Dental Management and Prosthetic Rehabilitation of Patients Suffering from Hypohidrotic Ectodermal Dysplasia: A Report of Two Case Histories

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The prosthetic restoration and dental long-term care of two oligodont male cousins suffering from X-linked hypohidrotic ectodermal dysplasia is described in two case histories. The first patient was provided with three removable prostheses between ages 5 and 12, and the second patient was restored twice by tooth- and implant-supported dentures within an observation period of 10 years. In both patients, implants were placed in the growing jaw (both arches) in order to enhance denture retention by single attachments and/or a palatinal bar. In one patient, loss of one maxillary implant occurred 3 years after implant exposure and 2 years after the provision of the implant-supported denture. In the second patient, after completion of growth and preceding bone augmentation, further implants facilitated an improved removable maxillary restoration. In both patients, an orthodontic alignment and reshaping of the conical anterior teeth by means of resin or full-ceramic crowns was applied to improve the esthetic appearance. Patients with HED require a consistent lifelong attendance comprised of oral hygiene, denture maintenance, and refitting. In oligo-/anodont children, the placement of implants in selected regions may be useful for the enhancement of denture retention. However, due to poor bone quality and volume, implant failure may occur. Int J Prosthodont 2018;31:552–557. doi: 10.11607/ijp.5857

The term ectodermal dysplasia (ED) comprises a large group of hereditary disorders characterized by anomalies in structures of ectodermal origin.1 X-linked ectodermal dysplasia (XLHED; OMIM # 305100) represents the most common form, with an incidence of 1 per 17,000 births and a prevalence of 1 per 100,000 men and 17.3 per 100,000 women.2 X-linked inheritance effects a typical clinical expression in boys and less impaired phenotypes in heterozygous women (carriers). EDA1 has been identified as the disease-causing gene in XLHED. It encodes ectodysplasin, a collagenous protein implicated in ectodermal structure and osteogenesis.3–5 Typical phenotypes of HED display a hypoplasia of sweat glands, sebaceous glands, seromucous glands of the respiratory and the gastrointestinal tracts, salivary glands, and Meibomian glands, with clinical manifestations such as a reduced ability to sweat; dryness of skin, mouth, and eyes; and a susceptibility to eczema, infections of the eyes, ears, nose, and throat, the upper and lower airways, and the gastrointestinal tract.6 Hypotrichosis is expressed in sparse body/scalp hair and eyebrows/eyelashes. Typical oral features are hypo-, oligo-, or anodontia (lack of up to five, more than five, or all primary or permanent teeth) with a severe atrophy of edentate alveolar ridges and tooth malformations such as cone shape in anterior teeth and taurodontism in posterior primary and permanent teeth.2,7,8 Typical craniofacial signs are maxillary hypoplasia, mandibular prognathism, facial concavity, a reduced lower facial height, lip eversion, a prominent forehead, and a saddle nose. A variety of mutations affecting various exons of the EDA locus have been described and assigned to various dental phenotypes and craniofacial traits.7,9

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Although typical facial features and the inability to sweat, which is a risk for life-threatening hyperthermia,\textsuperscript{10,11} may allow the recognition of HED immediately after birth or in early childhood, this syndrome is often diagnosed only when teeth fail to erupt on time, erupt in an unusual position, or show malformation. According to a Danish study, the diagnosis of XLHED was most frequently set at an age between 11 and 18 years.\textsuperscript{12}

Orofacial impairment has a tremendous impact not only on vital functions such as breathing and food intake (ie, mastication), but also on speech and socialization. Therefore, dental rehabilitation should start in early childhood with a multidisciplinary team including a prosthodontist, an orthodontist, a maxillofacial surgeon, and a speech therapist in order to stepwise define and accomplish treatment goals.\textsuperscript{13,14} Therapy options range from preliminary removable dentures at an early age (preferably before school enrollment) to implant-supported removable or fixed prostheses in adolescence or adulthood after eventual orthodontic alignment of existing teeth or after orthognathic surgery. In general, dental implants should not be placed before the dental and skeletal maturation has finished\textsuperscript{15}; however, in children with severe oligodontia or anodontia, the insertion of implants in selected regions may enhance the retention of preliminary removable dentures and impede skeletal dental maldevelopment, such as an anterior rotation of the mandible in patients with maxillary retrognathism.\textsuperscript{16,17}

The long-term prosthetic management of two boys diagnosed with XLHED and oligodontia is presented in the following report. Subperiosteal palatal implants and/or endosseous dental implants placed at a young age were used to assist with denture retention. In one patient, failure of an endosseous maxillary implant is reported.

**Case Report**

**Medical History and Family Anamnesis**

Patients 1 and 2, male cousins of Caucasian origin, were referred to the University Hospital for Dental Prosthetics and Restorative Dentistry, Medical University of Innsbruck (Innsbruck, Austria) for dental treatment. Detailed case histories of both patients revealed an intolerance to heat; dry and scaly skin; dry mucosal surfaces; and delayed tooth eruption. In addition, patient 1 had been repeatedly treated for infections of the upper and lower airways during infancy. Extraoral examination showed sparseness of scalp hair, eyebrows, and eyelashes; dry and rough skin; protuberant and everted lips; and small nasal wings in both patients.

Genetic analysis identified a deletion in exon 4 (c.659_676del) of the EDA gene in both patients. The patients’ mothers are sisters and XHLED carriers, and both have hypodontia and mildly sparse hair. Patient 1 has one older brother also displaying manifest XHLED and one younger sister with hypodontia and conically shaped anterior deciduous teeth at the age of 5. Patient 2 has an older brother free from signs of XHLED. Anamnestically, the boys’ maternal grandfather had dental prostheses at a young age and intolerance to heat.

Informed written consent for the publication of their medical history/data and intra- and extraoral photographs were given by both patients and the parents of patient 1.
At the age of 7, the patient outgrew the fragile, repeatedly repaired “primary teeth” prostheses. The second prosthetic restoration was accomplished with overdentures containing galvano copings and cast metal reinforcements, again using the patient’s teeth as primary conical crowns (Fig 2).

At the age of 8, the exfoliation of deciduous teeth 51 and 61 (World Dental Federation [FDI] system) and the eruption of the conically shaped permanent teeth 11 and 21 took place. The maxillary denture had to be relieved and perforated in order to allow complete eruption, which negatively impacted denture retention. Due to growth, the mandibular denture required continuous adjustment (relieving) as well. When the patient was 10, the steps for the next prosthetic treatment were planned in order to establish a sufficient denture retention together with a favorable appearance: The insertion of one implant (Regular Crossfit 8 mm/4.1 mm, Straumann) in each maxillary tuberosity; the correction of tooth shape in the maxillary incisors with (provisional) resin crowns as a basis for an orthodontic line-up; the fitting of a new maxillary (partial) prosthesis retained by Novaloc retention elements (Straumann); and a new mandibular prosthesis containing a metal cast frame with a clasp in the meanwhile-erupted tooth 36 and computer-aided design/computer-assisted manufacturing (CAD/CAM) resin friction copings (Tecno Med, Zirkonzahn) as attachments to the conical canines. Figure 3 documents the realization of this treatment plan. The patient was pleased with the stable occlusion and chewing ability given by his implant-retained maxillary and tooth-retained mandibular dentures and was delighted with the esthetic outcome. The diastemas between the

**Dental Examination, Treatment Planning, and Prosthetic Restoration**

Oral and radiologic examinations of patient 1 at the age of 5 years showed oligodontia (erupted teeth: two deciduous maxillary incisors and two permanent maxillary incisors or canines, two permanent mandibular incisors or canines; retained teeth: two permanent maxillary incisors and one permanent mandibular molar) and an underdevelopment of edentulous alveolar ridges. The patient was administered his first set of resin overdentures at the University Hospital for Dental Prosthetics and Restorative Dentistry by use of his (unprepared) conically shaped primary and permanent incisors as abutment teeth. Figure 1 illustrates the radiologic and clinical states around the age of school enrollment.
maxillary incisors were being closed by a fixed orthodontic appliance.

Three years after implant exposure (following a healing period of 6 months) and 2 years after provision of the implant-supported maxillary denture, the implant in the left tuberosity loosened and was removed. To compensate for the reduction of denture retention, the insertion of one implant in the median palatinal suture and the provision of another single attachment, together with a new removable denture, were planned as next steps.

After completion of growth, a reevaluation of craniofacial proportions will be made and new treatment goals will be defined. Depending on the patient’s demands and willingness to undergo major surgery, maxillary osteotomy and advancement may be a treatment option if a compensation of the midface hypoplasia is indicated/desired. In any case, more implants (after bone augmentation in the severely atrophic maxillary and mandibular alveolar ridges) will be helpful for establishing a stable occlusion with removable or fixed prostheses.

Patient 2 had been treated at the Bernhard Gottlieb School of Dentistry when he was a child: With only three existing anterior teeth in the maxilla and two molars in the mandible, he had been provided with two palatinal onplants (OnPlant, Nobel Biocare) and four root-shaped implants (Nobel Biocare) in the interforaminal region of the mandible to achieve a sufficient denture retention. Restoration was accomplished by means of a bar-retained maxillary prosthesis and an overdenture in the mandible by use of customized conical attachments, which functioned satisfactorily for many years.

At the age of 17 (Fig 4), the patient presented at the University Hospital for Dental Prosthetics and
Restorative Dentistry for prosthetic rehabilitation. He had limited compliance concerning oral hygiene and adherence to appointments. He wished to keep surgical intervention to a minimum and had restricted financial resources. Therefore, with the aid of a lateral cephalogram, cranial computed tomography, and mounted casts so as to determine the optimal lower facial height and the alveolar bone conditions with respect to implantation, the following customized treatment plan was developed:

1. In the mandible, preprostodontic orthodontic alignment with gap closure between teeth 21 and 22 by means of a spring mechanism attached to the removable denture;
2. Bilateral alveolar ridge augmentation with iliac crest graft and subsequent placement of two implants (Regular Neck 10 mm/4.1 mm, Straumann) on each side;
3. Fitting of a removable palate-free prosthesis attached by Locator abutments (Zest Dental Solutions);
4. Crowning of teeth 13, 21, and 22;
5. And, in the mandible, placement of full-ceramic onlays in the two molars and of Locator attachments to the existing implants and fitting of a removable prosthesis containing two molar clasps.

This plan was—with slight modifications—carried out within a period of 2 years (Fig 5). Because of dysparity, the implant in the region of tooth 43 was provided with a parallelized pre-existing conical abutment instead of a Locator abutment. All teeth were kept vital. The lithium-disilicate crowns (e.max, Ivoclar Vivadent) for teeth 21 and 22 were linked for a better stabilization. The patient denied surgical lengthening of the clinical crowns for teeth 21 and 22, which would have improved the esthetic outcome. To avoid further surgery, the palatal bar was removed, and the onplants were left “sleeping.” Follow-up sessions including professional hygiene and denture service in 3-month intervals will assure maintenance of oral health and function over the years. If desired at any time, a restoration using fixed prostheses with further implants remains a treatment option.

Discussion

The rehabilitation of young HED patients with dental deficits and facial traits represents a challenge and should be approached by a multidisciplinary team of specialists. In 2013, an international Delphi study was published to determine an agreed approach to the management of children with dental manifestations of ectodermal dysplasia, including the use of dental implants. Despite a relatively high (early) implant failure rate in HED children due to small bone volume and the extreme hardness of alveolar bone and despite unpredictability of implant behavior in the growing bone, there has been agreement upon the appropriateness of implant placement in the mandibular canine region at the age of 5 to 10 years and in the...
maxilla at the age of 6 to 10 years or more. However, implants should not be placed adjacent to teeth until the completion of growth. In children, removable dentures were consensually considered an appropriate treatment option, whereas in adults, removable or fixed implant-supported dentures were agreed upon as a feasible treatment modality (aside from removable complete or tooth-supported partial dentures). Orthodontic treatment such as interceptive orthodontics, distribution of edentulous spaces, and dentoalveolar or jaw growth modification by means of functional appliances was consensually considered a good treatment option, if necessary, with the help of provisional implants providing orthodontic anchorage.

In the two presented patients, these consensually determined treatment recommendations were applied. In patient 1, one maxillary implant failed. A reduced bone quantity and quality were the presumed reasons for implant loss. In patient 2, a bar attached to subperiosteal onplants actually designed for orthodontic anchorage accomplished preliminary denture retention during the growth period. The provision of onplants splinted with a versatile bar was thereby indicated as a provisional solution for several years that would not impair sutural growth. However, domestic hygiene of the palatinal bar was difficult, with the mucosa being prone to proliferation and mycosis in the constantly humid milieu underneath the denture. Moreover, the use of onplants has been proven insufficient and is contraindicated for permanent maxillary rehabilitation in adults.

In both patients, a consistent regimen comprising oral hygiene, denture maintenance, and refitting or renewal will be undertaken for oral health and functioning over the future years. Owing to an increase of lower facial height and to reshaping of conical anterior incisors and canines by resin or full-ceramic crowns (together with an orthodontic alignment), the two cousins’ esthetic appearance has much improved.

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References


