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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 complicated by hidradenitis suppurativa: a family study

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SUMMARY

A family is described in which five of the six members with the Jackson-Lawler type of pachyonychia congenita also had varying degrees of hidradenitis suppurativa. We suggest an association between this type of pachyonychia congenita and hidradenitis suppurativa.

Pachyonychia congenita is a rare genodermatosis which is characterized by a variety of ectodermal abnormalities. An autosomal pattern of inheritance is usual. Two major and a number of minor forms are recognized. The Jadassohn-Lewandowsky or type 1 variety consists of pachyonychia, palmar and plantar hyperkeratosis, hyperhidrosis, a propensity for the development of friction blisters, follicular hyperkeratoses and oral leukokeratoses. Distinctive features of the Jackson-Lawler, or type 2, variety are the presence of natal teeth and multiple cysts on the trunk, axillae, neck and scalp which are thought to represent epidermoid cysts and steatocystoma multiplex. Oral leukokeratoses are not generally observed, but the other abnormalities as seen in the Jadassohn-Lewandowsky type may occur.

CASE REPORTS

Six members of one family were identified as having pachyonychia congenita (Fig. 1). Each affected member had thickened, discoloured nails (Fig. 2) and hyperkeratosis with fissuring of the feet from childhood, leading to pain and difficulty in walking. With the exception of a 12-year-old girl (Fig. 1, III.1) all had hyperhidrosis, blistering of palmar and plantar surfaces and multiple epidermoid cysts on the neck and scalp and multiple steatocystoma of back, arms, axillae and chest. Two family members (Fig. 1, III.1 and III.3) also had numerous periorbital milia and one member (Fig. 1, III.4) had two incisors present at birth.

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Five of the six affected family members gave a history of recurrent abscesses particularly in the axillae, groins and anogenital areas (Table 1). Three patients had evidence of scarring, nodules and sinus tract formation in the axillae. Although two male family members with pachyonychia congenita had a past history of mild facial acne, none of the females gave a history of androgen-related disorders.

Investigations
Biopsies of small cysts on the ear and arm were obtained from two members of the family. The histological appearance was consistent with the clinical diagnosis of epidermoid cysts. Patient III.4 (Fig. 1) developed an inflammatory lesion on the pinna which was excised and the
**Pachyonychia congenita and hidradenitis suppurativa**

**Table 1. Suppurative skin conditions and treatments of affected family members**

<table>
<thead>
<tr>
<th>Case</th>
<th>Sex</th>
<th>Age</th>
<th>Inflammatory skin conditions</th>
<th>Age at onset</th>
<th>Sites</th>
<th>Treatment</th>
<th>Examination</th>
</tr>
</thead>
<tbody>
<tr>
<td>III-1</td>
<td>F</td>
<td>53</td>
<td>Recurrent abscesses</td>
<td>30</td>
<td>Axillae, Anogenital</td>
<td>Antibiotics</td>
<td>No active supuration</td>
</tr>
<tr>
<td>III-2</td>
<td>F</td>
<td>21</td>
<td>Recurrent abscesses</td>
<td>17</td>
<td>Axillae, Groins</td>
<td>Antibiotics</td>
<td>No active supuration</td>
</tr>
<tr>
<td>III-3</td>
<td>M</td>
<td>23</td>
<td>Recurrent abscesses</td>
<td>17</td>
<td>Axillae, Groins</td>
<td>Antibiotics</td>
<td>Hypertrophic, tethered scars and hyperpigmentation</td>
</tr>
<tr>
<td>III-4</td>
<td>F</td>
<td>25</td>
<td>Recurrent abscesses, Inflamed epidermoid cyst, Discharging sinuses</td>
<td>19</td>
<td>Axillae, Groins, Scalp</td>
<td>Antibiotics, excision and grafting of axillary skin</td>
<td>No active supuration</td>
</tr>
<tr>
<td>III-5</td>
<td>M</td>
<td>31</td>
<td>Recurrent abscesses, Discharging sinuses</td>
<td>21</td>
<td>Axillae</td>
<td>Antibiotics, Isotretinoin</td>
<td>Sinus tracks and scarring</td>
</tr>
</tbody>
</table>

Histology was suggestive of an infected epidermoid cyst. One patient (Fig. 1, III.4) underwent excision and grafting of the axillary skin. Histological examination of the excised areas was consistent with the clinical diagnosis of hidradenitis suppurativa.

The serum levels of IgG, IgM and IgA were found to be within normal limits. The total neutrophil and lymphocyte counts were normal. No abnormality was detected in the numbers of T helper, T suppressor or natural killer lymphocytes or in the total number of B cells. A number of neutrophil functions were examined: random migration, chemotaxis, phagocytosis, and intracellular killing. No significant difference was noted between subjects and controls. Nail cultures were negative for fungi, yeasts and bacteria.

**DISCUSSION**

The family described here have the typical features of pachyonychia congenita of the Jackson-Lawler type. In addition, recurrent and chronic suppurative was a consistent feature in five of the six affected family members. The apocrine gland-bearing areas including axillae, anogenital regions and groins were particularly affected and varying degrees of scarring were observed.

An association between hidradenitis suppurativa and pachyonychia congenita has not previously been documented. Hidradenitis suppurativa is characterized by chronic suppurative and eventual scarring of apocrine gland-bearing skin. Apocrinitis associated with abscess formation and development of sinus tracks occurs. The primary event, however, is generally thought to be ductal occlusion by hyperkeratosis at the mouth of the apocrine follicle.1,2

Defects in systemic immunity are generally not thought to be important in the pathogenesis of
hidradenitis suppurativa, although it was recently noted that a small subgroup of patients with moderate and severe disease had reduced levels of T-helper cells. We found no abnormality in neutrophil numbers and functions or in total T and B lymphocyte numbers. The subsets of T lymphocytes and immunoglobulin levels were also entirely normal. This suggests that the apparent predisposition of these family members to recurrent abscess formation and hidradenitis suppurativa was unlikely to be attributable to a shared immune defect.

There are four possible explanations for the apparent association between pachyonychia congenita and hidradenitis suppurativa. Inflammatory change occurring in sebaceous glands located in the axillae and groins may lead to the development of hidradenitis suppurativa in these areas. This could account for the development of hidradenitis suppurativa in two patients described by McDonald and Reed, who had natal teeth, epidermoid cysts and sebaceous cysts multiplex but no other features of pachyonychia congenita. An alternative explanation is that the follicular hyperkeratosis of pachyonychia congenita may predispose to the development of hidradenitis suppurativa by causing occlusion of the mouth of apocrine follicles. This seems improbable, as most patients with pachyonychia congenita or other hyperkeratotic conditions do not develop hidradenitis suppurativa. The coexistence of the two conditions in this family may of course be due to chance. However, since familial hidradenitis is recognized, the fourth and most likely explanation is that a genetic linkage exists between the two conditions.

REFERENCES