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ANHIDROTIC ECTODERMAL DYSPLASIA

MPS Sawhney, CH Gidwani, K Nagendra and SD Rajpathak

Two sisters had classical features of anhidrotic ectodermal dysplasia. The mode of inheritance in these cases seems to be autosomal recessive which is a very rare occurrence.

Key words : Ectodermal dysplasia, Anhidrotic ectodermal dysplasia.

Anhidrotic ectodermal dysplasia is a genetic disorder first recognised in 1848 by Thruman as suggested by Reed et al.[1] Prior to this, Wedderburn had studied a hindu family with 10 affected males in 1838. But his study was not published till 1875 when Charles Darwin included it in his classic book, "The variation of animals and plants under domestication".[1] The condition is characterised by the triad of anhidrosis, hypotrichosis and hypodontia and manifests itself principally by attacks of hyperpyrexia.[2] There may be associated endodermal abnormalities in the form of absence of submucous glands throughout the respiratory tract. Many other associated defects include, sensorineural deafness, corneal opacities, corneal dysplasias, congenital cataract, mental deficiency, short stature and atopy.[3]

Case Reports

Case 1

A 7-year-old girl was getting hospitalised repeatedly with intolerance to heat, hyperpyrexia and repeated chest infections since birth. She was third of the five children in her family, and had two brothers and two sisters. Case 2 is her younger sister. There was no history of consanguinity in the family. Clinical examination revealed height 102 cm (5th percentile), weight 12 kg (54% of the expected weight) and chest circumference 49 cm. General physical and systemic examination including CNS were normal. Dermatological examination revealed sparse, thin, hypopigmented and short scalp hair with patches of alopecia over the temporal region. Body hair were scanty with paucity of eyebrows and eyelashes. Ears were low-set but there was no conductive or sensorineural deafness. Incisors and canine teeth were widely spaced and peg shaped (Fig. 1). Forehead was square and prominent with flattened nasal bridge and thick everted lips. Periorbital skin was wrinkled with hyperpigmentation around

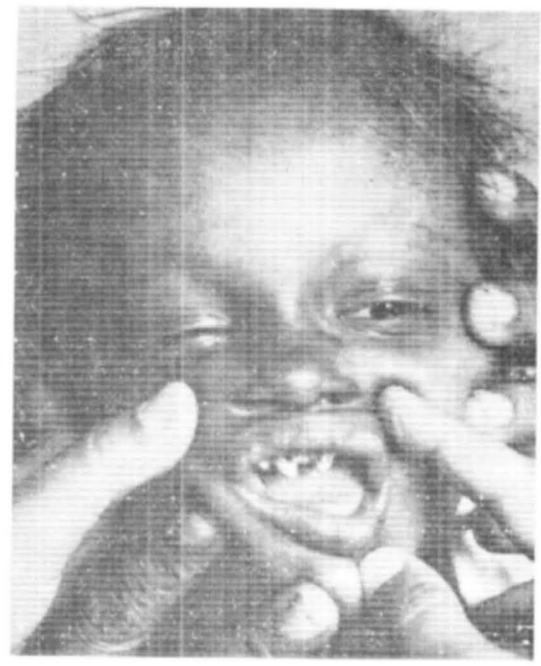

Fig. 1. Scanty scalp hair, paucity of eyebrows, prominent square forehead, flattened nasal bridge, thick everted lips and peg-shaped teeth.

From the Departments of Dermatology and Venereology and Paediatrics, Command Hospital (SC) Pune-411 040, India.

Address correspondence to : Maj MPS Sawhney.

the eyes and over the nasal bridge. Skin in general was thin, wrinkled, pale and anhidrotic with hyperkeratosis of the soles. Nails, external genitalia, nipples and mental development were normal. Routine urine, haematological parameters, serum proteins and skiagrams of skull were within normal limits. X-ray chest showed prominent broncho-vascular markings. Skin biopsy showed normal epidermis with no cutaneous appendages including sweat glands.

Case 2

A 3-year-old girl, younger sister of case 1 had similar complaints and similar clinical features. Her height was 81 cm (25th percentile), weight 7 kg (5th percentile) and head circumference 46 cm. Routine urine, haemotological parameters, serum proteins and fundus were within normal limits.

Examination of father, mother and brothers revealed no abnormality.

Comments

This condition has been thought to be an X-linked recessive disorder affecting predominently the males.[1] Female carriers may have reduced sweating and faulty dentition. Since some cases of anhidrotic ectodermal dysplasia affecting females also started appearing in the literature, some authors suggested that a mutant gene was transmitted as X-linked conditional dominance.[2] Lyon[4] postulated inactivation of either the maternal or paternal X-chromosome in transmission of X-linked abnormalities. Once a particular X-chromosome is inactivated, all descendants of that cell have the same inactive X-chromosome. This inactivation occurs in a random fashion, so that a vast majority of the females have a mixture of active paternal or maternal X-chromosome in their cells. A few females however, will have cells with an overwhelming number of paternal X-chromosome or reverse. If the normal X-chromosome is the one which is largely inactivated, the X-chromosome containing the mutant gene will be present in the majority of cells in that individual and will produce an affected female. The mutant gene may therefore be expressed partially or wholly depending on this proportion.

When two or more females in one family were affected by the disease, it was thought that the disease is transmitted also as an autosomal recessive disorder,[5] since the chance of a new dominant mutation occurring twice in one generation of one family is negligible. Reports have also appeared of male patients inheriting their disease as autosomal recessive disorder.[6,7]

This report of two sisters of classical anhidrotic ectodermal dysplasia favours autosomal recessive mode of inheritance though no history of consanguinity is available. Repeated chest infections in our patients point towards some abnormality of the sero-mucous glands of the respiratory tract as has been reported in many cases.[1]

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