Pachyonychia Congenita Type 1: Case Report and Review of the Literature

Praveen Kumar Rathore, Varun Khullar, Anupam Das

Abstract
The case of an 8-year-old boy is hereby reported, who presented with nail dystrophy, subungual hyperkeratosis, oral leukokeratosis, and numerous follicular papules all over the body. The features were consistent with a diagnosis of pachyonychia congenita type 1. The case is being reported for its rarity. We also discuss in a nutshell, the literature till date.

Key Words: Follicular papules, leukokeratosis, nail dystrophy, subungual hyperkeratosis

What was known?
Pachyonychia congenita is a rare genodermatosis with a wide array of cutaneous manifestations including nail dystrophy, subungual hyperkeratosis, follicular papules, oral leukokeratosis, palmar/plantar keratoderma, etc.

Introduction
Pachyonychia congenita (PC) is a rare autosomal dominant disorder of keratinization.[1] It was first documented by Muller in 1904.[2] followed by Jadassohn and Lewandowsky in 1906.[3] It is classified into four types, of which the two important ones include type-1 (Jadassohn-Lewandowsky type) and type-2 (Jackson-Lawler type). These are characterized by subungual hyperkeratosis, focal palmar/plantar keratoderma, oral leukokeratosis, which are usually present since birth.[4]

Case Report
An 8-year-old boy born of nonconsanguineous parentage, with normal developmental milestones for his age; presented with nail defects since birth along with numerous elevated skin lesions. Family history was unremarkable. Cutaneous examination revealed dystrophic, discolored, and thickened fingernails and toenails, along with massive subungual hyperkeratosis producing a distal elevation of nail plates and wedge-shaped deformity of the nails. This resulted in the upward growth of the distal edge of the nail plates, accompanied by angulation of the lateral border [Figures 1-3]. Besides, there were numerous pin-head sized follicular papules over the entire body, concentrated over the face, back, buttocks, abdomen, and gluteal region [Figures 4-6]. Besides, hyperkeratotic lesions were distributed throughout the body. Marked hyperhidrosis of the palms and soles was observed. Palmar/plantar keratoderma was present, along with painful hyperkeratotic plaques [Figures 7 and 8]. Mucosal examination was significant for the presence of asymptomatic oral leukokeratosis over the dorsum of the tongue [Figure 9]. Routine laboratory investigations including complete hematogram, hepatic profile, and renal profile were within normal limits. KOH microscopy and culture of nail clippings was negative. Skin biopsy from a hyperkeratotic lesion around elbow showed orthokeratosis and acanthosis [Figure 10]. No evidence of any malignancy was found during the thorough work up. Genetic and molecular biological studies could not be carried out due to lack of infrastructure facilities. Based on the above findings, he was diagnosed as pachyonychia congenita type 1. The child was prescribed Vitamin A and E along with emollients and keratolytics. He was prescribed oral Vitamin A at a dose of 25,000 IU, under a multidisciplinary approach after consultation with

Access this article online
Quick Response Code:  
Website: www.e-ijd.org  
DOI: 10.4103/0019-5154.177761

How to cite this article: Rathore PK, Khullar V, Das A. Pachyonychia congenita Type 1: Case report and review of the literature. Indian J Dermatol 2016;61:196-9.  
Received: September, 2015. Accepted: December, 2015.
Department of Pediatrics and Ophthalmology. The patient is under stringent follow-up every 2 weeks. He has been referred to the Department of Physical medicine and rehabilitation for weight control exercises.

**Discussion**

PC is a rare genodermatosis with autosomal dominant mode of inheritance. Heterozygous mutations involving keratins K6a or K16 are associated with PC-1 whereas those involving K6B and K17 are associated with PC-2.[18] Although, autosomal dominant mode is the most common mode of inheritance, there are reports of autosomal recessive inheritance as well.[4]
PC has been classified into four types, the common clinical findings in all of them being painful and debilitating plantar keratodermas, nail dystrophy and hypertrophy, oral leukokeratosis, palmoplantar hyperhidrosis, and a variety of epidermal cysts.[7]

Patients with type 1 PC (Jadassohn-Lewandowsky syndrome) are characterized by the presence of nail dystrophy since birth. This may be accompanied with painful paronychia, hyperkeratosis of palms and soles over the pressure sites, oral leukokeratosis, palmoplantar hyperhidrosis, and follicular keratotic papules distributed throughout the body.[7,8] Besides, painful blisters also develop over the palms and soles. Another characteristic finding is the presence of verrucous lesions over the elbows, knees, popliteal fossae, and ankles. In addition, hoarseness of voice is also an important feature of PC type 1.[8,10]

In addition to the above mentioned findings, type 2 PC (Jackson-Lawler syndrome) has the features of natal teeth, hair anomalies including pili torti, unruly hair, and bushy eyebrows. Oral leukokeratosis and palmoplantar keratoderma is milder in comparison to type 1 PC, but the development of epidermal cysts or sebocysts are the hallmark findings in type 2 PC. PC type 3 (Schafer-Branuer) has features of conical keratoderma.

Type 4 PC is termed as PC tarda, which is manifested in the second or third decade of life. It results from mutations in the keratins 16 and 17 genes.[11]

PC with unusual features has been noted. Rare cases of pachyonychia congenita tarda have been reported with symptoms developing in the fifth decade of life.[12] Recently, a case of pachyonychia congenita associated with B-cell lymphoma has been reported.[13] Cases with unusual dental findings have also been reported.[14] An interesting case of PC with woolly hair in a 10 month old patient has been also reported.[15] Cases with isolated involvement of nails have also been described.[16] Apart from the numerous oral manifestations, median rhomboid glossitis in association with PC has been documented.[17]

The clinical features in our case are suggestive of PC type 1 with characteristic subungual hyperkeratosis and follicular papules over the entire body since birth. Histological findings of orthokeratotic hyperkeratosis and acanthosis confirm our diagnosis. The patient was
prescribed Vitamin A and E along with emollients and keratolytics. Mahajan et al. reported a case of PC, which was treated with oral Vitamin A and E. Vitamin A stimulates differentiation and leads to normalization of accelerated epidermopieerosis of pathological keratinocytes of epidermis of skin and nail. With this background, we have prescribed Vitamin A to our patient. The upper limit of dose of Vitamin A in pediatric patients (1–8 years) is 17,500–35,000 IU. Hypervitaminosis A occurs when the consumption is more than or equal to 100,000 IU for months. Our patient was referred to Department of Pediatrics and Ophthalmology. Thereafter, he was prescribed Vitamin A at a dose of 25,000 IU. The patient is under stringent follow-up every 2 weeks, for systemic check-up including fundoscopy and signs of irritability. For management of pain and discomfort due to the palmoplantar keratoderma, patient was advised limitation of walking and standing, use of soft shoes, control of body weight, and use of appropriate clothing. The patient is under periodic follow-up. Mechanism of manipulating gene expression is now known which in future, may help to suppress or correct the genetic defect in the chromosomes. The only effective treatment though is nail surgery with radical excision of the nail bed and matrix and grafting at the site but patient being very young, this line of treatment was not accepted by the parents.

**Declaration of patient consent**

The authors certify that they have obtained all appropriate 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.