IDR International Dental Research

Original Article

Ectodermal dysplasia: A retrospective evaluation of the clinical findings of forty-four cases in the 0-16 years age

Ebru Akleyin[1], Cansu Osmanoğulları Sarıyıldız[2], İzzet Yavuz[3], Özkan Adıgüzel[4]

[1] Dicle University, Faculty of Dentistry, Department of Paediatric Dentistry, Diyarbakır, Turkey
[2] Diyarbakır Dental Health Center, Diyarbakır, Turkey
[3] Harran University, Faculty of Dentistry, Department of Paediatric Dentistry, Şanlıurfa, Turkey
[4] Dicle University, Faculty of Dentistry, Department of Endodontics, Diyarbakır, Turkey

Abstract

Aim: The aim of this study was to review the craniofacial anomaly results of children diagnosed with ectodermal dysplasia (ED) and to identify the oral requirements of ED cases.

Methodology: The data of this study were obtained by taking clinical examinations and radiographs on forty-four (44) children (22 females and 22 males), 0-16 years aged, who were admitted to the Dicle University, Faculty of Dentistry due to dental problems and were diagnosed with ED. The number of affected siblings was ascertained, and systemic findings were evaluated. Malformations in the hair, nails, nose, skin, lips, and teeth were clinically examined.

Results: The most common clinical findings were sparse hair, dry skin, sweating problems, respiratory difficulty, saddle nose, a history of fever, hearing loss, and deformation in the nails. In the intraoral and radiological examinations, findings were evaluated of conical teeth, protuberant lips, prosthetic rehabilitation, impacted teeth, and abnormal root resorption. The rates of ED in the siblings of the ED cases were determined as 39.2% in the siblings of male cases and 37.9% in the siblings of female cases.

Conclusion: The frequency rates of the anomalies seen in ED obtained in this study can be considered important as a guide for further studies of individuals with ED. When multiple missing teeth and conical teeth are encountered, the dental practitioner should investigate whether or not there are other symptoms of ED, and it must not be forgotten that the dentist may be the first step in the diagnosis of this genetic irregularity.

Keywords: ectodermal dysplasia, malformation, conical tooth, sparse hair

Correspondence:

Dr. Ebru AKLEYİN
Dicle University, Faculty of Dentistry,
Department of Pediatric Dentistry,
Diyarbakır, Turkey.
E-mail:dt.eakleyin@gmail.com

Received: 13 October 2021
Accepted: 27 June 2022

Access Online

DOI:
10.5577/intdentres.2022.vol12.no1.4

How to cite this article: Akleyin E, Sarıyıldız Osmanoğulları C, Yavuz İ, Adıgüzel Ö. Ectodermal dysplasia: A retrospective evaluation of the clinical findings of forty-four cases in the 0-16 years age. Int Dent Res 2022;12(1):21-6.
https://doi.org/10.5577/intdentres.2022.vol12.no1.4

Introduction

Ectoderm, which is one of the three germ layers in the developing embryo, is found in the central nervous system, peripheral nervous system, sweat glands, hair, nails, and dental enamel (1). Ectodermal dysplasia (ED) is defined as a hereditary irregularity characterized by a broad range of clinical findings and two or more tissue anomalies in tissues developing from the ectoderm,

such as hair, teeth, nails, and sweat glands (1, 2). ED was first reported by Danz in 1792, and of approximately 200 different ectodermal dysplasia, approximately 30 have been identified at the molecular level with the identification of the causative gene. Some studies in the literature have reported the genetic information of ED with clinical findings (3). The prevalence of ED is estimated to be at least 1 in 5000-10,000 live births (2, 4). However, this estimation is inaccurate as cases may be overlooked in infancy before the cardinal characteristics have become evident. It has been reported that all racial and ethnic groups are affected (4). In individuals with ED, the hair, eyebrows, and eyelashes are fine, sparse, or absent. There may be a projecting forehead, thick, extroverted, protruding lips, and a saddle nose. Fine lines around the nose and eyes give the face an elderly appearance. Due to dystrophic nails and the partial or complete absence of sweat and fat glands, the skin is soft, dry, smooth, and thin, and therefore hyperthermia may develop in hot conditions (1, 2, 5). Due to the absence of teeth, the alveolar crest cannot develop, there is a loss of height in the jaws, and retarded development is seen in the sagittal plane.

The palatinal arch is generally deeper; in some cases, cleft lip/palate may be seen. Dry mouth and dysphagia may be related to the insufficient working of the saliva glands associated with secretory gland anomalies. A number of anomalies ranging from hypodontia in the milk and permanent dentition to anodontia have been reported as conical-shaped incisor and canine teeth, diastema related to missing teeth, abnormal root shapes, resorption of permanent teeth roots, and enamel hypoplasia (6, 7). The most common form of ED is hypohydrotic ectodermal dysplasia (HED), which is characterized by hypo hidrosis (associated with hypoplasia of the sweat glands), hypotrichosis (sparseness of scalp and body hair), and hypodontia. The majority of individuals with HED have the X-linked form (8). Three basic characteristics are targeted in the management of this genetic irregularity. These targets have been reported as optimizing psychosocial development, creating optimal oral function, and reducing the effect of the clinical symptoms by which these genetic irregularities diminish the quality of life. Studies in literature have shown that oral rehabilitation is useful for psychological development in respect of the patient regaining self-confidence and appearing and feeling like their peers (1, 9). ED causes psychological problems in children as a result of impairments in function, phonation, and aesthetics because of anodontia or oligodontia, nutritional deficiencies, therefore retarded physical development.

The aim of this study was to evaluate the data obtained retrospectively and prospectively from pediatric ED cases and thereby determine the rates of anomalies occurring in different tissues, raise the awareness of dental practitioners about this genetic irregularity, and increase the quality of life in collaboration with the cases.

Materials and Methods

This retrospective study included forty-four cases (22 male, 22 female) who presented at Dicle University Faculty of Dentistry between 1997 and 2020 and were diagnosed with ED. The anamnesis and clinical and radiological examinations of individuals with ED were obtained and analyzed. The number of siblings affected, history of fever, and hearing loss were ascertained from the anamnesis. In the clinical examination, the sparseness of the hair, deformation in the nails, saddle nose, and dry skin were evaluated. The oral region was evaluated in respect of protuberant lips, conical teeth, respiratory difficulty, and oral prosthetic rehabilitations. Root resorptions and impacted teeth were evaluated with panoramic, and cone-beam computed tomography.

Statistical analysis

Analysis of the data was carried out with IBM SPSS Version 22 (IBM SPSS Inc., Armonk, NY, USA). Frequency analysis was applied, and data were reported in graph form.

Results

An evaluation was done of forty-four ED cases, comprising 22 females and 22 males, with a mean age of 9.14 years (range, 1,5 -16 years). All forty-four cases had missing teeth. The number of teeth present (including teeth and teeth germs) was a mean of 18.4 (range, 2 – 28). In 8 cases, the number of teeth was <6 (Fig. 1, 2).

The clinical findings determined in the cases were sparse hair at 97.7%, dry skin at 79.5%, sweating problems at 79.5%, respiratory difficulty at 70.5%, saddle nose at 70.5%, a history of fever at 68.2%, hearing loss at 52.3%, and deformation in the nails at 47.7%. The rates of extra-oral clinical findings of ED seen in the cases are shown in Table 1. In the intra-oral and radiological examinations of the cases, conical teeth were determined in 72.2% (Fig 3), protuberant lips in 68.2%, prosthetic rehabilitation in 20.5%, impacted teeth in 15.9%, and abnormal root resorption in 9.1% (Table 2). The rates of siblings with ED were determined as one sibling in 61.4% of the ED cases, two siblings in 27.3%, three siblings in 9.1%, and four siblings in 2.3% (Table 3). When the siblings of the ED cases were examined, the male ED cases had a total of 79 siblings, of which 31 had ED, a frequency rate of 39.2%. The female ED cases had a total of 95 siblings, of which 36 had ED, a frequency rate of 37.9% (Table 4).

Table 1. The ratio of extra-oral clinical findings of ectodermal dysplasia cases

Table 2. The ratio of intra-oral clinical findings of ectodermal dysplasia cases

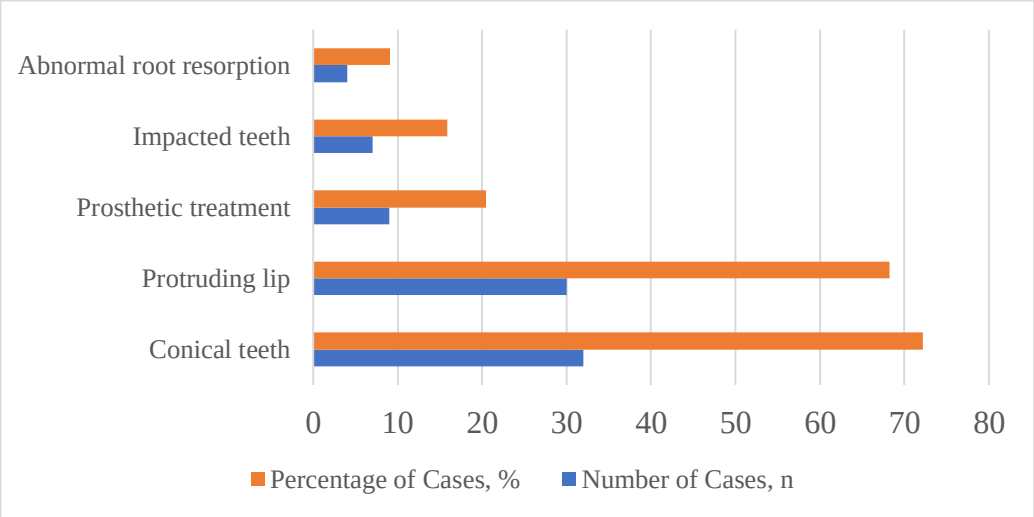


Table 3. The ratio and percentage of siblings of ectodermal dysplasia cases

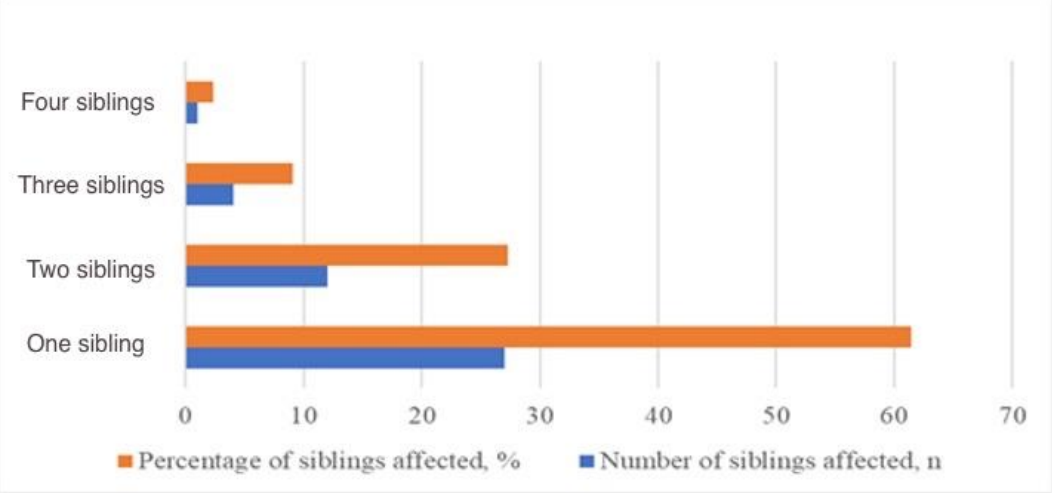
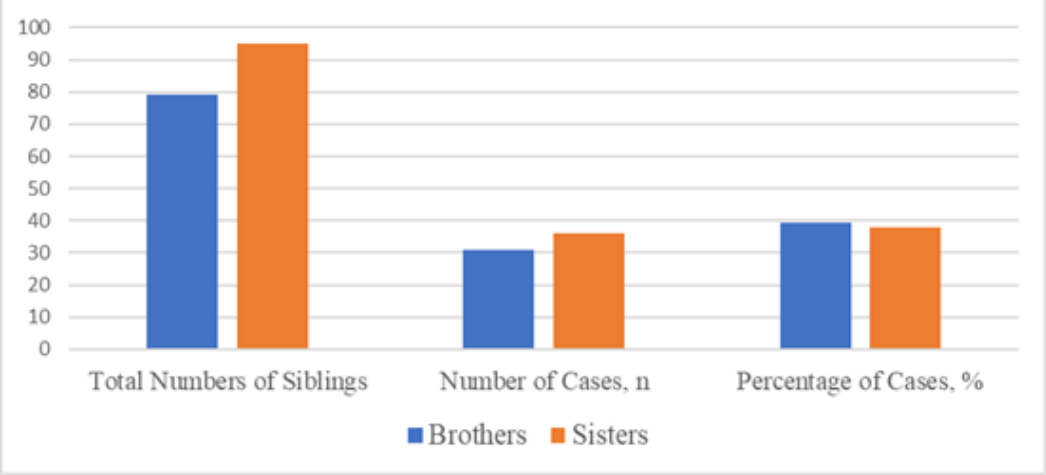


Table 4. The ratio and numbers of siblings of ectodermal dysplasia cases



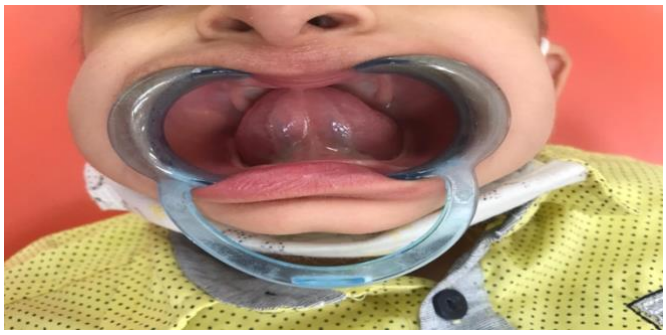


Figure 1. Our 5-year-old case with total tooth deficiency

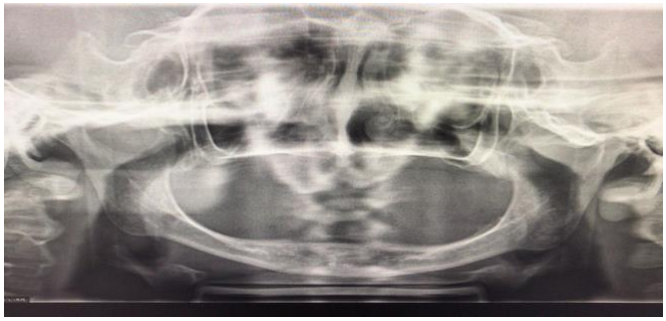


Figure 2. Our 5-year-old case with panoramic radiography

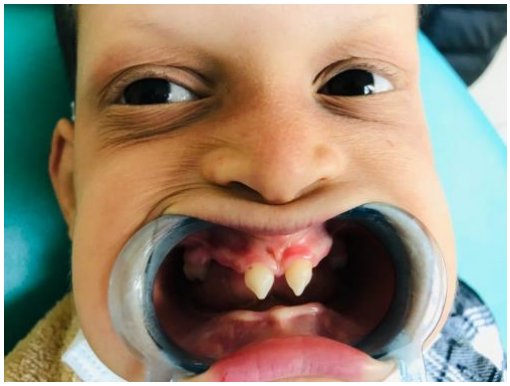


Figure 3. Conical teeth anomaly and oligodontic appearance in an ectodermal dysplasia case.

Discussion

Ectodermal dysplasia is an uncommon genetic irregularity, estimated to be seen in 7 per 100,000 births (3). In a study by Bondarets et al. (10), which examined 61 cases with a mean age of 11 years, there was reported to be severe hypodontia, with a mean of 15.4 (range, 6-28) missing teeth (excluding third molars). In the current study of forty-four cases with a mean age of 9 years, the number of teeth present (including teeth and teeth germs) ranged from 2 to 28 (mean 18.4), and in all cases, there were missing teeth and severe hypodontia, consistent with previous studies (1, 5, 10). In 8 of the current study cases, the number of teeth was <6, and severe oligodontia and chewing difficulty were determined in these cases. ED is a hereditary irregularity characterized by congenital dysplasia of one or more ectodermal structures. Common symptoms include fine and sparse hair follicles and eyebrows, collapsed bridge of the nose, and protruding lips. Cases may suffer from dry skin because of a lack of sweat glands, hyperthermia, and unexplained fever. Within the mouth, common findings are anodontia or hypodontia and conical teeth (11).

In a study of 75 ED cases, Mehta et al. (12) reported sparse hair (41%), heat intolerance (76%), saddle nose (44%), and hearing loss (48%). Yavuz et al. (1) evaluated 15 cases and reported findings of sparse hair (80%), dry skin (93%), sweating problems (86%), saddle nose (73%), a history of fever (80%), hearing loss (53%) and deformation in the nails (80%). In a study of 19 ED cases, More et al. (13) reported that the number of teeth presents ranged from 0 to 19, and the clinical findings were seen to be dry skin (94.74%), sparse hair (100%), saddle nose (57.89%), nail anomaly (52.63%), and abnormal sweat glands (78.95%). Yıldırım et al. (14) evaluated 23 cases, comprising 11 males and 12 females, ranging in age from 5-45 years, who were diagnosed with ED between 2006 and 2008, and reported hearing loss (39.1%) and saddle nose deformity (56.5%).

In the current study, no difference was determined between the genders. Consistent with the findings in the literature, the leading symptoms of the ED cases were seen to be sparse hair (hypotrichosis, 97.7%), thin, dry skin (79.5%), lack of or reduced sweating because of the partial or complete absence of sweat glands (anhydrosis or hypohydrosis, (79.5%), the respiratory difficulty associated with the absence (aplasia) or poor development (hypoplasia) of the mucous glands in the respiratory tract of the majority of the affected children (70.5%), a history of recurrent fever (68.2%), slow nail growth, protrusions in the back of the neck, collapse and dysplasia with concavities of varying degrees (47.7%), saddle nose (70.5%), and hearing loss (52.3%). In studies of ED cases, the finding of conical teeth was reported at the rate of 75.51% by Doğan et al. (15), 84.21% by More et al. (13), and 76% by Mehta et al. (12).

In the current study, conical teeth were determined at the rate of 72.2% in intra-oral and radiological examinations as the most evident dental anomaly seen in the ED cases, which was consistent with the findings in the literature. The gene disorders causing ED (Eda Edar, Edaradd) play an important role in dental development, the number of teeth, crown shape, and root formation. During root development, the Edar gene is expressed by the epithelial root sheath of Hertwig, and mutation causes root anomalies (16). In addition to missing teeth, all the cases in the current study were determined with shape anomalies of the teeth, impacted teeth, and abnormal root resorption. The most complaints of the cases were of anomalies related to the teeth.

The typical facial appearance of forward protrusion, thick lips, and a wide nasal bridge lead to social anxiety and diminished quality of life in individuals with ED. According to case reports of children and adolescents, they have stated that they are the targets of mockery, criticism, and exclusion from their peer groups. Previous studies have reported that surgical or non-surgical aesthetic correction has generally led to an improvement in quality of life and a general improvement in psychological health (17). Loss of vertical height because of missing teeth leads to an appearance of thick and protruding lips in ED cases. More et al. (13) reported that protuberant lips were seen at the rate of 57.89%. In the current study, this rate was higher, with protuberant lips seen in 68.2% of the cases. In children with anodontia or severe oligodontia, as the majority of cases are aged <5 years, early prosthesis treatment seems to be positive in respect of food intake, speech development, and socialization. Whatever the age, prostheses which are fully or partially mobile are a treatment method frequently applied in children. Oral hygiene, prosthesis adaptation, and a staged approach to the treatment goals form the basis of treatment success. Renewal of full prostheses is expected in 1-3 years and of partial prostheses in 1-6 years. The survival of mandibular implants in children aged 6-14 years has been reported to range from one year to 26 years (7). Although all the children in the current series had missing teeth, only 20.5% received prosthetic rehabilitation treatment. Traditional prosthetic treatments (full prostheses, combinations of fixed and mobile partial prostheses) have many problems because of the teeth that are present and the anatomic anomalies of the bone. In young cases, there is usually a need for the prosthesis to be re-structured as the child grows. For treatment planning at a young age, the current study participants were referred for a multidisciplinary approach, including a specialist pediatric physician, a specialist prosthodontist, an orthodontist, and a pedodontist.

Different types of ED have been identified, showing various clinical findings. The genetic carrier model includes autosomal dominant, autosomal recessive, and X chromosome-linked recessive characters. ED can be seen in both male and female children (18, 19). In this context, as the rates at which males and females are affected will be different according to the genetic carrier model, it was thought that there would be no significant relationship between the siblings with ED. There can be considered to be a need for gene mapping evaluation in these cases to be able to obtain more detailed information.

Conclusions

Dental practitioners should have sufficient knowledge and experience in respect of supporting the normal growth and development of patients with ED with the restitution of aesthetics, function, and phonation. As dental practitioners may be the first step in the diagnosis of this uncommon genetic irregularity, it is important that further studies are conducted to raise awareness of ectodermal dysplasia.

Ethical Approval: Ethics committee approval was received for this study from Dicle University, Faculty of Dentistry Ethics Committee, in accordance with the World Medical Association Declaration of Helsinki, with the approval number: 2021/23).

Peer-review: Externally peer-reviewed.

Author Contributions: Conception – E.A.; Design – C.O.S., İ.Y.; Supervision – Ö.A.; Materials – E.A., F.H.; Data Collection and/or Processing – C.O.S., İ.Y.; Analysis and/or Interpretation – Ö.A.; Literature Review – E.A., F.H.; Writer – F.H.; C.O.S; Critical Review – İ.Y.

Conflict of Interest: No conflict of interest was declared by the authors.

Financial Disclosure: The authors declared that this study has received no financial support.

References

1. Yavuz I, Baskan Z, Ulku R, Dulgergil TC, Darı O, Ece A, et al. Ectodermal dysplasia: retrospective study of fifteen cases. Arch Med Res 2006; 37(3): 403-9. (Crossref)
2. Reyes-Reali J, Mendoza-Ramos MI, Garrido-Guerrero E, Méndez-Catalá CF, Méndez-Cruz AR, Pozo-Molina G.

Hypohidrotic ectodermal dysplasia: clinical and molecular review. Int J Dermatol 2018; 57(8): 965-72. (Crossref)
3. Itin PH, Fistarol SK. Ectodermal dysplasias. Am J Med Genet 2004; 131(1):45-51. (Crossref)
4. Wright JT, Grange DK, Fete M. Hypohidrotic Ectodermal Dysplasia. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews [Internet]: University of Washington, Seattle; 1993-2018. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1112/
5. Wohlfart S, Hammersen J, Schneider H. Mutational spectrum in 101 patients with hypohidrotic ectodermal dysplasia and breakpoint mapping in independent cases of rare genomic rearrangements. J Hum Genet 2016; 61(10): 891-7. (Crossref)
6. Elhamouly Y, Dowidar KM. Dental management of a child with ectrodactyly ectodermal dysplasia cleft lip/palate syndrome: a case report. Special Care in Dentistry 2019; 39(2): 236-40. (Crossref)
7. Chrcanovic BR. Dental implants in patients with ectodermal dysplasia: a systematic review. J Cranio Maxill Surg 2018; 46(8): 1211-7. (Crossref)
8. Schnabl D, Grunert I, Schmuth M, Kapferer-Seebacher I. Prosthetic rehabilitation of patients with hypohidrotic ectodermal dysplasia: a systematic review. J Oral Rehabil 2018; 45(7): 555-570. (Crossref)
9. Clarke A. Hypohidrotik ectodermal dysplasia. J Med Genet 1987; 24(11): 659-63. (Crossref)
10. Bondarets N, Jones RM, McDonald F. Analysis of facial growth in subjects with syndromic ectodermal dysplasia: a longitudinal analysis. Orthod Craniofac Res 2002; 5(2): 71-84. (Crossref)
11. Itthagarun A, King NM. Ectodermal dysplasia: A review and case report. Quintessence Int 1997; 28(9): 595-602.
12. Mehta U, Brunworth J, Fete TJ, Sindwani R. Head and neck manifestations and quality of life of patients with ectodermal dysplasia. Otolaryngol Head Neck Surg 2007; 136(5): 843-7. (Crossref)
13. More, CB, Bhavsar K, Joshi J, Varma SN, Tailor M. Hereditary ectodermal dysplasia: A retrospective study. J Nat Sci Biol Med 2013; 4(2): 445-50. (Crossref)
14. Yıldırım M, Yorgancilar E, Gun R, Topcu I. Ectodermal dysplasia: otolaryngologic evaluation of 23 cases. Ear Nose Throat J 2012; 91(2): E 28-33. (Crossref)
15. Doğan MS, Ataş OU, Yavuz I, Tekin S. Investigation of clinical and radiogical in cases with ectodermal dysplasia. Fırat Med J 2019; 24(2):85-8.
16. Fons Romero JM, Star H, Lav R, Watkins S et al. The impact of the eda pathway on tooth root development. J Dent Res 2019; 96(11): 1290-7. (Crossref)
17. Deo K, Sharma YK, Shah B, Kothari P, Chavan, D, Sitaniya S, Gupta A. Improvement in the quality of life of a patient of ectodermal dysplasia with reconstructive surgeries. J Cutan Aesthet Surg 2019; 12(4): 244. (Crossref)
18. Över H, Akşit KS. Prosthetic rehabilitation patients with ectodermal dysplasia: case report. J Dent Fac Atatürk Uni 2012; 2: 180-4.
19. Itin PH. Etiology and pathogenesis of ectodermal dysplasias. Am J Medi Genet, 2014; 164(10): 2472-7. (Crossref)