

GOLDENHAR SYNDROME: A CASE REPORT

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ABSTRACT

To report an unusual variant of Goldenhar syndrome

We reviewed a 7 year old boy attending our OPD with left upper eyelid coloboma and multiple congenital deformities including auricular, facial and vertebral abnormalities like microtia, hemifacial microsomia, cleft palate, dental abnormalities, kyphoscoliosis and tilted optic disc. He underwent radiological imaging and thorough ocular & systemic examination which were suggestive of Goldenhar syndrome.

The most common ocular finding of Goldenhar syndrome, also known as oculo-auriculo-vertebral spectrum, is epibulbar dermoid, which was absent in our case, instead there was upper-eyelid coloboma and tilted optic disc. However other facial, auricular and skeletal deformities were in concordance with the spectrum of Goldenhar Syndrome.

Goldenhar syndrome consists of a large spectrum of congenital abnormalities and patients might not present with the most diagnostic ocular features. Hence, its diagnosis and treatment requires a multidisciplinary approach.

KEYWORDS: Goldenhar syndrome, Goldenhar-gorlin syndrome, Epibulbar dermoid, Eyelid coloboma, Hemifacial microsomia, Oculo, auriculo, Vertebral spectrum.

INTRODUCTION

This report highlights a peculiar case of a variant of Goldenhar Syndrome presenting with upper eyelid coloboma in the left eye along with the other symptoms of the syndrome. The presence of an isolated eyelid coloboma in Goldenhar syndrome, whose otherwise most common ocular manifestation is epibulbar dermoid, is rarely seen and thus makes this case an interesting study subject.

Goldenhar Syndrome as described by Maurice Goldenhar in his case collections, is a congenital mandibulo-facial malformation with epibulbar dermoids, auricular appendages and auricular fistulas. It is a variant of the Oculo-Auriculo-Vertebral spectrum, a name suggested by Gorlin as he also included vertebral anomalies as signs of this syndrome. It is a pleiotropic condition with a spectrum of cardiac, renal, and skeletal manifestations besides the typical facial and vertebral anomalies (1).

CASE REPORT

A 7 year old boy reported in ophthalmology OPD with a defect in left upper eyelid. There were no other ocular complaints. The child was born to non-consanguineous parents with uneventful prenatal and antenatal period. There was no relevant family history. His mental status was normal and there was no

evidence of developmental delay. Upon examination we found there was an upper eyelid coloboma on the left side figure 1. and further examination of the fundus revealed a tilted optic disc in the left eye.



Fig 1: Left Upper Eyelid Coloboma

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Other ocular findings were within normal limits. The patient also had several facial deformities on the left side like hemifacial macrosomia figure 2.



Fig 2: Hemifacial Macrosomia Of Left Side With An Underdeveloped Maxilla And Mandible

With an underdeveloped maxilla and mandible on the left side of the face and unilateral macrostomia, microtia with a preauricular skin tag associated with decreased hearing on the same side, figure 3.



Fig 3: Microtia With A Preauricular Skin Tag On The Left Side

A slightly deviated nasal septum, a high palatal vault, gingival hypertrophy and abnormal dental development with a defective speech. The patient also had kypho-scoliosis although there were no complaints suggestive of lung function compromise.

The patient was referred to the department of pediatrics of our facility for further evaluation of systemic features of the syndrome which were found to be within normal limits.

On axial Computerized Tomographic Scan of face absence of condyle and glenoid fossa with hypoplastic ramus and coronoid process of left side. There was evidence of subclinical bony cleft palate of right side and atretic bony external auditory canal and middle ear cavity figure 4.

Based on the clinical and radiological findings a diagnosis of Goldenhar syndrome was made.

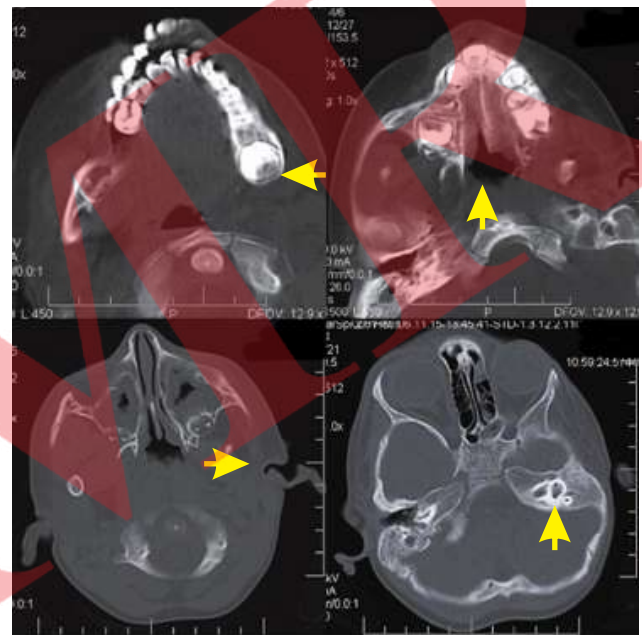


fig 4: Axial CT Scan Of Face Showing Hypoplastic Ramus And Coronoid Process Of The Left Side, Bony Cleft Palate Of The Right, Underdeveloped Maxilla And Bony Ear Cavities Of The Left Side

DISCUSSION

The incidence of Goldenhar syndrome vary between 1:3500 to 1:5600 live births with a male to female ratio of 3:2 (2).

The exact cause of Goldenhar syndrome is unknown but considered to be multifactorial, i.e. a combination of gene interactions and environmental factors that causes a mal-development of the first and second branchial arches during the first trimester of pregnancy.

Goldenhar syndrome is usually sporadic but can be familial having an autosomal dominant or autosomal recessive inheritance. The causative teratogenic factors include smoking, cocaine use during pregnancy; diabetic embryopathy, primidone, retinoic acid, or thalidomide use during pregnancy (3).

Symptoms of Goldenhar syndrome can vary, but may include one or more of these features (4).

A host of craniofacial abnormalities including hemifacialmicrosomia, particularly in the area affecting the ear, mouth and jaw areas (prominent forehead, hypoplasia of the zygomatic area, and maxillar and mandibular hypoplasia) is found. Preauricular skin tags or blind fistulas, microtia, or other external ear malformations (dysplasias, asymmetries, aplasias, and atresias of the external meatus), middle and internal ear anomalies, unilateral macrostomia, cleft lip or cleft palate, tongue cleft, unilateral tonguehypoplasia, and parotid gland aplasia are commonly seen. In the neck, branchial cartilage, branchial fistula, webbing, short neck, abnormalities of sternocleidomastoid muscle can also be found.

Ocular symptoms include epidermoid tumors which occur in 35 % of all cases and can be unilateral (50%) and bilateral (25%). They can occur at any location on the globe or in the orbit and can be dermoid, lipodermoid(25%), or dermis-like or complex. Other features include unilateral or bilateral blepharoptosis, elevated orbit, clinical anophthalmia or microphthalmia, retinal abnormalities like tilted optic disc, colobomas of the upper eyelid, iris, choroid and retina, ocular motility disorders (esotropia, exotropia, duane syndrome), microphthalmia, anophthalmia, cataract, amblyopia, antimongoloid obliquity of palpebral fissures, microcornea and congenital cystic eye.

Vertebral anomalies seen in Goldenhar syndrome are scoliosis, kyphosis or both, abnormal rib structure including missing or fused ribs, thoracic insufficiency syndrome due to diminished lung function.

The systemic features include Cardiac defects, Respiratory issues, Kidney and urogenital issues, Central nervous system defects.

The present case also had the classical features such as facial asymmetry, microtia with preauricular tags and hypoplasia of the left mandibular ramus, and condylar and coronoid processes resulting in facial asymmetry affecting the left side, thus leading to a diagnosis of Goldenhar syndrome.

The most common ocular finding which is considered to be a hallmark of Goldenhar syndrome is Epibulbar dermoid which occur in almost 60% of the cases. Interestingly it was absent in our case which instead

presented with an eyelid coloboma and tilted disc while other ocular findings were within normal limits.

Several reports of eyelid colobomas have been reported in Goldenhar syndrome but they have mostly been found to be associated with an epibulbar dermoid (2,5-6).

CONCLUSION

Goldenhar Syndrome presents with multiple craniofacial and vertebral defects and there is no specific criteria for the diagnosis. Dermoids have been considered to be the most common and typical ocular feature of Goldenhar Syndrome, but as seen in our case this may not always be true. Hence, it is important to consider the possibility of Goldenhar Syndrome even in the absence of the pathognomic ocular features of the syndrome and thorough ocular and systemic examination should be carried out before ruling out Goldenhar Syndrome.

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