

Oculo-Auriculo-Vertebral Dysplasia: A Rare Case Report

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ABSTRACT

Oculo-auriculo-vertebral dysplasia is also called as Goldenhar syndrome which is a rare syndrome developing from first and second pharyngeal arches during blastogenesis. Blastogenesis is a period which occurs in first 4 weeks after fertilization. This syndrome is characterized by abnormalities of face (hemifacial macrosomia), eyes (epibular dermoid), abnormalities of ear (microtia, anotia) and other external ear malformations. Here we present a case of a 25-year-old male patient with a classical feature of Ectrodactyly of ring finger of right hand, Syndactyly of left foot with middle finger, microphthalmia of right eye and hypoplasia of facial bone with facial asymmetry.

Keywords: Hemifacial, macrosomia, Microtia, Ectrodactyly

INTRODUCTION

Goldenhar syndrome was first documented in 1952 by Maurice Goldenhar. It is also known as Oculo-auriculo-vertebral syndrome or hemifacial macrosomia.¹The incidence of this syndrome is 1 in 5600 live births with male to female ratio of 3:2. There is lack of genetic linkage and sporadic occurrence which suggests a multifactorial etiology that includes nutritional and environmental factors that result in disturbance of blastogenesis.²

This syndrome presents a wide range of manifestations including craniofacial, vertebral, central nervous system etc. The Goldenhar syndrome is typically associated

with hemifacial macrosomia. The classic presentation of this syndrome is epibular dermoids, microtia, mandibular hypoplasia etc³

CASE REPORT

A 25-year-old male patient who reported to Department of oral medicine and Radiology of Swargiya Dadasaheb Kalmegh Smruti Dental college and Hospital, Nagpur with the chief complaint of pain in upper right back tooth region of jaw since 2 months. History of present illness revealed that he had a similar complaint 1 month back but the pain was relieved on taking medication. Further he revealed uncomplicated pregnancy of mother. His elder brother was normal. There was no history of trauma to head and neck region. No signs of mental retardation or impairment of cognitive functions were seen. Past medical history was not contributory to present case. Past dental history was he has done restoration with 46 three years back. And vital parameters were in normal range.

On general examination there was Ectrodactyly with ring finger of right hand (fig1). Syndactyly is seen on left foot with middle finger (fig 2). Partial clavicular stump was palpable on right side. IQ was also normal. A review of other systems appeared to be normal. Scars were seen on upper abdomen neck and back (fig 3).

On Extra Oral Examination, microtia of both of right and left side (fig 4) with flat nasal bridge (fig 5) and microphthalmia of right eye (fig 6) was observed.

On Intraoral Examination, Diastema is seen between 11 21, paramolar present between 16 17 in upper arch. Grossly decayed 17, Tenderness positive 17 (fig 7, fig

9). Microdontia seen 32 and 42, Missing 31 and 41, Restoration with 36 (fig 8).

On investigation, Chest X-ray revealed hypoplasia of right clavicle bone and inter-rib distance is short on right side as compared to left side (fig 10). Ectrodactyly of ring finger on right hand (fig 11) and Syndactyly on left foot with middle finger (fig 12).



Fig1: Ectrodactyly with ring finger of right hand

Fig 2: Syndactyly is seen on left foot with middle finger



Fig 3: Scars were seen on upper abdomen, neck, and back



Fig 4: Microtia of both of right and left side



Fig 5: Flat nasal bridge



Fig 6: Microphthalmia of right eye



Fig 7: Maxillary arch



Fig 8: Mandibular arch



Fig 9: Orthopantomogram



Fig 10: Hypoplasia of right clavicle bone



Fig 11: Ectrodactyly of ring finger on right hand



Fig 12: Syndactyly on left foot with middle finger

Management

The dental problem of patient was resolved by doing extraction with 17. Patient was further advised orthodontic corrections for diastema with 11 and 21 and prosthesis with 31 and 41, Extraction with paramolar, Oral prophylaxis was done and was also advised to consult dermatologist for scar.

DISCUSSION

Goldenhar syndrome is a rare disorder which is characterized by incomplete development of the ear, nose, soft palate, lip, and mandible. Classical feature of this disorder includes Hemi facial microsomia, Epibulbar dermoid and Deformity of ears with pre auricular appendage. Ocular manifestations include epibulbar dermoid in 75% of cases.¹⁻²

Involvement of vertebra and ribs are observed in 24% of cases³ which was seen in our case as the inter rib distance was short. Mandibular hypoplasia resulted in facial asymmetry. Facial asymmetry is reported in 65-75% of cases which was also feature of our case. Overall frequency of ear anomalies in 83% are seen was also a feature of our case and a frequency of eye malformations in 66% of patients. Ear abnormalities, ranging from anotia to preauricular tag are considered the most common malformation⁴. Abnormalities are unilateral in 85% of cases and bilateral in 10% to 33% of cases, and the right side is more frequently affected⁵ and in our case there was unilateral involvement was seen and the right side was also affected. Craniofacial anomalies, which include hemifacial microsomia and malar and

maxillary hypoplasias, are present in 50% of patients with this disorder⁶⁻⁷ Underdeveloped mandible (micrognathia), maxilla, and zygomatic bones with hypoplastic muscles for mastication and facial expression.⁸

Central nervous system manifestations are seen in 46% of cases. Diffuse cerebral hypoplasia, Dilated lateral cerebral ventricles (asymptomatic Hydrocephalus), Corpus callosum dysgenesis and frontal hypodensities were the most frequent abnormalities.⁹ Cardiac defects include Ventricular septal defects, Patent ductus arteriosus, Fallot's tetralogy, Coarctation of Aorta and pulmonary stenosis¹⁰. The reported prevalence of cardiovascular anomalies is 5-58%. The other cardiac anomalies reported are Ventricular inversion associated with double outlet Right ventricle, Pulmonary atresia with VSD, Double outlet right ventricle and infra diaphragmatic total anomalous pulmonary venous connections¹¹. In our case no cardiac anomaly was reported. Some researchers suggest that the disorder may be caused by the interaction of genes in combination with environmental factors. It has been suggested that there is a defect in branchial arch development late in the first trimester¹². The ingestion of some drugs such as cocaine, thalidomide, retinoic acid and tamoxifen by the mother were also related to the development of the Disease. Maternal diabetes has also been suggested as an etiologic factor¹³.

In summary, the molecular basis of GS is still unclear, and currently no specific diagnostic test is available. Patients with GS can have multiple congenital anomalies, and they need particular attention to internal abnormalities. Pediatric specialists should consult with ear-nose-throat, orthopedics, neurosurgery, and ophthalmology clinics to decide on the most appropriate treatment plan, which varies with age and systemic associations. Dental care with experienced multidisciplinary team of orthodontists and maxillofacial surgeons is also necessary for good results¹⁴⁻¹⁵

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How to cite this article: Lanjekar A, Bagde L, Sathwane R et.al. Oculo-auriculo-vertebral dysplasia: a rare case report. *International Journal of Research and Review*. 2020; 7(2): 187-191.
