Research Paper

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Christ –Siemens- Touraine Syndrome : A Rare Case Report

Medical Science

KEYWORDS : Hypohydrotic ectodermal dysplasia, Hypodontia, Oral Rehabilitation, Complete Denture

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ABSTRACT

Ectodermal dysplasia is a genetic disorder in which there are congenital birth abnormalities of 2 or more ectodermally derived structures which include teeth, hair,nails and sweat glands.Hypodontia of the primary and permanent dentition is the most common oral finding.Therefore,affected patients need dental prosthetic treatments during their developmental years.The management of ectodermal dysplasia is complicated due to wide array of dento-facial defects and because the individuals are quite young when they are evaluated for treatment.

SUMMARY:- This article reports a clinical case of a child affected by ectodermal dysplasia with hypodontia & reviews different dental treatment options for such patient.

INTRODUCTION

Ectodermal dysplasia (ED) is a congenital syndrome characterized mainly by tissue abnormalities of ectodermal origin,namely skin, nails, hair and teeth [1] . They constitute a wide spectrum of congenital diseases that was first described by Thurman [2,8]. The etiology of ectodermal dysplasia lies in genetic anomalies, which can be inherited through either parents or manifested via gene mutations [3]. It displays an autosomal dominant, autosomal recessive or X-linked pattern of inheritance[4,5]. Although more than 170 different subtypes of ectodermal dysplasia have been identified, these disorders are considered to be relatively rare with an estimated incidence of 1 case per 100,000[6].

ED can be classified into hypohidrotic,in which the sweat glands are absent or significantly decreased; and hidrotic, in which the sweat glands are normal [7].(1) Hypohidrotic or anhydrotic (Christ-Siemens-Touriane syndrome) in which sweat glands are either absent or significantly reduced in number; (2) Hydrotic (Cloustone syndrome) in which sweat glands are normal.Dentition and hair are involved similarly in both types but hereditary patterns of nails and sweat glands involvement are different[2,9].Hypohidrotic ectodermal dysplasia is the more severe form of the disease and is associated with sensitivity to heat, frequent high fevers and dento-facial defects. It is the most frequently reported ED variant with an X-linked recessive inheritance gene mapped to Xq12- q13. [4, 5]. HED affects men more severely and frequently,whereas female heterozygotes present minor defects [2] .

Its characterstic symptoms which leads to diagnosis include hypodontia (80%), [7, 12] anodontia or oligodontia (reduced number of teeth) with conically shaped teeth, hypohidrosis (decreased perspiration) with concomitant hyperthermia caused by a lack of sweat glands and hypotrichosis (sparse hair) [11] .

Case report

A 8 year old boy reported to the Department of Oral Medicine, Jaipur Dental College with chief complaint of missing teeth since childhood. He had a medical history of bald scalp, dry extremities and intolerance to heat. Hair growth in head region had started only 3 years back.In family history there was no record of any such abnormality among patients relatives/distant cousins.The child had kept a good oral hygiene.In general examination, the height and the weight of the patient was within the normal range.Extra oral examination revealed symmetrical facial profile, in posterior region of the head occipital bossing was present.Patient had depressed nasal bridge, pro-turbent lips,indistinct vermilion border, pronounced supra-orbital ridge, hypoplastic mid-face [fig 1 (A)] thin spars blonde hair and facial eyebrows (hypotrichosis) [fig 1(B,C,D)] and there was scaly skin over facial region. The lower extremities had smooth and dry skin indicating complete absence of sweat glands (hypohydrosis) and there was thick skin over palms with brownish pigmentation [fig 2].These findings matched typical features of hypohidrotic ectodermal dysplasia (Christ – Siemens – Touriane syndrome).The intra oral examination revealed resorbed upper and lower alveolar ridges, high arched palate, partially edentulous ridges (hypodontia), conical peg-shaped upper deciduous incisors, deciduous molars (55, 65) having an increased coronal diameter, reduced vertical bone height, loss of vestibular depth in the lower jaw.Thus, there were classical features of 3H – Hypotrichosis, Hypohydrosis, Hypodontia.

With the parents consent the patient was subjected for radiographic examination..The radiographic findings also confirmed the clinical diagnosis. Panoramic view and an Intraoral periapical radiograph for the upper deciduous incisors were taken for the patient.IOPAR revealed completely erupted conical shaped 51,61 (deciduous incisors) and unerupted permanent central incisors 11, 21 in the upper jaw. Alveolar area w.r.t. 51,61 indicated bone loss. Follicular space was present around unerupted permanent upper central incisors.Pulp chamber was wide open at periapex w.r.t. 51,61 [fig 3] OPG revealed partially edentulous maxillary region with completely edentulous mandibular jaw. There was decreased vertical bone height in respect to mandible and maxilla jaw and there was presence of deciduous tooth (51,61,55,65) [fig 4] in maxilla.

Treatment plan included psychological counselling for child and the parents, genetic counselling in order to ascertain any history of genetic abnormalities in the family.Patient was also advised for dermatological consultation.Complete prosthetic rehabilitation which involved a removable partial prosthesis in maxilla . Vestibuloplasty and ridge augumentation for mandible which was having severe hypoplastic ridges, to enhance the soft and hard tissue in lower jaw,for support of a removal complete mandibular denture.Oral prophylaxis and oral hygiene instructions were given to the patient and family members of the patients were advised to avoid direct sun exposure to the child and keep him in cold environment as much as possible.Parents were explained the benefits of implant once the patient completed 12 yrs of age.

Dental management of ectodermal dysplasia

The dental management of the ED patient requires knowledge of growth development, paedodontics, oral surgery, orthodontics & prosthodontics. Since ED patients usually present themselves at a very young age with a multitude of anomalies, a multidisciplinary approach is required [10].Clinicians have suggested that the child patient must have an initial prosthesis before he/she begins school so as to adapt and get accustomed to the prosthesis as well as for increase in self confidence of the patient in the social environment[7,14].

Behavioral management techniques like “tell-show- do” play an important role in conditioning young patients to conquer their

fear, anxiety and establish a trustworthy relationship with them [10,15,16]. Uncooperative young patients may need sedation for extensive prosthodontic procedures; however, Nowak stated that sedation compromises the patient's understanding and compliance, which are essential for the success of a prosthesis based treatment [10,13].

The oral rehabilitation of ED patients usually consists of complete or removable prosthesis in the development years, followed by a definitive prosthesis based on fixed partial dentures and/or endosseous implants,after the complete development of the alveolar process [10].

Removable prosthodontics is a common mode of treatment in ED as such patients are often associated with anodontia or hypodontia [10].Complete denture treatment is satisfactory for the functional and aesthetic rehabilitation of the patient but not in the case of severely hypoplastic ridges which may require vestibuloplasty and ridge augumentation[10,13,17].

Fixed prosthodontic treatment may be initiated for older patients but as most of the ED patients are quiet young, it should be avoided[10].

Dental implants are increasingly being used in the management of ED [7,18].However, concern has arisen regarding the placement of osseointegrated implants in developing alveolar bone [19]. Implants placed in young ED patients may get submerged due to the continuously growing alveolar process and investing tissues [7].The growth of the adjacent natural teeth relative to the implant supported prosthesis may cause infraocclusion over a period of time,which would require frequent remakes to correct the plane of occlusion or distraction osteogenesis for repositioning the submerged implant [7,20].The submerged implants may be redisposed to peri-implantitis and the inadequate crown/implant length ratios may contribute to increased horizontal forces [20].In case of severe bone atrophy,implant placement may not be possible without bone grafting [3]. Consequently,clinicians have contraindicated dental implants for children upto 6 yrs of age [11]. Dental implant treatment may be considered in situations of anodontia as a method of preserving the residual alveolar ridge [10, 28-31] and in patients above 13 yrs of age as most of the alveolar bone growth would have been completed by then [10,25].

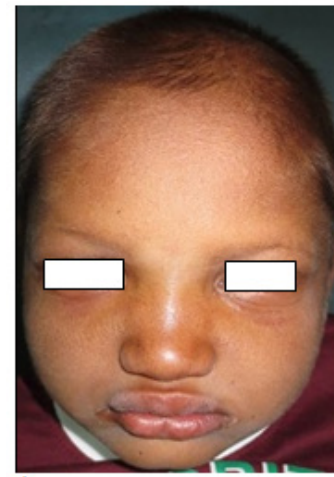
Clinicians have initiated prosthetic treatment for ED patients at the age of 5 yrs stating that it ensures functional, phonational, psychologic and esthetic rehabilitation of the child [7].Initiating prosthodontic treatment at an early age enhances masticatory muscles tonicity, delays alveolar bone resorption associated with the absence of teeth, compensates for the decrease in vertical dimension and prevents angular cheilitis [19] Early complete dentures treatment can lead to significant improvements in mastication, appearance, speech and satisfactory diet for the child.It instills self-confidence in the child, which is essential for normal psychological and social development [12].Failure to initiate complete denture therapy in young ED patients may cause reduction in the height of the lower third of the face, upward and forward displacement of the chin due to antero-rotation of the mandible and a predilection to Class III malocclusion [2,19].

Periodic recall and maintenance appointments in young ED patients are important as prosthesis modification or replacement will be needed due to continuing skeletal growth and development [10,13,17,27].

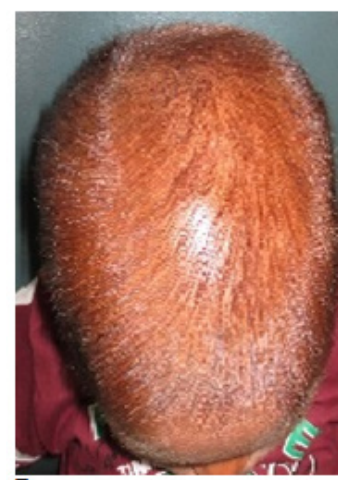
Conclusion

Management of clinical manifestations associated with ectodermal dysplasia presents a unique challenge for prosthodontists and pedodontists. Treatment of young edentulous patients with removable partial or complete denture is an acceptable, available and cost effective modality, which improves function,speech, esthetics and psychosocial condition. However, its long-term success depends on regular recall appointments and meticulous maintenance of oral and prosthetic hygiene.

LEGEND



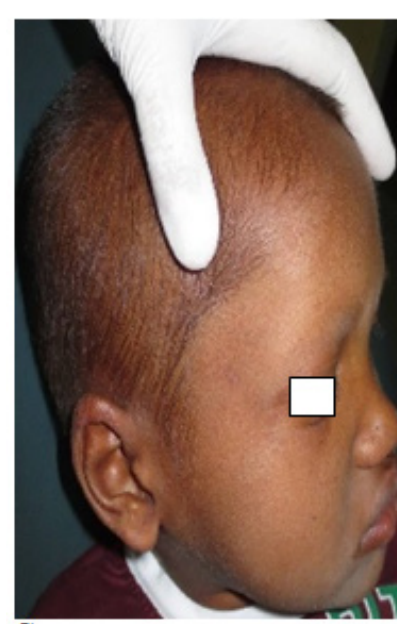
A



B

FIGURE 1:- 8 year old boy with hypohydrotic ectodermal dysplasia:

A. Frontal view B. Cranial view



C

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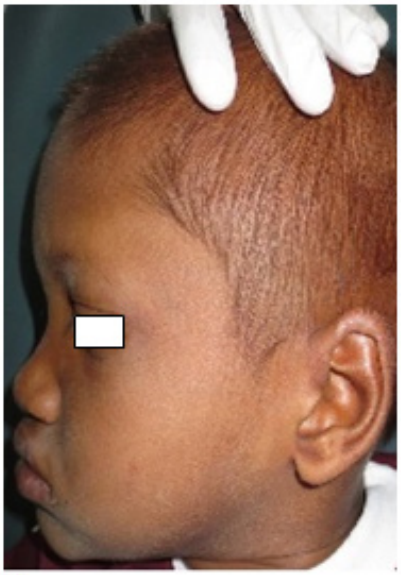

D

C & D. Profile View FIGURE 1

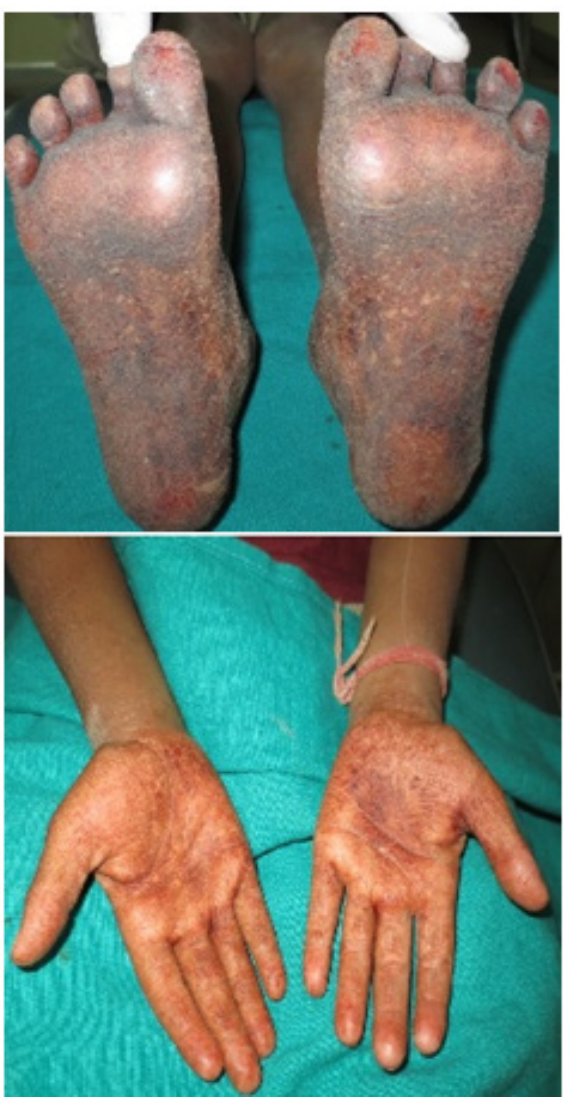

FIGURE 2:- Thick skin with brownish pigmentation over extremities

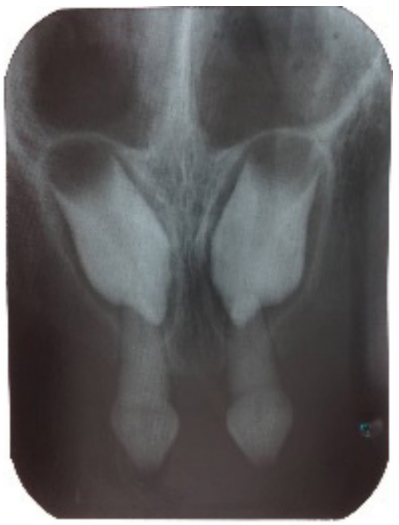

FIGURE 3:- Intraoral periapical radiograph

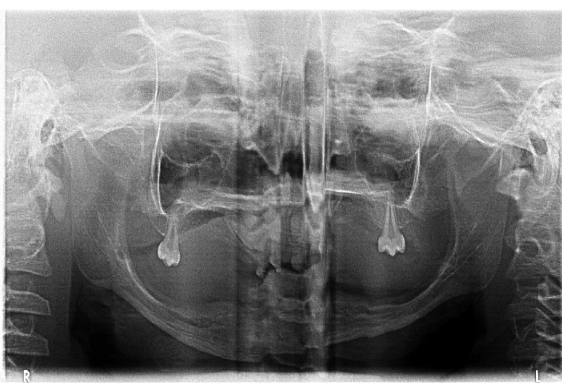

FIGURE 4:- Orthopantomograph

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