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FROM DEVELOPMENTAL DEFECT TO FUNCTIONAL DENTITION - A PEDIATRIC REHABILITATION JOURNEY

Dr. Sunil Kumar Mahadev[1], Dr. Sherin Arulraj[2], Dr. Vignesh Guptha Raju[3], Dr. P. Mahesh Kumar[4], Dr. Sudhakar S[5]

[1]MDS.,Professor, Department of Pediatric and Preventive Dentistry, Karpaga Vinayaga Institute of Dental Sciences, Chinnakolambakkam, Tamil Nadu, India, Mail: drmsk89@gmail.com, Ph .no:9962783848
[2*]MDS.,Post Graduate Student Department of Pediatric and Preventive Dentistry, Karpaga Vinayaga Institute of Dental Sciences, Chinnakolambakkam, Tamil Nadu, India.Orchid ID: 0009-0000-4095-9302, Mail: sherinarul0104@gmail.com, Ph.no:8124420360.
[3]MDS., PHD.,Professor,Department of Pediatric and Preventive Dentistry, Karpaga Vinayaga Institute of Dental Sciences, Chinnakolambakkam, Tamil Nadu, India.Mail: vigneshguptha88@gmail.com, Ph .no: 9840485769
[4]MDS.,Professor, Head of The Department, Department of Pediatric and Preventive Dentistry, Karpaga Vinayaga Institute of Dental Sciences, Chinnakolambakkam, Tamil Nadu, India. Mail: drmahesh1972@gmail.com, Ph .no:9841397411
[5]MDS.,Senior Lecturer, Department of Pediatric and Preventive Dentistry,Karpaga Vinayaga Institute of Dental Sciences, Chinnakolambakkam, Tamil Nadu, India, Mail: dr.sudhakar.0011@gmail.com, Ph .no:8760136632

ABSTRACT:
The Ectodermal dysplasia (ED) refers to a range of hereditary disorders that impact ectoderm-derived tissues, including sweat glands, teeth, hair, and nails. This syndrome is characterized by dental abnormalities such as hypodontia, anodontia, and conical-shaped teeth. The aim of this case report is to discuss the management of ectodermal dysplasia and clinical developments. The pediatric case of hypohidrotic ectodermal dysplasia of five-year-old boy is presented in this case report; has unique general features, functional and oral symptoms. The fixed partial denture provided for the missing teeth in upper arch and removable partial denture has been given to the lower arch. The aim of the case report is to discuss child unique, interdisciplinary dental care techniques, radiological characteristics, and clinical presentation and multidisciplinary dental management. To improve oral function, appearance, and psychosocial results, early diagnosis and management are crucial.

KEYWORDS: ectodermal dysplasia, groper's appliance, hypodontia, hypohidrosis, removable partial denture.

INTRODUCTION

Ectodermal dysplasia's (ED) are a group of inherited disorders affect the growth or homeostasis of two or more ectodermal derivatives, such as teeth, hair and nails, and also other structures including glands, retina, cochlea, central nervous system (CNS), and the adrenal medulla [1]. The incidence of ectodermal dysplasia is estimated to be seven per 100,000 making it a very rare disease [2]. The two most prevalent forms of ectodermal dysplasia are hidrotic ectodermal dysplasia and X-linked recessive hypohidrotic ectodermal dysplasia (HED) [3]. The classic triad of hypohidrotic ectodermal dysplasia (HED) comprises sparse hair (hypotrichosis), malformed or absent teeth (hypodontia/anodontia), and diminished or absent sweating (hypohidrosis/anhidrosis) [4,5]. Oral manifestations frequently include diminished asymmetric alveolar ridge height, maxillary retrusion, enamel hypoplasia, irregularly formed teeth, partial or complete anodontia, and a high palatal arch [6]. The exact pathogenesis of ectodermal dysplasia and their molecular basis of the disorders are remain under investigation [7]. Diagnosis is typically made in associated with delayed tooth eruption and is generally made quite late due to HED-related oral abnormalities are very common. As with any genodermatosis genetic counseling for the affected individual and their family members is crucial to provide information regarding the likelihood of disease manifestation or reoccurrence [8,9].

Case Presentation

A five-year-old boy presented with the chief complaint of missing teeth, noted by his parents since he was one years of age. Parents reported that the child experienced impaired aesthetics and difficulty in mastication which necessitates adherence to soft food diet. Patient had a history of wheezing occasionally. No prior similar family history. The oral hygiene regimen involves brushing his teeth once daily. The general examination findings indicate that the patient's height was 56 cm, weight was 17 kg, posture was characterized by a forward head, build was moderate, and the patient was afebrile with a pulse rate of 128 beats per minute (Figure1). Patient had restricted mouth opening with 32 mm. The Intra oral features with soft tissue examination reveals that buccal and labial mucosa has no abnormality, gingiva was coral pink in color with firm in consistency and with irregular and round contour. Clinically, oligodontia, conical (peg-shaped) canines (53, 73, and 83), delayed eruption of teeth, underdeveloped alveolar ridges in both the maxilla and mandible, and reduced alveolar bone development associated with missing teeth were observed (Figure2). Hard tissue examination via an orthopantomogram (OPG) (Figure 3) confirmed clinical findings of hypodontia. Multiple permanent tooth buds were missing, and primary teeth 55, 65, 73, and 83 were retained. The OPG further revealed a collapsed bite, a narrow U-shaped arch form, and hypoplastic alveolar ridges with reduced bone height and width. An impacted canine (63) was also observed. Tooth development appeared delayed compared to chronological age. Targeted genetic testing performed at a

separate facility revealed autosomal recessive inheritance pattern, and no history of similar illness was reported among other family members. This finding was significant for genetic counselling, as it indicated a 50% likelihood of siblings being carriers and a 25% risk of recurrence in future offspring. The patient and parents were counselled regarding the hereditary nature of ectodermal dysplasia (Figure4, 5).

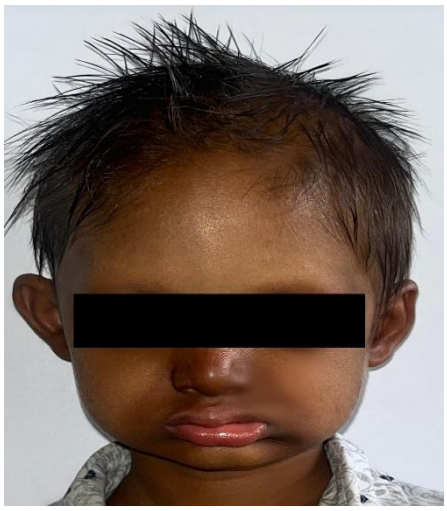

FIGURE 1: Extraoral Frontal View showing Sparse, fine, brittle scalp hair; absent eyebrows and eyelashes (alopecia); Frontal bossing (prominent forehead), Depressed nasal bridge, Thick, everted, protuberant lips, Large, low-set ears, diminished lower facial height.

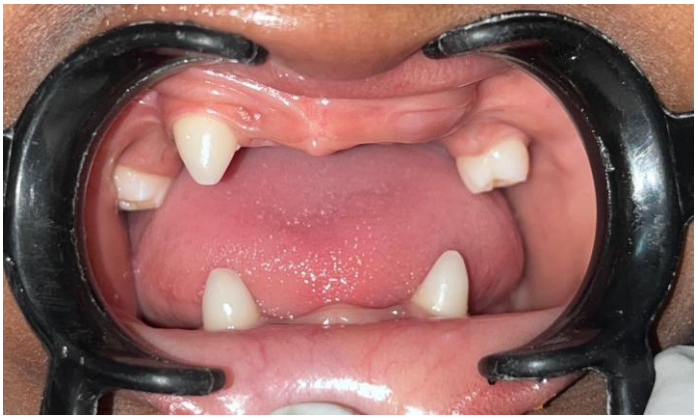

FIGURE 2: Intraoral findings which shows presence of teeth 55,53,65,73,83.

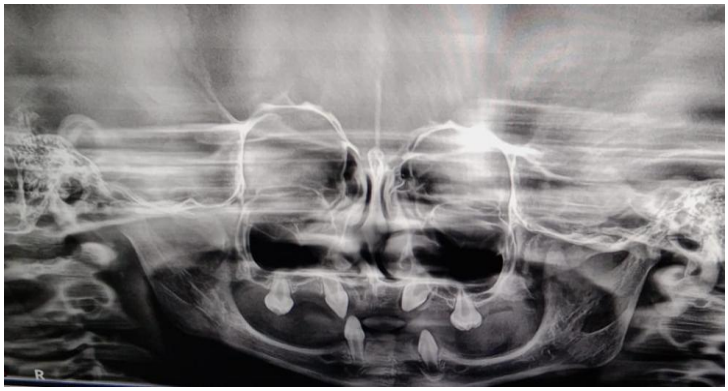

FIGURE 3: Orthopantomogram shows hypodontia of primary teeth, Impacted 63, complete absence of permanent tooth buds and hypoplastic alveolar ridges.

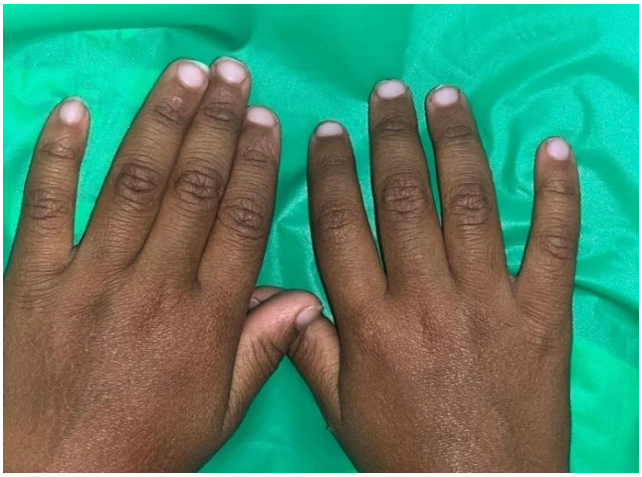

FIGURE 4: Cobblestone appearance of hand

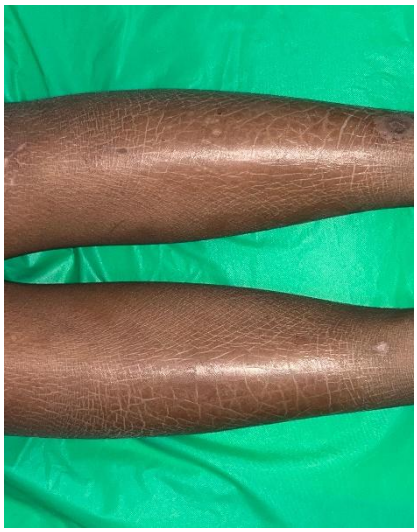

FIGURE 5: Cobblestone appearance of leg.

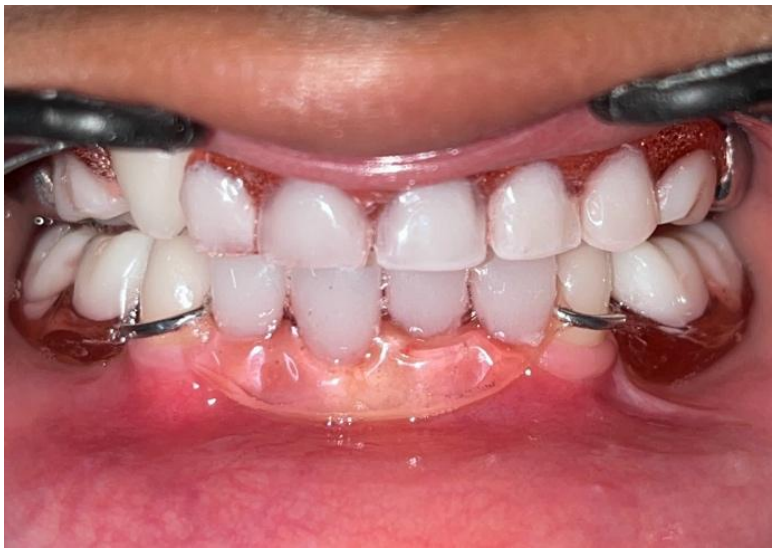

FIGURE 6: Intraoral Post-Operative Frontal View.

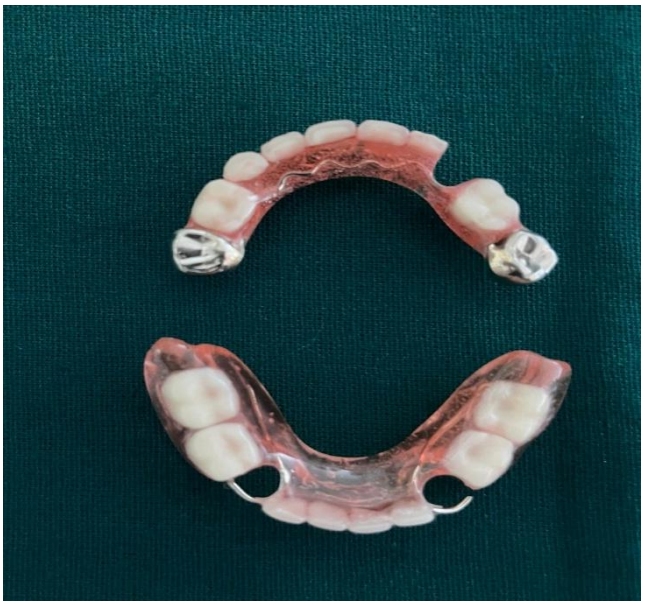

FIGURE 7: Figure showing Fixed Groper Appliance for the Maxillary arch and Removable Partial Denture with C-clasp in 73,83 for the Mandibular arch

Management:

During the initial visit, the consultation and proposed treatment plan were explained to the parents. The patient did not return for six months; upon their return, the treatment plan was reiterated, and both the patient and parents were motivated to proceed. Written parental consent and patient assent were obtained prior to commencing any procedures. Treatment initiated with tooth preparation and the placement of stainless-steel crowns on teeth 55 and 65 utilizing the Hall technique. Primary impressions of the upper and lower arches were recorded using silicone putty, followed by secondary impressions using a light-body addition silicone. Casts were poured, and acrylic teeth were arranged, processed, finished, and polished. For the maxillary arch, a fixed Groper appliance was fabricated and cemented to the stainless-steel crowns on 55 and 65. A wire framework provided support for the acrylic teeth, which replaced 54, 52, 51, 61, 62, 63, 64, and 65 (Figure 6). For the mandibular arch, a removable partial denture (RPD) was fabricated to replace teeth 85, 84, 82, 81, 71, 72, 74, and 75, utilizing C-clasp on teeth 73 and 83 for retention (Figures 7). Due to growth considerations, RPD is often selected for pediatric ED patients since children are still developing. Implants or fixed prostheses may impede the development of the jaw. As the child grows, the RPD can be altered or recreated to maintaining the remaining teeth. Many people with ED have few teeth therefore RPD aids in force distribution and tooth preservation. Poor Ridge Development makes fixed alternatives challenging. RPD provides greater flexibility, easy adjustment is possible. The RPD can be relined, repaired, expanded and useful throughout the time of mixed dentition.

DISCUSSION

The most prevalent kind of ED is called HED, and it manifests as defects in the growth of the exocrine glands, teeth, and hair [9]. In the literature, ED has been divided into over 190 subtypes, which are categorized according to the molecular pathway, kind of genetic mutation, or clinical characteristics. However, the two most prevalent forms of ED are hidrotic ED, in which the sweat glands are unaffected, and hypo hidrotic/anhidrotic ED, in which the sweat glands are inadequate [10]. Following a first dental examination, children with ED are identified when the eruption of their primary teeth is significantly delayed or when their first tooth erupts in an unusual shape [11]. Prosthetic rehabilitation accomplished timely helps affected children's social suffering and improves their maxillofacial development [12]. Groper's is regarded as a temporary cosmetic functional space maintainer for children with ED. It preserves arch integrity, prevents the migration of adjacent teeth, and maintains room for future prosthetic considerations. Assists in preserving vertical dimension. The maxillary anterior teeth are crucial for: Labiodental sounds (F, V), Sibilants (S, Z), Without anterior teeth: Lisping occurs, Air escape alters phonetics, Groper's restores phonetic guidance. Gropers can also be modified as patient jaw grows [13,14]. The Groper appliance was planned for this patient since patient had Cawood and Howell Class 4 classification that is thin, knife-edge alveolar ridges were seen. Since there was minimal bone support, lack of reliance on ridge anatomy gropers was planned as it does not rely on ridge anatomy for retention [15]. The Removable Partial Denture was planned for lower arch. since the patient had Cawood and Howell Class 5 classification. RPD restores facial height, replaces several teeth at once, and can be altered, relined, or recreated as jaws grow [16]. Child's psychological state is affected by the diagnosis, which is essentially clinical with a senile facial look. This Psychological well-being can be ensured by consulting with a psychologist, who can also provide psychological treatment if required. The care of the patient includes collaboration of dermatologist, ENT specialists, pulmonologists, and psychologists. otolaryngologist and a speech therapist are consulted to identify phonetic or articulation abnormalities [17- 20]. Thus, multidisciplinary collaboration is essential for the child's emotional development as well as functional rehabilitation. On 3 and 6 month follow up patient had ill fitted denture in lower arch which was corrected by adjusting c-clasp on either side. And we recommended the patient be followed up with every three months to monitor growth-related changes and to check the

canine eruption of 63 as it was impacted. And we have informed about the change that occur between the ages of 10 and 11, shedding of canine and molar and further upper and lower full dentures may be needed. After 18 years, depending on the width of the bone ridges we plan for an implant.

CONCLUSION

This case highlights the uncommon occurrence of hypohidrotic ectodermal dysplasia affecting multiple systemic and craniofacial structures in a pediatric patient. Timely and appropriate prosthetic intervention successfully improved the patient's masticatory function, perioral muscle tone, and basal bone growth trajectory. Beyond functional restoration, early intervention dramatically enhances nutritional intake, phonetic ability, and the child's self-esteem. Managing this disorder requires a dedicated, multidisciplinary approach to ensure optimal clinical outcomes, societal integration, and an improved overall quality of life.

Declaration of patient consent:
The authors certify that they have obtained all appropriate patient assent and parent consent forms. In the form the parent(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The parents/patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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REFERENCES

1. Dev A, Malhi K, Mahajan R. Ectodermal Dysplasia - An Overview and Update. Indian Dermatol Online J. 2024;15(3):405-414. doi: 10.4103/idoj.idoj_599_23.
2. Shamim H, Hanif S. Hypohidrotic Ectodermal Dysplasia: A Case Report. Cureus. 2023;15(10):e46530. doi:10.7759/cureus.46530
3. Albeik MT, Abdullah L, Almatroud MM. Hypohidrotic ectodermal dysplasia: a case report. Annals of Medicine and Surgery. 2023 Mar 1;85(3):561-4.
4. Visinoni AF, Lisboa-Costa T, Pagnan NA, Chautard-Freire-Maia EA. Ectodermal dysplasias: clinical and molecular review. Am J Med Genet A. 2009;149A(9):1980-2002. doi: 10.1002/ajmg.a.32864. PMID: 19681154.
5. Elgasmi FE, Rahmaoui M, Elarabi S, Badre B. Dental Management of Ectodermal Dysplasia: A Report of Two Clinical Cases. Cureus. 2025;17(5):e84031. doi: 10.7759/cureus.84031.
6. Bagdey SP, Moharil RB, Dive A, Bodhade A. Hypohidrotic ectodermal dysplasia: A case report with review and latest updates. J Oral Maxillofac Pathol 2022;26:S12-6.
7. Alshegifi H A, Alamoudi A M, Alrougi A, et al. Ectodermal Dysplasia: A Case Report. Cureus 2022;14(1): e21184. DOI 10.7759/cureus.21184
8. Wright JT, Fete M, Schneider H, Zinser M, Koster MI, Clarke AJ, Hadj-Rabia S, Tadini G, Pagnan N, Visinoni AF, Bergendal B, Abbott B, Fete T, Stanford C, Butcher C, D'Souza RN, Sybert VP, Morasso MI. Ectodermal dysplasias: Classification and organization by phenotype, genotype and molecular pathway. Am J Med Genet A. 2019;179(3):442-447. doi: 10.1002/ajmg.a.61045. Epub 2019 Jan 31.
9. Stecksén-Blicks C, Falk Kieri C, Hägg D, Schmitt-Egenolf M. Hair shaft structures in EDAR induced ectodermal dysplasia. BMC Med Genet. 2015;16:79. doi:10.1186/s12881-015-0227-5
10. Meshram GG, Kaur N, Hura KS. A case report of hypohidrotic ectodermal dysplasia: A mini-review with latest updates. J Family Med Prim Care 2018;7:264-6.
11. Cerezo-Cayuelas M, Pérez-Silva A, Serna-Muñoz C, Vicente A, Martínez-Beneyto Y, Cabello-Malagón I, Ortiz-Ruiz AJ. Orthodontic and dentofacial orthopedic treatments in patients with ectodermal dysplasia: a systematic review. Orphanet J Rare Dis. 2022;17(1):376. doi: 10.1186/s13023-022-02533-0.
12. Schnabl D, Grunert I, Schmuth M, Kapferer-Seebacher I. Prosthetic rehabilitation of patients with hypohidrotic ectodermal dysplasia: A systematic review. J Oral Rehabil. 2018;45(7):555-570. doi:10.1111/joor.12638
13. Sk Alima, Dey B, Chakraborty A, Pal A, Chakraborty S, Bhowmik K. Anterior fixed functional space maintainer: a case series. Int J Dev Res. 2025;15(5):68333-68336.
14. Jabin Z, Verma SD, Agarwal N, Anand A, Waikhom N. Groper's appliance for esthetic rehabilitation of anterior missing teeth: two case reports. J Interdiscip Dent. 2022;12(1):22-5.
15. Günaydin B, Bayraktar C, Şahin ND. Groper appliance rehabilitation of traumatically lost anterior teeth. Int Dent J. 2024;74(Suppl 1):S158.
16. Pigno MA, Blackman RB, Cronin RJ Jr, Cavazos E. Prosthodontic management of ectodermal dysplasia: a review of the literature. J Prosthet Dent. 1996;76(5):541-545. doi:10.1016/s0022-3913(96)90015-3
17. Joseph S, Cherackal GJ, Jacob J, Varghese AK. Multidisciplinary management of hypohydrotic ectodermal dysplasia - a case report. Clin Case Rep. 2015;3(5):280-6. doi: 10.1002/ccr3.209.
18. Bhakta P, Barthunia B, Nigam H, Pawar P. Ectodermal dysplasia - A rare case report. J Family Med Prim Care. 2019;8(9):3054-3056. doi:10.4103/jfmpc.jfmpc_625_19
19. Chappidi V, Voulligonda D, Bhogavaram B, Reddy PK. Ectodermal dysplasia: Report of two cases in a family and literature review. J Family Med Prim Care. 2019;8(3):1263-1265. doi:10.4103/jfmpc.jfmpc_48_19
20. Khedim N, Hali F, Chiheb S. Ectodermal Dysplasia: Series of 8 Cases. Arch Paediatr Dev Pathol 2022;5(1): 1024