



Pachyonychia Congenita Project

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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.

may also explain some blisters occurring in comatose patients (2).

Our patient, of necessity, spent a prolonged period with his head in the same position in the ICU because of cerebral edema. This resulted, in pressure-induced ischemic changes over the scalp occiput. Although the hair loss was perhaps minor, it is likely to be permanent and may lead to embarrassment in the future. Such alopecia is preventable by regularly moving the position of the head during and after general anesthesia or immobilization; however, regular movements may not always be practical.

We believe that such hair loss may not be uncommon, but is often unrecognized. A pertinent reason for the latter is that an occipital bald patch may remain unnoticed until long after a period of immobilization so that such an etiology may not even be considered.

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P. D. YESUDIAN, MRCP
J. L. VERBOV, FRCP
Liverpool, England

TWO CASES OF JUVENILE PEMPHIGUS VULGARIS: LONG-TERM FOLLOW-UP

To the Editor:

In 1991 we reported a case of juvenile pemphigus (JP) in a 13-year-old girl who was successfully treated with moderate doses of deflazacort 1 mg/kg/day slowly tapered to 0.1 mg/kg every other day (1). The therapy was maintained for 4 years without significant side effects. The patient has now been in complete remission and off treatment for 6 years. During this period, indirect immunofluorescence (IFF), initially performed every 3 months and subsequently once a year, has been negative.

In 1992 we observed another case of JP in a 17-year-old girl. The same treatment was given. The clinical course was similar to that of our first patient and the

response to therapy was good, although slower. Treatment was suspended after 5 years. Four years later, the patient is still without signs of the disease. IFF performed as in the first case was always negative.

The literature suggests that JP is a rare disease (2,3) and follow-up has seldom been reported (4). The course is variable, similar to adult pemphigus, although the prognosis of childhood pemphigus is good and helped by early therapy with moderate doses of systemic steroids (5). However, we do not have much or detailed information on the relationship between early treatment, clinical outcome and long-term prognosis. Long-term follow-up is important to detect flare-ups of the disease and to demonstrate that early diagnosis and prompt therapy are essential for a good prognosis (6).

In our two patients, diagnosis was early in the first and slightly later in the second, although both were within 6 months of onset. Treatment with moderate doses of steroids quickly controlled the skin manifestations and it was possible to completely suspend steroids after 4 and 5 years, respectively. These two cases seem in line with other reports. We suggest that detailed follow-up reports in diseases such as JP would be useful for a precise clinical prognostic profile.

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ELISA PIANIGIANI, MD
FRANCESCA IERARDI, MD
ANDREA ANDREASSI, MD
MICHELE FIMIANI, MD
Siena, Italy

PACHYONYCHIA CONGENITA: A CASE REPORT

To the Editor:

The term pachyonychia congenita (PC) was first used by Jadassohn and Lewandowsky to denote symmetrical thickening of all nails associated with palmoplantar

hyperkeratosis, blistering of the feet, oral and sometimes laryngeal leukokeratosis, hoarseness, palmar and plantar hyperhidrosis, follicular keratosis, and hair abnormalities (1,2). Most often the inheritance is autosomal dominant, but an autosomal recessive pattern has also been reported, reflecting genetic heterogeneity (2,3). Clinically there are probably four different syndromes (4,5). Type 1 is the classic form described by Jadassohn. In type 2, nail thickening is uniform and associated with chronic mucosal candidiasis. Keratoses are less severe. Type 3 has less severe nail thickening and keratoses and is associated with neonatal teeth and epidermal cysts (Jackson-Lawler type). In type 4, nail thickening and keratoses are associated with widespread flexural macular pigmentation. We describe a 15-year-old boy who presented with congenital pachyonychia and chronic oral candidiasis (type 2 PC) associated with woolly hair and recurrent pyodermas.

A 15-year-old boy born to consanguineous parents had thickened, angulated nails since 2 years of age. Initially he had recurrent painful erythematous swelling of the nail folds followed by shedding of the nails. This process ceased after a year and was followed by thickening and angular growth of the nails which was rapid and required frequent trimming. He had woolly hair since birth and oral white lesions since 3 years of age. He also complained of recurrent cutaneous pyogenic infections for the past 5 years. His physical and mental milestones were normal. None of the other siblings or family members had similar lesions.

On examination, all the fingernails and toenails were dystrophic, discolored, and thickened, with massive subungual hyperkeratosis producing a steplike deformity of the nail plate (Figs. 1 and 2). His hair was woolly and his tongue was thickened, with fissures on the dorsal surface (Fig. 3). Patches of white, creamy, pseudomembrane were present on the tongue and buccal mucosa along with angular cheilitis. He also had four episodes of furunculosis and deep-seated pyogenic abscesses over the back, abdomen, buttocks, and thighs during a 6-month follow-up period. His palms and soles did not show any hyperkeratosis or hyperhidrosis. His systemic examination was unremarkable.

A complete hemogram, serum biochemistry, and histology of skin biopsy specimens from the palm and back were normal. Tuberculin test was nonreactive. Potassium hydroxide (KOH) examination and culture of nail clippings were negative. *Candida albicans* was cultured from the mouth. *Staphylococcus aureus* was isolated from the recurrent pyogenic infections.

The oral candidiasis responded to weekly oral fluconazole (150 mg) and remission was maintained with topical clotrimazole. Episodes of pyogenic infec-



Figure 1. Bilateral and symmetrical thickening and dystrophy of nails.



Figure 2. Massive thickening of a fingernail.

tions responded well to oral cloxacillin. Pachyonychia showed minimal response to topical 10% salicylic acid ointment.

Symmetrical thickening of all nails is the most striking and consistent feature of pachyonychia congenital (1,2). As seen in our patient, thickening and discoloration of the nails is due to subungual hyperkeratosis, leading to upward angulation of the distal nail plate and incurving of the lateral borders (2). The presence of other abnormalities is not an absolute prerequisite for the diagnosis, as cases with involvement of nails alone have been reported (2,6). Our patient had features of type 2 PC, but the association of woolly hair and recurrent pyoderma has not been reported before in the literature.

PC is thought to be a state of abnormal keratinization of oral epithelium, nails, and skin (7). Keratin gene mutations in keratins K6/K16 and K17 might explain the clinical abnormalities in PC. Keratins K6/K16 are expressed in mucosal epithelia, follicular keratinocytes,

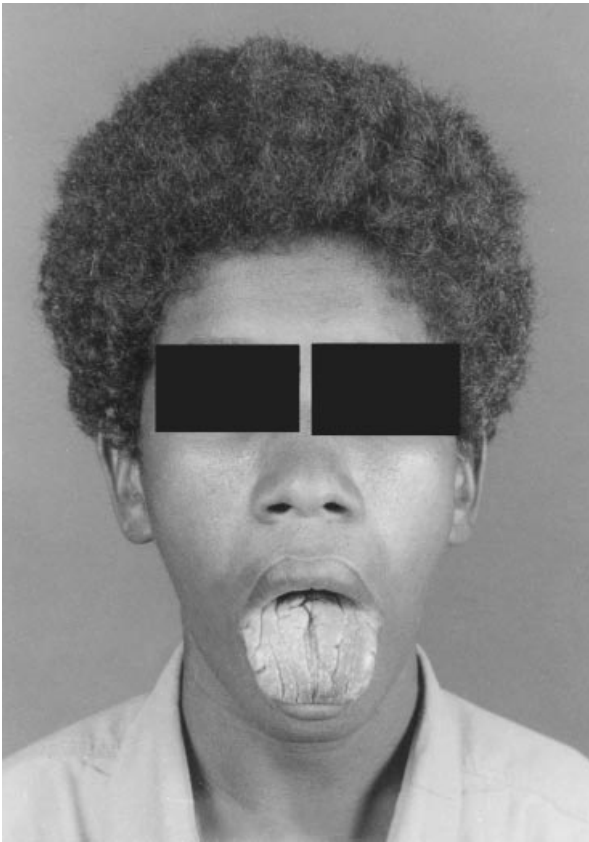


Figure 3. Face shows woolly hair and a fissured tongue.

and palmoplantar epidermis, whereas K17 is expressed in the pilosebaceous unit and basal appendageal keratinocytes (8). Our patient had involvement of the nails, hair, and oral mucosa, while palmoplantar keratoderma and other cutaneous changes were absent. This might reflect the genetic heterogeneity of the PC syndrome (1,6). Although parental consanguinity suggested an autosomal recessive inheritance in our patient, the possibility of a mutant gene couldn't be excluded due to the lack of a family history.

The common oral involvement in PC is leukokeratosis in the form of patchy or streaky, white, thickened areas on the tongue and oral mucosa (8). Our patient did not have leukokeratosis but had chronic oral candidiasis, which is the distinct clinical feature of type 2 PC (4,7). Impaired immunologic response to *Candida* antigen has been reported in some cases (7,8). It has been suggested that the inherited abnormality of the oral mucosa may predispose to infection with *C. albicans* in early infancy, which leads to the absence of a normal cellular response to *Candida* (7). The association of chronic oral candidiasis with recurrent pyoderma in our patient adds evidence in favor of an immune defect in type 2 PC.

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M. VIJAIKUMAR, MD
DEVINDER M. THAPPA, MD, DHA, MNAMS
CHANDRASHEKAR LAXMISHA, MBBS
Pondicherry, India

RECTORRHAGIA AND LICHEN SCLEROSUS IN CHILDHOOD

To the Editor:

An 8-year-old girl with precocious puberty had since 2 years of age pruritic genital lesions with occasional hemorrhagic blisters and bleeding, together with episodes of blood in the feces and rectorrhagia of unknown cause. Two rectocolonoscopies revealed only a linear fissure in the rectum, 2 cm from the anus. Previous treatment included mild topical corticosteroids and a high-fiber diet for prevention of constipation.

On evaluation at our institution, physical examination revealed lesions on the vulva, but a normal appearance in the anal and perianal areas (Fig. 1). A vulvar biopsy specimen showed findings characteristic of lichen sclerosus (Fig. 2). Other findings (including gynecologic findings) were unremarkable, and sexual abuse, bullous pemphigoid of the vulva, and lichen planus were ruled out. The rectorrhagic episodes without associated perianal cutaneous lesions were interpreted as unusual manifestations of lichen sclerosus. Treatment was initiated with a 0.05% clobetasol propionate cream nightly for 3 months, until the appearance of the vulva returned to normal, and then twice a week for 2 months. Exacerbations did not occur during intermittent maintenance treatment and it was then temporarily withdrawn until