Journal of Bangladesh College of Physicians and Surgeons
Vol. 28, No. 3, September 2010

Goldenhar Syndrome-A Case Report

MAR SIDDIQUE[a], J HOSSAIN[b], MJ ABEDIN[c], M PARVEZ[d]

Summary:

A 7 years old boy was diagnosed a case of Goldenhar Syndrome. He presented with swelling in the upper and outer part of the left eye as limbal dermoid associated with preauricular tags, hemifacial asymmetry, microtia and small chin since birth. His vertebral anomalies also detected by skiagram of the vertebral column as spina bifida. His ocular and auricular problems were solved by surgery without any complicatons. Patient is leading a normal life.

(J Bangladesh Coll Phys Surg 2010; 28: 193-195)

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 Varriants of craniofacial anomalies. It is unilateral in 70-80% of the cases [2]. 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 commmon anomalies of the condition. It is a rare condition characterized by the triad (usually unilateral) of craninofacial microsomia, ocular dermal cyst and spine anomalis. Age of onset during neonatal & infancy. Prevalence rate is in 1-9/100000 [3,4], incidence rate is 1 in 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 [5]. A few person manifested with autosomal recessive inheritance has been reported. Aetiology of the syndrome remains unclear. Currently a deficiency in mesodermal formation or defective interaction between neural crest or mesoderm is suggested as possible aetiology[5]. Different factors also contributed 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 about 50% of the case 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 & conductive hearing loss. Vertebral anomalies are combination of hemivertebra, fused ribs, kyphosis and scoliosis. Additional features [7,8] - cardiac, genito-urinary and pulmonary systems can also be affected. Cardio pulmonary distress within the few months of life is relatively common life threatening complication. The purpose of this article is to report a rare case of craniofacial anomalies and manage satisfactorily.

Case Report:

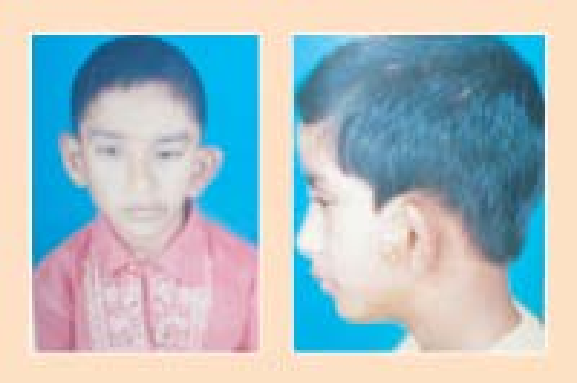
A 7 years old boy reported to BNS Patenga a Naval Hospital of Bangladesh Navy at Chittagong on 07 july 2008 with complains of swelling of the upper and outer part of the left eye associated with preauricular tags, hemifacial asymmetry, microtia, small chin and abnormalities in the spine. The swelling in the inferotemporal region of the left eye was gradually increasing and causing obstruction of the visual axis by the drooping of the eye lid. The patient was examined thoroughly. Ocular examination revealed a small soft mass of the left eye locating in the ineferotemporal region obscuring the visual axis. But his visual acuity was 6/6 in both eyes. Fundoscopic examination was found normal. ENT examination revealed preauricular tags present in the left ear and small ear present on the left side. No other abnormalities are detected. Systemic examination like cardiovascular, pulmonary and genito-urinary systems are done but no abnormalities are detected. CNS examination showed slow mental uptake. The laboratory investigations are within normal limit. ECG - normal, X-ray chest (postero anterior view) showed nothing abnormality detected, X-ray of the vertebral column showed spina bifida. The patient operated under GA for his visual and auricular anomalies. The result was satisfactory without any complications or no uneventful occurrence happened. Now the patient is cured and leading a normal life.

a. DR MAR Siddique
b. Dr. Colonel Jakaria Hossain, MBBS, DO (DU), FCPS (Oph), CMH Chittagong Cantonment, Chittagong
c. Dr. Mohd Jainal Abedin, MBBS, DO, FCPS (Oph), Chittagong Medical College Hospital, Chittagong
d. Dr. Masud Parvez, MBBS, MS (Oph), FCPS (Oph), Chittagong Medical College Hospital, Chittagong

Address of Correspondence: Dr. Lt. Col. Md. Abdur Rouf Siddique, Eye Spl and Surgeon, Dept. of Ophthalmology, BNS Patenga, Chittagong.

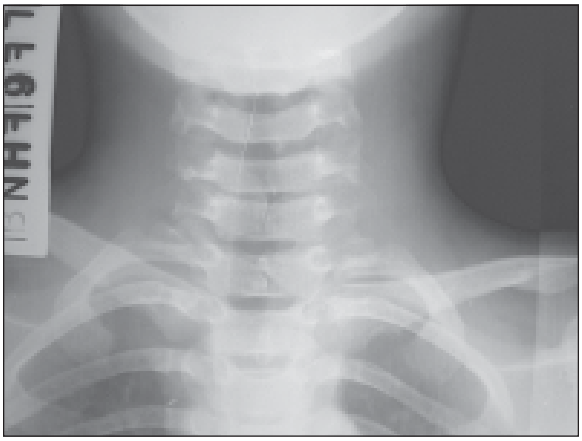
Received: 14 December, 2008 **Accepted:** 5 May, 2009

Photograph of the Patient

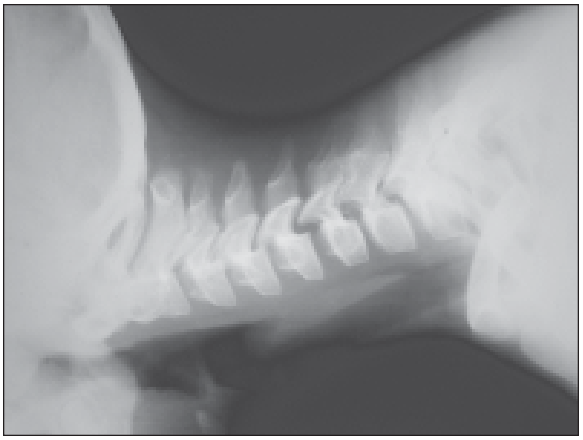


Photograph(Before operation): 1 Showed Limbar dermoid (Left eye & Preauricular tags (Left ear)

Photograph (After operation): 2- Showed exsion of Limbar dermoid (left eye) with lamellar keratoplasty and exsion of Preauricular tags (left ear).



Skiagram of the vertebral coulmn (Cervical Spine) (AP view)- 3 showed spina bifida involving C6- T1 Vertebrae.



Skiagram of the vertebral coulmn (Cervical Spine) (Lateral view)- 4 showed spina bifida involving C6-T1 Vertebrae.

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 manifestation 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 2 types of limbal dermoid - large & 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 occur in some cases. The central nervous system are 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 of swelling in the left eye lid, preauricular tags, difficulty in opening of the mouth and difficulty in walking occasionally. The most common findings are limbal dermoid or epibulbar dermoid in the upper and outer

part of the left eye. Other includes preauricular tags, microtia and hemifacial asymmetry. Vertebral anomalies are not obvious in this particular case. X-ray of the vertebral column is done to exclude vertebral anomalies. Only spina bifida is detected by skiagram which is 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 requires 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 malformation are severe speech therapy is required [13]. In this particular case there is anomalies of eye and ear that was corrected by surgical intervention without any comlications. Patient is now cured and leading a normal life.

References:

1. Jack J Kanaski, Clinical Ophthalmology, 5th edition, Butterworths London, 2005: Page 92, 607.
2. Basic AAO and clinical science course, 2003-2004, Section-6, Page 391-92
3. Bayraktar S, Bayraktar ST, Ataoglu E, Ayaz A, Elveli M. Goldenhar's syndrome associated with multiple congenital abnormalites. J Trop Pediatr. 2005;51(6):377-9.
4. Beck AE, hudgins L, Hoyme HE. Autosomal dominant microtia and ocular coloboma: new syndrome or an extension of the oculo-auriculo-vertebral spectrum? Am J Med Genet A. 2005;1;134(4):359-62.
5. Cohen J, Schanen NC. Branchial cleft anomaly, congenital heart disease, and biliary atresia: Goldenhar complex or Lambert syndrome? Genet Couns. 2000;11(2):153-6.
6. Gorlin RJ, et al. Oculo-auriculovertebral dysplasia. J Pediatr. 1963;63:991-999.
7. Goldenhar M. Associations malformatives de l'oeil et l'oreille, en particulier le syndrome dermoide epibulbaire-appendices auriculaires-fistula auris congenital et ses relations avec la dysostose mandibulo-fistula auris congenita et ses relations avec la dysostose mandibulo-faciale. J Genet Hum 1952;1:243-282.
8. "Oculoauriculovertebral Dysplasis." Online Mendelian inheritance in Man. WWW. ncbi.nlm.nih.gov/entrez/entrez/dispomim.cgi?id=164210.
9. Schaefer, Bradley G, Olney A, Kolodziej P. Oculoauriculo-vertebral Spectrum. ENT-Ear, Nose & throat Joumal. 1998;77:17-18.
10. Singer SL, Haan E, Slee J, Goldblatt J. Familial hemifacial microsomia due to autosomal dominant inheritance. Case reports. Aust Dent J. 1994;39(5):287-91.
11. Stoll C, Viville B, Treisser A, Gasser B. A family with dominant oculoauriculovertebral spectrum. Am J Med Genert. 1998;78(4):345-9.
12. Tasse C, hohringer S, Fisher S, Ludecke HJ, Albrecht B, et al. Oculo-auriculo-vertebral spectrum (OAVS): clinical evaluation and severity scoring of 53 patients and proposal for a new classification. Eur J Med Genet. 2005;48(4):397-411.
13. Tewfik TL, Alnoury Kl;Manifestations of Craniofacial Syndromes; eMedicine, October 2008.