

# Anhidrotic Ectodermal Dysplasia- A Report of Two Cases

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## Abstract

Ectodermal dysplasias (EDs) are a group of X-linked recessive inherited disorders characterized by primary defects in the development of two or more tissues derived from embryonic ectoderm. The tissues which are affected include skin, hair, nails, eccrine glands and teeth.

Here, we report two cases of anhidrotic ectodermal dysplasia who were male siblings.

## Introduction

The ectodermal dysplasias (EDs) are congenital, diffuse and non progressive disorders. More than 192 distinct disorders have been described till date. Most common of them are X-linked recessive anhidrotic (Christ-Siemens-Touraine syndrome) and hidrotic ectodermal dysplasias (Clouston syndrome).<sup>1</sup>

They are classified as either group A disorders having at least two of the four classic ectodermal structures defect with or without other defects and group B disorders having one classic ectodermal structure defect with a defect in other ectodermal structures i.e. ears, lips, dermatoglyphics; the four defects being trichodysplasias, dental abnormalities, onychodysplasias and dyshidrosis.<sup>1</sup>

The frequency of different EDs is highly variable. In the US, the prevalence of anhidrotic ED is estimated to be 1 case per 100,000 births. Collectively, the prevalence of ED is estimated at 7 cases per 10,000 births.<sup>1</sup>

We report two male siblings, both, a case of anhidrotic ectodermal dysplasia.

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## Case Report

A 3 year old male and his younger male sibling of 1½ years of age, both born of non-consanguineous marriage presented with sparse hair over the scalp and complete absence of sweating since birth. As they grew older, there was failure of teeth eruption, progressive hyper pigmentation around eyes and mouth and recurrent episodes of seizures due to hyperpyrexia and exposure to extreme heat. There was hair growth only on the eye lashes which were normal in colour and number. Their maternal grandfather had similar complaints.

Physical examination revealed recession of hair line all along with sparse, thin, light brown lusterless scalp hair (Fig. 1), multiple tiny skin coloured follicular papules on scalp and forehead, frontal bossing with prominent supraorbital ridges and low set ears. Eyelashes appeared normal. Hyper pigmented ill defined patches were seen periorbitally and periorally (Fig. 2). Lips were everted and prominent (Fig. 2). Dental examination revealed absence of teeth in both jaws without gum atrophy or hypertrophy. Other systems were within normal limits.

We received skin biopsies of both siblings which showed absence of adnexal structures including sweat glands, sebaceous glands and hair follicles (Figs. 3 and 4).

## Discussion

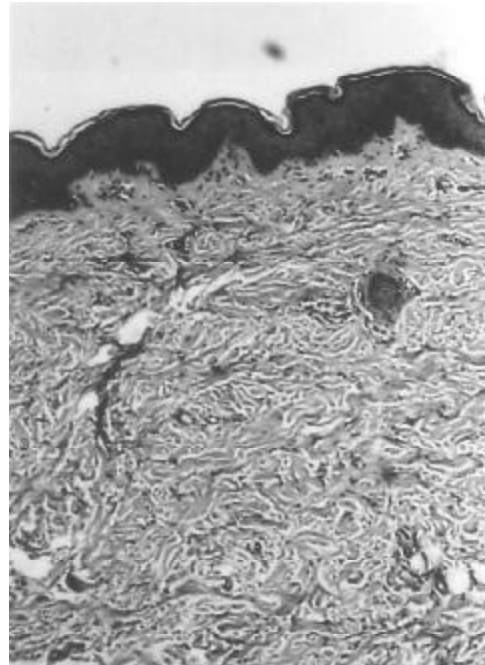
Thurnam published the first report of a patient with ectodermal dysplasia in 1848,<sup>2</sup> but the term ectodermal dysplasia was not coined until Weech termed it as so in 1929.<sup>3</sup> In 1984, Friere-Maia proposed the first



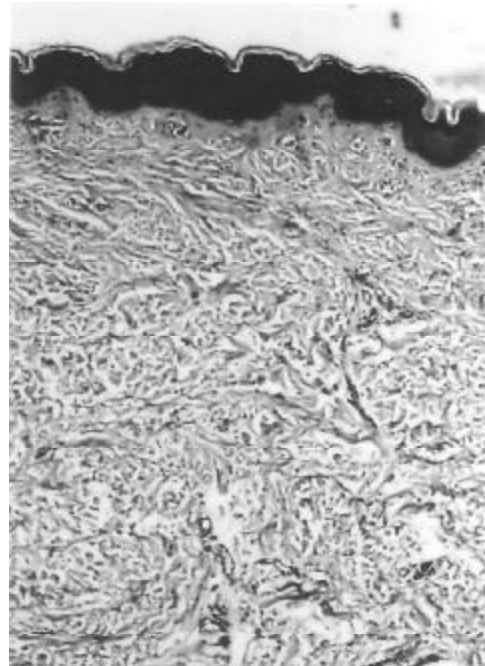
*Fig. 1 : Sparse, thin, lusterless scalp hair*



*Fig. 2 : Hyper pigmented ill defined patches seen periorally with everted and prominent lips.*



*Fig. 3 : Skin biopsy of one sibling showing absence of adnexal structures including sweat glands, sebaceous glands and hair follicles.*



*Fig. 4 : Skin biopsy of other sibling showing similar features.*

classification with updates in 1994 and 2001.<sup>4</sup>

With recent identification of the causative genetic defect for a number of the EDs, newer classification systems have been devised. In 2003, Lamartine reclassified the EDs into the following 4 functional groups based on the underlying patho-physiologic defect. 1) Cell to cell communication and signaling, 2) adhesion, 3) development and 4) other.<sup>5</sup> Similarly in 2005, Priolo and Lagana reclassified EDs into two main functional groups: 1. defect in developmental regulation/epithelial-mesenchymal interaction and 2. defect in cytoskeleton maintenance and cell stability.<sup>6</sup>

The most common type of ED is X-linked recessive anhidrotic ED (Christ-Siemens-Touraine syndrome). It has full expression only in males, although female carriers outnumber affected men but show little or no sign of the condition.<sup>1</sup> The typical facies is characterized by frontal bossing, sunken cheeks, saddle nose, thick everted lips, wrinkled hyper pigmented periorbital skin and large low set ears. Dental manifestations include conical or pegged teeth, hypodontia or complete anodontia and delayed eruption of permanent teeth. Fine, sparse, lusterless fair hair over scalp is seen in most of the patients. Onychodystrophy may be seen but is not common. Extensive scaling of the skin and unexplained pyrexia and heat intolerance due to anhidrosis occurs. Intelligence is normal.<sup>1</sup> Palmo-plantar keratoderma is a component of hidrotic ED but has been reported in anhidrotic ED.<sup>7</sup> Patients may have chronic nasal infections with foul smelling discharge and increased lung infections.<sup>8</sup>

Clinical recognition of ED varies from birth to childhood depending on the severity of symptoms and the recognition of associated complications. Dental, hair and nail

anomalies usually become evident during infancy or childhood.<sup>1</sup> A family history of similar clinical features is helpful.

In our case, the patients showed typical facies with dental manifestations, sparse scalp hair and heat intolerance. As both male siblings and their maternal grandfather were affected, we can ascertain the mode of inheritance to be X-linked recessive.<sup>9</sup>

The prognosis for most patients with ED is very good. If hypohidrosis is recognized in the neonatal period and managed appropriately, no evidence indicates that the life span for a person with one of the common types of EDs is shorter than average.<sup>1</sup>

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