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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.
AA—Hereditary Koilonychia

B.B. (JHH 1339988), a 39-yr-old male of “Lumbee Indian”* descent, was hospitalized for treatment of an astrocytoma and was found to have koilonychia.

Review of the family history showed that 4 generations were involved (Fig. AA-1). Consanguinity was denied.

Physical examination showed a normal appearing male with sparse, blond hair (secondary to radiation therapy for brain tumor). The teeth were normal. The nails of all digits were spoon-shaped, being more severely affected in fingers than toes (Figs. AA-2A and B). The nails were firm and slightly thicker than normal. No cataracts were present.

Florence Char

*Triracial, i.e. Indian-Negro-Caucasian, group from North Carolina, taking its name from the Lumbee River.

![Pedigree diagram]

Fig. AA-1. Pedigree.

Figs. AA-2A (top) and B (bottom). Hands and feet of B.B. The spoon-shape of the nails was most striking in the thumbs.

BB—Pachyonychia Congenita in Father and Son

The father I.L. (HLH 15922), was reported by Wright and Guequiere (Arch. Derm. Syph. 55:819, 1947). He was the son of unaffected, unrelated parents and in him the condition may represent the result of new dominant mutation. His son is identically affected.

When the father was first seen here at age 10 yr, ulceration at pressure points, including the feet, was a problem and ulceration occurred also in the mouth and particularly at the commissures of the lip (angular stomatitis). Most of the fingernails were removed surgically when the patient was in his teens.

In addition to striking changes in the nails (Figs. BB-1A and B) both father and son show extensive hyperkeratosis of the soles of the feet (Fig. BB-2), leukokeratosis of the tongue, buccal mucosa and curious tiny horns in many areas of the skin (Fig. BB-3).

Presently (1970) the father is 40 and the son 10 yr old (Figs. BB-4A and B). The father develops miliaria in a hot environment and suffers from hidradenitis suppurativa of the axillas, especially in hot weather. In the son, cracking of the hyperkeratotic soles leads to foot ulcers which are slow to heal.

Victor A. McKusick
Figs. BB-1A (top) and B (bottom). *LL*. Striking changes in the fingernails (age 10 yr) and toenails (age 35 yr) are shown.

Fig. BB-2. *LL*. Hyperkeratosis of the soles with extensive fissuring.

Fig. BB-3. *LL*. Note the small keratotic horns scattered over the upper arm and chest wall.

Figs. BB-4A (top) and B (bottom). The nail changes are striking in the 10-yr-old son of *LL*. 