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Case Report

Ectodermal Dysplasia-a report of two cases

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Abstract

Ectodermal dysplasia is a large hereditary group of disorders which is usually manifested as X-linked recessive Hypohidrotic Ectodermal Dysplasia (HED) and has a full expression in males, whereas females show little to no signs of the disorder. It is a genetic disorder affecting the development or function of the teeth, hair, nails and sweat glands. The two most common types of Ectodermal Dysplasias are the X-linked recessive Hypohidrotic Ectodermal Dysplasia (Christ-Siemens-Touraine syndrome), and Hidrotic Ectodermal Dysplasia (Clouston syndrome). Hypohidrotic ectodermal dysplasia is characterized by hypodontia, hypotrichosis and hypohydrosis. Here in, we report case series of this rare entity of ectodermal dysplasia in a 15year old female and 42 years old male showing characteristic features of hypodontia, hypohidrosis, hypotrichosis.

Keywords: Ectodermal dysplasia, Early Diagnosis, Hypotrichosis, Oligodontia, Hypohidrosis

Introduction

Ectodermal dysplasia is a congenital, diffuse Non -progressive hereditary disorder. First case of Ectodermal Dysplasia was reported by Thurman. [1] The triad of nail dystrophy, alopecia or hypotrichosis and palmoplantar hyperkeratosis is usually accompanied by a lack of sweat glands and a partial or complete absence of primary and/ or permanent dentition. Ectodermal dysplasia represents a large andcomplex group of diseases comprising more than 170 different clinical conditions.[2] The incidence of this condition is 1:100,000.[3]Most patients with Ectodermal dysplasia (EDA) have a normal life expectancy and normal intelligence

CASE SERIES

Case 1: Female patient of age 15 yrs was reported to the Department of Oral Med-icine and Radiology (Vishnu Dental College Bhimavaram , Andhra Pradesh, India) with a chief complaint of missing teeth . Patient reported missing teeth since birth. There is no relevant medical history as reported by the patient. Patient visited local dentist for the same problem 3 months back but did not underwent any treatment. Detailed family history was parents. There were no abnormal findings reported among any of the siblings or near relatives. The lack of many of primary and permanent teeth in the oral cavity resulted in dietary problems. On general examination all the vital signs are in normal limit with no relevant medical history. Patient also complained of dry skin and absence of sweat in her skin and she was intolerable to withstand hot water and hot environment.

On extra oral examination, patient had scanty eyebrows and eyelashes (fig-1)along with frontal bossing , thinning of upper lip is also evident. Intraoral examination revealed the partial absence of primary and permanent teeth,(fig-2) thin alveolar ridges, reduced vertical bone height, and loss of sulcus depth in the posterior regions of maxillary and mandibular jaws (fig-2) partial anodontia was noticed, there is grossly decayed teeth in relation to 65 .On soft tissue examination, an erythematous area is seen on hard palate in mid-palatine raphe region which is tender on palpation. Patient was subjected to further radiographic investigations (fig-2) Orthopantomograph (OPG) revealed generalized decrease in height of alveolar bone. Oligodontia was observed. Based on correlation of clinical and radiographic findings, a diagnosis of ectodermal dysplasia was made.

CASE 2: A 42 year male patient was reported to our out patient department (Vishnu dental college, Bhimavaram, Andhra Pradesh, India) with a chief complaint of missing teeth . Patient reported missing teeth since birth. Patient gave history of frequent fevers in childhood. Patient visited dental office one year regarding same problem but no treatment was done. Detailed family history was taken which revealed that he is the only child of his parents There were no abnormal findings reported among any of the siblings or near relatives. The lack of primary and permanent teeth in the oral cavity resulted in dietary problems. On general examination all the vital signs are in normal limit with no relevant medical history. Patient also complained of absence of sweat in his skin and he was intolerable to withstand hot water and hot environment.

On extra oral examination, patient had scanty eyebrows and eyelashes (fig-3) along with frontal bossing. Intraoral examination revealed the partial absence of primary and permanent teeth, thin alveolar ridges, reduced vertical bone height, and loss of sulcus depth in the posterior regions of maxillary and mandibular jaws. Patient was subjected to further radiographic investigations. OPG revealed generalized decrease in height of alveolar bone extending about 6-7cm below the cemento-enamel junction suggestive of generalized bone loss. (fig-4). Partial anodontia of permanent and Deciduous teeth was found. Based on correlation of clinical and radiographic findings, a diagnosis of hypohidrotic ectodermal dysplasia was made.

All primary teeth were extracted and patient was sent to department of Prosthodontics for fabrication of complete denture. Routine procedures were followed for the construction of the dentures. Preliminary impressions were made with irreversible hydrocolloid, and then custom trays were prepared for functional impression. Acrylic bases with wax rims were made on the master casts in order to establish maxillo-mandibular relations. After making the maxillo-mandibular records, the casts were mounted in an articulator. Over denture was fabricated. After the final insertion, routine hygiene instructions for the dentures were given to the patient (fig-5).

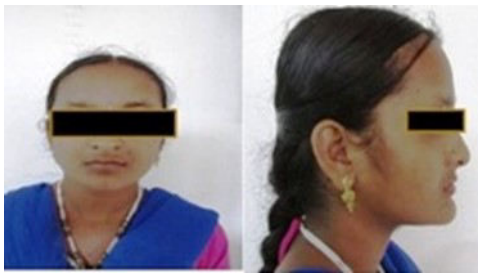

Fig 1: Case-1 profile and lateral view

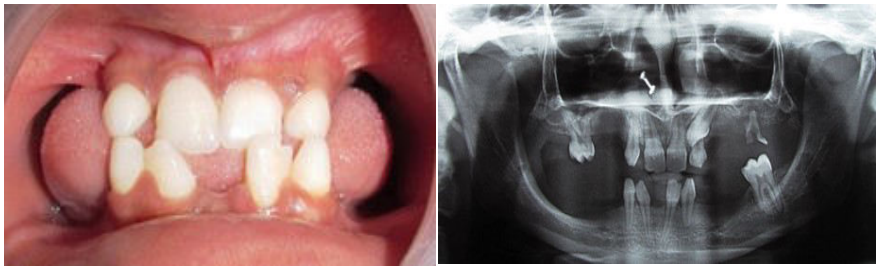

Fig 2: Case-1 Intraoral and Orthopantamograph

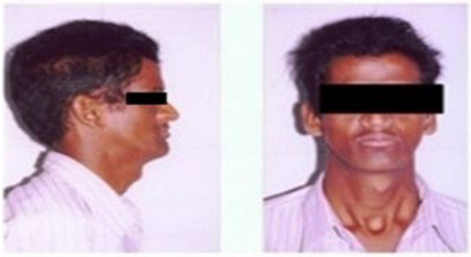

Fig 3: Case-2 profile and lateral view

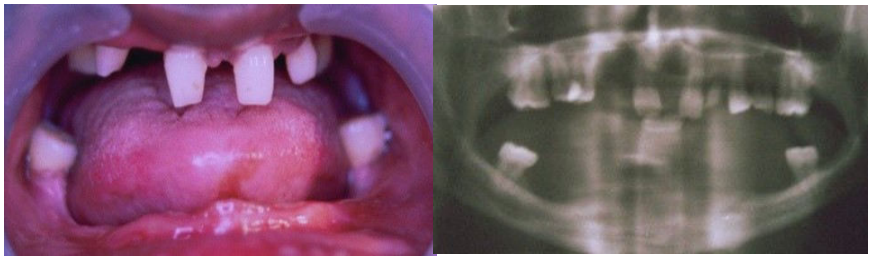

Fig 4: Case-2 Intraoral and Orthopantamograph

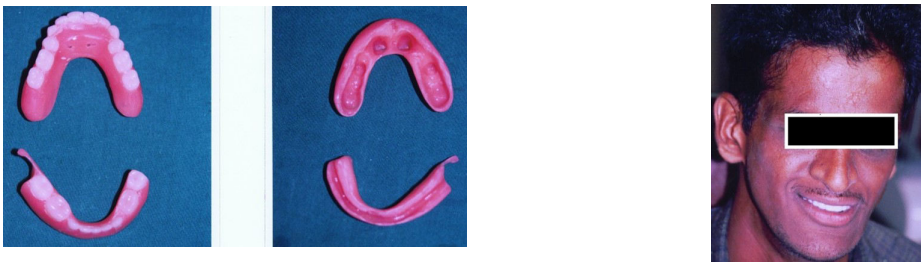

Fig 5: Case-2 Denture fabrication and post operative picture

Discussion

Freire-Maia and Pinheiro proposed the first classification system of the ectodermal dysplasias in 1982 [4].and provided additional updates in 1994 and 2001[5] The patients were classified into subgroups which were based on the presence or absence of the following ie trichondysplasia (abnormal hair), abnormal dentition, onchondysplasia abnormal nails) and dyshidrosis (abnormal or missing sweat glands) [6].

Hypohidrotic Ectodermal dysplasia (HED) is usually transmitted as an X-linked recessive trait in which the gene is carried by the female and manifested in male. In X -linked form carrier mothers exhibit minimal expression in the form of hypodontia or conical teeth and spottily reduced sweating. The unaffected female has 50% chance of transmitting this disorder to her male children and each female offspring has 50% chance of inheriting the defective gene, thereby being a carrier. Spontaneous gene mutation is possible and may occur in family without any previous history of the syndrome [5]. The prevalence in the population has been assessed as between 1:10,000 and 1:100,000 male live births. The differences were enlisted in (Table 1). The different types of Ectodermal Dysplasias are caused by the mutation or deletion of certain genes located on different chromosomes. Because ectodermal dysplasias are caused by a genetic defect they may be inherited or passed down the family line. In some cases, they can occur in people without a family history of the condition, in which case a *de novo* mutation would have occurred. Mutations in the EDA, EDAR and EDARADD genes cause HED. EDA is the only gene known to be associated with X-linked HED (XLHED). Ninety-five percent of individuals with HED have the X-linked form. The genes EDAR and EDARADD are known to be associated with both autosomal dominant and autosomal recessive forms of HED. Mutations in these genes account for 5% of HED [7].

Because of partial or complete absence of sweat and sebaceous glands, the phenotype has smooth, soft, dry and thin skin. Fine linear wrinkles and increased pigmentation are often present around the eyes and mouth. There may be hyperkeratosis of the palms of the hands and soles of the feet. There is absence of long hair, although beard is usually normal, auxiliary and pubic hair is generally sparse. The hair over the scalp is often blonde, fine, stiff and short. While the eyebrows and eyelashes are frequently missing. The nails appear to be normal or somewhat spoon shaped. In females, the mammary glands are aplastic or hypoplastic.

Impaired lacrimal gland function and occasionally glaucoma has been reported. Also there can increased susceptibility to allergic disorders, such as asthma or eczema. Other manifestations include variations in the shape of the skull which can resemble inverted triangle. The face looks smaller because of the frontal bossing and depression of the nasal bridge [8] .

Oral manifestations include oligodontia of the primary and permanent teeth. However, congenital absence of the primary teeth is relatively rare. Teeth that are present have abnormal crown form. Teeth in anterior region of the maxilla and mandible are conical shaped [7]. The number of missing teeth varies with higher incidence in mandible. In the primary dentition, maxillary 2nd molars, canines, central incisors and mandibular canines are the teeth commonly present.[5]Van Ramos et al. observed complete anodontia, reduced vertical dimension and overclosure in their patient [9] also reported a case of complete anodontia [10] Children with Ectodermal Dysplasia show maxillary retrusion due to sagittally underdeveloped maxilla, forward and upward displacement of mandible and collapsed anterior facial height. The palatal arch is frequently high and cleft palate may be present [4].

Diagnosis is based on the episodes of hyperpyrexia, lack or type of the hair, absence of teeth and tooth buds and tooth morphology. Peeling of the skin at birth, eczema, asthma, and frequent respiratory infections may be additional clues. Other diagnostic criteria have been dermatoglyphic analysis, characteristics of lacrimal secretion and distribution and pattern of scalp hair. Missing teeth can also be an important sign. Patients with ectodermal dysplasia associated with immunodeficiency may have hypogammaglobulinemia with impaired lymphocyte proliferation and cell-mediated immunity.

Dental management include fabrication of dentures, and as the child with ectodermal dysplasia grows, the denture will be modified or replaced. During the mixed dentition stage, the prosthesis will require modification to accommodate loss of exfoliated primary teeth and appearance of newly erupted permanent teeth. During the permanent dentition, the removable prosthesis may be replaced by fixed depending on the number and position of the teeth. Before providing the patient with the dentures, the tone of the perioral muscles can be trained by vestibular shield. Studies have shown that early prosthetic treatment helps in establishing normal craniofacial growth. It also improves harmonization of the position of the lips in relation to nose and chin. The other treatment option is the use of implants. Use of implants in adult pat-

patients is well-documented in literature. However, when placement of implants is the treatment of choice, bone grafting and sinus membrane elevation can be considered to facilitate placement [7].

In present cases, male patient was provided complete denture .Female patient was advised for oral prophylaxis and fabrication of complete denture prosthesis. These patients have to undergo regular dental checkups. Relining or change of dentures should be done every 1 or 2 years.

Conclusion

Patients afflicted with a hereditary deformity, such as ectodermal dysplasia suffer from poor psychological and physiologic development as a result of unacceptable esthetics and abnormal functions of orofacial structures. Optimal treatment requires a multidisciplinary collaborative efforts pediatric professionals, psychologist, ENT specialist and speech therapist. Dentists have responsibility to rehabilitate these patients to improve appearance, mastication and speech. Dental treatment depends on severity of the disorder .Global self-esteem is highly determined by assessment of one's own physical presentation, as well as comparison with the attractiveness, ability, intellectual skills and social acceptance of other people. Unusual facial features exacerbate the social challenges of meeting new people. Lowered self-esteem, speech defects, decreased academic performance and social isolation may result from merely looking different from one's peers. Therefore, cosmetic and prosthodontic measures should be instituted as early as possible.. It is also important to instill awareness among parents regarding early management. Since it is an hereditary disorder, research should be done to check any abnormality prenatally in mother's womb and procedures to correct abnormality. Since these patients are hypohidrotic extensive care should be taken to their skin in tropical countries like India.

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