X-linked Hypohidrotic Ectodermal Dysplasia: a ten-year case report and clinical considerations

S. DALL’OCA*, E. CEPPII**, G. POMPA***, A. POLIMENI*

ABSTRACT. Background Ectodermal Dysplasias (EDs) are a large group of syndromes that are heterogeneous under clinical and genetic aspects, and are characterised by anomalies in the structures of ectodermal origin. In EDs dental anomalies in shape and number (oligo-hypodontia) occur frequently and severely and can affect both the primary and permanent dentition. Case Report The oral habilitation of a child affected by X-linked Hypohidrotic-Ectodermal Dysplasia with oligodontia over a period of ten years is described. This report includes discussion of the aetiology of EDs as well as discussion of the long-term prognosis for the patient. Conclusion The success of treatment in the case reported is based on an early diagnosis and a correct timing of interventions addressing the preservation of the existing dental elements, early functional rehabilitation and aesthetic correction.

KEYWORDS: Ectodermal dysplasias, Classification, Hypodontia, Skeletal growth, Salivary glands.

Introduction

Ectodermal Dysplasias (EDs) are a large group of syndromes that are heterogeneous under clinical and genetic aspects, and are characterised by anomalies in the structures of ectodermal origin. They can be manifested in problems relating to hair, nails, teeth, sweat and sebaceous glands, epithelium, conjunctiva, nervous system etc. The EDs are not generally pure "one-layer diseases", mesodermal and, rarely, endodermal dysplasias coexist [Pinheiro et al., 1994; Itin et Fistarol, 2004].

The incidence of EDs is estimated as being 7 in 10,000 live births. They follow all possible Mendelian modes of inheritance (autosomal, dominant or recessive, x-linked dominant or recessive) but also sporadic cases are reported [Lamartine, 2003].

Freire-Maia and Pinheiro established a classification for EDs based on clinical criteria which distinguished these pathologies into two large groups, A and B.

Group A was assigned to all the conditions that present anomalies of at least two of the four structures most frequently mentioned in literature relating to ED: hair, nails, sweat glands, with or without malformations or other defects. Group B, on the other hand, is related to those conditions which, along with alterations of only one of the aforementioned structures, show the presence of another anomaly relating to ectodermic structure tissue [Pinheiro et al., 1994].

Group A is comprised of 192 syndromes: 41 of autosomal dominant transmission, 52 of autosomal recessive transmission, 8 are x-linked, and the aetiopathogenesis of the others is unknown and probably a result of casual genetic defects. A good 120 of the 192 EDs assigned to Group A present anomalies of dental tissue [Pinheiro et al., 1994].

Priolo et al. [2000] proposed a classification of EDs integrating both molecular-genetic data and corresponding clinical findings or related diseases. About 30 EDs have been explained at a molecular level by identification of causative genes, and according to Lamartine these genes can be categorized in four major functional groups: cell-cell communication and signaling; cell adhesion; transcription regulation, and development [Lamartine, 2003; Itin et Fistarol, 2004].
The most frequent phenotypical alteration of ED comprises reduction or absence of body hair and alterations of the scalp, hair appears sparse, lightly pigmented with a wooly appearance. In the hypohydrated form there is a deficit of sweat glands which gives a marked intolerance to heat and dry skin, localised increase of pigmentation on the hands and feet in conjunction with a flattening of the epidermal ridges. The absence or reduction of the mucous glands of the respiratory or gastroenteric apparatus leads to major susceptibility to infections and immune system disorders, which predisposes to allergic pathologies, asthma and eczema. Relapsing conjunctivitis with photophobia because of a reduction of lachrymation must also be mentioned. Nails can be dystrophic, hypertrophic, abnormally keratinised, thick, discoloured, striated, split or fragmented [Priolo et al., 2000; Nordgarden et al., 2003; Itin and Fistarol, 2004; Singh and Warnakulasuriya, 2004;].

The oral examination often shows hypo-oligodontia, anodontia is rare. The teeth are usually malformed and small with cone or peg-shaped aspect, prone to caries because of enamel defect or salivary gland malfunction with xerostomia, and taurodontism is frequently seen. The distribution of absent teeth deviates remarkably from the general population. The alveolar process fails to develop at edentulous sites [Rashedi, 2003; Suri et al., 2004; Penarrocha-Diago et al., 2004; Prager et al., 2006; Yavuz et al., 2006; Barbería et al., 2006; Bergendal et al., 2006].

The oligo-hypodontia causes a reduction of the vertical dimension of the lower third of the face, a missing physiological closure of the gonial angle, a primary rotation of the mandible with resulting stabilisation in III Class skeleton [Suri et al., 2004].

The aim of this study is to report the case of oral habilitation of a child affected by X-linked Hypohidrotic-Ectodermal Dysplasia with oligodontia over a period of ten years.

**Case report**

The patient was referred to the Department of Paediatric Dentistry in 1997, when he was 18 months old, for advice as only two teeth had erupted. His parents were non-consanguineous and his mother, who did not take potentially teratogenic drugs during pregnancy, reported to have many missing teeth, partially due to extractions, partially due to congenital agenesis.

His stature, growth and neurological development were normal. The skin was dry, delicate and prone to rashes, his hair was thin, light blond and wooly, eyelashes and eyebrows were poor (Fig. 1). The parents reported that their son suffered from heat intolerance and that his sweat production was poor, as confirmed by the test of perspiration with pilocarpine. The nails were normal. Only two malformed upper molars were present, they were reduced in size, with three conoid cusps (Fig. 2). The alveolar bone was small, the oral mucosa was normal and the salivation was physiological.

The diagnosis of X-linked Hypohidrotic-Ectodermal Dysplasia was confirmed by the geneticist. X-linked Hypohidrotic-Ectodermal Dysplasia-(OMIM #305100) is caused by mutation in the gene (map locus Xq12-q13.1) encoding ectodysplasin-A that is involved in cell-cell communication and signaling [Lamartine, 2003].

The parents were informed about the pathology and its modes of inheritance, about the importance of dietary control, fluoride supplementation and special consideration in oral hygiene. The geneticist, the paediatrician, the dermatologist and the
ophthalmologist supported us in medical counseling. The execution of a panoramic radiograph was not possible because of the inadequate patient’s cooperation, so it was postponed. As no caries were detected and the diet of the patient included only soft food, but he was well-nourished and well developed according to his age, any dental treatment was deferred.

At the age of three years the panoramic and cephalometric radiographs (Fig. 3, 4) were taken and they revealed the severe oligodontia and presence of 55 and 65 with tendency to taurodontism. The patient underwent removable prosthetic treatment (Fig. 5, 6). The maxillary denture was delivered and after two months the fabrication of mandibular prosthesis began, according to conventional prosthetic techniques. The teeth were reproduced from a cast of a child with complete deciduous dentition. For the first year the patient was not able to eat wearing his dentures, but he wanted to wear them for aesthetic reasons particularly when he was attending the kindergarten. After about one year he was able to eat with his removable prostheses. Recall appointments were scheduled every three months for the hygienisation of the prostheses, to check if revision of the prostheses was necessary due to maxillary and mandibular bone growth, to check plaque control and the conditions of the oral mucosa.

Major modifications to the denture bases, and occlusal adjustment, had been required 12 months after their fabrication. The fabrication of new dentures, which allowed the teeth to erupt, was necessary when the patient was five, eight and ten years old. Since the age of eight the prostheses were set to a permanent dentition.

The patient will be evaluated for the placement of endosseous implants in the lower jaw even though he has completely adapted to removable prostheses.

**Fig. 3 -** The orthopantomogram at three years of age shows severe oligodontia, scarce bone height of the upper alveolar process, the extensive sinuses contained within thin walls.

**Fig. 4 -** Cephalometric radiographs at three years of age.

**Fig. 5 -** Upper and lower prostheses with age-appropriate teeth.

**Fig. 6 -** Frontal view of the patient after treatment at the age of three. A slight hyper-pigmentation is evident in the periocular area and around the mouth.
There is no potential implant recipient site in the maxilla because of inadequate bone height and the extent of the nasal cavities and maxillary sinuses.

At the age of eleven years, when the patient wears his prostheses, an extraoral examination highlights a harmonious grow of the face, only a slight depression of the bridge of the nose is evident (Fig. 7, 8). The skin is dry and hyper-pigmented in the periocular area and around the mouth, his hair is thin, light blond and wooly, eyelashes and eyebrows are poor.

**Discussion**

In the case reported in this study, the early rehabilitation of the masticatory function started when the patient was three years old. At this age the patient cooperated and we could take the necessary impressions for the manufacture of prostheses which were functional and aesthetic. The re-establishment of a correct vertical height of the lower third of the face facilitated a harmonious growth both of the skeletal components and muscular tone. In addition, we agree with other authors that in serious cases of oligodontia, the earlier the application of a prosthesis, the better it is accepted by the patient [Rashedi, 2003].

We also wish to underline the importance of frequent recall appointments and the particular attention that we paid regarding inflammation due to plaque deposits on the prostheses and to intolerance toward resin. In fact patients affected by X-linked Hypohidrotic-Ectodermal Dysplasia are prone to suffer from allergies, in addition to reduced salivary clearance and delicate mucosa [Nordgarden et al., 2003; Singh and Warnakulasuriya, 2004].

Unfortunately for our patient the possibility of habilitation using prosthetic implants are few considering that the alveolar bone is severely reduced in the areas where agenesis occurred. Some authors report that endosseous implants can be successfully placed even in very young children affected by ED. Early implants are destined to become submerged because of adjacent alveolar development and prosthetic revision and placement of longer abutments are necessary over the years [Ruhin et al., 2001; Alcan et al., 2006]. As our patient has completely adapted to removable prostheses, we decided to re-evaluate him for implants after adolescence.

**Conclusion**

The case reported highlights that the paediatric dentist has an important role in the diagnosis and therapy of EDs. He can be called by other specialists to highlight signs of these syndromes in the oral-maxillary-facial region, and sometimes, he himself can be the first to recognize certain signs which could induce further investigations. The success of treatment in the case reported is based on an early diagnosis and a correct timing of interventions addressing the preservation of the existing dental elements, early functional rehabilitation and aesthetical correction.

**References**

