

# Oculo-auriculovertebral Spectrum with Radial Anomaly: A Rare Case Report

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### Abstract

Oculo-auriculovertebral spectrum (OAVS) or the Goldenhar syndrome is a rare developmental disorder with plethora of congenital anomalies which mostly affects structures arising from the first and the second branchial arches. The affected structures include cheekbone, jaws, mouth eyes, ear and vertebrae. The case which we are presenting here had classical features such as microsomia, unilateral microtia, microphthalmia and facial nerve palsy along with vertebral anomalies with additional signs as unilateral hearing loss and radial limb anomalies. OAVS with radial defect and limb abnormalities is a rare presentation and thus reported..

**Key words:** Goldenhar syndrome, ear malformation, hemifacial microsomia, microphthalmia, radial defect, facial nerve palsy, vertebral anomalies.

### Introduction:

First reported by Goldenhar in 1952 as a syndrome which presents with multiple congenital anomalies as malformation of ear and eyes, narrow or missing ear canal (atresia), peri-auricular tags, hemifacial microsomia, hypoplasia of zygomatic, mandible and maxilla bones, vertebral anomalies, epibulbar dermoid and therefore also the name oculoauriculovertebral spectrum (OAVS). Prevalence estimated to be 1 in 4500 live births and male to female ratio of 3:2. It is considered to be the result of blastogenesis defect that involves structures arising from the first and second branchial arches<sup>1,2</sup>. It is mostly sporadic condition and inherited very rarely, therefore aetiology in most cases is difficult to assess<sup>3</sup>. Some cases appear genetic while other occurring in sporadic manner most probably due to environmental factors<sup>4</sup>.

OAVS with radial defects and ectrodactyly is a rare presentation and till date only 32 cases have been reported<sup>5</sup>. Hence we are reporting one such case.

### Case Report:

A 16-year-old Indian rural boy, student of standard X, presented to Physical Medicine and Rehabilitation OPD with complaints of facial asymmetry, malformed external right ear (Fig 1) with hearing loss, neck and upper back deformities and deformed both hands (Fig2), since birth. He was born of non-consanguineous marriage to a 28-year-old, non diabetic mother, at term by normal vaginal route. There was no history of drug intake and exposure to x-rays by mother during her antepartum period. However, there was an episode of bleeding per vagina at 8th week of gestation, for which she took no medical consultation and which stopped on its own. Intrapartum and postpartum periods were uneventful. Developmental milestones were achieved at appropriate time. He had 4 siblings and none in family had any similar complaints of malformations.

General examination of the subject revealed a below average built and nutrition with low weight and a short stature. Height was 145 cm (<50th percentile) and weight 52 kg (<50th percentile). Intelligence was normal. Tone and gait were normal and he had a typical face with facial asymmetry, where the maxillary, temporal and malar bones on the right side were flattened with mandibular hypoplasia. Ophthalmic examination revealed microphthalmia on the right side. There were

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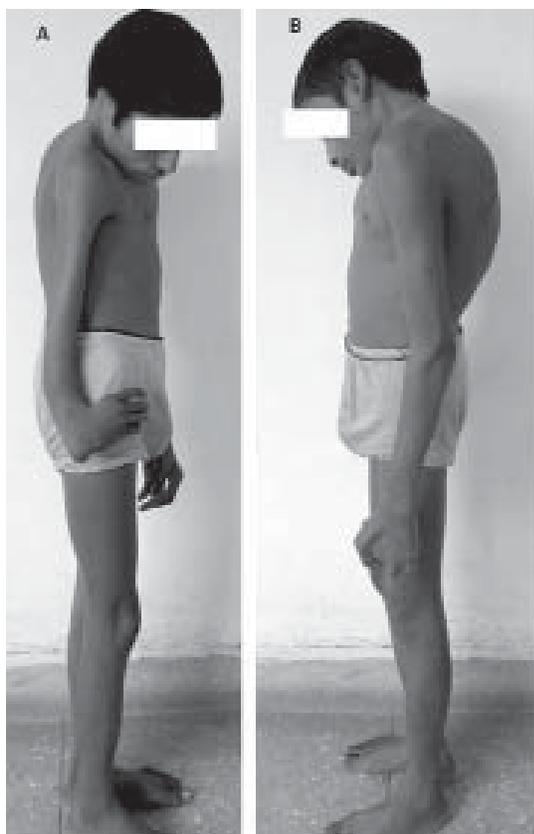
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**Fig 1 - Right Lateral View of the Subject (A) Showing Radial Club Hand and Microtia, and Left Lateral View of the Subject (B) Showing Kyphoscoliosis and Ectrodactyly.**



**Fig 2 - Left Hand of the Subject (C) Showing Ectrodactyly and Right Hand of the Subject (D) Showing Radial Club Hand.**

no epibulbar dermoids, lipodermoids or coloboma. Ipsilateral microtia was seen. CNS examination was normal except facial nerve palsy on the right side with positive Bell's phenomenon. Along with cervicodorsal kyphoscoliosis was seen. Inspection of upper limbs revealed radial club hand on the right side with absent thumb and ectrodactyly on the left side with absence of middle ray and a hypoplastic thumb (Fig 2). No abnormality was found in the lower limbs.



**Fig 3 - Digital Skiagram of the Cervicodorsal Spine of the Subject Showing Features of Kyphoscoliosis.**



**Fig 4 - Skiagram of Left Hand of the Subject Anteroposterior View (E) and Lateral View (F) and Right Hand of the Subject Anteroposterior View (G) and Lateral View (H).**

Hematological and biochemical findings of the patient were found to be within their normal limits. Skiagram of cervicodorsal spine showed left cervicodorsal kyphoscoliosis, with multiple hemivertebrae and segmentation defects (Fig 3). Skiagram of right forearm and wrist with hand showed absence of radius and 5th ray and left hand skiagram showed absence of middle ray (Fig 4).

MRI of cervicodorsal spine showed multiple fusion and segmentation anomalies maximum at upper cervical and upper dorsal region leading to severe kyphoscoliosis. Signal intensity alterations suggestive of focal syrinx at C6-7 levels. No abnormality was detected on MRI brain and CT head. Radiological examination of right ear showed external, middle and inner ear malformations with canal atresia and audiological examination revealed moderately severe conductive hearing loss. NCV of facial and trigeminal nerve revealed right facial and trigeminal neuropathy. There was no cardiovascular, gastro-intestinal, renal and genito-urinary system involvement. However genetic study could not be performed.

Children with OAVS should be thoroughly investigated to know about all the deformities and anomalies present. Treatment of the disorder varied with the age and systemic associations and is mainly cosmetic in uncomplicated cases. The structural anomalies of eyes and ear can be corrected by plastic surgery. Reconstruction of external ear can be one at 6-8 years, and mandibular defect reconstruction by rib bone graft. Use of a hearing aid device is necessary early in life. Good oral hygiene is also emphasised. Prognosis is good in uncomplicated cases.

### Discussion:

OAVS or Goldenhar syndrome is characterised by a variable degree of unilateral or bilateral involvement of the craniofacial structures which arise from the first and the second branchial arches. OAVS with radiological defect characterises a subset within OAVS which also affects the limb primordium<sup>5</sup>, chiefly manifesting as ear malformations, facial asymmetry mandibular hypoplasia and radial defects.

It was first recorded by a German physician Carl Ferdinand Von Arlt in 1845<sup>6</sup>. In 1952, Goldenhar described a patient with triad of accessory tragus, mandibular hypoplasia and ocular dermoid and gave the name Goldenhar syndrome<sup>7</sup>. Not much is known regarding the aetiology of the syndrome but studies suggest that exposure to viruses during pregnancy, abnormal vascular supply to the first arch and abnormal first branchial arch development may all lead to this syndrome. Some authors reported that the disorder may be due to multifactorial inheritance. Baun and Feingold<sup>4</sup> reported this syndrome to be sporadic in over 90 % of the cases while positive family history was also reported in the maternal grandmother and mother of two cases in the series. Maternal diabetes is also thought to be an aetiological factor<sup>8</sup>.

Classical abnormalities include ocular changes such as microphthalmos, epibulbar dermoids, lipodermoids and coloboma. Aural features as periauricular tragi, hearing loss and microtia vertebral anomalies such as scoliosis, hemivertebrae and cervical fusion<sup>9,10</sup>. These abnormalities are unilateral in 85% of the cases and bilateral in 10-33%<sup>11</sup>. Our case had microphthalmos, microtia and scoliosis and multiple hemivertebrae defects.

One study<sup>12</sup> reported 46% incidence of conductive hearing loss in their study. The present case had severe conductive hearing loss on the right side along. Among the craniofacial anomalies which include maxilla malar and mandibular hypoplasia macrostomia, cleft lip and palate this patient had facial asymmetry with malar, mandibular and maxillary hypoplasia. Wang *et al*<sup>8</sup> reported 33% patients with craniofacial abnormalities had facial nerve palsy which was found in the present case too.

Radial club hand and ectrodactyly are rare associations with OAVS. Vendramini *et al*<sup>5</sup> reported that most common anomalies observed in OAVS were hypoplasia of thumb and hypoplasia/agenesis of radius. Tasse *etal*<sup>13</sup> found that anomalies of extremities such as thumb hypoplasia were significantly associated with the short stature. The low weight and short stature in this case shows that these clinical findings are a part of the phenotypic spectrum of OAVS with radial defects.

About 40-60% of cases have congestive heart disease as teratology of Fallots, dextrocardia, transposition of great vessels and right bundle branch block, 70% have renal problems as renal agenesis, ectopic kidney and urethral duplication. However the present case did not have any such anomalies. Other conditions with similar features as Nager syndrome, Holt-Oram syndrome, radial-renal syndrome, facio-auricular radial dysplasia, Fanconi's anaemia and vertebral, anal, cardiac, tracheal, oesophageal, renal and limb association (VACTERL) should also be considered for differential diagnosis.

### Conclusions:

In conclusion we postulate that the patient presented with radial defects apart from the other classical OAVS findings of facial asymmetry, ear malformation, hearing loss and anomalies of first and second branchial arches may characterise a rare subset within OAVS spectrum. This report suggests a rare variety of OAVS which requires molecular analysis and chromosomal analysis for better understanding of the spectrum of the disease.

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### Conflicts of Interest:

There are no conflicts of interest.

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