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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*

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In 1906, Jadassohn and Lewandowski for the first time reported in the iconographia dermatologica a peculiar anomaly of the finger and toe nails which they termed pachyonychia congenita. The patient was a girl of fifteen and showed, in addition to the changes in the nails, other abnormalities of the skin and mucous membranes. These changes in many respects resembled those found in keratosis follicularis of Darier, from which disease pachyonychia congenita was distinguished by the authors on account of the peculiar lesions of the nails and tongue. For the past year a case which is almost identical to the one described by Jadassohn and Lewandowski has been under our care, and this similarity, as well as the apparent rarity of the condition, has led us to a careful study and report of the case.

Following is a description of the condition as originally given by Jadassohn and Lewandowski:

History.—E. C., fifteen year old girl, first seen June 15, 1905, presented a cornification anomaly of the skin and nails. The nail condition had been present since birth. The duration of the condition of the skin is not known. The parents and seven other sisters were free of this condition, but one brother showed abnormal changes, which will be described below.

Examination.—The nail plates of all the fingers and toes are greatly thickened and so hard that it is impossible to cut them with scissors, the finger having to take recourse to a hammer and chisel to trim them. The finger nails are of normal length and width, becoming thicker at the free borders and measuring three to five millimeters in depth. They are for the most part transparent and pale, with smooth and glistening surface, and at the tips are grayish-black in color. At the free borders here and there appear whitish-gray streaks.

The toe nails are all greatly thickened, especially the large toe nail which resembles a condition of onychogryphosis. The surfaces are not smooth but slow irregular transverse grooves. The consistency is the same as that of the finger nails. The growth of the nails is not especially rapid. The patient is not conscious of any impediment to finer movements of the fingers.

On the face, around the nose and the chin, are bright red, somewhat pointed papules, and on the nose are several vesicles with clear fluid, alkaline in reaction and sterile. There is hyperdrosis of the skin of the nose, palms and soles. On the soles there are many callusities, which through maceration form whitish elevations. In the summer, and seldom in the winter, there appear on these callusities painful large bullae, the contents of which are alkaline in reaction and show no organisms of any kind. On the elbows and knees are definitely grouped pinhead-sized follicular papules. In their centers there is a horny plug which fits into the papule in a crater-like depression. The dorsum of the tongue is covered with thick, adherent, irregular shaped white plaques, sharply defined. At the borders and on the undersurface there are present white streaks and irregular, slightly raised, linear lesions. The mucous membranes are otherwise normal.

About four weeks after the patient was first seen, a definite eruption was noticed, which has persisted and which is present chiefly on the extensors of the arms and legs, on both scapulae, and in the region of the axillae and the gluteal region. The eruption on the elbows and knees is most marked. Most of the lesions are pinhead sized with intensely red bases containing a centrally located horny cone, whitish-gray in color. The skin between these papules appears normal. Removal of one of these horny cones left a slightly bleeding crater-like depression.

The above-mentioned four-year-old brother presents an analogous condition except that there is no involvement of the nails nor leukokeratosis of the tongue.

Histological Findings.—The examination of a lesion containing a horny cone (described above) shows a hyperkeratosis of the follicle surrounded by parakeratotic layers heaped up so as to encroach on the next lesion. There is decided thickening of the rete with marked infiltration of the neighboring rete pegs in the cutis. The rete cells are excessively vacuolated; here and there numerous and large keratin hyaline granules are present extending to the deeper layers. The cutis shows slight edema and a moderate increase in cells.

History.—Our own case is that of a boy five and a half years of age, born in the United States of Russian Jewish parentage. He was the first child and has one other brother two years of age, who is free of this anomaly. The boy was born of a normal labor, and there is no history of miscarriages. The family history on both sides is entirely negative. The condition was first noticed two weeks after birth when the nails on the hands grew out like talons and were cut off by the physician. At the age of six months, the nails of the hands and feet began to fall off at intervals of one to two months. This was preceded by an area of redness and bogginess surrounding the nail bed, all the nails being involved at one time or another. At eighteen months of age, the child began to develop lesions on both

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knees which the mother said looked like warts, this condition gradually spreading to the elbows, popliteal spaces, buttocks, legs and ankles.

During his infancy the patient had repeated attacks of vomiting, during which time he would be unable to retain any food for two or three days at a time. Since infancy the child has shown excessive sweating of the palms, soles, and of the face, and at intervals also developed herpetic lesions about the mouth.

Examination.—The boy appears normal in size for his age, and is apparently in very good health. He is somewhat backward mentally and replies to questions in a deep, guttural voice with single words. His hearing and vision are normal; teeth are all carious. The dorsum of the tongue is covered with a general white patchy coating, which is difficult to remove. On close examination, the condition consists of confluent plaques, irregular in shape and slightly elevated. The tonsils are greatly hypertrophied, and the patient presents the facies of an adenoid type of child. The posterior cervical glands, epitrochlear glands and inguinal glands are all palpable.

All the finger and toe nails are abnormal. The finger nails are of normal width but greatly thickened, the thickness increasing towards the free border where they measure about three to four mm. The surfaces are smooth and show diminution of lustre, being brownish gray in color. The free borders are grayish-black in color and show vertical striations. The nail plates are extremely hard and firmly attached to the nail-beds. Some of the nails show transverse grooves. The nails grow straight out and show no tendency towards upward or downward curvature. Patient is not conscious of any hindrance in the performance of fine movements.

Occasionally one of the nails becomes acutely inflamed, being surrounded by a boginess, which, after a period of one to two weeks, falls off. The nail-plate thus seen shows abnormal depth, increasing towards the free border, the underlying nail-bed being covered with a purulent, foul smelling exudate. The new nail then begins to grow as any other nail would.

Over the extensor surfaces of the forearms, elbows, knees, popliteal spaces and the anterior aspects of the ankles, buttocks and lumbar regions, there are follicular lesions varying in size from a pinhead to a pea. These are grayish-black keratotic papules, having in their centers horny cones which fit into corresponding crater-like depressions. The removal of these central cones is fairly easy, leaving a slightly bleeding cavity. The eruption on the outer aspects of the upper and lower extremities is also follicular, resembling keratosis pilaris. This latter condition is not constant and disappears spontaneously only to recur. The intervening skin between lesions is apparently normal.

At irregular intervals, usually after a history of undue exposure to rainy or snowy weather, the patient develops a series of yellowish waxy bullae. These occur on the planter surfaces of the toes, on the heels and along the borders of the feet. These bullae are painful; puncture reveals a clear watery fluid which is alkaline to litmus. At the present time the patient shows an oval bulla on each heel about three inches in length.

Histological Findings.—The striking feature of the specimen is a thickening of the epidermis due to acanthosis and parakeratosis especially pronounced about the pilosebaceous follicles. The rete pegs are lengthened and about the follicles the epidermal thickening causes funnel-shaped prolongations extending into the corium. The openings of the follicles are dilated and plugged with imperfectly cornified and somewhat degenerated horny material, and horny plugs are also present in the sweat pores. The basal-cell layer is irregular and the cells are swollen. There is granular degeneration in the prickle cells and the nuclei are deeply staining and some are crescentic, being pushed to one side of the cell by hydrops. They resemble the "corps ronds" found in Darier's disease. The papillary bodies between the rete pegs are elongated and in places the apices come near the surface of the skin. The blood vessels of the corium are dilated and surrounded by lymphocytes, mast-cells, connective-tissue cells and an occasional plasma-cell—
the appearance suggesting a mild inflammation from the pressure or irritation of the overlying thickened epidermis. The connective tissue seems to be normal.

In the course of a talk with Doctor Howard Fox, after we decided to present our case, he told us about a child with a similar eruption in his care, and strangely his case is almost a complete counterpart of the two just given. We are indebted to him for the privilege of including the following description of his case:

**History.**—S. W., three years, nine months of age, born in the United States of American parents, presented a peculiar anomaly of cornification affecting the nails and cutaneous surfaces. The lesions on the nails had been first noticed two weeks after birth, those on the skin, when the child was two years of age. At the age of one year, he was said to have suffered from an enlarged thymus gland, which had been interfered with and which had been treated by X-ray. He had suffered from diphtheria shortly before the appearance of the cutaneous lesions. The patient is the only child of young, healthy parents. No similar disease is present in any of his relatives, including twelve first cousins on the maternal and four on the paternal side.

**Examination.**—Examination shows the patient to be a well-developed, mentally active, fine-looking child, weighing 42 pounds. The eyes are blue and the hair blond. All of the finger and toe nails are abnormal. The finger nails are of normal length but greatly thickened, the depth increasing towards the free borders. The surface is smooth, presenting no pits, ridges or furrows. There is some diminution of lustre. The nails are grayish-brown in color, being increasingly discolored towards the distal ends, where there are some whitish streaks on the nails of the left hand. The nail-plates are extremely hard and firmly attached to the nail-beds. There are no paronychia and no abnormality on the skin of the fingers. There is no hindrance to the performance of fine movements. The rate of growth of the nails is normal and the direction straight. The toe nails show changes similar to those of the fingers with the exception that the growth, if uncut, is in a curved direction (onychogryphosis). No subjective symptoms are caused by the changes in either finger or toe nails. The scalp is normal. There is no hyperdrosis of the face and nothing to suggest granulosis rubra nasi. At the right angle of the mouth is a crusty area, apparently a simple herpes. The mother states that the patient often has cold sores in this location which last several weeks. The tongue is coated habitually but there is nothing to suggest leukokeratosis. The teeth are in good condition and show no abnormalities. On the extensor surfaces of the forearm there are pinpoint to small pinhead kerasites. Over each olecranon there is an irregularly shaped, warty mass, the size of a bean, surrounded by small pinhead warty, spinous lesions. On the lumbar and scaral regions, fairly numerous, grouped, small pinhead lesions are located, being rough to the touch and the largest having a red base and filiform projections. The genitalia are apparently normal. The lower extremities are affected chiefly above the knees to a less extent on the anterior aspects of the legs. The buttocks show a similar condition to that of the lumbar region but with more marked filiform spines. The thighs show a fairly profuse eruption of follicular keratoses, from a pinpoint to a pinhead in size, the largest having filiform spines. The skin between all of the above-described warty lesions is smooth and normal in appearance. On the internal aspect of the great toes, metatarsal regions and heel of the right foot there are pea to bean-sized yellowish callousities, under which are painful bullae. On puncture they yield a clear sterile, alkaline fluid.

**Discussion.**—Referring to the original case, Jadassohn and Lewandowski stated that a definite diagnosis was impossible, and that the essential follicular hyperkeratosis brought to their minds Darien's disease and keratosis (contagi-osa) follicularis of Brooke. The plantar and palmar hyperkeratosis was described in Darien's disease with additional localization on the nose and chin but leukokeratosis of the tongue was not described at that time in either affection. The nail changes in their case had never before been noted and the various dermatologists who saw their case were unable to classify it. For these reasons Jadassohn and Lewandowski chose the term pachyonychia congenita because in Brooke's disease as well as in Darien's disease no nail changes were at that time reported. Since their article, the condition of leukokeratosis has been described in Darien's disease and we feel, in view of the clinical and objective findings grossly and pathologically, that pachyonychia congenita is not a clinical entity but together with leukokeratosis is a cornification anomaly which forms a part of Darien's disease.