

HYPOHYDROTIC ECTODERMAL DYSPLASIA: A CLINICAL CASE REPORT

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Abstract

In times where rare diseases, mostly of genetic offspring, lead research to carry out genetic counselling to better understand the pathogenetic role of the diseases, with chances to develop alternative therapy respect than the multispecialistic and symptomatic approach, we report on a case of X linked Hypohidrotic Ectodermal Dysplasia, knowing the importance of research, basic to gene therapy, aware of the recent novelties which might drive to an improvement of the symptoms in an alternative way; still as Paediatric Dentists, awaiting for these magic result we carry out an oral rehabilitation reporting step by step the treatment achieving a very good compliance in term of smile, occlusion and aesthetics concerning the patient.

Case report (J Int Dent Med Res 2014; 7: (2), pp. 37-41)

Keywords: Ectodermal dysplasia, oral rehabilitation.

Received date: 21 December 2013

Accept date: 18 January 2014

Introduction

Ectodermal dysplasias (EDs) represent a large group of inherited disorders which affects at least two ectodermal-derived structures such as hair, nails, skin, sweat glands and teeth with or without the involvement of the other organ¹⁻³. Approximately 200 different conditions have been classified as an ectodermal dysplasia of which the hypohidrotic/anhidrotic ectodermal dysplasia (HED/EDA) represents the most common form and is characterized by the clinical triade of hypotrichosis (sparse hair), abnormal or missing teeth (anodontia or hypodontia) and defect sweating (hypohidrosis and anhidrosis). Different modes of inheritance have been described for HED. Mutations in ectodysplasin A1 (EDA-A1; MIM#300451)⁴ result in X-linked hypohidrotic

ectodermal dysplasia (XLHED; MIM#305100); while mutations in the ectodysplasin A receptor gene (EDAR; MIM #604095)^{5,6} and mutations in EDAR-associated Death Domain gene (EDARADD; MIM#606603) are implicated in both autosomal recessive (MIM#305100), and dominant (MIM#129490) forms of HED^{7,8}. Recently mutations in WNT10A (MIM#606268) and TRAF6 (MIM#602355) have been reported to cause HED-like phenotypes^{9,10}. An X-linked ectodermal dysplasia syndrome with immunodeficiency has been reported in patients with hypomorphic mutations in IKBKG (inhibitory B kinase gene)^{11,12}. People affected by the most common form of Ectodermal Dysplasia, called Christ-Siemens-Touraine Syndrome have a characteristic facies: frontal bossing prominent forehead, loss of the vertical height of the face, maxillary hypoplasia, flattened nasal bridge with thickened lips, prominent chin, periorbital wrinkled and hyperpigmented skin, dystrophic nails, or hypertrophic with abnormal keratinization, fragility of skin and sweat gland abnormalities associated with a defective thermoregulation¹³. Of dental interest are the changes in the oral cavity. In these patients,

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there is almost always a decrease in the number of salivary glands and sometimes hypoplasia of major salivary glands, resulting in dry mouth, difficulty in swallowing and sometimes easier possibility to develop infections of the oral cavity.

In addition, an analysis of saliva reveals a reduction in the buffering capacity of stimulated saliva and an increase in the bacterial flora with a predominance of beta-hemolytic streptococci. In the 80% of cases dental anomalies of both the deciduous dentition and the permanent one are found, from hypodontia to complete anodontia: the upper central incisors are present in 42% of cases, the maxillary first molars in 41% and 39% in the first mandibular molars, maxillary canines in 22%, while the group of mandibular anterior teeth is frequently absent¹⁴. Abnormalities are often present in the shape of teeth: dental crowns may appear smaller and the upper incisors and canines are always conically shaped, taurodontic, supernumerary teeth might be present, retention of deciduous teeth is observed; absence of teeth results in hypoplasia of the jaw associated with an atrophy of the alveolar ridges which leads to a reduction of the vertical dimension especially at the level of the lower third of the face^{15,16}.

Often the delayed eruption of teeth in the young child allow the diagnosis; the absence of evidence to deciduous at the age of 20-24 months should prompt the clinical suspicion of multiple agenesis, one of 3 cardinal sign of hypohidrotic ectodermal dysplasia. Numerous dental anomalies and jaw bone problems may bother little patients in functional, aesthetic and not least psychological aspect: the task of pediatric dentists is to try to give these young patients a smile and a facial aesthetics as harmonious as possible¹⁷.

Case Report

A 6 years old young male came to our attention already with clinical diagnosis of XLHED at the Department of Pediatrics Dentistry of ISI, together with parents and along with the request of an esthetic and functional oral rehabilitation. After an accurate inspection he revealed severe hypodontia in mixed dentition; an Orthopantomography has been carried out showing the following dental formula: 16, 55, 13, 51, 23, 65, 26, 47 (Fig 1 a,b,c). His general conditions were good along with his

collaboration. He presented the characteristic facies of hypohidrotic ectodermal dysplasia with frontal bossing, maxillary hypoplasia, protruding lips, sparse eyebrows, sparse hair, dry skin especially in the hands and feet, sweating heavily reduced.

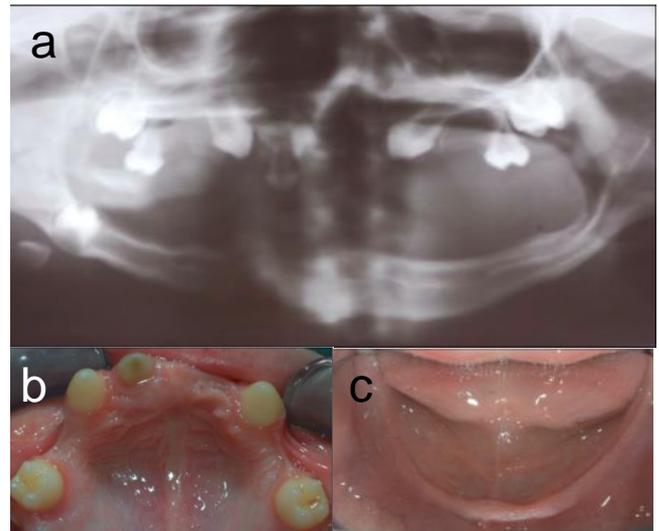


Figure 1 (a). Orthopantomography showing severe hypodontia; **(b).** Occlusal mandibular view showing anodontia in deciduous dentition, with atrophy of the alveolar ridge; **(c).** Occlusal mandibular view showing anodontia in deciduous dentition, with atrophy of the alveolar ridge.

The mother had milder signs of hypohidrotic ectodermal dysplasia, with hypohidrosis and agenesis of lateral mandibular permanent incisor. During the collection of medical records parents reported a recent episode of epistaxis.

Furthermore, the patient previously had already underwent to a series of investigations which are below reported in order to show how useful can be a formulation of a correct treatment plan. Also during this first visit impressions were taken to carry out the study models, photos were made intra- and extra-oral.

To better analyse the bone and choose the best treatment a CT DICOM 3D has been carried out; possible solution of implants placement in the intercanine region has been left as a last resort, but was anyway later abandoned because we did not have the authorization from the Ethics Committee of Referral Center. At this point, given the particularities of the disease, it was decided to establish a team comprising a Prosthodontist, an Orthodontist, a Maxillofacial

Surgeon and a Pediatric Oral Surgeon in order to establish a treatment plan immediately and the timing of successful treatment.

An orthodontic treatment device with ELN (envelope lingual nocturne) to harmonize the parameters through a cross-lingual re-education has been carried out (Fig. 2-3).

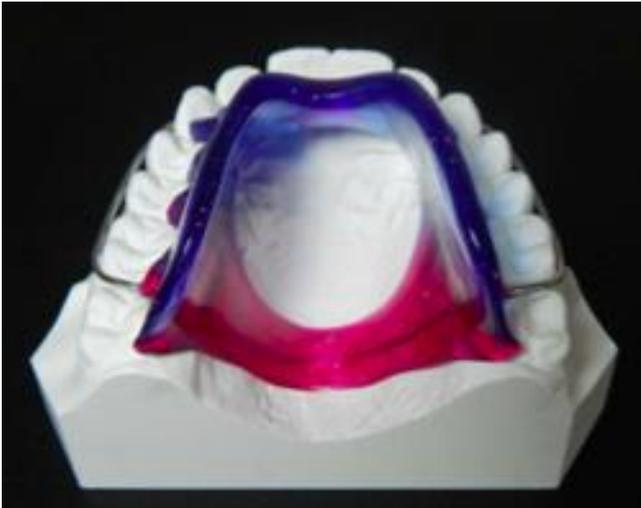


Figure 2. Appliance of ELN (envelope lingual nocturne) to harmonize transversal parameters through a lingual rehabilitation.



Figure 3. Facial arch realized in laboratory to reproduce a correct vertical height of the lower 1/3 of the face.

In the lower jaw as at the beginning an overdenture implant supported could be the ideal goal, considering the young age and after an accurate and fruitful decision with the patient and his relatives. Hence a removable total prosthesis has been carried out, tried in and after any occlusal parameters records fit in the mouth reaching a very good level of compliance, awaiting for the end of the growth and a fix solution (Fig. 4-6).



Figure 4. Intraoral fit of the superior and inferior to check the vertical height



Figure 5. (a). Occlusal view of the lower prosthesis; (b). occlusal view of the upper prosthesis; (c). lateral view of the upper and lower prosthesis; (d). lateral view of the upper and lower prosthesis.



Figure 6. Frontal view of the prosthesis.

Discussion and Conclusion

The prosthetic solution adopted although not ultimately has been chosen because it has

allowed us to optimize functionality and aesthetics aspects which were the initial requirements of the patient and his parents. In particular, comparing the pictures of the profile of the face is easily notable the return of harmony resulting from the re-establishment of an adequate vertical dimension after the full mouth oral rehabilitation (Fig.7c,d).

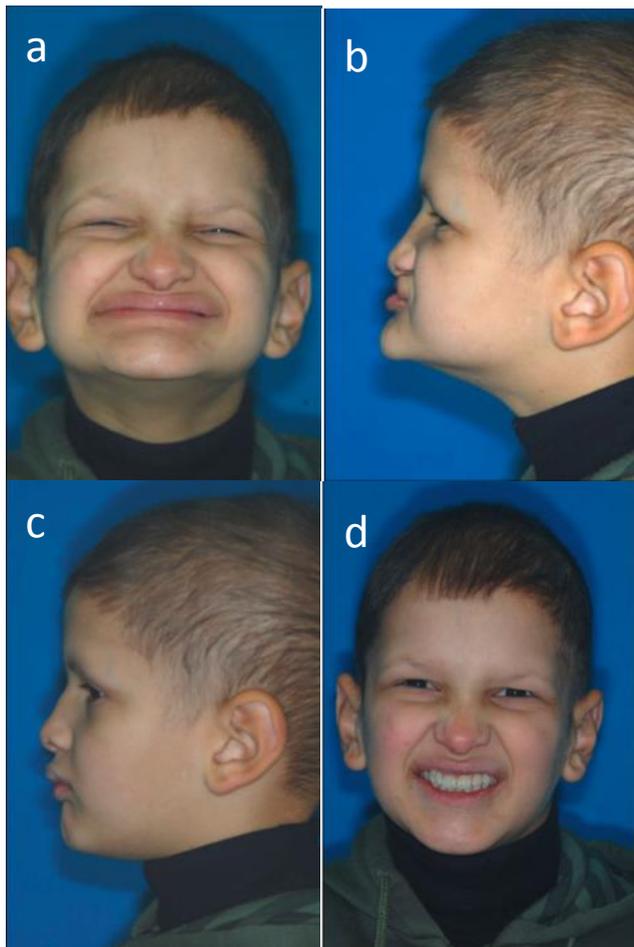


Figure 7. (a). Left profile showing reduce vertical height; (b). frontal view of the patient affected by XLHED; (c). lateral view showing harmonious vertical height, recuperated after prosthesis rehabilitation; (d). frontal view showing a good occlusion and the consequent smile enhancing the level of compliance)

This prosthetic solution cannot be considered final and it has been preferred to leave the residual growth of the jaw occurring in a harmonious way. Although we did not report in this study the patient's genotype, we report a case clinically solved, to be genotyped in the next future. In times where rare diseases, mostly of genetic offspring, lead research to carry out

genetic counselling to better understand the pathogenetic role of the diseases, with chances to develop alternative therapy respect than the multispecialistic and symptomatic approach, knowing the importance of research, basic to gene therapy, aware of the recent novelties which might drive to an improvement of the symptoms in an alternative way; still as Paediatric Dentists, awaiting for these magic result we carried out an oral rehabilitation reporting step by step the treatment achieving a very good compliance in term of smile, occlusion and aesthetics concerning the patient.

Final-note

The treatment presented has been made possible through a grant from the ISI Foundation.

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