Goldenhar syndrome presenting as limbal-epibulbar Lipodermoid tumour: A Case Report

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ABSTRACT
Goldenhar syndrome is a birth defect resulting from the maldevelopment of the first two brachial arches with incomplete development of the ear, nose, soft palate, lip and mandible. The phenotype is highly variable. It is also known as oculo-auriculo-vertebral (OAV) dysplasia. It is a rare condition characterized by triad of craniofacial microsomia, ocular dermoid or lipodermoid tumour, and spine anomalies. Here we report a case of Goldenhar syndrome in a 04-year-old Indian male with right eye limbal-epibulbar lipodermoid tumour with upper lid coloboma, with right side microtia with delayed development of speech and hearing with mandibular hypoplasia. Multidisciplinary treatment approach was advised; lipodermoid tumour was excised surgically and Paediatrician and ENT consultation was advised regularly. This case has been presented to increase the awareness about this rare entity, to highlight the importance of typical clinical and radiological findings and its association with othersystemic conditions and its treatment by surgical intervention.

Key words: Goldenhar, limbal-epibulbar lipodermoid, microtia, Mandibular hypoplasia.

INTRODUCTION
Goldenhar syndrome is a rare congenital disorder with occurrence of about 1 per 5800 birth and male: female ratio is 3:2. It is usually unilateral and the age of presentation is usually during neonatal & infancy. In 1952, Goldenhar was first to describe this rare disorder. Goldenhar syndrome is also known as oculo-auriculo-vertebral dysplasia. The abnormalities in organs developing from first and second brachial arches during blastogenesis causes morphological anomalies. It has a multifactorial etiopathology that include nutritional and environmental factors. Ocular anomalies occur in about 50% cases. Epibulbar dermoid and lipodermoid are the most common. Coloboma of upper lid may be present. Additional features include pre-auricular skin tag, microtia, anotia and vertebral and dental anomalies. Other systemic conditions such as cardiac, genito-urinary and pulmonary anomalies may be associated in rare cases.

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CASE REPORT
History
A 4-year-old male child reported at the ophthalmology department of SSG Hospital Vadodara on with complains of defect in upper lid and swelling in the lower and outer part of right cornea associated with absence of pinna on same side and hemi facial asymmetry. There was evidence of developmental delay in speech and hearing. The swelling in inferotemporal region of right eye was since birth which was painless and gradually increasing in size. There was no redness or discharge from eye. These symptoms were consistent with the diagnosis of Goldenhar syndrome. The child was born to non-consanguineous parents with uneventful prenatal and antenatal period. There was no relevant family history.

Examination
The patient was examined thoroughly. Ocular examination revealed a small soft swelling measuring about 3.5 x 2.5 cm near limbal-epibulbar area locating in inferotemporal region (Figure 1). Swelling was not obscuring the visual axis. Coloboma of right eye upper lid was also present in the form of notch (Figure 1). Patient was not much co-operative for distant vision but follows lights and objects easily. Patient having central steady fixation in each eye.
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independently. Both eyes pupils were reacting to light normally and funduscopic examination was found normal. The other eye was clinically normal (Figure 1).

**Figure 1 : RE showing limbal epibulbar lipodermoid with upper lid coloboma**

ENT examination revealed microtia in right side (Figure2) with hearing problems and bilateral enlarged tonsils and pharyngeal wall mucosal laxity with bilateral mobile vocal cords. Systemic examination like cardiovascular, pulmonary, and genito-urinary systems was done by paediatrician but no abnormalities were detected.

**Investigations**

The laboratory investigations were within normal limits. Electrocardiography, X-Ray chest and vertebral column were normal. CT SCAN head and neck (Figure 3&4) showed hypoplasia of right mandibular ramus with right submandibular gland appearing cranially displaced with obliteration of right Para pharyngeal space. Evidence of microtia is seen on right side with rudimentary pinna with congenital agenesis of right external auditory canal and hypoplasia of internal auditory canal and semi-circular canals.(Figure 3)

**Management**

Complete excision of lipodermoid tumour was done (figure5). The patient was operated under general anaesthesia. The lesion was dissected off the cornea with meticulous attention for preservation of normal tissue. A cleavage plane was fashioned, and the dermoid was removed from the cornea first, with movement towards the limbus and conjunctival side. Removed section was sent for histopathology which showed lipodermoid on microscopic examination. The exposed sclera was covered by conjunctival autograft taken from superotemporal region. The upper lid coloboma was also surgically repaired. The result was satisfactory without any complications and the postoperative course was uneventful. (Figure 5)
DISCUSSION
Goldenhar syndrome is known as oculo-auriculo-vertebral dysplasia. It is proposed to represent a variant of hemi facial microsomia group. It includes hemifacial hypoplasia, and first and second arch syndrome. The involvement is unilateral in 70%-80% cases. Sporadic, autosomal dominant and autosomal recessive modes of inheritance have been seen in most of cases along with association of Trisomy of 7 and 22. Goldenhar syndrome has been also found in children born to pregnant women who were exposed to various teratogenic agents like retinoic acid, primidone and thalidomide. Ocular manifestations are limbal dermoid (more common) or lipodermoid and occasional coloboma of the upper lid. Lipodermoid is usually present in inferotemporal quadrant and can be bilateral in 25% cases. There are two types of limbal lipodermoid—large and small. The larger one interfere with the visual axis causing astigmatism and predisposing to secondary strabismus from anisometropic amblyopia. Other associations are Duane retraction syndrome, cataract and iris abnormalities. Ear tags are common. Inner ear anomalies can occur in some cases. The central nervous system is occasionally affected. Hemi facial asymmetry and vertebral anomalies are common which include kyphosis, scoliosis and lumbosacral lordosis. Other findings include microtia, microsomia and mandibular anomalies. The clinical diagnosis is based on the obvious clinical finding and other laboratory and radiological findings. The most common complaints are swelling in eye, preauricular tags, difficulty in hearing, difficulty in opening of the mouth and difficulty in walking. The treatment of disease varies according to severity of manifestations. With regard to the rule of ophthalmology, it is aimed first to prevent amblyopia due to obstruction of visual axis, second at ocular exposure due to large coloboma or large limbal dermoid or lipodermoid preventing lid closure and third at working with craniofacial surgeon in case of severe muscular weakness that require reconstruction of upper face. Systemic treatment may be related for cardio renal or CNS malformation. Surgical treatment of condition related to large coloboma require surgical repair, large limbal dermoid needs excision of dermoid with lamellar keratoplasty. Severe anomalies of the mandible require reconstruction with bone graft. In case of microtia or other ear defect extensive ear reconstruction to be done within 6-8 year of age if the facial or congenital malformation are severe then speech therapy required. In this particular case anomalies of eye corrected by surgical intervention without any complications. Because small area of cornea was involved and deep excision was not performed, so lamellar keratoplasty was not done.

CONCLUSION
Goldenhar syndrome is a rare congenital abnormality associated with cosmetically unacceptable defects whose management may pose numerous challenges and requires multistage and multi-disciplinary approach for its management. Prognosis of this disease is good in otherwise uncomplicated cases without any systemic associations. Ocular dermoid or lipodermoid tumour can be excised surgically. The ophthalmologist should focus on visual consequence, early treatment and meticulous follow up of patient. Reconstructive oral and facial surgeries are required for malocclusion and bony deformity. This case has been presented to increase the awareness about this rare entity, to highlight the importance of typical clinical and radiological findings and its possible surgical correction.

REFERENCES
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