

Pachyonychia Congenita Associated with Oral Leukoplakia: A Rare Case Report with Review of Literature

¹Kamala KA, ²L Ashok, ³Rajeshwari G Annigeri

¹Senior Lecturer, Department of Oral Medicine and Radiology, Maratha Mandal's Nathajirao G Halgekar Institute of Dental Sciences and Research Center, Belgaum, Karnataka, India

²Professor and Head, Department of Oral Medicine and Radiology, Bapuji Dental College and Hospital, Davangere, Karnataka, India

³Professor and Head, Department of Oral Medicine and Radiology, College of Dental Science, Davangere, Karnataka, India

Correspondence: Kamala KA, Senior Lecturer, Department of Oral Medicine and Radiology, Maratha Mandal's Nathajirao G Halgekar Institute of Dental Sciences and Research Center, RS No. 47A/2, Bauxite Road, Belgaum, Karnataka, India
e-mail: kamsweetsmile@gmail.com; kamala.kamble@rediffmail.com

ABSTRACT

Pachyonychia congenita comprises a heterogeneous group of autosomal dominantly inherited conditions manifesting with characteristic nail thickening, palmoplantar keratoderma, follicular keratosis and oral manifestations like mucosal leukokeratosis. Less frequently epidermal cysts, hair-shaft abnormalities, natal teeth and laryngeal involvement may be seen. It is distributed almost evenly throughout the world affecting 1 in 100,000 populations. Here, we report a case of pachyonychia congenita type-1 with an added emphasis on dental findings in 45-year-old South Indian male patient.

Keywords: Pachyonychia congenita, Genodermatosis, Palmar plantar keratosis, Dental findings, Clinical features, Oral manifestations.

INTRODUCTION

Pachyonychia congenita (PC) is a group of autosomal dominantly inherited disease characterized by dystrophic nail, palmoplantar hyperkeratosis, accompanied by varying features of ectodermal dysplasia.¹ PC is a rare genodermatosis, usually inherited as an autosomal dominant trait with varying degree of penetrance. It is characterized by the marked subungual hyperkeratosis with thickening of the distal part of the nails. The other findings include palmar and plantar keratosis, hyperhidrosis, follicular hyperkeratosis and frequent blistering of the feet.² The oral manifestations in PC include leukokeratosis of tongue, leukoplakia, pilosebaceous cyst, neonatal teeth, angular cheilitis, and laryngeal involvement leading to the hoarseness of voice. Leukokeratosis of oral mucosa is a predominant feature of PC which mainly occurs on tongue, buccal mucosa and sometimes on gingiva.³ Only one case has been reported till today and to our knowledge, this is the first case report of PC associated with leukoplakia of right and left retrocommissural area, leukokeratosis of tongue, angular cheilitis and generalized periodontitis.

PC was first described by Muller and Wilsen in 1904, but it was Jadassohn Lewandowsky in 1906 who reported anomaly of nail, palmoplantar keratoderma, and ectodermal defects and gave the name PC.⁴

The manifestations of PC are chiefly subungual hyperkeratosis with marked thickening of the distal portions of the nails and severe disabling hyperkeratosis of the palms and

soles. The thickening results from the subungual hyperkeratosis with an upward angulation of the distal portion of the nail tip. The dorsal surface of the nail is smooth, whereas lateral borders often curve toward the center in pinched manner. Other manifestations include follicular hyperkeratosis, observed on face and on the extensor aspect of the proximal parts of the extremities, hyperhidrosis, particularly on palms and soles, corneal changes and epidermal inclusion cysts.⁵⁻⁷

PC usually begins in infancy, but late onset PC, referred to as pachyonychia congenita tarda, which begins in the fourth or fifth decade, has also been reported. It frequently affects Jewish and Yugoslavian race with female predominance.^{3,8,9}

CASE REPORT

A 45-year-old male patient was referred from the Department of Dermatology and Venerology, Chigateri General Hospital, to the Department of Oral Medicine and Radiology, Bapuji Dental College, Davangere, India, for the evaluation of oral lesions. Patient complained of bleeding gums and burning sensation of tongue from 3 months along with halitosis. The medical history of the patient revealed that he was diagnosed with PC type-1 after birth. Family history revealed that the patient had consanguineous marriage and his father, grandfather, brother and son also suffered from similar condition. All lesions of PC have started early in the life. His personal history revealed history of chewing raw tobacco 3 to 4 times daily for 5 years. He used to keep the tobacco quid on both right and left buccal vestibule.

General physical examination revealed a moderately built and nourished individual of normal gait with vital signs within the normal range. Nail examination showed hyperkeratotic fingernails (Fig. 1) and toenails with mild yellow discoloration (Fig. 2). Thickening of the toenails was more obvious than that of the fingernails. The affected nails showed thickening and hardening, subungual hyperkeratosis, and upward growth of the distal nail with hypercurvature and some of the nails had a 'pinched' appearance at the proximal aspect. The palms were hyperhidrotic, and marked hyperkeratosis was noted on the left palm. The follicular keratoses of both right and left elbows were also noted. Patient gave history of recurrent blister formation on his feet after prolonged walking, especially during summer. Patient also had photosensitivity and classic symptoms of PC, which included hyperhidrosis of palms and soles and hoarseness of voice. Hair and eyes were not affected. On palpation, all the lesions were nontender, firm in consistency and elevated.

Intraoral examination showed a feature of leukokeratosis of dorsal and lateral aspect of tongue with depapillation (Fig. 3) and bilateral angular cheilitis (Fig. 4). Grayish white patches were present on right and left retrocommissural area measuring approximately 1 × 1 cm on left side and 2.5 × 1 cm on right side (Fig. 5). Overlying mucosa was soft and

surrounding area appeared normal. On palpation the lesions were nontender nonscrapable and elevated above the surface. On periodontal examination (Fig. 6), local deposits with generalized severely inflamed, erythematous, and enlarged gingiva was observed with bleeding on probing. Loss of attachment with average probing depth of 5 mm, attrition and cervical abrasion were also observed.

Routine blood and biochemical investigations, like complete hemogram, urine analysis, liver function test, renal function test were carried out. All parameters were within the normal limits except for slightly raised erythrocyte sedimentation rate (ESR). To exclude onychomycosis (fungal infection of nails), potassium hydroxide examination of nail clipping was done and nail culture were examined for the presence of fungi which proved to be negative.

On routine radiographic examination, intraoral periapical radiographs showed no major changes and an orthopantomograph revealed only mild generalized bone loss. The radiographic examination of hands and feet showed acro-osteolysis, destructive changes in the distal phalangeal bone and bone erosion with soft tissue atrophy which was more marked in radiograph of feet (Fig. 7).



Fig. 1: Plantar and dorsal surface of hands. Hyperkeratosis of left palm and papules over dorsa of hands and subungual hyperkeratosis



Fig. 2: Sole and dorsum of feet. Keratoderma of soles and subungual hyperkeratosis with elevation of nail plate

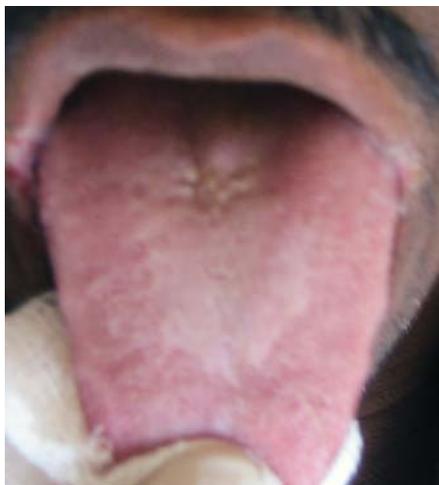


Fig. 3: Dorsal surface of tongue. Leukokeratosis affecting dorsum of the tongue along with surrounding depapillation



Fig. 6: Intraoral view. Photograph shows plaque, calculus, clinical loss of attachment and generalized gingival inflammation.



Fig. 4: Angular cheilitis. Fissures and cracks seen at the angle of the mouth bilaterally

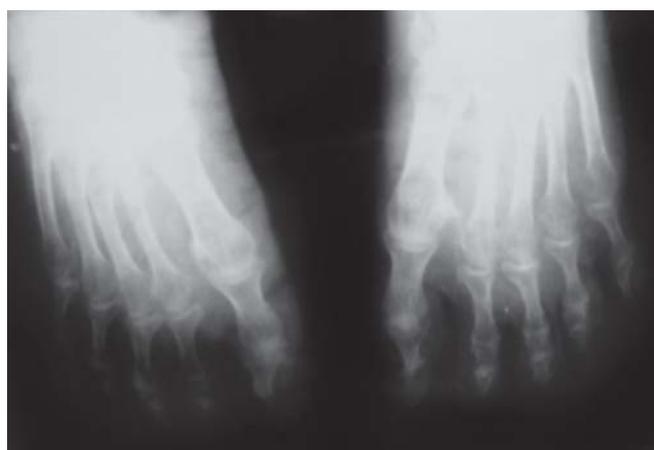


Fig. 7: Radiograph of feet. Radiograph showing acro-osteolytic bone changes



Fig. 5: Leukoplakia. Keratotic lesions on right and left retrocommissural area



An incisional biopsy taken from the retrocommissural area showed hyperkeratosis, parakeratosis, acanthosis and chronic inflammatory infiltrate suggestive of leukoplakia. A thorough scaling and root planing was carried out, and for burning sensation triamcinolone acetonide was prescribed and patient was advised to quit tobacco chewing habit.

DISCUSSION

Pachyonychia congenita is a rare genodermatosis, inherited as an autosomal dominant trait, characterized by variety of ectodermal abnormality that occurs in first month of life. Here we reported a case of PC type 1, with added emphasis on dental

findings. The familial nature of the disturbance was established in 1912, when Murray documented seven affected people in three generations of a family. In similar report Kumer and Loos in 1935 described 24 cases in a five-generation family. The phenotype was expanded, and the autosomal dominant mode of inheritance was documented in 1983 when Stieglitz and Centerwall published 17 affected individual in four generations.^{3,10}

We would also like to highlight the importance of taking family history with an emphasis on consanguineous marriage as these consanguineous marriages are being commonly practiced in this part of South India. Individual who are related through one or more common biological ancestors are called consanguineous relatives. Thus marriage among such relatives is called as consanguineous marriage. In our case report five individuals were affected in three generations.

In Greek pachy means thick and onyx means nail. Four clinical subtypes of PC have been described so far. PC-1, or Jadassohn–Lewandowsky type, named after the professor of dermatology at the University of Bern in Switzerland, Josef Jadassohn and his colleague Lelix Lewandowski, is associated with a heterozygous missense mutation in the helix initiation motif of keratin (K) 16 gene and keratin 6 isoform (K6a). PC-2, or Jackson-Lawler type, is associated with mutation in K17 and K6b.¹¹

PC-1 (56% of cases) is characterized by: (i) The distinctive and excessive thickening of all nails; (ii) palmar and plantar hyperkeratosis; (iii) follicular keratosis of the skin, especially on the knees and elbows; (iv) palmar and plantar hyperhidrosis; (v) leukokeratosis of the mucous membrane; and (vi) onset at infancy. PC-1 is also characterized by development of callus overplantar pressure points, blistering on walking and hair abnormalities. The upward angulation of distal end of the nail as a result of subungual hyperkeratosis is described as ‘door wedge’ distal hyperkeratosis. PC-2 (25% of cases) has additional features, such as multiple pilosebaceous cysts, neonatal teeth, pili-torti, and oral leukokeratosis occurs less frequent in PC-2. Presence of steatocytoma, multiple, uniform, yellowish cystic papules of 2 to 6 mm, located on anterior portion of the trunk, upper arms, axilla, and widespread pilosebaceous cysts following puberty is an important factor in PC-2.^{12,13}

PC-3 or Schafer-Branauer type (12% of cases), includes features of both PC-1 and PC-2 with additional features of angular cheilitis, corneal dyskeratosis and cataract. PC-4 (7% of cases) or pachyonychia congenita tarda has features of other three types with additional laryngeal involvement and mental retardation.^{8,14,15}

Leukokeratosis of oral mucosa is a predominant feature, mainly occurring on the tongue, buccal mucosa and sometimes on the gingiva. In some patients early tooth decay, periodontitis, enamel hypoplasia and talon cusps may be evident. The oral leukokeratosis is not a precancerous lesion and can be differentiated from leukoplakia or other dysplastic lesions by performing oral biopsy or recognizing its presence in patients with other expressions of PC.³

In the present case, on intraoral examination, grayish white, nonscrapable patches on right and left retrocommissural areas

were noted and it was attributed to the patient’s habit. Though the diagnosis of leukoplakia was made, a differential diagnosis of white sponge nevus, hyperplastic candidiasis, oral discoid lupus erythematosus and lichen planus were considered. A case of PC with oral leukoplakia has been reported in 1999.¹⁶ White sponge nevus occurs soon after the birth and is usually widely distributed over the oral mucous membrane. In contrast, leukoplakia is seen mostly in patients over 40 years of age and usually is not disseminated throughout the oral cavity, whereas multiple lesions which can be scrapable can favor the diagnosis of hyperplastic candidiasis. The oral discoid lesions share much in common with leukoplakia and lichen planus. The lesions of lichen planus are most difficult to differentiate from leukoplakia because both diseases usually affect patients over 40 years of age; however, leukoplakia more often affects men, whereas lichen planus occurs more frequently in women, and if the intraoral lesions take the forms of Wickham’s striae, the diagnosis is readily discernible.

In case report of 30-year-old female, on routine radiological examination multiple radiopacities were discovered in both jaws in premolar region with diagnosis of multiple localized idiopathic osteosclerosis. These radiopacities can be differentiated from, supernumerary teeth with aberrant morphology, complex composite odontomes and hyperostotic areas.³

At present there is no cure for PC. Treatment is aimed at providing symptomatic relief for the patient, such as soaking feet and hands in saline or in 50% propylene glycol solution followed by gentle debridement. Application of emollients or cream containing 10 to 20% salicylic acid is used to soften the nails prior to the paring down the excess along with curettage of matrix and nail bed. For skin lesions, sodium levothyroxine, gentian violet, ammonium lactate and salicylic acid can be applied. Certain drugs, such as oral retinoids (isotretinoin, etretinate), and keratolytic agents have also been used with no reports of long-term benefits. In case of oral lesions, extraction of natal and neonatal teeth and for periodontal conditions routine scaling and root planing along with curettage is mandatory.

Complications of PC are acro-osteolysis occurrence of destructive changes in the distal phalangeal bone and bone changes like, demineralization, thinning, premature closure of the epiphyses and increased curvature of long bones with osteopenia.

CONCLUSION

PC is hereditary disorder that runs in family. A dentist needs an exceptionally strong knowledge base in identifying the dental related problems there by to minimize adverse outcome secondary to provision of oral health. The management strategy should include both skin as well as oral lesion preventing the further complications of this disease.

REFERENCES

1. Stevens HP, Kelsell DP, Bryant SP, et al. Linkage of an American pedigree with palmoplantar keratoderma and malignancy (palmoplantar ectodermal dysplasia type III) to 17q24 literature survey and proposed updated classification of the keratodermas. *Arch Dermatol* 1996;132:640-51.

2. Feinstein A, Friedman J, Schewach-Millet M. Pachyonychia congenita. *J Am Acad Dermatol* 1988;19:190-93.
3. Pradeep AR, Nagaraja C. Pachyonychia congenita with unusual dental findings: A case report. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod* 2007;104:89-93.
4. Haber RM, Rose TH. Autosomal recessive pachyonychia congenita. *Arch Dermatol* 1986;122:919-23.
5. Anneroth G. Pachyonychia congenita. *Acta Dermatol Venereol* 1975;55:387-94.
6. Soderquist NA, Reed WB. Pachyonychia congenita with epidermal cysts and other congenital dyskeratosis. *Arch Dermatol* 1968;97:31-33.
7. Schonfeld PH. The pachyonychia congenita syndrome. *Acta Dermatol Venereol* 1980;60:45-49.
8. Paller AS, Moore JA, Scher R. Pachyonychia congenita tarda. A late-onset form of pachyonychia congenita. *Arch Dermatol* 1991;127:701-03.
9. Hannaford RS, Stpleton K. Pachyonychia congenita tarda. *Australas J Dermatol* 2000;41(3):175-77.
10. Murray FA. Congenital anomalies of nails. 4 cases of hereditary hypertrophy of nail bed associated with a history of erupted teeth at birth. *Br J Derm* 1921;33:409-12.
11. Drawber RPR, Barna R, de Berker D. Disorders of nails. In: Champion RH, Burton JL, Burns DA, Breathnach SM, editors. *Rook/Wilkinson/Ebling, Textbook of Dermatology* (6th ed). Oxford: Blackwell 1998;28-34.
12. Feng YG, Xiao SX, Ren XR, Wang WQ, Liu A, Pan M. Keratin 17 mutation in pachyonychia congenital type 2 with early onset sebaceous cysts. *Br J Dermatol* 2003;148:452-55.
13. Oh SW, Kim MY, Lee JS, Kim SC. Keratin 17 mutation in pachyonychia congenita type 2 patient with early onset steatocystoma multiplex and Hutchinson-like tooth deformity. *J Dermatol* Mar 2006;33(3):161-64.
14. Covello SP, Smith FJ, Smitt JH, Paller AS, Munro CS, Jonkman MF, et al. Keratin 17 mutation cause either steatocystoma multiplex or pachyonychia congenita type 2. *Br J Dermatol* 1998;139:475-80.
15. Clementi M, Cardini de Stafani E, De Rossi C. Pachyonychia congenital Jackson-Lawler type: A distinct keratodrome. *Br J Dermatol* 1986;114:367-70.
16. Lim TW, Paik JH, Kim NI. Case of pachyonychia congenita with oral leukoplakia and steatocystoma multiplex. *J Dermatol* 1999;26(10):677-81.