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Case Report

Goldenhar Syndrome –A Rare Case Report

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ABSTRACT
Goldenhar Syndrome or oculo auriculo vertebral spectrum is a complex syndrome characterized by an association of maxillomandibular hypoplasia, deformity of the ear, ocular dermoid and vertebral anomalies and the most severe form of hemifacial microsomia. We report a case of a 7 year old male child presented to us with bilateral epibulbar dermoids, mild conductive deafness, preauricular tags and operated cleft lip.
Keywords: *Goldenhar syndrome, accessory tragus, oculo auriculo vertebral dysplasia.*

Introduction

In 1881, the first observation of oculo-auriculo-vertebral dysplasia was reported by Von Arlt[1] and in 1952, Dr. Maurice Goldenhar, a renowned Swiss ophthalmologist classified the clinical features and named the malformation complex as Goldenhar Syndrome and described it as a congenital defect characterized by constellation of malformations classically involving the face, eyes and ears.[2]

In 1963, Gorlin et al. suggested the use of the term oculo auricular vertebral dysplasia to describe the syndrome characterized by epibulbar dermoids, auricular appendages, blind-ended auricular fistulas and vertebral anomalies.[3]

It is otherwise known as oculo auriculo vertebral syndrome, hemifacial microsomia, first arch syndrome, first and second branchial arch syndrome, Goldenhar–Gorlin Syndrome, lateral facial dysplasia, unilateral craniofacial microsomia, otomandibular dysostosis, unilateral intrauterine facial necrosis and auriculo-branchiogenic dysplasia., Facio-auricular dysplasias represent a single disorder with great variability of expression and an isolated ear malformation may represent the mildest expression of the disorder.[4]

Abnormalities are unilateral in 85% of cases and bilateral in 10–33% of the cases and the right side is more frequently affected.[5] The incidence of Goldenhar Syndrome has been reported to be varying from 1:3500 to 1:5600 live births and it is present in 1:1000 children with congenital deafness with a male to female ratio of 3:2.[5] The disease is seen as sporadic and its etiology is not fully understood; however, positive family

histories have been reported suggesting autosomal dominant or recessive inheritance. Some researchers have suggested that multifactorial inheritance are caused by the interaction of many genes, possibly in combination with environmental factors.[6,7]

Ingestion of drugs such as thalidomide, retinoic acid, tamoxifen, and cocaine by the pregnant mother may be related to the development of this syndrome. Maternal diabetes, rubella, and influenza have also been suggested as etiologic.[9]

Tetralogy of Fallot and ventricular septal defects are the most common cardiovascular anomalies associated with OAVS. 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 .[9]

Other syndromes associated with multiple pre-auricular tragi include Treacher-Collins syndrome, Wolf-Hirschhorn syndrome, Nager's acrofacial dysostosis, Wildervanck syndrome (cervicooculoacoustic syndrome), Townes-Brocks syndrome, and Delleman syndrome.[10,11,12]

Case Report

A 7-year-old boy, presented to paediatric eye OPD at Venu Eye Institute and Research Center (V.E.I.R.C), New Delhi with complaint of mass on both the eyes. Parents also gave history of operated cleft lip at the age of 4 years. Patient had no history of perinatal complications nad maternal drug intake.

On Examination patient was found to have bilateral epibulbar dermoid, pre-auricular appendices (Figure 1,2), bilateral asymmetry of face (Figure 3) and short stature. On further investigation, patient was found to have mild conductive type of deafness.

On ophthalmic examination, bilateral solid yellowish or pinkish white ovoid masses of approximately 8 x 5 mm in size, non mobile, non tender, with fine hairs over the surface suggestive of epibulbar dermoid (Figure 4). The dermoid in the left eye was extending to the inferior fornix and both the dermoids were covering half the pupilary area (Figure2). Rest of the ophthalmic examination was normal.

Correlating the history and clinical findings a provisional diagnosis of Goldenhar Syndrome as made. Parents were counselled about the syndrome and epibulbar dermoid excision with lamellar keratoplasty with or without amniotic membrane transplantation was advised.

Figure 1

Figure 2

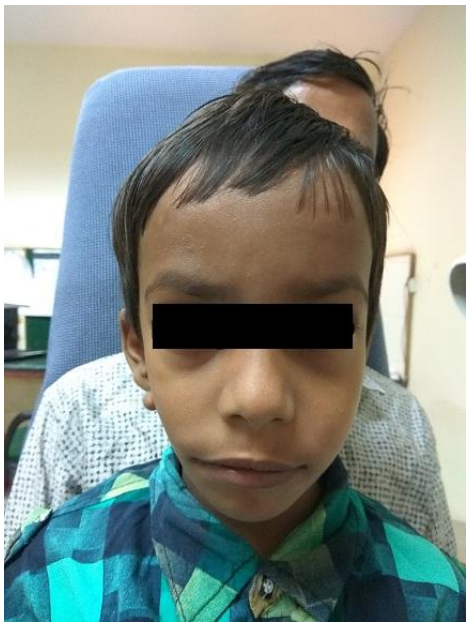

Figure 3

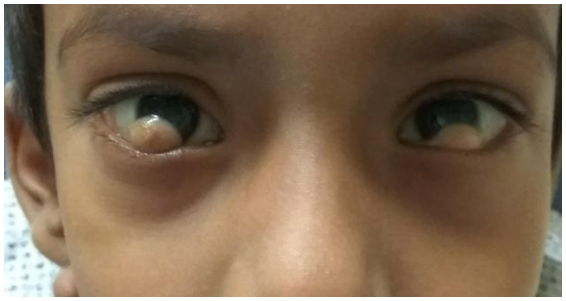

Figure 4

Discussion

Goldenhar Syndrome is a rare hereditary condition characterized by numerous anomalies affecting the first and second branchial arches of the first pharyngeal pouch, the first branchial cleft, and the primordia of the temporal bone.[13,14]

Affection occurs before the end of organogenetic period (7th or 8th week of embryonic life).[15]

The aetiology of this rare disease is not fully understood. However numerous hypotheses have been proposed to explain the etiopathogenesis of this syndrome. Gorlin and Pindborg 1964, suggested that some abnormal process affects the mesoblasts embryologically which affects the branchial and vertebral systems thereby resulting in the syndrome.

Baum and Feingold in 1973, stated that Goldenhar's syndrome may be a sporadic event that occurs early in embryogenesis which is explained by reduced penetrance, somatic mosaicism or epigenetic changes [18,20,21]

Also there are reports of familial cases in successive generation having history of consanguineous marriage that thus requires consideration of autosomal recessive, dominant, or multi factorial inheritance.[6,13,22]

Features of Goldenhar Syndrome

Ocular features-Epidermoid tumors occur in 35 % of all cases. They can be Unilateral (50%) and bilateral (25%). They appear as solid yellowish or pinkish white ovoid masses varying in size from that of a pinhead to 8–10 mm in diameter. They occur most often at the inferotemporal quadrant at the limbus. The surface is usually smooth and frequently has fine hairs. They can occur at any location on the globe or in the orbit and can be dermoid (white solid masses), lipo-dermoid(25%) (yellow, movable, conjunctival), or dermis-like or complex (mesoectodermal). Astigmatism and lipid infiltration of the cornea can lead to encroachment on the pupillary axis leading to vision impairment. Other features include unilateral or bilateral blepharoptosis, elevated orbit, clinical anophtha-lmia or microphthalmia, retinal abnormalities, Colobomas of the upper eyelid, iris, chorioidea, and retina, ocular motility disorders (esotropia, exotropia, duane syndrome),hypertelorism, microphthalmia, anophthalmia, cataract, antimongoloid obliquity of palpebral fissures, microcornea and congenital cystic eye.[17,23-27]

Facies - Unilateral macrostomia , Marked facial asymmetry, cleft lip, cleft palate[3]

Ear - Anomalous pinnae are seen bilaterally. Supernumerary ear tags, unilateral and bilateral preauricular tags of skin and cartilage, along with blind fistulas and sinuses are extremely common.[3]

Skeletal abnormalities - Skull defects like cranium bifidum, microcephaly, dolichocephaly, plagiocephaly. Spinal abnormalities like Spina bifida, hemi vertebrae, butterfly, fused and hypoplastic vertebrae, atlas occipitalization, synosto-sis, hemivertebrae, fused vertebrae, scoliosis, and bifid spine are also common. Radial limb anomalies may take the form of hypoplasia

or aplasia of radius and/or thumb and bifid or digitalized thumb.[3,13,28,29]

Other systemic abnormalitites[8,17]

Congenital Heart Disease – Ventricular septal defect, Atrial Septal Defect

Growth Retardation

Severe respiratory distress – Upper respiratory tract infection , tracheoesophageal fistula,

Urogenital system – Ectopic Kidneys, Ureeteropelvic junction obstruction

Central nervous system – occipital encepahlocele

Retardation of mental development

Various diagnostic aids such as ultrasonography, computed tomography and radiographic analysis should done to rule out the syndrome. Ultrasonography is done during pregnancy and can rule out severehypoplasia of mandible, severe abnormality of the auricle, and cleft lipand/or cleft palate. Computed tomography is done for the evaluation of hearing to see the middle ear bones and to rule out skeletal findings radiographic analysis can be carried out.[6]

Timing of the primary and secondary reconstructions plays an important role in the complex treatment. Primary reconstruction consists of a cleft repair, corrections of colobomas and ear deformities, and extirpation of the dermoids and preauricular tags.[13,17]

Guarded recommendations for surgical removal of ocular dermoids[30]

Grade of pediatric limbal/corneal dermoid	Recommended techniques
Grade I: <50 µm thickness and <1 mm diameter	Simple excision
Grade I: <100 µm thickness and <1 mm diameter	Keratectomy + AMT + ALSCA
Grade II and deeper Grade I	Keratectomy + AMT + ALSCA + PPG versus anterior or deep anterior lamellar keratoplasty ± AMT
Grade III	Total anterior segment reconstruction

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Conflict of interest: None

Abbreviations: AMT, amniotic membrane transplantation (multilayered); ALSCA, autologous limbal stem cell allograft; PPG, pericardial patch graft.

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