

## Pachyonychia Congenita Tarda with Only Nail Involvement: A Rare Occurrence

*To the Editor.* - Pachyonychia congenita (PC) is a rare genodermatosis characterized by hypertrophy of the nail; often associated with nail bed and hyponychial hyperkeratosis [1]. Four varieties have been described; PC-1 being the most common followed by PC-2. When the onset is later in life, it has been called Pachyonychia congenita tarda (PCT) [2]. Several cutaneous features are associated with these conditions which vary according to the type of PC. PC-4 or PCT is usually associated with flexural hyper pigmentation, cystic lesions on the upper torso [2] and hidratenitis suppurativa [3]. Isolated PCT, affecting only the nails is extremely rare; we present here such a case of isolated PCT.

A 38-year-old otherwise healthy gentleman, born of non-consanguineous parentage presented to us with complaints of gradually progressive thickening, brownish discoloration and atrophy of some of his nails, for the last 18 months. Initially the process involved two fingers of his left hand, and gradually it has spread to involve the other nails as well. The patient denied any history of itching, pain, swelling or redness around the nails; thus ruling out any infective or inflammatory reaction. However, he received prolonged antifungal treatment from outside; with no improvement. Even on further enquiry, no history of increased sweating, cutaneous blisters, natal teeth or ocular/oral abnormalities could be elicited. There was no history of similar illness affecting any of the family members. Dermatological examination revealed thickening, brownish discoloration and atrophy of almost all nails of his left hand (Figure 1), while a few nails of his right hand were also involved. The toe nails showed milder changes (Figure 2). Horny keratotic debris were found beneath the nail-plates which caused the ragged free edges to project upwards. His palms and soles were normal. No evidence of keratosis pilaris, palmoplantar blis-

tering, callosities, flexural hyper pigmentation (around neck, waist, axilla, knees etc.), and alopecia could be found. His mucosa, eyes and teeth revealed no abnormality. All routine biochemical investigations were within normal limits. KOH mount of the subungual debris showed no fungal elements. The patient refused a nail biopsy. Based on history, physical examination and the late age of onset; a provisional diagnosis of isolated Pachyonychia congenita was made. Curettage of the matrix and nail bed was done to provide some relief. The patient has been advised regular follow up, and oral Acitretin therapy is being contemplated in the future.

Pachyonychia congenita (PC); which was first described by Jadassohn and Lewandowski in 1906 [2], is characterized by hypertrophy of the nails which may be associated with nail bed and hyponychial hyperkeratosis. Barely 450 cases of PC have been reported since 1906, thus attesting its uncommon occurrence [4]. Although most cases are autosomal dominant in inheritance; recessive forms have also been reported [1]. Four types of PC have been described; each type has certain characteristic associated features apart from the obvious nail anomaly. PC-1 (Jadassohn-Lewandowski syndrome) is the commonest variant [2]; followed by Type II PC (Jackson-Sertoli syndrome); the other types being exceedingly rare. The four types of PC with their usual associated anomalies have been tabulated below (Table 1).

The first three types of PC usually arise within the first few months of life; however the fourth variety or Pachyonychia congenita tarda (PCT) usually arises during the second or third decade of life [3]. However, it can manifest as early as ten years [5] or as late as fifty-five years [6]. In our case, nail dystrophy started in the fourth decade, thus corroborating with a diagnosis of PCT.



**Figure 1.** Thickening, brownish discoloration and atrophy of almost all nails of his left hand



**Figure 2.** Mild thickening, brownish discoloration and atrophy of the nails of his right hand. Note the milder changes in the toe nails

**Table 1.** Associated Clinical Features with Different Varieties of Pachyonychia Congenita (PC) <sup>2,3</sup>

Type of Pachyonychia congenita(PC)	Associated clinical features
Type I PC ( Jadassohn-Lewandowski syndrome) (most common variety)	Palmoplantar hyperhidrosis, extensive follicular hyperkeratosis (also affecting the trunk), palmoplantar hyperkeratosis, blistering under the callosities, benign leukokeratosis of the mucosa, B-cell lymphoma (rare) [9] etc.
Type II PC (Jackson-Sertoli syndrome)	Almost same features as Type I with additional features of: Natal teeth and steatocystoma multiplex. (however, palmoplantar keratoderma is usually less severe and mucosal lesions may be absent).
Type III PC (Schafter-Branauer syndrome)	Corneal leukokeratosis, cataract.
Type IV PC (Pachyonychia congenita tarda) [our case]	Flexural hyper pigmentation (around neck, axilla, thighs, waist, flexural surface of knees etc.), cysts on the head, neck and chest at puberty, hidradenitis suppurativa.

PC-1 usually occurs as a result of genetic mutation affecting Keratin 6a or 16, while mutation affecting Keratin 6b or 17 results in PC-2. The mutation is hypothesized to adversely affect the functioning of small inhibitory RNAs (siRNA) and hedgehog signaling. While the exact genetic basis of PCT is not known, several authors speculate mutations in less critical sites of the keratins may be responsible for this delayed-onset variety [7].

All varieties of PC are associated with several features dyskeratosis of skin and mucous membranes other than the nail anomaly (Table 1). Isolated early-onset nail change is rare, and late-onset isolated nail dystrophy (PCT or PC-4) is even more unusual [3]. There are only a handful of reports in the English literature depicting isolated PCT [3, 7, 8, 9]. In our case also, the patient presented with this rare variety of isolated late-onset nail dystrophy or PC.

The common differentials to be considered in case of PCT are other focal palmoplantar keratodermas associated with oral leucokeratosis (our patient presented with normal palms, soles and mucosa), dyskeratosis congenita (onset of symptoms since birth), psoriasis (presence of typical erythematous skin lesions with micaceous scaling at characteristic sites), pityriasis rubra pilaris (presence of characteristic skin lesions with islands of sparing mainly over the joints), onychogryphosis congenita (onset at birth, hyperkeratotic nail with increased curvature), traumatic thickening of nails (history of trauma). Proper history and physical examination helped us to exclude these disorders.

Treatment is usually palliative and often frustrating. Avulsion of the nail may bring temporary relief. Vigorous curettage of the matrix and nail bed remains the most effective treatment (treatment adopted in our case). Recently some reports have suggested the possible role of Acitretin in the tre-

atment of PCT [2, 4]. However, the ideal permanent cure for PC would be a gene replacement therapy in which the defective PC gene would be replaced with a corrected version [5]; but this expensive treatment modality remains elusive in our resource-poor country.

Thus, here we present a rare case of isolated Pachyonychia congenita tarda (PCT); affecting only the nails. The rarity of such a case has prompted the present report.

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