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The Journal of Dental Panacea

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Case Report

Ectodermal dysplasia: An overview and a case report

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Abstract

Ectodermal dysplasia (ED) comprises a group of rare inherited disorders characterized by abnormal development of ectodermal structures such as skin, hair, nails, teeth, and sweat glands. This paper provides an overview of major ED subtypes, including hypohidrotic, hidrotic, and other syndromic forms, highlighting their genetic etiology, clinical manifestations, and particularly the significant dental anomalies they cause. The report emphasizes the importance of early diagnosis and multidisciplinary management, especially concerning dental rehabilitation. A detailed case report of a 14-year-old boy with hypohidrotic ectodermal dysplasia illustrates the clinical challenges and prosthodontic management strategies involved. The patient, presenting with severe oligodontia and facial anomalies, was rehabilitated with custom complete dentures tailored for psychological and functional improvement. The discussion underscores the critical role of prosthetic interventions in maintaining aesthetics, mastication, and speech, while accommodating growth. The article concludes by advocating for timely prosthodontic care and individualized treatment planning, paving the way for future implant-supported solutions post-skeletal maturity.

Keywords: Ectodermal dysplasia, Clinical manifestation, Management

Received: 22-04-2025; **Accepted:** 09-06-2025; **Available Online**: 25-07-2025

1. Introduction

Ectodermal dysplasia (ED) refers to a group of rare genetic disorders that affect the development of ectodermal structures, including the skin, hair, nails, teeth, and sweat glands. These disorders result from mutations in genes involved in ectodermal development, leading to abnormalities in these structures. ED is typically inherited in an autosomal dominant, autosomal recessive, or X-linked manner.

1. Hypohidrotic Ectodermal Dysplasia (HED) (Christ-Siemens-Touraine Syndrome)
   a. Etiology: Caused by mutations in the *EDA*, *EDAR*, or *EDARADD* genes, with the most common form being X-linked recessive.
   b. Signs and Symptoms:
      i. Reduced or absent sweating (hypohidrosis or anhidrosis), leading to overheating
      ii. Sparse, thin, and brittle hair (hypotrichosis)
      iii. Peg-shaped or missing teeth (hypodontia or oligodontia)
      iv. Dry skin and increased risk of infections
      v. Prominent forehead, depressed nasal bridge, and wrinkled skin around the eyes
2. Hidrotic Ectodermal Dysplasia (Clouston Syndrome)
   a. Etiology: Caused by mutations in the *GJB6* (connexin 30) gene; inherited in an autosomal dominant manner.
   b. Signs and Symptoms:
      i. Normal sweating ability
      ii. Thickened, ridged, and brittle nails (onychodystrophy)
      iii. Thickened skin on the palms and soles (palmoplantar keratoderma)
      iv. Sparse or absent scalp and body hair
      v. Normal teeth development

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https://doi.org/10.18231/j.jdp.2025.024

3. Tooth and Nail Syndrome (Witkop Syndrome)
   a. Etiology: Caused by mutations in the *MSX1* gene; inherited in an autosomal dominant manner.
   b. Signs and Symptoms:
      i. Hypodontia or missing teeth
      ii. Abnormal tooth shape (small, peg-shaped teeth)
      iii. Brittle or slow-growing nails
      iv. Normal sweating and hair growth
4. Ectrodactyly-Ectodermal Dysplasia-Clefting Syndrome (EEC Syndrome)
   a. Etiology: Caused by mutations in the *TP63* gene; inherited in an autosomal dominant pattern.
   b. Signs and Symptoms:
      i. Cleft lip and/or cleft palate
      ii. Ectrodactyly (split-hand or split-foot deformity)
      iii. Abnormal teeth, nails, and skin
      iv. Dry eyes due to underdeveloped tear glands
      v. Hearing loss and urogenital anomalies
5. Rapp-Hodgkin Syndrome
   a. Etiology: Also caused by mutations in the *TP63* gene; considered a variant of EEC syndrome.
   b. Signs and Symptoms:
      i. Similar features to EEC syndrome, including cleft lip/palate
      ii. Hypodontia and nail abnormalities
      iii. Sparse hair and dry skin
      iv. Eye abnormalities such as conjunctivitis

1.1. Dental features of ectodermal dysplasia

Dental abnormalities are among the most significant and characteristic features of ectodermal dysplasia (ED). These abnormalities can affect both primary (deciduous) and permanent dentition, leading to functional and aesthetic challenges. The severity of dental manifestations varies depending on the type of ED, with hypohidrotic ectodermal dysplasia (HED) being the most commonly associated form.(**Table 1**)

1.2. Management of dental issues

A multidisciplinary approach involving pediatric dentists, prosthodontists, and orthodontists is crucial to managing dental abnormalities in ED patients. Treatment options include (**Table 2**)

Table 1: Dental features of ectodermal dysplasia

S.No.	Type of abnormality	Dental features
1	Hypodontia, Oligodontia, and Anodontia	• Hypodontia: Partial absence of teeth (missing fewer than six teeth). • Oligodontia: Missing six or more teeth, a more severe form of hypodontia. • Anodontia: Complete absence of teeth (rare). • Clinical Impact: o Difficulty in chewing and speech development. o Aesthetic concerns, especially in the anterior region. o Delayed eruption of permanent teeth.
2	Abnormal Tooth Morphology	• Peg-shaped or conical teeth: o The most characteristic dental feature in ED. o The incisors and canines are often small, pointed, and widely spaced. • Reduced crown size: o Teeth appear smaller than normal. o Leads to spacing issues and malocclusion. • Enamel hypoplasia: o Thin, defective enamel, making teeth prone to cavities
3	Delayed Tooth Eruption	• Primary and permanent teeth may erupt later than usual. • Some teeth may remain unerupted due to lack of supporting structures. • Retained primary teeth due to absence of permanent successors
4	Malocclusion and Jaw Development Issues	• Reduced alveolar bone development: o Due to the absence of teeth, the jawbones (maxilla and mandible) may not develop properly. o Leads to a sunken midface appearance (increased vertical facial height). • Increased overjet and open bite: o Because of missing teeth, the lower jaw may appear more prominent (pseudo-prognathism). • Crowding or spacing issues: o Due to the abnormal size and number of teeth, significant spacing or malalignment occurs
5	Functional and Aesthetic Challenges	• Difficulty in mastication (chewing): o Leads to nutritional deficiencies and digestion problems. • Speech difficulties: o Lack of proper teeth placement affects pronunciation of certain sounds. • Aesthetic concerns:

		○ Missing or abnormal teeth can significantly impact self-esteem and social interactions

Table 2:

S.No.	Treatments	Indications
1	Dentures (Removable Prosthetics)	• Used in young children with anodontia or severe oligodontia. • Helps with aesthetics, speech, and mastication. • Replaced periodically as the child grows.
2	Dental Implants	• Used in older adolescents and adults once jaw development is complete. • Provides a permanent and functional solution. • Requires sufficient bone support, which may be limited in ED patients.
3	Fixed Prosthetics (Bridges and Crowns)	• Used for missing teeth or abnormal tooth morphology. • Bridges can help close gaps due to missing teeth. • Crowns protect and restore the shape of peg-shaped teeth.
4	Orthodontic Treatment	• Used to correct malocclusion, spacing issues, and misalignment. • Often combined with prosthetic rehabilitation for optimal results

2. Case Report

A 14-year-old boy reported to Department of prosthodontics, with difficulty in eating food due to absence of teeth. The parents wanted a prosthesis which could help the child in eating. The boy was diagnosed of Hypohidrotic Ectodermal Dysplasia at the age of two. There was a positive family history for ectodermal dysplasia.

On examination, the boy exhibited the classical features of ED: hypodontia, anhydrosis, hypotrichosis, prominent forehead, decreased lower anterior facial height, flat mandibular plane, saddle nose, thin upper lip, everted lower lip, prominent chin and a resultant concave facial profile as shown in **Figure 1**. The medical history was non-contributory, and no other systemic abnormalities were reported.

The intraoral examination showed the presence of 1 tooth: A conical maxillary central incisor. Mandibular arch was completely edentulous. The patient exhibited the loss of vertical dimension, the absence of alveolar processes and an anomalous development of alveolar ridges shown in **Figure 2**, **Figure 3**. OPG revealed aplasia of alveolar bone and absence of other teeth buds. Maxillary incisor showed closed apice.

Considering the age of the patient and the poor amount of alveolar bone present, treatment option considered cusil complete denture for maxillary arch after reshaping the conical maxillary central incisor with composite and complete denture for mandibular arch. This therapeutic approach provided for the psychological and social comfort for the patient.

Periodic recalls of young ED patients are also important because prosthesis modification or replacement will be needed as a result of continuing growth and development. Adjustments related to proper fit, the occlusion of a prosthesis should be monitored for period of time for changes because of jaw growth over the years.[1]

2.1. Prosthodontic management

After a detailed case history and examination a preliminary impression was made

1. Primary Impression: (**Figure 3**)
   a. A preliminary impression was made using alginate impression material(Zhermack Tropicalgin)
2. Secondary Impression:
   a. A special custom tray with uniform 2 mm full arch wax spacer covering the natural teeth was prepared. Peripheral border seal was established With Border molding . Performed using low-fusing compound to improve retention.
   b. Final impressions were taken with C silicone base and catalyst paste Zhermack oranwash L and indurant gel) for better adaptation (**Figure 3**, **Figure 4**)
3. Occlusal Registration:
   a. Wax rims were adjusted to restore proper vertical dimension of occlusion (VDO).
   b. Jaw relations were done by nick and notch method with help of Aluwax by manually guiding mandible into centric.(**Figure 6**)
4. Try-in Stage: Teeth arrangement was done with the consideration as per the age, sex and arch size of the patient. Modifications were done as per the patient and his family at the time of Trial (**Figure 7**)
   a. A wax try-in was performed with acrylic resin teeth to assess aesthetics and occlusion.
   b. The patient and guardians approved the alignment and appearance.
5. Denture Fabrication and Insertion:
   a. The dentures were processed using heat-cured acrylic resin (Dentsply lucitone denture base resin).
   b. After necessary adjustments, the final dentures were delivered.(**Figure 8**)

6. Post-Insertion Follow-Up:
   a. The patient was instructed on proper denture hygiene.
   b. Follow-ups were scheduled at 1 week, 1 month, and 6 months to monitor adaptation and function. (**Figure 9**)
      i. The patient adapted well to the dentures within a few weeks, reporting improved chewing ability and speech clarity.
      ii. Aesthetic concerns were addressed, significantly boosting the patient's confidence and social interactions.
      iii. Challenges included mild initial discomfort and salivation changes, which resolved with adaptation.
      iv. Given the progressive changes in jaw growth, periodic denture relining and replacement were planned until skeletal maturity for future implant placement.

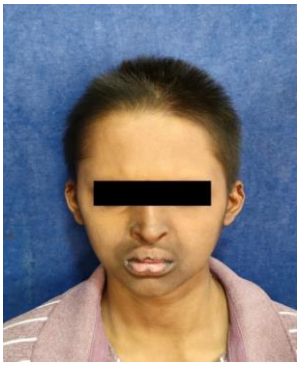

Figure 1: Frontal profile

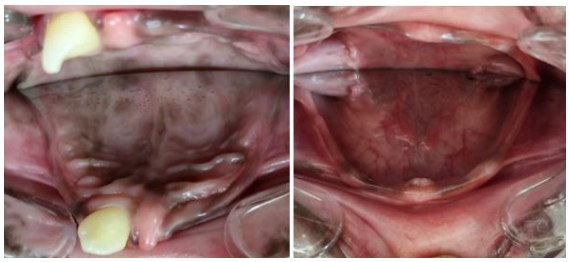

Figure 2: Intraoral preoperative maxillary arch

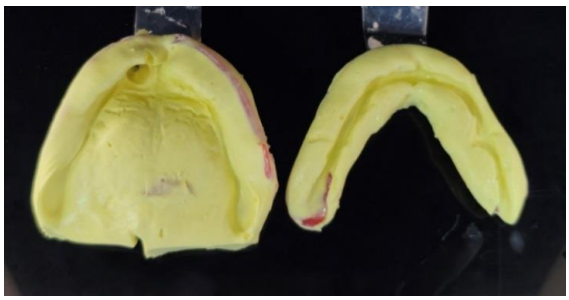

Figure 3: Preliminary Impression

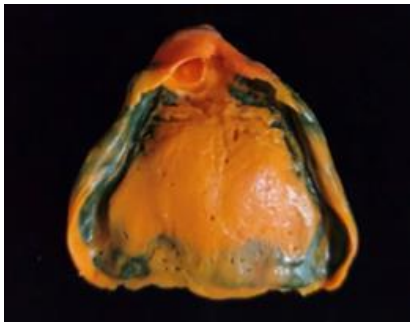

Figure 4: Maxillary final impression

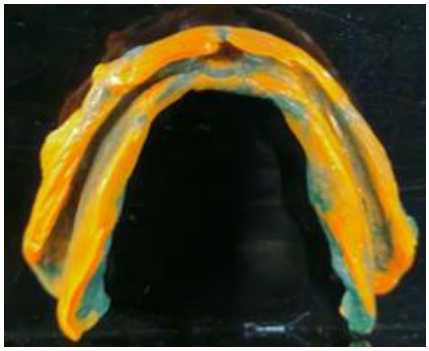

Figure 5: Mandibular final impression

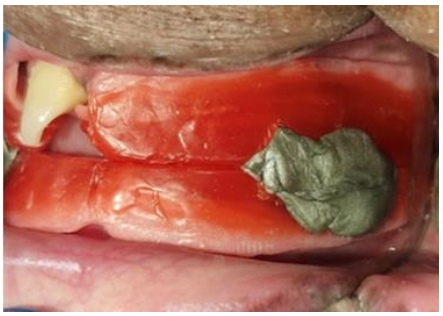

Figure 6: Jaw relation

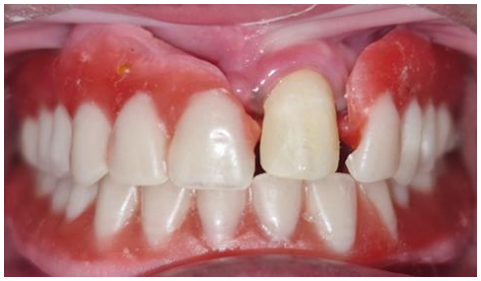

Figure 7: Try – in

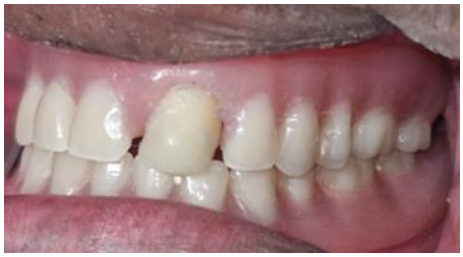

Figure 8: Denture insertion

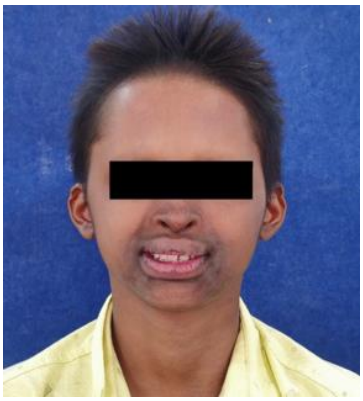

Figure 9: Postoperative

3. Discussion

The treatment for a patient with ectodermal dysplasia varies and needs comprehensive overall assessment including consideration of child's age, dental agenesis, stage of teeth development, degree of malformation of teeth, the growth and development of the stomatognathic system of the patient. Early Oral rehabilitation of patients with ectodermal dysplasia is necessary for improving their facial profile, aesthetics and to boost patients confidence. Whenever patients demand for implant therapy, the main problem is insufficient bone; implant placement may not be possible without bone grafting.

While the defects associated with ED are not typically life-threatening when properly managed, they can significantly impact quality of life. This is especially relevant in dentistry due to the numerous oral manifestations of ED. Tooth eruption is often affected, including agenesis of primary and/or permanent teeth, as well as irregular eruption order or timing. In addition to missing teeth, abnormalities in tooth morphology—such as undersized, conical, or fused teeth, and taurodontism—are common. Malpositioned teeth and abnormal spacing can further influence jaw and alveolar development. Given these extensive dental needs, genetic testing and counseling should be considered.[2]

As stated by Nowak, treating the pediatric patient with ectodermal dysplasia requires the clinician to be knowledgeable in growth and development, behavioral management, techniques in the fabrication of prosthesis, the ability to motivate the patient and parent in the use of the prosthesis, and the long term follow-up for the modification and/or replacement of the prosthesis.[3]

A complete denture prosthesis provided to the patient modifies alveolar height, enhances the musculocutaneous profile, and significantly improves mastication, esthetics, phonetics, and psychological well-being. The standard treatment for ectodermal dysplasia typically involves a sequence of complete or partial dentures during the dentofacial growth phase, followed by definitive rehabilitation once jaw growth is complete.[4]

As described by marques and till a prosthesis should be provided before the child starts school to ensure a natural appearance and allow time for adaptation. Early restoration of facial features is crucial for healthy psychological development.[5]

Restoring a natural and aesthetically pleasing appearance is essential for the psychological well-being and social integration of children with HED. While dentures are not ideal substitutes for natural teeth, they help maintain a balanced and adequate diet, which is crucial since lifelong dietary habits are formed during childhood.[6] Additionally, research has shown that nonfunctional speech issues improve when dentures are provided for edentulous children.[7]

Complete dentures or RPDs require periodic adjustments and should be replaced when growth leads to a reduced vertical dimension of occlusion and abnormal mandibular posture. Without dentures, the forward rotation of the mandible results in the chin shifting upward and forward, reducing the height of the lower third of the face and increasing the risk of Class III malocclusion. Dentures help facilitate a backward-downward mandibular rotation, promoting proper chin positioning.[8]

Achieving proper retention and stability of prostheses can be challenging. In HED patients, factors such as oral mucosal dryness and underdeveloped maxillary tuberosities and alveolar ridges hinder denture resistance and stability.[9]

Fabricating a dentures in these patients is critical, there should be a wide distribution of occlusal load fully extending the denture base. Careful consideration must be given to the impression technique. For complete dentures, support should extend beyond the denture base area to include the full vestibular sulcus reflection, ensuring a retentive base with an effective border seal.[10]

When natural teeth are present, overdentures are the preferred treatment option. They offer the added benefit of preserving the alveolar bone. Due to ongoing growth and development, regular adjustments or replacements of the prosthesis are necessary.[5,11] A systematic review revealed that dentists were dissatisfied with the results in up to 30% of cases, requiring prosthesis replacement approximately every 3.5 to 4 years.[12]

Fixed prosthodontic treatment is rarely used due to the limited number of abutments and the young age of the patient. FPDs with rigid connectors should be avoided, as they may hinder jaw growth. In young patients, a combination of individual crown restorations and direct composite restorations with a removable partial denture is often preferred.[5]

Although there are concerns about growth patterns and early implant placement should not be performed routinely, there are certain cases where implants may be indicated for young patients with anodontia or severe oligodontia.

Currently, evidence suggests that implant placement in these cases does not significantly impact craniofacial growth, though there may be a slight increase in the risk of implant failure.[13]

Definitive dental treatment of implant placement in the maxilla and posterior mandible should be best carried out after the completion of skeletal growth. A systematic review found success rates from 88.5- 97.6% for implants placed in ED patients.[14,15]

Removable prosthodontics is the most common treatment approach for managing ED. While complete dentures are a viable option, overdentures or RPDs supported by natural teeth are preferred for better alveolar bone preservation[5]. Individuals with ED typically undergo a series of dentures throughout their growth years, followed by definitive prosthetic rehabilitation once jaw growth is complete.[16]

4. Conclusion

The prosthetic rehabilitation of young ED patients **r**equires a comprehensive approach that considers growth, function, and aesthetics. Removable dentures serve as an effective transitional solution**,** improving quality of life until definitive treatment with implant-supported prostheses can be performed. Early intervention plays a crucial role in enhancing facial development and psychosocial well-being in children and adolescents with ectodermal dysplasia.

5. Source of Funding

None.

6. Conflict of Interest

None.

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Cite this article: Nirgulkar PM, Khalikar SA, Mahale K, Rajguru V, Mahajan SV, Tandle U. Ectodermal dysplasia: An overview and a case report. *J Dent Panacea*. 2025;7(2):110-115.