ORIGINAL RESEARCH PAPER

Dental Science

ECTODERMAL DYSPLASIA: A CASE REPORT

KEY WORDS: Ectodermal Dysplasia, Hereditary Disorder,oligodontia.

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ABSTRACT

Ectodermal dysplasia, a hereditary disorder is a triad of nail dystrophy, alopecia or hypotrichosis and palmoplantar hyperkeratosis is usually accompanied by a lack of sweat glands and a partial or complete absence of primary and/or permanent dentition.The two most common forms of the disease are hypohidrotic/anhidrotic ED and hidrotic ED.We present a case of a 11 year old child with hypohidrotic ED, who presented with oligodontia, dyshidrosis and raised body temperature. A multidisciplinary approach with physicians from several fields is required to provide comprehensive medical care to patients with ED.

INTRODUCTION

Ectodermal dysplasia is a hereditary disorder occurring as a consequence of disturbances in the ectoderm of the developing embryo.These tissues primarily are the skin, hair, nails, eccrine glands, and teeth.The incidence of ED is around 7 cases in 10,000 live births.The disease is broadly classified into hypohidrotic/anhidrotic (Christ–Siemens–Touraine syndrome) or hidrotic (Clouston syndrome), based on the presence or absence of sweat glands.[1] The common clinical features of ED include dental abnormalities, hypotrichosis, abnormal nails, and reduced ability to sweat.[2]

Oral traits of ectodermal dysplasia (ED) may be expressed as anodontia or hypodontia, with or without a cleft lip and palate. Freire-Maia and Pinheiro proposed the first classification system of ectodermal dysplasia in 1982.[1] They classified ectodermal dysplasia into different subgroups according to the presence or absence of (1) hair anomalies or trichodysplasia, (2) dental anomalies, (3) nail abnormalities or onychodys-plasia, (4) eccrine gland dysfunction or dyshidrosis.

CASE REPORT

A 12 year old boy was brought to thedepartment by his parents due to decreased sweating, dry skin, recurrent episode of high-grade fever, abnormally shaped teeth along with delayed eruption of other teeth .It was also brought to our notice that the child had intermittent episodes of fever in the past, associated with physical activity. Such episodes occured more frequently in hot climate, but no definite cause had been diagnosed. No other family menber reported similiar family history. Patient's vitals and systemic examination were normal.

On extraoral examination, parse and hypopigmented hair on the scalp were present and the eyebrows were sparse. The upper and lower eyelids showed sparse eyelashes. Fingers' examination revealed normal shaped fingers with thin, brittle nails. Patient had typical facies which was characterized by saddle nose, thick everted lips with midfacial hypoplasia (Fig.1).

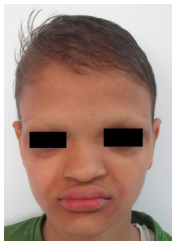

Fig.1: Extraoral view of the patient showing typical features of hypohydrotic ectodermal dysplasia.

Intraoral examination revealed dry mucous membrane, mandibular and maxillary oligodontia (Fig.2a,b) with four conical tooth erupted in anterior region and four posterior teeth in the maxilla and three teeth in the mandibular arch. Reduced vertical heights of both the arches were of considerable significance.

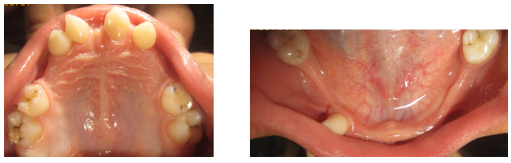

Fig.2a,b: Intraoral view of the patient revealing oligod ontia and resorbed ridges.

The clinical 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.[3] 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.[4]

Based on the history, clinical features, and examination, the child was diagnosed as a case of hypohidrotic ED. To improve appearance, mastication and speech, it was decided that removable complete maxillary and mandibular dentures would be appropriate for the patient.

DISCUSSION:

The literature describes more than 190 subtypes of ED, which can be classified according to the clinical features or the type of genetic mutation or the molecular pathway involved. Hypohidrotic ED is inherited by X-linked (most common), autosomal dominant, and autosomal recessive patterns.[1]

Clinically hypohidrotic/anhidrotic ED is characterized by hypotrichosis, hypo/adontia, dyshidrosis (abnormal sweating), and facial dimorphism[1] while hidrotic ED is characterized by the triad of onychodysplasia, hypotrichosis, and palmoplantar hyperkeratosis.[2] The clinical presentation of our patient was consistent with a case of hypohidrotic ED.

Mortality is as high as 30% in the first 3 years of life in children with hypohidrotic ED, due to numerous complications such as failure to thrive, pulmonary infections, and hyperthermia. After 3 years of life, life expectancy is normal.[5]

Oral rehabilitation of the ectodermal dysplasia patient is necessary 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.[6]

Although removable prostheses are the most common treatment method, dental implants are also considered to be a treatment option. Dental implants combined with implant supported dentures for adolescents over 12 years of age are recommended as a treatment choice in literature. In situations where implant therapy is indicated, the main problem is insufficient bone.[7] Improvement in the facial profile and expression should be significant with complete dentures and mastication, dietary patterns alongwith speech and communication skills should also improve.

CONCLUSION:

ED is a genetic disorder that affects not only the oral functions, but also normal body functions. A multidisciplinary approach is necessary for the rehabilitation of patients with ectodermal dysplasia.

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