Pachyonychia Congenita: A Case Report

Marwa A. Abo Elmagd, and Rasha I. Mohamed
Department of Dermatology, Venereology and Andrology, Faculty of Medicine, Sohag University

Introduction
Pachyonychia congenita (PC) is a rare autosomal dominant disorder of keratinization[1]. It was first documented by Muller in 1904[2] followed by Jadassohn and Lewandowsky in 1906[3]. It is classified into four types, of which the two important ones include type-1 (Jadassohn–Lewandowsky type) and type-2 (Jackson–Lawler type). These are characterized by subungual hyperkeratosis, focal palmoplantar keratoderma, oral leukokeratosis, which are usually present since birth.[4]

Case Report
An 7-year-old boy born of consanguineous parentage, with normal developmental milestones for his age except for natal teeth; also presented with nail defects since birth along with numerous skin lesions. Family history was unremarkable. Cutaneous examination revealed dystrophic, discolored, and thickened toenails and one fingernail along with massive subungual hyperkeratosis producing a distal elevation of nail plates and wedge-shaped deformity of the nails. This resulted in the upward growth of the distal edge of the nail plates [Figure 1],[Figure 2],[Figure 3]. Besides, there were numerous, hyperkeratotic lesions over the entire body, concentrated over both knees, legs and flanks [Figure 4],[Figure 5]. Marked hyperhidrosis of the palms and soles was observed. Palmoplantar keratoderma was present, along with painful ulcerated plaques [Figure 6]. Routine laboratory investigations including complete hemogram, hepatic profile, and renal profile were within normal limits. KOH microscopy and culture of nail clippings was negative. Skin biopsy from a hyperkeratotic lesion from the leg, showed orthokeratosis and acanthosis.[Figure 7]. No evidence of any malignancy was found during the thorough work up. Genetic and molecular biological studies could not be carried out due to lack of infrastructure facilities. Based on the above findings, he was diagnosed as pachonychia congenita type II.
Figure 2. Dystrophic fingernail with wedge-shaped deformity.

Figure 3. Natal teeth.

Figure 4. Follicular hyperkeratosis of knees.

Figure 5. Hyperkeratotic plaque.

Figure 6. Plantar keratoderma.

Figure 7. Showing orthokeratosis and acanthosis (H and E, ×10).
Discussion

Pachyonychia congenita (PC) is a rare, but well characterized autosomal dominant disorder of keratinization characterized by a triad of sub-ungual hyperkeratosis with accumulation of hard keratinous material beneath the distalportion of the nails, lifting the nails from the nail bed, keratosis palmaris et plantaris with thick callosities, especially on the soles andthick white areas on the oral mucosa [5]. According to these mutations, various clinical variants have been described .

PC type I (Jadassohn-Lewandowsky, PC-I) consists of palmoplantar hyperkeratosis, follicular hyperkeratosis, and oral leukokeratosis. Occasionally, bullous lesions, hoarse voice due to laryngeal involvement, warty lesions on knee and elbow, and hyperhidrosis may occur.

In PC type II (Jackson-Lawler, PC-II), the palmoplantar keratoderma and oral changes are of less importance or may be absent. In addition, history of natal teeth and the development of epidermal cysts or steatocysts are remarkable [6].

PC type III (Schafer-Brunauer, PC-III), includes combined features of types 1 and 2 with angular cheilitis, corneal dyskeratosis, and cataracts. Type IV includes features of type 1 and type 3 with laryngeal lesions, hoarseness of voice with mental retardation, hair abnormalities and alopecia. Rare variants include pachyonychia congenita tarda, characterized by isolated nail changes that usually begin in the second and third decades of life [7]. These different presentations are currently known to be due to mutations in variable genes encoding one of the paired epidermis keratins, K6α/K16 in PC-I and K6b/K17 in PC-II [8].

Complications like respiratory distress due to laryngeal leukokeratosis and acroosteolysis, malignant changes in palmoplantar lesions can occur in pachyonychia congenita [9]. In milder forms of pachyonychia congenita, local emollients and keratolytics have been used with considerable improvement. Oral retinoids have been demonstrated to improve the hyperkeratotic skin lesions. Retinoids given for long periods produce a reasonable degree of flattening of the nails [10]. The only effective treatment for nail lesions is surgery with radical excision of the nail, nail bed and nail matrix and skin implantation at the site of improved nail. Surgical treatment is also important in case of oral lesions with hoarseness or respiratory problems. When the familial mutation is known, genetic counseling can be done and if required, prenatal diagnosis can be done at early stage of pregnancy by chorionic villi biopsy [11].

References


11. Smith FJ, McKusick VA, Nielsen K et al., Cloning of multiple keratin 16 genes facilitates prenatal diagnosis of pachyonychia congenita type I. Prenat Diagn 1999; 19: 941-946. PMID: 10521820