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GOLDENHAR SYNDROME- A CASE REPORT

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Abstract

Goldenhar syndrome is a birth defect resulting from the maldevelopment of the first two branchial arches with incomplete development of the ear, nose, soft palate, lip and mandible. The phenotype is highly variable. Goldenhar syndrome is one of the variants of craniofacial anomalies. It is unilateral in 70-80% of the cases. It is known as oculoauriculo vertebral (OAV) dysplasia. The syndrome complex includes limbal dermoid or lipodermoid, pre-auricular tags, hemifacial asymmetry and vertebral anomalies. These are the common anomalies of the condition. It is a rare condition characterized by the triad of craninofacial microsomia, ocular dermal cyst and spine anomalies'. It is usually unilateral and the age of onset is usually during neonatal & infancy. The purpose of this article is to report a rare case of craniofacial anomalies and its management.

Key words: Goldenhars syndrome, limbal dermoid, epibulbar dermoid

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Introduction:

Goldenhar syndrome is a birth defect resulting from the maldevelopment of the first two branchial arches with incomplete development of the ear, nose, soft palate, lip and mandible. The phenotype is highly variable[1]. Goldenhar Syndrome is one of the variants of craniofacial anomalies. It is unilateral in 70-80% of the cases[2] . Prevalence rate is 1-9/per 100000 [3,4] and incidence rate is 1 per 25000-45000 births[5]. Male is more commonly affected than the female (ratio 2:1). Most of the cases of OAV are sporadic, autosomal dominant transmission is reported for 1% - 2% of the cases. A few cases with autosomal recessive inheritance have been reported. Aetiology of the syndrome remains unclear. Currently a deficiency in mesodermal formation or defective interaction between neural crest and mesoderm is suggested as possible aetiology. Different factors also contribute to the development of the disease such as: ingestion of some drugs (Cocaine, Thalidomide, Retinoic acid and temoxifen), environmental factors (Insecticides, Herbicides) and maternal diabetes [6]. Ocular anomalies occur in about 50% of the cases of OAV[6]. Epibulbar dermoid and lipodermoid are the most common. Coloboma of the upper eyelid may be present. Limbal dermoid or lipodermoid are mainly located in the inferotemporal region of the eye. Ocular defects are reported in 65% of the cases and include pre-auricular tags, microtia, anotia and

conductive hearing loss. Vertebral anomalies are combination of hemivertebra, fused ribs, kyphosis and scoliosis. Additional features [7,8] such as cardiac, genito-urinary and pulmonary systems can also be affected.

Case report:

An 8 years old girl reported at the Ophthalmology department of MIMSR medical college, Latur Maharashtra, on 03rd may 2013 with complains of swelling of the lower and outer part of the both eyes associated with preauricular tags, hemifacial asymmetry and abnormalities in the spine in the form of scoliosis. The swelling in the inferotemporal region of the both eyes was gradually increasing and causing obstruction of the visual axis. Coloboma of left eye upper lid was present in the form of a notch. These symptoms were consistent with the diagnosis of Goldenhar syndrome.

The patient was examined thoroughly. Ocular examination revealed a small soft mass of the left eye locating in the inferotemporal region obscuring the visual axis. But her visual acuity was 6/6 in both eyes. Fundoscopic examination was found normal. ENT examination revealed preauricular tags present in the left ear. No other abnormalities were detected. Systemic examination like cardiovascular, pulmonary and genito-urinary systems was done but no abnormalities were detected. The laboratory investigations were within normal limits. Electrocardiography was normal; X-ray chest (postero anterior view) showed nothing abnormality detected, while X-ray of the vertebral column showed scoliosis. Complete excision of the dermoid cyst was achieved. The patient was operated under general anaesthesia for his visual and auricular anomalies. The result was satisfactory without any complications. The postoperative course was uneventful and the child was transferred to Paediatrics department for multidisciplinary management of skeletal abnormalities. Now the patient is cured and leading a normal life.

Discussion:

Goldenhar syndrome is known as oculoauriculo vertebral dysplasia. It is proposed to represent a variant of hemifacial microsomia group. It includes hemifacial hypoplasia, oculoauriculo vertebral dysplasia and first and second arch syndrome. The involvement is unilateral in 70%-80% of cases [2,9]. Ocular manifestations are limbal dermoid or lipodermoid and occasional coloboma of the upper eye lid. Limbal dermoid is more common than lipodermoid. It is usually present in the inferotemporal quadrant and can be bilateral in 25% cases [7]. There are two types of limbal dermoid - large and small. The larger one interferes with the visual axis causing astigmatism and predisposing to secondary strabismus from anisometropic amblyopia. Other associations are Duane Retraction syndrome and lower incidence of decreased corneal sensation; cataract and iris abnormalities[10,12] Ear tags are common. Inner ear anomalies are occurring in some cases. The central nervous system is occasionally affected. Vertebral anomalies are common which includes kyphosis, scoliosis and lumber lordosis. Hemifacial asymmetry is also common. Other findings include microtia, macrosomia and mandibular anomalies. The clinical diagnosis is based on the obvious clinical findings and other laboratory and radiological findings. The most common complaints are swelling in the left eye lid, preauricular tags, difficulty in opening of the mouth and difficulty in walking occasionally. Only spina bifida is detected by skiagram does not significantly affects the child. Treatment of the disease varies according to the severity of the manifestation[13]. With regard to the rule of ophthalmology is aimed first at strong amblyogenic risk causing obstruction of the visual axis, severe astigmatism or strabismus, second at ocular exposure (due to large coloboma or large limbal dermoid preventing lid closure), third at working with craniofacial surgeon in case of severe muscular weakness that requires reconstruction of the upper face. Systemic treatment may be related for cardio-renal or CNS malformation[11]. Surgical treatment of the condition related to large coloboma requires surgical repair and spectacle correction, large limbal dermoid needs excision of the dermoid with lamellar keratoplasty. Severe anomalies of the mandible require reconstruction with bone graft. In

case of microtia or other ear defects needs extensive ear reconstruction to be done within 6-8 years of age[12]. If the facial or congenital malformations are severe speech therapy is required[13]. In this particular case there were anomalies of eye and ear that was corrected by surgical intervention without any complications. Patient is now cured and leading a normal life.

Conclusion:

Goldenhar syndrome is a rare congenital abnormality associated with cosmetically unacceptable defects whose management may pose numerous challenges and requires a multistage and multidisciplinary approach for its optimal management.

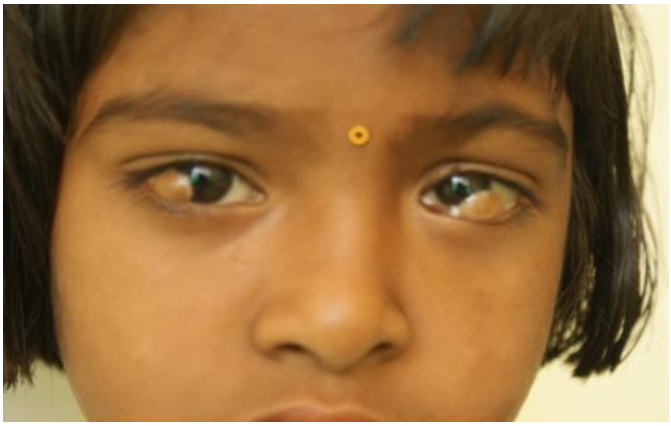

Fig-1: Limbal dermoid

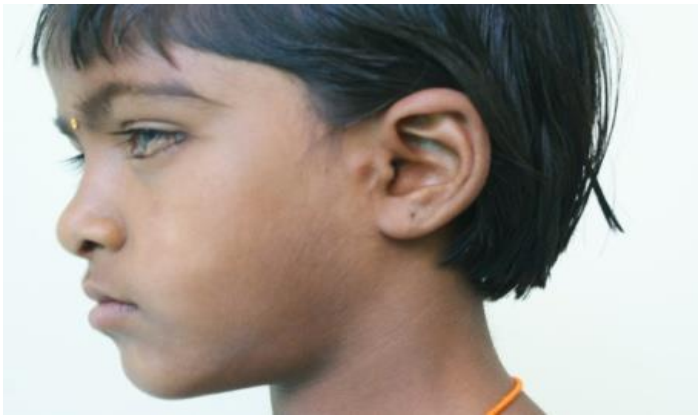

Fig-2: Pre- auricular skin tag

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