ISSN: 0975-833X

Available online at http://www.journalcra.com

International Journal of Current Research
Vol. 13, Issue, 02, pp.16384-16386, February, 2021

DOI: https://doi.org/10.24941/ijcr.40843.02.2021

INTERNATIONAL JOURNAL OF CURRENT RESEARCH

RESEARCH ARTICLE

ECTODERMAL DYSPLASIA- A CASE REPORT WITH LITERATURE REVIEW

[1,*]Dr. Sheetal Sharma and [2]Dr. Sapna Saini

[1]M.D.S in Oral Medicine & Radiology, Oral Medicine & Radiology, AIIMS, New Delhi, India
[2]B.D.S (Jodhpur dental college, Jodhpur), INTERN, Rajasthan dental college, Jaipur

ARTICLE INFO

Article History:
Received 05th November, 2020
Received in revised form
20th December, 2020
Accepted 19th January, 2021
Published online 28th February, 2021

Key Words:

Anodontia, Hypohidrotic Ectodermal Dysplasia, Multidisciplinary Approach, Prosthetic Rehabilitation,

ABSTRACT

Ectodermal dysplasia (ED) is a hereditary disorder involving two or more ectodermal structures which include the skin, hair, nails, teeth, and sweat glands. They are caused by the mutations of several genes. The two most common forms of the disease are hypohidrotic/ anhidrotic ED and hidrotic ED. We present a case of a 12-year-old boy with hypohidrotic ED. The oral rehabilitation of such cases is often difficult; particularly in pediatric patients. A multidisciplinary approach by a team consisting of physicians from several clinical modalities is required to provide comprehensive medical care to children suffering from ED.

Citation: ***Dr. Sheetal Sharma and Dr. Sapna Saini. 2021.*** "Ectodermal Dysplasia- A Case Report with Literature Review.", *International Journal of Current Research*, 13, (02), 16384-16386.

INTRODUCTION

Ectodermal dysplasia (ED) is a rare heterogeneous group of inherited disorders that involve primary defects in the development of two or more tissues derived from the ectoderm. The most commonly involved tissues are the skin, hair, nails, eccrine glands, and teeth. (1-2) The incidence of ectodermal dysplasia is 1 in 100,000 births. (1, 3) 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. (1-5) The common clinical features include dental abnormalities such as conical or pegged teeth, hypodontia/ complete anodontia, and delayed eruption of permanent teeth, hypotrichosis, abnormal nails, and reduced ability to sweat. (2) The disorders are congenital, diffuse, and nonprogressive. (3-4) other common signs are short stature, eye abnormalities, decreased tearing, and photophobia. (3-4) As multiple organs are involved, the treating physician team faces several therapeutic challenges. This case study with literature review aims to examine the oral manifestations observed in ED and make dental practitioners aware of the possible oral and other associated complications of ED, as well as the available treatment modalities.

***Corresponding author: Dr. Sheetal Sharma,**
M.D.S in Oral Medicine & Radiology, Oral Medicine & Radiology, AIIMS, New Delhi, India.

CASE REPORT

A 12-year-old year visited the department of oral medicine and radiology by his parents due to the absence of teeth. The parents revealed that the child had intermittent episodes of fever. No significant family history was present. On examination, the patient's vitals and systemic examination were normal. The hairs on the scalp were sparse and the eyebrows were absent. Both the upper and lower eyelids showed sparse eyelashes. Periorbital and perioral wrinkling and pigmentation were also present. The nasal bridge was depressed; consistent with a saddle-nose. (Figure 1, 2) His skin was dry, warm, and sensitive. Intraoral examination revealed the child had mandibular and maxillary hypodontia and reduced vertical bone height. (Figure 3) OPG shows multiple congenitally missing teeth along with the reduced height of the maxillary and mandibular body. (Figure 4) The patient's complete blood count, comprehensive metabolic panel, and urine analysis reports were normal. Based on the history, clinical features, and examination, the child was diagnosed as a case of hypohidrotic ED. Anticipatory guidance was given to his parents about the disease. Apart from medical management, he was referred to the pediatric department for dental reconstruction.

DISCUSSION

Ectodermal dysplasia was first described by Thurman, as a hereditary disorder occurring as a consequence of disturbances in the ectoderm of the developing embryo (1).

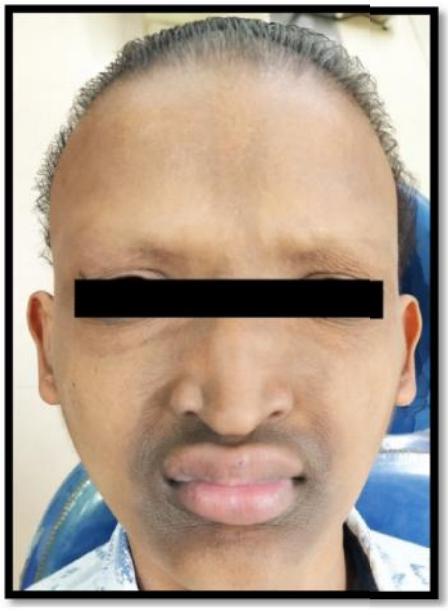

Fig 1. Photomicrograph showing fine, sparse scalp hair, scanty eyebrow and eyelashes, frontal bossing or depressed nasal bridge, and full and everted lips, with midface hypoplasia

Fig 2. Photomicrograph showing fine, sparse scalp hair

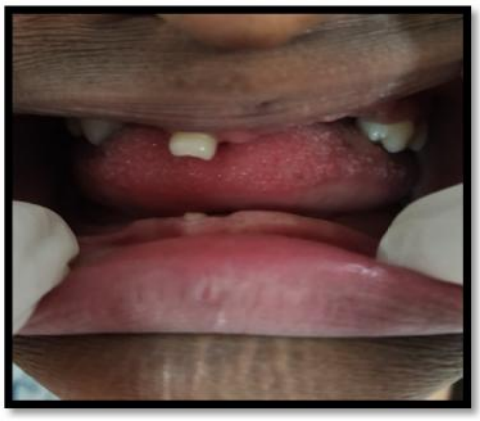

Fig 3. Photograph showing intraorally partial anodontia with upper and lower arch

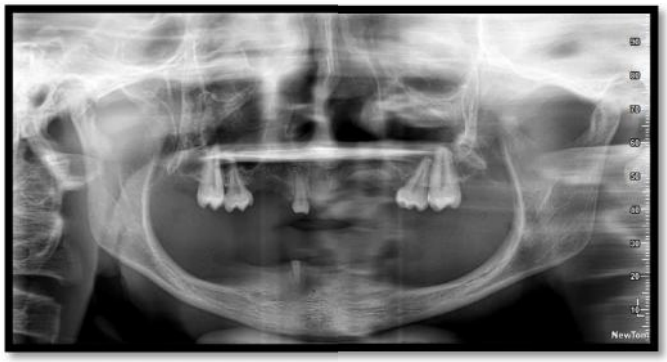

Fig 4. Panoramic radiograph confirming partial anodontia

Freire-Maia and Pinheiro proposed the first classification system of ectodermal dysplasia in 1982. 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 onycholysis-plasia, (4) eccrine gland dysfunction or dyshidrosis (4). The literature review describes more than 190 subtypes of ED, which can be classified based on the clinical features or the type of genetic mutation, or the molecular pathway involved. However, the two most common subtypes of ED are hypohidrotic/anhidrotic ED and hidrotic ED. (1-3) The genetic studies reveals that mutations in the ectodysplasin-A and ectodysplasin-A receptor genes are responsible for X-linked and autosomal hypohidrotic ectodermal dysplasia. (1) X-linked recessive hypohidrotic ectodermal dysplasia is caused by a mutation in EDA, which encodes for the ectodysplastin protein, a soluble ligand that activates the NF-kappa B and JNK- fos/c-jun signaling pathways. Hidrotic ectodermal dysplasia, which is an autosomal dominant disorder, is caused by a mutation in GJB, which encodes for connexin 30, a component of the intercellular gap junction. Autosomal dominant and recessive hypohidrotic ectodermal dysplasia is caused by a mutation in the DL gene, which encodes for the EDA receptor. (4) Molecular studies have described that the above-mentioned genes are responsible for the formation of several substrates required for the activation of the tumor necrosis factor α-related signaling pathway, the WNT-signaling pathway, and the nuclear factor-κB pathway, involved in ectoderm-mesoderm interactions, differentiation of ectodermal appendages, and organogenesis during the initiation of embryonic development (4-5).

Clinically hypohidrotic/anhidrotic ED is characterized by hypotrichosis, hypo/adontia, dyshidrosis (abnormal sweating), and facial dimorphism. The typical facies are characterized by frontal bossing, sunken cheeks, a saddle nose, thick and everted lips, wrinkled and hyperpigmented skin around the eyes, and large, low-set ears. On the other hand, hidrotic ED is characterized by the triad of onychodysplasia, hypotrichosis, and palmoplantar hyperkeratosis. (1-4) The clinical presentation of our patient was consistent with the case of hypohidrotic ED. The prenatal identification of this condition can be established by DNA-based linkage analysis and genetic tests for detecting mutations in EDA/EDAR/EDAEADD. In the second trimester of pregnancy, sonography, and fetal skin biopsy are suitable diagnostic tests. (4). Numerous clinical and psychological aspects need to be addressed in patients with hypohidrotic ED, a multidisciplinary approach is a key (4). The patients with hypohidrotic ED usually have elevated body temperatures or hyperpyrexia. Hence, cooling techniques such

as cold sponging, using cooling vests and chilled emollients, placing the child in a tub containing cold water, using air-conditioned, and consuming cold water, also to refrain from indulging into intense physical activities, and exposure to high temperatures (2,6,7). Oral rehabilitation of patients with ectodermal dysplasia is necessary to improve sagittal and vertical skeletal relationships during craniofacial growth and development as well as esthetics, speech, and masticatory efficiency. (8) The dental referral is warranted in all patients of hypohidrotic ED. In childhood, dentures are the primary line of treatment for missing teeth. Regular follow-up is as per the child's growth and development. Thus implant-supported prosthesis may be less favorable and therefore, the use of implants in young children should be considered carefully.

Early prosthetic treatment is generally recommended from the age of 5 years. (8-11) In older individuals dentures, dental implants, and orthodontia are usually the preferred treatment options. When implant therapy is indicated, the main problem is insufficient bone; if bone atrophy progresses in these already alveolar deficient patients, implant placement may not be possible without bone grafting. The most common treatment plan is a removable prosthesis. Our case was treated with partial dentures. In addition to this psychological aspects must be taken into consideration as patients with hypohidrotic ED may suffer from low self-esteem, insecurity, and depression due to their unusual physical appearances and lack social acceptance. (2, 7, 8) In this situation, special attention and psychological counselling should be advised. The use of wigs in patients with severe alopecia may improve their cosmetic appearance.

The newer therapy via Intravenous injection of recombinant EDA-A1 to newborn dogs with X-linked hypohidrotic ED has been found to restore the growth of their teeth, skin structures, and mucous glands. Furthermore,intra-amniotic injections of recombinant EDA-A1 to pregnant mice partially improved the phenotype of the X-linked hypohidrotic ED newborn mice. Recombinant EDA-A1 at present is in Phase-II clinical trials and is being administered to newborn males with hypohidrotic ED to hopefully alleviate some of their symptoms. (4) 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. Hence, additional care must be provided to infants and young children. After 3 years of life, life expectancy is normal. (3, 4, 6)

Conclusion

The patients with ectodermal dysplasia can have considerable social problems due to its clinical manifestations.

A multidisciplinary approach by a team consisting of physicians from several clinical modalities is required to provide comprehensive medical care to children suffering from ED. Management of clinical manifestations associated with ectodermal dysplasia presents a unique challenge for prosthodontists and pedodontists but it's of great value to these patients from functional, psychological, and psychosocial standpoints. However, its long-term success depends on regular recall appointments and meticulous maintenance of oral and prosthetic hygiene.

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