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Effects of Ectodermal Dysplasia on the Maxilla: A Study of Cone-Beam Computed Tomography

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Abstract

Background: This study aimed to examine the effects of ectodermal dysplasia (ED) on the transverse width of the maxillary bone.

Methods: The ED group was composed of seven people, while the control group consisted of retrospective cone-beam computed tomography images of seven individuals with skeletal class 1 relationship. Images on the sagittal planes were taken, and cross-sections were taken from the longest point of the Anterior Nasal Spine-Posterior Nasal Spine line. The distance between the distal anterior canine teeth from the right buccal cortical bone to the left buccal cortical bone was measured. At the posterior region, the distance between the right point where the pterygoid protrusions and the tuber maxilla fused and the left point was measured.

Results: The ED group has significantly narrower ($p < 0.05$) anterior region than the control group, and no significant difference in the posterior region width was found between the ED group and control group.

Conclusions: The quality of life should be improved by awareness of ED in dentistry, by using a professional approach and modern applications such as three-dimensional computed tomography when necessary, and by considering the morphological characteristics of the patients.

Keywords: ectodermal dysplasia, cone-beam computed tomography, maxilla, maxillofacial development

INTRODUCTION

Ectodermal dysplasia (ED) is a rare hereditary disease characterized by dysplasia of tissues of mostly ectodermal origin.[1] ED can have autosomal dominant, autosomal recessive, or X-linked genetic transmission. The most common types are hypohydrotic (Christ–Siemens–Touraine syndrome) and anhydrotic ED.[2–5] Previous studies have reported that the most common type of ED is X-linked hypohydrotic ED (HED), which commonly affect men, while women show varying symptoms due to X-chromosome inactivation.[6–8] In ED, in which ectodermal structures are affected, although a wide spectrum is formed clinically, the hair (hypotrichosis, partial or total alopecia), nails (dystrophic, hypertrophic, or abnormal keratinized), teeth (enamel defect or absence), and sweat glands (hypoplastic or aplastic) are usually affected, and it occurs in less than 1 in every 100,000 individuals.[9–15] At present, clinical studies on the treatment of X-linked HED are on-going, and preliminary results are expected in the coming years.[16]

Clinically, patients with ED classically show the triad of hypotrichosis (insufficient hair growth), hypohydrosis (insufficient sweat secretion), and cranial abnormalities. With the prominence of the forehead (frontal bossing), there is a concavity in the nasal area, despite having a small face. Owing to the absence of sweat glands, patients have a very smooth and dry skin, and hyperkeratosis in the hands and feet is observed as a clinical sign of hypohidrosis. Anodontics, hypodontics, and conical teeth are oral symptoms. Anodontics is also manifested by a developmental disability in the alveolar ridge.[17–19] Most permanent teeth have structures similar to the primary teeth, but patients may have few or no permanent teeth at all. Teeth are generally conical, giving an undesirable aesthetic appearance. The most common missing teeth are maxillary central incisors (42%), maxillary first molars (41%), mandibular first molars (39%), and maxillary canines (22%); the absence of other teeth is less common.[2] Patients with HED exhibit a skeletal class 3 trend as a result of mid-facial hypoplasia and show a flat or concave face in which mandibular protrusion can be traced, which is associated with maxillary retrusion.[20–22]

Yavuz *et al*. stated that cranial, facial, nasal, and maxillary widths were affected by gender in the transverse measurements performed on posteroanterior cephalometric X-ray images.[23] In their longitudinal study, Snodel *et al*. found that growth continued after age 18 years, except the maxillary width, in men (age 4–25 years); in women (age 4–20 years), skeletal growth was completed at age 17 years.[24] They found a correlation between maxillary width and maxillary inter molar width and an increase in maxillary intermolar distance that

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reflects the increase in the maxillary width. Transverse enlargement of the face is proportionally less than the sagittal or vertical enlargement of the face. A significant increase in maxillary width occurs in girls aged 6–12 and in boys aged 7–11 years, and the increase continues until age 16 years, albeit a little.[24]

A study examined the intercanine and intermolar widths of class 1, class II-1, class II-2, class III, and class II subdivision groups and found no significant difference between the intermolar and intercanine widths between the five malocclusion groups.[25] Staley *et al*. reported that individuals with normal occlusion had greater maxillary molar widths, canine interdental widths, and alveolar widths than individuals with class II-1 malocclusion, by working on plaster models and cephalometric X-rays.[26]

As regards the eruption of canines and molars, a significant increase was found in intercanine and intermolar arch widths from birth to mid-adulthood.[27] The increase in intercanine width lasts until mid-adulthood and then decreases relatively insignificantly.[28] The erupted upper canines significantly affect the distance between the maxillary canines.[29]

Posterior cross-bite was defined as one of the most common skeletal anomalies in the craniofacial region, in which the upper jaw has stenosis in the transverse direction.[30] Developmental disability in the transverse direction may occur due to genetic or environmental cause.[31] In the study using CT images and plaster model, the maxillary basal arch was insufficient in patients with skeletal class 3 malocclusion compared with individuals with normal occlusion.[32] Seven-year-old children with class l-II and class l-III occlusions have a smaller maxillary width of 2.5 and 4 mm, respectively, than individuals with normal condition.[33] Dental compensation results from skeletal incompatibilities in the posterior segments of individuals with class II and III maxillary occlusions.[34]

Çoban *et al*. evaluated the pharyngeal airway volume in three dimensions in individuals with different malocclusions. Total and oropharyngeal airway volumes of class l-II cases were significantly lower than that of class l-I and class l-III cases. No significant difference was found between the airway volumes of class l-I and class l-III individuals.[35] While maxillary growth decreases in individuals with ED, the maxilla increases by 2 mm in the anterior region in the transverse direction and 5 mm in the hamular region between age 6 and 10 years.[36] Dental agenesis in individuals with HED results in oligodontia, which causes severe atrophy of the alveoli and underdevelopment of the maxilla/mandible.[37]

ED significantly affects all craniofacial structures, including hard and soft tissues. Especially, the mid-facial area is severely affected in the sagittal direction. The maxilla, which is among the morphological structures that make up the mid-facial region, is also significantly affected. Very few cone-beam CT (CBCT) studies have focused on ED to date. Thus, this study aimed to examine the effects of ED on the transverse width of the maxillary bone, which is located in the mid-facial region of individuals, by using CBCT.

METHODS

Before the study commenced, ethics committee approval was obtained from the local ethics committee. The experimental group was composed of seven individuals (4 women and three men with ED), while the control group consisted of seven individuals (three women and four men with skeletal class 1 (ANB 0-4) relationship). The mean ages of the female and male group were 20.25 and 18.67 years, respectively. CBCT images of 14 individuals were investigated. Patients had multiple missing teeth, while some had retained primary teeth and incisors; canines of one patient have conical shape (Table 1).

The linear distance between the right buccal cortical bone and the left buccal cortical bone between the anterior canine fossa was measured on the transverse plane from the place where the pterygoid protrusions first meet with the maxillary tuberosity (from bottom to top), parallel to the Frankfurt horizontal plane, on CBCTs. In the posterior region, the linear distance between the right point where the pterygoid protrusions and the maxillary tuberosity first meet and the left point was measured.

The exclusion criteria of the respondents were those with history of orthodontic treatment or surgical operations in the head and face area. This study categorized the respondents into two groups. The first one consists of respondents who were diagnosed with ED, and the second group was the control group. The inclusion criteria of the control group were those without airway pathology or craniofacial syndrome, without multiple missing teeth (n > 4) that may affect the vertical size, and have skeletal class 1 (ANB 0-4) relationship.

Reference points, lines, planes, and metric measurements (Figures 1–3)

• Frankfurt horizontal plane [plane passing through the middle of the porion and both orbital points on both sides]

• Sagittal plane (Figure 1), frontal plane (Figure 2), and transverse plane (Figure 3)

• Linear metric distances in the transverse plane (anterior region width, posterior region width)

Statistical analysis

In this study, descriptive statistics are given as number of individuals (n), mean, mean ranks, and sum of rank values. Distribution of the normality of measurements was evaluated with the Shapiro–Wilk normality test. The non-parametric Mann–Whitney U test was applied to look for the mean difference between two independent groups and to determine difference or equality between the groups. Moreover, 95% confidence intervals were used, and $p < 0.05$ in the results were considered significant.

Data are presented as number of observations (n), mean ± standard deviation, and range. Results of homogeneity (Levene's test) and normality (Shapiro–Wilk test) tests were used to decide the statistical methods for comparing study groups. Among normally distributed groups with homogeneous variances, dependent groups were compared using Student's t-test. According to the test results, parametric test assumptions were not available for some variables; therefore, independent groups were compared using the Mann–Whitney-U test.

TABLE 1. Age, dental status, and date CBCT images were taken from each patient with ectodermal dysplasia

Gender	Patient	Age	Dental status	Conical teeth
Female	Patient 1	24	Only #13 present	-
	Patient 2	22	Only #16 and #11 present at right side #15,18 impacted #21,23,26 present at left side	Centrals
	Patient 3	18	#16,13,11 present at right side #26,23,21 present at left side	Centrals and canines
	Patient 4	17	#11,21,23,25 present at right side #21,22,23,V,26 present at left side	Centrals
Male	Patient 5	22	#16,V,13,II,11 present at right side #21,23,V,26 present at left side	Centrals
	Patient 6	19	#16 and #26 present #13 and 23 implants	-
	Patient 7	15	#16,15,13,11,IV present at right side #21,23,25,26 present at left side	-

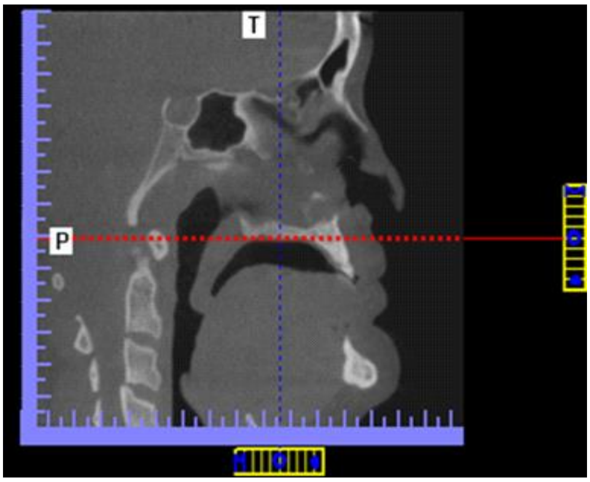

FIGURE 1. Reference plane on the sagittal plane, parallel to the Frankfurt horizontal on the maxilla

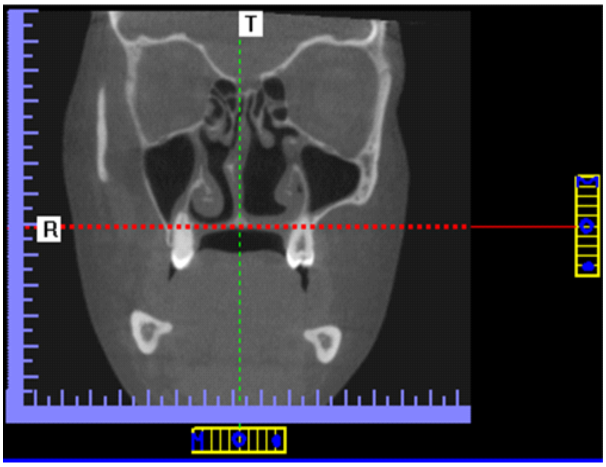

FIGURE 2. Appearance with defined distance between the reference points of the maxilla on the frontal plane

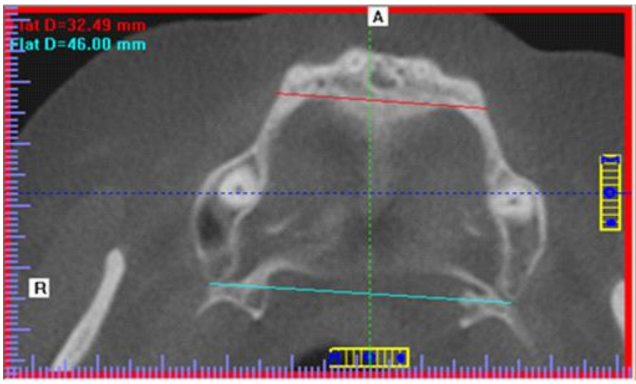

FIGURE 3. Linear metric measurements made from the maxilla in the anterior and posterior regions on the axial plane

RESULTS

According to the non-parametric Mann–Whitney-U test result between the control and ED groups in terms of the linear metric distances (the anterior region width between the specified points of the maxilla, on the cross-section taken in the transverse plane, from where the pterygoid processes first meet the maxillary tuberosity (from bottom to top), parallel to the Frankfurt horizontal plane), the difference between groups was significant (p = 0.041). The width of the anterior region of the ED group was significantly smaller than that of the control group (Table 2).

The linear metric distance, which is the posterior width between the specified points of the maxilla, was used to reveal differences between the control and ED groups on the cross-sections taken in the transverse plane, from where the pterygoid processes first meet the maxillary tuberosity (from bottom to top), parallel to the Frankfurt horizontal plane. Mann–Whitney U test revealed no significant difference between the two groups (p = 0.701). The width of the posterior region in the ED group was not significantly narrower than that in the control group (Table 2).

TABLE 2. Comparison and findings of the anterior and posterior maxillary width of the ectodermal dysplasia group and control group according to the Mann–Whitney Test

Groups	n	Mean	Mean Ranks	Sum of Ranks	p
Anterior width (mm)					
Ectodermal dysplasia	7	5,21	5,21	36,50	0,041*
Control	7	9,79	9,79	68,50	
Posterior width (mm)					
Ectodermal dysplasia	7	7,07	7,07	49,50	0,701
Control	7	7,93	7,93	55,50	

DISCUSSION

The use of CBCT in dentistry has led to a significant progress in diagnosis and treatment planning. In previous studies, three-dimensional structures were evaluated using two-dimensional X-ray images. With the development of three-dimensional imaging systems, disadvantages of two-dimensional imaging systems, such as distortion, magnification, and superimposition, have been eliminated. With the three-dimensional diagnostic methods, patient's problems and treatment alternatives are determined appropriately, and the physician can predict the treatment prognosis more accurately.[38] In the three-dimensional evaluation of the maxillofacial morphology, the use of not only cephalometric radiographic drawings but also of high-resolution analysis methods was recommended.[39]

Severe non-syndromic hypodontics cause underdevelopment of the lower anterior face height, anticlockwise rotation of the mandible, and mandibular prognathism due to insufficient occlusal support.[40,41] Endo *et al*. reported that the maxillary first molars were in the supra-position in non-syndromic hypodontic cases.[42] Nakayama *et al*. reported that although hypodonia in ED is a remarkable finding in all patients, the maxillary first molars were in the supra-position, resulting in a shorter anterior upper face height.[43] Insufficient vertical and anteroposterior growths of the maxilla, which is one of the typical facial features of ED, may be related to this situation. Johnson *et al*. compared healthy individuals and those with HED and found that those with HED experienced developmental disability of maxillary growth.[44] Saksena and Bixler noted that the maxillary transverse size was also reduced.[41]

Lexner *et al*. evaluated boys and girls with X-linked HED and found short maxilla in both groups; however, boys with HED had significantly shorter and retrognathic maxilla than girls with heterozygous HED, and the facial height was small.[46] Briefly, when the anterior cranial base and maxillary relationship in men was considered, the maxilla had a shorter, retrognathic, and more clockwise inclined position. In girls, when the relationship between the anterior cranial base and maxilla was compared, the maxilla was shorter and retrognathic but has normal inclination. The craniofacial morphology of individuals with ED is different from that of the normal population, and deviations are more pronounced in men.[46] Studies have stated that the cranial structures and facial, nasal, and maxillary widths are affected by gender[23,24]; for this reason, our study groups were formed with close homogeneity in terms of gender.

In their CBCT study, Karadede *et al*. found no significant difference between the maxillary volumes of class 1, 2, and 3 face type groups, and the maxillary volume in the class 2 and 3 short face type group was significantly larger than that in the long face type group.[47] In the present study, the control participants were selected from the normal population, but by nature, individuals with ED had a short face type. Therefore, intergroup differences may have been monitored less.

While some researchers have found no difference between intermolar and intercanine widths,[25] others reported that individuals with malocclusion had smaller maxillary molar, canine interdental, and alveolar widths.[26,32–34] As regards the eruption of canines and molars, a significant increase was found in the intercanine and intermolar arch widths from birth to mid-adulthood.[27–29] Dental agenesis in individuals with HED results in oligodontia, which causes severe atrophy of the alveoli and underdevelopment of the maxilla and mandible.[37] While functioning structures develop, functional structures become atrophied. For this reason, congenital tooth deficiencies, which are often seen in individuals with ED, may cause a delay in the buccal alveolar bone development and emergence of transverse deficiencies in that region. According to Moss' functional matrix theory, in individuals with ED, a significant decrease in the anterior width of the maxilla was thought to be due to the negative effect of congenital tooth deficiency concentrated in the premolar region on maxillary alveolar development.

In our study, in the linear metric measurements applied to cross-sections taken in the transverse plane, a significant decrease in the anterior region was found in the ED group but not significant decrease was found in the posterior region. The posterior region is less affected by the negative effect of tooth deficiency on the alveoli. Bhalha *et al*. examined children aged 6–10 years and found that the maxilla transversely increases by 2 mm in the anterior region and 5 mm in the hamular region.[36] Moreover, results of Johnson *et al*. and Saksena and Bixler are consistent with those of our study.[44,45]

Conceivably, one of the important reasons that the posterior region width is less affected by the anterior region is that the maxillary tuberosity fuses with the lateral pterygoid protrusions extending downward of the os sphenoidale and the suture palatina media is not yet fully fused. As a result, the transverse development of the cranial base is whipping the posterior transverse development of the maxilla.

CONCLUSIONS

The jaw structure of individuals with ED, which is characterized by a large number of tooth deficiencies, is affected by this condition. Moreover, unused organs atrophied and alveolar crests are affected by tooth extraction. Bone formation in areas with congenital tooth deficiency is also adversely affected. As a result, transverse distances are insufficient, especially in the anterior region. However, the posterior region associated with lateral pterygoid protrusions is positively affected by the transverse development of the skull base.

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CONFLICT OF INTEREST

The authors declare that there is no conflict of interest.

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ETHICAL COMMITTEE APPROVAL

The study protocol was approved by the Health Research Ethics Board of İzmir Katip Çelebi University, School of Medicine, a report of ethics committee decision numbered 0052. The study was conducted in accordance with the principles of the Declaration of Helsinki. In this study, CBCT images, which was previously recorded for diagnostic and therapeutic purposes, were used from the archives of İzmir Katip Çelebi University Faculty of Dentistry Department of Radiology.

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