

Do you know this syndrome?

Você conhece esta síndrome?

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CASE REPORT

Case 1 – A 22-year-old female patient with anonychia of all fingers and toes at birth. When the patient was only a few weeks old, the condition evolved with thick nails, severe keratosis pilaris of the limbs, diffuse and transgressive plantar hyperkeratosis. Hoarseness and recurrent otalgia were observed during her childhood.

Medical history - see genogram.

Upon examination, we observed thick and curved nails, plantar thickening, severe keratosis pilaris of the limbs, palpable cysts in the neck, armpits and bosom, and white patches on the tongue (Figure 1).

Case 2 - A 20-year-old female patient, with thickening of the nails of all fingers and toes, plantar hyperkeratosis and keratosis pilaris since childhood, but to a lesser degree. A few years ago the condition evolved with the formation of cysts on the trunk, especially in the neck region (Figure 2).

Medical history - see genogram. (Figure 3).

Upon examination, we observed plantar hyperkeratosis in areas of higher pressure, thick and curved nails, keratosis pilaris of the upper limbs, multiple cysts on the trunk, absence of patches in the oral mucosa.

Suspected diagnosis: Pachyonychia Congenita.

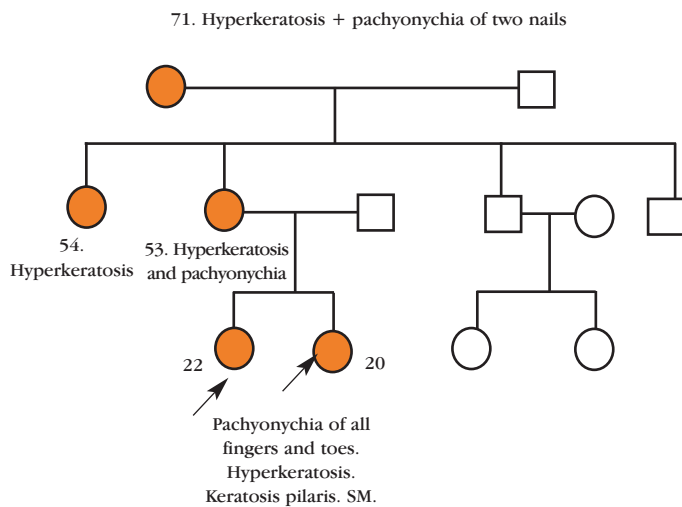


FIGURE 1: Genogram. Age and clinical manifestations of affected individuals. Clinical variability and peculiarity of exclusive involvement in women



FIGURE 2: Patient 1 - pachyonychia and oral leukoplakia



FIGURE 3: Patient 2 - plantar hyperkeratosis in areas of pressure and SM

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DISCUSSION

Pachyonychia congenita (PC) is classified into two major subtypes: PC-1 and PC-2.^{1,3} Both can manifest as nail dystrophy, hyperhidrosis and palmoplantar keratoderma, oral leukoplakia and keratosis pilaris.³ Steatocystoma multiplex (SM) is a rare disease characterized by multiple dermal cysts involving the pilosebaceous glands, being considered an exclusive manifestation which distinguishes PC-2.^{2,3} Oligosymptomatic forms of PC are frequent.^{2,4} Also known as Jackson-Lawler syndrome, PC-2 can be confirmed by KRT6B (*encoding keratin, type II cytoskeletal 6B*) and KRT17 (*encoding keratin, type I cytoskeletal 17*) gene research. Phenotypic variations of unknown cause within the same family are reported and subtypes may overlap.^{2,4} Half of the cases are due to de novo mutations and the other half is inherited as an autosomal dominant disease, with 100% penetrance, with the recessive form being described.⁵ Identical mutations in the keratin 17 gene may manifest only with SM and/or PC-2.^{6,7} More than 11 distinct

mutations are described, but the resulting phenotype does not depend on the type of mutation.⁶

One of the lesions was excised and an anatomopathological study was conducted in case 2. Involvement of the tympanic membrane is rarely reported. It was confirmed in one of the patients, which could justify her recurring otalgia, but it is traditionally described in PC-1.¹ Hoarseness, in turn, is described in both subtypes.^{1,3} Intriguingly, in this family of three generations affected and in which only women were affected, only two cases presented with SM. No sex predilection has been described.^{8,9} There is great variability of expression among family members and there are early symptoms, more severe and diverse in patient 1.

Symptomatic treatment of manifestations and family orientation as to the role of genetics were adopted. The use of retinoids in the most exuberant cases was discussed. □

Abstract: Pachyonychia Congenita is a rare genodermatosis of keratinization, first described in 1906 by Jadassohn and Lewandowsky. Besides not being well known, phenotypic variability and oligosymptomatic subtypes make the diagnosis difficult. We report a family with three generations affected, until recently not diagnosed. The active search for familial cases in patients with suspicious manifestations and identification of peculiar characteristics of its subtypes, as multiplex steatocystoma, provide early clinical diagnosis. In addition, nurture the family counseling and informations about prognosis.

Keywords: Genetic variation; Keratin-17; Oligosymptomatic patients; Pachyonychia congenita

Resumo: A paquioníquia congênita é uma rara genodermatose da ceratinização, primeiramente descrita em 1906 por Jadassohn e Lewandowsky. Além de pouco conhecida, a variabilidade fenotípica e as formas oligossintomáticas dificultam o diagnóstico. Relatamos uma família com três gerações afetadas, até recentemente sem diagnóstico. A busca ativa por casos familiares em pacientes com quadro suspeito e a identificação de manifestações peculiares de seus subtipos, como esteatocistoma múltiplo, permitem diagnóstico clínico precoce. Além disso, oportunizam a orientação familiar e de prognóstico ao portador.

Palavras-chave: Pacientes oligossintomáticos; Paquioníquia congênita; Queratina-17; Variação genética

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