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We hope that making available the relevant information on Pachyonychia Congenita will be a means of furthering research to find effective therapies and a cure for PC.
Pachyonychia congenita with unusual dental findings: a case report

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Pachyonychia congenita is a rare genodermatosis, usually inherited as an autosomal dominant trait, characterized by a variety of ectodermal abnormalities. The most characteristic finding of affected patients is 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 development of friction blisters. The oral findings in pachyonychia include leukokeratosis of the palms and soles, and palate, angular stomatitis, and presence of natal or neonatal teeth. To our knowledge, this is the first report of pachyonychia congenita associated with unusual dental findings, such as presence of multiple localized idiopathic osteosclerosis, multiple retained primary roots, multiple talon cusps, and mesiodens.

Pachyonychia congenita (PC) is a rare form of hereditary palmoplantar keratodermia. It was first documented by Muller in 1904, but it was Jadassohn and Lewandowsky in 1906 who reported palmoplantar keratoderma and ectodermal defects. It is usually inherited as an autosomal dominant trait with varying degree of penetrance, although autosomal recessive forms have also been described.

The manifestations in PC are chiefly subungal hyperkeratosis with marked thickening of the distal portions of the nails and severe and disabling hyperkeratosis of the palms and soles. Other possible manifestations include follicular hyperkeratosis observed on the face (e.g., temples, eyebrows) and on the extensor aspect of the proximal parts of the extremities, hyperhidrosis particularly on palms and soles, cornal changes, and epidermal inclusion cysts.

The oral findings in pachyonychia include leukokeratosis of tongue or buccal mucosa, scalloped edges of the tongue, angular cheilitis, and dental abnormalities, including enamel hypoplasia, neonatal teeth, hypodontia, and periodontitis and severe caries. Pachyonychia congenita usually begins in infancy, but late-onset PC, referred to as pachyonychia tarda, which begins in the fourth or fifth decade, has been reported.

CASE REPORT

A 30-year-old female patient was referred from the Department of Skin and Venereology, Victoria Hospital, Bangalore, India, to the Department of Periodontics, Government Dental College and Hospital, Bangalore, India, for treatment of poor periodontal condition, with bleeding gums and halitosis. The medical history of the patient revealed that she was diagnosed with PC within 2 months of birth. Family history revealed that the maternal grandfather also suffered from the condition.

General physical examination showed hyperkeratotic fingernails and toenails (Figs. 1 and 2). Fingernails were affected more than the toenails. The affected nails showed thickening and hardening, subungal hyperkeratosis, and upward growth of the distal nail with hypercurvature. Patient gave a history of blisters on her feet after prolonged walking, especially during summers, blepharitis, and photosensitivity and exhibited classic symptoms of PC, which included hyperhidrosis of palms and soles (Fig. 3) and hoarseness of voice.

In the preceding month, patient had developed few clusters of papules on her forearm and back. Based on the appearance of pearly white papules with central indentation these were diagnosed as molluscum contagiosum (Fig. 4), and the diagnosis was confirmed microscopically by the presence of molluscum bodies seen on hematoxylin and eosin staining.

Intraoral examination showed a striking feature of leukokeratosis of the right dorsal and lateral aspect of the tongue (Fig. 5), right buccal mucosa, and angular cheilitis on the right side, and all the 6 maxillary anterior teeth showed talon cusps (Fig. 6). A mandibular left retained deciduous canine and buccally erupted left permanent canine was also seen. On periodontal examination, prominent local deposits and generalized severely inflamed, erythematous, and enlarged gingiva with bleeding on probing and an average probing depth of 5 mm were seen. However, because no clinical attachment loss was observed, the pockets were considered as pseudo-pockets (Fig. 7). Periodontal indices were recorded: Plaque index9 score was 2.6, and modified gingival index10 score was 2.3.

Routine blood chemistry and biochemical investigations, which included random blood glucose, serum phosphorous, urea, and creatinine were carried out. All values were within normal limits except for a raised erythrocyte sedimentation...
rate of 87 mm/h (normal 15 mm/h in women, 6.5 mm/h in men) which was possibly due to lower respiratory tract infection, for which she was under medication. To exclude onychomycosis, nail cultures were examined for the presence of fungi and proved to be negative.

On routine radiologic examination, an interesting finding was observed: elongated radiopacities were seen in the posterior regions in all quadrants of maxilla and mandible. The radiopacities were surrounded by a thin radiolucent rim, which in turn was surrounded by a sclerotic lamina dura like trabeculation, suggesting retained deciduous roots (Fig. 8) in the mandible, but in the maxillary arch the radiopacity was densely homogeneous and did not show a perilesional radiolucent rim, thus giving an impression of idiopathic osteosclerosis (Fig. 9).

An orthopantomograph revealed an unerupted mesiodens. A thorough scaling and root planing was carried out, and the patient was put on maintenance therapy.

DISCUSSION

Pachyonychia congenita is predominantly an autosomal dominant group of ectodermal dysplasias characterized by hypertrophic nails and other ectodermal changes that occur with the first months of life.

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 Felix 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 mutations in K17 and K6b.11

PC-1 (56% of cases) is associated with oral leukokeratosis, palmoplantar keratoderma and follicular keratosis. PC-2 (25% of cases) has additional features such as multiple pilosebaceous cysts, neonatal teeth, and pilitorti, and oral leukokeratosis occurs less frequently.
Presence of widespread pilosebaceous cysts following puberty is an important distinguishing factor in PC-2.

PC-3, or Shafer-Brunauer type (12% of cases), includes features of both PC-1 and PC-2 with additional features of angular cheilitis, corneal dyskeratosis, and cataracts. PC-4 (7% of cases) has features of the other 3 types with additional laryngeal involvement and mental retardation. The defective gene responsible for PC is located on chromosome 17q and associated with mutations in K16 and K17.12

The familial nature of the disturbance was established in 1921, when Murray13 documented 7 affected persons in 3 generations of a family. In a similar report, Kumer and Loos in 193514 described 24 cases in a 5-generation family. The phenotype was expanded, and the autosomal dominant mode of inheritance was documented in 1983 when Stieglitz and Centerwall15 published details of kindred with 17 affected individuals in 4 generations.

Leukokeratosis of oral mucosa is a predominant feature, mainly occurring on the tongue, buccal mucosa, and sometimes the gingiva. In some patients early tooth decay, periodontitis, and enamel hypoplasia 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.

Presently 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. Nails can be managed by application of emollients or cream containing 10%-20% salicylic acid to soften the nails prior to paring down the excess. Certain drugs have also been used with no reports of long-term benefits, such as dilantin, fluorouracil, oral retinoids such as isotretinoin, etretinate, and keratolytic agents. In the present case, on routine radiologic examination multiple radiopacities were discovered in both jaws in the premolar regions. Though a diagnosis of multiple localized idiopathic osteosclerosis in the maxilla and mandible was made, a differential diagnosis of retained roots of deciduous predecessors in the mandible, supernumerary teeth with aberrant morphology, complex composite odontomes, or hyperostotic areas was also considered.

Initially the radiopacities were thought to be aberrant supernumerary teeth, but the radiopacities did not show any coronal component, which had a radiodensity of enamel, and their shape did not suggest that a crown was present. Also, there was no evidence of a follicle, follicular cyst, or dentigerous cyst associated with the superior aspect of the radiopacities.

Odontomes were also ruled out for the same reason,
because they did not exhibit radiodensity of enamel, and their shape was also not similar to a tooth as in the case of compound composite odontomes. Although odontomes, which bear no structural resemblance to a tooth, are classified as complex composite odontomes, such odontomes appear more globular in nature, with radiodensity similar to tooth structure and frequently associated with an unerupted tooth.

Hyperostotic areas were excluded from the diagnosis because observation of radiographs under higher magnification showed that the radiodensity was homogeneously devoid of trabeculation.

The presence of focal radiopaque area associated with the existing teeth, and the absence of fragments of lamina dura surrounding the radiopacity led to the diagnosis of localized idiopathic osteosclerosis.

Idiopathic osteosclerosis is a focal deposition of bone which occurs even in the absence of trauma or any systemic conditions. It is more common in the mandible than in the maxilla. In the present case focal type of idiopathic osteosclerosis was seen both in the maxilla and in the mandible. Generally, once the osteosclerosis develops, it does not show any regression and no treatment is required.

Although an interesting finding in this patient was the presence of radiopaque areas radiographically, this, in all likelihood, is not related to PC but rather is a coincidental finding.

Another interesting feature in the present case is the presence of multiple talon cusps. The prevalence of talon cusps is usually 1% to 8%. Its occurrence is most common in the lateral incisors (55%) than central incisors (33%) and least on canines (6%). In the present case, talon cusps were seen on all 6 maxillary anterior teeth, thereby being a very rare and interesting finding.

Thus this is a case report of a rare hereditary genodermatosis with very unusual dental findings which had not been previously reported.

REFERENCES

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