Case Report

A case report - goldenhar's syndrome

Bindu C B[1*], Srinivasa B S[2], Bhagawan[3]

[1]Assistant Professor, Department of General Medicine, [2]Assistant Professor, Department of Pediatrics, [3]Professor and HOD, Department of Dentistry, Hassan Institute of Medical Sciences, Hassan, Karnataka, INDIA.
Email: srinivasabindu76@gmail.com

Abstract

A boy aged 2 years 6 months was reported to our district hospital with a rare syndrome of goldenhar having multiple symptoms. He had facial asymmetry, scoliosis, eye problems like limbaldermoid, hypoplastic mandible with bilateral preauricular skin tag and there is history of delayed dentition and the baby was operated during neonatal period for tracheoesaphagialfistulla. Child also had absent Right Kidney.
Keywords: Goldenhar syndrome, Facial asymmetry, Preauricular tag.

***Address for Correspondence:**
Dr. Bindu C. B., # 73 7th Floor, Doctors Quatrus, SC Hospital, HIMS Campus, Hassan Karnataka-573201, INDIA.
Email: srinivasabindu76@gmail.com
Received Date: 29/01/2020 Accepted Date: 09/03/2020

Access this article online

Quick Response Code:

Website: www.statperson.com

Volume 10
Issue 2

INTRODUCTION

Goldenhar syndrome is a rare hereditary condition due to multifactorial etiopathology leading to disturbance of blastogenesis[1]. Goldenhar first described this condition in 1952 as a combination of several anaomolies such as , dermal epibalbartumours. Preauricular appendices, facial asymmetry. Gorlin in 1963 also described the Goldenhar syndrome in association with heart disease, hypoplasia the zygomatic, mandibular and maxillary bones[2], eye anomolies like coloboma of upper eyelid, iris, choroidea and retina, ear anomolies like microtia, anotia, preauricular skin tags, blind fistulas and other external ear manifestaions[3, 4, 5]. The incidence of this condition varies from 1 in 3500 to 1 in 5600 live births [3]. The Male: Female ratio is approximately 3:2. Cases may report with malformations of cardiac, renal, vertebral, ear, central nervous system and others skeletal anomolies[6]. A few cases of Orodentalanomolies have also been described. We report a case of a with Goldenhar syndrome with various clinical manifestations as described above.

CASE REPORT

A 2 years 6 months old boy was brought to outpatient department with a history of cough and cold, on examination child looks comfortable, afebrile, runny nose, respiratory rate of 20 cycles / min, pulse rate of 106/min and BP 90/60 mm of Hg in right upper limb. Facial asymmetry with right side hypoplasia with limbaldermoid (opthalmologist opinion) which was not disturbing his vision, Bilateral preauricular skin tag without any sinuses with no hearing disturbance. There was a right sided delayed eruption of mandibular teeth and there was a gross scoliosis towards the left side which was also evident by X-rays spine. There was no cardiac abnomilities, respiratory sounds were normal and child was neurologically normal but ultrasonography of the abdomen shows absent right kidney. The child was operated for tracheoesophageal fistula during neonatal period and the child's speech and hearing was intact. The parents were nonconsanguinously married without any family history of similar complaints and this is the only child. No maternal history of any medications and exposure to radiations.

Investigations

Blood routine was normal and renal functions were within normal limits.2D Echo was normal, X-ray of face showed right mandibular hypoplasia. Ultrasound of abdomen showed absent right kidney. Dentist opinion was taken and advised for correction of malocludedteeths.

How to cite this article: Bindu C B, Srinivasa B S, Bhagawan. A case report - goldenhar's syndrome. *International Journal of Recent Trends in Science and Technology* April to June 2020; 10(2): 37-38 http://www.statperson.com

DISCUSSION

The patient presented to us with various clinical manifestaions in the form of occulofacioauricular and vertibralanomolies as described above as in Goldenhar syndrome[8]. Facila asymmetry and hypoplasia of the mandible are typical features of Goldenharsyndrome[9].Which were present in our case. On the other hand the presence of epibulbardermoid tumour is variable [2] and it is usually associated with bilateral preauricular skin tags which is observed in our case. There were no hearing abnomolities and no facial nerve palsy[10] in this syndrome cardiac abnormality was observed in 5 to 58 percent of patients[11] which was not found in our case. Renal anomolies were common in this syndrome [12] and in our case there was absent right kidney with normal renal functions. There was no family history and this case could be a sporadic. Drugs like primidone, thalidomide, vitamin -A, cocaine exposure in pregnancy and also tobacco and herbicides have been associated with this syndrome[13].

REFERENCES

1. Altamar Rios J. Síndrome de Goldenhar – A propósito de umcaso. An OtorrinolaringolIber Am 1998; XXV: 491-497.
2. Oski FA, de Angelis CA, Feigin RD, Warshaw JB. Síndromescomuns com anormalidadesmorfológicas.Princípios e Prática de Pediatria. Rio de Janeiro: Guanabara Koogan; 1990. p 482.
3. Kokavec R (2006) Goldenharsyndrome with various clinical manifestations. Cleft Palate Craniofac J 43,628-634.
4. Garcia de Paula e Silva FW, de Carvalho FK, Diaz-Serrano KV, de Freitas AC, Borsatto MC, de QueirozAM (2007) Solitary median maxillary central incisor in association with Goldenhar's syndrome: a case report. Spec Care Dentist 27, 105-107.
5. Vinay C, Reddy RS, Uloopi KS, Madhuri V, Sekhar RC (2009) Craniofacial features in Goldenhar syndrome. J Indian SocPedodPrev Dent 27, 121-124.
6. Bielicka B, Necka A, Andrych M (2006) Interdisciplinary treatment of patients with Goldenhar syndrome – clinical reports. Dent Med Probl 43, 458-462.
7. Pinheiro AL, Araújo LC, Oliveira SB, Sampaio MC, Freitas AC (2003) Goldenhar's syndrome – case report. Braz Dent J 14, 67-70.
8. Rodríguez JI, Palacios J, Lapunzina P. Severe axial anomalies in the oculo-auriculo-vertebral (Goldenhar) complex. Am J Med Gen 1993; 47:69-74.
9. Schaffer AJ, Avery ME. Doenças do Recém-Nascido. 4th ed. São Paulo: Interamericana; 1979. p 803.
10. Carvalho GJ, Song CS, Vargervik K, Lalwani AK. Auditory and facial nerve dysfunction in patients with hemifacialmicrosomia.ArchOtolaryngol Head Neck Surg 1999; 125:209-212.
11. Nakajima H, Goto G, Nakata N. Goldenhar syndrome associated with various cardiovascular malformations. JnpCirc J 1998; 62:617-620.
12. Ritchey ML, Norbeck J, Huang C, Keating MA, Bloom DA. Urologic manifestations of Goldenhar syndrome. Urology 1994; 43:88-91.
13. Kumar R, Balani B, Patwari AK, Anand VK, Ahuja B. Goldenhar syndrome with rare association. Indian J Pediatr 2000; 67:231-233.

Source of Support: None Declared
Conflict of Interest: None Declared