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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PACHYONYCHIA CONGENITA

Gerson Carakushansky

Cardinal Clinical Features

Piled-up, horn-like thickening of the nails and other abnormalities of keratinization, including thickened, hyperkeratotic skin and callosities of the feet.

Clinical Picture

In 1906 Jadassohn and Lewandowsky[1] described the abnormality of the nails to which they gave the name pachyonychia congenita. It is characterized by distinctive, excessively thickened nails. These patients also have palmar and plantar hyperkeratoses, follicular keratosis of the skin and leukokeratosis of the oral mucous membranes. Of these manifestations, only the nail changes are constantly present; they usually represent the earliest manifestation of the disorder.

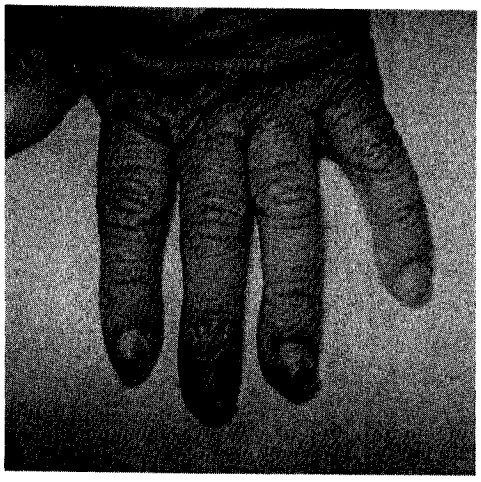
At birth the nail may appear normal, but shortly thereafter the nail bed develops a yellow-brown discoloration. There is uplifting of the nail. The nails may be smooth and of normal appearance at the base, but at the free end they are wedge shaped and raised by a horny mass of material in the hyponychium. This leads to an angular projection up from the nail bed that is particularly prominent in the fingernails. The nail plates are extremely hard and are firmly attached to the nail beds. Ordinary measures were not sufficient to trim the nails of the patient originally reported by Jadassohn and Lewandowsky,[1] and a hammer and chisel were used for this purpose. The mother of the child described in the Case Report used a saw to reduce the size of the heaped-up, horny projections of his nails. Sohrweide[2] reported a patient in whom the nails were 0.7–1 cm in thickness at their free margins. Because of the projections, these nails are subject to injury. Paronychia may be a problem. Some babies with pachyonychia congenita have attracted attention because they were born with erupted teeth. Murray[3] first noted the association of erupted teeth at birth and pachyonychia congenita. Jackson and Lawler[4] and Soderquist and Reed[5] also observed this association, which may be familial and may be transmitted as a dominant through three or more consecutive generations.

Skin involvement may be noticeable in the presence of keratotic patches on the palms and soles. The entire skin may appear thickened. Follicular hyperkeratosis with well-developed keratin plugs may occur on the extensor surfaces around the large joints. There may be bullae, which may be associated with hyperhidrosis of the palms and soles. Areas such as the heels may readily form blisters. Bullae may develop over callosities and may be painful. In rare instances breakdown of the skin on weight-bearing surfaces may be so severe that admission to hospital and skin grafting are required. Occasionally an unusually dry kinky hair is seen on the scalp of white patients.[6] Nodular epithelial cysts have been described.[5]

Oral leukokeratosis is not found in all patients or kindreds but, when present, may be an early manifestation in infants. In some instances the oral lesions, which may be seen as early as at birth, have been mistaken for thrush. These lesions take the form of diffuse, white or gray-white plaques, most commonly involving the dorsal surface of the tongue. The lateral borders of the tongue may appear especially thickened. Other mucous membranes have on occasion shown leukokeratosis, including the nasal, tympanic and perianal mucosa.

Other reported defects, such as mental deficiency,

Fig. 1.–J. A. S. (Case Report). The nails were typical of pachyonychia congenita. Note the claw-like incomplete pointed cylinder or cone.



corneal changes and diffuse gastrointestinal polyposis, do not appear to be consistent features of the syndrome.

Differential Diagnosis

The fully developed syndrome is seldom mistaken for anything else. The differential diagnosis includes hereditary onychogryposis, which is also inherited as a dominant trait, but there are no abnormalities other than the nail changes. In epidermolysis bullosa, blister formation may be associated with nail thickening, but the picture is really very different.

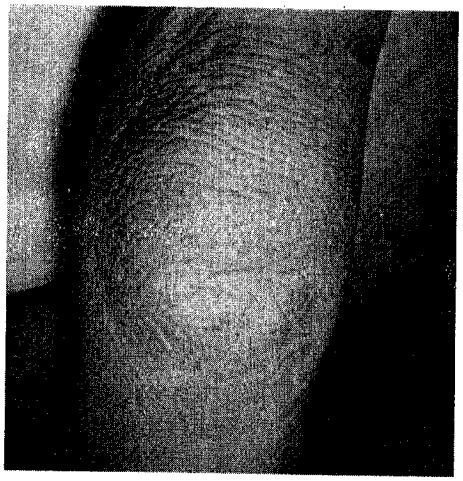
Case Report

J. A. S., a 3-year-old black male, was admitted to the Pediatric Service of the Federal University of Rio de Janeiro because of a chronic condition involving the skin and nails. He was the second child of healthy parents. The first child was free of the condition. In the second week of life all of the patient's fingernails and toenails had turned yellow. By 1 year of age the nails were grossly thickened and had begun to curl. At the time of admission the nails were remarkable. The piled-up nail growth had led to curved, claw-like or horn-like, incomplete pointed cylinders or cones. Our first pictures were lost; Figure 1 represents the appearance after the mother trimmed the nails with a saw. Follicular keratotic lesions were present over the knees (Fig. 2) and at the base of the toes (Fig. 3). This patient's entire skin was thickened and dry and had an ichthyotic appearance. The ear was unusual (Fig. 4).

Genetics

Cockayne[7] observed that the syndrome was in many cases inherited directly from a parent who suffered from the condition and that parental consanguinity was absent. A single autosomal dominant gene with wide expressivity would appear to satisfy the evidence of the pedigrees described.[4] Kumer and Loos[8] found 24 affected members in a pedigree comprising five generations. The varied expression in different kindreds suggests that there may be a number of different dominant genes responsible for similar but distinct syndromes.

Fig. 2.—J. A. S. Close-up of the knee showing follicular keratosis.



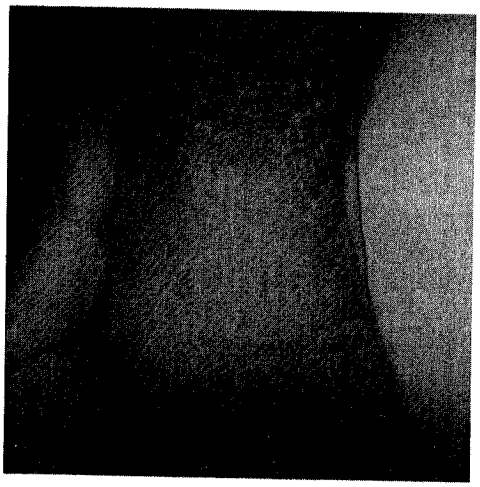
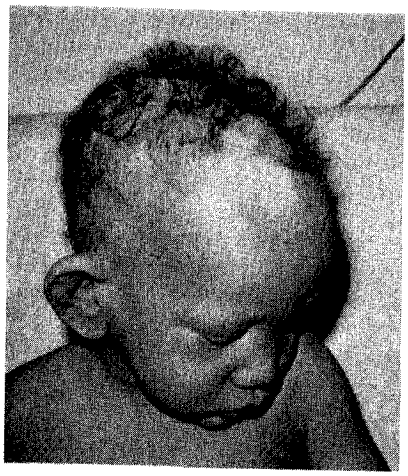


Fig. 3.—J. A. S. The foot illustrates the presence of keratotic lesions at the base of the toes.

Treatment

Pachyonychia of the nails is of only cosmetic importance but is difficult to treat. Some rather vigorous attempts have been made because of the possible psychologic problems. Children are cruel, and patients with this disorder have been labeled as "witches." Even avulsion of the nail brings only temporary relief. Amputation of the distal phalanges has been used, but this does not appear justified. There is evidence that

Fig. 4.—J. A. S. This patient also had an unusual ear.



the distal matrix produces excessive amounts of keratin and therefore produces a material similar to that of a horse's hoof. Even after deep surgical excision of the nails,[9] a small portion of the distal matrix usually is left and nail again forms at the site.

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