Hypohydrotic Ectodermal Dysplasia: A Case Report and Review
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Abstract
Ectodermal dysplasias are a large hereditary group of disorders which are usually manifested as X-linked recessive hypohidrotic ectodermal dysplasia (HED) and has a full expression in males, whereas females show little to no signs of the disorder. The two most common types of ectodermal dysplasias are the X-linked recessive hypohidrotic ectodermal dysplasia (Christ-Siemens-Touraine syndrome), and hidrotic ectodermal dysplasia (Clouston syndrome). Hypohidrotic ectodermal dysplasia is characterized by hypodontia, hypotrichosis and hypohydrosis. We present a case of a 17 year old male patient suffering from hypohydrotic ectodermal dysplasia and a review on hypohydrotic ectodermal dysplasia discussing the etiology, clinical features, diagnosis and treatment plan.

Keyword: Hypohydrotic Ectodermal Dysplasia, Hypodontia, Hypohydrosis, Prosthetic Therapy

Introduction:
Ectodermal dysplasia is a congenital, diffuse and nonprogressive hereditary disorder first described by Thurman1,2 which occurs due to disturbances in the ectoderm of the developing embryo. It is basically of two types i.e hypohidrotic ectodermal dysplasia also known as Christ-Siemens-Touraine syndrome, and hidrotic ectodermal dysplasia also known as Clouston syndrome.3 The clinical features associated with Ectodermal dysplasia most commonly consists of onchodysplasia, alopecia or hypotrichosis, hypohydrosis and hypodontia.2,4-6

Case Report:
A 17yr old male reported to our institution with a chief complaint of missing tooth in upper and lower anterior region. Patient gave no history of exfoliation or extraction of teeth but gave a history of delayed eruption of teeth. When asked about a similar history in the family patient gave a positive history of his sister suffering from a similar complaint. Patient also complained of dry skin (Figure No. 1) and absence of sweat in his skin (Figure No. 2) and he was intolerable to withstand hot water and hot environment. Patient also gave a history of dry mouth. On extra oral examination, patient had scanty eyebrows and eyelashes (Figure No. 1) along with frontal bossing and a saddle nose was evident.

Intra orally patient was partially edentulous (Figure No. 3). Maxillary anterior teeth appeared to be conical, high arched palate was seen on clinical examination (Figure No. 4). His OPG was relevant to the above findings (Figure No. 5). So with the above intraoral and extraoral clinical findings regarding the complain of absence of sweat, a diagnosis of Hypohydrotic Ectodermal Dysplasia was made.

Discussion:
The ectodermal dysplasias are a heterogenous group of inherited disorders which occur approximately one in every 100,000 births which are caused by primary defects in the development of two or more tissues derived from the embryonic ectoderm.7
Table No. 1: Differences between the Hydrotic and Hypohydrotic Forms of Ectodermal Dysplasia

<table>
<thead>
<tr>
<th></th>
<th>Hydrotic</th>
<th>Hypohydrotic</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mode of Inheritance</td>
<td>Most often autosomal dominant</td>
<td>Most often autosomal recessive</td>
</tr>
<tr>
<td>Scalp Hair</td>
<td>Soft, dawny, color is darker</td>
<td>Fine in texture, fair and short</td>
</tr>
<tr>
<td>Teeth</td>
<td>Anodontia to hypodontia</td>
<td>Anodontia to hypodontia</td>
</tr>
<tr>
<td>Lips</td>
<td>No Abnormality</td>
<td>Protruding</td>
</tr>
<tr>
<td>Sweat glands</td>
<td>Active</td>
<td>Reduced to absent</td>
</tr>
<tr>
<td>Nasal bridge</td>
<td>No Flattening</td>
<td>Underdeveloped</td>
</tr>
<tr>
<td>Nails</td>
<td>Dystrophic nails</td>
<td>No Abnormality</td>
</tr>
<tr>
<td>Eyebrows</td>
<td>Frequently Absent</td>
<td>Absent</td>
</tr>
<tr>
<td>Eyelashes/Pubic/Axillary hairs</td>
<td>Scanty/absent</td>
<td>Variably affected</td>
</tr>
</tbody>
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Figure No. 1 & 2: Extraoral Examination of the Patient

Figure No. 3 & 4: Intraoral Examination of the Patient
More than 300 cases which had undergone genetic studies revealed that X linked mode of inheritance with its gene locus being Xq11-21.1, is carried by the female but manifested only in the male. Improvements in esthetics, speech, masticatory efficiency as well as to improve both sagittal and vertical skeletal relationship during craniofacial growth and development has to be provided by oral rehabilitation.

Freire-Maia and Pinheiro proposed the first classification system of the ectodermal dysplasias in 1982, and provided additional updates in 1994 and 2001. The patients were classified into subgroups which were based on the presence or absence of the following ie trichondysplasia (abnormal hair), abnormal dentition, onchondysplasia (abnormal nails) and dyshidrosis (abnormal or missing sweat glands).

Ectodermal dysplasias were classified into either group A or group B disorder. Group A disorder are manifested by at least two of the four classic defects of ectodermal structures as defined above, with or without other defects, group B disorders, were manifested by a defect in one classic ectodermal structure (1-4 from above) in combination with, a defect in one other ectodermal structure (i.e., ears, lips, dermatoglyphics). In group A eleven subgroups were defined, each with a distinct combination of two or more ectodermal defects (e.g., 2-4, 1-3, 1-4 from above). The group B disorders were indicated as 1-5, 2-5, 3-5 or 4-5 (from above). Table No. 1 shows differences between the hidrotic and hypohidrotic forms of ectodermal dysplasia.

It results from developmental defect of embryonic ectodermal structures and the genetic defects responsible for approximately 30 of the ectodermal dysplasia have been identified. The two types of Ectodermal dysplasia i.e. X linked recessive hypohidrotic ectodermal dysplasia (EDA or Christ-siemens-Touraine syndrome) are caused by maturation in EDA, which encodes for ectodysplasin protein and hidrotic ectodermal dysplasia, which is an autosomal dominant disorder, is caused by mutation in GJB, which encodes for connexin 30. Mutation in the DL gene, which encodes for the EDA receptor causes autosomal dominant and autosomal recessive hypohidrotic ectodermal dysplasia.

Ectodermal dysplasia is basically divided into two broad categories ie hypohidrotic form(Christ-Touraine Syndrome) which is X-linked and is characterized by the classical triad of hypodontia, hypotrichosis and hypohidrosis and the other category ie hidrotic form(Clouston Syndrome), which also affects the teeth, hair and nails sparing the sweat glands.

Extraorally fine, sparse, lustreless fair hair is seen over the scalp along with extensive scaling of the skin and unexplained pyrexia and heat intolerance most commonly occurs due to anhidrosis. Normal intelligence is observed. The other extra–oral features are frontal bossing, sunken cheeks, depressed nasal bridge, thick everted protuberant lips, wrinkled
hyper pigmented periorbital skin and a large low set of ears.21

Intraorally missing permanent teeth are most commonly present, the maxillary central incisors and canines present with a conical crown form. In rare instances, one or both jaws may be edentulous and the alveolar processes may not develop due to the absence of teeth.20

Diagnosis is most commonly based on family history, thorough clinical and radiographic examination. History of hyperpyrexia and the tooth form and tooth morphology leads to the diagnosis. However the diagnosis in early infancy is difficult as the manifestations involving teeth, hair and inability to sweat are difficult to detect.23 Diagnostic criteria regarding the structural and biochemical characteristics of hair, dermatoglyphic analysis, characteristics of lacrimal secretion and missing teeth have also been studied.24,25

Treatment should be initiated in children suffering from ectodermal dysplasia by providing dentures as early as 2 years of age.26 Due to difficulty in mastication and speech, implantation of prosthetic teeth in adults is necessary. A positive self image outlook and an overall oral health must be incorporated by providing aesthetic dental interventions at proper time in patients with ED.27-31

Conclusion:
The clinical manifestations of ectodermal dysplasia cause considerable social problems in individuals affected by the condition. It not only affects the oral functions of the patient but the normal body functions also get hampered. A correct diagnosis & treatment by the multidisciplinary approach can be helpful for the successful management of the patient.

References:


