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Goldenhar Syndrome: Case Report

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Abstract

Goldenhar syndrome, also known as oculo- auriculo-vertebral (OAV) syndrome, is a congenital defect in the development of the first and second branchial arches resulting in incomplete development of the ear, nose, soft palate, lip, and mandible on one side of the face [4].

OAVS affects males more frequently than females by an approximate 3:2 ratio. There is some disagreement concerning the disorder's rate of occurrence. Reported estimates range from one in 3000 to 5000 live births up to one in 25,000-40,000 live births. Most of the physical characteristics associated with OAVS are apparent at birth (congenital), with the possible exception of facial asymmetry, which may not become apparent until approximately four years of age in many cases [5].

In this article, we present a case of male newborn presented with simple TTN (transient tachypnea of newborn) associated with facial and organic anomalies diagnosed clinically and by imaging studies as Goldenhar syndrome.

Keywords: Early diagnosis; Goldenhar syndrome; Oculo-auriculo-vertebral spectrum; Multidis-ciplinary approach; Malformations of ear, eye, nose, soft palate, lip, mandible, vertebral and internal organs (heart, kidney, brain, esophagus, etc.)

Introduction

Goldenhar syndrome, also known as oculo-auriculo-vertebral syndrome (OAVS), is a rare congenital condition arising from defects in the first and second branchial arches. It was first described by Dr. Maurice Goldenhar in 1952 [1]. 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]. The etiology of this condition is not yet fully established. Abnormalities of chromosomes, neural crest cells, environmental factors during pregnancy like ingestion of drugs, such as cocaine, thalidomide, retinoic acid, 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 [3]. Clinically, the patient may present with a variety of features ranging from facial abnormalities, ear abnormalities, eye abnormalities, vertebral defects, congenital heart problems and internal organs defects.

In this article, we report a case of Goldenhar syndrome along with discussion on clinical features, importance of early diagnosis, and interdisciplinary approach to managing it.

Case Report

We report a case of a full-term male baby (38 weeks) born via cesarean delivery to a healthy mother, Gravida 8 Para 6 Aborta 2. Maternal history was negative, no pregnancy complications were mentioned, no drugs or radiation exposure were noticed. The mother denies a family history of similar diseases. The baby was born with an APGAR score of 7, 8 at one and five minutes respectively, vigorous, active, pink tonic, crying. No resuscitation was needed upon delivery. On physical examination, he had facial abnormalities: an absent right ear (Figure 1), an ear tag or accessory tragi on the left ear (Figure 2), ipsilateral facial underdevelopment or hemifacial microsomia (Figure 3), a left facial fistula near the upper lip, mandibular hypoplasia or micrognathia (Figure 4). In addition, the baby developed tachypnea, retractions, and grunting. The baby was otherwise healthy with normal weight, height, and head circumference. He was admitted to the neonatal intensive care unit for transient tachypnea in newborns, congenital malformations, and further investigations.

Labs were done and were all within normal ranges (Table 1 and 2).

Screening for any associated anomalies was done, including chest X-ray, KUB, echocardiography, echo abdominal pelvis, and CT brain. Subspecialties were consulted (ENT, cardiologist, neurologist).

ENT wise: bilateral ear malformations with right auricular absent and narrowing of the right ear canal, left ear tag (accessory tragi), left facial fistula near the upper lip, ipsilateral facial underdevelopment (hemifacial microsomia), mandibular micrognathia.

Cardiology wise: small PDA restrictive 1,5mm shunt left to right, gradient 30mmhg. Small perimembranous VSD with 2mm shunt left to right, gradient max of 30 mmhg.

Neurology wise: left partial facial palsy, CT brain showed mild cortical atrophy mainly at the posterior aspects of parietal lobes is seen, mild dilatation of the ventricular system is noted, there is congenital absence of the right external ear. CT scan of the inner ear, MRI of brain, EEG and karyotype are recommended for better evaluation.

Systemic intravenous antibiotics (ampicillin and cefotaxime) were given for 7 days. After one week, the baby was discharged home with detailed instruction and follow up regarding Karyotype, MRI of brain and EEG.

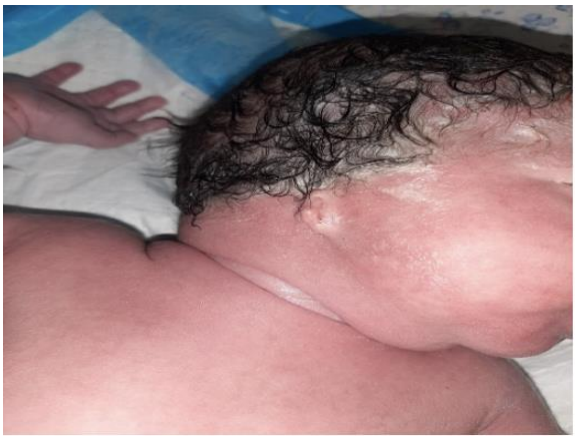


Figure 1: Absent right ear.

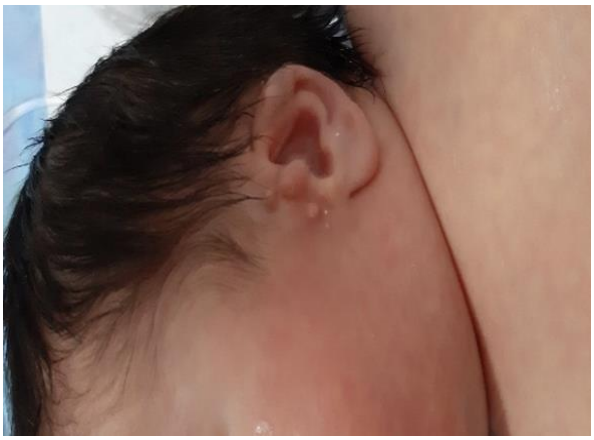
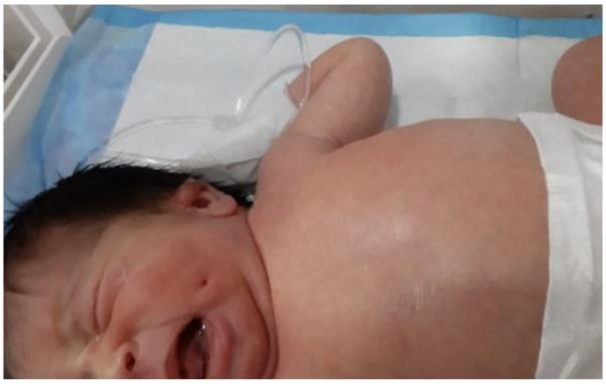


Figure 2: Ear tag or accessory tragi.



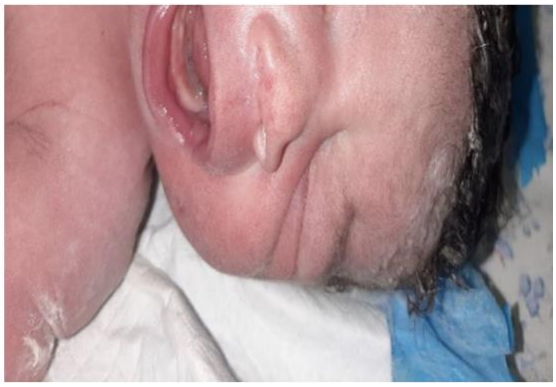


Figure 3: Ipsilateral facial underdevelopment, hemifacial microsomia (left).

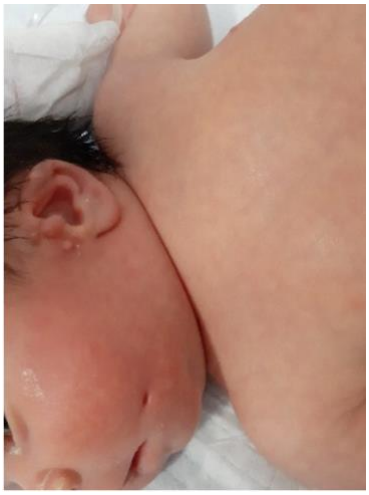


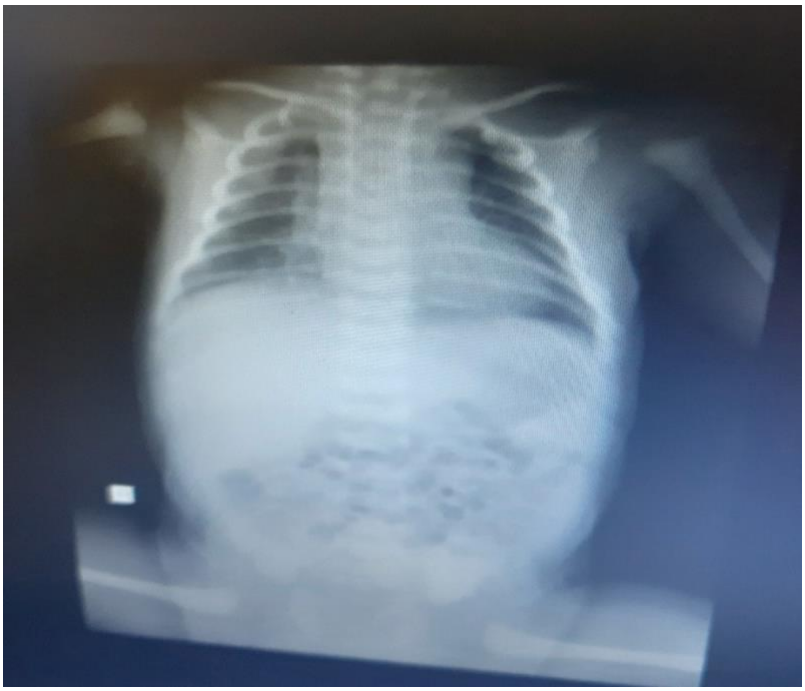
Figure 4: Left facial fistula near upper lip with mandibular hypoplasia (micrognathia).

Table 1: Labs done on admission.

CBCD	
WBC	16200 CU/MM
RBC	3.38 CU/MM
Hemoglobin	12.2 G/DL
Hematocrit	36.30%
Neutrophils	49%
Lymphocytes	42%
Platelets	243000 CU/MM

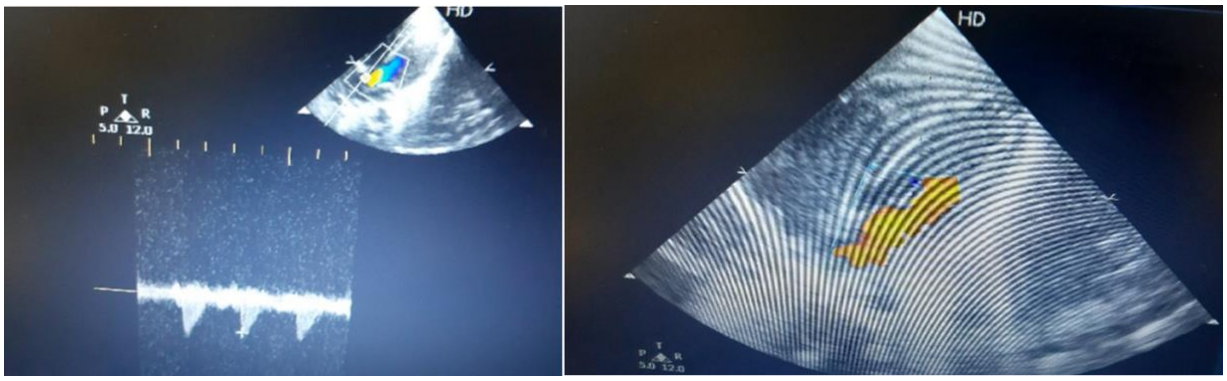
Table 2: Labs done on admission.

BIOCHEMISTRY	
Glucose	62 MG/DL
Urea	25 MG/DL
Creatinine	0.95 MG/DL
Bilirubin Total	4.11 MG/DL
Bilirubin Direct	0.27 MG/DL
SGOT	19 U/L
SGPT	12 U/L
Sodium	133.3 MMOL/L
Potassium	4.59 MMOL/L
Chloride	100.9 MMOL/L
Calcium	9.05 MG/DL
Magnesium	1.82 MG/DL
CRP	< 5 MG/L (Negative)



C-XRAY: Mild bilateral basal infiltrates are seen, the heart is normal in size, the trachea is central, the thoracic cage is intact.

KUB: Bowel distention is seen, No air-fluid levels are noted.

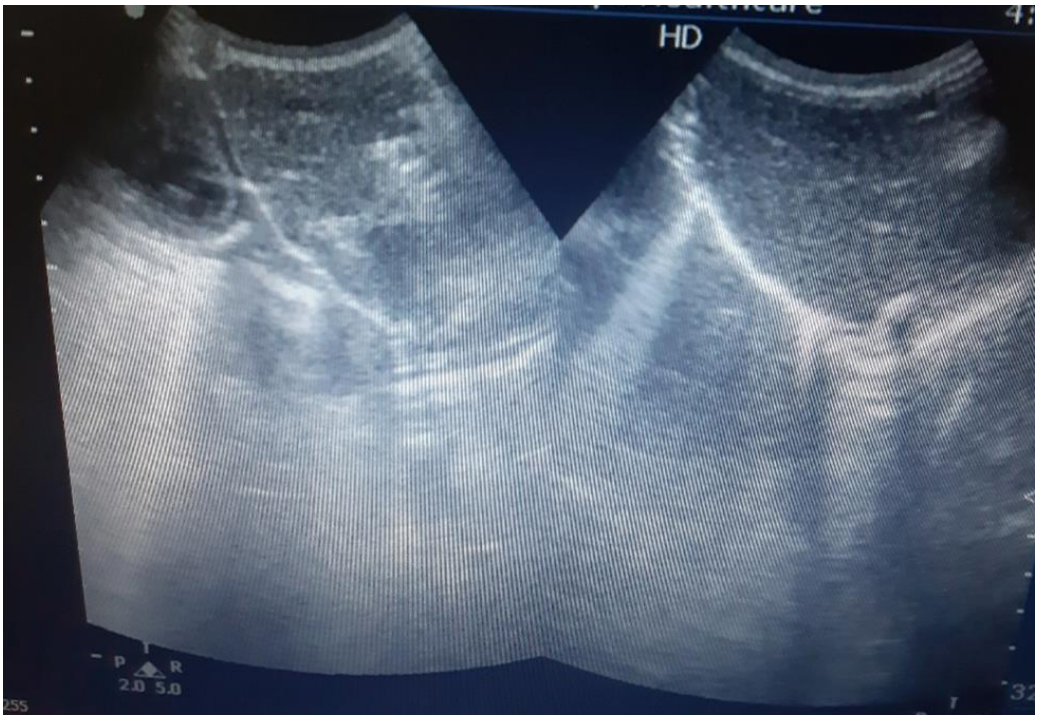


Transthoracic Echocardiogram TTE:

Small perimembranous VSD (2mm) shunt left ----- right, gradient max 30mmhg.

Small PDA restrictive (1,5mm) shunt left ----- right, gradient 30mmhg.

Follow up after one month is recommended.



Echo Abdominal and Pelvis:

The left kidney was not visualized and probably congenitally absent or ectopic.

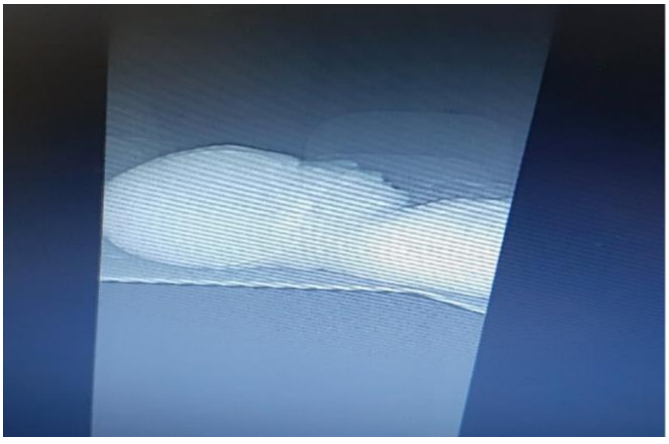
The right kidney is normal in size and echotexture with no evidence of hydronephrosis.

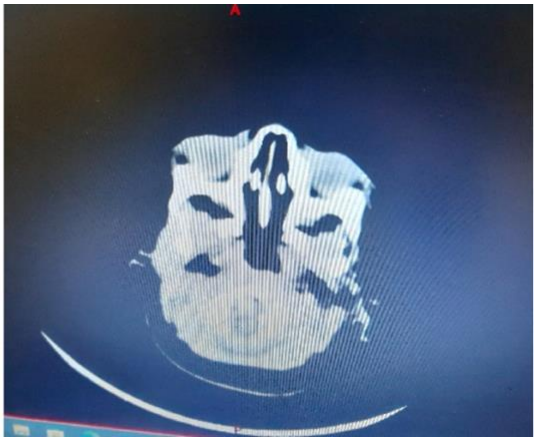
The liver and spleen are normal, the gall bladder is not visualized and probably contracted, No biliary ductal dilatation is seen.

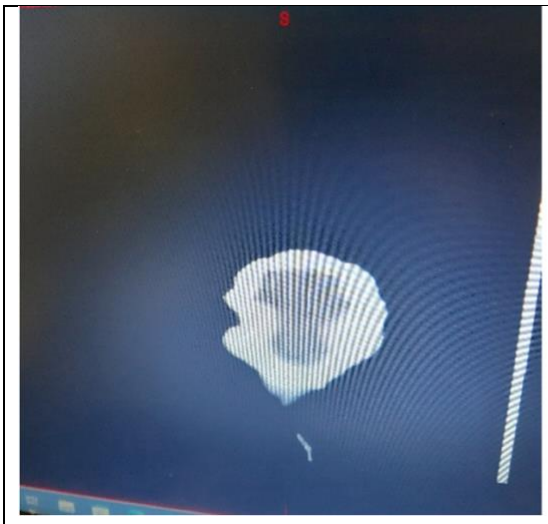
No abnormality is seen in the urinary bladder.

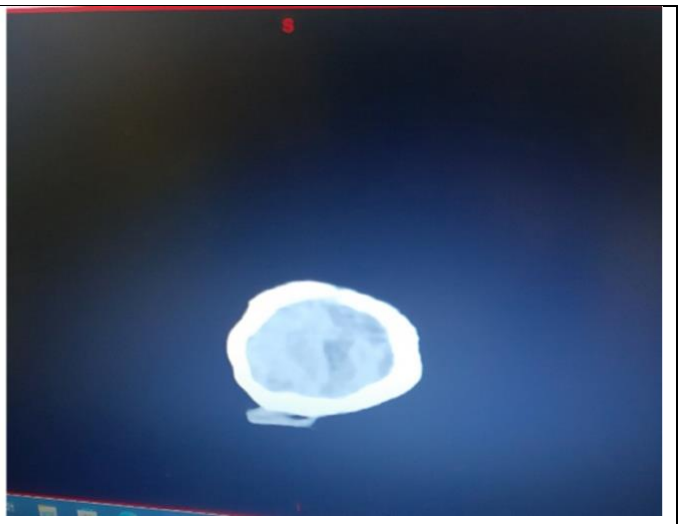
No free fluid in the cul-de sac is seen.

Follow up ultrasound is recommended to assist the left kidney adequately if it is absent or ectopic.









CT Brain:

Mild cortical atrophy mainly at the posterior aspects of parietal lobes is seen.

Mild dilatation of the ventricular system is noted.

There is congenital absence of the right external ear.

Narrowing of the right external auditory canal in the medial aspect is noted.

No masses, no bleed, no subdural collections are noted.

CT scan of the inner ear is recommended for better evaluation.

Discussion

Goldenhar syndrome is a rare congenital defect characterized by complex craniofacial abnormalities associated with vertebral, cardiac, renal, central nervous system, and gastrointestinal malformations [10]. In medical literature, this syndrome is also known as oculo-auriculo-vertebral spectrum, Goldenhar-Gorlin syndrome, and oculo-auriculo-vertebral dysplasia with hemifacial microsomia. It is characterized by a triad of accessory tragi, mandibular hypoplasia, and ocular dermoids, as presented in our case. It is an extremely rare, inherited syndrome with unclear etiology most cases are sporadic, as in our case. In some cases, positive family histories have been found to be present that have suggested autosomal dominant or recessive inheritance. Some researchers suggest a multifactorial mode of inheritance due to the interaction of several genes, possibly in combination with environmental factors. Drugs, such as cocaine, thalidomide, retinoic acid, and tamoxifen, ingested during pregnancy have also been suggested as etiologic factors [11]. Maternal diabetes and infections caused by rubella and influenza during pregnancy may be related to the development of this syndrome. Abnormal vascular supply of embryo, disrupted mesodermal migration can lead to defective formation of branchial arche and vertebral system [6]. Goldenhar syndrome affects one out of every 3,500 to 25,000 children at their time of birth. The formational flaws appear in the spine, eyes, face, and ears of people with Goldenhar syndrome. In some cases, this condition can also cause harm to the internal organs. The severity of Goldenhar syndrome depends on the individual case. The condition is categorized under the group of disorders called craniofacial microsomia. The characteristic combination of external ear anomalies and ipsilateral facial underdevelopment is the hallmark of Goldenhar syndrome, as seen in our case [7].

The condition is mostly unilateral in occurrence in 85% of cases, with the right side being more frequently affected than the left, with a ratio of 3:2 [8]. In our case, unilateral facial involvement on the left side was seen, which made it a more rare category, baby had left facial underdevelopment (hemifacial microsomia), with a facial fistula on the left side near the upper lip, accessory tragi on the left ear. Mandibular hypoplasia is usually seen in patients, as in our case. Systemic involvement in Goldenhar syndrome can vary widely too. Among cardiovascular anomalies, tetralogy of Fallot and ventricular septal defects are most commonly associated with OAVS [9]. In our case, the baby has PDA and VSD. In addition to brain and renal anomalies, the baby has mild cortical atrophy, mainly at the posterior aspects of the parietal lobes is seen, mild dilatation of the ventricular system is noted, and the left kidney is congenitally absent or ectopic. Eye abnormalities in Goldenhar syndrome include anophthalmia, microphthalmia, eyelid and iris coloboma, epibulbar limbal dermoids, strabismus, and retinal abnormalities [12]. Cleft lip and palate, macrostomia, micrognathia, webbing of the neck, a short neck, tracheoesophageal fistulas, and abnormalities of the sternocleidomastoid muscle may be associated. The vertebral abnormalities associated with Goldenhar syndrome are supernumerary vertebrae, hemivertebrae, and fused vertebrae. These abnormalities occur more commonly in the cervical spine. Sometimes they are associated with rib anomalies, scoliosis, kyphosis, and skull abnormalities [14].

The diagnosis of Goldenhar syndrome is made by physical examination, imaging studies, and laboratory tests [15]. Intranatal diagnosis can be confirmed ultrasonographically by the detection of abnormalities. Sometimes the diagnosis of Goldenhar syndrome can be difficult [16]. Thus, Martelli Junior et al. (2010) [16] reported a mean age at the time of diagnosis of 7.15 years (ranges 3 months to 12 years). So, the diagnosis of Goldenhar syndrome is clinical, as there is no specific genetic defect described yet in this developmental disorder.

The treatment of this syndrome is multidisciplinary team including pediatricians and surgeons. Surgical treatment of this syndrome generally involves the surgical correction of facial cosmesis and improvement of hearing and sight loss. Cleft lip and palate surgical repair and surgical orthodontic reconstruction of dental malocclusion, if they are present.
In our case, the treatment was only medically for the respiratory distress syndrome, systemic intravenous antibiotics (ampicillin and cefotaxime) were given for 7 days. After one week, the baby was discharged home with detailed instruction and follow up regarding Karyotype, MRI of brain and EEG.

Conclusion

Goldenhar syndrome is a rare congenital defect also known as oculo-auriculo-vertebral syndrome (OAVS), arising from defects in the first and second branchial arches. characterized by complex craniofacial abnormalities associated with vertebral, cardiac, renal, central nervous system and gastrointestinal malformations. Patients have to be examined by a multidisciplinary team of specialists, in order to detect the visceral anomalies, that may be associated with this syndrome. In Goldenhar syndrome early diagnosis, adequate management, and continued monitoring of the patient are very important to optimal long-term outcomes. The course and prognosis of Goldenhar syndrome may be favorable but depends on the severity of abnormalities.

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