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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.
Section of Dermatology

President F R Bettley FRCP

Meeting February 17 1966

Cases

The results from sweat distribution tests on these heterozygous females were consistent with a quantitative defect of sweat glands and this is suggestive evidence of tissue mosaicism as predicted on the inactive X-chromosome theory (Lyon 1961). Confirmation of this assumption requires more precise quantification of active sweat glands.

Full details of this family will be published elsewhere (Kerr et al. 1966).

REFERENCES

Pachyonychia Congenita

Alan B Shrank MRCP (for E J Moynahan FRCP)
(Hospital for Sick Children, Great Ormond Street, London)

N W, girl aged 4
Born with six 'gristly' teeth in the lower jaw, which were shed after a month. When she was 10 days old all her nails began to thicken (Fig 1). Her scalp hair became coarse and sparse. At the age of 3 follicular hyperkeratosis appeared on her elbows and knees, and occasional bullae developed on her soles. In September 1965 her left knee became painful and swollen.

Several relatives (Fig 2) developed the same stigmata though the number of teeth present at birth varied from four to eight; in addition, at puberty many cysts appeared all over the body.

Fig 1 Pachyonychia congenita: thumb nails
On examination: All the nails were wedge-shaped with a nearly normal proximal part, a thick free edge and a smooth surface. There was sparse coarse scalp hair and follicular hyperkeratosis of the elbows and knees. The lower central incisors and molars were absent but the rest of the primary dentition appeared normal. The eyes were reported normal and sweating was normal. The left knee was stiff and the adjacent muscles wasted.

Her mother (III.1) and grandmother (II.3) exhibited the same lesions as well as a normal permanent dentition and many cysts of all areas, suggestive of sebaceous cysts. Investigations: Hb 66%; ESR (Westergren) 39 mm in 1 hour; latex test positive; ANF test weakly positive. Chest radiograph normal. Biopsy of knee-joint showed rheumatoid changes.

Comment

This family was first reported by Jackson & Lawler (1951), and the propositus is the first affected member of the fourth generation. The syndrome manifested in this family differs from that first described by Jadassohn (1906) as leukokeratosis oris lacking in addition to the characteristic nail dystrophy, follicular hyperkeratosis and bullae on the soles, there are other features such as erupted teeth at birth, a hair disorder and multiple cysts from puberty.

There appears to be some variation from family to family in the clinical pictures reported as pachyonychia congenita, while within each family there is little or no variation. The autosomal dominant mode of inheritance shown by this disorder is explicable on the basis of a single gene, but it is difficult to understand how a single aberrant gene could be responsible for such interfamilial differences. It may be that closely linked genes are responsible or that the gene is different in each family.

REFERENCES

Jackson A D M & Lawler S D (1951) Ann. Eugen. 16, 142
Jadassohn J (1906) Verh. dtsch. Ges. 9, 381

Dr P R Montgomery: I saw a case like this at Guy's Hospital. She had follicular hyperkeratosis, thickened nails, blisters on the soles, and these cysts. The histology was that of sebocystomatosis with the sebaceous glands stretched out over the cyst wall. The cysts of Dr Shank's case look clinically like those of sebocystomatosis.

Dr R S Wells: From the pedigree of Dr Shank's case, and from others in the literature, I would think the condition is due to a single pleiotropic gene inherited as a mendelian dominant trait.

Bullous Ichthyosiform Erythrodermia

R S Wells MD (for C D Calnan FRCP) (Institute of Dermatology, London)

Y C, girl, aged 4

History: It was reported that during delivery the skin was stripped off giving the appearance of a macerated stillbirth. She lost a great deal of fluid, was treated with tetracycline and cortisone, and recovered. Later she developed the appearance of a collodion baby, which was followed by widespread ichthyosis. She still has marked bullae formation from time to time. Sweating has been noted in the axillae and groins.

Family history: Several members of her family are affected and the pedigree is compatible with an autosomal dominant mode of inheritance (Fig 1). On examination: There is extensive gross hyperkeratosis with some sparing of the lower arms and lower legs. The palms of the hands and soles of the feet show thickening and there is marked hyperkeratosis of the elbows and knees. The lower part of the face is more severely affected than the upper. The mother shows a very similar appearance and distribution of her lesions.

Histology: Histology from the mother is typical for bullous ichthyosiform erythrodermia (Dr E Wilson Jones).

Comment

When Brocq first described ichthyosiform erythrodermia he noted that a minority had bullae. I have examined nine families with bullous ichthyosiform erythrodermia with more than one affected member and there is no doubt that this form is inherited as an autosomal dominant, whereas the non-bullous is an autosomal recessive trait. Even if the patients are only mildly affected, the histology of the bullous form is diagnostic, with vacuolation and cavitation in the upper layers of the epidermis. The non-bullous form shows changes similar to those seen in sex-linked ichthyosis, with an increase in thickness of the upper layers of the epidermis.

Generalized Hyperkeratosis - ? Ichthyosis Erythroderma. Psoriasis

R S Wells MD (for F Ray Betley FRCP) (St John's Hospital for Diseases of the Sk, London)

E J W, woman, aged 53

History: This patient was told that she of her skin all her life. She has further blistering episodes, but has always been markedly ichthyotic with keratosis of the flexures. For the last six years she has had psoriasis.

Family history: She is of short stature and now dead, had dry, flaky skin all his life. His flexures were also the most severe areas. In addition he had gross hyperkeratosis of the palms and soles. There are no other members in the family and there is no sanguinity.

On examination: Generalized mild ichthyosis uniform hyperkeratosis of the palms and rather warty lesions on the buttocks and dorsum of the feet. There is smooth hyperkeratosis of the wrists, antecubital fossae, axillae and groins.