CASE REPORT

Hypohidrotic Ectodermal Dysplasia In Two Siblings: Case Report And Review Of Literature

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ABSTRACT

Introduction: Ectodermal dysplasia (ED) represents a heterogeneous group of disorders characterized by developmental dystrophies of ectodermal structures. These disorders are congenital, diffuse, non-progressive and incurable in nature. It is characterized by the triad of signs comprising sparse hair (atrichosis or hypotrichosis), absent or reduced sweating (hypohidrosis) and abnormal or missing teeth (anodontia or hypodontia). The lack of teeth is seen along with truncation or cone shaped formation of the teeth present.

Case Report: We present a report of Hypohidrotic Ectodermal dysplasia in 2 brothers in a family, showing most of the characteristic features of the disorder, which is a relatively uncommon occurrence.

Conclusion: A facial appearance of frontal bossing, saddle nose with deficient naso-maxillary complex and protrusion of mandible are of major concern. Since there is no definitive treatment for the condition, the affected individuals with dental defects can be managed by subjecting to early dental evaluation and intervention with dentures. Thus, prosthetic rehabilitation helps in improving the function, esthetics and further quality of life.

Keywords: Ectodermal dysplasia, Hypohidrotic, Hypodontia, Familial.

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INTRODUCTION

Ectodermal dysplasias (ED) are a group of X-linked inherited disorders characterized by dysplasia of tissues of ectodermal origin with prevalence of approximately 1:1,00,000 live births. Aberrant development of ectodermal derivatives in early embryonic life results in Ectodermal dysplasia. It is mapped by genetic defects in ectodysplasin in signal transduction pathways located at the proximal area of the long arm of band Xq12-q13.1. The clinical features showing the most common syndromes are hypohidrotic (anhidrotic) ED and hidrotic ED. Hypohidrotic ectodermal dysplasia (Christ-Siemens-Touraine syndrome) characterized by the classical triad of hypodontia, hypotrichosis, and hypohidrosis including dental manifestations like conical or peg shaped teeth, hypodontia or complete anodontia, and delayed eruption of permanent teeth. X-linked recessive is the usually seen trait of inheritance. Hidrotic ectodermal dysplasia affects only the hair and nail while the sweat glands remain unaffected. Autosomal dominant is the usually seen trait of inheritance.[2] 117 varieties of ectodermal dysplasia with various combinations of abnormal ectodermally derived structures were described by Freire-Maia and Pinheiro. 150 distinct disorders have been described till date.[3] The term anhidrotic was replaced with hypohydrotic by Felsher in 1944 as these are persons who are not devoid of sweat

glands.[2] Dry scaly skin and unexplained pyrexia and heat intolerance occur due to anhidrosis due to their diminished ability to sweat. Here we report 2 cases of Hypohydrotic Ectodermal dysplasia observed in 2 siblings.

CASE REPORT

A 26 year old male reported to the department with the chief complaint of missing teeth since childhood. Extraoral examination revealed concave facial profile with deficient naso-maxillary complex. Forehead showed frontal bossing, prominent supraorbital ridges with depressed nasal bridge and sunken cheeks. Scalp hair and eyelashes were thin, lustreless and sparse. The periorbital and perioral skin showed wrinkling with hyperpigmentation. The mandible is found to be protruding with dry, cracked and everted lips. The lack of teeth formed a reduced vertical facial height. The overlying skin surface was dry, scaly and thick. Nails and palmoplantar surfaces were found to be normal. Patient also reported decreased sweating with mild intolerance to heat. Intraoral examination revealed oligodontia with attrited maxillary right and left first molars and mandibular right first molars the only teeth present. Macroglossia and ankyloglossia was seen. Inflamed oral mucosa with reduced salivation was also observed(Fig.1). Medical and dental history was non-contributory. Complete radiographic examination revealed the edentulous maxillary and mandibular ridges with maxillary right and left first molars and mandibular right first molar as the only teeth present. Cephalometric analysis showed prominent supraorbital ridge with frontal bossing, absence of nasal bridge, deficient naso-maxillary complex, concave facial profile with deficient midface and protruding mandible. Deficient nasal prominence and everted lips was also evident, thus confirming all the clinical findings. Family history revealed the presence of similar features in his younger sibling. On examination of his sibling, a concave facial profile with deficient naso-maxillary complex, frontal bossing of forehead and prominent supraorbital ridges with depressed nasal bridge were observed. Scalp hair, eyebrows and eyelashes were thin, lustreless and sparse. The periorbital and perioral skin showed wrinkling with hyperpigmentation. Dry, cracked and everted lips were seen. The lack of teeth formed a reduced vertical facial height. The overlying skin surface was dry, scaly and thick. Nails and palmoplantar surfaces were found to be normal. Intra oral examination revealed maxillary right and left first and second molars and mandibular right and left second molars witha conically shaped maxillary left central incisor were the only teeth found to be present. Radiographic investigations further confirmed the clinical findings(Fig.2). Considering the characteristic features we arrived at a final diagnosis of Hypohydrotic Ectodermal dysplasia. Patient was scheduled for a surgical reconstruction of maxillary and mandibular arch with a full mouth prosthetic rehabilitation.

DISCUSSION

A well-functioning dentition is essential for the survival of many animals. Although not usually life threatening to humans, dental anomalies are challenging problems in clinical dentistry. Oligodontia refers to the lack of several, usually more than six teeth and is usually associated with other congenital defects such as ectodermal dysplasias. Ectodermal dysplasias comprise of a heterogenous group of hereditary disorders, which mainly involves structure of ectodermal origin including teeth, hair and sweat glands. Palms and soles are hyperkeratotic with presence of pseudorhagade around the eyes.[1] The facial features characteristic of ED include frontal bossing, prominent supraorbital ridges with depressed nasal bridge. Prominent and obliquely set ears with depressed midface are also seen. The lower third of the face appears small due to lack of alveolar bone development, and protuberant lips. These characteristic features were seen in our cases too. As stated by Besserman-Neilsen, xerostomia and the drying and cracking of the protuberant lips in these conditions, is due to hypoplasia of the salivary glands including the intraoral accessory glands.[1] A mortality rate of up to 30% is found in infancy or early childhood because of intermittent hyperpyrexia due to decreased sweating. The morbidity and mortality rate depends upon the absence or presence of eccrine and mucous

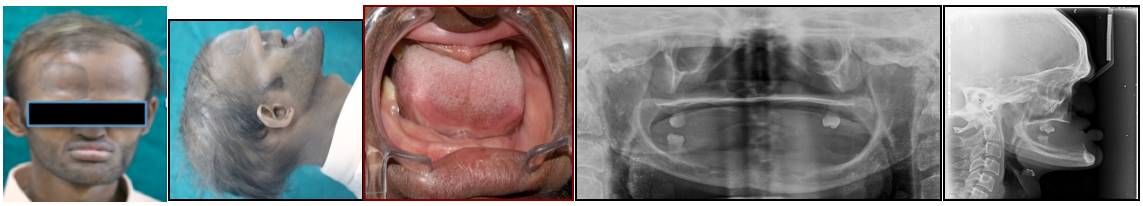

Figure-1: Clinicaland radiographic images of patient

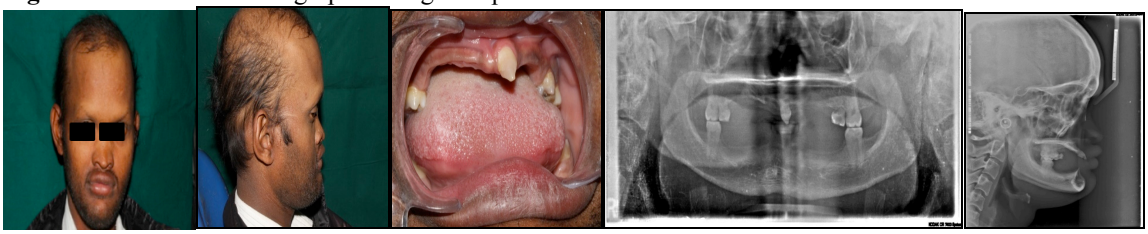

Figure-2: Clinical and radiographic images of patient's sibling

glands.[5] Freire-Maia [1971, 1977] defined Ectodermal dysplasia as congenital disorders characterized by alterations of two or more ectodermally derived structures, at least involving one amongst hair, teeth, nails, or sweat glands. It was first described by Thurnam in 1848 and was coined by Weech in 1929.[1] These are classified as group A and group B disorders as follows: group A disorders, which are manifested by defects in at least two of the four classic ectodermal structures (1-2-3-4: hair-teeth-nails-sweat glands), with or without other defects. Group B disorders, which are manifested by a defect in one classic ectodermal structure in combination with a defect in another ectodermal structure (5:i.e., ears, lips, dermatoglyphics)[2] It is a condition associated with deformation of one or more ectodermal derivatives which are typically inherited as a cross-linked recessive trait so that the frequency and severity of the condition is more pronounced in males than in females. Most common mode of inheritance of ED is X-linked. But both recessive and dominant forms do exist. Autosomal recessive disorders are phenotypically indistinguishable from the X-linked forms.The molecular pathogenesis of hypohidrotic ectodermal dysplasia (HED) is poorly understood. Mutations in gene EDA causes the X-linked recessive hypohidrotic ED, or anhidrotic ectodermal dysplasia and immunodeficiency (EDA-ID) which encodes the ectodysplasin protein. This in turn activates the NF-kappaB and essential modulator (NEMO), resulting in conical teeth, sparse hair, anhidrosis or hypohidrosis, and recurrent bacterial infections.[6] The EDA1 gene has been mapped to Xq12-q13.1 and encodes ectodysplasin A, a transmembrane protein. Thus it affects the development of keratinocytes and cause aberrations in the hair, sebaceous glands, eccrine and apocrine glands, nails and teeth. For the formation of structures originating from the ectoderm which include the skin, hair, teeth, sweat glands, interaction between the Ectoderm-mesoderm is essential. This interaction is prevented by the mutation in the EDA, EDAR or EDARADD gene which impairs the normal development of these structures leading to characteristic features of HED[7]. In the present cases the edentulous alveolar ridges was resorbed and thin giving knife-edge appearance with reduced ridge height as compared to the alveolus surrounding the present teeth, which also supports the theory that alveolar process fails to develop in edentulous areas of the ridge. The management of the case varies depending upon the severity. Identification of the way of transmission is of utmost importance to give reliable genetic counselling to the family and to address molecular studies. The key to clarify intrafamilial genetic transmissions only by complete examination of relatives of patients with HED and identification of carriers of partial forms of the disorder in their families.[8]

MANAGEMENT

The most commonly recommended treatment is early detection and placement of partial or full dentures from the age of two or three year

onwards. Though removable complete/partial dentures still remains the main stay of treatment in many cases, FPDs and implant-supported prostheses are to be considered when viable.[9]

CONCLUSION

Ectodermal dysplasia is a rare genetic disorder with involvement of the ectodermally derived tissues including teeth, skin and hair. A thorough and careful examination will lead to an accurate diagnosis which is of utmost importance. Restoration of normal function and esthetics should be the main concern in these patients. Since there is no definitive treatment for the condition focus shall be on increasing the overall quality of life in these patients through esthetic and functional rehabilitation.

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