Oral Findings of Ectodermal Dysplasia and Literature Review

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Abstract

Ectodermal dysplasias represent a large and comlex group of diseas charracterized by various defects in hair, nails, teeth sweat glands. This paper proposes the possible craniomaxillofacial deformative consequences associated with ectodermal dysplasias and embriyonic malformations.

Fifteen ectodermal dysplasia patients had a clinical examination and underwent radiographic, 3D cone beam dental volumetric computerized tomography and photographic assessment.

All patients had tooth ageneses (from hypodontia to anodontia), associated with cutaneous dyshidrosis and hair and nail dystrophy. Most of the them had a short face, sparse or absent hair, unusual facial concavity, a maxillary retrusion and a relative mandible protrusion also they had visual problems, respiratory problems.

Dental treatment is often necessary in patients with some forms of ED and some children may need dentures as early as 2 years of age.19 It is important to seek dental advice early as maintenance of the alveolar ridge is important for later dental intervention. (*International Dental and Medical Disorders 2008; 1: 43-49)*

Keywords: Ectodermal dysplasia, dental volumetric CT, Anodontia, Hypodontia.

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Introduction

The ectodermal dysplasias (EDs) comprise a large, heterogeneous group of inherited disorders that are defined by primary defects in the development of 2 or more tissues derived from embryonic ectoderm. The original constructional theme encoded in ectoderm diverges into epidermis, hair, sweat and milk glands, and the mineralized crystalline anvils of teeth, under the direction of local signals emanating from the underlying mesoderm. The intimate origins of these diverse ectodermal structures account for the wide spectrum of dysplasias. Clinically the hair (hypotrichosis, partial or total alopecia), nails (dystrophic, hypertrophic, or abnormally keratinized), teeth (enamel defects or absence), and sweat glands (hypoplastic or aplastic) are usually affected. [1,2,3,4.]

An estimated incidence of ED is about 7 in 10,000 births and all mendelian modes of inheritance have been reported.[5] Of more than 190 EDs described, the molecular basis has been elucidated for more than 30 of them.[6]

The most frequently reported manifestation of ectodermal dysplasia is hypohidrotic dysplasia, also termed Christ-Siemens- Touraine syndrome and anhydrotic dysplasia. Patients with this form of ectodermal dysplasia exhibit the following clinical traits: hypothricosis, hypohidrosis, and cranial abnormalities.[7] X-linked hypohidrotic ED (Christ-Siemens-Touraine syndrome) is the most frequent form; the diagnosis is usually made with the identification of hypotrichosis, characteristic facial features, hypohidrosis (and more rarely anhidrosis), and teeth abnormalities. The nails are usually normal. Abnormalities in the development of tooth buds result in hypodontia and peg-shaped or pointed teeth. [8]

Dental defects represent a core clinical feature of many EDs: anodontia, polydontia, dysplastic teeth, retained primary teeth, deficient enamel development (amelogenesis imperfecta), dentine deficiency (dentinogenesis imperfecta), and underdevelopment of the alveolar ridge.[9] In some EDs, the number of erupted teeth is reduced, the

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spacing of the teeth disrupted, and the peridontium affected.

Dental volumetric CT has attracted considerable attention as a new diagnostic. Various radiographic imaging techniques are available to visualize anatomical and pathological details. Although periapical and panoramic radiography produce acceptable details in the mesial-distal direction, the observation of details in the bucco-lingual dimension is inadequate. Three-dimensional computerized tomography (CT) is used for the diagnosis and treatment planning of periapical pathosis. [10,11.] Dental volumetric CT was used for diagnosis and treatment planning in this study.

Patients and Methods

This retrospective study was carried out on 15 patients (X males, X females, aged 5-25 years) with a diagnosis ED, in the Faculty of Dentistry, Dicle University

Clinical and radiographical examination for diagnosis and therapy was conducted and it is included the skull, face, hair, teeth, nails, skin, lungs, sweat glands. Also three of patiens radiographies were taken by dental volumetric CT (iCAT) (Figure 1a, 1b, 2a, 2b, 3a, 3b)

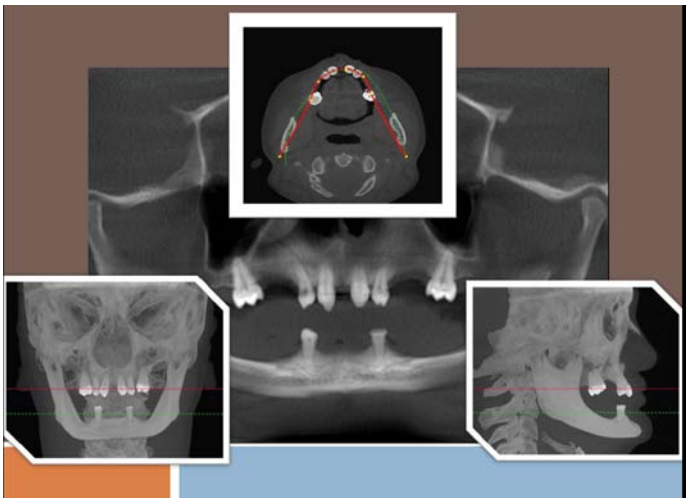

Fig.1a 3D Dental volumetric CT sample (Case 1)

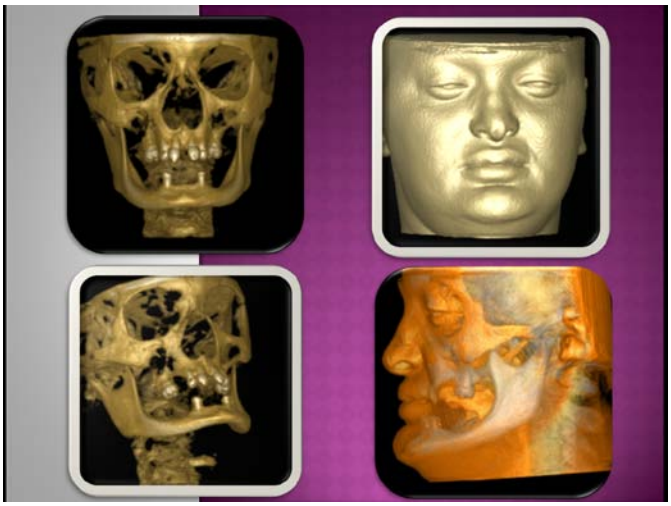

Fig.1b 3D Dental volumetric CT sample (Case 1)

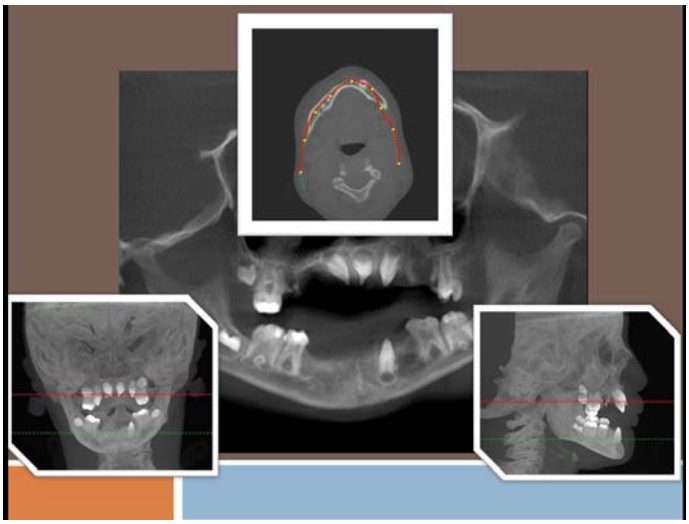

Fig.2a 3D Dental volumetric CT sample (Case 2)

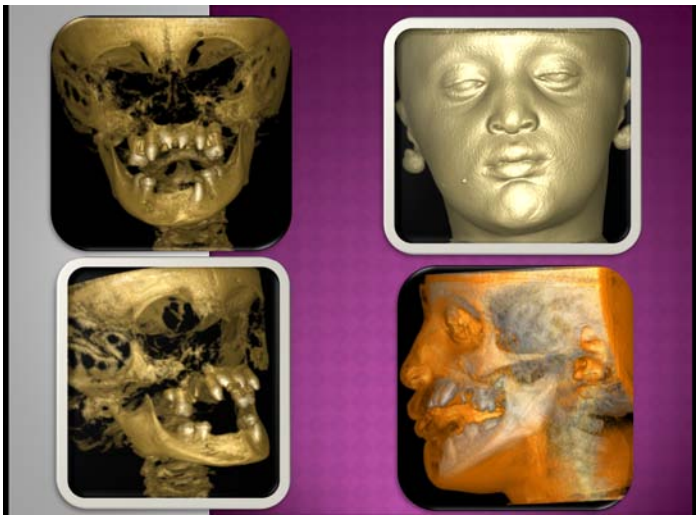

Fig.2b 3D Dental volumetric CT sample (Case 2)

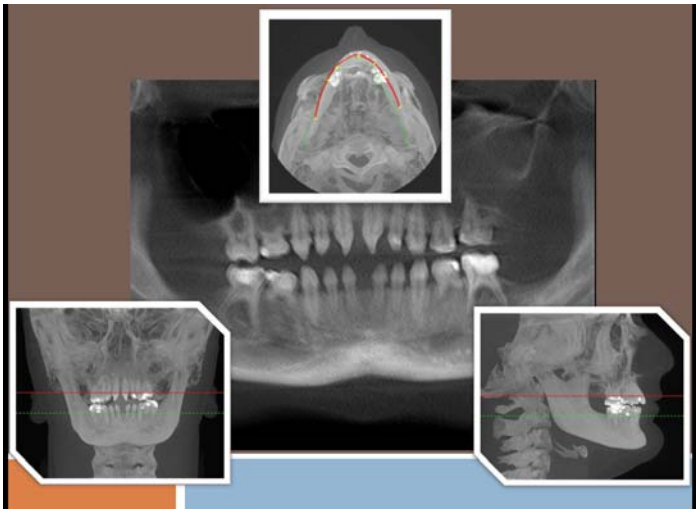

Fig.3a 3D Dental volumetric CT sample (Case 3)

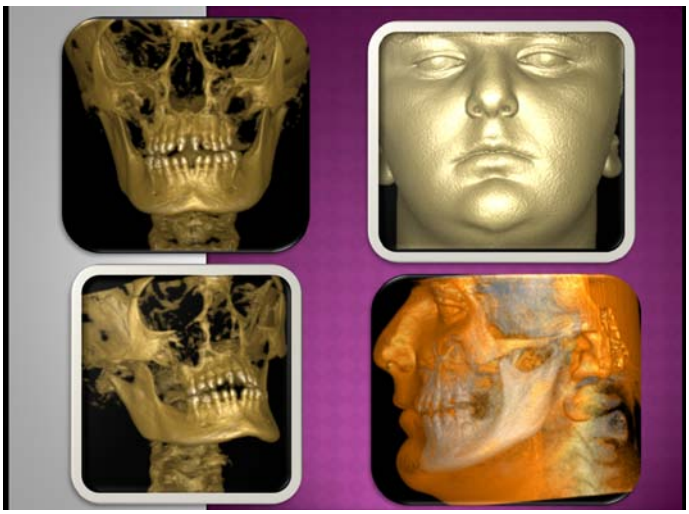

Fig.3b 3D Dental volumetric CT sample (Case 3)

All major sign of ED were studied, such as sparse hair (trichodysplasia), smooth skin (hypohidrosis), abnormal finger and toe nails, skull and face abnormalities and the pedigree of the patients were researched. The hair is generally blond and scanty. Hypotrichosisand partial or total alopecia are frequently reported. Body hair follicles are usually diminished or absent (Fig 4, 5). The nails may be dystrophic, hypertrophic, abnormally keratinised, thickened, discoloured, striated, split or fragmented. The epidermis was dry, fine and smooth, hypopigmented, with patches of hyperkeratosis and:or eczematous (Fig 6a, 6b, 7a, 7b).

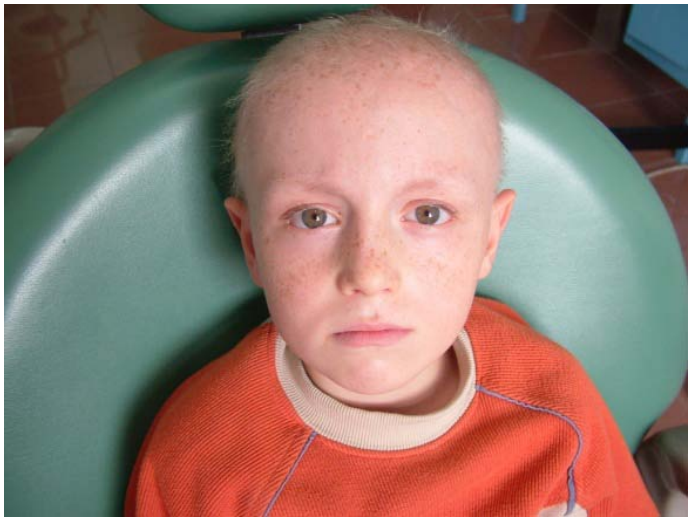

Fig. 4 Extra-oral appearance.

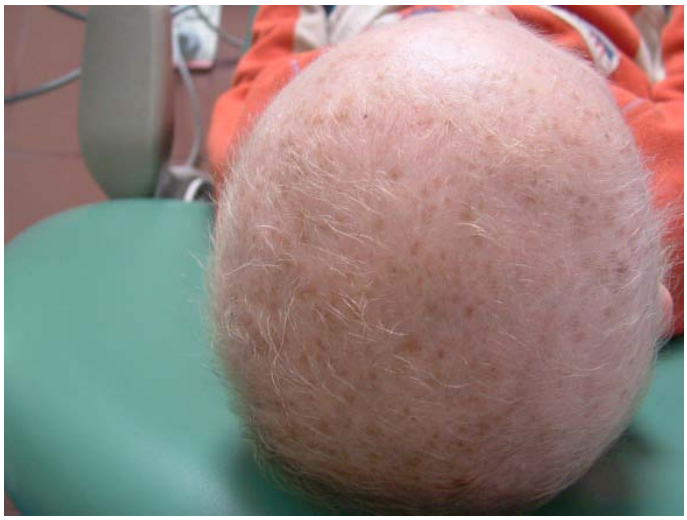

Fig. 5 Appearences of diminished or absent hair follicles.

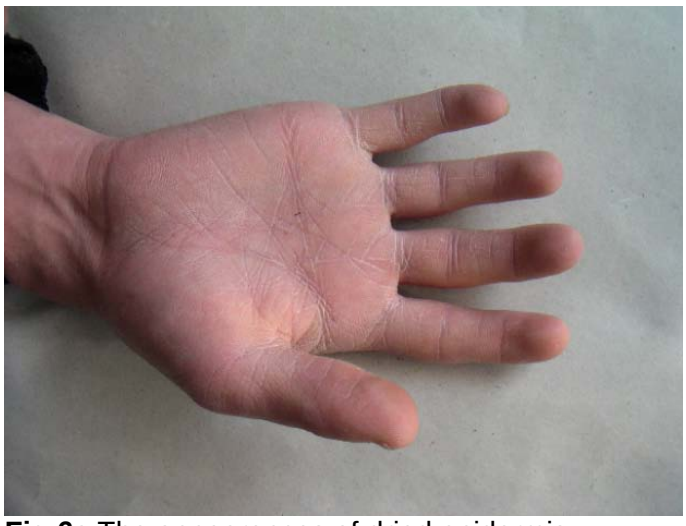

Fig.6a The appearences of dried epidermis.

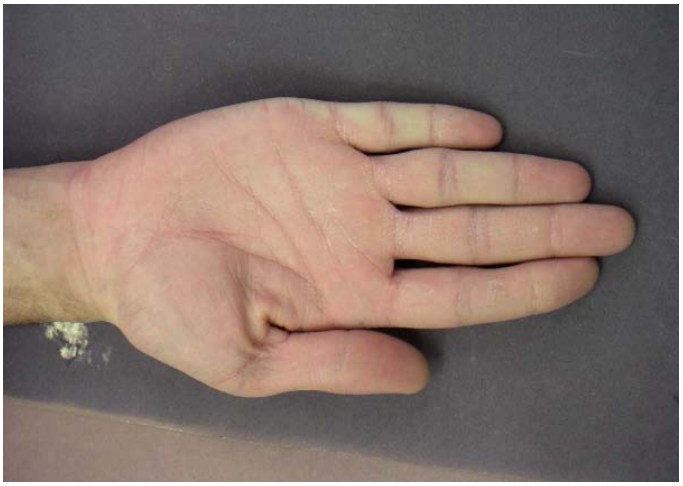

Fig.6b The appearences of dried epidermis.

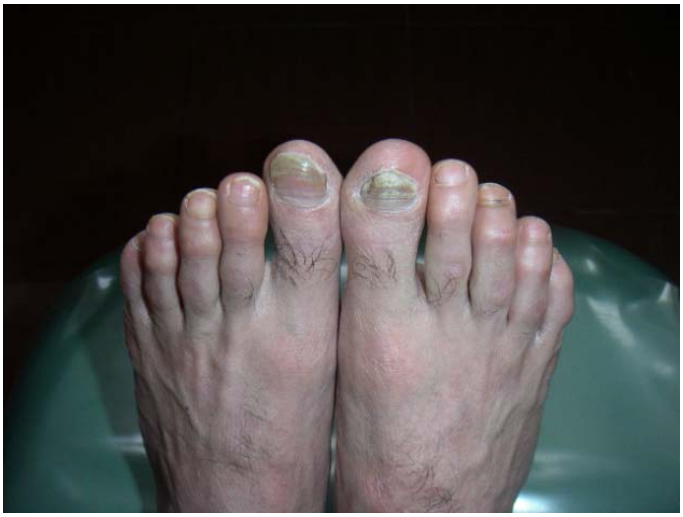

Fig.7a The appearences of dystrophic, hypertrophic, abnormally keratinized foot nails.

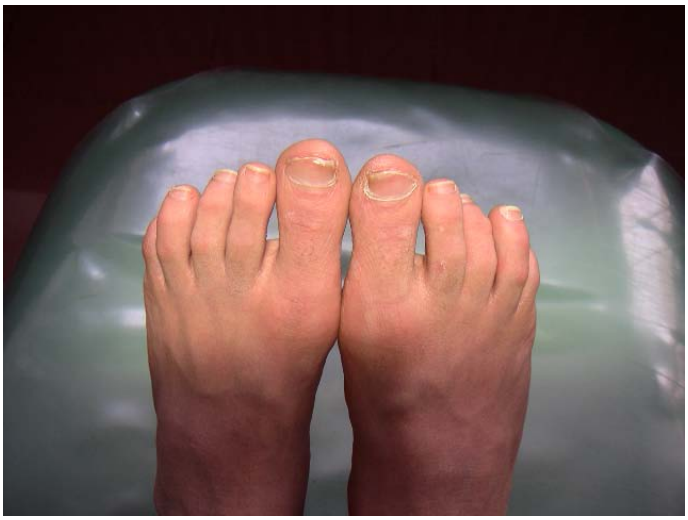

Fig.7b The appearences of dystrophic, hypertrophic, abnormally keratinized foot nails.

Each patient had the benefit of a rigorous clinical examination for diagnosis and therapy: minor or major abnormalities had to be detected in both patients and family. Oligodontia or anodontia are very common features, but rudimentary or conical teeth and enamel dysplasia may be observed (Fig 8a, 8b, 9a, 9b, 10a, 10b).

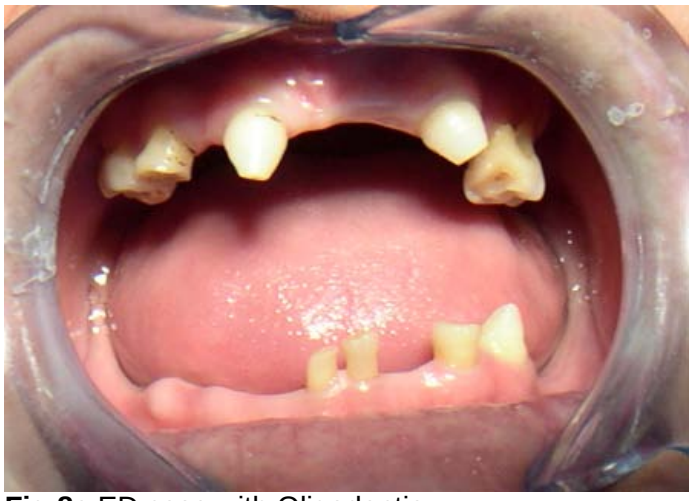

Fig.8a ED case with Oligodontia.

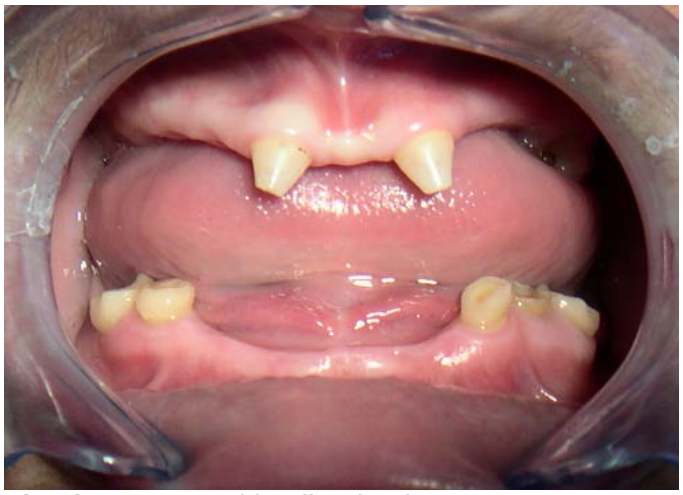

Fig.8b ED case with Oligodontia.

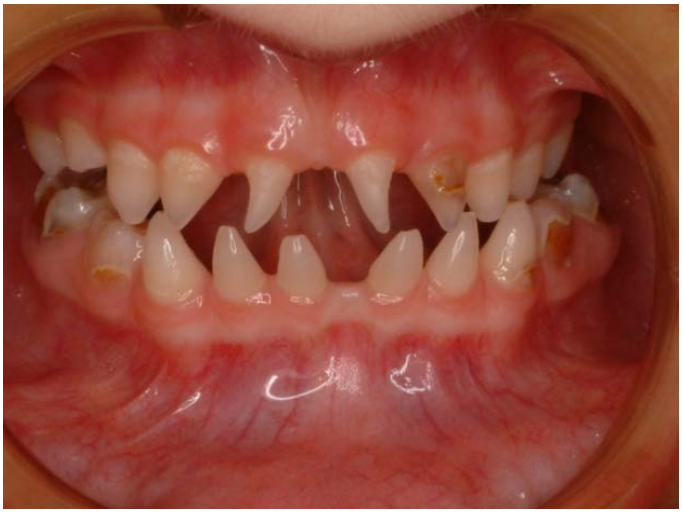

Fig.9a ED case with conical teeth.

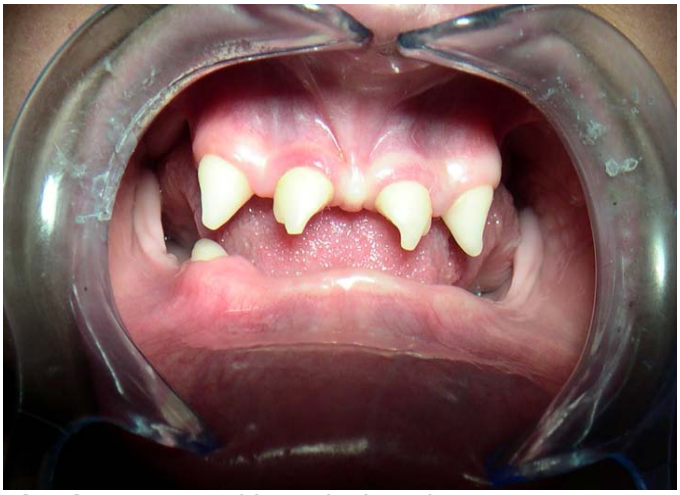

Fig.9b ED case with conical teeth.

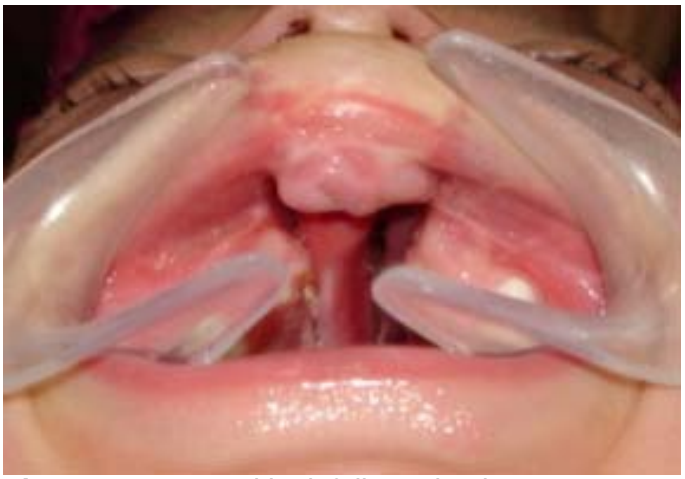

Fig.10a ED case with cleft lip and palate.

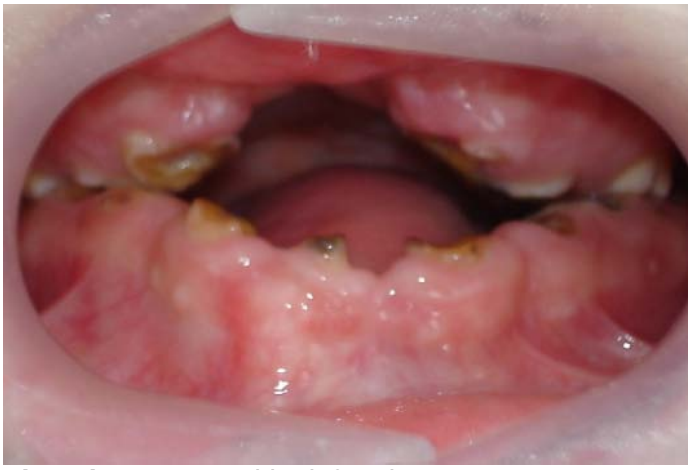

Fig.10b ED case with cleft palate.

Discussion

Clinical diagnosis of ectodermal dysplasia may be difficult. Hypohidrotic ectodermal dysplasia is a rare inherited multisystem disorder that belongs to the group of diseases known as ectodermal dysplasias. ED patients typically affect the hair, teeth, nails, and/or skin. ED is primarily characterized by partial or complete absence of certain sweat glands (eccrine glands), causing lack of or diminished sweating (anhidrosis or hypohidrosis), heat intolerance, and fever; abnormally sparse hair (hypotrichosis); and absence (hypodontia) and/or malformation of certain teeth. Many individuals with HED also have characteristic facial abnormalities including a prominent forehead, a sunken nasal bridge (so-called "saddle nose"), unusually thick lips, and/or a large chin. The skin on most of the body may be abnormally thin, dry, and soft with an abnormal lack of pigmentation (hypopigmentation).

In many cases, affected infants and children may also exhibit underdevelopment (hypoplasia) or absence (aplasia) of mucous glands within the respiratory and gastrointestinal (GI) tracts and, in some cases, decreased function of certain components of the immune system (e.g., depressed lymphocyte function, cellular immune hypofunction), potentially causing an increased susceptibility to certain infections and/or allergic conditions. Many affected infants and children experience recurrent attacks of wheezing and breathlessness (asthma); respiratory infections; chronic inflammation of the nasal passages (atrophic rhinitis); scaling, itchy (pruritic) skin rashes (eczema); and/or other findings.

The oral and nasal mucous glands, the salivary glands and the mammary glands may be hypoplastic or absent. The mucous glands may be absent in the whole respiratory tree. Affected patients may suffer from recurrent infections of the respiratory ways. The absence of the salivary glands may lead to xerostomia. [3]

The gastrointestinal mucous glands may be absent. Affected patients may present with dysphagia . [3]

In one case-controlled study of 68 persons with oligodontia, 57% had disturbances in hair, nails, and/or sweat production in addition to defective teeth, and were classified as having ED.[13] Disturbance of the enamel matrix may occur, making the teeth more susceptible to caries, and altering the shape of the teeth, leading to a pegged appearance and additional accessory cusps. X-linked hypohidrotic ED (Christ-Siemens- Touraine syndrome) is the most frequent form; thediagnosis is usually made with the identification of hypotrichosis, characteristic facial features, hypohidrosis (and more rarely anhidrosis), and teeth abnormalities. The nails are usually normal. Abnormalities in the development of tooth buds result in hypodontia and peg-shaped or pointed teeth.[14] The hypodontia varies in each case, but usually only 5 to 7 permanent teeth are present, the teeth are smaller than average, and the eruption of teeth is often delayed.[15]

Dental treatment is often necessary in patients with some forms of ED and some children may need dentures as early as 2 years of age.[16,17,18]

It is important to seek dental advice early as maintenance of the alveolar ridge is important for later dental intervention. Prosthetic teeth are implanted in adults for mastication and speech. Importantly, aesthetic dental interventions in patients with ED and malformed teeth and malocclusion helps with the development of a positive self-image and overall oral health.

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