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 Hemivertebral, 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]

KEYWORDS

Goldenhar syndrome, case series, Tetralogy of Fallot

INTRODUCTION

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.

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 atroventricular 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 Hydrenephrosis. Prenatal growth deficiency and Normal intelligence in not all cases.

Genetic counseling is important in these cases as the recurrence risk in patient’s sibling is of 2%. Management of these cases involves 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 2- Features of Case 2

Figure 3- Features of Case 3

REFERENCES