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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
Surgical Management of the Nail Changes

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The fingernails of a woman with pachyonychia congenita were ablated by several different methods. The most effective, most rapidly performed, and most acceptable method was vigorous curettage and electrofulguration of the matrices and beds of the nails. The responses to treatment suggest that the matrix, rather than the bed, is the site from which the abnormality of the nails develops in pachyonychia congenita. Removal of the nail beds is not necessary to prevent recurrence of deformed nails, but it may reduce hyperkeratosis in their sites.

INTRODUCTION

Pachyonychia congenita is a rare, autosomal dominant disorder characterized by thickened finger and toe nails, benign leukoplakia of the oral mucosa, keratosis pilaris-like lesions, plantar and palmar hyperkeratoses, and painful bullae on the feet. The most bothersome consequences to patients with the condition are the bullae and the abnormal nails. The thickened nails reduce dexterity, are cosmetically embarrassing, and painful when traumatized, as they easily are.

Several surgical methods of managing the abnormal nails of pachyonychia congenita have been reported. Simple avulsion provides only temporary improvement, because the nails grow back to their original forms. Wright and Guequierre reported a patient in whom they amputated the entire distal phalanx of each finger and thereby rid their patient of the troublesome nails. We consider that method excessively and unacceptably radical. Cosman et al. treated a patient with complete surgical removal of the nail matrices and nail beds, except for small distal portions, followed by coverage by full-thickness grafts. They had excellent results except for hyperkeratinization on the residual portions of the nail beds. They consequently recommended complete removal of the matrix and nail bed. White and Noone treated a patient by complete excision of the matrices and nail beds followed by coverage with split-thickness grafts, and achieved excellent results.

We recently had the opportunity to treat the nails of a patient with pachyonychia congenita by various methods, and we herewith report what we deem to be the simplest and best method, and what is the site of the faulty production of nail.

CASE REPORT

A 37-year-old woman was referred to us for removal of her fingernails. She was very troubled by the thickness of her fingernails because they interfered with the dexterity of her fingers and she thought that the condition was the cause of her dismissal from two jobs. Previous removal of the nails and ablation of their matrices from the great toes had produced results gratifying to her. The patient’s paternal great grandfather had, and her father, two sisters, and son all have fingernails characteristic of pachyonychia congenita.

On examination, all ten fingernails appeared similar. The plates seemed to be of normal thickness, but were elevated by markedly excessive keratosis beneath the distal half of each nail (Fig. 1). There were several calluses on the hands and two areas of oral leukoplakia. Neither follicular hyperkeratosis nor bullae were found.

A biopsy specimen of the proximal nail fold, matrix, and the proximal portion of nail bed were found to be histologically normal, but another of the distal nail bed showed hyperkeratosis, irregular acanthosis, papillomatosis, and focal areas of benign dyskeratosis with ovoid eosinophilic bodies in the hyperkeratotic stratum corneum (Fig. 2). Whereas normally the nail bed does not have a granular layer, sections from our patient showed a granular layer in the distal portion of the nail bed, which in places consisted of ker-
atohyaline granules that varied markedly in size. In those places too there were some vacuolated keratinocytes, but no tiers of parakeratotic nuclei over the papillary tips as are seen in verrucae vulgares.

Several different surgical procedures were performed on the fingernails. On the thumb, index finger and middle finger of the left hand, the entire nail matrix and nail bed were excised by cold steel in the manner described by White and Noone, except that the underlying tissue was allowed to granulate and re-epithelize spontaneously. On the left ring finger, only the nail matrix was removed as described by Johnson and Ceil-ley. On the left small finger, only the nail bed was removed completely; the distal one-third of the matrix was removed solely to make certain that all of the nail bed had been removed. In each case, tissue was removed to the depth of underlying bone and the wounds were allowed to granulate and re-epithelize spontaneously. The procedures were done in an outpatient facility under local anesthesia with a 1% solution of lidocaine.

After nine weeks, good healing was evident in all fingers. There was some recurrence of spicules of nail on the left middle finger. On the left small finger, where

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**FIGURE 1.** Classical appearance of the fingernails of the patient showing subungual hyperkeratosis and elevation of the nail plates.

**FIGURE 2.** Photomicrograph of a specimen of the bed of a nail affected by pachyonychia congenita showing a focus of a granular layer and ovoid eosinophilic bodies in the hyperkeratotic stratum corneum. (H&E. ×80)

**FIGURE 3.** Clinical appearance of the left little finger showing regrowth of a typical nail of pachyonychia congenita after removal of the nail bed and distal one-third of the matrix.
the proximal two-thirds of the matrix had been left, there was growth of a new nail which was like that of the previously existing nail with the same markedly excessive hyperkeratosis beneath the distal portion of the nail plate (Fig. 3). Subsequently, that finger and the five fingers of the right hand were all treated by complete ablation of the matrices by vigorous curettage and fulguration in depth to periosteum and laterally to fatty tissue. The nail beds were also curetted and fulgurated, but not all the way to periosteum. Healing was permitted to proceed by secondary intention. The patient herself cared for the surgical wounds by gentle cleansing with a solution of 3% hydrogen peroxide followed by application of polysporin ointment three times daily.

Examination two months later revealed good healing of all fingers. Three fingers had small spicules of regrown nail (Fig. 4), which were so easily controlled by clipping that the patient did not desire further destruction of them. Slight hyperkeratosis of the nail bed developed on the left ring and right middle fingers. The rest of the nail beds were smooth (Fig. 5). The new skin over what were originally nail beds was bound down, but less so on the fingers treated by curettage and electrofulguration. The patient thought that the
healing on the fingers treated by curettage and electrofulguration was more satisfactory. Early postoperatively, pain was a little more intense after curettage and fulguration, but it was less persistent than after excision by cold steel. At follow-up, 15 months later, the appearance of the patient's fingers were as pictured in Figures 6 and 7; the patient said she was pleased with the result.

A summary of procedures done and results achieved are tabulated in the table. The patient is presently working as a nurse's aide and plays the piano, activities she was not able to engage in before the operations.

**DISCUSSION**

The precise location from which the pathological alteration in the nails of pachyonychia congenita develops is not known. The possibilities most often discussed cite the nail bed, which produces such marked hyperkeratosis that the otherwise normal nail is pushed up, or the matrix is abnormal in producing nail plate, or both bed and matrix are abnormal. Pinkus and Kelly believe that the very distal nail bed (which they refer to as the terminal nail matrix) contributes loose keratin to the ventral nail plate in normal digits. It is their belief that in pachyonychia congenita this area is hyperplastic and produces abundant keratin that causes the nail deformities. We believe that our experience with the left ring and small fingers disproves Pinkus and Kelly's theory.

Forsli et al. studied a patient with pachyonychia congenita by a micro-radiographic technique. They found no pathologic changes in the nail plates, but did find marked increase in thickness and dry mass of nail beds. They concluded that the defect was solely in the nail bed and not in the germinal matrices. Again, we think that our experiences with the left ring and small fingers disproves this theory.

Joseph observed that the proximal nail folds in pachyonychia congenita resemble those of a three-month fetus in that they are tilted upward. He suggested that the abnormality may be due to arrested development of the nail folds, which continue to slant so that nail plates are angled away from the nail beds. He then speculated that the hyperkeratosis of the nail bed may in part be due to altered keratinization that occurs whenever nail beds are not tightly covered by nail plates. A granular layer in the distal portion of nail beds in pachyonychia congenita may develop and produce hyperkeratosis secondary to the elevation of overlying nail plates. During early fetal life, nail beds have granular layers until nail plates grow out and firmly cover them. When a nail plate is avulsed and its nail bed no longer has a tightly attached covering, it once again develops a granular layer. The surgical results in our patient do not prove Joseph's theory of a tilted matrix, but they are consistent with it.

As of now, it appears that satisfactory treatment of the abnormal nails of pachyonychia congenita require elimination of nail matrices. We found that removal of the matrix alone by excision or physical destruction was effective and sufficient because there were then no nail plates. Treatment of nail beds does not seem necessary. Our patient preferred curettage and electrofulguration to conventional surgery. Moreover, curettage and electrofulguration are quicker and easier to do. Chemical destruction of the matrix would probably also be effective. Perhaps a procedure could be developed to correct the lie of the proximal nail folds in pachyonychia congenita that causes plates to grow off and away from beds that then become hyperkeratotic. If that becomes technically possible and if Joseph's theory is correct, properly adherent plates ought to develop and subungual hyperkeratosis ought not supervene.

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<td><strong>Summary of Procedures Performed and Results Achieved</strong></td>
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<td><strong>Fingernails</strong></td>
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<td>Of the left thumb, index, and middle fingers</td>
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<td>Of the left ring finger</td>
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<td>Of all of the fingers of the right hand</td>
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REFERENCES