ISSN: 0976-3031

Available Online at http://www.recentscientific.com

CODEN: IJRSFP (USA)

International Journal of Recent Scientific Research
Vol. 9, Issue, 11(C), pp. 29670-29672, November, 2018

International Journal of Recent Scientific Research

DOI: 10.24327/IJRSR

Case Report

GOLDENHAR SYNDROME: A CASE REPORT

Nikhil Achanta* and Anand Sude

Deptt.of Pediatrics,. D.Y. Patil University, School of Medicine, Nerul, Navi Mumbai, Maharshtra, 400 706

DOI: http://dx.doi.org/10.24327/ijrsr.2018.0911.2904

ARTICLE INFO

Article History:

Received 12th August, 2018
Received in revised form 23rd September, 2018
Accepted 7th October, 2018
Published online 28th November, 2018

Key Words:

Preauricular sinus, epicanthal folds, accessory tragus

ABSTRACT

Alsoknownas'Oculo-auriculo-vertebral syndrome', Goldenhar syndrome is a rare condition which is characterized by a multitude of congenital anomalies involving the vertebrae, craniofacial structures, internal organs, usually with unilateral involvement. Here we are presenting a case of 21 month old female child with classical features of this rare condition i.e. microtia, pre auricular sinus, accessory tragus, epicanthal folds and mild facial asymmetry. This patient had many classical signs of the syndrome along with few rare ones. We have discussed various aspects of this extremely rare conditionwith emphasis on early diagnosis and multidisciplinary approach to managing the same.

INTRODUCTION

Goldenhar syndrome is an extremely rare congenital condition arising from defects in the first and second brachial arches. It was first described by Dr. Maurice Goldenhar in 1952 [1]. In 1963, Gorlin *et al*. suggested the use of the term oculoauricularvertebral dysplasia to describe the syndrome[2]. The incidence of Goldenhar syndrome has been reported to be 1:35,000-1:56,000 with a male to female ratio of 3:2 [3]. The etiology of this condition is multifactorial. Many factors are implicated including chromosomal abnormalities, anomalous neural crest cell development, other factors like ingestion of drugs, such as cocaine, thalidomide, retinoic acid, intake of alcohol by the mother during pregnancy. Maternal diabetes has also been implicated in the development of the disease[4]. Patients may present with a variety of dysmorphic features ranging from facial abnormalities, ear or eye abnormalities, or with problems with development as seen in this case.

In this article, we report a case of Goldenhar syndrome along with discussion on clinical features, importance of early diagnosis, and interdisciplinary approach needed to manage it.

CASE REPORT

21 month old female child was born with bilateral microtia (right side affected more than the left),left sided pre auricular sinus, right sided accessory tragus and bilateral epicanthal folds via spontaneous vaginal delivery to a 28 year old G3P2L2 mother at 37 completed weeks of gestation. Antenatal fetal USG were unremarkable and the subsequent delivery was uncomplicated.

The patient presented to a tertiary care centre in Navi Mumbai at 21 months of age with dysmorphic features and inability to open her mouth completely. All milestones were appropriate for age except for language delay. The patient could only speak bisyllables at 21 months of age.

Examination: On examination vitals were stable, there was bilateral Microtia (Fig.1) with bilaterally visualised ear canals, Preauricular sinus on the left side (Fig.2); additionally the Right Post auricular region showed a firm flesh coloured nodule consistent with accessory tragus (Fig.3). Ophthalmic examination revealed bilateral epicanthal folds (Fig.1) and epibulbardermoids. There was mild facial asymmetry present. Oralexamination was normal (no cleft lip/palate). Clinical diagnosis of Goldenhar syndrome was rendered.

Investigations: CT Temporal Bone showed moderate narrowing of both external auditory canals and bilateral semicircular canal-vestibular dysplasia.

Corresponding author:* **Nikhil Achanta
Deptt.of Pediatrics,. D.Y. Patil University, School of Medicine, Nerul, Navi Mumbai, Maharshtra, 400 706

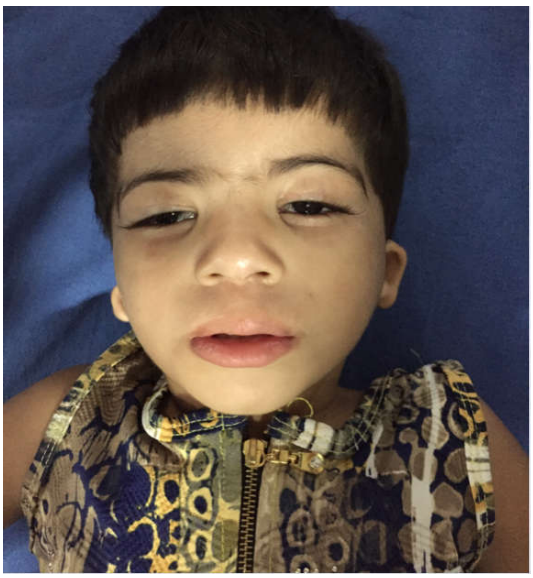

Fig 1

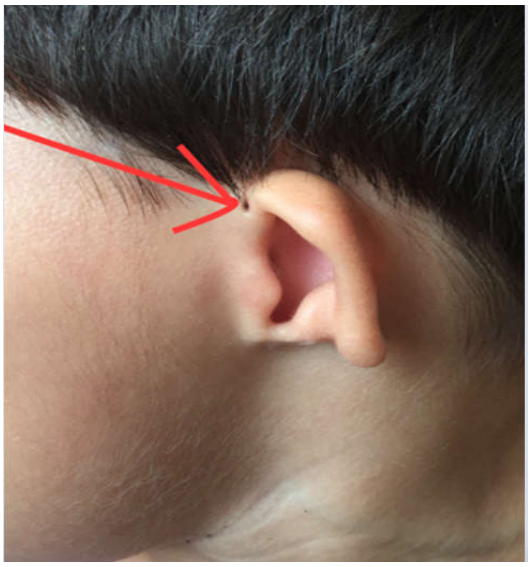

Fig 2

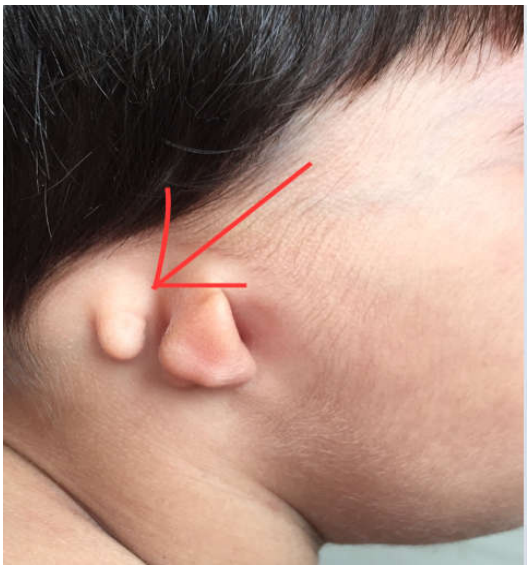

Fig.3

Hearing assessment was suggestive of moderate conductive hearing loss in both ears.

Xray whole spine, 2D Echo, USG Abdomen and KUB were normal.

Course and treatment options

The treatment of the disease varies with age and systemic associations and is mainly cosmetic in uncomplicated cases such as this. Reconstruction surgeries of the external ear may be performed at the age of 6 to 8 years, for which the patient has been asked to follow up. 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.

DISCUSSION

Goldenhar Syndromeincludes a wide spectrum of anomalies that include epibulbardermoids and/or lipodermoids, auricular appendages, auricular fistulas, facial and vertebral anomalies; and as such several terms have been used to describe this condition over the years, one of these terms being oculoauriculovertebral dysplasia. However, due to significant overlap in the clinical features ofhemifacial microsomia, oculoauriculovertebral dysplasia and GS; the term oculo-auriculovertebral (OAV) spectrum is best used to describe the patients affected by this association. Still, the term Goldenhar Syndromeis more widely recognized and traditionally used despite OAV spectrum being the more inclusive and descriptive of the two.

As seen with our case, most of the patients reported within the OAV spectrumare sporadic.[2] Positive family histories have been presentin some cases that have suggested autosomal dominant or recessive inheritance. Some researchers suggested multifactorial mode of inheritanceinvolving interaction of several genes, and other factors leading to defective formation of brachial arches and vertebral system including but not limited to disruption of mesodermal migration and abnormal vascular supply of embryo[5]

The condition is mostly unilateral in occurrence in 85% cases, with the right side more frequently affected than the left [6]. Our case showed bilateral involvement which makes it a more rare presentation. Our case demonstrates some of the classical features seen in Goldenhar Syndrome. These involve aural defects, such as preauricular tags, anotia, microtia and hearing loss, ocular anomalies such as anophthalmia, microphthalmia, epibulbar dermoidtumors, and eyelid colobomas, vertebral abnormalities, such as scoliosis, hemivertebrae and cervical fusion, and mandibular hypoplasia.[7,8,9] Systemic involvement in Goldenhar syndrome is varied. Among cardiovascular anomalies, tetralogy of Fallot and ventricular septal defects are most commonly associated with Oculo Auriculo Vertebral Syndrome. Cleft lip and palate, macrostomia, micrognathia, webbing of the neck, a short neck, tracheoesophageal fistulae, and abnormalities of sternocleidomastoid muscle may also be associated. The frequency of cardiovascular alterations and conductive hearing loss has been reported to occur with an overall frequency of approximately 50%[10] and our patient had evidence of bilateral conductive hearing loss.GS is clinically heterogeneous, and there is no agreement in the literature on the minimal diagnostic criteria. The diagnosis ismade mainly

based on clinical features supported by relevant radiologic investigations.

The differential diagnosis is broad and syndromes from which Goldenhar syndrome needs to be differentiated include Treacher-Collins syndrome, Wolf-Hirschhorn syndrome, Nager's acrofacial dysostosis, Wildervanck syndrome (cervicooculoacoustic syndrome), Townes-Brocks syndrome, and Dellemansyndrome.[11]

Treacher Collins syndrome is associated with maxillary and mandibular hypoplasia and much more marked degree of facial asymmetry as compared to Goldenhar syndrome but is not associated with ocular and aural anomalies.[5]

The treatment of the disease relies heavily on prompt and earlydiagnosis. Treatment options vary with age and systemic associations and treatment is mainly cosmetic in uncomplicated cases. Reconstruction surgeries of the external ear may be performed at the age of 6 to 8 years. Jaw reconstruction surgeries can be done in the early teens in patients with milder involvement; Surgical excision of epibulbardermoids, and correction of structural anomalies of the eyes and ears by plastic surgery can be done. Prognosis of the disease is good in otherwise uncomplicated cases without any systemic associations. A multidisciplinary approach involving otolaryngologists, ophthalmologist, pediatrician, dermatologist, orthopedician is central to successful treatment of these patients.

CONCLUSION

The effect of Goldenhar Syndrome becomes more and more evident as the child grows, because of delays in the growth and the development of the affected areas. The lack of development of the ear canals can hinder speech development, as seen in this case. Furthermore, malformations of the jaws can cause breathing problems, as well as dental malocclusion which requires multidisciplinary approach[12]. Early diagnosis is paramount to ensure the best possible quality of life for the patient and efforts need to be made by the medical fraternity to diagnose and manage this condition at the earliest, so as to minimise the physical, emotional, and financial burden of living in these special children.

References

1. Goldenhar M. Associated malformations of eye and ear, particularly dermoid syndrome epibulbar-appendices, congenital auricular fistulas and its relations with Manibulofacial Dysostosis. *J Genet Hum*. 1952; 1:243-82.
2. Gorlin RJ, Jue KL, Jacobsen U, Goldschmidt E. Oculoauriculovertebral dysplasia. *J Pediatr*. 1963; 63:991-9.
3. Ashokan CS, Sreenivasan A, Saraswathy GK. Goldenhar syndrome-review with case series. *J Clin Diagn Res*. 2014 Apr;8(4):ZD17-ZD19
4. Araneta MR, Moore CA, Onley RS, Edmonds LD, Karcher JA, McDonough C, Hiliopoulos KM, Schlangen KM, Gray GC. Goldenhar syndrome among infants born in military hospital to Gulf war veterans. Teratology. 1997 Oct; 56(4):244-251.
5. Kulkarni VV, Shah MD, Parikh AA. Goldenhar syndrome: a case report. *J Postgrad Med*. 1985 Jul; 31(3):177-179.
6. Kokavec R. Goldenhar syndrome with various clinical manifestations. Cleft *Palate Craniofac J*. 2006 Sep; 43(5):628-634.
7. Rollnick BR, Kaye CI, Nagatoshi K, Hauck W, Martin AO. Oculoauriculovertebral dysplasia and variants: phenotypic characteristics of 294 patients. *Am J Med Genet*. 1987; 26:361-375.
8. Stromland K, Miller M, Sjogreen L, Johansson M, Joelsson BM, Billstedt E, *et al*. Oculo-auriculo-vertebral spectrum: associated anomalies, functional deficits and possible developmental risk factors. *Am J Med Genet* A. 2007; 143A:1317-1325.
9. Tasse C, Bohringer S, Fischer S, Ludecke HJ, Albrecht B, Horn D, *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; 48:397-411.
10. Touliatou V, Fryssira H, Mavrou A, Kanavakis e, Kitsiou-Tzeli S. Clinical manifestations in 17 Greek patients with Goldenhar syndrome. Genet Couns. 2006;17:359-370
11. Mehta B, Nayak C, Savant S, Amladi S. Goldenhar syndrome with unusual features. *Indian J Dermatol VenereolLeprol*. 2008;74:254-6.
12. Bielicka B, Necka A, Andrych M. Interdisciplinary treatment of patients with Goldenhar Syndrome - Clinical reports. Dent Med Probl. 2006; 43:458-62.

How to cite this article:

Nikhil Achanta and Anand Sude.,2018, Goldenhar Syndrome: A Case Report. *Int J Recent Sci Res.* 9(11), pp. 29670-29672. DOI: http://dx.doi.org/10.24327/ijrsr.2018.0911.2904

****