Prosthodontic Management of Children with Hypohidrotic Ectodermal Dysplasia: Case Reports and a Review of the Literature

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Abstract: Hypohidrotic Ectodermal Dysplasia (HED) is a hereditary syndrome characterized by congenital defects in ectodermally derived structures as teeth, hair, skin and nails. Dental manifestations include abnormalities of form and number (hypodontia or anodontia) with subsequent aesthetic, psychological and functional consequences.

Objectives: The purpose of this paper was to show through cases report that dental management help to improve the patient's quality of life and to optimize their social integration.

Methods: Two clinical cases about a boy and a girl, 8 and 6 years old respectively, with HED were exposed. They had a senile facial expression and presented a hypodontia with smooth dry skin, a hypotrichosis and an onychodystrophy.

Management: After clinical and radiographic examination, a prosthetic rehabilitation was planned. Instructions on denture wear, hygiene and maintenance were given.

Conclusion: In children with HED, an early oral management is essential because it gives a psychological boost and enhances conditions for normal growth and orofacial development. A close medical and psychological follow-up is needed.

Keywords: Child, dental prosthesis, disease management, ectodermal dysplasia, hypohidrotic, hypodontia.

1. INTRODUCTION

The Hypohidrotic Ectodermal Dysplasia (HED) constitutes one of the multiple types of the Ectodermal Dysplasia group (ED), defined as genetic disorders in structure or function of ectoderm-derived tissues including hair, skin, sweat glands, teeth and nails [1, 2].

Literature has reported more than 200 distinct varieties of Ectodermal Dysplasias (ED) with a multiple combinations of abnormalities [3]. Friere-Maia listed these types in two main groups, Hidrotic and Hypohidrotic, where teeth and hair are similarly affected but the hereditary pattern and the manifestations in sweat glands and nails are different [4].

Several classifications based either on clinical findings or molecular genetics were developed, in order to integrate the most recent clinical and molecular information available [10]. However, an International Conference held in 2008 on ED, proposed a new classification supporting the precise diagnosis of patients with an ED, helping clinicians with treatment planning and pushing forward the frontiers of research [5].

Among all the varieties of ED, the HED is the most frequent and the most common form. Its diagnosis is fundamentally based on the physical examination which revealed a classic triad: partial or complete absence of teeth (hypodontia or anodontia), sparse hair (hypotrichosis) and reduced sweat secretion (hypohidrosis). Nail dysplasia (onychodysplasia), frontal bossing and periorbital pigmentation are often associated with the triad. The patient may also produce a saddle nose, prominent lips and a pointy chin, exhibiting, therefore, a distinctive senile facies [6-9].

Major consequences have been noted in patients who suffer from HED, like functional, aesthetical and psychological impairment that alters the patient's quality of life and compromises his social integration [10].

The HED is relatively rare with an incidence of about 1 case per 100,000 newborns, although all racial groups can be affected all over the world by such a syndrome; it is mostly encountered in males and it is usually inherited in an X-linked recessive pattern (95%). The literature also reported autosomal dominant and autosomal recessive patterns, which explains that girls can also be affected as it will be shown in our second clinical case [11-15].

This paper describes the symptoms of two cases in children with HED and discusses the dental management through a recent literature review.

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2. PATIENTS AND METHODS

2.1. First Case

2.1.1. Presentation

An 8-year-old young boy, was referred to Monastir University Dental Clinic, Department of Paediatric Dentistry and Oral Prevention, accompanied by his parents for a prosthodontic treatment. The chief complaint was an aesthetical, psychological and functional impairment since the patient presented missing teeth and a delayed eruption.

The medical questionnaire revealed neither parental consanguinity nor family history of any orofacial anomalies. The patient's mother had a normal pregnancy, a natural childbirth at the time and controlled vaccinations; however she noticed a delayed eruption of her child's primary teeth as well as recurrent episodes of hyperthermia since infancy. She also reported that her child did not sweat and was unable to tolerate a hot environment. He went to school and no mental retardation was noted.

The physical examination showed a smooth dry skin, a hypotrichosis with a thin, silky and sparse hair, eyebrows and eyelashes, associated with a periorbital hyper-pigmentation and easily breakable nails (onycho-dystrophy). The patient developed a senileappearing facies (Figure **1**) and was suffering from nearsighted-dness (myopia). He also showed a depressed nasal bridge, a decreased lower facial height and prominent lips.

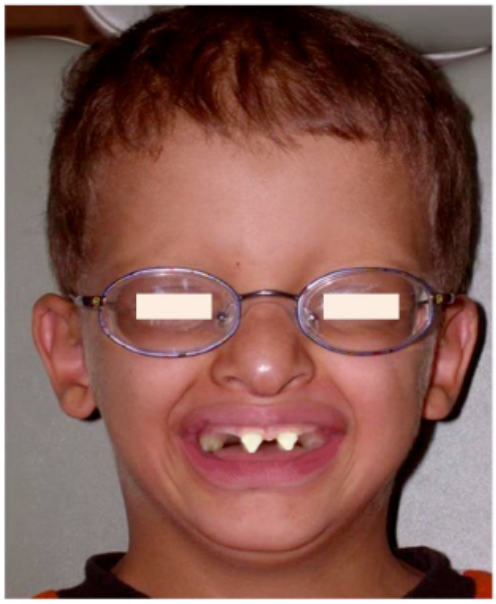

Figure 1: Facial features of 8-year-old boy affected by HED presenting the classic triad of hypotrichosis, hypohidrosis and hypodontia.

The intra-oral examination revealed a partial anodontia with only six primary teeth, lower canines, upper second molars and two upper cone-shaped teeth separated by a large diastema that occupies the sites of central incisors (Figure **2**), were present in the dental arches. The partial anodontia was associated with an atrophy of both maxillary and mandibular alveolar bone and thin residual alveolar ridges. A relative macroglossia and a dry mucosa were also noted. The orthopantogram (OPG) investigation confirmed the partial anodontia with absence of other primary and permanent tooth buds (Figure **3**).

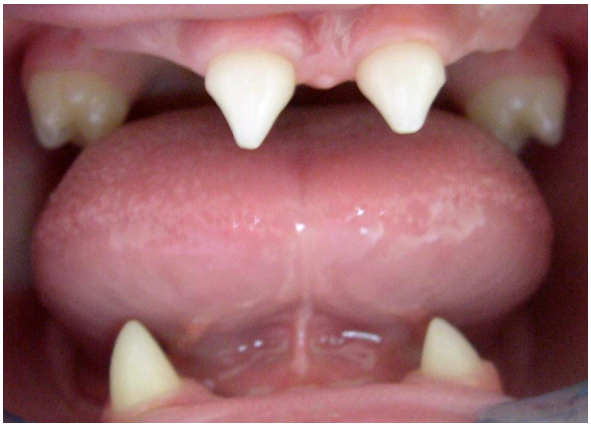

Figure 2: Intraoral view showing a partial anodontia with two upper cone-shaped teeth separated by a large diastema.

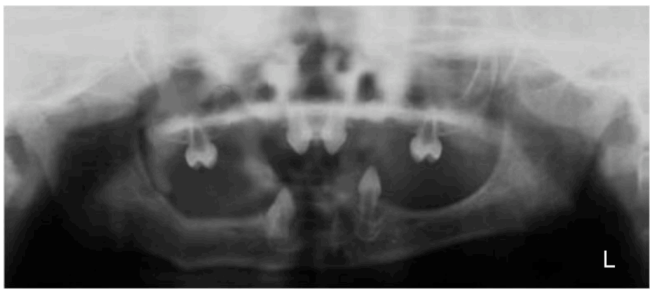

Figure 3: The orthopantogram confirmed the agenesis of many primary and permanent teeth.

According to the ectodermal dysplasia's new classification which integrates the multiple classification systems and the most recent clinical and molecular information, the patient was diagnosed with Hypohydrotic Ectodermal Dysplasia (HED) associated with a partial anodontia [5, 16, 17].

2.1.2. Management

In the present case of partial anodontia, as suggested by Johnson *et al.* [18], maxillary and mandibular complete removable dentures can be planned as well as removable partial dentures associated with a transformation of the conical upper

anterior teeth into incisors using composite resin. The first solution was privileged.

After taking preliminary impressions using alginate, personalized trays were realised with autopolymerising acrylic resin (BMS® 017 for trays self-curing). These special trays were used to perform secondary impressions with a heavy silicone (Optosil® and Xantopren® Comfort system of Heraeus Kulzer). Wax record bases of jaw relations were used for mounting casts on the articulator. Appropriate moulds (3M™ ESPE™ Polycarbonate Crowns) were chosen to cover natural teeth in the upper anterior region and a toothcoloured acrylic resin (Duracryl®, self curing crown resin) in the lower anterior region. Trial dentures had been checked for retention, occlusion, phonetics and aesthetics, before Heat-Cured Acrylic Resin dentures were processed (Figures **4**, **5**).

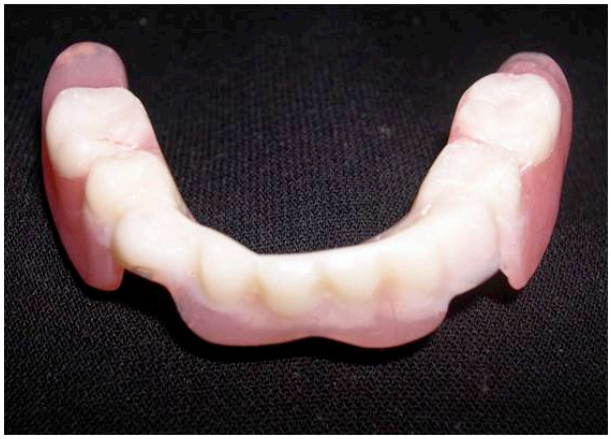

Figure 4: Mandibular prosthesis, the primary canines were covered by aesthetic acrylic resin.

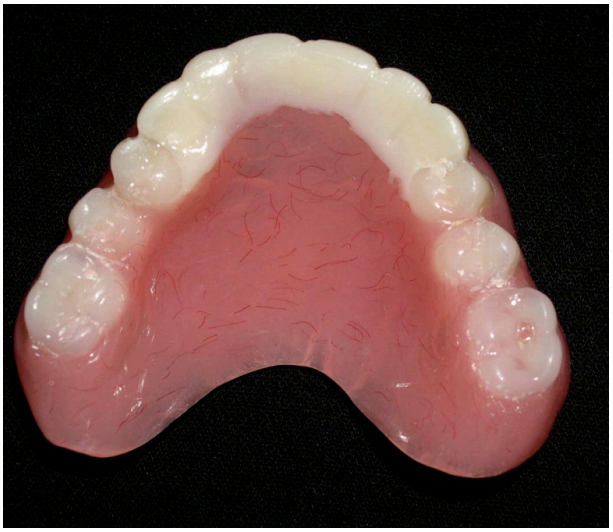

Figure 5: Maxillary prosthesis processed using Heat-Cured Acrylic Resin. Considering the extra space, the first primary molars were replaced each by two premolars.

The patient and his parents were trained how to place and remove the dentures and were given instructions for feeding, speech, hygiene and maintenance (Figure **6**). The child was called back for follow-up visits.

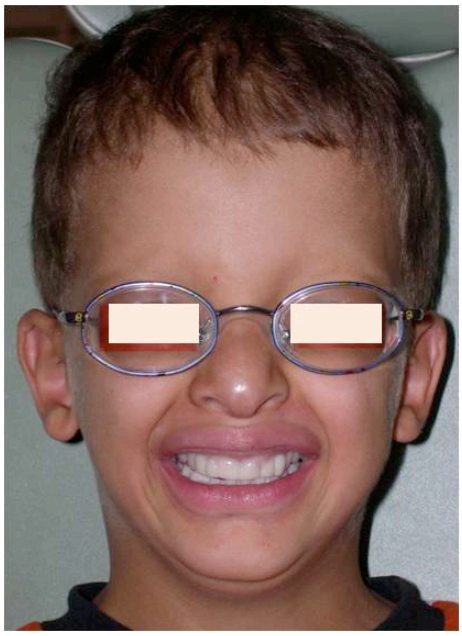

Figure 6: Final prosthesis was checked for retention, occlusion, phonetics and aesthetics.

The parents were aware that they have to stay in contact with paediatrician to prevent and treat all episodes of hyperthermia along with other subsequent complications. They were also put on their guard to check up the dentures at least every six months for adaptation, functional control and any other relevant or notified issues (pain, fracture of dentures, functional limitation or psychological rejection...).

2.2. Second Case

2.2.1. Presentation

A 6-year-old girl, showed up with her father, was examined in the same Department. She suffered from functional, aesthetical and psychological impairment because of her missing and malformed teeth. The patient lived in the South of Tunisia and revealed a relatively low socioeconomic status.

A parental consanguinity was stated but no orofacial impairment was reported in the medical family history. The patient suffered from fever each time she was exposed to sun or heat.

The physical examination revealed light hypotrichosis affecting the patient's hair, eyelashes and eyebrows. A dry skin, a broader nose and prominent

lips were also noted with breakable nails. The lower facial height was preserved in spite of the missing teeth (Figure **7**).

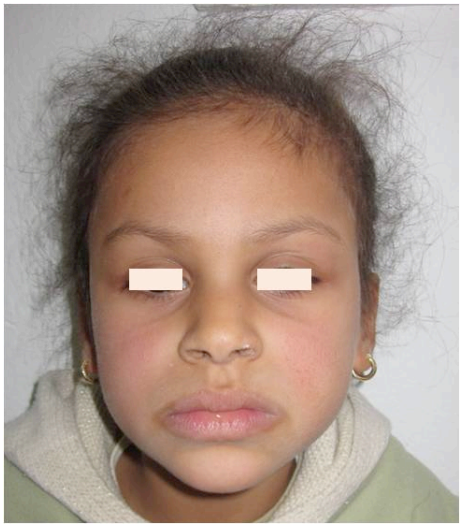

Figure 7: Facial characteristics of a 6-year-old girl presenting HED with sparse hair and prominent lips.

The intraoral examination showed a large diastema between two upper abnormally shaped teeth located at the central incisors positions. We also found that seven primary teeth were missing (the lateral upper incisors, the four lower incisors and the first right upper molar). Alveolar bone atrophy was detected in the mandible along with a relative macroglossia (Figures **8**, **9**).

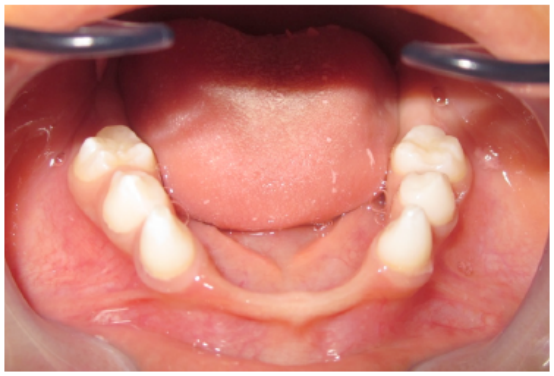

Figure 8: Overall View of the mandible with four incisors missing and thin alveolar ridge.

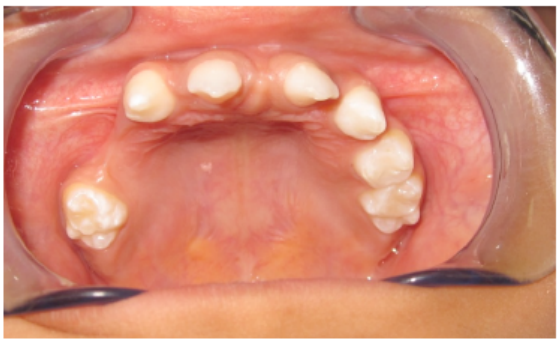

Figure 9: Intraoral view Maxilla with first right primary molar and incisors missing.

The orthopantogram examination (OPG) confirmed the agenesis of almost all permanent tooth buds except for those of the first permanent molars, the central upper incisors and two abnormally shaped buds in the areas of lower lateral incisors (Figure **10**).

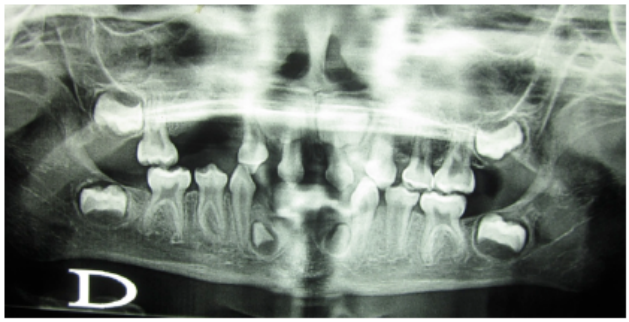

Figure 10: The orthopantogram examination shows a hypodontia with agenesis of seven primary teeth and almost all permanent teeth except for the first permanent molars, central upper incisors and two lower anterior elements.

In the light of these clinical and radiological findings, the diagnosis of Hypohydrotic Ectodermal Dysplasia (HED) associated to a hypodontia was evoked.

2.2.2. Management

The father told us that he cannot return for a second appointment. After a briefing we decide to realize a temporary prosthesis on the same day aiming to enhance the rehabilitation process. Our objective was especially functional so orthodontic resin dentures (BMS® 016 For orthodontia self curing) were processed after taking the alginate impressions and making plaster casts (Figures **11**, **12**).

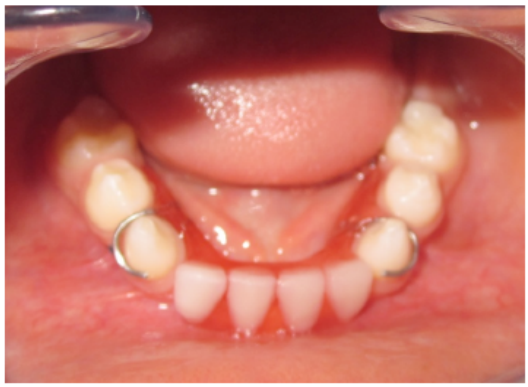

Figure 11: Mandibular prosthesis processed in pink orthodontic resin.

Instructions about oral hygiene and training for denture insertion were given to the parent with a

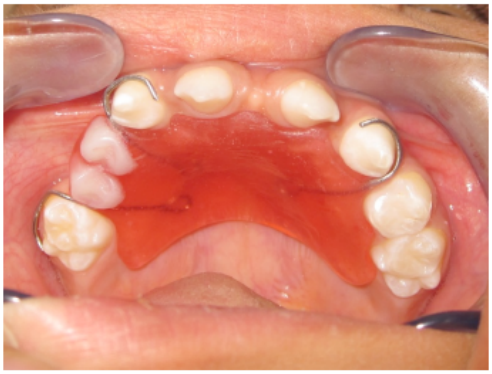

Figure 12: Maxillary prosthesis.

referral letter to the nearest dentist in their hometown who must try to improve aesthetics in anterior upper region. However, we lost sight of the patient as expected.

3. DISCUSSION

The ectodermal dysplasias, first described in 1848 by Thurnam, constitutes a wide, heterogeneous group of congenital anomalies which are characterized by developmental defects of some or all of the ectoderm derived tissues [19, 20].

Although the pathogenesis of HED is still not totally clarified, many authors think that a genetic disorder of the EctoDysplasin-A protein (EDA A); which takes part in the signalling interaction between the ectoderm and the mesoderm, alters the normal development of the ectoderm derivatives [21, 22].

The diagnosis of HED is generally established at a young age by the family doctor or pediatrician on occasions of repeated and unexplained fever and must be confirmed by the histological examination which is an important diagnostic argument, revealing that sweating glands and hairy follicles are reduced [1]. Besides, Sepulveda *et al.* have suggested a possible prenatal diagnosis of HED by Three-dimensional Ultrasonography through the identification of the distinct facial features at 30 weeks gestation [23].

The differential diagnosis should be performed between HED and the other several syndromes pertaining to the numerous varieties of Ectodermal Dysplasias that result from affected ectoderm originated tissues. We may state the tooth and nail syndrome, characterized by a severe nail dystrophy, yet a normal sweating; the multiple congenital agenesis and the Ellis-Van Creveld syndrome, the diagnosis of which is based on three major symptoms: small stature, hexadactylism and ectodermal dysplasia. A hydrotic form of ectodermal dysplasia, called Clouston syndrome, was also reported in Literature [24].

Throughout the review of literature, it appears that even if the HED syndrome is relatively rare, it must be well known by health professionals and needs to be diagnosed early and treated in order to minimize the heavy consequences and their complications [13].

Keeping in mind the limitations of the case report design, we can never conclude, based upon these two observations, that our management strategy is effective for all patients with the same conditions.

An efficient management of HED needs a multidisciplinary team including the pediatrician, the pediatric dentist, the orthodontist, the dermatologist, the otorhino-laryngologist, the psychologist... [25-27].

Some supports were given by Foundations for Ectodermal Dysplasia that were set up in some developed countries, to provide reliable information for people affected by HED and connect them together through an international network.

In children, the oral treatment plan should be performed in agreement of the other medical specialists and must be well exposed to their parents before being applied for forensic reasons. This management often includes an immediate prosthodontic rehabilitation in order to restore function and, notably, a satisfactory daily diet [28-30]. According to the changes of oral structures because of continuous development and growth in young patients, periodic recalls are needed to adjust prosthodontic dentures that must be checked and changed approximately every six months [28].

Concerning our patients, the extraction of malformed teeth must be avoided due to the risk of alveolar ridge reduction and aggravation of psychological consequences. Oral hygiene measures should be carefully applied; otherwise the over-denture indicated for the first patient will encourage dental plaque accumulation, gingivitis and dental caries (Figure **6**).

In spite of the bone atrophy in the alveolar ridge associated with tooth agenesis, some authors used dental implants for supporting prostheses in children with HED; but the literature review reported growth related problems after placing implants in young patients [31, 32]. In fact, Guckes showed, in his clinical case report, that the endosseous dental implant can becomes deeply embedded in with the growth phenomenon [33].

The treatment with fixed dental prostheses or implant-supported prostheses will be indicated later in adulthood after associated examinations like conebeam computed tomography and adopting other treatment options such as bone grafting...[31, 32].

4. CONCLUSION

This paper allows drawing the main lines of treatment plan for patient with HED, sets forth the necessary recommendations and precautions to achieve a social integration and a better quality of life. An early oral management is essential in children because it gives them a psychological boost and enhances the conditions for growth and orofacial development.

The cooperation of the child and parents and the multidisciplinary collaboration guarantee the success of the treatment. Eventually, gaining the parents confidence and seeing the smile on the child's face will always be our invaluable award.

CONFLICT OF INTEREST STATEMENT

The authors declare that they have no conflict of interest.

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