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# Prosthetic Rehabilitation of Ectodermal Dysplasia in a 14 Year Old Child: A Case Report

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## ABSTRACT

Ectodermal dysplasia is a diverse group of genetic disorders that involve defects of the ectodermally derived structures and their accessory appendages including hair, nails, teeth, skin and glands. Other parts of the body, such as the eyes or throat, may be affected as well. Oral signs and symptoms are present in most of patients. The aim of this article is to report a case of child affected by ectodermal dysplasia with partial anodontia and to summarize some of the literature on current knowledge of oral manifestations and orofacial function as dental manifestations can be crucial for differential diagnosis of this disorder.

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***Keywords:*** Hair and nails abnormalities, hereditary ectodermal dysplasia, skin, teeth.

## INTRODUCTION

Ectodermal dysplasias are a heterogeneous group of hereditary, non-progressive disorders characterized by developmental dystrophies of more than one ectodermal structure. They are genetic disorders, which mean caused by mutations in various genes; the mutations may be inherited from a parent, or normal genes may become mutated at the time of fertilization.[1] The genes responsible for this syndrome might be either mutated or deleted; some of them are Xq-12-q 13.1, PVRL1 and GJB6. The risk for parents to have an affected child depends on the inheritance pattern of the specific type of ectodermal dysplasia with which the individual in the family has been diagnosed.[2] The different types of inheritance patterns that occur depending on the specific type of ectodermal dysplasia in the family include x-linked recessive, autosomal dominant, and autosomal recessive.[3] About ninety-five percent of affected individuals with this disorder have the X-linked recessive inheritance.[4] Thurnam published the first report of a patient with ectodermal dysplasia in 1848, but the term ectodermal dysplasia was coined by Weech in 1929.[5] Prevalence of the disorder is approximately 1 in 1, 00,000 live birth. More than 192 different varieties of these clinical conditions have been recognized. The most common form of ectodermal dysplasia usually affects men.[6] Other forms of the disease affect men and women equally. Some significant syndromes include Rapp-Hodgkin hypohidrotic ectodermal dysplasia, ectrodactyly ectodermal dysplasia, ectrodactyly-ectodermal dysplasia-clefting syndrome, trichorhinophalangeal syndrome, oral-facial-digital syndrome, nail dystrophy-deafness syndrome, trichodento-osseous syndrome, Johanson-Blizzard syndrome, Strandberg-Ronchese's Syndrome, Rosseli-Gulienetti Syndrome and various others.[7]

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## CASE SERIES

A 14-year-old boy was referred to the Department of Oral Medicine, Diagnosis & Radiology, and Institute of Dental Education & Advance Studies with chief complaint of missing teeth in both jaws. Patient gave history of unerupted deciduous and permanent dentition. On general examination nails and palm were found to be normal. There was a history of reduced sweating and heat intolerance. On extra oral examination, sparse fine hair, dry skin, reduced vertical height of facial lower third and prominent chin and lips were observed. The nasal bridge was depressed. [Figure 1]. Intraoral examination revealed left permanent second molar tooth was only present in oral cavity. The oral mucosa & palate was normal. A clinical diagnosis of hereditary ectodermal

dysplasia was made based on the history and cutaneous features. No other sibling had similar clinical features. None of the family members was involved in a similar manner in previous generations. There was no history of consanguineous marriage of parents. A digital panoramic radiograph was taken revealing missing dentition and absence of impacted teeth. [Figure 2] Treatment plan included making removable partial prosthesis for maxillary and mandibular jaws to improve appearance, function and speech. In order to fabricate removable partial denture, two impressions were made with a hydrocolloidal material (alginate); first with stock tray and then with custom tray. Jaw relationship was recorded using temporary base and wax rim. The arranged teeth were verified in the mouth during the try-in appointment. After laboratory processing, the denture was delivered to the patient [Figure 3]. In the delivery appointment, instructions were given to maintain proper oral hygiene. Continuous follow-ups every six months were planned for adjustment of existing denture.

**Figure 1: Clinical photograph depicting sparse hair, eyebrows, depressed nasal bridge & everted lips**

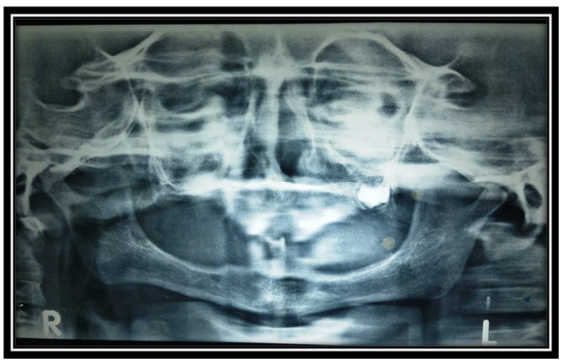


**Figure 2: Panoramic radiographic reveals only tooth present 27**

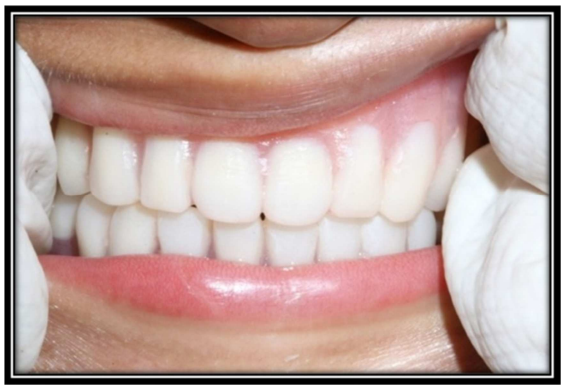

**Figure 3: Prosthetic rehabilitation**

## DISCUSSION

Hereditary congenital dysplasia of one or more ectodermal structures and their accessory appendages is Ectodermal dysplasia. Charles Darwin cited Wedderburn as having found the disorder in an Indian from the subcontinent.[8] The first classification system of the ectodermal dysplasias was proposed by Freire-Maia and Pinheiro in 1982 with additional updates in 1994 and 2001.[9] Their original classification system classified the ectodermal dysplasias into 10 different subgroups according to the presence or absence of (1) trichodysplasias or hair anomalies, (2) dental abnormalities, (3) onychodysplasias or nail abnormalities or, and (4) dyshidrosis or eccrine gland dysfunction.[10] Perabo et al reported that Danz recorded it in 1972.[10] In 1913, Christ further defined it as congenital "ectodermal defect".[11] In 1929 because of non functional sweat glands Weech coined term "anhidrotic ectodermal dysplasia".[12] Ectodermal dysplasia is broadly classified into two broad categories. One is the hypohidrotic form which is X-linked, characterized by the classical triad of hypodontia, hypotrichosis and hypohidrosis, which is also termed as Christ-Touraine Syndrome. The other category is that of the hidrotic form described by Cloustan which usually spares the sweat glands and can affect the teeth, hair and nails.[13] Felsher in 1944 changed the adjective anhydrotic to hypohydrotic because the persons with hypohydrotic form are not truly devoid of all sweat glands.[14] Patients affected with this disorder have a tendency to develop hyperthermia with physical exertion or exposure to a warm environment because of their severely diminished ability to sweat, and affected infants often present with recurrent high fevers. The scalp hair, eyebrows, and eyelashes are sparse, fine, and oftentimes lightly pigmented. Our patient had loss of eyebrows and sparse, thin, lightly pigmented scalp hair Other cutaneous features include peeling of the skin during the neonatal period, hyper pigmentation of periorbital region and wrinkles, facial sebaceous hyperplasia, and eczematous dermatitis. Symptoms also include

disfigured nails, toenails may be absent, thick, abnormally shaped, discoloured, ridged, slow growing, or brittle difficulties with the nasal passages and ear canals, lack of breast development and recurrent infections. Skin may be parched or satiny smooth, prone to rashes, and slow to heal. In contrast to several other types of ectodermal dysplasia, nails were normal.[15] Oral manifestation includes conical and pointed teeth usually affecting incisors and/or canines, anodontia or hypodontia, generalized spacing, taurodontism of deciduous molars, cleft palate and lip. Clinical differential diagnosis include: focal dermal hypoplasia syndrome, incontinentia pigmenti, Naegeli-Franceschetti-Jadassohn syndrome, pachyonychia congenital, alopecia areata & aplasia cutis congenital.[16] Esthetics, speech and masticatory efficiency is of prime concern for oral rehabilitation of patients with ectodermal dysplasia. Therefore is necessary to improve sagittal and vertical skeletal relationships during craniofacial growth and development. Removable prosthesis is the most common treatment plan. For adolescents Implant-supported denture is also suggested as the ideal reconstruction modality. The main problem in implant placement is bone density. Bone grafting is essential in case of implant placement in case bone atrophy progresses. In addition to the psychological effects particularly in young children, implant surgery is accompanied by a higher risk of failure compared to that of more conservative prosthetic treatments. Implants act similar to ankylosed teeth if placed in a growing child & may lead to some cosmetic problems. Along with the craniofacial growth, implant over-structures may not be in occlusion with opposite teeth and even the adjacent teeth may tilt into the space. Thus implant supported prosthesis may be less favourable, and therefore, the use of implants in young children should be considered carefully. Early prosthetic treatment is generally recommended from the age of 5 years. Regular adjustments should be done by dentists for removable partial or complete dentures due to continuous growth of paediatric patient and should be replaced when a decreased vertical dimension of occlusion and an abnormal mandibular posture are detected. Retention and stability for the prostheses is essential & it's quite difficult to obtain as parched oral mucosa and the under-developed maxillary tuberosities and alveolar ridges are problematic factors for resistance and stability of the dentures. For harmonious occlusion care should be taken about distribution of occlusal loads and extension of denture base while fabricating dentures. Anterior teeth that are of atypical conical shape may not be suitable for removable partial denture stability; they may be used as abutments for over dentures. Special attention must be paid to the impression making technique in such instances. Heat intolerance especially during warm climate and strenuous physical activity is difficult to be managed. As patients are susceptible to pulmonary infections they should be managed with great care. Regular visits to an Ear, Nose & throat surgeon may be necessary for the management of the nasal and aural concretions.

Affected individuals must have access to an adequate supply of water and a cool environment in case of extreme temperature. Cooling vests, air conditioning, a wet T-shirt, and/or a spray bottle of water make temperature bearable. However, external cooling is less effective in these patients because their heat transfer from the core to the skin is also reduced, presumably due to poor capillary dilatation.[17]

## CONCLUSION

Management of clinical manifestations of paediatric patients and adults associated with ectodermal dysplasia is of great challenge for dentists. Treatment with removable partial or complete denture is comfortable and cost effective modality, which improves function, self esteem, speech, esthetics and psychosocial condition of young edentulous patients. However, its long-term success depends on regular recall appointments and meticulous maintenance of oral and prosthetic hygiene.

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