Case Report

Indian Journal of Maternal-Fetal & Neonatal Medicine
Volume 9 Number 1, January - June 2022
DOI: http://dx.doi.org/10.21088/ijmfnm.2347.999X.9122.3

Goldenhar Syndrome: A Case Report

Habibur Rehman[1], Sushruta Kumar[2], Sunil Natha Mhaske[3], Abhijeet shinde[4]

How to cite this article:
Habibur Rehman, Sushruta Kumar, Sunil Natha Mhaske, Abhijeet shinde/Goldenhar Syndrome: A Case Report /Indian Journal of Maternal-Fetal & Neonatal Medicine; 9(1):29–31.

Authors Affiliation:
[1]Postgraduate Student, [2,4]Assistant Professor, [3]Dean, Dr. Vithalrao Vikhe Patil Medical College and Hospital, Ahmednagar, Maharashtra 414111, India.

Correspondence Author:
Sushruta Kumar, Assistant Professor, Dr. Vithalrao Vikhe Patil Medical College and Hospital, Ahmednagar, Maharashtra 414111, India.

E-mail: Sushruta-Kumar@gmail.com

Received on: 15.09.2021

Accepted on: 19.10.2021

Abstract

Goldenhar syndrome is an uncommon disorder characterised by a variety of defects including craniofacial tissues, vertebrae, and internal organs. It preferably affects one side of the body. The origin of this illness is unknown because it is genetically variable and can be caused by a variety of factors. We present a case with Goldenhar syndrome with microtia, as well as systemic involvement, that was evaluated clinically and radiographically. The patient had many common symptoms of the illness as well as a few unusual ones. The many elements of this rare disease have been examined, with a focus on early detection and a multidisciplinary management strategy.

Keywords: Goldenhar Syndrome; Radiographically; Chromosome abnormalities; Microcornea.

Introduction

Oculo-auriculo-vertebral syndrome (OAVS), sometimes known as Goldenhar syndrome, is a rare congenital disorder caused by abnormalities in the first and second brachial arches. Dr. Maurice Goldenhar was the first to describe it in 1952.[1]

The aetiology of this illness is still unknown. Chromosome abnormalities, neural crest cells, and environmental influences during pregnancy, such as medication ingestion, thalidomide, retinoic acid, and alcohol consumption by the mother, were all linked to the disease's development. Diabetes in the mother has also been proposed as an etiologic cause.[2] In terms of clinical features, the patient may have face abnormalities, ear abnormalities, eye abnormalities, vertebral malformations, and congenital cardiac difficulties.

We present a case of Goldenhar syndrome in this article, along with a discussion of clinical symptoms, the necessity of early identification, and a multidisciplinary approach to treatment.[3]

Case Report

3 hrs old male child born to primigravida of non-consanguineous marriage at 38 weeks of gestational age via caesarean section due to meconium-stained liquor with birth weight of 2900 gm referred to our hospital I/v/o meconium aspiration o/e: facial features: no facial asymmetry present, hypertelorism, antimongoloid slant of eyes, depressed bridge of the nose and anteverted nostrils.

Eyes: There was no microcornea, micro-ophthalmia, ptosis, coloboma of the iris or choroid or a cataract.

There were no skull, spinal, rib or limb anomalies on examination ; strabismus was present.

Ears: both ears had 3rd degree microtia

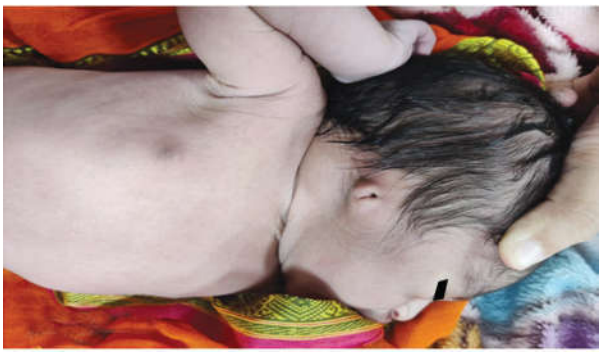
Spinal examination: scoliosis was present clinically confirmed on xray.

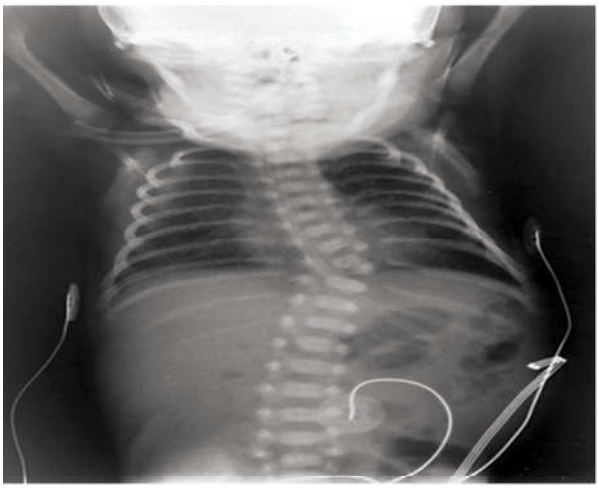
systemic examination revealed murmur on cardiovascular examination later on 2D echo findings were: tiny PDA, moderate TR, ventricular septal defect of 3mm.

Abdominal ultrasound revealed structurally and functionally normal kidneys.

On audiometry test: results found to be suggestive of bilateral otoacoustic emissions absent in both side cochleae

Neurosonogram revealed: no abnormality





Discussion

Goldenhar syndrome has been estimated to have a prevalence of 1:3500 to 1:5600.[4] The specific cause of the disease is unknown. However, aberrant embryonic vascular supply to the first arch is thought to be the cause[5] and aberrant mesoblastic development influencing branchial and spinal system creation. The majority of the incidents were sporadic. There have also been suggestions for autosomal dominant, autosomal recessive, and multifactorial inheritance mechanisms.[6]

It is an exceedingly rare, genetic syndrome with an unknown cause that occurs seldom. Positive family histories have been found in some cases, indicating autosomal dominant or recessive inheritance.[7] According to some experts, multifactorial inheritance occurs as a result of the interplay of numerous genes, sometimes in combination with environmental variables. Defective creation of the brachial arches and spinal system can be caused by abnormal vascular supply of the embryo and altered mesodermal migration.[8]

The condition is mostly unilateral in occurrence in 85% cases, with the right side more frequently affected than the left with a ratio of 3:2.[9]

The characteristic combination of external ear anomalies and ipsilateral facial underdevelopment is the hallmark of Goldenhar syndrome as seen in our case.[10]

Patients with mandibular hypoplasia are common, however no maxillary insufficiency was seen in our case. Goldenhar syndrome can also have a wide range of systemic involvement. Tetralogy of Fallot and ventricular septal abnormalities are the most prevalent cardiovascular anomalies linked with OAVS, as demonstrated in our patient.[11]

Cleft lip and palate, macrostomia, micrognathia, webbing of the neck, a short neck, tracheoesophageal fistulas, and sternocleidomastoid muscle anomalies have all been linked to cleft lip and palate. Although clinical diagnosis is the primary focus, radiographic examinations can assist in confirming the clinical diagnosis. Ultrasound, which may reveal evident anomalies, can be used to get a precise prenatal diagnosis.

Prenatal deoxyribonucleic acid testing cannot be utilised to diagnose this syndrome because no particular genes have been linked to it. It must be separated from other syndromes such as Treacher Collins syndrome (TCS), Nager syndrome, and Townes-Brocks syndrome because the differential diagnosis is so vast. In Goldenhar syndrome, facial

involvement is usually unilateral, resulting in facial asymmetry, whereas in TCS, involvement is frequently bilateral. The TCOF1 gene mutation on human chromosome 5q31-34 is connected to TCS and aids in the ultimate diagnosis. Plastic surgery can repair the structural abnormalities of the eyes and ears in Goldenhar syndrome. Plastic surgery is usually postponed until the growth of the structure is completed.

The epibulbar dermoids should be removed. They may regrow and recurrence has been observed as long as 12 years after original surgery. Orthodontic treatment may be required for maloccluded teeth. Hearing defect should be looked for and if detected, may be improved by the use of hearing aids early in life, provided it is because of conductive deafness.

Conclusion

There should be multidisciplinary approach via otorhinolaryngologist, cardiovascular thoracic surgeon, physiotherapy, occupational and speech therapist. Role of a pedodontist is significant to provide optimum oral health treatment for such syndromic patients from birth through adolescence since often they have complex unmet dental needs. Paediatric dentists and paediatricians should liaise with sound referral services for prompt treatment of the affected children.

Medical and dental professionals should make every effort to diagnose and treat this illness as soon as possible, easing the emotional, physical, and financial burdens that these children experience. The cardiac anesthesiologist faces a difficult task when dealing with patients who have this unusual illness. The kind, extent, and severity of craniofacial vertebral abnormalities, the severity of the cardiovascular condition, the underlying metabolic abnormality, and the nature of surgery all influence perioperative therapy for cardiac surgical patients with GH syndrome.

References

1. Kannur A, Talpallikak N, Chavan P. Goldenhar syndrome-a case report. National Journal of Medical and Allied Sciences. 2013;2(2):32-5.
2. Araneta MR, Moore C, Olney RS, Edmonds LD, Karcher JA, McDonough C, Hiliopoulos KM, Schlangen KM, Gray GC. Goldenhar syndrome among infants born in military hospitals to Gulf War veterans. Teratology. 1997 Oct;56(4):244-51.
3. Goswami M, Bhushan U, Jangra B. Goldenhar Syndrome: a case report with review. International Journal of Clinical Pediatric Dentistry. 2016 Jul;9(3):278.
4. Grabb WC. The first and second branchial arch syndrome. Plastic and reconstructive surgery. 1965 Nov 1;36(5):485-508.
5. McKenzie J. The first arch syndrome. Archives of disease in childhood. 1958 Oct;33(171):477.
6. Collins ET. Case with symmetrical congenital notches in the outer part of each lower lid and defective development of the malar bones. Trans Ophthalmol Soc UK. 1900;20:191-2.
7. Bhuyan R, Pati AR, Bhuyan SK, Nayak BB. Goldenhar Syndrome: A rare case report. Journal of oral and maxillofacial pathology: JOMFP. 2016 May;20(2):328.
8. Kokavec R. Goldenhar syndrome with various clinical manifestations. The Cleft palate-craniofacial journal. 2006 Sep;43(5):628-34.
9. Vinay C, Reddy RS, Uloopi KS, Madhuri V, Sekhar RC. Craniofacial features in Goldenhar syndrome. Journal of Indian Society of Pedodontics and Preventive Dentistry. 2009 Apr 1;27(2):121.
10. Werler MM, Starr JR, Cloonan YK, Speltz ML. Hemifacial microsomia: from gestation to childhood. The Journal of craniofacial surgery. 2009 Mar;20(Suppl 1):664.