

Case Report

Goldenhar syndrome in an infant with possible familial association: a case report

Pranjali Varshney*, Prithi Inamdar, Ankitha Ponathil

Department of Pediatrics, Dr. D. Y. Patil Hospital, Nerul, Navi Mumbai, Maharashtra, India

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*Correspondence:

Dr. Pranjali Varshney,

E-mail: pranjaliwarshney.me@gmail.com

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ABSTRACT

Goldenhar syndrome, is a rare congenital disorder characterized by craniofacial anomalies, auricular malformations, and vertebral defects. We report the case of an 8-month-old female presenting with an ocular mass, which was confirmed to be an epibulbar dermoid. Further evaluation revealed bilateral anotia/microtia and facial asymmetry. Systemic evaluation did not demonstrate vertebral, cardiac, or renal anomalies. Notably, the mother exhibited mild craniofacial anomalies, suggesting a possible familial association. This case highlights the importance of early diagnosis, thorough systemic evaluation, parental counselling, and consideration of recurrence risk and prenatal detection in future pregnancies.

Keywords: Goldenhar syndrome, Oculo-auriculo-vertebral spectrum, Epibulbar dermoid, Anotia, Craniofacial anomalies, Prenatal diagnosis

INTRODUCTION

Goldenhar syndrome, first reported by Maurice Goldenhar, is part of the oculo-auriculo-vertebral (OAV) spectrum and results from abnormal development of the first and second branchial arches.^{1,2} The condition demonstrates marked clinical heterogeneity, with common features including epibulbar dermoids, auricular anomalies such as microtia or anotia, facial asymmetry, and vertebral defects. The estimated incidence ranges from 1 in 3,500 to 1 in 5,600 live births, with a male to female ratio of 3:2.¹

Most cases are sporadic; however, familial occurrences have been described, suggesting a possible genetic component with variable expressivity.² This article presents a case report of Goldenhar syndrome in an 8-month-old female patient. The patient shows symptoms affecting multiple organ systems. These include an epibulbar dermoid in the eyes, marked facial asymmetry, right sided anotia, left sided microtia, atresia of external auditory canal, pre auricular tags, and microcephaly. Notably, the mother exhibited facial asymmetry and prominent bilateral pre auricular tags, suggesting possible

familial occurrence, which makes this case rare. Early recognition in such cases is essential for appropriate multidisciplinary management, and might have some value in pre-natal counselling.^{3,4}

CASE REPORT

An 8-month-old female infant was brought to the paediatric outpatient department (OPD) with chief complaints of a mass in the eye possibly present since birth, but noticed by parents 2 months back. There was no history suggestive of ocular trauma or infection. The patient was a full-term baby with birth weight of 2.5 kg, and there was no history of any maternal illnesses during pregnancy apart from hyperemesis for 6 months. However, the mother had not consumed any iron or folic acid tablets before conception or during pregnancy and, hadn't vaccinated the baby beyond 6 weeks of age.

On head to toe examination, the child had marked facial asymmetry with a deviation of mouth towards left side and bilateral external ear malformations. Microtia and pre auricular tags were present on both sides, and both external

auditory canals appeared to be missing. There was also mild mandibular hypoplasia.

Ophthalmological evaluation confirmed the lesion to be an epibulbar dermoid and an otorhinolaryngology evaluation confirmed bilateral auricular malformations. Subsequently, the patient underwent a comprehensive radiographic evaluation which included an X-ray of the spine to screen for any associated vertebral anomalies, and no vertebral anomalies were detected.

High-resolution computed tomography of the temporal bones revealed right sided grade IV microtia/anotia and left sided grade III microtia, with bilateral external auditory canal atresia and hypopneumatised mastoid air cells. Bilaterally eroded ossicles were found, and bilateral facial nerve showed an abnormal anterior course. Both inner ears, however, were normal.

BERA was suggestive of bilateral moderately severe hearing loss.

A comprehensive systemic evaluation was performed to rule out any associated anomalies. 2D echo did not reveal any cardiac abnormalities. Ultrasound abdomen and KUB did not reveal any renal or visceral malformations. Ultrasound skull was also not significant of any deformity.



Figure 1: Epibulbar dermoid.



Figure 2: Facial asymmetry.

Notably, the mother showed significant facial asymmetry with a deviation to left without any history of trauma, and bilateral pre auricular tags were present.

Based on clinical and radiological findings, a diagnosis of Goldenhar syndrome was established. Patient was advised bone-conduction auditory brainstem response, and parents were counselled regarding bone conduction hearing aids and reconstructive therapy.

A plan to do whole genome sequencing was suggested, to look for possible gene variations causing these symptoms, but the patient's family did not consent to that.



Figure 3: Auricular malformation.



Figure 4: Preauricular appendages.

DISCUSSION

OAVS arises due to abnormal embryonic development of derivatives originating from the first and second branchial arches.² The classical triad of Goldenhar syndrome includes ocular dermoids, auricular anomalies, and vertebral defects; however, the presentation is highly variable, and there are no guidelines currently for the minimum diagnostic criteria for this syndrome.^{2,3}

Although prenatal environmental factors like maternal and fetal hypoxia, maternal hypertension, maternal diabetes, viral infections such as influenza or rubella, malnutrition, smoking, exposure to ionizing radiation during the pregnancy through radio-diagnostic procedures, as well as exposure to certain drugs like cocaine, tamoxifen, ibuprofen, and retinoic acid, and consanguineous marriage have been hypothesised as potential causal agents for Goldenhar syndrome, despite extensive research, etiology remains elusive.^{1,2}

Goldenhar syndrome commonly presents with various eye symptoms including epibulbar dermoids, microphthalmia, strabismus, cataracts, coloboma of the iris and retina, as well as coloboma of the upper eyelid.^{1,5} Among ear changes, it is possible to observe auricular appendage, asymmetry of the ears, atresia of the auditory external canal, and preauricular fistulas.^{1,5} In addition to the eye symptoms mentioned earlier, Goldenhar syndrome individuals may also present with hemifacial microsomia, facial asymmetry, mandibular and/or maxillary hypoplasia, dental alterations such as supernumerary teeth, Fallot's tetralogy, dextrocardia, transposition of the great vessels, gastrointestinal tract atresia, scoliosis, microcephaly, hydrocephalus, hypoplasia of the corpus callosum, and several other associated abnormalities.^{1,2,5} Children affected by Goldenhar syndrome may also exhibit characteristics such as low height, delayed psychomotor development, intellectual disability, speech disorders, psychosocial challenges, as well as autistic behaviors. These additional features can contribute to the complex clinical presentation and overall impact on the affected individuals.^{1,3,5}

In the present case, the patient demonstrated key features including epibulbar dermoid, bilateral anotia/microtia, and facial asymmetry, without associated vertebral, cardiac, or renal anomalies. This emphasizes the variable expressivity of the condition and the importance of clinical diagnosis even in the absence of systemic involvement.²

A particularly important aspect of this case is the presence of craniofacial anomalies in the mother. While most cases of OAV spectrum are sporadic, familial occurrences have been reported, suggesting possible genetic contributions with incomplete penetrance and variable expressivity.² The maternal findings in this case raise the possibility of a familial association, although definitive inheritance cannot be established without genetic studies.²

This case highlights the importance of early diagnosis, especially in outpatient settings where subtle findings such as epibulbar dermoids or auricular anomalies may be overlooked. Early recognition facilitates timely multidisciplinary intervention, including ophthalmologic care, hearing assessment, developmental evaluation, and reconstructive planning.^{3,4}

Equally crucial is parental counselling. In cases with suspected familial features, parents should be informed about the uncertain but potential risk of recurrence in future pregnancies. Referral for genetic counselling should be considered to discuss inheritance patterns, phenotypic variability, and available screening options.^{2,4}

Regarding prenatal detection, advances in antenatal imaging have improved the ability to identify features of OAV spectrum in utero.^{6,7} Targeted anomaly scans using high-resolution ultrasonography may detect craniofacial asymmetry, microtia, or mandibular hypoplasia. Fetal

MRI can provide additional anatomical detail in selected cases.⁷ However, mild forms may go undetected, and there are no definitive genetic markers routinely used in clinical practice for prenatal diagnosis. Therefore, in at-risk pregnancies, careful and targeted fetal imaging remains the mainstay of early detection.^{6,7}

CONCLUSION

This case underscores the importance of recognizing early clinical signs of Goldenhar syndrome, even in the absence of systemic anomalies. The presence of suggestive maternal features highlights the need to consider possible familial association. Early diagnosis, multidisciplinary evaluation, and appropriate parental counselling are essential, along with awareness of the potential role and limitations of prenatal detection in future pregnancies.

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REFERENCES

1. Fu Y, Yu H, Zhang J, Zhou N. Goldenhar syndrome with limbal neoformation, microtia and skeletal deformities: a case report and literature review. *BMC Ophthalmol.* 2024;24:81.
2. Tingaud-Sequeira A, Trimouille A, Sagardoy T, Lacombe D, Rooryck C. Oculo-auriculo-vertebral spectrum: new genes and literature review on a complex disease. *J Med Genet.* 2022;59(5):417-27.
3. Vong A, Funamura J. Multidisciplinary management of oculo-auriculo-vertebral spectrum. *Curr Opin Otolaryngol Head Neck Surg.* 2018;26(4):234-41.
4. Puri RD. Clinical dysmorphology: an approach to bedside diagnosis. In: Gupta P, Menon PSN, Ramji S, Lodha R, editors. *PG Textbook of Pediatrics.* Volume 1. 1st edition. New Delhi: Jaypee Brothers Medical Publishers; 2015:33-8.
5. Schmitzer S, Burcel M, Dascalescu D, Popteanu IC. Goldenhar syndrome-ophthalmologist's perspective. *Rom J Ophthalmol.* 2018;62(2):96-104.
6. Barisic I, Odak L, Loane M, Garne E, Wellesley D, Calzolari E, et al. Prevalence, prenatal diagnosis and clinical features of oculo-auriculo-vertebral spectrum: a registry-based study in Europe. *Eur J Hum Genet.* 2014;22(8):1026-33.
7. Ribeiro B, Igreja J, Goncalves-Rocha M, Cadilhe A. Goldenhar syndrome: a rare diagnosis with possible prenatal findings. *BMJ Case Rep.* 2016;2016:bcr2016215258.

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