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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Q. Ectodermal Dysplasias

PACHYONYCHIA CONGENITA SYNDROME

Thick Nails, Hyperkeratosis, Foot Blisters

Pachyonychia congenita is an ectodermal dysplasia described by Jadassohn and Lewandowsky,[1] in which there is excessive keratin and occasionally extra teeth.

ABNORMALITIES

Nails. Progressive thickening of anterior half. The nails may eventually be hypoplastic or even absent.

Skin. Patchy to complete hyperkeratosis of palms and soles, callosities of feet that blister easily, keratosis pilaris with tiny cutaneous horny excrescences.

Epidermal cysts filled with loose keratin on face, neck, and upper chest.

Mucous Membranes. Leukokeratosis of mouth and tongue, especially in positions of increased trauma.

Dentition. Erupted teeth at birth, lost by four to six months.

OCCASIONAL ABNORMALITIES. Corneal thickening, cataracts, thickening of tympanic membrane, hyperhidrosis. Dry and sparse hair. Osteomata, intestinal diverticula.

NATURAL HISTORY. The complications are obvious. Usually the nails are grossly thickened by one year of age. Complete surgical removal of the nails is sometimes merited, although any matrix left behind will reform abnormal nails.

ETIOLOGY. Autosomal dominant, with wide variability in expression. Predominantly found in Slavs and Jews of Slavonic origin. The basic mechanism of the disease is unknown; however, vacuolization of the cytoplasm of nail matrix cells may be of significance.

REFERENCES

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Negro infant showing al
and lateral palate.