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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

Manifestations for the Otalaryngologist

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When discovered, leukoplaikia of the oral cavity is commonly an acquired lesion. Pachyonychia congenita is a member of a rare group of disorders in which congenital white lesions of the oral cavity are present. Additional findings of note to the otalaryngologist are described as well.


White lesions of the oral cavity are, for the most part, acquired by products of the multitude of events occurring daily in the oral cavity. Less commonly, they may appear as manifestations of either local or systemic diseases such as lupus erythematosus and others. Rarely encountered is a group of congenital white lesions of the oral cavity herein referred to as oral genodermatoses. One of these disorders, pachyonychia congenita (PC), is presented along with some of its potential otalaryngologic head and neck manifestations.

Pachyonychia congenita was first described by Jadausn and Lewandowsky in 1906 as an anomaly in which dystrophic fingernails and toenails occurred together, acquired by specific cutaneous and mucous membrane changes that may first be noted at birth or in early childhood.

The remarkable fingernails (Fig 1) in these patients are the sine qua non of this disorder. Steiglitz and Centerwall have found that approximately 90% of cases reviewed have dystrophic nails with all fingernails and toenails being affected. Together with marked thickness, these nails take on a yellow-brown discoloration with pinched margins and an upward angulation of the distal tips. These hypertrophic nails can lead to difficulties in digital dexterity, and emotional remains a lingering concern.

Cutaneous findings sometimes resemble plantar and palmar hyperkeratosis in conjunction with hyperhidrosis. Follicular keratosis of the extensor surfaces of the knees and elbows may also be present. They are usually of the sessile type. These keratotic lesions may at times appear on the buttocks and lumbar regions as well. Plantar bullae secondary to hyperkeratosis and hyperhidrosis may become infected and lead to additional difficulties.

Gorlin and Chaudhry described the oral lesion as white, opaque thickenings of discrete or confluent areas of the oral cavity. The entire buccal mucosa may at times be involved. An associated overgrowth of Candida albicans has been noted with some of these leukoplakic lesions. Angular cheilitis has been described as well. The tongue may often appear with scalloped edges (Fig 2), and may be partially or totally covered by leukoplakic blanket. Aphthous ulcerations of the oral cavity have been noted to occur with PC, and neonatal teeth have been described in a number of affected individuals.

A number of other affected sites occur with less frequency including the larynx, eyes, teeth, hair, and nails. The likelihood of one of these comonly affected sites being involved will depend, for the most part, on the pedigrees under study. A recent presentation in one pedigree may support a common finding in another. Gorlin and Chaudhry noted the existence of white plaque-like lesions involving the interarytenoid area of the larynx.

There are a number of significant questions that may arise. Firstly, is the affected individual one of the few who have all three affected ectodermal, mesodermal, and endodermal anomalies? The likelihood of one of these occur in the future of the affected individual.

A number of the oral manifestations described by Wickup and Gorlin were noted by Wickup and Gorlin and others. Wickup and Wickup found similar findings to those described by Wickup and Gorlin.


Pachyonychia Congenita—Hersh

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of thickened tympanic membranes, with a resultant conductive hearing loss.

Pachyonychia congenita has been divided into various types based on clinical presentation. Kummer and Leon proposed the following division: type 1, symmetrical keratosis of the hands and feet along with follicular keratosis of the body; type 2, oral bulla; and type 3, oral bulla plus keratosis of the hands and feet. Additional subdivisions have been proposed in the literature but may, in fact, be artificial in nature as the subdivisions may represent possible variations in phenotypic expression of a single disease entity.

REPORT OF A CASE

This 45-year-old man was seen in consultation for a totally unrelated problem. On routine otorhinolaryngologic examination, a rather remarkable-looking tongue was noted (Fig 2), which obviously led to further questioning. His fingernails were remarkably well developed as well (Fig 1). He was aware that he had some form of genetic illness as his son had similar findings. To the best of his knowledge, no other family members were affected. Permission to obtain a biopsy specimen from the tongue to obtain additional investigations was refused.

Pathologic Findings

There do not appear to be any microscopic features that are strictly pathognomonic of PC. A number of the genodermatoses may have histologic features that are unidentifiable from those found in PC. Bright and Cesterow have found that all three germ layers may be involved: the epidermis, nails, skin, hair, teeth, and oral mucosae; the endoderm, larynx, and the mesoderm, joints, and tracheal cartilage. It was then concluded that the histologic picture of the disparate sites was essentially similar.

Withen and Gurlin performed exfoliative cytotologic analyses of the oral mucosa and tongue. They noted that the epithelium was orthostratified with uniform intraepithelial exocytosis. Interestingly, the intercellular bridges were not present in the prickle cell layer. A marked parakeratosis was also evident, while the stratum granulosum was lacking. Gurlin and Chaufour observed representative biopsies of the tongue and oral mucosa with findings similar to those obtained by Withen and Gurlin. Caux et al found the ultrastruc-

Fig 1.—Hyperplastic gingiva with characteristic rounded elevation of distal tip.

Fig 2.—Marked keratolytic scaling of the tongue along with scalloped perimeter.

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In Other AMA Journals

AJDC
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