Hereditary Ectodermal Dysplasia - A Case Report

Corresponding Author:
Dr. Subhas G Babu,
Professor and Head, AB Shetty Institute of Dental Sciences, Nitte University, 575018 - India

Submitting Author:
Dr. Shishir Shetty,
Assistant Professor, Department of Oral Medicine and Radiology, AB Shetty Memorial Institute of Dental Sciences - India

Article ID: WMC001711
Article Type: Case Report
Article URL: http://www.webmedcentral.com/article_view/1711
Subject Categories: DENTISTRY
Keywords: Hypodontia, Anodontia, Anhidrosis.


Source(s) of Funding:
Nil

Competing Interests:
Nil
Hereditary Ectodermal Dysplasia - A Case Report

Author(s): Babu S G, Castelino R L, Shetty S R, Rao K A

Abstract

Ectodermal dysplasia is an extremely rare genetic disorder characterized by faulty development of ectodermal structures. The tissues in which the primary defects occur are the skin, hair, nails, exocrine glands, and teeth. It usually affects the males and females are the carriers. The clinical features include sparse hair, abnormal or missing teeth, and inability to sweat due to lack of sweat glands. One such case report of ectodermal dysplasia is presented here.

Introduction

Ectodermal dysplasias are a heterogeneous group of disorders characterized by developmental dystrophies of ectodermal structures. It is an X linked recessive disorder. It is characterized by the triad of signs which comprises of sparse hair (atrichosis or hypotrichosis), abnormal or missing teeth (anodontia or hypodontia) and inability to sweat due to lack of sweat glands (anhidrosis or hypohidrosis). The etiology of ectodermal dysplasia appears to be genetic in nature [1].

Case Report(s)

A 32-year-old male patient reported to our institution with the complaint of multiple missing teeth since childhood. The patient also gave a history of delay in the eruption of deciduous and permanent teeth, intolerance to heat and reportedly less sweat production. There was no history of consanguineous marriage between the parents. On extra oral examination, the patient had dry skin with periocular area being hyperpigmented and wrinkled with sparse hair on the body and scalp. Hair present were fine in texture & lighter in color. Prominent supraorbital ridges, small and outwardly placed ears and flattened nasal bridge was also present. Both upper and lower eyelids showed sparse eyelashes [Figure 1 and 2]. The skin was warm and dry.

Intra oral examination revealed multiple missing teeth in the maxillary and mandibular arches [Figure 3]. Salivary Flow rate was estimated at 0.1 ml in 5 min. The similar findings were present in the maternal uncle, as revealed by the patient. Based on these findings a diagnosis of hereditary ectodermal dysplasia was made. An orthopantomograph was made which revealed multiple missing teeth [Figure 4]. The teeth with bad prognosis were extracted and the replacement of missing teeth was done using a fixed prosthesis in the maxillary and mandibular arches [Figure 5].

Discussion

The Ectodermal dysplasias are a group of inherited disorder that share in common developmental defects involving at least two of the major structure classically hold to derive from the embryogenic ectoderms hair, teeth, nails, sweat glands [2]. The incidence of ectodermal dysplasia in males is estimated at 1 in 100,000 births [3]. Thurman first described this entity in 1848[4]. More than 192 distinct disorders have been described till date. Freire Maia and Pinheiro published an exhaustive review and classification system for these disorders using a numeric system of 1 (hair), 2 (teeth), 3 (nail), 4 (sweat glands) for characterization [5]. The complete syndrome does not occur in females but females may show dental defects, sparse hair, reduced sweating and dermatoglyphic abnormalities [2]. The major concern seen in these patients is the lack of teeth and the special appearance, as seen in our case [1]. The most characteristic findings is the reduced number and abnormal shape of teeth. The delay in eruption of teeth is often the first step in the diagnosis. The men have an easily recognizable facies, also referred to as an old man facies. Some infants have a premature look with scaling of the skin. This can also form a clue to the diagnosis [1]. The extra oral features seen in this disorder are frontal bossing with the forehead appearing square in shape, prominent supraorbital ridge, depressed nasal bridge (saddle nose) as seen in our case. The other features include midface hypoplasia, pointed chin and protruberent and everted lips, however these features were not seen in our case [6]. Abnormalities of skin, nails and teeth are also noted, which was also seen in our case [1]. Prenatal diagnosis of ectodermal dysplasia has occasionally been reported which has been diagnosed by foetal skin biopsy, obtained by fetoscopy by 20 weeks gestation [7]. The characteristic facies is pathognomonic but may not be recognized in infancy. In partial forms, the pointed...
conical teeth provide the most valuable indication and should suggest the need for sweat test and a skin biopsy [2]. Both autosomal dominant and recessive mode of inheritance has been described. another variety of hydrotic ectodermal dysplasia called as Clouston syndrome is inherited in an autosomal dominant manner, was described by Clouston in 1929 and Lowrey et al in 1966, which is found in Canadian families of French descent. This syndrome usually spares the sweat glands [8]. The treatment usually comprises of complete restoration of function and aesthetics normalise the vertical dimension and provide adequate support to the facial soft tissues.

Conclusion

Ectodermal dysplasia is a rare genetic disorder with involvement of various tissues in the body. A careful and a thorough examination will lead to an accurate diagnosis. Restoration of normal function should be the main concern in these patients.

References

Illustrations

Illustration 1

Extra oral photograph showing sparse hair on the face and scalp.

Illustration 2

Sparse body hair
Illustration 3

Intra oral photograph showing multiple missing teeth

Illustration 4

Orthopantomograph showing multiple missing teeth
Illustration 5

Post treatment photograph of the patient
Disclaimer

This article has been downloaded from WebmedCentral. With our unique author driven post publication peer review, contents posted on this web portal do not undergo any prepublication peer or editorial review. It is completely the responsibility of the authors to ensure not only scientific and ethical standards of the manuscript but also its grammatical accuracy. Authors must ensure that they obtain all the necessary permissions before submitting any information that requires obtaining a consent or approval from a third party. Authors should also ensure not to submit any information which they do not have the copyright of or of which they have transferred the copyrights to a third party.

Contents on WebmedCentral are purely for biomedical researchers and scientists. They are not meant to cater to the needs of an individual patient. The web portal or any content(s) therein is neither designed to support, nor replace, the relationship that exists between a patient/site visitor and his/her physician. Your use of the WebmedCentral site and its contents is entirely at your own risk. We do not take any responsibility for any harm that you may suffer or inflict on a third person by following the contents of this website.