

# Goldenhar Syndrome- A case report

Renita Lorina Castelino<sup>1</sup>, Subhas Babu<sup>1</sup>, Fazil Ka<sup>2</sup>, Preethi Balan<sup>2</sup>, Anusha Rangare Laxmana<sup>3</sup>,  
Harish Shetty<sup>4</sup>

<sup>1</sup>Department of Oral Medicine and Radiology, Nitte University, AB Shetty Memorial Institute of Dental Sciences

<sup>2</sup>Department of Oral Medicine and Radiology, Sree Anjaneya Institute of Dental Sciences, Kerala University of Health Sciences, Calicut

<sup>3</sup>Department of Oral Medicine and Radiology, Century International Institute Of Dental Sciences And Research Centre, Kerala University of Health Sciences

<sup>4</sup>Department of Ophthalmology, Nitte University, K S Hegde Medical Academy

Yazışma Adresi / Address reprint requests to: Renita Lorina Castelino  
Department of Oral Medicine and Radiology, Nitte University, AB Shetty Memorial Institute of Dental Sciences  
Elektronik posta adresi / E-mail address: renita.castelino@yahoo.com  
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## ÖZET

Goldenhar sendromu- Bir olgu raporu

Goldenhar Sendromu, ilk iki brankiyal yarığın gelişim bozukluğu sonucunda oluşan, gelişimi tamamlanmamış kulak, burun, yumuşak damak, dudak ve mandibula ile karakterize doğumsal bir hastalıktır. Gelişim bozukluklarına neden olan Goldenhar Sendromu, beslenme ve çevresel faktörleri içeren çok bileşenli etyopatolojiye sahip nadir görülen kalıtsal bir hastalıktır. Goldenhar bu durumu ilk defa 1952'de mikroti, hemifasiyal mikrozomi, kulak önünde akrokordon, epibulber dermoid ve vertebral malformasyonlar gibi çeşitli anomalilerin kombinasyonunu gösteren bir hastalık olarak tanımlamıştır. Bu olgu sunumunda hafif Goldehar Sendromu varyantı olan 30 yaşında erkek hastanın durumunu ilgili literatür özeti ile beraber bildirmektediriz.

**Anahtar sözcükler:** Mandibula, sendrom, kulak, akrokordon

## ABSTRACT

Goldenhar syndrome- A case report

Goldenhar syndrome is a birth defect resulting from the maldevelopment of the first two branchial arches with the incomplete development of the ear, nose, soft palate, lip and mandible. Goldenhar syndrome is a rare inherited condition, which has a multifactorial ethiopathology that includes nutritional and environmental factors which can result in developmental disturbances. Goldenhar first described this condition in 1952 as a disease that presents a combination of several anomalies such as microtia, hemifacial microsomia, pre-auricular skin tags, epibulbar dermoids, and vertebral malformations. In this case report we report a case of 30 year old male patient with a mild variant of Goldenhar syndrome with review of literature.

**Key words:** Mandible, syndrome, ear tags

## INTRODUCTION

Goldenhar syndrome, also known as oculoauriculo-vertebral spectrum (OAVS) is a developmental anomaly involving the structures derived from first and second branchial arches (1). It is also called as Franceschetti Goldenhar syndrome, facio auriculo vertebral spectrum (FAV), first and second branchial arch syndrome, or oculoauriculovertebral (OAV) syndrome (2). The syndrome was first recorded by German physician Carl Ferdinand Von Arlt in 1845, but Maurice Goldenhar described its various characteristic features in 1952 and the credit of discovery went to him. In 1963, Gorlin named this syndrome as oculo auriculo vertebral (3,4). Smith (1978) used the term facio-auriculo-vertebral sequence to include both Goldenhar syndrome and hemifacial microsomia (4). The incidence of this syndrome is 1:3500 to 1:5600 with a male to female ratio of 3:2 (2). Most of the cases are sporadic in nature but

autosomal dominance inheritance has also been described. There is no geographic or racial predilection suggested (5). The case reported here is a mild variant of goldenhar syndrome with review of literature.

## CASE REPORT

A 30 year old male patient visited to the department of oral medicine and radiology with the complaint of root pieces in the lower right and left back region of the jaw since six months. There were no associated symptoms reported by the patient. On extra oral examination, the patient had a convex profile with competent lips (Figure 1). The patient had midface protrusion with mandibular hypoplasia and steep mandibular angle on both right and the left side. The patient had strabismus and gave history of diminished eyesight on further questioning (Figure 2). The patient had malformations of the ear with preauricular ear tags present



**Figure 1:** Extra oral photograph of the patient



**Figure 2:** Clinical photograph of eye showing strabismus



**Figure 3:** Bilateral malformations of the ear with pre auricular ear tags

bilaterally (Figure 3). The patient also had speech problems. The patient was the older of 2 siblings and was born to non-consanguineous parents at full term. Antenatal history and family history were non contributory. The general physical examination revealed that the patient was well nourished with normal mental status. Intra orally the patient had a high arched V-shaped palate (Figure 4) and generalised spacing



**Figure 4:** Intra oral photograph showing high arched palate



**Figure 5:** Panoramic radiograph showing hypoplastic condyle, coronoid, ramus and mandibular body on both the sides but more on the right side

of teeth was seen. The buccal and the labial mucosae and the tongue appeared to be normal. There were multiple missing teeth, root stumps with soft and edematous gingiva. As a part of radiographic examination, a panoramic radiograph was made which revealed hypoplastic coronoid and the condylar processes ramus, and mandibular body on both the sides but more on the right side along with multiple missing teeth and root stumps in the maxilla and the mandible. There was also generalised bone loss present (Figure 5). The lateral cephalogram showed a steep mandibular plane (Figure 6). Audiologist consultation was obtained and hearing deficit was elicited on the both the sides. An ophthalmological examination was also conducted which revealed diminished eyesight on both the eyes. Based on the clinical and radiographic features a provisional diagnosis of mild variant of Goldenhar syndrome was made.



**Figure 6:** Lateral cephalogram showing steep mandibular plane

The dental treatment was planned for the patient along with treatment for the deformity, however the patient declined the treatment for the deformity citing financial reasons.

## DISCUSSION

Goldenhar syndrome was named in 1952 when Goldenhar described a number of facial problems that tend to occur together. These include opening of mouth larger and extended towards the ear on one side, under development of the muscles of the face, cheek bones and skin, small or misshapen ears, skin tags or pits usually in front of the ear in line with the mouth opening, mouth problems such as lack of saliva, problems in tongue shape or use, hemispinal vertebrae which are small or not completely formed on one side. Other problems that may occur in some but not all cases are eye defects, deafness, cleft palate, heart, limb or kidney (6). The few of the above features like underdevelopment of muscles of face, cheek bones and skin, misshapen ears, pre auricular skin tags, strabismus and difficulty in speech were present in our case.

The study of this condition is still controversial because the symptoms and the physical features may vary greatly in range and severity from case to case (3).

The syndrome encompasses a range of other features; craniofacial features are highly characteristic and make an oral physician an important portal in the diagnosis of such syndromes (2). The etiology of this condition is not yet fully established, although it is known that it has genetic origin (7). Apart from the triad of features this syndrome may also present heart diseases (5-58% of the patients) (8), hypoplasia of the zygomatic, mandibular and maxillary bones, muscle hypoplasia, anatomical and morphological abnormalities of

the tongue, vertebral anomalies, cleft palate, disturbance of the central nervous system and other visceral anomalies (9,10). Craniofacial anomalies, including mandibular, zygomatic and/or maxillary hypoplasias are found in 50% of patients with Goldenhar syndrome (11,12). The differential diagnosis of Goldenhar syndrome can be challenging because of the variety of clinical signs overlapping other conditions like Collins and Wildervanck syndrome (syndrome cervico oculoacusticum) which can be considered. The treatment of Goldenhar syndrome usually requires a multidisciplinary approach. Severe anomalies of the mandible require reconstruction with bone graft. In cases of microtia or other ear defects, extensive ear reconstruction has to be done within 6-8 years of age (13). If the facial or congenital malformations are severe speech therapy is required (14). The management of the deformity represents a long-term approach and involves multiple procedures performed by a group of specialists. The treatment includes plastic surgery to fix the jaw, cheeks and ears, specialized dental care like distraction osteogenesis along with functional orthodontics has been tried in growing age, hearing aids, speech therapy, physiotherapy may be required and prevention of the psychosocial aspects of the malformation. The timing of the primary and secondary reconstructions plays an important role. The treatment is usually based on the pathogenetic, psychosocial, and structural problems. The primary reconstruction typically consists of a cleft repair, corrections of colobomas and ear deformities, and extirpation of the dermoids and preauricular tags (2,4,6). The prognosis of the condition is usually good if systemic complications are absent. However, our patient agreed only for dental correction and was not willing for other treatment owing to financial problems.

Goldenhar syndrome is characterized by bony abnormalities of the face and jaw with various clinical manifestations. The early recognition and prompt treatment by a team of specialists helps in reducing the esthetic and functional disability associated with the syndrome. Counselling of families with sporadic occurrence should be done as the syndrome is found to have a possible genetic etiology. The purpose of the paper was to highlight the various features of Goldenhar syndrome to aid in the diagnosis of this syndrome which has various overlapping clinical features with other conditions like hemifacial microsomia.

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