



# Pachyonychia Congenita Project

15 March 2005

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We hope that making available the relevant information on Pachyonychia Congenita will be a means of furthering research to find effective therapies and a cure for PC.

## Case report

# Pachyonychia congenita tarda: very late onset

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A 55-year-old woman presented with changes involving all of her nails of 4 years' duration (Fig. 1a). The patient had no associated skin eruption, palmo-plantar hyperhidrosis, oral mucosal lesions, abnormal dentition, nor eye symptoms. She had been previously treated with multiple prolonged courses of systemic antifungals to no avail, but had not received any treatment over the past 2 years. Her parents were nonconsanguineous and her family history was negative for similar nail changes. The patient had diabetes for which she had been maintained on metformin for the past 8 years. She also underwent a hysterectomy a few months prior to the onset of her nail changes.

On examination, all 20 nails exhibited a green-yellow discoloration and severe subungual hyperkeratosis, which led to variable degrees of nail plate elevation. Only the fifth fingernails of both hands exhibited subungual hyperkeratosis covered by intact nail plates. The rest of her nails showed variable degrees of nail plate dystrophy. Interestingly, complete sparing of the portion of the nail plate overlying the lunula was noted in all the nails where the lunula was visible (the great toenails and most fingernails) (Fig. 1a inset, Fig. 1b). Examination of the rest of her skin and mucosal surfaces was nonrevealing. No corneal abnormalities were present on ophthalmologic examination. Direct microscopic examination of keratinous material from subungual debris and plantar skin (potassium hydroxide preparation) were negative. Periodic acid-Schiff stain of nail clippings revealed no fungal elements.

### Discussion

The appellation pachyonychia congenita (PC) was first coined by Jadassohn and Lewandowsky in 1906. It is now used to refer to a rare group of ectodermal dysplasias that are usually

inherited as an autosomal dominant trait. Yet, autosomal recessive and sporadic cases have been described.<sup>1-3</sup> The most widely accepted classification of PC defines four different subtypes. These have in common pathognomonic nail changes affecting all 20 nails, but differ in the associated ectodermal features.<sup>2,4</sup> The nails in PC exhibit variable degrees of elevation secondary to marked subungual hyperkeratosis, transverse over-curvature, discoloration, and dystrophy of the nail plates. Additional clinical features vary according to the PC subtype. Patchy palmo-plantar keratoderma (at times with blister formation), follicular hyperkeratosis, and oral leukokeratosis occur in type 1 PC. Natal teeth, multiple steatocystomas, epidermal and eruptive vellus hair cysts, and palmo-plantar hyperhidrosis characterize type 2 PC. Type 3 PC may have features of both types 1 and 2, in addition to corneal leukokeratosis and cataracts. Finally, laryngeal lesions with hoarseness, hair abnormalities, and mental retardation are characteristic of type 4 PC. Types 1, 2, and 3 are also referred to as Jadassohn-Lewandowsky, Jackson-Lawler, and Schafer-Brunauer, respectively.<sup>5</sup>

Pachyonychia congenita tarda (PCT) is thought to represent a delayed form of PC type 1 and may have similar underlying genetic defects affecting the keratin 6a/16 pair.<sup>5</sup> It is characterized by the onset of nail changes during the second or third decades of life. Since it was first described by Paller *et al.* in 1991, more than 10 cases of PCT, both familial and sporadic, have been reported in the literature.<sup>6-10</sup> The age of onset ranged between teenage years and 44 years.<sup>4,6-10</sup> Some cases of PCT were associated with palmo-plantar keratoderma,<sup>4,6-8</sup> leukokeratosis,<sup>4,9</sup> and cutaneous cysts,<sup>4,9</sup> while others presented with changes limited to the nails.<sup>4,7,10</sup> The reasons behind the late onset of PC in some patients as well as the exclusive involvement of the nails in others are not yet understood.



**Figure 1** (a) All fingernails and toenails showing the characteristic nail changes of PC. (a) inset: sparing of the lunula in the great toenail. (b) Sparing of the lunula in the fingernails. (c) Side view showing the marked elevation of the nail plates

The onset of PCT in our patient occurred at the age of 51 and manifested itself exclusively in the form of nail dystrophy. Other conditions including severe onychomycosis, subungual warts, crusted scabies, contact dermatitis, lichen planus, psoriasis, acrokeratosis paraneoplastica and Darier's disease may lead to extreme subungual hyperkeratosis.<sup>5</sup> However, the absence of previous personal and family history of psoriasis, the absence of other associated findings, the symmetrical involvement of all 20 nails, the lack of response to systemic antifungals, and the negative testing for fungi all supported the diagnosis of PCT.

Furthermore, since the expression of keratins 6a/16 in the nail unit is mainly confined to the nail bed with much lower levels being expressed in the nail matrix, one would expect PCT to affect predominantly the nail bed and to spare the nail matrix and its product, the nail plate. This is in accordance with the complete sparing of the lunula (which represents the visible part of the nail matrix) noted in the great toes and most fingers in our patient. The normal nail plates covering the marked hyperkeratosis in the fifth fingernails are also in support of this.

Our case is a good illustration of PCT limited to the nails. It also represents, to the best of our knowledge, the latest onset of PCT reported in the literature. PCT should be kept in mind when facing severe subungual hyperkeratosis affecting all 20 nails regardless of the age of the patient. In addition, in view of the wide differential diagnosis for subungual hyperkeratosis, we propose that the observed sparing of the

lunula may serve as a clinical sign in support of PC when associated symptoms are lacking and further testing is not available.

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