ANHIDROTIC ECTODERMAL DYSPLASIA WITH PECTUS EXCAVATUM, MITRAL VALVE PROLAPSE AND PALMO-PLANTAR KERATODERMA

C Balachandran, BC Ravi Kumar, Munish Paul, L Sabitha

Anhidrotic ectodermal dysplasia (AED) is a rare x-linked recessive disorder characterised by the triad of hypotrichosis, anodontia and anhidrosis. Here we report a case of AED with associated features of pectus excavatum, mitral valve prolapse and palmoplantar keratoderma.

key words: Ectodermal, Pectus, Mitral prolapse

Introduction

Anhidrotic ectodermal dysplasia (AED) is an inherited disease, mode of transmission by an x-linked recessive gene. The hallmark of the disease is absence of sweating, resulting in fever and heat intolerance associated with various other defects. We are reporting a patient with AED who had associated features of palmo plantar keratoderma, mitral valve prolapse and pectus excavatum.

Case Report

A 23-year-old male born of a non consanguineous marriage presented with inability to tolerate heat, complete absence of sweating, absence of eyebrows, eyelashes, pubic hair and hair over the trunk and limbs since birth. He had sparse hairs over the scalp, moustache, beard and axillae. Patient had no history of having temporary teeth and subsequently had abnormal dentition, developed few permanent teeth at 9 years of age. He also had difficulty in chewing food and complained of occasional chest pain. There were no complaints of epistaxis, sexual dysfunction, atopy or abnormal behavior. No other family member had similar complaints.

Examination revealed sparse hairs on the scalp, bossing prominent supraorbital ridges,prominent chin and diffuse hyper pigmentation over the face. (Fig 1). Eyebrows and eye lashes were absent. Axillary hairs were sparse and there were no hairs on the body. Plams and soles showed keratoderma. Nails, eyes and ears were normal. Dental examination (fig.2) revealed conical teeth in place of incisors and canines; retained deciduous molars; permanent first molar and all other teeth were missing. There was no alveolar ridge in the lower jaw. Apart from these findings the chest showed a precardial bulge and hollowing of the lower one third of the presternal area. Systems were within normal limits. Skin biopsy

From the Department of Skin and STD,
Kasturba Medical College, Manipal - 576 119, India.
Address correspondence to:
Dr C. Balachandran

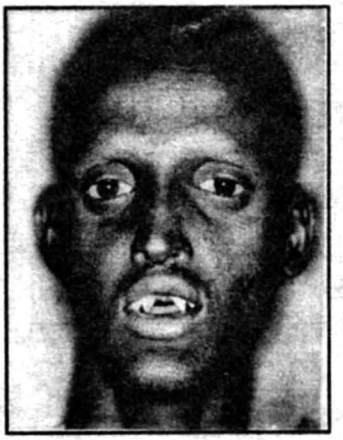

Fig. 1 - Typical facies of AED with abnormal dentition and hypotrichosis.

Fig.2 - Conical teeth

showed complete absence of sweat glands and was consistent with the diagnosis of AED. Echocardiography revealed mitral valve prolapse.

Discussion

AED also known as Christ-Siemens-Toraine syndrome is characterized by partial or complete absence of sweat glands, hypotrichosis and hypodontia associated with a host of other defects such as sensorineural deafness, corneal opacities, congenital cataracts, mental deficiency, mitral valve prolapse, cleft lip and cleft palate.[1] The only skeletal abnormality described so far in AED to our knowledge is kyphosis of lower cervical and upper thoracic spine.[2] Though AED is inherited predominantly by X-linked mode, a few cases with autosomal dominant mode of inheritance have been reported.[1] However as none of the other family members were affected in our case, it is difficult to ascertain the exact mode of inheritance in our patient. Monitoring the subsequent generations may throw some light in this regard.

References

1. Rapp RS, Hodgkin WE. Anhidrotic ectodermal dysplasia: autosomal inheritance with palate and lip anomalies. J Med Genet 1968;5: 269- 272.

2. Shah KC, Umrigar DD. Unusual cutaneous manifestations of anhidrotic ectodermal dysplasia; a case report. J Dermatol 1990 ;17:380-384.