

## CASE REPORT

### **Pachyonychia congenita tarda presented in a 63-year old patient**

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#### **ABSTRACT**

Pachyonychia congenita (PC) is a rare autosomal dominant inherited dermatosis that appears in early infancy. A late onset of the disease (pachyonychia congenita tarda, PCT) has been described. We herein report a case of PCT that appeared in a 63-year old patient.

#### **INTRODUCTION**

##### **CASE REPORT**

A 63-year old woman presented with a rapidly progressive thickening and brownish discolouration of nails. Thickening and discolouration started in the index finger of the right hand then spread to involve all fingers and toe nails within 3 months. The patient sought medical advice and she received repeated courses of systemic antifungal therapy with no improvement. Hyperkeratosis of both feet occurred two months later. The patient had no history of cutaneous blisters, natal teeth or corneal dystrophy. There is no history of similar nail conditions in other family member.

Examination revealed subungual hyperkeratosis, thickening, hypercurvature of the transverse axis of nail plates together with brownish discolouration. Fingernails were more affected than toe nails (Fig. 1, 2). Both feet showed hyperkeratosis (Fig. 3) and small areas of hyperkeratosis were found on the hypothenar eminences (Fig. 4).

KoH examination and culture for fungus were negative on repeated occasions. The patient refused to undergo nail plate biopsy but skin biopsy was taken from the hyperkeratotic area of the



**Fig. 1, 2** Subungual hyperkeratosis and thickening, transverse overcurvature with brownish discolouration of fingernails and toe nails.

hand to exclude other disorders of keratinisation. Histopathological examination of the skin biopsy revealed orthokeratosis and mild acanthosis.

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**Fig. 3** Planter hyperkeratosis.

## DISCUSSION

Pachyonychia congenita (PC) is a rare autosomal dominant genodermatosis that occurs due to mutation in one of 4 keratin genes.<sup>1</sup> PC has been subdivided into four groups, based on the clinical features associated with nail changes.<sup>2</sup> These have in common pathognomonic nail changes affecting all 20 nails, but differ in the associated ectodermal features.<sup>3</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.<sup>4</sup> Common to almost all patients who have been described, regardless of the form of inheritance or subclassification of the disorder, is the onset of pachy-

nychia in infancy.<sup>5</sup>

A late onset of the disease was first described by Paller et al in 1991,<sup>6</sup> in which symptoms are presented in second, third or later decades of life. This is called pachyonychia congenita tarda (PCT). Both familial and sporadic forms, have been reported.<sup>7</sup> Some cases of PCT were associated with palmoplantar keratoderma,<sup>8</sup> while others presented with changes limited to the nails.<sup>6,9</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. No established classification mentions a late onset form of this disorder.<sup>10</sup>

The onset of PCT in our patient occurred at the age of 63 which is, to our knowledge, most delayed reported age of onset for PCT. The previous such delayed onset of PCT was reported by Bahady et al. in 2008 in which PCT was manifested as isolated nail changes in a 51 year-old patient.<sup>11</sup> Other conditions including severe onychomycosis, subungual warts, crusted scabies, contact dermatitis, lichen planus, psoriasis and Darier's disease may lead to extreme subungual hyperkeratosis.<sup>12</sup> However, the absence of previous personal and family history of psoriasis, the absence of other associated findings, the symmetrical involvement



**Fig. 4** Areas of hyperkeratosis over the hypothenar eminences.

of all 20 nails, the lack of response to systemic antifungals, the negative testing for fungi and the histopathological picture of skin biopsy, all supported the diagnosis of PCT.

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