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Case Report

Goldenhar syndrome: a rare entity

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ABSTRACT

Goldenhar syndrome was first described by Maurice Goldenhar in 1952. It involves abnormalities in the first and second branchial arches. This syndrome appears to have varying degree of presentation from mild to severe form. Different pathophysiological mechanisms have been proposed. Oculo auriculo vertebral syndrome which involves reduction in blood flow resulting in focal haemorrhage in the first and second branchial arches. In addition, there may be cardiac, vertebral and CNS defects. A male baby 30-32 wks, preterm SGA was admitted in NICU for evaluation of congenital malformations which includes swelling of the anterior border of both eyes with b/l preauricular ear tags, malformed pinna, high arched palate and micrognathia. Ophthalmological examination revealed b/l limbal dermoid inferotemporal aspect. ECHO done revealed ASD 4mm. CT scan showed obstructive hydrocephalus. Treatment of the condition varies according to the severity. Ophthalmic treatment is aimed first because of amblyogenic risk. Craniofacial surgery in view of severe micrognathia systemic treatment may be related to cardiac, renal and CNS malformations. Goldenhar Syndrome is a rare congenital anomaly, with cosmetic defects whose treatment may pose numerous challenges with multidisciplinary approach for optimal management.

Keywords: Goldenhar syndrome, High arched palate, Malformed pinna, Preauricular tag

INTRODUCTION

Goldenhar syndrome, a type of craniofacial syndrome, is a rare congenital birth defect causes abnormalities in the formation of the face and head.[1] Von Arlt in 1881 reported the first observation of oculo-auriculo-vertebral (OAV) dysplasia and Dr. Maurice Goldenhar, a popular Swiss ophthalmologist in 1952 classified the clinical features and named the abnormality complex as Goldenhar syndrome characterized by epibulbar dermoids, auricular appendages, blind-ended auricular fistulas and vertebral anomalies.[2] The occurrence of Goldenhar syndrome has been reported to be varying from 1 in 3500 to 5600 new borns and presently in 1 in 1000 children with congenital deafness with a male to female ratio of 3:2.[2] Goldenhar syndrome occurs due to complications occurring during development of fetus within the mother's womb, which involves abnormalities in the first and second brachial arch. Goldenhar syndrome may also affect the different organs including heart, lungs, kidneys, and central nervous system.[3] Some reports stated the use of drugs including thalidomide, retinoic acid, tamoxifen, and cocaine at pregnancy may be related to the development of such syndrome. Diseases like diabetes, rubella, and influenza in mother may be considered as etiologic factors.[4]

The etiology is heterogenous. This syndrome which includes reduction in blood flow resulting in focal haemorrhage in the first and second branchial arches. The disease varies from mild to severe form.[4] In 85% of cases defects are unilateral and in 10-33% of the cases are bilateral. The right side is more commonly affected.[2] Studies have reported facial asymmetry in approximately 70% of the cases, while ear anomalies in 83% and eye

malformations in 66%. Ear deformities, ranged from anotia to preauricular tag.[5]

CASE REPORT

A male baby 30-32-weeks, preterm small for gestational age (SGA) infant was admitted in NICU on 22nd October 2016 for evaluation of congenital malformations which includes swelling of the anterior border of both eyes with b/l preauricular ear tags, malformed pinna, high arched palate and micrognathia. Opthalmological examination revealed b/l limbal dermoid in inferotemporal aspect. ECHO done revealed ASD 4 mm. CT scan showed obstructive hydrocephalus.

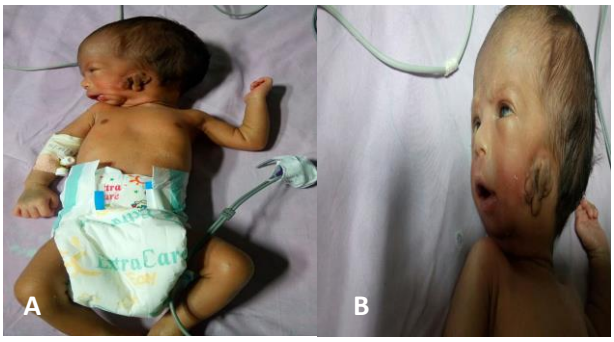

Figure 1 (A and B): Swelling of the anterior border of eyes, preauricular ear tags, malformed pinna, high arched palate and micrognathia ophthalmology.

DISCUSSION

Goldenhar syndrome or facio-auriculo-vertebral dysplasia or oculo-auriculo-vertebral syndrome, is an autosomal dominant inherited genetic sporadic condition characterized by hemi-facial microsomia, epibulbar dermoid, defect of ears with pre auricular appendage, coloboma of upper "eye lid," limb dermoids, cardiac abnormalities and hemi vertebra in cervical region.[5,6] This syndrome is accompanied with abnormal growth of the first and second branchial arches during blastogenesis.[6] Ocular manifestation are limbal dermiods and lipo dermoid, limbal dermoid is more common and can interfere with visual axis causing astigmatism, secondary strabismus from anisometric amblyopia. Ear tags are common. Other findings include microtia and mandibular abnormalities.

Treacher-Collins syndrome (TCS) is a similar type of syndrome having similar features like abnormalities in the first and second branchial arches during development of embryo. Distinguishing features amongst Goldenhar syndrome and TCS include facial asymmetry and hypoplasia of the malar bones.[5,7] Other syndromes associated with multiple pre-auricular tragi include Nager's acrofacial dysostosis, Wildervanck syndrome (cervicooculoacoustic syndrome), Wolf-Hirschhorn syndrome, Townes-Brocks syndrome and Delleman syndrome.[4]

Clinical diagnosis is based on obvious clinical findings and other laboratory and radiological findings. The evaluation includes identification; prenatal, perinatal, postnatal and complete family history, followed by complete general and systemic scrutiny of the external ear and facial features.

Speech audiometry, pure tone audiometry, tympanometry, stapedius reflex measurement, and brainstem evoked response are some audiological assessments to be performed when needed. Other related tests which are performed include X-rays of skull, facial, upper limbs, and spine. 2D ECHO for cardiac abnormalities; temporal bone computed tomography (CT) scan, CT spine, magnetic resonance imaging as per the condition of the patients. Patient is followed up.

The management varies with age and systemic associations according to the severity and is mostly by cosmetic surgery in uncomplicated cases. Reconstruction surgeries of the external ear may be done at the age of 6-8 years. Craniofacial surgery in view of severe micrognathia systemic treatment may be related to cardiac, renal and CNS malformations. In case of microtia, reconstructive surgery may be done at 6 to 7 years of age. In patients having milder deformities, reconstruction surgeries of jaws can be performed in the early ages; epibulbar dermoids should be surgically excised.[4]

In mildly affected patients of 2-4 years of age, no treatment is necessary. If patient is severely affected and having underdeveloped mandible, then enlargement of underdeveloped mandible is recommended by a bone distraction device and teeth modifications can be done with the assistance from orthodontics. In 6-8 years of age, the surgery of the external ear is done over the period of 6-12 months. Plastic surgery is recommended for modification of structural abnormalities of the eyes and ears.

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