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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.
Picture of the Month

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The statements listed below are best associated with which of the figures: (a) This is a condition of autosomal-dominant inheritance affecting all nails. (b) This is a condition of autosomal-dominant inheritance affecting only one or two nails. (c) This is not a hereditary condition and affects all nails.

Figure 1.

Figure 2.

Figure 3.

Figure 4.

Figure 5.

Figure 6.

Accepted for publication January 18, 1991.
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—Bennett

AJDC—Vol 145, November 1991  Picture of the Month—Cohen & Hebert 1301
Pachyonychia Congenita

Figs 1 and 2.—A patient with congenital malalignment of the great toenail in which the right hallux nail is deviated in a lateral direction and has multiple transverse ridges on its surface.

Figs 3 and 4.—Thin, friable nails with loss of nail luster, multiple pits, excess longitudinal ridging, and distal notching that splits the nail into layers in a child with 20-nail dystrophy of childhood.

Figs 5 and 6.—Distal hypertrophy of the nails with marked subungual hyperkeratosis in a boy with pachyonychia congenita.

(a) Pachyonychia congenita is a rare genodermatosis of autosomal-dominant inheritance with a high degree of penetrance and variable phenotypic expression. This disorder, first described by Jadassohn and Lewandowsky in 1906, has also been referred to as Jadassohn-Lewandowsky syndrome. The onychodystrophy typically appears within the first 6 months of life, and may be present at birth. It is characterized by hypertrophy and distal subungual hyperkeratosiıs of all nails. Secondary complications include nail shedding, chronic paronychial infections, and increased susceptibility to local traumatic injuries. The only effective treatment of the nail dystrophy has been surgical removal of the entire nail. Radical excision of the nail, nail bed, and nail matrix, followed by vigorous curettage and electrodessication of the bed and matrix with subsequent skin implantation at the site of the removed nail has provided good functional and esthetic results. In addition to onychodystrophy, mucosal and cutaneous manifestations associated with pachyonychia congenita include palmoplantar hyperkeratosis, leukokeratosis of the tongue and oral mucosa, follicular keratosis (especially on the elbows and knees), bullae and erosions on the palms and soles, natal or neonatal teeth, and angular cheliosis. Hair anomalies, corneal dyskeratosis, hoarseness, cataracts, mental retardation, and alopecia are less frequently noted features of pachyonychia congenita.

(b) Congenital malformation of the great toenails is an autosomal-dominant inherited onychodystrophy with variable expression that affects the nail of one or both halluces. The disorder is typically present at birth and characterized by lateral deviation of the nail with respect to the long axis of the phalanx. Transverse ridging is often present across the entire surface of the nail, and, less commonly, the nail may be discolored, thickened, shortened and/or onycholytic (characterized by loosening or separation of the nail from the nail bed). Complications of congenital malformation of the great toenails include ingrown toenails and hemionychogryphosis (a hooked or incurved nail). Although spontaneous improvement has been observed in a few children, the disorder usually persists into adulthood if not corrected earlier. Surgical realignment of the entire nail may be necessary for individuals with either marked nail deviation or condition-related disabling sequelae. The clinical differential diagnosis of congenital malformation of the great toenails may be made based on the presence of bacterial and candidal paronychia, ingrown toenails, congenital hypertrophy of the lateral nail folds of the hallux, dermatophyte infection, and onychogryphosis.

(c) In 1977, Hazelrigg et al described six healthy children with an onychodystrophy that uniformly affected the nails of all 20 digits: 20-nail dystrophy of childhood. There was no evidence of psoriasis vulgaris, lichen planus, alopecia areata, dermatitis, or ectodermal defects. Therefore, this condition was assumed to be idiopathic. In 20-nail dystrophy of childhood, the onychodystrophy is asymptomatic and appears insidiously. The nails are characterized by excessive longitudinal striation and loss of nail luster. The nails are thin, fragile, and have distal notching that splits the nails into layers. Regular manicuring and application of nail lacquer are the most effective palliative treatments. Similar features of this nail dystrophy have been noted in patients with lichen planus, alopecia areata, psoriasis vulgaris, and IgA deficiency. Hence, it is perhaps most appropriate to reserve the diagnosis of 20-nail dystrophy of childhood for children with idiopathic onychodystrophy that affects all of the nails.

The descriptive term for rough nails, trachyonychia, should be used when the nail changes traditionally found in 20-nail dystrophy of childhood are present in individuals with dermatologic or systemic illness that has also been associated with similar nail findings or in children or adults in whom less than all 20 nails are involved.

References