Ectodermal Dysplasia: Clinical Diagnosis

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Abstract

The aim of this article is to review and show the possible cranio-maxillofacial deformities consequences associated with ectodermal dysplasia, which include dental ageneses, and describe the oral clinical aspects.

Twenty three ectodermal dysplasia patients had a clinical examination and underwent radiographic and photographic assessment.

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

Dentists must have experience and be aware about his/her ectodermal dysplasia cases before conduct a treatment way to these patients in order to improve their dental, masticator, growing and orthognathic conditions.

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Introduction

Ectodermal dysplasia (ED) is not a single disorder, it is a large and complex group of disorders defined by the abnormal development of two or more structures derived from the embryonic ectoderm layer. The most frequently reported manifestation of ED is hypohidrotic dysplasia (HED), also termed Christ-Siemens-Touraine syndrome, and anhydrotic dysplasia, that can be due to the HED could be detect easily in clinic. The ectoderm, one of three germ layers present in the developing embryo, gives rise to the central nervous system, peripheral nervous system, sweat glands, hair, nails, and tooth enamel.[1,2,3]

As a result, patients of ED exhibit the following clinical sign: hypotrichosis, hypohidrosis, and cranial abnormalities. The patients often exhibit a smaller than normal face because of frontal bossing, a depressed nasal bridge, the absence of sweat glands results in very smooth, dry skin and/or hyperkeratosis of hands and feet. Oral traits may express themselves as anodontia, hypodontia, and conical teeth. Anodontia also manifests itself by a lack of alveolar ridge development.[2,4,5]

The earliest recorded cases of ED were described in 1792.2 Since then, nearly 200 different pathologic clinical conditions have been recognized and defined as ED. These disorders are considered relatively rare, 1 in 10,000 to 1 in 100,000 births.[2-4,6,7]

The clinical manifestations of ED also cause considerable social problems in the affected patients. Dental treatments of the clinical traits of ED can have a profound impact on these patients. The ability to look and feel like their peers is imperative for the psychological development of these patients. The literature has demonstrated the

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benefits that corrective dentistry has for the self-esteem and social well-being of these patients.[2,8,9]

Our major goals of providing dental and medical clinical images with different aspect of our cases for provide experience to especially dentists and other clinicians.

Cases and Method

This retrospective study was carried out on the patients applying to our university dental clinic from 1997 to 2008. In our faculty, twenty three (aged 3 to 45 years) with a diagnosis of HED were included.

All major sign of ED were studied, such as sparse hair (trichodysplasia), smooth skin (hypohidrosis), abnormal finger- and toenails, skull and face abnormalities and the pedigree of the patients were researched (Fig. 1- .

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. Examination included the skull, face, hair, teeth, nails, skin, lungs, sweat glands, et cetera.

Discussion

ED is genetically transmitted, a rare, multi-system disorder. At our cases, after a discussion of the family and medical history, it was found that the parents of some of the patients were related to each other and they had a similar features of ED in their parents confirming the hereditary nature of ED (Such as; skin, sparse hair and difficulty in sweat).

Clinical diagnosis of ED is difficult because the identification of the precise syndrome could be a challenge[10,11] without collaboration between the patient and the different specialties concerned. Diagnosis of ED, without any other diagnostic precision, would be difficult at best.

Steiner analyses are useful for reveal a facial height reduction and concavity. It can be reval to maxillar reduction, labial retrusion, chin prominence and nasolabial and chin reinforcement. But the researchers should be in attention that these measures may be unreliable because they vary according to tooth ageneses and the severity of ED. The consequence of that dental ageneses could curb bone growth[1,12,13].

ED is primarily characterized by partial or complete absence of certain sweat glands (eccrine glands), causing a lack of or diminished sweating (anhidrosis or hypohidrosis), heat intolerance, and fever.

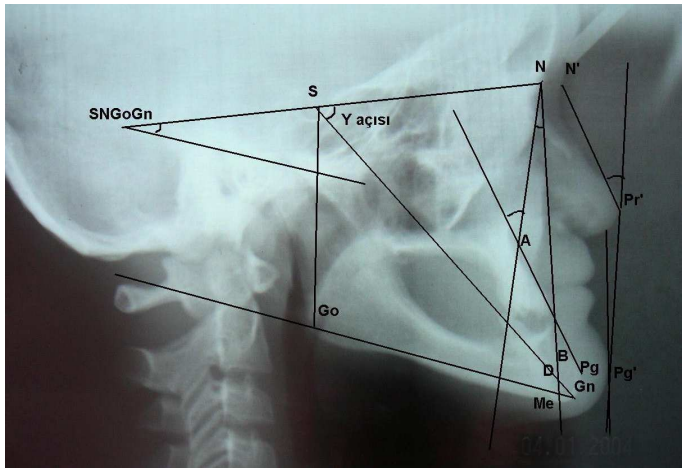


Fig.1 Sample teleradiographs and Steiner's cephalometric analysis

Also ED is characterized by absence and/or malformation of certain teeth (from hypodontia to anodontia and conical shape) (Figs. 2,3,4,5).

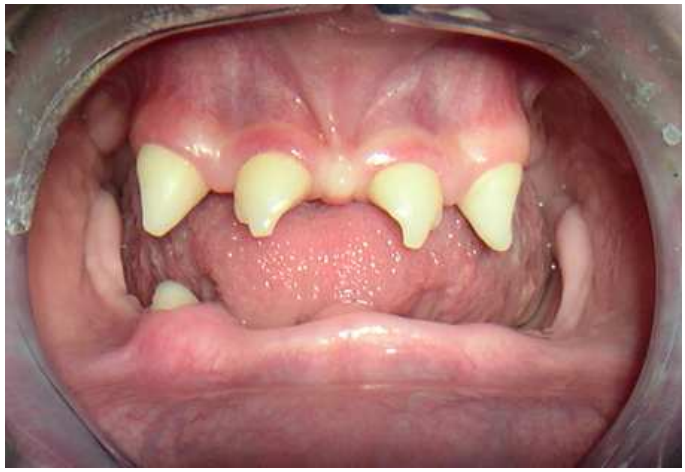


Fig. 2a Absence and/or malformation of certain teeth.

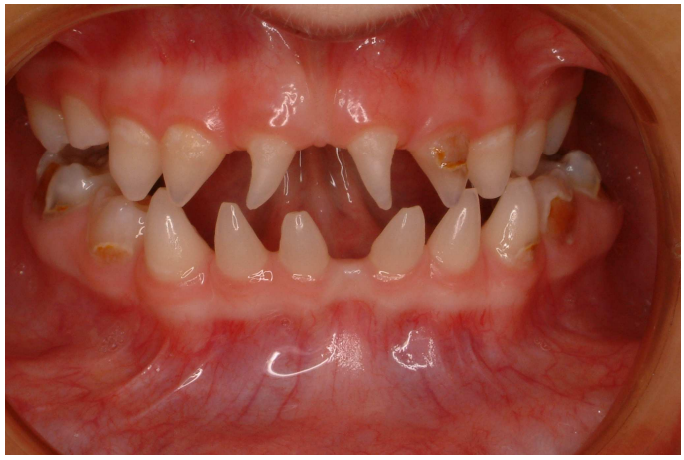


Fig. 2b Absence and/or malformation of certain teeth.

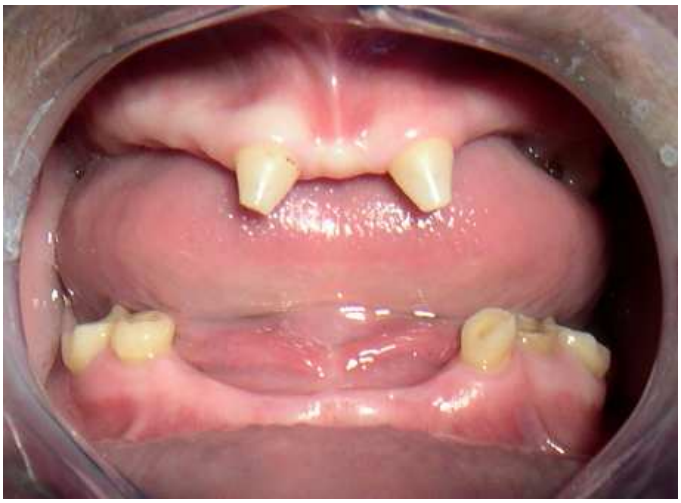


Fig. 2c Absence and/or malformation of certain teeth.

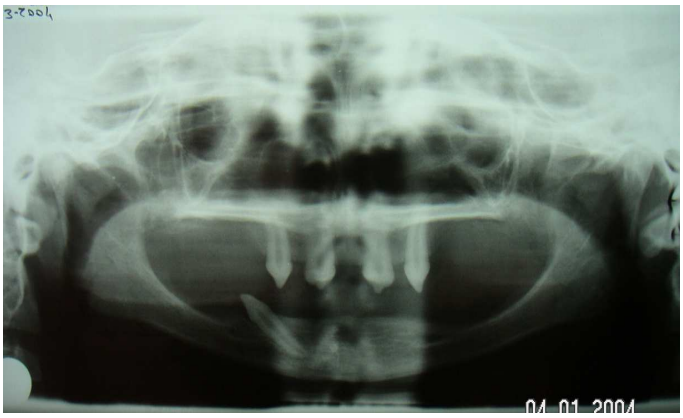


Fig. 3b Panoramic radiography is showing absences and/or malformation of certain teeth.

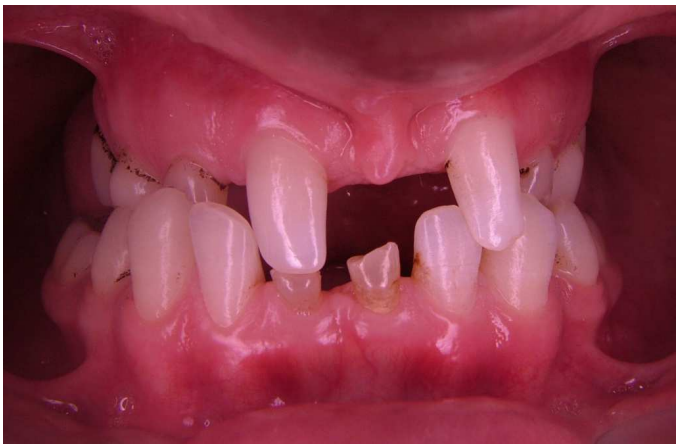
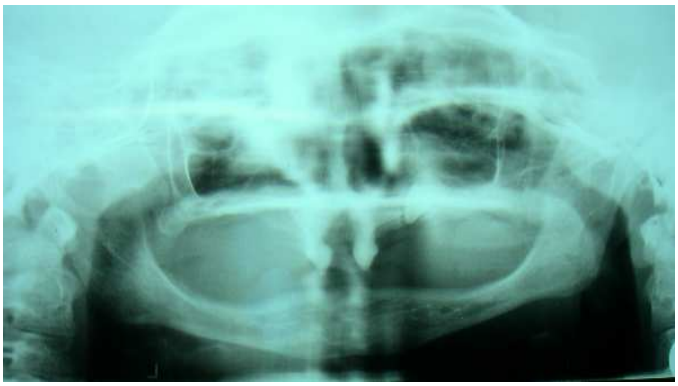


Fig. 2d Absence and/or malformation of certain teeth.



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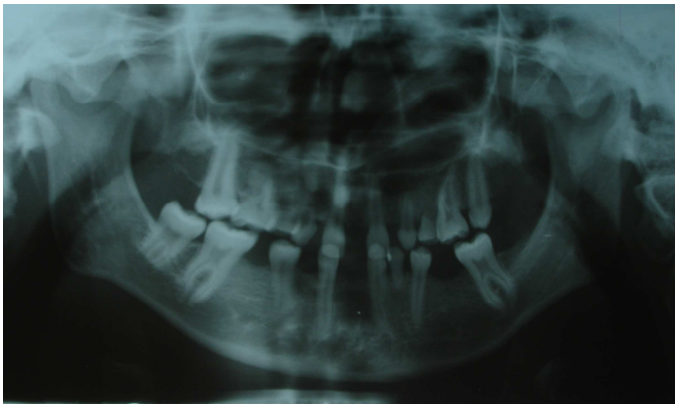

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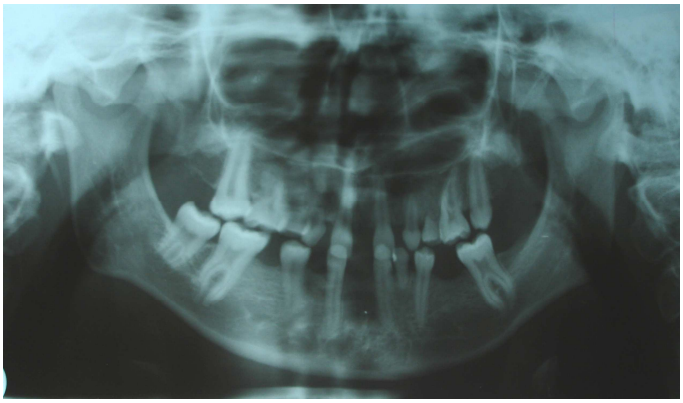

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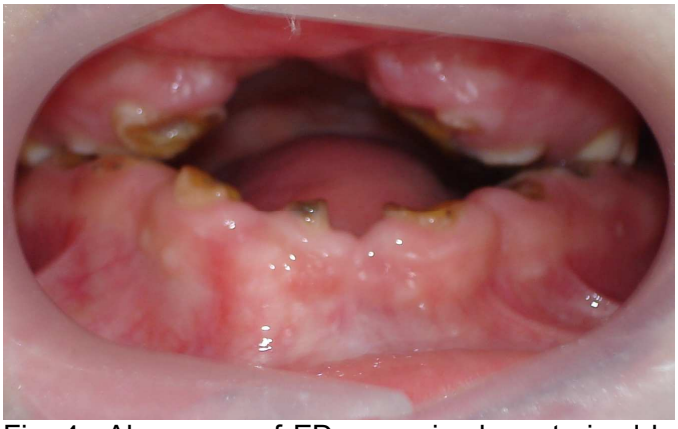


Fig. 4a Also some of ED cases is characterized by cleft lip and palate.

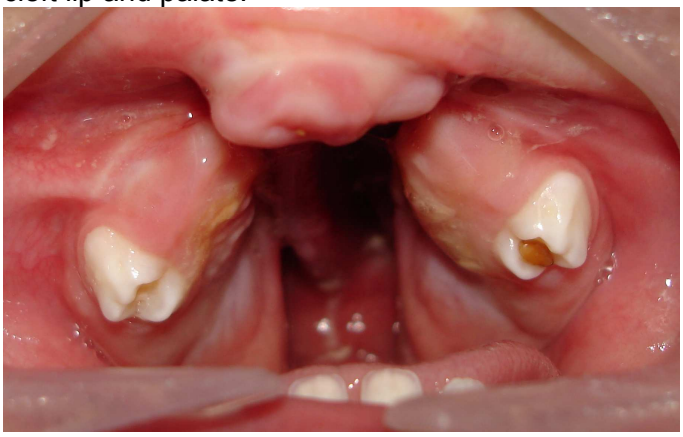


Fig. 4b Also some of ED cases is characterized by cleft lip and palate.

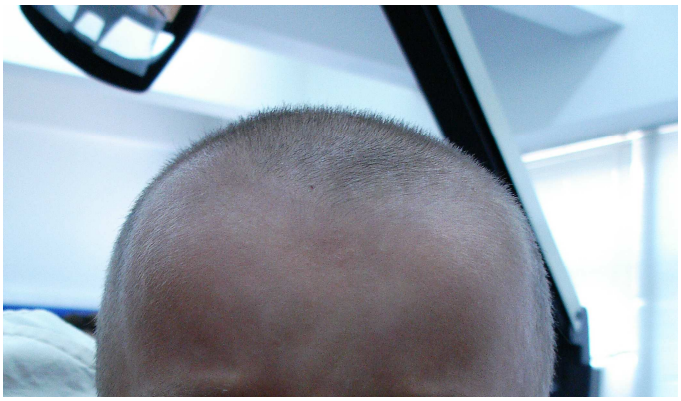


Fig. 5a Also ED is characterized by facial abnormalities, including a prominent forehead.

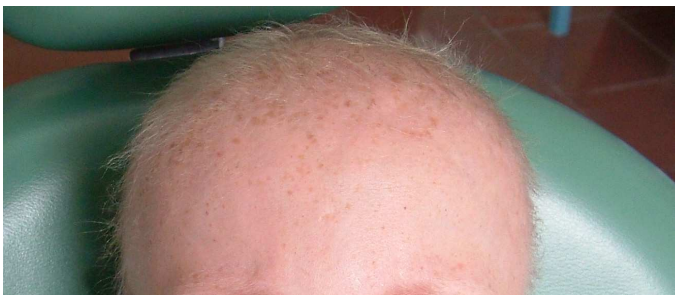


Fig. 5b Also ED is characterized by facial abnormalities, including a prominent forehead.

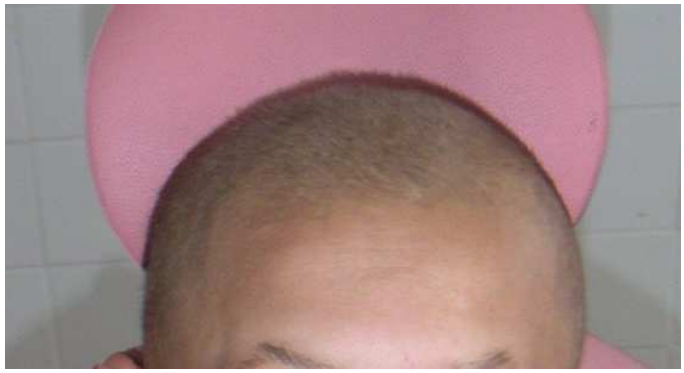


Fig. 5c Also ED is characterized by facial abnormalities, including a prominent forehead.

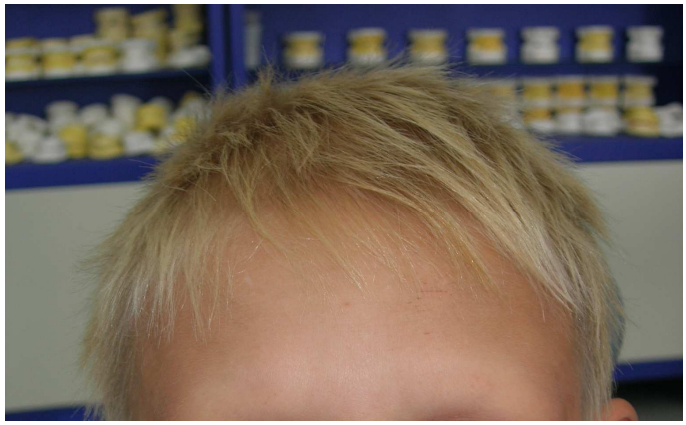


Fig. 5d Also ED is characterized by facial abnormalities, including a prominent forehead.

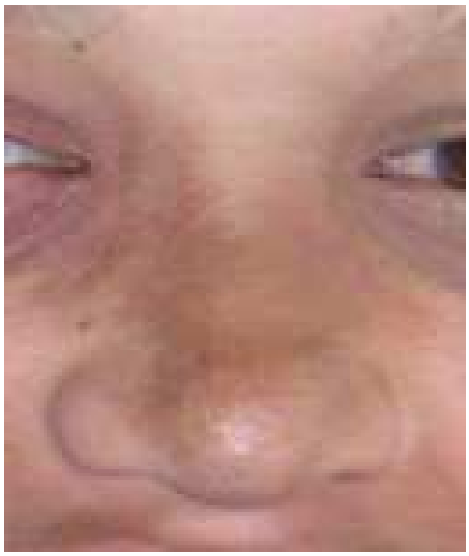


Fig. 6a Also ED is characterized by a sunken nasal bridge (so-called "saddle nose").

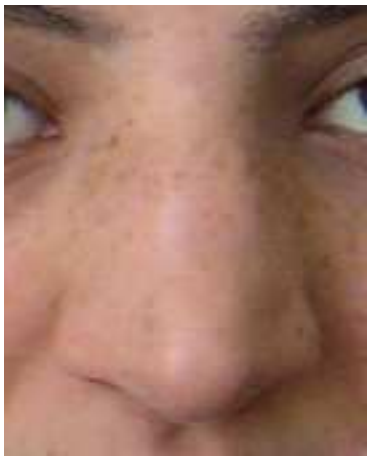


Fig. 6b Also ED is characterized by a sunken nasal bridge (so-called "saddle nose").

Fig. 6c Also ED is characterized by a sunken nasal bridge (so-called "saddle nose").

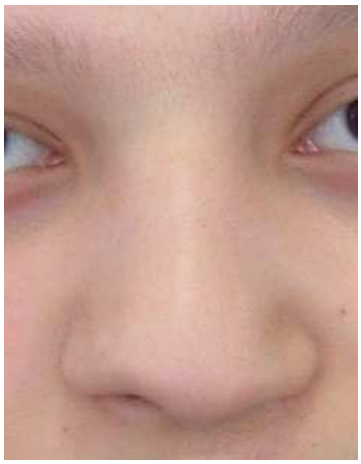


Fig. 6d Also ED is characterized by a sunken nasal bridge (so-called "saddle nose").

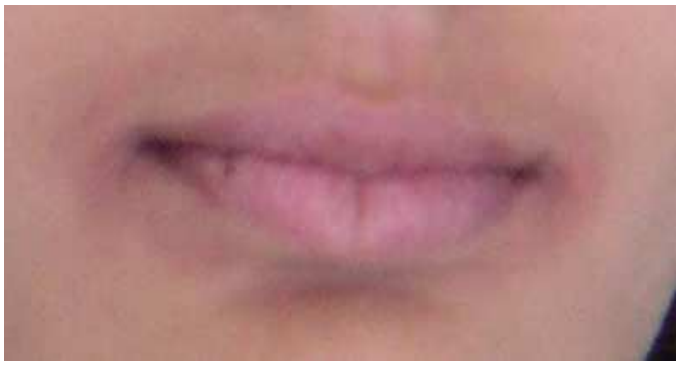


Fig. 7a Also ED is characterized by an unusually thick lips.

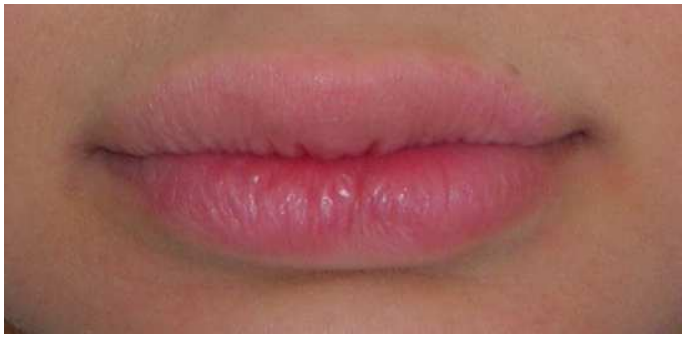


Fig. 7b Also ED is characterized by an unusually thick lip.

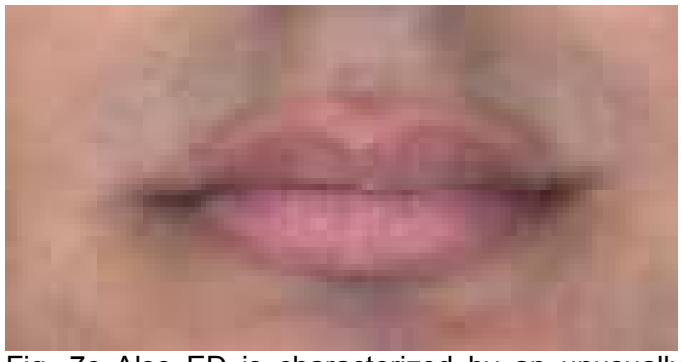


Fig. 7c Also ED is characterized by an unusually thick lip.

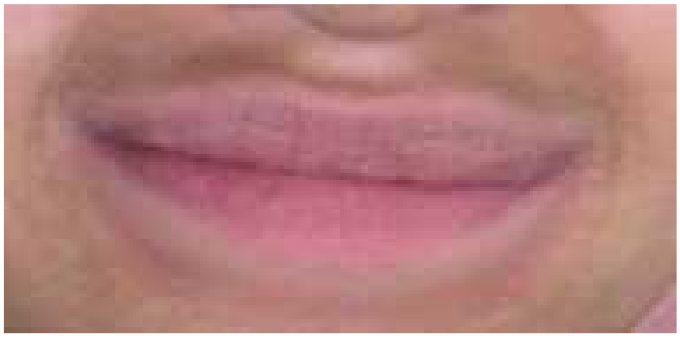


Fig. 7d Also ED is characterized by an unusually thick lip.

Also ED is characterized by skin on most of the body may be abnormally thin, dry, and soft with an abnormal lack of pigmentation (hypopigmentation), however, the skin around the eyes (periorbital) can be darkly pigmented (hyperpigmentation) (Fig. 8a-8d).

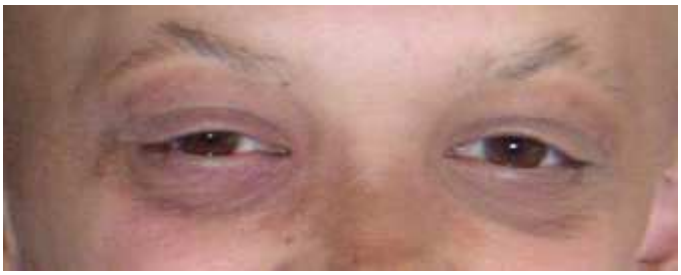


Fig. 8a Skin around the eyes (periorbital) have been darkly pigmented (hyperpigmentation).

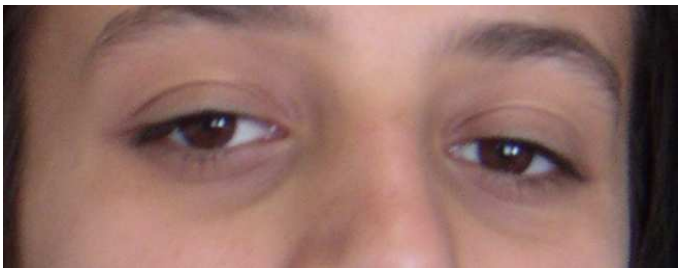
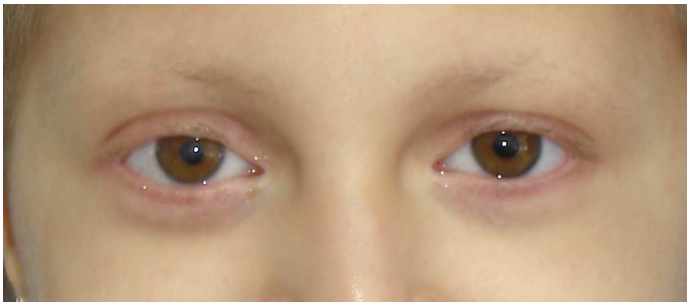


Fig. 8b Skin around the eyes (periorbital) have been darkly pigmented (hyperpigmentation).



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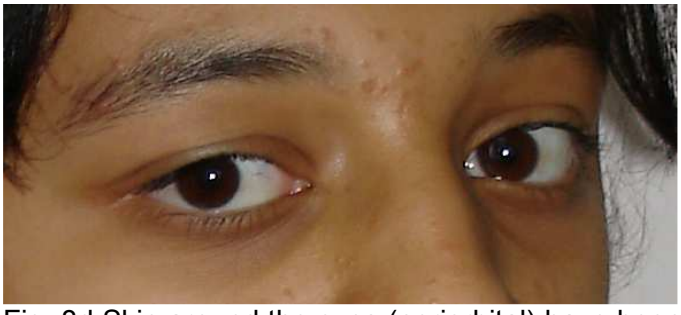

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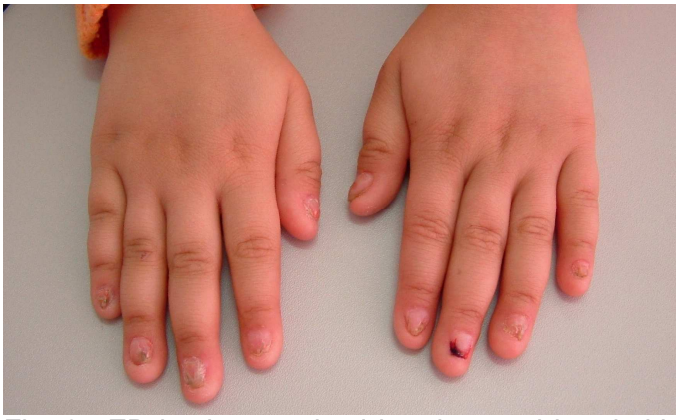


Fig. 9a ED is characterized by abnormal hand skin and nails.

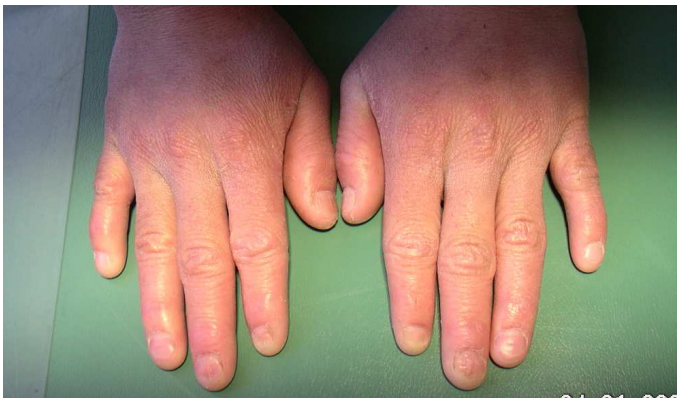


Fig. 9b ED is characterized by abnormal hand skin and nails.

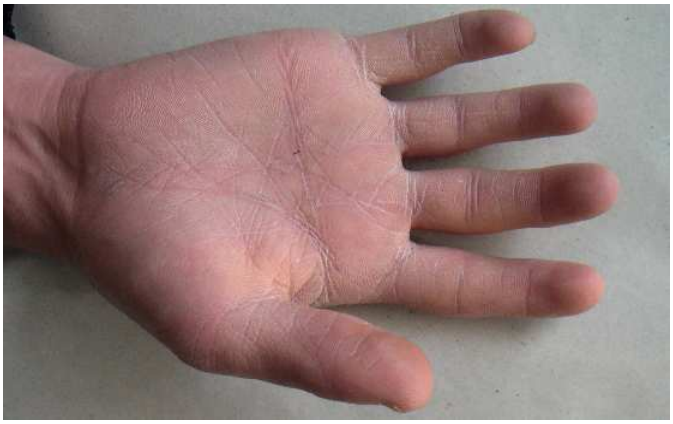
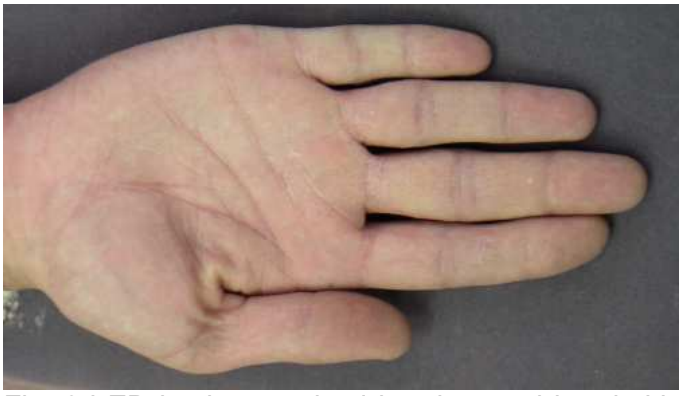


Fig. 9c ED is characterized by abnormal hand skin and nails.



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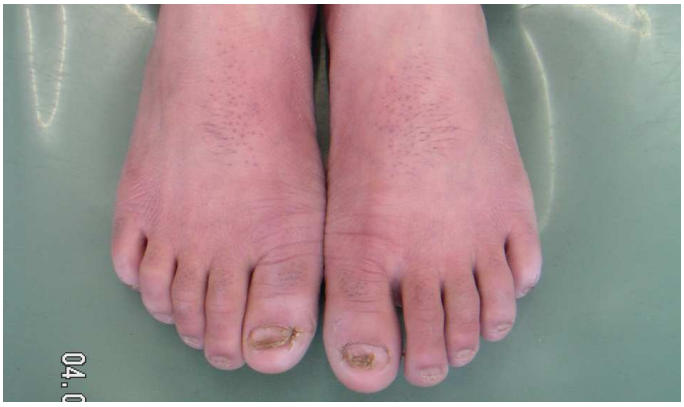
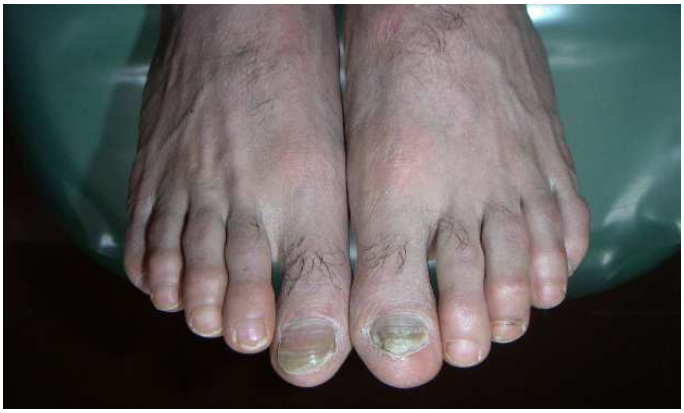


Fig. 10a Also ED is characterized by abnormal foot skin and nails.



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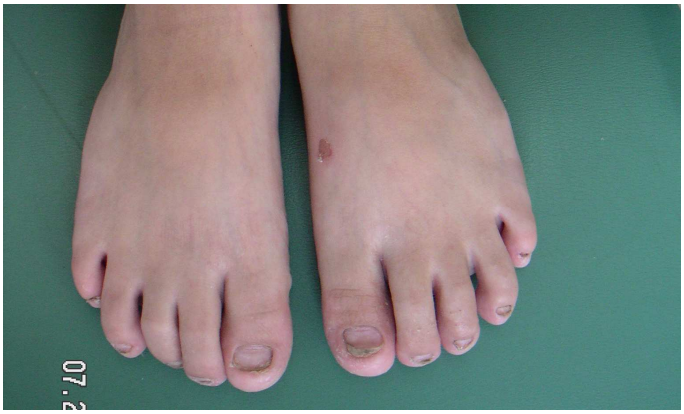

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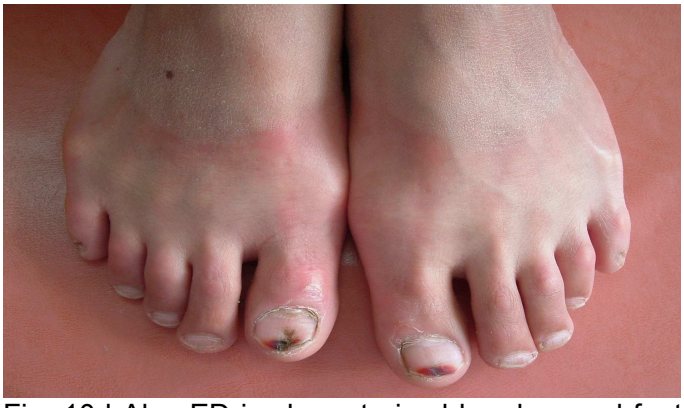

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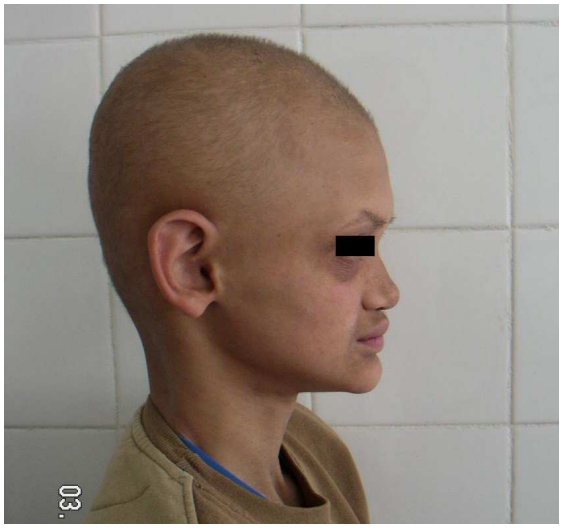
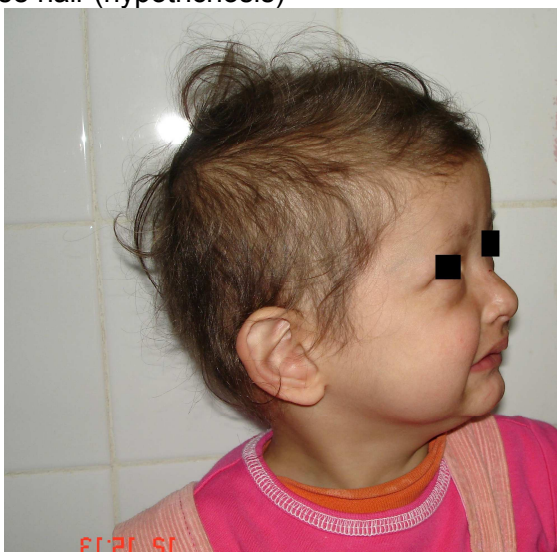


Fig. 11a Also ED is characterized by abnormally sparse hair (hypotrichosis)



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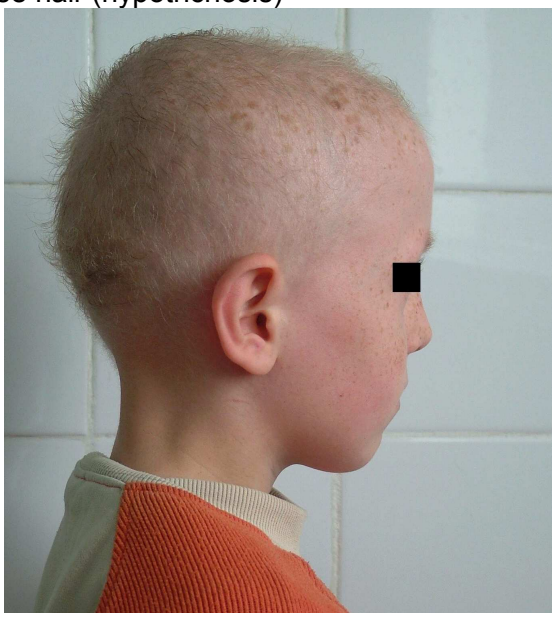

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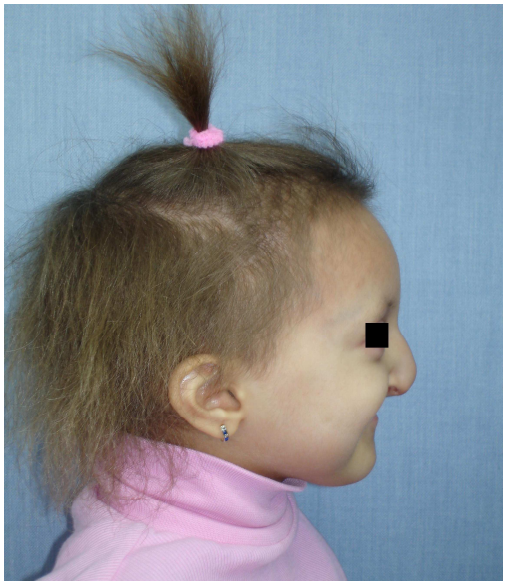

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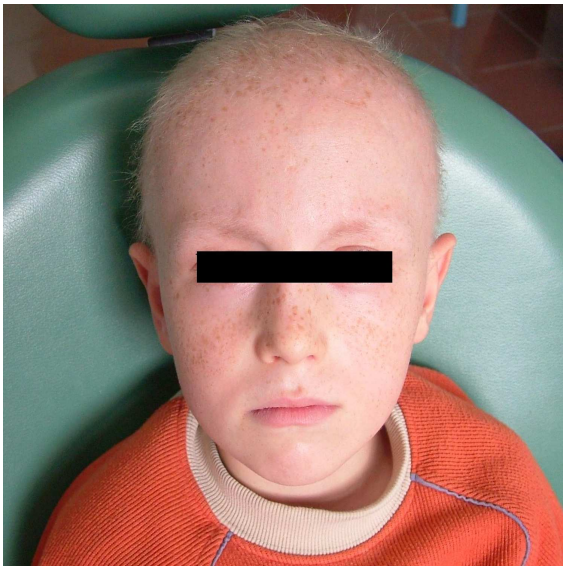
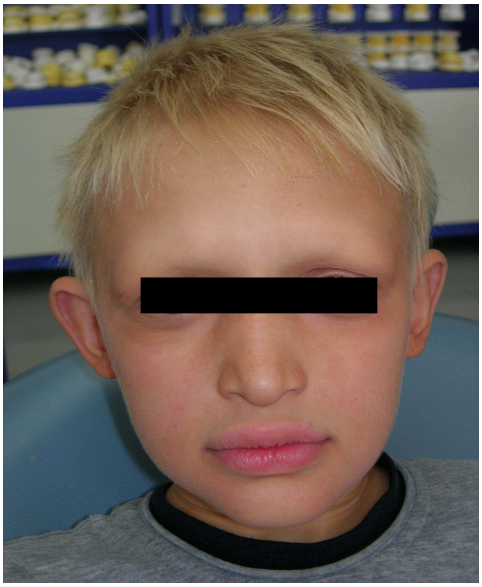


Fig. 12c Clinical appearance of ED.



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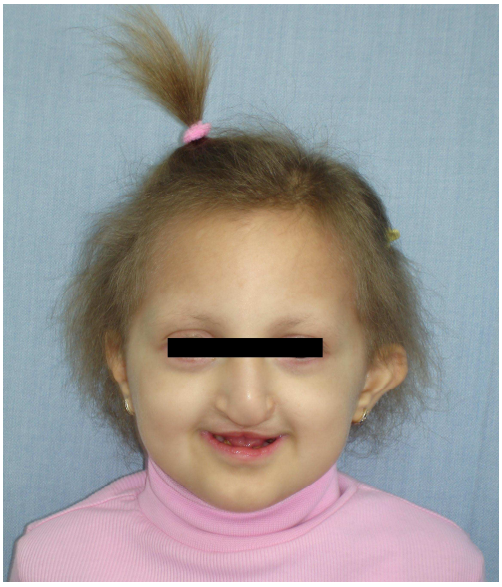

Fig. 12d Clinical appearance of ED.

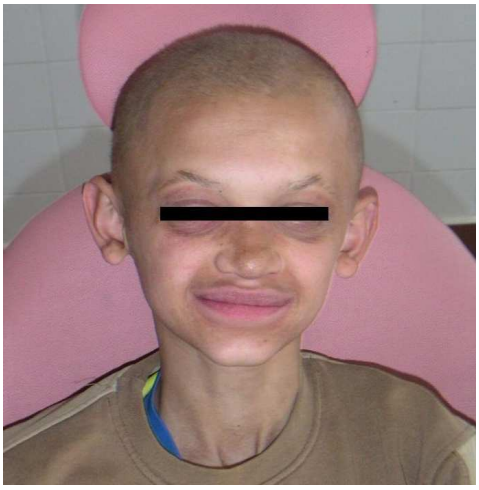

Fig. 12b Clinical appearance of ED.

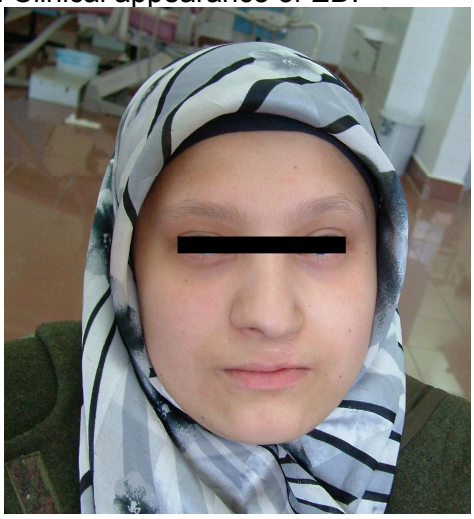

Fig. 12e Clinical appearance of ED.

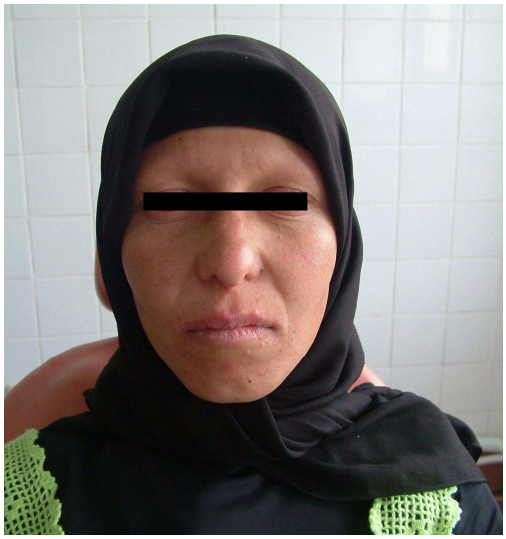

Fig. 12f Clinical appearance of ED.

All of these giving to the ED cases as appearing prematurely aged. These agrees with past research.[1-3,5,9-11,14-21]

In many cases, affected infants and children may also exhibit underdevelopment (hypoplasia) or absence (aplasia) of mucous glands within the respiratory tracts and, in some cases, decreased lung capacity and function, potentially causing an increased susceptibility to certain infections and/or allergic conditions1. Many affected patients experience recurrent attacks of wheezing and breathlessness (asthma), and respiratory infections.

ED is usually inherited as an X-linked recessive genetic trait; in such cases, the disorder is fully expressed in males only. However, females who carry a single copy of the disease gene (heterozygote carriers) may exhibit some of the signs and findings associated with the disorder. Also ED could appear to be inherited as an autosomal recessive genetic trait. In such cases, the disorder is fully expressed in both males and females[1,16,17,21,22].

Despite the great number of ED cases described so far, fewer than 30 have been explained at the molecular level with identification of the causative gene[1-4,14].

At this point, scientists should have a molecular and biochemical background, with a scientific multidisciplinary approach and equipment. These conditions are what makes a clinical diagnosis of ED difficult.

A multidisciplinary approach is required in modern dentistry for diagnosis and treatment. This study has enabled us to demonstrate a relationship between all major symptoms of ED, including hypodontia, thin hair (hypothricosis) and smooth skin (hypohidrosis).

Conlusions

When confronted with multiple dental ageneses, the clinician should look for an association of ED signs, because of ED could also be detected.

The principal aim of this study is to get experiences to determine patients who affected by ED.

Finally, we believe that all clinical research will supply to improve the knowledge, experiences to determine ED patients.

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