



Goldenhar Syndrome – A Case Report

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ABSTRACT

Goldenhar syndrome (GHS) is also known as 'Oculo-auriculo-vertebral syndrome' (OAV).^[1] It is a spectrum of rare disorder which is apparent at birth. Dating back in 1950s it was initially comprised of malformations of ears and ocular abnormalities, only in 1963, the vertebral abnormalities were included as a sign of this syndrome. It is characterised by combination of anomalies like limbaldermoid, epibulbar cysts, auricular appendices, malformation of ears and hypoplasia of bones of mandible. GHS is a rare developmental disorder affecting the first and second branchial arches. Its also known as facio-auriculo-vertebral anomaly. The aetiology remains unclear in this syndrome. We present a case of 8 month male child with classical features with variable presentation.

Keywords – Goldenhar syndrome, oculo-auriculo-vertebral syndrome.

Synonym – Hemifacial Microsomia,^[1]

FAV spectrum, OAV spectrum,

INTRODUCTION

Goldenhar syndrome (GHS) is a congenital defect characterised by asymmetrical malformations classically involving face, eyes and ears. This condition was first described by Maurice Goldenhar in 1952.^[2] Gorlin in 1963 described this syndrome with vertebral anomalies.^[2] It is also associated with anomalies of CNS like calcification of falx cerebri, visceral, Cardiac and GIT anomalies.

The precise incidence of Goldenhar syndrome is unknown, but estimate are from 1 in 35,000 to 1

in 56,000.^[3] The male-female ratio is 3:2.^[3] Sporadic nature occurring randomly with no apparent cause. Positive family history have been described that have suggested autosomal recessive or dominant and multifactorial inheritance.^[4] Drugs like thalidomide, tamoxifen, retinoic acid and cocaine by pregnant mothers may be implicated. Heavy alcohol consumption during pregnancy^[5] and rubella, influenza and maternal diabetes have also been suggested as etiological factors.^[6]

The diagnosis of this syndrome is mainly based on clinical aspect. Most consider presence of ear anomalies are essential for diagnosis.

CASE HISTORY

8 Month old male child brought to us with complaints of cough, rhinorrhea and fever since 8 days increasing in intensity. On examination, he had tachycardia, tachypnea and slightly high blood pressure diagnosed to have bronchiolitis. On physical examination s/o bilateral microtia with narrow external ear canal, abnormal shaped skull with increased head circumference, right hypoplastic thumb. On ophthalmic examination s/o bilateral cataracts, coloboma of iris and disc. On radiological examination, left sided renal agenesis (USG finding) was found. Warm humidified oxygen and nebulisation was given. The patient improved after 8 days and discharged.

DISCUSSION

Goldenhar syndrome was first described by German physician, Carl Ferdinand VON ARLT in 19th century. Goldenhar M described this syndrome with triad of accessory tragus, mandibular hypoplasia and ocular-dermoid-goldenhar syndrome. Gorlin described this condition as oculo-auriculo-vertebral syndrome.

The exact etiology of this syndrome is not known. It is sporadic in nature.

Family history has been reported. Some authors have said that GHS may be due to the interaction of genes possibly in combination with environmental factors, many others suggested a possible link between genetic causes such as vascular disruption due to fetal hemorrhage in first and second branchial arches when blood supply switches from stapedial artery to external carotid artery (ECA).

The symptoms and physical features vary in severity from case to case. External ear anomalies and ipsilateral facial underdevelopment; right side is affected more than left side but in 30% to 50% both sides are affected with one side affected more. Other anomalies also associated with

classical features like mandibular hypoplasia, ophthalmic anomalies like coloboma of the eyelids/iris/retina/choroid, strabismus, dermoid cyst, anophthalmia, microphthalmia, blepharoptosis, vertebral abnormalities like hemivertebra or block vertebra, congenital heart diseases like tetralogy of Fallot and ventricular septal defects are most common, renal anomalies (agenesis/hypoplasia), CNS (intracranial lipoma, cranial nerve dysgenesis, hydrocephalus), GIT anomalies has been reported. In our case the patient showed pre auricular tags i.e. bilateral microtia with narrow external ear canal, abnormal shaped skull with increased head circumference, right hypoplastic thumb^[5], bilateral cataracts, coloboma of iris and disc, mild right sided facial asymmetry, and left sided renal agenesis.

There is no single test to diagnose Goldenhar's syndrome and diagnosed clinically.

In these cases, the treatment of the syndrome varies with age and systemic associations and mainly cosmetic.^[5] Craniofacial reconstruction, reconstructive surgery of the external ear, jaw reconstructive surgery may be needed for some patients.

Supportive multidisciplinary management is important.

Differential diagnosis specially for facial anomalies should be considered -

- I. Treacher Collins syndrome.
- II. Craniosynostosis.
- III. Romberg disease.
- IV. Hemifacial microsomia.

Conflict of interest- none

Clinical Photographs-



BIBLIOGRAPHY

1. Gorlin RJ, Pindberg JJ, Cohen MM. Oculo-auriculo-vertebral dysplasia In: Syndromes of the Head and Neck. A Blackiston Division. McGraw-Hill: New York, London and Johannesburg; 1964. p546-52
2. Goldenhar M. Association malformatives de l'oeil et de l'oreille, en particulier le syndrome dermo-epibulbaire-appendice-auriculaires-fistula auris-congenita et ses relations avec la dysostose mandibulo-faciale. J Genet Hum 1952;1:243-82.
3. Grabb WC. The first and second branchial arch syndrome. Plast Reconstr Surg 1965;36: 485-508.
4. Collins ET. Case with symmetrical congenital notches in the outer part of each lid and defective development of malar bones. Trans Ophthalmol Soc UK 1900; 20:190.
5. Das A, Ray B, Das D, et al A case of Goldenhar-Gorlin syndrome with unusual association of hypoplastic thumb. Indian J Ophthalmol. 2008 Mar-Apr;56(2):150-2.
6. Mutanabbi M, Rahman MA, Mamun AA, et al Goldenhar syndrome - a case report. Mymensingh Med J. 2014 Jul;23(3):586-9.