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# Ectodermal Dysplasia- A Case Report

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**Abstract**

Hypohidrotic ectodermal dysplasia is a congenital, non-progressive disorder characterized by hypodontia, hypohidrosis and hypotrichosis. It is inherited in an autosomal dominant, autosomal recessive, or X-linked patterns. The diagnosis is established by genetic tests or after infancy, based on physical features. In some patients, the pattern of inheritance is determined by family history and in others by molecular genetic testing. Characteristic changes in teeth in these patients are: both deciduous and permanent teeth are affected, the alveolar ridges are hypoplastic, missing teeth or retarded growth of teeth, peg-shaped, tooth enamel is also defective. Dental treatment is necessary and children as young as 2 years may need dentures. Through this manuscript, we report a case of hypohidrotic ectodermal dysplasia.

**Keywords**

Ectodermal Dysplasia; Genetic; Hypodontia; Prosthodontic Rehabilitation

The ectodermal dysplasia (ED) encompasses a colossal, heterogeneous group of hereditary disorders that are defined primarily by defects in the development of two or more tissues derived from embryonic ectoderm [1]. The tissues primarily involved are the skin, hair, nails, eccrine glands, and teeth. Although Thurnam published the first report of a patient with ectodermal dysplasia in 1848 [2], the term ectodermal dysplasia was not coined until 1929 by Weech [3].

The most common ectodermal dysplasias are X-linked recessive hypohidrotic ectodermal dysplasia (Christ-Siemens-Touraine syndrome), and hidrotic ectodermal dysplasia (Clouston syndrome). Current classification of ectodermal dysplasias is based on clinical

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features. Pure ectodermal dysplasias are manifested by defects in ectodermal structures alone, while ectodermal dysplasia syndromes are defined by the combination of ectodermal defects in association with other anomalies [4].

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Initially all three elements are divided into two groups hidrotic and anhydrotic. One of the more common types of ectodermal dysplasia also called anhidrotic ectodermal dysplasiaand Christ-Siemens-Touraine syndrome [5]. The condition is inherited as an X-linked recessive.

Characteristic changes in teeth in these patients are: both deciduous and permanent teeth are affected, the alveolar ridges are hypoplastic, missing teeth or retarded growth of teeth, peg-shaped, tooth enamel is also defective. Dental treatment is necessary and children as young as 2 years may need dentures [6].

## Case Report

A 7 year-old girl (EE), Albanian nationality, live in the village Svilare near Skopje city. Family history reveal that mother have hipodontia of upper lateral incisors. Orthopanthomogram investigations revealed presence of several deciduous teeth and four first permanent molars with
immature root growth (Figure 1) After two years another was taken (Figure 2). At that time the development of the first permanent molars were finished, but right low first permanent molar developed dental caries.

**Figure 1:** Orthopantomograph Showing Four First Permanent Molars with Immature Root Development

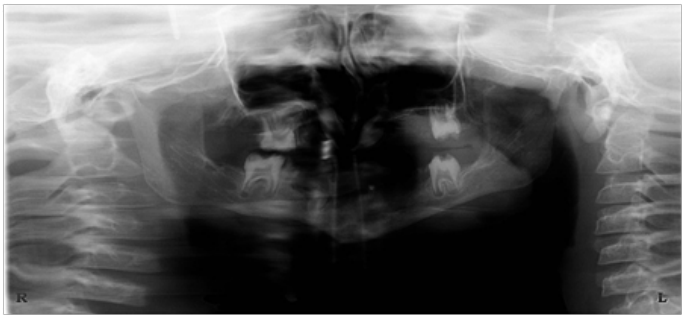
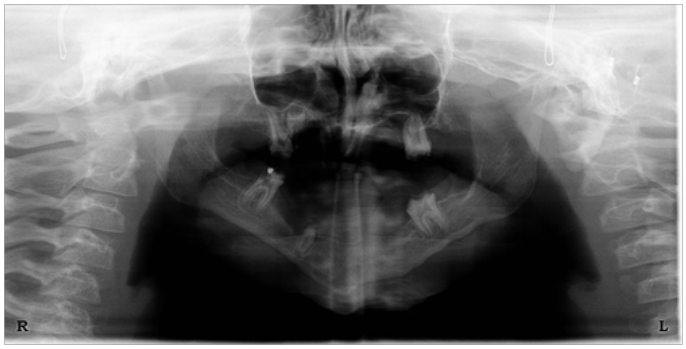


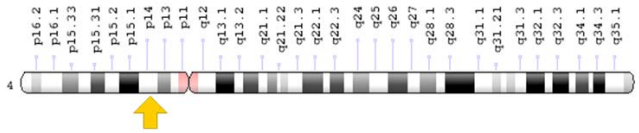

**Figure 2:** Orthopantomograph Took Two Year Later, Showing That the Development of The First Permanent Molars Was Finished




She had dry and sensitive skin since birth. Her scalp hair and eyebrows were absent and she wears a wig (Figure 3). The skin was dry, scaly, lichenified and excoriated. The nasal bridge was depressed, consistent with a “saddle nose”. She had maxillar and mandibular hypodontia with typical conical incisors and perioral erythema. Intraoral examination revealed undeveloped maxilla with poorly expressed tubers, flat palatal vault with slightly prominent and wide palatal tori, hypertrophic gingivobuccal plicas. Alveolar ridges were rather atrophic (knife - ridge) except in the areas where teeth were present. The color of alveolar mucosa and gingiva was normal. Severe hypodontia was present with missing most of the primary and buds of the permanent teeth. Underdevelopment of alveolar ridges was also confirmed by orthopantomogram (Figure 1) that revealed two permanent first molars teeth in the maxilla and two in mandible, as well as only two developing permanent teeth in the frontal region of the maxilla and one in mandible. Routine blood and urine laboratory tests were normal.

**Figure 3**: 7-Year Old Girl with Wig on Her Head

**Figure 4:** Chromosome 4 at Position 14




Since patient was only 7 years old, with undeveloped alveolar ridges, making of new maxillary and mandibular mobile dentures could be considered as a treatment. Preliminary impressions were made with appropriate stock trays and irreversible hydrocolloid material (Hydrogum soft, Zhermack, Italy). Casts were prepared using dental stone and custom trays (Plaque Photo Light - curing hybrid composite resin for making individual trays, Dentabiz, Sweden) were fabricated respectively. Border moulding was done with a thermoplastic material (Hoffmann's Impression Compound green, Germany) while the final (functional) impressions were made with light body polyvinyl siloxane impression material (Low viscosity C - Silicone Oranwash L, Zhermack, Italy). Final casts were made using hard dental stone and temporary bases (Hoffmann's Shellac Base Plates, Germany) with wax rim (Modeling wax, Dentaurum, Germany) were made respectively. Maxillo - mandibular relations were established, vertical dimension of occlusion and centric relation were recorded. Then the casts were mounted on a semi adjustable articulator and artificial teeth (NT Ünay acrylic resin teeth, Toros Dental, Turkey), reshaped considering the child's age, were arranged according to a balanced occlusion. Final trial was taken to verify vertical and centric relations, occlusion, phonetics and aesthetics. The maxillary and mandibular prosthesis (Figure 2) were fabricated in the conventional heat cure acrylic resin (SR Triplex Hot, Heat - curing denture base material, Ivoclar Vivadent, Schaan Liechtenstein). The dentures were then inserted in the patient's mouth and adjusted carefully.

## Discussion

Removable prosthesis made by acrylic resin (complete dentures or partial dentures) are the most frequently reported treatment modality for the dental management of ED in childhood; these are cost effective, and can be easily readapted and modified (relaying) during periods of rapid growth.

Because the absence of teeth predisposes the child to a lack of alveolar process growth, the construction of dentures is complicated. A deficiency in sweat glands causes a predisposition to increased body temperature, and children with hypohidrosis/anhidrosis are extremely uncomfortable during hot weather. Many of them must reside in cool climates (McDonald 2). Orofacial characteristics of this syndrome include anodontia or hypodontia, hypoplastic conical teeth, underdevelopment of the alveolar ridges, frontal bossing, depressed nasal bridge, protuberant lips, and hypotrichosis [7]. The case report of ectodermal dysplasia by Gupta et al demonstrated prosthetic management of ED through the strategic use of telescopic retainer in the mandibular arch and fixed prosthesis in the maxillary arch [8]. Škrinjarić I et al. noted that when used in conjunction with other methods the anthropometrics pattern profile analysis can considerably enhance detection of gene carriers for HED and increase objective assessment of the craniofacial region in HED patients [9]. Rathee M et al. in her study presented 6 year-old boy and concluded that dental restoration aids the patient in developing proper speech, deglutition, and mastication, and may have dramatic social and psychological benefits for these patients [10].

Chaiban R et al. state that gaining self-confidence after dental rehabilitation contributed tremendously to the development of this patients [11]. Cranioectodermal dysplasia can affect additional organs and tissues in the body. A kidney disorder in this patients can lead to a life-threatening failure of kidney function known as end-stage renal disease. Abnormalities of the liver, heart, or eyes also occur in people with cranioectodermal dysplasia. Cranioectodermal dysplasia is caused by mutations in one of at least four genes: the WDR35, IFT122, WDR19, or IFT43 gene. About 40 percent of people with cranioectodermal dysplasia have mutations in one of the four known genes. The cause of the condition in people without mutations in one of these genes is unknown.

The IFT-A complex is essential for proper regulation of the Sonic Hedgehog signaling pathway, which is important for the growth and maturation (differentiation) of cells and the normal shaping (patterning) of many parts of the body, especially during embryonic development. The exact role of the complex in this pathway is unclear.

## Cytogenetic Location

4p14, which is the short (p) arm of chromosome 4 at position 14

## Molecular Location

Base pairs 39,182,404 to 39,285,810 on

chromosome 4 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI) s figure 4) This case report describes a method for mobile prosthesis treatment of patient with ectodermal dysplasia. Excellent oral hygiene is crucial to the successful treatment of these patients. The patient should use topical fluoride daily for prophylaxis against caries during the treatment.

## Conclusion

This case highlights the positive effect of oral rehabilitation on the physical, emotional and social life of the patients with ED. Considering an age when patients should be dentally treated, making removable dentures is a rational, reasonable, acceptable and cost effective option.

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## Statement of Conflict of Interest

The authors declare no conflict of interest.

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