ORIGINAL RESEARCH PAPER

Volume-7 | Issue-9 | September-2018 | PRINT ISSN No 2277 - 8179

INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

HYPOHYDROTIC ECTODERMAL DYSPLASIA: A CASE REPORT AND LITERATURE REVIEW

Dental Science

Muhamood Moothedath — Assistant Professor (Public Health Dentistry), College of Applied Health Sciences in Ar Rass, Qassim University, Al-Qassim, Kingdom of Saudi Arabia.

Muhaseena. M* — Lecturer, Department of Biomedical Dental Sciences, College of Dentistry, Imam Abdulrahman Bin Faisal University, P.O. Box. 2435, Dammam, Kingdom of Saudi Arabia. *Corresponding Author

Muzammil — Assistant Professor (Periodontics), College of Applied Health Sciences-Ar Rass, Qassim University, Al-Qassim, Kingdom of Saudi Arabia.

Akshata. G. Kamath — Reader, Department of Periodontics, Century International Institute of Dental Sciences and Research Centre, Poinachi, Kasaragod, Kerala, India.

ABSTRACT

Ectodermal dysplasia is a hereditary disorder characterized by developmental defects of the ectodermal derivatives. It is characterized by aplasia or hypoplasia of structures such as skin, hair, nails, teeth, nerve cells, sweat glands, parts of the eye and ear and other organs. The most prevalent form is hypohydrotic ectodermal dysplasia (HED), also known as Christ-Siemens-Touraine syndrome (CST). This case report describes the manifestations and the dental management of patients with hypohydrotic ectodermal dysplasia. Literature review on ectodermal dysplasia types are also highlighted.

KEYWORDS

Ectodermal Dysplasia, Hypohydrotic, Hypodontia, Hydrotic.

INTRODUCTION

Ectodermal dysplasias (EDs) coined by Weech in 1929 [1] represents a heterogeneous group of disorders characterized by developmental dystrophies of more than one ectodermal structure. These disorders are usually congenital, diffuse and non-progressive in nature.[2] It was Thurnam in 1848 first described it as an ectodermal disturbance of the developing embryo.[3] Prevalence of this disorder is approximately estimated as 1 in 1, 00,000 live birth [2] and can occur during the first trimester of pregnancy. Hypohydrotic Ectodermal Dysplasia (HED) also known as Christ-Siemens-Touraine syndrome is the most common phenotype and is typically inherited as an X-linked recessive trait with gene mapping to Xq12-q13.[4] Hence, men are more susceptible than women. The dentition is affected before the sixth week of embryonic life. Consequently, after eighth week, other ectodermal structures may be affected.[5] Ectodermal dysplasia exhibits a classic triad of symptoms: they are Hypohydrosis, Hypotrichosis and Hypodontia.[6]

In patients with HED, the skin is often dry, either with complete or partial absence of sweat glands (Hypohydrosis). As a result, they cannot sweat normally and may exhibit heat intolerance. Hair follicles and sebaceous glands are often defective or absent and the hair of the scalp and eyebrows tends to be fine and scanty (hypotrichosis). Oral manifestations include partial or complete absence of teeth (hypodontia or anodontia) affecting both primary and permanent dentition. If teeth are present, they are conical in shape and malformed. [7]Our case report aims to present the clinical features and the dental treatment modalities of HED along with review of literature.

CASE REPORT

A 20-year-old male patient reported to our dental clinic with a chief complaint of several missing teeth in the upper and lower jaw since birth. He was concerned about aesthetics, difficulty in speech and mastication. Detailed history revealed that the patient also complains of dry skin, decreased sweating and he was intolerable to withstand hot water and hot environment. He did not give any history of exfoliation or extraction of teeth, however he reported of delay in eruption of presently existing teeth. Medical history and family history were non-contributory. On general examination, the patient was co-operative and well oriented. Clinical examination revealed a fine and sparse scalp hair, eyebrows, and eyelashes. Scalp hair was light in colour. Other features observed were dry skin, depressed nasal bridge, frontal bossing, and sunken cheeks with prominent supraorbital ridges (Fig:1). On intraoral examination, partially edentulous maxillary arch was observed (Fig:2). Deciduous canines, first and second molars were present on either side of the maxillary arch. Canines were conical in shape. Lower arch revealed anodontia (Fig:3). Other oral findings included high arched palate, dryness of the mouth and macroglossia due to anodontia. Radiographic investigation was done and orthopantomogram (OPG) revealed multiple missing teeth with absence of impacted teeth (Fig:4). Only the existing six deciduous teeth in the maxillary arch were observed and the mandibular arch was totally edentulous. Therefore, based on the above clinical findings of hypohydrosis, hypotrichosis, hypodontia and radiographic findings we gave a diagnosis of Hypohydrotic Ectodermal Dysplasia. A mandibular complete denture and maxillary removable partial denture was fabricated and delivered (Fig:5).

Fig 1: Clinical photograph showing classical features of ectodermal dysplasia.

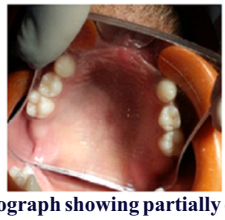

Fig 2: Clinical photograph showing partially edentulous maxillary arch.

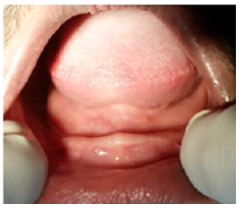

Fig 3: Clinical photograph showing anodontia of mandibular arch.

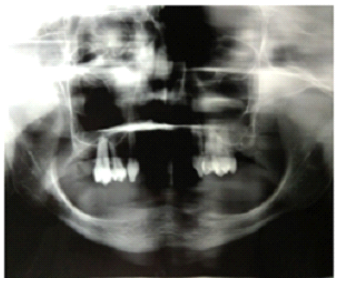

Fig 4: Orthopentamograph of the affected patient.

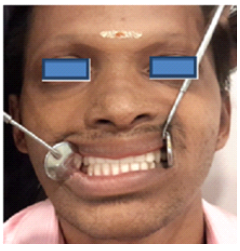

Fig 5: Clinical photograph with removable prosthesis.

DISCUSSION AND REVIEW OF LITERATURE

Ectodermal dysplasias represents a large group of hereditary conditions in which two or more ectodermal derivatives fail to develop. Freire-Maia described it as a developmental defect which at embryonic level affects the ectoderm and therefore the tissues and structures derived from it.[8] Two major groups are distinguished based on the sweat gland involvement: 1) Hypohydrotic or anhidrotic (Christ-Siemens-Touriane syndrome) in which sweat glands are either absent or significantly reduced in number; (2) Hydrotic (Clouston's syndrome) in which sweat glands are normal. 4 Dentition and hair are involved similarly in both the types but hereditary patterns of nails and sweat glands involvement are different. The numerical classification system was proposed by Freire Maia and Pinheiro for these disorders. A numeral was assigned for every involved ectodermal structure:[9]

ED1: Trichodysplasia (hair dysplasia)
ED2: Dental dysplasia
ED3: Onychodysplasia (nail dysplasia)
ED4: Dyshidrosis (sweat gland dysplasia)

Hypohydrotic ED (Christ-Siemens-Touriane syndrome) is more common and is characterized by a triad of signs comprising sparse hair (hypotrichosis), abnormal or missing teeth (hypodontia or anodontia), and an inability to sweat because of the lack of sweat glands (anhydrosis or hypohydrosis).10 It is inherited as an X-linked recessive, autosomal dominant, or autosomal recessive manner and is caused by mutation in the EDA, EDAR, and EDARADD gene, respectively. These regulate the EDA A protein, which cause signal interaction between ectoderm and mesoderm. Mutation in this gene results in defective EDA A and hence impairs normal development of hair, sweat gland, and teeth.11 In X-linked recessive mode of inheritance, only males are affected whereas an autosomal dominant inheritance female are also affected.12 According to Sidhu et al,13 the anhydrotic variant of ED may occur without any family history of this disease due to genetic mutations. In our case report, the proband was the only child of his parents. No similar case of ectodermal dysplasia has been identified among the family and relatives, which suggests that the propositus was probably due to new mutation or due to translocation of genes. In hydrotic ectodermal dysplasia (Clouston's syndrome), the mode of inheritance is most often autosomal dominant, and the gene identified is the GJB6, which encodes for connexin-30. BJ-6 has been mapped to the pericentromeric region of chromosome 13q. Mutations of the gene PVRL1, encoding a cell-to-cell adhesion molecule/herpes virus receptor, have been reported in those with cleft lip/palate ectodermal dysplasia.14 Severity is more pronounced in males than in females and thus females show only minor defects. The sweat glands are active and normally functioning unlike hypohydrotic ED. The nails of patients with hydrotic ED are often described as small and dysplastic and may be spoon shaped/concave15 whereas, there is no nail abnormality noted in hypohydrotic ED cases. In our case nail abnormality was not observed. The most characteristic dental manifestation is the reduced number (partial or complete anodontia) and abnormal shape of teeth.4 Delayed teeth eruption is often seen these patients. The present case also reports partially edentulous maxillary arch and complete anodontia of mandibular arch and pointed conical teeth. The affected individuals show similar facial appearance. The extraoral features seen in this disorder include frontal bossing with the forehead appearing square in shape, depressed nasal bridge (saddle nose) and prominent supra orbital ridge which were also noted in our case. The other features include midface hypoplasia, pointed chin and protuberant and everted lips.16 The distinguishing features of hypohydrotic and hydrotic ED are summarised and presented in table 1

Table 1. Distinguishing features of Hypohydrotic and Hydrotic types of Ectodermal Dysplasia.

	Hypohydrotic Ectodermal Dysplasia	Hydrotic Ectodermal Dysplasia
Mode of inheritance	Most often autosomal recessive	Most often autosomal dominant
Gene mutation	EDARADD gene	GJB6 gene
Syndrome	Christ-Siemens-Touriane syndrome	Clouston's syndrome
Prevalence	1 in 1,00,000 live births	1-9 in 100,000 live births
Scalp hair	Fine in texture, light coloured	Soft, dark coloured
Eyebrows	Absent	Mostly absent
Eyelashes/pubic/axillary hair	Scanty / variably affected	Scanty/absent
Nasal bridge	Underdeveloped	No visible flattening
Lips	Usually protruding	No visible abnormality
Nails	No visible abnormality	Dysplastic/spoon-shaped/ concave
Sweat glands	Reduced or absent	Active
Teeth	Anodontia or hypodontia/ Abnormal tooth morphology	Anodontia or hypodontia

MANAGEMENT

Definitive treatment is not yet established for ED patients and the management depends on the structures involved to improve the aesthetics and function. Sweat test and skin biopsies are reliable in obtaining an accurate diagnosis for patients who present with features of ED. Sweating response can be demonstrated by applying yellow starch-iodine powder on palmar or dorsal skin in a sweating room (sauna) for 10 minutes. Skin biopsy is done to demonstrate absence or hypoplasia of sweat glands.[17] Use of artificial tear and skin moisturizers are recommended for these patients and they should avoid excessive exposure to warm climates and heavy physical activities.[16] ED patients should be advised to use light clothing, a cool-water spray bottle and an air conditioned or cooler environment. Patients with severe alopecia may go for hair implant to improve their appearance and confidence. Oral rehabilitation of patient with ED is a multidisciplinary team approach with the consideration of growth and development.[18] The treatment usually comprises of complete restoration of function and aesthetics to normalise the vertical dimension and provide adequate support to the facial soft tissues. Removable prosthesis which includes complete dentures, partial dentures and over-dentures are the most frequently used treatment modalities for the dental management of children suffering from ED in growing age.[19] Due to anodontia, the alveolar bone of affected patients will be hypoplastic, which affects the development of jaw(s).[20] In younger patients the best treatment modality includes the use of removable prosthesis as they require repeated change of prosthesis because of the continuous growth of basal bone. Hence replacements should be done till the permanent dentition stage after which implant supported dentures should be included in the treatment plan.

CONCLUSION

Hereditary ectodermal dysplasias though rare in occurrence has no definite treatment but require symptomatic management depending upon the ectodermal structures affected. Dental professionals play a vital role in contributing to the overall appearance and wellbeing of the affected patients.

REFERENCES

1. Weech AA. Hereditary ectodermal dysplasia (congenital ectodermal defect): A report of two cases. Am J Dis Child. 1929; 37 (4):766-790.

2. Ammanagi R, Keluskar V, Bagewadi A. Ectodermal dysplasia: Report and analysis of eleven south Indian patients with review of literature. J Ind Acad Oral Med Rad. 2010; 22:93-8.
3. Murthy J V, Vaze R. Prosthetic Management of an Ectodermal Dysplasia: A Case report. People's Journal of Scientific Research. 2010; 3(2):37-40.
4. Hekmatfar S, Jafari K, Meshki R, Badakhsh S. Dental Management of Ectodermal Dysplasia: Two Clinical Case Reports. J Dent Res Dent Clin Dent Prospect 2012; 6(3):108-112.
5. Bonilla D E, Guerra L, Luna O. Over denture prosthesis for oral rehabilitation of hypohidrotic ectodermal dysplasia: A case report. Quintessence Int. 1997;28 (10):657–66.
6. Tarjan I, Gabris K, Rozsa N. Early prosthetic treatment of patients with ectodermal dysplasia: a clinical report. J Prosthet Dent. 2005; 93:419-424.
7. Shashibhushan KK, Viswanathan R, Sathyajith Naik N, Subba Reddy VV. Hypohidrotic Ectodermal Dysplasia with Total Anodontia: A Case Report. J Clin Exp Dent. 2011;3:352-355.
8. Itthagarun A, King NM. Ectodermal Dysplasia: a review and case report. Quintessence Int. 1997 Sep;28 (9):595-602.
9. Babu, S.G., Castelino, R.L., Shetty, S.R., Rao, K.A. Hereditary ectodermal dysplasia A case report. Web. Med. Central Dent. 2011.2(3):WMC001711.
10. Singh R, Lele G S. Hypohidrotic Ectodermal Dysplasia: A Case Report. International Dental and Medical Disorders. 2008; 1: 11-14.
11. Goyal M , Pradhan G, Gupta S, and Kapoor S. Hypohidrotic ectodermal dysplasia with ankylosis of temporomandibular joint and cleft palate: A rare presentation. Contemporary Clinical Dentistry. 2015;6(1): 110–112
12. Cockayne Cockayne EA. Inherited abnormalities of the skin and its appendages. London: Oxford Uni Press; 1933.
13. Sidhu M, Kale A D, Kotrashetti VS. Karyotyping, dermatoglyphic, and sweat pore analysis of five families affected with ectodermal dysplasia .Oral Maxillofac Pathol 2012 16 (3) ; 380-387.
14. Arora KS, Hundal KD, Kaur RD, Sharma SK. Hidrotic Ectodermal Dysplasia: Case Report and Review. Journal of Oral Medicine, Oral Surgery, Oral Pathology and Oral Radiology. 2015;1(3):156-159.
15. Chitty LS, Dennis N, Baraitser M. Hidrotic ectodermal dysplasia of hair, teeth, and nails: case reports and review. J Med Genet. 1996;33:707-10.
16. Seraj B and Nahvi A. Hydrotic or Hypohydrotic Ectodermal Dysplasia: Diagnostic Dilemmas. Int.J.Curr.Microbiol.App.Sci. 2015;4(8): 778-783.
17. Arfan Ul Bari, Simeen Ber Rahman. Hypohidrotic ectodermal dysplasia: a case report and literature review. Journal of Pakistan Association of Dermatologists. 2007; 17: 52-55.
18. Kaul S, Reddy R. Prosthetic rehabilitation of an adolescent with hypohidrotic ectodermal dysplasia with partial anodontia: Case report.J Indian Soc Pedod Prev Dent. 2008;26(4):177-81.
19. Prithviraj DR, Harshamayi P, Madan V, Kumar GC, Shruthi DP. A complete prosthodontic solution for patient with ectodermal dysplasia: a review. Int J Therap Appl. 2014;17:16-20.
20. Hickey AJ, Vergo TJ Jr. Prosthetic treatments for patients with ectodermal dysplasia. J Prosthet Dent. 2001 ;86(4):364-8.