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CASE REPORT

Ectodermal Dysplasia In A Six-Year-Old Bahraini Child - Oral and dental aspects and the management - A Case Report

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Abstract

Ectodermal Dysplasia (ED) is a heterogeneous group of genetically determined disorders affecting two or more ectodermal derivatives, including skin, teeth, nails, hair, and sweat glands. Other structures, including the lips, ears, and eyes-also of ectodermal origin-may be involved. This report presents a 6-year-old Bahraini boy with ED who presented to the Paediatric Dental Clinic at the Dental and Maxillofacial Centre (DMFC), Royal Medical Services (RMS), with concerns regarding his appearance due to numerous missing teeth. Initial management focused on preventive care to preserve the existing dentition, followed by restorative procedures and comprehensive prosthetic rehabilitation. A multidisciplinary team approach was essential to establishing a practical treatment pathway.

Keywords: Ectodermal dysplasia, Hypodontia, Hypohidrosis, Hypotrichosis, Multidisciplinary approach

Introduction

Ectodermal dysplasia (ED) is a rare genetic disorder characterized by abnormalities in the development and function of ectodermal tissues, including hair, teeth, nails, and sweat glands. More than 192 types of ED have been identified in the literature, with an estimated frequency of 7 per 100,000 births.[1,2]

ED is classified into nine types based on the structures involved. Examples include X-linked hypohidrotic ED with immunodeficiency, hypohidrotic ED, ankyloblepharon-ectodermal defect-cleft lip/palate syndrome, tricho-dento-osseous syndrome, hidrotic ED, focal dermal hypoplasia, MIDAS syndrome, and focal facial dermal dysplasia.[3] The most common type is X-linked hypohidrotic ED.

Signs and symptoms vary with the affected structures and typically become evident in infancy or childhood, as they may not be apparent at birth.[4] ED is more commonly seen in males, while females may show only minor features.[5] Mortality is higher during infancy or early childhood due to episodes of hyperpyrexia.[6]

The four main organs commonly affected in ED are detailed in Table 1.

Dental abnormalities such as hypodontia, anodontia, and conical teeth are often key diagnostic indicators, particularly in childhood. Early identification of these signs is crucial for timely diagnosis and comprehensive care planning. Management typically includes prosthetic rehabilitation, orthodontic treatment, and restorative procedures to restore oral function and aesthetics and to support psychosocial development. The management primarily focuses on addressing the specific symptoms and challenges that affect individuals. Since the condition affects multiple ectodermal-

derived structures, a multidisciplinary approach involving various healthcare professionals is crucial.[8]

Table 1: Signs and symptoms of ED.[7]

Hair	• Body and scalp hair could be light-colored, sparse, and thin. • twisted, curly, coarse, or overly brittle.
Nails	• Toenails and fingernails can be brittle, thick, unevenly shaped, colored, ridged, or growing slowly. • Nails may be absent • Cuticles could be prone to infection
Teeth	• Abnormal tooth development that results in the growth of pointed or peg-shaped teeth or missing teeth • Tooth enamel might be defective • Taurodontism
Sweat Glands	• Sweat glands may function abnormally or not at all if eccrine sweat glands are absent or sparse • The body is unable to regulate temperature without regular sweat production appropriately. • Recurrent high fevers in children can cause seizures and neurological issues. • One common issue is overheating, especially in warmer climates.

Case Presentation

A six-year-old Bahraini boy, medically fit and well, presented with his mother to the Paediatric Dental Clinic at DMFC-RMS, concerned about his poor smile due to multiple missing teeth and experiences of bullying at school. The parents were non-consanguineous. Two male siblings (aged 6 and 19) were diagnosed with ED, while a 17-year-old female sibling had no signs or history of the disorder. Genetic testing was not performed.

This was the patient's first dental visit, and he had not received prior dental treatment. He brushed twice daily with adult toothpaste and had no parafunctional habits.

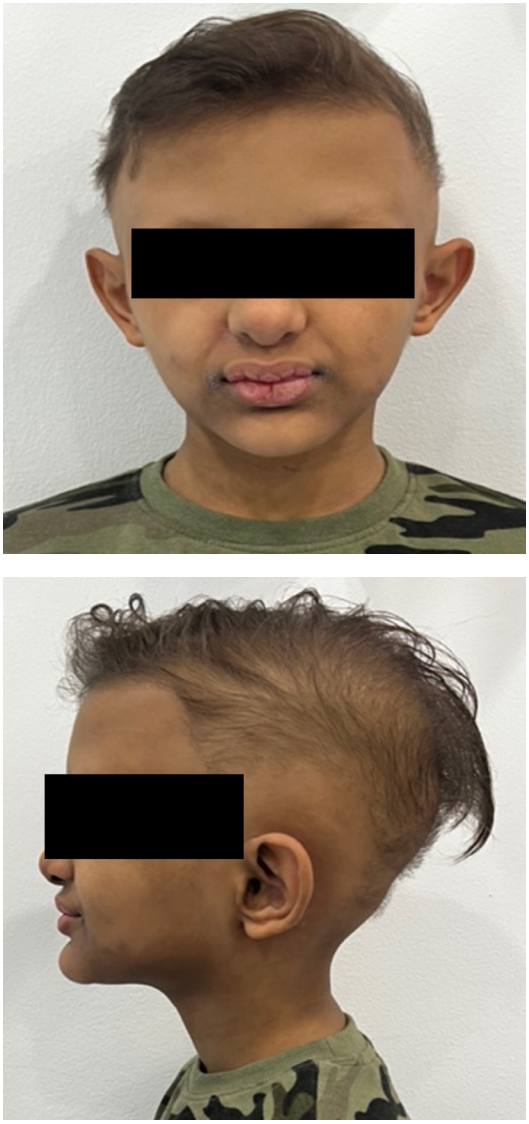

Figure 1: Physical examination. (A) Frontal View (B) Lateral View

Physical examination: Revealed decreased facial height, sparse fine brown hair, protuberant lips, prominent ears and supraorbital ridges, a saddle nose, scanty eyebrows and eyelashes, and dry, pigmented periorbital skin. Nails appeared normal (Figure 1A and 1B).

Intraoral examination: Oral hygiene was fair, as per the simplified oral hygiene index. The alveolar ridge was underdeveloped with only four visible teeth-two in each arch-likely 53, 51, 73, and 83 (Figure 2). A fifth tooth, likely tooth 21, began to erupt during follow-up. All present teeth exhibited a peg-shaped morphology.

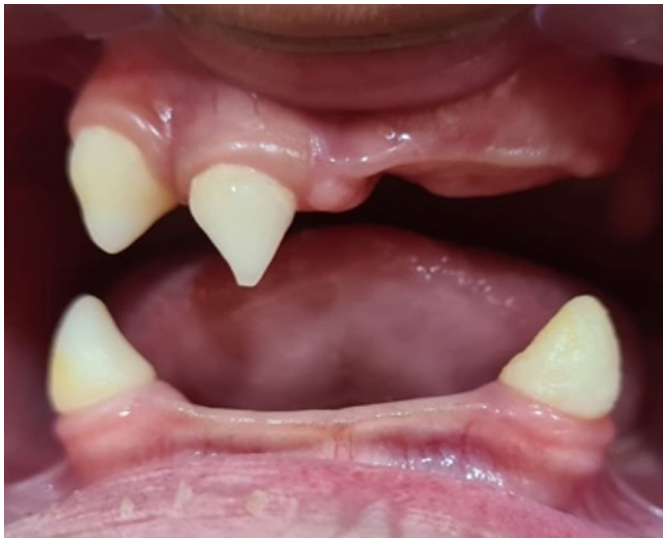

Figure 2: Intra-oral Examination showing partially edentulous arches

Investigations

An orthopantomogram (OPG), taken at age eight despite some movement during exposure, revealed severe bone loss and hypodontia. Present teeth appeared to be 53, 51, 73, 83, and 21 (Figure 3).

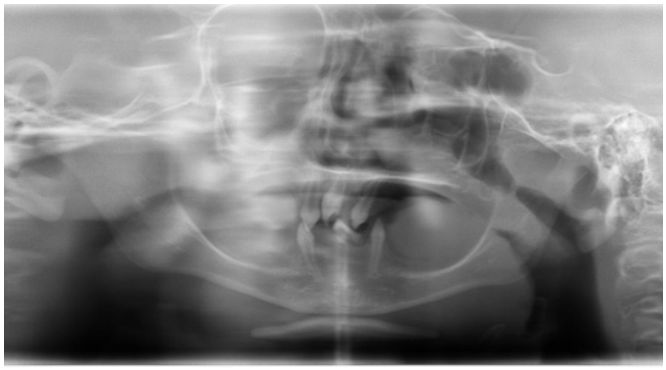

Figure 3: Intra-oral Examination showing partially edentulous arches

Treatment Plan

The treatment adhered to the Department of Health Preventive Toolkit.[9] Emphasizing a positive dental experience aimed to promote cooperation and long-term oral health. Non-pharmacological behavior management techniques, particularly Tell-Show-Do and positive reinforcement, proved to be effective.[9]

Current treatment plan

Prevention plan: Oral hygiene instructions were provided to the patient at age 7, including a demonstration of toothbrushing techniques and an explanation of the importance of regular follow-up.

Prosthetic plan: Multiple visits were scheduled to fabricate upper and lower overdentures as temporary prostheses to improve esthetics and function until complete tooth eruption.

- Visit 1: Preliminary impressions of both arches were taken using alginate (Tropicalgin®, Zhermack, Italy) and small perforated metal dentulous trays (API Instruments, Germany) to create study casts.
- Visit 2: Secondary impressions were made using the same alginate material. Bite registration was recorded using bite registration wax (Aluwax®, USA).
- Visit 3: Try-in of the wax denture setup was performed to assess fit, esthetics, and occlusion.
- Visit 4: Denture delivery was completed. The patient expressed satisfaction with comfort and appearance. (Figure 4)
- Visit 5: Review was performed 1 month after denture delivery.

Future restorative and prosthetic plan: Composite build-up will be performed for teeth 51 and 21 after complete eruption, using composite shade A2 (3M™ Filtek™ Z350 XT, USA) in a single visit to improve esthetics and function. Subsequently, in coordination with the prosthodontics department, the patient will receive upper and lower removable dentures rather than overdentures.

Future definitive treatment plan: Future consultation with orthodontic, prosthodontic, and conservative dentistry specialties will be considered. After age 18, the patient may undergo an implant procedure, followed by an implant-supported denture, after consultation with the oral surgery team.

Discussion

Among ED types, hypohidrotic and hydrotic are the most clinically significant, primarily differentiated by sweat gland function. Hypohidrotic ED, the most common form, follows X-linked inheritance (EDA gene, Xq12-q13.1), while autosomal dominant/recessive types involve EDAR and EDARADD genes. Hydrotic ED results from mutations in GJB6 on chromosome 13 (13q11-q12).[6,10]

The patient exhibited classic features of hypohidrotic ED: hypotrichosis, hypodontia, and hypohidrosis. Early diagnosis in at-risk families can be achieved through genetic analysis, fetal skin biopsy, and second-trimester sonography.[11] Diagnosis is often clinical before the age of 3.

Management must be individualized based on patient needs, with an emphasis on temperature regulation, dental rehabilitation, and multidisciplinary coordination. Heat intolerance due to absent sweat glands necessitates prompt medical evaluation for fever and overheating. Immediate interventions include cold sponging, cooling vests, emollients, air-conditioning, and hydration.[10,11] Parents should be advised to limit the child's exposure to heat and physical exertion. Other typical ED symptoms that should be treated symptomatically include atopic dermatitis, xerostomia, and dryness of the eyes and nose. Children with hypohidrotic ED may experience up to 30% mortality in the first three years due to failure to thrive, hyperthermia, and infections. After this critical period, life expectancy is typically normal.[10]

Skin care measures, such as moisturizers and sun protection, can help manage dryness and sensitivity. In cases of hearing loss or eye abnormalities, appropriate interventions, such as hearing aids or corrective lenses, may be necessary.

Dental abnormalities are a prominent feature of hypohidrotic ED. Individuals may have missing or malformed teeth, which can affect their appearance, speech, and ability to chew food. Dental interventions aim to address these issues. They may include dentures, dental implants, orthodontic treatment, and other dental prosthetics to improve aesthetics and function.[10,12]

In this case report, the patient is young and has ongoing craniofacial growth, which makes fixed prosthetics or implants unsuitable at this stage, as these could interfere with jaw development and require frequent adjustments. Dentures provide a reversible, noninvasive solution that restores essential functions, such as mastication and speech, while improving aesthetics, which is critical for the child's psychosocial development. Additionally, dentures allow for easy modification or replacement as the patient grows. This approach aligns with established protocols for managing pediatric patients with hypodontia secondary to ectodermal dysplasia and provides a foundation for more definitive treatment in adolescence or adulthood.

ED can significantly impact psychosocial well-being, particularly due to visible physical differences and dental anomalies.[10] The condition may affect self-esteem, body image, and social interactions. Psychosocial support and counseling can help individuals and their families cope with these challenges. The cosmetic appearance of patients with severe alopecia may be improved by recommending wigs and the early use of dental prostheses. If phonetics and word articulation abnormalities are noticed, consultation with a speech therapist and otolaryngologist is recommended.

Conclusion

Ectodermal dysplasias are a diverse group of genetic disorders affecting ectodermal tissues. Its management requires a multidisciplinary approach. Pediatricians must address acute complications such as infections and hyperpyrexia, whereas pediatric dentists play a vital role in restoring function and aesthetics and in supporting psychosocial development. Other specialists, including dermatologists, psychologists, otolaryngologists, and speech therapists, may be involved as needed.

Conflict of Interest

Nil

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