

# Ectodermal dysplasia: Report of two cases in a family and literature review

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### ABSTRACT

Ectodermal dysplasia (ED) is an inherited genetic disorder with manifestations of abnormalities in more than one ectodermal derivatives like skin, hair, nails, exocrine glands and teeth. There are more than 150 different variants of ED described in literature. The condition is thought to occur in approximately 1 in every 100,000 live births. It mainly manifests in two types i.e. Hypohidrotic (Anhidrotic) type and Hydrotic type depending on degree of sweat gland function. This report presents two cases within a family, a 4 year old boy and a 6 year old girl with typical features of Hypohidrotic Hereditary ED i.e. hypodontia, hypohidrosis and hypotrichosis.

**Keywords:** Ectodermal dysplasia, hypodontia, hypohidrotic, hypotrichosis

### Introduction

Ectodermal dysplasia (ED) represents a group of inherited conditions in which two or more ectodermally derived anatomic structures fail to develop. There are more than 150 different variants of ED described in literature. The condition is thought to occur in approximately 1 in every 100,000 live births. Individuals affected with ED have abnormalities in different ectodermal derivatives like skin, hair, nails, teeth, sweat glands, parts of the eyes, ears, neural and adrenal tissues to various degrees. Dental, hair and nail anomalies are evident during infancy or childhood.

ED mainly manifests in two types, i.e. hypohidrotic (anhidrotic) type and hydrotic type depending on sweat gland function.<sup>[1]</sup> The classical triad of hypodontia, hypohidrosis and hypotrichosis is seen in the X-linked hypohidrotic form (Christ–Siemens–Touraine syndrome), whereas the hydrotic form (Clouston's syndrome) does not involve the sweat glands

and is inherited as autosomal trait. X-linked hypohidrotic ED is caused by alterations in the gene ectodysplasin (EDA or EDA1) located at Xq-12-q13.1.<sup>[2]</sup> Hydrotic ED is caused by a mutation in connexin gene, GJB6 or connexin-30.<sup>[3]</sup>

The present article reports two cases of hypohidrotic type of ED in a family.

### Case Report 1

A 4-year-old boy accompanied by his father reported to the Department of Oral Medicine and Radiology, Government Dental College, Hyderabad with a chief complaint of missing teeth since birth. He also gave a history of delayed eruption of milk teeth and mentioned that the child was intolerant to heat, had an inability to sweat and suffered from frequent bouts of fever. He had dry skin with prominent cracks on the bridge of nose, nasolabial folds and periorbital region. He had frontal bossing, depressed nasal bridge, periorbital pigmentation, protuberant incompetent lips, scanty hair on the scalp and eyebrows [Figure 1a] and scanty body hair. Nails appeared thin

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and short. Intraoral examination revealed a relatively dry mucosa. Both the arches were partially edentulous with multiple missing deciduous teeth [Figure 2a]. The anterior teeth were conical in shape and the crown morphology of the posterior teeth was altered. Orthopantomograph revealed multiple missing permanent tooth buds [Figure 3a].

## Case Report 2

A 6-year-old girl, elder sister of the first patient had similar symptoms of heat intolerance, inability to sweat and frequent bouts of fever. Her general and extraoral examination revealed the typical features of ectodermal dysplasia as aforementioned [Figure 1b]. Intraoral examination revealed a relatively dry mucosa with multiple missing teeth in the maxillary and mandibular arches [Figure 2b]. The teeth present had altered tooth morphology. Orthopantomograph revealed few impacted deciduous teeth and permanent teeth in different stages of development [Figure 3b]. Based on the history, clinical and radiologic examination, the girl was diagnosed with hypohidrotic ED with partial anodontia.

Both the patients were subjected to prosthetic evaluation for improvement of mastication, speech and esthetics.

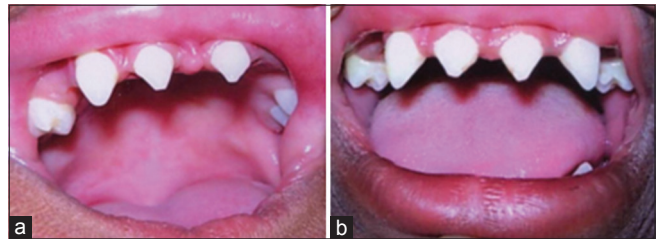
## Discussion

Ectodermal dysplasia results from the aberrant development of ectodermal derivatives in early embryonic life. ED is classified into two subcategories as hypohidrotic or hydrotic depending upon the degree of sweat gland function. X-linked hypohidrotic ED has been mapped in the long arm of band Xq-12-q13.1.<sup>[2,4]</sup> X-linked hypohidrotic form (Christ–Siemens–Tauraine syndrome) is characterized by the classical triad of hypodontia, hypohidrosis and hypotrichosis with characteristic dysmorphic facial features. Other signs of this disorder include fine sparse blond hair including a reduced density of eyebrow and eyelash hair.<sup>[5]</sup> The periorbital skin may show a fine wrinkling with hyper-pigmentation, and mid-face hypoplasia resulting in protuberant lips. Nails may also appear dystrophic and brittle.<sup>[6]</sup> These features were akin with the typical features seen in both the patients. Affected individuals with hypohidrotic form typically display heat intolerance because of reduced number of sweat glands. Sometimes the diagnosis is made during infancy because the baby appears to have a fever of unknown origin.<sup>[3]</sup> These findings were typically seen in both the patients reported.

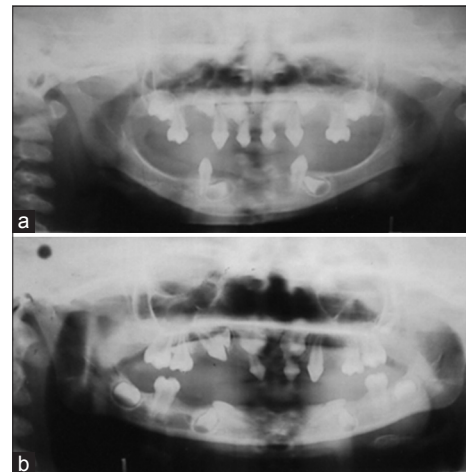
Lack of alveolar ridge development is seen wherever the teeth are absent. As a result, the vertical dimension of the lower face is reduced and gives a typical senile look to the patient. Varying degrees of xerostomia have been reported in few patients as the salivary glands are ectodermally derived. In the present report, both patients exhibited a relatively dry mucosa. The teeth are markedly reduced in number and their crown shapes are characteristically abnormal. The incisal crowns



**Figure 1:** (a and b): A 4-year-old boy and a 6-year-old girl with features of ED



**Figure 2:** (a and b): Intraoral view showing partially edentulous arches in both the cases



**Figure 3:** (a and b): Orthopantomograph showing multiple missing and impacted teeth in both the cases

usually appear tapered, conical or pointed, and the molar crowns are reduced in diameter.<sup>[6]</sup> Complete lack of tooth development has also been reported, but this appears to be uncommon.<sup>[6]</sup> Although the parents of the present cases were not consanguineously married and were not affected with the condition, a history of consanguineous marriage among the parents of affected individuals has been implicated in many reports.

Early prenatal diagnosis in patients with a family history of hypohidrotic ED can be established by DNA-based linkage analysis and genetic tests for detecting mutations in ectodysplasin (EDA or EDA1) gene. Sonography and fetal skin

biopsy are suitable diagnostic tests in the second trimester of pregnancy.<sup>[7,8]</sup>

Additional care must be provided to infants and young children with hypohidrotic ED by the treating physician to reduce the mortality rate as these patients suffer with numerous complications such as failure to thrive, pulmonary infections and hyperthermia.<sup>[9]</sup>

Patients affected with ED suffer from poor psychological and physiological development as a result of unacceptable esthetics and abnormal function of orofacial structures. Although there is no specific treatment for the disease, affected individuals should be subjected to early dental evaluation including prosthetic rehabilitation for good esthetics, phonetics and masticatory comfort.<sup>[10]</sup> The prosthesis must be periodically modified in young adults as alveolar growth, erupting teeth and rotational jaw growth changes.

## Conclusion

ED is a heterogeneous group of inherited disorders with familial tendencies. In this report, two siblings from a South Indian Hindu family showed features of hypohidrotic ED. Dentists play a significant role in the early diagnosis of the condition, thereby providing early interventions which is a key to the effective management of the patient.

## Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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## Conflicts of interest

There are no conflicts of interest.

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