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Goldenhar syndrome with preeclampsia: A rare case report

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Abstract

Goldenhar Syndrome also known as oculo-auriculo-vertebral spectrum (OAVS) is a rare syndrome characterized by hemifacial microsomia, microtia, epibulbar dermoids, ear malformations and hemivertebrae. The aim of this presentation is to present this young mother with oculo-auriculo-vertebral spectrum and related problems. Most of the individuals with this syndrome do not notice themselves before the time of diagnosis. This work reports a case of Goldenhar's syndrome in an 20-year-old female, primigravida, who presented to us with preeclampsia and all classical signs of this rare condition. The enlightening of these patients with this syndrome is very important for the evaluation of this family and for the future pregnancies.

Keywords: Goldenhar syndrome, preeclampsia, oculo-auriculo-vertebral

Introduction

Goldenhar syndrome, also known as facioauriculovertebral spectrum (FAV), first and second branchial arch syndrome, or oculo-auriculovertebral (OAV) spectrum is a rare congenital malformation which encompasses various morphological and functional abnormalities. Maurice Goldenhar described its various characteristic features in 1952. It consists classically of the triad of (Usually unilateral) mal development of the first and second branchial arches, ocular dermoids, and vertebral anomalies^[1, 2]. Reported incidence of this syndrome is 1:3500 to 1:5600 with a male to female ratio of 3:2^[1]. Although most cases are sporadic, autosomal dominance inheritance has also been described.

Though, the etiology of Goldenhar syndrome is not well established, it is thought to be due to exposure to various viruses or chemicals during pregnancy. Some researchers also suggested gestational diabetes mellitus as one of the cause. The MSX homeobox genes play a crucial role in the pathogenesis^[2, 3].

Various clinical features have been described like

- Epibulbar dermoid or lipodermoid (Mostly bilateral); colobomas of the upper eyelid, iris, chorioidea, and retina, or other eye anomalies (e.g. microphthalmia, anophthalmia, cataracts, astigmatism, antimongoloid obliquity of palpebral fissures, and blepharophimosis).
- Preauricular skin tags or blind fistulas; microtia, or other external ear malformations (Dysplasias, asymmetries, aplasias, and atresias of the external meatus); middle and internal ear anomalies.
- Unilateral facial hypoplasia, prominent forehead, hypoplasia of the zygomatic area, and maxillar and mandibular hypoplasia.
- Neck: Branchial cartilage, branchial fistula, webbing, short neck, abnormalities of sternocleidomastoid muscle.
- Unilateral macrostomia (Lateral facial cleft).
- Back: Pilonidal dimple, kyphoscoliosis, Sprengel's deformity
- Hands / Fingers: clubbing, polydactyly, clinodactyly, single palmar crease
- Vertebral column anomalies (Atlas occipitalization, synostosis, hemivertebrae, fused vertebrae, scoliosis, and bifid spine)^[4].

In this report we describe a case of 20 year old pregnant woman complicated with pre eclampsia, in whom classical features prompted us to make a diagnosis of Goldenhar syndrome.

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Case report

A 20 year old woman, G1P0 at 38 week 5 days gestation was referred to our tertiary care hospital located in Indore, Madhya Pradesh, India with a diagnosis of term pregnancy with severe pre-eclampsia. On admission, her general condition as average, Pulse Rate -106/min, Temperature- afebrile, Blood Pressure-170/110, SPO2 90% with oxygen (86% in room air), central cyanosis present, urine albumin+2, urine output-100 cc clear. Her physical examination revealed clubbing in all digits (figure 1 and 2), pre-auricular skin tag on right and left ear (figure 3), facial asymmetry (figure 4), congenital coloboma on upper lid of right eye (figure 5), mal-occlusion of jaw, severe kyphoscoliosis (figure 6). On per abdominal examination uterus was 30 week size with breech presentation, liquor was less, uterus was relaxed, fetal heart sounds were regular. On per vaginal examination- cervical os was closed, not effaced, no show. Her investigations revealed HB-19.5 g%, TLC-10500/cu.mm., plt-2.23 lacs, serum creatinine-1.6mg/dl, urea- 51.3mg/dl.

By 1st trimester scan, she was 37 weeks 3 days. Obstetric USG was performed in our institution which showed-single live intrauterine fetus of 31 weeks 5 days pregnancy breech of 1.72 kg baby weight with AFI-3.9 cm which furthermore confirmed the diagnosis of oligohydramnios with intra uterine growth restriction. Her abdominal ultrasonography revealed multiple right renal cysts with left mild hydronephrosis.

Opinion from ophthalmologist was taken which confirmed our initial diagnosis of right upper lid coloboma, left limbal dermoid (possibility of OSSN i.e. ocular surface squamous neoplasia) and presence of micro aneurysm on temporal side of right eye. Examination by a physician revealed high arched palate, positive wrist sign, positive thumb sign, grade 3 clubbing, s1s2 heart sounds wide split, pan systolic murmur at tricuspid area, and early diastolic murmur at aortic area. Her echo revealed pulmonary arterio-venous fistula.

Difficult intubation was expected because of facial and oral abnormalities. Multidisciplinary approach was taken in consultation with anesthetist and cardiologist and her Cesarean section was done in view of severe pre-eclampsia with breech with severe oligohydramnios and IUGR. Baby was male 1.54 kg and was being shifted to NICU in view of very low birth weight. Her baby had a normal phenotype with no signs of facial and skeletal abnormalities.

She was diagnosed as a sporadic case of OAVS as there were not any similar finding in any other family member. She denied a history of in utero exposure to known teratogenic agents or consanguinity between her parents. She demonstrated age appropriate mental and speech development with no signs of mental retardation. She was kept under close monitoring post operatively. Her vital signs were normal. She was discharged on 7th day after stitch removal with advice on regular follow up visits at our hospital.

Discussion

Goldenhar syndrome was classically described by Maurice Goldenhar as a triad of accessory tragic, mandibular hypoplasia and ocular dermoids. A number of case reports of Goldenhar syndrome have been described in literature. Most cases are sporadic but Tsai and Tsai reported a family in which seven members in three successive generations were diagnosed with Goldenhar syndrome [5]. Goldenhar syndrome has been seen in association with cranial anomalies. Anderson and David reported spinal anomaly in seven patients with wide range of

abnormalities including butterfly vertebrae, hemivertebrae, kyphosis and rib anomalies [6]. Friedman and Saraclar presented a review of cardiac findings and revealed a high frequency of congenital heart disease [7]. Bayraktar *et al.* reported a case of 79-year-old patient of Goldenhar syndrome with multiple congenital anomalies [8]. Abe *et al.* described a case of Goldenhar syndrome associated with cardiac abnormalities such as single ventricle, atresia of pulmonary artery and patent ductus arteriosus [9].

Our patient was provisionally diagnosed as Goldenhar syndrome but a number of other first and second arch syndromes were considered in the differential diagnosis [10].

1. Treacher-Collin syndrome- external ear deformities are extreme and there is anti-mongoloid slant of eyes with absence of zygoma in radiograph.
2. Hallermann-Streiff syndrome (mandibulo-oculo-dyscephaly) - patient has stunted growth, characteristic facial appearance with beaked nose, small mouth, irregular dentition and microphthalmia.
3. Cockayne's syndrome- photophobia and light sensitive skin are prominent features with cataracts, coarse skin and mental retardation.
4. Seckel syndrome- has extreme microcephaly, short stature and beak nose.
5. Delleman syndrome- includes orbital cysts or microphthalmia, focal skin defects and central nervous system cysts and/or hydrocephalous [11].

The frequency of cardiovascular malformation in Goldenhar syndrome is 5-58%, showing great variability [12]. The common cardiovascular malformations are ventricular septal defect and tetralogy of Fallot. OAVS should be kept in mind when cardiac malformations accompanies dysmorphic facial ocular and vertebral features. The case reported here had pulmonary arterio-venous fistula with normal systolic functions.

In the medical literature there are hypothesis concerning development of this syndrome, in 1998 Nakajima *et al.* reported that the cause is the abnormalities of the first and second branchial arches. In 1981 Russell *et al.* pointed out that Goldenhar syndrome might result from mesodermal deficiencies caused by impairment of primitive streak cell migration, because the organs showing the dysplasia spectrum of this syndrome are derived from the mesoderm [13]. It is also reported that reported autosomal dominant inheritance of Goldenhar syndrome, but there is no family history of this syndrome in the present case.

The safety of assisted reproductive technology (ART) is also a major concern as Beksac *et al.* reported that OAVS might be associated with ART [14]. Horsthemke *et al.* reported that there is a bidirectional relationship between twinning and OAVS [15]. In conclusion, we have reported the antenatal course and postpartum period of a pregnant woman with Goldenhar syndrome. The presented case had a spontaneous pregnancy. The fetal imaging studies showed normal developing fetus without any anatomic defect. During cesarian section epidural block was performed by an expert anesthesiologist due to risk of difficult intubation. After delivery, the baby and the mother did well and discharged at 3rd postpartum day. Because patients with Goldenhar syndrome may have associated cardiac defects and performance of anesthesia to these patients requires expert anesthesiologists and cardiologists, they should be referred to tertiary hospitals.



Fig 1 and 2: Showing clubbing in all the digits.



Fig 3: Lateral view of face showing preauricular tags on both right and left side.



Fig. 4: Frontal view of the face showing marked facial asymmetry and malocclusion.

(The patients signed informed consent authorizing the publication of all these pictures)



Fig 5: Frontal view of the face showing coloboma of the upper right eye



Fig 6: Posterior sitting photograph of the patient, demonstrating severe kyphoscoliosis.

(The patients signed informed consent authorizing the publication of all these pictures)

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