CASE REPORT
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ECTODERMAL DYSPLASIA WITH TRUE ANODONTIA-A RARE CASE REPORT

Dentistry

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ABSTRACT

Ectodermal dysplasia is a genetic disorder caused by defects in the ectoderm that is developing in the embryo. The distinction between the hidrotic and hypo hidrotic kinds of ectodermal dysplasia is based on the presence or lack of sweat glands. A case of a 6-year-old male child with hypo hidrotic ectodermal dysplasia is given, together with the complete anodontia of both the primary and secondary dentitions. The youngster was under weight and was of short stature. The absence of sweat glands and a partial or total absence of primary and/or permanent dentition are frequently seen along with the triad of nail dystrophy, alopecia or hypotrichosis, and palmoplantar hyperkeratosis.

KEYWORDS

Ectodermal dysplasia, Anodontia, Hypohidrotic

INTRODUCTION

Ectodermal dysplasia is a very uncommon inherited congenital condition. Ectodermal dysplasia is a set of genetic diseases in which the development of two or more anatomic components that are derived from the ectoderm fails.[1]

It can be autosomal dominant, autosomal recessive and X-linked. There are more than 170 clinical illnesses that make up the broad and complex category of diseases known as ectodermal dysplasias.[2] The most commonly occurring ectodermal dysplasia is called as hereditary hypohidrotic (anhidrotic) ectodermal dysplasia.

Thurman was the first to characterise ectodermal dysplasia as a hereditary condition that results from defects in the embryo's developing ectoderm. The absence of sweat glands (hypohidrosis), primary and/or permanent dentition, and palmoplantar hyperkeratosis typically accompany the triad of nail dystrophy (onchodysplasia), alopecia or hypotrichosis (scanty, fine light hair on the scalp and eyebrows), and these conditions.[3]

1 in 100,000 people have this illness, and male children under 3 years old who die from it have a mortality rate of 28%.[5] The patient is identified as having ectodermal dysplasia syndrome when at least two different aberrant ectodermal characteristics, such as missing teeth and very little hair, are present.

According to the number and functionality of the sweat glands, there are two main types of this condition: (1) X-linked anhidrotic or hypohidrotic, in which the sweat glands are absent or significantly diminished in number (Christ-Siemens-Touraine syndrome), and (2) hidrotic, in which the sweat glands are normal and the condition is inherited as autosomal dominant (Clouston's syndrome), in which the sweat glands are normal.[6]

A male predominance is typically observed since this condition appears to have an X-linked pattern and has a gene that maps to Xq12-q13.1. Patients that are female may only partially exhibit this faulty gene. Affected individuals display the triad of anhydrosis/hypohydrosis, hypotrichosis, and dental hypoplasia.[7]

Case Report

A 6-year-old male patient reported to outpatient department with the chief complaint of missing teeth in relation to upper and lower arch since birth. History of present illness reveals congenitally missing deciduous and permanent, maxillary and mandibular teeth since birth. Medical history was non-contributory. However, there was positive family history of similar dental condition in his elder brother, maternal uncle and maternal grandfather. On general examination, built of child was asthenic with short stature and with normal IQ.

On extraoral examination, facial profile was straight with saddle nose, scanty and greyish eyebrows, sparse hair, sunken cheeks, hypertelorism, mid facial hypoplasia, depressed zygomatic arch, protruded lips, dry and crusted sole and lips(Figure 1). Hypofunction of sweat glands. Nails were thin and brittle.

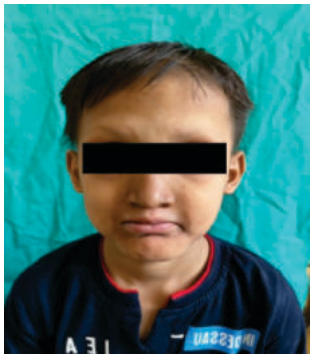


Figure 1 showing straight profile with hair abnormality

On intraoral examination, edentulous maxillary and mandibular arches were seen with flat palate, decreased alveolar process vertical height, underdeveloped mandible and deformed maxillary ridge (figure 2).

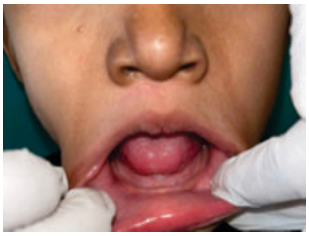


Figure 2 Showing Clinically Edentulous Maxillary And Mandibular Arches

Orthopantomogram (OPG) was taken which revealed complete absence of primary teeth and absence of permanent tooth buds as well as underdevelopment of alveolar ridges of maxillary and mandibular arches with reduction in height of alveolar bone (figure 3).

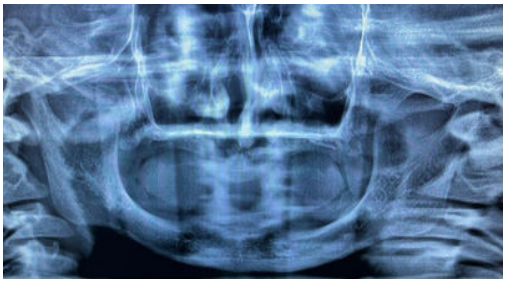


Figure 3 OPG of patient showing complete absence of teeth

As family history was positive an orthopantomogram (OPG) of his elder brother was also taken which revealed complete absence of both primary and permanent teeth.

Hence, based on history, clinical findings and radiographic findings a final diagnosis of hereditary ectodermal dysplasia was made.

Treatment plan- In order to improve mastication and aesthetics fabrication of maxillary and mandibular dentures was advised in the department of pedodontics.

DISCUSSION-

The Ectodermal Dysplasia are a diverse set of inherited illnesses that affect one in 100,000 births and are brought on by primary developmental abnormalities in two or more genes generated from embryonic ectoderm tissues.

The aberrant development of cutaneous and/or oral embryonal ectoderm (i.e., hair, nails, teeth, and exocrine glands) is the cause of ectodermal dysplasia. Anomalies of the mesoderm can also be seen in some types. Characteristic features include the following:

1.Hair defects: A decrease in the number of hair follicles as well as structural abnormalities in the hair shaft may be present. Anomalies in the creation of the hair bulb can cause structural abnormalities in the hair shaft, such as longitudinal grooves, twisting of the hair shaft, and cuticle ruffling. Small, bifid, or deformed hair bulbs are possible.[8]

2. Exocrine defects: In patients with hypohidrotic ectodermal dysplasia, exocrine sweat glands may be missing or scant and primitive.[9]

3.Various other secretory gland defects- Hypoplasia of the lacrimal, sebaceous, and salivary glands. Mucous glands in the bronchi, oesophagus, and duodenum as well as in the upper respiratory tract may not be present in some people.

4. Dental problems include abnormal tooth morphogenesis, tooth absence, and enamel flaws.[10]

5. Nail dystrophy: Improper nail plate development can lead to brittle, thin, ridged, or noticeably malformed nails.

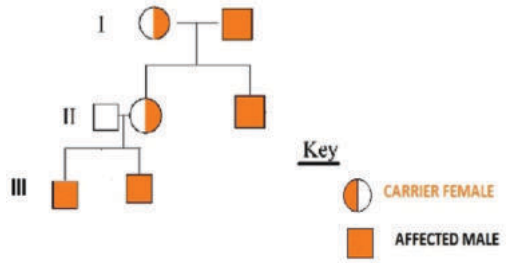
Even if there are certain Ectodermal Dysplasia syndromes with no known genetic causes, the proportion of Ectodermal Dysplasia syndromes with a clear genetic cause is growing. In 2009, 62 Ectodermal Dysplasias were connected to 64 genes and three chromosomal locations.[11]

Making proteins (ectodysplasin A) that collaborate throughout embryonic development is guided by the EDA, EDAR, and EDARADD genes.

The relationship between the ectoderm and the mesoderm, two cell layers, depends on a signalling system that ectodysplasin A is a part of these cell layers create the groundwork for many of the body organs and tissues in the early embryo. Ectoderm-mesoderm interactions are crucial for the development of a number of ectoderm-derived structures, such as the skin, hair, nails, teeth, and sweat glands. Defective ectodysplasin A is the result of mutations in the EDA, EDAR, or EDARADD gene a structure that prevents the ectoderm and mesoderm from interacting normally, impeding the development of teeth, sweat glands, and hair normally. The defining characteristics of hypohidrotic ectodermal dysplasia result from the incorrect development of these ectodermal structures.[12]

Family pedigree-

As it is an autosomal recessive disorder and there was familial history of the disease in his elder brother, maternal uncle and maternal grandfather, so the patient was considered to be third generation sufferers.



CONCLUSION –

Ectodermal Dysplasias are uncommon genetic illnesses with a variety of overlapping characteristics, making it challenging to categorise them. The clinical side effects of Ectodermal dysplasia result in serious social issues for those who experience them. It interferes with the patients' natural bodily processes as well as oral processes. Quick diagnosis and prosthetic rehabilitation using a multidisciplinary approach are essential to the therapy of Ectodermal dysplasia.

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