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Case Report **Pediatric Dentistry**

Ectodermal Dysplasia: A Case Report

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Abstract

Ectodermal Dysplasia (ED) is a rare genetic disorder characterized by the aberrant development of ectodermal structures, presenting complex dental, dermatological, and psychological challenges. This case report focuses on a 7-year-old male with ED, delving into his prenatal, natal, and postnatal history, medical and dental records, clinical examination findings, and the subsequent treatment plan. The patient exhibited classical ED features, including missing teeth, dry and fragile skin, prominent lips, cranial abnormalities, reduced facial height, and an enlarged tongue. A multidisciplinary team approach was pivotal in addressing the intricate dental complications associated with ED. Preventive measures such as meticulous oral hygiene, periodic high-fluoride applications, and resin-modified glass ionomer fissure sealants were administered. Prosthodontic interventions were designed to restore function and aesthetics, including upper partial denture and lower overdenture. This case underscores the significance of early and comprehensive dental intervention in ED patients, emphasizing the vital roles played by pediatrics, orthodontics, prosthodontics, surgery, and periodontics in achieving favorable outcomes. Furthermore, it highlights the psychosocial challenges faced by ED individuals, emphasizing the need for holistic care and support.

Keywords: Ectodermal Dysplasia, Genetic Disorder, Multidisciplinary Approach, Dental Management, Prosthodontics.

INTRODUCTION

Ectodermal Dysplasia (ED) is a rare and complex genetic disorder characterized by the defective development of ectodermal structures, including teeth, hair, nails, and sweat glands [1]. It presents many challenges across multiple domains, necessitating a comprehensive and multidisciplinary approach for effective management. In recent years, ED has garnered increased attention due to its impact on the oral health and quality of life of affected individuals [2].

The etiology of ED is multifactorial, encompassing various genetic mutations that affect the formation and function of ectodermal-derived tissues. While fewer than 20% of specific gene mutations and their chromosomal locations have been identified, more than 170 subtypes of ED have been recognized [3]. One of the most common forms is hypohidrosis ectodermal dysplasia (HED), often associated with X-linked inheritance, primarily affecting males. This condition reduces sweating, leading to heat intolerance and occasional fatalities in infancy due to hyperthermia [4].

The clinical manifestations of ED are diverse and can include fine and sparse hair, decreased eyebrow and eyelash density (Trichondysplasia), periocular skin changes such as fine wrinkling and hyperpigmentation, hypoplastic maxilla, and protuberant lips [5]. Additionally, individuals with ED commonly exhibit xerostomia, an abnormal dryness of the mouth, which contributes to an increased risk of dental caries. Dental anomalies are a hallmark of ED, with affected individuals presenting with oligodontia or hypodontia, characterized by a reduced number of primary and permanent teeth. These teeth often display distinctive conical shapes and constricted crowns, further complicating oral health management [6].

The challenges posed by ED extend beyond the clinical and biological aspects. Psychosocial implications play a significant role, as individuals with ED may experience social and psychological difficulties due to their abnormal appearance and functional limitations [7]. Rejection by peer groups during childhood may lead to long-term psychological issues,

including aggression, delinquency, and mental health problems in adulthood.

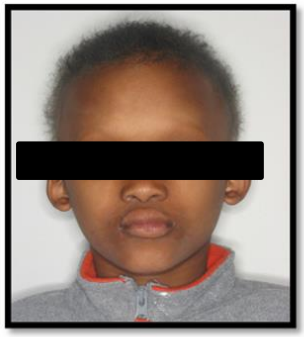
Managing ED requires a multidisciplinary team approach involving various dental specialists, pediatricians, orthodontists, prosthodontists, surgeons, and periodontists. This approach encompasses various stages of intervention, including early dental treatment throughout childhood, interceptive orthodontics, and prosthodontic solutions like partial dentures, complete dentures, and overdentures [8]. Additionally, the possibility of using dental implants, which may require bone augmentation, is considered in older individuals with ED.

In this context, this report aims to provide a comprehensive understanding of Ectodermal Dysplasia, emphasizing the importance of early intervention, interdisciplinary collaboration, and psychosocial support in managing this intricate condition. It delves into a specific case study of a 7-year-old male with ED, detailing the clinical examination findings, treatment plan, and the broader implications of ED on the individual's life. Through this exploration, we seek to highlight the significance of holistic care in enhancing the overall well-being of individuals with ED.

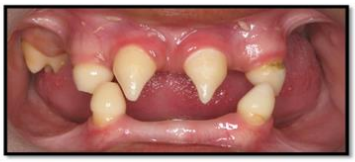
CASE PRESENTATION

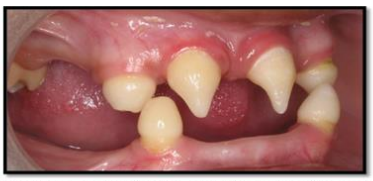
The patient in question is a 7-year-old male who presented with a chief complaint voiced by his mother. She expressed concerns about her child's missing teeth and a strong desire to restore both function and esthetics for him. The prenatal history of the patient revealed an uneventful pregnancy, with no reported sickness, where the mother primarily consumed fruits and vegetables. The patient was born full-term without complications or injuries, and there were no congenital abnormalities. In the postnatal phase, the patient was breastfed for two months and subsequently consumed a diet consisting of milk (naan), Infacare, supplemented with two teaspoons of sugar until age 2. Notably, the patient has been diagnosed with Ectodermal Dysplasia (ED), setting the stage for a distinctive clinical profile. At the age of 3, the patient underwent the extraction of teeth 51 and 61, carried out under sedation. The child's behavior stands out as positive, with satisfactory progress at school and harmonious relationships with peers. During the clinical examination, the following findings were noted: Present teeth include 16, 53, 11, 21, 63, 73, and 83, while others were missing due to underlying ED. The teeth were conical in shape, tilted and spaced Panoramic radiograph showed impacted upper and lower canines, atrophic under developed ridges.

Additionally, distinctive features associated with ED were observed, including missing teeth in both primary and permanent dentitions, dry and fragile skin leading to hypo or anhydrosis and associated risk of hyperthermia, protuberant lips, frontal bossing, reduced lower facial height, and macroglossia. Based on the clinical findings and medical history, the patient was diagnosed with Ectodermal Dysplasia (ED). The proposed treatment plan is structured around preventive measures, encompassing oral hygiene instructions, frequent fluoride applications, fissure sealants, and prosthodontics. The goal is strengthening the enamel, reducing caries risk, and restoring oral function and esthetics.

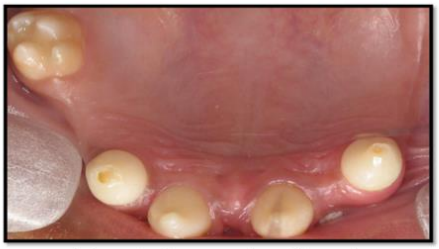


Features of ED are the prutrubrent lips, scanty hair, pale skin, saddle nose and reduced eyebrows.

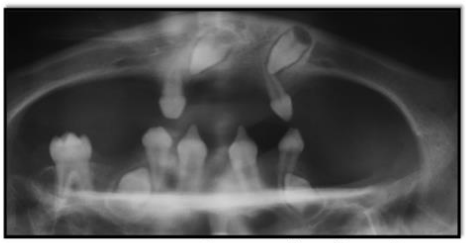




Conical shape (incisors) and (hypoplastic molars)



Extra-Oral panoramic radiograph



Extra-Oral panoramic radiograph

Figure 1: Increase in Lower facial height after placement of upper and lower over-dentures (Before Treatment)

Treatment Goals:

- Restore oral function, allowing for improved chewing and speaking abilities.
- Enhance aesthetics and boost patient confidence.
- Address the underlying oral health issues, including decay and periodontal disease.
- Provide a comfortable and stable prosthesis for long-term use.

TREATMENT PLAN

Initial Assessment:

- Comprehensive oral examination, including X-rays and diagnostic imaging.
- Medical history review
- Open communication with the parents to understand their concerns and treatment expectations.

Treatment Options Discussion:

- Presented various treatment options, explaining the advantages and disadvantages of overdentures.

Decision Making:

- Collaborated with the parents of a 7-year-old male patient in the decision-making process, considering factors such as cost, long-term benefits, and personal preferences.
- Mutual agreement on pursuing overdentures.

Impressions and Model Fabrication:

- After the initial assessment, removable upper and lower complete overdentures were planned.
- Primary impression was done with a stock metal tray with polyvinyl siloxane putty impression material.
- Final impression was done with light body polyvinyl siloxane impression material and master casts were poured.
- Face-bow records, vertical and horizontal jaw relations were taken as per standard protocols. Articulation and tooth arrangement were done on a semi-adjustable articulator.
- Bilateral balanced occlusion was provided on average values on the semi-adjustable articulator

Try-In Stage:

- The 7-year-old male patient experienced a try-in stage where a wax prototype of the overdenture was tested for fit, comfort, and aesthetics.
- Adjustments made to achieve optimal results.

Final Overdenture Fabrication:

- Fabricated the final overdenture, focusing on proper alignment, bite, and aesthetics.

Final Fitting and Patient Education:

- Fitted the final overdenture in the patient's mouth.
- Provided detailed instructions on oral hygiene, maintenance, and potential adjustments.

Follow-Up Appointments:

- Scheduled regular follow-up appointments to monitor adaptation and address any concerns.
- Follow-up was done after 1 week to check for the fit of denture. The patient was recalled after 15 days, 3 and 6 months. Parents were also told that child will need rebasing or change of denture as the growth of the jaws continues. Therefore, they were advised a follow-up after every 6 months.
- Conducted thorough examinations to ensure the health of oral tissues and the stability of the overdentures.

Ongoing Maintenance:

- Emphasized the importance of routine dental check-ups, professional cleanings, and proper oral care practices.

OUTCOME

The 7-year-old male patient successfully adapted to his overdentures, experiencing improved oral function and a boost in confidence. Regular follow-up appointments revealed optimal oral health and the stability of the prosthetic, confirming the success of the comprehensive overdentures treatment plan.

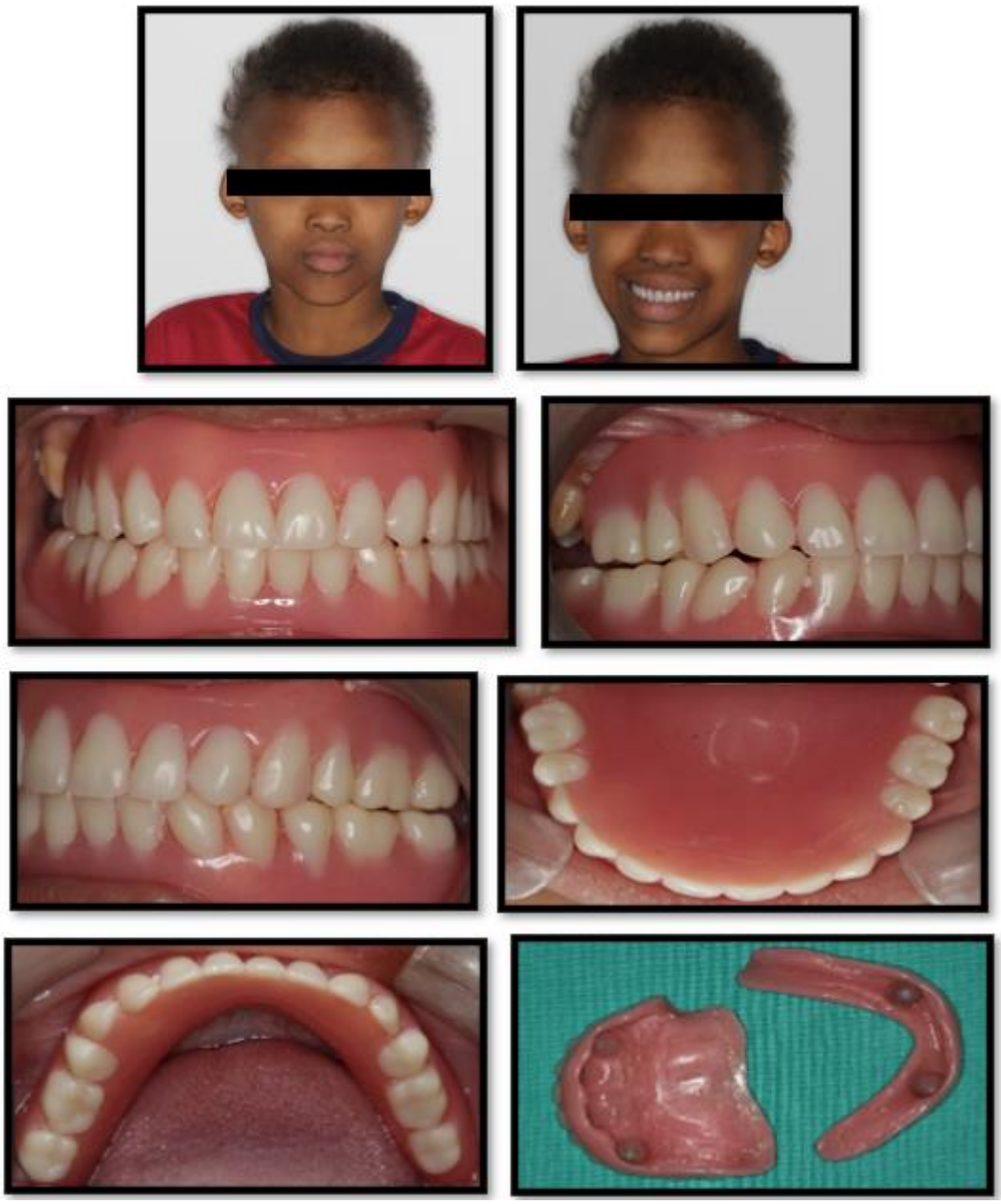


Figure 2: Increase in Lower facial height after placement of upper and lower over-dentures (After Treatment)

DISCUSSION

Ectodermal Dysplasia (ED) represents a complex and multifaceted genetic disorder that necessitates a holistic approach for comprehensive management. This case exemplifies the challenges encountered when dealing with a 7-year-old male patient diagnosed with ED. The diagnosis of ED, supported by distinctive clinical features, guides the formulation of an effective treatment plan, primarily focusing on preventive measures and prosthodontics [9]. One of the distinctive features noted during the clinical examination is the absence of several teeth in both primary and permanent dentitions, a common hallmark of ED. This phenomenon, referred to as oligodontia or hypodontia, significantly reduces the number of teeth present [10]. The affected teeth often exhibit a unique conical shape with constricted crowns, making oral health management exceptionally challenging. These dental anomalies are further exacerbated by xerostomia, a condition characterized by abnormally dry mouth due to reduced salivary flow, significantly increasing the risk of dental caries.

The primary goal of the treatment plan is to address these challenges through a combination of preventive measures and prosthodontic interventions. The preventive strategies encompass oral hygiene instructions (OHI) and oral health education (OHE), aimed at empowering the patient's mother with the knowledge and skills required for effective oral care. Additionally, frequent Durashield fluoride applications are incorporated to enhance enamel strength and

resistance to caries, given the high-risk classification associated with xerostomia and hypodontia [11].

Furthermore, fissure sealants, specifically resin-modified glass ionomer, seal fissures in the permanent molars. This strategy aims to release fluoride and withstand masticatory forces, contributing to caries prevention in compromised molars [12]. The multidisciplinary approach emphasizes prosthodontics as a critical aspect of ED management. The plan outlines the construction of a partial denture for the upper arch and an overdenture for the lower arch, utilizing teeth 73 and 83 for stabilization and retention. However, it's essential to note that the placement of these dentures is deferred due to the presence of erupting teeth, which adds complexity to the treatment process and necessitates accommodation [13].

The case underscores the significance of a collaborative and interdisciplinary approach involving dental specialists, pediatricians, orthodontists, prosthodontists, surgeons, and periodontists. This approach recognizes that ED management extends beyond the clinical aspects, encompassing psychosocial challenges that individuals with ED may encounter throughout their lives. The abnormal appearance resulting from missing teeth and other characteristic features of ED may lead to social and psychological difficulties, emphasizing the importance of psychosocial support and holistic care.

Implant placement was not considered as treatment of choice because of atrophic under-developed ridges and patient's economic status. Overdenture was a suitable choice in this specific case as anterior atypical conical teeth used as abutments to compensate the drawbacks related to retention, stability and support in the case of removable partial or conventional complete dentures.

CONCLUSION

The comprehensive management of Ectodermal Dysplasia (ED) involves a multidisciplinary approach considering the unique dental and psychosocial challenges of this condition. Preventive strategies, including oral hygiene and fluoride applications, are pivotal in mitigating dental complications. Prosthodontic solutions are essential for restoring oral function and esthetics. Moreover, recognizing the psychosocial impact of ED underscores the importance of holistic care and support to enhance the overall well-being and quality of life for affected individuals. Ongoing research and collaboration are vital for refining ED management strategies.

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