Perspective

# A Brief Discussion on Goldenhar Syndrome

**Sreekanth P Kunjumon***

*Department of Oral Medicine and Radiology, Rajiv Gandhi University of Health Science, Bangalore, Karnataka, India*

**Corresponding author:**
Sreekanth P Kunjumon,
Department of Oral Medicine and Radiology,
Rajiv Gandhi University of Health Science,
Bangalore, Karnataka, India,
E-mail: sreekanthpkunjumon@gmail.com
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## Abstract

Oculo-auriculo-vertebral distortion and hemi facial macrosomia are additional names for Goldenhar Syndrome (GS). The oral cavity, eyes, ear, and vertebrae are the biggest areas affected by this disease. It is a very uncommon genetic anomaly. With a short outline of its etiology, clinical and radiographic features, differential diagnosis, and management, we describe a case of GS here. Goldenhar syndrome is a rare condition which is characterized by a multitude of anomalies involving craniofacial structures, vertebrae, internal organs and usually occurs unilaterally. The etiology of this syndrome is unclear since it varies genetically and is linked to a plethora of reasons. Common clinical manifestations include limbal dermoids, preauricular skin tags and strabismus. It is associated with anomalous development of the irst branchial arch and second branchial arch. The main sign and symptoms are facial asymmetry (one side of the face is different from the other), a partially formed ear (microtia) or totally absent ear (anotia), noncancerous (benign) growths of the eye (ocular dermoid cysts), and spinal abnormalities. Goldenhar disease may also affect the heart, lungs, kidneys, and central nervous system. It is due to problems that occur when the fetus is forming within the womb of the mother, in structures known as the irst and second brachial arch. These structures will develop to form the neck and the head. The cause is still unknown. Goldenhar syndrome is part of a group of conditions known as craniofacial microsomia. It is not known whether the conditions included in the group really are different conditions or part of the same problem with different degrees of severity.

**Keywords:** Goldenhar syndrome; Branchial cleft; Epibulbar dermoid; Hemifacial macrosomia; Blind fistulas

## Introduction

The term "Goldenhar Syndrome" (GS) is also used to refer to OAV abnormalities. It is regarded as a subtype of hemifacial macrosomia [1]. Carl Ferdinand von Arlt made the first observations of and records of GS. Maurice Goldenhar was the first to thoroughly characterize the syndrome, leading to the naming of the illness as GS. Hemifacial macrosomia was described as a disease that mainly affects the development of the mandible, the oral cavity, and the ears in the 1960's. Due to extra vertebral anomalies and epibulbar dermoids, GS was regarded as a subtype of this complex. After Gorlin et al., included the vertebral anomalies in 1963, the term OAV dysplasia was proposed. It's believed that GS is an uncommon congenital anomaly. Aural fistula, accessory auricular appendages, and epibulbar dermoid are the three anomalies that make up the condition. A malformation complex with different degrees of severity is GS. It includes the primordia of the temporal bone, the first pharyngeal pouch, the first branchial cleft, and the structures arising from the first and second branchial arches. The following are some examples of the distinctive clinical findings: Colobomas of the upper eyelid, iris, choroidea, and retina as well as other eye abnormalities like microphthalmia and exophthalmia are examples of epibulbar dermoid or lipodermoid, which are typically bilateral. Blind fistulas, preauricular skin tags, microtia, and other ear abnormalities of the middle and inner ear hypoplasia of the zygomatic region, hypoplasia of the brow, mandibular and maxillary hypoplasia, and unilateral facial hypoplasia one sided macrostomia abnormalities of the vertebral column, such as synostosis and bifid spine [2,3].



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## Description

OAV dysplasia and hemifacial macrosomia are additional names for GS. Clinical manifestations of this disease can take many different forms. Anomalies of the craniofacial, vertebral, cardiac, renal, and central nerve systems are among the clinical symptoms. An uncommon inherited condition, GS is. Its multifactorial etiopathology can lead to disturbances in blast genesis due to dietary and environmental variables. Unfortunately, the precise aetiology is not well known. Many different theories have been put forth. Gorlin and Pindborg proposed that the syndrome results from an abnormal embryological process that affects myoblasts and, in turn, the branchial and vertebral systems in 1964. According to Baum and Feingold, GS may be a sporadic event that happens early in embryogenesis. Reduced penetrance, somatic mosaicism, or epigenetic modification could all be to blame for this there have also been reports of cases involving families with a history of consanguineous unions. According to reports, there are between 1:35,000 and 1:56,000 cases of GS per 100,000 people, with a 3:2 male to female ratio. It frequently has right-side predominance and is unilateral [4,5].

## Conclusion

The patient's daily activities and social life may be impacted by severe GS. Later in life, difficulties can be avoided with early identification. These patients will be more likely to experience psychosocial problems. Various aesthetic operations may be financially supported by social workers or particular groups. Families of such patients may receive counselling and moral assistance to help them accept their family and relatives who have such conditions.

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