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Hypohidrotic ectodermal dysplasia: A rare case report

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Abstract

Ectodermal dysplasia is a rare congenital disorder caused by primary defects in the growth of embryonic ectoderm tissues and its accessory appendages including skin, hair follicles, nails, teeth, eccrine glands, and sebaceous glands. Ectodermal dysplasia is caused by mutations in specific genes. The clinical characteristics of ectodermal dysplasia are sparse scalp hair, missing teeth, thin and brittle skin and deformed nails, and inability to sweat due to lack of sweat glands. The present study reports a case of a 12-year-old girl with ectodermal dysplasia

Keywords: Ectodermal dysplasia; Congenital disorder; Mutation

Introduction

Ectodermal dysplasia (ED) is a rare heterogeneous group of genetic disorders that are categorized by major defects in the growth of two or more tissues resulting from the embryonic ectoderm. The prevalence of ectodermal dysplasia is about 1 in 10,000-100,000 live births (1). The clinical characteristics of ectodermal dysplasia are sparse scalp hair, missing teeth, thin and brittle skin and deformed nails, and inability to sweat due to lack of sweat glands (2).

Hypohidrotic ED (Christ-Siemens-Touraine syndrome) and hidrotic ED (Clouston syndrome) are the most common forms of ED. Hypohidrotic ED is the most phenotype of ED and is considered as the inherited X-linked recessive disorder (the disorder *is located on the* X chromosome) (3). ED is typically characterized by hollow cheeks, saddle-nose deformity, unusually prominent forehead, and thick and reversed lips (4). *D*ental manifestations of ED include peg teeth or microdontia, the absence of one or more teeth (anodontia, hypodontia), and delayed tooth eruption of the permanent dentition (5). *The sweat glands in patients with ED, specially hypohidrotic ED, may function abnormally* due to the absence of eccrine sweat glands. There may be genetic hair disorders and abnormalities in the hair structure including hair-shaft torsion and distorted and ruffled cuticle (3). Other clinical manifestations of ectodermal dysplasia are brittle and thin nails, eye abnormalities, tearing problem, and abnormal light sensitivity and intolerance to visual perception of light (3). *The m*ilder forms of the ED may remain undetected until signs of disorder start to disturb the child's daily life or development. Genetic assessment, radiological examination, and clinical examination help to determine and confirm the clinical diagnosis ectodermal dysplasia. The present study reports a case of a 12-year-old girl with ectodermal dysplasia.

Case Report

A 12-year-old female patient with the complaint of missing teeth and difficulty in eating was referred to the department of oral and maxillofacial radiology, School of Dentistry, Ahvaz Jundishapur University of Medical Sciences for cone-beam computed tomography (CBCT) imaging. The marriage history of her parents revealed that they had a consanguineous marriage. There was no history of ED in her family.

Clinical examination showed decreased hair growth and thinning hair, scanty eyebrows, thick and prominent lips, protrusion of the forehead, sunken nasal bridge (saddle nose), and decreased midface depth (Figure 1).

Figure 1. Extraoral view of patient

The patient suffered from dry skin and reduced ability to sweat (hypohidrosis). There was no abnormality in the nails and fingers and toes (ectrodactyly). The intelligence quotient (IQ) of the patient was at the normal level. The cone beam 3D reformatted images showed that the patient had a total of 5 teeth (three right maxillary central and lateral incisors and two mandibular anterior teeth) and a retained root within the jawbone and 27 missing permanent teeth. The teeth were cone-shaped, pointy, and smaller than the average (Figure 2 and 3).

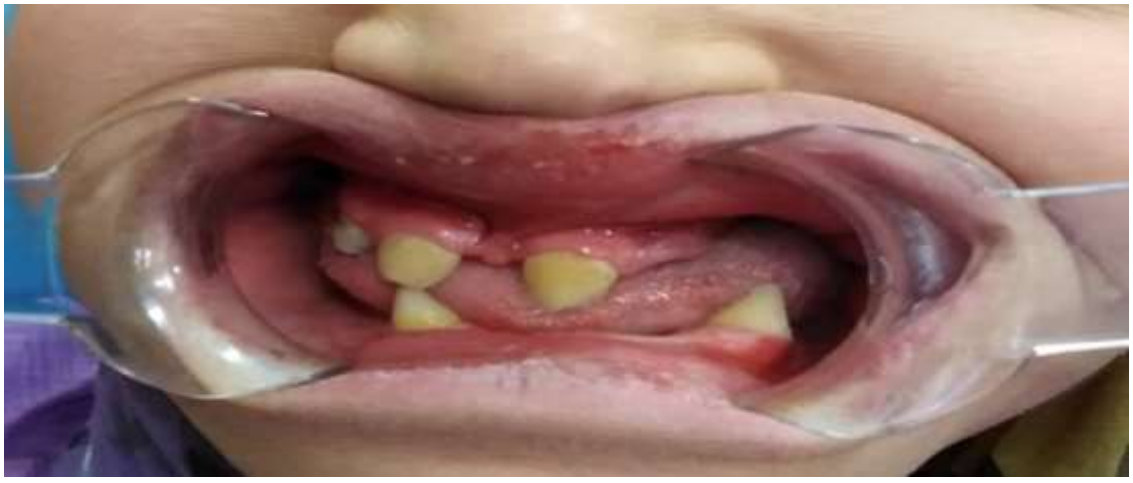

Figure 2. Intraoral view of the patient

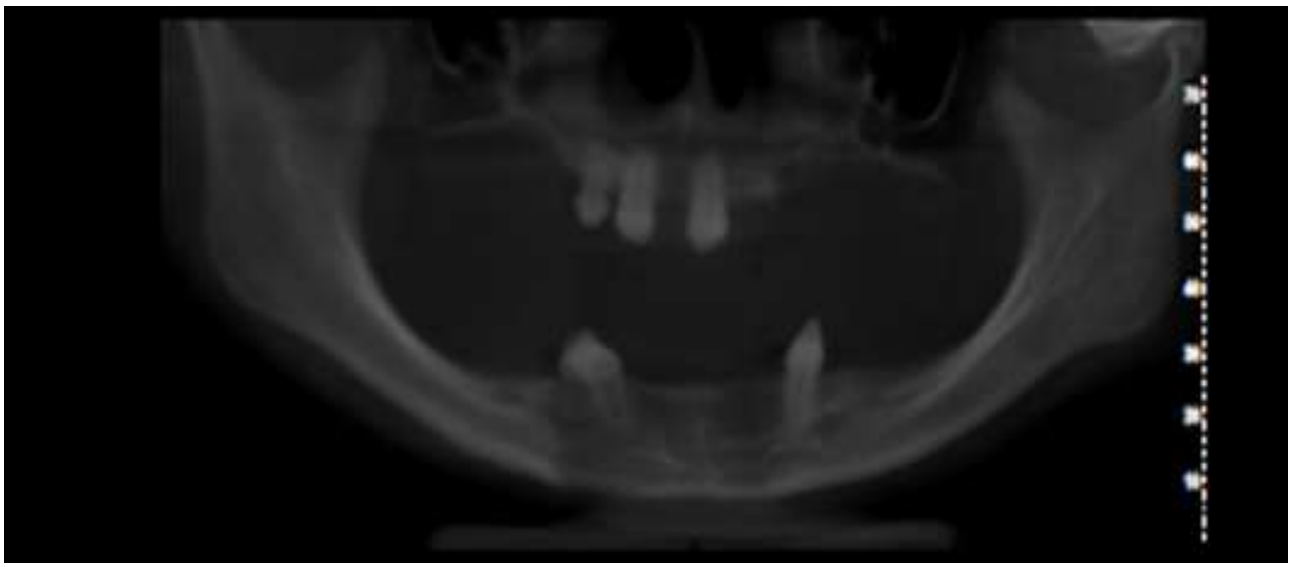

Figure 3**.** Dentomaxillofacial imaging with panoramic views

Incomplete evolution of alveolar ridges with a significant reduction in the amount of alveolar bone in the vertical and horizontal dimensions behind the maxilla and mandible was observed (Figure 4 and 5). The extraoral and intraoral clinical examination confirmed the presence of developing ED. Reconstruction and grafting the atrophied ridges and implant placement in the grafted areas were recommended for the treatment plan.

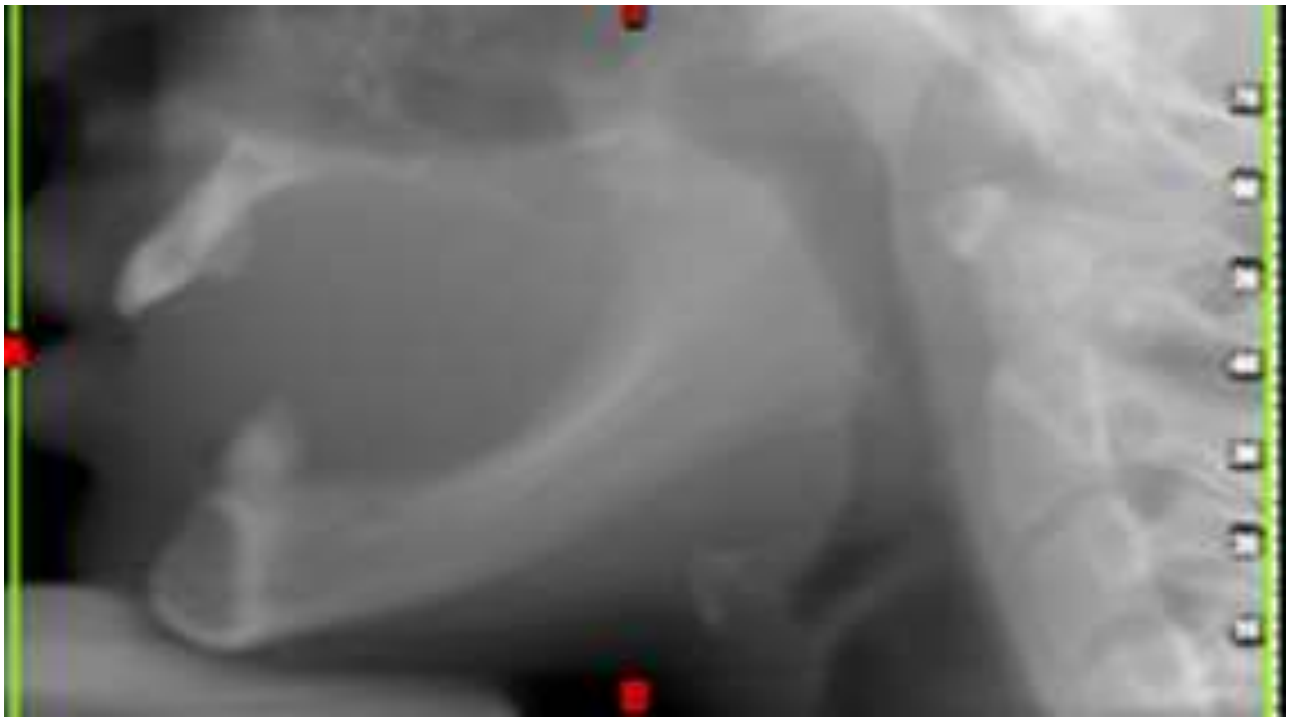

Figure 4. Lateral radiographic view

Figure 5.Three-dimensional CBCT image

Discussion

The ectodermal dysplasias are a hereditarily varied group of disorders that include 186 discrete diseases with 64 gene mutations (5). Ectodermal dysplasia is basically divided into two broad groups, including the hypohydrotic form (Christ Touraine syndrome) and the hydrotic form (Clouston syndrome). The most common form of hypohydrotic is characterized by the classic triad of hypodontia, hypotrichosis, and hyperhidrosis (abnormal sweating). The hydrotic form is characterized by defects in the function and development of the teeth, hair, nails and/or sweat glands. In most cases, sparse and lusterless fair hair, a temperature above the normal range (*pyrexia),* and inability to sweat (*anhidrosis*) are detectable (6,7). These patients have IQ scores within the normal range. The most frequent extraoral manifestations include large and low set ears, unusually prominent forehead, sunken appearance of cheeks, sunken nasal bridge, thick everted lips and pointed chin, and wrinkled darkly pigmented skin (7). The most common oral manifestation of ED includes hypodontia or anodontia of deciduous and permanent teeth followed by conical-shaped teeth (8). In rare instances, the mandible and/or maxilla may be edentulous and the alveolar processes may not develop in the base of the jaws due to anodontia (6). Ectodermal dysplasia is diagnosed according to clinical examination and family history (9).

In the present study, the patient had several maxillary and mandibular missing permanent teeth, and conical-shaped teeth were observed in radiological examination. In clinical examination, sunken cheeks, sparse and fair hair, frontal bossing, and scanty eyebrows were evident. The results of panoramic and CBCT examinations confirmed the diagnosis of ectodermal dysplasia.

Bhakta et al. reported a rare case report of ectodermal dysplasia with clinical manifestation similar to the present study, except fusion of the 2nd and 3rd digit of the right foot with nail dystrophy (3). Meshram et al. reported a case of hypohidrotic ectodermal dysplasia with clinical manifestation, except the hairs on the scalp were sparse and hypopigmented (2).

In children, in most cases, the preferred treatment for hypodenia dental disorders is removable partial dentures along with composite veneer in malformed and peg-shaped teeth that allow the child to eat properly, look natural, and speak correctly. Furthermore, the treatment plan improves their social lives.

In adult patients with ED and hypodontia and oligodontia, alveolar ridges are severely hypotophic, Therefore, a treatment plan including bone augmentation followed by implant-retained overdenture will be satisfactory and optimizes esthetics and function of teeth set-up (10). Dentists and oral and maxillofacial radiologists are the first groups to identify ectodermal dysplasia patients, so they should be fully aware of the clinical and intraoral manifestations of the disease and be able to guide the patient to health services for appropriate treatment.

Conclusion

Clinical manifestations of ectodermal dysplasia may cause a variety of socio-psychological and functional problems and disrupt daily life. The role of oral and maxillofacial radiologists in the early detection and successful management of ED is very important. The early and prompt diagnosis of the disease can provide appropriate care and management

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