Ectodermal dysplasia, anhidrotic

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Abstract :

Male, 8 years of age, presented with heat intolerance since early infancy. Examination revealed dry skin, generalized scant hair, absent sweating and anodontia. Syndrome is characterized by the triad of partial or complete absence of sweat glands, anomalous dentition and sparse body hair. Multidisciplinary team approach is necessary to provide care to these syndromic children.

Keywords: *Anhidrosis, anodontia, hypotrichosis*

Introduction

Male, 8 years of age, presented with persistent low grade fever and failure of teeth eruption since early infancy. History of heat intolerance was present. Born of non-consanguineous marriage, younger of two siblings, 11 year old sister was unaffected. He was undernourished: Weight: 21.7 kgs, height: 118 cms, occipitofrontal circumference: 48cms, chest circumference: 52 cms, febrile 99°F, pulse 76 /min, respiratory rate: 20/min, BP: 100/60mm Hg. (Examination of Face [Fig.1, 2] Feet [Fig. 3]: Scalp hair: Sparse, unruly, lightly pigmented with

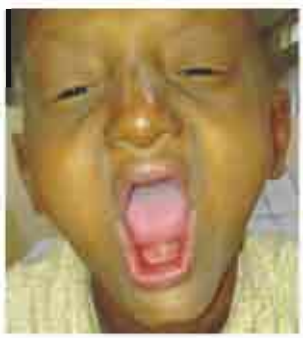

Fig. 1 : Examination of Face : Saddle nose, sparse eyebrows, absent teeth.

frontal bossing, prominent low set ears, flattened nasal bridge, a saddle nose, sparse eyebrows and lashes, wrinkled and hyperpigmented periorbital skin, malar hypoplasia due to **absent teeth** (anodontia), thick everted lips (eclabion) convex nails, **sparse hair** and **dry**

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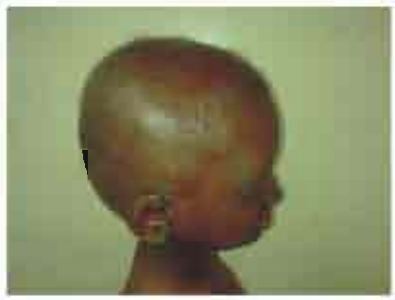

Fig.2: Examination of Head: Sparsehair, prominent low set ears

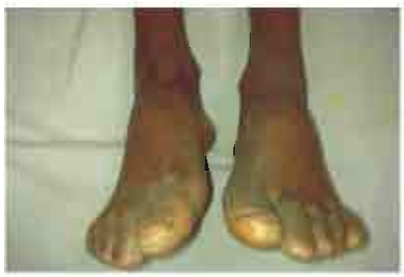

Fig. 3 : Examination of Feet : Lustureless scaly skin

lusterless scaly skin present over the entire trunk and limbs [Fig. 3]. Eye and ENT evaluations were within normal limits. Hematological investigations: Hb: 11.7 gm/dl, TLC: 8700/cumm, Platelet count: 2,06,000/ cumm. Biochemical investigations: Liver and renal function tests were normal. Histopathology: *Site:* Left palm hypothenar eminence. *Microscopy:* Only epidermis lined by stratified squamous epithelium with hyperkeratosis was seen. No dermis was present. Prosthodontics provided the dentures essential for nutrition and appearance. Parents were instructed to provide cool ambient temperature, plenty of fluids, especially so in hot weather, a nutritious diet, upgrade immunization and regular follow up.

Discussion

Ectodermal dysplasia anhidrotic (EDA) is diagnosed by the presence of the triad of hypotrichosis, hypohidrosis and hypodontia; with the characteristic facial appearance as seen in this boy. Born of noncosanguineous parentage with unaffected sister, suggests X linked recessive inheritance Type 1[1]. Unexplained fever and dry sloughing skin, helps in diagnosis in the neonatal period. Dental defects and heat intolerance in early childhood bring most of these patients to attention[2].

Incidence of EDA is 1-7:100,000. Risk of recurrence in patient's sibling and in patient's progeny mandates the need for genetic counseling[3]. Sex ratio of Male: Female is 1:1. Female carrier of X-linked recessive form has mild manifestations including abnormal sweat test but autosomal recessive carrier is not detectable[4]. Prenatal diagnosis by linkage analysis is possible as the EDA defective gene has been mapped as Xq12-q13.1. Differential diagnosis include hidrotic ectodermal dysplasia and ectrodactyly ectodermal dysplasia syndromes.

Treatments is restorative or preventive in nature and includes provision of dental implants, periodic removal of nasal secretions and ear wax, provision of hearing aids, ointment emoderm for dry skin, use of wig for enhanced appearance and artificial tears to prevent corneal damage[5,6].

Overall prognosis of EDA is good with respect to function, intelligence and lifespan. But high grade fever in childhood can result in permanent brain damage with consequent learning disability. In some cases morbidity can be due to presence of complications like respiratory and gastrointestinal infections because of poor development of mucous glands.

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