Pachyonychia Congenita Project

15 March 2005

Use of Articles in the Pachyonychia Congenita Bibliography

The articles in the PC Bibliography may be restricted by copyright laws. These have been made available to you by PC Project for the exclusive use in teaching, scholarship or research regarding Pachyonychia Congenita.

To the best of our understanding, in supplying this material to you we have followed the guidelines of Sec 107 regarding fair use of copyright materials. That section reads as follows:

> Sec. 107. - Limitations on exclusive rights: Fair use
> Notwithstanding the provisions of sections 106 and 106A, the fair use of a copyrighted work, including such use by reproduction in copies or phonorecords or by any other means specified by that section, for purposes such as criticism, comment, news reporting, **teaching (including multiple copies for classroom use), scholarship, or research**, is not an infringement of copyright. In determining whether the use made of a work in any particular case is a fair use the factors to be considered shall include - (1) the purpose and character of the use, including whether such use is of a commercial nature or is for nonprofit educational purposes; (2) the nature of the copyrighted work; (3) the amount and substantiality of the portion used in relation to the copyrighted work as a whole; and (4) the effect of the use upon the potential market for or value of the copyrighted work. The fact that a work is unpublished shall not itself bar a finding of fair use if such finding is made upon consideration of all the above factors.

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.

2386 East Heritage Way, Suite B, Salt Lake City, Utah 84109 USA
Phone +1-877-628-7300 • Email—Info@PC-Project.org
www.PC-Project.org

oral medicine

Editor:
ALVIN L. MORRIS, D.D.S., Ph.D.
Vice-President for Administration
University of Kentucky
Lexington, Kentucky 40506

Pachyonychia congenita

A long-term evaluation of associated oral and dermal lesions

Louis L. Young, D.D.S., M.S.D., and John A. Lenox, 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 dermal 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 continuing dental treatment in relation to the current oral disease is proposed to avoid undue 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.[1-7] 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 palmoplantar pressure areas, making movement painful and difficult. Pachyonychia congenita appears to be inherited genetically as a simple mendelian dominant characteristic with incomplete penetrance. Jadassohn and Lewandowski,[1] in 1906, described the syndrome which included oral leukokeratosis, thickened nails, hyperhidrosis, and palmoplantar and follicular keratosis.

Gorlin and Chaudhry[2] 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

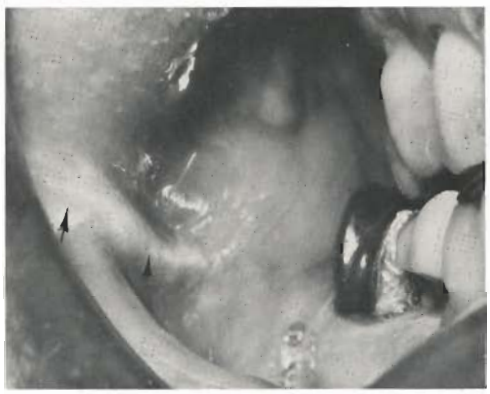

Fig. 1. Hyperkeratotic area along the interdental line of the buccal mucosa is indicated by the arrows.

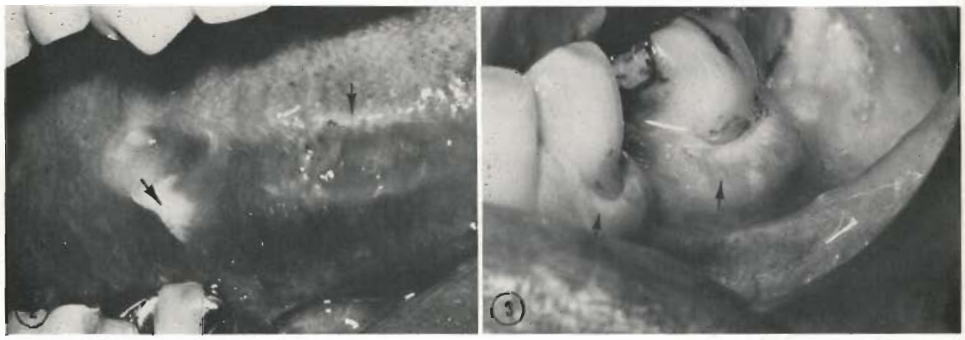

Fig. 2. Hyperkeratotic areas on the lateral border of the tongue appear to follow above and below the scalloped area, which corresponds to the lingual aspects of the teeth.

Fig. 3. Hyperkeratotic areas of the attached gingiva appear as a diffuse stria and are confined to attached gingiva (above the mucogingival junction). It is noted that the patient had been vigorously brushing this tissue with a hard brush for the past several years.

true dyskeratotic changes. Mucosal keratotic changes may occur in the larynx, nose, tympanic membrane, and cornea.[2] Jackson and Fowler[3] have described the occurrence of neonatal teeth in this syndrome.

CASE REPORT

This report provides a 36-year follow-up of a case of pachyonychia congenita in a female patient which was first reported by Tauber, Goldman, and Classen in 1936.[4] When seen and documented in 1936, the patient was 20 years old and had pachyonychia, palmar and plantar keratosis, generalized ichthyosis, 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 described as being 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 plainful bullae on the feet. These were apparently due to friction from shoes, and therefore the patient was advised to wear soft cotton slippers.

The patient, who is now 56 years old, was seen for evaluation of oral "leukoplakia" and periodontal disease. She related a prior history of multiple oral biopsies of the tongue and buccal mucosa along with extensive periodontal therapy. The patient was aware of the presence

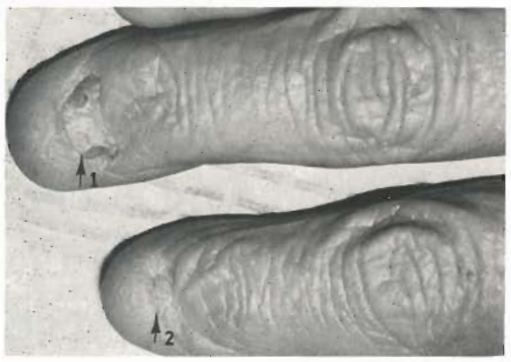

Fig. 4. The nails appear as keratotic areas with a vestigial bed *(1)* or are completely absent *(2)*. The skin has a thickened appearance and a firm texture.

of pachyonychia congenita but was concerned over the recent biopsy reports of "leukoplakia" which had led to a subsequent cancerphobia.

The current oral examination revealed opaque "white lesions" on the buccal mucosa at the interdental line area (Fig. 1) and on the lateral borders of the tongue (Fig. 2), as well as diffuse white striae on most of the attached gingiva (Fig. 3). The dorsum of the tongue was unremarkable.

Periodontal disease was evident, with generalized pocket depth up to 8 mm. Mandibular bilateral tori were also present, and for several years the patient had been infrequently wearing a lower removable partial "overlay" denture and cosmetic acrylic veneer gingivae in 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 intracellular vacuolization of the epithelium. Acanthosis and parakeratosis 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 curettage. Selected extractions and reconstruction of edentulous areas with fixed bridgework was also advised.

Because of the locations of the intraoral white 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 dermal reaction has become stable along with the loss of the nails. The skin, although thickened clinically, does not cause problems. The follicular keratosis 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 36 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

1. Jadassohn, J., and Lewandowski, K.: *In* Neiser, A., and Jacobi, E.: Keratosis linguae, idonographia dermatologia, Berlin, 1906, Urban Schwarzenberg, p. 29.
2. Gorlin, R. J., and Chaudhry, A. P.: Oral Lesions Accompanying Pachyonychia Congenita, ORAL SURG. 11: 541-543, 1958.
3. Jackson, A. D. M., and Fowler, S. D.: Pachyonychia Congenita: A Report of Six Cases in One Family, With a Note on Linkage Data, Ann. Eugenics 16: 142-146, 1951-52.
4. Tauber, E. B., Goldman, L., and Classen, H.: Pachyonychia Congenita; Case Report With Discussion, J.A.M.A. 107: 29-30, 1939.
5. Andrews, G. C., and Strumwasser, S.: Pachyonychia Congenita, N. Y. State J. Med. 29: 747-749, 1929.
6. Sohrweide, A. W.: Pachyonychia Congenita; Report of a Case, Arch. Dermatol. Syph. 33: 183-184, 1936.
7. Wright, C. S., and Gueguierre, J. P.: Pachyonychia Congenita, Arch. Dermatol. Syph. 55: 819-827, 1947.

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