

# ECTODERMAL DYSPLASIA: TWO CASE REPORTS

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## ABSTRACT:

The ectodermal dysplasia are a large and complex nosological group of diseases, first described by Thurnam in 1848. It is ordinarily considered to be a triad of clinical findings – hypodontia, hypotricosis and hypohidrosis and is frequently associated with other components resulting from defective development of structure of ectodermal origin. The dental findings are of particular interest, since patient with this abnormality invariably manifest true anodontia or hypodontia, with frequent malformation of any teeth present. There is no known treatment for the condition itself.

**Key words:** Hypotricosis, hypohidrosis, anodontia, nosology



## INTRODUCTION:

Dental professionals commonly observe congenitally missing teeth during the process of care. There are over 120 syndrome in which oligodontia is one of associated findings. Those most commonly associated with missing teeth are ectodermal dysplasia and down syndrome. <sup>(1)</sup>

Ectodermal dysplasia is a hereditary disorder that occurs as a consequence of disturbances in the ectoderm of the developing embryo. <sup>(2)</sup> Although Thurnam

published the first report of a patient with ectodermal dysplasia in 1848, the term ectodermal dysplasia was coined by Weech in 1929. <sup>(3)</sup> There are more than 180 different ectodermal dysplasia which are recognized by the combination of physical features an affected person has and the way they are inherited. <sup>(4)</sup> X-linked hypohidrotic ectodermal dysplasia is the most common syndrome constituting 80% of those affected by the condition. <sup>(1,4)</sup> It is characterized by hypoplasia of hair, teeth and sweat glands. (Mc Kusick, 1994) <sup>(5)</sup> Incidence of this condition is

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estimated as one in 100000 births worldwide. <sup>(1)</sup>

Patients with hereditary ectodermal dysplasia usually exhibit a fine, smooth, dry skin with partial or complete absence of sweat glands. The bridge of nose is depressed and supraorbital ridges are pronounced. Cephalometric studies by Sarnat *et al* indicate that apart from defective alveolar growth, jaw and facial development is essentially normal. <sup>(6)</sup> Already in 1936, Tannhauser stated that the characteristic deformity of the cranial bones of all affected patients is such that the resemblance among the patients is bigger than when compared with their own unaffected sib (Bergendal *et al.* 1998). <sup>(5)</sup> This article presents two cases of such condition reported to us.

### CASE DETAIL:

#### CASE 1:

A 7 yrs old male patient along with his parents reported in our clinic with the complaint of missing teeth. He has history of mild hyperthermia but no history of any medical evaluation. Patient does not have any family history. On general examination patient shows increase in body temperature and dry skin. On extraoral examination patient shows presence of sparse hairs, protuberance of lips, sparse eyelashes while intraorally patient shows oligodontia with teeth present are 16 55 12 11 21 22 65 26 in upper arch and 36 75 73 83 85 46 in lower arch. Teeth appear cone shaped with spacing in upper anterior region and edentulous, thin

alveolar ridge seen in lower anterior region.

#### CASE 2:

6 yrs old patient reported in our clinic with the complaint of missing teeth. He has not given any significant medical history. Family history was also not significant. On general examination patient appears of thin built. Extra orally patient shows scanty hairs, sunken cheeks, protruded lips. Intraorally patient shows oligodontia with teeth present are 11 55 16 21 65 26 36 31 41 46. Radiographic examination reveals presence of 11 55 16 21 65 26 36 31 41 46 teeth with enlarged pulp chamber, conical roots and tooth buds of 17 27 37 47.

### DISCUSSION:

Ectodermal dysplasia syndrome is a large heterogeneous group of inherited disorder, the manifestations of which could be seen in more than one ectodermal derivatives. The most common syndrome within this group are hypohidrotic (anhidrotic) ectodermal dysplasia and hidrotic ectodermal dysplasia. <sup>(7)</sup> All the ectodermal dysplasias appear to be genetic in etiology. The recent cloning of the gene has led to the identification of a novel transmembrane protein "ectodysplasin" (TNF family ligand) and receptor "edar" (TNF receptor). <sup>(5)</sup> The disease is generally much more apparent in affected men than in carrier women. <sup>(8)</sup>

During childhood the diagnosis is more easily made on the basis of history and clinical examination. <sup>(9)</sup> Affected individuals display heat intolerance due to reduced

number of sweat glands (hypohidrosis). Other signs of this disorder include fine, sparse blonde hair including a reduced density of eyebrow and eyelash hair (hypotricosis). Midface hypoplasia is observed resulting in protuberant lips.<sup>(10)</sup> In our cases also both patient have sparse hair over head and eyelash. They also have complaint of hyperthermia.

Dental abnormalities are most common complaint which include oligodontia (reduced number of teeth) or anodontia (complete absence of teeth) and delayed eruption of permanent teeth. The anterior teeth frequently have conical crowns.<sup>(11)</sup> Radiographs are helpful for determining the extent of hypodontia, taurodontism or elongation of the pulp chamber is more common in molar teeth. Roots are also in conical shape.<sup>(12)</sup> In our cases also oligodontia can be appreciated with conical anterior teeth crown while radiographically in second case enlarged pulp chamber is present with conical roots.

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In contrast with other findings seen in ectodermal dysplasia our patient have normal nail structure ,no frontal bossing and mild midface depression. The diagnosis made on clinical and radiographic findings suggest the presence of Ectodermal dysplasia. The management of children and adults with ectodermal dysplasia is a challenge because of their heat intolerance and because of their susceptibility to pulmonary infections.<sup>(13)</sup>

The dental problems are best managed by prosthetic replacement of the dentition with complete dentures, overdenture or fixed appliances. Dental implants can be considered for patient older than 5 years of age.<sup>(10)</sup>

## CONCLUSION:

Dentists are often the first who diagnose these patients. This will be helpful in proper diagnosis, early interventions and appropriate therapies for these patients.

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## FIGURES:

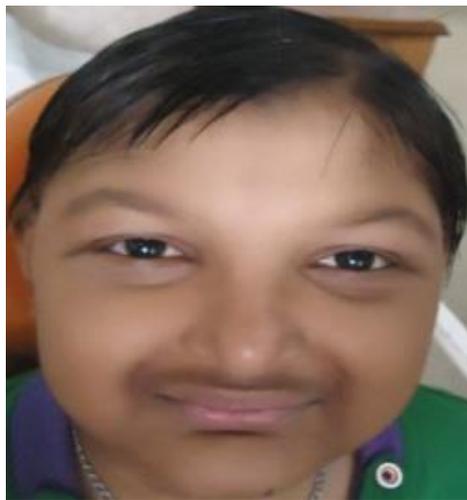


Fig 1: Showing extraoral appearance with sparse eyelashes protruded lips.



Fig 2: Showing sparse hairs over the head



Fig 3: Showing lower arch with missing lower anterior teeth.



Fig 4: Showing upper arch with oligodontia and spacing in anterior region.



Fig 5 : Showing extraoral appearance with sunken cheeks and protruded lower lip.



Fig 6: Showing upper arch with oligodontia teeth present are 16 15 11 21 25 26.



Fig 7 : Showing oligodontia in lower arch with teeth present are 36 31 41 46.



Fig 8: Panoramic view showing oligodontia with missing 12 13 14 22 23 24 32 33 34 35 42  
43 44 45