Pachyonychia Congenita Project

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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.

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Natal Teeth and Steatocystoma Multiplex Complicated by Hidradenitis Suppurativa

A New Syndrome

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• A new syndrome, consisting of natal or defective teeth, or both, steatocystomas of the skin, and epidermal cysts of the scalp, is described in several generations. One member of the family had eruptive molars. Male-to-male transmission suggests autosomal dominant inheritance. This syndrome should be separated from pachyonychia congenita I and II.

(*Arch Dermatol* 112:1132-1134, 1976)

Pachyonychia congenita is divided into two groups of disorders, both autosomal dominant in inheritance. The first (type I) involves the congenital thickening of the nails, oral leukokeratosis, palmar-plantar keratosis and hyperhidrosis.[1-4] The second (type II) also has congenital thickening of the nails, hyperhidrosis, and palmar-plantar keratosis (sometimes with blistering), plus steatocystomas of the body, epidermal cysts of the scalp, but no mucosal involvement or premature dentition.[5-10]

We report a new syndrome of epidermal cysts (mainly steatocystomas) and premature dentition in a family (Fig 1). The condition resembles pachyonychia congenita, except for the absence of nail involvement.

REPORT OF CASES

CASE 1.—A 58-year-old woman had been born with two lower incisors and with her body encased in a membrane described by her mother as shaped like waxy grains of rice that washed off with soap and water. During infancy, the patient had multiple small cysts on her face and body. She had rickets from questionable malnutrition at age 1. She presently has many cysts on the underside of her forearms, all around her head, breasts, trunk, neck area, and on the back of her thighs. Occasionally, a cyst erupts and becomes inflamed, but she denies having had hidradenitis suppurativa. She says that cysts have been removed from all over her body, and that they were called steatocystomas. Her nails and hair were normal. She has refused biopsies and photography.

CASE 2.—A 30-year-old man, the son of patient 1, had numerous "cysts" on the chest, back, face, scalp, axillae, groins, and arms since early puberty. He had had numerous surgical procedures for "infected cysts" in the axillae, and had been told that he had hidradenitis suppurativa. The cysts scattered about his trunk and head were described to him as "wens", but a biopsy was never performed.

This patient had a high birth weight, and had two molars at birth. His face was covered with scattered small white lesions during infancy.

Physical examination showed a well-developed man with numerous steatocystomas of the trunk, arms, and head (Fig 2 and 3). There were scars from surgical intervention in both axillae and in the groins. His nails, teeth, and hair were normal.

Biopsy of a cyst on his head revealed an epidermal cyst, and a biopsy of a lesion on his chest confirmed the diagnosis of steatocystoma (Fig 4). In some areas, there was abnormal collagen in the walls of the cyst, with an apparent attempt to produce hair follicles.

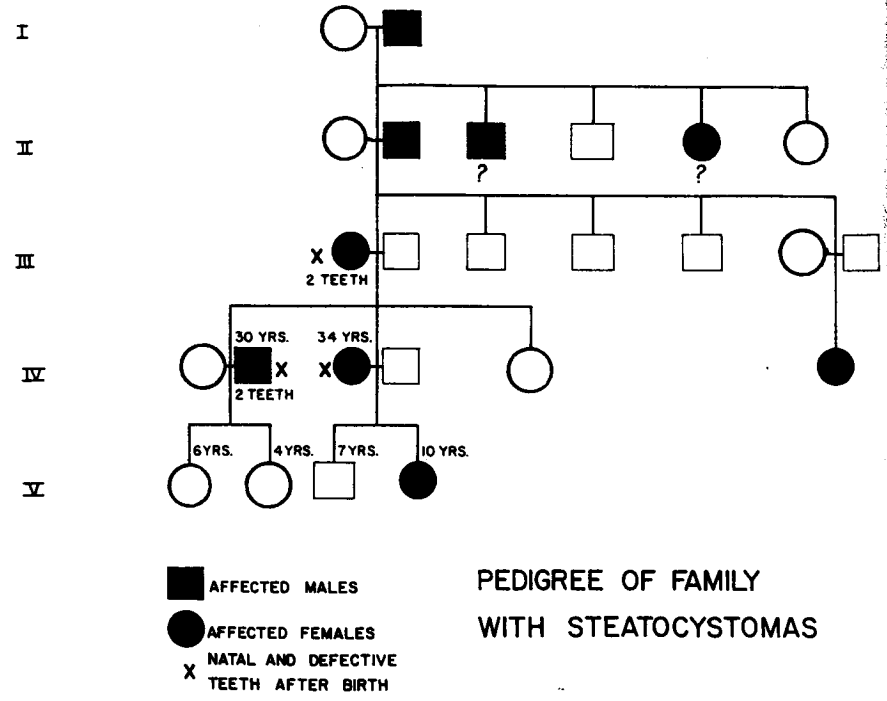
CASE 3.—A 34-year-old woman, the daughter of patient 1 and sister of patient 2, also has had numerous "cysts" of the chest, back, axillae, groins, face, and scalp since early puberty. Biopsy had never been performed. She has had numerous surgical procedures in the groin and axillae for hidradenitis suppurativa.

She had a normal birth weight, but at birth was covered "with tiny cysts all over her body," according to her mother. She was not born with teeth, but her dentition was abnormal, and by age 1 year she had to have four pitted and cracked anterior teeth removed.

Physical examination showed a normally developed woman; the upper part of the chest and back, face, and scalp were covered with scattered steatocystomas. There was evidence of surgical intervention in her axillae and groin, and there was a large, oval, red sterile abscess in her right groin, which was clinically typical hidradenitis suppurativa. Her nails, hair, and teeth appeared normal.

A biopsy of a chest cyst revealed a typical steatocystoma. No biopsy of the "wens" of her head were performed (Fig 5

Fig 1.—Genetic chart showing autosomal dominant inheritance in five generations.



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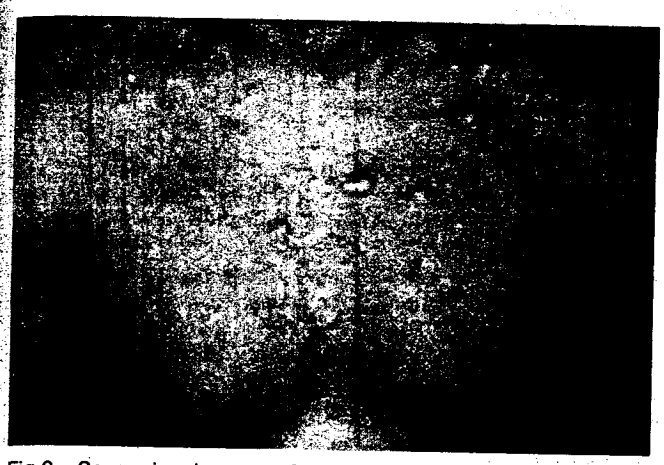

Fig 2.—Severe involvement of steatocystomas on chest of patient 2.

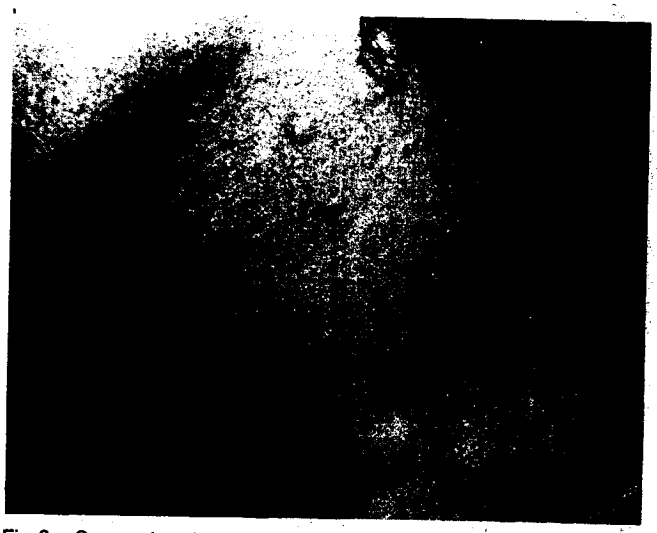

Fig 3.—Severe involvement of steatocystomas on neck of patient 2.

and 6). Her affected daughter was not examined.

COMMENT

Natal or neonatal teeth are rare, with an incidence of about one in 3,000 births.[11] Type II pachyonychia congenita has been reported with natal and neonatal teeth, but always without mucosal involvement. All such patients had epidermal cysts and palmar-plantar keratosis, and some had severe hyperhidrosis and blistering of the palms and soles. This type must be separated from the better known type I pachyonychia congenita.[1-4]

The first report of congenital anomalies of the nails associated with erupted teeth at birth was by Murray in 1921.[5] He delineated a three-generation history of pachyonychia congenita, with four of five siblings in one generation having several erupted incisors at birth. The age of onset of steatocystomas varied from 8 months to 20 years.

Jackson[6] reported six cases of pachyonychia congenita in one family spanning three generations, with each affected patient having dyskeratosis of the nails of the hands and feet and erupted teeth at birth. Three members had epidermal cysts of the face and neck.

Vinegard and Scott[7] reported eight cases in four generations with steatocystoma multiplex and pachyonychia congenita. Their skin lesions were on the body and scalp. A diagnosis of steatocystoma multiplex was not confirmed histologically, since no sebaceous glands were connected with the cystic structures. The cystic nodules on serial sections did not communicate with the surface or with the pilosebaceous apparatus. An autosomal dominant inheritance was not proved, since male-to-male transmission was not observed.

Fig 4.—Typical histopathological picture of steatocystoma with sebaceous glands lining the cyst wall (hematoxylin-eosin, original magnification ×40).

Soderquist and Reed[8] reported three patients in three generations. The youngest had moderately severe hyperhidrosis with epidermolysis bullosa of the nondystrophic type. The hair was dry, broken, and twisted, and appeared by microscopic examination to be similar to puli torti. However, with age, her hair has improved. Biopsy specimens from the scalp of all three patients revealed only epidermal cysts. None of these patients had mucosal involvement, but all three had teeth at birth.

Boxley and Wilkinson[9] reported six individuals in four generations with both natal and neonatal teeth and abnormal hair—unruly hair and eyebrows that stuck straight out. The histopathologic features of the nodules of the chest consisted of multiple epidermal hamartomas (sebaceous gland cells, abnormal hair follicles haphazardly in the dermis with irregular collagen bundles). These findings are more consistent with the two biopsy specimens from the chests of our patients 2 and 3.

Two of the clinical aspects of these families have a hereditary basis, namely, natal teeth, neonatal teeth, or both,[11] and steatocystoma multiplex.[12,13] Steatocystoma multiplex was observed by Noojin and Reynolds[12] in

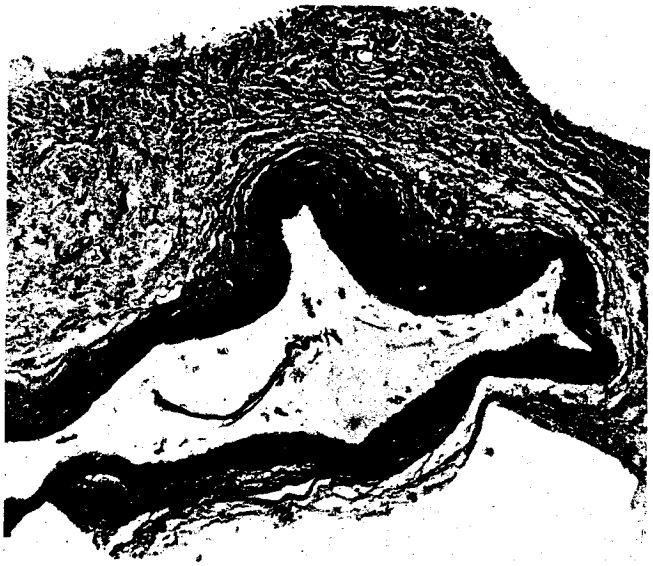

Fig 5.—Steatocystoma showing lining of sebaceous gland cells with projection into lumen (hematoxylin-eosin, original magnification ×50).

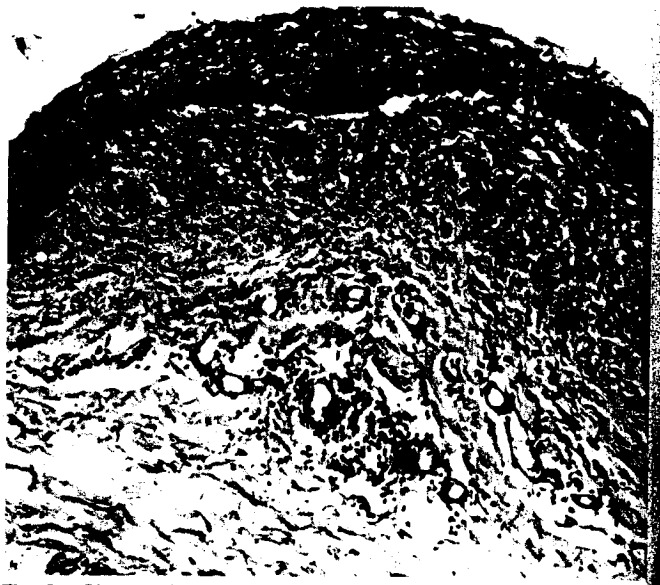

Fig 6.—Closer view of projection showing increased fibrosis (hematoxylin-eosin, original magnification ×200).

12 individuals in three generations. In typical cases, the patient may exhibit 100 to 200 round or oral cystic tumors widely distributed on the back, anterior part of the trunk, scrotum, and thighs. Stephens (1959)[13] reported sebaceous cysts, clinically "wens", in a very large number of individuals in five generations in a definite pedigree.

Natal or neonatal teeth are also found in two other conditions besides type II pachyonychia: chondroectodermal dysplasia or Ellis-van Creveld syndrome, which is autosomal recessive, and oculomandibular dyscephaly with hyperhidrosis, or Hallerman-Strieff syndrome, which is a questionable hereditary disorder.[11,14]

Ellis-van Creveld syndrome is characterized by bimanual ulnar polydactyly, chondrodysplasia of the long bones resulting in dwarfism, hidrotic ectodermal dysplasia that principally affects the nails, teeth, and hair, and less often, causes heart malformations.[15] The Hallerman-Strieff syndrome includes dyscephaly with a parrot nose, mandibular hypoplasia, proportionate nanism, hypotrichosis, blue sclerae, and congenital cataracts. Natal or neonatal teeth may also be found in association with such developmental anomalies as cleft palate, cleft lip, and cyclopia.[11]

Hidradenitis suppurativa has, as far as we know, no hereditary basis, but severe acne vulgaris does occur in families. The presence of hidradenitis suppurativa in patients 2 and 3 is probably a coincidental complication, but it might be an inherent part of the syndrome, and the lesions may be inflamed steatocystomas.

We regard the association of natal teeth and steatocystoma multiplex as a new autosomal dominant genetic disorder. It resembles the pachyonychia congenita disorders only in part, and can be readily differentiated by the lack of nail involvement. However, in a recent article, type I pachyonychia congenita with natal teeth has been reported in a large Swedish family.[16] This is in variance with Moldenhauer and Ernst,[17] who analyzed 96 cases of type I pachyonychia congenita and found that 96.8% exhibited nail lesions, 72% had palmar-plantar keratosis, 58.5% had follicular acneiform keratosis, and 54.4% had oral leukokeratosis but no natal teeth.

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