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# An oculo-auriculo-vertebral spectrum: A radiological case report and literature review of Goldenhar syndrome with hemifacial microsomia

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**Abstract**
Goldenhar syndrome, or the oculo-auriculo-vertebral syndrome, is an uncommon congenital condition associated with a broad range of anomalies involving the craniofacial structures, typically unilateral, and the vertebrae, but may also show systemic involvement. Many times, the syndrome may be diagnosed late or misdiagnosed due to overlapping features with other congenital syndromes, commonly the Treacher-Collins syndrome.
This case report highlights a 6-year-old child who presented with classical features of hemifacial microsomia with vertebral anomalies and summarizes the features seen in Goldenhar syndrome to achieve a prompt radiological diagnosis.

**Keywords:** Oculoauriculovertebral, Goldenhar, microsomia, preauricular, microtia

## Introduction
The most severe form of hemifacial microsomia, “Goldenhar syndrome” also referred to as oculoauriculovertebral dysplasia, is a complex congenital disorder with various associations including maxillomandibular hypoplasia, ear deformities, dermoid, ocular, and vertebral anomalies. Other various eponymous names for this condition are First and second branchial arch syndrome, Oto-mandibular dysostosis, Facio-auricular dysplasia, Auriculo-branchiogenic dysplasia, Unilateral intrauterine facial necrosis, Unilateral craniofacial microsomia, and Lateral facial dysplasia [1]. Goldenhar syndrome was first described by the Swiss ophthalmologist Maurice Goldenhar in 1952 as an oculo-auricular dysplasia. In 1963, Gorlin and colleagues incorporated vertebral anomalies to characterize the syndrome, proposing the name “oculoauriculovertebral dysplasia” [2]. The incidence of Goldenhar syndrome has been reported to be 1: 3500 to 1: 5600 with a male-to-female ratio of 3: 2 [3]. This syndrome may range from mild to severe, with unilateral involvement being more common, however, cases of bilateral involvement have been documented, with severe expression of one side and the right being the most common side to be involved [1].
We report a case of Goldenhar syndrome, discussing its clinical and imaging features.

## Case
A 6-year-old male child was referred to the Department of Plastic and Reconstructive Surgery with complaints of microphthalmos in the right eye and nostril deformity. On external examination, the patient has a nasal deformity in the form of an atretic right nostril, however, the nasal pit is present. External ear malformation was noted in the form of right microtia and the presence of a preauricular tag. Ophthalmic examination revealed right incomplete cryptophthalmos with skin continuous over the right eyeball, absence of palpebral fissure with presence of eyelashes (Figure 1). Intra-orally, the child showed a small or absent uvula with an asymmetrical soft palate. The child is second by birth order, born out of a non-consanguineous marriage with the age of the mother being 20 years at the time of birth. His antenatal history was uneventful with no history of trauma or maternal exposure to any teratogenic drugs. The child was born as a full-term normal vaginal delivery with an immediate cry after birth and no NICU admission, however, the child developed difficulty in latching due to cleft lip for which he was operated at 6 months of age. The patient had no family history of known birth defects, and both parents had normal craniofacial development. Overall, the features are suggestive of an oculo-auricular syndrome.
High-Resolution Computed Tomography (HRCT) of the paranasal sinuses was performed

using axial sections with thin slice thickness (typically 0.5 mm) and high spatial resolution algorithms on a Toshiba Aquilon Prime 160-slice CT machine. The scan coverage extended superiorly from the vertex of the skull to inferiorly up to the level of inferior vena cava (IVC) formation, thereby including the cervical spine and adjacent soft tissues. Coronal and sagittal reformats were obtained in bone and soft tissue windows.

The computed tomography revealed a small deformed right pinna with pre-auricular tag and moderate to severe stenosis of the osseous part of the right external auditory canal, suggestive of type 2 microtia (Figures 2 & 3). There was complete aplasia of the right-sided maxillary, ethmoid, and frontal sinuses, as well as the sphenoid sinus (Figure 4). The nasal septum was deviated to the right with non-visualization of all the right-sided nasal turbinates, causing significant narrowing of the right nasal cavity (Figure 4). The right nasal ala appeared deformed with stenosis of the right anterior nare (Figure 4). The right nasolacrimal duct was absent (Figure 4).

The right orbit was hypoplastic with the absence of the right globe, which was replaced by an ill-defined soft tissue with a tiny air focus within (Figure 5). A well-defined hypodense ovoid lesion was noted along the medial wall of the right orbit, inferomedial to the above-mentioned soft tissue, with resultant mild scalloping of the medial wall of the right orbit, suggestive of a dacrocystocele (Figure 5). The right optic nerve was not seen, and the same-sided optic canal was hypoplastic (Figure 5). The right half of the hard plate and also the right-sided styloid process were not seen (Figure 6). Dental malocclusion with dysmorphic teeth were seen predominantly on the right side (Figure 6). This collection of imaging findings was indicative of Hemifacial microsomia.

Vertebral anomalies were seen, such as fusion of the right laminae of C6 and C7 vertebral bodies and non-fusion of the posterior arch of C6 vertebra (Figure 7).

Thus, radiologically, oculo-auriculo-vertebral syndrome was determined, and keeping in mind the triad of congenital anomalies, i.e., preauricular tag/ microtia, hemifacial microsomia, and vertebral anomalies together, a diagnosis of Goldenhar syndrome was made.

Informed consent was obtained from the mother of the patient involved in the study regarding potential publication in a scientific journal.

## Figures


**Fig 1:** Clinical photograph of a child demonstrating unilateral craniofacial anomalies. The inset at the lower left corner shows the side profile of the child to highlight microtia and preauricular tag

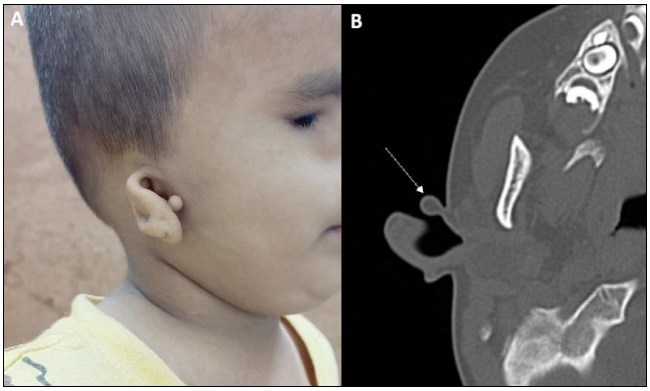


**Fig 2:** Image A is the side profile of the child demonstrating external ear anomalies, and Image B is the axial section computed tomography image in bone window setting at the level of the external ear, showing preauricular tag (marked by white line arrow)

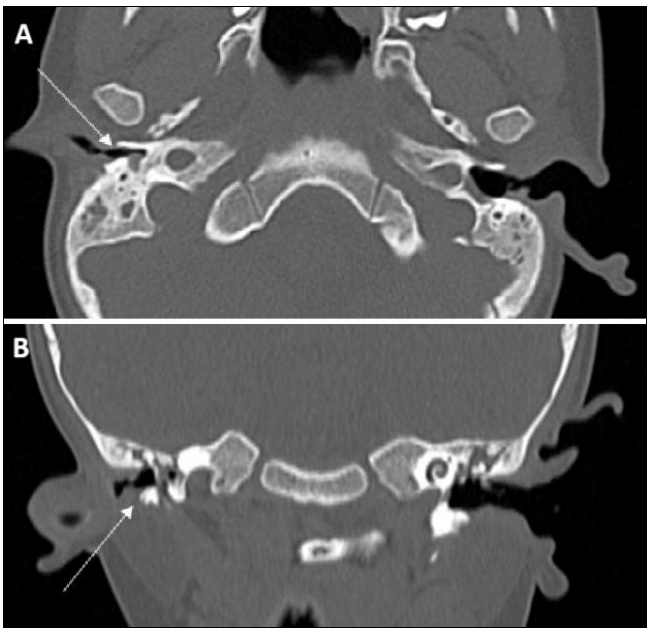


**Fig 3:** Axial section computed tomography (Image A) and coronal section MPR reconstructed (Image B) images in bone window setting show moderate to severe stenosis of the osseous part of the right external auditory canal (marked by white line arrow)

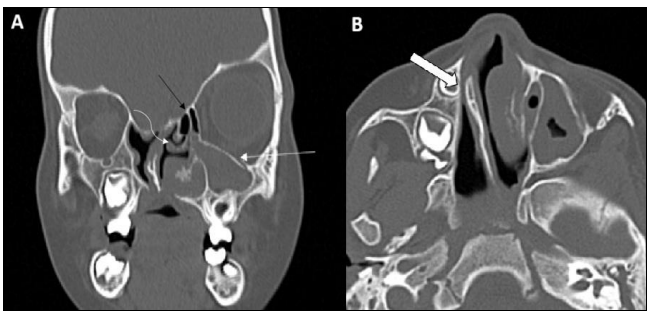


**Fig 4:** Computed tomography coronal section MPR reconstructed image in bone window setting (Image A) shows normal left-sided maxillary (marked by white line arrow) and ethmoid sinuses (marked by black line arrow), and normal nasal turbinates on the left side (marked by white curved arrow), however the sinuses and nasal turbinates are absent on right side. Axial section image (Image B) shows a normal anterior nare on the left side, however, atretic on the right side (marked by white block arrow). Also in image B, the right-sided nasolacrimal duct is seen, which is absent on the left side


**Fig 5:** Computed tomography coronal section MPR reconstructed image in bone window setting (Image A) shows hypoplastic right orbit (marked by curved white arrow). Images B, C, and D are axial sections in soft tissue window where the right globe is not visible and is replaced by an ill-defined soft tissue (marked by white line arrow). Image C shows a dacryocystocele in the medial wall of the right orbit (marked by white block arrow). In Image D, the normal optic nerve is seen on the left side (marked by white star, which is not visible on the right side. Image E is a coronal section MPR reconstructed image in bone window setting showing hypoplastic right optic canal (marked by black line arrow) as compared to the left optic canal.

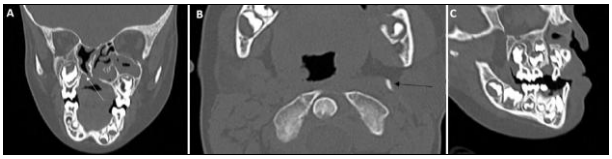


**Fig 6:** Computed tomography coronal section MPR reconstructed image in bone window setting (Image A) shows a normal hard plate on the left side; however, not seen on the right side (marked by white line arrow). Image B is an axial section image in soft tissue window, showing the normal left styloid process (marked by black line arrow), but not seen on the right side. Image C is a curved MPR reconstructed image showing dental malocclusion with dysmorphic teeth

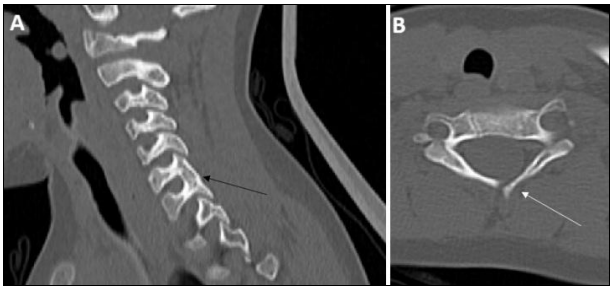


**Fig 7:** Images A and B are sagittal section MPR reconstructed image and axial section image. Image A shows fusion of right laminae of C6 and C7 vertebral bodies (marked by white line arrow). Image B shows non-fusion of the posterior arch of C6 vertebra (marked by black line arrow).

## Discussion

Goldenhar syndrome, also referred to as oculo-auriculo-vertebral (OAV) dysplasia, is a complex and poorly understood developmental disorder of unknown cause, characterized by variable involvement of the eyes, ears, face, spine, and other structures in each individual [4]. This condition was first noted by German physician Carl Ferdinand von Arlt in 1845, but it was not identified as a distinct syndrome until 1952, when Dr. Maurice Goldenhar described it as a disorder characterized by a combination of anomalies such as dermal epibulbar tumors, preauricular appendages, and mandibular hypoplasia [5]. In 1963, the term 'oculo-auriculo-vertebral (OAV) syndrome' was coined by Gorlin *et al*. due to the identification of associated vertebral anomalies, leading to its alternate name, Goldenhar-Gorlin syndrome [5].

The estimated incidence of Goldenhar syndrome ranges from 1 in 3,500 to 5,600 live births, with a male preponderance showing a male-to-female ratio of approximately 3:2 [4]. Several chromosomal defects have been associated with the syndrome, including del(5p), del(6q), trisomy 7 mosaicism, trisomy 9 mosaicism, trisomy 18, recombinant chromosome 18, del(8q), del(18q), ring 21 chromosome, trisomy 22, del(22q), dup(22q), 47 XXY, 49 XXXXX, and 49 XXXXY [6]. However, normal karyotyping has also been reported in some cases, making genetic karyotyping an unreliable tool for definitive diagnosis.

According to the literature, the term 'hemifacial microsomia' is used when the malformations predominantly affect the jaw, mouth, and ears with unilateral presentation [5]. When vertebral and/or ocular abnormalities are also present, the condition is more specifically referred to as Goldenhar syndrome [5]. Our patient showed malformations involving the mouth, ears, eyes, and vertebral column, leading to the diagnosis of Goldenhar syndrome.

There is a constellation of various archetypal features seen in cases of Goldenhar syndrome. Peculiar ocular anomalies include epibulbar dermoid or lipodermoid tumors, eyelid colobomas, microphthalmia, and anophthalmia, with other less specific findings being cataract, blepharophimosis, astigmatism, and anti-mongoloid obliquity of palpebral fissures [2, 6]. Our patient presented with skin covering the nasal side of the unilateral eyeball with loss of palpebral fissure and presence of eyelashes, together suggestive of incomplete cryptophthalmos. Also, the patient showed a unilateral hypoplastic orbit with absence of globe and presence of a dacryocystocele. Along with this, the same-sided optic canal was hypoplastic with an absent optic nerve. The common associated auricular abnormalities are anotia, microtia, preauricular tags, external ear malformations like aplasias, dysplasias, and atresias of the external meatus [2, 6]. Our patient had unilateral microtia with preauricular tag and moderate-to-severe stenosis of the external auditory canal. The patient has not suffered any hearing disturbances or facial nerve dysfunction. Facial abnormalities are usually unilateral and comprise unilateral facial hypoplasia, cleft lip, cleft palate, macrostomia, high-arched palate, dental malformations, facial bone hypoplasia, etc [5]. Of these, our patient had a cleft lip, for which he was operated on earlier, an absent hard palate and styloid process with features of dental malformations, all on one side. Supernumerary teeth, gingival hypertrophy, delayed tooth development, and enamel and dentin malformations are a few dental abnormalities seen [6]. Scoliosis, fused vertebrae, hemivertebrae or butterfly vertebrae, and short stature are a few of the many vertebral abnormalities that form the third component of this syndrome [2].

In addition to these primary abnormalities, this syndrome is associated with other systemic malformations, including the cardiovascular system (congenital heart diseases like atrial and ventricular septal defects, transposition of great vessels, tetralogy of Fallot, as well as anomalies of pulmonary arteries), the nervous system (encephalocele, Arnold Chiari malformation, falx calcification, Corpus callosum lipoma or

agenesis associated with cognitive decline), the gastrointestinal system (Umbilical hernia, imperforate anus, accessory spleen), the urogenital system (Renal hypoplasia or agenesis, ectopic kidney or pelvi-ureteric junction obstruction, hypoplastic vagina) and the pulmonary system (pulmonary atresia, trachea-esophageal fistula, esophageal atresia, bronchogenic cyst) [3, 4, 5, 6]. In 2005, Tasse *et al*. reported that the presence of eye anomalies or orofacial clefts like cleft lip or cleft palate in patients with Goldenhar syndrome may predict an increased risk of brain malformations [7].

Various classification systems have been proposed to clinically categorize patients with Goldenhar syndrome or oculo-auriculo-vertebral spectrum and assist in prognosis, with the recently introduced system by Tasse *et al*. being straightforward and clinically practical [7]. Using this classification system, our patient m*et al*l three minimal diagnostic criteria-unilateral microtia with preauricular tag, hemifacial microsomia, and vertebral anomalies, placing the patient in group 3u.

There is a wide range of differential diagnoses for Goldenhar syndrome, including various congenital conditions like CHARGE syndrome, Parry Romberg syndrome, Townes-Brocks syndrome, Nager syndrome, and Treacher-Collins syndrome with Treacher-Collins being the closest mimic; however, facial involvement in Treacher-Collins is typically bilateral, whereas it is usually unilateral in Goldenhar syndrome [4].

The treatment for this syndrome depends on the patient's age and associated systemic abnormalities, however, it requires a multidisciplinary approach. Several surgical methods are available for correcting hemifacial microsomia, including traditional reconstruction with bone grafts and distraction techniques followed by later orthodontic treatment, while structural abnormalities of the eyes and ears are addressed through reconstructive surgery [5]. In severe cases of Goldenhar syndrome, multiple aspects of the patient's livelihood may be affected. For instance, patients may experience severe obstructive sleep apnea due to airway abnormalities, jaw issues that restrict diet and cause malnutrition, and eyelid defects that can result in vision problems [3]. Such cases require immediate intervention from birth. Generally, without any systemic associations, the prognosis is favourable, however, one should be vigilant about the development of signs and symptoms indicating any systemic involvement [8].

Although primarily the diagnosis is clinical, radiological investigations are useful in supporting and confirming the clinical findings [2]. Ultrasound can enable prenatal diagnosis with good accuracy by identifying obvious structural abnormalities. In 1996, De Catte *et al*. demonstrated that Goldenhar syndrome could be detected sonographically in a foetus at 15 weeks of gestation by identifying a maxillary cleft alongside unilateral microphthalmia [9]. As no specific genes have been associated with this syndrome, prenatal DNA testing is not applicable for its diagnosis [2].

## Conclusion

Goldenhar syndrome is a complex congenital developmental disorder linked to abnormalities in multiple organ systems, particularly involving the eyes, ears, face, and vertebrae. This case report emphasizes the crucial role of the radiologist in diagnosing Goldenhar syndrome. There is often a diagnostic delay in the absence of clear characteristic features and a lack of understanding. Therefore, this article sums up the key features of patients with Goldenhar syndrome, which will help in enabling early planning and implementation of interdisciplinary interventions. Although Goldenhar syndrome cannot be completely cured, treatment and management of the disorder's specific symptoms and features are often effective.

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