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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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A case report of pachyonychia congenita is presented. The literature is reviewed for etiology, clinical findings, pathology and treatment. The clinical findings are defined in detail to provide a guide for correct diagnosis and treatment.

Jadassohn and Lewandowski used the term pachyonychia congenita to describe an autosomal dominantly inherited congenital disorder of nails, skin and mucous membranes. Dystrophic changes involving all fingernails and toenails is the hallmark of this disease. Skin changes include palmar and plantar hyperhidrosis with keratoderma, plantar bullae, keratosis pilaris, pilar cysts and partial to complete alopecia. Leukokeratosis of the oral, nasal or otic membranes and corneal dyskeratosis with cataracts can occur. Natal teeth are not uncommon and two cases of permanent hoarseness have been reported. The purposes of this paper are to review the literature on pachyonychia congenita, present a case report and discuss the author's diagnosis.

Clinical Manifestations

Symmetrical involvement of all fingernails and toenails is present at birth or within the first 6 months of life. The distal nail plate is a dark yellowish-brown while the proximal portion is normal to slightly discolored. The free edge of the nail is elevated by a thick, keratinous nail bed. There is an increased incidence of paronychia with shedding of the nail once or twice yearly. By age 3 or 4, skin colored follicular papules with a central keratotic plug called keratosis pilaris appear on the extensor surfaces of the extremities. The lesions often become worse during the cold months. Sometimes the papules coalesce to form verrucoid lesions.

Hyperkeratotic lesions may occur on the thenar and hypothenar eminences, volar surface of the fingers and weight-bearing surface of the feet. Frequently there is associated hyperhidrosis of the palms and soles. Large painful bullae on the plantar surface of the feet are not uncommon, especially during the summer months. These lesions can be severe enough to inhibit normal walking. Leukokeratosis of the oral, nasal and otic mucous membranes are commonly seen by puberty. This appears as striate or diffuse white thickened patches. This should be differentiated from leukoplakic lesions of Darier-White’s disease, hereditary benign intraepithelial dyskeratosis and white sponge nevus of Cannon. All except leukokeratosis oris of pachyonychia congenita have diagnostic cytopathic features.

Natal teeth are not uncommon in persons with this disease. Usually there are four to eight gritty teeth in the mandible at birth. These teeth are shed before age 6 months and secondary dentition occurs normally.

Corneal dyskeratosis and cataracts are sometimes seen. Severity varies, but blindness has been reported. Appearance is not consistent with any age group.

Epidermal cysts of the face, neck and especially lower anterior central chest are sometimes seen. Steatocystoma is the term most often used to name these lesions, but a more correct term is pilar cyst, as they arise from the pilary apparatus. Size ranges from a few millimeters to 2 cm. in diameter. Aspiration or incision produces either a syrupy, odorless liquid or a white, cheesy, malodorous exudate.

Congenital alopecia is a rare finding and can be partial or complete. Existing hair is generally dry, lusterless, kinked and sparse.
There are two reported cases, within one family, of permanent hoarseness. Laryngoscopic examination showed a thickening of the posterior laryngeal commissures. 

Case Report

A 3-year-old black female was examined for a chief complaint of unsightly finger and toenails. The condition had been present since age 3 months. On several occasions the nails had shed spontaneously. This was followed by regrowth of a similarly deformed nail.

Past medical history was unremarkable. Pregnancy was uneventful and there was a normal, spontaneous delivery. The child was of average weight, and growth and development were normal.

The family history was relatively unremarkable. The maternal grandfather, who is deceased, had a history of a chronic skin condition and possibly a similar nail problem.

Positive physical findings were confined to the nails. All were dark brown and elevated distally by a thick, keratinous cheesy-looking nail bed (Figs. 1 to 3). Cultures of the nail bed and plate were negative for bacterial or fungal infection. A diagnosis of pachyonychia congenita was made.

Treatment consisted of debridement of the nail.

Figure 1

Figure 2

Nail lesions have been treated by plate avulsion and deep excision of the bed and matrix. Extreme cases have been treated by distal phalangeal amputation and plastic skin repair. Mechanical debridement of the nails with a drill and bur has been effective, temporary relief of pain and disfigurement. High doses of vitamin A (100,000 units/day) has a questionable effect on nails. Steroids are occasionally used for paronychia.

Hyperkeratoses and bullous lesions have been treated with high doses of vitamin A and with plastic surgery. Both methods give questionable results. The most successful treatment has been use of molded shoes to accommodate the deformity.

Keratosis pilaris has been treated with vitamin A, wet dressings and compresses. Treatment for this is not always necessary.

Pilar cysts are generally excised surgically if only a few exist. When several cysts are present, incision and drainage with phenolization gives cosmetically good results.
with a podiatry drill (Fig. 4). The improved appearance of the nails could not be maintained by use of Keratyl Gel, salicylic acid cream or Carmol cream. Surgical removal of the nail plate with deep excision of the matrix was refused by the patient's mother.

The patient was seen 5 months later at which time flesh-colored follicular papules with central keratotic plugs were noticed over the flexor and extensor surfaces of the legs (Fig. 5). These lesions were diagnosed as keratosis pilaris. No treatment was instituted as involvement was minimal and presented no problem to the patient.

Discussion

The key to making a diagnosis of pachyonychia is the nail pathology. All cases reviewed in the literature report smooth surfaced, discolored nails elevated at the free edge by a thickened nail bed.

Involvement extends to all finger and toenails between birth and 6 months.

The case presented here is unusual in that on initial examination, the deformed nails were the only clinical findings. In a report on 6 cases within one family, Jackson states that nail involvement can occur alone; however, lesions of the nails, skin and mucous membranes are usually seen.

Fitzpatrick et al. list 18 congenital and hereditary diseases involving nails, but only two besides pachyonychia congenita cause hypertrophy. In broad thumbs and broad great toes with facial abnormalities and mental retardation, the name describes the clinical findings which in no way resemble those of pachyonychia. Hemifacial or unilateral hypertrophy (congenital hemihypertrophy, Curtius syndrome, Steiner syndrome, hemigigantism, hemimacrosomia, partial gigantism) only affects one side of the body. Two other diseases, hidrotic ectodermal dysplasia and dyskeratosis congenita, are often confused with pachyonychia congenita although clinically they present with nail atrophy (Table 1).

The case reported here is further confirmed by the finding of keratosis pilaris. According to other cases in the literature, this is a secondary finding not commonly seen before age 3 or 4. The age of the patient at this time was 3 years and 5 months.

Clearly, at the time of examination this patient did not exhibit many of the clinical findings usually associated with pachyonychia congenita. There was no history of natal teeth or congenital alopecia. Nasal, lingual, oral and otic membranes were clear and hyperkeratosis with hyperhidrosis and epidermal cysts were not present. In support of the diagnosis, however one must appreciate the expected timetable and incidence of occurrence for each of these lesions. As mentioned under “Clinical Manifestation,” many of these findings are considered
Table 1

<table>
<thead>
<tr>
<th></th>
<th>Pachyonychia Congenita</th>
<th>Ectodermal Dysplasia</th>
<th>Dyskeratosis Congenita</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Origin</strong></td>
<td>Hereditary</td>
<td>Hereditary</td>
<td>Hereditary</td>
</tr>
<tr>
<td><strong>Onset</strong></td>
<td>Birth to 6 months</td>
<td>Birth</td>
<td>Birth</td>
</tr>
<tr>
<td><strong>Nails</strong></td>
<td>Nail bed hypertrophy</td>
<td>Nail atrophy</td>
<td>Nail atrophy</td>
</tr>
<tr>
<td><strong>Palm/soles</strong></td>
<td>Hyperhidrosis, hyperkeratosis, possible bullae on soles</td>
<td>Palmar keratosis</td>
<td>Hyperhidrosis, hemorrhagic bullae on palms and soles</td>
</tr>
<tr>
<td><strong>Skin</strong></td>
<td>Keratosis pilaris, pilar cysts</td>
<td>Dry skin, sebaceous hypoplasia</td>
<td>Leukokeratosis oris and leukoplakia oris (premalignant) ulceration</td>
</tr>
<tr>
<td><strong>Mucous membranes</strong></td>
<td>Leukokeratosis of oral, lingual, nasal, otic membranes</td>
<td>None</td>
<td>None</td>
</tr>
<tr>
<td><strong>Eyes</strong></td>
<td>Corneal dyskeratosis, cataracts</td>
<td>None</td>
<td>Constant tearing, keratinization of tear ducts</td>
</tr>
<tr>
<td><strong>Teeth</strong></td>
<td>Occasional natal teeth (mandible), normal secondary dentition</td>
<td>Minimal dental abnormalities</td>
<td>Minimal dental abnormalities</td>
</tr>
<tr>
<td><strong>Hair</strong></td>
<td>Thin, dry, lusterless, kinky; complete alopecia possible</td>
<td>Normal to slight alopecia</td>
<td>Alopecia of scalp, eyebrows, eyelashes</td>
</tr>
<tr>
<td><strong>Voice</strong></td>
<td>Permanent hoarseness, a rare finding</td>
<td>None</td>
<td>None</td>
</tr>
</tbody>
</table>

rare while others are not usually seen before puberty or adulthood.

Laboratory studies were completely normal in this patient. This is pertinent to the diagnosis as there are no known laboratory findings consistent with the disease. Certain histopathologic findings are common as was mentioned under “Pathology.”

**Summary**

Pachyonychia congenita is a rare disorder inherited as an autosomal dominant gene with incomplete penetrance.\(^2\,^3\) Zeigman states that fewer than 60 cases have been reported in the literature.\(^5\) The disease can express itself as changes in nail morphology only, but this is extremely rare. More often the disease is a widespread dysplasia of nails, skin and mucous membranes.\(^6\) Usually manifestations are similar among affected members of the same family, but interfamilial differences are not uncommon.\(^7\)

Diagnosis is based on clinical findings. Family history is often inconsistent and laboratory tests are not helpful. Gross pathology of all fingernails and toenails at or soon after birth is the sine qua non of pachyonychia congenita.

Cure of the disease is unknown. Palliation and cosmesis are the goals of treatment. Lesions persist throughout life and except for ocular lesions, usually cause no serious disorders.\(^5\)

**References**


**Additional References**