

UROGENITAL ANOMALIES IN GOLDENHAR SYNDROME – A RARE CASE REPORT

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ABSTRACT

Goldenhar syndrome is one of the rare congenital developmental anomalies involving the structures developing from first and second branchial arches. The classic features of Goldenhar syndrome include ocular changes such as microphthalmia, epibulbar dermoids, lipodermoids and coloboma. Aural features such as preauricular tragi, hearing loss and microtia or even anotia may be present. Costo-vertebral anomalies such as scoliosis, hemivertebrae and cervical fusion are usually present. Micrognathia may also be present which may cause feeding difficulties in neonates. Unusual features like high arched palate, gingival hypertrophy and malaligned teeth may rarely be present. In addition to these various other anomalies involving cardiovascular system, Central nervous system and genitourinary malformations may also be present. We present here a rare case of 3 day old neonate who presented with absence of external auditory canal, multiple accessory ear tags. Ophthalmological examination revealed dermoid cyst in the right eye. Other anomalies included genitourinary abnormalities like hypospadias and micropenis. In view of constellation of abnormalities baby was diagnosed to be having Goldenhar Syndrome. This was a rare case of goldenhar syndrome with urogenital anomalies.

KEYWORDS: Goldenhar Syndrome, microtia, ocular dermoid, dilated pelvicalyceal system.**INTRODUCTION**

Due to involvement of peculiar structures, Goldenhar syndrome is also called oculoauriculovertebral dysplasia with hemifacial microsomia. The characteristic facial abnormalities sometimes associated with vertebral anomalies and involvement of eyes in the form of ocular dermoid is called Goldenhar syndrome. Maurice Goldenhar, a French ophthalmologist first described this syndrome in 1952.^[1] Males are more commonly affected than females. The incidence reported is 1:3500 and 1:5600.^[2] The exact etiology is unknown but disrupted mesodermal migration and abnormal embryonic vascular supply are some of the postulated factors leading to characteristic facial and systemic abnormalities of Goldenhar Syndrome.^[3] Though most cases are sporadic there are various case reports of occurrence of Goldenhar syndrome in families which point towards autosomal recessive and autosomal dominant inheritance patterns.^[4] Also there is evidence to suggest that Ingestion of drugs such as vitamin A, primidone, thalidomide, tomoifen, and cocaine by the pregnant mother has been associated with the development of this syndrome.^[5] Maternal rubella and influenza have also been suggested as etiologic factors.^[6] There are also case reports of Goldenhar syndrome in infants of diabetic mothers.^[7] Microphthalmia, epibulbar dermoids, lipodermoids and coloboma. Aural features such as preauricular tragi, hearing loss and microtia or even anotia may be present. Costo-vertebral anomalies such as scoliosis, hemivertebrae and cervical fusion. About 50% of

patients may have systemic abnormalities. Most common systemic abnormalities include cardiovascular anomalies like ventricular septal defect and Tetralogy of Fallot. Other anomalies may include urogenital anomalies, cleft lip and palate, micrognathia, webbing of neck, short neck, tracheoesophageal fistula and abnormalities of sternomastoid muscle.

CASE REPORT

A 3 day old neonate was referred to our hospital in view of multiple facial abnormalities. Birth history revealed that he was a full term IUGR baby delivered to a primigravida mother. Baby cried immediately after birth. There were no maternal high risk factors like pre eclampsia or gestational diabetes etc. At the time of admission the baby was irritable. There was no apparent respiratory distress in the form of subcostal or intercostals retractions. Head to toe examination revealed abnormally developed ears with multiple accessory ear tags and absent external auditory meatus.



Fig 1: Visible Ear Tags with absent External auditory meatus.



Fig 2: on ocular examination Ocular Dermoid was seen.



Fig 3: hypospadias and micropenis visible on urogenital examination.

Ocular examination revealed dermoid cyst in the right eye, genitourinary examination revealed hypospadias and micropenis. Peripheral pulses were felt. Anterior fontanelle was bulging. Septic screen was positive. CRP was raised. Lumbar puncture was done. CSF examination was suggestive of pyogenic meningitis. Baby was kept NBM. IV antibiotics and IV fluids were started. Blood culture and CSF culture was sent before starting antibiotics. Baby responded well to antibiotics. Orogastric feeding was started and later the baby was shifted to breast feeding. On day 10 of admission the baby was taking breast feeding well with no features of sepsis. USG skull was normal. USG-KUB revealed left pelvicalyceal dilatation. 2 D ECHO was done to rule out any cardiovascular anomaly which was normal. BERA test was done to rule out any defect in hearing which was

normal. Patient was discharged with an advice to follow up in pediatric and plastic surgery OPD.

DISCUSSION

Goldenhar syndrome was first described by Maurice Goldenhar who was a French ophthalmologist.^[1] The classic features of Goldenhar syndrome include microphthalmia, epibulbar dermoids, lipodermoids and coloboma. Aural features such as preauricular tragi, hearing loss and microtia or anotia. Costo-vertebral anomalies such as scoliosis, hemivertebrae and cervical fusion may be present. Micrognathia may also be present which may cause feeding difficulties in neonates. Unusual features like high arched palate, gingival hypertrophy and malaligned teeth may rarely be present. Associated anomalies like cardiovascular anomalies in the form of VSD or TOF may be present. Various urogenital anomalies like ectopic and/or fused kidneys, renal agenesis, vesicoureteral reflux, ureteropelvic junction obstruction, ureteral duplication, and multicystic kidney are also reported.^[8] Wang et al conducted a study in 2002 in which they reported that goldenhar syndrome is more common in infants of diabetic mother. Further they recommended infant of diabetic mother with features consistent with Oculo-Auriculo-Vertebral Spectrum should undergo a workup including hearing evaluation, skeletal survey, echocardiogram, renal ultrasonogram, and immunodeficiency workup if clinically indicated.^[9] Ritchey ML et al reported common urogenital anomalies present in goldenhar syndrome to be ectopic and/or fused kidneys, renal agenesis, vesicoureteral reflux, ureteropelvic junction obstruction, ureteral duplication, and multicystic kidney.^[10] Our case also represents one of the rare cases of Goldenhar syndrome with urogenital anomalies in the form of pelvicalyceal system dilatation, hypospadias and micropenis.

The treatment of goldenhar syndrome depends upon age and systemic involvement. Facial abnormalities may need plastic surgical procedures like pinna reconstruction etc. Systemic involvement like cardiovascular anomalies may need surgical correction. Vesicoureteric reflux may need prophylactic antibiotics or surgical correction. Cases in which systemic involvement is absent or minimal the prognosis is fair.

CONCLUSION

Though Goldenhar syndrome is rare, Any patient presenting with features consistent with Oculo-Auriculo-Vertebral Spectrum (Goldenhar syndrome) should undergo a workup including hearing evaluation, skeletal survey, echocardiogram, renal ultrasonogram, and immunodeficiency workup if clinically indicated. Our case report emphasize that rare urogenital anomalies should not be missed while dealing with these patients.

Conflict of interest: None

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