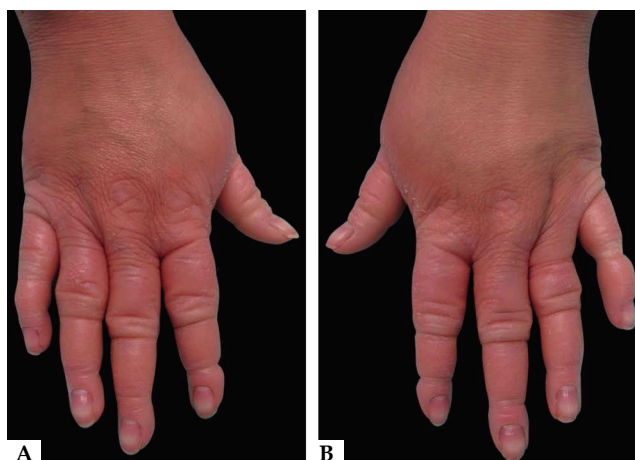


FIGURE 4: Patient's mother presenting digital constrictive bands (pseudoainhum)



The differential diagnosis of VS includes other keratodermas presenting with digital autoamputation, such as Mal de Meleda, Olmsted syndrome, acral keratoderma, pachyonychia congenita, palmoplantar keratoderma of Sybert, and palmoplantar keratoderma of Gamburg-Nielsen, as well as acquired dermatoses that can lead to the appearance of constricting bands, such as leprosy, tertiary syphilis, ainhum, scleroderma, amniotic bands, Raynaud syndrome and syringomyelia.¹


The aim of treatment is to relieve hyperkeratosis with emollient and keratolytic topical therapy and to prevent autoamputation of digits with therapeutic release of constricting bands.^{4,7} The use of retinoids, such as acitretin, isotretinoin and etretinate, has been demonstrated to result in resolution of pseudoainhum and reversal of keratoderma by decreasing epithelial cell cohesion and inhibiting pathological keratinization.^{1,8,9} □

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Approval of the final version of the manuscript, Design and planning of the study, Preparation and writing of the manuscript, Critical review of the literature	
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