Pachyonychia Congenita Project

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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.

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ORAL LESIONS ACCOMPANYING PACHYONYCHIA CONGENITA

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PACHYONYCHIA congenita is an unusual congenital and sometimes familial malady that is characterized by dystrophic changes in the fingernails and toenails, hyperkeratoses or callosities of the palms and soles, follicular keratosis of the acneiform type particularly about the knees and elbows, and hyperhidrosis or excess sweating of the hands and feet. Dystrophic changes are also occasionally observed in the hair or cornea. Verrucose lesions may appear on the elbows, but they are especially common on the lower extremities about the knees, popliteal fossae, lower legs, and ankles. Plantar bullae are not uncommon.

Oral lesions are nearly always present. They take the form of white opaque thickenings. These thickenings may be focal, involving only a portion of the buccal mucosa or tongue, or they may be quite generalized, covering the entire mucosa of the tongue, lip, and cheek. Angular cheilosis is commonly present.

The disease was recognized as an entity and originally described by Jadassohn and Lewandowsky in 1906.[1] No more than a dozen cases have been cited in the domestic literature since the original report.[2-10] Analysis of these cases revealed that males are more often affected (seven of eleven cases) and that the nails appear abnormal at birth or very soon after birth. It is not uncommon for the skin lesions to appear when the child is about 18 months to 2 years old, although skin abnormalities have been observed at birth. White patches on the oral mucosa have been noted at birth in most patients, although several case reports reviewed did not specify the age at which leukokeratosis was initially observed. Although some investigators have described these thickenings as "leukoplakia," it is likely that this term was used in the literal rather than the microscopic sense, as their photomicrographs indicate hyperkeratosis or pachyderma oralis rather than dyskeratosis.

Case Report

The present case concerns a 17-year-old, somewhat thin, white girl who was first seen at the University of Minnesota Hospital when she was less than 2 years of age. At that time, on the basis of her presenting signs of deformed darkened fingernails and toenails and a rash limited to her lower legs, her condition was diagnosed as pachyonychia congenita. Her nails had been abnormal from birth (Fig. 1). At the age of 2 months a rash appeared about the lower legs; this rash cleared up upon treatment. When the patient was about

From the Division of Oral Pathology, University of Minnesota, School of Dentistry.

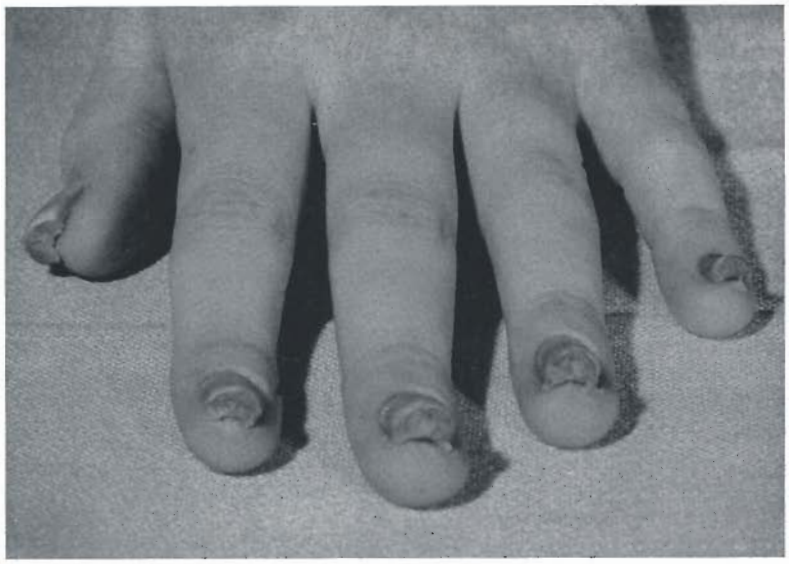

Fig. 1.—Note thickened, darkly colored nails. This change was evident from birth.

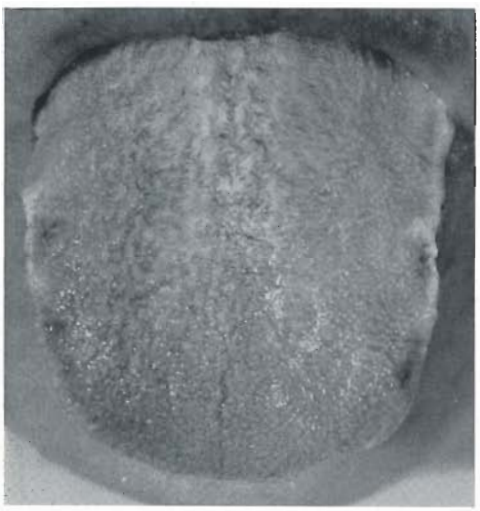

Fig. 2.—The patient's tongue was covered with a heavy white coat that did not rub off. This white thickening was especially apparent about the lateral margins.

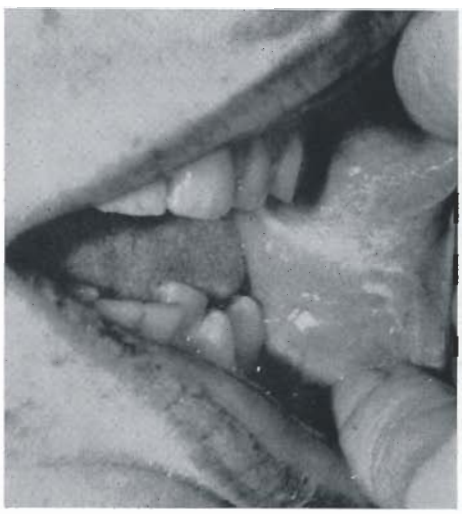

Fig. 3.—The buccal mucosa was involved bilaterally with an identical whitish thickening. It was most prominent about the third molar area.

18 months old, a scaly papular rash appeared on the knees, elbows, and buttocks. The rash consisted of tiny follicular hyperkeratotic papules. Subsequently she developed "sores on her feet" which took the form of deep, long-lasting bullae. Callosities gradually developed on the palms and soles, and especially between the toes. These broke down and became secondarily infected. Walking became increasingly difficult. The condition has not improved during the intervening years. It seemed to become worse with increased perspiration, especially during the summer, although hyperhidrosis has been constantly present. Family history was entirely negative. Oral examination revealed white thickenings bilaterally on the buccal mucosa, especially along the interdental line and a diffuse white covering of the tongue, especially along the lateral margins (Figs. 2 and 3). Laryngeal examination revealed two small whitish plaques in the interarytenoid area, which were responsible for a mild husky voice change.

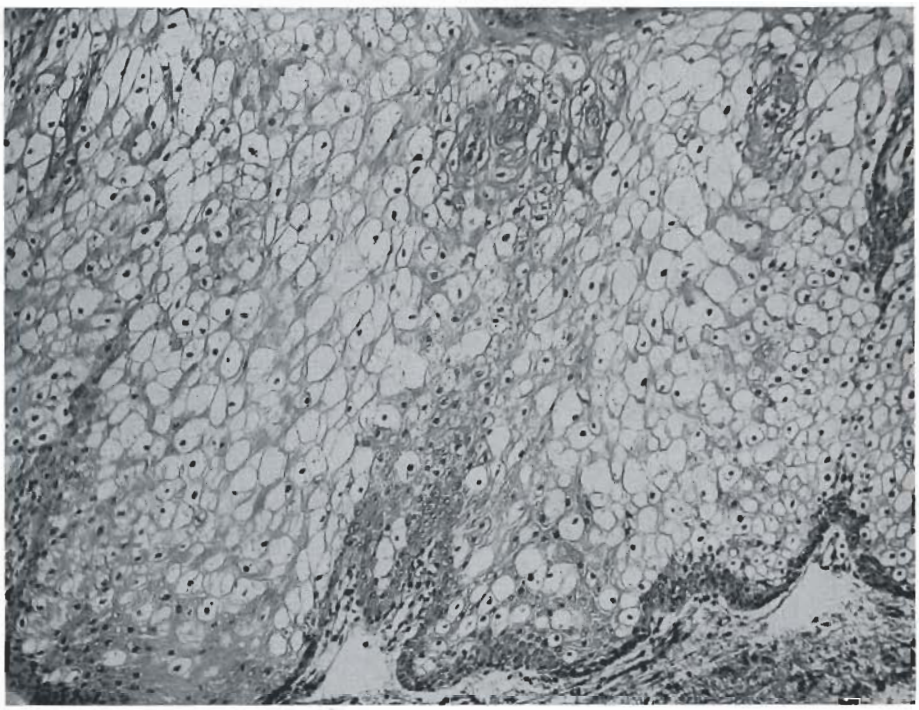

Fig. 4.—Photomicrograph of buccal mucosal lesion. This was identical to lingual change. Note "washed-out" appearance of epithelial cells. This was uniform throughout the whole section.

Microscopic examination of the oral mucosa from the tongue and buccal surfaces revealed identical changes. The epithelium was acanthotic with a uniform intracellular vacuolization. This was uniform throughout all the involved epithelium. The intercellular bridges in the prickle-cell layer were missing. There was marked parakeratosis and absence of stratum granulosum. A Schiff stain did not reveal any Schiff-positive material within the epithelium (Fig. 4).

Summary

A case of pachyonychia congenita, with characteristic gross and microscopic oral changes, has been presented.

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