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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 With Epidermal Cysts and Other Dyskeratotic Lesions

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Three individuals representing three successive generations of one family are presented demonstrating pachyonychia congenita involving multiple nails of hands and feet, epidermal cysts, callosities of feet with blistering, hyperhidrosis, unusual hair texture, and erupted teeth.

**PACHONYCHIA CONGENITA**, first described by Jackson and Lowenstokowsky in 1908, is a rare disorder transmitted as a single mendelian dominant characteristic with incomplete penetrance. Its association with other hereditary dyskeratoses, such as planar and palmar callosities, bullae, keratotic plaques, ichthyosis, hyperhidrosis, leukokeratotic oris, and steatocystoma multiplex, has been well documented. Murray first noted the correlation between the presence of erupted teeth at birth and pachyonchla congenita. The association of this condition with steatocystoma, however, is very rare. Jackson reported six cases in one family spanning three generations, with each affected member demonstrating dyskeratosis of the nails of the hands and feet and erupted teeth at birth. Three members had epidermal cysts of the face and neck.

Three individuals representing three generations of the same family were recently studied. Each of the three patients had a remarkably similar clinical picture (Fig 1).

**Report of Cases**

Case 1—A 55-year-old woman has had thickened and distorted fingernails and toenails since 1 year of age, callosities with frequent "blisters" on her feet, hyperhidrosis, and multiple epidermal cysts of the neck and upper chest since age 13. She has no history of any similar problems in either parent or sibling. She was born at term and delivered vaginally without any complications. Her prenatal care was uneventful, and there were no significant illnesses in her childhood. She has had no major illnesses since childhood.

Fig. 1.—Pachyonychia congenita of three generations: feet of patient 1, top, 2, center, and 3, bottom.
This Baby Born
With Six Teeth

Fig. 2.—Newspaper clippings representing individuals of three generations with erupted teeth at birth.

10. The finger nails had been surgically removed but the thickened nails subsequently recurred. In 1938 she was operated upon for a fistula in the distal third of the descending colon. Biopsy exams and upper gastrointestinal (GI) studies in 1935 revealed a large diverticulum of the duodenal loop. Diverticula of the descending colon, and an anastomosis of the fourth lumbar vertebra with the partial formation of an extra vertebra. She reportedly had three teeth erupted at birth (Fig 2).

Pusillium examination revealed marked thickening and friability of the distal half of the first three finger nails of both hands and all the toenails of both feet. Calculiaries along the borders of the great toes were present. Multiple firm yellowish round to oval nodules, up to 1 cm in diameter and attached to the overlying skin, were noted scattered over the neck and upper chest. Skull x-ray films in February 1967 revealed a fine outline of the frontal bone. Bumps of masses varied from the anterior chest revealed multiple cystic lined with stratified squamous epithelium and filled with loose keratin arranged in laminated layers, consistent with the diagnosis of epidermal cysts.

Case 2.—A 25-year-old man, the only son of patient 1, was noted to have six erupted teeth at birth. Symmetrical thickening of the finger nails and toenails was also noted during the first year of life. Nodular cysts of the faeces, chest, and back developed after adolescence and were worsened with adolescence, and were prone into adulthood. During childhood he was plagued by recurrent calculiaries of the lower bunion. These calcallaries have developed on the weight-bearing areas of the feet.

Moderate dystrophy of all nails with prominent thickening of the toenails was evident by physical examination. Soft cysts, nodules up to 2 cm in diameter and attached to the overlying skin, were particularly prominent on the fingers but widely distributed over the face, neck, chest, and back (Fig 3). Hair x-ray films in February 1967 were normal in appearance.

This patient, the youngest son of patient 2, had six teeth erupted at birth. Dystrophic thickening of all finger nails appeared in the first year of age. The development of calcallaries along the borders of the great toes, the frequent eruptions of blisters, and hyperhidrosis of palms and soles were early complaints. She was initially diagnosed as having epidermal nodules of the acanthoma type. The patient’s mother reported that the child was an agable hair that has improved with age. The patient’s 8-year-old daughter is not affected.

Physical examination revealed marked

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Fig 5.—Large epidermal cysts of patient 2 present at age 1 and age 10; no significant dental abnormalities were noted.

Skinfold infoldable nails of the hands and feet and callos formation along the borders of the eustachian tube. The patient's blonde scalp hair was unusually dry, lusterless, and kinky. Petalium hyaline veinmout and cultures on Sab- raud medium were negative for pathogenic fungi. Skull x-ray films in February 1967 were normal.

Comment

Pachyonychia developed in all five patients prior to age 1, and both adult patients reported the onset of epidermal cysts just prior to adolescence. In one recent study of eight individuals involving four generations, the time of onset of the stenocystomi- um varied from eight months to 20 years. The dystrophic nails are known to have several common distinguishing features: geometrical involvement, distal free edge mottled by thick keratogenous debris, and a relatively normal base. Although the presence of hyperhidrosis, callosities, and frequent blistering of the feet were uniformly found among the patients described in this report, other dyskeratosis involving the mucous membranes and eyes were not.

The formation of bullae on the feet from irritation and hyperhidrosis lends first to a diagnosis of epidermolysis bullosa of the nonstaphylococcal type. The inheritance in such cases is also autosomal dominant, and these patients may also demonstrate calcification and hypophosphatemia.

The abnormalities dry, kinky hair of the scalp of patient 3 was of interest. Very similar findings were recently reported by Shosne's. Teeth present at birth which fall out at four to six weeks, as well as other malformations of dentition, are part of the hereditary dermatoses.

The relationship of diffuse GI polyposis and ectodermal changes involving the skin and nails has been recently reported. In our first case, the findings of a congenital diverticulum of the small intestine, cutaneous blebs of the frontal bone, and the epidermal cysts initiated a possible association with Gardner's syndrome. Multiple polyposis was ruled out, however, by x-ray films. Skull x-ray films of patients 2 and 3 and the brain enema study of patient 2 were within normal limits.

Biopsies of the cysts revealed a histologi- cal picture of epidermal cysts, not of steno- cystomi- um. The designation of stenocystomatis with pachyonychia congenita is thus probably incorrect.

References