

Navigating the Diagnosis of Ectodermal Dysplasia: A Case Series and Review

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Abstract

Ectodermal dysplasia (ED) is a genetic disorder affecting the development or function of the teeth, hair, nails, and sweat glands. ED is a diverse group of inherited disorders that have many overlapping features like absence of sweat glands; the affected individuals may display heat intolerance and elevated body temperature. Patients affected with ED suffer from poor psychological and physiologic development because of unacceptable esthetics and abnormal functions of orofacial structures.

Keywords: Anodontia, Diagnosis, Ectodermal dysplasia, Hereditary disorder, Hypohidrosis, Hypotrichosis.

INTRODUCTION

Ectodermal dysplasia (ED) is a congenital, diffuse, and non-progressive hereditary disorder first described by Thurman which occurs due to disturbances in the ectoderm of the developing embryo.^[1] Mutation occurs in the first trimester of the pregnancy; this is carried by females and usually manifested in males. The different types of ED are caused by the mutation or deletion of certain genes located on different chromosomes. It is a genetic disorder affecting the development or function of the teeth, hair, nails, and sweat glands. Depending on the syndrome, ED can also affect the skin, the lens, or retina of the eye; parts of the inner ear; the development of fingers and toes; the nerve; and other parts of the body.^[2,3] In this report, we discuss three cases of ED who presented to outpatient department with typical features.

CASE REPORTS

Case 1

A 20-year-old male patient with a chief complaint of missing teeth in the upper and lower jaw region was referred to Faculty of Dental Science, DDU, Nadiad. The patient also had a complaint of dry skin and absence of sweating. Familial history revealed that no close relatives including grandparents on both mother and father were affected by the same abnormality.

Extraoral examination, patients were observed to have frontal bossing, prominent supraorbital ridge fine and sparse hair, a prominent chin, and lips were very thick and elevated.

(Figure 1a). Patient's facial profile was observed to be concave. Nasal bridge was flat and the patient had absence of perspiration and so had dry skin. Hyperkeratosis is seen on fingers (Figure 1c). Beard is normal. Nails are normal.

In intraoral examination (Figure 1b), it was observed that the patient had completely edentulous lower arch, and upper arch was observed to have 6 teeth that were namely 15, 16, 17 and 25, 26, 27. Bud-shaped teeth are seen. Mandibular and maxillary ridge was thin and highly resorbed with the presence of dry oral mucosa.

Case 2

A 14-year-old boy with a chief complaint of congenitally missing teeth in the upper and lower jaw region was referred to the Faculty of Dental Science, DDU, Nadiad. The patient complained of malformed teeth since childhood. A history of delayed tooth eruption in the patient was provided by his parents, missing lower front teeth and was edentulous for years after birth. He was born of a normal delivery. Examination of his parents and siblings did not reveal any similar abnormality. There were no similar features among other members of the family.

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Extraoral examination (Figure 2a) revealed hair abnormalities including sparse eyelashes and thin eyebrows. He had a square forehead and frontal bossing. His lower and upper lips were protruded. His skin appeared to be dry and rough. The hands did not show keratosis (Figure 2c). The boy was moderately built and poorly nourished.

Intraoral examination (Figure 2b) disclosed a comparatively dry mucosa and included malformed and missing teeth. The presence of 11 and 21 conical anterior teeth and bud-shaped 16, 26, and 27 posterior teeth are seen. The alveolar ridge appeared to be very thin and short resulting in the appearance of vestibule to be shallow.

Case 3

A 17-year-old girl reported with a chief complaint of congenitally missing teeth to the Faculty of Dental Science, DDU, Nadiad. The patient gave a history of delayed tooth eruption, multiple missing permanent teeth, and malformed teeth. No contributing family history was found.

Extraoral examination revealed no hair abnormalities. She had a square forehead with frontal bossing and supraorbital ridge (Figure 3a). The girl's body was moderately built and poorly nourished and her fingernails were brittle with keratosis on the distal phalanges (Figure 3c) with skin appeared dry and rough.

In intraoral examination (Figure 3b), it was observed that the patient had 10 teeth with mixed dentition with malformed teeth that were namely 11, 52, 16 and 21, 62, 26 in the maxillary arch and 36, 36 and 45, 46 in the mandibular arch. Anterior teeth appeared conical and posterior teeth were bud shaped. The alveolar ridge of the mandibular and maxillary arch appeared very thin and highly resorbed with the presence of dry mucosa.

Orthopantomograms (Figure 4a-c) show malformed teeth in the maxilla and mandible and multiple missing tooth buds confirming hypodontia in both arches. The alveolar ridges were underdeveloped and atrophic.

ED was diagnosed based on history and clinic-radiographic evaluation in all three cases presented here. These patients



Figure 1: (a) Shows the Extraoral photograph showing frontal bossing, prominent supraorbital ridge, thick lower lip, sparse hair. (b) Intraoral photograph showing six permanent teeth were present- maxillary right second premolar, first molar, second molar; maxillary left second premolar, first molar, second molar. (c) Palmer surface shows Hyperkeratosis on fingers.



Figure 2: (a) Shows the Extraoral photograph showing frontal bossing, thick upper and lower lips, sparse hair, thin eyebrows. (b) Intraoral photograph showing six permanent teeth were present- maxillary right central incisor, first molar; maxillary central incisor, first molar, second molar. (c) Palmer surface of hands showing brittle nails.



Figure 3: (a) Shows the Extraoral photograph showing frontal bossing, prominent supraorbital ridge. (b) Intraoral photograph showing mixed dentition with malformed teeth. (c) Dorsal surface shows Hyperkeratosis on fingers.

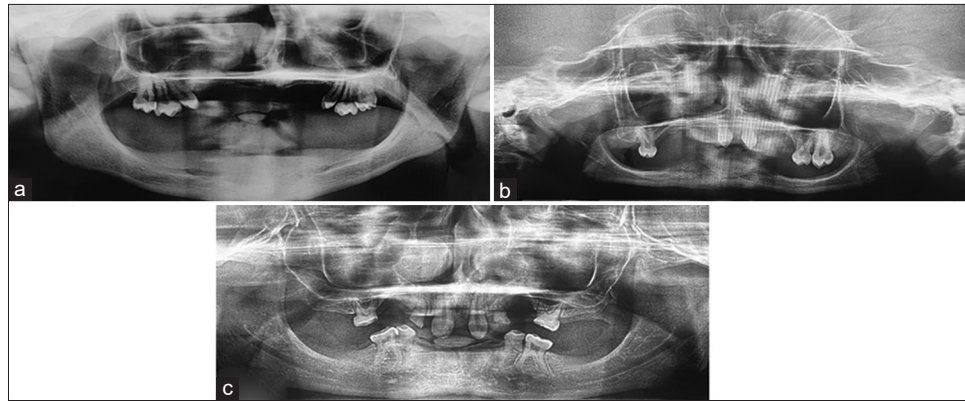


Figure 4: (a-c): Showing malformed teeth in the maxilla and mandible, multiple missing tooth buds confirming hypodontia in both arches. The alveolar ridges were underdeveloped and atrophic.

were referred to the prosthodontia department for rehabilitation and advised to consume water frequently to maintain proper hydration.

DISCUSSION

EDs are an inherited group of disorders that share in general developmental defects concerning a minimum two of the major structures characteristically held to derive from embryonic ectoderm – hair, teeth, nails, skin, and sweat glands. Sweat glands may be absent or reduced in number or non-functioning due to which the affected individuals may display heat intolerance and elevated body temperature. The triad of nail dystrophy (onchodysplasia), phalacrotrichosis or hypotrichosis (scanty, fine light-weight hair on the scalp and eyebrows), and palmoplantar hyperkeratosis is typically accompanied by a scarcity of sweat glands (hypohidrosis) and a partial or complete absence of primary and/or permanent dentition.^[4-6]

Freire-Maia and Pinheiro proposed the first classification system of the ED in 1982 and provided additional updates in 1994 and 2001. The patients were classified into subgroups which were based on the presence or absence of trichondysplasia (abnormal hair), abnormal dentition, onchodysplasia (abnormal nails), and dyshidrosis (abnormal or missing sweat glands).^[1,8]

According to them, ED is classified into different subgroups^[1,5]

1. ED1: Presence or absence of hair anomalies or trichodysplasia
2. ED2: Dental anomalies
3. ED3: Nail abnormalities or onychodysplasia
4. ED4: Eccrine dysfunction or dyshidrosis.

ED is basically divided into two broad categories, i.e., hypohidrotic form (Christ–Touraine syndrome) which is X-linked and is characterized by the classical triad of hypodontia, hypotrichosis, and hypohidrosis and the other category, i.e., hydrotic form (Clouston syndrome), which also affects the teeth, hair, and nails sparing the sweat glands.^[1,9]

Patients affected with a hereditary deformity, such as ED, suffer from poor psychological and physiologic development because of unacceptable esthetics and abnormal functions of

orofacial structures.^[2,8,10] It was suggested that the pediatric dentist is best suited to manage these special needs as their major goal is to provide the child with an oral apparatus that provides optimal esthetics and function to allow the patient to develop physically, emotionally, and socially.^[1,8]

In cases presented here, we found clinical features of congenitally missing teeth (hypodontia), conical-shaped teeth, sparse hair, dry skin, and radiographically atrophic ridges. Based on clinico-radiographic evaluation, the diagnosis of ED is kept.

CONCLUSION

ED is a diverse group of inherited disorders that have many overlapping features. The clinical manifestations of ED cause significant social problems in individuals affected by it as it disturbs the oral functions and normal bodily functions of the patients. An earlier diagnosis and rehabilitation by multidisciplinary collaborative efforts are the key to successfully manage ED.

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