ANHIDROTIC ECTODERMAL DYSPLASIA

A CASE REPORT

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ABSTRACT

The anhidrotic ectodermal dysplasia is a group of rare inherited disorders that affect various tissues of ectodermal origin like hair, nails, teeth, skin and sweat glands. The management of the patient with anhidrotic ectodermal dysplasia requires multidisciplinary specialists. Oral and facial rehabilitation is the prime concern for all dental professionals. For rehabilitation it is necessary to consider the age, no of teeth present, the state of growth and aspects of social integration of the patient. The review of literature of anhidrotic ectodermal dysplasia and the prosthodontic rehabilitation of a 9-year-old boy is presented and discussed.

INTRODUCTION

Hereditary anhidrotic ectodermal dysplasia is a specific syndrome characterized by a congenital dysplasia of one or more ectodermal structures and their accessory appendages, manifested primarily by hypohidrosis, hypotrichosis and hypodontia.[1] Thurman first described the anhidrotic ectodermal dysplasia in 1848.[4,7] It is also known as hypohidrotic ectodermal dysplasia or Christ-Siemens-Touraine Syndrome (Friere-Maia and Pinheiro, 1980).[2,3,4] It has been proposed that a decreased expression of epidermal growth factor receptor plays a causal role in the anhidrotic ectodermal dysplasia (Vargas et al. 1996).[3,4] The incidence of anhidrotic ectodermal dysplasia is estimated to be 1 per 10,000 to 1 per 1,00,000.[5] The anhidrotic ectodermal dysplasia is a recessive X-linked disease. There are also autosomal recessive and autosomal dominant forms of the disorder. Sex predilection is more towards males.

REVIEW OF THE LITERATURE

X-linked anhidrotic ectodermal dysplasia is found mostly in males than in females (Carola Duran-McKinster).[3]

Pinheiro and Friere-Maia[4] (1979) reported hereditary hypohydrotic ectodermal dysplasia in a large Brazilian Kindred with multiple affected individuals over 6 generations. 13 males and 27 females were variably affected.

Nakata et al.[4] (1980) reported the clinical findings suggesting hereditary anhidrotic dysplasia in 23 affected males from 15 families as well as 21 mothers. 7 other male family members were also reported to be affected.

Saksena and Bixler[4] (1990) found 13 families associated with this disease among which 16 males were affected.

Crawford et al.[4] (1991) reported the similar cases in 34 British families.

Thomas and Glenn[6] (2002) reported a 20 year old male diagnosed with hypohidrotic ectodermal dysplasia.

Guckes and colleagues[6] found that the permanent teeth most likely to be present in 52 ectodermal dysplasia patients were the maxillary central incisors (42%), followed by maxillary first molar (41%) and mandibular first molar (39%). Mandibular anterior teeth were the least likely to be present.

Though anhidrotic ectodermal dysplasia are common in male than in female, there has been many cases being reported affecting the latter.

Singh et al.[4] (1962) described a severe case in a 27 year old Sikh woman in India. Richards and Kaplan[4] (1969) described a female infant with neonatal pyrexia due to anhidrotic ectodermal dysplasia.

Gerald and Brown[4] (1974) noted a girl with severe manifestation of the disease.

Clark et al.[4] (1990) found abnormal skin temperature pattern consistent with altered peripheral vascular perfusion in heterozygotes for X-linked hereditary ectodermal dysplasia.

MacDermot and Hulten[4] (1990) diagnosed a girl with hypohidrotic ectodermal dysplasia with moderately severe mental retardation.

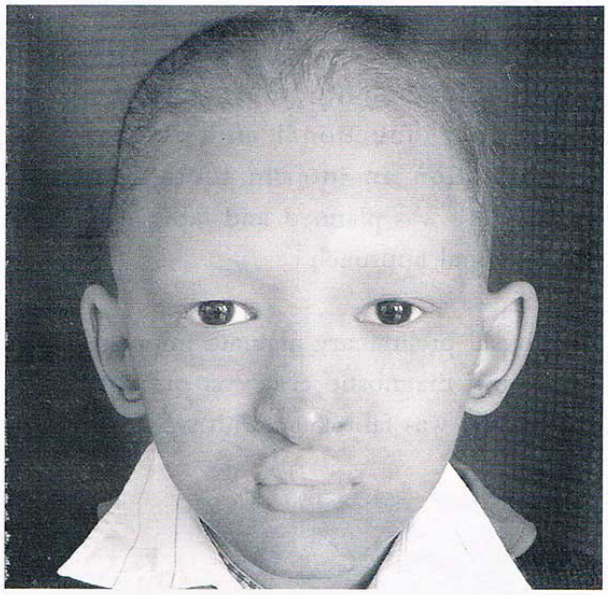
Zank et al.[4] (2001) also described female monozygotic twins with this disease.

Ghosh and Das[5] (2004) also reported this disease in 2 daughters, one 4 years of age and another 1 year of age from Muslim family in West Bengal, India.

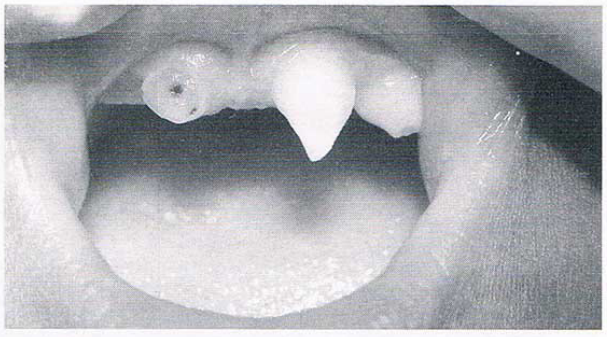
CASE REPORT

A 9-year-old boy from Lagan, Kathmandu reported to the Department of Pedodontics and Preventive Dentistry, Peoples' Dental College and Hospital with the chief complaint of missing teeth and difficulty in chewing foods. He was then referred to the Department of Prosthodontics for the needful rehabilitation. The child parents stated that he was also psychologically disturbed because his friends used to tease him on his abnormal appearance and speech.

The family history revealed that none of the other family members and their primary or secondary relatives being affected by similar illness. There was no prior history of hyperpyrexia, rhinitis and/or pharyngitis and use of antibiotics. The patient experienced burning sensation on taking hot and spicy foods.



The extra-oral examination revealed prominent frontal bone and supraorbital ridges, depressed nasal bridge, absence of eyebrows, few and fine eye-lashes. The other findings were large, low set ears, protuberant lips, sunken cheeks, soft, dry skin and sparse, fine, short, brown hair on the head while the nails were thin and brittle. The eyes were also dry.



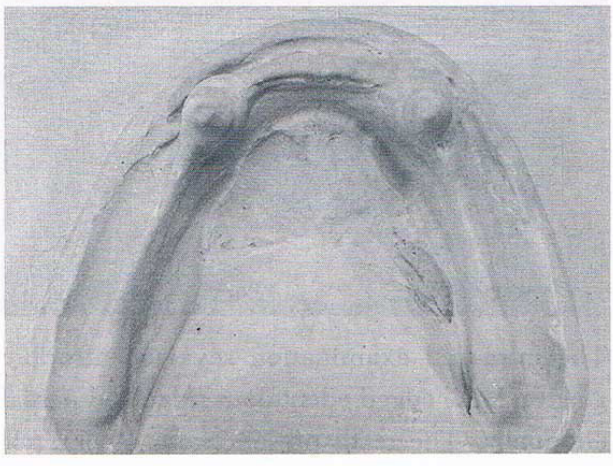
The intra-oral examination revealed atrophic gingiva, knife edged edentulous ridge and partial anodontia. The teeth present were 16, 11, 21,

23, 33 and 43. All these teeth were malformed. All the anterior teeth were conical and pointed while the molar was smaller in size.

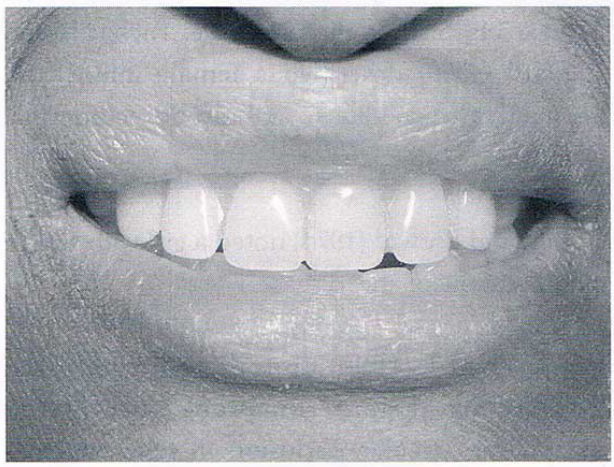
Advanced investigations (biopsy of the mucus membrane, biopsy of the skin, genetic testing and radiographic examination) which were mandatory for the confirmed diagnosis of the disease were planned for later date due to the unwillingness of the parents. All the clinical findings were suggestive of anhidrotic ectodermal dysplasia.

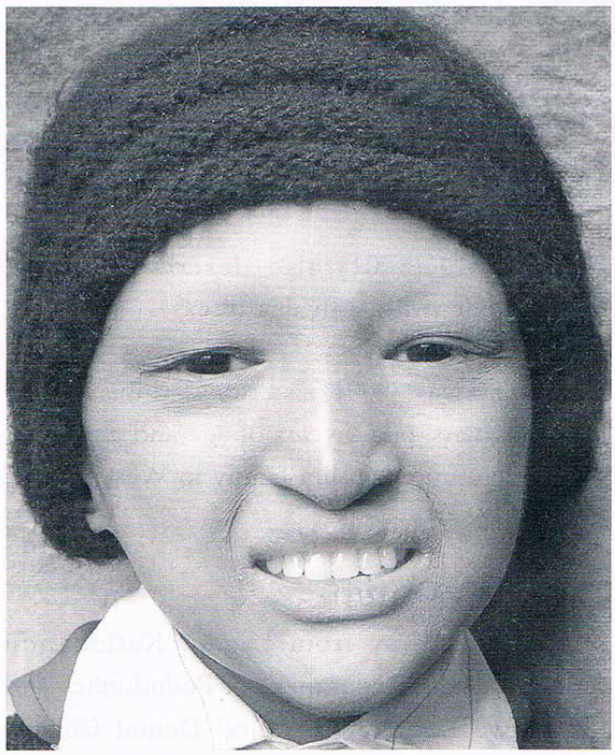
For the immediate improvement of his appearance, functional and psychological rehabilitation an interim tooth supported overdenture was planned and fabricated in the conventional approach.

First of all preliminary alginate impression was made and diagnostic cast was prepared. The special tray was fabricated with self cure acrylic without stops and spacers. The border molding was done with greenstick compound and the dual impression was made with light body elastomer and alginate.



The maxillomandibular relation was recorded initially with posterior occlusion rim and waxing of anterior segment was done later to harmonize labial fullness and visibility. Selection of teeth, arrangement and try-in was done in conventional manner. Finally the interim overdenture denture was inserted.





Instructions after care including limitation of denture was properly explained to the patient and his parent.

Follow up was done at the regular basis, the patient until this date is learning to masticate, speak, smile and adapt to his new prosthesis.

He will be kept on thorough follow up for the period of 6 months till 16-18 years of age after which the treatment will be planned accordingly. Then he will be given either overdenture (tooth/ implant supported) or telescopic denture.

DISCUSSION

Anhidrotic ectodermal dysplasia is inherited as an X-linked recessive genetic trait and boys who only have 1 X chromosome often have pronounced symptoms. Girls who have two X chromosomes and if one of these carries the predisposition for the syndrome it is compensated for by the other, normal chromosome. As a result women with the predisposition for the syndrome usually have less pronounced symptoms.[7]

The characteristics clinical manifestations of anhidrotic ectodermal dysplasia include dry skin, hypotrichosis, and total or partial anodontia (with frequent malformation of the teeth present). The bridge of the nose is depressed, the supraorbital ridges and frontal bones are pronounced and the lips are protuberant.[1,2,4,7,9]

The management of this syndrome is multidisciplinary one. Till date there is no pharmacological treatment available for the cure of anhidrotic ectodermal dysplasia. The dental treatment of the patient with anhidrotic ectodermal dysplasia should be planned in such a way that many possibilities for the treatment could be used in the furture. Dental management also requires several follow up for good esthetics and functional results if treatment is initiated in growing individual. If dentures are given they require multiple adjustments and replacements as the child grows. When the growth of the patient ceases an overdenture (tooth/implant supported) should be considered as a better option.

The patient is advised for restriction of the physical exertion. Since individuals with the syndrome do not have a satisfactory normal system for heat regulation, it is necessary for them to drink liquids in large quantities and to avoid physical exertion.

Avoidance of the warm climates is suggested. The importance of light clothing, a cool water spray bottle should be discussed with the parent. The actopic eczema can be treated with cortisone ointment. The patient also needs psychological support from their friends and teachers.[3,7]

Artificial tears can be used to prevent damage to the cornea in patients with defective lacrymation.[7]

There is hypoplasia of nasal and pharyngeal mucous membranes which leads rhinitis and pharyngitis.[1,3,4] The dry mucus membranes may make it difficult to blow one's nose, and may result in the development of hard scabs and an unpleasant odour. It may therefore be necessary to rinse the nose with saline solution morning and evening, or see an ENT specialist for occasional rinsing.[3,7]

Patients with severe alopecia may wear wigs to improve their appearance.[3,7]

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