

## **Early Prosthodontics Rehabilitation of a Patient with Hypohidrotic Ectodermal Dysplasia: A Case Report**

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**Abstract:** Ectodermal dysplasia is a hereditary disorder that occurs as a consequence of disturbances in the ectoderm of the developing embryo. Hypohidrotic ectodermal dysplasia is a congenital syndrome, characterized by hypotrichosis (hair is sparse, fine and weak; anomalies in the skin and nails), hypohidrosis (due to the paucity of sweat glands which in turn gives rise to sweat disorders) and hypodontia (partial, and occasionally total, absence of primary and/or permanent dentition). A case of a ten year old girl with hypohidrotic ectodermal dysplasia associated with partial anodontia is presented. A prosthetic rehabilitation in the form of a removable acrylic prosthesis was made, achieving excellent esthetics, functionality and adaptation.

**Key words:** Hypohidrotic ectodermal dysplasia, removable prosthesis, hypodontia, hypohidrosis.

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### **I. Introduction**

Hereditary ectodermal dysplasia is characterized by defective formation of one or more structures derived from ectoderm. It was first described by Thurnam in 1848 and was coined by Weech in 1929.<sup>1,2</sup> The clinical expression of ectodermal dysplasia varies, depending on the specific syndrome. The group was defined by Friere-Maia [1977] as expressing at least two of the following traits: trichodysplasia, abnormal dentition, onchodysplasia, and dyshidrosis. The disease may be inherited by autosomal-dominant, autosomal-recessive, or X-linked genetic transmission. These disorders, relatively rare, occur in 1 in 10000 to 1 in 100,000 births [Neville, 1995; Megarbane et al., 1998].<sup>3</sup> Ectodermal dysplasia is a group of disorders defined by the abnormal development of two or more structures derived from the ectodermal layer. The ectoderm, one of three germ layers present in the developing embryo, gives rise to the central nervous system, peripheral nervous system, sweat glands, hair, nails, and enamel of the teeth [Larson, 1993; Neville, 1995; Perabo et al., 1956]. A definitive classification of ectodermal dysplasia (ED) is difficult to formulate since many of the syndromes that involve ED have overlapping features. A simple attempt made by Nelson included five categories, namely Hypohidrotic (anhidrotic), Hydrotic (Clouston's syndrome), EEC (Ectodactyly ectodermal dysplasia) syndrome, Rapp-Hodgkin syndrome and Robinson's disease. The most frequently reported manifestation of ectodermal dysplasia is hypohidrotic dysplasia, also termed Christ-Siemens-Touraine syndrome and anhidrotic dysplasia.<sup>3,4</sup> The prosthodontic management of a ten year old child with hypohidrotic ectodermal dysplasia and partial anodontia is described here.

### **II. Case Report**

A ten year old girl reported to the department of Pedodontics and preventive dentistry with the chief complaint of missing teeth in both upper and lower dentition. Patient also complained of dry skin and absent of sweat in her skin. The child presented with features of Hypohidrotic ectodermal dysplasia and had been diagnosed much earlier by the paediatrician. Her family history and past medical history was insignificant and non-relevant, except the history of intolerance to heat. General examination of the patient exhibited the classical features of Hypohidrotic ectodermal dysplasia, characterized by hypohidrosis, hypotrichosis and hypodontia.<sup>5</sup> In addition to these signs and symptoms, the patient presented with dry skin, sparse eyebrows and hair. Extraoral examination revealed chin and supraorbital ridges were prominent. Frontal bossing and saddle nose present. Hypertonicity of the peri-oral and masticatory muscles caused speech and masticatory difficulties. The vertical dimension of the lower face was reduced and the lips were protuberant, leading to the characteristic senile appearance. The lower facial height decreased due to over closed profile, making lips more prominent and protuberant (Fig 1). Nail examination revealed no abnormality. The child was unable to tolerate hot climates and the parents revealed that the child used to wear wet clothes in summer to combat the heat.



Figure 1: Pre-operative extra oral view



Figure 2: Pre-operative intra oral view



Figure 3: Orthopantomograph

Intra orally palate was shallow and patient was partially edentulous. There was presence of maxillary permanent first and deciduous second molars bilaterally and presence of deciduous maxillary canines bilaterally and also presence of permanent mandibular first molars bilaterally. Partial anodontia was confirmed in an orthopantomograph (Fig 2 and 3). Examination of the mother showed no abnormality and no tooth was missing. Based on the typical clinical and radiographic feature, it was diagnostically concluded a case of hypohidrotic ectodermal dysplasia. After thorough examination, treatment plan to fabricate a removable partial denture was made. The parents and the patient were explained about the treatment plan. The objective was to preserve the health and restore function of the stomatognathic system. Considering the clinical situation and the age of the patient, a maxillary and mandibular removable partial denture were determined to be the treatment of choice and to improve inter arch relationship, as well as to provide improvements in esthetics, speech and masticatory efficiency. The maxillary and mandibular primary impression was made using polyvinyl siloxane putty impression material supported on an adhesive coated stock metal tray. Custom trays were fabricated with autopolymerized acrylic resin. Single step border molding was done using polyvinyl siloxane putty impression material. Conventional border molding using low fusing compound was avoided for better patient compliance, avoidance of discomfort, irritation and injury to the mucosa. Final impression were made using injection type polyvinyl siloxane impression material and master casts were poured. Temporary acrylic record bases were fabricated on the master casts and wax rims were fabricated. The maxillo-mandibular relationship was recorded conventionally, and the casts were mounted in an articulator. Smaller size acrylic teeth were obtained

and trimmed according to the esthetic requirements. Try in was done during which the patient and the parents were satisfied, the waxed-up dentures were processed in heat-polymerizing acrylic resin. Later the removable partial dentures were inserted (Fig 4, 5, 6) and instructions were given. It gave the patient a natural appearance. The removable partial dentures were delivered and instructions were given to patient and parents, regarding its use. Periodic recall visits were scheduled at one week, first, third & sixth month post-insertion and follow up was done.



Figure 4: A upper and lower removable acrylic partial denture



Figure 5: Post-operative view with maxillary and mandibular removable partial denture



Figure 6: Post-operative view of patient

### III. Discussion

Transmission of hypohidrotic ectodermal dysplasia is, in general, X-linked (females carry the responsible gene, and males suffer from the disease, although the carrying mothers usually bear some typical characteristic feature of the disease) and at times in autosomal recessive form. Mosaic expression is rare. Currently, hypohidrotic ectodermal dysplasia is related with a mutation of the protein ectodysplasin-A, related with the EDA gene in the q12-q13 locus of the X chromosome (consisting of 12 exons, 8 of which are responsible for encoding the EDA-A1 transmembrane protein which is related with ectodermal growth).<sup>6,7</sup> The diagnosis of ectodermal dysplasia can usually be done based on clinical findings, the sequencing of a gene for ectodermal dysplasia has given new possibilities for the detection of carriers of XLHED by molecular genetics.<sup>8</sup> Optimal treatment for children with hereditary ectodermal dysplasia requires multidisciplinary approach and knowledge of behaviour management of pediatric patients.<sup>13</sup> Nowak suggests “Tell Show Do” technique for the child management.<sup>9,10</sup> In the present case partial anodontia and the age of the patient dictated the fabrication of removable partial denture. The pathogenesis of anodontia lies in the aplasia of the dental lamina. Complete aplasia implies total absence of both deciduous and permanent dentitions.<sup>10</sup> The thermostatic mechanism of the

body is disturbed in these patient because of the hypohidrosis and subsequent decrease in sweating. This results in irritability and fever, especially during summer months.<sup>10</sup> The dysfunction accompanying HED imposes a considerable burden on affected children from the first days of life. The lack of teeth is a severe limitation, the effect of which becomes crucial as the child reaches school age. Early intervention with easily adapted and frequently replaced removable appliances is beneficial because it restores esthetics and function, thus assisting the child in social and emotional maturation.<sup>11</sup>

Usually hypodontia can be managed by removable partial dentures, fixed prosthesis and complete anodontia by complete dentures and implant supported dentures. However the use of implants in children has not been fully investigated and their use should be postponed in children under 13 years.<sup>12</sup> This is because of implant movement caused by jaw growth.<sup>6</sup> This recommendation is well supported by the survival rate study of the 243 implants placed in the anterior mandible, 221 (91%) survived. Of the 21 implants placed in the anterior maxilla, 16 (76%) survived. Fourteen of the 51 (27%) subjects had a failed implant.<sup>13</sup> Different authors have proposed different rehabilitation possibilities for these patients. In general, almost all agree in recommending the use of removable prostheses during the first stages of growth (3-5 years), allowing the adjustment of the vertical dimension or maxilla / mandible interrelationship, so as to later option where possible for provisional fixed prostheses until the patient finishes growing when a more stable and fixed situation is established and the possibility of implant treatment can be considered.<sup>6</sup>

Periodic recalls are often necessary in young patients as a result of continuing growth and development that mandates adjustments in the prostheses. Early rehabilitation of children with ectodermal dysplasia will go a long way in helping them interact normally and integrate them with their peers. Restoration or prosthesis should offer dentition that match the age of the patient. The principal aim of dental treatment is to restore missing teeth & bone, since it provides good esthetic, phonetic & masticatory comforts. It also help patients develop good psychological good image.<sup>14</sup>

#### IV. Conclusion

In our case, the patient showed typical clinical appearance with partial anodontia, sparse hair & heat intolerance suggestive of ectodermal dysplasia. Early diagnosis & dental treatment is vital in restoring the mastication, speech & esthetic in patient with ectodermal dysplasia, thus associated emotional & psychological distress is minimized in these patients.

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