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Case Report

# X-LINKED HYPOHIDROTIC ECTODERMAL DYSPLASIA MANAGEMENT WITH REMOVABLE PROSTHESIS IN A PEDIATRIC PATIENT. CASE REPORT.

## Manejo de displasia ectodérmica hipohidrótica ligada al cromosoma X con prótesis removible en un paciente pediátrico. Reporte de caso.

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**ABSTRACT:**

Introduction: Ectodermal dysplasia is a rare genetic disorder that affects structures derived from ectoderm such as teeth, hair, nails, and sweat glands. Oligodontia is a common finding that affects the chewing, smiling and self-esteem of these pediatric patients.

Case Report: We present a case report of a 7-years-old pediatric patient who consulted with his mother showing the same condition. The intervention consisted of a removable prosthesis with satisfactory results.

Conclusión: The X-linked hypohidrotic ectodermal dysplasia represents a challenge for a pediatric dentist who offers rehabilitation to the patient according to craniofacial development, age, socioeconomic status, and the number of missing teeth.

**KEYWORDS:**

*X chromosome; Ectodermal dysplasia; Pediatric dentistry; Dental prosthesis; Denture, partial, removable; Anodontia.*

**RESUMEN:**

**Introducción:** La oligodoncia es un hallazgo común que afecta la masticación, la sonrisa y la autoestima de estos pacientes pediátricos.

**Case Report:** Presentamos un reporte de caso de un paciente pediátrico de 7 años que consultó con su madre presentando la misma condición. La intervención consistió en una prótesis de remoción con resultados satisfactorios.

**Conclusión:** La displasia ectodérmica hipohidrótica ligada al cromosoma X representa un desafío para un odontopediatra que ofrece rehabilitación al paciente de acuerdo con el desarrollo craneofacial, la edad, el nivel socioeconómico y el número de dientes faltantes.

**PALABRAS CLAVE:**

*Actitud frente a la salud; Atención odontológica; Salud bucal; Autoinforme; Hábitos; Higiene bucal.*

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## INTRODUCTION.

Ectodermal dysplasias (ED) are genetic conditions affecting the development and/or homeostasis of two or more ectodermal derivatives, including hair, teeth, nails, and certain glands. The molecular causes of these diverse conditions involve many genes and multiple developmental pathways and components of complex molecular structures required for the normal formation, structure, and function of the ectodermal derivatives. In X-linked hypohidrotic ectodermal dysplasia (XLHED) cases, there are disturbances of epithelial-mesenchymal interaction affecting endodermal and ectodermal structures, such as the defects of mucous gland formation in the lung and the colon.[1] It is characterised by hypohidrosis, sparse hair, and teeth abnormalities. In addition, infants with XLHED have an increased risk of death by hyperpyrexia.

XLHED is the most common form of hypohidrotic ectodermal dysplasia; however, no population-based prevalence estimates are available.[2] The overall prevalence of ED syndromes is unknown but appears rare, with a presumed cumulative incidence of approximately 1/1,429. More than 120 clinically and/or genetically distinct ED have been cataloged.

Thurnam first described the EC case in 1848. More than 170 different pathological clinical conditions have been described in the last ten years, all sharing common anomalies of the hair, teeth, nails, and sweat glands. Many are associated with abnormalities in other organs and systems and, in some conditions, related to mental retardation.

The abnormalities affecting the epidermis and epidermal appendages are extremely variable and clinical overlap is present among the majority of EDs. Particular clinical signs define most ED. Priolo *et al.*,[3] in 2001, classified the ED into two main functional groups: defects in developmental regulation/epithelial-mesenchymal interaction and defects in cytoskeleton maintenance and cell stability.

In the last decade, the study of the human genome has expanded the knowledge of the genes and molecular pathways causing the ectodermal dysplasias (ED), a diverse group of inherited disorders that share developmental abnormalities of two or more of the following: hair, teeth, nails, sweat glands and other ectodermal structures. In addition, several numbers of patient-oriented studies have provided insights into the cognitive abilities, growth and nutrition, allergies and asthma and women's symptoms in patients affected with EDs.[4]

There are dental implications that affect the appearance and function of ED patients. The teeth are markedly reduced in number (oligodontia or hypodontia) and often manifest abnormal development in the form that may appear tapered, conical, or pointed at the incisors. Molars can be seen reduced in size.[5] Failure to form tooth buds causes hypoplastic alveolar bone, leading to reduced vertical dimension of occlusion. Therefore, an aged appearance is common in affected individuals.[6] There is not much information on this topic in literature, which creates a knowledge gap that results in many dentists not being sufficiently trained in critical appraisal.

## CASE REPORT.

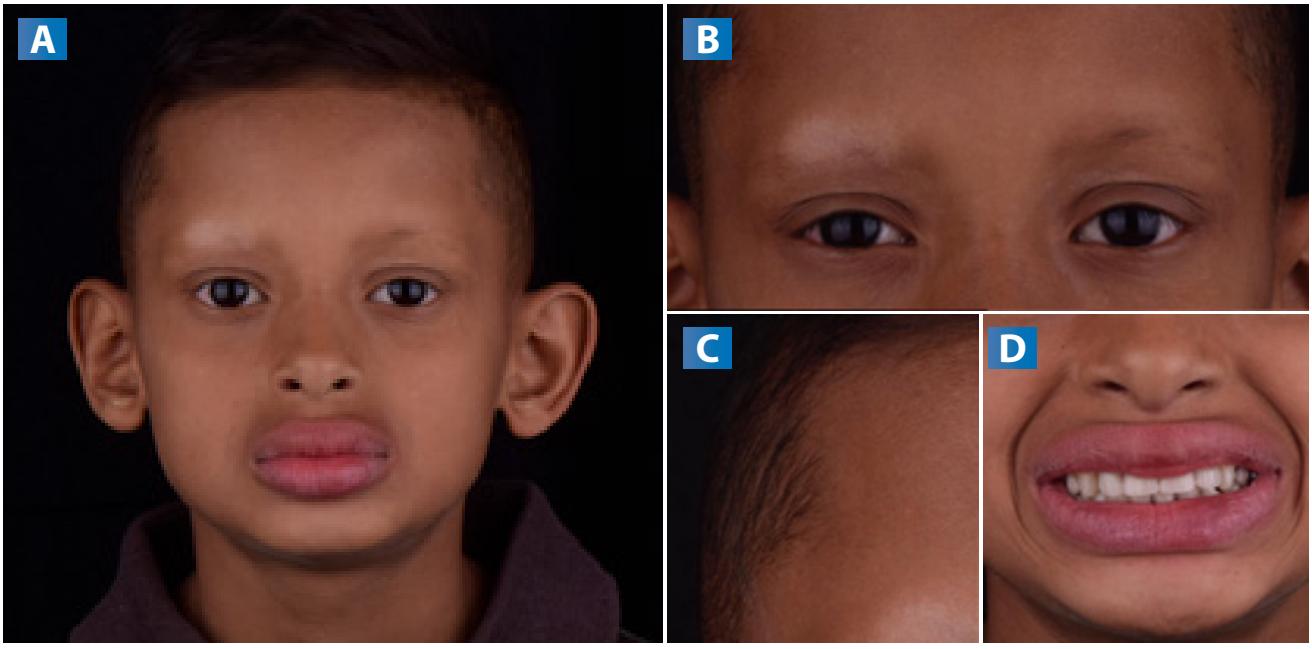
We report a seven-year-old male patient and his mother aged 35 with a chief complaint of lack of smiling by the child. The mother signed the informed consent to share the records for this case report. Patient is the product of the first pregnancy of non-consanguineous parents. Neither medical nor other systemic disease finding was reported. The mother stated no other member in the family had the condition. In the past he had not received any intervention, neither did the mother. Patient presents normal cognitive development without mental retardation or any related disorders.

A limited quantity of hair was observed upon physical examination. The eyebrows had alopecia and eyelashes were scarce; no ophthalmological disorder was found. The patient presented tiny white spots on the forehead (Figure 1), normal ear implantation, and ands, fingers and nails were normal without ectrodactyly (Figure 2). The patient reported no sweating.

At the moment of consultation, the patient had only nine teeth; no extractions were done. Oral hygiene was good and gingival condition acceptable, no caries found; no temporomandibular joint disorders were detected.

Radiographic examination with a panoramic radiograph confirmed 21 teeth were developmentally missing (including third molars) (Table 1 and Figure 3): The mother provided a panoramic radiograph of her teeth which confirmed the ab-sence of six teeth: upper lateral incisors (2), molars (2), lower central incisor (2), (Figure 4).

**Figure 1.** The patient presented tiny white spots on the forehead.




**A:** Frontal photograph. **B:** Slight white spots in the forehead with alopecia in eyebrows. **C:** Low amount of hair. **D:** Patient with removable prosthesis appliance.

**Figure 2.** Hands and nails of the pediatric patient.

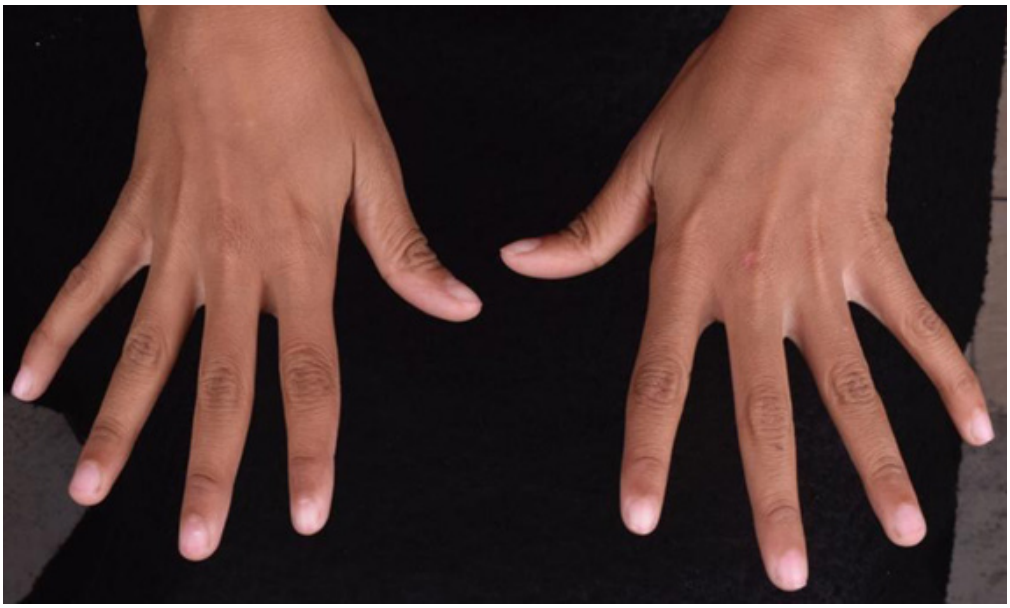



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**Figure 3.** Orthopantogram (panoramic radiograph) of the child where it can be seen the agenesis of the following teeth: upper central incisors (2), upper lateral incisors (2), upper premolars (4), third molars (2), lower central incisor (2), lower premolars (4), lower molars (5).

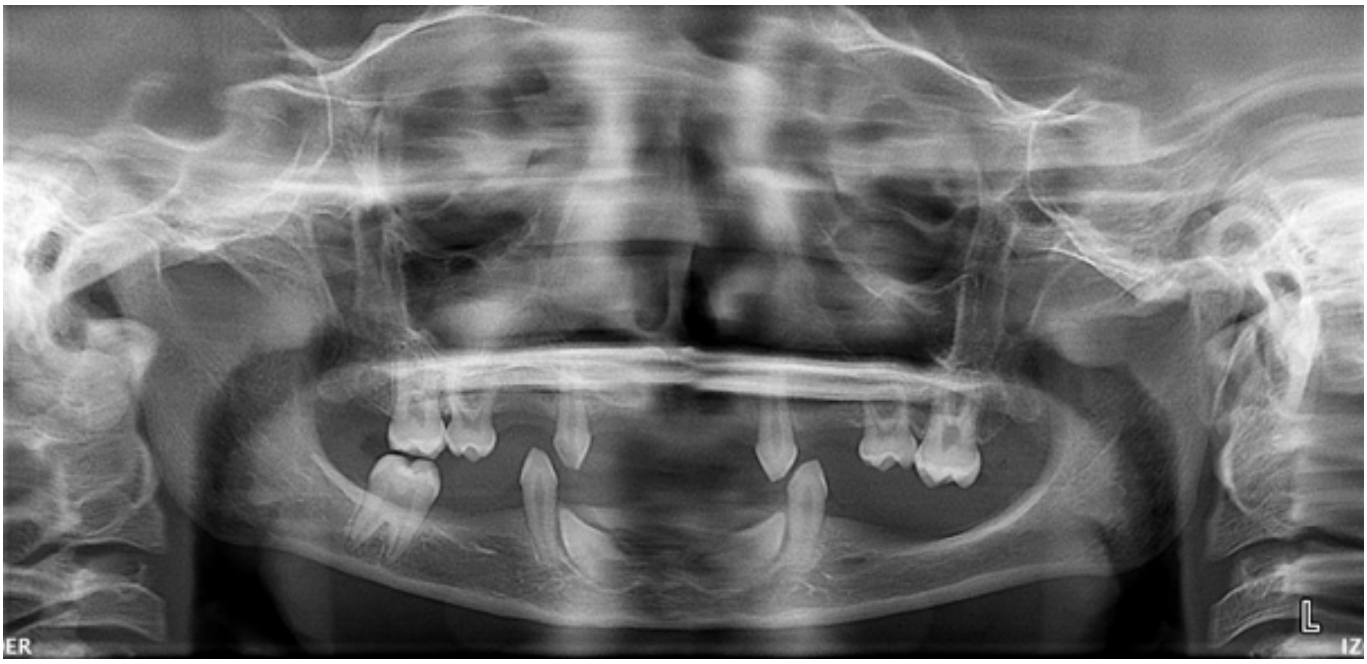


**Figure 4.** Orthopantogram (panoramic radiograph) of the patient´s mother where the agenesia of the following teeth can be observed: upper lateral incisors (2), molars (2), lower central incisor (2).

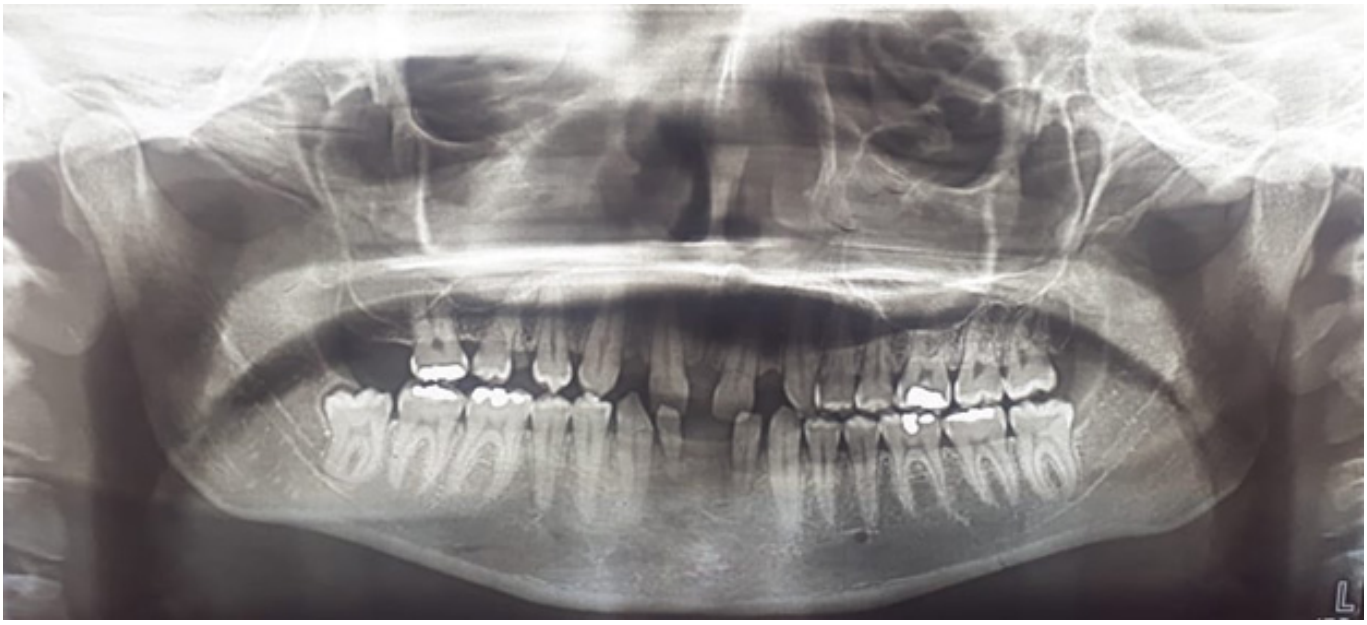

**Figure 5.** Treatment with removable prosthesis of the pediatric patient.

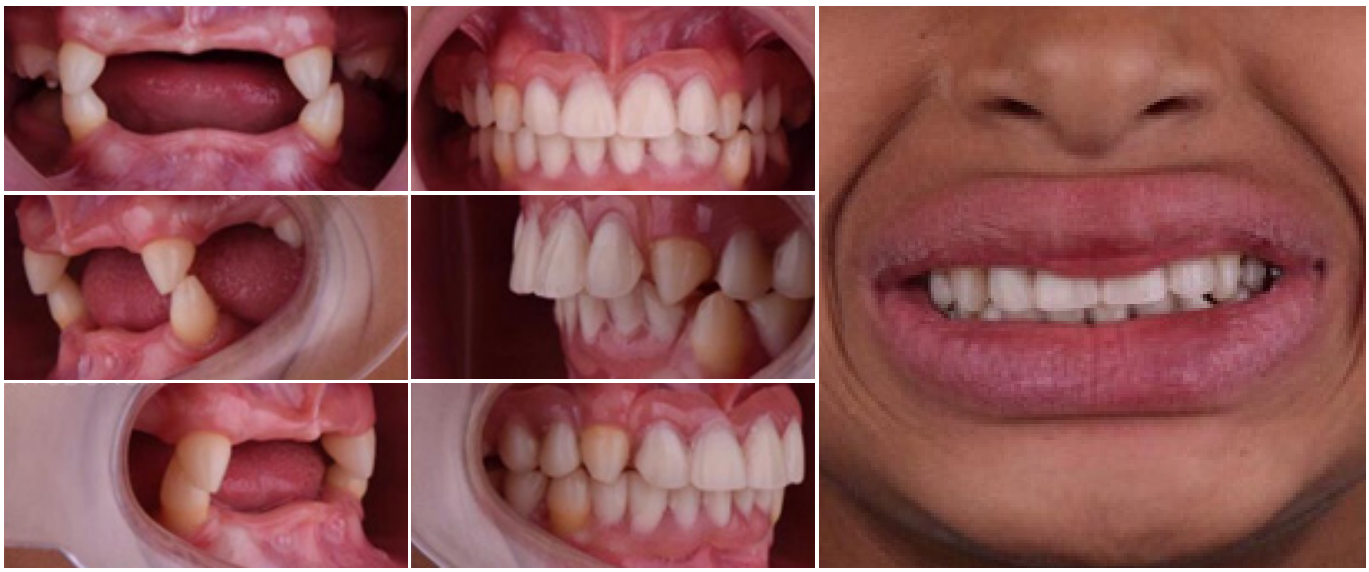

**Table 1.** Developmentally missing teeth identified from panoramic radiograph.

| Tooth type | Number of missing teeth |
|---|---|
| Upper central incisors | 2 |
| Upper lateral incisors | 2 |
| Upper premolars | 4 |
| Upper third molars | 2 |
| Lower central incisor | 2 |
| Lower premolars | 4 |
| Lower molars | 5 |

A consultation with a geneticist was done resulting in the diagnosis of X-linked hypohidrotic ectodermal dysplasia with the phenotypic features: alteration in ectodermal structures like skin, hair, sweat glands and teeth. Mother and son were both affected; however, the mother had a reduced phenotype.

## Intervention

The treatment proposed for this case was a removable prosthesis which has to be changed according to the growth and development of the pediatric patient. Therefore, appointments were scheduled four months apart to readjust the prosthesis (Figure 5).

## Follow up

The patient has been evaluated three times since the installation of the removable appliances. The positive outcomes were higher than expected. The patient improved his social integration, had better phonation and chewing and an overall better quality of life.


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## DISCUSSION.

Hypohidrotic ectodermal dysplasia is the most common type of ectodermal dysplasia, which encompasses a large group of syndromes that share several phenotypic features such as missing or malformed ectodermal structures, including skin, hair, sweat glands, and teeth. X-linked hypohidrotic ectodermal dysplasia (XL-HED) is associated with mutations in ectodysplasin (EDA1). Hypohidrosis due to hypoplastic sweat glands and thin, sparse hair are phenotypic features that significantly affect the daily lives of XL-HED individuals and therefore require systematic analysis.[7]

In the present case report, the consultation motive was the missing teeth and the smile impact. Hypodontia, or tooth agenesis, is the most prevalent craniofacial malformation in humans. It may occur as part of a recognised genetic syndrome or as a nonsyndromic isolated trait. Excluding third molars, the reported prevalence of hypodontia ranges from 1.6% to 6.9%, depending on the population studied.[8] Oligodontia is used to describe more severe forms of tooth agenesis, typically the absence of more than six teeth and the entire dentition.[9] These missing teeth marked the life of this child, who didn't want to talk, smile or interact with his mates. It's important to highlight that he only had nine teeth in his mouth, diminishing his quality of life.

The hypohidrotic ectodermal dysplasia has different etiologies, and it is more frequently associated with an X-linked pattern of inheritance caused by pathogenic variants of the EDA gene in Xq13.1. EDA encodes the protein ectodisplasin A, a signal molecule that participates in epithelium and mesenchymal development of the skin.[10]

The results of the study by Kohli *et al.*,[11] found that people with ED in the older age group (15 to 19 years old) perceived more functional problems than the younger age group (11 to 14 years old) and females who had ED presented more emotional problems than males who had ED. However, caregivers tended to report poorer oral health-related quality of life (OHQoL) for their children than did the children. No significant differences were found between children's and parents' total OHQoL and individual domain median scores. Thus, among children affected by ED, the understanding of oral health and well-being may vary by age and gender. One should be cautious while using caregivers as proxies for their children in assessing the child's OHQoL.[11]

Likewise, Hanish *et al.*,[12] designed an anonymous epidemiological survey study among people with ED to evaluate oral symptoms, satisfaction with the health system and their respective OHRQoL. When asked about oral symptoms, 110 participants provided responses, 109 (99.09%) of which described oral symptoms. The average age of the female participants at the time of diagnosis was 17.02 years (range: 0 to 48 years), the average age of men was 5.19 years (range: 0 to 43 years). Difficulty in finding a dentist ($p$=0.001), and dissatisfaction with the health system ($p$= 0.007) negatively impacted the OHRQoL.

It was especially noticeable that male participants reported being diagnosed earlier than female participants. One explanation could be that most female patients are affected by externally inconspicuous subtypes and are diagnosed later. However, an additional study would be necessary to evaluate this clinically. People with EC rate their OHRQoL worse than is usually prevalent in the normal participants.[12]

Patients with ED present with multiple restorative issues. A multidisciplinary approach involving various clinical modalities is required to render comprehensive dental care. Treatment decisions of the dental team depend on the patient's needs, wishes, and willingness to undergo minor or major treatment and on their economic possibilities.

Different prosthetic options may be indicated, and the patients need to be encouraged for appropriate treatment. Deciding factors for endos-seous implants are the patient's systemic health, adequate bone volume, and socioeconomic conditions.

Prosthetic rehabilitation may include removable or fixed prosthesis, which is a cost-effective treatment modality. Though implants are ideal for partial anodontia in adult patients, this case was treated in a conservative and minimally invasive method. Implants are not recommended at younger ages.

Orthodontic treatment in combination with prosthetic rehabilitation of the patient will support the normal function, esthetics, and psycho-social well-being. Eventually, the patient was satisfied with the prosthesis as function was re-established and esthetics were not compromised. Long-term success depends on regular recall appointments and meticulous maintenance of oral and prosthetic hygiene.[13]

Marked hypodontia demands coordinated treatment planning and appropriate timing of the delivery of care by various dental specialties. Management of hypodontia in adolescent patients permits optimum orthodontic control of the developing occlusion. Multidisciplinary referral or consultation is vital in treatment planning. Planning for space management is best carried out before initiating orthodontic treatment.

A diagnostic orthodontic setup is an essential adjunct to the treatment planning process. Tooth size measurements provide valuable data for evaluating the final tooth position and morphology. Careful consideration should be given to the timing of extraction of primary teeth and, if possible, extraction should coincide with implant insertion.[14]

Chrcanovic in a systematic review, assessed the clinical outcome and survival rate of oral implants placed in individuals with ED. A total of 90 publications were included, reporting 228 ED patients that received 1472 implants. The probability of failure was 4.5% (95%CI 3.5%-5.6%, $p<0.001$). Additional treatments performed were Le Fort I osteotomy (99 implants, 20 patients, 3.5% failed), grafting (497 implants, 77 patients, 5.2% failed), distraction osteogenesis (79 implants, 16 patients, 10.1% failed).

Mean follow-up was 42.9 ± 41.9 months (min-max, 2-240). The conclusion was dental implants placed in ED patients, either infants or adults, presented a high survival rate, in 20-year 84.6%.[15]

It is recommended while deciding the optimal individual time point of implant insertion, the status of skeletal growth, the degree of hypodontia, and extension of related psychological stress should be taken into account, in addition to the status of existing dentition and dental compliance of a pediatric patient.[16]

Furthermore, it is possible to combine orthodontic appliances and prostheses. Celli *et al.*,[17] proposed a treatment with a device, in which upper and lower (two parts) were partially removable and partially fixed. The patient, a six-year-old boy with hypohidrotic ectodermal dysplasia with severe oligodontia, was prepared to receive dental implants for definitive oral rehabilitation. The intervention began with a heat-cured acrylic resin removable appliance with an expansion screw in the maxilla and the mandible.

Afterwards, an innovative orthodontic/prosthetic modular device was made in the maxilla and in the mandible, fixed with bands on the first permanent molars. An expansion screw and a telescopic screw followed and supported the resin prosthetic teeth during the orthopedic expansion. The resin prosthetic teeth are removable from the metallic fixed structure of this appliance. The patient was followed for ten years from the beginning of treatment.[17]

No consensus exists on the ideal age for the beginning of prosthetic rehabilitation. Prosthetic rehabilitation must be done at the earliest age possible to maintain and correct the oral functions and prevent growth anomalies. It makes the subsequent treatment steps easier. Early rehabilitation and follow-up seem to be one of the keys to a successful treatment that help these children overcome their handicap and integrate them into society.[18]

Management of clinical manifestations associated with EC presents a unique challenge for


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prosthodontists and pedodontists. Treatment of young edentulous patients with a removable partial or complete denture is an acceptable, available and cost effective modality that improves function, speech, aesthetics and psychosocial condition. However, its long-term success depends on regular recall appointments and meticulous maintenance of oral and prosthetic hygiene.[6]

Although this case report has the limitation that the genetic test was not carried out, the clinical findings of mother and child confirm the phenotype of the condition. Therefore, the importance of molecular analysis to confirm the diagnosis and genetic assessment for the family cannot be denied. Even the mother had oligodontia, but with a milder impact compared to the son. Expressiveness in the mother occurs by the inactivation of one of the X chromosomes.

This case report showed the strength to enhance the work of a multidisciplinary team consisting of a pediatric dentist, an orthodontist, a geneticist and a psychologist in rare diseases such as X-linked hypohidrotic ectodermal dysplasia. The smile expression begins from a very young age and is a mechanism by which the individual expresses himself and receives gratifications.

Later concepts such as self-concept and self-esteem are consolidated according to what the subject sees or the evaluations he receives from his parents, family and colleagues. Oral health comprises a comprehensive concept where these variables must be taken into account, understanding that it is a fundamental pillar in the quality of life of children and adolescents. Individuals with dental-oral conditions have more acceptance and socialization problems than their unaffected peers, and the dentist can provide changes and improvements in self-image and self-esteem.[19]

The proposed treatment allowed the patient to smile, eat, pronounce words and socialize as he had not done previously.

## CONCLUSION.

This case report adds value to the literature, because it deals with an early-age patient with ED, offering him a minimally invasive treatment that contributes to the patient's development and can be performed by a pediatric dentist. This approach provides the patient with a better quality of life, by allowing him to express himself, smile and improve his masticatory function.