

HYPOTRICHOTIC ECTODERMAL DYSPLASIA -A CASE REPORT

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ABSTRACT

Ectodermal dysplasia is a rare hereditary disorder. Its Hypohidrotic (HED) variant is also known as Chirst-Siemens-Touraine syndrome. It is inherited as an X- linked trait. Such Patients are characterized by the clinical manifestations of Hypodontia, Hypotrichosis, Hypohidrosis and a highly characteristic facial physiognomy. This article, reports a typical case of Hypohidrotic Ectodermal Dysplasia (HED) and management.

KEYWORDS: Ectodermal dysplasia, Hypohidrotic ectodermal dysplasia, Hypodontia, Hypotrichosis, Hypohidrosis

INTRODUCTION

The national foundation of Ectodermal dysplasia (NFED) defines Ectodermal dysplasia (ED) as a genetic disorder in which there are congenital birth defects (abnormalities) of two or more ectodermal structures. These structures include skin, hair, nail, teeth, nerve cells, sweat glands, parts of eye and ear and parts of other organ¹. The condition is hereditary and non progressive. More than 192 distant disorders have been described².

CASE REPORT

A 10 year old male patient reported with the chief complaint of malformed teeth and delay in eruption of other teeth. There was no family history of consanguinity. The patient had been full term baby and delivery was uneventful. He had inability to sweat and added he could not tolerate heat well.

The extra oral examination revealed frontal bossing, depressed nasal bridge and protuberant lips due to absence and mal developed teeth. The patient also presented with sparse scalp hair, missing eyelashes and eyebrows. The skin appeared soft, thin and dry with a hyper pigmentation around the eyes and the mouth. A deep mentolabial fold was noticed. The nails were normal.

Intraoral examination revealed conically shaped maxillary incisors and canine and Hypodontia of maxillary and mandibular arches. The palate was shallow and the oral mucosa was healthy with a slight dry appearance. The tongue was relatively large, but no signs of macroglossia could be detected. The flow of saliva appeared to be reduced. The alveolar ridge in the region of absent teeth was flat and atrophied.

Panoramic view confirmed the intraoral findings pertaining to shape of the crown and multiple missing tooth buds. It also revealed short roots with enlarged pulp chambers.

By considering the presence of triad of Hypohidrosis, Hypodontia and Hypotrichosis, with normal nails, diagnosis of hypohidrotic ectodermal dysplasia was made.

Keeping in mind the physiological and psychosocial reasons, patient was provided with dentures. The interdental spaces between upper central incisors were reduced with elastics and semipermanent cantilever type partial fixed dentures were given with minimal crown preparation. Care was taken to allow alveolar growth by providing gap in midline. Additionally removable partial dentures were provided in upper and lower arches. Patient was

asked to come for follow up for modifications in prosthesis in every 6- 12 months to cope up with the ongoing growth processes.

DISCUSSION

Ectodermal dysplasia consists of clinical and genetic heterogeneous group of disorders, characterized by either absence, complete or delayed development of one or more of the appendages derived from epidermal tissue (hair, sweat gland and nails) or of oral ectodermal origin during embryogenesis³. It was 1st described by Thurman in 1848.

Several classifications exist with some based on clinical features and other on genetic components of the disorder. Clinically there are two board groups:

- (a) Group A – includes those cases in which there is a defect in atleast 2 of the 'classic' structures, i.e. Hair, nails, teeth and sweat glands with or without other defects.
- (b) Group B – encompasses those cases in which only 1 of the 'classic' structure is affected, but there co-exist at least another ectodermal defect.

All cases can be further subdivided into

- I) Pure Ectodermal dysplasia – in which there are only ectodermal signs
- II) Ectodermal syndrome – in which there are ectodermal, features as well as other malformations⁴.

Two different forms are clinically distinguished, CLOUSTAN'S SYNDROME (hidrotic form) and CHIRST- SIEMENS- TOURAINE SYNDROME (hypohidrotic form)⁵.

The genetic basis is largely unknown, but recent evidence implicates a genetic defect in the signaling pathway that regulates ectodermal organogenesis. Chirst-Siemens-Touraine Syndrome (Anhidrotic/ hypohidrotic ectodermal dysplasia) is mainly inherited through an x – linked recessive trait. The causative gene (EDA gene) for this has been identified on chromosome Xq 12- q13 and encodes a novel transmembrane protein, ectodysplasin, which seems to play a critical role in epithelial –

mesenchymal interactions during hair follicle morphogenesis. There also exist, Autosomal dominant and recessive forms. Gene responsible for these forms have been isolated and termed as downless (DL). Males are mainly affected and Female being carriers.⁶. It is well known that the chance of being homozygote affected in this type transmission increased with consanguineous marriages, which fortunately not there in our case.

Hidrotic variant of ED is a rare Autosomal dominant genodermatosis. Recently multipoint linkage analysis has mapped the disease to the pericentromeric region of chromosome 13q (13q 11-12)⁷.

Most of the clinical findings in this report were typical characteristics of Chirst-Siemens-Touraine Syndrome as reported by various authors^{8, 9, 10} particularly with respect to the tetrad of hypohidrosis, Hypotrichosis, Hypodontia and characteristic facies. The first feature among the tetrad is hypohidrosis/ anhidrosis which means, complete or partial absence of sweat glands making skin smooth, thin, dry with reduced or absent sweating and often present with episodes of hyperthermia or unexplained fever in infancy or childhood. Rhinitis, pharngitis, respiratory infections, diarrhea and recurrent otitis can also occur due to defective mucus gland function of nasal, pharangeal, respiratory system, GIT and ear canal respectively. Hypoplasia of salivary gland including intraoral accessory gland result in xerostomia and the protuberant lips may be dry and cracked with pseudorhagades formation.

Hypotrichosis is characterized by sparse and short hair of scalp, eyebrow, and eyelashes and represent lanugo. The mustache and beard are usually normal in appearance.

Intraoral hard tissue findings reported in our case are also well documented. It includes Hypodontia of both arches with tooth crown of conical or truncated shape, giving Dracula appearance. The roots are short and pulp chambers are large.



Fig 1. – Facial view of the patient showing the characteristic physiognomy



Fig 3. 4– Intraoral views



Fig 2. – Lateral profile showing depressed nasal bridge, frontal bossing and protuberant lips

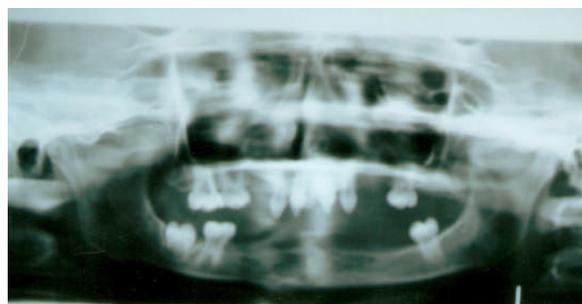


Fig 5. – Panoramic radiograph showing severe Hypodontia in maxillary and mandibular arches



Fig 6. – Intraoral view after rehabilitation

Due to Hypodontia the alveolar bone growth is decreased and the alveolar ridges are not fully developed, resulting in a loss of vertical dimensions¹¹.

In most complete forms, patient have a distinctive path gnomonic facies with sparse hair, saddle nose, frontal bossing and protuberant lips as also evident in the present case.

Dystrophies of nails, being thickened, striated and often discoloured with palmoplantar keratoderma are prominent features of hidrotic ectodermal dysplasia. Above mentioned Signs were not seen in the present case.

Treatment for the dry skin involves daily bathing with "super fatted" soap followed by the use of moisturizing lotions/ creams. Sunscreen with a moisturizing base is recommended when the child is exposed to the sun

Treatments for the hyperthermic episodes are a part of preventive and part reactive in nature. Limiting physical activity in warm / hot weather, increasing fluid intake, and proper dressing will lower the incidence of such events. When the child is overheated, he/she should be given a lukewarm or sponge bath to reduce the body temperature.

Treatment of Hypotrichosis includes use of gentle or protein coating shampoos. Wigs can improve quality of life in severe cases.

Treatment options for dystrophic nail include, lubrication of nail, keeping them short and smooth and consulting physician in case of persistent fungal infection¹¹.

Oral rehabilitation is important from a functional, esthetic and psychological standpoint. A team approach that include input from pediatric dentist, an orthodontist, a prosthodontist and a oral and maxillofacial surgeons necessary for a successful outcome. Ideally, restoration should be placed before child starts school. The principal aims of dental treatment are to restore missing teeth and bone, establish the normal vertical dimensions, and

provide support for facial soft tissues. Treatment generally include removable and / or fixed partial denture, a complete denture prosthesis^{12]} and or an implant retained^[13, 14] prosthesis or retained with magnets^[2].

In the present case, a triad of symptoms of Chirst-Siemens-Touraine Syndrome or hypohidrotic ectodermal dysplasia was observed and so the diagnosis of the same is made. The other disorders to be considered in differential diagnosis were Congenital Insensitivity to Pain with Anhidrosis, Incontinentia Pigmenti, Rapp-Hodgkin Syndrome, and Ellis-Van-Creveld Syndrome. They could be ruled out because of sensitivity to pain, regular color of skin, normal fingers, normal palate and normal limbs respectively.

CONCLUSION

Hypohidrotic ectodermal dysplasia (HED) or Chirst-Siemens-Touraine syndrome linked to chromosome X is a hereditary disorder characterized by abnormal development of tissues derived from the ectoderm. The diagnosis of HED in the neonatal and early infancy period may be difficult inasmuch as sparse hair and absent teeth are normal at this stage. In addition, dysmorphic facial features, including prominent supraorbital ridges, frontal bossing and a depressed nasal bridge, may be deemed normal variants. In childhood, the diagnosis is more easily made on the basis of history and clinical examination.

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