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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 Jackson-Lawler type: a distinct malformation syndrome

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SUMMARY

A family with three members in two generations affected by pachyonychia congenita, hyperkeratosis and hyperhidrosis of the palms and soles, follicular keratosis, neonatal teeth and epidermoid cysts (Jackson-Lawler syndrome) is described.

The nosological autonomy of this condition is proposed and a further heterogeneity is suggested on the basis of histopathological changes in the subcutaneous cysts.

Pachyonychia congenita is a genetically determined ectodermal dysplasia inherited as an autosomal dominant trait. On the basis of phenotype analyses, Gorlin et al. (1976) proposed that two distinct entities could be recognized: the more frequent Jadassohn-Lewandowsky syndrome characterized by pachyonychia congenita, hyperkeratosis and hyperhidrosis of the palms and soles, follicular keratosis and oral leukoplakia, and the less frequent Jackson-Lawler syndrome (Jackson & Lawler, 1951), characterized by neonatal teeth and epidermoid cysts in addition to the same nail and skin abnormalities, but no oral leukokeratosis; both forms have never been observed in the same family.

We report a family in which three members in two generations were affected by the rarer form of pachyonychia congenita, and describe in detail the clinical picture of this rare malformation syndrome.

CASE REPORT

Case 1

The proband, a 21-year-old female and the second child of unrelated 39-year-old parents, was born at term following an uneventful pregnancy and delivery. Birth weight was 3.8 kg (90th percentile), and neonatal teeth were present. At one month of age, nail dystrophy of hands and feet was noted. Armpit folliculitis, and cysts in the metacarpal and elbow regions were observed at puberty. Growth and psychomotor development were normal, and scholastic performance was good.

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When she came under our observation, her weight was 56 kg (25th-50th percentile), her height was 160 cm (25th-50th percentile), and head circumference was 53 cm (10-25th percentile). Her hair was dry, bristly and sparse in the parietotemporal region; the eyebrows were very sparse. Except for malaligned teeth there were no oral abnormalities; she had a hoarse voice. Subcutaneous nodules adhering to the overlying normal skin were present on the left elbow (two nodules, 1 cm and 2 mm in diameter), on the dorsal surface of the proximal phalanges of the 3rd and 4th fingers of the left hand, and 2nd and 4th fingers of the right hand (several, 1-2 mm), and on the right knee (several, 1-2 mm). Follicular dermatosis was localized mainly on the back, armpits and pubic region. Palmoplantar hyperkeratoses with multiple foci particularly evident at the pressure points, and marked palmoplantar hyperhidrosis were also present.

The opaque, thickened nails of both hands and feet curved upwards, and were formed by yellowish grooved laminae; subungual keratosis was evident. Corneal dystrophy was not observed and fundus oculi was normal.

Histological examination of the elbow skin cyst disclosed a unilocular epidermoid cyst with encysted lamellated keratin (Fig. 1).

**Case 2**

The proband's mother was a 61-year-old housewife. She had had neonatal teeth, and nail dystrophy was observed during the first year of life. At our examination her weight was 61 kg, height was 165 cm, and head circumference was 54 cm (50th percentile). Her hair was thin and dry, and eyebrows were absent. She had no dysmorphic facial features nor oral or ocular abnormalities; her teeth were malaligned, but normal. Her voice was hoarse.

Several subcutaneous nodules and nodules adhered to the overlying normal skin on the elbows (from 0.5 to 2.5 cm in diameter), on the dorsal surface of the proximal phalanges of the 2nd and 3rd fingers of the right hand and the 1st and 2nd fingers of the left hand (1-2 mm), on the middle phalanx of the 3rd finger of the left hand (3 mm) (Fig. 2), on the knees and on the pretibial regions (0.5-2.5 cm).

Pustular folliculitis was present, mainly in the armpit and pubic regions; palmoplantar hyperkeratosis with multiple foci associated with marked palmoplantar hyperhidrosis were also
observed. The nails of hands and feet showed the same changes seen in the proband. Microscopic examination of one elbow cyst revealed a unilocular epidermoid cyst with encysted lamellar keratin.

Case 3
The proband’s sister had died thirty years previously at six months of age from pneumonia. She is reported to have had nail dystrophy since birth and neonatal teeth, but no skin cysts were observed.

Discussion
Gorlin et al. (1976) distinguished two different patterns of pachyonychia congenita; they called the association of pachyonychia congenita with palmoplantar hyperkeratosis and hyperhidrosis, follicular keratosis, neonatal teeth and subcutaneous cysts the Jackson-Lawler syndrome. However, McKusick (1983) in his catalogue, stressed that the genetic relationship between the two syndromes (no. 16720; no. 16721) is unclear. Goodman and Gorlin (1983) described the two syndromes as distinct, but they were doubtful regarding their legitimate separation; Smith (1982) considers the two syndromes as one entity.

Eleven families with one or more members affected by the Jackson-Lawler syndrome are described in the literature; members of the original family reported by Jackson and Lawler (1951) have also been described by Shrank (1966), and Besser & Moynahan (1971). The condition is inherited as an autosomal dominant trait with complete penetrance and variability of expression; only two sporadic cases have been reported (de Groot, 1966; Moldenhauer & Seidel, 1973). In the family we describe, three members in two generations were affected: the proband and her mother with full expressivity, while only the incomplete syndrome was present in the sister; although it must be emphasized that expressivity is, in part, age-dependent, i.e., skin cysts and keratosis are more evident in older patients. In the nine families described, more than 50 affected members are reported, but in only a proportion of these were clinical details
included. The most frequent clinical findings are thickened nails, cutaneous cysts, palmoplantar hyperkeratosis and hyperhidrosis, neonatal teeth, and dry thin hair. Other anomalies such as a hoarse voice and corneal dystrophy are less frequently described; leukokeratosis of mouth and tongue has not been reported previously.

Histological examination of the subcutaneous cysts has disclosed epidermoid cysts in three previous cases (Soderquist & Reed, 1968; Besser & Moynahan, 1971; Boxley & Wilkinson, 1971) and in two of three affected members of the family we describe; steatocystoma multiplex was detected in 6 cases (Vineyard & Scott, 1961; de Groot, 1966; Engel & Pinzer, 1966; Ionescu, Wolfshaut & Cernaianu, 1968; Moldenhaver & Seidel, 1973; Hodes & Noris, 1977). In two families no histological examination of the cutaneous cysts was performed (Sertoli, 1949; Schonfeld, 1980).

The concomitant presence of ectodermal dysplasia and subcutaneous cysts, with no mucous membrane involvement in probands or their affected relatives, strongly supports the hypothesis that a single autosomal dominant gene with complete penetrance and age-dependent expressivity is responsible for the pachyonychia congenita syndrome of the Jackson-Lawler type.

The histopathological difference in the skin cysts suggests a further genetic heterogeneity in this syndrome. For this reason, histological examination of subcutaneous cysts should be performed in all affected family members in order to ascertain whether histopathological heterogeneity between and within families exists.

REFERENCES


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