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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Cameo

Pachyonychia Congenita with Tuberous Sclerosis

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A 7-year old girl who had irregular thickened nails since birth presented with a 4-year history of recurrent headache, effortless vomiting associated with slow rhythmic, convulsive movements of the right arm, head, and neck. Examination revealed marked nail plate thickening with subungual hyperkeratosis, lifting the distal end of the nail plates and involved all the twenty nails of hands and feet (Fig. 1). The soles showed hyperkeratosis of the pressure areas and the medial border of the great toes. In addition, there were multiple, skin-colored, acuminate follicular papules scattered over the extensor surface of the elbows, knees, and the buttocks (Fig. 2). There were multiple 2–4 mm brown papules over the chin, nose, and right cheek (angiofibromas) and a shagreen patch over the left nasolabial fold (Fig. 3). The fundus showed phakomas in the supranasal and temporal quadrants of the right eye. Hair and mucous membranes were normal. Systemic examination did not reveal any abnormality. Her intelligence was subnormal (IQ, 65) and there was no family history of epilepsy, mental retardation, and nail dystrophy.

Laboratory studies and x-rays of the head and trunk were normal. The electroencephalogram (EEG) showed features of generalized seizures with localization to left frontal, central and parietal cortex. The histopathology of the brown papule over the cheek was consistent with angiofibroma, and that of the plaque over the left nasolabial fold, with shagreen patch. The histologic examination of the follicular papules on the elbow showed hyperkeratosis, follicular plugging, and mild acanthosis of epidermis. There was mild perivascular lymphomononuclear infiltrate in the upper dermis.

Comments

Pachyonychia congenita is a rare genodermatosis, chiefly affecting the boys.[1] It is usually transmitted by an autosomal dominent gene,[2] however, a recessive pattern of inheritance also has been described.[3] This condition is characterized by prominent nail changes, but may be associated with palmoplantar hyperkeratosis, follicular keratosis, steatocystoma multiplex, and leukokeratosis of mucosae. There may be patchy alopecia,[4] prenatal teeth, hair abnormalities, and corneal leukokeratosis.[5] Pachyonychia has been reported in association with multiple hamartomas,[6] steatocystoma multiplex[2-4] and cutaneous amyloidosis,[7] all inherited in an autosomal dominant manner. Tuberous sclerosis is also an autosomal-dominant inherited disorder that is commonly manifested in the skin and the central nervous system, but affects several other systems as well.[1] It has been reported in association with number of other diseases, such as neurofibromatosis, Struge-Weber syndrome, cutis verticis gyrata, partial albinism, mucoviscidosis, situs-inversus, familial infantile myoclonic epilepsy, arthrogryposis multiplex congenita, coloboma of iris, and catatonic schizophrenia.[8]

The association of these two autosomal dominant

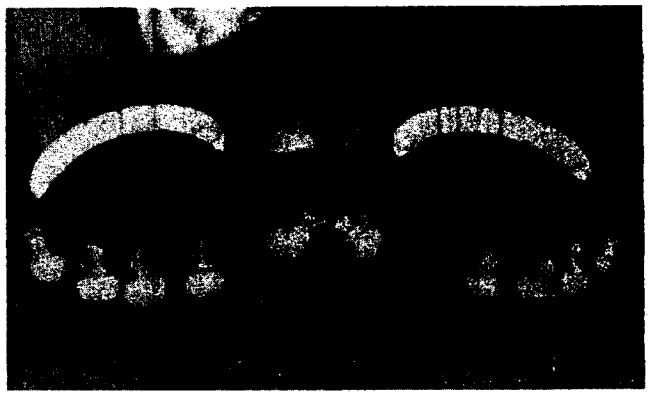

FIG. 1. Fingernails showing thickened nail plate and subungual hyperkeratosis.

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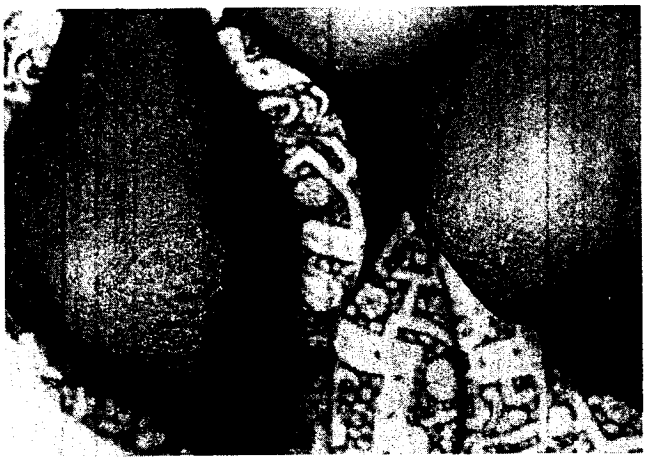

FIG. 2. Acuminate follicular papules over the elbows.

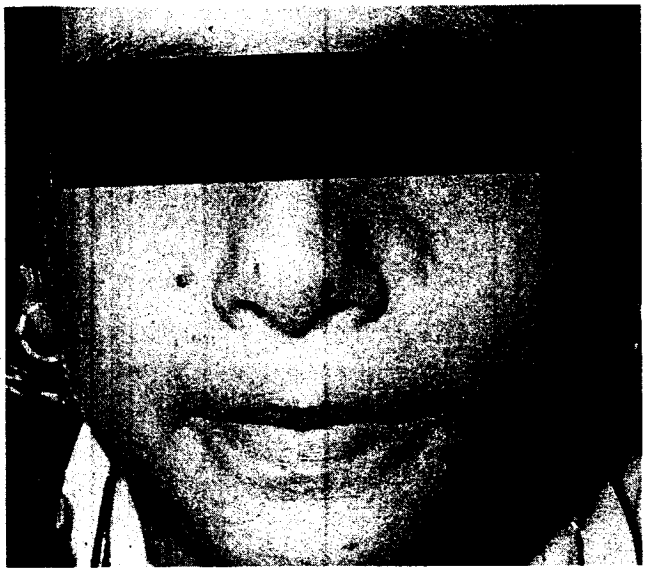

FIG. 3. Face showing angiofibromas and shagreen patch.

genodermatoses has not been noted previously; however neurofibromatosis, cutis verticis gyrata, and partial albinism, all autosomal dominant disorders, have been found to be associated with tuberous sclerosis.[8] Similarly, steatocystoma multiplex and cutaneous amyloidosis, both autosomal-dominant inherited disorders have been reported with pachyonychia congenita.[2-4] There is no apparent causal relationship between these two conditions, and their coexistence is perhaps incidental. It may have resulted from simultaneous mutation at two different loci because there was no family history of either tuberous sclerosis or pachyonychia congenita in our patient.

Acknowledgment

Dr. B. D. Radotra, M.D., Lecturer, Department of Pathology, Postgraduate Institute of Medical Education and Research, Chandigarh interpreted the histopathology specimens.

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Hypersensitivity to Allopurinol

A syndrome of severe toxicity in patients following allopurinol intake has been described. Main features include generalized pruriginous exanthema, fever, liver dysfunction, eosinophilia, and renal function impairment. Most patients had previous mild renal failure and were receiving a thiazide diuretic.

Two patients with clinical criteria for this syndrome are presented. One patient was a woman with previous renal failure who was receiving thiazide diuretic. She developed the clinical picture following allopurinol intake. The second patient was an extremely obese woman with hyperventilation syndrome who developed severe toxicity after receiving the drug. A cutaneous biopsy specimen taken from this patient disclosed leukocytoclastic vasculitis.—*Martin RL, Cuchet MJO, Bauza FM, Sebastian FV, Diez LI. Severe hypersensitivity caused by allopurinol ingestion: report of two cases. Actas Dermatol Sif 1987;78:511–514 (Spanish). Submitted by Yehudi M. Felman, M.D., Brooklyn, NY.*