Case Report

Building self-esteem by prosthetic rehabilitation in a child with hypohidrotic ectodermal dysplasia, role of pediatric dentist: A case report

Amit Khatri[1], Namita Kalra[2], Rishi Tyagi[3], Neetu Garg[4], Puja Sabherwal[5], Mayank Sharma[5]

From [1]Professor, [2]Professor (SAG) and Head, [3]Professor (SAG), [4]Post Graduate Student, [5]Senior Resident, Department of Paedodontics and Preventive Dentistry, University College of Medical Sciences and GTBH, New Delhi, India

ABSTRACT

Ectodermal dysplasia (ED) is a congenital disorder exhibiting multiple disorders that affect ectodermal tissues. Over 150 different presentations of the syndrome have been reported, of which the most commonly encountered are hidrotic and hypohidrotic variants. In the present paper, we report the case of an 8-year-old male who was diagnosed with hypohidrotic ED by a physician. The extraoral and intraoral findings were recorded and found in accordance with the diagnosed variant. A comprehensive therapy was initiated which included child and parent counseling, familiarizing the child with dental setup, and delivery of a removable partial denture for the upper arch and a removable complete denture for the lower arch. The importance of follow-up and newer dentures as per the growth of the patient was also explained to the parents. The article highlights the key role of a pediatric dentist in managing the child and parents in syndromes as such.

Key words: Ectodermal dysplasia, Hypohydrotic ectodermal dysplasia, Prosthesis, Rehabilitation

Ectodermal dysplasia (ED) is a congenital, diffuse, and non-progressive heterogeneous group of disorders that are derived from embryonic ectoderm and result from the abnormal development of two or more tissues at a time [1]. First reported in 1792 by Danz, the disorder was identified in the literature again when Wedderburn described 10 similar cases to Charles Darwin in a letter [1]. Weech coined the term inherited ED and proposed the term "anhidrotic" for cases in which sweating was completely absent [2]. Later on, Felsher altered the term anhidrotic to hypohidrotic in 1944 as the idea of complete absence of sweat glands was challenged [2,3]. Today, the prevalence of X- linked ED is reported to be one in 10,000 to one in 100,000 male births and the mortality rate is up to 28% in males up to 3 years of age and 17.3 in 100,000 female births [4]. The most commonly encountered X-linked variant, that is, recessive hypohidrotic ED (HED) (EDA or Christ-Siemens-Touraine syndrome) is caused by a mutation in the EDA gene that encodes for the ectodysplastin protein, a member of the TNF family which is directly related to the development of skin and other ectodermal tissues [5,6]. Hidrotic ED, the autosomal dominant variant, is due to mutation in GJB, which encodes for connexin 30, a critical element of intercellular gap junctions [7].

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CASE REPORT

An 8-year-old male reported to the Department of Pedodontics and Preventive dentistry with a complaint of inability to eat due to the lack of teeth. On recording medical history, the patient was found to be a known case of HED. No history of ED was obtained in the last three maternal or paternal generations. The parents reported having a non-consanguineous marriage. The present visit was recorded as the child's first appointment at any dental clinic/hospital. Drug history was irrelevant. The child was classified as Frankel 3 on behavior rating (Fig. 1a).

General examination revealed normal vitals but a mild rise in body temperature (99.4° Fahrenheit). The height and gait were normal for the age group as per the World Health Organization (WHO) standards. The patient was, however, underweight (18 kg) which could be related to the nutritional deficiency arising because of compromised masticatory functioning. Palmoplantar keratosis was observed alongside very coarse skin (Fig. 1c and d). Minimal sweating and salivation (frequent drying of mouth) were reported by the parents. On extraoral clinical examination, alopecia, scanty eyebrows and eyelashes, prominent frontal bossing, and the depressed nasal bridge were noted along with full and everted upper and lower lips (Fig. 1a and b). Intraoral examination under natural light

Correspondence to: Dr. Neetu Garg, Department of Paedodontics and Preventive Dentistry, University College of Medical Sciences and GTBH, New Delhi, India. E-mail: neetugarg11@gmail.com

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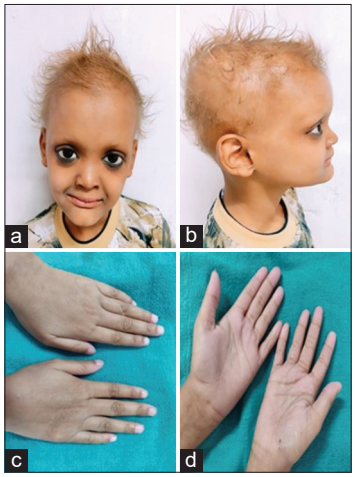

Figure 1: (a) Extraoral frontal view; (b) Extraoral sagittal view; (c) Extensor surface; and (d) Flexor surface of palms

revealed complete anodontia of primary teeth and hypodontia of permanent teeth. The alveolar ridges were knife-edged and grossly ill-formed (Fig. 2b-d).

Orthopantomogram (OPG) showed complete mandibular anodontia and maxillary hypodontia with both permanent first molars erupted and two-thirds of root formation completed. Tooth buds of both permanent maxillary second molars were also noted (Fig. 2a). Functional activities such as speech and mastication were partially impaired due to missing teeth and underdeveloped jaws. Parents reported decreasing interest shown by the child to attend school and social affairs due to his unique appearance and speech defects.

A comprehensive approach was opted for managing the case. Child and parental counseling were done at every visit. The child was accompanied by either of the parents during recall visits. A removable complete denture was planned for the mandibular arch and the partial denture for the upper arch with a decision to save the natural teeth. The first visit involved familiarizing the child with the dental operatory and a primary impression of both arches (Fig. 3a and b). The second visit was conducted after 3 days when a secondary impression using a custom tray and border molding was done (Fig. 3c-f). The child was cooperative throughout the procedure. A wax trial was done during the third visit (Fig. 3g and h - 4a and b). After which, the final acrylic denture was delivered (Fig. 4c). The speech was checked along with a social smile (Fig. 4d and e). Three-month and 6-month follow-ups over teleconsultation and a 14-month follow-up in the department were done. The child presented with coordination in school teaching activities and improved grades at school. The child became more interactive, playful, and lively (Fig. 4f and g).

The parents were given proper instructions for maintaining the dentures and oral health of the child. Information was provided regarding renewing the denture from time to time as per the jaw growth of the child as well as implant-based fixed prosthesis when the individual is of age. A monthly visit was recommended to check for lesions and other abnormalities.

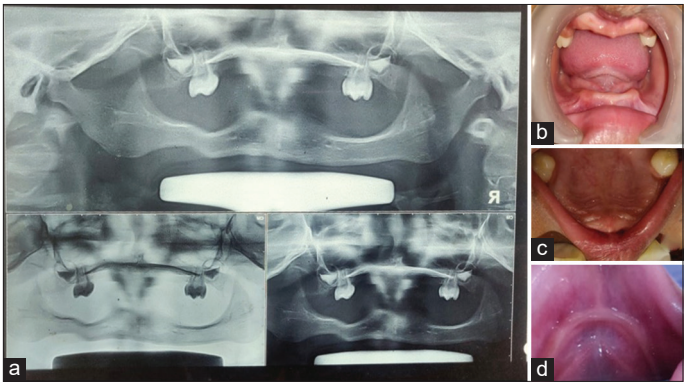

Figure 2: (a) Orthopantomogram for diagnostic evaluation; (b) Intraoral buccal view; (c) Maxillary, and (d) Mandibular occlusal view

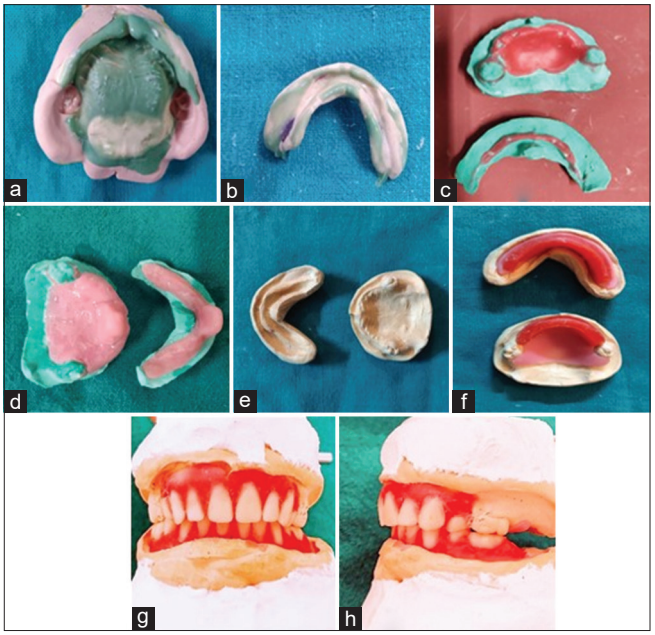

Figure 3: (a) Maxillary and (b) mandibular arch pick-up impressions; (c) Spacer and (d) custom tray fabrication; (e) Master cast preparation; (f) Occlusal rims for jaw relations; (g) Buccal, and (h) Occlusal view of teeth-setting arrangement for try-in

DISCUSSION

Hypohidrotic or anhidrotic ED, being the most common amongst this large group of hereditary disorders, affects one in seventeen thousand people globally. The cardinal features of classic hypohidrotic ED are hypohidrosis (reduced ability to sweat), hypotrichosis (sparse hair on scalp and body), and hypodontia (congenital absence of teeth) [1,6].

Mutations in the DL gene, which encodes for the EDA receptor are the primary cause of autosomal dominant and autosomal recessive HED [8]. Pathogenic changes in any of these genes can generate a clinical phenotype that is comparable to XLHED. Nearly half of the disorders previously categorized as EDs have a genetic cause, and the causative genetic changes underlying most of the more common ED conditions are now known [9]. ED is known to be inherited through all Mendelian modes of inheritance, as well as by spontaneous mutations [10].

Figure 4: Try-in records- (a) frontal and (b) sagittal extraoral views; (c) Final denture intraoral view; Extraoral (d) frontal view and (e) sagittal view; (f) 14 months follow-up; (g) Child with raised self esteem

The disorder is most likely to manifest in the first trimester of the gestational period but in severe cases of ED, the onset occurs before the 6th week of embryonic life as a result of which both primary and permanent dentition gets affected [2]. However, the complete absence of deciduous teeth is a rare finding [11].

On the basis of histologic analysis of fetal skin collected by second-trimester fetoscopy-guided skin biopsy, prenatal identification of this syndrome has previously been documented in high-risk pregnancies. With the use of chorionic villi in the first trimester, DNA-based linkage analysis has also made the diagnosis possible [12]. Postnatal diagnosis is primarily made by the presentation of symptoms and familial history. In patients having oral implications of ED, quality of life gets largely affected. Studies by Jabarifar *et al*. (2009) and Farrazzano *et al*. (2020) clearly state that oral health-related quality of life was significantly improved after full mouth rehabilitation under general anesthesia in pediatric patients [13,14].

Systemic management of ED depends on its manifestations. In the above case report, the patient was advised of frequent consumption of cool fluids for adequate thermoregulation and hydration since the temperature of the body was slightly elevated and the acrylic dentures were provided to aid the child in his daily life activities. A partial or complete denture in cases of anodontia or hypodontia is frequently the most accepted first line of treatment option for patients with ED and is indicated for children aged two or three who are capable of coping with the treatment [15]. Some clinicians recommend starting the prosthetic treatment when a child is 3–4 years old, while others argue that the first prosthesis should be supplied to the child before he or she starts elementary school to allow for adequate adaption. Another concern with denture providing in HED patients is developing resistance to anteroposterior and lateral displacement, which is exacerbated by underdeveloped tuberosities and ridges, as well as dry mucosal tissues [16]. In the reported case, the prosthetic treatment and counseling had a noticeable impact on speech, mastication, and esthetics as well as the child's social presentation. He had become more interactive with significant improvement in his grades at school.

CONCLUSION

A pediatric dentist not only diagnoses and provides appropriate age-based treatment to patients but also helps in building up self-esteem. The present article concludes that in syndromic patients like ones with ED, the role of a dentist is a multiverse. Prosthetic treatment begins at a very early age that also requires behavior guidance and continues to adulthood wherein the final therapy involves implant-based fixed prosthesis.

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