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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: A REPORT OF SIX CASES IN ONE FAMILY

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WITH A NOTE ON LINKAGE DATA

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The term pachyonychia congenita was coined by Jadassohn & Lewandowsky (1906) to describe a particular type of congenital dysplasia of the nails and skin. Since then about fifty cases of this condition have been described in the American and European literature, and the cutaneous features are now sufficiently well defined to warrant their separation, as a distinct entity, from other similar ectodermal dysplasias.

The distinguishing feature, which is common to all cases, is the condition of the nails, which are affected symmetrically. The base of the nail appears normal, but the free margin is raised by a thick, horny mass. In addition, most of the recorded cases have shown changes in the skin consisting of follicular keratosis, affecting mainly the extensor surfaces of the fingers, xeroderma with plantar and palmar hyperhidrosis; and hyperkeratosis and the formation of bullae on the soles of the feet. There is often leukokeratosis of the tongue and buccal mucous while a few cases have shown abnormalities of the cornea and malformations of the teeth. The clinical picture is thus one of widespread dyskeratosis.

The description which follows is of a family, five of whose members were examined recently at the Hospital for Sick Children and were found to have the features of this disease together with two other associated abnormalities. A sixth member, deceased, is considered from the family history to have suffered from the same condition. The pedigree of this family is shown in Text-fig. 1. The condition is illustrated by the photograph (Pl. 1).

CASE RECORDS

The maternal grandfather, I. 2. At birth he was thought to have had some teeth present. All nails were thickened from an early age, and he suffered from blisters on the soles of his
Hands of case of pachyonychia congenita (III. 1).
He had many small cysts, probably sebaceous, in the skin. He is said to have been normal in other respects and died some years ago at the age of 72 from a lung abscess.

The mother, II. 2. This woman was also born (16 July 1915) with teeth, but the exact number is not known. All her nails were thickened by the age of 1 month, although they had appeared to be normal at birth. She is frequently inconvenienced by whitlows which develop at the sides of the nails. From the age of about 5 years she has had a dry skin except on the palms and soles, where it is excessively moist. At this time she developed reddish spots on the limbs which have tiny greasy plugs in the centre. For many years she has had patches of thickened skin on the soles of the feet accompanied by painful blisters in the hot weather. When she was about 12 years old several small cysts appeared in the skin of her face and neck. These cysts sometimes burst and exude a cheesy material.

On examination all her nails showed the typical appearances of pachyonychia. On the undersurface of their distal ends were thick, hard, yellowish brown masses which caused them to project upwards at an angle of about 30° from the nail-bed and produced an accentuation of their normal lateral convexity. The dorsal surfaces of the nails were smooth but slightly discoloured. The skin of the palms was moist, and there were patches of hyperkeratosis on the soles of the feet. On the extensor aspects of the limbs there were many reddened follicles plugged with tiny sebaceous horns (i.e. follicular keratosis), and there were several small sebaceous cysts in the skin of the face and neck. The mouth, tongue and teeth were normal.

The eldest child, III. 1. A girl, who was born at full term (22 February 1938) with eight teeth in the lower jaw. These teeth were not very firmly fixed and only the cutting edges seemed normal, the remainder being soft and translucent, 'like gristle'. They had all fallen out by the age of 6 weeks and were replaced between the sixth and eighth years by a normal second dentition. The rest of her teeth were normal and had erupted at the normal times.

The nails appeared to be normal at birth, but by the age of 1 month they had all become deformed and have remained so. The extreme hardness of these nails renders manicure a problem, and although some of them have been removed surgically they have grown again as mightily as ever. She, too, suffers from recurrent paronychia.

Her skin has always been dry except on the palms and soles, where it is unusually moist. By the age of 5 years she, too, had developed thickened skin on the soles of the feet with painful blisters in the hot weather. During the first year of life she had developed some small red spots on the elbows and knees, and recently several cysts had appeared on her face.

On examination she was a healthy girl with normal hair, teeth and eyes. There were no lesions on the tongue or elsewhere in the mouth. All her nails showed exactly the same features as have been described in the mother. The palms and soles were particularly moist, although the rest of the skin was dry. On the soles of the feet there were patches of hyperkeratosis with some broken vesicles on the heels and under-surfaces of the toes. Some small sebaceous cysts were present on the face and neck, while on the extensor surfaces of the limbs there was a typical follicular keratosis. There were some freckles present on the forehead. Apart from these lesions there were no abnormal physical signs.

The second child, III. 2. This boy was born (11 November 1940) with four teeth in the lower jaw which had fallen out by the age of 4 months and which had shown the same characteristics as those of his sister. His further history, too, was substantially the same as hers.
On examination there were no sebaceous cysts, but to a lesser degree all the other features of familial abnormality were present, and apart from some freckles on the face he was otherwise normal.

The third child, III. 8. Another girl who was born (7 January 1942) with four teeth in lower jaw. Here again the history was similar, but an additional feature in this sibling was symptom of hoarseness of the voice which had been present since infancy. Repeated laryngoscopy during previous years had shown only thickening of the posterior commissure of larynx.

On examination she also showed similar defects, although her skin lesions were only slight; there were no sebaceous cysts. There were an unusual number of freckles on the face.

The fifth child, III. 5. This was a small boy, born 31 December 1943, who had had four in the lower jaw at birth and they had fallen out only a few weeks later. His nails were affected by the second month, and he has recently developed the red spots on the limbs.

On examination his primary dentition was complete except for the lower incisors, which were missing. His nails were severely affected and there was quite extensive follicular keratosis of the arms and legs and at the back of the neck. He had no hyperhidrosis or hyperkeratosis of the soles and there were no sebaceous cysts. His voice had a slight, but definite, hoarse quality, but there were no other abnormalities.

The fourth, III. 4, and sixth, III. 6, children, born 4 June 1947 and 22 September 1951 respectively, had no teeth present at birth and showed no abnormalities whatsoever. The father, II. 1, and the two maternal aunts, II. 3 and 4, were also normal, healthy individuals. All the affected siblings showed a marked physical resemblance to their mother, although the normal child of 3 did not. The infant is as yet too young for her appearance to be compared with that of the rest of the family. None of the remaining members of the family was born with teeth. This fact and the normality of their skin and nails are vouched for by the mother, who is a reliable witness. Her two nephews, III. 7 and 8, are aged 7 and 5, so that any potential defect in these children would have become obvious by now had they been affected.

**DISCUSSION**

Pachyonychia congenita is not to be confused with hereditary onychogryphosis, in which lesions of the nails are similar (Videbaek, 1948). It will be seen from the illustration, however, that the appearances of pachyonychia are sufficiently distinctive to render differential diagnosis simple. The condition is inherited as a dominant character, and in the cases quoted by Cockayne (1933) there was a preponderance of males of Slav origin, but this has not been so in all reported since then.

There are only two other families on record in which the abnormality has been found in more than two generations. Kumar & Loos (1935) record the pedigree of a family in which, out of some seventy members in five generations, no fewer than twenty-three had both the nail and skin lesions of pachyonychia. None of these had either erupted teeth at birth or sebaceous cysts. Murray (1921) described a family in which the affected individuals showed both thickened nails and erupted teeth at birth but no skin lesions. In this family there were four out of five siblings affected as well as their mother, a maternal aunt, and the maternal grandfather.

The familial incidence of sebaceous cysts is rare, but Stauffer & Simmons (1942) give an account of a family showing both sebaceous cysts and a dystrophy (quite different from pachyonychia) of the nails of the great toes.
The presence of erupted teeth at birth is unusual in itself, but according to Cockayne its occurrence in more than one member of a family is extremely rare. It is usual for such teeth to be abnormal in appearance and to fall out at an early age. The association of prematurely erupted teeth with various types of dystrophy of the nails has been recognized previously (Cockayne, 1933; Gates, 1948).

The family described in this paper would appear to be particularly unusual. It displays all the common features of pachyonychia congenita, with the exception of leucokeratosis oris, together with premature eruption of teeth at birth and multiple sebaceous cysts arising at puberty. The hoarseness of the voice, which was noted in two of the siblings, was present in several members of the family recorded by Kumer & Loos and also in the case described by Radda & Marill (1948). It is interesting to note that in the latter case laryngoscopy was performed and showed thickening of the posterior commissure of the larynx.

Cockayne suggests that the abnormality of the skin and nails in pachyonychia depends on the presence of two genes, neither of which alone causes any defect, and Gates (1946) ascribes the combination of nail dystrophies with premature eruption of teeth to a possible gene linkage. These theories appear to be unnecessarily complicated, and it would seem more probable that the multiple defects in this family and the lesions of pachyonychia generally are due to a single gene.

**Summary**

The features of pachyonychia congenita are described.

An account is given of a family in which six members were affected by this disease; they also had erupted teeth at birth and sebaceous cysts developing at puberty.

Some points regarding the genetics of the condition are discussed.

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