Case Report

Goldenhar Syndrome—Two Case Report

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Introduction

Goldenhar syndrome (oculo-auriculo-vertebral dysplasia with hemifacial microsomia) is a rare congenital developmental anomaly involving the first and second branchial arches [1]. The incidence of Goldenhar syndrome has been reported to be between 1:3500 and 1:5600, with a male: female ratio of 3:2 [2]. The exact etiology is not known. However, it is possible that abnormal embryonic vascular supply, disrupted mesodermal migration or some other factor leads to defective formation of the branchial and vertebral systems [2,3]. Most of the cases have been sporadic. Autosomal dominant, autosomal recessive and multifactorial modes of inheritance have also been suggested [2].

The classic features of this syndrome include ocular changes such as microphthalmia, epibulbar dermoids, lipodermoids, and coloboma; aural features such as pre-auricular tragi, hearing loss, and microtia; and vertebral anomalies such as scoliosis, hemi vertebrae, and cervical fusion [1,4]. The abnormalities are found to be unilateral in 85% of cases and bilateral in 10-33% cases [4]. 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 [2].

In addition to facial, vertebral, ophthalmic malformations, goldenhar syndrome also associated with cardiovascular, CNS, genitourinary malformations.

Other systemic features are found in about 50% of the patients [5]. Tetralogy of Fallot and ventricular septal defects are the most common cardiovascular anomalies associated with OAVS

Other syndromes associated with multiple pre-auricular tragi include Treacher-Collins syndrome, Wolf-Hirschhorn syndrome, Nager’s acrofacial dysostosis, Wildervank syndrome (cervicooculoacoustic syndrome), Townes-Brocks syndrome, and Delleman syndrome [1]. Treacher Collins syndrome is associated with maxillary and mandibular hypoplasia but is not associated with ocular and aural anomalies [2].

Case 1

A one month-old girl, born of non-consanguineous parents, was referred to our centre for the treatment of coloboma of eyelid and assessment of dysmorphic facial features. The patient was full-term normal vaginal delivery with no perinatal complications. Patient had coloboma of eyelid on right side of the face (Figure 1). Left-sided epibulbar lipodermoid, (Figure 1) B/L pre-auricular tags (Figure 1), malformed pinna of the ear, malformed and hypoplastic right side of face, pulled up angle of mouth, mandibular hypoplasia on right side. No vertebral anomalies were noted. This patient was referred to ophthalmology outpatient department regarding coloboma of eyelid and epibulbar lipodermoid. The patient was given treatment symptomatically and was asked for regular follow-up.

Case 2

A one and half year old girl of non-consanguineous parents of middle class family was referred to me for her assessment of dysmorphic facial features. The patient was full-term, normal vaginal delivery with no perinatal complications. She had asymmetry of the face (Rt side deviation) but no coloboma of iris or eyelid and epibulbar lipodermoid. Bilateral pre-auricular tags,
malformed pinna of the ear, malformed and hypoplastic right side of face, pulled up angle of mouth, mandibular hypoplasia on right side. No vertebral anomalies were noted. The child was developmentally normal and there was no family history of same type of illness. This patient was treated symptomatically and was asked for regular follow-up.

**Fig 2:** (Case No .2) shows facial asymmetry, pre-auricular tag (arrow) and squint.

**Discussion**

Goldenhar syndrome or Oculo-Auriculo-Vertebral Syndrome is a rare disorder characterised by Hemi facial microsomia, Epibulbar-dermoid and Deformity of ears with pre auricular appendage [5,6]. It is associated with anomalous development of the first and second branchial arches during blastogenesis. The syndrome was first described in 1952 by the French ophthalmologist Maurice Goldenhar [1]. Tsai and Tsai reported this syndrome in three consecutive generations in a family [5]. In our case all the features mentioned above were seen. Ocular manifestations include epibulbar dermoid in 75% of cases, bilateral at infratemporal quadrant [6]. Otherocular manifestations are coloboma, microphthalmos, cataract, iris anomalies, anophthalmos, optic nerve hypoplasia and squint [5]. Dacrocystitis has been reported in some cases. Otherocular manifestations are coloboma, microphthalmos, cataract, iris anomalies, anophthalmos, optic nerve hypoplasia and squint [5]. In our case left unilateral epibulbar Dermoid in the infratemporal quadrant was seen. Auricular manifestations are preauricular kintags, accessory auricle [5]. Anotia is rare and has been reported by Jaison and Batra [5]. Bilateral preauricular tags were seen in our case. Involvement of vertebra and ribs are observed in 24% of cases [6,7]. Spina bifida is the least severe of all vertebral anomalies. Association of post axial polydactyly with GHS is described [8]. Central nervous system manifestations are seen in 46% of cases. Diffuse cerebral hypoplasia, Dilated lateral cerebral ventricles (asymptomatic Hydrocephalus), Corpus callosum dysgenesis and frontal hypodensities were the most frequent abnormalities. Arnold Chiari formation and facial nerve palsy have also been reported [9]. Cardiac defects include Ventricular septal defects, Patent ductus arteriosus, Fallot’s tetralogy, Coarctation of Aorta and pulmonary stenosis [10]. The reported prevalence of cardiovascular anomalies is 5-58% in our case VSD congenital heart disease was seen.

**References**