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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

REPORT OF A CASE

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In 1906 Jadassohn and Lewandowsky described in “Ikonographia dermatologica” a cutaneous rarity that they believed to be an unclassified dermatosis, a disease sui generis to which they gave the name pachyonychia congenita. This original paper had the subtitle “Keratosis Disseminata Circumscripta, Tylomata and Keratosis Linguae.” A review of the earlier literature reveals nothing apparently similar, and though the entity is one with a definite clinical picture it is not well known, probably because it is infrequently mentioned in the standard dermatologic works. Only a few reports of genuine cases of pachyonychia congenita can be found in the American literature. The cases described by Howard Fox in 1928, by Andrews in 1929 and by Diao in 1934 fulfill the clinical requirements of the disease. Perhaps cases are occasionally encountered in every clinic that pass undiagnosed, but in the available American literature these three reports afford the only basis for comparative study with the case reported here.

REPORT OF CASE

On Oct. 16, 1933, J. S., a healthy boy 8 years of age, the son of healthy Jewish parents, was admitted to the Stuyvesant Square Clinic because of thickening of the nails of the hands and feet and an eruption on the feet and ankles. The mother stated that the cutaneous manifestations were not evident until the age of 18 months, at which time there developed bilaterally about the ankles two or three moderate-sized bullae with clear contents, and that from this time the lesions as described were more or less constantly present, with marked exacerbation each summer. Two other children were well in every respect. There were no other irregularities in the patient’s history.

From New York Skin and Cancer Hospital, service of Dr. Binford Throne.
2. Fox, Howard, cited by Andrews and Strumwasser.
## Review of Cases Reported in the Literature

<table>
<thead>
<tr>
<th>Author and Year</th>
<th>Age, Years:</th>
<th>Sex:</th>
<th>Nationality</th>
<th>Duration</th>
<th>Subj. Symptoms</th>
<th>Localization</th>
<th>Color</th>
<th>Clinical Picture</th>
<th>Histologic Picture</th>
<th>Complications</th>
<th>Comment</th>
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</thead>
<tbody>
<tr>
<td>Jackson &amp; Lewandowsky 1906</td>
<td>Since birth</td>
<td>F</td>
<td>German</td>
<td></td>
<td>Hyperhidrosis of palms and soles</td>
<td>Nails, feet, hands, elbows, knees, axillae, and sacral regions</td>
<td>Gray-yellow papules</td>
<td>Marked thickening of all nails; stria- tions of white-slate color in distal third of tongue; oval-sized follicular plugging on knees, sacrum and elbows</td>
<td>Hyperkeratosis of follicles; vacula- tion of rete cells; keratotic hyaline granules extending to deeper layers</td>
<td>Fungating tuberculosis of skin</td>
<td>A brother had similar involve- ment; seven children in family</td>
</tr>
<tr>
<td>Fox 1928</td>
<td>Two weeks after birth</td>
<td>M</td>
<td>American</td>
<td></td>
<td>Bullae were painful</td>
<td>Finger-nails, extensor surface of forearms, lumbar and sacral regions</td>
<td>Red papules with yellow apices</td>
<td>Thickening of finger-nails; oval-shaped papules on red base in sacro- lumbar region and extensor area of arms; bullae on ankles</td>
<td>None-reported</td>
<td>Habitu- ally coated tongue</td>
<td>Tendency to coloboma at right angle of mouth</td>
</tr>
<tr>
<td>Andrews &amp; Strumwasser 1929</td>
<td>Two weeks after birth</td>
<td>M</td>
<td>Jewish</td>
<td></td>
<td>Hyperhidrosis of palms and soles</td>
<td>All finger-nails, toes-nails, elbows, knees, buttocks, and lumbar region</td>
<td>Brown-gray nails, gray-black papules</td>
<td>Thickened toes-nails and finger-nails; follicular keratosis on elbows, knees, buttocks and lumbar regions</td>
<td>Acanthosis and parakeratosis about plucked- ovaries; irregular basal layer; granular degeneration of prickle cells</td>
<td>All teeth carious; tongue coated</td>
<td>Tendency to herpetic lesions about mouth; backward mental elements</td>
</tr>
<tr>
<td>Diaz 1934</td>
<td>Five years of age</td>
<td>M</td>
<td>Italian</td>
<td></td>
<td>Slight deafness; hyperhidrosis of palms and soles; epiphora</td>
<td>Finger-nails, toes-nails; generalized xerotic skin</td>
<td>Grayish-yellow cornification</td>
<td>General xerotic skin; smallness of scalp; thin nails; obstruction of right lateral duct</td>
<td>Thick horny layer; thick prickle cell layer; large lumen of sweat glands; cornification of follicular openings</td>
<td>Coated tongue; slight deafness</td>
<td>Patient treated for syphilis; marked scal- liness of scalp</td>
</tr>
<tr>
<td>Sobrwarda 1934</td>
<td>Since birth (nails); skin lesions 18 months later</td>
<td>M</td>
<td>Jewish</td>
<td></td>
<td>Hyperhidrosis of palms and soles</td>
<td>Bullae about heels, soles; keratosis of legs, arms, forearms; stria- tions of tongue</td>
<td>Yellow-gray nails, reddish follicular keratosis</td>
<td>Follicular keratosis on legs, sacrum, arms, forearms; bullae on ankles; coated tongue</td>
<td>Parakeratosis and acanthosis; follicular plugging and cornification; basal cell layer irregular; 11/9/33</td>
<td>None</td>
<td>Two other children, free from similar trouble</td>
</tr>
</tbody>
</table>
Physical examination revealed that in addition to the "blisters" there were irregular patchy areas of hyperkeratosis cushion-like about both heels and on the plantar surfaces of all the toes. The bullae on the left foot were distributed about the heel, that just below the left inner malleolus being 1 3/4 inches (4.4 cm.) wide and 3 inches (7.6 cm.) long. On the right foot a bulla of similar size was present in the same location, but in addition there was a small straw-colored bulla about 1 3/5 inches (3.7 cm.) long around the distal border of the third and fourth toes. The contents of the bullae were sterile on culture. There was hyperhidrosis of the palms and soles.

Fig. 1.—Photograph showing maceration about the heels, knees and legs. Note the separation of the lesion on the right ankle and the bullet-shaped plugs in relief on the left knee.

A nutmeg-grater-like area of keratosis pilaris was present on the anterolateral aspect of the legs in their central portion, the distinct, horny, bullet-shaped plugs being diffusely scattered. On the knees was a marked area of follicular keratosis, some of the plugs being from 2 to 3 inches (5 to 7.6 cm.) in diameter. Both elbows showed changes identical to that noted on the knees, together with occasional elevated, vesicular papules containing a gray-yellow plug; about the base of some of the papules there was moderate erythema. On the extensor surfaces of the arms and forearms irregular patches of the same type of keratosis were
There were also multiple lesions found on the toes, distributed mainly on the first and fourth (4.4 cm.) toes. The lesion size was variable, ranging from a small red bulla to a large, thickened lesion on the fourth toe. The condition resembled hereditary thickened toe-nails.

Fig. 2.—Photograph showing the lesions on the ankles and thickening of the toe-nails.

Fig. 3.—Photograph showing a characteristic lesion on the knee and a front view of the thickened toe-nails.
present as were seen on the legs, and though distinct they were not as prominent as those on the elbows and knees.

The changes in the nails were striking and characteristic. The free margin was from 7 mm. to 1 cm. in diameter. In all the nails the color was slightly darker than normal, and in some, particularly on the thumb and index fingers, the thickened portion was dark yellow-gray. The lunula was absent in the bases of all the nails, and viewed dorsally rugae were seen running in a longitudinal direction from the nail bed to the free margin. The nails were arched in the center, and the underlyng cuticle was firmly pressed against by the hypertrophied nail to form a small knoblike tab of flesh at the distal margin. There was no impairment of the circulation or of the fine movements of the ends of the fingers.

Fig. 4.—Photograph showing scattered papules about the elbows and thickened finger-nails.

The skin over the nose showed nothing abnormal, in contrast to the case presented by Jadassohn in which occasional follicular plugs were noted diffusely spread over the nose, glabella and lower part of the forehead.

A feature found in all previously reported cases was duplicated in our own. The tongue in the entire posterior two thirds was crossed irregularly by whitish striations limited by the margin of the tongue and heavily coated with a grayish-yellow film. This coating was firmly adherent and could not be removed by lifting or abrasion.

The scalp was clean.

Observations at biopsy agreed essentially with those of previous investigators. Since in none of the available literature was there a report of studies of endocrine function or of the blood in a case of pachyonychia congenita, investigation
was directed along these lines. On October 16 the patient's basal metabolic rate was -21.9, suggesting a possible endocrine deficiency. On October 18, an examination of the blood for the retention of heavy metals, according to the method of C. N. Myers of the Stuyvesant Square Hospital, recorded the following values: arsenic, 0.093 mg. per hundred cubic centimeters; solids, 2.06 mg., and lead, 0.0 mg. Examination of the urine revealed specific gravity, 1.024; solids, 4.75 mg. per hundred cubic centimeters, and arsenic, 0.081 mg. On November 3 examination of the blood three and a half hours after a meal revealed: sodium chloride, 503.3 mg. per hundred cubic centimeters; sugar, 117 mg.; urea nitrogen, 9.1 mg.; uric acid, 2.24 mg., and carbon dioxide, 61.1 cubic centimeters.

![Fig. 5.—Photograph showing keratotic plugging and crusted wet lesion about the elbow and a lateral view of the thickened finger-nails.](image)

![Fig. 6.—Photograph showing the thickened finger-nails.](image)

As revealed by the examinations, arsenic was present in relatively small amounts both in the blood and in the urine. Further experimentation on patients with proved cases of pachyonychia congenita might demonstrate the importance of this observation with reference to the production of general symptoms. The reasons for anomalous growth are many; most frequently it is caused by simple external factors such as pressure and both general and local infection. In pachyonychia congenita, however, heredity is a certain factor, and when sufficient data have been collected the part played by endocrine dysfunction and arsenical retention will be settled.
COMMENT

Since the fact is well known that allergic cutaneous manifestations are the result not of quantity but of individual sensitivity to certain substances, the premise is offered that this extraneous metal may be the reason for part, at least, of the hyperkeratosis so typical of pachyonychia congenita. At any rate, further biochemical study is necessary, for in this field of research may lie the answer to the alleviation of this distressing condition.

SUMMARY

A case of pachyonychia congenita having all the clinical features is described.

Chemical studies, not before reported in such a case, were made on the basis of new observations, and possible interpretations were made. The opinion is advanced that arsenical retention and endocrine dysfunction are possible factors in aggravating, if not causing, this condition.