

Ectodermal Dysplasia: Diagnosis and Dental treatment

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Abstract

Ectodermal dysplasia is characterized by the absence of or defect of 2 or more ectodermally derived structures. It is characterized by the triad of signs comprising sparse hair (hypotrichosis), abnormal or missing teeth (anodontia or hypodontia) and inability to sweat due to lack of sweat glands. Most patients with ED have a normal life expectancy and normal intelligence. However, the lack of sweat glands may lead to hyperthermia, followed by brain damage or death in early infancy, if unrecognized. Thus, an early diagnosis is important. This article consists of 2 case reports of ectodermal dysplasia. Common dental, oral and physical conditions were taken into account. Clinical management includes removable or fixed partial denture to promote better functioning of stomatognathic system.

Keywords: Anodontia; Ectodermal Dysplasia; Hypodontia; Hypotrichosis

Introduction

Anodontia, which represents the congenital absence of all teeth in the primary and/or the permanent dentition, is a rare condition [1]. Dental agenesis is an important clinical and public health problem [2]. Patients with missing permanent teeth may suffer from a reduced chewing ability, inarticulate pronunciation and an unfavorable aesthetic appearance [3]. Agenesis of the teeth is because of the ectodermal tissues do not proliferate and differentiate into highly specialized cells which make up the tooth buds [4,5].

There is no single etiology for anodontia. It has been proposed that partial or complete anodontia is an evolutionary stage which will result in mankind not having teeth [6]. Three syndromes characterized by partial or complete anodontia are oculomandibulodyscephaly, mesoectodermal dysplasia, and ectodermal dysplasia [7]. Oculomandibulodyscephaly is characterized by microphthalmia, blue sclera's and microcephaly. In addition, permanent teeth are absent, but the primary teeth endure. Symptoms of mesoectodermal dysgenesis include a wide face, eye deformity, muscle dystrophy, underdeveloped premaxilla, and sometimes complete hypodontia [8]. Although these two conditions are extremely rare and very few cases have been reported so far.

Ectodermal dysplasia is also a rare group of inherited disorders characterized by aplasia or dysplasia of tissues of ectodermal origin, such as hair, nails, teeth and skin [9]. The X- linked recessive ED (Christ-Siemens-Touraine syndrome) is the most common disorder (80%of EDs); it affects males and is inherited through female carriers. Genetic studies regarding the etiology of ED reveal that mutations

in the ectodysplasin-A and ectodysplasin-A receptor genes are responsible for X-linked and autosomal hypohidrotic ectodermal dysplasia. It is characterized by the triad of signs comprising sparse hair, abnormal or missing teeth (anodontia or hypodontia) and inability to sweat due to lack of sweat glands. The lack of teeth and the special appearance were reported to be major concerns. Most patients with EDs have a normal life expectancy and normal intelligence. However, the lack of sweat glands may lead to hyperthermia, followed by brain damage or death in early infancy, if unrecognized. Thus, an early diagnosis is important. Families with ED should therefore be offered genetic counseling [10].

The present article reports 2 cases of ectodermal dysplasia and their management.

Case 1

A 20 years old male patient reported to the Department of Periodontics, CSMS Dental College and Hospital, Aurangabad with a complaint of inability to chew food and also complained of retained milk teeth and missing permanent teeth. His family history showed that parents were normal but his younger brother affected (discussed as case 2 below).

The skin of the patient was dry and rough. Extra oral examination showed a senile facial appearance. His eyebrows and eyelashes were sparse. Additionally, he showed a prominent forehead and everted lips in the profile. However, the shape of the nails of the fingers and toes seemed to be normal. Patient profile was concave with a relative prognathic mandible (Figure 1). Patient was also having problem of lack of sweating. Intraoral clinical examination revealed the over-retained deciduous maxillary canines, first molars and maxillary right second molar which were firm and healthy (Figure 2). There were no teeth in mandibular arch (Figure 3). Dentition was characterized with generalized spacing.



Figure 1: Case 1 facial profile.

Panoramic radiographs and cephalogram revealed complete absence of permanent tooth buds and atrophy of the alveolar process of the mandible (Figure 4).



Figure 2: Case 1 maxillary arch.



Figure 3: Case 1 mandibular arch.

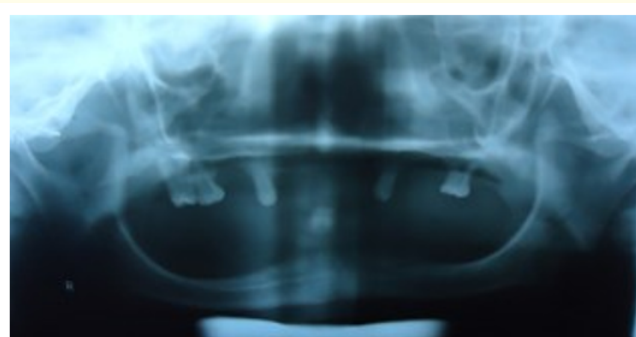


Figure 4: Case 1 OPG.

The Patient was rehabilitated with removable partial denture for maxilla and mandible (Figure 5).



Figure 5: Case 1 after denture placement.

Case 2

A 16 years old male patient, brother of patient mentioned in case #1, reported to the Department of Periodontics, CSMSS Dental College and Hospital, Aurangabad with a complaint of missing teeth and inability to chew food.

The skin of the patient was dry and rough. The patient showed a senile facial appearance and fine sparse hair on the scalp, especially on the temporal region. His eyebrows and eyelashes were also sparse, with a prominent forehead, a saddle nose and everted lips (Figure 6). Shape of the nails of the fingers and toes appear to be normal. This patient too had hypohidrosis.



Figure 6: Case 2 facial profile.

Intraoral clinical examination revealed that the presence of maxillary deciduous central incisors and molars. Both the deciduous maxillary central incisors showed slight distolabial rotation (Figure 7). There was complete absence of mandibular teeth (Figure 8).



Figure 7: Case 2 maxillary arch.



Figure 8: Case 2 mandibular arch.

Panoramic and cephalometric radiographs showed complete absence of permanent teeth and atrophy of the alveolar process of the mandible (Figure 9). Removable partial denture for maxilla and mandibular complete denture was given (Figure 10).

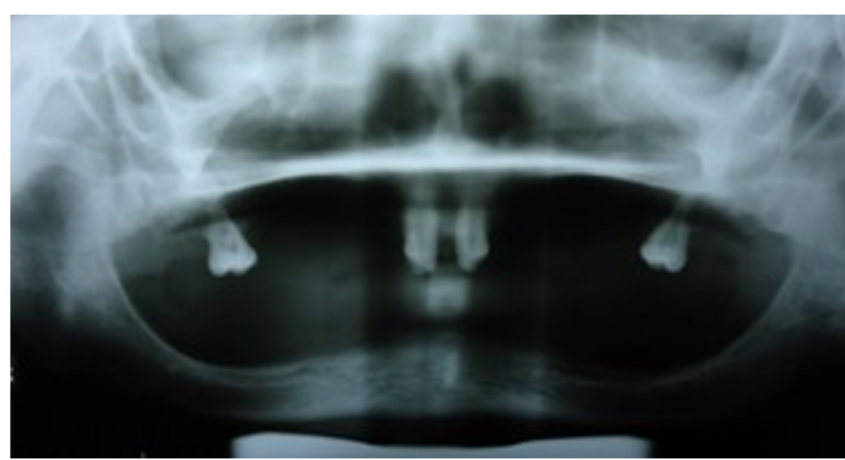


Figure 9: Case 2 OPG.



Figure 10: Case 2 denture placement.

Discussion

Charles Darwin first documented the earliest accessible account of Ectodermal Dysplasia in English in the 1840s after he had received some correspondence from a Medical Officer in the Indian army. The Officer had observed families in Punjab whose grandfathers and grandsons were affected by the lack of hair and sweat glands, which is a major hazard when working out in the fields in India. These people survived the heat by tipping buckets of water over each other. There may possibly be earlier accounts than Darwin's, but these do not appear to have been documented. However, in the mid-19th century it appears nobody understood the sex-linked inheritance and it wasn't until 1910 that this began to be understood [11].

Ectodermal Dysplasia is an extremely rare condition. The prevalence of ED is unknown; however, the incidence in male is estimated at 1 in 100,000 births although the condition is usually overlooked in infants [12]. This X-linked recessive disorder affects males and is inherited through female carriers. This carriers-incidence is probably 17.3 in 100,000 women [13].

Guckes and colleagues [14] found that the permanent teeth most likely to be present in 52 ED patients were the maxillary central incisors (42%), followed by the maxillary first molars (41%) and mandibular first molars (39%).

Cautely R [15] reported the case of a young man aged 20 years, who had only deciduous teeth present. Both jaws were abnormal, the mandible especially, being small, underdeveloped and out of proportion to the rest of the face. His hair and sweat glands were quite normal. He had a brother and sister who were similarly affected. The mother and father and former generations as far as could be ascertained were affected. Similarly, present case 1 and 2 were siblings.

Thomas Balshi, *et al.* [16] reported 20 years old male patient diagnosed as a case of hypohydrotic ectodermal dysplasia. The patient initially presented with a 2 maxillary permanent teeth (central incisors), 6 maxillary primary posterior teeth and 6 mandibular primary teeth (both canines and all first and second molars). Thomas Balshi, *et al.* extracted all the teeth and simultaneously placed the implants.

Treatment should be commenced as soon as possible in order to avoid possible resorption and atrophy of the alveolar ridges, and to control the vertical dimension, which can be severely affected by the total or partial lack of teeth.

Lifetime preservation of the primary teeth and alveolar ridges is a problem rarely encountered by the clinician. Based on the number of reported cases, complete anodontia is more frequent than anodontia of the permanent teeth only. The objective of treatment is to maintain the primary teeth by protecting the thin enamel cap and dentin from occlusal abrasion and trauma. This protection may be achieved by restoring the posterior teeth with full crowns and prescribing a mouthguard. In addition, esthetic facings may be placed on the anterior teeth to increase the mesial-distal diameters and to close diastemas [17]. In both the cases all the measures were taken to preserve deciduous teeth and the remaining teeth were replaced by removable partial denture.

Beierle and Jorgenson [18] found 14 cases of complete anodontia of primary and permanent teeth. They also described 3 cases in which primary teeth were present but the permanent teeth were missing. In present case reports, in both cases complete anodontia was not found.

Mehmet Bani [19] reported 8 years and 3 years old male patients as a case of ectodermal dysplasia's. The lack of primary and permanent teeth in the oral cavity resulted in dietary problems of both the patients. Both the patients were given removable dentures as a treatment part. As both the patients were having deciduous teeth, they had no problem of mastication in early childhood.

The fabricating procedure of dentures for the young patient was not so complicated, although the treatment requires knowledge of growth and development, behavioral management and the skills for fabricating dentures [20].

The future treatment planned for these patients after the exfoliation of primary dentition was the construction of complete dentures to carryout regular functions of the oral cavity and musculature of mouth.

Conclusion

Ectodermal dysplasia is an extremely rare condition in which complete or partial anodontia and hypotrichosis (sparse hair) and hypohidrosis (lack of sweating) are main features. Here two such typical cases are presented (Table 1) along with their management. While managing patients with Ectodermal dysplasia, only rehabilitative treatment can be provided.

Characteristic features	Case 1	Case 2
Anodontia	✓	✓
Palmer Planter Hyperkeratosis	✓	✓
Hypotrichosis	✓	✓
Hypohydrosis	✓	✓
Anhydrosis	✗	✗

Table 1: Some characteristic features of both cases.
(✓ indicates presence of feature, ✗ indicates absence of feature).

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