ISSN (O):2395-2822; ISSN (P):2395-2814

Concomitant Presence of Ectodermal Dysplasia with Taurodontism: A Rare Entity.

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Received: January 2018
Accepted: January 2018

ABSTRACT

Ectodermal dysplasia (ED) is a rare genetic disorders characterized by faulty development of ectodermally derived structures ranging from primarily hairs, nails teeth, exocrine glands. Most of the patients are males because of the X linked recessive inheritance. We report a case of ectodermal dysplasia along with taurodontism. These cases along with ectodermal dysplasia and taurodontism are more likely to be associated with a Hypohydrotic form of ED. Dentists are first to encounter these patients because of the presence of hypodontia so proper diagnosis, early intervention and appropriate approach is necessary for these patients. The authors have also reviewed the literature in the present context. Ectodermal dysplasia are group of disorder mainly associated with a defect in the development of two or more ectodermally derived structures. Early oral intervention and rehabilitation in such patients helps in normal social and psychological development

Keywords: Ectodermal dysplasia,taurodontism, Hypodontia, hypohydrosis.

INTRODUCTION

Ectodermal dysplasia(ED) is a phenotypically heterogenous group of malady first described by Thurman, characterized by one or more ectodermally derived structures & occasionally non ectodermal origin.[1,2] Almost 192 types of different disorders are recognized as ED, out of them most frequently encountered are X linked recessive hypohidrotic and hidrotic type.[3,4]

The most frequent phenotype includes hypotrichosis (sparse hairs), hypohydrosis (deficiency of the sweat glands). Intraorally mostly there is presence of oligodontia or anodontia along with conical teeth and generalized spacing.[3,5] Other features include frontal bossing with prominent supraorbital ridges, nasal bridge depression, and protuberant lips. Patient perhaps suffers with hyperthermia, and unexplained high fever because of the result of the deficiency of sweat glands.

Many of the times Dentist encounters these patients because oligodontia might be a prominent feature. Proper diagnosis, early rehabilitation and genetic counseling should be done for these patients to improve the quality of life.

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Here we documented an uncommon case of ED along with taurodontism affecting all permanent dentition.

CASE REPORT

A male patient aged 20 years reported to the clinic with missing teeth. From clinical history, it was found that the condition is present both in his upper and lower jaws since childhood. Also similar condition was evident in the family (mother's brother). The patient also complains of difficulty in chewing and eating food.

Upon clinical examination, extraorally, patient presented with sparse hair (hypotrichosis) [Figure 1], eyelashes and eyebrows were scanty [Figure 2] and skin appeared to be rough, lusterless and dry (hypohydrosis) [Figure 2]. Both upper and lower lips were everted and prominent along with saddle shaped depressed nose and undeveloped bridge of the nose [Figure 2]. Profile view shows under development and hypoplasia of middle third of the face along with frontal bossing [Figure 3].

On intraoral examination, oligodontia was evident along with crown malformation in all his four quadrants. Atrophy of alveolar ridge was evident. Overall clinical features and family history given by the patient lead to a provisional diagnosis of anhidrotic ED.

Investigations

Intraoral periapical radiograph (IOPA): on IOPA examination of peculiar teeth in all the quadrants

there in evidence of some degree of taurodontism [Figure 4a,4b].

Panoramic radiograph: panoramic radiograph revealed underdevelopment of alveolar ridge along with taurodontism is clearly evident which is most prominently in a left mandibular permanent molar. [Figure 5].

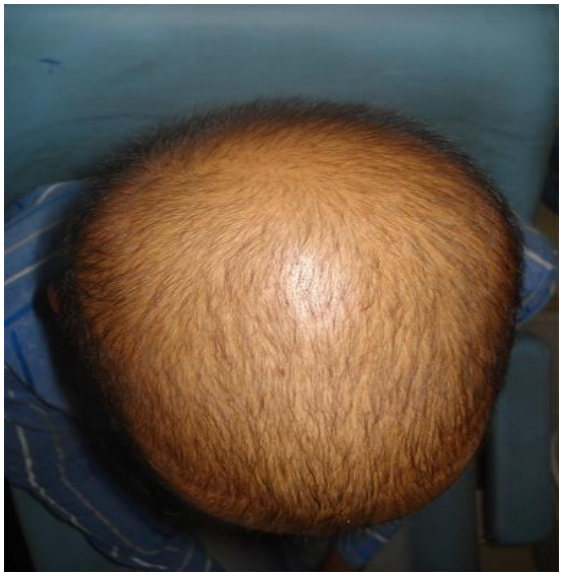

Figure 1: Showing very sparse hair (hypotrichosis) on scalp.

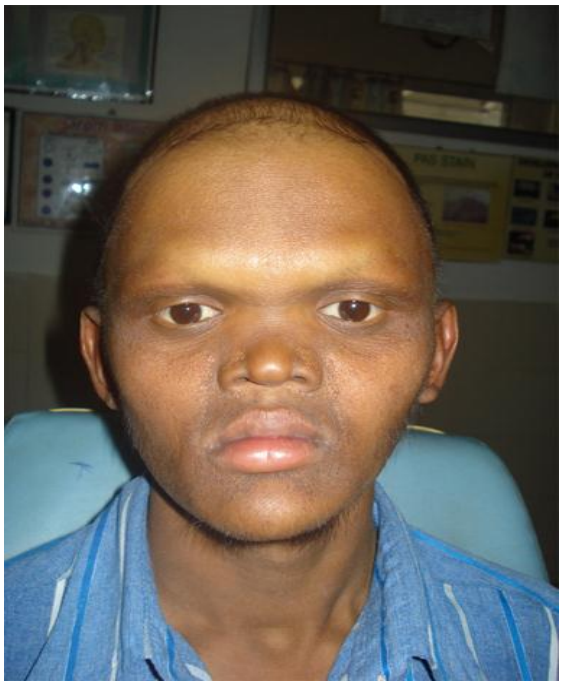

Figure 2: showing lustreless dry skin, depressed nose bridge and everted lips.

Differential Diagnosis

Syndromic partial anodontia and trichodento-osseous syndrome, Non-specific partial anodontia,

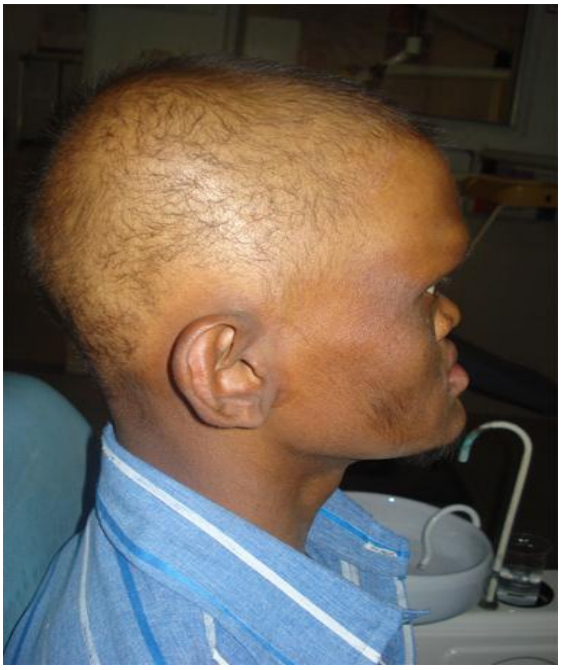

Figure 3: profile view showing underdeveloped middle third of face with sparse scalp hair

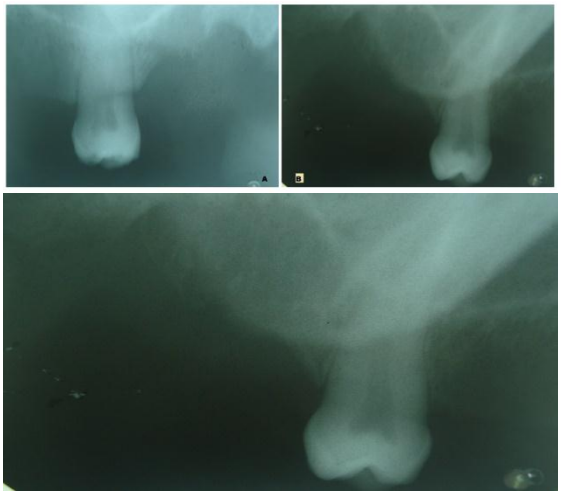

Figure 4: figure 4a and 4b showing taurodontism in upper maxillary tooth

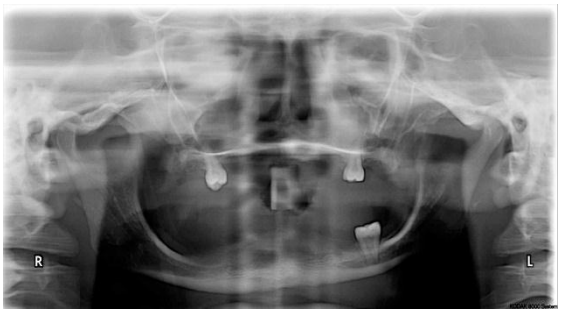

Figure 5: panoramic radiograph showing taurodontism in lower left molar

Treatment & Follow Up

A comprehensive approach is taken towards patient's awareness about the education of the condition and motivated for prosthetic treatment for

missing teeth. The patient also educated about the absence of the sweat glands related hyperthermia, and hence advised to avoid long term activities in the increased temperature environment. The patient also educated about the genetic inheritance of the present condition and appropriate counselling for the same is done.

The patient was kept under follow-up for 1 month, during this period prosthetic treatment for the the patient is started.

Table 1: classification system for Ectodermal dysplasia.[7]

Major Groups	Sub-Groups
ED1: Trichodysplasia (hair dysplasia)	Subgroups 1-2-3-4
ED2: Dental dysplasia	Subgroups 1-2-3
ED3: Onychodysplasia (nail dysplasia)	Subgroups 1-2-4
ED4: Dyshidrosis (sweat gland dysplasia)	Subgroups 1-2
	Subgroups 1-3
	Subgroups 1-4
	Subgroups 2-3-4
	Subgroups 2-3
	Subgroups 2-4
	Subgroups 3
	Subgroups 4

Table: 2 Conditions associate with Taurodontism and their prominent features.[14-19]

Syndrome	Prominent features
Amelogenesis imperfecta	Enamel hypoplasia, hypomaturation
Down's syndrome	Macroglossia, Delayed eruption, Absence of tooth germs
Ectodermal dysplasia	Orodental dysplasia, Cranioectodermal dysplasia, and Rapp-Hodgkin syndrome
Klinefelter syndrome	Cleft soft palate, Missing premolars,Delayed development of the permanent tooth germs
Tricho-dento-osseous syndrome	Hypoplastic enamel
Lowe syndrome	Severe bone loss,Jaws underdevelopment,Gross periodontal disease,Permanent teeth Impaction
Mohr syndrome	Cleft palate,Small tongue,Notching of the upper lip
Wolf-Hirschhorn syndrome	Microdontia,Severe hypodontia,Cleft lip and/or palate Taurodontic primary molars,Delayed dental development
Smith-Magenis syndrome	Tooth agenesis,Root dilaceration
Williams syndrome	Wide mouth,Smaller size of teeth,Aberrant shape of teeth
McCune-Albright syndrome	Oligodontia,Malocclusion,Tooth rotation
Apert syndrome	Anterior open bite,Dental malocclusion,Delayed tooth eruption,Crowding of the dental arch

DISCUSSION

History

Thurnam, first reported Ectodermal dysplasia (ED) was first reported by Thurman and the term ED was coined in 1929 by Weech. EDAs is congenital, diffuse and non progressive.[3,6]

Classification:

Presently almost 150 different types of ectodermal dysplasias has been identified. There are mainly defects of hair, tooth,nails and sweat glands. On the basis of these four primary ectodermal defects an arbitrary classification of ectodermal dysplasia is given [Table 1].[7]

Hypohydrotic/anhydrotic ectodermal dysplasia is most common found ectodermal dysplasia, which comes under subgroups 1-2-3-4 followed by hydrotic ectodermal dysplasia which is subgroup 1-2-3.[7]

Aetiology and pathogenesis

Mostly ED is genetically related and EDA (ectodysplastin), EDARADD (ectodysplasin receptor associated death domain) and EDAR(ectodysplasin receptor) are the genes responsible. Ectodermal dysplasia can be autosomal dominant, autosomal recessive or X-linked recessive.[7] Hypohidrotic ectodermal dysplasia (HED) transmitted mainly as an X-linked recessive trait in which the gene is carried by the female and manifested in male.[8]

In our case patient was not subjected to the genetical analysis because of time and money constraints.

Clinical Presentation

The earliest manifestation could appear during intrauterine life, mainly first trimester. Dentition might be affected if severity appears to be before 6th week of Intrauterine life.[9]

Extraoral findings:One of the main distinctive features of ectodermal dysplasia is hypohydrosis and manifests as soon as first year of life. Clinically hypohydrosis presented as repeated episodes of unknown pyrexia. The complete or partial hypohydrosis leads to dry, soft, smooth and thin skin. Apart from hypohydrosis there is presence of sparse hairs (hypotrichosis) over scalp, axilla, pubic hairs. The nails appear to be spoon shaped.[10]

Other ectodermal structures involved are facial bones. Skull shape and size is variable and resemble triangular. The middle third of the face is underdeveloped (hypoplastic) and profile view of the patients shows depression of the nasal bridge along with frontal bossing. Lips are thick and become protuberant.[10]

Most of the characteristic features are present in our patient, but the hypohydrosis, hypotrichosis and hypoplasia of middle third of the face were most prominent.

Intraoral findings

The most striking is the partial (oligodontia) or complete absence of the teeth (anodontia) in both primary as well as permanent dentition. Apart from the number the shape of the tooth is also altered in some cases, mainly conical in shape.[9] Along with that taurodontism is also present in few cases. Taurodontism is not a striking feature as it is present in various other syndromes also as shown in [Table 2]. The incidence of missing teeth is more in mandible as compare to the maxilla. Absence of teeth is a major concern for esthetics in these patients and prompt treatment should be perform for this.

Maxilla is underdeveloped in these patients leading to prognathic mandible, retruded maxilla and decreased anterior facial height.[10-12]

In our case oligodontia, taurodontism and maxillary hypoplasia were most evident features observed.

Diagnosis and Treatment

Diagnosis is mainly based on family history, clinical features and investigations. There are various investigations like radiographs, sweet pore count to rule out hypohydrosis, Pilocarpine iontophoresis, skin biopsy and genetic tests are avaialible.[13]

In our case the diagnosis is mainly based on radiographs, family history and clinical examination done by us. The goal of treatment for ectodermal dysplasia patient is mainly for esthetic and involve a team approach consist of pediatricians, pediatric dentist, prosthodontist, dermatologist, otolaryngologist, speech therapist and psychologist.

CONCLUSION

Ectodermal dysplasia are group of disorder mainly associated with a defect in the development of two or more ectodermally derived structures. Oral and extraoral features, hypohidrosis, hypotrichosis, anodontia (partial or total) are useful in the early recognition of individuals with this state and female are only carriers. Early oral intervention and rehabilitation in such patients helps in normal social and psychological development. Counselling may prove to be cautious in family planning and help reduce the incidence of these kinds of cases.

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How to cite this article: Dagrus K, Manjunatha BS, Shah V, Shah SN. Concomitant Presence of Ectodermal Dysplasia With Taurodontism: A Rare Entity. Ann. Int. Med. Den. Res. 2018; 4(2):DE14-DE17.

Source of Support: Nil, **Conflict of Interest:** None declared