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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

A long-term evaluation of associated oral and dermal lesions

Louis L. Young, D.D.S., M.S.D., and John A. Lanct, D.M.D.,
Lexington, Ky.

DEPARTMENT OF PERIODONTICS, UNIVERSITY OF KENTUCKY COLLEGE OF DENTISTRY

This article reports a long-term follow-up of a patient with pachyonychia congenita. The dental and oral manifestations of this disease are described. The current oral changes include multiple areas of hyperkeratosis in areas susceptible to trauma. A rationale of obtaining dental treatment in relation to the current oral disease is proposed to avoid further trauma to the oral mucosa.

Pachyonychia congenita, by definition, refers to a congenital hypertrophy of the nail bed and is an uncommon congenital dysplasia which has rarely been reported in the literature. Shortly after birth the nail bed becomes yellowish brown, discolored, and elevated. This elevation of the nail upward instead of outward progresses in severity until finger or toe movement is restricted. Plastic surgical correction and removal of nails are possible forms of treatment, but in either case the nails are ultimately lost. Skin lesions generally appear after birth and are seen as hyperhidrosis, follicular keratosis, and hyperkeratosis of the palms and soles. Bullae may also occur in the palmar plantar pressure areas, making movement painful and difficult. Pachyonychia congenita appears to be inherited genetically as a simple mendelian dominant characteristic with incomplete penetrance. Jafarsohn and Lewandowski, in 1966, described the syndrome which included oral leukokeratosis, thickened nails, hyperhidrosis, and palmar plantar and follicular keratosis. Gorlin and Chaudhury summarized the available literature in 1958 and concluded that the oral findings may include white, opaque thickening of the dorsum of the tongue and along the interdental line. They concluded that these thickenings are due to hyperkeratosis or parakeratosis of the epithelium rather than to
true dyskeratotic changes. Mikulicz keratotic changes may occur in the larynx, nose, tympanic membrane, and cornea. Jackson and Poole have described the occurrence of neonatal teeth in this syndrome.

**CASE REPORT**

This report provides a 2-year follow-up of a case of Polyosis epidermodica congenita in a female patient who was first reported by Tunes, Goldman and Chassin in 1968. When seen and documented in 1968, the patient was 20 years old and had polyoid lesions, palmar and plantar keratoses, generalized alopecia, and some white opaque thickening along the lateral borders of the tongue. The nails had been deformed since birth, and some had been surgically removed. The skin was dry and scaly since birth. No oral lesions were observed other than those already mentioned on the lateral border of the tongue. The patient's chief complaint was painless blisters on the feet. There were apparently due to friction from shoes, and therefore the patient was advised to wear soft cotton slippers.

The patient, who is now 26 years old, was seen for evaluation of oral "lactophlebitis" and periodontal disease. She related a prior history of multiple oral lesions of the tongue and buccal mucosa along with extensive periodontal therapy. The patient was aware of the presence of these lesions at an early age and had undergone multiple debridements and explantations. She had also undergone multiple dental procedures, including the extraction of teeth with lesions of the oral mucosa. The patient was referred to a dermatologist for further evaluation and management of her condition.
of Pachyonychia congenita but was concerned over the recent biopsy reports of "leukoplakia" which had led to a subsequent esophagitis.

The current oral examination revealed opaque "white patches" on the buccal mucous at the incisal line area (Fig. 1) and on the lateral borders of the tongue (Fig. 2), as well as diffuse white areas on most of the attached gingiva (Fig. 3). The gums of the tongue was unremarkable.

Periodontal disease was evident, with generalized pocket depth up to 8 mm. Mandibular bilateral gingival expansion were also present, and for several years the patient had been frequently wearing a lower removable partial "overlay" denture with cosmetic acrylic veneer gingivae on the upper arch.

The nails were almost completely absent on both fingers and toes (Fig. 4); they had been either removed surgically or shed spontaneously.

Histopathologic tissue sections from recent biopsies of oral mucosa revealed no dyskeratotic changes but, rather, a generalized intercellular vacuolization of the epithelium. Anatomic and paraanatomic sections of epithelium were also evident. It was decided that, because of the adequacy of previous pathologic slides, subsequent diagnosis, and description of the clinical features, a new biopsy was contraindicated.

The patient was reassured of the nonmalignant nature of the oral lesions. The proposed treatment involved conservative maintenance therapy of the periodontal pockets with overbridges. Selected extractions and reconstruction of edentulous areas with fixed bridgework was also advised.

Because of the locations of the internal oral lesions on pressure-bearing areas or areas subject to trauma, it was believed that it would be beneficial to avoid these areas with any removable dental appliances.

**DISCUSSION**

This case is of interest because of its prior documentation and subsequent long-term follow-up.

It appears that the dental reaction has become stable along with the loss of the nails. The skin, although thickened clinically, does not cause problems. The following keratin is apparently not evident at this time, although when the patient was younger this was a significant clinical problem. The formation of bullae on the feet has been avoided in previous years by the patient's awareness that trauma is a predisposing factor.

The oral manifestations, however, have become more overt and are evident.
in areas in the mouth subject to the most trauma. In the past 30 years progressive white lesions, which probably represent hyperkeratotic areas, have developed along the lateral borders of the tongue, attached gingiva, and interdental line areas, which are the areas most likely to be traumatized during mastication.

The proposed treatment is purposely conservative in nature to prevent surgical trauma and avoid removable prostheses which may add further insult to the oral mucosa.

REFERENCES

Reprint requests to:
Dr. Louis L. Young
Department of Periodontics
College of Dentistry
University of Kentucky
Lexington, Ky. 40506