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ANHIDROTIC ECTODERMAL DYSPLASIA- AUTOSOMAL RECESSIVE FORM

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Anhidrotic ectodermal dysplasia with classical features in 2 sisters is reported. The mode of inheritance in these seems to be autosomal recessive; which is a very rare occurrence.

Key Words : Anhidrotic ectodermal dysplasia, Autosomal recessive.

Introduction

Anhidrotic ectodermal dysplasia (AED) is a rare genetic disorder characterized by faulty development of ectodermal structures, resulting most notably in anhidrosis, hypotrichosis and abnormal dentition.[1] This condition has been thought to be an X-linked recessive disorder affecting predominantly the males.[2]

In the present communication AED as autosomal recessive form is reported, which is very rare occurrence.

Case Report

Two sisters born to consanguineously married parents aged 8 years and 5 years were referred to skin OPD from paediatric department for repeated history of intolerance to heat since their early childhood. Cutaneous examination of both sisters revealed - sparse, thin, hypopigmented and short scalp hair with paucity of eyebrows and eyelashes. They had square faces with prominent frontal bossing, prominent supraorbital ridges, pointed chin, flattened nasal bridge and thick everted lips. Ears were lowset and anteriorly displaced. Periorbital skin was wrinkled with hyperpigmentation. Skin in general was thin, dry, pale and finely wrinkled. The incisors and canine teeth were peg shaped and widely spaced (Fig.1). Palm, sole and nails were normal. External genitalia, nipples and mental development were normal. Other systemic examination including CNS were normal. Other siblings were normal.

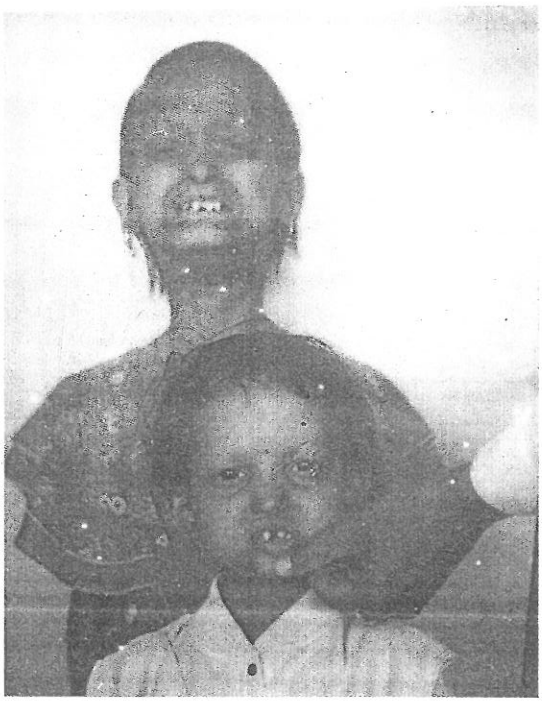

Fig. 1. Typical AED facies with peg shaped incisors & canine teeth.

Routine urine, haematological parameters, skiagram of skull and chest were within normal limits. Skin biopsy showed normal epidermis with no cutaneous appendages. A diagnosis of AED was made.

Comments

A number of cases of anhidrotic ectodermal dysplasia (AED) have been

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reported in which autosomal recessive (AR) inheritance of the syndrome seems probable.[3,4] There are as yet no known clinical features by which this form of the disease can be differentiated from the sex linked recessive form, except that the complete syndrome occurs in both sexes.[4] Sawhney, et al[5] from India reported 2 sisters with AED, which favours an AR inheritance, though no history of consanguinity was available. Infact, consanguinity is one of the chief historical markers of recessive inheritance[6] as in the present case. Their explanation is that - when two or more females in one family were affected by the disease, it was thought that the disease is transmitted also as an AR disorders, since the chance of a new dominant mutation occurring twice in one generation of one family is negligible.[7]

It has been claimed that in autosomal recessive form of AED sweat glands are reduced in number but are not absent.[8] For want of facilities study of sweat pores was not done in the present case, hence the above view neither be contradicted nor supplemented, though biopsy finding in the present case and case reported by Sawhney et al[5] showed absence of sweat glands.

This report of two sisters with comple... syndrome of AED favours AR inheritance ... history of consanguinity available.

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