Pachyonychia congenita tarda: A rare case report

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Abstract

Pachyonychia congenita is a rare, but well-characterized autosomal dominant disorder of keratinization. It usually begins within the first few months of life. Here, we are presenting a rare case, which started at the age of 10 years of life and is known as pachyonychia congenita tarda. The case is being reported for its rarer occurrence as the patient had oral leukokeratosis and angular cheilosis present in the same type of the syndrome (Jadassohn-Lewandowsky syndrome), which is still uncommon.

Keywords: Angular cheilosis, nails, oral leukokeratosis, pachyonychia congenita

Introduction

Pachyonychia congenita (PC) is a group of rare, inherited ectodermal dysplasias associated with mutations in keratin genes of K6a, K6b, K16 or K17.[1,2] The most prominent clinical features of PC are nail dystrophy and dyskeratosis of skin and mucous membranes.[1-3]

PC was first described by Muller in 1904 and Wilson in 1905; although, the association of the disorder with palmoplantar keratoderma and other ectodermal defects was reported by Jadassohn and Lewandowsky in 1906.[1,2]

Common to almost all patients who have been described, regardless of the form of inheritance or subclassification of the disorder, is the onset in infancy. Very few cases have been reported so far with late onset and considered as a rare variant of PC. The term pachyonychia congenita tarda (PCT) has been suggested to describe this late onset form of PC.[1-3] It is characterized by nail dystrophy, palmoplantar hyperkeratosis, leukokeratosis of mucous membranes, follicular hyperkeratosis, hyperhydrosis of palms and soles.[1-3] We herein report one such rarer case with oral leukokeratoses and angular cheilosis as components of the same syndrome.

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Case Report

A 45-year-old male patient was referred from Department of Dermatology, Government District Hospital for examination of whitish lesions in the mouth. History of present illness revealed that oral lesions along with thickening of nails and callosities on palms and soles were present since 10 years of age. These changes were spontaneous in origin and gradually increasing in severity. Soles were severely affected causing discomfort and pain on walking. Associated excessive sweating of soles and palms present. Whitish plaques in the mouth were asymptomatic. The patient was born of non-consanguineous parents.

Patient's dental and medical histories were non-contributory. His family history revealed that his father, brother and son have a similar condition, but declined clinical examination. No tissue abusing or parafunctional habits reported. General examination revealed that patient was moderately built and nourished for his age. Patient had an altered gait. Nails were extremely hard, thickened, opaque brown, lusterless and free edges were raised by a thick horny masses of subungual keratosis. Palmoplantar keratosis was more marked on pressure areas and elbows [Figures 1]. All vital signs were within normal limits.

Extraoral examination revealed no abnormalities. Solitary right and left submandibular lymph nodes were palpable, 1 cm × 1 cm, mobile and firm. On intraoral examination, full complement of teeth was present. Labial mucosa, ventral surface of tongue, floor of the mouth, vestibular mucosa and oropharynx revealed no abnormalities.

Leukokeratosis characterized by diffuse whitish, non-tender and non-scrapable plaque such as lesions of the both right and left buccal mucosae [Figure 2],

left retromolar region [Figure 2], dorsal aspect of the tongue [Figure 3], hard and soft palate and gingivae [Figure 4] were present. Angular cheilosis was present bilaterally [Figure 5].

All routine investigations such as complete hemogram, urine analysis and fasting blood sugar were within the normal limits. Potassium hydroxide examination of nail clippings and fungal culture of the nails were negative. Biopsy from palmar lesion was suggestive of palmoplantar keratoderma.

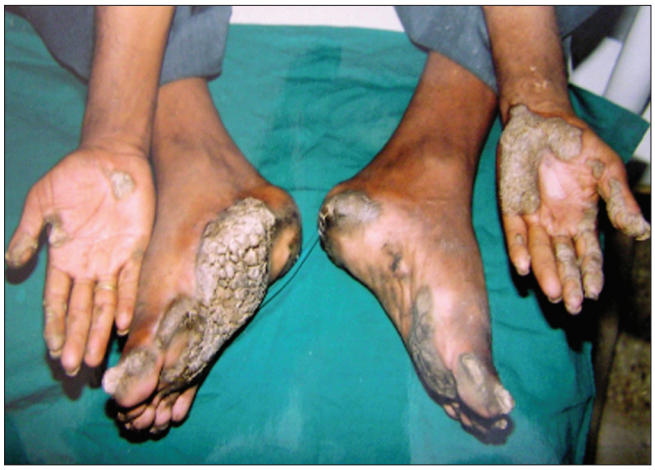

Figure 1: Palms and soles changes

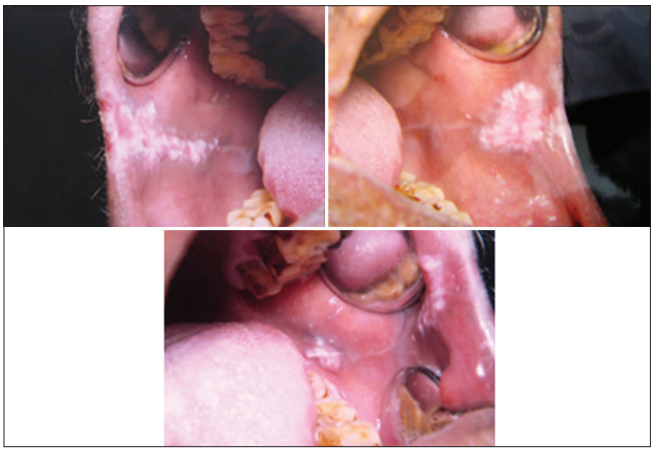

Figure 2: Leukokeratoses of buccal mucosae and retromolar region

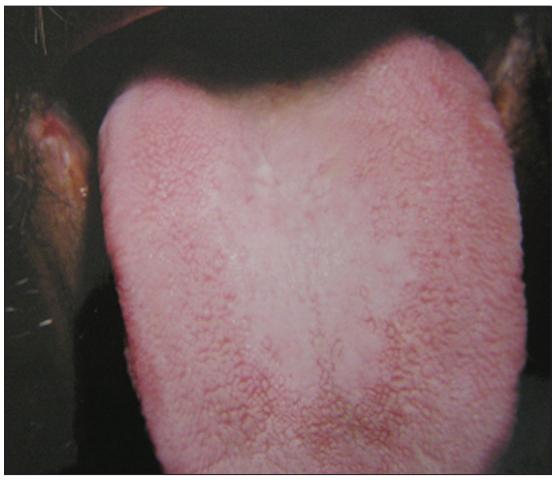

Figure 3: Leukokeratoses of tongue

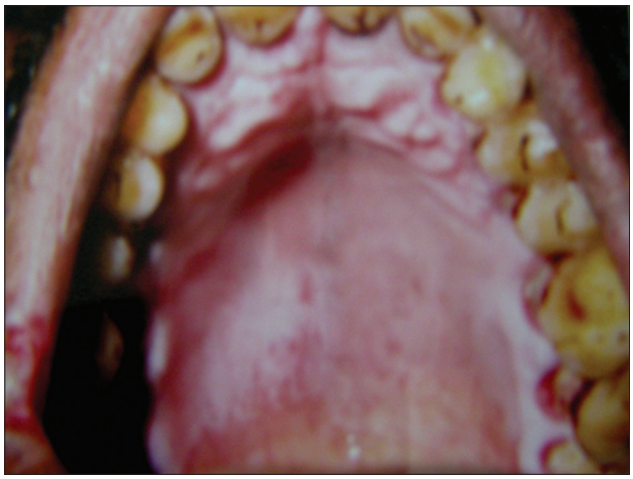

Figure 4: Leukokeratoses of the palate and gingivae

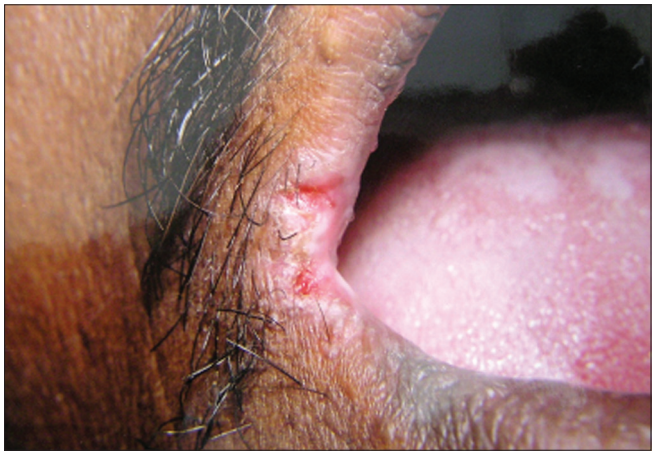

Figure 5: Angular cheilosis

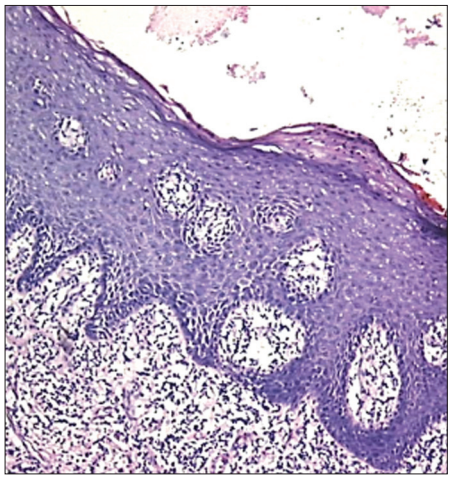

Figure 6: Histopathological features suggestive of leukokeratoses

Incisional biopsy was carried out on the left buccal mucosa and histopathological features were suggestive of oral leukokeratosis [Figure 6].

Based on the characteristic clinical presentation and history, a diagnosis of PCT was made. There is currently no specific treatment for PC. Available treatments generally are directed at specific manifestations of the syndrome. Oral lesions do not require treatment. For symptomatic treatment of extraoral lesions the patient was referred to the Department of Dermatology, Government District Hospital where he was put on oral vitamin A, keratolytic agents and emollients.

Discussion

The present case reports a patient with PCT as characteristic clinical changes had started in the second decade of life. Though dystrophy of all nails is the main feature of the syndrome and usually presents within the first month of life, recently few patients have reported with the onset of the characteristic nail changes during the second and third decades of life.[1-3] The term PCT was proposed to describe this rare subset of PC, representing exceptions that result from mutations elsewhere in the keratins 16 and 17 genes.[2-6]

The inheritance pattern of our case is highly suggestive of an autosomal dominant mode of inheritance as his father, brother and son have a similar condition and therefore consistent with the other families with PCT described.[3]

In classical PC hypertrophy and distortion of nails, hyperkeratotic skin lesions, such as palmoplantar keratoderma, follicular keratoses, varicosities over knees, elbows, buttocks and popliteal area and oral leukokeratoses, have been well documented to be variably present.[1-4,7] Other associated features, which may occur include bullae on palms and soles, hyperhidrosis of the palms and soles, natal or neonatal teeth, angular cheilosis, steatocystoma multiplex, hair anomalies, alopecia, corneal dyskeratosis, hoarseness, laryngeal lesions, cataract, polydactyly and mental retardation.[2,4,6]

These above mentioned clinical manifestations may vary and or could overlap as it is in our case. The case reported here has angular cheilosis in the Type I PC (Jadassohn-Lewandowsky syndrome). This could be because of PC has both autosomal dominant and autosomal recessive modes of inheritance with variable penetration, reflecting genetic heterogeneity.[2-4]

Complications like respiratory distress due to laryngeal leukokeratosis and acroosteolysis and malignant changes in palmoplantar lesions can occur in PC. Hence, patients with PC should be thoroughly investigated and treated accordingly as early as possible.[1,4] When familial mutation is known, genetic counseling and if required, prenatal diagnosis can be performed at the early stage of pregnancy, enabling new options for suitable therapeutic regimens. It even offers the hope of curing such type of skin diseases by means of somatic cell gene therapy.[1,4]

There are no reports available at the moment on the specific therapy of PCT. In classical PC, retinoids, oral vitamin A, keratolytic agents, emollients and surgery have been reported to be effective in alleviating some signs and symptoms of the disease.[3,4,8] Oral lesions do not require treatment. Angular cheilitis and fissures are usually treated with heavy emollients.[8,9] The ideal solution for a permanent cure for PC would be a gene therapy replacement procedure in which the defective PC gene would be replaced with a corrected version that would be regulated in identical fashion to the wild type gene.[4,10]

Conclusion

The classical as well as late-onset variants of PC are characterized by heterogeneity in expression of a number of associated features. It seems rational to distinguish PCT from PC by the differences in age of onset. Dentists should be able to recognize this condition in its early stages and advice appropriate investigations and management as dentists may be the first to see and diagnose this condition.

References

1. Kaur T, Gupta RR, Mahajan BB, Sachdeva R. Pachyonychia congenita type 1: Jadssohn Lewandowsky syndrome. Egypt Dermatol Online J 2010;6:1-6.
2. Leachman SA, Kaspar RL, Fleckman P, Florell SR, Smith FJ, McLean WH, *et al*. Clinical and pathological features of pachyonychia congenita. J Investig Dermatol Symp Proc 2005;10:3-17.
3. Lucker GP, Steijlen PM. Pachyonychia congenita tarda. Clin Exp Dermatol 1995;20:226-9.
4. Caproni M, Fabbri P. Pachyonychia congenita. Firenze, Italy: Orphanet Encyclopedia; 2003. p. 1-3.
5. Sharma VM, Stein SL. A novel mutation in K6b in pachyonychia congenita type 2. J Invest Dermatol 2007;127:2060-2.
6. Bansal A, Sethuraman G, Sharma VK. Pachyonychia congenita with only nail involvement. J Dermatol 2006;33:437-8.
7. Auluck A. Dyskeratosis congenita. Report of a case with literature review. Med Oral Patol Oral Cir Bucal 2007;12:E369-73.
8. Milstone LM, Fleckman P, Leachman SA, Leigh IM, Paller AS, van Steensel MA, *et al*. Treatment of pachyonychia congenita. J Investig Dermatol Symp Proc 2005;10:18-20.
9. Jayanthi P, Ranganathan K. Differential diagnosis of white lesions of oral mucosa. J Orofac Sci 2010;2:58-63.
10. Kaspar RL. Challenges in developing therapies for rare diseases including pachyonychia congenita. J Investig Dermatol Symp Proc 2005;10:62-6.

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