Pachyonychia Congenita Project

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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.

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PACHYONYCHIA CONGENITA

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Jadassohn and Lewandowski in *Ikonographia Dermatologica,* Fasc. 1, 1906 page 28, tab. VI, Figs. 8, 9. described a patient, girl, 15 years of age under the title, pachyonychia congenita; keratosis disseminata circumscripta (follicularis); tylomata; leukokeratosis linguae. Individual case reports and reviews of the literature have been made in the past forty years. The clinical symptoms are unmistakable and the history almost repetitious. The mystery of the etiology remains. Therapy has seemed unavailing. The relation of the condition of the nails to the definite pathology of the skin has not been clarified. The possibility of failure of hormonal output has been suggested. The relationship to lack of vitamin had not been previously cited in the case reports studied. The appearance of the patient offered the opportunity of consultation with the director of the New York City Nutrition Institute, and Dr. Norman Jolliffe after blood studies reported all findings normal except for ascorbic acid. The findings here were 0.3 mgms percent with normal given as .6 mgms percent.

It was possible also to secure abstracts from two hospitals where the patient had been under observation when much younger. The case presentation of Dr. Cipollaro is on this patient. For ten years, the patient was under intermittent observation. In those ten years, his nail condition was recognized. Despite diagnosis, treatment was unavailing.

Joseph P. American born youth of Italian parentage now 17 years of age, was the youngest of four children He knows many cousins and other relatives. He is the only one with the skin and nail disturbance to be described.

The nails were noted to be abnormal at birth. They were thick, with rounded upper surface, and extended beyond the tip of the fingers and toes. All the nails were lost when the infant was 40 days old. The new nails grew thicker, harder anl longer than normal nails. A physician consulted said there would be great difficulty with them later in life. The child always had repeated loss of nails. Each new growth of nail was more stubby and thicker than the previous. From about the age of two to about ten or eleven years, inflammation about the nails caused repeated loss of the nails. The nails always regrew with the same abnormalities. It was noted the nails of the two fifth fingers most closely approximated normal nails. They were the thinnest. The normal activities of the boy and youth have never been curtailed by the presence of the abnormal nails. He can write, work with tools and draw. Only in infancy and early childhood has it been possible to cut or trim the nails with scissor or clippers. A contrivance of grinding wheel on shaft of a hobby motor has been utilized to trim the nails for many years. From two to four years of age the patient had kidney trouble and diseases of childhood. The boy has noted the presence of chicken skin or permanent goose pimples, on the extremities. Wart-like projections formed on the tips of the elbows and less prominent warty-like ones on the knees.

When the patient was seven or eight years of age, he developed new skin troubles. After wearing a pair of rubber sneakers, he noted a larger blister on one foot. He leaned on the other foot to balance himself and blisters formed on both feet. He has never been without blisters since. About eighteen months ago, when fifteen and one-half years old, he noticed pimples on his face with enlarged pores. Fever blisters appear from time to time at the corners of the lips. When presented before the Atlantic Dermatologic Conference by Dr. Anthony C. Cipollaro, March 9, 1940, changes were noted in the oral mucous membranes. The inner surfaces of the lips and cheeks and on the dorsum and edges of the tongue, dull whitish gray patches, irregular in shape and slightly elevated, were described. The patient mentioned these in the historical survey of his condition as present since four years of age following very severe whooping cough. The mother insists patient was 'nearly dead' and survived by means of strong medicine, chiropractic treatment and inhalation from steam kettle. The mother believes strong medicine given caused the white wasting and mushroom of the tongue and cheeks and perianal

regions. The mother gave Keppler's cod liver oil regularly—says more than 200 bottles.

Appearance of the skin—March, 1945:

The extensor surface of upper arms and forearms present non-inflammatory perifollicular hyperkeratosis. Normal skin exists between the projections. The hyperkeratotic lesions cannot be removed by expression but can be rubbed off with firm agitation. The projections are highest on the tips of each elbow, and only slightly less marked over the knee caps. On the extensor surfaces of the legs, the hair projects from the keratotic lesion.

An erosion is present below the left external malleolus. It measures 2 x 2 cm. The edges are limited by soggy masses of corneum. On the dorsum of the left foot is an erosion 6 x 2 cm. with thin overhanging epidermis at the periphery. Another erosion, 3 x 3 cm. exists on the inner side of the foot below the malleolus. The soggy corneum extends from its lower border to the sole. An erosion, 3 x 3 cm. was present below the external malleolus of the right foot. The callus-like periphery extended to the sole and to the Achilles tendon. A soggy adherent mass of corneum was preesnt on the inner malleolus of the right foot. It was impossible to remove the keratin. The sole of the right foot at the arch showed an erosion, 3 x 4 cm. with irregularly thick soggy keratin at the periphery. Erosions were present between the first and second toes of each foot, and between the second and middle toe of each foot with extensions to the contiguous skin of the sole.

The surface of the tongue was white, and thick with definite linear thickening at the periphery of the upper surface and the sides of the tongue. The sides and the under surface of the tongue were pink, smooth and moist. At the mid-point of the tongue extending to each side of the median raphe was an erosion, or smooth section about 1 x 1 cm. in area. The other papillae seemed exaggerated in height, but forcible agitation did not remove any of them. The inner surface of each cheek evidences white irregular patches along the line of teeth closure. In the discussion of Dr. Cipollaro's presentation, Dr. A. B. Cannon suggested the resemblance of the mucous membrane lesions to his "white sponge nevus of the mucosa."

The skin of the buttocks was very keratotic. The perianal mucosa showed whitish raised lesions individually and conglomerate but slightly different from the mucous membrane lesions of the surface of the tongue and the inner surfaces of the cheeks.

A hospital in Brooklyn where the patient had been under observation in 1936 wrote as follows:

Joseph No. 150262 9½ years. Admitted: 11/20/36. Discharged: 12/23/36.

Chief complaint: Skin eruption for past 7 years.

Family history: No family history of disease similar to patient's.

Past history: Birth, feeding and development normal.

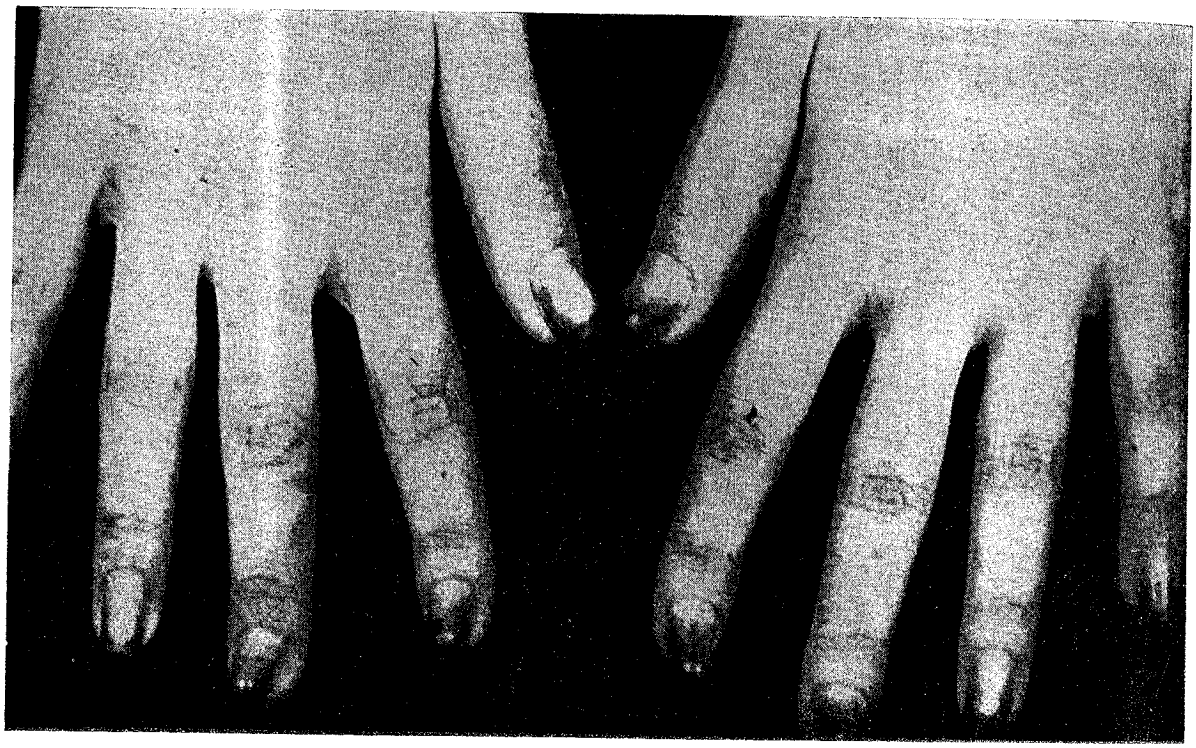

FIG. 1.
Pachyonychia

The child has been vaccinated and given toxoid. At 7 months of age the patient had a first attack of a urinary infection and was unable to void for 3 days. On subsequent visits to a hospital in Brooklyn the mother was told that the trouble was still active over a period of 4 years. At 14 months he had diphtheria, followed by pneumonia, and the patient has been subject to bronchitis since. At two years the patient had measles, pertussis at 3½ years. Since the fourth year the patient has been in fairly good health, generally.

Present illness: In infancy nails were noted to be very small; when they first grew out they were thickened and repeatedly fell off. Since the age of two years the patient has had lesions on his skin. These began from measles rash and spread over his body with breakdown of large areas of skin. Since that time new lesions have occurred, beginning with discrete yellow pimples occurring singly, then in crops with coalescence to form an open sore which drains pus. The lesions are made worse by cold weather and seem to have some relation to fatty foods and sweets. They are irritated by clothing. Various ointments have been used on the lesions, but clearing is

thought to have been spontanous. They have occurred particularly on the back, flanks, hands and feet. With the thickened nails he has had frequent infections of the nail beds. Last winter the patient had the worst period in several years, and had raw areas over both feet, right thumb and right knee when admitted to a hospital in Brooklyn. Last summer while in the hospital, the beds of several nails were curetted under anesthesia and the other nails trimmed with bone shears. He had been discharged from St. John's Hospital one week before admission here.

Physical examination: A fairly well developed, poorly nourished white male, appearing chronically ill. No toxicity or respiratory distress. There is moderate discomfort from lesions on the right foot. Intelligence normal. There were several carious teeth. No general glandular enlargement.

Skin—(1) A crusted ulceration surrounds both nostrils and part of the upper lip. (2) The skin of the trunk and extremities is thickened, with increased pigmentation, particularly in the lumbo-sacral region and on the outer surfaces of the arms and legs. (3) Scalp, face and neck normal in texture. Hair follicles and sebaceous glands show hard papular lesions of varying size, some having the appearance of large comedones. (4) There are isolated areas of scaly hypertrophy on the thighs and elbows. (5) In the gluteal fold there is a patch of papilliform structures, raised on end 1 to 2 mm. above the surface of surrounding skin. (6) The dorsum of the right foot and the area between the toes are covered by a granulating, healing raw area. (7) All of the nails are corrugated longitudinally, and thickened to 1 to 5 mm. (8) Buccal mucosa is pale, with a thick white coating sharply limited to the upper surface of the tongue and the area above the line of the teeth.

The remainder of the physical examination is negative.

Laboratory data: Schick, Kline and Mantoux—negative.

Blood count—Hemoglobin 12.6 gm. RBC 4.06 million. WBC 5,100; Mature polys 16, immature polys 4, lymphocytes 68, monocytes 2, eosinophiles 10%.

Urine—negative except for trace of sugar on one occasion, and 2 to 3 WBC on a subsequent occasion.

Blood chemistry—Urea N 13. Cholesterol 275 on 12/1 and 305 12/13.

Basal metabolic rate—+3.

Stool culture—B. coli. Negative for typhoid and dysentery.

Culture from mouth—Hem. staph. aureus, micrococcus catarrhalis; negative for fungi.

Scrapings from foot—negative for fungi.

Culture from foot—Staph. aureus and diphtheroid bacilli; negative for fungi.

Course in the hospital: The patient was seen by Dr. Grace, Dermatologist, who was of the opinion that the picture fitted in with descriptions of keratosis follicularis (Darier's disease). The unusual feature was an absence of lesions from the scalp, palms and soles. It was thought that no therapy of any value could be provided beyond local care of the broken down areas on the skin of the foot. Whitfield's ointment 1/4 strength was applied to this, and ammoniated mercury was used on the nose and upper lip with improvement of both sites. Before discharge boric ointment was used on the foot. The use of x-ray therapy was thought inadvisable by Dr. Levine and Dr. Schloss. An effort was made to determine the patient's reaction to physostigmine, which has been found helpful in scleroderma in cases where the blood pressure and the pulse show a rise rather than a fall. There was no consistent change noted in these observations. At the time of discharge the patient's right foot was healed except between the first and second toes. Otherwise his condition was unchanged.

Dr. Levine was of the opinion that this was not a true case of Darier's disease, but represented congenital anomaly of the skin and nails.

As of 3-30-45, the child has not been see here since discharge from the hospital.

A second hospital sent the folowing case report:

The above named patient registered in our skin department on 7-12-37 at which time a diagnosis of Darier's disease (?) Pachyonchia congenitalis (?) was made. Local medication was prescribed.

Patient, Joseph P.—On February 7, 1940 the patient returned to clinic after a lapse of 3 years and a diagnosis of "dermatitis" was made. The patient made routine visits to clinic through August 23, 1940. During this time the patient received 6 Alpine treatments to the toes, and seven, 1/4 skin units of unfiltered x-ray to the outer lateral of right foot, dorsum of all toes. Biopsy done on 3-4-40 reported: "Eczema with contact dermatitis."

On January 6th, 1943, the patient returned to clinic with an eruption on the right ankle. Local medication was given. Another visit was made on March 29, 1943. Patient has not been seen in clinic since that time.

Resumé: A youth of 17 was observed for some months in whom the clinical diagnosis of pachyonychia congenita was made. The condition had been recognized for years. The skin eruption of the youth was one found associated with pachyonychia congenita, and had features of both keratosis follicularis and epidermolysis bullosa. Studies for vitamin content of the blood indicated subnormal ascorbic acid content, with other findings normal. Treatment was of no help. The patient was the same as discussed in a case presentation of Dr. Cipollaro.

No solution to the problem of pachyonchia congenita and the associated skin and mucous membrane disorders has been found.

Note: The discrepancy of history given by the mother in 1936 and ten years later should be noted. The 1946 history was taken in stages, repeated, and given to the mother to amend after consultation with other members of the family.

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*POMPHOLYX**

Its etiology, pathogenesis and vaccine therapy, with particular reference to its military importance and post-war significance.

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**Formerly Lt. Colonel, Medical Corps, Army of the United States.

*This paper was ready for publication in October, 1944; however, due to statute of limitation it could not be published during the war. The text has remained substantially unchanged except for amplification based on recent literature and on additional observations made in the Central and Western Pacific, while the author was stationed at Oahu, Territory of Hawaii and on Okinawa, the Ryukyus, Japan with the 233rd General Hospital.