GOLDENHAR Syndrome with Tetrology of Fallot-A Case Report

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ABSTRACT

The oculo-auriculo-vertebral spectrum (OAVS) or goldenhar syndrome is a non-random association of microtia, hemifacial microsomia with mandibular hypoplasia, ocular epibulbar dermoid, and cervical vertebra malformations. Congenital heart defects (CHDs) have been reported in 5-58% of these patients.We report a 20 year old male patient diagnosed with tetrology of fallot and oculo auriculo vertebral syndrome.

Key words: Goldenhar syndrome, oculo-auriculo-vertebral spectrum, tetrology of fallot

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INTRODUCTION

Goldenhar syndrome is a rare genetic syndrome with multisystem involvement. External phenotype is quite characteristic with pre auricular tags and oculor dermoids. Prompt screening of such patients for congenital hear disease is important.

CASE REPORT

A 20-year-old Male with conductive type of deafness was referred for evaluation of cardiovascular system due to cyanosis and clubbing. On examination, he was found to have Bilateral epibulbar dermoid[Figure 1], Bilateral multiple pre-auricular tags [Figure 2], scoliosis of thoracic spine .

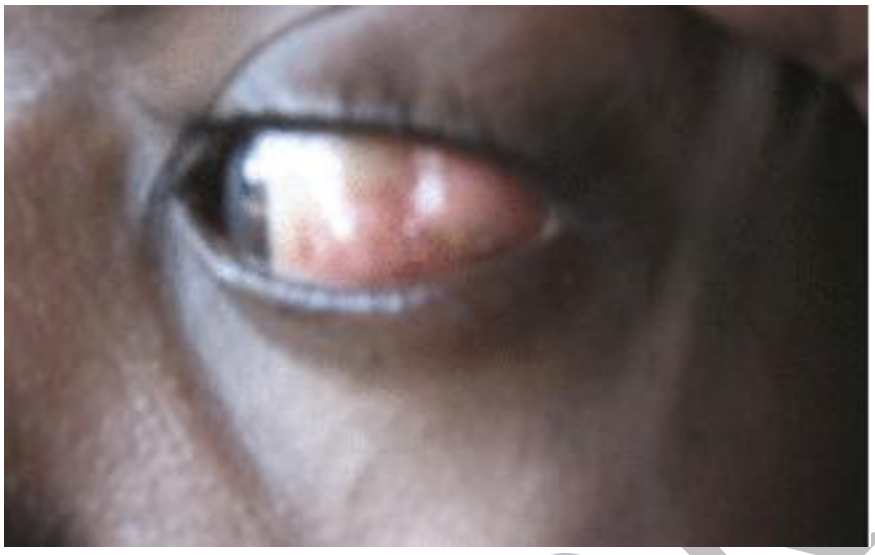

Figure 1: Epibulbar dermoid

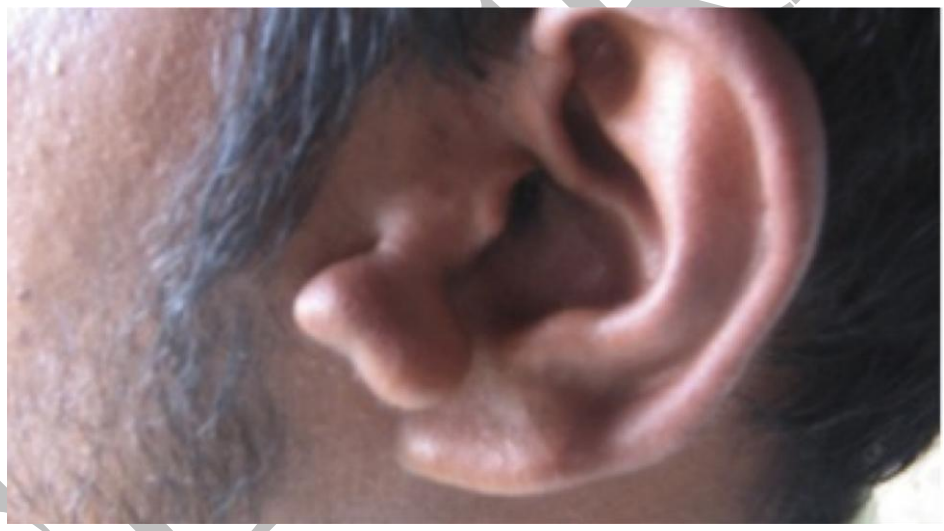

Figure 2-Pre auricular tags

Patient was also having mal-aligned teeth, high arched palate,pulled up angle of mouth on left side of face [Figure 3].

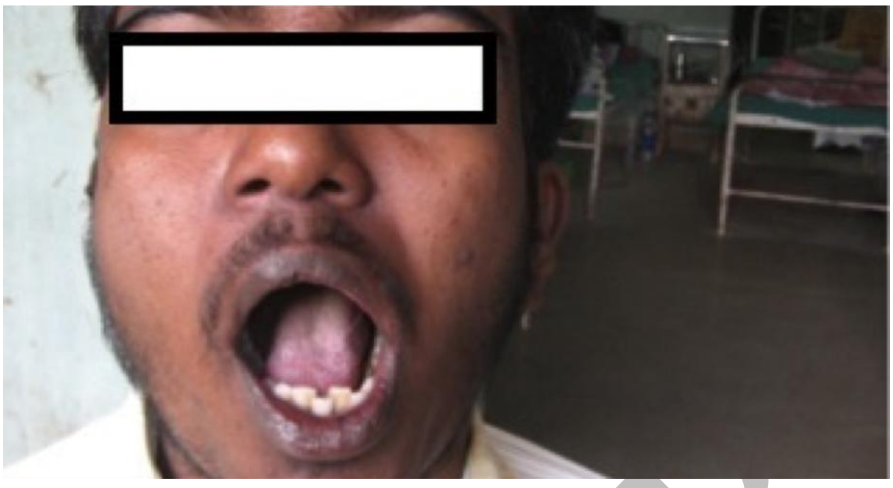

Figure 3: Pulled up angle of mouth

He was born of non-consanguineous marriage, and mother gave history of squatting equivalents since childhood. His resting saturation was 87% with pandigital clubbing. Cardiac examination revealed a relatively silent precordium with Right ventricular apex and single second heart sound (aortic component), findings consistent with tetrollogy of fallot.ECG,chest Xray and 2D echocardiogram confirmed the diagnosis of tetrology of fallot with a large subaortic ventricular septal defect, overriding of aorta..He was advised total corrective repair but refussed due to personal reasons.

DISCUSSION

Goldenhar syndrome (oculoauriculovertebral dysplasia with hemifacial microsomia) is a rare congenital anomaly involving the first and second branchial arches, where the patient's facial features are incompletely developed on one side, resulting in eye, ear, and jaw abnormalities. When these facial abnormalities are associated with vertebral malformations in the spine and more severe involvement of the eyes, this collection of symptoms is called Goldenhar syndrome.

In 85% of patients only one side of the face is affected.. The syndrome was first described in 1952 by the French ophthalmologist Maurice Goldenhar.[1] The incidence of Goldenhar syndrome has been reported to be between 1:3500 and 1:5600, with a male:female ratio of 3:2.[2] It is possible that abnormal embryonic vascular supply, disrupted mesodermal migration or some other factor leads to defective formation of the branchial

and vertebral systems.[2,3] Most of the cases have been sporadic. Autosomal dominant, autosomal recessive, and multifactorial modes of inheritance have also been suggested.[2]

Ingestion of drugs such as thalidomide, retinoic acid, tamoxifen, and cocaine by the pregnant mother may be related to the development of this syndrome. Maternal diabetes, rubella, and influenza have also been suggested as etiologic factors.[4]

The classic features of this syndrome include ocular changes such as microphthalmia, epibulbar dermoids, lipodermoids, and coloboma; aural features such as pre-auricular tragi, hearing loss, and microtia; and vertebral anomalies such as scoliosis, hemivertebrae, and cervical fusion.[1,4] The abnormalities are found to be unilateral in 85% of cases and bilateral in 10-33% cases.[4]

In Goldenhar syndrome, ocular anomalies especially bilateral dermoids are seen in 60% of the cases, vertebral anomalies in 40% of the cases, and ear anomalies also in 40% of the cases.[2].Other systemic features are found in about 50% of the patients.[5]

Congenital heart diseases are seen in 40 to 60% of patients with Goldenhar's syndrome, especially Tetralogy of Fallot, VSD, ASD, patent ductus arteriosus (PDA), and coarctation of the aorta of which TOF is more common[4] Cleft lip and palate, macrostomia, micrognathia, webbing of the neck, short neck, tracheoesophageal fistula, abnormalities of sternocleidomastoid muscle, umbilical hernia, inguinal hernia, urologic anomalies, hypoplastic vagina, and anal anomalies may be associated.[4]

Other syndromes associated with multiple pre-auricular tragi include Treacher-Collins syndrome, Wolf-Hirschhorn syndrome, Nager's acrofacial dysostosis, Wildervanck syndrome (cervicooculoacoustic syndrome), Townes-Brocks syndrome, and Delleman syndrome.[6].

The treatment of the disease varies with age and systemic associations and is mainly cosmetic in uncomplicated cases. Reconstruction surgeries of the external ear may be performed at the age of 6 to 8 years. In patients with milder involvement, jaw reconstruction surgeries can be done in the early teens; epibulbar dermoids should be surgically excised. Structural

anomalies of the eyes and ears can be corrected by plastic surgery.[2]

Prognosis of the disease is good in otherwise uncomplicated cases without any systemic associations. Successful treatment requires a multidisciplinary approach involving otolaryngologists, ophthalmologist, pediatrician, dermatologist, orthopedician, cardiologist..

CONCLUSION

Goldenhar syndrome is a rare disorder, presents with multi-organ involvement, presence of pre auricular tags and occulor demoids should always prompt the physician to screen such patients for this syndrome and all such patients should be promptly evaluated for cardiac defects.

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