

Case Report

Hereditary Ectodermal Dysplasia – A Case Report

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ABSTRACT

Ectodermal dysplasias (EDs) are a large and complex group of diseases characterized by congenital dysplasia of one or more ectodermal structures and other accessory appendages. It is a genetic defect which is typically inherited as a cross-linked recessive trait where males are usually affected and females are carriers. The tissues in which the primary defects occur are the skin, hair, nails, exocrine glands, and teeth. Oral rehabilitation is a major challenge in such patients as it requires both functional and esthetic correction of face. Here, one such case report of hypohidrotic ectodermal dysplasia is presented.

KEY WORDS

Hypohidrotic, Oligodontia, ectoderm

INTRODUCTION

Ectodermal Dysplasias are a group of X linked recessive inherited disorders characterised by congenital dysplasia of one or more ectodermal structures and other accessory appendages. The tissues primarily are the skin, hair, nails, ecrine glands and teeth. The term ectodermal dysplasia was first reported by Thurman^[1] in 1848 and was first coined by Weech in 1929.^[2] 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.^[3]

It 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.^[3] Most common of them are X linked recessive anhidrotic (Christ-Siemens-touraine syndrome) and hidrotic ectodermal dysplasias (Clouston Syndrome).^[4] The frequency of different ectodermal dysplasia is highly variable as the condition is thought to occur in 100,000 births.^[5]

The patient usually requires both functional and esthetic correction of face. Oral rehabilitation is a major challenge in such patients; one such case report of hypohidrotic ectodermal dysplasia is presented here.

CASE REPORT

A 23 year old male patient reported to the department of Oral Medicine Diagnosis & radiology with the chief complaint of difficulty in mastication due to congenital missing teeth, lack of aesthetics, sparse hair on scalp and absence of sweating. History revealed as he grew older there were absence of teeth, sparse hair, progressive

hyperpigmentation and heat intolerance. Family history revealed non consanguineous marriage of his parents and the maternal uncle had similar complaint. Physical examination revealed recession of hair line all along with sparse thin, light brown brittle hair. On extraoral examination, prominent forehead, sunken cheeks with prominent supraorbital ridges, thin eye brows, hyperpigmentation around eyes and thick everted lip were noted. (Fig. 1) The skin was dry and scaly. On Intraoral examination, partial anodontia, hypoplastic peg shaped upper conical anterior teeth, generalised spacing, complete absence of mandibular teeth and underdeveloped alveolar ridges. (Fig. 2&3) Other findings include carious 53 and root stump w.r.t. 63. All routine investigations were normal. His OPG was relevant to the above said findings. (Fig. 4) With above history and clinical findings a diagnosis of hypohidrotic ectodermal dysplasia was put forth. The treatment option preferred was extraction of root stump wrt 63 and composite restoration of 53. The peg shaped lateral teeth were modified with composite restoration which helped in enhanced retention of maxillary denture. The removable partial denture was given for maxillary missing teeth and complete denture for mandibular missing teeth.



Fig. 1



Fig. 2



Fig. 3



Fig. 4

DISCUSSION

Ectodermal Dysplasias are congenital, diffuse, inherited and non progressive disorders.^[1] Current classification of Ectodermal dysplasia 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.^[6] From clinical aspect two main forms of Ectodermal Dysplasia are distinguished i.e. Hypohidrotic/ Christ Siemen's Tourian Syndrome and Hidrotic form/ Clouston syndrome.^[7] The basic differences between two forms are described in Table: 1.

Clinical features	Hidrotic Type	Hypohidrotic type
Hairs		
• Scalp Hairs	Soft Downy & Darker in color	Short, fine & fair in color
• Pubic & Axillary hairs	Scanty/ Absent	Not affected
Nails	Dystrophic nails	No abnormality
Nasal bridge	No Flattening	Under developed
Teeth	Anodontia - oligodontia	Anodontia - oligodontia
Lips	Not affected	Protuberant
Sweat glands	Not affected	Reduced to absent

Table 1: Differences between Hidrotic and Hypohidrotic forms of Ectodermal Dysplasia

Genetic studies of more than 300 cases revealed X linked mode of inheritance with its gene locus being Xq11-21.1; the gene is carried by the female but manifested in males.^[8] It is typically inherited as a cross linked recessive trait so that frequency and severity of the condition is more in males.^[7]

In developed countries diagnosis pertains to laboratory identification of genes but this may be difficult in developing countries like India where such facilities are insufficient. Therefore, above mentioned case is in accord with previous findings of most common type i.e. X linked recessive Hypohidrotic Ectodermal dysplasia. In this 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.^[9] In our case, the presentation of facial deformity, dry skin and sparse hair are due to anomalies in hair follicles, sweat glands and sebaceous glands.^[10] The heat intolerance is due to absence of sweat glands.^[11]

The most important aspect to be considered in these patients is the psychological impact on patient due to absence of teeth. The principal aim of dental treatment is to restore missing teeth and bone since it provides good esthetics, phonetics and masticatory efficiency.^[7] Generally, the time of treatment planning is not a definitive to begin with but Pigno et al.^[10] 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.^[11] When the patient is in the last stage, the removable prosthesis may be replaced by a fixed type restoration using osseointegrated implants.^[12] The prognosis of most patients is good as no evidence indicates that the lifespan for a person with common types of Ectodermal dysplasia is shorter than average.

CONCLUSION: Clinical recognition of Ectodermal dysplasia varies from birth to adulthood depending on severity of symptoms and recognition of associated complications. Once diagnosed the immediate appropriate care needs to be rendered to maintain oral functions and esthetics of the patient.

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