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Case Report

Hypohidrotic hereditary ectodermal dysplasia: A case report

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ABSTRACT

Hypohidrotic Ectodermal Dysplasia (HED) is characterized by a triad of hypotrichosis (sparse hair), hypodontia (multiple missing teeth), and hypohidrosis (inability to sweat adequately). Consequently, patients' quality of life is severely affected due to impairments in mastication, speech, and esthetics. Timely oral rehabilitation of patients with HED can enable them to lead a normal life by helping them perform their functions adequately and prevent the psychological ill-effects; in which dental professionals play a key role. The present case report describes the oral rehabilitation and management of a 7-year-old male patient with HED.

1. Introduction

Ectodermal dysplasias (EDs) are a heterogeneous group of hereditary disorders characterized by certain shared structural and functional abnormalities in tissues derived from the ectoderm. The ectoderm gives rise to various structures such as hair, teeth, nails, and the central nervous and peripheral nervous systems. Various glands including the sweat glands, sebaceous eccrine, mammary, and pituitary also arise from the ectoderm.[1] As the name implies, abnormalities in the embryogenesis of the ectoderm give rise to defective structure and function of these derivatives which collectively constitute the spectrum of ED.

Mutations in ectodysplasin-related genes (EDA1, EDAR, EDARADD) and the WNT-10A gene are responsible for ectodermal defects ED.[2] The number of alleles affected and the different extent of involvement of the various derivative structures give rise to numerous varieties of ED, of which 117 were identified by Freire-Maia in the 1970s.[3] Even with all the types combined, the incidence of ED is as low as 1 out of 1,00,000 live births, and therefore, it is considered a rare condition. Hidrotic Ectodermal Dysplasia (Clouston's syndrome) and Hypohidrotic Ectodermal Dysplasia (HED) also known as Christ-Siemens-Touraine Syndrome are the most frequent of all the types.

Records of the earliest cases of ED date back to 1792, although the first case was described by Thurman in 1848.[4] The term Hereditary ED was suggested by Weech in 1929, and because of the inability of the patients to perspire, he also added the adjective anhidrotic.[5] Since all the affected patients are never completely devoid of sweat glands, the adjective 'hypohidrotic' was discerned to be more apt by Felsher in 1944.[3]

While the syndrome only affects males due to its X-linked recessive pattern of inheritance, carrier females may display some of the characteristics of the disease.[6] True hypodontia due to agenesis of teeth, along with atrophy of the maxillary and mandibular alveolar bone are classically present in children with ED. Consequently, patients'

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quality of life is severely affected due to impairments in mastication, speech, and esthetics. The mortality rate of the condition is 28% up to 3 years of age.[7]

Timely oral rehabilitation of patients with ED can enable them to lead a normal life by helping them perform their functions adequately and prevent the psychological ill-effects; in which dental professionals play a key role. The present case report describes the oral rehabilitation of a pediatric patient with ED.

2. Case Report

A known case of 7-year-old male with HED was reported to the institutional department with multiple missing teeth. A history of absence of sweating since birth and delay in tooth development was elicited from the patient's mother. The patient had a history of absence of sweating even in hot summers, frequent rise of body temperature since early infancy, and getting micturition reflex frequently. The patient also complained of difficulty in mastication and speech. Family history revealed consanguineous marriage of parents. Parents and other family members were normal. The patient had multiple white papules over the upper back and buttocks. Hyperpigmented scaly plaques were present over both elbows and abdomen with bilateral upper and lower limb hypohidrosis. The skin was pale and dry in general with noticeable peeling from the palms, soles, and hands.

Extra oral examination of the face revealed multiple skin-colored papules around the eyes, cheeks, and forehead. Periorbital skin showed wrinkling and hypermelanosis. Frontal bossing with sparse hair over the scalp and the eyebrows was noted. The patient had a saddled nose with a long philtrum extending from unusually thick, everted lips. (Figure 1)

Intraorally, oligodontia was noted with only seven teeth being present. These included the maxillary deciduous second molars, peg laterals, and permanent first molars bilaterally; while only a partially erupted right first molar was present in the mandible (Figure 2A-C). The maxillary lateral incisors were characteristically conical in shape and transposed in the position of central incisors (Figure 2A). The deciduous maxillary molars were affected with severe caries while the maxillary right first molar had mild pit and fissure caries. The maxillary and mandibular arches exhibited severe ridge resorption.

The hemoglobin levels were found to be low (9.5 gm/dl), and the total leukocyte count was slightly above the normal level (13900/μl). The rest of the hemogram parameters including the platelet count were normal. Serum Calcium, Phosphorous, total protein, and albumin levels were also within the normal range.

The radiographic investigations included Orthopantomogram (OPG) and a hand-wrist radiograph. OPG showed the presence of peg laterals in the upper arch, developing permanent maxillary central incisors, deciduous maxillary second molars bilaterally, erupting permanent maxillary first molars bilaterally, and permanent right mandibular first molar (Figure 2D). Radiolucency was seen involving enamel, dentin, and pulp with respect to deciduous maxillary second molars bilaterally. The OPG also revealed reduced height of the mandible, which is an expected finding in a patient with ED.

Using a hand-wrist radiograph and after reviewing Fisherman's[8] skeletal maturation indicators, it was seen that the skeletal growth was not complete and hence no definitive prosthesis could be advised for another 10-11 years.

Given the history, and clinical and radiological features, hypohidrotic hereditary ectodermal dysplasia was evident.

In order to improve the appearance, mastication, and speech, the treatment plan included a removable prosthesis with both arches. The parents were informed about the procedures involved in the fabrication of the dental prostheses and the need for continuing reevaluation. Behavioral management techniques such as tell-show-do were used throughout the treatment.

2.1. Management

Planned approach for oral rehabilitation was undertaken as follows:

1. The pit and fissure caries of the permanent maxillary right first molar were treated with an atraumatic restorative treatment using Glass ionomer cement.
2. Pulpectomy was performed for the deciduous maxillary second molars bilaterally followed by full coverage restoration with stainless steel crowns.
3. Strip crowns were provided for the peg-shaped maxillary lateral incisors (Figure 3).
4. The prosthodontic phase was initiated after the restorative procedures were completed.
5. A removable partial denture was planned for the patient.
6. The primary impression of the maxillary arch was taken with alginate impression material. Owing to severe ridge resorption, the mandibular arch primary impression was taken with impression compound. (Figure 4 A)
7. The final impression (Figure 4B) of both the arches was taken with Hydrophilic vinyl polysiloxane monophase impression material- medium body (AvueGum Mono Heavy dental impression material)
8. The master casts (Figure 4C and D) were mounted on a semi-adjustable articulator, and the maxillo-mandibular relation was recorded. A balanced occlusal scheme was used to arrange the teeth.
9. The maxillary and mandibular prostheses were fabricated in heat-cure acrylic resin after a try-in and careful evaluation. After that, the removable prosthesis

was placed in the patient's mouth. (Figure 5)

2.2. *Follow-up*

The patient and his parents were taught how to properly insert and remove the prostheses, as well as how to maintain the dentures properly. The parents were reassured and speaking exercises were taught to the child (counting, reading aloud) to help train his oral musculature to accommodate the new appliance. After 24 hours, a recall was scheduled to make the necessary modifications. Follow-up visits were planned every 6 months to review the bone growth and to reline the dentures. The appliances remained stable with no appreciable bone loss or gingival irritation. The parents reported that the patient had significant improvement in speech and masticatory function. The patient had well-adapted to the dentures after a one-year follow-up

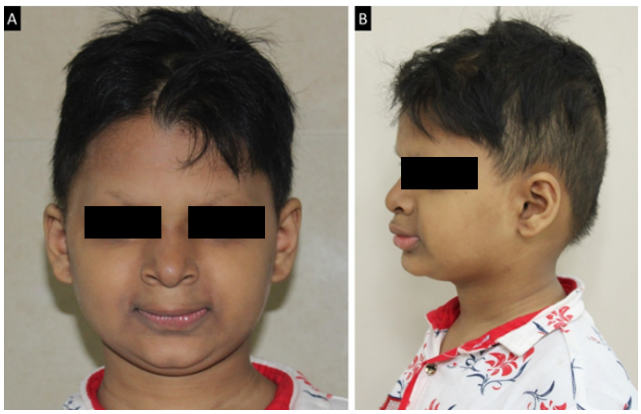

Fig. 1: Periorbital wrinkling, saddle nose, long philtrum, thick lips, and sparse hair on scalp and eyebrows noted on extraoral examination from **A:** Front profile, and **B:** Lateral profile.

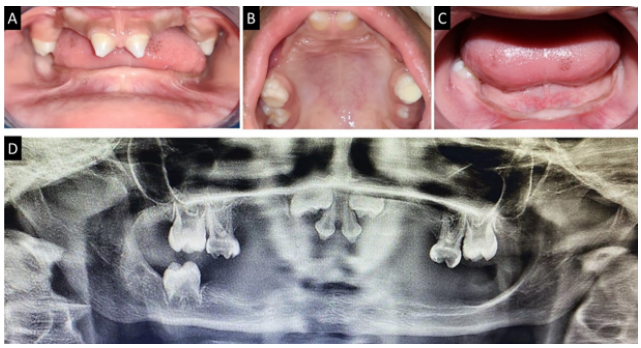

Fig. 2: A: and **B:** Maxillary arch with six teeth present including characteristic cone-shaped incisors; **C:** Mandibular arch with only one tooth present and severely resorbed ridge; **D:** Orthopantomogram confirming the clinical findings and revealing impacted teeth in the anterior maxillary region

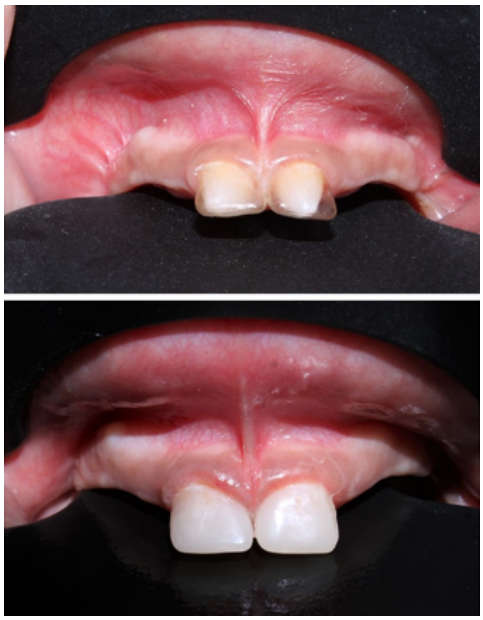

Fig. 3: Strip crowns for the peg laterals

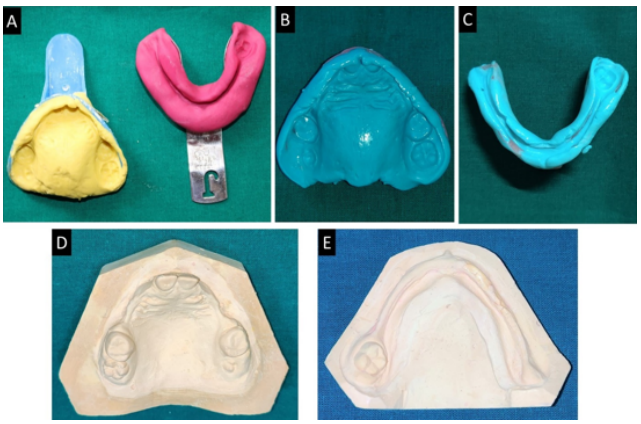

Fig. 4: A: Primary impression of both the arches; Final impression of the **B:** maxillary and **C:** Mandibular arch; Master casts of the **D:** mandibular and **E:** Maxillary arch.

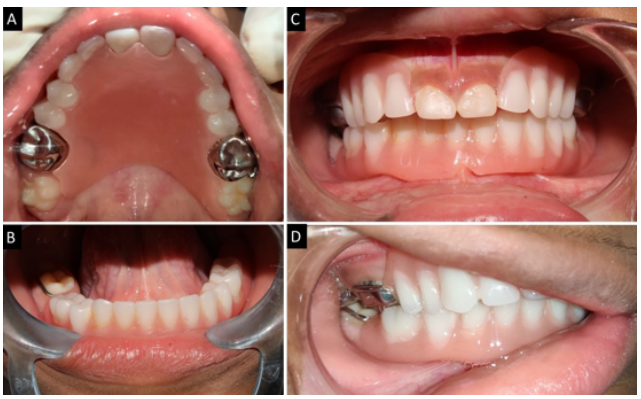

Fig. 5: Dentures in position– **A:** and **B:** Maxillary and mandibular dentures in position respectively; **C:** and **D:** Dentures in occlusion

3. Discussion

HED is characterized by a triad of hypotrichosis (sparse hair), hypodontia (multiple missing teeth), and hypohidrosis (inability to sweat adequately). A history of consanguineous marriage was reported in 68% of cases of ED in a retrospective study by More et al., which was also present in our case[7] Although not mandatory, a biopsy from the skin can help in proving the paucity/absence of the eccrine glands. Therefore, a skin biopsy was advised in the present case to increase the positive predictive value of the diagnosis. However, the patient's mother did not consent to a biopsy procedure.[9]

The mortality rate has dropped down from 30% to about 13% over the past few decades. The most common reasons for death included hyperthermia and respiratory infections, particularly in the early years. The high leukocyte count in the present case was indicative of HED-associated dysimmunoglobulinemia due to a possible infection. Therefore, the patient was prescribed broad-spectrum antibiotics and salbutamol nebulization before the commencement of the oral rehabilitation phase.[10]

The effects of the deformities in patients with HED are not only limited to physical form and function but also affect their psychological well-being. In the years of development, a child's self-esteem is much dependent on their perception of their attractiveness, physical presentation, intellectual capability, and social acceptance. It has been reported that a child's self-esteem begins to take shape by about 4 years of age.[3] The paucity of hair and teeth have an unattractive appearance, thus, leading the patients with HED to not get accepted by other children. The child develops a very low self-image owing to social isolation, physical defects, and impaired speech. Therefore, dentists have a crucial responsibility to rehabilitate patients with HED as early as possible. Nowak stated that pediatric dentists are the most suitable professionals for the management of patients with HED.[3]

Although an optimal time for beginning the treatment has not been clearly defined, Till and Marques recommended that a temporary prosthesis must be provided when the child starts going to school. Removable partial dentures and overlay dentures are generally considered the treatment of choice for the replacement of missing teeth. They are affordable and can be easily adjusted by relining to match the patient's changing oral anatomy.[5] The use of implants is not indicated in patients with HED, because of incomplete skeletal growth, less amount of bone available, and increased chances of infection which may lead to implant failure.[11] Previous reports using Cephalometric analysis have demonstrated favorable growth of the maxillary and mandibular alveolar bone following the placement of dentures.[3] Therefore, a removable partial overlay denture was provided for the present case.

There is no standard treatment for the skin lesions, dermatitis, or periocular hyperpigmentation associated with ED. An increased risk of developing melanoma is also associated with hyperpigmentation and other skin lesions.[10] The patient was recommended to undergo a full physical examination at least once a year, and report immediately in case of a suspicious lesion. Iron and zinc supplementation was started for three months.

The absence of sweat glands leads to a defective thermoregulatory dermal function. Generally, this results in complications in patients with HED, particularly in neonates. The patients may experience headaches, nausea, vomiting, dizziness, excessive tiredness, and muscle cramps.[10] Thus, the patient was advised to avoid visiting regions with high temperatures and refrain from activities requiring severe physical exertion. The patient's family was aware of the various techniques for reducing his body temperature should the need arise, including immersion in water, the application of a sponge, and air conditioning. Water sports were recommended for the maintenance of the physique.

4. Conclusion

HED is a rare yet dreadful condition that can drastically affect the patient's quality of life. A multidisciplinary approach involving the pediatrician, dermatologist, psychiatrist, and pediatric dentist is essential for the management of patients with HED. Timely rehabilitation of the oral cavity plays a pivotal role in restoring the masticatory function, aesthetics, and speech of the patient, thus improving the self-esteem and overall quality of life of the patient.

5. Conflict of Interest

None.

6. Source of Funding

None.

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