Case Report

Varied Ocular Manifestations of Goldenhar Syndrome

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INTRODUCTION

Goldenhar syndrome is a rare congenital anomaly involving the first and second branchial arches. This syndrome was first described in 1952 by the French ophthalmologist Maurice Goldenhar.[1] It is a disorder where the patient’s facial features are incompletely developed on one side resulting in eye, ear, and jaw abnormalities. The syndrome is bilateral in 10%–33% of the patients. 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 unknown. Ingestion of drugs such as thalidomide, retinoic acid, and tamoxifen during pregnancy may be related to the development of this syndrome. Maternal diabetes, rubella, and influenza have also been suggested as the etiologic factors.[3]

The classic features of this syndrome include ocular changes such as, limbal dermoids, lid coloboma, and microphthalmia. Auricular features include preauricular skin tag, hearing loss, and microtia. Vertebral anomalies such as scoliosis, hemivertebrae, and cervical fusion may also be seen.

Goldenhar syndrome, also known as oculo-auroiculo-vertebral syndrome, is a rare congenital disorder due to defect in the development of the first and second branchial arches during blastogenesis. This syndrome consists of epibulbar dermoids, eyelid coloboma, auricular abnormalities, preauricular appendages, and mandibular hypoplasia. There can also be associated cleft palate, malocclusion of teeth, and involvement of the axial skeleton (vertebrae and ribs). Congenital heart disease and anomalies of the genitourinary system are some rare manifestations of this syndrome. We report two cases of Goldenhar syndrome with varied ocular manifestations. The first patient was an 18-year-old female who presented with an inferotemporal limbal dermoid with protruding hair involving the inferotemporal cornea. She also had external ear malformation, preauricular skin tag, and mandibular hypoplasia. The second patient was a 3-year-old boy with low intelligence who presented with an upper eyelid coloboma and a temporal limbal dermoid in the left eye with telecanthus and epicanthal folds. There were associated mandibular hypoplasia and preauricular skin tag. Both the patients did not have any vertebral anomaly or congenital heart defect.

KEYWORDS: Eyelid coloboma, Goldenhar syndrome, limbal dermoid

Tetralogy of Fallot and ventricular septal defects are the most common congenital heart diseases associated with oculo-auroiculo-vertebral syndrome (OAVS).[3] Cleft lip and palate, macrostomia, tracheoesophageal fistula, umbilical hernia, inguinal hernia, urologic anomalies, hypoplastic vagina, and anal anomalies are some rare associations of the disease.[1]

CASE REPORTS

Case 1

An 18-year-old female presented to the ophthalmology outpatient department (OPD) with complaints of a fleshy mass in the left eye since birth. She was born of nonconsanguineous marriage between her parents with no maternal history of any illness or drug intake during pregnancy. On clinical examination, there was left-sided facial hypoplasia [Figure 1a]. She had a 3 cm × 2 cm...
limbal dermoid with protruding hair in the inferotemporal quadrant involving 2 mm of the peripheral cornea from 3 to 6 o’clock position in the left eye [Figure 1b]. There was +2.25 D of against-the-rule astigmatism in the left eye. Her best-corrected visual acuity was 6/6 in both eyes. Fundus of both eyes was within normal limits. There was right ear malformation with preauricular appendage [Figure 1c]. Malocclusion of the teeth was noted. There were no vertebral anomalies. Based on the clinical findings, a diagnosis of Goldenhar syndrome was made. She was prescribed convex glasses and given carboxymethyl cellulose eye drops to prevent exposure keratopathy. The patient was not willing for excision of the limbal dermoid.

Case 2
A 3-year-old boy with subnormal intelligence was referred from the pediatric OPD with the presence of abnormal left upper eyelid. Antenatal and perinatal histories were uneventful. Clinical examination revealed left-sided facial hypoplasia [Figure 2a]. An upper eyelid coloboma was noted at the junction of the medial 1/3rd and lateral 2/3rd in the left eye. A 1 cm × 1 cm temporal limbal dermoid was present in the left eye [Figure 2b]. Telecanthus and epicanthal folds were noted in both eyes. Dilated cycloplegic refraction with atropine revealed a +1.0 D of against-the-rule astigmatism in the left eye. Fundoscopy was normal in both eyes. A broad nose with a small cutaneous sinus was noted. Microtia of the left ear with preauricular skin tag was present [Figure 2c]. Skeletal and cardiovascular system examination was normal. Based on the clinical findings, a diagnosis of Goldenhar syndrome was made. The patient was prescribed glasses, given carboxymethyl cellulose eye drops, and is on regular follow-up.

Discussion
Goldenhar described a patient with a triad of accessory tragus, mandibular hypoplasia, and limbal dermoids and called this constellation of features, the Goldenhar syndrome. Gorlin named this syndrome, oculo-auriculo-vertebral dysplasia, due to the presence of additional vertebral anomalies. The incidence of Goldenhar syndrome has been reported to be between 1:3500 and 1:5600, with a male: female ratio of 3:2. The exact etiology is not known. However, it is possible that the abnormal embryonic vascular supply, disrupted mesodermal migration, or some other factor leads to abnormal formation of the branchial arches and vertebral systems.

The main ocular feature of Goldenhar syndrome is epibulbar choristoma. It consists of a dermoid or a lipodermoid located on the inferotemporal or superotemporal part of the limbus. Corneal invasion by tumors is rare and leads to against-the-rule astigmatism. Other ophthalmological symptoms have also been reported such as eyelid coloboma, anophthalmia, microphthalmia, motility disorders, strabismus, blepharoptosis, coloboma of the iris or choroid, iris atrophy, polar cataract, anomalies of the lacrimal drainage system, and retinal and optic nerve anomalies.
Exposure keratopathy may be seen due to the presence of dermoid or eyelid coloboma.\[4]\n
The treatment of the disease varies with age and systemic associations. Ocular treatment includes early correction of refractive errors to prevent the development of amblyopia later in life. Epibulbar dermoids are classified into three grades according to the size and degree of the involvement of cornea. Grade I limbal or epibulbar dermoids are lesions with a size <5 mm. Grade II limbal dermoids are larger in size and extend into the corneal stroma down to the Descemet’s membrane. Grade III limbal dermoids involve the entire cornea and structures of the anterior chamber. Medical management is generally reserved for Grade I dermoids inducing only mild astigmatism of <1 D. In the presence of irregular astigmatism or if the patient is not compliant with wearing of spectacles, surgical excision is indicated. Surgery is indicated for Grade II and III limbal dermoids. Simple excision is usually sufficient for Grade I limbal dermoid. Grade II dermoid is treated by keratectomy followed by amniotic membrane transplantation and limbal stem cell allograft. Grade III dermoids need complete anterior segment reconstruction.\[5]\n
Treatment is mainly cosmetic in uncomplicated cases. Corneal protection and cosmesis are common indications for repairing eyelid coloboma, and its treatment depends on the size of defect.\[6]\n
Reconstruction surgeries of the external ear can be done at the age of 6–8 years. Jaw reconstruction surgeries can also be done in patients with milder disease.\[2]\n
The most frequently reported ear anomalies in OA VS are external and middle ear abnormalities. Thus, secondary conductive hearing loss predominates in them. Bilateral profound hearing loss is rare in these patients. These hearing losses may cause impaired language and speech development. Hearing loss is generally unilateral in OA VS, and the level of hearing in patients enables them to be socially active. However, patients with bilateral mixed profound hearing loss or pure sensorineural hearing loss require more advanced forms of treatment, such as cochlear implants or bone-anchored hearing aids. Prognosis of the disease is good without any systemic associations. Life-threatening anomalies should be assessed and treated in infancy followed by any necessary ophthalmologic intervention, vertebral corrections, auricular reconstructions, and jaw surgeries. Patients with Goldenhar syndrome require an early diagnosis, a skilled multidisciplinary approach to treatment, and counseling for the best chance of a successful long-term outcome.

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/ her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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**Conflicts of interest**

There are no conflicts of interest.

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