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Case Report

Ectodermal dysplasia: a rare entity featuring complete anodontia: Case report and review of literature with a note on genetics

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Abstract
Ectodermal dysplasias are a large, multifarious group of inherited disorders, characterized by a flaw in the tissues derived from the embryonic ectoderm. The incidence of ectodermal dysplasia is rare. Various alterations in the genes coding for proteins like EDA, EDAR, EDARADD are the causes for the manifestations of ectodermal dysplasia. Oligodontia or hypodontia of the deciduous and/ or permanent dentition is the most common intra-oral finding. Ectodermal dysplasia is not only physically devastating to the patients but also emotionally demoralizing. It is essential that they be treated at an early age to improve their quality of life. The following case report aims at describing the condition in a patient with complete anodontia and discusses the genetic component of the disorder. Oral rehabilitation was achieved by using removable prosthesis.

Keywords: Ectodermal Dysplasia; EDA; EDAR; EDARADD; Anodontia.

INTRODUCTION

Thurman, first described ectodermal dysplasia, as a hereditary condition occurring as a consequence of disturbances in the ectoderm of the developing embryo. Ectodermal dysplasia is a complex group of disorders comprising of more than 170 different clinical conditions. The prevalence in males is estimated at 1 in 100,000 births. Onchodysplasia, hypotrichosis and palmo-plantar hyperkeratosis accompanied by hypohidrosis with hypodontia or oligodontia of the primary and/or permanent dentition, are the usual identifying signs (1,2).

This case report presents a patient with ectodermal dysplasia with complete absence of teeth followed by a review aimed at informing the reader on the genes that are mis-functioning behind the scenes.

CASE REPORT

A 19-year old male patient reported to the Department Of Oral Medicine And Radiology, with chief complaint of missing teeth from childhood. The patient did not give any history of extractions. The patient was the only child and there was no positive family history for the same.

During extra-oral examination, the patient exhibited the typical features of anhidrotic ectodermal dysplasia. The bridge of the nose seemed to lack its normal prominence. Lips were protuberant and everted.

On closure of the mouth the thickened lips pouted and gave the patient an aged appearance. Hair on the scalp and body was scanty and eyebrows were absent. Skin was soft and dry with no apparent sweating seen inspite of it being peak summer. Increased pigmentation in the peri-oral region was noticed. The toe and fingernails were present and normal (Figure 1). The remainder of the physical examination revealed no abnormalities.

Intraoral examination revealed the complete absence of teeth, thin alveolar ridges, reduced vertical bone height, and loss of sulcus depth in the posterior regions of maxillary and mandibular jaws; complete anodontia was also confirmed by panoramic radiography (Figure 2).

In order to improve appearance, mastication, and speech, removable complete maxillary and mandibular dentures were determined to be the best treatment choice (Figure 3).

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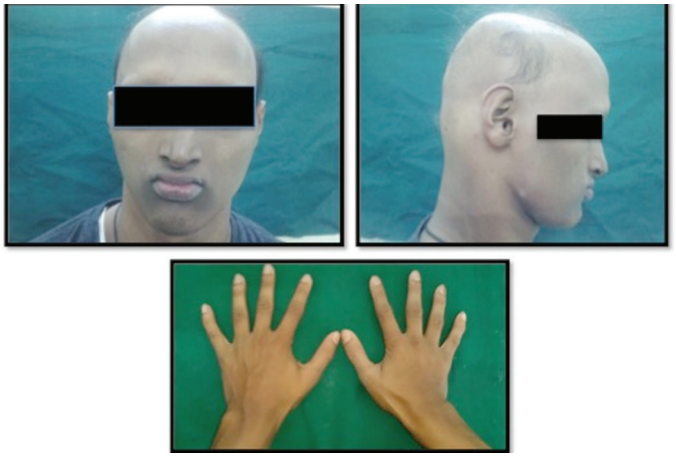

Figure 1. Extra Oral Photo Showing Scanty Hair, Absent Eyebrows, Protuberant Lips and Unaffected Hands and Nails

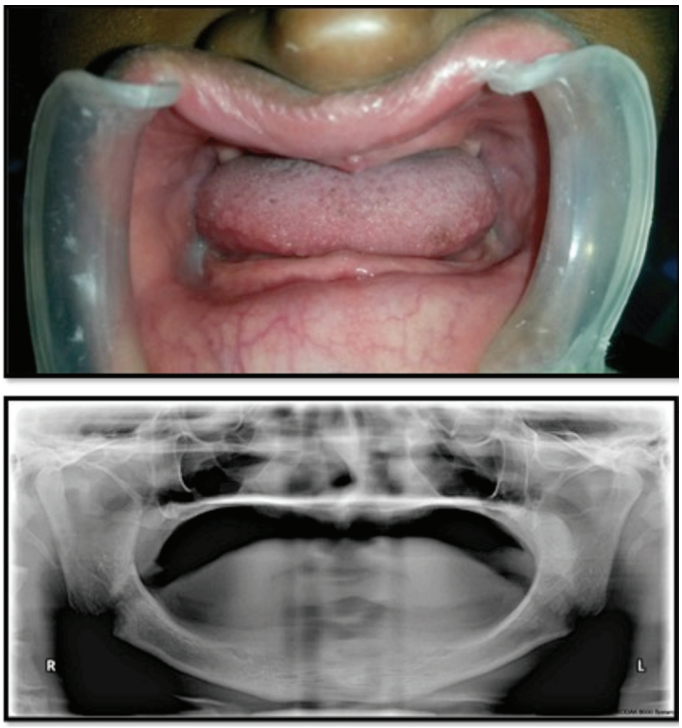

Figure 2. Poorly Formed Alveolar Ridges And OPG Showing Completely Edentulous Arches

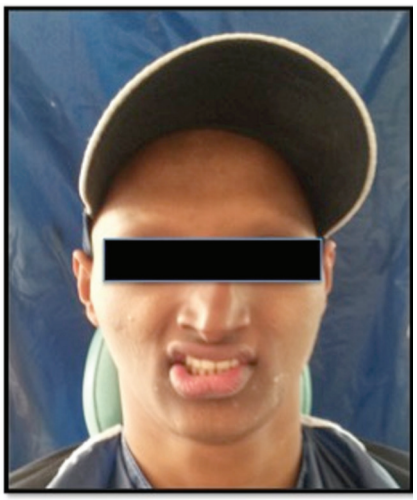

Figure 3. Post Complete Denture Insertion

DISCUSSION

Ectodermal dysplasias are a large, multifarious group of inherited disorders, characterized by a flaw in the tissues derived from embryonic ectoderm. Based on the ability to sweat, they are termed hidrotic ectodermal dysplasia or Clouston syndrome if the functioning of sweat glands are normal and hypohidrotic/anhidrotic ectodermal dysplasia or Christ-Siemens-Touraine syndrome where the ability to sweat is either impaired or absent; as was seen in the case reported here. Various alterations in the genes coding for proteins like EDA, EDAR, EDARADD are the causes for the manifestation of ectodermal dysplasia like hypoplasia of sweat glands, tooth buds and hair follicles. These hypoplastic defects are manifested as hypohidrosis, hypodontia/ anodontia and alopecia (3,4).

Positioned on the long (q) arm of the X chromosome at position 13.1, the EDA gene is responsible for the coding of the protein called Ectodysplasin A (EDA), which plays a vital role in the development of ectodermal tissues. EDA interacts with a protein produced by EDAR gene called the ectodysplasin A receptor. When these two proteins are linked, they activate a series of chemical signals. This signaling cascade controls the formation, development and maturation of ectodermal structures namely hair follicles, sweat glands, and teeth.

Either Mutations in the EDA gene, or deletions have been identified in people with hypohidrotic ectodermal dysplasia. These changes cause production of abnormal EDA, which cannot carry out the required tasks, without which ectodermal structures do not form properly, leading to the characteristic features of hypohidrotic ectodermal dysplasia. Bi-directional full gene sequencing and quantitative polymerase chain reaction (PCR) can be used to test these deletions (5).

Coding for the protein ectodysplasin A receptor, the EDAR gene is positioned on Chromosome 2 on the long (q) arm at position 13. EDAR is required for the attachment of EDA to the cell surface. Deletions and mutations in the EDAR gene have been recognized, which prevent the proper attachment of EDA to the surface, thus preventing proper ectoderm–mesoderm interactions, resulting in the improper or nil signaling resulting in the improper formation of structures responsible for the occurrence hypohidrotic ectodermal dysplasia. These changes or mutations can be identified using full sequencing of the EDAR gene (5).

Ectodysplasia A Receptor Associated Death Domain (EDARADD), which is also known as Ectodysplasin A Receptor Associated Adapter Protein, is located on the long (q) arm of Chromosome 1, between positions 42.3 and 43. The EDARADD gene is responsible for the production of EDAR-associated death domain protein. This protein interacts with ectodysplasin A receptor and acts as an adapter initiating chemical signals and thus causes cell division, growth, and maturation. All these interactions

take place at a region called the death domain that is present in both proteins (6-8).

In its true sense ectodermal dysplasia must include only those structures of ectodermal origin or derivation. But as some complex structures are derived from more than one germ layer, varied presentations of the condition are observed. Various classifications have been put forth. Centered on the classical signs related with EDs, Freire-Maia and Pinheiro based their classification on presence of trichodysplasia, dental defects, onychodysplasia and dyshidrosis dividing into 2 Groups. Group A featuring those disorders with manifestations of any 2 of the classical structures like hair, teeth, nails and sweat glands. Group B includes disorders with manifestations of any one of the classical signs associated with another ectodermal sign. The case reported here fulfilled 3 criteria under Group A and hence can be classified under it (9-11).

In 2001, Priolo and Laganà proposed a clinico-functional classification, dividing ectodermal dysplasias into 2 main functional groups: Group 1 with defects in the epithelial-mesenchymal interaction and Group 2 with defects in cytoskeleton maintenance and cell stability (12).

The main concern from the dental point of view is hypodontia, which is almost always present in ectodermal dysplasia and in severe cases, complete anodontia is seen. The complete absence of primary dentition and permanent dentition is a rare phenomenon. In this case, the patient's history and clinical and radiographic examination revealed the absence of primary and permanent dentition. Acikgoz et al., and Vieira et al., have put forth cases with complete absence of primary dentition where as Pirgon et al., reported complete absence of teeth in both primary and secondary dentitions. Primary dentition must be present for the development of the permanent successors (13-15).

Complete oral rehabilitation is required, especially prosthodontic rehabilitation, if when initiated at an early age not only enables in development of normal appearance, phonation, mastication and TMJ development, but also improves the confidence of the child and enables him/her to be socially acceptable. Although implants can be placed, they have low success rates. In the case presented here, the patient did not have bone quantity that was favorable for implant placement and thus complete denture was the treatment of choice (16).

CONCLUSION

Ectodermal dysplasias are a large, group of inherited disorders, characterized by a flaw in the tissues derived from embryonic ectoderm. Various alterations in genes coding for proteins like EDA, EDAR, EDARADD are the causes for the manifestation of ectodermal dysplasia. Complete oral rehabilitation is required at an early age, which needs to be followed up regularly.

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