

Rare disease

Oral rehabilitation of children with ectodermal dysplasia

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The aim of this study was to describe the clinical treatment of young patients, affected by ectodermal dysplasia (ED), and to possibly establish clinical guidelines. The study design was case series. ED syndromes (EDs) are a heterogeneous group of inherited diseases characterised by abnormal development of tissues of ectodermal origin. The most common form of EDs is X linked hypohidrotic ED (HED). Characteristic triad of HED is oligo-anodontia, hypotrichosis, hypo-anhidrosis. Oligo-anodontia is one of the most severe impairment, since it affects chewing, swallowing, speech, esthetics and social relation. Early prosthetic rehabilitation (at 2–3 years of age), with partial or complete dentures, is essential to improve oral function and reduce the social impairment.

BACKGROUND

The aim of this study was to describe the clinical treatment of young patients, affected by ectodermal dysplasia (ED), and to possibly establish clinical guidelines.

Early prosthetic rehabilitation (at 2–3 years of age), with partial or complete dentures, is essential to improve oral function and reduce the social impairment.

CASE PRESENTATION**Introduction**

ED syndromes (EDs) represent a rare group of inherited disorders characterised by dysplasia of at least two tissues of ectodermal origin (hair, nails, teeth, skins and glands).^{1 2} Clinically, it may be divided into two broad categories: the X linked hypohidrotic form and the hidrotic form. Hypohidrotic ED (HED) is characterised by the triad oligo-anodontia, hypotrichosis, hypo-anhidrosis (Christ–Siemens–Tourane syndrome)^{3 4} and its incidence is about 1/100000 live borns.¹ Mutations in the ectodysplasin-A (EDA) and EDA receptor genes are responsible for the X linked and autosomal dominant and recessive forms of HED.^{2 4 5} The clinical features include sparse and fine hair, congenital missing of teeth and anomaly of shape as conical-shaped teeth, sweat and eccrine glands decreased in number and function and hypoplastic skin.

Heat intolerance at increased ambient temperature or during exercise might rise high fever, which is one of the most important and dangerous aspects of patients affected by HED.^{3 6} Dental abnormalities are the most common patients complaint; complete or partial anodontia and teeth malformations are the most frequent dental findings. Moreover, incisors and canines are often conical-shaped while primarily second molars, if present, are mostly affected by taurodontism.⁷ The diagnosis of HED in the neonatal and early infancy periods may be difficult since sparse hair and absent teeth are normal findings at this age. In childhood, the diagnosis is mostly made on the basis of history and clinical examination.^{6 8}

Treatment is symptomatic and includes protection from heat exposure, skin, hair, ear, nose and nail care. Genetic counselling is available for the patients and their families and often reveals the gene involved.⁸ In addition, early prosthetic rehabilitation is recommended as essential in the management of HED because it is important from the functional, esthetic and psychological point of view.^{8 9} However, a successful outcome is only guaranteed when a team approach is provided by a paediatric dentist, an orthodontist, a prosthodontist and an oral-maxillofacial surgeon.^{9 10}

Conventional prosthodontic rehabilitation in young patients is often difficult because of the anatomical abnormalities of the existing teeth and alveolar ridges, since the conical shaped teeth and ‘knife-edge’ alveolar ridges result in poor retention and stability of dentures. Moreover, denture should allow jaws expansion and a correct growth-pattern.^{7 11 12}

The aim of this article is to present the early prosthetic rehabilitation of 20 HED patients treated at the Department of Oral Science (Service of Dentistry for Special Care) of the University of Bologna (Italy).

Case reports

Patients with ED presented chewing difficulties and esthetic problems. Clinical evaluation revealed children with soft, thin and dry skin. Most of them could not tolerate excessive heat. Frontal bossing, depressed midface, prominent lips and depressed nasal bridge were typical features (figure 1A).^{6 10} Almost all children showed thin, sparse and blond hair, with sparse eyelashes and eyebrows. In several cases the mother was a carrier and showed some of the clinical features (hypotrichosis, hypodontia and mild hypohidrosis).^{5 6 10} One patient showed hypodontia (6+missing teeth), 18 (89.4%) oligodontia (five or less missing teeth) and one anodontia (missing of all teeth). Table 1 shows the general characteristics of each of the 20 cases we report here.



Figure 1 (A) Typical facies of a patient with ectodermal dysplasia. (B) Dental status before treatment. (C, D) Prosthesis with expanding screws. (E) Final prosthesis in situ: a good esthetic and function result has been obtained. (F) Prosthesis was rebased with soft siliconic resin.

After the medical history recording, a complete examination was performed, including complete-mouth radiographs (orthopantomogram and lateral cephalometric films), diagnostic casts and clinical examinations. Intraoral examination showed dry and sticky oral mucosa, oligodontia both in the mandible and maxilla, reduced alveolar bone volume associated with the lack of development of the alveolar ridge.^{13 14} The teeth were often conical in appearance.⁶ Moreover, all patients showed normal cephalometric measurements.

TREATMENT

Due to the young age of the patients and the poor amount of alveolar bone, a prosthetic approach was chosen and the prosthetic rehabilitation (removable dentures) was carried out as soon as possible after diagnosis, to allow normal a physiological development, a correct masticatory function^{9 15} and to improve esthetics, preventing mandibular prognathism due to hypo/anodontia,^{8 9 16} in order to reduce social impairment.

On the first appointment, initial maxillary and mandibular impressions were made with the smallest stock trays and irreversible hydrocolloid (Zhermack, Rovigo, Italy).

Casts were built with type II dental stone for custom tray fabrication. Custom trays were manufactured with autopolymerising acrylic resin. On the second appointment, the individual trays were border moulded in the patient's mouth with a softened thermoplastic modelling compound, and a final impression was made with light-bodied polysulfide rubber base impression material (Permlastic, Kerr).

A master cast was poured in the final impression with type II dental stone. A base of autopolymerising resin was built up and rimmed with wax.

On the third appointment, the patient's vertical dimension of occlusion was determined through phonetic and esthetic criteria. Preliminary occlusal relations were recorded using autopolymerising bases with wax rims and a Fox's plane. Maxillary and mandibular casts were then mounted on the articulator.

On the fourth appointment, acrylic resin teeth specific for children dentures (bambino tooth, Major, Moncalieri, Italy), shade A2, were selected. Teeth were mounted to allow the resin teeth to be set over existing and yet maintain proper occlusal and esthetic relationships. The dentures were tried in, and required adjustments were carried out. After proper adjustments the dentures were inserted.^{9 15}

The upper and lower prosthetic devices were delivered on the fifth and sixth appointment, respectively. The patients were monitored clinically every month to assess prosthetic fit. Orthopantomography and cephalometric radiographs were taken every 6 months with the prostheses in place, in order to evaluate correct pattern of growth. A radio-opaque paint was layered over the prosthetic teeth to outline the occlusal surface on the cephalometric radiographs. After 2 or 3 months, according to the developmental growth, expander screws (three-way screw in the upper denture and two-way in the lower one) were inserted, after adequate sectioning of each denture, to allow the expansion of the denture following the jaws growth. The three-way screw of the upper denture, through cyclical activation, allowed the expansion of the palatine bone and the premaxilla. Parents were instructed to turn the expansion screws every 2 weeks following the pattern of growth proposed by Biörk *et al.*^{7 11 12} In order to improve the retention and comfort, the prosthesis was rebased with soft siliconic resin (Sofreliner, Tokuyama, Japan) (figure 1F).

If conical teeth were present, composite crown were realised and luted in order to improve the esthetics and phonesis of patients.^{6 10 17 18}

OUTCOME AND FOLLOW-UP

Prosthetic acceptance, masticatory improvement, esthetic improvement and phonetic improvement were evaluated (table 2).

DISCUSSION

Early oral rehabilitation improves oral function, phonesis and esthetics, reducing social impairment (figure 1B–E).^{6 10} The psychological effect of an oral rehabilitation on these children and their parents was very remarkable, because patients could follow the primary school without aesthetic complexes towards their schoolmates.^{8 10} All treated patients reported a significant improvement in their own diet because the oral rehabilitation also allowed them to eat meat, fish, fibrous vegetables and cheese.^{6 10} The acceptance of the removable prostheses was higher in patients with more own teeth. Fixed prosthesis showed a higher level of acceptance than removable prosthesis, probably because of the absence of micro-movements, and to improve retention and stability during mastication and phonesis. The majority of patients showed a good improvement of masticatory function after prosthetic rehabilitation. Phonesis improvement was good or very good for most patients. Prostheses were well accepted by the patients and their

Table 1 General characteristics of the patients

Patient no.	Date of birth	Gender	Medical history	Facial features	Total amount of teeth	Peg-shaped teeth
1	11/2002	Male		Depressed midface, prominent lips	7	Yes
2	12/2002	Male	Eczema, sinusitis and nasopharyngitis	Frontal bossing, depressed midface, prominent lips and depressed nasal bridge	4	Yes
3	06/2001	Male	Eczema, sinusitis and nasopharyngitis	Frontal bossing, depressed midface, prominent lips and depressed nasal bridge	7	Yes
4	11/2002	Male		Frontal bossing, depressed nasal bridge	13	Yes
5	04/2000	Male	Eczema, sinusitis and nasopharyngitis	Frontal bossing, depressed midface, prominent lips and depressed nasal bridge	6	Yes
6	12/1998	Male	Eczema, sinusitis and nasopharyngitis	Frontal bossing, depressed midface, prominent lips and depressed nasal bridge	4	yes
7	10/1999	Male	Nasopharyngitis	Frontal bossing, depressed midface	2	Yes
8	06/2001	Male	Eczema, sinusitis and nasopharyngitis	Frontal bossing, depressed midface and depressed nasal bridge	12	Yes
9	04/1997	Male	Eczema, sinusitis and nasopharyngitis	Frontal bossing, depressed midface and depressed nasal bridge	0	Yes
10	11/2001	Male	Eczema and nasopharyngitis	Depressed midface, prominent lips	14	Yes
11	09/1998	Male	Eczema	Depressed midface, prominent lips and depressed nasal bridge	6	Yes
12	01/1991	Male	Eczema, sinusitis and nasopharyngitis	Frontal bossing, depressed midface, prominent lips and depressed nasal bridge	2	Yes
13	12/1997	Male		Frontal bossing, prominent lips	18	No
14	04/1999	Male	Nasopharyngitis	Depressed midface, prominent lips	0	Yes
15	01/2005	Male	Nasopharyngitis	Frontal bossing, depressed midface, prominent lips	4	Yes
16	06/2001	Male	Eczema and nasopharyngitis	Depressed midface, prominent lips and depressed nasal bridge	7	Yes
17	7/2003	Male	Eczema, sinusitis and nasopharyngitis	Frontal bossing, depressed midface, prominent lips and depressed nasal bridge	5	Yes
18	9/1999	Male	Eczema, sinusitis and nasopharyngitis	Frontal bossing, depressed midface, prominent lips and depressed nasal bridge	2	Yes
19	09/2004	Male	Eczema, sinusitis and nasopharyngitis	Frontal bossing, depressed midface, prominent lips and depressed nasal bridge	7	Yes
20	12/2000	Male	Eczema and nasopharyngitis	Frontal bossing, depressed midface, prominent lips and depressed nasal bridge	9	Yes

Table 2 Prosthetic treatment and its effects on mastication, esthetics and phonetics

Patient no.	Age at first prosthetic rehabilitation	Prosthetic treatment	Prosthetic acceptance*	Masticatory improvement*	Esthetic improvement*	Phonetic improvement*
1	3 years	Removable prostheses	++	++	++	+
2	4 years 6 months	Removable prostheses	+	+	++	++
3	3 years 5 months	Removable prostheses	++	++	+	++
4	7 years 8 months	Orthodontic treatment with fixed prosthesis	+++	++	++	+
5	2 years 6 months	Removable prostheses	++	++	++	+++
6	4 years 10 months	Removable prostheses	++	+++	+++	++
7	3 years 2 months	Removable prostheses	+	+++	++	+++
8	3 years 2 months	Removable prostheses	++	++	+++	++
9	3 years 9 months	Removable prostheses	++	+++	+++	++
10	4 years 6 months	Removable prostheses and fixed crowns	+++	+	+	+
11	4 years 4 months	Removable prostheses	++	++	++	++
12	2 years 6 months	Removable prostheses	+	+++	+++	+++
13	6 years 8 months	Orthodontic treatment with fixed prosthesis	+++	+	+	+
14	3 years 8 months	Removable prostheses	+++	+++	+++	+++
15	3 years 2 months	Removable prostheses	++	+++	+++	++
16	2 years 6 months	Removable prostheses	++	+++	++	+++
17	3 years 4 months	Removable prostheses	++	++	+++	++
18	3 years 1 month	Removable prostheses	++	+++	+++	++
19	2 years 9 months	Removable prostheses	++	+++	++	++
20	3 years 7 months	Removable prostheses	++	+++	++	+++

*Evaluations were made at least 5 years after the first prosthetic rehabilitation; + fairly good, ++ good, +++ very good.

parents.^{8 15 19} Different kinds of prosthetic rehabilitation with removable dentures are suggested in Literature, but none of these studies employ expansion screw as proposed in the present study. The literature reports that the growth of the symphysis stops about at 2 years.²⁰ The clinical experience gained in more than 15 years of oral rehabilitation in growing patient with EDs shows transversal changes of the anterior mandible that make necessary the realisation of a new lower denture about every 6 months. The placement of a two-way expanding screw could tell us if a real transversal growth occurs in anterior mandible and could to follow the transversal expansion of anterior mandible. Three-way expansion screw allows a maxillary growth on both the transversal and sagittal planes, without influencing the palatine suture and the premaxilla, while a two-way expanding screw allows the mandibular growth on the transversal plane.

Learning points

- ▶ Early oral rehabilitation improves oral function, phonesis and esthetic, reducing social impairment.
- ▶ The placement of a two-way expanding screw could tell us if a real transversal growth occurs in anterior mandible and could to follow the transversal expansion of anterior mandible.
- ▶ Two-way expanding screw permits the mandibular growth in the transversal plane without locking it.

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Competing interests None.

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