

A familial case of pachyonychia congenita

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Abstract Pachyonychia congenita (PC) comprises a group of rare autosomal genodermatosis caused by mutation in any of the four genes KRT6A, KRT6B, KRT16 or KRT17. Classically, it is subdivided into two major variant types, PC-1 (Jadassohn-Lewandowski syndrome) and PC-2 (Jackson-Lawler syndrome). We hereby report a case of 22-year-old, married woman with progressive thickening and discoloration of all 20 nails, multiple, hyperkeratotic lesions present all over the body with oral lesions since childhood. She had a 2-month-old male baby (the only child) who presented with similar lesions of yellowish discoloration and nail thickening of both nails and foot since birth. She was diagnosed as PC type 1

Key words

Hyperkeratotic lesions, oral lesions, nails, pachyonychia congenita.

Introduction

Pachyonychia congenita (PC) is a group of inherited ectodermal dysplasia in which the most prominent feature is hypertrophic nail dystrophy.¹ Muller and Wilson described the first case of PC and one year later similar case was reported by Jadassohn and Lewandowsky.^{2,3} There are two main types of PC recognized: (1) pachyonychia congenita type - 1 (Jadassohn-Lewandowsky type) and, (2) pachyonychia congenita type - 2 (Jackson-Lawler type). PC type 1 is the more common subtype. PC type 2 is distinguished by the development of natal teeth, widespread steatocystomas, and occasionally pili torti. A third variant, pachyonychia congenita tarda has also been described and is characterized by a later onset that ranges from late childhood to middle age.

Case Report

A 22-year-old, married woman, born of non-consanguineous marriage, presented in our skin OPD with progressive thickening and discoloration of all 20 nails, multiple, hyperkeratotic lesions all over the body with oral lesions since childhood (**Figure 1**). Painful thickened lesions were also present over her palms and soles since last 10 years (**Figure 2**). She complained of burning sensation and difficulty in walking which aggravated on friction and cold climates. Sweating was more marked over the foot than the hands. She also had frequent nail infections. There was no history of natal teeth. She had no known medical problems and her growth and development was normal. She had a 2-month-old male baby (only child) who presented with similar lesions of yellowish discoloration and nail thickening of both hands and feet since birth with no neonatal teeth; face covered with tiny small white lesions since 7 days (**Figure 3**).

On examination white patches were seen on the dorsum of tongue and multiple, scattered, dirty looking, hyperpigmented hyperkeratotic papules

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Figure 1 Multiple, hyperkeratotic lesions present all over the body with oral lesions.



Figure 2 Painful thickened lesions (focal keratoderma) were also present over her palms and soles.



Figure 3 Two-month-old baby with miliaria crystallina on the face yellowish discoloration of the all twenty nails and subungual hyperkeratosis



Figure 4 All 20 nails were yellowish brown, thickened (wedge shaped) with subungual hyperkeratosis.

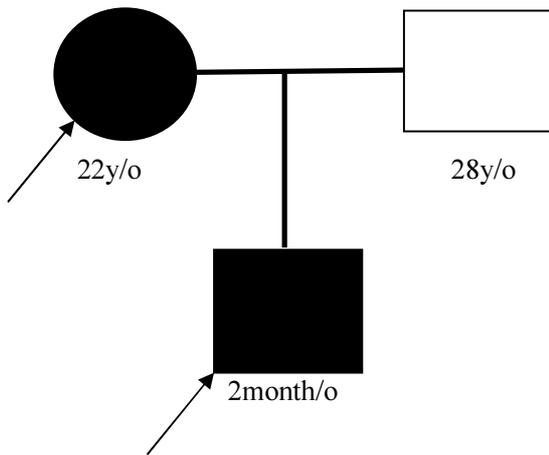


Figure 5 Patient's family tree.

over face, trunk and extremities with some skip lesions. All 20 nails were yellowish brown, thickened (wedge shaped) with subungual hyperkeratosis (**Figure 4**). There was also painful focal keratoderma at pressure points on palms and soles.

All routine investigations were within the normal limits. Potassium hydroxide (KOH) mount was done to rule out fungal etiology. Ophthalmological tests and otolaryngeal examination were within normal limits. Skin biopsy was not done due to unwillingness of the patient and genetic study could not be done due to the unavailability in our tertiary care centre. With all these clinical findings and family history (**Figure 5**), a diagnosis of PC type 1 (Jadassohn-Lewandowsky) was made and patient was managed conservatively by keratolytics like urea and lactic acid preparations. Her child had miliaria crystallallina for which no treatment was given as it was self-limited.

Discussion

PC is a rare, autosomal dominant disorder characterized by 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.^{4,5} Other associated features which may occur include keratosis pilaris, hyperkeratotic follicular papules on the sites of friction, hair abnormalities and hyperhidrosis of the palms and soles. These disorders have been suggested to be due to mutations in paired keratins, K6A/K16 (in PC1) and K6B/K17 (in PC2).^{6,7} According to these mutations, various clinical variants have been described:⁷

Type 1 - PC1: Jadassohn-Lewandowsky syndrome which is characterized by focal palmoplantar keratoderma (PPK), follicular keratosis mainly on the trunk and oral leucokeratosis.

Type 2 - PC2: Murray-Jackson-Lawler syndrome is the most common form associated with mild focal PPK, pili torti, natal teeth and multiple epidermal cysts. Angular cheilitis, hoarseness, follicular keratosis may be present in both types,

Type 3: includes combined features of types 1 and 2 with angular cheilitis, corneal dyskeratosis, and cataracts.

Type 4: comprises features of type 1 and type 3 with laryngeal lesions, hoarseness of voice with mental retardation, hair abnormalities and alopecia.

Other rare variants include pachyonychia congenita with only nail involvement and pachyonychia tarda that is pachyonychia congenita with onset in teenage years.^{8,9} Complications like respiratory distress due to laryngeal leucokeratosis and acroosteolysis, malignant changes in

palmoplantar lesions can occur in pachyonychia congenital.⁷

Treatment options for PC fall into four broad categories: (1) non- invasive (mechanical) e.g. abrasion with some hand tool; (2) invasive (surgical) e.g. electrofulguration, excision; (3) chemical methods using urea, propylene glycol, alpha hydroxy acid; and (4) pharmacological (vitamin A, retinoids), all basically targeted at reducing the hyperkeratosis involving different sites.¹⁰

In our case we gave emollients and keratolytic agents such as urea and lactic acid. Vitamin A preparations, topical retinoids or oral acitretin, could not be given as the patients was in the lactational period.

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 villous biopsy. For all these reasons, a patient with PC should be thoroughly investigated and treated accordingly.

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