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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.
SEBOCYSTOMATOSIS WITH CONGENITAL PACHYONYXIA

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Sebocystomatosis or steatocystoma multiplex is a genodermatosis transmitted as an autosomal dominant trait and is defined by the presence of numerous epidermal cysts widely distributed throughout the body. Pachyonychia congenita is also a hereditary condition, characterized by the thickening of the distal part of the nail and associated with other keratotic lesions. Few cases of the coexistence of the two conditions have been reported,1-2 but the presence of epidermal cysts has been noticed in patients with pachyonychia.3 4 5

Here we report the clinical and hereditary findings in three families, which through several generations have had sebocystomatosis associated with congenital pachyonychia. Some comments are made on the nature of the lesions and the possible genetic relationship between the two conditions.

The three families (Fig. 1) are from the east region of the Department of Antioquia, Republic of Colombia. Some of the patients were studied at the Instituto Colombiano de los Seguros Sociales. We were able to study their cases personally; through them the others were located.

Clinical Examinations

The lesions of sebocystomatosis varied in number and size among patients and developed most rapidly during puberty. In some cases they were few, small and inconspicuous, while in others they were large (3 cm) and confluent. Cysts were located on the forehead, in the retroauricular region, on the chest, neck, back, genitalia and proximal portions of the extremities (Fig. 2, 3). Occasionally a cyst would open and drain a creamy material.

The nail lesions could be observed at birth; nails were hyperconvex, dark and thick (Fig. 5). Later the free edge of the nail became thick, irregular and fragile. Both fingernails and toenails were affected, but in every case two or three fingernails or toenails remained free of lesions.

The children of families BB and Z, according to their parents, were all born with teeth (Fig. 7), usually central incisors. These deciduous teeth were shed by the second or third month, and were replaced in the seventh year when the second dentition started.

The following tests were performed: complete blood count, serology, serum cholesterol, blood glucose and chest X-rays. All of the results were normal except for the serum cholesterol of one member of family BB, which was 367 mg per cent (normal 180-230). Examinations for fungous infections in several cases gave negative results. Several histologic studies of the cysts showed an inner layer of malpighian epithelium. In some cases, sebaceous glands were present in the walls of the cysts (Fig. 6). No hair or sweat glands were observed. One of the children of family BB had keratotic plus in the abdomen (Fig. 7).

Comments

The nature of the cysts of sebocystomatosis is not clear. They have been considered retention cysts.6 However, some investigators refute this theory by pointing
out the lack of relationship between sebocystomatosis and comedons. Some authors give them a neoplastic origin of nevoid type, while others classify them as hamartomas because of the presence of hair follicles, sebaceous glands and sweat glands in the cyst wall in direct continuity with its epithelium. Pinkus in comparing the epidermoid cysts and the trichofoliculoma, considered the former as derived from the upper part of the follicle and the trichoemric one from its medial portion.

Others suggest a defect in lipid metabolism as a causative factor. We have not attempted to analyze this in our study, and in only one individual was the serum cholesterol level above normal.

Pachyonychia congenita was described in 1906 by Jadadson and Lewandowsky, who noticed its incidence in families. Kummer and Loos, according to the symmetric and genotypic lesions, and Type I, II and Type III: lesions. In normality, presence of the finding present in normal, not have a.

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Fig. 1—Pedigrees of Families BB, OO, and Z. (In family OO, there were 6 abortions.)

Fig. 2—Lef
and Loos,\textsuperscript{12} tracing the condition through five generations, identified three types according to the predominant lesions: Type 1, symmetrical keratosis of palms and soles and generalized follicular keratosis; Type 2, lesions of Type 1 plus leukokeratosis oris; and Type 3, lesions of Type 1 plus corneal lesions. Intestinal polyposis and skeletal abnormalities have been reported\textsuperscript{4,5} and the presence of erupted teeth at birth is a common finding.\textsuperscript{13} This last abnormality was present in two of our families and was also present in members of a family who did not have nail lesions (Fig. 7).

The three pedigrees reveal autosomal dominant transmission for both conditions, with predominance for neither sex. Pachyonychia was not present in all family members. It varied among individuals: thus, the nail changes were present only in three members of family OO, three members in family BB, and most of the descendents in family Z. The nail lesions could be observed prior to one year of age in fingers and toes, but generally not all fingernails or toenails were affected.

Several genetic possibilities could explain the association of the two conditions. One of them is that the association is due to a single gene with pleiotropic effects manifested in skin, nails, intestines, teeth and bones\textsuperscript{4,5} with symptoms varying among individuals. Another possibility suggests the existence of two separate genes,\textsuperscript{13, 14, 15} one for each entity, but linked on the same chromosome. A statistical analysis of more cases would be necessary to prove this hypothesis.

Pedigrees of families BB and OO show consanguinous marriages. In case 1, both
this case the high frequency of affected chil-
dren could be explained by the possibility 
that the mother was a carrier. A cross be-
tween two heterozygotes carries with it the 
possibility of 50 per cent heterozygous, 25 
per cent pathologic homozygous, and 25 per 
cent normal homozygous. Since the homo-
yzous state could be lethal for the fetus, 
this could account for the high number of 
abortions in the consanguinous marriage 
in both families, six in family OO and three 
in family BB.

Summary

A study of the three families, in which 
the association of sebocystomatosis and 
pachyonychia congenita was found in sev-
eral individuals, is reported. The two con-
ditions are transmitted as autosomal dominant 
traits and two possibilities are considered for 
their coexistence: that they are due to a 
single gene with pleiotropic effects, and that 
they are transmitted by two genes linked 
on the same chromosome.

Résumé

On décrit trois familles dont la sebocystoma-
tose et la pachyonychie congenita étaient présen-
dans plusieurs membres. Ces deux conditions 
sont héréditaires comme des traits dominants; il 
'y a deux possibilités pour expliquer leur coex-
istence: le deux sont due à un seul gène qui a

Fig. 8—Lesions of sebocystomatosis in the genitalia.

parents had sebocystomatosis and the 
mother had pachyonychia. In family OO, 
only the father was affected with sebocysto-
matosis and the mother was normal. In

Fig. 4—Lesions of pa-
chyonychia in adult and 
child.

Fig. 5—Hist opy of t

Fig. 6—E
acy of affected child by the possibility of a carrier. A cross between heterozygous, and 25 per cent, since the homozygous fetus, he high number of ignitary marriages, family OO and three

des effects pleiotropic, ils sont transmis par deux gênes lies dans le meme chromosome.

Resumen

Se presentan los hallazgos clínicos y hereditarios de tres familias en las que se asocian las lesiones sebocistomatosas con paquionichia congénita. Tal asociación puede explicarse de dos maneras: las dos condiciones clínicas son producidas por un solo gene con manifestaciones pleiotrópicas, cada una de las alteraciones es producida por un gene diferente pero ligados en el mismo cromosoma.

Referencias

2. Nasemann T, Bandmann HJ: Sebocystomatosis associated with epidermolyis bullosa simplex which at present is quiescent. XIII Congressus Internationalis Dermatologie, 1967