

Hereditary Ectodermal Dysplasia: A Case Report

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Abstract

Ectodermal dysplasia (ED) is a hereditary disease characterized by anomalies in the structures of ectodermal origin. The disease affects skin, saliva, sebaceous and sweat glands (anhidrosis or hypohidrosis), hair (atrichosis or hypotrichosis), nail and teeth (anodontia or hypodontia). Oral rehabilitation is important from a functional, esthetic and psychological standpoint. A team approach, that includes input from Peadodontist, an Orthodontist, a Prosthodontist and an Oral and maxillofacial surgeon, is necessary for a successful outcome. This paper discusses a case of Hypohidrotic Ectodermal Dysplasia and its management.

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Introduction

Hereditary Ectodermal Dysplasia is a hereditary ^[1], clinically diverse, genetically heterogeneous group of conditions, characterized by developmental defects in the tissues of the embryonic ectoderm. Ectodermal dysplasia may be inherited by all *Mendelian* means of inheritance including spontaneous mutations. The ectoderm is one of the three germinal cell layers that form the early embryo. It eventually develops into the epidermis (surface skin), nails, hair, tooth enamel, sweat glands, sebaceous glands, and nerves. In addition, other derivatives of ectoderm include keratinocytes, melanocytes, endocrine glands, apocrine glands, ears, nipples, mucosa, the lens of the eye, the central nervous system, the anterior pituitary and the adrenal medulla^[2]. Any of the above tissues that is formed abnormally (dysplastic) may be characterized by the term ectodermal dysplasia. This simply implies that an end product of the ectoderm has not formed properly.

The earliest recorded cases of ED were described in 1792. Since then, nearly 200 different pathologic clinical conditions have been recognized and defined as Ectodermal Dysplasia. These disorders are considered relatively rare, 1 in 10,000 to 1 in 100,000 births. Most of the patients have normal life expectancy and normal intelligence^[3]. It was first described by Thurman in 1848. In 1875, Charles Darwin documented it amongst a Hindu family of *Scinde* where ten men in the course of four generations were affected.

Case Report

A 15 years boy reported to the Department of Prosthodontics and Crown & Bridge, with chief complaint of inability to chew food properly due to absence of teeth.

During extra-oral examination a facial *physiognomy* typical of Hereditary Ectodermal Dysplasia was observed. The patient presented with slight frontal bossing, depressed nasal bridge (saddle nose) and exhibited sparse scalp hair and eye lashes which were fine textured and light brown in colour. The skin was soft, thin and dry. The finger nails were normal while toe nails were dystrophic. The patient also presented with retracted mid face and poorly developed malar prominence. Detailed history revealed that the patient had reduced sweating, recurrent episodes of unexplained fever, intolerance to heat and used to eat large amount of ice many a times daily to keep himself cool.

Intra-oral examination revealed complete absence of teeth. The palate was shallow. Brownish pigmentation of oral mucosa with slight dry appearance was noticed. The tongue was relatively large. Evaluation of the diagnostic casts showed underdeveloped alveolar ridges with minimal height and width. OPG of the patient showed absence of all teeth in maxilla and mandible except for a developing molar in left maxillary posterior region.

Family history revealed that his father had syndactyly of left hand (Figure 1) and patient also had deformities of hands and feet. (Figure 2 & 3)



Figure 1: Picture showing patient's father had syndactyly of left hand



Figure 2 & 3 : Picture showing deformities of hands and feet of patient

Treatment plan: Since the boy was completely edentulous, so to improve the patient's appearance, masticatory efficiency and speech, he was provided with removable complete maxillary and mandibular dentures. The socioeconomic status of the patient precluded implants as the other treatment option. The basic principles of removable Prosthodontics were followed for the fabrication of complete dentures.

- 1. The primary impression was made in impression compound (Pyrax) and custom trays were fabricated based on selective pressure theory given by Boucher on the resultant primary cast.
- 2. The maxillary and mandibular custom trays were border moulded with green stick compound wax (Tracing sticks, DPI). Then the secondary impression was made with ZOE impression paste (DPI).
- 3. Maxillary and mandibular occlusal rims were fabricated on the master cast and then checked for accuracy in patient's mouth. The horizontal and vertical jaw relations were recorded and transferred to an articulator using a face bow.
- 4. Artificial teeth (Acryrock) were arranged in class-I molar relationship with minimal overjet and overbite.
- 5. Try-in was done to check stability, phonetics, jaw relation and esthetics. The trial dentures were then processed in heat polymerizing denture base resin and characterized also to match the patient's mucosal colour (pink+clear, heat cure acrylic, *Travelon HI, Dentsply*).
- 6. After finishing and polishing the dentures were inserted in patient's mouth.
- 7. Patients was given post insertion instructions and recalled for follow up assessment.

Discussion

EDs are a heterogenous group of disorders characterized by a group of findings involving defects of two or more of the teeth, skin and appendageal structures including hair, nails and eccrine and sebaceous glands ^[3,4]. It is commonly transmitted as an *X-linked recessive* disorder ^[5,6]. However rarely autosomal recessive and autosomal dominant inheritance have also been seen. In most of ED patients in addition to the delay in teething, the teeth are abnormal in shape and structure. Not only is the shape abnormal but also the number. Some cases have congenital anodontia. In the case of this patient, diagnosis was made on the basis of the classical clinical manifestation of the hypohidrotic ectodermal dysplasia.

The most common types are Hypohidrotic or Anhidrotic Ectodermal Dysplasia (Chirst-Siemens-Touraine Syndrome) and Hidrotic Ectodermal Dysplasia (Cloustan's Syndrome)^[7]. The hidrotic type was first defined in 1929 by Clouston. The term Ectodermal Dysplasia was coined by Weech in 1929.

Oral rehabilitation is important from a functional, esthetic and psychological standpoint. A team approach^[8], that include input from Pedodontist, an Orthodontist, a Prosthodontist and an Oral and maxillofacial surgeon, is necessary for a successful outcome. There is no definite time to begin the treatment but *Till and Marques (1992)* recommended that the initial prosthesis could be delivered when the child starts school, so that the child would have a better appearance and will also have the time to adapt to the prosthesis^[9].

The Prosthetic treatment should be carried out on an individual basis, aimed always towards providing good occlusal stability. Removable prostheses, including treatment partial dentures and overlay dentures, are often the treatment of choice to replace missing teeth and/or restore vertical dimension of occlusion prior to definitive treatment^[10]. Removable prostheses are easily modified or remade during the growth period, offering an easy, affordable, and reversible method of dental rehabilitation. Also, the removable prostheses are an economic and conservative alternative for patients who cannot afford other treatment options or prefer to avoid invasive surgical procedures associated with bone grafting and implant placement^[11]. As in this case, the patient was completely edentulous, we had two treatment options; conventional complete dentures or implant supported overdentures. But due to lack of well formed alveolar ridges and poor financial condition of the patient, we decided to fabricate removable complete dentures.

Kramer at al.^[12] reported the case of a boy at the age of 8 years with ectodermal dysplasia who exhibited a severe hypodontia and who was treated with implants inserted into the anterior mandible and recommended the early insertion of dental implants in children with severe hypodontia. Growing implant patients present a unique age-related problem regarding implant positioning and prosthetic outcomes. *Guler at al.* reported that dental implants with or without bone grafts can be used in patients over 12 years of age. Implant treatment is normally deferred until the jaws have stopped growing, to avoid growth related problems, which means in the very late teens or perhaps early 20's. Most patients of this age are also better able to make the lifetime commitment which this type of treatment requires^[13].

There are reports that implant treatment should be ended before puberty for optimum functional and psychosocial development (*Giray et al.* 2003). Nevertheless, reports in the literature describe placement of implants as early as 3 years (*Guckes et al.* 1997)^[14] or 5 years of age (*Hickey AJ*)^[15].

According to *Vieira at al.*^[6], prosthetic management is important for ED patients because it provides good esthetics, phonetics, and masticatory comfort, maintains healthy supporting tissues throughout a lifetime of denture wearing experience and helps the patient develop a good psychologic self-image. The prosthesis must be periodically modified in young adults as alveolar growth, erupting teeth and rotational jaw growth changes.

Conclusion

Treatment for the patient with hypohidrotic ectodermal dysplasia needs the effort of a team approach. Dental treatment should begin as early as 2-3 years. Removable denture is the treatment of choice initially in an attempt to rehabilitate the affected patients both functionally and psychologically to make them able to enjoy their normal life. However, under all circumstances the treated patient needs to be under observation long after.

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Competing Interests

None declared.

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