JALALABAD MEDICAL JOURNAL

Case Report

Goldenhar Syndrome: A Rare Case Report

Naznin Akther[1], Kaniz Fatema[2], Jannatul Ferdush Chowdhury[3], Tofayel Ahmed[4], Zainab Rahman[5], Rabaka Sultana[6]

[1,3,4]Assistant Professor, Department of Paediatrics, Jalalabad Ragib-Rabeya Medical College, Sylhet.

[2]Assistant Registrar, Department of Paediatrics, Jalalabad Ragib-Rabeya Medical College Hospital, Sylhet.

[5]Registrar, Department of Paediatrics, Jalalabad Ragib-Rabeya Medical College Hospital, Sylhet.

[6]IMO, Department of Paediatrics, Jalalabad Ragib-Rabeya Medical College Hospital, Sylhet.

ABSTRACT

Goldenhar syndrome is a rare congenital condition characterised by a range of anomalies, including ear malformations, hemifacial microsomia, epibulbardermoids, vertebral anomalies, and potential internal organ defects. This syndrome arises from maldevelopment of the first and second branchial arches during early embryonic stages. Here we report a case of Goldenhar syndrome with congenital heart disease, bilateral microtia, and hemifacial microsomia.

Keywords: *Goldenhar syndrome, Microtia, Hemifacial microsomia, Epibulbar dermoid.*

[Jalalabad Med J 2025; 22 (1): 37-39]; DOI: https://doi.org/10.3329/jmj.v22i1.86510

INTRODUCTION

Goldenhar syndrome, also known as oculoauriculovertebral spectrum (OAVS), is a developmental anomaly affecting structures that originate from the first and second branchial arches[1]. This condition is sometimes referred to as Franceschetti-Goldenhar syndrome, facioauriculovertebral spectrum (FAV), or first and second branchial arch syndrome[2]. The syndrome was first documented by German physician Carl Ferdinand Von Arlt in 1845, but it was Maurice Goldenhar who detailed its distinctive features in 1952 and the credit for the discovery went to him[3,4].

Goldenhar syndrome is quite rare, with an estimated incidence of 1 in 5,800 live births and a male-to-female ratio of approximately 3:2. Most cases are sporadic, although autosomal recessive, autosomal dominant, and multifactorial inheritance patterns have also been suggested[5]. Certain maternal factors, such as the ingestion of drugs like thalidomide, retinoic acid, tamoxifen, and cocaine have been linked to the development of the syndrome, along with maternal diabetes as a potential etiological factor[6].

Goldenhar syndrome can lead to a wide range of oral and systemic manifestations, and the severity of symptoms varying greatly among individuals. Classical features include the absence or underdevelopment of the auricles and associated middle or inner ear impairments, which can result in hearing loss. Other common characteristics are hemifacial microsomia and hypoplasia of the maxilla or mandible. Congenital scoliosis is present in about 50% of cases, and epibulbar dermoids can pose risks to visual acuity, are also recurrent feature[7,8,9,10]. In addition to OMENS (orbit, mandible, ear, facial nerve, soft tissue) anomalies, patients may experience a range of cardiac, respiratory, central nervous system, gastrointestinal, skeletal, and renal issues[7,11].

Address of correspondence:

Dr Naznin Akther, Assistant Professor, Department of Paediatrics, Jalalabad Ragib-Rabeya Medical College, Sylhet. Email: drnazninakter27@gmail.com.

CASE REPORT

A 3-day-old female baby, the fourth issue of non-consanguineous parents, presented with fever for 2 days and convulsion one time for 1 day, accompanied by auricular abnormalities. The infant was delivered at term via elective lower uterine caesarean section (LUCS) due to a previous caesarean delivery and cried immediately after birth. Mother, aged 24, was healthy during her pregnancy, although she received irregular antenatal care. She had a history of two abortions.

The maternal history was unremarkable for cutaneous eruptions, teratogenic drug exposure, or radiation. There was no family history of similar conditions. On examination, the neonate had facial asymmetry like hemifacial microsomia and right-sided facial nerve palsy, bilateral microtia with external auditory canal stenosis, and a short neck. Vital signs indicated tachypnea, and sucking and moro reflexes were poor. Cardiovascular system examination revealed a pansystolic murmur best heard at the left lower sternal border, without any radiation. Echocardiography demonstrated a small perimembranous ventricular septal defect (3 mm) and an atrial septal defect (4 mm). There were no clinically or radiologically detectable vertebral anomalies, and both clinical and ophthalmological examinations showed no ocular abnormalities. A cranial ultrasound revealed normal findings. A complete blood count revealed leukopoenia with thrombocytopenia. CSF study, serum electrolytes,

Figure-1,2: *Bilateral microtia of the baby.*

Figure-3: *Facial asymmetry of the baby*

serum calcium and random blood sugar revealed normal. Based on the clinical presentation and investigation results, a diagnosis of Goldenhar syndrome with early-onset neonatal sepsis (EONS) was established. The infant was managed conservatively. A consultation with an otolaryngologist was obtained, and a Brainstem Auditory Evoked Response (BAER) test was advised.

DISCUSSION

The study of this condition remains controversial due to the significant variability in symptoms and physical features from case to case. The aetiology of this condition is not fully understood, though it is recognized to have a genetic basis[3,6]. Most individuals with Goldenhar syndrome exhibit microtia, a malformation of the outer ear. Approximately one third of cases show bilateral microtia. In this child, microtia was observed bilaterally. Associated conditions may include underdevelopment of the skull and face, facial asymmetry, hemifacial microsomia and lower motor neuron-type facial nerve palsy. Abnormalities tend to be unilateral in 85% of cases, with bilateral occurrences ranging from 10-33%; asymmetric involvement usually affects the right side more than the left[5,6]. Our reported case exhibited all these features. The frequency of cardiovascular abnormalities in Goldenhar syndrome varies between 5-58%[5,6], including atrial septal defect (ASD), ventricular septal defect (VSD), tetralogy of Fallot (TOF), and ventricular hypertrophy[5]. This baby presented with cardiac lesions in the form of ASD and VSD. Ocular anomalies, particularly bilateral dermoids, are noted in 60% of cases, while vertebral abnormalities occur in 35-60% of cases[6]. Our case had the characteristic ear, facial, and cardiac features, but we surprisingly found no ocular or vertebral anomalies, which are frequently reported in the literature. High-quality ultrasound, high-resolution computed tomography and magnetic resonance imaging may aid in diagnosis. Management typically involves plastic surgery, along with specialised dental care, hearing aids, speech therapy, and physiotherapy as needed. Specific treatments are required for systemic abnormalities. Patients with Goldenhar syndrome can generally expect a normal lifespan, though low IQ and systemic involvement may indicate a guarded prognosis[6].

CONCLUSION

This case report highlights the complexities associated with Goldenhar syndrome, emphasising its multifaceted nature and the importance of a multidisciplinary approach to management. Early diagnosis and intervention are crucial for optimising developmental outcomes and addressing the unique challenges faced by affected individuals. This case not only contributes to the existing literature on Goldenhar syndrome but also underscores the need for ongoing research to better understand its aetiology and to develop tailored treatment strategies.

REFERENCES

1. Gharehbaghi MM, Ghaemi MR. Goldenhar syndrome in an infant of diabetic mother. Iran J Pediatr 2010; 20 (1): 131-4.
2. Saxena R, David MP. Goldenhar syndrome- a rare case report. J Genet Syndr Gene Ther 2012; 3: 2.
3. Castelino RL, Babu S, KaF, Balan P, Laxmana AR, Shetty H. Goldenhar syndrome- a rare case report. J Marmara Uni Inst Health Sci 2015; 5 (3): 211-4.
4. Holla V, Kini R, Kumar P, Kashyap R, Shetty D, Jha K. Goldenhar syndrome- A rare case report. Cumhuriyet Dental J 2019; 22 (1): 131-5.
5. Sen S, Maity D, Koley A. Atypical presentation of Goldenhar syndrome. Bengal J Otolaryngol Head Neck Surg 2021; 29 (1): 98-101.
6. Das AC, Bhattacherjee A, Choudhury TJ, Akter H, Deb B, Hasan R. Goldenhar syndrome- a case report. Sylhet Med J 2011; 34 (1): 57-9.
7. Fu Y, Yu H, Z Jiajia, Z Nan. Goldenhar syndrome with limbal neoformation, microtia and skeletal deformities: a case report and literature review. BMC Ophthalmol 2024; 24: 81: 1-7.
8. Bogusiak K, Puch A, Arkuszewski P. Goldenhar syndrome: current per spectives. World J Pediatr 2017; 13 (5): 405-15.
9. Schmitzer S, Burcel M, Dascalescu D, Popteanu IC. Goldenhar Syndrome- ophthalmologist's perspective. Rom J Ophthalmol 2018; 62 (2): 96-104.
10. Vong A, Funamura J. Multidisciplinary management of oculo-auriculo-vertebral spectrum. Curr Opin Otolaryngol Head Neck Surg 2018; 26 (4): 234-41.
11. Pariza PC, Dima V, Grigoriu C, Berceanu C, Georgescu TA, Mihai BM, et al. Diagnostic challenges in Goldenhar syndrome-a case report. Romanian J Pediatr 2021; 70 (4): 269-72.