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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.
Steatocystoma multiplex and pachyonychia congenita are uncommon genodermatoses, judging from the scarcity of reports of either condition in the medical literature. The observation of a patient in whom both conditions exist and can be traced through 4 generations appears to represent a combination of hereditable dermatoses never before reported. The purpose of this paper is to report 8 cases of these combined disorders and present the results of dermabrasion therapy in the management of one patient.

Report of a Case

A 35-year-old white man came to the dermatology clinic complaining of numerous “bumps on his skin and thick nails.” He stated the thickened nails were first noted when he was approximately 7 months of age. His skin was essentially clear until he was 20 years old when numerous yellow, elevated nodules developed on his face. These lesions have progressively increased in size and number and have since appeared over the scalp, axillae, chest, and back.

Family History.—The patient stated his paternal grandfather had many similar cysts and thickening of his nails. Subsequently, 7 other members of the family were similarly afflicted with this combination of dermatoses, including the patient and 2 of his 3 children. The time of onset of the steatocystoma varied from 8 months to 20 years of age, but all individuals involved had the onset of thickened nails before they were a year old (Fig. 1). No member of the family had either of these conditions without the other also appearing. Both sexes were involved, but the majority of those affected were males. No history of consanguinity could be elicited.

Laboratory Studies.—Complete blood count and urinalysis were within normal limits. Serologic test for syphilis was negative. Cultures and potassium hydroxide preparations from several toenails and fingernails were negative for fungi.

Physical Examination.—Examination was entirely within normal limits, except for the skin and nails. Scattered over the face, scalp, chest, axillae, back, and neck, there were approximately 100 yellowish nodules varying from 0.2 to 1.5 cm. in diameter. These nodules were cystic in consistency and were firmly adherent to the overlying skin. The vertex of the scalp showed an irregular alopecia overlying the lesions. The fingernails and toenails were thickened and markedly thickening of the proximal interphalangeal joint and with fragmentation of the free edge. There were numerous linear scars over the axillae, and trunk where cysts had been removed.

Fig. 1.—Genealogic chart (dark areas show those affected): 1 is the patient's grandfather; 2, the patient's father; 3, the patient; 4, the patient's brother; 5, the patient's sister; 6, the patient's son; 7, the patient's daughter; 8, the patient's nephew.

Fig. 2.—Appearance of face before excision.

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Steatocystoma Multiplex with Pachyonychia Congenita

Eight Cases in Four Generations

Submitted for publication July 17, 1961.
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MULTIPLE STEATOCYSTOMA

The arms and legs were free of nodules (Figs. 2, 3, and 4).

Biopsy.—The epidermis was somewhat flattened with loss of rete ridges. The dermis revealed a cystic, epithelial tumor consisting of flattened, stratified squamous epithelium peripherally, with a central cavity filled with keratinous debris. No sebaceous glands were connected with the structure, and the cystic nodule, on serial sections, did not communicate with the surface or with a pilosebaceous apparatus. The surrounding corium was replaced by loose fibrous stroma.

Course in the Hospital.—Initially, one lesion from the chest was excised for pathologic confirmation and a test site was dermabraded on the

Vineyard—Scott
Comment

Figure 1 shows the pattern of involvement in the 4 generations of our family group. The pachyonychia appeared in all patients before the age of 1 year while the steatomata were highly variable at the time of onset. Neither condition has been expressed singularly in any member of the family. Therefore, it appears these genetic traits are linked in their transmission.

Both dermatoses have been considered to be transmitted as a simple mendelian dominant with incomplete penetrance. Our cases coincide with this mode of transmission.

While several authors report the presence of various ectodermal defects associated with steatocystoma multiplex, to our know-

edge the coexistence of pachyonychia congenita has not been previously reported. None of the individuals involved manifested the leukokeratosis or hyperkeratotic eruption associated with pachyonychia congenita. The expression of this genodermatosis, however, may be limited to the nails.  

Burks advocated the use of dermabrasion in the therapy of multiple sebaceous cysts. Our patient was materially improved cosmetically by this procedure. No attempt was made to completely remove the deep cystic lesions, but thorough marsupialization was undertaken. Further observation will be necessary to determine the degree of regeneration of the cysts after dermabrasive surgery.

Fig. 7.—Steatocystoma in a son and daughter (age 12 years).

Summary

Steatocystoma multiplex associated with pachyonychia congenita is rare, occurring in 4 generations of our family involving 8 individuals.
The response of one patient to facial dermabrasion is presented.

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REFERENCES


Fig. 7.—Steatocystoma in axilla of patient's daughter (age 12 years).

Summary

Steatocystoma multiplex associated with pachyonychia congenita is reported as occurring in 4 generations of one family and involving 8 individuals.
Steatocystoma multiplex with pachyonychia congenita. Eight cases in four generations.

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PMID: 13926103 [PubMed - OLDMEDLINE for Pre1966]