

## Direct Composite Resin Application, and Prosthetic Management in a Patient with Hypohidrotic Ectodermal Dysplasia: a Case Report

E.Caner Tumen<sup>1</sup>, Nihal Hamamcı<sup>2</sup>, Yalcin Deger<sup>3</sup>, Demet Suer Tumen<sup>4</sup>, Engin Agackiran<sup>5</sup>

<sup>1</sup>Assist. Prof. DDS PhD Dicle University, Faculty of Dentistry Department of Pedodontics Diyarbakir/TURKEY.

<sup>2</sup>Assist. Prof. DDS PhD Dicle University, Faculty of Dentistry Department of Orthodontics/Diyarbakir/TURKEY.

<sup>3</sup>Assist. Prof. DDS PhD Dicle University, Faculty of Dentistry Department of Prosthodontics/Diyarbakir/TURKEY.

<sup>4</sup>Research Assistant DDS Dicle University, Faculty of Dentistry Department of Orthodontics Diyarbakir/TURKEY.

<sup>5</sup>Research Assistant DDS Dicle University, Faculty of Dentistry Department of Pedodontics Diyarbakir/TURKEY.

### Abstract

Ectodermal dysplasia is a rare group of disorders affecting the nails, hair, teeth, and sweat glands to a variable degree. The most common form of ectodermal dysplasia is X-linked hypohidrotic ectodermal dysplasia, which affects males more severely, while heterozygous females exhibit variable severity, ranging from mild to severe, because of inactivation of the X-chromosome.

The 7-year, 3-month-old boy presented the classical features of hypohidrotic ectodermal dysplasia, including diffusely sparse hair, eyelashes and eyebrows, severe hypohidrosis, and subsequent problems with thermoregulation, dry skin and fingernail defects. Considering the clinical situation, the age and potential growth of the young patient, a maxillary RPD and mandibular complete denture were determined to be the treatment of choice.

HED is usually a difficult condition to manage prosthodontically, because of the typical oral deficiencies and afflicted individuals are quite young to receive extensive prosthodontic treatment, which restores their appearance, for the development of a positive self-image.

(*Journal of International Dental and Medical Research 2009; 2: (1), pp. 19-24*)

**Keywords:** Hypohidrotic ectodermal dysplasia, hypohidrosis, composite resin restoration, prosthetic management.

Received date: 15 December 2008

Accept date: 25 February 2009

### Introduction

Ectodermal dysplasia (ED) is a congenital dysmorphogenesis of epithelial derivatives. This group of rare inherited disorders is characterized by the abnormal development in the embryonal stage.<sup>1,2</sup> It comprises over 150 distinct conditions of pathogenesis.<sup>2</sup> The commonly known signs of nail dystrophy (onchodysplasia), alopecia or hypotrichosis (scanty, fine light hair on the scalp and eyebrows), and palmoplantar hyperkeratosis<sup>3</sup> is usually accompanied by a lack of sweat glands (hypohidrosis) and a partial or complete absence of the primary and/ or permanent dentition.

Orofacial characteristics of this syndrome include hypoplastic conical teeth, underdevelopment of the alveolar ridges, frontal bossing, a depressed nasal bridge and protuberant lips.<sup>4,5</sup> The most frequently reported ED syndrome is X-linked recessive hypohidrotic ectodermal dysplasia<sup>4,6</sup> (HED; OMIM#305100) where males are usually more severely affected, and female carriers show variable severity ranging from mild to severe because of X-chromosome inactivation.<sup>2,6</sup>

The X-linked form of HED is caused by mutations in the *ED1* gene, located at Xq12-q13.1.<sup>2</sup> HED also known as Christ-Siemens-Touraine syndrome affects 1 to 7 individuals per 10.000 live births.<sup>7</sup> Most patients with HED have a normal life expectancy and normal intelligence. However, the lack of sweat glands may lead to hyperthermia, followed by brain damage or death in early infancy, if unrecognized. Thus an early diagnosis is important.<sup>8</sup>

Dentures typically are needed at an early age and can be problematic because of poorly developed alveolar ridges.<sup>9</sup> Oral rehabilitation of the HED patient is recommended to improve both the sagittal and vertical skeletal relationship during craniofacial growth and development<sup>7</sup>, as well as to

#### \*Corresponding author:

Dr. E. Caner TÜMEN  
Dicle University  
Faculty of Dentistry  
Department of Pedodontics  
Diyarbakir / TURKEY

Phone: + 90. 412. 2488101/3406

Fax: + 90. 412. 2488100

E-mail: emin.caner@isnet.net.tr

provide improvements in aesthetics, speech and masticatory efficiency. Prosthetic solutions for HED patients have consisted of various combinations of RPDs, fixed partial dentures and endosseous implants.<sup>3,9-12</sup>

The aim of this paper is to present the early prosthetic oral rehabilitation of a young boy with HED associated with severe hypodontia; only the maxillary primary centrals were present and total lack of teeth in the mandible.

## CASE REPORT

A 7-year, 3-month-old boy was referred to the Department of Pediatric Dentistry, Dicle University, Diyarbakir, Turkey for examination, evaluation and treatment. The chief complaints of the patient, who came to our clinic from an orphanage, accompanied by a nurse, were difficulty in eating because of a lack of teeth and mockery from his peer group.

The child presented the classical features of HED, including diffusely sparse hair, eyelashes and eyebrows, severe hypohidrosis, and subsequent problems with thermoregulation, dry skin and fingernail defects.

The facial profile showed a sunken nasal bridge, with prominent forehead and everted lips. An overclosed profile with decreased lower face height and creased oral commissures gave the impression of an older, more "aged" appearance<sup>12</sup> (Figs.1,2).



Fig. 1 Frontal view of 7-year-old patient with hypohidrotic ectodermal dysplasia before treatment.



Fig. 2 Pretreatment lateral facial photograph shows extremely decreased vertical dimension.

Clinical oral examination revealed large tongue, a slightly dry and sticky oral mucosa, and severe hypodontia; only the maxillary primary centrals were present and total lack of teeth in the mandible (Fig. 3).



Fig. 3 Intraoral examination revealed severe hypodontia; maxillary conical shaped primary centrals are present.

He exhibited aplasia of alveolar bone in the edentulous area. Also, the two conical shaped maxillary permanent centrals, and the right maxillary canine, were evident on the panoramic radiograph (Fig. 4).



Fig. 4 Panoramic radiograph. Right maxillary permanent canine, 2 permanent centrals and 2 permanent laterals.

primary centrals are present and total lack of teeth in the mandible.

The nurse stated that it was difficult for the child to speak and eat properly because of the lack of teeth. Considering the clinical situation, dental and skeletal maturity and the early age of the patient, a maxillary RPD and mandibular complete denture were determined to be the treatment of choice.

Cephalometric analysis findings revealed that the patient, with severe low angle pre-prosthetic rehabilitation was restored to a mesofacial status post-rehabilitation. The patient's anterior facial height was determined to have increased from 91 mm to 107 mm, and as a result of analysis of the jaws' relationship to the cranial base, the patient's facial depth angle was determined to have decreased from 91 mm to 86 degrees (ideal normative value  $86^{\circ}\pm 3$ ). At the same time, SNB angle decreased from 76 to 69,5 degrees and the relationship between the mandible and cranial base was restored to normal. Mandibular plane angle was observed to have risen from 17 to 26 degrees (ideal normative value  $17-28^{\circ}$ ) and nasolabial angle to have increased from 67 to 106 degrees (ideal normative value  $102^{\circ}\pm 8$ ), thus providing an aesthetically acceptable lip fullness (Figs. 5A, B).

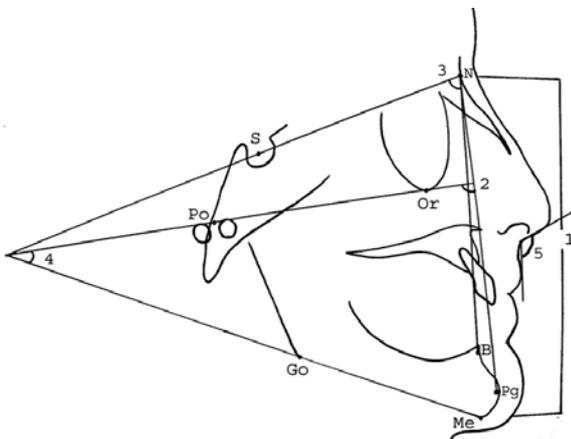


Fig. 5A Cephalometric landmarks, linear and angular measurements used in this case report. (N), Nasion: Most anterior point on fronto-nasal suture; (S), Sella: Midpoint of sella turcica; (Pg), Pogonion: The most anterior point on the mandibular symphysis; (Me), Menton: The lowermost point on the shadow of the mandibular symphysis; (Go), Gonion: The most outward point on the angle formed by the junction of the ramus and body of the mandible on its posterior, inferior aspect; (Po), Porion: The superior aspect of the external auditory meatus; (Or), Orbitale: The lower border of the orbit of the eye; (B), Point B: The

deepest (most posterior) midline point on the bony curvature of the anterior mandible. (1) Anterior facial height: The measurement between nasion and menton points. Representing anterior face development of the patient. (2) Facial depth angle: The angle between facial plane (N-Pg) and frankfort horizontal plane (Po-Or). Representing the horizontal development of the chin. (3) SNB angle: The angle between S-N plane and B point. Determining mandibular position according to cranial base. (4) Mandibular plane angle: The angle between frankfort horizontal plane (Po-Or) and mandibular plane (Go-Me). Representing the vertical development of the patient. (5) Nasolabial angle: The angle formed by the intersection of the columella tangent and the upper lip. Determining the upper lip position according to the nose.

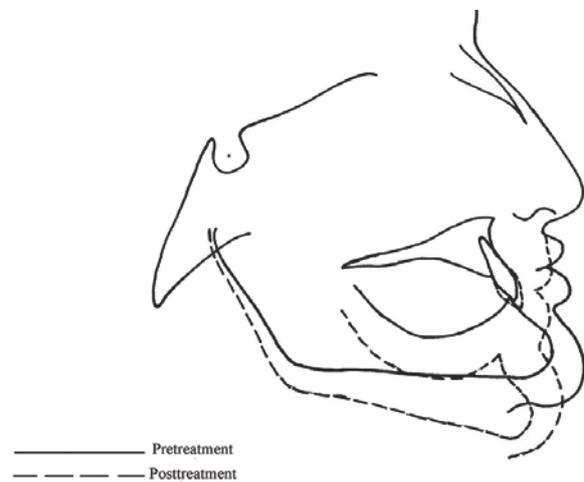


Fig. 5B Cephalometric superimposition.

In addition to function, particular attention was paid to appearance of the anterior region of the patient, who had presented with aesthetic complaints. Therefore, the maxillary primary centrals were initially morphologically restored with composite resin. Preliminary impressions were made with irreversible hydrocolloid (Cavex, CA37, Cavex Holland B.V. of Haarlem, The Netherlands).

Custom trays were fabricated with autopolymerized acrylic resin (Duracryl; Spofa Dental, Prague, Czech Republic), and definitive impressions (Elite, Zhermack, Rovigo, Italy) were made. Maxillomandibular records were made, and the casts were mounted in an articulator. The artificial teeth were arranged in wax for trial evaluation.

The occlusion and position of the prosthetic teeth were evaluated intraorally, and the necessary corrections were made before processing the dentures. The completed dentures gave the patient a natural appearance (Figs. 6-8A,B).



Fig. 6 Lateral facial photograph with dentures shows harmonious proportion of face.



Fig. 8A Pretreatment photograph.



Fig. 8B Posttreatment photograph.



Fig. 7 Frontal view of the patient after treatment.

Instructions were given to the child and the nurse to maintain a soft diet for the first few days to facilitate accommodation; also, the necessity of regular cleaning and maintenance was explained. The patient was instructed to remove the dentures at night and to present the following day and once a week for a period of 2 months for inspection and possible corrections and adjustments.<sup>13</sup>

The child was monitored every 3 months, and the nurse reported only minor problems during the adaptation period, and the accommodation to both partial and complete dentures occurred relatively rapidly, with considerable improvements in speech, aesthetics and general well-being, including the social adaptation of the child.

The denture must be periodically modified as alveolar growth, erupting teeth and rotational jaw growth change both the alveolar, occlusal and basal dimensions. As a result, the patient's dentures had to be remade 7 months later due to the growth of his jawbones.

## Discussion

Children with ED present many and different clinical problems from early childhood through adolescence and also present a life-long need for maintenance care and revisions. The principal aims of dental treatment for HED patients are to restore missing teeth and bone, establish the normal vertical dimension and provide support for the facial soft tissues.

The treatment modalities generally used include operative and prosthodontic treatment. For the hypoplastic teeth common with ED, direct composites or crowns often are used to restore proper contours to the teeth.<sup>9,14</sup> Treatment also can involve fixed, removable or implant prosthodontics, singly or in combination. Removable prosthodontics is the most frequent modality used for dental treatment of ED.<sup>15</sup> Although complete dentures are an acceptable form of treatment, overdentures that are supported by natural teeth will preserve the alveolar bone. Implant-supported restorations can improve physiological and psychosocial function to a greater degree than can complete dentures.<sup>9</sup>

Oligodontia or total lack of teeth associated with ED is often characterized by underdeveloped alveolar bone structures with missing or reduced alveolar ridges. This results in less volume of bone for support of conventional prosthetic dentures, and it can also affect the bone volume available for the placement of dental implants.<sup>10</sup> In developing the optimal surgical and prosthetic approach, the patient's age, dental and skeletal maturity, and the bone volume that is available at the time of intervention must be considered.<sup>16</sup>

In the present case report, considering the clinical situation, the age and potential growth of the young patient, a maxillary RPD and mandibular complete denture were determined to be the treatment of choice. Required dental implant application to provide an optimal treatment is postponed until the patient is fully grown. Early prosthetic rehabilitation of ED patients is crucial. Some authors recommend that the initial prosthesis be delivered before the child begins school so that the child has time to adapt to it.<sup>14</sup> This results in significant improvement in aesthetics, masticatory and phonetic functions.<sup>12</sup> In addition, the restoration of a natural and pleasing appearance is important for the normal psychological development of children with HED and their future social integration. Although dentures are poor alternatives to healthy teeth, they create conditions for the maintenance of a normal, satisfactory daily diet for the child. This is very important, considering that the establishment of lifelong dietary patterns occurs during

childhood.<sup>17</sup>

In HED patients, dryness of the oral mucosa and the underdeveloped maxillary tuberosities and alveolar ridges are problematic factors for resistance and stability of dentures. Therefore, retention and stability of the prostheses are difficult to obtain. When planning dentures in these patients, care should be taken to obtain a wide distribution of occlusal load fully extending the denture base. The remaining anterior teeth, due to their atypical conical shape, may not be suitable to stabilize RPDs. However, they may be used as abutments for overdentures.<sup>11,13</sup> Also, special attention must be paid to the impression technique; for complete dentures, support should not be limited to the denture base area but should also include the entire vestibular sulcus reflection for a retentive base construction with border seal.<sup>13</sup> For RPDs, the occlusion should be compatible with the patient's occlusion; generally, an occlusal scheme utilizing linear occlusal contact is recommended to preserve the existing teeth and to create freedom of movement.<sup>3</sup> In this case report, the patient had only minor problems related to extremely reduced alveolar process, which generally result in having poor retention, stability and inability to use prosthesis; moreover, the accommodation to both partial and complete dentures occurred relatively rapidly.

Periodic recalls of young HED patients are also important because prosthesis modification or replacement will be needed as a result of continuing growth and development.<sup>18,19</sup> In addition to adjustments related to fit, the occlusion of a prosthesis must be monitored for changes due to jaw growth.<sup>19</sup> Other problems related to removable prostheses are speech difficulties, dietary limitation, and loss of the prosthesis.<sup>18</sup>

## Conclusions

Young children with anodontia caused by hypohidrotic ectodermal dysplasia not only have difficulties in eating and speaking but can also sense that their appearance is different than others.

Early rehabilitation of children with hypohidrotic ectodermal dysplasia will go a long way in helping them interact normally and integrate with their peers. But one must remember that any form of restoration or prosthesis should provide dentition confirming with the age of the patient.

## References

1. Johnson EL, Roberts MW, Guckes AD, Bailey LJ, Phillips CL, Wright JT. Analysis of craniofacial development in children with hypohidrotic

- ectodermal dysplasia. *Am J Med Genet* 2002;112:327-334.
2. Kere J, Srivastava AK, Montonen O, et al. X-linked anhidrotic (hypohidrotic) ectodermal dysplasia is caused by mutation in a novel transmembrane protein. *Nat Genet* 1996;13:409-416.
  3. Bonilla ED, Guerra L, Luna O. Overdenture prosthesis for oral rehabilitation of hypohidrotic ectodermal dysplasia: a case report. *Quint Int* 1997;28:657-665.
  4. Levin LS. Dental and oral abnormalities in selected ectodermal dysplasia syndromes. *Birth Defects Orig Artic Ser* 1988;24:205-227.
  5. Champlin TL, Mallory SB. Hypohidrotic ectodermal dysplasia: a review. *J Ark Med Soc* 1989;86:115-117.
  6. Freire-Maia N, Pinheiro M. Ectodermal dysplasia: a clinical and genetic study. New York: Alan R. Liss;1984: 25-31.
  7. Buyse ML. Birth defects encyclopedia. St. Louis: Blackwell Publishing,1990:597-598.
  8. Mortier K, Wackens G. Ectodermal dysplasia syndrome. *Orphanet encyclopedia*. September 2004.
  9. Pigno MA, Blackman RB, Cronin RJ Jr, Cavazos E. Prosthodontic management of ectodermal dysplasia: a review of the literature. *J Prosthet Dent* 1996;76:541-545.
  10. Guckes AD, Scurria MS, King TS, McCarthy GR, Brahim JS. Prospective clinical trial of dental implants in persons with ectodermal dysplasia. *J Prosthet Dent* 2002;88:21-25.
  11. O'Dwyer MR, Renner RP, Ferguson FS. Overdenture treatment – one aspect of the team approach for the EEC syndrome patient. *J Pedod* 1984;8:192-205.
  12. Ramos V, Giebink DL, Fisher JG, Christensen LC. Complete dentures for a child with hypohidrotic ectodermal dysplasia: a clinical report. *J Prosthet Dent* 1995;74:329-331.
  13. Dyson JE. Prosthodontics for children. In: Wei SHY, editor. *Pediatric dentistry and orthodontics: total patient care*. Philadelphia: Lea & Febiger,1988: 259-274.
  14. Till MJ, Marques AP. Ectodermal dysplasia: Treatment considerations and case report. *Northwest Dent* 1992;71:25-28.
  15. Murdock S, Lee JY, Guckes A, Wright JT. A costs analysis of dental treatment for ectodermal dysplasia. *J Am Dent Assoc* 2005;136:1273-1276.
  16. Imirzalioglu P, Uckan S, Haydar SG. Surgical and prosthodontic treatment alternatives for children and adolescents with ectodermal dysplasia: a clinical report. *J Prosthet Dent* 2002;88:569-72.
  17. Kravitz E, Pollack RL, Mueller DH. Nutrition during pregnancy, infancy, childhood, and adolescence. *J Pedod* 1983;7:182-195.
  18. Nowak AJ. Dental treatment for patients with ectodermal dysplasia. *Birth Defects Orig Artic Ser* 1988;24:243-252.
  19. Nomura S, Hasegawa S, Noda T, Ishioka K. Longitudinal study of jaw growth and prosthetic management in a patient with ectodermal dysplasia and anodontia. *Int J Paediatr Dent* 1993;3:29-38.