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Case Report

A variant of Goldenhar syndrome

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ABSTRACT

We report a case of 10 months old child with a variant of Goldenhar syndrome in the form of microtia of right ear, microsomia of the right-side face, right facial nerve palsy with cardiac anomaly, renal anomaly and sensorineural hearing loss on the right side which were rare associations prompted to report this case.

Keywords: Clinical manifestations, Goldenhar syndrome

INTRODUCTION

Facioauriculovertebral syndrome was first recorded by German physician, Carl Ferdinand Von Arlt.[1] Goldenhar syndrome (oculoauriculovertebral dysplasia with hemifacialmicrosomia) is a rare congenital anomaly involving the first and second brachial arches.[2] It is also known as the oculoauriculovertebral syndrome (OAVS) because of the association of eye anomalies with or without vertebral and ear anomalies.[3]

Oculoauriculovertebral dysplasia, consisting of preauricular appendages, fistulas and epibulbardermoids, was first described in 1952 by the Swiss ophthalmologist Maurice Goldenhar. Gorlin et al, included vertebral anomalies as signs of the syndrome and suggested the name Oculoauriculovertebral (OAV) dysplasia for this condition. Smith used the term facio-auriculo-vertebral sequence to include both Goldenhar syndrome and hemifacialmicrosomia. The classic triad is mandibular hypoplasia resulting in facial asymmetry, ear and/or eye malformation and vertebral anomalies. Goldenhar's syndrome is often characterized by the combination of anomalies including dermal epibulbar appendices and malformation of the ears. We present a case of a variant of Goldenhar Syndrome.

CASE REPORT

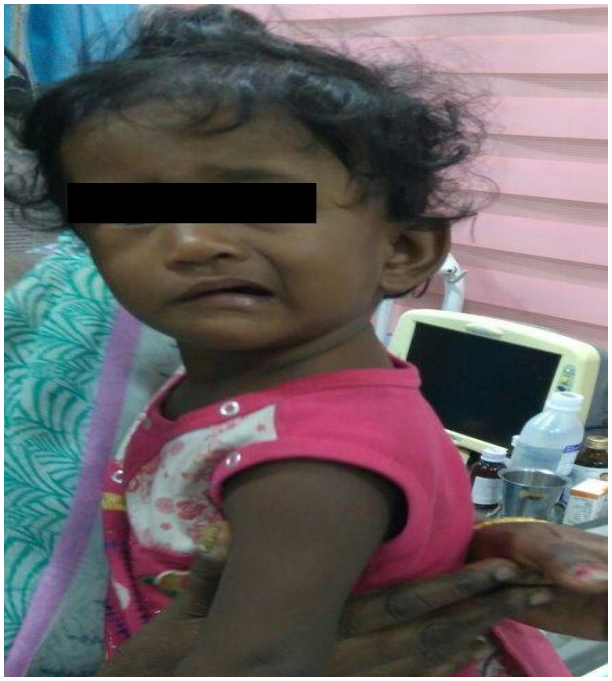

Figure 1: Right side facial macrosomia, right facial asymmetry with deviation of angle of mouth to the left side.

Study report a 10-month-old female child who was admitted in our hospital with diagnosis of Bronchopneumonia. This child was second born to non consanguinous healthy parents with no significant antenatal, natal, postnatal and family history. Developmental milestones were normal.

Physical examination revealed right side facial microsomia,right facial asymmetry with deviation of angle of mouth to the left side (Figure 1) and inability to close right eyelid completely, microtia of right ear and microcephaly (Figure 2).

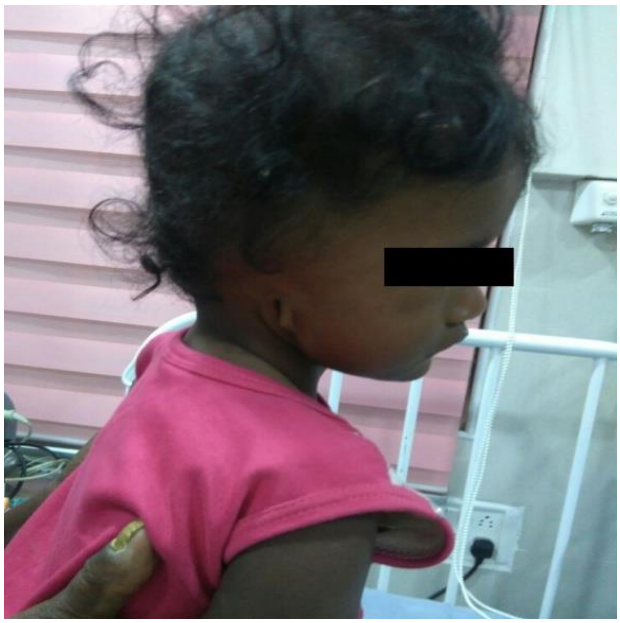

Figure 2: Microtia of right ear and microcephaly.

Systemic examination revealed cardiovascular system with soft systolic murmur, respiratory system- bilateral air entry present and bilateralcrepitations present. CNS and Per abdomen- NAD.

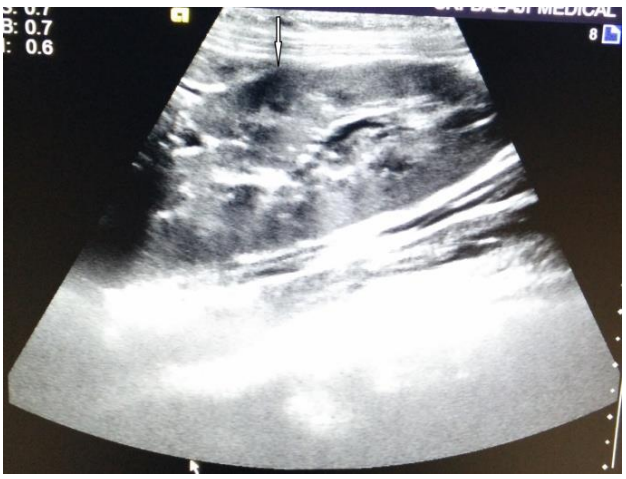

Figure 3: Right kidney not seen in right renal fossa, but seen across the midline fused with the upper pole of left kidney.

Investigations showed chest X ray suggestive of bronchopneumonia, ECHO showed situs levocardia, small patent ductus arteriosus left to right shunt with small defect width 2 mm, USG Abdomen suggestive of right kidney not seen in right renal fossa, but seen across the midline fused with the upper pole of left kidney (Figure 3). BERA was done which showed Sensorineural hearing loss on right side. X-ray Spine was normal. MRI Brain was done which showed Normal Study. Fundus examination was normal.

We diagnosed the patient as a case of rare variant Goldenhar syndrome on the basis of physical findings, systemic examination and investigations.

DISCUSSION

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.[1] 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.[3] Most of the cases have been sporadic. Autosomal dominant, autosomal recessive and multifactorial modes of inheritance have also been suggested.[3] Chromosomal studies have not revealed any abnormality.[4]

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.[5,6] In our case there was no history of maternal intake, any febrile illness or diabetes during pregnancy.

The classic features of this syndrome include ocular changes such as microphthalmia, epibulbardermoids, lipodermoids and coloboma; aural features such as preauricular tragi, hearing loss and microtia; and vertebral anomalies such as scoliosis, hemivertebrae and cervical fusion.[2,3] The abnormalities are found to be unilateral in 85% of cases and bilateral in 10-33% cases.[5] In Goldenhar syndrome ocular anomalies especially bilateral dermoids are seen in 60% of the cases, vertebral anomalies in 40% of the cases and ear anomalies also in 40% of the cases.[3] In our case the child had microtia of the right ear, right facial nerve palsy, microcephaly and right facial microsomia.

Systemic features are found in about 50% of the patients.[7] Tetralogy of fallot and Ventricular Septal defects are the most common anomalies associated with OAVS.[8] But in our case the child had small PDA left to right shunt, defect width 2mm. Cleft lip and palate, macrostomia, micrognathia, webbing of the neck, short neck, tracheoesophageal fistula, abnormalities of sternocleidomastoid muscle, umbilical hernia, inguinal

hernia, urologic anomalies , hypoplastic vagina and anal anomalies may be associated.[2,5,7] Anopthalmos, facial palsy, calcification of the falxcerebri, undescended testis and association of goldenhar syndrome with Turners syndrome and glaucoma are the rarer reported associations.[4] In our case the child had crossed fused ectopic right kidney. BERA was done which showed sensorineural hearing loss on the right side. MRI brain was done showing normal study.

The treatment of this disease varies with age and systemic associations and is mainly cosmetic in uncomplicated cases. In patients with Mandibular hypoplasia, reconstruction can be done with rib grafts and an under developed maxilla can be lengthened by a bone distraction device. Reconstruction surgeries of the external ear may be performed at the age of 6 to 8 years. In patients with milder involvement, jaw reconstruction surgeries can be done in early teens; epibulbardermoids should be surgically excised3,5. Structural anomalies of the eyes and ears can be corrected by plastic surgery.[3]

Prognosis is variable and depends on the presence and severity of associated cardiovascular, neurological and other complications.

CONCLUSION

The outlook for children with Goldenhar syndrome varies. Once treatment has been administered, the majority can expect to have a normal life span and normal level of intelligence.

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