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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.
Siemens' Keratosis Multiformis Idiopathica (Jadassohn-Lewandowsky's Pachonychia Congenita)
by
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With six pictures

(Received on January 28, 1961)

Numerous case studies of pachyonchia congenita have been published (by Kumer and Loos; Touraine and Granjon; Goldberg; Touraine; Franklin; Anderson; Wright and Quequierre; Wise; Vermenouze; Wright and Gross; Duperrat and Poncelet; Brain; Mullins, Murray, and Shapiro; Krepler; Kern) since 1906, when Jadassohn and Lewandowsky defined pachyonchia congenita as a characteristic systemic syndrome involving the skin and its adnexa separate from other congenital dyskeratoses.

The original name for the syndrome, which is still commonly used today, refers to the characteristic nail abnormalities that are present at birth or develop shortly thereafter. However, they are only one part of a generalized keratinization anomaly. As patients get older, but usually while they are still children, skin dyskeratosis, and possibly disorders of the mucosa as well, follow. This is why Siemens refers to a keratosis multiformis idiopathica (KMI). We would like to use this name as well, despite the fact that "keratosis" should actually be replaced by "dyskeratosis." In addition to nail abnormalities and keratinization disorders affecting the skin and mucosa, hyperhidrosis, hair and tooth anomalies (Brain), a predisposition to blistering, bone growth disorders, and reduced intelligence have also been observed in some cases. The clinical picture changes depending on when symptoms appear, and in what combination they appear, so that "no two cases are alike" (Moncorps). Therefore, it is not surprising that the literature has described other diseases, particularly congenital nail anomalies, as pachyonchia congenita. Touraine mentions a number of "formes frustes" that are not clearly classifiable as keratosis multiformis idiopathica.

It is not clear how the syndrome is inherited. Kumer and Loos report on a family in Upper Austria with 23 people with KMI (out of 74 total family members) across five generations. Fischer reports on eight KMI sufferers in five generations, and Touraine observes 37 KMI patients in one family that had only 39 healthy, unaffected people. While KMI seems to be dominant within these families, other researchers reported only individual cases, with no other family members affected (Grace; Sohrweide, Franklin; Vermenouze). There is no known consanguinity in the affected families. Race also does
not seem to play a role. According to Kumer and Loos, the syndrome strikes more men than women.

Kumer and Loos find it helpful to differentiate between three different types of KMI:

1. Pachyonychia-like nail abnormalities, symmetric keratomas--some rounded, some elongated--on the palms and soles of the feet, follicular skin keratoses.
2. Pachyonychia-like nail abnormalities with symmetric keratomas, follicular keratoses, and oral leukokeratosis (the mouth, gums, tongue, and in isolated cases, the laryngeal mucosa is affected). The literature often refers to this as the "Riehl" type illness, because the abnormalities agree with the "unusual palmar and plantar keratosis with nail abnormalities" described by Riehl.
3. Pachyonychia-like nail abnormalities with symmetric keratomas and follicular skin keratoses, and mucosal and keratinization abnormalities.

Because Type 2 has been described most frequently, Type 3 is considered to be extremely rare.

Due to the special course of the syndrome, the authors' own observations will be described here in more detail:

Family history. Father and mother are both healthy. No consanguinity in the family bloodline. No known congenital anomalies or skin conditions in the family. Because the rest of the family lies in Bavaria, the information provided by the parents had to suffice.

Before she gave birth to the sick child, the mother had a stillbirth following a normal pregnancy. The cause of death remained unexplained, but the stillborn baby was reportedly not deformed in any way. The second pregnancy was completely normal.

Personal history. At birth, the child had noticeable abnormalities in the fingernails and toenails, which appeared thickened and frayed. The skin was not affected. Shortly after the child's sixth birthday, small tubercles appeared on both thighs and proceeded to spread to the knees, elbows, palms, and soles of the feet. Particularly in the palmar and plantar areas, the nodules ran together into dark red plaques. The child's voice, which had already been rough for a long time, became hoarse. The child had no other major illnesses.

**Findings upon admission in December 1958**

The general clinical examination did not reveal any pathological findings other than the hoarseness.

Affected are: 1. all nails. 2. knees, forearms and lower legs, top of the foot, insides of the hands and soles of the feet. 3. Eyelids, lips, nostrils, corners of the mouth, tongue, hard palate.

Figure 1: Nail abnormalities, December 1958
The nails are thickened, dull, and without recognizable lunulae. They are a discolored yellow. The free edge seems frayed. Parts of the surface have a convex curvature. The tubercles on the palms and soles of the feet overlap, range from pinhead-size to lentil-size, are arranged symmetrically, and are raised; some are shiny and coated with yellow flaky material.

Both eyelids are thickened, red, and heavily keratinized. The mucosa around the vestibule of the nose exhibit scabs. Lingua plicata, which is bright red at the edges and tip. Almond-sized, sharply delimited redness on the hard palate. The hair is conspicuously dry, downy, relatively sparse, and flaxen-blonde in color. Sweat gland function normal.

Histology (Prof. Herzberg). 1. Hard palate mucosa. Some vacuolous degeneration of the epithelial cells, general acantholysis with well-formed germ ridges\(^1\) that are certainly too long, exoserosis, moderate exocytosis, formation of perinuclear cavities. The high-reaching papules are edematous and have a raised bulbous shape. Sparse basal-cell infiltrate around the flexuose wide capillaries. The appearance of concentrically arranged, coreless keratinocytes in the bottom of the germ ridges with an indication of granulous cells speaks for the dyskeratosis.

2. From the skin of the left lower leg. The section contains two large parakeratotic scales, one fibrin-saturated with leukocyte infiltration. The epidermis in the region of this parakeratosis is free from granulous cells, only slightly acanthotic, and more block-shaped than wavy. Fibroblastic, papillary and band-cell infiltration that is loose, sub-basal, and perivascular, solely in the areas of desquamation. Cutis otherwise normal.

Figure 2: Leukokeratosis on the hard palate

Other clinical examination findings

ENT: Nose has many scabs. Pharyngonasal cavity and larynx unobstructed. Mucosa is unremarkable. A pfennig-sized lesion on the hard palate was excised.

Dental: Jaw slightly undershot. Tooth eruption and X-ray findings are appropriate for the patient's age.

Psychiatry: Intelligence appropriate to the patient's age.

Eyes: Normal. In particular, no changes to the cornea.

Lab findings: Lues serum reactions: negative. Vitamin A 120 IU/100 mL serum, Carotin: 440 -%. Blood counts: Unremarkable, with the exception of 10% eosinophils.

Course of illness

We observed the girl, who in 1958 presented with a full-blown case of keratosis multiformis idiopathica, for two years. The favorable developments in her case, which were not known to us from the literature, justify a detailed description of the course of her illness. The degree to which hormone therapy with prednisone and administration of vitamin A improved the patient's condition cannot be determined.

Figure 3: Prepared section from left lower leg

\(^1\) Translator's note: Literal translation of German "Keimleisten". Expression unknown at present.
Our treatment consisted of long-term daily administration of 5 mg prednisone and 50,000 U of vitamin A (Retinol drops) with a local application of 2% salicylic vaseline and boric ointment. After six months, the condition was noticeably improved. The affected areas on the knees and elbows were reduced to individual follicular hyperkeratoses, but the large-area hyperkeratoses on the palms and soles of the feet were unchanged. Over the following six months, the knees and elbows were symptom-free (cf. December 1959 photo), while the palmar and plantar keratoses remained unchanged. The patient's vitamin E level was 175 IU/100 ml. Continuation of the therapy seemed justified. The child also experienced good mental development and started school at Easter 1960.

At our last examination in September 1960, the dermatological symptoms were limited to nail abnormalities and the palmar and plantar hyperkeratoses, which no longer showed any signs of infection.

**Summary**

We report on a case of keratosis multiformis idiopathica in a five year-old girl. Over a period of two years, long-term treatment with vitamin A and prednisone contributed to nearly complete resolution of symptoms, with the exception of the nail abnormalities.

**Literature**