

Anhidrotic Ectodermal Dysplasia

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Abstract

Anhidrotic ectodermal dysplasia (AED) is a rare disorder characterised by a constellation of defects involving the teeth, skin and appendageal structures. We report a child who had typical features of this disorder.

Key Words

Ectodermal dysplasia, Anodontia, Hypotrichosis

Ectodermal dysplasia are a heterogeneous group of disorders characterized by a constellation of findings involving defects of two or more of the following: the teeth, skin and appendageal structures including hair, nails and eccrine and sebaceous glands (1,2).

Anhidrotic ectodermal dysplasia (AED) is a rare disorder also known as Christ-Siemens-Touraine syndrome (3). It is commonly transmitted as an X-linked recessive disorder. However, rarely autosomal recessive and autosomal dominant inheritance have also been seen (4). This syndrome manifests as a triad of defects: partial or complete absence of sweat glands, anomalous dentition and hypotrichosis (1). A recent review shows only seven cases of AED published in the European medical literature during 1970-2001 (3). In view of the rarity of this entity, we report a classical case of anhidrotic ectodermal dysplasia.

Case Report

A 2-year old male child first in birth order born to non-consanguineous parents presented with fever on

and off and intermittent cough since birth. Fever used to be high grade and more so during summer and would subside with medication. He had history of recurrent chest infections. On examination, the child was emaciated weighing 7 kg. He had sparse, light hair on the scalp while hair were absent on eye brows and eye lashes (Fig. 1). There was frontal bossing, depressed nasal bridge and malar hypoplasia. Wrinkled hypopigmented skin around periorbital region was noted. There was no teeth in the oral cavity. Skin was rough and dry with no sweat on it. Chest examination showed signs of consolidation on right side. Other systems were normal.

Laboratory examinations revealed hemoglobin of 11g/dl and white blood cell count of 8000/mm³, with 60% polymorphs and 40% lymphocytes and with normal platelets. Renal and liver function tests were normal. Urine analysis was normal. The ESR was 40 mm/hr. Chest X-ray revealed patch of pneumonitis in right upper and middle zone. Montoux test and gastric lavage for AFB were negative.

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In view of history of recurrent chest infections and fever, more so during summer months, facial features, anodontia, hypotrichosis, absence of sweat glands, the clinical diagnosis of anhidrotic ectodermal dysplasia was made.

In addition to adequate dietary rehabilitation, vitamin and mineral supplementation, antibiotics in the form of ampicillin and cloxacillin for two weeks were administered.

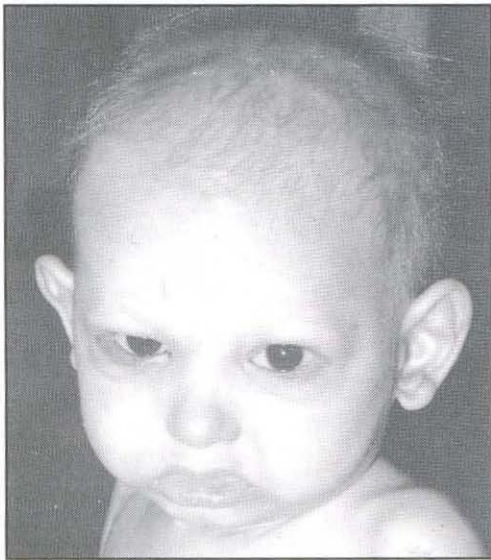


Fig : 1 Note the sparse hair on scalp, loss of eyebrows, lack of eyelashes and absent teeth.

Discussion

Anhidrotic ectodermal dysplasia, also known as Christ-Siemens-Touraine syndrome, commonly transmitted as an X-linked recessive disorder, is very rare. The complete syndrome occurs in males whereas females are carriers. However, autosomal recessive (3) and autosomal dominant (4) modes of transmission have also been reported.

Hypohidrotic (anhidrotic) ectodermal dysplasia (HED), skin disorder is particularly common in French-Canadian population of south-west Quebec. HED gene has been mapped to the pericentric region of chromosome 13q using linkage analysis in eight

French-Canadian families (6). This has also been mapped to the pericentric region of chromosome 13 q1. to a 2.4 cm interval flanked by markers D13S1828 and D13S1830 (7).

Hypohidrotic ectodermal dysplasia manifests as a triad of defects, partial or complete absence of sweat glands, anomalous dentition and hypotrichosis. Affected children, unable to sweat, may experience episodes of high fever in warm environment and may be mistakenly considered to have fever of unknown origin. The typical facies is characterised by frontal bossing, malar hypoplasia, a flattened nasal bridge, recessed columella, thick everted lips, wrinkled hyperpigmented periorbital skin and prominent low set ears. The skin over the entire body is dry, finely wrinkled and hypopigmented and often with prominent venous return. Anodontia or hypodontia with widely spaced, conical teeth are consistent features. Poor development of mucous gland in the respiratory and gastro-intestinal tract may result in increased susceptibility to respiratory infections, purulent rhinitis, dysphonia and diarrhoea (1).

The case reported have classical facies, anodontia, scanty hair and thin dry skin. Clinical profile was characteristic enough for the AED and investigative support for the diagnosis was not considered necessary. Presence of recurrent chest infections in our case can be explained on the basis of absence of mucous gland in the airways in this entity.

Incontinentia pigment has been reported with hypohidrotic ectodermal dysplasia (8). A case of 9-year old male child has been reported with complete anodontia. In this child, dental problem was best managed by prosthetic replacement of dentition (9).

Cardiomyopathy has been reported with ectodermal dysplasia which included hair shaft dystrophy (10). A 16-year old male with end stage lung disease secondary to chronic severe respiratory

infection has been reported in association with AED. He was subjected to bilateral sequential lung transplant (11).

Approximately 30% of affected boys die during the first 2 years of life as a result of hyperpyrexia or fulminant respiratory infections (1). The sweating deficit is a reflection of hypoplasia or absence of eccrine glands which may be diagnosed by skin biopsy. The palmar skin is an appropriate site for biopsy. Linkage analysis has been used for prenatal and early neonatal diagnosis.

Treatment of these children includes protecting them from exposure to high ambient temperature. Early dental evaluation is necessary so that prosthesis can be provided for cosmetic reasons and for adequate nutrition (1).

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