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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.
PACHYONYCHIA CONGENITA (JADASSOHN-LEWANDOWSKI SYNDROME)

Case Report

RALEIGH R. WHITE, W. M.D. AND R. BARRETT NOONE, M.D.


*Pachyonychia congenita,* first described in 1906, is a rare genodermatosis characterized by nail dystrophies. Although the manifestations of the syndrome usually are diagnosed and treated by dermatologists, surgical treatment offers the best permanent relief of the severe nail deformity. Our recent experience with the surgical treatment of *pachyonychia congenita* is the basis for this report.

**CASE REPORT**

A 15-year-old black male was referred to us by his dermatologist for surgical evaluation. At birth he had abnormally heavy nails on all fingers and toes. As he grew older, the nails became increasingly thickened, opaque, and malformed.

He had been thoroughly evaluated twice for chronic fungal infection of the nails, and the fungal work-up was repeatedly negative. Systemic and topical antifungal therapy, as well as topical corticosteroid medication, had been unsatisfactory. He had suffered repeatedly from paronychia and chronic infection of the nail-finger complex, requiring medical treatment each time. His dermatologist had removed one hyperkeratotic nail plate by conservative excision, as a therapeutic trial. Within a few weeks, the nail was returning to its former hyperkeratotic condition.

At examination, the nails of his fingers were so thick that he experienced difficulty in putting on conventional winter gloves. His toenails were similarly abnormal, and the nail length was controlled only by use of a heavy-duty file (Fig. 1).

![Fig. 1. Preoperative appearance. The fingernails are extremely distorted and thickened, and the combined nail plate and hyperkeratotic nail bed is approximately 6 to 8 times its normal thickness.](image1)

![Fig. 2. Postoperative appearance. The split-skin grafts were placed immediately on the abraded insertion of the extensor tendons, after complete excision of the entire nail-matrix complex.](image2)

From the Department of Plastic and Reconstructive Surgery of the Harrison Department of Surgical Research of the School of Medicine at the University of Pennsylvania.
He had difficulty with the fine manipulation of small objects, such as picking up a coin from the table top, both because the usual advantages of a well-formed nail were absent and because fingertips-to-fingertip contact was impaired by the bulky nails. In addition, a severe social problem was rapidly developing for him, because of peer-group rejection and ridicule.

We also noted several associated stigmata of pachyonychia congenita, including acelomiform follicular keratosis of the skin of his knees and elbows, hyperkeratosis of the soles of his feet, and leukokeratosis (sponge nevi) of the tongue and buccal mucosa. His hair growth appeared normal, and he did not complain of palmar or plantar hyperhidrosis. He did not demonstrate corneal dyskeratosis or abnormal thickening of the tympanic membrane. His voice quality was normal, and he gave no childhood history of hoarseness or unusual upper airway distress. His somatic growth had always been below standard; at the age of 15 years, he was of slight build (38.1 kg), and he appeared distinctly younger than his stated age.

Although the condition is thought to be a genodermatosis transmitted as a dominant autosomal gene with incomplete penetrance, our patient's family history revealed no known occurrence of pachyonychia congenita in the previous two generations. (Such a case has been reported as representing a new dominant mutation.)

Because of sporadic reports of successful definitive treatment by radical excision of the nail plate, nail bed, and germinal matrix and covering the wound by a split-skin graft, we offered the patient such an operation. He chose to have all fingers of the left, non-dominant hand operated on as a trial.

The operation was done under general anes-
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Anesthesia, with a pneumatic tourniquet. Six months postoperatively, no regeneration of the fingernails was seen in the treated hand (Fig. 2). The surgical result was highly acceptable to the patient and to his family. He requested similar surgical treatment for his right hand.

OPERATIVE RATIONALE AND TECHNIQUE

Recent studies from Sweden suggest that the hard nail plate itself is not the primary site of the pathological process in pachyonychia congenita. The nail plate appears quite normal on histological examination. However, the supporting nail bed (also called the ventral nail plate) is markedly abnormal in pachyonychia congenita (Fig. 3), measuring about 6 times its normal thickness. The matrix of the nail bed is markedly more cellular than normal, and the connective tissue just beneath the nail bed is characterized by pronounced fibroblastosis. As the nail bed thickens, it progressively elevates and distorts the overlying nail plate.

Considering both the failure of the previous conservative resection of the nail plate in our patient, and the concept of the nail bed as the primary site of pathology, we concluded that radical treatment was indicated. Our resection included excision of the hard nail plate, the abnormal germinal matrix, and the entire thickness of the nail bed (Fig. 4).

Wound coverage was accomplished with split-skin grafts. The skin grafts were placed on the exposed extensor tendon insertion and the associated periosteum of the terminal phalanx, because all the tissue dorsal to these structures had been excised. The dystrophic nail complex was totally excised; no nail regeneration ensued, and the patient was well pleased with the final result.

SUMMARY

We report an illustrative case of pachyonychia congenita, with a description of the presenting complaints, the associated physical findings, the rationale for surgical treatment, and the technical aspects of the surgical treatment.

Raleigh R. White, IV, M.D.
Scott and White Clinic
Temple, Tex. 76501

Dr. Noone is Chief of the Section of Plastic and Reconstructive Surgery at the Pennsylvania Hospital, Philadelphia. Dr. White is now at the Scott and White Clinic in Temple, Texas.

REFERENCES


Adduction of the following two conditions of the finger: 'man-in-a-hole' and 'man-in-the-cap.' The former condition consists of a ridge along the side of the middle finger. The latter consists of the pollicis, an adductor muscle acting on the side of the finger. Following a complete contraction, the finger is straightened and the structure of the adductor muscle is achieved. To perform this maneuver, the thumb is adducted to the side of the middle finger. This is achieved by a transverse incision in the skin and covering the graft with a skin flap. The flap is then sutured to the skin flap.

From the Orthopedic Research Center.