Original Research Paper Medical Science

A Case Series of Goldenhar Syndrome with the Additional Finding of Tetralogy of Fallot in one Case

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ABSTRACT

Goldenhar syndrome includes a spectrum of ocular, facial and aural malformations with vertebral, renal, respiratory and cardiac manifestations seen uncommonly. We report 3 cases of Goldenhar syndrome with some of these rarely reported findings. One of the cases also had TOF and BT shunting was done for the same.

KEYWORDS Goldenhar syndrome, case series, Tetralogy of Fallot

INTRODUCTION

Goldenhar in 1952 reported a pair of monozygotic twins with hemifacial microsomia, mandibular hypoplasia, auricular malformations, and epibulbar dermoids. [1]

Gorlin et al. in 1963 coined the term "oculoauriculovertebral dysplasia" to describe patients with mandibular hypoplasia, microtia, epibulbar dermoids, and vertebral anomalies. [2]

The incidence varies from 1 in 3,500 to 1 in 5,600 live births. [3] It has a male predominance, the ratio being 3:2. Majority of cases are unilateral (85%) as compared to both sides (10 % to 33%). The right side ear involvement is being more common. [4]

Major clinical features are Ocular manifestations, Epibulbar dermoids, microphthalmia, Upper eyelid coloboma, Strabismus, Diminished visual acuity, Optic nerve hypoplasia. Ear anomalies, Microtia, Preauricular tags and/or pits, Middle ear anomaly, Inner ear defects, Variable deafness. Vertebral defects such as Hemivertebrae, Hypoplasia of vertebrae, usually cervical. Hemifacial microsomia, Ipsilateral hypoplasia of malar, maxillary, and mandibular region, especially temporomandibular joint, Ipsilateral hypoplasia of the facial musculature. [5]

DETAILS OF THE CASES

Case 1

A 1 year old female child was refered to us with facial asymmetry, right sided facial hypoplasia and facial asymmetry, left sided anotia and preauricular tag as shown in Fig. 1. This was the second child of a non consanguineous marriage. There was history of cyanotic spells since 3 months of age, which on evaluation showed TOF with infundibular and valvular pulmonary stenosis. At 10 months of age child was operated palliatively with BT shunt and later refered for reconstructive surgery.

Case 2

A 2 day old male neonate was admitted to our NICU with complains of respiratory distress, feeding difficulties, facial asymmetry due to hypoplasia of left sided facial musculature, maxillary, malar and mandibular region. Left microphthalmia and left sided preauricular tag with bilateral microtia as shown in Fig. 2. Cardiovascular evaluation showed large ostium secundum ASD with multiple fenestrations and moderate pulmonary arterial hypertention. XRay Chest showed bilateral pneumonitis post aspiration with no tracheo-esophageal fistula. After recovery from the respiratory distress child was refered for oromaxillofacial surgery.

Case 3

A 10 years old male child presented with facial hypoplasia and asymmetry, right sided maxillary, malar and mandibular hypoplasia, right sided upper eyelid coloboma, right sided congenital cataract, bilateral preauricular tags and right sided microtia with hearing loss as shown in Fig. 3. On evaluation his IQ was normal with right sided conductive deafness and normal ECHO. CT Brain showed right optic nerve atrophy.

DISCUSSION

Apart from the characteristic findings discussed above, other associated features are Cranial nerve palsy; Cleft lip/palate, Low scalp hair line and Brachial cleft remnants in anterior-lateral neck and CNS anomalies.

Congenital heart diseases (5–58% depending on ascertainment of cases) include Tetralogy of Fallot (with or without right aortic arch) and VSD (account for over half of the cases with congenital heart diseases), Pulmonary stenosis, Patent ductus arteriosus, Coarctation of aorta, Total atrioventricular canal defect, ASD, Transposition of great vessel, Rare isolation of the left innominate artery infradiaphragmatic total anomalous pulmonary venous connection, Wolf-Parkinson-White syndrome.

Respiratory tract anomalies include incomplete lobulation of the lung and gastrointestinal anomalies esp. esophageal atresia. Renal anomalies include renal agenesis and Hydronephrosis. Prenatal growth deficiency and Normal intelligence in noted in most cases.

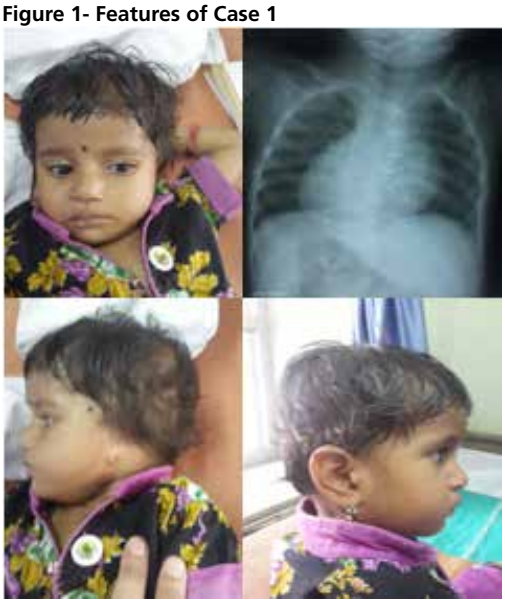
Genetic counseling is important in these cases as the recurrence risk in patient's sibling is of 2%. Management of these cases involve Surgical removal of epibulbar dermoids, if needed, Cleft lip/palate repair, Combined surgical-orthodontic approach for dental occlusion, Traditional osteotomies, followed by acute orthopedic movement and osseous fixation for adult patients with

maxillomandibular hypoplasia, facial asymmetry, congenital micrognathia and hemifacial microsomia. Alternative procedure is distraction osteogenesis. It's important to assess cervical spine for instability before undergoing any general surgery as C1–C2 fusion or occipitocervical Fusion may be needed. Be-

fore anesthesia anticipate airway obstruction and difficulty In tracheal incubation, resulting from a combination of micrognathia, unilateral mandibular hypoplasia and vertebral anomalies including vertebral fusion and odontoid elongation. [5]

FIGURES

Figure 1- Features of Case 1



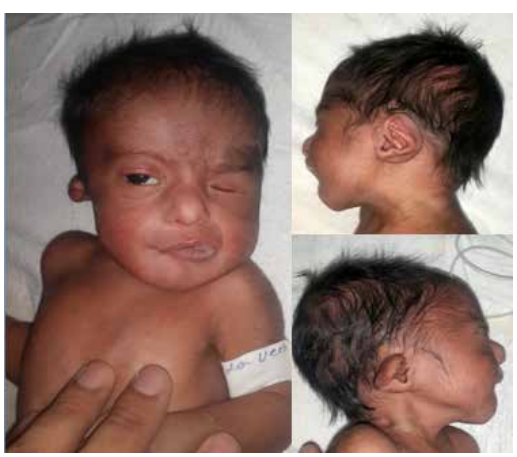


Figure 3- Features of Case 3

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