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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.
ORAL LESIONS ACCOMPANYING PACHYONYCHIA CONGENITA

ROBERT J. GOOBIN, D.D.S., M.S., AND A. P. CHAUDHRY, B.D.S., Ph.D.,
MINNEAPOLIS, MINN.

PACHYONYCHIA congenita is an unusual congenital and sometimes familial
malady that is characterized by dystrophic changes in the fingernails and
 toenails, hyperkeratosis or cornification of the palms and soles, follicular keratosis
of the areoliform type particularly about the knees and elbows, and hyperhidrosis
or excess sweating of the hands and feet. Dystrophic changes are also occa-
sionally observed in the hair or cornae. Verrucous 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
plaque 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 cheilitis is commonly
present.

The disease was recognized as an entity and originally described by
Meesmann and Lewandowski in 1906. 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 "leukokeratosis," it is likely that this term was used in the literal
rather than the microscopic sense, as their photomicrographs indicate hyper-
keratosis or pachydermia orals rather than dyskeratosis.

Case Report

The present case concerns a 17-year-old, 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 present ing signs of deformed discolored 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.

441
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 peel. This with the tongue ulcers closely resembled the lesions described.

Fig. 3—The buccal mu cosa was involved with an identical whitish thickened mucous membrane protruding about the third molar area.

A sepal.
At 11 months old, a scaly papular rash appeared on the knees, elbows, and buttocks. The rash consisted of tiny follicular hyperkeratotic papules. Subsequently, she developed "bumps on her face" which took the form of deep, long-lasting papules. Callouses gradually developed on the palms and soles, and especially between the toes. These breaks dreg 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 consistently present. Family history was entirely negative. Oral examination revealed white thickenings thickening on the buccal mucosa, especially along the interdental line and a diffuse white stinging of the tongue, especially along the lateral margins (Fig. 2 and 3). Laryngeal stroboscopy revealed two small whitish plaques in the interarytenoid area, which were responsible for a mild hoarse voice change.

![Image of a lesion on the tongue](image)

**Fig. 4.** Photomicrograph of buccal mucosal lesion. This was identical to lingual changes. Note "studded-out" appearance of epithelial cells. This was uniform throughout the whole lesion.

Microscopic examination of the oral mucosa from the tongue and buccal surfaces revealed identical changes. The epithelium was acanthotic with a uniform intercellular vacuolation. This was uniform throughout all the involved epithelium. The intercellular spaces were dilated, and the keratinizing layer was absent. There were marked parakeratosis and absence of normal granulocytes. 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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