

Pachyonychia congenita with late onset (PC tarda)

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ABSTRACT

Pachyonychia congenita is a rare type of ectodermal dysplasia further classified into 4 types. Cutaneous manifestations seen in most of the cases of Pachyonychia congenita include palmoplantar keratoderma, follicular hyperkeratosis, wedge shaped nails, oral leukokeratosis and woolly hair. A 25-year-old male presented to us with thickened nails and scanty scalp hair. On examination, we noticed hyperkeratotic plaques over both the soles, palmoplantar hyperhidrosis and yellowish discoloration, wedging with subungual hyperkeratosis of all the nails. Follicular hyperkeratotic papules and steatocystoma multiplex were also observed over the scalp and face. The patient had history of natal teeth and on dental examination, lower central incisors were absent. All cutaneous changes in our case had manifested first in the 2nd decade except for natal teeth. All the above features suggested the diagnosis of pachyonychia congenita with late onset (PC tarda), which is an infrequently reported rare variant.

Key words: Late onset pachyonychia congenita, pachyonychia congenita, Pachyonychia congenita tarda

INTRODUCTION

Pachyonychia congenita (PC) is a rare type of ectodermal dysplasia with defects in keratin. Inheritance is autosomal dominant with incomplete penetrance but autosomal recessive and sporadic cases have been reported.^[1,2] It was first documented by Muler in 1904 and later published by Wilson in 1905, and it was Jadassohn and Lewandowsky who first described the association of palmoplantar keratoderma with ectodermal dysplasias.^[3] Pachyonychia congenita has been classified into two types. PC-1 (Jadassohn and Lewandowsky type) is the most common type characterized by palmoplantar keratoderma, follicular hyperkeratosis, wedge-shaped nails, oral leukokeratosis, and hyperhidrosis. In PC-2 (Murray–Jackson–Lawler type), apart from features of type 1, natal teeth, dry kinky scalp hair and steatocystoma multiplex are present. Rarely, onset can be delayed till adult life, which is termed as PC tarda. We herein report a case of late-onset type of pachyonychia congenita who presented with hyperkeratotic plaques over soles, wedge-shaped nails, woolly hair, and follicular papules along with steatocystoma multiplex over scalp.

CASE REPORT

A 25-year-old man born out of a non-consanguineous marriage presented to us with thickened nails that had developed around 12 years of age. Over next few years, he developed multiple, small raised lesions over the scalp, eyebrows and beard area followed by gradual loss of hair over these areas. History of natal teeth, development of blisters on palms and soles following minor trauma, and focal thickening of palms and soles was present. No other family members had similar complaints.

On general examination, there was no pallor, icterus, clubbing, cyanosis or lymphadenopathy.

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Cite this article as: Sravanthi A, Srivalli P, Gopal K, Rao TN. Pachyonychia congenita with late onset (PC tarda). Indian Dermatol Online J 2016;7:278-80.

Access this article online

Website: www.idoj.in

DOI: 10.4103/2229-5178.185463

Quick Response Code:



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Systemic examination was normal. On dermatological examination, there was a hyperkeratotic plaque over the left sole with surface showing ulceration [Figure 1] and was associated with palmoplantar hyperhidrosis. All finger and toe-nails were wedge-shaped with yellowish discoloration and subungual hyperkeratosis [Figure 2]. Over the scalp, multiple soft discrete shiny nodulocystic lesions were present [Figure 3]. Follicular papules and sparse woolly hair were seen over scalp, beard, and eyebrows [Figure 4]. The central lower incisors were absent as natal teeth had fallen prematurely and had not been replaced [Figure 5]. Oral leukokeratosis was absent.

Nail clippings for KOH mount and fungal culture were negative. Skin biopsy of hyperkeratotic lesion on the sole showed orthohyperkeratosis, parakeratosis, and acanthosis. Complete blood picture, routine blood biochemistry, and urine analysis were within normal limits. On puncturing a cystic lesion over scalp, sebaceous material was obtained. Histopathological examination of follicular papules over eyebrows revealed follicular orthokeratotic keratin plug and mild perivascular mononuclear infiltrate suggestive of follicular keratoses. Due to lack of infrastructure, genetic testing could not be conducted. The patient was managed with Acitretin 25 mg per day and his nails were pared for symptomatic relief. He was also prescribed 40% urea cream for thickened nails and iontophoresis was performed regularly for hyperhidrosis.

DISCUSSION

Pachyonychia congenita is a rare genodermatosis and is classified into two types.^[4] Type 1 ((Jadassohn and Lewandowsky type) is due to mutations in keratin 6a and 16. Nails are normal at birth but within months they become discolored and progressively thickened and are associated with palmoplantar hyperhidrosis, acral bulla, and oral leukokeratosis. Type 2 (Murray–Jackson–Lawler syndrome) is due to mutations in keratin 6b and 17 and is distinguished from type 1 by natal teeth, steatocystoma multiplex, and woolly hair with other manifestations being less severe.^[5,6] Pachyonychia congenita tarda is a rare variant with onset later in life ranging from the second decade to middle age.^[7]

Mutations in K6A, K6B, K16, K17 genes cause pachyonychia congenita.^[8] These genes encode tough fibrous proteins called keratins, which form a network that provides strength and resilience to the skin, hair, and nail tissues. Mutations in any of the above keratin genes cause structural alterations in keratin proteins, which are unable to form a stable network within the cells. Without this network, skin cells become fragile and susceptible to friction and minor trauma resulting in the formation of severe painful blisters and callosities. Defective keratin also results in abnormal growth and function of cells in the nails and hair follicles leading to discolored, abnormally thickened nails.

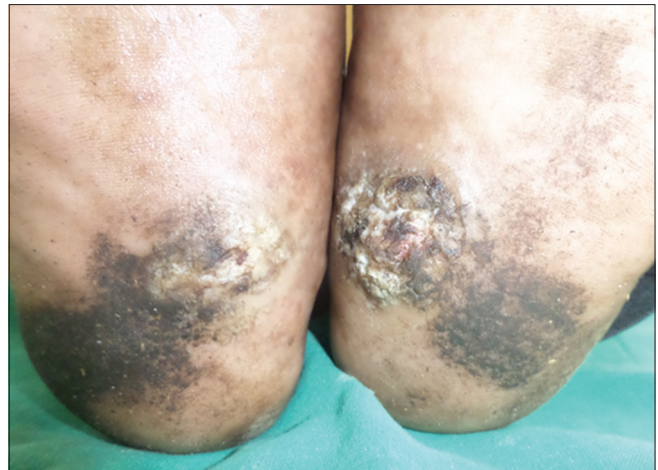


Figure 1: Focal hyperkeratotic plaques over both soles with dry ulcerated surface



Figure 2: Thickened discolored wedge-shaped finger nails with subungual hyperkeratosis



Figure 3: Multiple nodulocystic lesions over the scalp

In the present case, the patient complained of thickened discolored nails and was concerned about his sparse unruly



Figure 4: Follicular papules and sparse woolly hair over scalp, beard and eyebrows

hair; poor hair growth; and extensive skin eruptions over the scalp, eyebrows, and beard region. On detailed examination, we noticed that the patient had focal hyperkeratotic plaques over the soles, palmoplantar hyperhidrosis, and extensive steatocystoma multiplex and follicular keratoses. All the cutaneous changes in our case manifested in the second decade along with steatocystoma multiplex, which favored a diagnosis of late onset pachyonychia congenita. Pachyonychia congenita tarda has been suggested as the late-onset variant of pachyonychia congenita, which is very rare with only a few cases reported till date.^[9,10] The common form of inheritance in pachyonychia congenita is autosomal dominant and infrequently, autosomal recessive, and sporadic cases have been reported. Our case can be considered as sporadic as there is no consanguinity or family history of similar abnormalities.

Declaration of patient consent

The authors certify that they have obtained all appropriate



Figure 5: Absent lower central incisors

patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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