Case Report

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Hereditary Hypohidrotic Ectodermal Dysplasia: Report of a Rare Case

Dentistry Section

GEETHA PARAMKUSAM[1], VENKATESWARLU MEDURI[2], LAKSHMI KAVITHA NADENDLA[3], NAMRATHA SHETTY[4]

ABSTRACT

Hereditary Hypohidrotic Ectodermal Dysplasia (HHED), an X-linked, recessive, Mendelian character, is seen usually in males and it is inherited through female carriers. It is characterised by congenital dysplasia of one or more ectodermal structures and it is manifested by hypohidrosis, hypotrichosis and hypodontia. It results from abnormal morphogenesis of cutaneous and oral embryonic ectoderm. Here, we are presenting a rare case of HHED in a 19 year female with classic features of this condition.

Key words: Hereditary hypohidrotic ectodermal dysplasia (HHED), Hypotrichosis, Anodontia, Hypodontia, Hypohidrosis

INTRODUCTION

'Ectodermal Dysplasia' comprises of a large, heterogeneous group of inherited disorders that are defined by primary defects in the development of 2 or more tissues which are derived from embryonic ectoderm. This term was first described by Thurnam in 1848 and it was coined by Weech in 1929 [1,2]. Genetic studies which have been done on more than 300 cases have revealed an X -linked mode of inheritance, with its gene locus being Xq11 -21.1; this gene is carried by females but it is manifested in males [1,2]. It is remarkable that no instance of a daughter being affected has occurred [2].This disorder might occur during the first trimester of pregnancy. If it is severe, it appears before the sixth week of embryonic life and consequently, the dentition will be affected. After eighth week, other ectodermal structures may be affected [3].

These disorders have been considered to be relatively rare, with an estimated frequency of 7 cases occurring in every 10,000 births [4]. The most common syndromes within this group include hypohidrotic (anhydrotic) ectodermal dysplasia (also known as Christ-Siemens Touraine syndrome) and hidrotic ectodermal dysplasia (Clouston syndrome), with hypohidrotic ectodermal dysplasia being the more common phenotype [5,6].

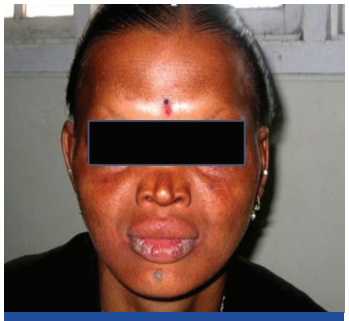
The disease is characterized by deformity of at least two or more of these tissues, which primarily involves skin, hair, nails, eccrine glands, and teeth [6], which makes it difficult for the patient not only to masticate food, but it also causes a psychological impact due to partial edentulism. Here, we are presenting a rare case of HHED with typical features in a 19 year old female.

CASE REPORT

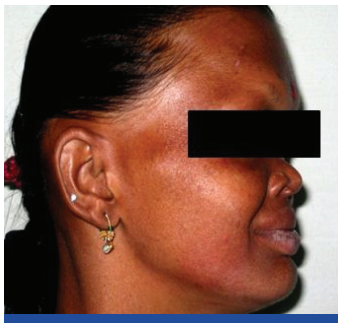
A female patient who was aged 19 years came with a chief complaint of missing teeth. Patient gave a history of (H/O) exfoliation of deciduous teeth 12 years back, after which permanent teeth had failed to erupt, due to which she had difficulty in chewing food. She gave a history of difficulty in tolerating high temperatures, especially during summers. She also gave a H/O a decreased flow of tears and photophobia. Patient's family history revealed that her grandmother from maternal side also had a similar complaint. Patient gave a H/O a consanguineous marriage between her parents.

An extra-oral examination revealed a receding hairline, scanty eyebrows, frontal bossing, [Table/Fig-1] a depressed nasal bridge [Table/Fig-2] and prominent supra-orbital ridges. Lips were protuberant and dry. Skin was dry and parched and it gave a scaly appearance [Table/Fig-3]; palms and soles appeared to be dry.

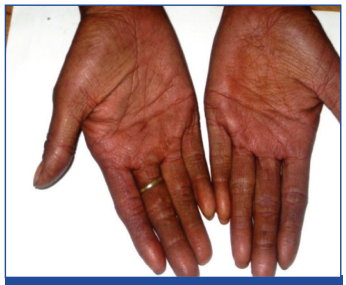
On intra-oral examination, many teeth were found to be missing. The only teeth which were present were upper left and right first



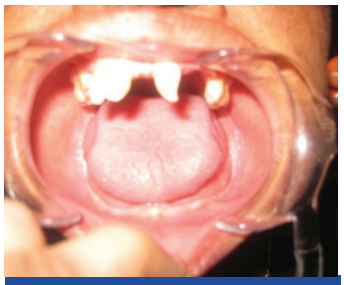
[Table/Fig-1]: Facial profile of the patient showing receding hair line, scanty eyebrows and frontal bossing



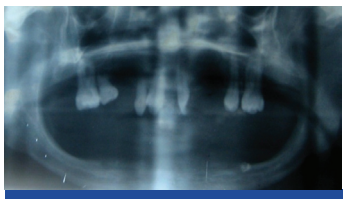
[Table/Fig-2]: Lateral profile of the patient showing depressed nasal bridge



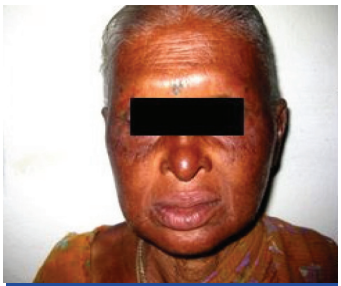
[Table/Fig-3]: Palm showing dry appearance



[Table/Fig-4]: Intraoral picture showing completely edentulous lower arch and few teeth in upper arch



[Table/Fig-5]: OPG showing absence of impacted teeth



[Table/Fig-6]: Maternal grandmother with similar features

molars, retained upper right deciduous first molar, and first premolar of left side. The anteriors were cone-shaped. The mandibular ridge was completely edentulous and it appeared to be flat [Table/Fig-4].The overall vertical dimension of face was also reduced. An orthopantomogram was advised to look for any impacted teeth [Table/Fig-5]. On radiographic examination, no impacted teeth were observed, though the roots of teeth which were present were found to be short and conical. Taking all these features into consideration; a diagnosis of HHED was made. Patient's grandmother who accompanied the patient showed similar features [Table/Fig-6] .

DISCUSSION

HHED is a X-linked recessive Mendelian character which is seen usually in males and it is inherited through female carriers. Males are affected severely, while females show only minor defects [6]. But the present case was a female who was affected, with major defects which were suggestive of HHED, which is very rare presentation [7].

Clinically, ectodermal dysplasia may be divided into two broad categories. One is the hypohidrotic form which is X-linked, which is characterized by the classical triad of hypodontia, hypotrichosis and hypohidrosis, which is also termed as Christ-Touraine Syndrome.

The other category is the hidrotic form which was described by Cloustan, which usually spares the sweat glands and can affect the teeth, hair and nails [8]. The various syndromes which are associated with ectodermal dysplasia are Rapp-Hodgkin Syndrome, Strandberg- Ronchese's Syndrome, Rosseli-Gulienetti Syndrome and various others [7].

The typical facies is characterized by fine, sparse, lustreless fair hair over scalp, which is seen in most of the patients. Extensive scaling of the skin and unexplained pyrexia and heat intolerance occurs due to anhidrosis. Intelligence is normal [7-9]. The reported case also presented with similar features of dry skin, sparse hair, intolerance to heat and hyperthermia. These features are caused by anomalies of the skin appendages, which include the hair follicles, sweat glands and sebaceous glands [8-10]. There may be partial or total absence of these glands [8,9].

Other extra–oral features include frontal bossing and sunken cheeks. A depressed nasal bridge gives added emphasis to the small size of the face, thick everted protuberant lips, wrinkled hyper pigmented periorbital skin and large low set ears [7]. Our case also presented with a marked frontal bossing, sunken cheeks, a depressed nasal bridge and thick protuberant lips. Periorbital skin did not show any hyperpigmentation and the ears were also normally positioned.

The most striking oral abnormality is missing permanent teeth. Maxillary central incisors and canines usually have a conical crown form. Very rarely, one or both jaws may be edentulous. The alveolar processes do not develop in the absence of teeth [8]. The present case also had missing permanent teeth and conical shaped anterior teeth. Due to absence of teeth, and reduced vertical height, the lips are protuberant. In reported case, the lower jaw revealed a complete edentulous arch.

Though the physiognomy is distinctive, there are several facets of the syndrome that remind the observer of other diseases or symptom complexes [10]. [Table/Fig-7] enlists differential diagnoses of Hypohydrotic ectodermal dysplasia (HED), that have few similarities as compared to HED [10].

Congenital syphilis
Chondro ectodermal dysplasia
Incontinentia Pigmenti
Rothmund – Thomson syndrome
Progeria
Idiopathic hypoparathyroidism – Addison's disease – moniliasis syndrome

[Table/Fig-7]: Differntial Diagnosis of HED [10]

An early identification of the condition and a multidisciplinary approach which involves oral and maxillofacial surgeons, a pedodontist, an orthodontist, and a prosthodontist, is required, for a comprehensive dental management of such cases [4]. The appearances of the patients can be greatly enhanced by making removable prosthodontic appliances and dental implants, which may be successfully employed to support and retain teeth [9].

In conclusion, the most important aspect which has to be considered in these patients is the psychological impact which is caused by absence of teeth. An early diagnosis and dental treatment is therefore an essential part of the management of ectodermal dysplasia.

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PARTICULARS OF CONTRIBUTORS:
1. Professor & HOD, Department of Oral Medicine and Radiology, Kamineni Institute of Dental Sciences, Sreepuram, Narketpally, Nalgonda District, Andhra Pradesh – 508254, India.
2. Professor, Department of Oral Medicine and Radiology, Kamineni Institute of Dental Sciences, Sreepuram, Narketpally, Nalgonda District, Andhra Pradesh – 508254, India.
3. Senior lecturer, Department of Oral Medicine and Radiology, Kamineni Institute of Dental Sciences, Sreepuram, Narketpally, Nalgonda District, Andhra Pradesh – 508254, India.
4. PG Student, Department of Oral Medicine and Radiology, Kamineni Institute of Dental Sciences, Sreepuram, Narketpally, Nalgonda District, Andhra Pradesh – 508254, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:
Dr. Namratha Shetty,
PG Student, Department of Oral Medicine and Radiology , Kamineni Institute of Dental Sciences, Sreepuram, Narketpally, Nalgonda District, Andhra Pradesh – 508254, India.
Phone: 07702832370, E-mail: namrata11@gmail.com

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