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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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Pachyonychia congenita is the name originally given in 1907 by Jadassohn and Lewandowsky (1) to this congenital anomaly. Siemens (2) in 1922 reported on this disease as an example of congenital dyskeratosis under the title “Keratosis Follicularis.” Heller (3) (1927) called attention to the close resemblance between pachyonychia congenita reported by Jadassohn and Lewandowsky and the syndrome described by Erich Schafer (4) in 1924 under the name “Congenital Dyskeratosis.” Fox (5) (1928) and Andrews and Strumwasser (6) (1929) reported cases of pachyonychia congenita. In 1934 Diasio (7) reported a case under the name “Pachyonychia Congenita Jadassohn - A variety of Ichthyosis (Pachyonychia Ichthyosiformis) involving chiefly the nails.” In 1935 Sobrweide (8) reported a case and used the term coined by Diasio. In 1936 Taubar, Goldman and Claassen (9) reported a case that combined all the features of pachyonychia congenita. Their case was overlooked for a long time because it was considered as an atypical type of epidermolysis bullosa presenting nail deformities, chiefly because of the prominence of the bullae on the extremities. In 1940 Anderson (10) reported pachyonychia congenita in a mother and her daughter. Cipallaro (11) (1941), Michelson (12) (1941), Wright (13) (1943), Wright and Gross (14) (1943), Wright and Guequierre (15) (1947) and Wise (16) & (17) (1948 and 1949) reported and presented cases of pachyonychia congenita in the different meetings of the American Dermatologic Society. In 1950 Garb (18) reported on the treatment of pachyonychia congenita. In 1961 Alkiewicz and Lepioda (19) reported 2 cases, and discussed the clinical and histological data of this disease. In the same year Jannasch and Wiseman (20) reported a case of this disease under the name “Keratosis multiformis idiopathica (Siemens)”.

Ormsby (21) recognized the title “Pachyonychia Congenita” and summarized the characteristics as follows: dystrophic changes in the nails, palmar and plantar hyperkeratosis anomalies of the hair, leukoplakia, follicular keratoses of the acniform type particularly about the knees and elbows and dyskeratosis of the cornea. Verrucous lesions are described as occurring on the knees, elbows, popliteal regions, buttocks, legs and ankles. Bullae are common and occur chiefly on the plantar surfaces of the feet. Generalized ichthyosis of variable degree may or may not be present. The affection occurs chiefly with the male sex. In all of the reported cases pachyonychia has been present. All of the associated symptoms above mentioned have occurred in whole or in part in the various cases recorded. Therefore, the title of Jadassohn and Lewandowsky's
paper "Keratosis disseminata circumscripta.
Tyfomata and Keratosis Linguae" shows the
essential features of this condition.

Kumer and Loos (22) have studied a family
of 5 generations and found 34 cases. From
their cases and from a survey of the literature,
they believed that pachyonychia congenita can
be divided into 3 classes:

1. Type I: Symmetrical keratoses of
hands and feet with follicular keratoses of
body.

2. Type II: Symmetrical keratoses of
hands and feet, follicular keratoses of body
and leukokeratoses oris; this is the common-
est type (Typus Riehl).

3. Type III: Symmetrical keratoses,
follicular keratoses of the body and corneal
changes.

There are 3 phases of pachyonychia con-
genita that distress the affected person: (1)
the dry hyperkeratotic skin with acneform
follicular keratoses; (2) the bullous and
crusted lesions that appear from time to time,
chiefly on the feet, and (3) the greatly thick-
ened nails.

It is quite possible, however, that many
cases are overlooked, and they may be
diagnosed as a simple congenital deformity of
the nails. Most of the cases reported have
been in children, for the reason that this
condition is a particular form of congenital
abnormality.

Report of a Case

This is the first report on this disease in
the U. A. R. The presented case is a girl
aged 14 years (Figs. 1, 2, 3 and 4) who was
born with deformed nails of fingers and toes.
Other changes on the knees, elbows and soles
occurred soon after birth and they are becom-
ing progressively worse.

Fig. 1

There was no family history of disease of
nails or skin. There was no record of con-
genital disturbances in the family. Parents
are not related. She presents changes of all
nails of the fingers and toes. They are elon-
gated, dry, hard with smooth surface, curved

Fig. 2
and thickened and enlarged resembling onychogryphosis. The thickening of the nails increases towards the free border. She has hard, thick, painful, hyperkeratotic, lemon-coloured circumscribed growths on the soles, heels and toes. She has leukoplakic patches on the buccal mucosa and also on the tongue (leukokeratosis). The hair appears normal. The extensor surfaces of both arms and knees show multiple, tiny, follicular, hyperkeratotic papules.

**Investigation:**

Hb. was 100% (14.5 gms.)

R. B. C. 5,120,000

W. B. C. 8,000 with normal differential count.

Urine was normal.

Total proteins in serum 7.5 gms. per 100 ml. serum with normal A/G ratio.

The serum cholesterol was 161 mg./100ml.

B. M. R. was +20 per cent.

Roentgenographic examination of the gastrointestinal tract was normal.

Scrapings from the skin and nails were negative for fungi in fresh specimen and in culture.


**Histopathology:**

Specimens for biopsy were taken from the skin opposite the right knee from an area that showed hyperkeratosis with keratotic plugging of the follicles. Sections were stained with Hx. and Eosin, others with the elastic tissue stain and on microscopic examination they showed (Figs. 5, 6, 7, 8, 9 & 10):

There was marked thickening of the epidermis due to acanthosis and parakeratosis and this was particularly marked in the horny layer which was composed of loose horny lamellae. The thickening of the prickle cell
layer could be considered relative since the cells were decreased in volume owing to the premature keratinization. The rete pegs were lengthened, and about the follicles the epidermal thickening caused funnel-shaped prolongations extending into the corium. There was cornification of the follicular openings but not of the sweat pores. In some areas the rete pegs were so elongated that the apices of some of them were so near to the surface of the skin. The granular layer was present and pronounced throughout, though less marked in some areas than in others. The basal cell layer was irregular and its cells were swollen. There was granular degenerations in the
The nuclei stained deeply, and some were crescentic, being pushed to one side of the cell. The blood vessels of the corium were dilated and surrounded by lymphocytes, mast cells, connective tissue cells, and an occasional plasma cell. The appearance was suggestive of mild inflammation, probably from the pressure or irritation of the overlying thickened epidermis. Except for some hypertrophy of collagen bundles the corium seemed normal. There was no loss of elastic tissue or fat. In some parts of the section large size of the lumen of the sweat glands and small size of the sebaceous glands could occasionally be seen.

A section of one of the nails revealed a swelling of the horn cells making up the nail plate. The horn cells retained their globular

outline and presented evidence of degeneration.

**Discussion**

The case of pachyonychia congenita, presented showed pachyonychia, localized and circumscribed plantar keratoses, leukoplakia of the mucous membrane of the mouth especially on the tongue and follicular keratosis of the body particularly on the extensors of elbows and knees. It belongs to the group of abnormal dyskeratosis in which the normal keratinization is disturbed.

The formation of keratin is a specific function of the epithelial cells. Dyskeratosis is an underlying disturbance in this specific function, i.e., there is an interference with the aging process of epithelial cells. According
to Diasio (4) the abnormal cornification may be characterized by (1) faulty development or (5) abnormal behavior. The change is most marked in the horny layer.

The dyskeratotic process (i.e., the disturbance in the evolution of epithelial cells with special reference to cornification) may be accelerated, resulting in a premature senility of epithelial cells, or it may be retarded, causing delayed juvenility of these cells. In the former one finds the dry, and in the latter the wet, type of dyskeratosis. In both types the basal cell layer may be normal. The disturbed keratinization may affect isolated cells or groups of cells, sections, layers or even the entire epidermis. Moreover, it may or may not be accompanied by inflammatory manifestations in the cutis, epidermis or both.

Diasio (1) stated that pachyonychia congenita of Jadassohn affects chiefly males and appears to be most prevalent among Slavs and Jews of Slavonic origin. He mentioned also that it is rarely represented in the sibships of twins, the notable exception being the boy seen by Mendes da Costa and Van der Valk (23) who was one of triplets.

Review of the literature to date showed that the great majority of reported cases were males. Our patient being a female seems to be a rare case. The disease appears to be rare in our country, since this is the first reported case in the U. A. R.

Andrews and Strumwasser’s (6) patient began to show pachyonychia changes two weeks after birth. In our case the nail defects were present at birth and the skin lesions soon followed.

Associated defects have been described. In the patients seen by Bettmann (24), Jadassohn, and Lewandowsky (1), Strandberg (25) and Schafer (4), bullae appeared from time to time on the thickest parts of the soles.

In the majority of cases keratosis of the mucous membranes of the mouth (leukoplakia) has been noted. In Burns (26) patient, a boy, the nasal mucosa was similarly affected while the tympanic membranes were so thickened and retarded that the boy was totally deaf. Opacities and partial blindness due to dyskeratosis of the cornea have been reported by Brettauerg (27), Brunauer (28) and Burns (26). The boy seen by Schafer (4) had bilateral cataract and twinning of the incisor teeth. Mental deficiency was noted by Brunauer (28). Riehl’s (29) patient had abnormally long bones, excessive motility of the joints and the striped or linear form of palmar keratosis.

Of the above mentioned associated defects, only leukoplakia of the tongue and mucous membrane was present in our case.

Alkiewicz and Leboda (19) in 1961 reported on the clinical course and histology of two cases of pachyonychia congenita. They studied the histology of the nails in these patients and found that the nail plate appeared somewhat thickened. Its cell-structure was unchanged. On the palmar side of the nail plate there was a small band of closely attached horny layers, the cells of which were elongated and ran parallel to the nail plate. In the deeper layers the horn masses form an undulant structure, these layers contained amorphous lumps arranged in nests. These lumps gave positive Periodic Acid Schiff, and ninhydrin Schiff reactions and appeared gram negative. In the horny layers many cells showed extensive colloid degeneration.

The study of a section of one of the nails in our case revealed swelling of the horn cells making up the nail plate, the horn cells

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Wright and Guequierre (15) reported two cases of pachyonychia congenita and found that massive doses of vitamin A with infrequent bathing and topical use of emollients ameliorated symptoms referable to the dry skin. In one of their cases wet dressings with buffered cysteine hydrochloride solution resulted in prompt healing of eroded lesions. In the same case excision of the distal phalanges of all fingers of both hands gave excellent functional results. Previously, the hands have been almost useless because of painful overgrowth of the nails. Simple removal of the nail plate was ineffective.

Garb (15) reported on the treatment of pachyonychia congenita and claimed regression of plantar lesions on patients wearing specially made rubber base molds and shoes.

Porter (20) in 1951 found a low mean value for vitamin A in plasma of four cases with pachyonychia congenita and he claimed that 2 of these had benefited with vitamin A therapy.

In our case large doses of vitamin A over a reasonably long period were given without noticeable improvement. Emollients gave temporary relief. The method advised by Garb was not tried in our case.

Summary

(1) The first report of pachyonychia congenita in the U. A. R. is presented.

(2) The rarity of onset of this congenital dyskeratosis in females in the literature is stressed—our case is a female.

(3) A detailed study of the clinical, histological and laboratory findings in this case is given.

(4) A detailed review of the literature on this very rare disease is forwarded.

Acknowledgement

We should like to express our thanks to Dr. A. Elwi, Professor of Pathology, Faculty of Medicine, Cairo University for his valuable aid in the interpretation of the histopathologic findings. Again our thanks are due to the Translation Department of the National Research Center, Cairo, for translating foreign literature other to English.

References


5) Fox, H. (1928) cited Andrews and Strumwasser (Ref. 6).


27) Brettaufer, cited Heller (Ref. 3).