

Ectodermal Dysplasia – A Case Report

Abstract

Ectodermal dysplasia (EDs) are a large and complex nosological group of diseases characterized by congenital dysplasia of one or more ectodermal structure and other accessory appendages. The tissues in which the primary defects occur are the skin, hair, nails, exocrine glands, and teeth. In last 10 years more than 170 different pathological clinical conditions have been recognized and defined as EDs. Oral rehabilitation is a major challenge in such patients. One such case report of a 14 year old child with hypohidrotic ectodermal dysplasia is presented here.

Key Words

Hypohidrotic, Hidrotic, Ectodermal dysplasia, Oligodontia

Introduction

Ectodermal dysplasia (ED) is defined by National foundation for ectodermal dysplasia as a genetic disorder in which there are congenital birth defects of two or more ectodermal structures. These tissues primarily are the skin, hair, nails, eccrine glands, and teeth.^[1] Charles Darwin first documented the earliest accessible account of ectodermal dysplasia in English as early as 1840s. To date more than 150 distinctive phenotypes have been described with all possible modes of inheritance. The most common group are hypohidrotic (anhidrotic) ED also known as Christ-Seimens –Touraine syndrome or hidrotic ED (Clouston syndrome).^[2] Thurnam published the first report of a patient with ectodermal dysplasia in 1848, the term ectodermal dysplasia was not coined until 1929 by Weech. In hypohidrotic (anhidrotic) ectodermal dysplasia clinically there is anhidrosis, hypodontia and hypotrichosis. Other features include frontal bossing, thick lips, broad depressed nasal bridge of nose and deformed ears. Inheritance is usually X linked recessive trait. Ectodermal dysplasia results in aberrant development of ectodermal derivatives in early embryonic life. Hidrotic include the same without dental anomalies.^[3] Dental manifestations include conical teeth, hypodontia or complete anodontia, and delayed eruption of permanent teeth. ED is caused by mutations in any of the three EDA pathway genes: ectodysplasin (EDA) ectodysplasin receptor (EDAR), and EDAR associated death domain (EDARADD) which encode a ligand, a receptor, and an intracellular signal

mediator of a single linear pathway, respectively.^[2] Morbidity and mortality are related to absence or presence of eccrine and mucous glands. In affected patients, the extensive lack of both deciduous and permanent teeth results in an atrophy and a reduced growth rate of the affected alveolar processes.^[4]

Case Report

A 14 year old male patient reported to the department of Oral Pathology and Microbiology in Bhojia Dental College And Hospital, Baddi with the chief complaint of difficulty in mastication due to congenital missing teeth, lack of aesthetics, sparse hair on scalp and



Fig 1 - Sparse hair on scalp and thick everted lips



Fig 2 – Nails appear normal.

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absence of sweating. Extra oral examination revealed thin eyebrow, fine linear wrinkles and sparse hair and thick everted lower lip (Fig 1). Past medical and treatment history was insignificant and no positive family history was found. He was the first of the two children in his family and was the only affected member of his family. Nails appeared normal (Fig 2). Intraorally the patient was partially edentulous. Tooth number 51, 61 appeared conical and tooth number 52, 53, 62, 63, 71, 72, 73, 81, 82, 83 were absent (Fig 3). Upper and lower alveolar ridges appeared normal. His OPG was relevant to the above said findings (Fig 4). The combined dental and clinical findings pointed towards hypohidrotic ectodermal dysplasia. The treatment plan preferred was removable partial denture for both upper and lower arches and the upper



Fig 3 – Tooth number 51, 61 appears conical and tooth number 52, 53, 62, 63, 71, 72, 73, 81, 82, 83 are absent.



Fig 4 – Partially edentulous on intra oral examination.

midline diastema closure using elastic traction and restoration with composite resin (Fig 5).

Discussion

The ectodermal dysplasias comprise a large, heterogenous group of inherited disorders that are defined by primary defects in the development of two or more tissues derived from the embryonic ectoderm. The condition is thought to occur in approximately one in every 100,000 births. Genetic studies of more than 300 cases have revealed X linked mode of inheritance with its gene locus being Xq11-21.1, the gene is carried by the female but manifested in the male.^[1] Current classification of ectodermal dysplasias is based on clinical 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. Freire-Maia and Pinheiro proposed the first classification system of the ectodermal dysplasias in 1982,^[5] with additional updates in 1994 and 2001.^[6] The patients were stratified into subgroups based on presence or absence of the following:

1. Trichondysplasia (abnormal hair)
2. Abnormal dentition
3. Onchondysplasia (abnormal nails)
4. Dyshidrosis (abnormal or missing sweat glands)

Overall, the ectodermal dysplasias were classified into either group A disorders, which were manifested by defects in at least two of the four classic ectodermal structures as defined above, with or without other defects, and group B disorders, which were manifested by a defect in one classic ectodermal structure (1-4 from above in combination with (5), a defect in one other ectodermal structure (i.e., ears, lips, dermatoglyphics). HED or anhidrotic ectodermal dysplasia is the most common syndrome among this large group of hereditary disorders. Hypohidrosis, hypotrichosis and



Fig 5 – Removable partial denture for both upper and lower arches and upper midline diastema closure using elastic traction.

hypodontia constitute the main symptoms of the syndrome.^[7] In the hypohidrotic form, the skin is soft, thin, and dry. The sebaceous glands are also defective or absent. Palms and soles are hyperkeratotic, pseudorhagades are present around the eyes. In the oral cavity, the most striking feature is oligodontia. The teeth that are present have abnormal crown form. Teeth in the anterior region of maxilla and mandible are conical in shape.^[8] In the above mentioned case this feature was prominent. Oligodontia with only presence of tooth number 51 and 61 was seen. There was a wide midline diastema. The characteristic facial features like frontal bossing, depressed nasal bridge, prominent supraorbital ridges, prominent and obliquely set ears were also present in this case. Also the midface is depressed, the lower third of the face appears small due to lack of alveolar bone development, and lips are protuberant.^[9] The molecular pathogenesis of hypohidrotic ectodermal dysplasia (HED) is poorly understood. The gene responsible for X-linked HED, EDA, produces ectodysplasin-A, a protein that is important for normal development of ectodermal appendages including hair, teeth, and sweat glands. Evidence is accumulating that ectodysplasin-A is important in several pathways that involve ectodermal-mesodermal interactions during embryogenesis. Defects in the molecular structure of ectodysplasin-A may inhibit the action of enzymes necessary for normal development of the ectoderm and/or its interaction with the underlying mesoderm.^[10] The dental literature describes the dental treatment and management for the patient with anodontia or severe oligodontia. There is not a definitive time to begin treatment, but Pigno . et al.^[11] suggest that an initial prosthesis should be delivered before school age of the patient. As the child grows, the denture will have to be

modified and replaced, because it is indicated in the longitudinal studies of anodontia that the growth of jaws is independent of existence of teeth.^[12] When the patient is in the last stage, the removable prosthesis may be replaced by a fixed type restoration using osseointegrated implants. The implant-supported dentures reported are excellent in stability and retention but the placement of dental implants in young children generates several problems.^[13]

Conclusion

Young patients with ED need to be evaluated by a dental professional to determine the oral ramifications of the condition. When indicated, appropriate care needs to be rendered throughout the child's growth cycle to maintain oral functions as well as to address the aesthetic needs of the patient.

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