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Case Report

Goldenhar syndrome

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ABSTRACT

Goldenhar syndrome is an oculoauriculovertibral dysplasia representing the error in morphogenesis of the first and second brachial arches. Presence of preaxial polydactyly cleft lip preauricular skin tag, dextrocardia and limbal dermoid without lid and iris coloboma prompted us to report this case.

Keywords: Goldenhar syndrome, Oculoauriculovertibral dysplasia, Morphogenesis, Limbal dermoid

CASE REPORT

A 10 month female born of non-consanquineous marriage presented with complaints of whitish cystic swelling in the right eye with abnormal looking face and ears. Child was born of a full term normal delivery. There was no history of exposure neither to known teratogenic drug nor maternal diseases during the pregnancy. All other family members including two other siblings are normal but patient's cousin (Uncle's son) has anophthalmos with cleft palate.

Clinical examination revealed right sided torticollis, facial asymmetry, hypoplasia of mandibles, cleft lip, malformed ears with preauricular tags and preaxial thumb. Cardiovascular system revealed dextrocardia. Skeletal examination showed cervical scoliosis. Cleft lip operated and corrected by plastic surgeon.

Ocular examination showed presence of soft pinkish brown nodule in infero temporal quadrant on the bulbar conjunctiva extending till limbus identified as epibulbar dermoid in right eye. Other ophthalmological symptoms have been reported such as anophthalmos, micophthalmos, coloboma of eyelid iris choroid, polar cataract, anomalies of retina optic nerve and lacrimal drainage system was not present in our case.

Child was having bi oblique astigmatism. Ocular motility and Fundus both eyes normal. Dermoid removed under general anaesthesia. Histopathological report confirmed dermofibroma.

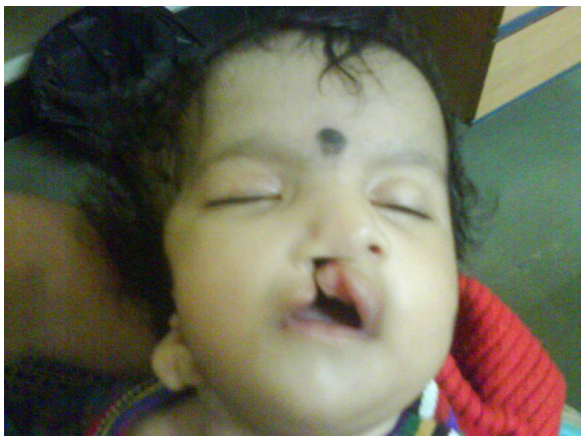

Figure 1: Goldenhar syndrome (Cleft lip with accessory auricle seen).

Figure 2: Post-operative dermoid.

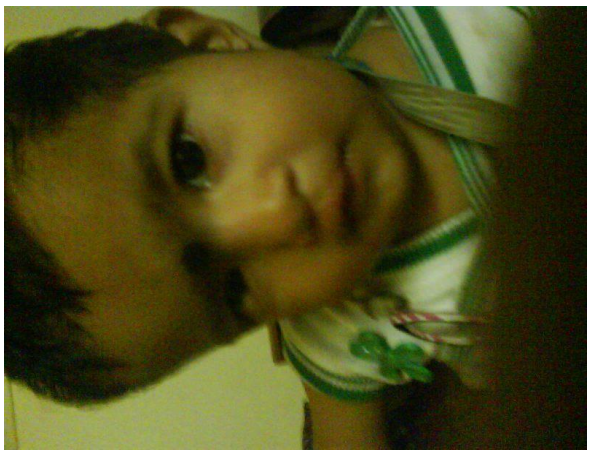

Figure 3: Cleft lip corrected.

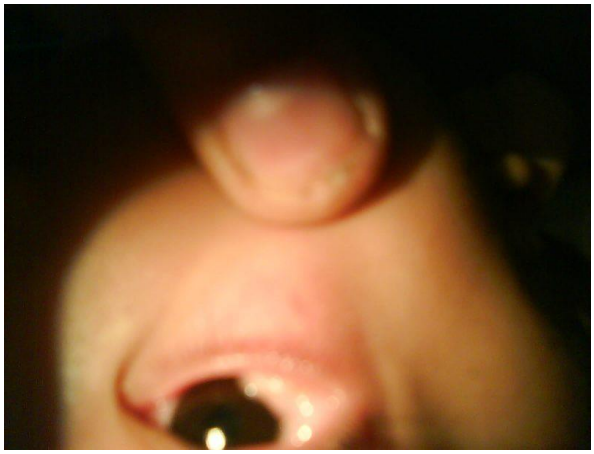

Figure 4: Preoperative dermoid.

DISCUSSION

The Goldenhar syndrome was documented in 1952 by Maurice Goldenhar (1924-2001) an American ophthalmologist. In 1963 Gorlin suggested the name Oculoauricularvertibral syndrome (OAV).

Frequency of occurrence is estimated to be 1:3000 to 1:5000. Males are more affected than female (3:2).[1] It is thought to be multifactorial although there may be genetic component which would account for general familial pattern. Though most of the GS cases are sporadic, both autosomal dominant and autosomal recessive pattern are described. After reviewing literature by Hartfield it was suggested that GS is the result of some type of vascular perturbation during embryogenesis associated with development of the first and second brachial arch.[2,3] In around 25% of cases of GS epidermal dermoid have been observed. Bilateral presence of defect in the organs that is either the organ not present on one side or under developed, incidence is 10%. Reported frequency of cardiovascular malformation of this syndrome is 5-58%.[5-7] Dextrocardia and preaxial polydactyly are reported in one study each.[8,9]

The treatment is usually confined to surgical intervention as may be necessary to help the child to develop e.g. jaw distraction/bone graft ocular dermoid debulking, repairing cleft palate/lip repairing heart malformation spinal surgery, hearing aid.

In our case surgical repair includes removal of dermoid, correction of refractive error.

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Conflict of interest: None declared
Ethical approval: Not required

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