Evaluation of patient with Holt-Oram syndrome in terms of oral and maxillofacial findings

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ABSTRACT

The Holt-Oram syndrome (HOS) is a genetic disorder with autosomal dominant inheritance associated with anomalies in upper extremities and heart and affects one out of every 100,000 live births. Maxillofacial development is also affected by these skeletal abnormalities. Although there are many studies about the HOS in the literature, the data about the development of oral and maxillofacial development are very few. In this study, evaluation of a child with the HOS is made in terms of dental and gingival health, oral and maxillofacial formation, and cephalometric analysis measurements are made for the 1st time in the literature and identified findings are discussed in company with the literature.

Key words: Atriodigital dysplasia, Cephalometric analysis measurement, Findings, Heart-hand syndrome, Holt-Oram syndrome

Access this article online

Website: www.jpediatrdent.org

DOI: 10.4103/2321-6646.174932

Quick Response Code:

INTRODUCTION

The Holt-Oram syndrome (HOS) is a genetic disorder with autosomal dominant inheritance associated with anomalies in upper extremities and heart and affects one out of every 100,000 live births.[1,2] It was identified and defined for the 1st time in 1960 by Holt and Oram in 9 individuals affected in four generations of a family.[3] It is also called atriodigital dysplasia, Heart-hand syndrome, upper extremity cardiovascular syndrome, cardiac extremity syndrome, and cardiomegalic syndrome.[4] Skeletal deformities cover upper extremities only, and bilateral thumb anomalies are typical symptoms of the HOS. The incidence of cardiac anomalies is 95%, and the cardiac septal defect is observed more. Some researchers reported that the mutation in gene TBX5 causes this disease.[4,5]

Although only cardiac and upper extremity deformities are looked for characteristically in individuals who are diagnosed with the HOS, it is reported that there are specific characteristics in maxillofacial areas.[6] In the studies, wide lower jaw, apparent protruding forehead, narrowing at the temples, and close-set eyes are observed, whereas micrognathia is reported in some patients.[6,7]

Although there are a great number of studies about the HOS in the literature, only few mentions the oral and maxillofacial findings of this syndrome. Allanson conducted the first systemic study about the maxillofacial findings of the HOS in the literature. Allanson performed anthropometric measurements on 25 patients with the HOS and identified facial characteristics about the syndrome.[6] Studies other than Allanson's are not systemic and cover case sharing and oral and maxillofacial findings of individuals with the HOS.[1,7]

How to cite this article: Arslanoglu Z, Bilgiç F, Kale E, Arpağ OF, Altan H. Evaluation of patient with Holt-Oram syndrome in terms of oral and maxillofacial findings. J Pediatr Dent 2016;4:24-8.

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In this study, evaluation of a child with the HOS is made in terms of dental and gingival health, dental plaque, oral and maxillofacial formation, and cephalometric analysis measurements are made for the 1st time in the literature and identified findings are discussed in company with the literature.

CASE REPORT

An eleven-year-old male patient, who was diagnosed with the HOS applied to our clinic with complaints of ache in his decayed teeth. After the anamnesis was taken, we learnt that the patient has a congenital cardiac anomaly, had an associated operation 2 years ago, and had a series of operations about his forearm skeletal structure and finger structure. In the patient's family history, no consanguineous marriage was observed, and it was identified after a genetic test performed using the amniotic fluid in the 3rd month of pregnancy that a sibling of the patient had the same syndrome, and the baby was aborted at the own request of the family. Forearm anomaly was found out in the extra oral examination, and the history of mild mental retardation and heart disease was learnt from the patient's history [Figures 1 and 2]. During the preliminary intraoral examination, it was identified that the patient, compared with the normal patients, has a narrower upper dental arch, more posterior cross bite, increased overjet, decreased overbite, and retrognathic and narrower mandibula [Figure 3]. Long facial structure, which is also called adenoid type, was observed associated with the patient's failure to make nasal breathing comfortably. In addition, an increase in the dentoalveolar height, V-shaped narrow and deep palate, small nostrils, and strong buccinator muscles were monitored in the patient [Figures 4 and 5].

The cephalometric film of the patient was taken, and maxillofacial skeleton and malpositioned teeth were analyzed with the VistaDent cephalometric analysis software [Figure 6 and Table 1].

As a result of the analysis, it was found out that vertical direction size has increased, upper and lower front facial height has increased equally, maxillary deep bite exists, mandibular hypoplasia exists, ramus length has

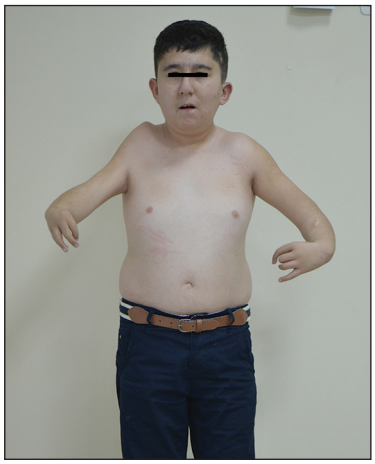

Figure 1: Front view of the patient

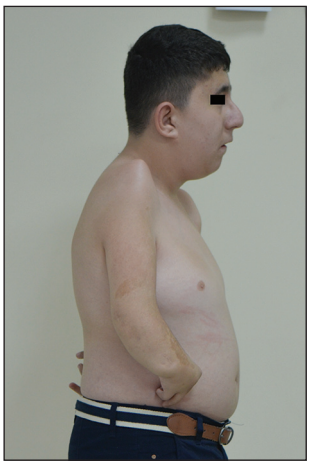

Figure 2: Right side view of the patient

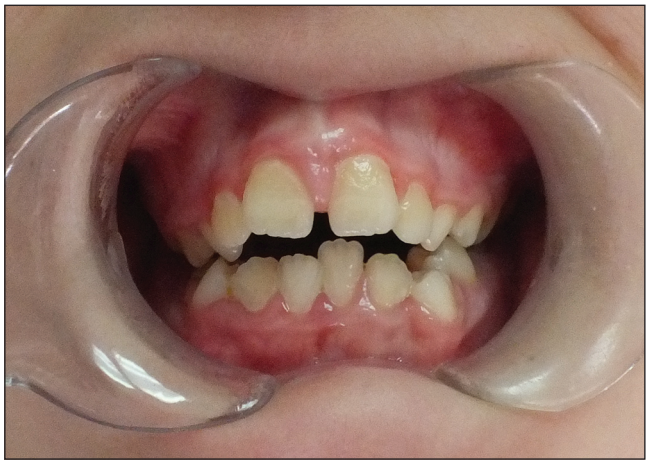

Figure 3: View of increase in the dentoalveolar height

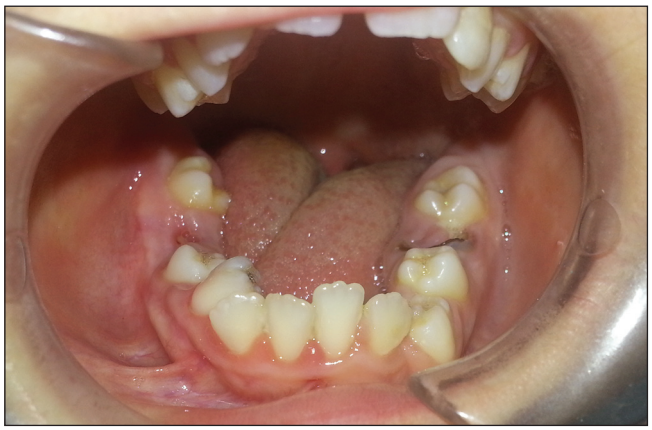

Figure 4: View of retrognathic and narrower mandibula

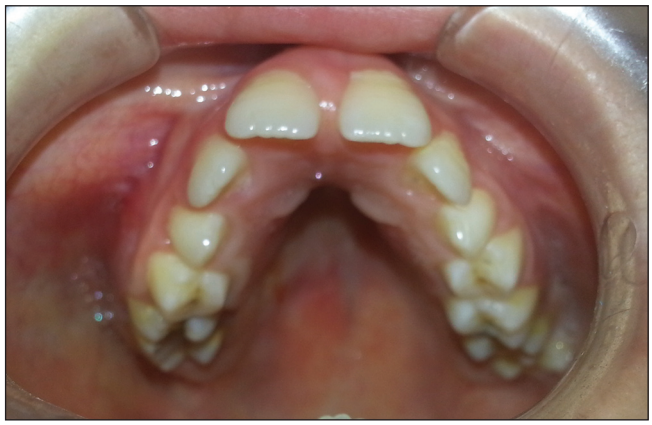

Figure 5: View of V-shaped narrow and deep palate

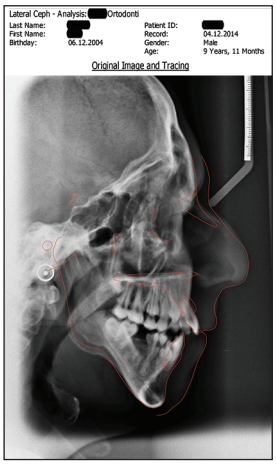

Figure 6: Image of lateral cephalometric film

shortened, mandibular plane has posterior rotation, anterior skull base has shortened, corpus length has shortened, posterior skull base has shortened, maxilla is positioned at the back compared with its skull base, upper incisors are protrusive, lower incisors are retrusive, and convex has a soft tissue profile.

The patient was subjected to periodontal examination, and plaque index (PI) and gingival index (GI) was taken. Accordingly, an intensive plaque formation was observed when evaluated based on the PI. The PI was found to be 1.27 on an average. The GI value was measured 1.44. The values were observed between 2 and 7 mm in probing pocket depth. When the amount of attached gingiva was measured, attached gingiva was observed ranging between 3 and 10 mm. The highest level of attached gingiva was found 10 mm in the upper canine-premolar area. An average amount of attached gingiva in upper incisor areas was 6.25 mm, whereas it was found 6 mm in lower incisor area. In general, the average amount of attached gingiva in the maxilla was 6.28 mm, whereas it was found 4.85 mm in the mandibula.

In the intraoral examination, it was identified that the teeth number 16, 36, and 46 had deep decay, and the fistula was found in soft tissue in line with the root in teeth 36 and 46. The extraction of teeth number 36 and 46 was made, after consultation with the pediatric cardiologist who followed the patient. The composite restoration was applied with glass ionomer base to the tooth number 16. Scaling and curettage procedures were carried out. The patient and his relatives were informed about the oral hygiene.

DISCUSSION AND CONCLUSION

The HOS is defined as a genetic disease with autosomal dominant inheritance with accompanying preaxial radial anomalies in upper extremities and congenital heart disease.[1,2] Skeletal deformities cover upper extremities only, and bilateral thumb anomalies are typical symptoms of the HOS. The incidence of cardiac anomalies is 95%, and the cardiac septal defect is observed more.[4,5] Findings of our patient comply with the definition of the syndrome, and it was reported by the orthopedics and cardiology clinics that he had a series of operations and is still followed by these clinics.

Although the knowledge is that there is no findings in the literature about the "refractive error in HOS,"[8] myopic visual impairment and mild strabismus were identified in our case. In his study, Allanson states that the finding of close-set eyes is a specific feature of individuals with the HOS.[6] Our case is also compatible with this data.

In his article, Turleau reported mental retardation in the patient with HOS.[9] No data are found about mental status in individuals with HOS in the literature other than said case. In our case, it was identified that the patient has mild mental retardation and is being followed by the neurology and psychiatry clinics.

In his article, Lichiardopol shared the findings of micrognathia, dental malpositions, and hypoplastic mandible with retrognathia in his patient with the HOS.[1]

Arturo identified hemifacial microsomia on the right side, depressed nasal bridge, and micrognathia in his patient with the HOS.[7]

In this context, Allanson conducted the most comprehensive study. In his study, Allanson made craniofacial measurements in 25 patients and presented some findings about the structure of jaw as well. Allanson mentions that the face is square shaped, the nose is long, and mandibula is wide in the patients with HOS. Our patient is matching with these findings of Allanson.

Regarding the gingiva and surrounding dental tissues, there is no information in the literature about the HOS. In a

Table 1: Vistadent cephalometric analysis table of patient

Lateral Ceph - Analysis: Orthodontics

Last Name: [redacted]
First Name:
Birthday: 06.12.2004

Patient ID: [redacted]
Record: 04.12.2014
Gender: Male
Age: 9 years, 11 months

Variable	*Value*	*Clinical Norm*	*Difference*	*Deviation*
Vertical				
SNGoMe	47°	36°	+11	
Saddle angle	114°	123±5°	-4	
Articular angle	148°	143±6°	0	
Gonial angle	146°	130±7°	+9	
Sum of Angles	407°	396±5°	+6	
Jarabak ratio	60%	62..65%	-2	
ANSMe/Nme	65%	60%	+5	
Maxillary height	61.3°	53.4±3%	+4.9	
Facial Axis Angle	78°	90±3°	-9	
SAr/Ramus	102%	75%	+27	
FMA	44°	16..35°	+9	
Y-axis (FH-GnS)	70°	59.4±3.8°	+6	
Occlusal P. / SN	15.5°	14.0°	+1.5	
Palatal P / SN	0°	11±4°	-7	
Palatal P. / Mand. P.	47°	25±5°	+17	
Occlusal P./ Mand. P.	31°	20±5°	+6	
Sagittal				
SNA	79.5°	80.0..89.0°	-0.5	
SNB	77.5°	75.0..82.0°	0.0	
ANB	2.0°	2.0..4.0°	0.0	
Wits	-1mm	0..4mm	-1	
N-S (Anterior Cranial Base)	61mm	71±3mm	-7	
M. Corpus	59mm	71±5mm	-7	
S-Ar (Posterior Cranial Base)	37mm	32±3mm	+2	
S-L	38mm	51mm	-13	
S-E	15mm	22mm	-7	
Nper-A	-6mm	1.1±2.7mm	-5	
Maxillary Depth	82.5°	90.0±3°	-4.5	
Convexity Angle	3.4°	4.2±5.8°	0.0	
Cond-A	71mm			
Cond-Gn	104mm	97..100mm	+4	
Dental Measurements				
1-SN	106.4°	108.0°	-1.6	
1-NA	27.0°	22.0°	+5	
1-NA	10mm	4mm	+6	
1-FH	111°	90±3°	+18	
IMPA	70°	84..92°	-14	
1-NB	14.9°	25.0°	-10.1	
1-NB	5mm	4mm	+1	
Pog-NB	0mm			
Holdaway ratio	5mm	0..2mm	+3	
I-I	136.2°	130.0..150.0°	0.0	
Overjet	7.1mm	2.5±2.5mm	+2.1	
Overbite	-1.0mm	2.5±2.0mm	-1.5	
Soft Tissue Relations				
Upper Lip-E	0mm	-4mm	+4	
Lower Lip-E	-1mm	-2mm	+1	
Naso-Labial Angle	121°	90..110°	+11	
Soft Tissue Convexity	153°	161°	-8	

healthy individual, average gingival pocket depth is 1.8 mm. In our patient, average pocket depth was found 2.95 mm. This finding can be explained as the development of gingival inflammation and increase in pocket depth based on already high plaque scores of the individual. When our patient was examined in terms of the amount of attached gingiva, it was observed that the patient had attached gingiva above the average. The average level of attached gingiva in a healthy individual is between 1 and 9 mm, whereas it was ranged between 3 and 10 mm in our patient.[10]

Since there is no similar study about the HOS in the literature, cephalometric analysis results of the patient were compared with those of healthy individuals. Accordingly, it was found out that vertical direction size has increased, upper and lower front facial height has increased equally, maxillary deep bite exists, mandibular hypoplasia exists, ramus length has shortened, mandibular plane has posterior rotation, anterior skull base has shortened, corpus length has shortened, posterior skull base has shortened, maxilla is positioned at the back compared with its skull base, upper incisors are protrusive, lower incisors are retrusive, and convex has a soft tissue profile.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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