PACHYONYCHIA CONGENITA- A RARE GENODERMATOSIS

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ABSTRACT

Background: Pachyonychia Congenita is a rare autosomal dominant disorder, typically affecting nails, skin, oral mucosa, larynx, hair and teeth, but some forms of autosomal recessive cases are also reported, characterized by hypertrophic nail changes and nail dystrophy which is usually presented at birth or develop within 1 year after birth.

Case Report: A 16 years old female patient reported to our dental OPD with the chief complaint of bleeding on brushing, discoloration of teeth and multiple missing teeth. On examination palmar-plantar keratosis, thickened nails with transverse curvature, hoarseness of voice were present.

Discussion: Our patient presented with some distinct clinic-pathological pictures with the presence of retained deciduous teeth, numerous hypoplastic teeth, with incompletely formed roots giving the features of mutilated dentition.

Conclusion: The dentists often get consulted for the oral leukokeratosis usually present in Pachyonychia Congenita. Awareness of its benign nature can alleviate the apprehension in patient. Advancements in delivery systems, availability of RNA reduction agents, gene slicing approaches may improve, make the ultimate goal of target gene correction of PC more feasible in future.

KEYWORDS: Keratosis, Mutilated Dentition, Leukokeratosis.

INTRODUCTION - Pachyonychia Congenita (PC) is an uncommon autosomal dominant keratin disorder that typically affects a number of ectodermal structures including the nails and palmo-plantar skin, and often involves the oral mucosa, tongue, larynx, teeth and hair¹²³. Clinical features are usually present at birth or early infancy. It is estimated that there are 1000–10,000 cases of PC worldwide⁴. It was first described by German dermatologists Josef Jadassohn and Felix Lewandowsky in 1906, which identified a 15-year-old girl with unusual keratinization of the skin and the tongue, and extremely thickened nails of the fingers and toes⁵.

It is a group of rare, inherited ectodermal dysplasias associated with mutations in keratin genes of K6a, K6b, K16 or K17. The most prominent clinical features of PC are nail dystrophy and dyskeratosis of skin and mucous membranes. 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.

PC was historically classified into at least two subtypes according to the clinical features: PC type 1 (MIM #167200, PC-1, or Jadassohn-Lewandowsky syndrome), the more common variant, is characterized mainly by nail changes, palmoplantar keratoderma (PPK), follicular keratosis and oral leukokeratosis, while PC type 2 (MIM #167210, PC-2, or Jackson-Lawler syndrome) includes the features of PC-1 plus natal teeth, epidermal inclusion cysts, pilosebaceous cysts such as steatocystomas and vellus hair cysts, and hair abnormalities, including alopecia, pili torti (twisted hair) and unruly hair. A third variant, PC tarda, has also been described and is characterized by a later onset that ranges from late childhood to middle age. Pathogenic mutations in the genes encoding keratin 6A (KRT6A) or its expression partner keratin 16 (KRT16) are associated with the PC-1 phenotype, whereas mutations in PC-2 occur in KRT6B or its expression partner KRT17. These keratins are constitutively expressed in keratinocytes of the nail, palmoplantar skin, mucosa, and hair, leading to the manifestations of the disorder at these sites. However, as a considerable overlap between PC subtypes exists and distinct phenotypic differences can occur in the same variant, a new nomenclature based on genotyping has been proposed, as follows: PC-6a, PC-6b, PC-16 and PC-17.

CASE REPORT
A 16 years old female patient reported to Dr. Ziauddin Ahmad Dental College, AMU, Aligarh with chief complaint of bleeding on brushing, abnormal dentition (Fig.1), numerous missing teeth, difficulty in chewing and hypersensitivity to cold. There was no significant medical history and as well as no history of consanguineous marriage of the parents was reported. She had no harmful personal habits and menstrual history was normal. She had never visited to any dentist. There was no history of natal or neonatal teeth and early loss of deciduous dentition. She had difficulty in speech articulation and hoarseness of voice. On general examination nails were thickened with transverse curvature (Fig.2) and palmo-plantar keratosis (Fig.3) was present. On intra-oral examination multiple abnormal teeth with hypoplastic enamel, missing mandibular canine, retained deciduous teeth and some of the teeth had sharp enamel projections on occlusal surfaces. There were no oral blisters or white patches inside the oral cavity (Fig.4). On radiographic examination it was revealed that she had multiple missing teeth and retained deciduous mandibular anteriors and impacted mandibular right canine and some of the teeth were hypoplastic with undeveloped roots (Fig.5). Routine blood and urine examinations were within the normal limits.

Based on the characteristic clinical presentation and history, a diagnosis of pachyonychia congenita 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, Jawahar Lal Nehru Medical College where she was put on oral vitamin A,
keratolytic agents and emollients. This case of PC had some distinct picture with presence of retained deciduous teeth, numerous hypoplastic teeth with incompletely formed roots giving the features of mutilated dentition.

DISCUSSION

We discuss a rare case of PC with distinct oral and dental findings. The most common clinical feature in PC is hypertrophic nail dystrophy in association with painful palmoplantar keratoderma, which progresses with age. Nail changes include transverse overcurvature, distal onycholysis, subungual hyperkeratosis, and variable discolouration. Other common findings are oral leukokeratosis (67%), follicular keratosis (53%) and cysts (64%). PC-1 and PC-2 can be distinguished clinically as oral leukokeratosis occurs in PC-1, and steatocystomas/pilosebaceous cysts, vellus hair cysts, hair abnormalities (alopecia, pili torti) and natal/neonatal teeth occur in PC-2, usually. Also, hoarseness due to laryngeal involvement is generally associated with PC-1.

The features of PC-1 are found in a variety of autosomal dominant keratodermas that have oral lesions, such as focal palmoplantar and gingival keratosis, focal non-epidermolytic palmoplantar keratoderma with oral-genital-follicular lesions, and focal palmoplantar keratoderma associated with esophageal cancer. These all involve focal and pressure-related palmoplantar hyperkeratosis and oral hyperkeratosis. In focal palmoplantar and gingival keratosis, lesions appear in the attached gingiva. Palmoplantar keratodermas exhibit oral lesions in the buccal mucosa, palate and occasionally the gingiva. Hyperkeratotic lesions are more commonly seen in oral regions and the tongue is characteristically affected with buccal mucosa in PC.

With the exception of the previously described keratodermas, there are a number of differential diagnoses for PC. Hyperkeratotic nail thickening is seen in onychomycosis, similar to PC, but fungal infection does not affect all nails in early infancy. Clouston syndrome, a rare autosomal dominant genetic disorder, is characterized by the major triad of features: nail dystrophy, generalized hypotrichosis, and palmoplantar hyperkeratosis. This is similar to PC; however, alopecia is a common feature of Clouston syndrome, which is not typically seen in PC. Dyskeratosis Congenita is a rare inherited fatal disorder that mimics the skin, nail and oral features of PC. Unlike PC, progressive bone marrow failure is one of the most well-known symptoms of Dyskeratosis Congenita and the primary cause of mortality.

There is no effective treatment for PC. Several clinical studies and clinical trials are underway, sponsored by the patient advocacy group Pachyonychia Congenita Project (PC Project). Currently, treatment of PC is primarily symptomatic. Emollients (moisturizers) and keratolytics (products containing alpha-hydroxy acids) provide little improvement for the hyperkeratosis and mechanical removal of the callus several times a week is usually necessary. Routine grinding of the nail plates can minimize their interference with function. Oral retinoids such as Accutane have no positive effect for those with PC. The retinoids Soriatane, Tigason or Neo-Tigason have minimal effect, but may allow some slight relief especially of palmar...
keratoderma when used in low dosage or variable dosage. Retinoids are used cautiously due to their known bone toxicity and other complications.

**CONCLUSION**

Pachyonychia congenital (PC) is a rare genetic disorder of autosomal dominant trait. The dentists often get consulted for the oral leukokeratosis usually present in PC. Awareness of its benign nature can alleviate the apprehension in patient. 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.

![Figure 1: Abnormal Dentition](image1)

![Figure 2: Thickened Nails with Transverse Curvature](image2)

![Figure 3: Palmo-Plantar Keratosis](image3)

![Figure 4: No Oral Blisters or White Patches](image4)

![Figure 5: Hypolplastic Teeth with Undeveloped Roots](image5)
REFERENCES


