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Radiological Diagnosis of Atypical Case of Goldenhar Syndrome with Pulmonary Hypoplasia and Dextrocardia: A Case Report

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Abstract **Case Report**

Goldenhar Syndrome, also synonymously known as oculo-auriculo-vertebral spectrum is a rare congenital malformation involving face, eyes, ears and vertebra. The exact etiopathogenesis is not clear but maldevelopment of first and second pharyngeal arches is mentioned in literature. This syndrome is associated with abnormalities in cardiac, renal, pulmonary and nervous system. Some cases present with classic abnormalities where diagnosis is usually clinical. However, atypical cases have also been reported. We report a case of one year old boy presenting with recurrent lower respiratory tract infection and feeding diaphoresis. Extensive examination and investigations revealed left preauricular tag with radiological evidence of butterfly deformity of sixth cervical vertebra, right pulmonary hypoplasia, dextrocardia, patent foramen ovale and pulmonary artery stenonis. These abnormalities are rare but associations have been reported.

Keywords: Goldenhar Syndrome, oculo-auriculo-vertebral spectrum, feeding diaphoresis, dextrocardia.

INTRODUCTION

Goldenhar Syndrome was first described by ophthalmologist Maurice Goldenhar in 1952 as a rare congenital malformation involving face, eyes and ears (Bogusiak *et al*., 2017) (Chaudhary *et al*., 2017). Later, Gorlin *et al*., coined the term oculo-ariculo-vertebral spectrum (OAVS) to encompass the associated vertebral abnormalities (Gawrych *et al*., 2014). The words are usually used synonymously. Although the exact etiopathogenesis is not clear, genetic abnormalities, maternal factors and environmental factors are associated with the syndrome (Schmitzer *et al*., 2018).

Clasically, the features of eye involvement are epibulbar dermoid and coloboma. Auricular abnormalities like microtiaa, preauricular appendages or fistula can be present. Hemifacial microsomia or cleft lip and palate can be present. Vertebral anomalies like hemivertebra, fused vertebra or butterfly vertebra can be seen. Cardiovascular system involvement can present as ventricular septal defect, atrial septal defect, Fallot tetralogy or dextrocardia among many others (Bogusiak *et al*., 2017). Pulmonary hypoplasia is a rare association (Guo *et al*., 2022).

Cases can present with classical symptoms or atypically (Gawrych *et al*., 2014) (Jayaprakasan *et al*., 2023) (Chaudhary *et al*., 2017).. We present an atypical case of one year boy with auricular and vertebral involvement who had pulmonary hypoplasia with dextrocardia. This case highlights the role of radiology in adding evidence to diagnosis in atypical cases.

CASE PRESENTATION

We present a case of one year old male child born of non-consanguineous marriage who presented to pediatric outpatient department with features of lower respiratory tract infection. He had history of recurrent episodes of pneumonia and feeding diaphoresis since birth. There was no history of cyanosis, loss of consciousness, abnormal movement of body and inconsolable crying. He seemed apparently normal at birth. Antenatal, intra-natal and postnatal periods were uneventful. He was third child in the family and previous two siblings were normal compared to peer groups.

Extensive examination and investigations were done as the patient presented with recurrent disease to

find out co-existing abnormalities in other systems. On examination, the patient had vital signs within normal range. He had left sided pre-auricular appendage. Both ears had normal morphology with bilaterally normal hearing. Bilateral eye examination revealed no external abnormality with normal fundus.

Bilateral audiometric examination revealed no hearing loss. Spine X-ray showed presence of butterfly vertebra deformity of sixth cervical vertebra (C6) (Figure 1). Chest radiography showed dextrocardia and right pulmonary hypoplasia (Figure 2 and 3). Echocardiography revealed Patent Foramen Ovale (PFO) with left to right shunt. Interventricular septum was intact. Right pulmonary artery was hypoplastic but biventricular function was normal. Final diagnosis of Goldenhar Syndrome with right pulmonary hypoplasia and dextrocardia was made.

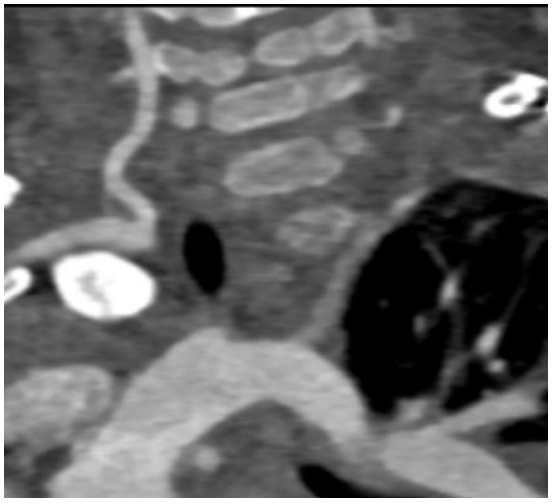

Figure 1: Cervical spine X-ray showing butterfly vertebra deformity of sixth cervical vertebra

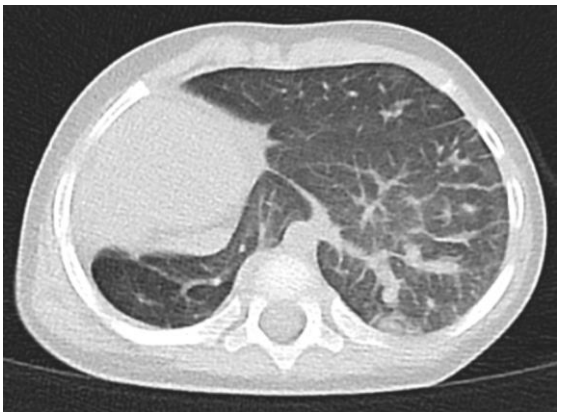

Figure 2: Computed Tomography of thorax (axial section) showing right pulmonary hypoplasia with dextrocardia

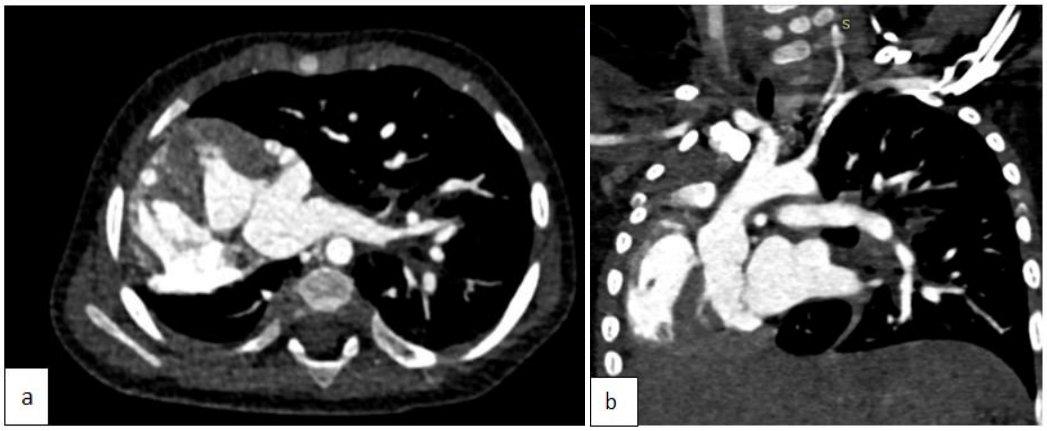

Figure 3: Contrast enhanced computed tomography of thorax ('a' axial section and 'b' coronal section) showing right pulmonary hypoplasia with dextrocardia

DISCUSSION

Goldenhar Syndrome was first described by ophthalmologist Maurice Goldenhar in 1952 as a rare congenital malformation involving face, eyes and ears (Bogusiak *et al*., 2017) (Chaudhary *et al*., 2017). Later, Gorlin *et al*., coined the term oculo-ariculo-vertebral spectrum (OAVS) to encompass the associated vertebral abnormalities (Gawrych *et al*., 2014). Goldenhar Syndrome is associated with anomalies in cardiovascular, urogenital, respiratory and nervous system (Bogusiak *et al*., 2017) (Al Ghamdi, 2022). Its incidence is one in every 3500 to 5000 live birth with 3:2 male to female ratio (Jayaprakasan *et al*., 2023).

It is suggested that maldevelopment of first and second pharyngeal arches during embryonic period leads to this syndrome (Chaudhary *et al*., 2017). Goldenhar syndrome has been described to have multifactorial origin. It usually occurs sporadically but familial cases have also been reported (Choudhury & Kapoor, 2017).. Familial cases have been noted to occur in consanguinous marriage with autosomal dominant, recessive or multifactorial inheritance pattern (Gawrych *et al*., 2014) (Chaudhary *et al*., 2017) (Muñoz-Pedroza & Arenas-Sordo, 2013). Genetic defect and chromosomal anomalies have been reported. Association with maternal drug intake (ibuprofen, isotretinoin), maternal diabetes, environmental factors and antepartum hemorrhage is also reported (Schmitzer *et al*., 2018). In our case, there was no familial association with no exposure to maternal factors as described earlier. Genetic analysis was not performed.

Goldenhar Syndrome can present with wide spectrum of abnormalities which is usually unilateral but can be bilateral in some cases. Facial, ocular, auricular and vertebral anomalies co-exist in classical cases of Goldenhar Syndrome. Facial abnormality can range from mild to severe facial asymmetry with hemifacial macrosomia, mandibular hypoplasia, high arched palate and cleft lip and palate among many others. Ocular abnormalities like epibulbar dermoid, coloboma and microphthalmia are described. Auricular involvement can present as preauricular appendages, microtia, atresia of external auditory canal with occasional involvement of middle and inner ear. Vertebral malformations include fused vertebra, hemivertebra, atlas occipitalization, butterfly vertebra, and many others (Bogusiak *et al*., 2017) (Gawrych *et al*., 2014). Although diagnosis is usually clinical, radiology plays vital role in finding associated abnormalities. In our case radiological analysis helped to come to conclusion as most of the features were not apparent on physical examination. Children can present with wide range of severe clinical manifestations (Gawrych *et al*., 2014). However, some atypical cases have also been reported (Jayaprakasan *et al*., 2023) (Chaudhary *et al*., 2017) Jayaprakasan *et al*., reported a case in which the child presented with unusual features of developmental and speech delay. She had ocular and auricular features but classical facial and vertebral abnormalities were absent (Jayaprakasan *et al*., 2023). Chaudhary *et al*., reported a case with absence of vertebral abnormalities with presence of eye, ear and facial abnormalities (Chaudhary *et al*., 2017). We present an atypical case with presence of auricular and vertebral involvement as preauricular tag and butterfly vertebra respectively. There is absence of typical facial and ocular abnormality in our case.

Abnormalities of multiple organs have been described in literature. Pulmonary hypoplasia with dextrocardia in Goldenhar Syndrome has been reported in few cases. All cases had auricular involvement in some form while three cases were associated with preauricular tags. All four cases had right sided pulmonary hypoplasia. All males had right ipsilateral facial/ocular/auricular abnormalities while the female had left sided lesion (Chaudhary *et al*., 2017) (Fan *et al*., 2015) (Maymon *et al*., 2001) (Guo *et al*., 2022). Our case is a male with left sided auricular involvement with right sided pulmonary hypoplasia. This contradicts with the proposed theory of laterality in males (Guo *et al*., 2022). Cardiovascular abnormalities are seen in about 5 to 58% cases (Choudhury & Kapoor, 2017) (Nakajima *et al*., 1998). Ventricular septal defect, atrial septal defect, Tetralogy of Fallot and conotruncal abnormalities are commonly associated (Bogusiak *et al*., 2017). Our patient had Patent Foramen Ovale with hypoplastic pulmonary artery. Pulmonary artery abnormality is seen in cases of pulmonary hypoplasia either in the form of dilatation, stenosis or ectopic origin of artery (Guo *et al*., 2022) (Chaudhary *et al*., 2017) (Maymon *et al*., 2001). Association with urogenital and central nervous system has also been reported (Bogusiak *et al*., 2017). Such abnormalities were not noted in our case.

CONCLUSION

Goldenhar syndrome is a rare congenital abnormality which can present with wide spectrum of facial, ocular, auricular and vertebral abnormalities. Patients can present with classical or atypical symptoms. Any patient with suspected Goldenhar syndrome should undergo extensive examination and investigations to look for abnormalities in other systems. Radiological evaluation thus plays vital role in every case with special role in atypical cases. Cardiovascular and respiratory system involvement is commonly seen. Pulmonary hypoplasia with dextrocardia and associated pulmonary artery abnormalities are rare but can occur in Goldenhar Syndrome.

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