ANHIDROTIC ECTODERMAL DYSPLASIA WITH PALMO-PLANTAR KERATODERMA

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Summary

A patient with anhidrotic ectodermal dysplasia and palmo-plantar keratoderma is presented. Palmo-plantar keratoderma is an unusual association with this disease. Atopic dermatitis was another associated condition in this patient.

KEY WORDS: Anhidrotic ectodermal dysplasia, Palmo-plantar keratoderma, atopic dermatitis.

Anhidrotic ectodermal dysplasia (AED) is characterised by partial or complete lack of eccrine sweat glands and other cutaneous appendages, by dental anomalies, and by other genetically transmitted congenital defects[1].

Most patients are males (more than 90% of the 300 reported cases), a sex-linked (x-linked) recessive pattern being the most common mode of transmission[1]. The responsible gene appears to exert its inhibitory effect at about the third month of embryonic life, when appendageal differentiation is well under way.

Clinically, patients with AED present with characteristic facies showing high cheek bones, prominent supra-orbital ridges, saddle nose, large conspicuous nostrils, thick muscular upper lip and prominent chin. Scalp hair is sparse with decreased pigmentation. Skin and eyes also show poor pigmentation. Dentition may be defective especially the incisors, and canines are conical[2]. There may be onychodystrophy but palmo-plantar keratoderma (PPK) is rarely seen. Very high incidence of atopic dermatitis has been noted. Mental retardation may be present. Life expectancy is usually normal.

Case Report

A 9 years old male child from a Sikh family of Muktsar, Punjab, presented with the complaints of generalised weakness, and itchy lesions in the cubital and popliteal fossae. Parents had noticed absence of sweating over the entire body and intolerance to heat since the time of birth. At the age of 2 months, patient had severe itching and developed erythematous papules in the cubital and popliteal fossae. Similar lesions appeared subsequently on the neck, lips and eyelids. The PPK was also observed at an early

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age. Voice was hoarse but there was no dysphagia. There was complete alopecia till 2 years of age, when sparse, brownish hair were first noticed on the scalp. Gradually the hair growth improved, though it remained sparse all along. The two upper incisors developed at 3-4 years of age and upper premolar at 5-6 years of age.

Examination showed a young boy of normal intelligence, whose growth appeared to be retarded. His facies was characteristic with frontal bossing, saddle nose, prominent nostrils, protruding and fissured lips, radiating furrows at the buccal commissures and large ears. Skin was soft, smooth, shiny and devoid of hair. It was wrinkled periorbitally and presented a prematurely aged appearance. Circumoral and circumorbital skin was hypermelanotic. Scalp showed partial alopecia. Eyebrows and eyelashes showed very sparse hair (Fig. 1). Well defined, dull red erythematous lichenified plaques were present at the flexural areas especially popliteal and cubital fossae (Fig. 2) PPK was uniform with a well defined border (Fig. 3). Nails were normal. There was no polydactyly.

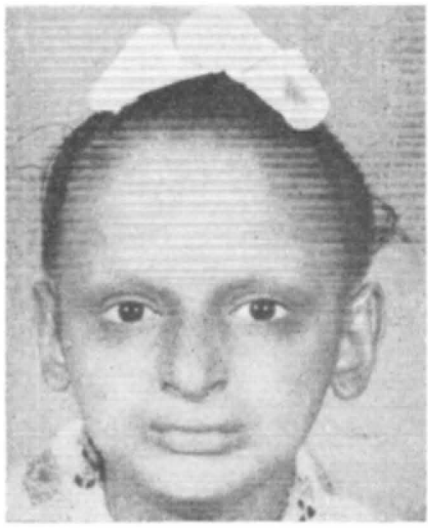

Fig. 1 Showing characteristic facies

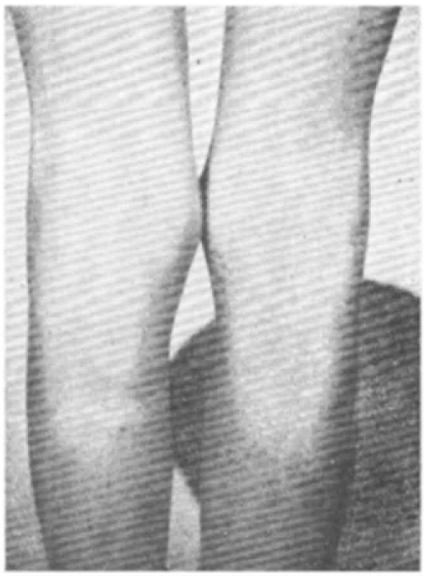

Fig. 2 Showing lichenified plaques over popliteal fossae

Two conical upper incisors and premolars were seen. Genitalia was normal.

Investigations

Routine blood, urine and stool tests were normal. Blood VDRL, Blood sugar values, Serum cholesterol, X-rays of chest and skull were normal. Skin biopsy showed atrophy of the epidermis. Sweat glands, sebaceous glands and hair-follicles were absent.

Discussion

AED is a rare familial disease generally affecting male children and transmitted mostly as a sex linked recessive trait. Rarely an autosomal recessive inheritance is also seen when females may be affected. Our patient had an affected male relation, his maternal aunt's grandson having the same disease. Amarjeet Singh et al[3] reported AED in a female from a Sikh family.

The association of AED with PPK is very rare. Thickening of skin of palms and soles is an unusual feature in hidrotic ectodermal dysplasia, an autosomal

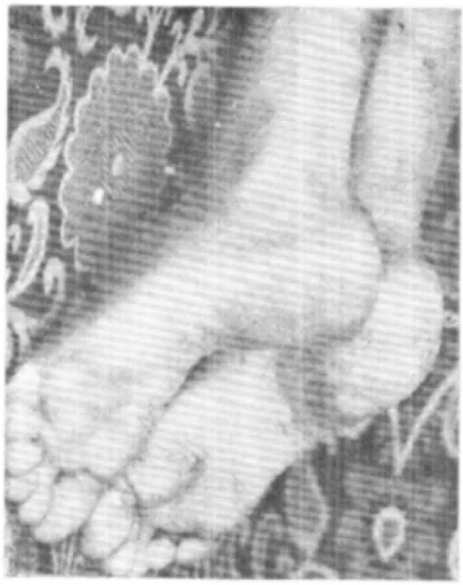

Fig 3 Showing uniform and well marginated plantar keratoderma

dominantly inherited condition. PPK in association with AED has been reported by Indian authors[3,4]. Such an association has been documented in a whole family by Gudin et al[5].

Onychodystrophy occuring in almost half of the patients was absent in our case.

Atopic dermatitis is reported to be a common association of anhidrotic ectodermal dysplasia. Such an association was evident in our patient who had a chronic skin problem from the age of 2 months and exhibited characteristic skin changes in the flexures at the time of the examination.

Approximately 40% of AED patients are mentally retarded[6]. Kermal[7] described one case of AED with idiocy. In our patient, the intelligence was considered to be normal.

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