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Case Report

A rare case of Goldenhar syndrome: oculo-auriculo-vertebral spectrum

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ABSTRACT

Franceschetti-Goldenhar syndrome or Goldenhar syndrome, also known as facio-auricular-spectrum (FAV), first and second branchial arch syndrome, or Oculo-Auriculo-Vertebral (OAV) spectrum is a rare congenital malformation which encompasses various morphological and functional abnormalities. The incidence of Goldenhar syndrome has been reported between 1:3500 to 1:5600 children, with a male: female ratio of 3:23. The incidence is higher, about 1 in 1000 children with congenital deafness. The exact etiology is not known. However, it is possible that abnormal embryonic vascular supply, disrupted mesodermal migration or some other factors leads to defective formation of the brachial and vertebral system. Most of the cases have been sporadic. Autosomal dominant, autosomal recessive and multifactorial modes of inheritance have also been suggested. Chromosomal studies have not revealed any abnormality. Authors report a case of a neonate with hemifacial microsomia, bilateral cleft lip and cleft palate, right deformed pinna, right facial palsy, single umbilical artery and congenital heart disease.

Keywords: Goldenhar syndrome, Hemifacial microsomia, Oculo- auriculo-vertebral spectrum

INTRODUCTION

Goldenhar syndrome, also known as Oculoauriculo-vertebral spectrum (OAVS) is a developmental anomaly involving structures derived from first and second branchial arches.[1,2] It was first described by Dr. Maurice Goldenhar in 1952.[3] 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.[4] Smith used the term Facio-auriculo-vertebral sequence to include both Goldenhar syndrome and hemifacial microsomia. The exact etiology of OAVS is not known. It may have a multifactorial etiology. Abnormal vascular supply disrupted mesodermal migration or disturbance of neural crest cells may be the cause of the disease.[1,5] Ingestion of drugs such as vitamin A, primidone, thalidomide, tamoxifen, and cocaine by the pregnant mother has been associated with the development of this syndrome.[5,6] Maternal rubella and influenza have also been suggested as etiologic factors.[5] Autosomal dominant, autosomal recessive and multifactorial modes of inheritance have been reported in 1-2% of cases.[5,7,8]

Clinically, the patient may present with a variety of features ranging from facial abnormalities, ear abnormalities, eye abnormalities, vertebral defects, and congenital heart problems.[9] Here authors report an infant of diabetic mother with Goldenhar syndrome.

CASE REPORT

A live late preterm male baby weighing 2.577 kg was born by emergency caesarean section in our institution at 35 weeks ±6 days gestation to a 30-year-old primigravida with gestational diabetes mellitus, preeclampsia and hypothyroidism.

Figure 1: Bilateral cleft lip, cleft palate, pseudomacrostomia (R) angle of mouth, (R) facial palsy.

This child was the first born of a non-consanguineous marriage.

Figure 2: Right deformed pinna, micrognathia.

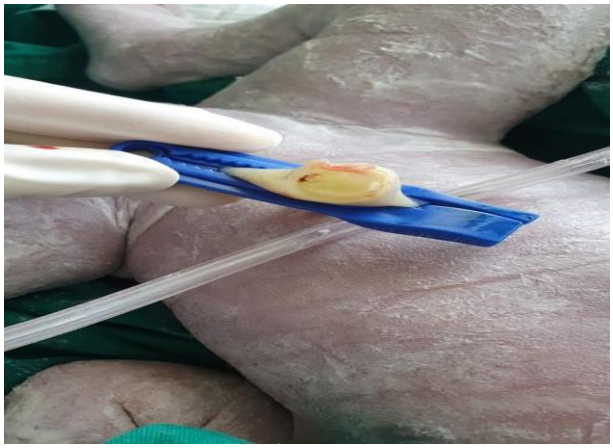

Figure 3: Single umbilical artery.

On examination the baby had bilateral cleft lip and cleft palate, psuedomacrostomia of right corner of mouth, (R) deformed pinna, right sided facial palsy (LMN), left sided choanal atresia, natal teeth, micrognathia (Figure 1and 2) and single umbilical artery (Figure 3). On examination the baby had bilateral cleft lip and cleft palate, psuedomacrostomia of right corner of mouth, (R) deformed pinna, right sided facial palsy (LMN), left sided choanal atresia, natal teeth, micrognathia (Figure 1 and 2) and single umbilical artery (Figure 3). The child was noted to have a continuous murmur on third day of life.

Table 1: Lab Investigations.

Investigation	Results
Complete blood count	Normal
Renal function test	Normal
Thyroid function test	Normal
Serum calcium	10.8mg/dl (normal)

The child was subsequently evaluated with basic investigations, echocardiogram, karyotyping and tandem mass spectroscopy and the reports are as per Table 1, 2 and 3.

Table 2: Radiological Investigations.

Investigations	Results
Chest X-ray	Lung fields clear, no cardiomegaly
MRI Brain	Normal
USG abdomen	Normal
Echo	Situs solitus, levocardia, patent foramen ovale with L-R shunt with additional tiny interatrial communication near IVC
	Small anterior muscular VSD with L-R shunt
	Large 2 mm PDA with L-R shunt peak PDA gradient 17 mmHG.
	Severe pulmonary hypertension (neonatal pulmonary pressures)

Ophthalmologist opinion has been taken in view of right eye lagophthalmos and advised eye care with lubricant eyedrops and bandaging of eyes.

Table 3: Genetic studies.

Genetic testing	Results
Karyotyping	Normal 46 XY No of cells karyotyped -10
	Estimated band resolution 400 Bphs
	Banding method GTG
Tandem mass spectroscopy	Normal

ENT opinion was sought and BERA was suggested at 3 months of age. Genetic specialist opinion was sought from the Department of Human Genetics, MGR university, Chennai, Tamil Nadu, India and the child was diagnosed as a case of Goldenhar syndrome which comes under Oculo-Auriculo-Vertebral Spectrum (OAVS) of malformation.

DISCUSSION

Goldenhar Syndrome or oculoauriculovertebral spectrum is a complex syndrome characterized by an association of maxillomandibular hypoplasia, deformity of the ear, ocular dermoid and vertebral anomalies and the most severe form of hemifacial microsomia.[10] In patients with OAVS, there is a wide spectrum of abnormalities including preauricular tags and pits, malformations of ears, lateral cleft-like extension of the corner of the mouth, cleft lip and palate, facial muscle hypoplasia, hypoplasia of mandibular or maxillary bones, and facial nerve paralysis.[1,11,12] In present case there was cleft lip, cleft palate, right facial nerve paralysis. The characteristic combination of external ear anomalies and ipsilateral facial underdevelopment is the hallmark of Goldenhar syndrome.[9] In present case there is right external pinna deformity and right facial palsy-lower motor neuron type. Pseudomacrostomia is a specific but relatively rare manifestation of Goldenhar syndrome as seen in our case.[13] Congenital heart defects have been reported in 5-58% of patients with OAVS. There are reports of congenital heart disease (CHD) in 32% of patients which were atrial and ventricular septal defects in 32% of cases.[14] In another study there were 36.7% cardiovascular defects, of which 45% were conotruncal defects. In our case there was PDA and VSD.

Ingestion of drugs such as thalidomide, retinoic acid, tamoxifen and cocaine by the pregnant mother may be related to the development of the syndrome. Maternal diabetes, rubella and influenza have also been suggested as etiologic factors. Women with diabetes are at risk for the development of fetal anomalies. It is suggested that a disturbance of neural crest cells in IDM (infant of diabetic mother) acts as the cause of malformations. Wang reported that IDM with preconception diabetes had a 1.5-fold increased risk for OAVS, but this odd ratio did not reach statistical significance. They concluded that OAVS occurs with higher incidence in IDM than in general population.[13] Although this lesion is most specific for diabetes, it is rare. The rate of malformation is related to glucose control during organogenesis. It is suggested that an increased supply of substrate in diabetic mothers leads to an oxidative stress on the developing fetus which in turn generates excess formation of free oxygen radicals that may be teratogenic.[15] Tight metabolic control during organogenesis with close perinatal care reduces the rate of anomalies. The effect of Goldenhar Syndrome is more evident as the child grows, because of delays in the growth and the development of the affected areas. The lack of development of the jaws can cause breathing problems, as well as dental malocclusion which requires multidisciplinary approach. Timing of the reconstruction plays an important role in the treatment. Primary reconstruction typically consists of a cleft repair, corrections of colobomas and ear deformities and removal of dermoids and preauricular tags. The complex treatment is focused not only on dental care, articulation and hearing but also on the prevention and treatment of the psychosocial aspects of the malformation. Treatment requires constant follow-up and reassessment of the results.[10] Children with Goldenhar syndrome tend to have normal IQ.

CONCLUSION

Authors presented this case due to its rarity since the prevalence of Goldenhar Syndrome in Indian population is very low. Maintenance of euglycemia prior to conception and throughout gestation is highly recommended to reduce the incidence of maternal, fetal and neonatal complications. The treatment of this disease varies with age and systemic associations and is mainly cosmetic in uncomplicated cases. Reconstruction surgeries of the external ear maybe performed at the age of 6 to 8 years. Structural anomalies of the eyes and ears can be corrected by plastic surgery.

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