

Hypohidrotic Ectodermal Dysplasia in Two Siblings with Missing Teeth: A Dental Perspective

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ABSTRACT

Hereditary ectodermal dysplasia (ED) is a rare inherited disorder involving skin, hair, nails, sweat glands and teeth. Different combination of defects may give rise to variable phenotypes. ED is divided into two common types depending whether sweat glands are involved: hypohidrotic form and hidrotic form.

Hypohidrotic ED in two brothers aged 12 and 9 years is being reported. The importance of early prosthetic management in restoring function, esthetics and facilitating social interaction has been discussed.

Keywords: Ectodermal dysplasia, Partial denture, Hypodontia.

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CASE REPORT

A male patient aged 12 years, studying in 6th class, reported to the Oral Health Center, Government Medical College and Hospital, Chandigarh, India with missing teeth. The child and his parents were concerned with esthetics and speech due to missing anterior teeth. In addition, the parents reported that the child was uncomfortable in hot and humid season.

Physical Examination

Physical examination revealed a thin built child with height, weight and body mass index (BMI) of 129 cm (below 5th percentile), 25 kg (below 5th percentile) and 15 (below 5th percentile) respectively. The patient showed fine hair in the scalp and eyebrows, large and oblique set of ears, frontal bossing and brittle, thin nails (Figs 1 to 3). The external genitalia and intelligence was normal.

Family History

The child was third in birth order with two elder sisters and one younger brother. The eldest sister aged 18 years, was not available for examination, did not have any dental, sweating and nail related complaints as told by her parents. The height, weight and BMI of the second sister, aged 16 years, was 140 cm, 38 kg, and 19.3 respectively with no history of delayed dentition and any other abnormality. The height, weight and BMI of the younger brother aged 9 years,

was 120.5 cm (below 5th percentile), 21 kg (below 5th percentile) and 14.4 (below 5th percentile) respectively. His intelligence was normal. He showed fine hair in the scalp, large and oblique set of ears, thin eyebrows, brittle nails and frontal bossing (Figs 4 and 5).

Oral and Radiographic Examination

Oral examination of the patient and his brother showed hypodontia in the maxillary and mandibular arches. Small



Fig.1: Facial appearance of the patient



Fig. 2: Brittle and thin nails of the patient



Fig. 3: Ridging of the toe nails of the patient

conical teeth, loss of occlusal vertical dimension (OVD) and underdeveloped alveolar ridges were detected intraorally (Figs 6 and 7). Orthopantomogram (OPG) of the patient (Fig. 8) showed congenitally missing permanent teeth 11, 12, 13, 15, 16, 17, 18, 22, 23, 25, 26, 27, 28, 31, 32, 33, 34, 35, 36, 37, 38, 41, 42, 43, 44, 45, 46, 47 and 48. Primary teeth present were 53, 55, 62, 63, 65, 72, 73, 74, 75, 83, 84 and 85. OPG of his brother (Fig. 9) showed congenitally

missing permanent teeth 12, 13, 15, 17, 18, 22, 23, 24, 25, 27, 28, 31, 32, 35, 37, 38, 41, 42, 43, 45, 47 and 48. Primary teeth present were 52, 53, 55, 62, 63, 65, 71, 72, 73, 74, 75, 81, 82, 83, 84 and 85.

Esthetics and speech were a concern for the patient. Considering the age of the child, partial dentures (Figs 10 and 11) were planned to restore the esthetics, phonetics, mastication and psychological well being. Since, the child



Fig. 4: Facial appearance of the sibling



Fig. 7: Dentition of the sibling

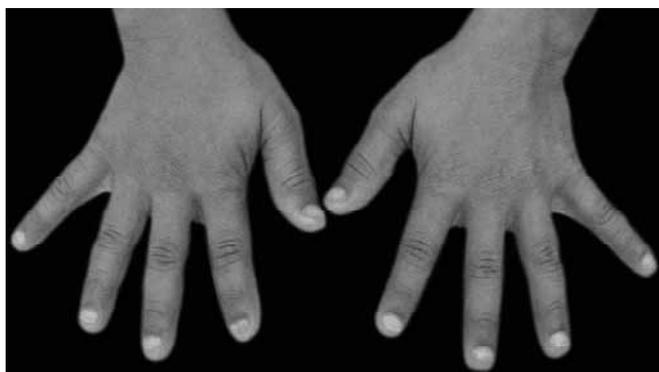


Fig. 5: Brittle and thin nails of the sibling

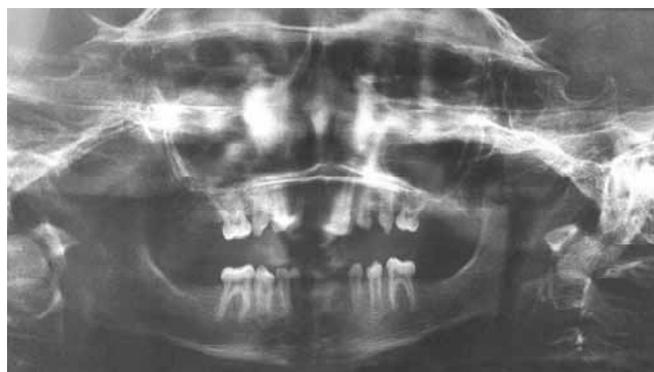


Fig. 8: OPG of the patient showing congenitally missing teeth



Fig. 6: Dentition of the patient



Fig. 9: OPG of the sibling



Fig. 10: Patient with partial denture—frontal view



Fig. 11: Patient with partial denture—lateral view

had several missing teeth and it was not possible to restore functional dentition with orthodontic intervention. Upper and lower impressions were made for the child and acrylic partial dentures were constructed. A review 1 week post insertion revealed that the child had adapted well to the dentures.

Patient's brother did not need any prosthetic management at that time as missing teeth were less in number and his esthetic and masticatory functions were not compromised. Consideration for fixed partial denture and/or implants was planned which would be reviewed after eruption of the permanent dentition.

DISCUSSION

Hereditary ectodermal dysplasia (ED) is the term used to describe defective formation of one or more tissues derived from embryonic ectoderm.¹ The tissues primarily involved are the skin, hair, nails, sweat glands and teeth. These disorders are relatively rare, with an estimated prevalence of seven cases per 10,000 births.² The first report of a patient

with ED was published by Thurman in 1848. In 1929, the term ED was coined by Weech.

To date, more than 192 different subtypes of EDs have been defined with several modes of inheritance.^{2,3} ED is divided into two common clinical types depending whether sebaceous glands are involved.³

1. Hypohidrotic (anhidrotic) ED (Christ-Siemens-Touraine syndrome) and
2. Hidrotic ED (Clouston syndrome).

Hypohidrotic (anhidrotic) ED exhibits the classic triad—hypodontia or anodontia, hypotrichosis and hypohidrosis (reduced sweating).⁴ It is more common phenotype, X-linked recessive condition and is found in all races. Female carriers outnumber affected men but females show little or no signs of the condition. Affected males usually have less and fine hair on the scalp, prominent supraorbital ridges, frontal bossing, thin or missing eyebrows, fine-linear wrinkles, prominent and oblique set ears, protuberant lips, saddle nose and defective nails.^{2,4,5} Due to the altered anatomy in the lower third of the face, they resemble edentulous elderly individuals. Oral characteristics include complete (anodontia) or partial absence (hypodontia) of teeth, impacted teeth, loss of OVD, malformed and peg-shaped or conical teeth and lack of alveolar growth. Maxillary retrusion due to sagittally underdeveloped maxilla, forward and upward displacement of the mandible and collapsed lower anterior facial height is seen in patients with hypohidrotic ED.^{1,6}

In the hidrotic form, the sweat glands are usually spared. It is usually inherited as an autosomal dominant trait. Other inheritance modalities like autosomal recessive have also been reported.^{1,6}

Diagnosis is primarily based upon the history, clinical features presented by the patient or parents, facial features and oral findings. ED types can be mild, moderate or severe.⁷ Each patient may have different combination of defects. Some patients may manifest defects in ectodermal structures alone, while others may show the combination of ectodermal defects in association with other anomalies of ear, lips and dermatoglyphics.

Various other diagnostic methods such as histopathology, radiographic evaluations (panoramic and cephalometric) and genetic testing, etc. may be done to assist in confirmation of the diagnosis of ED.

Early prosthetic treatment in children with ED results in significant improvement in esthetics, masticatory and phonetic functions. As the child's self-image is fairly completed by 4 to 5 years of age, cosmetic and prosthodontic measures should be instituted as early as possible to have the child resemble his peers.⁶ Necessary changes need to be done in denture teeth so that the appearance is always appropriate to age and similar to other children. Prosthetic

rehabilitation includes a removable and/or fixed partial denture, a complete denture, and/or an implant-retained prosthesis. Early implant placement in patients aged up to 5 to 6 years has been reported in the literature.⁸ However, this may present age related problems regarding positioning and prosthetic outcomes.

To ensure early and adequate care of a child with ED, a multidisciplinary team comprising of a pediatrician, pedodontist, dermatologist, speech therapist, psychologist and social worker, would help in facilitating and coordinating the diagnosis, treatment and monitoring of the child's progress. Also, thorough follow-up is mandatory to make suitable changes in the denture till the patient attains complete growth.

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