Case Report

Ectodermal dysplasia - A rare case report

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Abstract

Ectodermal dysplasia (ED) is a rare genetic disease caused by developmental disturbances of embryonic ectoderm derived tissues, organs, and other accessory appendages. The congenital missing of teeth is usually bilateral. Anodontia or hypodontia may be associated with other ectodermal disturbances, such as anhidrosis, asteatosis, hypotrichosis, and salivary glands defects. One such case report of ED is demonstrated here.

Keywords: Anhidrosis, hypodontia, hypohidrosis

Introduction

Ectodermal dysplasia (ED) is one of the rare hereditary disorders that demonstrate primary developmental defects of two or more tissues derived from the ectoderm. Primarily affected tissues are the skin, hair, nails, eccrine glands, and teeth. The disorders are congenital, diffuse, and nonprogressive.[1,2] The most common syndromes within this group are hypohydrotic (anhidrotic) ED and hidrotic ED. Hypohydrotic ED (also known as Christ--Siemens--Touraine syndrome) is the more common phenotype and is usually inherited as an X-linked recessive trait. It is characterized by several defects (e.g., hypohidrosis, anomalous dentition, onychodysplasia, and hypotrichosis). Typical facies are characterized by frontal bossing, sunken cheeks, a saddle nose, thick and everted lips, wrinkled and hyperpigmented skin around the eyes, and large, low-set ears. Dental manifestations include conical or pegged teeth, hypodontia or complete anodontia, and delayed eruption of permanent teeth. Eccrine sweat glands may be absent or sparse and rudimentary, particularly in those with hypohydrotic ED. In some cases, mucous glands are absent in the upper respiratory tract and in the bronchi, esophagus, and duodenum. Other common signs are short stature, eye abnormalities, decreased tearing, and photophobia.[3]

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A 16-year-old female patient came to outpatient department of OMDR at Pacific Dental College and Hospital, Udaipur, with the chief complaint of multiple missing teeth in relation to upper and lower arch. She also gave history of loss of hair growth (alopecia), dryness and decreased sweating. On clinical examination, she had frontal bossing, a prominent supraorbital ridge, sunken cheeks, thick lower lip, sparse hair, scanty eyebrows, and low-set and overfolding ears [Figure 1]. She also had fusion of 2nd and 3rd digit of right foot with nail dystrophy [Figure 2a and b]. Upon intraoral examination, Sixteen permanent teeth were absent including right and left lower and upper central incisors, lower and upper third molars, right lower canine and second molars, left lower lateral incisor, left upper canine, right and left upper first premolar, right upper second premolar, left upper second molar, and she also had a cone-shaped teeth [Figure 3]. Orthopantomogram (OPG) [Figure 4] was taken which revealed multiple missing teeth, generalized interdental bone loss, and retained tooth 75. Hand wrist radiograph [Figure 5] revealed

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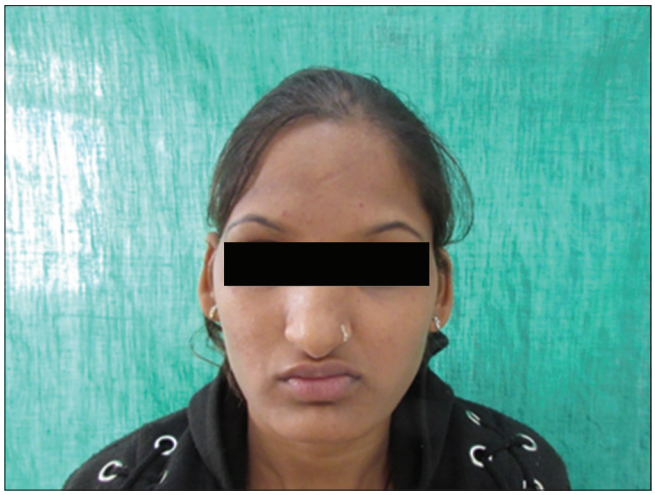

Figure 1: Extraoral view of patient

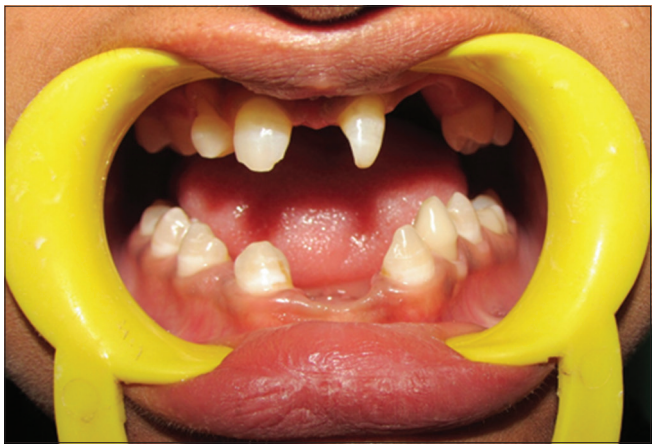

Figure 3: Intraoral view of the patient

shortened middle and distal phalanges of all fingers. On the basis of clinical and radiographic features, ED was diagnosed.

Discussion

The EDs are a heterogeneous group of hereditary disorders which occur approximately one in every 100,000 births which are caused by primary developmental defects of two or more embryonic ectoderm derived tissues.[4]

ED is basically divided into two broad categories, i.e., hypohidrotic form (ChristTouraine Syndrome) which is X-linked and is characterized by the classical triad of hypodontia, hypotrichosis, and hypohydrosis and the other category, i.e., hydrotic form (Clouston syndrome), which also affects the teeth, hair, and nails sparing the sweat glands. Extraorally fine, sparse, lusterless fair hair is seen over the scalp along with extensive scaling of the skin and unexplained pyrexia and heat intolerance most commonly occurs due to anhidrosis. Normal intelligence is observed.[5-7] The other extraoral features are frontal bossing, sunken cheeks, depressed nasal bridge, thick everted protuberant

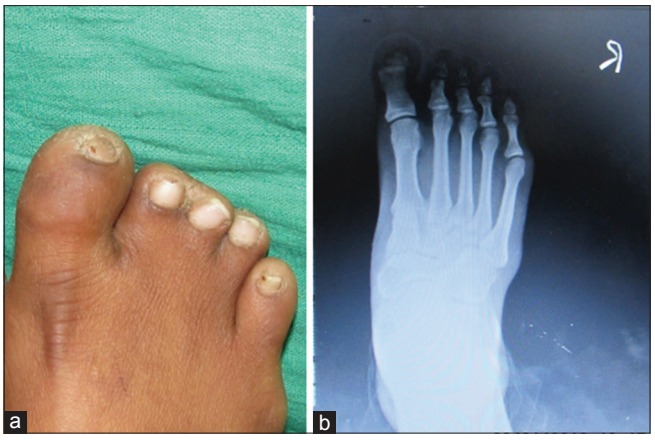

Figure 2: (a) Extraoral view of foot (b) Radiographic view of foot

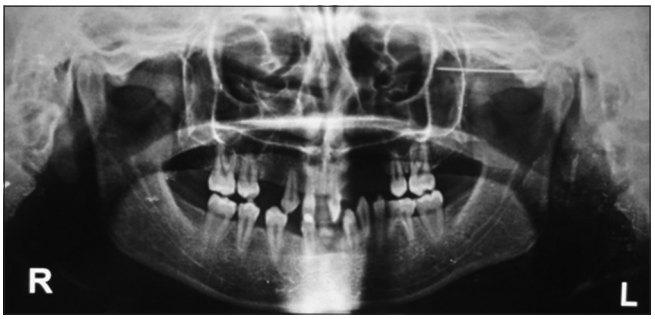

Figure 4: OPG of the patient

lips, wrinkled hyperpigmented periorbital skin, and a large low set of ears.[6] Hypodontia or anodontia of deciduous and permanent dentition associated with conical-shaped teeth is the most common oral symptom.[8] The patient reported here had involvement of hair, sweat glands, and she also had conical shaped teeth. In addition, she had frontal bossing, sunken cheeks, nail dystrophy, and low set ears. These clinical features were supportive in diagnosing hydrotic ED.

Intraorally missing permanent teeth are most commonly present, the maxillary central incisors and canines present with a conical crown form. In rare instances, one or both jaws may be edentulous and the alveolar processes may not develop due to the absence of teeth.[5] Diagnosis is based on family history, thorough clinical and radiographic examination.[9] The patient reported here had multiple missing permanent teeth in upper and lower jaws and she had bilateral conical crown-shaped lateral incisor and canine. In our case, radiographs like OPG and hand wrist radiographs were taken to confirmed the diagnosis.

Conclusion

EDs are rare genetic disorders that have many overlapping features and it is difficult to classify them. The clinical manifestations of ED cause significant social problems in affected individuals. It disturbs both oral functions and normal body functions of the patients. The key of success for management of ED is quick

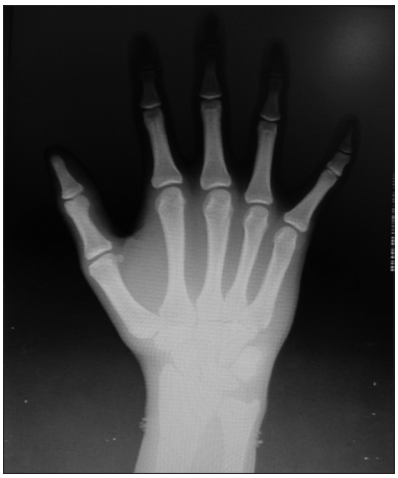

Figure 5: Hand wrist radiograph of the patient

diagnosis and prosthetic rehabilitation by the multidisciplinary approach.[10] The role of pedodontists is very important for successful management of ED because pedodontists are better trained in child psychological and behavioral management.[8]

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Conflicts of interest

There are no conflicts of interest.

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