

Prosthetic Rehabilitation of an Adult Patient with Ectodermal Dysplasia: A Case Report

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Key Clinical Message

A prosthetic approach towards rehabilitating ectodermal dysplasia patients with removable prostheses not only restores and improves the function of the stomatognathic system but also enhances self-confidence and improves the quality of life (QoL).

ABSTRACT

Ectodermal dysplasia (ED) is a rare inherited disorder affecting the structures originating from ectoderm. The hypohydrotic variant is the most common form of ED transmitted through an X-linked recessive trait. This condition manifests as sparse hair, hypodontia of primary and permanent dentitions, improper maxillo-mandibular relationship, reduction in the vertical dimension of the lower face, disappearance of the vermilion border, and prominent lips. Such manifestations devastate the patient's aesthetic and function, thus affecting their psychology and quality of life (QoL). This case report discusses the management of a 20-year-old male with ED. The Patient was rehabilitated using upper and lower dentures, restoring the vertical dimension of the face and improving aesthetics and the functioning of the stomatognathic system.

[Keywords: Ectodermal dysplasia, hypodontia, prosthodontic rehabilitation, prosthetic treatment, prosthodontics]

Introduction

Ectodermal dysplasia (ED) is a hereditary disorder associated with malformation secondary to errors in signaling transmission, affecting ectoderm coding. Various genetic patterns have been proposed for its transmission, such as autosomal recessive, autosomal dominant, and X-linked.¹ Freire-Maia defined ED as the syndrome that exhibits at least two characteristics: i.e. trichodysplasia, onchodysplasia, and dyshidrosis. Till date, more than 150 variants of ED have been reported in the literature.² Thurnam published the first report of a patient with ectodermal dysplasia in 1848; then, Darwin also reported a case of ED in 1875. However, Weech coined the term ectodermal dysplasia in 1929.³

Hypohydrotic/ Anhydrotic ED, also referred to as Christ-Siemens-Touraine syndrome, is an X-linked recessive genetic disorder.³ This medical condition predominantly affects males, while females act as carriers. Studies have suggested that the prevalence of ED in men is 1:100,000 live births, whereas carriers have an incidence of around 17:100,000 women.⁴ Diagnosis of ED is usually based on clinical signs and symptoms, along with maxillofacial radiographic findings. Various genetic tests are available for assessment and diagnosis of subtypes of ED.⁵ Patients with ED often have disfigured facial appearance, which has a definitive impact on their appearance, particularly regarding their facial profile and missing/ malformation of teeth.⁶ Studies have also highlighted that ED tends to have devastating emotional effects on the patient.^{7,8} Thus, it is essential to have a time bond rehabilitation approach for these patients to improve their quality of life (QoL).

Comprehensive management of ED requires a multidisciplinary approach involving a team of oral health care providers such as oral surgeons, prosthodontists, orthodontists, pedodontists, and periodontists. Experience has suggested that prosthetic rehabilitation is the most effective treatment option, enhancing the aesthetics and function of the patients. Through this case report, we would like to highlight the details of the prosthetic

rehabilitation of a 20-year-old male patient with hypohidrosis ectodermal dysplasia with partial anodontia. The treatment options were discussed with the oral health care providers, the Patient, and his family. After a fruitful discussion with all the stakeholders, partial dentures for maxillary and mandibular arches were finalized.

Case History/ Examination

A 20-year-old male patient was referred to the Department of Prosthodontics complaining of an unpleasant facial appearance and multiple missing teeth in the upper and lower jaws, leading to difficulties in chewing food. His father accompanied him during the prosthodontic consultation. There was no history of parents' consanguineous marriage. The Patient's family history revealed a non-conclusive finding for a similar condition. No other sibling had identical clinical features associated with ED. None of the family members was involved similarly in previous generations.

Clinical Findings and Observation

The clinical manifestations presented by the Patient were suggestive of hypohidrotic ectodermal dysplasia. The patient appears nervous and lacks confidence; his father mentioned the same. He also mentioned psychological and emotional issues he faced during his childhood at school and in the neighborhood. This has affected his performance through low grades and his reluctance to attend school.

A clinical extra and intra-oral evaluation was done, supplemented by extra-oral radiographic assessment. The extra-oral assessment suggested sparse hair of the scalp, characterized by fragile, hypochromic, and fragile hair, a prominent forehead, sparse eyelashes and eyebrows, pigmented and altered texture of the skin over the face and neck, protrusive and prominent lips contributed to a senile appearance of the face (Fig. 1). Apart from these findings, the skin over the upper and lower limbs was dry, rough, and scaly. Finger and toenails were brittle and thick and short.

The clinical intra-oral evaluation showed multiple clinically missing permanent teeth, leading to the maxilla and mandible's underdeveloped alveolar ridges (atrophic) (Fig.1). Mandibular teeth were absent, and four maxillary teeth were present (i.e., 11,16, 26, and 27) with peg-shaped 11 (Fig.1). Panoramic radiograph confirms the clinical findings; apart from that, it also showed incomplete formation of the roots of 16 and 26, peg-shaped 11; and associated atrophy of the alveolar ridges in the maxillary and mandibular arches secondary to congenital absence of teeth (Fig.2). The upper and lower alveolar ridges were thin. The oral mucosa appeared pale pink with rough texture.

Differential Diagnosis

The following medical disorders were considered as differential diagnoses for the medical and dental conditions of this patient (Table. 1):

Table 1. Medical conditions as a differential diagnosis for ectodermal dysplasia.

Medical condition	Clinical presentation
Alopecia Areata ^{9,10}	An autoimmune disorder that affects the hair follicles
Aplasia Cutis Congenita ¹¹	Rare congenital skin disorder leading to a focal or ext
Focal Dermal Hypoplasia Syndrome (Goltz syndrome) ^{12,13}	A genetically inherited disorder that can affect the de
Incontinentia Pigmenti (Bloch-Sulzberger syndrome) ^{14,15}	An inherited disorder (a rare X-linked dominant geno
Pachyonychia Congenita ^{16,17}	It is a group of rare, inherited disorders of keratinizat

Based on case history, clinical assessment, and radiological findings, the diagnosis was confirmed as ectodermal dysplasia (ED) of the hypohidrotic variant.

Outcome and Follow-Up

After careful patient assessment, a detailed, comprehensive treatment plan for corrective rehabilitation was

planned out. The patient and his father were given two options:

First prosthetic implant-supported prosthesis.

Second, removable dentures for maxillary and mandibular arches.

The Patient and his family discussed the pros and cons of each treatment option, and it was decided to proceed with removable dentures for the maxillary and mandibular arches. The patient was informed about the future implant-supported prosthesis in the upper and lower arches.

Before the commencement of treatment, consent was obtained from the patient. He was given the option of reshaping 11, which he declined. A frenectomy was done for the maxillary labial frenum, which was necessary to provide a better fit for the prosthesis. Irreversible hydrocolloid impressions (Tropicalgin, Zhermack S.P.A, Italy) were made for maxillary and mandibular arches, and casts were poured using dental stone (Kalabhai Kalstone, Kalabhai Karson Pvt Ltd, India). Special trays were fabricated, and border moulding was done with a thermoplastic material (DPI[®] Pinnacle, Dental Products of India, India) for a better marginal seal and fit. Functional wash impressions were made using light body polyvinyl siloxane impression material (Aqual[®] Ultra⁺ Dentsply, Milford, DE, USA), impressions were poured with dental stone, and casts were obtained. Maxillo mandibular relationships were established, followed by a recording of the vertical dimension of occlusion and centric relation. This was followed by mounting the cast on a semi-adjustable articulator setting of the teeth and clinical trial. The final trial also allows the clinician to verify centric and vertical facial relations, occlusion, phonetics, and retention of the plates. Maxillary and mandibular dentures were fabricated using a cross-linked heat cure acrylic resin (DPI[®] Heat Cure, Dental Products of India, India). The denture was placed inside the patient's mouth and adjusted (Fig. 3). There was an immediate improvement in the patient's vertical facial profile, thus improving his facial profile (Fig. 4). The Patient was provided information on how to insert and remove the dentures, maintenance of hygiene, and usage.

Follow-up was done after one week to check the fit of the maxillary and mandibular dentures. This was followed by a three-month follow-up for the next two visits and 6 months after. The Patient is motivated to maintain good oral and denture hygiene at each follow-up appointment. Regular follow-up visits and check-ups have been emphasized at each appointment to ensure the success of the treatment.

The patient was successfully rehabilitated with maxillary and mandibular removable dentures. He was happy and satisfied with his improved facial aesthetic and oral functions. This has increased his self-esteem and confidence.

Discussion

The case under discussion was diagnosed with ED based on distinctive clinical features and radiological findings. One of the most important intra-oral findings was multiple clinically missing permanent teeth; the teeth that were present exhibited abnormal shape, making oral rehabilitation challenging. Oral rehabilitation of ED patients is demanding and requires a multidisciplinary approach. The primary goal of such an approach is the combination of active preventive measures and prosthodontic interventions. Preventive measures include reinforcement of oral hygiene instructions and oral health education. To achieve the patient's aesthetic, speech and masticatory functionality as far as the treatment modality is concerned is based on age, skeletal growth, development of the stomatognathic system, degree of malformation, dental agenesis, and patient motivation.

Prosthetic implant is the treatment of choice for adult patients with ED.¹⁸ Studies have suggested an increase in the prevalence of failure of prosthetic implants in the maxillary anterior.¹⁹ In partial or complete anodontia of the maxillary and/or mandibular arch, the height of the alveolar bone is compromised, further complicating the rehabilitation. Further, such approaches are contraindicated in growing patients. Oral rehabilitation with removable partial and/ or complete prostheses and composite resin restorations is the treatment of choice in growing or young adult patients. Removable prostheses fabricated with cross-linked heat-cured acrylic resin are frequently reported in the literature due to their ease of fabrication and cost-effectiveness. After considering all the limiting factors, such as biological and economic factors, a removable prosthesis for

the maxillary and mandibular arches is needed. Patients are usually advised to be on regular follow-up appointments to adjust the appliance and its maintenance for optimal function.

Marked improvement in the facial aesthetic was evident following the insertion of the removable prostheses; apart from this, the patient noted an improvement in the form and functions, such as speech and masticatory efficiency.

Conclusions

The comprehensive and holistic oral rehabilitation of ED patients is challenging and requires a multidisciplinary approach. Conventional maxillary and mandibular removable dentures effectively rehabilitate the patient by improving the patient's stomatognathic function, aesthetics, and self-confidence.

Author Contributions

Pallawi Sinha : conceptualization, formal analysis, resources, super-vision, writing – original draft. **Nagaveni S Somyaji** : conceptualization, writing – original draft, writing – review and editing. **Subash C Nayak** : formal analysis, writing –review, and editing. **Sudipto Poddar**: formal analysis, writing –review, and editing. **Jitendra Sharan** : conceptualization, formal analysis, supervision, visualization. **Anand Marya** : formal analysis, writing – review and editing, supervision, visualization.

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Ethics Statement

The authors have nothing to report.

Consent

Written informed consent was obtained from the patient to publish this report in accordance with the journal's patient consent policy.

Conflicts of Interest

The authors declare no conflicts of interest.

Data Availability Statement

The data supporting this study's findings are available from the corresponding author upon reasonable request.

References

1. Arte S. Phenotypic and genotypic features of familial hypodontia (Doctoral dissertation). University of Helsinki, Finland; 2001.
2. Bhalla G, Agrawal KK, Singh K, et al. A preliminary study to analyze the craniofacial growth of an ectodermal dysplasia patient after prosthetic rehabilitation. *J Indian Prosthodont Soc* .2013; 13, 43-48. doi:10.1007/s13191-012-0167-0.
3. Sholapurakar AA, Setty S, Pai KM. Total anodontia in a patient with hypohidrotic ectodermal dysplasia: Report of a rare case of Christ-Siemens Touraine Syndrome. *N Y State Dent J*. 2011;77:36-39.
4. Deshmukh S, Prashanth S. Ectodermal dysplasia: A genetic review. *Int J Clin Pediatr Dent*. 2012; 5:197-202. doi:10.5005/jp-journals-10005-1165.
5. Peschel N, Wright JT, Koster MI, et al. Molecular pathway-based classification of ectodermal dysplasias: first five-yearly update. *Genes* .2022;3:2327. doi: 10.3390/genes13122327
6. Bergendal B. Orofacial manifestations in ectodermal dysplasia-A review. *Am J Med Genet Part A* . 2014;164:2465-71. doi:10.1002/ajmg.a.36571.

7. Saltnes SS, Jensen JL, Sæves R, Nordgarden H, Geirdal AØ. Associations between ectodermal dysplasia, psychological distress and quality of life in a group of adults with oligodontia. *Acta Odontol Scand.* 2017;17;75:564-72. doi:10.1080/00016357.2017.1357189.
8. Saltnes SS, Jensen JL, Sæves R, Nordgarden H, Geirdal AØ. Associations between ectodermal dysplasia, psychological distress and quality of life in a group of adults with oligodontia. *Acta Odontol Scand.* 2017;75:564-72. doi:10.1080/00016357.2017.1357189.
9. Pratt CH, King LE, Messenger AG, Christiano AM, Sundberg JP. Alopecia areata. *Nat Rev Dis Primers.* 2017;16;3:1-7. doi:10.1038/nrdp.2017.11.
10. Gilhar A, Etzioni A, Paus R. Alopecia areata. *N Engl J Med.* 2012;366:1515-1525. doi: 10.1056/NEJM-ra1103442.
11. Frieden IJ. Aplasia cutis congenita: A clinical review and proposal for classification. *J Am Acad Dermatol.* 1986;14:646-660. doi: 10.1016/S0190-9622(86)70082-0.
12. Grzeschik KH, Bornholdt D, Oeffner F, et al. Deficiency of PORCN, a regulator of Wnt signaling, is associated with focal dermal hypoplasia. *Nature Genet.* 2007;39: 833-835. doi: 10.1038/ng2052.
13. Wang X, Sutton VR, Peraza-Llanes JO, et al. Mutations in X-linked PORCN, a putative regulator of Wnt signaling, cause focal dermal hypoplasia. *Nature Genet.* 2007;39: 836-838. doi: 10.1038/ng2057.
14. Poziomczyk CS, Recuero JK, Brighenti L, et al. Incontinentia pigmenti. *An Bras Dermatol.* 2014;89:26-36. doi: 10.1590/abd1806-4841.20142584.
15. Swinney CC, Han DP, Karth PA. Incontinentia pigmenti: a comprehensive review and update. *Ophthalmic Surg. Lasers Imaging Retina.* 2015;46:650-657. doi: 10.3928/23258160-20150610-09.
16. Leachman SA, Kaspar RL, Fleckman P, et al. Clinical and pathological features of pachyonychia congenita. *J Invest Dermatol.* 2005;10:3-17. doi: 10.1111/j.1087-0024.2005.10202.x.
17. McLean WI, Hansen CD, Eliason MJ, Smith FJ. The phenotypic and molecular genetic features of pachyonychia congenita. *J. Invest. Dermatol.* 2011;131:1015-1017. doi: /10.1038/jid.2011.59.
18. Guckes AD, McCarthy GR, Brahmin J. Use of endosseous implants in a 3-year old child with ectodermal dysplasia: a case report and 5-year follow up. *Pediatr Dent.* 1997;19:282-285.
19. Sweeney IP, Ferguson JW, Heggie AA, Lucas JO. Treatment outcomes for adolescent ectodermal dysplasia patients treated with dental implants. *Int Paediatric Dent.* 2005; 15:241-248. doi:10.1111/j.1365-263X.2005.00610.x.

Legends

Legend for table

Table 1. Medical conditions as a differential diagnosis for ectodermal dysplasia.

Legend for figures

Figure 1. Pre-treatment extra-oral and intra-oral photographs depicting characteristic findings of ED. **Figure 2.** Orthopantomogram (OPG) of the craniofacial region of the patient. **Figure 3.** a) Finished maxillary and mandibular removable denture. b) Maxillary and mandibular dentures in patient mouth,. c) Post-treatment enhanced the facial appearance of the Patient. **Figure 4.** The improved facial aesthetic of the Patient: pre and post-treatment.



