Pachyonychia Congenita Project

15 March 2005

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 The fact that a work is unpublished shall not itself bar a finding of fair use if such finding is made upon consideration of all the above factors.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.

2386 East Heritage Way, Suite B, Salt Lake City, Utah 84109 USA
Phone +1-877-628-7300 • Email—Info@PC-Project.org
www.PC-Project.org

Clinical Notes, New Instruments and Techniques

PACHYONYCHIA CONGENITA

A Review and New Approach to Treatment

J. FRED MULLINS, M.D.
NEVILLE MURRAY, M.D.
and
EDWARD M. SHAPIRO, M.D., Galveston, Texas

One of the most striking and distressing of the congenital dyskeratotic skin lesions is the rare and peculiar condition of pachyonychia congenita. It is distressing, not only to the patient because of the severe disability involved, but also to the physician because he faces a difficult problem in rehabilitating the persons who are unfortunate enough to have this condition. The scope of this paper includes (1) a review of the various therapeutic modalities in pachyonychia congenita, (2) the presentation of a case, and the use of hypnosis in the treatment. The selection of this method of therapy was stimulated by the report of Mason in which suggestive therapy was of great benefit in a case of congenital ichthyosiform erythroderma of Brocq.

Pachyonychia congenita is a type of dyskeratosis with lesions involving chiefly the nails and occasionally the skin and mucous membranes. Transmission is probably by a single dominant gene (Jackson, 1951) although it has been reported more often in males, especially in those of Jewish extraction.

The original and classic report of this entity has been attributed to Jadassohn and Lewandowsky and was presented by the latter before the Ninth Congress of the German Dermatological Association in 1907. Many of the cases cited in the literature bear a striking resemblance to this case, in which a 15-year-old girl had nail changes from birth, i. e., the nail plates were greatly thickened and so hard they could be cut only with difficulty. The free borders were also greatly thickened and showed grayish-white streaks while the tips were darkened in color. In addition to the nail changes, she had many callosities of the soles which were associated with large painful bullae during the hot summer months. There was an associated hyperhidrosis of the palms, soles, and the nose. Bright red papules with central follicular plugs were found on her elbows, knees, chin, and on the tip of the nose. The tongue was coated with irregular leucokeratotic plaques and had whitish streaks on the borders and the ventral surface. A younger brother presented only the reddish papules with follicular plugging over the knees and elbows.

A review of the cases reported in the English language reveals that the treatment of pachyonychia congenita has been mostly unsatisfactory or merely temporarily palliative with only a few notable exceptions. Garb reports good results obtained in an 18-year-old boy by using a specially designed rubber base foot mold. This method also helped the mother, whose lesions had recurred after full-thickness skin grafts. The lesions in these cases, however, were confined to the soles. Vitamin A has been said to have a beneficial effect on the dystrophic nail changes in this condition when used in high doses for long periods of time (Porter, 1950); however, this was not of benefit to the hyperkeratotic lesions of the soles. Andrews has reported benefit from the surgical removal of the nails, followed by the scraping of the matrix to prevent regrowth, whereas Wright and Guequierre reported the removal of the distal phalanges with no resulting disability. Wright and Guequierre also reported the healing of painful broken bullae with wet dressings of buffered cystine hydrochloride. When the condition did recur, as it frequently did, the therapy was again instituted, and the lesions healed.

REPORT OF A CASE

J. L. N., an 11-year-old white boy, was normal at birth and remained so until one year of age, at which time a thickening of the skin on the palms and soles was noted. One year later his fingernails started to thicken and elevate from the matrix. At this time he began to develop large bullae on his palms and soles, the rupture of which would lead to the formation of a thick layer of hyperkeratotic epidermis.

By the time of admission, the condition was so painful that he could not walk without the use of

From the Department of Dermatology (Dr. J. Fred Mullins, Director), The University of Texas Medical Branch, Galveston.

crutches. While at home he would crawl about on his hands and knees in order to avoid pressure to his exquisitely tender soles.

Physical examination revealed a 58 in. (147 cm.), 96 lb. (43.5 kg.), slightly hypersthenic white boy, who did not appear acutely or chronically ill. The nails of his hands and feet were elevated from the matrix approximately 0.5 cm. by subungual, keratogenous material. There was a tendency toward a medial deviation of the great toenails and a lateral deviation of the nails on the other toes. The nails were extremely hard and could be pared only with bone-cutting instruments. The entire plantar surfaces were covered with a yellowish hyperkeratotic material, which in some places reached a thickness of 2.5 cm. The palmar surfaces were covered with a similar material reaching 1 cm. in thickness with linear extensions to the distal phalanges of each finger. There were erythematous papules with central horny plugs on the elbows and knees, and several callosities were noted on the extensor surface of his forearms. A few whitish striae ran vertically on the anterolateral aspect of the tongue; however, the mucous membranes were free of any leucokeratoses. The hair was normal in distribution and texture, and the remainder of his integument was smooth and free of abnormalities.

The patient had previously been treated with numerous topical and systemic agents, including fungicides and 10 x-ray treatments of unknown dosage to each foot. Along with these measures, he had received 50,000 units of vitamin A daily for several years, but he had received no benefit from this therapy.

Cases and Treatment of Pachyonychia Congenita Reported in English Language

Patient	Year	Author	Treatment	Results
5½-yr.-old Jewish boy	1929	Andrews, G. C. & Strumwasser	Cutting of nails	Not reported
3-yr. 9 mo.-old boy	1934	Fox, H.	Nails surgically removed and matrix scraped; ultraviolet light to follicular keratoses; drainage of bullae; dressings of methylrosaniline chloride and boric acid	Improved
9-yr.-old boy	1934	Diasio, E. A.	Thyroid extract; bland cream; weak sulfur ointment	Unimproved
8-yr.-old Jewish boy	1935	Solweida, A. W.	Not reported	Not reported
20-yr.-old Jewish woman	1936	Tauber, E. B.	X-ray	Not reported
9-yr.-old boy	1937	Grace, A. W.	Ammoniated mercury; cystine	Not reported
14-yr.-old boy	1937	Felden, B. F.-W.	Removal of toenails	Unimproved
20-yr.-old woman	1937	Goldberg, L. C.	Buffered cysteine hydrochloride	Improved
12-yr.-old white boy	1943	Wright, C. S.	Methylrosaniline chloride; choroazodin (Azochloramid) compresses; $KMnO_4$ soaks; 5% ammoniated mercury; soaking in acetone and paring nails with scalpel	Unimproved
6-yr.-old white boy	1947	Wright, C. S.	Wet dressings of buffered cysteine hydrochloride; removal of nail plates; removal of distal phalanges	Unimproved
5-mo.-old girl	1947	Wright, C. S.	Massive doses of vitamin A; mild emolient	Not reported
9-yr.-old girl	1950	Porter, A. D.	Vitamin B, 1 mg. t. d. s.; vitamin A, to 75,000 U. per day	Slight improvement
18-yr.-old boy	1950	Garb, J.	Thyroid extract; vitamin A, 25,000 U. per day; Vitamin E; 40% salicylic acid plaster; podophyllum resin 20% in castor oil and oxycholesterol-petrolatum ointment (Aquaphor) triolein ozonide in olive oil; rubber base foot mold	Improved
Mother of above patient	1950	Garb, J.	Full-thickness skin graft; aluminum chloride 25%; rubber base foot mold	Improved

On admission to John Sealy Hospital, July 16, 1953, he was unable to walk and moved about the ward in a wheel chair. He was treated with daily soaks and petrolatum, which brought about moderate maceration. Some of the hyperkeratosis could then be removed with scissors, scalpel, and, later, with sandpaper. When removal was faster than regrowth, painful bullae arose. A very marked bromidrosis was present, which was probably due to hyperhidrosis and degeneration of keratin. On Aug. 14, it was decided to try hypnotherapy, inasmuch as the aforementioned related congenital dyskeratotic disease, congenital ichthyosiform erythroderma, had responded favorably to such treatment.

All other local and systemic therapy was discontinued, and the patient was seen by one of us

emainder of his integument was abnormalities.

previously been treated with and systemic agents, including x-ray treatments of unknown . Along with these measures, he) units of vitamin A daily for ne had received no benefit from

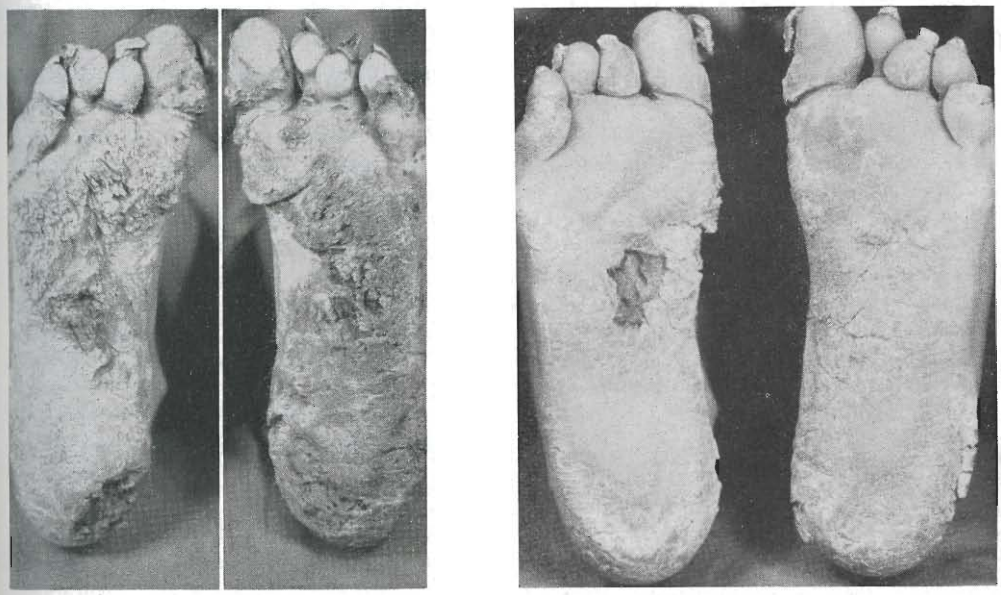
n English Language

	Results
	Not reported
rix scraped; toses; drain- hylrosaniline	Improved
weak sulfur	Unimproved
	Not reported
	Not reported
	Not reported
	Unimproved
	Improved
zodin (Azo- soaks; 5% acetone and	Unimproved
eine hydro- removal of	Unimproved
molient	Not reported
to 75,000 U.	Slight improve- ment
U. per day; aster; podo- l and oxy- (Aquaphor) r base foot	Improved
um chloride	Improved

John Sealy Hospital, July 16, to walk and moved about the air. He was treated with daily m, which brought about mod- me of the hyperkeratosis could th scissors, scalpel, and, later, en removal was faster than re- ae arose. A very marked bro- t, which was probably due to generation of keratin. On Aug. try hypnotherapy, inasmuch as related congenital dyskeratotic hthyosiform erythroderma, had to such treatment.

d systemic therapy was discon- tient was seen by one of us

(N. M.) at one- to three-day intervals during the remainder of his hospital stay. During these sessions an attempt was made to investigate the patient's personality structure and to induce abreactive phenomena by means of hypnotic procedures. The patient was at the same time reassured, as well as encouraged to express and act out various aggressive and destructive traits which had been noted as being present by prior psychometric examinations. Furthermore, posthypnotic suggestions were made to the effect that clearing of the lesions would take place, at first, in the left hand. Within three days there was noticeable softening of the keratotic material on the left hand and considerable subjective improvement. By Aug. 24, the 10th day of treatment, he was able to tolerate a pair of leather shoes. Aug. 27, he stood on his feet without pain for the first time in his life that he could remember, and by Aug. 31 he was able to walk the length of the ward without pain. He was discharged on Sept. 8 to attend school, and psychological therapy was continued at biweekly intervals. After two months this therapy was discontinued. When he was last seen five months later, he was walking with only slight impairment.



J. L. N., before and after hypnotherapy.

COMMENT

The concept that there may be psychic influences over the condition of the skin is not surprising or new. Sydenham (1681) wrote of "hysteric diseases," in which he described angioneurotic edema affecting one leg. Many attempts have been made more recently to demonstrate a causal relationship between psychic stresses and skin disorders. Becker and Obermayer (1947) list many diseases which they feel have a more or less psychic component, and recently Hall-Smith and Norton report significant psychiatric factors in 78.57% of dermatologic patients selected at random. In view of the fact that marked benefit resulted from suggestive hypnotherapy in this case of pachyonychia congenita and in one instance of Brocq's ichthyosiform erythroderma, perhaps this form of treatment should be tried in other congenital dyskeratoses when more conservative treatment has been of no avail.

SUMMARY

The treatment of pachyonychia congenita as reported in the English language is reviewed, and a case of pachyonychia congenita in a 13-year-old boy is reported. This case was treated by suggestive therapy with improvement after failure with the more conservative methods of therapy.

BIBLIOGRAPHY

Andrews, G. C.: Pachyonychia Congenita, in Society Transactions, Arch. Dermat. & Syph. **32**:501-502, 1935.

——and Strumwasser, S.: Pachyonychia Congenita, New York J. Med. **29**:747, 1929.

Becker, S. W., and Obermayer, M. E.: Modern Dermatology and Syphilology, Ed. 2, Philadelphia, J. B. Lippincott Company, 1947.

Diasio, F. A.: Pachyonychia Congenita Jadassohn, Arch. Dermat. & Syph. **30**:218, 1934.

Felden, B. F.-W.: Pachyonychia Congenita, in Society Transactions, Arch. Dermat. & Syph. **37**:1078, 1937.

Garb, J.: Pachyonchia Congenita, Arch. Dermat. & Syph. **62**:117-124, 1950.

Goldberg, L. C.: Resistant Erosive Lesions in Pachyonychia Congenita of Jadassohn, Arch. Dermat. & Syph. **36**:331-334, 1937.

Grace, A. W.: Pachyonychia Congenita, in Society Transactions, Arch. Dermat. & Syph. **36**:1255-1256, 1937.

Hall-Smith, S. P., and Norton, A.: Psychiatric Survey of a Random Sample of Skin Out-Patients, Brit. M. J. **2**:417-421, 1952.

Jackson, A. D. M., and Lawler, S. D.: Pachyonychia Congenita: Report of 6 Cases in One Family, with Note on Linkage Data, Ann. Eugenics **16**:142-146, 1951.

Jadassohn, J., and Lewandowsky, F.: Keratosis Disseminata Circumscripta Tylomata and Keratosis Linguae, Ikonographia Dermatologia I 29, 1910.

Mason, A. A.: A Case of Congenital Ichthyosiform Erythroderma of Brocq: Treated by Hypno Brit. M. J. **2**:422-423, 1952.

Porter, A. D., and Haber, H.: Vitamin A in C of Acquired Localized Keratosis Palmaris Plantaris and One of Acquired Pachyonych Brit. J. Dermat. & Syph. **62**:355-358, 1950.

Tauber, E. B.; Goldman, L., and Claassen, H. Pachyonychia Congenita, J. A. M. A. **107**: 30, 1936.

Wright, C. S.: Congenital Ectodermal Dysplasia Pachyonychia Congenita in Society Transactions, Arch. Dermat. & Syph. **47**:435, 1943

Wright, C. S., and Guequierre, J. P.: Pachyonych Congenita, Arch. Dermat. & Syph. **55**:819-82 1947.