ORIGINAL RESEARCH PAPER

Volume-8 | Issue-7 | July - 2019 | PRINT ISSN No. 2277 - 8179

INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

HEREDITARY HYPOHIDROTIC ECTODERMAL DYSPLASIA: REPORT OF 3 CASES WITH REVIEW OF LITERATURE

Dental Science

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ABSTRACT

The ectodermal dysplasias comprise a large, heterogeneous group of inherited disorders that are characterized by primary defects in the development of 2 or more tissues derived from embryonic ectoderm. The two most common types of ectodermal dysplasias are hypohidrotic ectodermal dysplasia (Christ Siemens-Touraine syndrome) and hidrotic ectodermal dysplasia (Clouston syndrome). The tissues primarily involved in hypohidrotic ectodermal dysplasia are skin, hair follicles, eccrine glands, sebaceous glands, nails and teeth. Hypohidrotic hereditary ectodermal dysplasia hence manifests as sparse hair, oligodontia, and reduced sweating. Here we present 3 cases of Hypohidrotic hereditary ectodermal dysplasia of which 2 male patients were siblings and one 6 years old male child patient.

KEYWORDS

Heterogeneous disorder, Hypohidrotic, Hereditary, Siblings

INTRODUCTION:

Ectodermal dysplasias (EDs) was coined by Weech in 1929.[1]Ectodermal dysplasia is a group of rare, inherited disorders caused by dysplasia of tissues of ectodermal origin-primarily, nail, teeth and skin, and occasionally, dysplasia of mesodermally derived tissues. Clinically, two main forms of ectodermal dysplasia have been distinguished: the Hypohidrotic form / Siemens Tourian Syndrome (x-linked recessive) and the Hydrotic form / Clouston syndrome (Autosomal inherited). The Hypohidrotic ectodermal dysplasia is more common form and exhibit classic triad of sparse hair (hypotrichosis), abnormal or missing teeth (hypodontia or anodontia), and complete or partial absence of sweat glands (Hypohydrosis) as a result of which patients cannot sweat normally and may exhibit heat intolerance.[2]

Case 1:

A male patient of age 25 years visited to the department of oral medicine and radiology of our institution with chief complaint of several missing teeth since birth.

Fig 1: Clinical photograph showing scanty hair, saddle nose, protuberant lips

Detailed history revealed that the patient had not undergone any extraction of any teeth and no history of exfoliation of any teeth was also reported. Patient also complained of dry skin and decreased sweating. Clinical examination revealed stunted growth of scalp hair, scanty eyelashes and eyebrows, prominent forehead, saddle nose, prominent lips and multiple missing teeth in upper and lower jaws of the patient. Radiographic findings showed absence of multiple missing teeth in upper and lower jaw suggestive of Oligodontia. There were also retained deciduous teeth present.

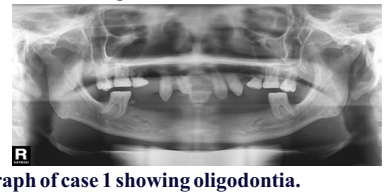

Fig 2: Radiograph of case 1 showing oligodontia.

Case 2:

This male patient was **sibling** of the first case aged 30 years.

Fig 3: Clinical photograph of (sibling) showing similar features

He also reported with the chief complaint of several missing teeth since birth. He also gave history of no extraction or exfoliation of any teeth. On extraoral examination similar features were noted as in first case and on intraoral examination several missing teeth were noted in upper and lower jaw.

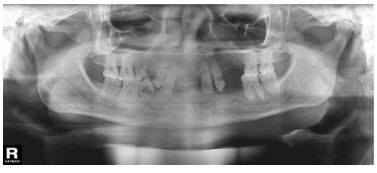

Fig 4: Radiograph of case 2 showing similar features as case 1

Case report 3 :(child patient)

A six year old boy reported to the department of oral medicine and radiology of our institution with the complaint of absence of teeth and inability to eat. The general medical history and family history was non-contributory. The child showed typical features of hypohidrotic ectodermal dysplasia: anodontia, hypohidrosis, hypotrichosis, prominent forehead, saddle nose, diminished lower facial height, and sparse scalp hair, absence of eyelashes and eyebrows and protuberant lips.

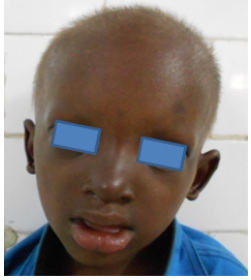

Fig 5: Clinical photograph of case 3 showing hypotrichosis, protuberant lips and saddle nose

The extraoral examination showed skin with scaling and palmoplantar hyperkeratosis. The intra-oral examination revealed complete absence of teeth except one conical shaped tooth in upper anterior region, as a result of which the edentulous alveolar ridges were deficient in both height and width. Furthermore patient had heat intolerance and suffered from recurrent fever since birth. Radiographic findings showed one upper central incisor with stunted root and one lower left molar tooth bud.

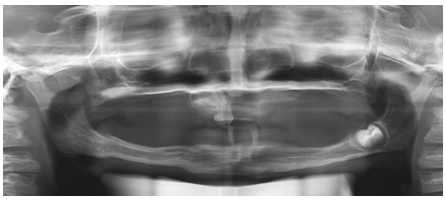

Fig 7: radiograph of case 3 showing deficient alveolar height of both maxillary and mandibular jaws.

DISCUSSION:

Thurman published the first report of a patient with ectodermal dysplasia in 1848, but the term ectodermal dysplasia was coined by Weech in 1929.[3]In 1875, Charles Darwin documented it among a Hindu family, where 10 men in the course of four generations were affected, which was communicated to him by Mr. W. Weddenburn. Both jaws taken together, only four small and weak incisor teeth, eight posterior molar were present.[4,5] Hypohidrotic Ectodermal Dysplasia or anhidrotic ectodermal dysplasia is the most common syndrome among this large group of hereditary disorders. Hypohidrosis, hypotrichosis and hypodontia constitute the main symptoms of the syndrome. HED affects at least one in 17000 people worldwide.[6] In the hypohidrotic form, the skin is soft, thin, and dry. The sebaceous glands are also defective or absent. Palms and soles are hyperkeratotic; pseudorhagades are present around the eyes. In the oral cavity, the most striking feature is oligodontia. The teeth that are present have abnormal crown form. Teeth in the anterior region of maxilla and mandible are conical in shape . The characteristic facial features are frontal bossing, depressed nasal bridge, prominent supraorbital ridges, prominent and obliquely set ears, midface is depressed, the lower third of the face appears small due to lack of alveolar bone development and lips are protuberant.[6,7] The tissues that are commonly involved in ectodermal dyplasia are the skin, hair, nails, eccrine glands, and teeth. The Hypohidrotic ectodermal dysplasia is a more common phenotype and is also known as Christ-Siemens-Touraine syndrome. The dermatological changes in ectodermal dysplasia include Hyperkeratosis of palms and soles. Scalp and facial hair is generally sparse, short, fine, dry although in some cases complete absence of hair has been reported. Dyshidrosis (abnormal or missing sweat glands) often leads to dry skin, unexplained pyrexia and heat intolerance. Anodontia leads to atrophy of alveolar ridge and reduction in vertical dimension of the lower face is reduced.[7] Mutations in EDA (ectodysplastin A is a protein), EDAR (ectodysplastin A receptor) and EDARADD (ectodysplastin A receptor associated death domain) genes are now identified to cause Hypohidrotic ectodermal dysplasia. These genes provide instructions for making proteins that work together during embryonic development. These proteins form part of a signaling pathway that is critical for the interaction between the two germ layers the ectoderm and mesoderm. In the early embryo these cell layers form the basis for many of the body's organs and tissues. Ectoderm-mesoderm interaction is essential for the formation of several structures that arise from the ectoderm including skin, hair, nails, teeth and sweat glands.[3]

Possible diagnosis of ectodermal dysplasia:

1) Prenatal diagnosis of this condition has been reported previously in high-risk pregnancies on the basis of histologic analysis of fetal skin obtained by second-trimester fetoscopy-guided skin biopsy. The diagnosis was achieved by identification of the distinct facial features at 30 weeks' gestation on three-dimensional (3D) ultrasonography.[8] In subjects with a family history of hypohidrotic ectodermal dysplasia, the diagnosis of this rare condition can be established noninvasively by sonography in the second trimester of pregnancy.[9]

2) Starch-iodide paper palm imprints identified a higher likelihood of diminished or absent sweat in the affected group, but this test had a low sensitivity (44%) and an imperfect specificity (93%). Palms that lack eccrine structures are diagnostic of HED.[10]Patchy distribution of sweating with help of Starch and iodine sweat testing leaving v-shaped pattern of pattern of streaks (Blaschko lines).[11]

Oral management of Ectodermal dysplasia:

Ectodermal dysplasia is heterogenous disorder; hence management aims at improving aesthetics and function. Early dental examination is required, and dental radiography and measurement of salivary flow should be carried out. Reduced salivary flow is associated with dental caries, so preventive treatments, including fluoride-therapy and regular periodic examinations should also be performed.[12] The oral rehabilitation of ED patients usually consists of complete or removable prosthesis in the developing years of the patients, followed by a definitive prosthesis after the complete development of the alveolar process. Orthodontic treatment needs to be considered for mal aligned teeth to be positioned favourably prior to removable or fixed partial denture treatment.[4]

CONCLUSION:

Family history should not be considered essential factor in diagnosis with respect to ectodermal dysplasia, as condition shows multiple modes of inheritance.[6] Thorough examination for accurate diagnosis and treatment by multidisciplinary approach is helpful for patients with ED.[4] Dental professionals play an essential role in contributing to the overall appearance and wellbeing of the affected patients.[1]

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