Diagnostic Imageology of Goldenhar Syndrome: Report of a Rare Case

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Abstract

Goldenhar syndrome is a rare disorder that normally affects just one side and is distinguished by a variety of anomalies in internal organs, vertebrae, and craniofacial tissues. Although this sickness varies genetically and has been linked to a variety of factors, its etiology is unknown. We describe a case of hemifacial microsomia linked with Goldenhar syndrome that was clinically and radiographically investigated using cone-beam computed tomography. Several classical indications of the condition were present in the patient along with few uncommon ones. The many facets of this uncommon disease have been covered, with a focus on early detection and a multidisciplinary approach to treatment.

Keywords: *Cone-beam computed tomography, Goldenhar syndrome, hemifacial macrosomia*

Introduction

Oculo-auriculo-vertebral syndrome (OAVS), commonly known as Goldenhar syndrome, is a rare congenital disorder caused by abnormalities in the first and second branchial arches. In 1952, Dr. Maurice Goldenhar gave the first explanation of it as a congenital condition that primarily affects the face, eyes, and ears and is characterized by a constellation of malformations. Dr. Maurice Goldenhar, a famous Swiss ophthalmologist, first defined its clinical symptoms and designated the malformation complex as Goldenhar syndrome.[1] This condition is also known as OAVS, first arch syndrome, first and second branchial arch syndrome, Goldenhar-Gorlin Syndrome, lateral facial dysplasia, unilateral craniofacial microsomia, otomandibular dysostosis, unilateral intrauterine facial necrosis, and auriculo-branchiogenic dysplasia. The incidence of Goldenhar syndrome has been reported to be 1:35,000–1:56,000 with a male-to-female ratio of 3:2.[2] There is no agreement on the incidence of Goldenhar syndrome in the literature.[3] Reports vary between 1:3000–5000 and 1:25,000–40,000. A recent population-based data linkage study of the epidemiology of rare craniofacial abnormalities in Western Australia found a birth prevalence of 15.8 (11.6–20.9) per 100,000 live and stillbirths in the decade from 2000 to 2010. The disease is considered to be sporadic and its etiology is not fully understood; however, positive family histories have been reported suggesting autosomal dominant or recessive inheritance. Abnormalities of chromosomes, neural crest cells, and environmental factors during pregnancy such as ingestion of drugs, such as cocaine, thalidomide, retinoic acid, and intake of alcohol by the mother were also related to the development of the disease. Maternal diabetes has also been suggested as an etiologic factor.[4] The right side is more often afflicted, and abnormalities are unilateral in 85% of cases and bilateral in 10% to 33% of instances.[5] In 85% of cases, the illness is typically unilateral, and in a 3:2 ratio, the right side is more frequently damaged than the left. Our case fits into the more uncommon category as only the left side of the face was unilaterally affected. We are reporting a case of Goldenhar syndrome in a 33-year-old male patient.

Case Report

A 33-year-old male patient reported to the Department of Oral Medicine and Radiology, Subbaiah Institute of Dental Sciences, Shivamogga, with the chief complaint of difficulty in mouth opening since childhood. On eliciting the history, there was no history of trauma to the head-and-neck region or maternal exposure to teratogenic agents. No signs of mental retardation or impairment of cognitive

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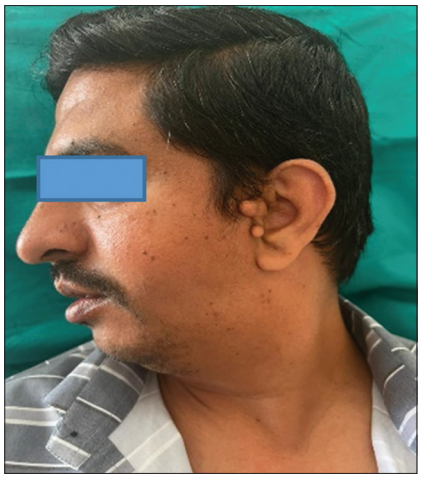

Figure 1: Profile view showing gross facial asymmetry with hypoplasia of the left mandible with accessory ear tags in the preauricular region on the left side

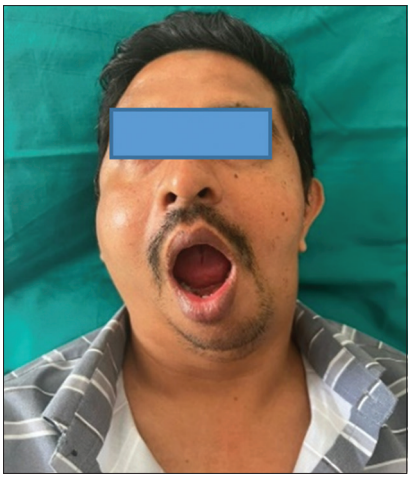

Figure 2: Deviation of mandible toward affected side

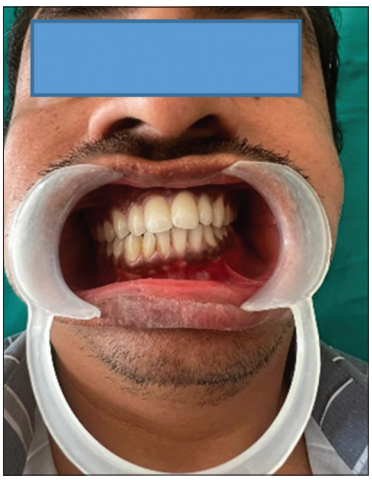

Figure 3: Slanting of occlusal plane

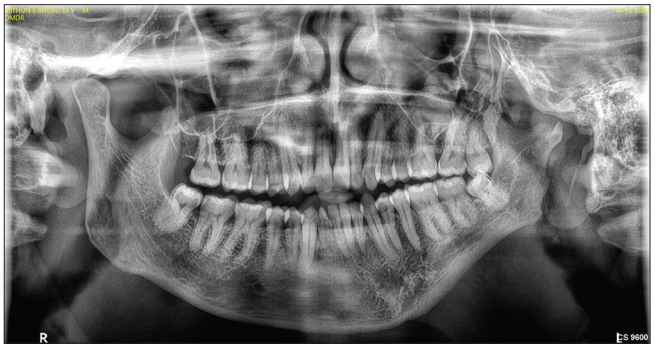

Figure 4: Orthopantomogram showing short ramal height and width, small condylar head, reduced depth of sigmoid notch, reduced height of body of mandible, and prominent antegonial notch on the left side

function were seen. Medical history was insignificant with no history of congenital heart diseases. The patient's family history was significant with similar complaints with his brother. General physical examination revealed facial asymmetry was present with a deviation of the angle of the mouth toward the left side with loss of malar prominence [Figures 1 and 2]. Distal aspect of the left eye demonstrated epibulbar dermoids with preauricular tags and raised the level of the orbital floor on the left side. No disturbance of the central nervous system, cleft lip/cleft palate, developmental dental disturbances, mental retardation, vertebral and congenital heart anomalies, growth abnormalities, pulmonary abnormalities, and labyrinthine, tracheoesophageal, and renal and genitourinary abnormalities were elicited. On intraoral examination, deviation of the mid-line toward the left side was noted with tilting of the occlusal plane and lower anterior crowding [Figure 3]. Radiographic investigations were carried out using orthopantomogram (OPG) and cone-beam computed tomography (CBCT) scans. OPG revealed mandibular deficiency on the left side, with steeping of the gonial angle and reduction in the size of the left mandibular ramus region with hypoplasia of the condylar and coronoid region. [Figure 4]. CBCT scan measurements were in agreement with OPG findings [Figures 5-8]. Correlating the history with clinical and radiographic findings, a provisional diagnosis of Goldenhar syndrome was given. The differential diagnoses considered were congenital unilateral Temperomandibular joint (TMJ) ankyloses of the left side, Treacher Collins syndrome (TCS), Pierre Robin syndrome, Parry–Romberg syndrome, and hypoplasia of the left mandibular condyle.

Discussion

The first description of Goldenhar syndrome, which includes a classical triad of epibulbar dermoids, fistulas, and preauricular appendages, was made in 1952 by Swiss ophthalmologist Maurice Goldenhar. Gorlin *et al*. proposed the name OAV dysplasia for this disorder and included vertebral deformities as symptoms of the syndrome.[6] Facio-auriculo-vertebral sequence, as used by Smith (1978), encompasses both hemifacial microsomia and the Goldenhar syndrome.[7] It is a sporadic genetic syndrome with a very low prevalence and an unknown

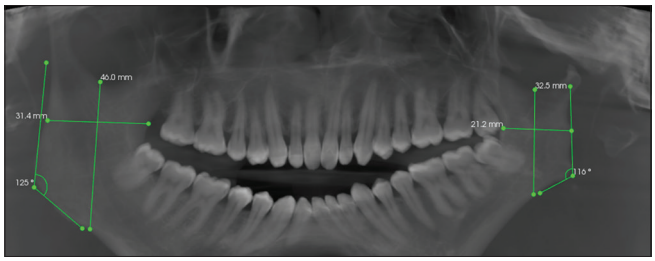

Figure 5: Cone-beam computed tomographic reconstructed panoramic image showing reduced ramal height and width and reduced height of body of mandible

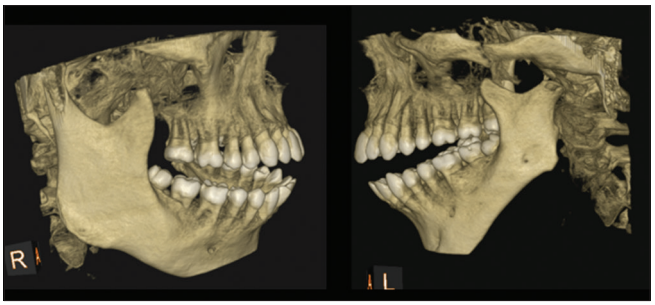

Figure 6: Three-dimensional reconstructed cone-beam computed tomography view showing hypoplasia of the left condyle in comparison to the right

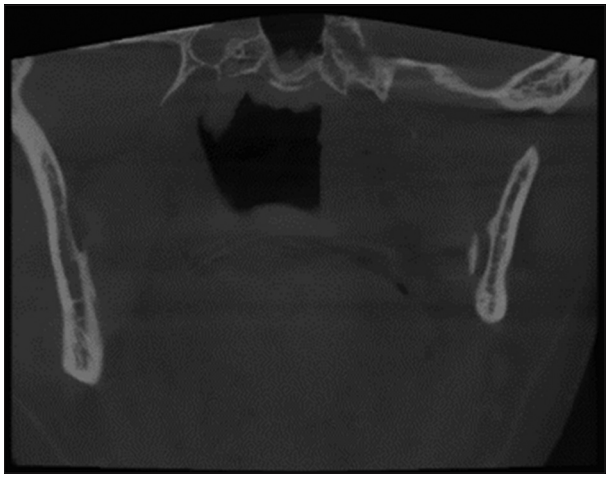

Figure 7: Coronal section

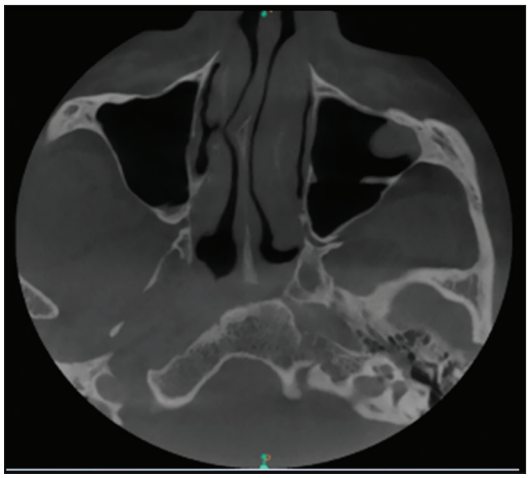

Figure 8: Axial section

cause. Positive family histories that supported autosomal dominant or recessive inheritance have occasionally been found. According to some scientists, multifactorial inheritance is the consequence of several genes interacting, maybe in addition to environmental variables. Alterations in mesodermal migration and aberrant embryonic vascular supply may result in brachial arch and spinal system defects. Some other etiologic factors include maternal vasoactive medication use (especially in conjunction with smoking) in the first 10 weeks of gestation, primidone, retinoic acid, and thalidomide embryopathy and maternal (preexisting or gestational) diabetic embryopathy. However, in this case, the patient did not give any such known maternal history except that his brother had similar complaints giving the implication of a genetic correlation. The hallmark of Goldenhar syndrome, as demonstrated in this case history, is the coexistence of ipsilateral facial underdevelopment and external ear abnormalities. In 85% of cases, the illness is typically unilateral, and in a 3:2 ratio, the right side is more frequently damaged than the left.[7] Our case fits into the more uncommon category as only the left side of the face was unilaterally affected. Goldenhar syndrome is characterized by a triad of accessory tragi, mandibular hypoplasia, and ocular dermoids.

Principal deformities of the Goldenhar syndrome are often combined with various malformations, such as:

i. Cleft lip and/or palate tongue cleft, unilateral tongue hypoplasia, and parotid gland aplasia
ii. Rib anomalies and anomalies of the extremities
iii. Congenital heart disease (ventricular septal defects), anomalies of the urogenital and gastrointestinal system (ectopic kidneys, uretropelvic junction obstruction, and imperforate anus), anomalies of the central nervous system (occipital encephalocele), and anomalies of the larynx and lungs (fistula and esophageal atresia).

In the present case, a triad of accessory tragi, left mandibular hypoplasia, and ocular dermoid were observed.

Although clinical diagnosis is the main focus, radiographic examinations help to support it. Ultrasound scans can be used to diagnose this condition at a much early stage of pregnancy with a high degree of accuracy. However, since no specific genes have been linked to this disease, prenatal deoxyribonucleic acid testing cannot detect it. Due to the wide scope of the differential diagnosis, it must be separated from other syndromes such as Townes–Brocks syndrome, Nager syndrome, and TCS. Unlike TCS, which results in facial asymmetry, involvement in Goldenhar syndrome often occurs unilaterally. TCS is specifically connected to the TCOF1 gene mutation on human chromosome 5q31–34, which aids in the precise diagnosis.

The treatment for this syndrome varies with age and systemic associations. Rib grafts can be used for reconstruction in patients with mandibular hypoplasia, and a bone distraction device can be used to stretch the undeveloped maxilla.[8] The structural problems with the ears and eyes can be treated through plastic surgery. Cleft lip and palate can be treated surgically, and after the jaw has finished growing, the correction can be made with orthodontics. Airway anomalies and inadequate jaw growth in the patient might cause severe obstructive sleep apnea, which can result in a restricted diet and malnutrition. Due to malocclusion, restricted mouth opening, and intellectual disability, Goldenhar syndrome patients have difficulty maintaining good oral hygiene, which makes them more susceptible to dental caries and gingivitis. Hence, management calls for a multidisciplinary strategy, although in straightforward situations, like the one in our case study, esthetic concerns are the primary focus. The prognosis is cautious in situations with systemic involvement, but favorable in other cases devoid of any systemic linkages.

Conclusion

Even though the diagnosis is mainly clinical, radiographic investigations play an important role in supporting the diagnosis. This case report highlights the role of an oral and maxillofacial radiologist in the diagnosis of an uncomplicated case of Goldenhar syndrome where the patient was unaware of the condition. The role of an oral medicine radiologist is significant to ensure optimum oral health care for such syndromic patients since they have complex unmet dental needs. The medical and dental communities should work together to identify and treat this problem as soon as possible to decrease the emotional, physical, and financial strain on those who are affected.

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.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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