

PACHYONYCHIA CONGENITA JADASSOHN-LEWANDOWSKY: A DISORDER OF KERATINIZATION

Jens Thormann and Takasi Kobayasi

*From the Department of Dermatology, University of Copenhagen, Rigshospital,
Copenhagen, Denmark*

Abstract. A 15-month-old boy with pachyonychia congenita is described. The patient also had follicular keratosis, leukokeratosis of the tongue, and blisters on the soles. Histopathological examination of the follicular keratosis showed hyperkeratosis and acanthosis. Horny plugs were located in sweat pores. By electron microscopy abnormal keratinization was demonstrated.

Key words: Pachyonychia congenita; Genodermatosis; Sweat pore; Abnormal keratinization

Pachyonychia congenita is characterized by remarkable nail changes, in particular dirty brown thickenings of nails and especially of the free edges, in newborn babies (5, 6, 7, 8, 12, 13). This rare disease is considered to result from congenital ectodermal dysplasia (13). The disease was described by Wilson in 1905 (11) and by Jadassohn & Lewandowsky in 1906 (6). Diasio (5) described the disease as a variant of ichthyosis and called it pachyonychia ichthyosiformis.

Besides pachyonychia, this condition is accompanied by several manifestations in the skin and mucous membranes. Moldenhauer & Ernst (8), reviewing 93 cases described previously by other authors, presented 3 patients and summarized their symptoms. Follicular keratosis, one of the skin manifestations, is characterized by horny plugs in the follicular openings (1, 14).

CASE REPORT

The patient is a 15-month-old boy. The family history is entirely negative. He was born with brownish-yellow nails. A few weeks after his birth, the mother noticed that all the nails were thickening in the upward and outward directions. At the age of 8 months some of the nails became inflamed and dropped off. Horny spines devel-

oped on his knees, elbows and abdomen. A clinical examination showed normal growth and normal mental as well as somatic condition. All nails (Fig. 1) showed marked thickening, some of them measuring 0.6 mm. Some of the nail beds showed inflammatory changes. On the extensor surfaces of the knees (Fig. 2) and elbows, follicular keratosis was found. On the abdomen and the buttocks, discrete follicular horny spines appeared. In the right-hand corner of the mouth, a keratotic fissure was found, from which candida albicans was cultivated. The centre of the dorsum of the tongue was smooth and covered by a thin greyish keratotic film. The development of the teeth was normal for the age of the patient. The eyes were normal. At a follow-up examination, at the age of 18 months, large tense bullae had developed on the soles of the feet.



Fig. 1. Typical thickened nail of pachyonychia congenita.

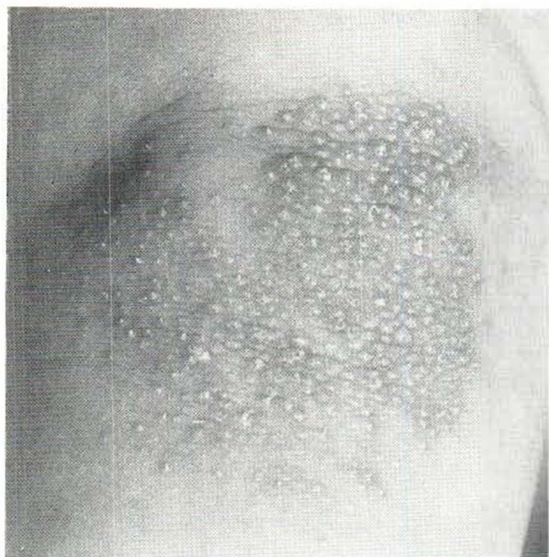


Fig. 2. Follicular hyperkeratosis on the right knee.

Light microscopy

Skin biopsies were removed from the involved areas of the knees, fixed in neutral formol, and serial paraffin sections were prepared. The sections were stained with hematoxylin-eosin, a 0.1% aqueous solution of toluidine blue and the van Gieson-Hansen method. A thick horny layer covered



Fig. 3. Hyperkeratosis, thick stratum granulosum and acanthosis of lesion of the right knee. Hematoxylin-eosin stain. $\times 220$.

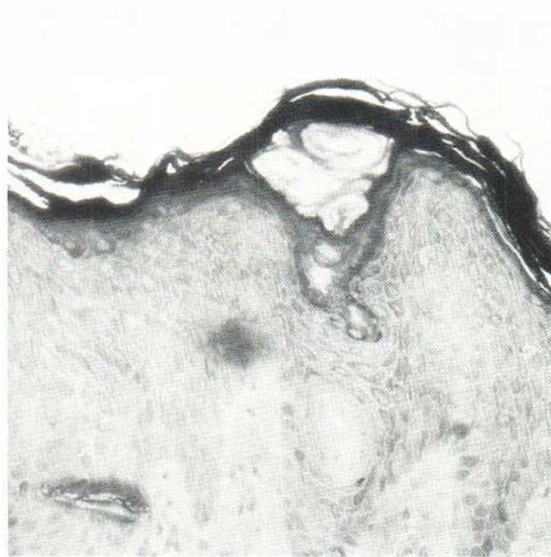


Fig. 4. Funnel-like sweat pore with thick stratum granulosum cells and a horny plug. The opening of the duct is preserved. $\times 435$.

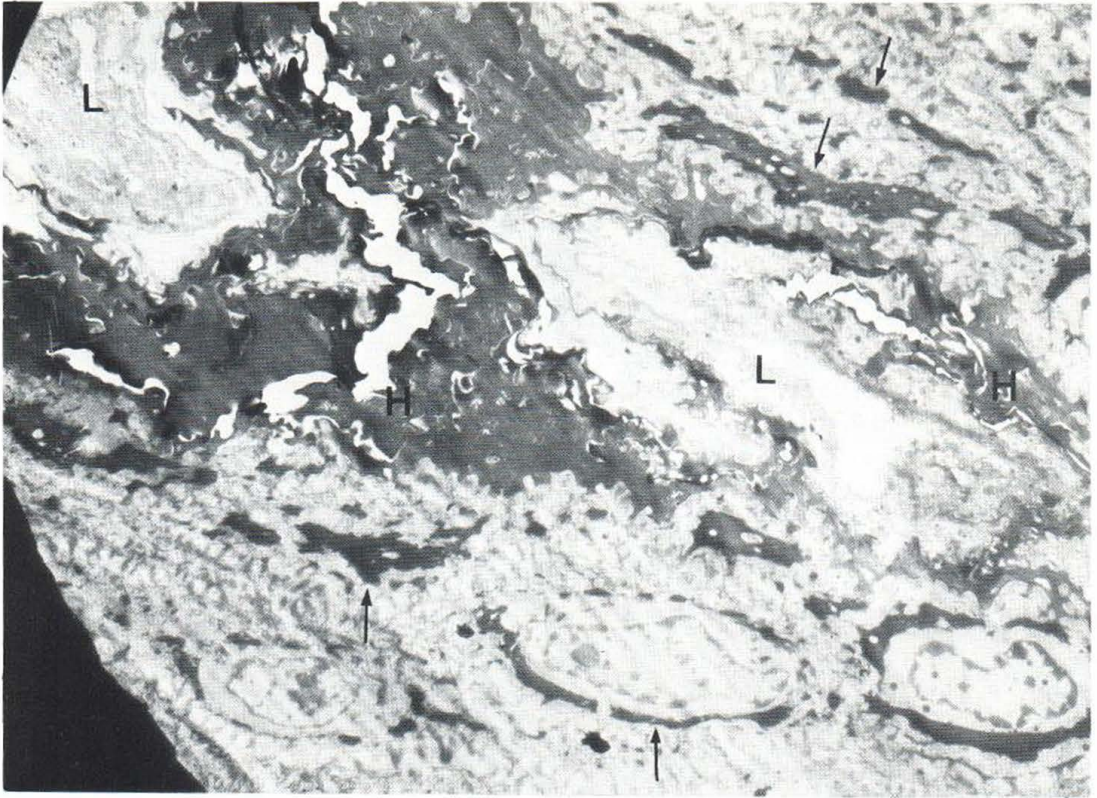
the epidermal surface and the walls of funnel-shaped sweat pores, the cavities of which were filled with horny masses. No hair follicles were seen in the sections. No retention cysts were found, neither was there any parakeratosis. The stratum granulosum was thickened. Wide elongated rete ridges invaded the upper corium, separated by long thin papillae. Squamous and basal epidermal cells appeared normal. The lower parts of the intra-epidermal sweat ducts were normal. The dermis showed a minimum of cell infiltration (Figs. 3, 4).

Electron microscopy

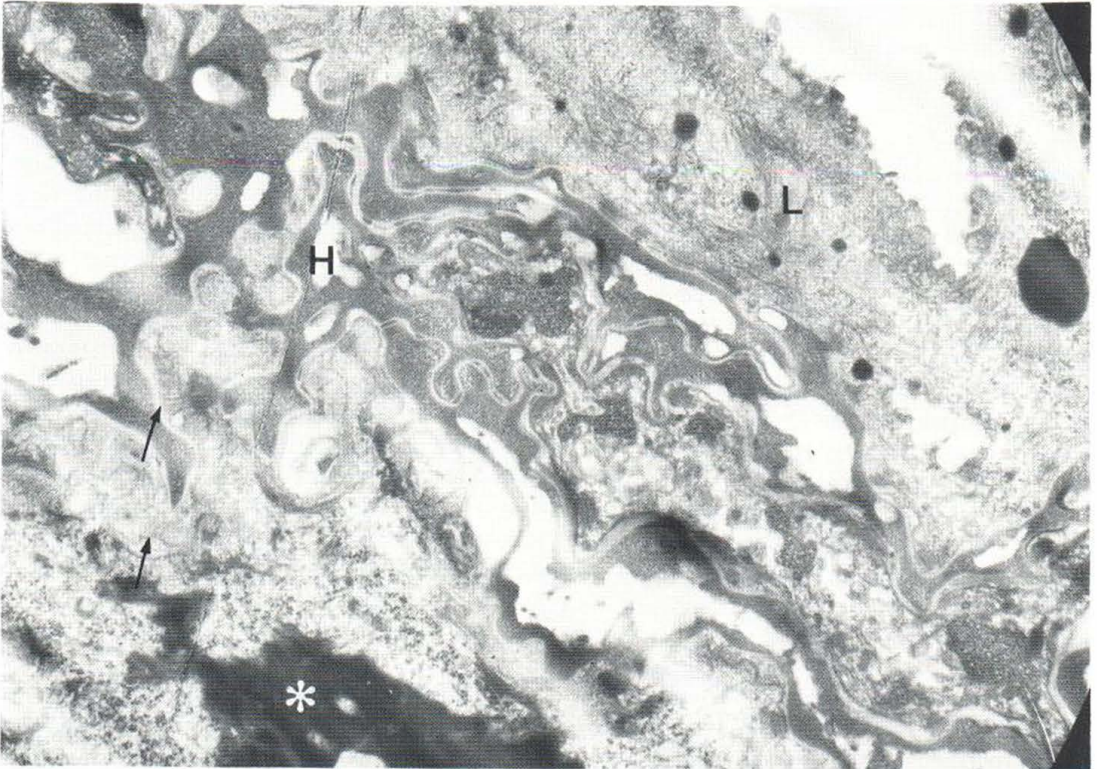
Skin specimens removed from the same area as for light microscopy were fixed in 6% glutaraldehyde in Veronal acetate buffer, pH 7.2, with 7.5% sucrose. The specimens were osmicated, dehydrated and embedded in Epon 812. Ultrathin sections were stained with uranyl acetate and lead

Fig. 5. Funnel-like sweat pore with a horny plug. Horny cells (*H*) are seen under the luminal cells (*L*). Thick keratohyalin granules (*arrows*) surround the perinuclear areas. The lumen of the sweat duct is preserved. $\times 4000$.

Fig. 6. In the deeper part of the funnel-like sweat pore, luminal cells (*L*) show normal structures, while horny cells (*H*) and thick keratohyalin granules (*asterisk*) appear under the cuticle. Keratinosomes (*arrows*). $\times 40000$.



5



6

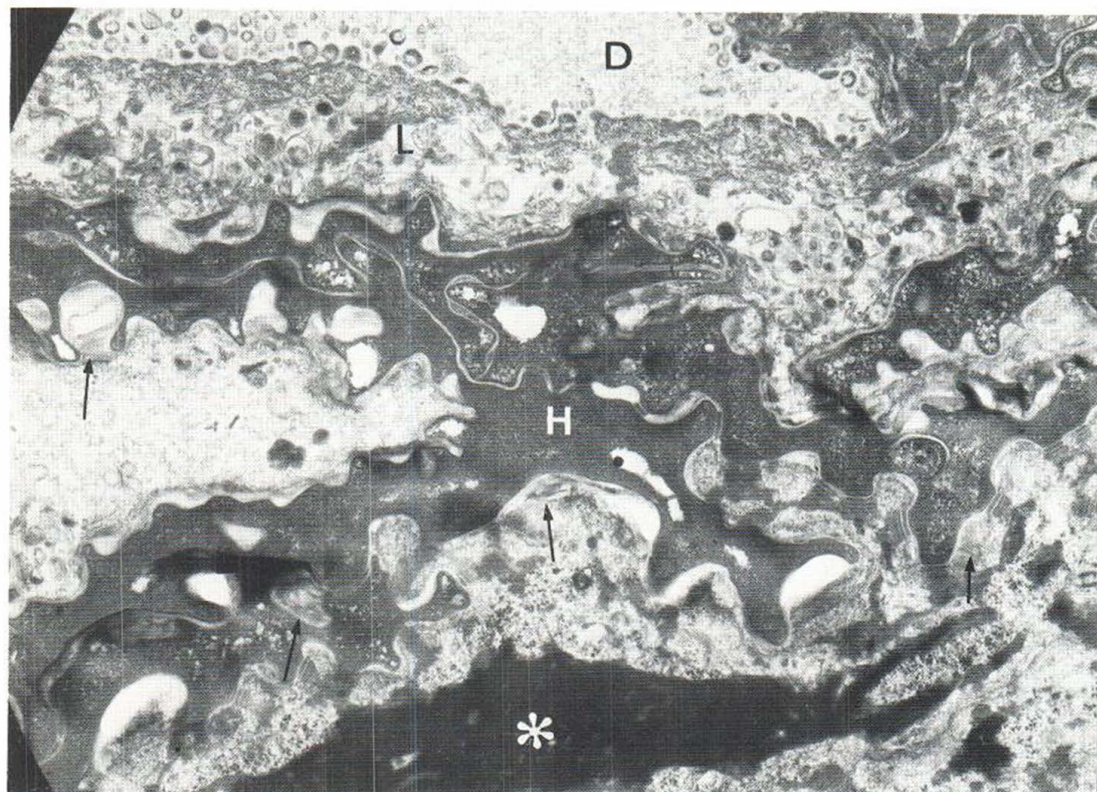


Fig. 7. Luminal cells (L) with round particles and filament bundles. Duct (D). Horny cells (H). Keratohyalin granule (asterisk). Keratinosomes (arrows). $\times 40\,000$.

citrate and observed in a Siemens electron microscope (Elmiskop 1A) at 80 kV.

Several layers of dense horny cells with lacunae covered the outer surface of the epidermis. The intercellular spaces of the horny layer were filled with lucent homogeneous material, remnants of keratinosomes and dense homogeneous desmosomes. Stratum granulosum cells in 3–4 layers contained thick tonofilament bundles with irregular masses of keratohyalin and numerous keratinosomes. The keratinosomes had a homogeneous content with indistinct transverse stripes. The cells lay closely packed and were connected by interdigitations and distinct desmosomes. Epidermal cells in the lower Malpighian and basal layers were connected by desmosomes crossing more or less dilated intercellular spaces. The cells contained thick bundles of tonofilaments in the periphery of the cytoplasm and mitochondria and ribosomes in the perinuclear areas. Glycogen particles were occasionally found among the cell organelles.

The walls of the funnel-like sweat pores showed normal luminal cells (Fig. 5) surrounded by several layers of horny cells (Fig. 6). The individual horny cells and intercellular spaces were identical with those of the surrounding epidermis. Below the horny cells, the stratum granulosum cells showed large keratohyalin granules and keratinosomes with indistinct inner structures, quite similar to those of the surrounding epidermal cells. The horny plugs of the sweat pores consisted of several dense horny cells. Luminal cells were seen in a cleft in the centre of the plug. The sweat pores were not closed by the plug (Fig. 7). These luminal cells contained horny masses, filamentous masses and keratinosome-like particles in their cytoplasm.

DISCUSSION

According to Moldenhauer & Ernst (8), palmar and plantar hyperkeratosis, follicular keratosis, leukokeratosis of the tongue and blister formation on the

soles were the most common symptoms accompanying pachyonychia congenita. Pachyonychia and leukokeratosis of the tongue are often noted in affected newborn babies. Later on, follicular keratosis starts on the elbows and knees and spreads to other parts of the body (8, 13). Histopathological examination reveals hyperkeratosis, acanthosis and horny plugs in follicular orifices (1, 13). Andrews (1) described horny plugs in sweat pores. The tortuous part of the epidermal sweat duct in normal skin does not contain horny cells under the luminal cells (4, 14). Keratin masses in luminal cells first appear in the horny layer. Compared with the structural aspects of the normal sweat duct, the present findings indicate that keratinization takes place in the outer part of the epidermal sweat duct. The detailed structure of keratinizing cells of the epidermal surface as well as in sweat pores is similar, though in the sweat pore, lacunae of horny cells and keratohyalin granules dominate. The other skin manifestations of the disease, viz. plantar hyperkeratosis and ichthyosis, show similar structures by electron microscopy (10). However, these changes of keratinization are different from those seen in normal keratinization, particularly the lacunae in the horny cells and the lack of separation of desmosomes between horny cells, the large indistinct keratinosomes, the increased numbers of keratohyalin granules, and the thick tonofilament bundles. In such horny cells, keratohyalin granules and tonofilament bundles resemble those of ichthyosis congenita (2, 9) and ichthyosis hystrix (3). However, distinct keratinosomes are not seen in this disease. Congenital ichthyosiform erythroderma shows clumps of tonofilaments around the nucleus (11). In hyperkeratotic skin, after ultraviolet irradiation, the horny cells show round lacunae (9), which are not seen in this disease.

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T. Kobayasi, M.D.
Department of Dermatology
Rigshospital
Blegdamsvej 9
DK-2100 Copenhagen
Denmark