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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.
Oral manifestations of pachyonychia congenita

Report of a case


BENSALEM PEDIATRIC-MEDICAL CENTER

Few cases of pachyonychia congenita are reported in the dental and medical literature because of the rarity of the disease. This article presents a review of the literature and adds a new case history. Examination of a 4-year-old boy revealed the presence of the disease, which was also present in the mother and a newborn sibling.

Pachyonychia congenita is a relatively rare congenital disease. In addition to changes on the nails, other abnormalities of the skin and mucous membranes are seen. In the area of particular interest to the dentist are the abnormalities present in the keratotic layer of the tongue and, in some cases, the commissure of the lip.

According to Andrews, the disease was first mentioned in the literature by Jadassohn and Lewandowski in 1906. They first described a peculiar anomaly of the nails. The nail changes are characterized by a great thickening which increases toward the free border. The nail plates are extremely hard and are attached firmly to the nail beds. On the extensor surfaces of the extremities and buttocks and lumbar regions there are follicular, keratotic, grayish black papules which have in their centers horny cones that fit into corresponding craterlike depressions. Removal of the central cones is fairly easy, leaving a slightly bleeding cavity. The eruption of the outer aspects of the upper and lower extremities is also follicular, resembling keratosis pilaris. This latter condition is not constant and disappears at times. Jadassohn and Lewandowski also mentioned that leukokeratosis of the tongue and plantar and palmar hyperhidrosis may be present.

In 1921, Murray cited four cases of “hereditary hypertrophy of the nail bed...”
Table 1

<table>
<thead>
<tr>
<th>Author</th>
<th>Year</th>
<th>No. of cases</th>
<th>Symptoms</th>
</tr>
</thead>
<tbody>
<tr>
<td>Wilson</td>
<td>1905</td>
<td>1</td>
<td>Hyperkeratosis of nail bed</td>
</tr>
<tr>
<td>Jadassohn and</td>
<td>1906</td>
<td>1</td>
<td>Hyperkeratosis of nail bed</td>
</tr>
<tr>
<td>Lewandowsky</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Murray</td>
<td>1921</td>
<td>4</td>
<td>Hypertrophy of nail bed, neonatal teeth</td>
</tr>
<tr>
<td>Dassio</td>
<td>1934</td>
<td>1</td>
<td>Dry scalp, hyperkeratosis of nail bed</td>
</tr>
<tr>
<td>Andrews</td>
<td>1935</td>
<td>1</td>
<td>Dry scalp, hyperkeratosis of nail bed</td>
</tr>
<tr>
<td>Kumer and Loos</td>
<td>1935</td>
<td>34</td>
<td>Keratosis of hands and feet, follicular keratoses of body, leukokeratosis oris, corneal changes</td>
</tr>
<tr>
<td>Tauber, Goldman, and Classen</td>
<td>1936</td>
<td>1</td>
<td>Keratosis of hands and feet</td>
</tr>
<tr>
<td>Wright and Guequierre</td>
<td>1947</td>
<td>2</td>
<td>Bullae of feet and hands, hypertrophy of nail bed, keratosis of buccal mucosa</td>
</tr>
<tr>
<td>Mullins, Murray, and Shapiro</td>
<td>1955</td>
<td>1</td>
<td>Bullae on sole, nail keratosis</td>
</tr>
<tr>
<td>Kelly and Pinkus</td>
<td>1958</td>
<td>1</td>
<td>Callosities on toes</td>
</tr>
<tr>
<td>Gorlin and Chaudhry</td>
<td>1958</td>
<td>1</td>
<td>Leukokeratosis of tongue, nail hypertrophy</td>
</tr>
<tr>
<td>Jackson</td>
<td>1961</td>
<td>6</td>
<td>Blister of feet, leukoplakia, hyperhidrosis of palm and foot</td>
</tr>
<tr>
<td>Joseph</td>
<td>1964</td>
<td>5</td>
<td>Leukoplakia, keratosis of nails</td>
</tr>
<tr>
<td>Cosman, Symonds, and Cricklair</td>
<td>1964</td>
<td>1</td>
<td>Parakeratosis of nails</td>
</tr>
<tr>
<td>Solerquist and Reed</td>
<td>1968</td>
<td>3</td>
<td>Neonatal teeth, thickening of nails</td>
</tr>
</tbody>
</table>

associated with a history of erupted teeth at birth," Gorlin and Pindborg also mention the possibility of neonatal teeth. They cited gernination of teeth, but this was probably due to chance. In 1958 Gorlin and Chaudhry described the oral manifestations of pachyonychia congenita. The oral lesions took the form of white opaque thickenings. These thickenings were said to be focal, involving only a portion of the buccal mucosa or tongue, or generalized, covering the entire mucosal surface of the tongue, lip, and cheek. Angular cheilosis is also commonly present. At the time of Gorlin's case report, it was mentioned that not more than one dozen cases had been cited in the domestic literature since the original report in 1929 (Table 1).

In 1964 Joseph reported that there may be three types of pachyonychia congenita. Type I presents a symmetrical keratosis of the hands and feet with follicular keratosis of the body; Type II is like Type I but with leukokeratosis oris (Biehl type); and Type III is like Type I but with corneal changes. Joseph also states that the disease is inherited as a dominant characteristic. Gorlin mentions that while the disease appears to be autosomal dominant, penetrance does not appear great. He also mentions there may be a somewhat higher frequency among Jews.

**CASE REPORT**

This present case involves a 4-year-old white boy. When the child was seen for an initial dental examination, deformed fingernails were observed in addition to what appeared to be some type of fungal infection about the face. The mother had noted on the medical history that the child had pachyonychia congenita and that she was also affected.

When the child was 14 months old, he was admitted to the local community hospital because of persistent fever and rash. The following day he began to have sores in the mouth and
Symptoms

is of nail bed
of nail bed, neonatal teeth
keratosis of nail bed
keratosis of nail bed and hands, follicular
f bais, leukokeratosis oris, nips
ands and feet
ands, hypertrophy of nail
sis of buccal mucosa, nail keratosis
oes
of tongue, nail hypertrophy
leukoplakia, hyperhidrosis
foot
keratosis of nails
of nails
h. thickening of nails

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Fig. 1. Pedigree of family manifesting pachyonychia congenita.

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Fig. 2. Nails of sibling at 2 months of age, already showing changes of pachyonychia congenita.

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in the diaper area. There were lesions resembling impetigo on the face, tongue, lips, and ears. There was also a monilial type of rash of the perianal area. The child's nails appeared to have a fungal infection, which was noted shortly after birth. A diagnosis of pachyonychia congenita and severe monilia of the mouth, face, and perianal area was made. The child's course in the hospital was of slow progress. The lesions bled freely when traumatized and seemed to be slow in healing. At the time of discharge there were signs that the lesions were healing and the child appeared to be doing well. He was receiving Mycostatin four times a day along with Fungisone lotion four times a day to the tongue, face, and perianal area. The boy was in the hospital 9 days.

At the time of his visit to the dental office the child appeared well nourished and healthy. Consultation with the dermatologist involved in this case disclosed that the child was seen
Fig. 3. Nails of patient at the age of 4 years with gross changes of pachyonychia congenita.

Fig. 4. Keratotic changes to the dorsum and lateral border of tongue of the patient.

periodically and at present the monilia rash was being treated with a Mycostation lotion. The dermatologist also stated that a younger sibling (2 months of age) had now shown signs of pachyonychia congenita in the nail region.

Intraoral examination revealed twenty primary teeth, with numerous areas of caries evident. There was much material alba present on the mandibular anterior teeth. Examination of the tongue revealed several white patches on the lateral borders. The patient had no complaints with regards to burning sensations or loss of taste.

Examination of the mother revealed nail signs of pachyonychia congenita. Oral examination revealed a full maxillary and mandibular denture. There were white keratotic patches on the lateral border of the tongue as well as a white thickening on the buccal mucosa at the level of the interdental line. Only nail changes were observed in the 2-month-old sibling. No natal or neonatal teeth were present (Fig. 1).

The patient was seen for a periodic recall 6 months after the restorative dentistry was completed. Extraoral examination revealed healing of most of the ulceration at the commissure of the lips. Examination of the tongue revealed that the hyperkeratosis had migrated slightly to the anterior lateral border. There were no remarkable changes evident to the nail bed. Examination of the younger sibling revealed no oral manifestation of the disease; however, the nail beds continued to be affected (Figs. 2 to 5).

Fig. 5. Keratotic change the time of a 6-month recall.

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Possible histopathology
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Possible treatment and ramifications
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SUMMARY
A case of pachyonychia reported. A review of the condition.

REFERENCES
1. Andrews, G. C.: Diseases
3. Gorlin, R. J., and Pied Anatomy
4. Gorlin, R. J., and Chan ORAL SURG. I: 541, 195
Fig. 5. Keratotic changes to the dorsum and lateral border of the tongue of the patient at the time of a 6-month recall examination. Note the anterior migration.

In consultation with the dermatologist, it was decided that the child should not be subjected to any biopsy procedure. The white areas on the tongue were clinically and histologically similar to the white sponge nevus (congenital leukokeratosis mucosae oris) and had no tendency to malignant degeneration.7

Possible histopathology

The nail beds showed marked hyperkeratosis. As in the normal nail bed, there was no granular layer. Oral histopathology would show thickening of the oral epithelium with extensive intracellular vacuolization. The connective tissue under the epithelium might show plasma cells and lymphocytes. The lesion is benign and should be left untreated.8

Possible treatment and ramifications

According to Butterworth and Stenman,9 sufferers of this disease usually have average intelligence. Many are able to walk with great difficulty and discomfort because of the blebs and thick calluses on the soles. Among associated findings have been atrophy of the tongue, abnormally long bones, twinning of the incisor teeth, and excessive mobility of the joints. Treatment with vitamin A has been recommended, although not universally successful. It is administered orally, 100,000 units daily, and applied as ointment to the beds of nails which have fallen off or been surgically removed. A successful surgical procedure involves removal of the nails, followed by thorough scraping of the matrix to prevent regrowth.

SUMMARY

A case of pachyonychia congenita, with characteristic gross changes, has been reported. A review of the literature is also presented, detailing the changes seen with the condition.

REFERENCES

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Cornwells Heights, Pa. 19020

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Report of a toothbrushi

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Donald L. E:
DEPARTMENT:
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jaw. Only a few c:
ported. These occurr to the mucosa,* or a
A single case is brushing trauma. W

*Postdoctoral Stips Mexico (CONACYI), York University.
**Professor, Depart
***Associate Profes