EKTODERMAL DİSPLAZİNİN PROTETİK REHABİLİTASYONU: OLGU SUNUMU

PROSTHODONTIC REHABILITATION OF ECTODERMAL DYSPLASIA: A CASE REPORT

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Özet

Bu vakanın amacı estetik, fonksiyonel ve fonetik yararlar, sosyal hayatında daha iyi bir dış görünüş kazandırılan hipohidroitk ektodermal displazili 20 yaşındaki kadın hastanın protetik tedavisini sunmaktır.

20 yaşındaki kadın hasta eksik dişler şikayeti ile sevk edildi. Hastanın konuşma ve çiğneme güçlüğü mevcuttu. Tedavi olarak, estetik, fonksiyonel ve fonetik yararları kazandırmak için maksillada full mouth restorasyon, mandibulada hareketli bölümlü protez seçildi. Böylelikle, hasta daha iyi bir görünüş ve psikolojiye sahip oldu.

Ektodermal displazi (ED) iki veya daha fazla ektodermal doku gelişimindeki defekt ile karakterize nadir, konjenital bir hastalıktır. Bu hastalık hipodonti veya anadonti gibi multipl diş anomalileri, gömülü dişler, çivi şeklinde ya da konik anterior dişler ve normal alveolar sırt gelişimindeki yetersizlikten ötürü dişhekimleri için önemlidir. Erken teşhis eksik dişlerden doğan negatif psikolojik durumu düzeltmek için önemlidir.

Anahtar Kelimeler: Ektodermal displazi, protez, hipodonti.

Abstract

The aim of this case is to report prosthodontic treatment of 20 year-old female patient with hypohidrotic ED who is provided esthetic, functional and phonetic benefits, better outlook in her social life.

A 20 year-old female patient was referred with a complaint of missing teeth. There was difficulty in speaking and mastication. As treatment, full mouth restoration in maxilla, removable partial denture in mandible was selected to provide esthetics, functional, phonetic benefits. Thus, the patient had better appearance and psychology.

Ectodermal dysplasia (ED) is a rare, congenital disease that is characterized by defects in the development of two or more ectodermal tissues. This disease is important for the dentist because of multiple tooth abnormalities such as hypodontia or anodontia, impacted teeth, peg-shaped or conical anterior teeth and deficiency of normal alveolar ridge development. Early diagnosis is very important to correct negative psychological situation which results from missing teeth.

Key words: Ectodermal dysplasia, denture, hypodontia.

Introduction

Ectodermal dysplasia (ED) is a heterogeneous group of disorders which are chacterized by developmental dystrophies of more than one ectodermal tissues (1). Prevalence of ED is approximately 1 in 100.000 live birth. More than 192 different varieties of this condition have been defined (1). But The ED has two main categories: Hidrotic (Clouston's Syndrome) and Hypohidrotic (Christ-Siemens-Touraine Syndrome) forms. Sweat gland manifestations are the difference between the two types. Hydrotic ectodermal dysplasia has normal sweat glands which are absent in Hypohidrotic type (2). Hypohidrotic type that is also called as anhydrotic type, is seen more commonly than hydrotic type and has often an X-linked inheritance (3). X linked recessive disorders are inherited through female carriers. The incidence of this carrier is 17.3 in 100.000 women.Males have more severe symptoms while females have only minor defects (3).

Orofacial symptoms of this syndrome involve anadontia or hypodontia, hypoplastic conical teeth, underdevelopment of the alveolar ridges, frontal bossing, a depressed nasal bridge, protuberant lips and hypotrichosis (4).

While the treatment is planning, some factors such as age of patient, stage of growth in conjunction with the missing teeth, malformed dentition, soft tissue defects and

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psychological condition must be considered (5). ED has multipl treatment choices but in young patients removable prosthodontics are most common treatment (6).

Our major purpose of dental and medical treatment was to provide comfort to patient in her social life. The aim of this case is to report a case of 20 year-old female patient with hypohidrotic ED who is treated with fixed and partial denture.

Case Report

A 20 year-old female patient referred to Oral and Maxillofacial Radiology Department with complaints of multiple missing teeth in maxilla and mandible. She had difficulty in speech and mastication. She reported about existence of sweating and didn't give any information about family history. She had no mental retardation and her vital signs were normal.

On extraoral examination nail thickening (Figure 1), dry skin, absent eyebrows, trichodysplasia, loss of eyelashes, depressed nasal bridge, frontal bossing and hypertelorism were determined. There was hyperpigmentation on the nose and around the eyes (Figure 2).

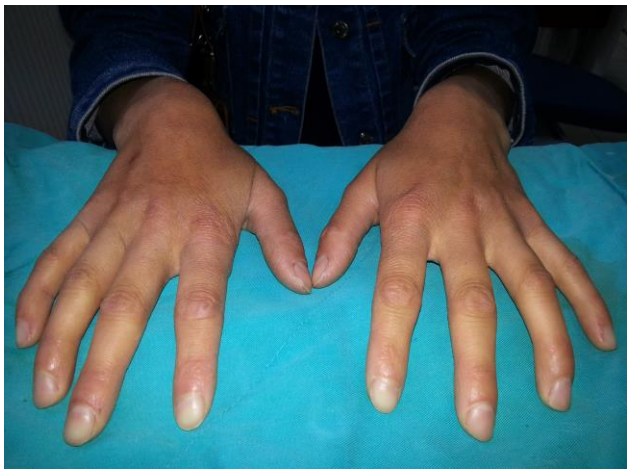

Figure 1. View of nails thickening

Intraoral examination revealed conical anterior teeth, resorption of alveolar ridge and hypodontia was seen. Permanent upper right central incisor was present in our case and it is a rare finding. Vertical dimensions were reduced depends on lack of occlusion (Figure 3-4).

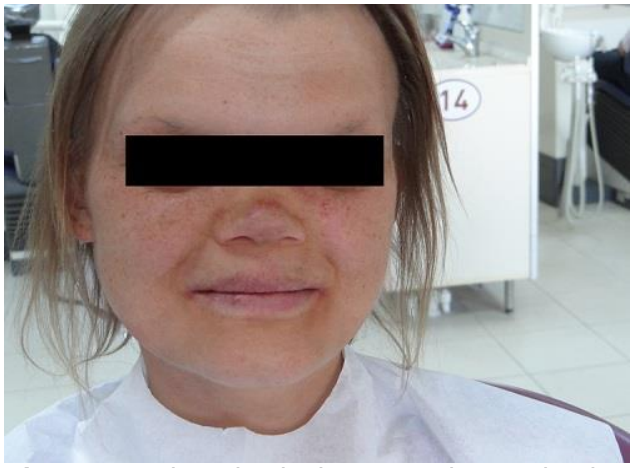

Figure 2. The physical extraoral examination showing dry skin, absent eyebrows, trichodysplasia, loss of eyelashes, depressed nasal bridge, frontal bossing and hypertelorism, hyperpigmentation on the nose and around the eyes.

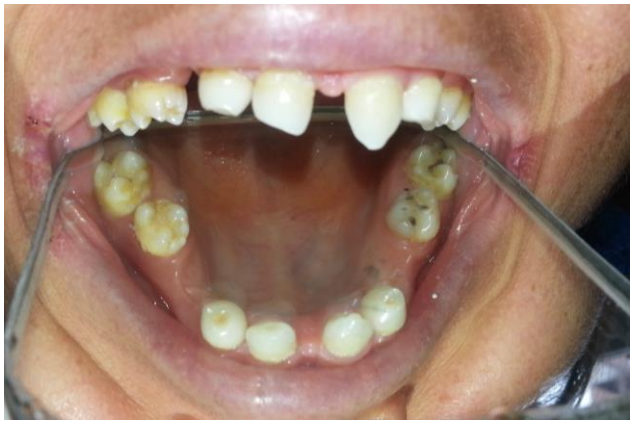

Figure 3. Intraoral view showing conical anterior teeth, resorption of alveolar ridge and hypodontia.

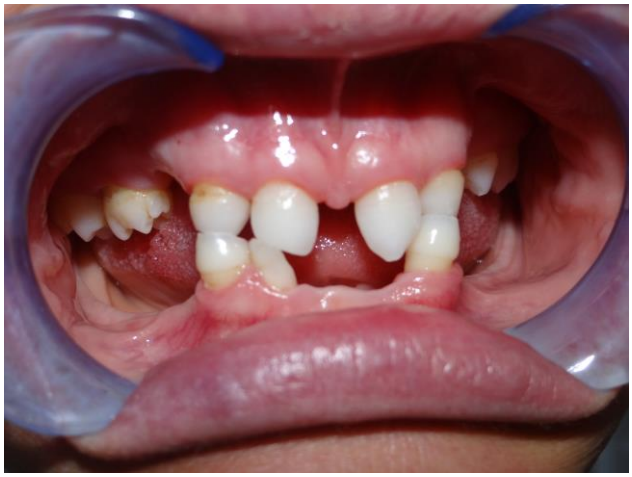

Figure 4. Intraoral view showing conical anterior teeth, resorption of alveolar ridge and hypodontia.

Panoramic radiograph showed presence of maxillary incisors, right and left maxillary first and second molars, right mandibular lateral incisor, right and left mandibular canine teeth with complete root formation, oligodontia, malformed dentition and underdevelopment of alveolar ridges (Figure 5).

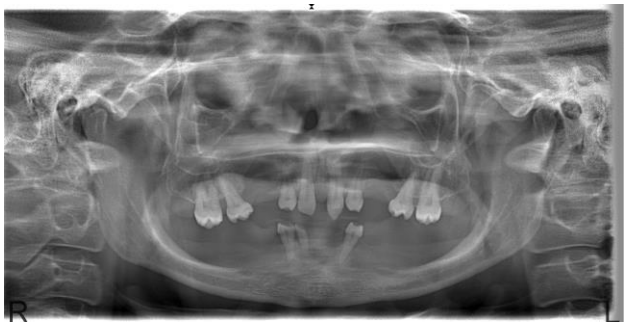

Figure 5. Panoramic radiograph showing oligodontia, malformed dentition and underdevelopment of alveolar ridges.

Based on the clinical findings, she was diagnosed with hypohidrotic ED. The patient was referred to Department of Dermatology. After the diagnosis, the patient was consulted to Department of Prosthodontia and treatment was planned to correct shape abnormality of upper anterior teeth and compensate difficulty in mastication and speech. Root canal therapy was performed in the mandibular right lateral incisor, right and left canine teeth because of the excessive loss of teeth tissues and enlargement of periodontal ligament space. Full mouth restoration in maxilla, removable partial denture in mandible was planned. Maxillary teeth were prepared for the metal supported fixed denture. Mandibular right lateral incisor and canine teeth were restored with metal supported fixed dental prostheses using extracoronal attachments. In mandible, Kennedy Class 1 toothless areas were restored with precision attachment-retained removable partial dentures (Figure 6-7).

Prothesis wounds depends on the excessive atrophy in the toothless areas, removed using soft denture lining materials (UFI GEL, VOCO). Thus, the patient gained esthetic, functional and phonetic benefits, better outlook in her social life and oral hygiene instructions were given to the patient. The patient was advised to be fed with soft foods for the first few days and to remove the dentures at night to support the healing of oral tissues. It was said that continued follow up was necessary for modification or replacement of the dentures depends on developing maxilla and mandible of the patient.

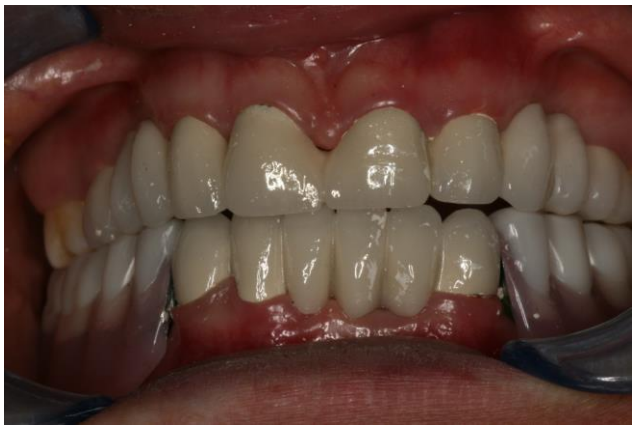

Figure 6. Frontal view of the patient showing improved facial appearance after the prosthetic rehabilitation.

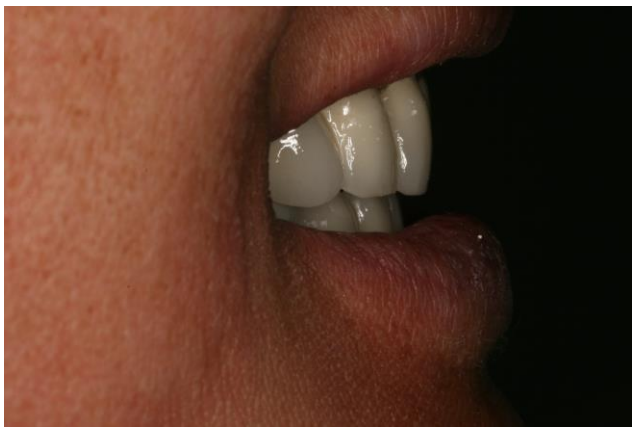

Figure 7. Sagittal view of the patient showing improved facial appearance after the prosthetic rehabilitation.

Discussion

ED was first described by Danz in 1792 (7) and later in 1838, ED in a letter that described 10 cases of Hindu male family members, was documented to Charles Darwin by Wedderburn (8). In 1895 Nicolle and Hallipre described firstly hydrotic ED in a French-Canadian family (9). In 1913, Christ rendered hypohidtotic ED as a congenital ectodermal defect (7). In 1921, X-linked nature of inheritence was described by Siemens. The term of ED was found by Weech in 1929 (10).

In 1936, Touraine described the wide range of features in ED. Therefore, hypohidrotic ED is also termed as Christ-Siemens-Touraine syndrome (11). The term of anhydrotic ED was used by Clouston in 1939 (12). Felsher in 1944 changed the term of anhydrotic to hypohydrotic because the individuals who was termed as anhydrotic weren't truly lack of sweat glands (13). Clouston in 1939 and Lowry et al. in 1966 described ED as a genetic entity (14). Hydrotic ED is also termed as Clouston's syndrome (12). Freire-Mara defined the nosologic group of ED that include at least two of the following situations:

1) Trichodysplasia (abnormal hair)
2) Abnormal dentition
3) Onchodysplasia (abnormal nails)
4) Dyshidrosis (abnormal or missing sweat glands) (4).

The causative gene has currently been identified in about 30 different EDs (15,18). Due to understanding of the genetic basis of this syndrome, newer classification, which benefit from moleculer information as the starting point and are based on the defects in cell-cell communication and signalling, transcription regulation, adhesion and development are suggested, such as the Priolo and Lagana classification in 2001 and the Lamartine classification in 2003 (16,18).

Mutations of EDA (ectodysplastin A protein), EDAR (ectodysplastin A receptor) and EDARADD (ectodysplastin A receptor related to death domain) are now described to cause hypohidrotic ED. These genes makes proteins that work together during embryonic development and these proteins form part of a signaling pathway which is critically important for the interaction between the ectoderm and mesoderm. These cell layers comprise many of the organs and tissues. Ectoderm- mesoderm interaction is necessary for the formation of several structures that originate from ectoderm such as skin, nails, hair, teeth and sweat glands. Mutations of these gens prevent the normal interaction between ectoderm and mesoderm and impairs the development of hair, teeth, sweat glands. The unproper formation of the ectodermal structures form the characteristic features of Hypohidrotic ectodermal dysplasia (19). Most abnormalities in craniofacial morphology can be attributed to the absence of teeth, although some authors have suggested that changes in embryonic morphogenesis could also be responsible (20).

The Minor test or iodide-starch test is very useful for confirming the absence of sweating, which is generalized inaffected individuals and patchy in female carriers, who have normally functioning eccrine glands alternating with eccrine glands with reduced function in areas with a Blaschkoid distribution.

This mosaic distribution can be more quickly revealed by the examination of large areas of the body, such as the back. Moreover, such an examination is useful for differentiating between X-linked HED carriers and females affected by autosomally transmitted HED, in whom the function of the sweat glands is almost completely absent (21).Other methods for assessing sweating include iontophoresis after applying pilocarpine to the forearm, sweat pore count, and measurement of skin conductance or temperature; such methods are useful for screening but are less sensitive in patients with residual gland function (22). Skin biopsy is not normally essential for confirmation, but the deficiency of eccrine glands has a positive predictive value and diagnostic specificity of 100% (23) Molecular analysis is the only way of determining which gene is included, detecting carriers, and confirming the type of inheritance. This information is vital for genetic counseling (24).

In a retrospective study of More et al. total nineteen cases of ED were observed and these cases were in the age group of 4-30 years with the mean age of 12.89 years. This study showed that ED was more prevalent in males with a ratio of 1.7:1 (25). Conversely in our case, the patient is female.

Consanguineous marriage have higher risk of birth defects because the couple sharing a common harmful gene and passing it on to the child. The risk varies according to how closely the couple is related (26). The marriage of parents of all cases revealed that consanguineous marriages were more than nonconsanguineous marriages and when the patients with ED were analysed, consanguineous marriage was directly proportional to ED (25). The retrospective study of More et al (25). showed that %66.67 of parents had consanguineous marriage and had %68.42 offspring's affected with ED; whereas %33.33 had non-consanguineous marriage and had %31.58 offspring's affected

with ED. But in our case, the patient didn't give any information about consanguineous marriage of her parents.

Hypodontia is a frequent finding of ED in %80 of cases (27,28) Oligodontia which refers to anodontia of six or more teeth, is a rare situation with prevalence of % 0.3 in permanent dentition (29). In this case, we observed oligodontia. According to Becktor et al (30), maxillary lateral incisors, second premolar and molars, mandibular incisors, second premolar and molars are usually absent. Balshi and Wolfinger (31) reported that mandibular anterior teeth are usually nonexistence in patients who has ED. In our case, we observed maxillary lateral incisors, right and left maxillary first and second molars, right mandibular lateral incisor, right and left mandibular canine teeth.

Abnormal dentition and decreased salivary flow cause difficulty in mastication, deglutition of food and speech (32). We found similar findings in our patient. Because of inadequate lubrication of vocal cords and more frequent acute laryngitis, the possibility of developing cord nodules may increase. Speech therapy is suggested for improving voice quality (32). We noticed that our patient has worse voice quality than the normal.

The time to begin dental treatment isn't definitive but Pigno et al (4). suggested that prosthodontic treatment should be provided before school age of the patient. But in our case, the patient was diagnosed late because she hadn't referred to any medical clinic.

Removable prosthesis (total dentures, partial dentures or overdentures) is the most commonly used treatment of choice for the oral rehabilitation because these treatment modalities can be easily modified during periods of rapid growth (4,33). In our case full mouth restoration in maxilla and removable partial denture in mandible were planned and retained teeth were prepared for the fixed denture to compensate microdontia. Dental implants may be one of the treatment modalities because of growth of adult patients is stabilized and implants can be used thus the prothesis was supported, retained and stabilized (28). In our patients dental implants weren't thought because of the alveolar ridge resorption and the economical status of the patient.

There is no treatment for the skin disorders or periocular hyperpigmentation, and outbreaks of atopic dermatitis may be difficult to treat. Some authors have suggested that the risk of melanoma increases in these patients , so a full physical examination is advised once a year (34).Early dental care to prevent maxillary hypoplasia and gum atrophy, which if severe may prevent chewing and language development in addition to being a remarkable aesthetic problem. Other specialists may also be involved the treatment of these patients for example, ear-nose-throat specialists when nasal and cerumen secretion is a problem, ophthalmologists when eye dryness or problems with the eyelids are present, pulmonologists in the event of respiratory tract infections and, in some case, psychologists (35). Gene therapy with recombinant EDA is still in the experimental phase, but it may be hope for these patients in the future (36,38).

In our case, the patient was comfortable with her fixed and partial denture and was satisfied with the esthetics of denture. Her parents reported significant progress in point of mastication and speech.

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