

## CASE REPORT

# ECTODERMAL DYSPLASIA – A REVIEW AND CASE REPORT

### Abstract:

A 3 year old male child described in this report exhibited features of ectodermal dysplasia along with partial anodontia .The treatment attempted to improve his appearance and restore oral functions with fixed functional space maintainers. The results were a significant improvement in confidence, esthetics, speech and function of child and parental satisfaction. Also ability of child to improve dietary habits was noted. This article aims to provide the reader an insight into various modalities of managing child patients with ectodermal dyaplasia.

### Keywords:

Ectodermal dysplasia; anhydrotic; hypodontia; anodontia; space maintainer

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## Introduction:

Ectodermal dysplasia is group of hereditary disorders characterised by developmental dystrophies of ectodermal derivatives<sup>[1]</sup>. Freire - Maia- Pinheiro have described 154 patterns of ectodermal dysplasias ,divided them into 11 subgroups ,and then classified them according to the involved structures.(hair,teeth,some or all of the sweat glands)<sup>[2]</sup>. It was first described by Thurnam<sup>[3]</sup> in 1848 and later in the 19th century by Darwin<sup>[4]</sup>. It is an X-linked recessive disorder<sup>[5]</sup>. The condition is thought to occur in 1-7 per 100000 live births<sup>[6]</sup>. Around 90% of cases are males and the complete syndrome does not occur in females<sup>[7]</sup>. The clinical findings in carrier females are the same as those in affected males. One third of the carriers appears healthy, another third of them is showing mild symptoms, and the last third exhibits significant symptoms, but often milder than the affected males (Sofaer et al. 1981)<sup>[8]</sup>. A much rarer form of anhidrotic ectodermal dysplasia inherited as an autosomal recessive trait has been described<sup>[9]</sup>, where phenotypically, the features are indistinguishable from those of the X-linked form except that complete syndrome occurs in females<sup>[10]</sup>. It is characterized by a triad of signs comprising sparse hair, abnormal or missing teeth and inability to sweat. Anodontia or hypodontia is the most striking dental manifestation<sup>[11]</sup>. In severe hypodontia, there is lack of alveolar development with consequent protrusion and eversion of the lips<sup>[12]</sup>. Christ-Siemens-Touraine syndrome, also known as anhidrotic ectodermal dysplasia is characterized by partial or complete absence of sweat glands, hypotrichosis and hypodontia .

## Disease name and synonyms

Ectodermal dysplasia anhidrotic (EDA)

Anhidrotic ectodermal dysplasia

Hypohidrotic ectodermal dysplasia (HED)

Christ-Siemens-Touraine syndrome

**Incidence and inheritance pattern:** The prevalence of EDA is unknown; however, the incidence in male is estimated at 1 in 100,000 births although the condition is usually overlooked in infants (Bergendal et al. 1998)<sup>[13]</sup>. This X-linked recessive disorder affects males and is inherited through

female carriers. This carriers-incidence is probably 17.3 in 100,000 women (Sofaer 1981)<sup>[8]</sup>. (EDA) is the most common Ectodermal dysplasia (80%); it is characterized by hypoplasia of hair, teeth and sweat glands. (Mc Kusick, 1994)<sup>[14]</sup>. Since there is not a complete lack of sweat glands the term hypohidrotic is more adequate than anhidrotic. The anhidrotic hypohidrotic) ectodermal dysplasia is often inherited as an X-linked disorder (XLEDA). The differential diagnostic problem is the distinction of the autosomal recessive form of HED (AR-HED) from X-linked HED. AR-HED is considerably less common than XLHED. The clinical features are quite similar in both conditions but due to the different mode of inheritance AR-HED affects both males and females and the heterozygotes have no signs at all. For adequate genetic counseling it is thus important to recognize XLHED heterozygotes by dental examination and sweat tests. To distinguish between AR- and XL-forms, the HED diagnosis should be followed by careful family history for ectodermal manifestations both in male and female and by tests for heterozygote identification. The findings of equally affected males and females in single sibships, as well as the presence of consanguinity, support an autosomal recessive mode of inheritance (Munoz et al 1997)<sup>[15]</sup>.

## Case report :

A 3 year old boy was referred to the department of Pedodontics, St. Gregorios Dental College, Kothamangalam; with a chief complaint of pain and swelling in upper front teeth. Parents were also concerned as child just had upper primary canines, primary second molars apart from root stumps of primary central incisors in the upper arch and primary second molars in the lower arch.

On examination coronal portion of central incisors were lost due to caries and the root stumps were pulpally involved. Single sitting pulpectomies were carried out for the incisors as patient was already on medication with antibiotic and analgesic.

Subsequently after treatment parents raised their concerns over absence of teeth unlike his peers. The child himself was upset over joining school without teeth like other children at home. He presented facial features of scanty eyebrows, rounded nose, prominent chin, brittle and fine black hair, protruded lips,

partial anodontia, loss of vertical dimension and anomalous development of alveolar ridges. The skin of his extremities was dry and wrinkled.

#### **Radiographic findings:**

OPG revealed the presence of upper molars, conical canines and root stumps of central incisors in the upper arch. Erupted mandibular second molars and mandibular primary canines embedded in the lower arch with half of root formation were seen.

#### **Treatment done:**

At the next visit crowns of 51, 61 were built up with composite with strip crowns, Preliminary impressions of upper and lower arches were made. OPG and lateral cephalogram were taken.

As the child was very young and lacking the dexterity needed to maintain a removable space maintainer, it was decided to give him fixed banded type of space maintainers for the time being with a lot of stress on the need for regular recall. Special trays were made and secondary impressions were made with rubber base impression material. Bands were fabricated on the upper and lower molars. Acrylic teeth were mounted on acrylic flow into the fixed space maintainer design.

This was cemented onto the patient's molars with luting GIC on the 3rd visit. Patient and parents were happy with the esthetics, and restoration of masticatory function.

They are now on a 3 month recall visit program for the last one and a half years where the functional space maintainers on both arches are removed and cleared of residual food debris. On their last visit the lower primary canines were seen to erupt accordingly the arches were relieved of the acrylic component also to facilitate a self cleansing action.

As the child ages the fixed type of space maintainers will be changed to removable ones so as to give adequate rest to the periodontal tissues and maintain them in good health.

#### **Discussion:**

Missing teeth or the delay in teething often starts to worry the parents and leads to the diagnosis of EDA in the second year of life (Pirinen et al. 1996). A den-

tist should not hesitate to radiographically examine a patient whose teeth have not erupted by the appropriate age in order to exclude EDA. The screening limit for the first tooth to erupt is 15 months ((Pirinen et al. 1996)<sup>[16]</sup>.

Ohno K and Ohmori reported cases of anodontia and the fabricating procedure of full dentures for a young child was described<sup>[17]</sup>.

Gerard Kearns et al studied the feasibility of placing endosseous implants in children and adolescents with ectodermal dysplasia and to assess the position and stability of such implants during growth. This article reports on 6 subjects with long-term follow-up. There was no evidence that implant placement or prosthetic rehabilitation resulted in restriction of transverse or sagittal growth. They observed that maxillary implants placed in a partially dentate jaw became submerged because of adjacent alveolar development and required placement of a longer abutment. They concluded that endosseous implants can be successfully placed and can provide support for prosthetic restoration in patients with hereditary ectodermal dysplasia<sup>[18]</sup>

Third molars and maxillary lateral incisors are the most common congenitally missing teeth. Complete anodontia of both deciduous and permanent dentition is rarely reported. Açıkgöz et al. reported the absence of all primary and permanent teeth except the bilaterally unerupted maxillary permanent canines. This shows that the permanent tooth can develop in the absence of its predecessor<sup>[12]</sup>.

Even in the case of complete anodontia, the general facial growth pattern is normal in these children. This implies that the development of the jaw does not depend to the presence of teeth. Nevertheless, the alveolar process does not develop in the absence of teeth, and the vertical dimension is reduced, which explains the protuberant everted appearance of lips in these patients<sup>[19]</sup>. In this case, missing teeth were more in the mandible than in the maxilla. Therefore, the underdevelopment of alveolar process in mandible was more evident in clinical and radiographic features.

Alterations in bone structure, such as hyperdensity of medullary bone, have been found in mandibular symphysis area and other jaw locations of these

patients. Although changes in the alveolar bone can be related to oligodontia, changes in the bone structure seem to be tooth independent and suggest a direct effect of genetic defect on bone formation and/or remodeling in this syndrome<sup>[20]</sup>.

If hypohidrosis is managed appropriately, the prognosis for most patients will be very good. Treatment of these children is protecting them from high temperature. In most cases, the preferred treatment option for dental disorders is a removable partial denture, which could also be associated with direct composite restorations. This allows the child to have adequate nutrition, normal appearance, and speech with significant psychosocial benefits. When child reaches teenage years, orthodontic treatment will prepare the mouth for a fixed partial denture or implants in future. In some patients, alveolar ridges are severely hypotrophic due to oligo- or anodontia. This can seriously affect a young person physically and psychologically. In these cases, augmentation of jaws by the use of bicortical corticocancellous bone blocks from hip and delayed implant placement seems to be a suitable treatment option. Immediate loading has also been applied successfully in patients with ectodermal dysplasia<sup>[20]</sup>.

#### **Broad treatment guidelines to the condition**

The course of the treatment is to restore the function and the aesthetics of the teeth, normalise the vertical dimension and support the facial soft tissues. As long as there are no physical, psychological or social burdens, no treatment is necessary. Early placement of partial or full dentures is commonly recommended from the age of two or three years onwards. The denture must be periodically modified as alveolar growth; erupting teeth and rotational jaw growth, change both the alveolar, occlusal and basal dimensions. In children, breakage and even loss of removable prostheses is quite common. They have also a limited retention and stability, a fastened bone destruction of an already hypoplastic alveolar process and the middle of the upper jaw is covered and so it blocks the sutural growth. For this reason, in young children we prefer a treatment with crowns and bridges. Prior to that it is generally advantageous to modify the crowns of the existing teeth with direct or indirect composite crowns. When conical

anterior teeth are crowned the appearance of the child is very much normalised. Restoration of facial height improves both facial aesthetics and speech<sup>[21]</sup>.

#### **Conclusion:**

Dentists are often the first who diagnose these patients. Therefore, they should be aware of the clinical manifestations of this syndrome. This will be helpful in proper diagnosis, early interventions, and appropriate therapies for these patients.

As children with anodontia present with an insufficient amount of alveolar bone, implantation reconstruction surgery is subjected to a greater risk of failure in comparison to more conservative prosthetic treatment. To restore function as well as esthetics bonded space maintainers were used in this patient as a management modality. It is essential to provide continuous evaluation of such patients to monitor the state of the alveolar ridge and accommodate erupting teeth. The main aim of the treatment was to improve psychological development and to promote better functioning of the stomatognathic system.



Fig 1: Pre Operative facial Photograph showing scanty eyebrows, rounded nose, prominent chin, brittle and fine black hair, protruded lips and loss of vertical dimension



Fig 2 : Wrinkled appearance of hands



Fig 3: Dry appearance of legs and feet



Fig 4: Pre Operative Upper Jaw with primary incisor root stumps, conical canines and second molars only.



Fig 5: Pre Operative lower jaw showing primary canines and second molars only



Fig 6: OPG shows presence of tooth buds of permanent first molars in all four quadrants along with the teeth clinically present.



Fig 7 : Post operative view of fixed, banded space maintainers



Fig 8: Post Operative view of face



Fig 9 : Lower space maintainer modified for eruption of canines

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