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Case Report

Hypohidrotic Ectodermal Dysplasia with Soft Palate Cleft in a 32-Year- Old Male

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ABSTRACT:
Ectodermal dysplasia's (EDs) are a rare, genetically heterogeneous group of disorders involving structures derived from the embryonic ectoderm, including hair, sweat glands, teeth, and nails. Hypohidrotic ectodermal dysplasia (HED), the most common form, presents with the classical triad of hypotrichosis, hypohidrosis, and hypodontia or anodontia.[1] This report describes a 32-year-old male with no family history, who presented with frontal bossing, sparse scalp and body hair, heat intolerance due to absent sweating, complete anodontia, a punched- nose deformity, disk-shaped face, depressed mandibular ridge, blanching of the soft palate mucosa, and a unilateral cleft of the soft palate. Diagnosis was established clinically and radiographically using panoramic radiography (OPG) as the only available diagnostic tool. This case is unique because of the presence of a soft palate cleft in adulthood, a rare finding in isolated HED. Multidisciplinary management strategies are proposed, including dental rehabilitation, palatal surgery, dermatologic care, and psychosocial support.
Keywords: Ectodermal dysplasia; Hypohidrotic ectodermal dysplasia; Cleft palate; Anodontia; Genetic disorder.

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INTRODUCTION

Ectodermal dysplasia's (EDs) comprise more than 200 distinct hereditary conditions, unified by defective development of two or more ectodermal derivatives.[1,2] These conditions may affect hair, teeth, nails, sweat glands, skin, and mucous membranes.[3] Hypohidrotic ectodermal dysplasia (HED), also known as Christ-Siemens-Touraine syndrome, is the most prevalent subtype, affecting approximately 1 in 17,000 live births.[4] It is usually inherited in an X-linked recessive pattern due to mutations in the *EDA* gene, though autosomal dominant and recessive variants involving *EDAR* and *EDARADD* genes are also reported.[5]

Etiopathogenesis of Ectodermal Dysplasia

Genetic Mutation
├──► Commonly Affected Genes:

- EDA (X-Linked)
- EDAR (Autosomal)
- EDARADD
- WNT10A

▼

Defective EDA/EDAR/NF-κB Signalling Pathway

▼

Abnormal Ectoderm–Mesoderm Interaction

▼

Failure Of Normal Development Of Ectodermal Appendage

- Hair Follicles → Sparse, Thin Hair (Hypotrichosis)
- Sweat Glands → Reduced/Absent Sweating (Hypohidrosis)
- Teeth → Hypodontia/Anodontia, Conical Teeth
- Nails → Dystrophic Or Brittle Nails
- Sebaceous & Salivary Glands → Dry Skin, Xerostomia.

Clinically, HED is defined by the triad of hypotrichosis, hypohidrosis, and hypodontia or anodontia.[6] Patients often display a characteristic facial appearance, including frontal bossing, depressed nasal bridge (saddle or punched-nose deformity), prominent lips, and a reduced lower facial height due to alveolar bone underdevelopment.[7] Skin is typically thin and dry, and affected individuals are unable to regulate body temperature effectively because of absent or reduced sweat glands.[8] Oral manifestations are central to diagnosis, with hypodontia or anodontia leading to impaired mastication, phonetics, and aesthetics.[9]

Cleft palate is an uncommon but documented finding in certain ectodermal dysplasia syndromes, such as EEC (ectrodactyly-ectodermal dysplasia-cleft) and AEC (Ankyloblepharon
-ectodermal defects-cleft).[10] However, its occurrence in isolated HED is extremely rare[11]. When present in adulthood, cleft poses unique challenges in speech, swallowing, and prosthetic rehabilitation.

Radiographic evaluation is essential in diagnosis and treatment planning. In resource-limited settings, panoramic radiograph (OPG) is frequently the only available modality to demonstrate tooth agenesis and alveolar ridge morphology[12]. This report describes an adult male with HED, notable for the rare coexistence of unilateral soft palate cleft, complete anodontia, and absence of family history, and outlines potential treatment approaches.

CASE PRESENTATION

A 32-year-old male presented to Department of Oral Medicine and oral Radiology at College of Dental Sciences and hospital, Amargadh with complaint of inability to tolerate heat, absence of sweating, and difficulty in mastication and speech. He reported progressive scalp hair thinning since adolescence. There was no significant medical history or family history of similar disorders.

Extraoral Examination

- Facial features: Frontal bossing, depressed nasal bridge with punched-nose deformity, prominent lips, and a disk-shaped face.
- Hair/skin: Sparse, fine scalp hair and body hair; alopecia on vertex; dry, thin skin with absence of sweating, absence of eyebrow hair.
- General features: Heat intolerance since childhood otherwise, healthy.

Intraoral Examination

- Dentition: Complete anodontia in mandibular arche and presence of 11,12,22 of conical shape in maxillary arch.
- Alveolar ridges: Severely resorbed and depressed mandibular ridge.
- Soft tissues: Blanching of oral mucosa, especially on the soft palate.
- Palate: Unilateral left-sided cleft of the soft palate. Based on the clinical features the provisional diagnosis were:

1. Ectodermal dysplasia with partial Anodontia & Unilateral cleft Palate.
2. Oral Submucous Fibrosis (Grade 1).
3. Cloustan syndrome.

Investigations

A panoramic radiograph (OPG) revealed:

- Complete absence of permanent teeth in lower arch.
- Underdeveloped alveolar ridges.
- No pathologic lesions noted.

No advanced imaging or genetic analysis could be performed due to resource limitations.

Based on classical clinical features and radiographic evidence, a diagnosis of hypohidrotic ectodermal dysplasia with soft palate cleft was established.

Figure: 1 The extraoral features of patient showing- Frontal bossing, depressed nasal bridge with punched-nose deformity, prominent lips and absence of eyebrow hair.

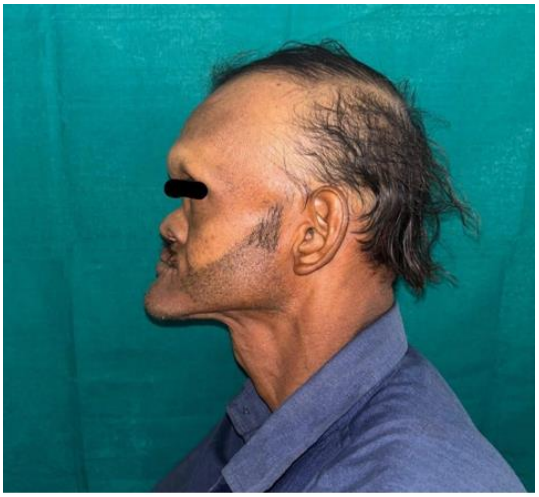

Figure 2: The extraoral features of patient showing - Frontal bossing, depressed nasal bridge with punched-nose deformity, prominent lips, thin sparse hair and a disk-shaped face.

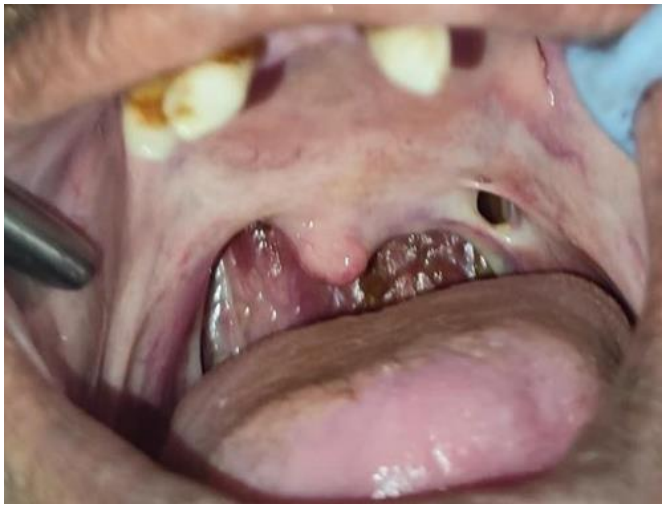

Figure 3: The Intraoral features showing presence of 11,12,22 in maxillary arch.
Soft tissues: Blanching of oral mucosa, especially on the soft palate.
Palate: Unilateral left-sided cleft of the soft palate.

Figure 4: The extraoral features showing thin sparse skin .

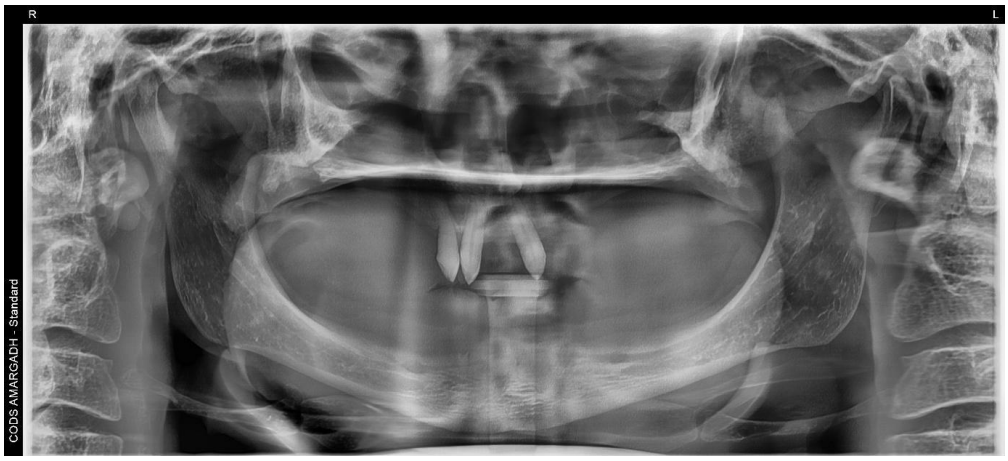

Figure 5: A panoramic radiograph (OPG) revealed:

- **Complete absence of permanent teeth in lower arch.**
- **Presence of 11,12,22 in maxillary arch**
- **Underdeveloped alveolar ridges.**
- **No pathologic lesions noted.**

DISCUSSION

Clinical Spectrum

The patient's findings correspond to classical HED, including hypotrichosis, anhidrosis, and anodontia[6,7] Craniofacial anomalies such as frontal bossing and saddle-nose deformity are common in HED due to midface hypoplasia[8]. The thin mucosa with blanching reflects the underlying ectodermal defect in epithelial development[13].

The presence of a cleft palate is noteworthy. While orofacial clefts are commonly associated with syndromes such as EEC and AEC, they are rarely reported in isolated HED[10,11.] This suggests phenotypic variability or a possible overlap between syndromic entities.

Diagnostic Considerations

Genetic confirmation, though ideal, is not always feasible in developing regions. Thus, diagnosis often relies on phenotypic features[14]. Panoramic radiography remains crucial in visualizing anodontia and assessing bone for prosthetic rehabilitation[12].

Management Strategies

A comprehensive, multidisciplinary treatment approach is essential:

1. **Dermatologic and systemic management**
   - Regular use of emollients to combat xerosis.
   - Education on temperature regulation strategies: cooling devices, hydration, avoidance of prolonged sun exposure[15].

2. **Oral rehabilitation**
- **Interim prostheses**: Complete removable dentures to restore function and aesthetics.
- **Definitive options**: for adults with ectodermal dysplasia include **implant- supported fixed prostheses**, planned according to bone availability and esthetic demands. **Conventional endosseous implants** are preferred when bone volume is adequate, while **basal (cortical) implants** offer a graftless alternative in severely resorbed ridges. In cases with inadequate bone, **augmentation procedures** such as onlay grafts, sinus lifts, or distraction osteogenesis may be required. Treatment should be individualized considering skeletal maturity and patient expectations [16.]
- **Challenges**: Severely resorbed ridges and mucosal blanching complicate prosthetic retention[17].

3. **Surgical management**
- **Palatoplasty**: Repair of the soft palate cleft to improve velopharyngeal function, speech, and swallowing[12].
- ENT evaluation for recurrent infections and speech therapy as adjuncts.

4. **Psychological support**
- Counselling to address psychosocial difficulties related to facial appearance and functional impairment[18].

Prognosis

With appropriate prosthetic and surgical rehabilitation, patients with HED can achieve significant improvements in mastication, phonetics, aesthetics, and quality of life[19]. Long-term follow-up is essential due to progressive ridge resorption and prosthetic adaptation needs.

CONCLUSION

This case illustrates a rare adult presentation of hypo hidrotic ectodermal dysplasia with soft palate cleft and no family history. Diagnosis was based on clinical findings and OPG, underscoring the importance of careful clinical examination in resource-limited settings. The unusual occurrence of soft palate cleft highlights the phenotypic variability of HED. A multidisciplinary treatment plan involving prosthodontic rehabilitation, palatal surgery, dermatologic care, and psychosocial support is critical to improving functional outcomes and quality of life.

REFERENCES

1. Wright JT, Fete M, Schneider H, Zinser M, Koster MI, Clarke AJ, et al. Ectodermal dysplasias: classification and organization by phenotype, genotype and molecular pathway. Am J Med Genet A. 2019;179(3):442-7.
2. Priya V, Muthu MS, Sehgal K, Reddy VV. Hypo hidrotic ectodermal dysplasia: Clinical features and management. J Indian Soc Pedod Prev Dent. 2013;31(1):58-61.
3. Itthagarun A, King NM. Ectodermal dysplasia: a review and case report. Quintessence Int. 1997;28(9):595-602.
4. Bergendal B. Prosthetic habilitation of children with ectodermal dysplasia and oligodontia. Swed Dent J Suppl. 2001;156:1-63.
5. Cluzeau C, Hadj-Rabia S, Jambou M, Mansour S, Guigue P, Masmoudi S, et al. Clinical and molecular characterization of 63 families with hypo hidrotic ectodermal dysplasia. Am J Med Genet A. 2011;155A(4):789-97.
6. Freire-Maia N, Pinheiro M. Ectodermal dysplasias: a clinical classification and a causal review. Am J Med Genet. 1988;31(1):153-62.
7. Rashedi B, Baba NZ, Ciaramicoli MT, Fernandes R. Prosthodontic treatment of ectodermal dysplasia: A clinical report. J Prosthet Dent. 2003;89(6):588-92.
8. Kishore M, Panat SR, Aggarwal A, Jha A, Upadhyay N. Hypo hidrotic ectodermal dysplasia: A case series. J Indian Soc Pedod Prev Dent. 2014;32(1):69-72.
9. Kupietzky A, Houpt M. Hypo hidrotic ectodermal dysplasia: characteristics and treatment. Quintessence Int. 1995;26(4):285-91.
10. Rosenmann A, Argov S, Goodman RM. Ectrodactyly, ectodermal dysplasia and cleft palate (EEC syndrome). Clin Genet. 1976;9(3):347-52.
11. Otsuki Y, Ito Y, Okuno H, Yoshimoto S, Fujimoto T. Intermediate phenotype between AEC syndrome and ectodermal dysplasia with cleft lip/palate. Plast Reconstr Surg Glob Open. 2016;4(12):e1172.
12. Malvankar DD, Manjunath K, Sinha R, Kurien M. Ectodermal dysplasia and cholesteatoma: a cross-sectional study. Int J Pediatr Otorhinolaryngol. 2017;94:1-5.
13. Kupietzky A, Belenky E. Hypo hidrotic ectodermal dysplasia: prosthetic management of an anhidrotic child. ASDC J Dent Child. 1993;60(4):282-6.
14. Vieira KA, Teixeira MS, Guirado CG, Gaviao MB. Prosthodontic treatment of hypo hidrotic ectodermal dysplasia with complete anodontia: case report. Quintessence Int. 2007;38(1):75-80.
15. Itthagarun A, King NM, Wong KH. Oral rehabilitation of hypo hidrotic ectodermal dysplasia patient: a multidisciplinary approach. Quintessence Int. 1999;30(7):461-6.
16. Hiremath V, Nyamati SB, Nair SJ, et al. Dental management of a patient with ectodermal dysplasia using basal implants: a case report. *J Adv Med Dent Scie Res.* 2025 Jul;13(7):88-92.
17. Hickey AJ, Vergo TJ Jr. Prosthetic treatments for patients with ectodermal dysplasia. J Prosthet Dent. 2001;86(4):364-8.
18. Bal E, Yavuz I, Gorur I, Sen A. Psychological profile and quality of life in hypo hidrotic ectodermal dysplasia. J Oral Rehabil. 2009;36(12):895-902.
19. Schnabl D, Grunheid T, Schupp W. Prosthetic rehabilitation of patients with hypo hidrotic ectodermal dysplasia: A systematic review. J Oral Rehabil. 2018;45(7):555-70.