Use of Articles in the Pachyonychia Congenita Bibliography

The articles in the PC Bibliography may be restricted by copyright laws. These have been made available to you by PC Project for the exclusive use in teaching, scholarship or research regarding Pachyonychia Congenita.

To the best of our understanding, in supplying this material to you we have followed the guidelines of Sec 107 regarding fair use of copyright materials. That section reads as follows:

Sec. 107. - Limitations on exclusive rights: Fair use
Notwithstanding the provisions of sections 106 and 106A, the fair use of a copyrighted work, including such use by reproduction in copies or phonorecords or by any other means specified by that section, for purposes such as criticism, comment, news reporting, teaching (including multiple copies for classroom use), scholarship, or research, is not an infringement of copyright. In determining whether the use made of a work in any particular case is a fair use the factors to be considered shall include - (1) the purpose and character of the use, including whether such use is of a commercial nature or is for nonprofit educational purposes; (2) the nature of the copyrighted work; (3) the amount and substantiality of the portion used in relation to the copyrighted work as a whole; and (4) the effect of the use upon the potential market for or value of the copyrighted work. The fact that a work is unpublished shall not itself bar a finding of fair use if such finding is made upon consideration of all the above factors.

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.
PLASTIC SURGERY IN PACHYONYCHIA CONGENITA AND OTHER DYSKERATOSES

CASE REPORT AND REVIEW OF THE LITERATURE


garden cosman, l.d.h., m.c., usn., francis c. symonds, jr.,

m.d., and george f. chikelair, m.d.

new york, new york

Pachyonychia congenita is a comparatively rare hereditary ectodermal dyskeratosis. It is characterized by markedly thickened nails on all of the extremities, scattered recurrent bullous skin lesions, leukokeratosis of the oral membranes, and, occasionally, dystrophic changes in the hair and other ectodermal appendages. One of the most constant features is the presence of large, curved, raised, dark nails, convex laterally, and with heaped up hypochromia. This aspect of the condition appears very early in life. The bullae, when they occur on weight-bearing surfaces, may break down and become painful. The nails and the bullae constitute the chief complaints of the patients. The successful removal of the nails is of functional and emotional importance. This paper describes a case in which this was attempted and adds the condition to those other ectodermal dyskeratoses in which plastic surgical techniques offer amelioration of symptoms.

CASE REPORT

PH 1306586. This 141/2-year-old white girl was seen in consultation by the Plastic Surgery Division of the Department of Surgery, Presbyterian Hospital, concerning the removal of the nails of both hands. She had first been seen at the Medical Center at the age of 1 month. Two days following birth a whitish crust had appeared in her mouth and “thrush” had been diagnosed. In the second week of life all of her fingernails and toenails had turned yellow, thickened and had begun to curl (fig. 1).

The child had otherwise appeared well. The mother gave a history of having had thickened nails starting at age 6 months. The nails had been removed at age 9. Osteomyelitis of all the distal phalanges had ensued with sequestration of bone fragments over the course of a year thereafter (fig. 2). The mother had had many recurrent bullae with occasional ulceration of such lesions on her feet and had been treated for this before the birth of her child (fig. 3).

Multiple cultures of the child’s thurn-like mouth lesions failed to demonstrate Monilia. A course of Dioceratin therapy following a tentative diagnosis of acrodermatitis enteropathica produced no change. In view of the familial history, a diagnosis of pachyonychia congenita was made and the child followed in clinic. At the age of 2 years, skin bullae appeared, completing the clinical picture of pachyonychia. At this time, also, a male sibling was born whose nails also thickened at the age of 2 weeks (fig. 4).

At age 3 years, the mother requested removal of her daughter’s nails before the child became disturbed by the comments of others. The child’s nails had grown in size and could not be cut with the usual instruments. The nails were removed by excision through the nail bed. Despite the fact that the nails were thickened the procedure had little discomfort.

Removal of the nails was performed for both hands. The nails were quite thick and had parakeratosis in the nail plate.
for ectodermal dysplasia, or all of the extremities or of the oral membrane, or of other ectodermal structures of large, curved, ingrowing onychomycosis. This condition usually occurs when they occur during pregnancy. The nails and skin symptoms are the successful reports. This paper describes a case of a patient with a successful amelioration of symptoms.

Fig. 1. Patient's finger nails at age 1 month

Fig. 2. Mother's hands showing deformed finger tips resulting from osteomyelitis of distal phalanges following nail removal.

not be cut with heavy scissors. The mother recalled many miserable years prior to the removal of her nails, and had other children refusing to sit near "the witch" in the schoolroom. Despite the healing difficulties she had had, and her residual deformities, she felt the procedure had been well worth while.

Removal of the child’s nails was first attempted on the Surgical Service. The nails of both hands were avulsed, the matrix scraped, and punch grafts applied to the nail beds. The pathology report on a portion of nail matrix stated that, "Epidermis shows a papillary structure with very elongated and thin papillae. Keratin layer is extremely thick and parakeratosis is evidenced by nucleated squames in upper keratin layer. Thick keratin
Fig. 3. Ulcerated bullae on mother's foot. Note partial losses of distal toe phalanges.

Fig. 4. Recent appearance of brother's hands, at age 4 years, showing untreated nails.

Layer has no regular shades of staining. The right hand showed otherwise little atrophic action.

At the mother's Service. The condition was:

There were scattered bullae and abdomen involved the venous system.

Operation was dorsal rectangular flap with the split section could be that point distally excised. The phalanges were cured.

Since the graft and creeping subcutaneous thymus graft abdominal donor wound but regrew. The condition then operated up and there had been...
layer has no regular distribution or maturity of different layers as evidenced by various shades of staining (fig. 5). Within 2 months, all of the nails had regrown. Those on the right hand showed no effect of surgery. The nails on the left hand were fragmented but otherwise little affected.

At the mother's urgent request, the child was readmitted and seen by the Plastic Surgery Service. The condition of the nails was as described. The toenails were similarly deformed. There were scattered bullae and resolving crusted areas over the dorsum of the hands, feet, knees and abdomen. Whitish plaques were present on the buccal mucosa and bullous lesions involved the vermilion (figs. 6 and 7).

Operation was carried out on the right hand under tourniquet ischemia. Proximally based dorsal rectangular flaps were elevated, taking care to leave the epithelium underlying the flap with the specimen. Dissection was carried proximally until the extensor tendon insertion could be made out. The nail and all the subjacent tissue lying over the bone from that point distally was excised, save for a small distal portion of the "sterile" matrix. The excision was carried laterally and volarward until finger pulp fat bulged. The exposed phalanx was covered, The dorsal flap was then replaced, and the remainder of the exposed phalanx was covered with full-thickness skin grafts obtained from the innominate fold.

Since the grafts were small, it might be argued as to whether contraction of the wound and creeping substitution from the edges of graft take had occurred. In the case of the larger thumb graft, however, it seemed clinically certain that the graft had taken. The abdominal donor site incision healed well; a large bulla formed over the rectal part of the wound but regressed spontaneously without changing the clinical course of wound healing.

The condition of the right hand at 2 months was satisfactory (fig. 8). The left hand was then operated upon. However, 10 months after operation small spicules of nail were present, and there had been considerable piling up of keratin on the residual areas of sterile matrix.
Fig. 6. Patient's right hand with thick tubular nails and healing bullous lesion on dorsum of thumb.

Although the mother was pleased with the result and the fingers were of good shape and functioned well, it seemed likely that still further attempts at removal of nail growing tissue might be needed in the future.

**DISCUSSION**

Pachyonychia congenita was first described by Judassohn and Lewandowsky in 1906. While doubt was initially raised as to whether the defect was a specific entity or a part of other dyskeratoses, a consensus now accepts pachyonychia as a single syndrome. The condition is generally considered to have as its primary manifestations nail thickening starting very early in life, skin bullae occurring without necessary relation to trauma, and leukokeratosis of the oral mucosa. Follicular plugging, especially on extensor surfaces, is frequently recorded. Dystrophic changes in the hair, the teeth, and other ectodermal appendages may or may not occur. Other reported defects, including cataracts and mental disorders, do not appear to be consistent parts of the disease.

Whether the condition has a specific histology remains a moot point. Andrews and Strumvasser described appearances much like those of Darier's disease with which they concurred in the same in every case the syndrome considered the syndrome occurred in the hand, while unaffected siblings suggested an autosomal, nait reporting a fam concluded that a s dominant trait. The mother an
with which they initially classified pachyonychia. Other observers have not concurred in these findings. The histology of the nail matrix has not previously been reported.

The heredity of the defect is incompletely worked out and may not be the same in every case. Cockayne, in 1933, in a review of pedigrees, pointed out that the syndrome could not be recessive since, in many cases, one of the parents had suffered from the condition and parental consanguinity was denied. On the other hand, while appearing to be transmitted as a dominant, the reports of many unaffected siblings and the paucity of pedigrees extending through many generations suggested that the defect depended upon the presence of two genes, both autosomal, neither of which caused abnormality alone. In a more recent review, reporting a family with several involved generations, Jackson and Lawler concluded that a single dominant gene could explain the observed distributions. A dominant trait certainly appeared to be involved in the family reported here. The mother and the male sibling had the identical condition; there was no paternal consanguinity.
When this syndrome is fully developed, there is little that can be mistaken for it. The differential diagnosis is less easy when only one or two of the clinical features are present. Thus, in the postnatal period, when leukokeratotic lesions of the oral mucosa developed the diagnosis of “thrush” was made. Cultures at that time, and subsequently, were negative for Candida albicans, and gentian violet did not alter the lesions.

When, at 2 weeks, the nails began to grow and thicken, an onychomycotic process was thought of. Had the nail changes appeared alone, hereditary onychogryphosis might have been considered. This condition is also inherited as a dominant trait, but other symptoms are not usually associated with it.²

The resistant thrush-like oral lesions and the dystrophic nails raised the diagnostic possibility of acrodermatitis enteropathica. This serious childhood disease appears to have a familial tendency, is associated with Monilial infection of skin and mucous membranes and involves severe gastrointestinal disturbances, mental changes, and nail dystrophy secondary to paronychia. Prior to the use of diodomin, very high mortality rates were reported and the disease remains a dangerous one.⁴ Our patient received a brief course of diodoquin but was, clearly, never appropriately ill. With the appearance of bullae she finally showed the full syndrome of pachyonychia congenita.

The thickened finger nails constitute one of the chief complaints of those with the disease. The nails are clumsy and unsightly and represent a functional and cosmetic impairment. Sohrweide³ reported a case in which the nails were 0.7 to 1 cm. in thickness at their free margins. Ordinary measures do not suffice to trim these claws hammer and chisel.
trim these claws. The patient seen by Jadassohn and Lewenbowsky\(^4\) required hammer and chisel for the purpose. In addition, the nails are variably discolored; they were an unpleasant dark yellowish brown in this case.

While nail removal is indicated, it is by no means easy. Simple surgical excision has most often been followed by distorted regrowth of the nails.\(^1\)\(^,\)\(^3\)\(^,\)\(^8\)\(^,\)\(^9\) X-ray of the nail bed with later infection and avulsion of the nails led to some improvement but with continued nail growth in a case reported by Diasio.\(^10\)

The most radical treatment recorded was that by Wright.\(^11\) Initial removal of the nail plates was followed in 2 years by recurrence of the nails. Thereafter, first one, then all the distal phalanges were amputated. A remarkable 20-year follow-up indicated the patient's satisfaction.\(^12\) The mother in our family had had osteomyelitis of all the distal phalanges with sequestration over the course of a year after excision of her nails. It is testimony to the distress the original nail growth caused that she felt the treatment with the year of invalidism and the residual deformities was justified.

The difficulty of removing active nail growing tissue arises from the anatomy
of the nail. The germinal matrix is commonly represented as extending from the distal margin of the lunula back proximally to an area in close relationship to the insertion of the extensor tendon at the base of the terminal phalanx. The remainder of the nail bed is termed the sterile matrix. The distinction is ascribed to Quenu, who emphasized the removal of the former and the lack of need to excise the latter in cases of ingrown nail.

Research by Lewis suggests a more complex situation. The nail is formed of a dorsal, an intermediate, and a ventral component (fig. 10). The dorsal layer extends to the free margin of the nail in the fetal fetus but is of lesser extent in the adult. It arises from the proximal half of the roof of the nail fold and, at times, from up to \( \frac{1}{6} \) of the proximal portion of the floor of the nail fold. The intermediate nail is the major part of the nail. In addition to its growth from the nail fold floor extending from the lunula proximally, it may also arise from up to \( \frac{1}{6} \) of the nail fold roof. Growth also occurs from the lateral walls of the nail fold. The ventral component arises from epidermal ridges running longitudinally along the entire nail bed distal to the lunula. Although not contributing to forward growth of the nail, it does add to the thickness.

Surgical appreciation of these several areas of nail growth is increasing. Although we removed the epithelium on the roof of the nail fold, the distal portion of the sterile matrix was left intact with the thought that it would give more stability to the finger tip and would be responsible for no significant nail growth. This proved incorrect. Considerable keratin formed on the residual matrix and was of the peculiarly tough and resistant nature of the previous nails.

The degree of vertical growth in the nails of similar patients led Wright and Guequiere to hypothesize that nail growth was away from the nail matrix rather than from the nail "root." Our experience did not confirm this. The forward growth of the nail matrix has been a recent and active participatory nursing feature. Total or subtotal nail matrix recommended in these cases.

The problem of impaired growth in this patient also led to the development of breakdors (keratoses), which ulcerated areas have not healed. Hypnosis have been useful.

Several other noteworthy cases in Ichthyosis hystrix have been noted. The lateral elevations in the axilla and lateral distribution of several cases of lateralis. Treatment was by skin graft. Axillary flaps, as well as flap repair, have been performed.

Tylosis palmaris and plantaris, thickening of the palms and soles, is a disorder as a dominantly inherited trait, and subsequent function limited in split-thickness skin grafts. Full-thickness skin grafts are required. Other apparent keratoses are permittted keratoses. Striata and papules of its clear-cut patterns, keratoses of symmetrical \( \frac{1}{6} \) of the plantar surface and \( \frac{1}{6} \) of Pata. It is a tissue replaceable keratoses.

A child with \( \frac{1}{6} \) of the nails excised was noted. These problems involve impaired in pat

![Fig. 10. Nail divisions and sites of nail growth](image-url)
growth of the nail was arrested by the excision of the nail fold. However, the active participation of the “sterile” matrix in forming nail thickness was a striking feature. Total removal of the sterile as well as the germinal matrix is therefore recommended in pachyonychia.

The problem of the bullae on weight bearing surfaces has not yet become acute in this patient although her mother has had three hospital admissions for treatment of breakdown areas on the feet. Healing has occurred with bed rest. Such ulcerated areas have been successfully skin grafted. Rubber sole moulds and hypnosis have been reported to alleviate symptoms.

Several other congenital dyskeratoses have received surgical attention. Ichthyosis hystrix and tylosis palmaris et plantaris, with its subvarieties, are the noteworthy examples. Ichthyosis hystrix is characterized by hypertrophic papillary elevations involving large areas of the body. Sometimes occurring in unilateral distribution or in more limited areas, it is also known as nevus unius lateris. Treatment by shaving or deep abrasion had been as successful as replacement by skin grafts. Kazanjian reported a case in which all of these methods as well as flap rotations had been employed to remove the condition from the axillary and perineal areas.

Tylosis palmaris et plantaris is characterized by symmetrically distributed thickening of the horny layer of the epidermis of the palms and soles. It is transmitted as a dominant. Cracks occur in the epithelial surface of the hands and feet, and subsequent low grade infection leads to chronic inflammation with function limited by pain. Excision of affected plantar skin and replacement with split-thickness skin grafts have been successful in alleviating the condition. Full-thickness skin graft replacement of the palms has also been reported. Other apparent varieties of tylosis include mal de meleda (a recessively transmitted dyskeratosis), symmetrical keratoderma, and keratosis palmoplantaris striata and papulosa. The last two are similar in appearance to tylosis but lack its clear-cut pattern of inheritance. A case of keratoderma plantaris treated by excision and split-thickness skin graft was reported by Wynn-Williams. Cases of symmetrical palmoplantar keratoderma have been treated by replacement of the plantar surfaces by abdominal tubes and cross leg pedicle flaps by Pupo and Farina. It is clear, therefore, that all of the major forms of skin and soft tissue replacement have been successfully used in the treatment of these dyskeratoses.

SUMMARY

A child with pachyonychia congenita was presented in whom bilateral finger-nail excision was carried out. The signs and symptoms of this hereditary dyskeratosis were pointed out, and conditions to be considered in differential diagnosis were mentioned. The indications for nail removal, the method employed, and the problems involved were discussed. A brief review of other dyskeratoses whose symptoms have proved amenable to plastic surgical techniques was given. From these reports and from our experience, it would appear that healing as such is not impaired in patients with these congenital dyskeratoses. Skin grafts appear to
take in the usual way. Excision of involved areas and skin graft or skin flap replacement is therefore feasible.

Dr. Cosman
150 East Washington Avenue
New York 22, New York

REFERENCES


MALIGNANT TUMORS

Submaxillary space neoplasms are of general respects and are called "mixed tumors." They are drawn in regard to their formations on the parotid gland, which results in the following facts:

1. The very fact that there is a lymph node is more frequent than those present in other parts of the body and, perhaps, others that are not present in the parotid gland.

2. The histology of the neoplasms is entirely of serous and mucous tissues, as well as serous and mucous glands.

3. Embryologically, the development of the submaxillary gland is first as branched tubules, and the submaxillary space is the result of the combination of these tubules.

4. On the other hand, the submaxillary space is undoubtedly the result of the combination of these tubules.

5. The incidence of mixed tumors in the submaxillary space is the result of the combination of these tubules.

* From the DeBakey Surgical Research Foundation,