Hypohidrotic ectodermal dysplasia: A case report

Hipohidrotik ektodermal dysplasia: Bir olgu sunumu

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Abstract
Ectodermal dysplasia is a hereditary disorder exhibiting developmental abnormalities of ectodermal structures. Patients with these disorders usually exhibit multiple missing teeth, sparse hair and inability to sweat with other manifestations. Early recognition and treatment by clinicians will prevent development of psychological trauma in patient and gives confidence to the patient. The current article presents a case of a 28 year old male with hypohidrotic ectodermal dysplasia who presented with rare unusual hyperpigmentations.
Keywords: Christ-Siemens Touraine syndrome, ectodermal dysplasia, hypohidrosis; missing teeth

Özet
Ektodermal displazi, ektodermal yapıların gelişimsel anomalilerini sergileyen kalıtsal bir hastalıktır. Bu hastalığı olan bireyler, genellikle diğer belirtiler ile birlikte birden fazla kayıp diş, seyrek saçlar ve yetersiz terleme belirtileri gösterirler. Klinisyenlerin erken tanı ve tedavisi, hastalarda psikolojik travma gelişimini önleyecek ve hastaya güven verecektir. Bu makalede, nadir sıra dışı hiperpigmentationlu, hipohidrotik ektodermal displazi olan 28 yaşındaki erkek bir olgu sunulmuştur.
Anahtar kelimeler: Christ-Siemens Touraine sendromu, ektodermal displazi, hipohidroz, eksik dişler

Introduction

Ectodermal dysplasia (ED) are group of rare X linked inherited disorders characterized by dysplasia of tissues of ectodermal origin-primarily teeth, skin and nails and occasionally, dysplasia of mesodermally derived tissues (1). The disease has prevalence of approximately 1 in 100,000 live births (2). ED is clinically classified as:

1. Hypohidrotic ectodermal dysplasia (HED) which is also called as Christ-Siemens-Touraine syndrome and
2. Hidrotic ectodermal dysplasia also called as Clouston syndrome.

The hypohidrotic form exhibits the classic triad-hypohidrosis, hypotrichosis and hypodontia. Males are affected severely, while females show only minor defects and in patchy distribution. The disease is usually inherited as autosomal recessive disorder. In the hidrotic form teeth, hair and nails are affected and sweat glands are usually spared. It is usually inherited as an autosomal dominant trait. Other inheritance modalities like autosomal recessive have also been reported (3).

Case report

A 28- year- old male patient came with a chief complaint of multiple missing teeth since childhood. The patient gave a history of absence of permanent teeth, intolerance to heat, less sweat and saliva production and difficulty in chewing. There was no history of consanguineous marriage between the parents nor there similar problems in any of his family members. On extra oral examination, the patient exhibited dry skin with periocular and periorbital area with hyperpigmentations. The skin was wrinkled with sparse hair on the body, face and scalp. There were also diffuse pigmentations seen on forehead and frontal scalp area. Hairs present were fine in texture & lighter in color. Prominent supraorbital ridges, frontal bossing, thick everted lips; small and outwardly placed ears and flattened nasal bridge were observed. Complete absence of eyebrows and sparse eyelashes were found (Fig.1). The skin was warm and dry. The palmar surfaces were dry and pigmented (Fig.2). Nails and patients intelligence were normal.

Intra oral examination revealed completely edentulous mandible. Apart from 11, 21, 22, rest all the teeth in maxilla were absent. Teeth present were conical in shape and smaller in size with slight loss of alveolar ridge height and width (Fig.3 and 4). Oral mucosa was normal in color and texture. Orthopantomogram (OPG) revealed multiple missing teeth with absence of impacted teeth (Fig.5). A diagnosis of hypohidrotic ectodermal dysplasia was made and patient was referred to expert prosthodontist for management of patient's chief complaint. Patient was also advised to consult dermatologist for skin problem.

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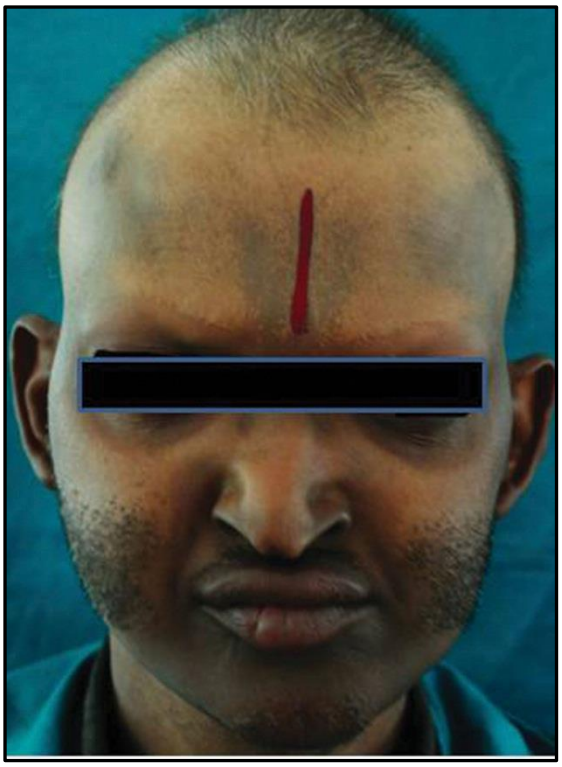

Figure 1. Extraoral examination shows sparse light hair, absence of eyebrows, frontal bossing, depressed nasal bridge, diffuse perioral and periorbital hyperpigmentation also involving forehead and frontal scalp, thick everted lips and pointed chin.

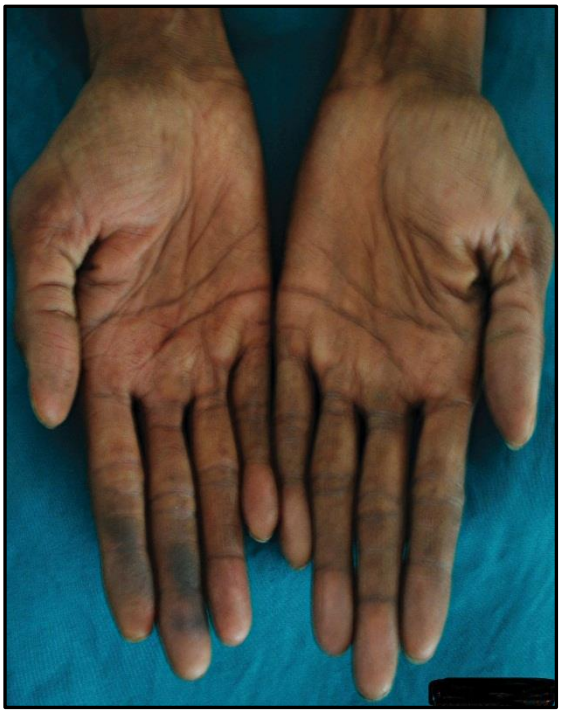

Figure 2. Dry and pigmented palmar surfaces.

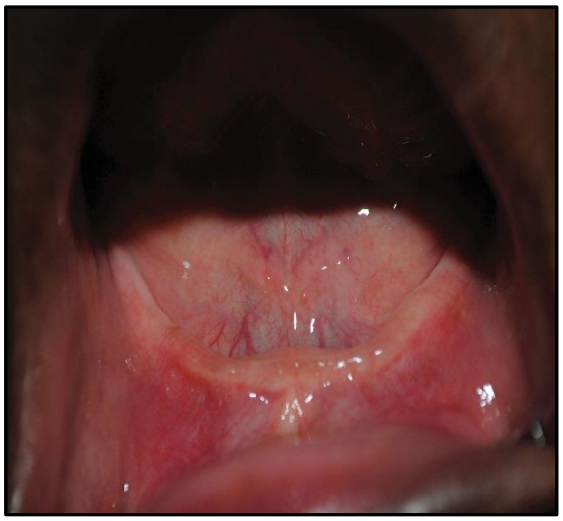

Figure 3. Completely edentulous mandible.

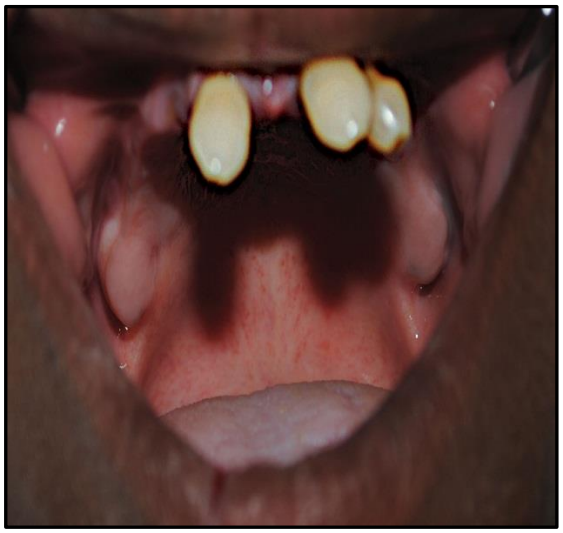

Figure 4. Conical shaped maxillary incisors with partially edentulous maxilla

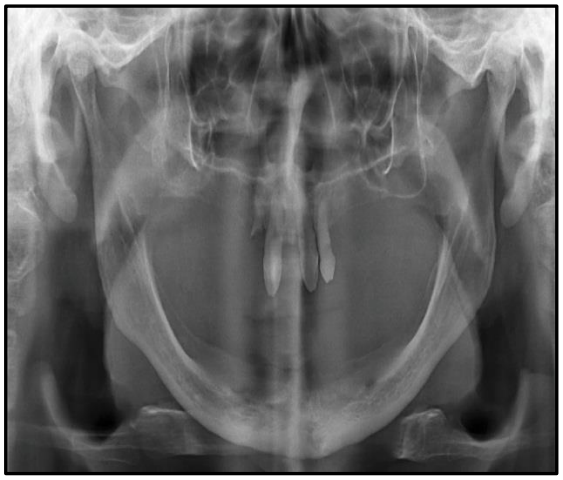

Figure 5. Orthopantomogram shows missing permanent teeth and absence of any impacted teeth.

Discussion

ED was first described by Thurman in 1848 and was coined by Weech in 1929 (4). Genetic defects in ectodysplasin signal transduction pathways located at Xq12-13 results in aplasia, hypoplasia, or dysplasia of these structures. Epithelial cells in developing tooth, hair follicle, and eccrine sweat gland utilize this pathway during morphogenesis (5, 6). The characteristic extraoral features of this disease are frontal bossing with the forehead appearing square in shape, prominent supra orbital ridge, depressed nasal bridge (saddle nose), sunken cheeks, pointed chin, protruberent and thick everted lips, wrinkled hyper pigmented periorbital and perioral skin and low set ears. The midface is depressed and hypoplastic, giving it a "dished-in" appearance. Other abnormalities of skin and nails may also be noted. The hyperpigmentations seen in this case is a rare finding in ED. This might have occured because of chronic damage to the sensitive skin due to this disease. Most patients have fine, sparse, lusterless, fair hair. Extensive scaling of the skin, dry skin and unexplained pyrexia and heat intolerance occurs due to anhidrosis or hypohidrosis. Patients usually exhibit normal intelligence (6-8).

Dental markers of ED are present in almost all patients. It includes multiple congenitally missing teeth especially permanent teeth, conical-shaped crowns, delayed teething and reduced saliva flow. Teeth may manifest abnormalities in structure, composition and size of crowns and roots. Microdontia is frequently observed in affected individuals and conical pointed teeth are commonly seen. Cases of ED with one or both jaws edentulous are a rare finding. The alveolar processes do not develop in the absence of teeth resulting in reduced vertical height. Due to this lip becomes protuberant, thick, everted and chin becomes pointed. The characteristic clinical features of this disease are often sufficient to a reasonable diagnosis of this disease. Molecular marking, family history and histopathology may also be helpful. OPG may be helpful to detect abnormalities like impacted teeth, supernumerary teeth, taurodontism and other unexpected abnormalities (9-13).

The major psychological concern seen in these patients are the lack of teeth and their appearance that is their face resembles that of an older person. Treatment involves symptomatic management. Maintenance of cool and ambient temperature is important to prevent hyperthermia as it may be life threatening (5, 8). Patients should receive early education about this disease and how to combat and cope with it. Patients should be assured and given confidence that they can thrive well as any of the normal persons upon treatment. Preventive procedures such as oral hygiene control of bacterial plaque, removal of deleterious habits should be performed regularly. Procedures of oral rehabilitation such as complete dentures, partial dentures, dental implants, dental restorations, crown and bridge, etc should be carried out regularly depending upon the peculiarity of the case. Patients often require multiple and frequent periodic follow up visits for life time. Early prosthetic rehabilitation of HED patient improves esthetic and oral functions, gives psychological, social comfort, confidence and contributes to the overall well-being of the patient.

Conclusions

Clinicians should consider ED in patients with anomalies of tooth number, form and structure. The clinical features are characteristic for this disease and cases should receive early intervention and treatment. The most important aspect which has to be considered in these patients is the psychological impact which is caused by absence of teeth. Although restorative management may be challenging, the importance of basic prevention should be stressed. An early identification of the condition and a multidisciplinary approach involving oral and maxillofacial surgeons, a pedodontist, an orthodontist, and a prosthodontist is required for comprehensive dental management of such cases with continuous reassessment, correction and follow up.

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