Case Report

Goldenhar Syndrome: A Rare Case Report.

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ABSTRACT:

Goldenhar syndrome or oculo-auriculo-vertebral syndrome is a sporadic or autosomal dominant, inherited genetic rare syndrome that includes limbal dermoid or lipodermoid, pre-auricular tags, hemifacial asymmetry and vertebral anomalies. The phenotype is highly variable. Goldenhar syndrome is one of the variants of craniofacial anomalies. The etiology of this rare disease is not fully understood, as it has shown itself variable genetically and of unclear causes. It is usually unilateral and the age of onset is usually during neonatal & infancy. The purpose of this article is to report this rare syndrome of craniofacial malformations, which may aware physicians to make its diagnosis.

Key words: *Goldenhar syndrome, oculo-auriculo-vertebral syndrome*

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INTRODUCTION:

Goldenhar syndrome is also known as facio-auriculovertebral spectrum (FAV) or oculo-auriculovertebral (OAV) spectrum is a congenital defect characterized by malformations classically involving face, eyes and ears. This condition was first described by Maurice Goldenhar in 1952. Goldenhar Syndrome is one of the variants of craniofacial anomalies. It is unilateral in 70-80% of the cases.[1]

Goldenhar syndrome is a birth defect resulting from the mal-development of the first two branchial arches with incomplete development of the ear, nose, soft palate, lip and mandible. The phenotype is highly variable.[2]

Reported incidence of this syndrome is 1:3500 to 1:5600 with a male to female ratio of 3:2,[3] although most cases are sporadic, autosomal dominance inheritance has also been described. There does not seem to be any geographic or racial predilection.[4]

Aetiology of the syndrome remains unclear. It is a multifactorial disease that includes genetic, nutritional and environmental factors. Chromosomal abnormalities and disturbance of the neural crest cell are suggested as possible aetiologies.[5,6] Some teratogens such as cocaine, thalidomide, retinoic acid, and tamoxifen taken by the mother during pregnancy were also related to the development of the disease.[7] Maternal diabetes has also been suggested as an etiologic factor.[8] The MSX homebox genes also play a crucial role in the differentiation of first branchial arch.[9]

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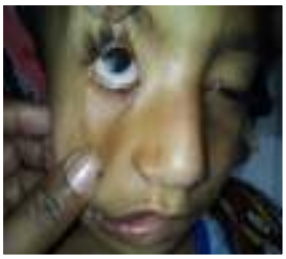
Therefore, this study was taken up on children in whom clinical features prompted us to make a diagnosis of Goldenhar syndrome and plan for its subsequent management.

Case record: A 5 years old male presented in the Pediatric OPD, of 250 Beded General Hospital Tangail, with speech and hearing difficulties with common cold. He was born to non-consanguineous parents with low birth weight at home. Antenatal, natal, post-natal history was uneventful and also there was no history of taking any offending drugs by mother during her pregnancy period. Other sib was healthy; no family history was relevant regarding this case. No mental retardation was detected during examination. Physical examination of the child showed undernourishment and weighed 13kgs (>-2Z). The child had short stature. Facial asymmetry was present; frontal bossing, congenital left facial paralysis (lower motor neuron lesion) [Figure 1], macrostomia, short neck, bilateral microtia [Figure 3, 4] with meatal stenosis with sensory neural hearing loss. On ophthalmic examination, there was coloboma on both upper eye lids and dermoid on right eye [Figure 2] and malocclusion of teeth. There were no cardiac anomalies on echocardiography and renal function revealed normal.

X-ray showed maxilla and mandibular hypoplasia on (Rt.) side. [Figure 5] X-ray cervical spine lateral view showed fusion of 2 vertebrae with tonguing of the subsequent vertebra.

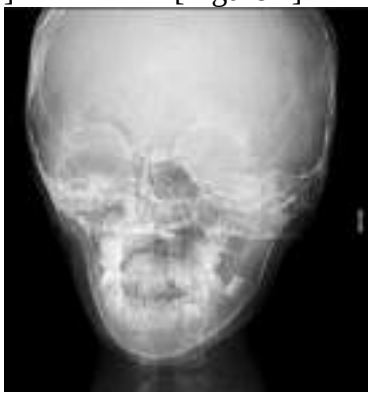
[Figure 1]



[Figure 2]

[Figure 3]

[Figure 4]



[Figure 5]

DISCUSSION:

Goldenhar syndrome was classically described as a triad of accessory tragic, mandibular hypoplasia and ocular dermoids,[3] in approximately 10 to 33 percent of affected individuals, malformations are bilateral affecting both sides of the body, with one side typically more affected than the other (leading to asymmetry). In the majority of such cases, the right side is more severely affected than the left.[10] This study showed right side of the face was more severely affected than left side.

Ocular anomalies occur in about 50% of the cases of Goldenher syndrome.[11] 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. Approximately 65% of the cases also found ear anomalies; include pre-auricular tags, microtia, anotia and conductive hearing loss. In this study we found microtia, pre-auricular tag and sensory neural hearing loss. Vertebral anomalies are combination of hemi-vertebra, fused ribs, kyphosis and scoliosis.

Additional features such as cardiac, genitourinary and pulmonary systems can also be affected.[12,13] Despite the reported frequency of cardiovascular alterations ranging from 5 to 58% in the reported patient.[14] There was no cardiac anomalies in our study.

Reports indicate that several teratogen agents such as retinoic acid, primidone, temoxifen and thalidomide, environmental factors such as insecticides, herbicides and maternal diabetes have produced this syndrome in infants born to pregnant women exposed to these agents.[15,16] In our case there was no history of maternal drug intake, any febrile illness or diabetes during pregnancy.

Trisomy of 7, 22 and juvenile glaucoma with turner syndrome have been described in association with Goldenhar syndrome.[17] Due to absence of genetic analysis and other advanced diagnostic aids, we based our diagnosis on various clinical and radiological features and came to a conclusion of Goldenhar syndrome.

Our patient was provisionally diagnosed as Goldenhar syndrome with most important clinical findings along with routine baseline investigation, but a number of other first and second arch syndromes were also considered in the differential diagnosis.[18] Although this syndrome presents some similarities with the Treacher-Collins syndrome, it is now considered a distinct entity. The features of Treacher-Collins syndrome are most frequently bilateral without ocular and aural anomalies.[19] Other syndromes like- Hallermann-Streiff syndrome (mandibulo-oculo-dyscephaly), Cockayne's syndrome, Seckel syndrome, Delleman syndrome were also differential with Goldenher syndrome.[20] Children with FAV syndrome should be assessed for both vision and hearing.[21]

Treatment for Goldenhar syndrome varies greatly depending on the needs of the individual with a multidisciplinary approach. The treatment of the disease varies with age and systemic associations and is mainly cosmetic and reconstructive surgery. In some mild cases, no treatment is needed. Children may need to work with a hearing specialist or speech therapist for hearing issues, or may need a hearing aid. If there are vision problems, corrective surgery or glasses may be needed. Surgery may also be needed to correct heart or spinal defects. Children with an intellectual disability may need to work with education specialists. Prognosis of this disease is good in otherwise uncomplicated cases without any systemic associations.

CONCLUSION:

We concluded our diagnosis Goldenhar syndrome on the basis of various clinical and radiological features. Goldenhar syndrome is an uncommon congenital deformity that requires a multistage and multidisciplinary approach for its best possible management.

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