



## ECTODERMAL DYSPLASIA WITH ANODONTIA; A CASE REPORT

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**ABSTRACT** Ectodermal dysplasia comprises a hereditary disorder which occurs as a consequence of disturbances in the ectoderm of the developing embryo. The triad of nail dystrophy, alopecia or hypotrichosis and palmoplantar hyperkeratosis is also accompanied by a lack of sweat glands and a partial or complete absence of primary and/or permanent dentition. A case reports illustrating the prosthetic rehabilitation of a young girl with anhidrotic ectodermal dysplasia associated with complete anodontia is presented. Since the oral rehabilitation of such case is often difficult; particularly in pediatric patients, treatment should be administered by a multidisciplinary team involving pediatric dentistry, orthodontics, prosthodontics and oral-maxillofacial surgery

**KEYWORDS :** Anodontia, hypodontia, oligodontia, oral rehabilitation, removable prosthesis.

**INTRODUCTION**

Ectodermal dysplasia, as first defined by Thurman,<sup>1,2</sup> is a hereditary disorder occurring as a consequence of disturbances in the ectoderm of the developing embryo. The triad of nail dystrophy (onchodysplasia), alopecia or hypotrichosis (scanty, fine light hair on the scalp and eyebrows), and palmoplantar hyperkeratosis is often accompanied by a lack of sweat glands (hypohidrosis) and a partial or complete absence of primary and/or permanent dentition.<sup>2,5</sup> Ectodermal dysplasia represents a large and complex group of diseases comprising more than 170 different clinical conditions.<sup>3</sup> The incidence of this condition is 1:100,000, with a mortality rate of 28% in males up to 3 years of age.<sup>1</sup> When at least 2 types of abnormal ectodermal features are present, such as malformed teeth and extremely sparse hair, the patient is diagnosed with ectodermal dysplasia syndrome.<sup>3,5</sup> There are 2 major types of this condition that depends on the number and functionality of the sweat glands: (1) X-linked anhidrotic or hypohidrotic, where sweat glands are either absent or significantly reduced in number (Christ-Siemens-Touraine syndrome), and (2) hidrotic, where sweat glands are normal and the condition is inherited as autosomal dominant (Clouston's syndrome).<sup>1,3,5</sup> The dentition and hair are equally affected in both types, but the hereditary patterns and nail and sweat gland manifestations tend to differ.<sup>2</sup>

Christ-Siemens-Touraine syndrome, with X-linked recessive inheritance, is the most frequently reported manifestation of ectodermal dysplasia.<sup>2,7,8</sup> Depending on the severity of clinical manifestations, Christ-Siemens-Touraine syndrome can be classified as either hypohidrotic or anhidrotic ectodermal dysplasia.<sup>8</sup> Oral manifestations of ectodermal dysplasia (ED) may be expressed as anodontia or hypodontia, with or without a cleft lip and palate. Anodontia also manifests itself by a lack of alveolar ridge development,<sup>7</sup> as a result of which, the vertical dimension of the lower face is reduced, the vermilion border disappears, existing teeth are malformed, the oral mucosa becomes dry, and the lips become prominent. The face of an affected child often has the appearance of old age.<sup>7,9</sup> Genetic studies regarding the etiology of ED reveal that mutations in the ectodysplasin-A and ectodysplasin-A receptor genes are responsible for X-linked and autosomal hypohidrotic ectodermal dysplasia.<sup>2</sup> The final diagnosis is based on clinical and radiological manifestations such as the number and distribution of sweat pores, the

amount of sweat produced, the structural and biochemical characteristics of hair, skin biopsy, and characteristics of lacrimal secretions.<sup>10</sup> Alopecia areata, incontinentia pigmenti, Werner syndrome, focal dermal hypoplasia, familial simple anhidrosis, and dyskeratosis congenita are some of the conditions through which the differential diagnosis is made.<sup>11</sup> The most common prosthetic treatment for the dental management of ectodermal dysplasia is removable prosthodontics. Since alveolar bone development is dependent on the presence of teeth, children with ectodermal dysplasia have little or no bone ridge upon which to construct dentures; therefore, restoring function and esthetics is more challenging than usual.<sup>3</sup> Follow-up by a multidisciplinary team involving pediatric dentistry, orthodontics, prosthodontics, and oral-maxillofacial surgery specialists is advocated to be the most accurate approach in such cases.<sup>8</sup> In this case report; it is aimed to describe the prosthetic rehabilitation of a young girl with anhidrotic ectodermal dysplasia associated with severe anodontia.

**CASE REPORT**

**Figure 1:**

A 13-year old female patient was referred to Deptt. Of Prosthodontics, Sh. J.N Kapoor D.A.V (C) Dental college and Hospital, Yamunanagar due to lack of teeth as well as speech and mastication problems. The lack of primary and permanent teeth in the oral cavity resulted in dietary problems; in addition, the child did not accept any previous dental intervention due to anxiety. During extraoral examination, the child exhibited the typical features of anhidrotic ectodermal dysplasia: saddle nose; soft, dry and light colored skin; increased pigmentation; as well as thin, linear wrinkles in the peri-oral region (Figure 1).



Figure 2



Figure 3

Intraoral examination revealed the total absence of primary and permanent teeth, thin alveolar ridges, reduced vertical bone height, and loss of sulcus depth in the posterior regions of maxillary and mandibular jaws (Figures 2 and 3). In order to improve appearance, mastication, and speech, removable complete maxillary and mandibular dentures were determined to be the best treatment choice. Routine procedures were followed for the construction of the dentures. Preliminary impressions were made with irreversible hydrocolloid impression material, and then custom trays were prepared for functional impression. Acrylic bases with wax rims were made on the master casts in order to establish maxillo-mandibular records. After establishing the maxillomandibular relation, the casts were mounted on an articulator. Rather than primary tooth forms, permanent tooth forms were selected so as to provide better static and dynamic occlusion. Primary tooth forms were not adequate to fulfill the ideal vertical dimensions as the patient is in the transition period from mixed to permanent dentition. After the final insertion, routine hygiene instructions for the dentures were given to both the child and his parents. (Figures 4 and 5)



Figure 4:



Figure 5:

The patient was advised to maintain a soft diet for the first few days, and to remove the denture at night so as to promote healing of the oral tissue. Despite the initial lack of compliance, the child tolerated the dentures quite well. In order to accommodate growth and development, the child was scheduled for ongoing follow up visits every 3 months. At recall appointments, good retention was observed and the parents reported a significant improvement in terms of speech and mastication; in addition, they discovered that she enjoyed wearing the dentures. Continued follow-up is suggested for modification or replacement of the dentures to fit the patient's developing maxilla and mandible.

#### DISCUSSION

Oral rehabilitation of the ectodermal dysplasia patient is essential to improve both the sagittal and vertical skeletal relationship during craniofacial growth and development as well as to provide improvements in esthetics, speech, and masticatory efficiency.<sup>2</sup> Although removable prostheses is the most common treatment method, dental implants can also be considered to be a treatment option. In situations where implant therapy can be used employed, the main hindrance is insufficient bone; if bone atrophy progresses in these already alveolar-deficient patients, placing an implant may not be possible without bone grafting.<sup>7</sup> Conversely, implantation reconstruction surgery poses a greater risk of failure compared to more conservative prosthetic treatment, besides its psychological aspects particularly in young children.<sup>8,12</sup> With the vertical development of the jaws, implant over-structures might not meet with the teeth of the opposing jaw, and might result in prosthetic infraocclusion.<sup>7,13</sup> Therefore, the use of implants in young children should be carefully evaluated, taking into account the above-mentioned issues, especially dental and skeleton maturation as compared to the chronologic age of the patient. In the abovementioned case, implant therapy was not the treatment of choice due to insufficient alveolar bone support. It is well-known that dental manifestations in ectodermal dysplasia may range from hypodontia to anodontia of the primary or permanent teeth. However, the congenital absence of primary teeth is relatively rare;<sup>5,9,10</sup> nevertheless, complete anodontia involving primary and permanent dentitions was observed in this case. There is little research regarding the impact of extensive hypodontia or anodontia on young children;<sup>14</sup> The prosthetic intervention with complete denture in this case is thought to be beneficial in terms of psychological development. Although the definite time to initialize treatment is still controversial, Till and Marquez<sup>15</sup> recommend that an initial prosthesis can be fitted when the child starts school, so that he may enjoy better aesthetics and will also have time to adapt to the prosthesis. Due to the lack of compliance in this case, the patient did not receive any dental treatment prior to his referral to our department. However, consequences of the delay in regards to speech, mastication, and psychological development were not determined to be significant. After employing appropriate behaviour management techniques, it was eventually possible to achieve prosthetic treatment.

Although dentures are poor alternatives to healthy dentition, they create conditions for the maintenance of a normal, satisfactory diet for

the child. This is very important, considering that the establishment of lifelong dietary patterns occurs during childhood.<sup>2</sup> Our observation in this case complies with this view. Dental prostheses also improves the tone of the muscles of mastication and compensates for the reduced vertical dimension.<sup>10</sup> Difficulty with mastication has been referred to as a major problem arising from loss of teeth.<sup>2</sup> As seen in this case, the facial profile and expression improved significantly with complete denture; and along with it, mastication and dietary patterns also improved. Regarding the dental treatment, it was possible to improve speech and communication skills. Thereby, a higher self-esteem and better social acceptance was promoted with the fabrication of complete dentures. This case report highlights the importance of accurate treatment planning as well as the influence of anodontia on the diagnosis of ectodermal dysplasia. As the oral rehabilitation of such cases is often difficult, treatment must be administered by a multidisciplinary team involving pediatric dentistry, orthodontics, prosthodontics, and oral-maxillofacial surgery.

## CONCLUSION

The clinical manifestations of ectodermal dysplasia cause considerable social problems in individuals affected by the condition. In this case report, the prosthetic rehabilitation of a young girl with anhidrotic ectodermal dysplasia associated with severe anodontia was described. Since oligodontia or complete anodontia leads to atrophy of the alveolar bone, prosthetic treatment is of great importance to these patients from functional, psychological, and psychosocial standpoints.

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