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Pachyonychia Congenita: A Case Report

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Abstract

Observations: Pachyonychia congenita is an autosomal dominant genodermatosis characterized by subungual hyperkeratosis of the distal nails and focal palmoplantar hyperkeratosis. It is classified as pachyonychia congenita type 1 (*Jadassohn-Lewandowsky*) and pachyonychia congenita type 2 (*Jackson-Lawler*) syndromes. Herein a case of Pachyonychia congenita type 1 in a middle aged Kashmiri female is reported in view of the clinical rarity of this condition.

Introduction

Pachyonychia congenita (PC) is a rare genodermatosis. *Müller* was the first to document this condition in 1904, following reports published in 1905 by *Wilson* and by *Jadassohn* and *Lewandowsky* in 1906 [**1, 2**]. There are two main types of PC which have been recognized: (1) pachyonychia congenita type 1 (*Jadassohn-Lewandowsky* type) and, (2) pachyonychia congenita type 2 (*Jackson-Lawler* type). It can affect all races and equally affects both sexes. The lesions are not life threatening but can be disfiguring. The classic type 1 (*Jadassohn-Lewandowski* syndrome) is the most common variant and is due to mutations of the K6a and K16 genes that disrupt assembly of the keratin filament. The characteristic clinical features include hypertrophy and distortion of nails, palmoplantar keratoderma, follicular keratoses, varicosities over knees, elbows, buttocks and popliteal area and oral leukokeratoses are variably present [**3, 4**]. Other associated features which may occur, include bullae on palms and soles, hyperhidrosis of the palms and soles, natal or neonatal angular cheilosis, steatocystoma multiplex, hair anomalies, alopecia, corneal dyskeratosis, hoarseness, laryngeal lesions, cataract, polydactyly and mental retardation [**3, 4**]. Pachyonychia congenita type 2 (*Jackson-Lawler* type) is associated with mutations in the K6b and K17 genes. Symptoms are similar to PC type 1; however, there is increased evidence of cyst formation (steatocystoma multiplex) and natal teeth in PC type 2 [**4**].

Case Report

A 45-year-old female, born of a consanguineous marriage presented with chief complaint of hypertrophic brittle nails since her childhood. Her finger and toe nails both were affected and the nail beds were hypertrophied with discoloration and longitudinal fissuring. She also gave a history of thick hypertrophic plaques over palms and soles. The patient had no history of cutaneous blisters, natal teeth or any ocular abnormality. She was otherwise well except for a recent history of

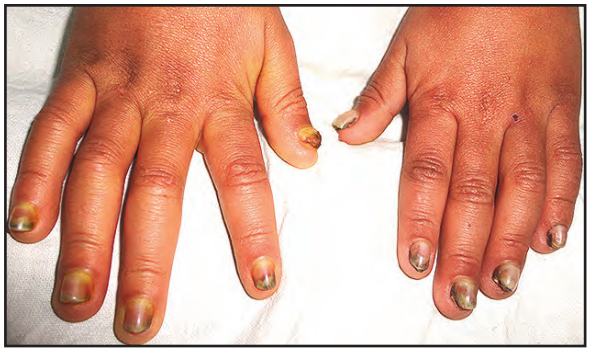

Figure 1. Yellowish discoloration subungual hyperkeratosis and marked thickening of the fingernails

paronychial infection. Soles were severely affected causing discomfort and pain on walking. There was a history of associated excessive sweating of palms and sole. She had a family history of the same disorder in her mother and aunt. Patient's dental and medical histories were non-contributory. General physical examination as well as systemic examination was normal.

Cutaneous examination revealed yellow discoloration and marked thickening of the fingernails and toenails, with subungual hyperkeratosis (**Figure 1**). There was an associated palmoplantar keratoderma with mild palmoplantar hyperhidrosis (**Figure 2 and 3**). There was no oral leukokeratosis. Results of KOH examination of skin scrapings as well as nail clippings were negative. Fungal culture from the nails yielded no organisms. X-rays of hands/feet did not reveal any abnormality. Patient refused a skin biopsy. With all these clinical findings, a diagnosis of pachyonychia congenita type 1 (*Jadassohn Lewandowsky* syndrome) was made and patient was put on oral vitamin A, keratolytic agents and emollients.

Discussion

Pachyonychia congenita (PC) is a rare, but well characterized autosomal dominant disorder of keratinization characterized by a triad of subungual hyperkeratosis with accumulation of hard keratinous material beneath the distal portion of the nails, lifting the nails from the nail bed, keratosis palmaris et plantaris with thick callosities, especially on the soles and thick 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 chielitis, 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

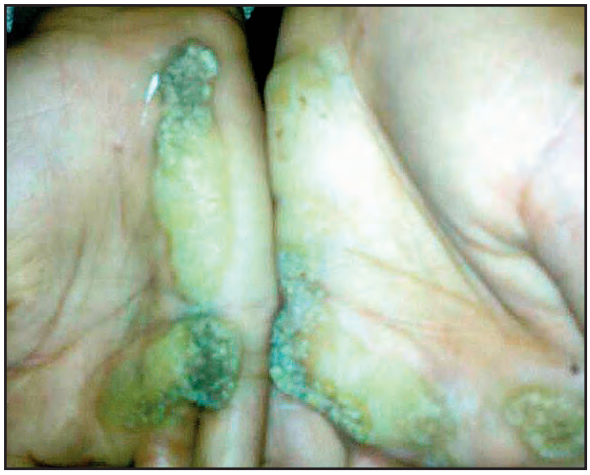

Figure 2. Palmar keratoderma

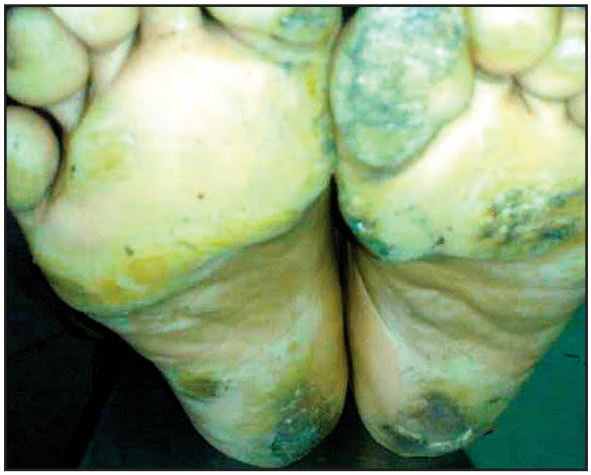

Figure 3. Plantar keratoderma

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, K6a/K16 in PC-I and K6b/K17 in PC-II [**8**].

Complications like respiratory distress due to laryngeal leucokeratosis and acro osteolysis, 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**].

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