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DOI: 10.23804/ejpd.2018.19.04.11

Interceptive treatment in ectodermal dysplasia using an innovative orthodontic/prosthetic modular appliance. A case report with 10- year follow-up

Aim *The treatment of a complex case of hypohidrotic ectodermal dysplasia (HED) with severe oligodontia.*
Case report *A 6 years old boy with HED, was treated with an orthodontic/prosthetic modular appliance. The device is custom made and consists of two parts, upper and lower, which were partially removable and partially fixed. The patient was prepared to receive dental implants for definitive oral rehabilitation. The treatment begun with heath-cured acrylic resin removable appliance with expansion screw in the maxilla and in the mandible. Afterwards, an innovative orthodontic/prosthetic modular appliance was made in the maxilla and in the mandible, fixed with bands on the first permanent molars, with expansion screw and telescopic screw that follow and support the resin prosthetic teeth during the orthopaedic expansion. The resin prosthetic teeth are removable from the metallic fixed structure of this appliance.The patient was followed for 10 years from the beginning of treatment.*

Conclusions *The modular appliance here described and our therapeutic approach showed to be efficient and durable in the achievement of many goals in the treatment of a complex case of HED. The objectives were not only just orthodontic, but also prosthetic and psychological.*

Keywords *Hypohidrotic ectodermal dysplasia, Interceptive treatment, Oligodontia, Orthodontic/prosthetic appliance.*

Introduction

The ectodermal dysplasias (ED) represent a large heterogenous group of diseases that affect not only the superficial ectodermal layer but also the mesectodermal layer derived from the neural crest [Dellavia et al., 2010; Coston et al., 1988].

The degree of dysfunction of sweat glands distinguishes EDs into hidrotic and hypohidrotic variants. Hypohidrotic ectodermal dysplasia [HED], also called Christ-Siemens-Touraine syndrome, is the most frequent ED variant with a prevalence of 1 every 100,000 births and it is identified by hypotrichosis, hypohidrosis, and hypodontia [Dellavia et al., 2010; De Aquino et al., 2012]. Common phenotypic forms of HED are frontal bossing, everted lips, saddle nose, hypopigmented skin, reduced vertical facial height and cranial base width, small hard tissue palate, high-sets orbits, and a prominent chin, conferring to the patients' face a typical aged aspect [Dellavia et al., 2010; De Aquino et al., 2012; Clauss et al., 2014]. Moreover, patients present agenesis of both deciduous and permanent teeth and hypoplasia of the alveolar processes. Indeed, oligodontia (the agenesis of more than six permanent teeth) is associated with growth disturbances of the maxillofacial skeleton, leading to atrophy and a reduced development of the stomatognathic system. The severe facial and dental deficiencies affects the masticatory function and aesthetics, with subsequent psychological problems of self-confidence [Dellavia et al., 2010; El Osta Chaiban et al., 2011].

The ideal treatment option depends on age, growth status and degree of hypodontia. In young ED patients, removable partial and complete dentures can be easily fitted with regular adjustments (approximately every year) and replacements, to compensate the growth of the jawbones [Iannidou-Marathiou et al., 2010]. Clinical management should consider a multidisciplinary approach with psychologists and speech therapists to increase the self-esteem of HED patients for their social integration [El Osta Chaiban et al., 2011].

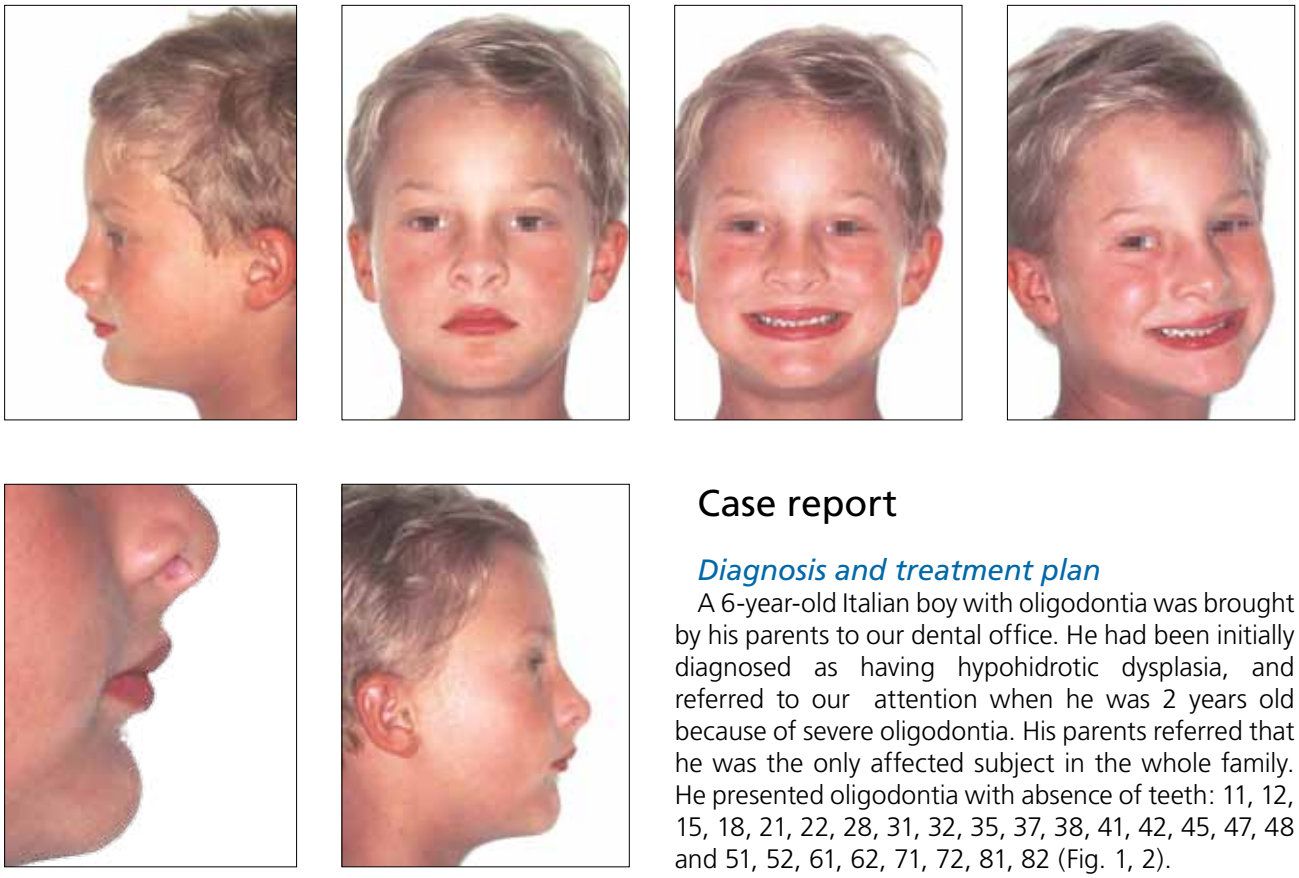


FIG. 1 Extra-oral pictures of the patients at 6 years of age.

Case report

Diagnosis and treatment plan

A 6-year-old Italian boy with oligodontia was brought by his parents to our dental office. He had been initially diagnosed as having hypohidrotic dysplasia, and referred to our attention when he was 2 years old because of severe oligodontia. His parents referred that he was the only affected subject in the whole family. He presented oligodontia with absence of teeth: 11, 12, 15, 18, 21, 22, 28, 31, 32, 35, 37, 38, 41, 42, 45, 47, 48 and 51, 52, 61, 62, 71, 72, 81, 82 (Fig. 1, 2).

Cephalometric investigation was carried out by comparative analysis by means of the Steiner and

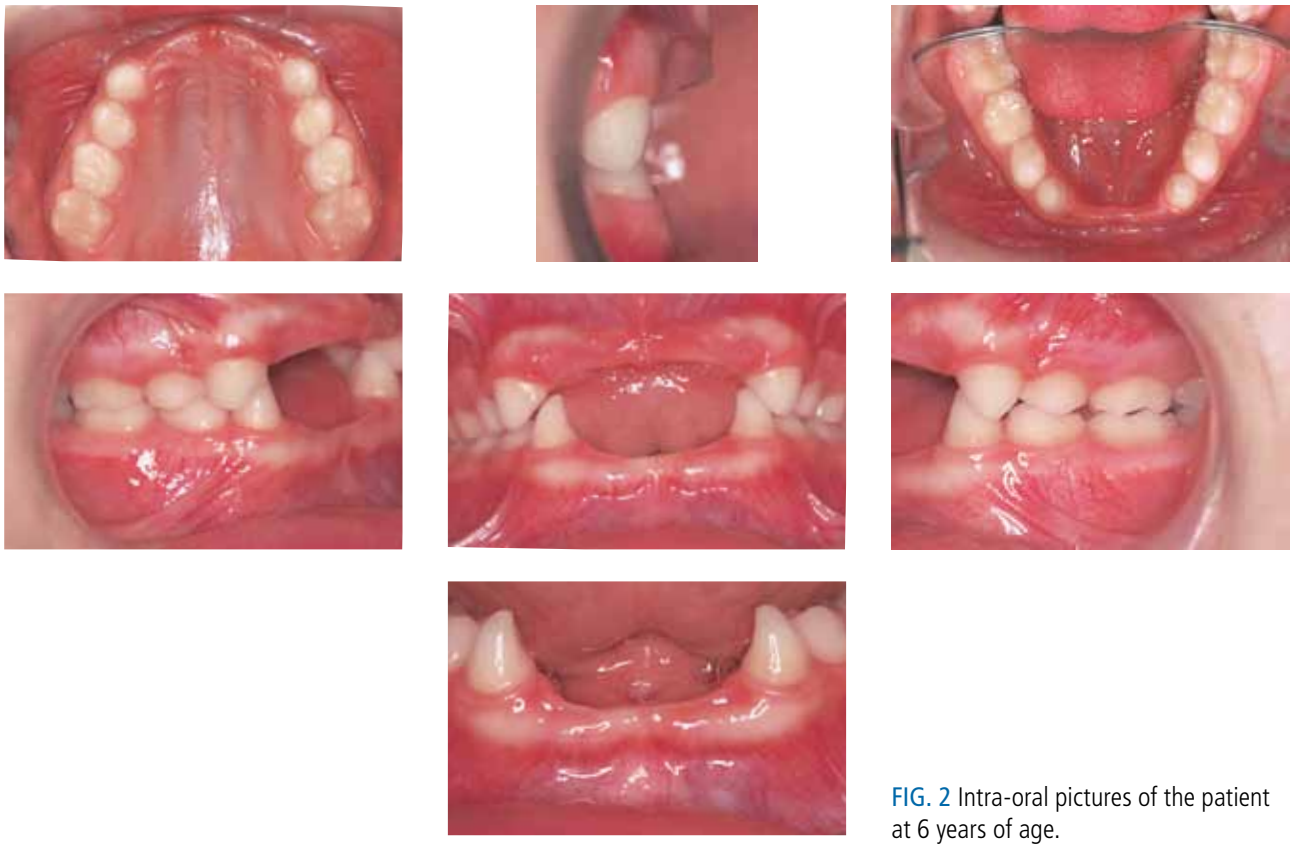


FIG. 2 Intra-oral pictures of the patient at 6 years of age.

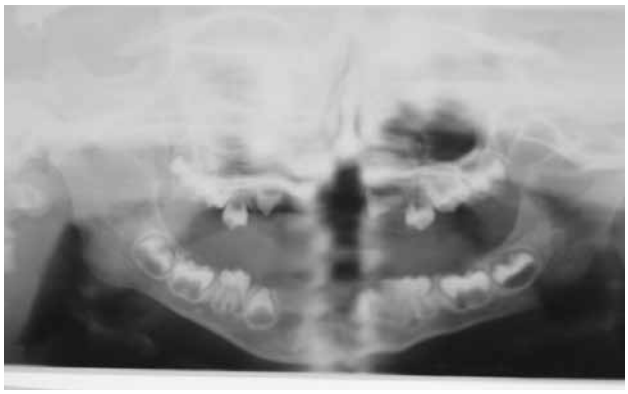


FIG. 3 Orthopantomography of the patient at 2 years of age.

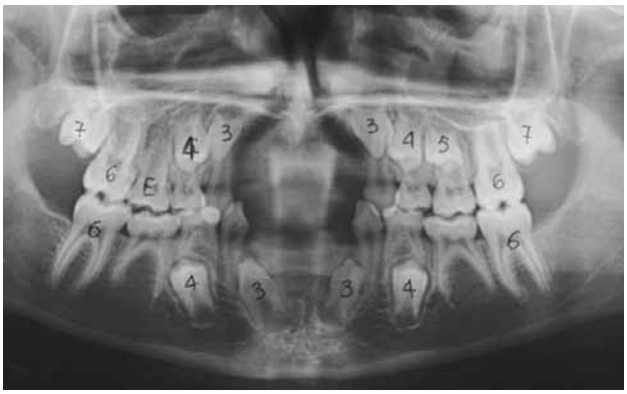


FIG. 4 Orthopantomography of the patient at 6 years of age.

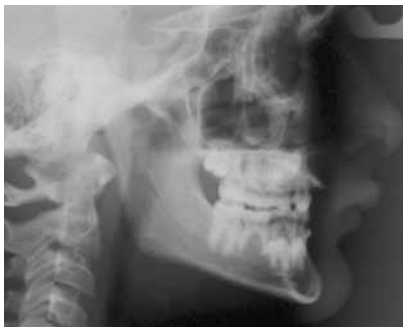
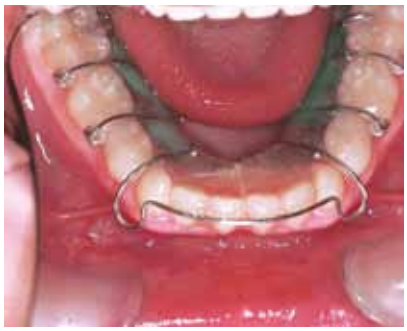


FIG. 5 Teleradiography of the patient at 6 years of age.



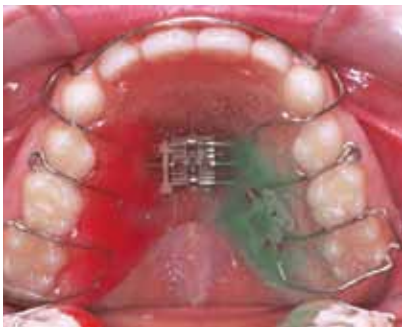


FIG. 6 First removable appliance.

Ricketts methods for basal classification. The patient showed a skeletal Class I with a Class III tendency, as well as maxillary and mandibular retrognathism. The vertical skeletal pattern was hypodivergent (Fig. 3).

The patient had Class I molar and canine relationship. Overjet and overbite could not be estimated because all front teeth were absent (Fig. 4, 5).

Extraoral examination revealed generalised hypotrichosis, frontal bulging, a depressed nasal bridge, lip thickening, and a prominent chin. The skin of the body appeared extremely dry and hairless. The intraoral examination showed complete upper and partial inferior edentulism, and severe maxillary and mandibular atrophy.

In addition to periodic caries prevention and dental hygiene instruction, partial upper and lower dentures were made at the age of 6 years. In the maxilla was placed a heath-cured acrylic resin appliance with expansion screw (Fig. 6).

The prosthetic rehabilitation with osteointegrated implants have to be postponed until the patient reaches adult age, because implants behave as ankylosed teeth during growth (Lamartine J. 2003). Therefore the main goal of the treatment was to maintain the space of the absent teeth and to provide a correct masticatory function and acceptable dental and facial aesthetics.

Since the patient had a Class III skeletal tendency with maxillary retrognathism, a rapid maxillary expansion was planned in order to produce an advancement of the cephalometric A point.

A new appliance, which could obtain all these targets, was designed and fabricated.

Appliance design and use

The device consisted of two parts, upper and lower, which were partially removable and partially fixed (Fig. 6). The device had to permit the maxillary rapid expansion without loosing stability. Furthermore it had to be easy to use for the patient. The maxillary rapid expander was produced by soldering the screw to the bands of the upper second deciduous molars and also by lengthening the arms up to the intercanine zone. The external spherical attachments were placed horizontally on the occlusal plane. The female attachment was made in a resin thermoplastic material modelling the hooks as guides on the upper deciduous canines (Fig. 7, 8). On the front teeth sufficient space was left in order to place the central screw, which would allow expansion of the maxillary bone (Fig. 9). A lingual arch was soldered to the bands of the lower second deciduous molars,using a telescopic system in the front to adapt to the development of the jaw. Both upper and

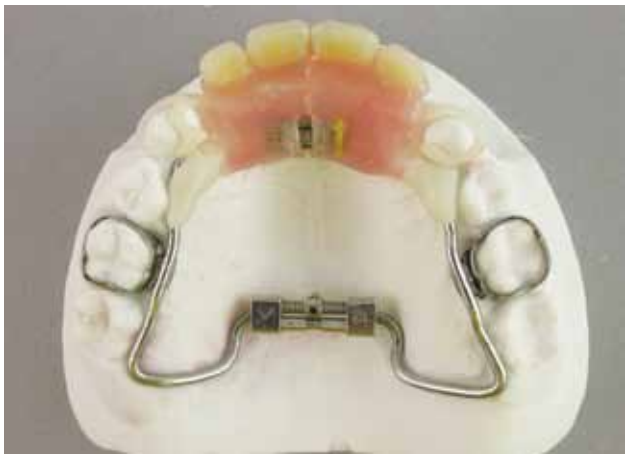


FIG. 7 The modular fixed/removable appliance on cast model of upper maxilla.

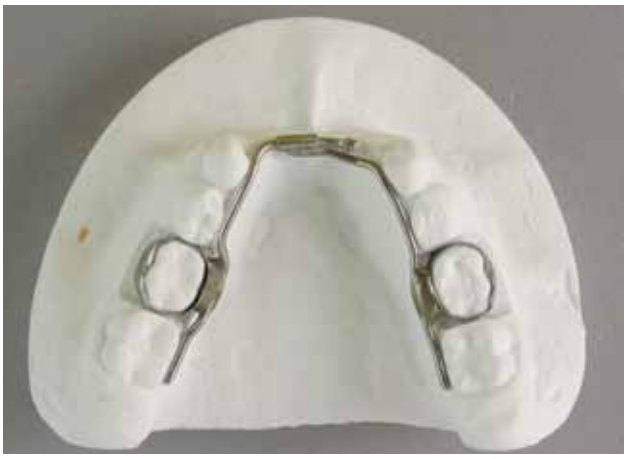
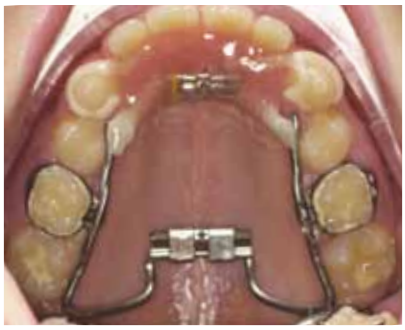


FIG. 8 The modular fixed/removable appliance on cast model of lower maxilla.

FIG. 9 Front wiev of the modular appliance on cast model.



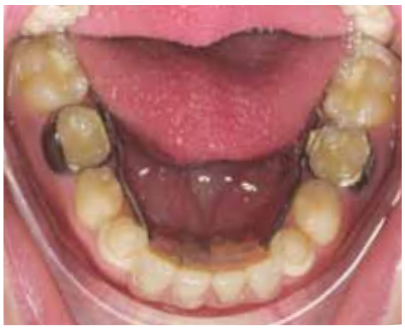


FIG. 10 Modular fixed/removable appliance with expansion screw *in situ*

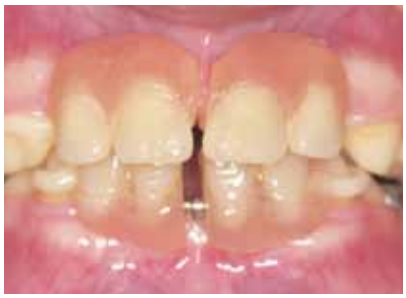
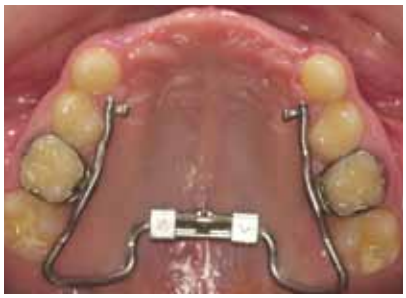


FIG. 11 Modular appliance after expansion.



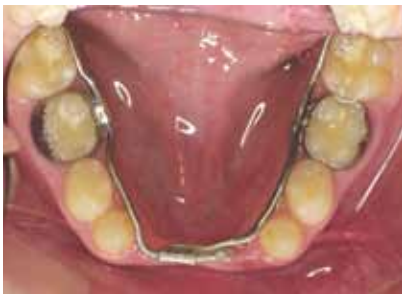


FIG. 12 Modular fixed/removable appliance without the front teeth.

lower appliances were split in the middle to follow the activations of the maxillary rapid expander (Fig. 10-14).

Treatment results

The treatment started at age of 6 years, with a removable heath-cured acrylic resin removable appliance with expansion screw in the maxilla and in the mandible. Afterwards, an orthodontic/prosthetic innovative modular appliance was prepared for the maxilla and the mandible, fixed with bands on the first permanent molars, with expansion screw and a telescopic screw that follow and support the resin prosthetic teeth during the orthopaedic expansion. The prosthetic resin teeth can be detached from the metallic fixed structure of this modular innovative appliance.

Discussion

Owing to the high clinical variability of EDs, accurate

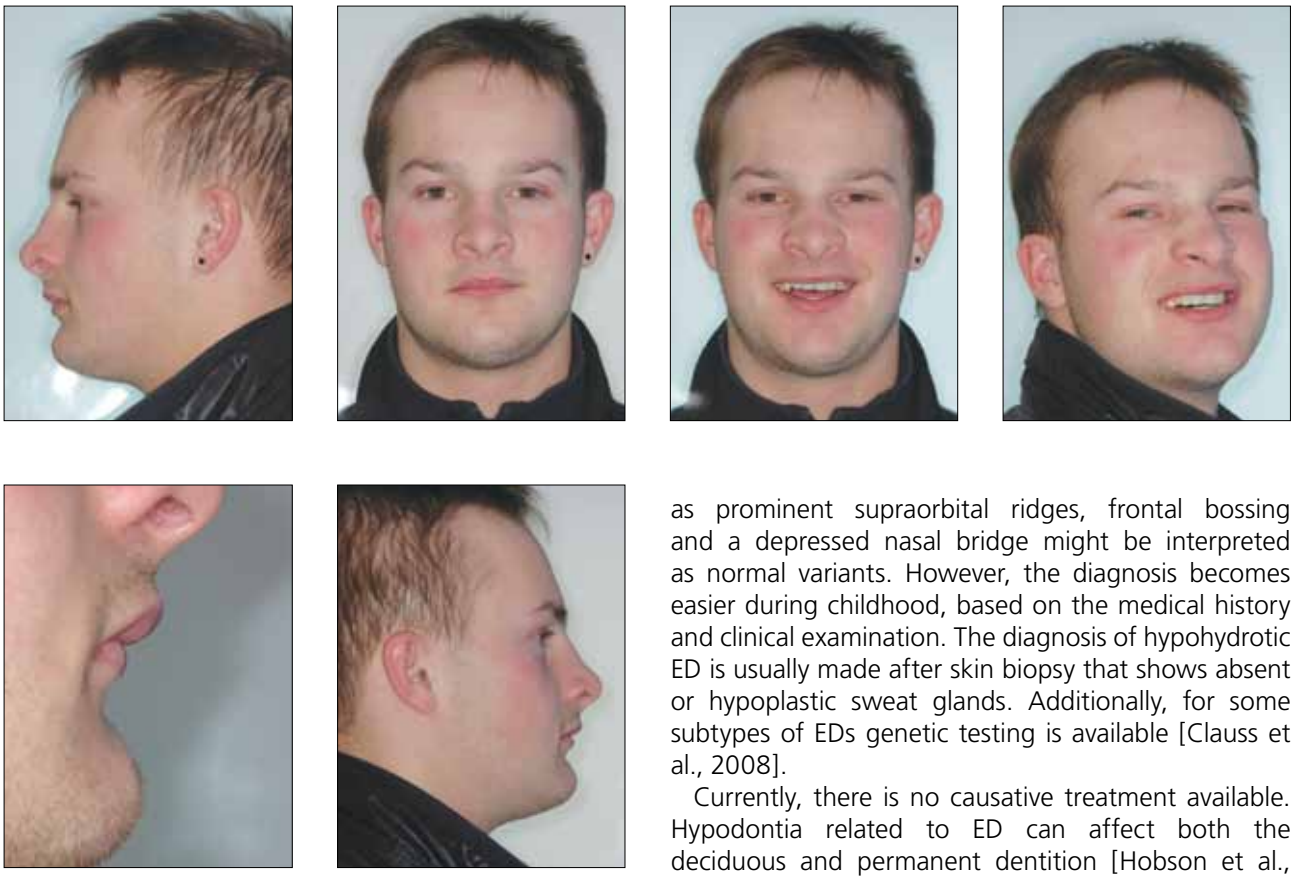


FIG. 13 Extra-oral wiev of the patient after ten years of treatment.

diagnosis can be difficult. There are over 170 clinically distinct hereditary syndromes in which ED is present [Lamartine, 2003]. EDs are rare diseases with an estimated incidence of seven every 100,000 births for all EDs [Itin et al., 2004]. Several classifications of EDs have been proposed from a clinical point of view [Pinheiro et al., 1994], with molecular genetic attributes [Priolo et al., 2000] and based on identified causative genes that most often are involved in processes of intercellular communication and signalling [Lamartine, 2003]. The clinical diagnosis of hypohidrotic EDs in the neonatal period and in the early infancy is difficult because sparse hair and absent teeth are often normal findings at this stage. Dysmorphic facial features such as prominent supraorbital ridges, frontal bossing and a depressed nasal bridge might be interpreted as normal variants. However, the diagnosis becomes easier during childhood, based on the medical history and clinical examination. The diagnosis of hypohydrotic ED is usually made after skin biopsy that shows absent or hypoplastic sweat glands. Additionally, for some subtypes of EDs genetic testing is available [Clauss et al., 2008].

Currently, there is no causative treatment available. Hypodontia related to ED can affect both the deciduous and permanent dentition [Hobson et al., 2003]. The permanent teeth most often present in hypohidrotic ED include the maxillary central incisors, maxillary first molars, mandibular first molars and maxillary canines [Guckes et al., 1998]. It was reported that especially in mild forms of ED, the most common concern in childhood and adolescence is about the dental anomalities and facial appearance [Siegel et al., 1990]. Consequently, the dentist, orthodontist or the maxillofacial surgeon are probably the first medical professionals to be confronted with complaints of EDs.

The conventional prosthodontic treatment of patients with severe hypodontia presents considerable problems [Boj et al., 1993]. The hypodontic mandible of children with ED exhibits underdeveloped alveolar ridges [Rashedi, 1993] and is therefore an area in which it is very difficult to gain adequate retention and support for conventional prostheses [Oesterle et al., 2000]. A prudent clinician should always attempt to use a conventional prothesis to gather functional

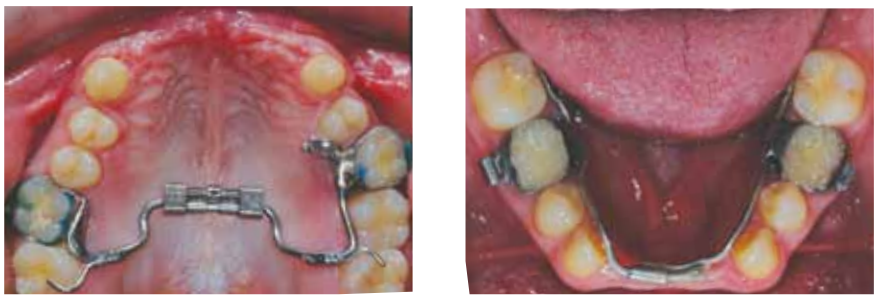


FIG. 14 Intra-oral view of the appliance after ten years of treatment without removable teeth.

and aesthetic information to aid in the design of the final prosthesis and to allow as much growth as possible before initiating the implant-assisted phase of treatment [Oesterle et al., 2000]. In treatment planning for implant placement in these patients, extra care must be taken to determine whether adequate bone level to receive the implants is present and whether there is adequate vertical dimension of bone to support the implants. Reduced bone volume may limit the success of implants, especially in the maxilla [Rad et al., 2007].

In ectodermal dysplasia the reduced bone volume available for implants, and the limited success of implants in the upper maxilla limits the possibilities of

fixed rehabilitation; as a result, it is not possible to place implants with primary fixation using the conventional technique, as indicated by Mattson et al. [1999].

Hypodontia associated with ED makes these patients candidates for dental implant reconstruction [Sweeney et al., 2005].

The main factors guiding the decision towards the orthodontic and prostethic choice are: the presence of posterior natural teeth, facial aesthetics, lip support, number and size of existing natural teeth, and the occlusal vertical dimension [Maroulakos et al., 2016].

However, in this particular condition, the lack of bone volume in young patients, owing to failure of development of the alveolar ridges, is a major challenge in providing future implant treatment.

A removable partial modular denture is a non-invasive, reversible, cost-effective, viable, and efficient treatment for a child with HED [Koscielska et al., 2017].

Successful treatment of the patient described in this paper is often possible thanks to a good relationship between the clinicians and the child's parents, who presented a positive attitude towards the process, as stated in another recent paper [Pinto et al., 2016].

Conclusion

When oligodontia occurs and all the incisors are absent, the interceptive treatment should combine the orthopaedic/orthodontic targets such as palatal expansion with the needs of the young patient, who requires a correct masticatory function and a good facial aesthetics in order to develop normal dietary habits and to have a rapid social integration.

The modular appliance described in this paper proved to be durable and effective in the achievement of all treatment goals, which were not only orthodontic, but also prosthetic and related to the self-esteem of the patient.

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