

Case Report

***Situs Inversus Totalis* with Complex Cardiac Malformations in Goldenhar Syndrome**

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ABSTRACT

Goldenhar syndrome is characterized by a derangement in the morphogenesis of the first and second branchial arches. Vertebral, ocular and cardiac anomalies are

occasionally encountered in such children. A rare association of Goldenhar syndrome with *situs inversus totalis* and multiple cardiac malformations is reported.

KEYWORDS: branchial arches, cardiac anomalies

INTRODUCTION

Goldenhar syndrome, also known as Oculoauriculovertebral dysplasia, represents a spectrum of defects associated with the development of the first two branchial arches^[1,2]. Congenital heart disease is reported as an 'occasional' feature in Goldenhar syndrome^[3]. Ventricular septal defect, patent ductus arteriosus, Fallot's tetralogy and coarctation of aorta occur with decreasing frequency^[3]. Cardiac malposition and abnormal situs are extremely rare in this disorder. Here we report a baby with several features of the syndrome in addition to *situs inversus totalis* and multiple cardiac lesions.

CASE REPORT

A twelve-day old male infant born of non-consanguineous parentage was admitted with tachypnoea and cyanosis since birth. He had a downward slant of the eyes, bilateral microtia with accessory tragus, preauricular pits, facial asymmetry due to depressor anguli oris muscle deficiency and macrostomia (Fig. 1). Clinical features were consistent with that of Goldenhar syndrome.

Peripheral pulses were palpable normally. Cardiac apical impulse was located in the fourth right intercostal space in the midclavicular line. The second heart sound had a wide fixed split. A grade two ejection systolic murmur was audible in the second right intercostal space. There was no evidence of cardiac failure.

Barium contrast radiograph of chest and abdomen confirmed dextrocardia with situs inversus (Fig. 2). Ultrasonogram of abdomen confirmed the abnormal situs and ruled out renal anomalies. Electrocardiogram showed inverted P

waves in limb leads with extreme right axis QRS morphology, suggestive of dextrocardia. Septal Q waves were absent in corrected lead positions, indicating defective trans-septal activation. Echocardiography demonstrated D-loop dextrocardia, atresia of tricuspid valve, secundum atrial septal defect, a large ventricular septal defect and pulmonary stenosis.

DISCUSSION

Goldenhar syndrome is a heterogenous constellation of malformations, classically involving the face, eyes and ears^[4]. The characteristic combination of external ear anomalies and ipsilateral facial underdevelopment is the hallmark of the syndrome. It usually occurs in a sporadic pattern^[5]. But autosomal dominant inheritance can rarely be encountered, as reported by Summitt in 1969^[6].

The abnormalities are usually asymmetric and unilateral in 70% of cases. The characteristic facial features are malar, maxillary and mandibular hypoplasia. There may be macrostomia and deficiency of depressor anguli oris muscle, as in this baby^[1,2,7]. Malformations of external ear include accessory preauricular tags and pits, as in the present case. Others, such as microtia, middle ear deafness and labyrinthine malformation may also occur^[1,2,8,9]. Vertebral anomalies like hemivertebrae and hypoplasia of cervical vertebrae were absent in this infant^[10].

Ocular anomalies are less common and include epibulbar dermoid, notched upper eyelid, strabismus and microphthalmia^[11]. However, except for the antimongloid slant, the eyes of this baby were normal. An ultrasonogram has ruled out genitourinary malformations like renal agenesis,

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Fig. 1: Macrostomia and malformed ear with accessory tragus and preauricular pits



Fig. 2: Roentgenogram showing dextrocardia and barium contrast depicting stomach on the right side

ectopic kidneys, urethral duplication and vesicoureteric reflux^[12].

Cardiac involvement is described as an 'occasional' feature of Goldenhar syndrome. The usual lesions are ventricular septal defect, patent ductus arteriosus, Fallot's tetralogy and coarctation of aorta^[1,3,13]. Interestingly, this baby had a situs inversus totalis with multiple cardiac malformations.

The occurrence of situs inversus along with other cardiac lesions like tricuspid atresia, secundum atrial septal defect, ventricular septal

defect and pulmonary atresia has seldom been reported in either the first or the second arch syndromes. This case is reported because of the complex and unusual pattern of cardiovascular involvement, unlike other children with Goldenhar syndrome.

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