

Case series of ectodermal dysplasia and evaluation of oral findings: A literature review

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Abstract

Ectodermal dysplasia (ED) is a hereditary disease characterized by anomalies in the structures of ectodermal origin. This report aims to determine the oral features of patients with ED syndrome and to present treatment approaches. Because of nutrition, speech and aesthetical problems due to the lack of teeth 22 patients with ED syndrome referred to faculty dentistry clinic. Facial physiognomy was typical for ED and the facial height was decreased due to the vertical dimension and the delayed exfoliation of primary teeth. Also anodontia, hypodontia, delayed eruption, diffuse enamel hypoplasia, conical tooth structure, talon cusps, microdontia, and transposition in teeth were seen in patients. In most cases, prosthesis were applied to improve the facial aesthetics, speech and oral function due to the common hypodontia. The primary goals of the dental treatment of patients with ED are enhancing aesthetics and improving masticatory function. The oral rehabilitation of the patients' needs multidisciplinary dental treatment.

Keywords: Ectodermal dysplasia; tooth loss; pediatric dentistry; dental prosthesis

INTRODUCTION

Ectodermal Dysplasia (ED) is an inherited syndrome in which ectoderm structure is affected by anomalies including skin, nails, sebaceous and eccrine glands, hair and teeth (1). It has a triad of hypotrichosis, anodontia and anhidrosis, and is a condition seen every 100,000 births worldwide (2,3).

It was first described by Darwin in 1860 (4). There are 192 subtypes and its etiology is genetic. The syndrome can be seen only in males, but in female cases a heterozygous with a normal X chromosome and a diseased chromosome is present and often shows symptoms of the disease (2,3).

Hair, eyebrows and eyelashes are thin, short and less than normal; skin is soft, flat and dry in these patients. The forehead and lips are protruding and the nose is submerged (saddle-shaped nose). Thin line wrinkles around the eyes and mouth, and also decreased vertical face dimension give patient a typical senile appearance. Thickening, discoloration, dark pigmentation and deformities can be seen on the nails. These are the main features of this rare hereditary disease (5-9).

As intra oral findings of the disease, the jaw development of the patients is not affected, but the alveolar crest is thin due to lack of teeth. Failure in the development of the alveolar crest leads to a decrease in the vertical dimension and thereby it causes a bulging appearance on the lips. The palatal arch is generally deep and the cleft palate can be seen. Hypodontia, anodontia or deformities in teeth are recognizable features. Dry mouth is not found in all patients, because the complete absence of salivary glands is a rare condition (6,7,9,10).

In addition to oral clinical examination, patient's medical history and panoramic radiographs; genetic analysis are used in the accurate diagnosis of the syndrome (9).

Approximately 200 different types of ectodermal dysplasia have been reported and there are different classifications for it (11-14). Deficiencies in nails, hair, teeth and sweat glands can be occurred individually or in combination. Ectodermal dysplasia is named according to these symptoms and combinations in affected individuals (15).

Some types of ED are hypohidrotic dysplasia (HED) (Christ-Siemens-Touraine syndrome, anhidrotic dysplasia) (16),

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Ectrodactyly-Ectodermal Dysplasia-Cleft Lip/Palate (EEC Syndrome) (17), Oculodentodigital Syndrome (ODD Syndrome) (18), Tricho-dento-osseous Syndrome (TDO Syndrome) (19), Ellis-Van Creveld Syndrome and Witkop Syndrome (4,20).

HED is the most frequently seen type of ED. It has three types named X-Linked Hypohidrotic Ectodermal Dysplasia (XLHED), Autosomal Recessive Hypohidrotic Ectodermal Dysplasia (ARHED) and Autosomal Dominant Hypohidrotic Ectodermal Dysplasia (ADHED) (16).

Ectrodactyly-Ectodermal Dysplasia-Cleft Lip/Palate (Ectrodactyly-ectodermal dysplasia-clefting (EEC) syndrome) is characterized by ectrodactyly (congenital absence of fingers and toes), cleft lip and palate, hypodontia, sparse hair, dystrophic nails and abnormalities in eyes and ears (17).

Oculodentodigital Syndrome (ODD Syndrome) is a disease that seen characteristic facial appearance and miscellaneous eye, tooth, finger, skin, nose and ear abnormalities and neurological disorders such as microcephaly, enamel hypoplasia, taurodontism, hypodontia, sparse hair, thick and dry skin, syndactyly, cataract, glaucoma and deafness (18).

Tricho-dento-osseous Syndrome (TDO Syndrome) is characterized by hair, nail, teeth and skeletal anomalies like curly hair, brittle nails, taurodontism, amelogenesis imperfecta, increased bone density, abnormal shape anomalies (19).

Ellis-Van Creveld Syndrome (Chondroectodermal dysplasia; Mesoectodermal dysplasia) is characterized by skeletal dysplasia (short arms and legs, polydactyly, clinodactyly), congenital heart defects, oral and dental abnormalities (taurodontism, supernumerary teeth, accessory labiogingival frenula, conical shaped teeth and malocclusion) (20).

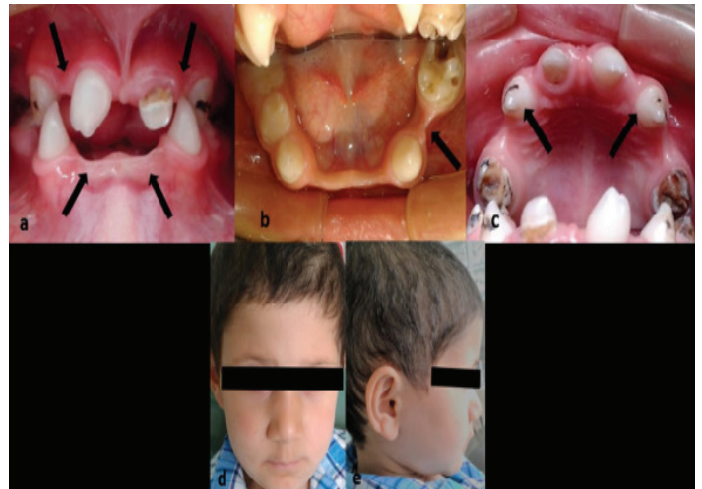
Witkop Syndrome (Tooth/Nail Syndrome, Nail Dysplasias with Hypodontia) is characterized by nail and tooth abnormalities (lack of germ in permanent teeth, conical shaped tooth, delayed eruption, hypodontia, brittle and fragile nails). Hair and facial features are commonly normal (4).

The aim of this report is to describe clinical and radiological findings of ectodermal dysplasia patients and to present treatment modalities.

CASE REPORTS

The ethical approval was obtained before the study. Patients applied to the faculty dentistry clinic with ED were included in this study. In accordance with the Declaration of Helsinki, patients and their families were informed and consent form signed by parents for under the age of 18. Intra oral and extra oral photographs of patients were taken before and after the treatment. A total of 22 patients (13 males and 9 females) were examined. The age range of patients was between 3 and 21. The mean age of the patients was 10.54 ± 4.59 . Medical history, familial pedigree

were recorded and clinical and radiographic examinations were obtained.



Figur 1: 3-year old boy patient with ED, intra oral and extra oral photographs

a. Lack of germs (maxillary primary lateral teeth, mandibular incisor teeth), hypodontia, diastema, dentin caries in left maxillary primary central and primary canine teeth and deep dentin caries in maxillary primary molars are seen.

b. Lack of germ (left mandibular first primary molar)

c. Conical tooth structure, lack of germ (maxillary first primary molars) and deep dentin caries in maxillary second primary molar teeth are seen.

d. Patient's front side photograph. Sparse eyebrows and hair are seen.

e. Patient's lateral side (profile) photograph. Sparse hair is seen.

Facial physiognomy was typical for ED and the facial height decreased due to the vertical dimension were seen in all patients. The delayed eruption finding was in found in 8 patients. The delayed exfoliation for primary teeth was observed in all cases. Diffuse enamel hypoplasia was seen in 3 patients' permanent teeth. Deep dentine caries was observed in only 4 cases (Figur 1 and Figur 2) and generally low incidence of caries was found. Conical tooth structure in 8 teeth of 4 cases (Figur 1 and Figur 2), talon cusps in 3 teeth of 2 cases, microdontia in 8 teeth of 6 cases (Figur 2), transposition in 12 teeth of 9 cases (especially canine teeth) were found. While hypodontia was observed commonly in lower incisors (31-32-41-42) was observed commonly (85%) (Figur 3), at least it was seen in first primary molars. In 22 cases, a total of 332 teeth were found to be missing which 149 of them were in maxilla, 183 of them were in mandible. In a girl patient, ED was seen in her sister, too. In most cases, child prostheses were applied to improve the facial aesthetics, speech and oral function due to the common hypodontia (Figur 3 and Figur 4). Lack of germ in the permanent teeth according to maxilla and mandible are seen in Figur 5 and Figur 6. Anodontia in lower jaw can be observed in Figur 7.

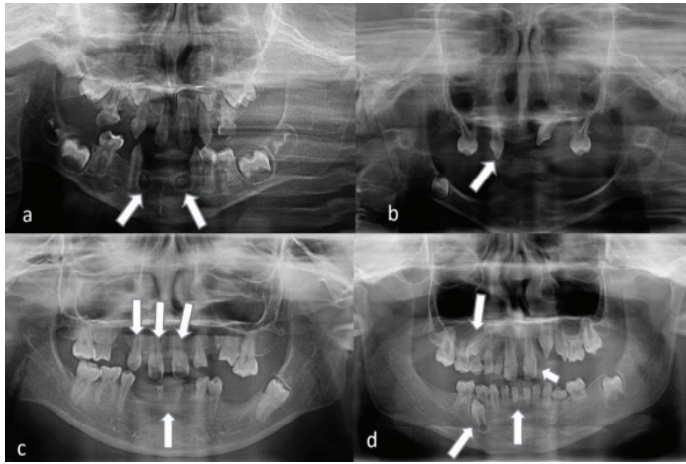


Figure 2. Panoramic radiographs of ED patients

- a. Diastema, lack of germs, micro germs and hypodontia
- b. Lack of germs, conical shaped teeth and oligodontia
- c. Diastema, lack of germs, microdontia and hypodontia
- d. Microdontia, diastema and transposition

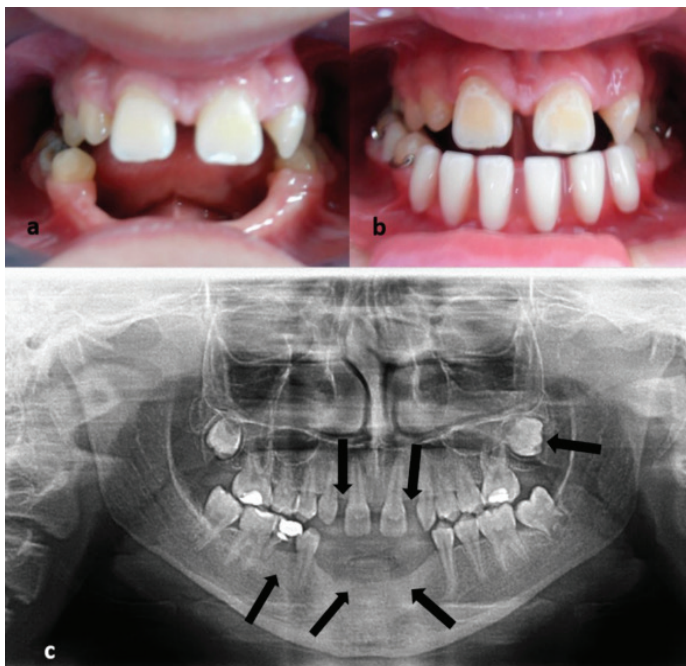


Figure 3. 14-year old girl patient's intra oral photographs and panoramic radiograph

- a. Patient's intra oral photograph before the treatment
- b. Patient's intra oral photograph after the treatment and applied removable space maintainer with tooth
- c. Patient's panoramic radiograph before the treatment. Lack of germs (maxillary permanent lateral incisors, mandibular incisors and right mandibular second premolar tooth), hypodontia, diastema and horizontal eruption direction of left second permanent molar tooth are seen. As a treatment, right mandibular second primary molar tooth was extracted due to mesial root resorption



Figure 4. Pre-treatment and post-treatment extra oral photograph of a patient with ED.

a. Pre-treatment extra oral photograph of the patient (vertical size loss, concave and elderly facial appearance, thick and outward-looking lips, saddle nose, lack of glands, dark pigmentation around the eyes, sparse hair, skin dryness, scaly skin-eczema, lack of tooth)

b. Post-treatment extra oral photograph of a patient

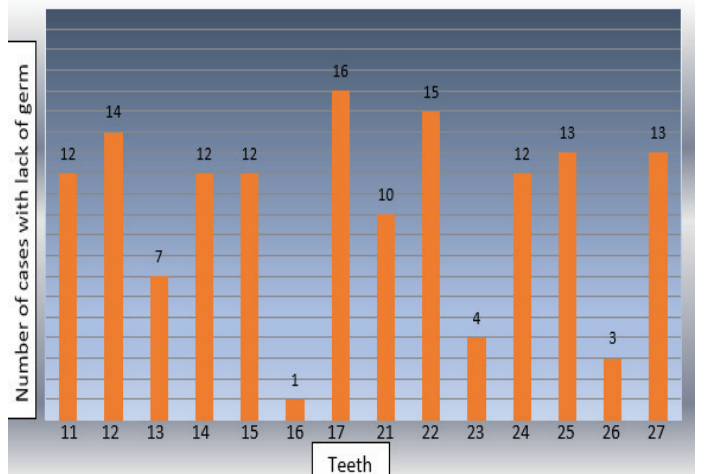


Figure 5. Lack of germ in the permanent teeth according to maxilla

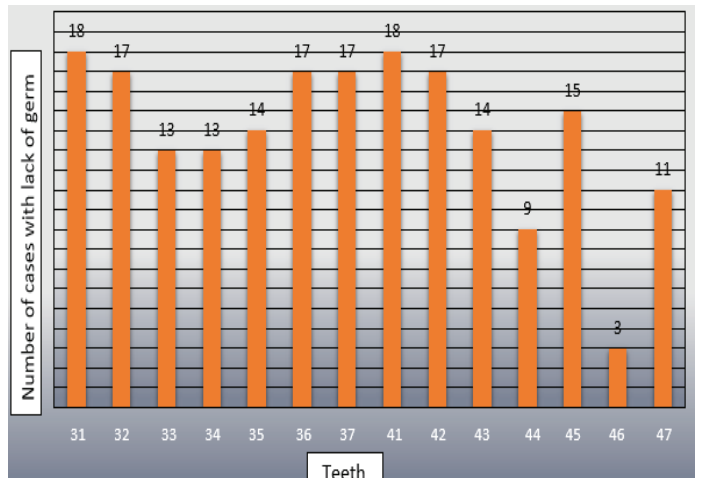
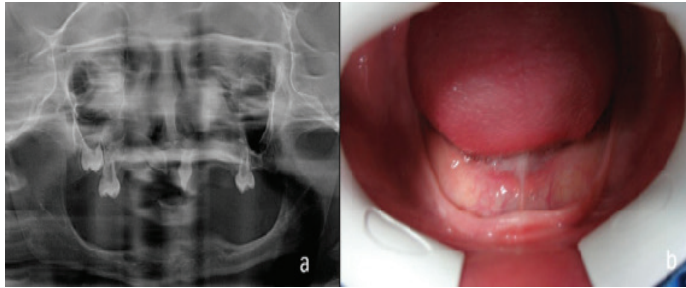


Figure 6. Lack of germ in the permanent teeth according to mandible



Figur 7. Panoramic radiograph and intra oral photograph of a patient with ED.

- a. Panoramic radiograph of the patient.
b. Intra oral photograph: Anodontia in patient's lower jaw.

DISCUSSION

In most ED patients, in addition to tooth deficiencies; structure and deformities in existing teeth are observed (10). Beside these, there are quantity anomalies. In some cases, anodontia was observed (11). In this study, the presence of many missing teeth and conical shaped teeth are also compatible with previous case reports about ED (21-24). Hypodontia and oligodontia were observed in 15 (68%) patients. Lack of germs were seen in mandible more than in maxilla compatible with the studies of Yavuz et al., Tuna et al. and Lexner et al (16,25,26).

In present study, as morphological anomalies (totally 63%), microdontia was observed in 6 (27%) patients, conical shaped teeth were observed in 6 (27%) patients and talon cuspid was observed in 2 (9%) patients. In the study of Doğan et al., conical shaped teeth were indicated in 75.5% of the patients (27). In the study of Tuna et al., crown tooth malformations were mentioned in 12 (75%) patients and peg shaped and conical shaped teeth were seen in 11 primary and 10 permanent teeth (25).

In this study, most commonly missing permanent teeth were in lower incisors (31-32-41-42) compatible with the study that indicated basically affected mandibular incisors (9), while in another study it was found permanent lateral incisors and secondary premolars (12-22-35-45) (25).

In all our patients, decreased vertical dimension of facial height was observed. In several studies, it was mentioned that reduced vertical size due to anodontia or oligodontia, disrupting aesthetics and giving an old appearance cause disruption of chewing functions as well as psychological problems in children and causes physical development disorders along with feeding problems (5,27,28). Our findings are consistent with these previous studies.

In cases of dental deficiency, oral rehabilitation may be achieved by overdenture, full or partial removable prosthesis or fixed prosthesis (29-31). The most recommended treatment modality is removable prosthesis. Implant treatments can also be planned (29,32), but this may be difficult in cases where the age of the child, the bone development is not complete and in the knife edge

bone type, additional treatment is required such as bone graft or sinus lifting (9,33,34). Temporary implants in pediatric patients may be applied as periodic treatment (35). Dentists and especially pediatric dentists usually take part in the early diagnosis of ED patients and provide the necessary guidance. Patients should be monitored periodically according to their growth and development stages and their prostheses should be routinely modified and controlled to match their growth (9). In our cases, implants weren't applied to pediatric patients; mostly removable appliances (space maintainers or acrylic total prostheses for children) were applied to them.

In some ED patients, because of the ectoderm origin of the salivary gland, dry mouth is observed (36,37). In our 3 patients, high caries (DMF/dmf) values were found caused by dry mouth. In another study, 75% of patients were complain of ophthalmologic problems. These were hyperpigmentation (88.9%), eyelid problems (88.9%), and dry eyes (61.1%) (38). In this study, we evaluated the oral findings of ED patients, but addition to oral problems, ophthalmologic problems can effect the quality of life negatively.

Oral agenesis and dental malformations are frequently observed in patients with ectodermal dysplasia. These anomalies can affect both primary and permanent teeth (8,29). The delayed exfoliation of primary teeth and delayed eruption of permanent teeth were observed in our cases coincide with other studies (9,39).

These dental and general findings of ED may psychologically adversely affect the patients. Therefore, early diagnosis and dental treatment have a great importance for improving the quality of life (24).

CONCLUSION

The primary goals of dental treatment of patients with ED are enhancing aesthetics and improving masticatory and phonetic function. The oral rehabilitation of the ED patients' needs a multidisciplinary dental treatment. Prosthetic treatment is a great value to these patients from the functional standpoint as well as for psychological and psychosocial reasons. In these cases, dentists, pediatric dentists, orthodontists, prosthodontists should work with coordination and multidisciplinary approaches should provide true guidance for patients.

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