The Turkish Journal of Pediatrics 28:77 - 84, 1986

INTERRUPTED AORTIC ARCH, MIRROR-IMAGE DEXTROCARDIA WITH SITUS INVERSUS AND SPRENGEL'S ANOMALY*

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Key words : aortic arch, congenital abnormalities, dextrocardia, scapula, Sprengel's deformity

Interrupted aortic arch (IAA) is an uncommon cardiac anomaly occurring in 19 per million live births[1]. Three locations of the arch interruption have been described: distal to the left subclavian artery (Type A, 42% of cases); distal to the left common carotid artery (Type B, 53%); and between the innominate and left common carotid arteries (Type C, 4%)[2,3].

Although many cases of IAA in association with other cardiac anomalies have been reported, the combination of IAA, mirror-image dextrocardia and Sprengel's anomaly has not yet been described in the literature.

The purpose of this communication is to report what we believe to be the first case of IAA in association with mirror-image dextrocardia and Sprengel's anomaly.

Case Report

A seven-month-old girl baby was admitted to the Pediatric Cardiology Unit of the Gülhane Military Academy of Medicine, with a high fever and respiratory difficulty which had been present for the preceding six days. According to the history, she had twice been treated with antibiotics, once at four months and once at five months, because of suspected pulmonary infections. There was no known family history of malformation. The pregnancy and the term delivery had been unremarkable and the birth weight was 3.75 kg.

Physical examination revealed a pale, tachypneic child who was underdeveloped for her age; weight was 6 kg and height was 65 cm. On oscultation, the cardiac apex was right-sided and there was a grade 2/6 systolic murmur along the right sternal border. The second heart sound was accentuated, all peripheral pulses

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were normal and there was no remarkable difference between brachial and femoral blood pressures. Fine crepitations were noted over both lung fields and breath sounds were diminished over the base of the left lung. The liver was palpable 4 cm below the left costal margin. An electrocardiogram showed the typical findings of mirror-image dextrocardia and the chest X-ray revealed dextrocardia with situs inversus, pulmonary plethora and a large consolidation in the lower lobe of the left lung. In addition, there was Sprengel's deformity of the left scapula associated with hemivertebrae in the sixth and seventh cervical segments and some segmentation anomalies in the left-sided ribs (Figure 1).

Key to Figures

L	: Liver	RV	: Right ventricle
SB	: Stomach bubble	CS	: Coronary sinus
DA	: Ductus arteriosus	LV	: Left ventricle
PA	: Main pulmonary artery	RA	: Right atrium
RPA	: Right pulmonary artery		

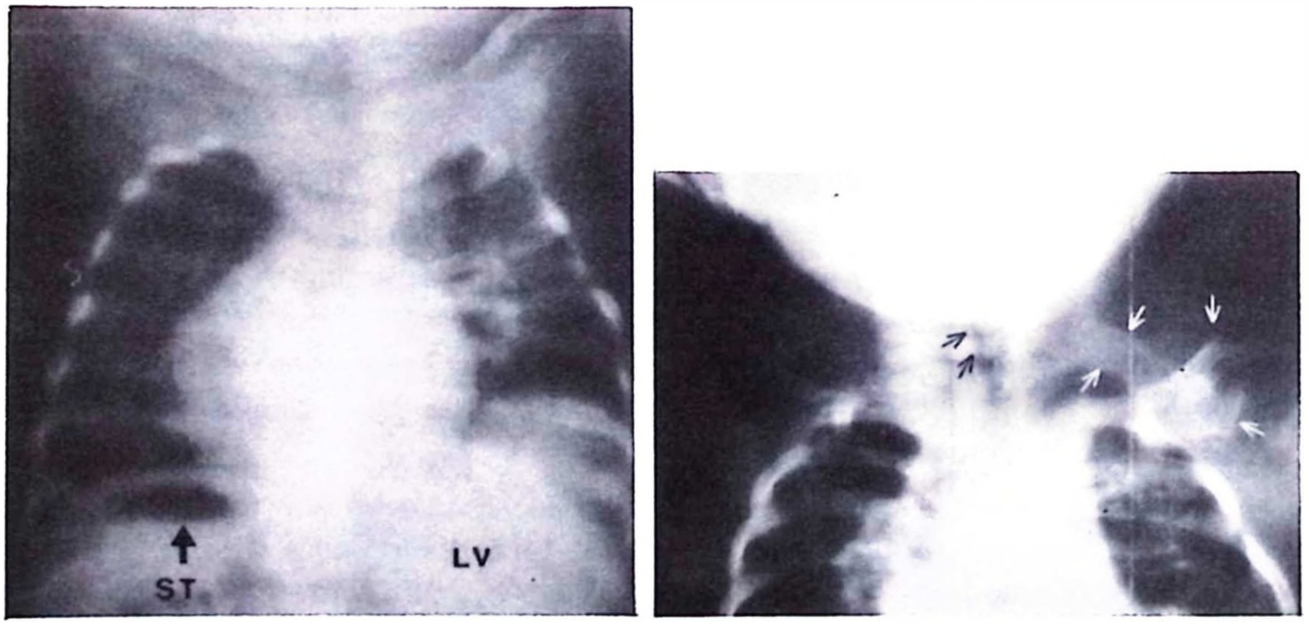

Figure 1

(a) Chest roentgenogram of the patient showing the dextrocardia with situs inversus and anomalies of left-sided ribs. (b) Sprengel's anomaly of the left scapula and hemivertebrae in the sixth and seventh cervical segments. (White arrows indicate the position of the left scapula, black arrows indicate hemivertebrae).

Other investigations including intravenous pyelography, a complete radiologic examination of the gastrointestinal system and a scintigraphic examination of the liver and spleen revealed no abnormality except a situs inversus of the abdominal viscera. There was no chromosomal abnormality, the serum calcium level and T-lymphocyte functions were normal and a tuberculin skin test showed six mm induration.

Treatment with antibiotics, digoxin, diuretics and oxygen was started. The general condition of the patient improved gradually and the consolidation in the left lung was reduced within twenty days.

Cardiac catheterization (Table I) and angiocardiography confirmed the diagnosis of mirror-image dextrocardia with situs inversus. Injection of contrast medium into the ascending aorta clearly showed the interruption of the aortic arch distal to the

TABLE I

Cardiac Catheterization Data

	O_2 Saturation (%)	Pressure (mmHg)
IVC	72	
SVC	69	
RA	70	(8)*
RV	75	85/6**
PA	78	85/50(70)
DAo	78	85/50(70)
LA	86	(15)
LV	86	95/10**
AAo	86	95/60(75)
Q_p / Q_s	2/1	
PVR	6.2 mmHg/L/min/m^2	

* Values in parentheses: mean pressures

** End-diastolic pressures.

AAo: Ascending aorta, DAo: Descending aorta, IVC: Inferior vena cava, LA: Left atrium, LV: Left ventricle, PA: Pulmonary artery, PVR: Pulmonary vascular resistance, Q_p / Q_s: Pulmonary-to-systemic flow ratio, RA: Right atrium, RV: Right ventricle, SVC: Superior vena cava.

right subclavian artery (Figure 2-a). The aortic valve, with its three cuspids, was normal and there was no pressure gradient through it. The descending aorta was filling via a large persistent ductus arteriosus during right ventricular injection (Figure 2-b). In addition, a large subpulmonic ventricular septal defect and two

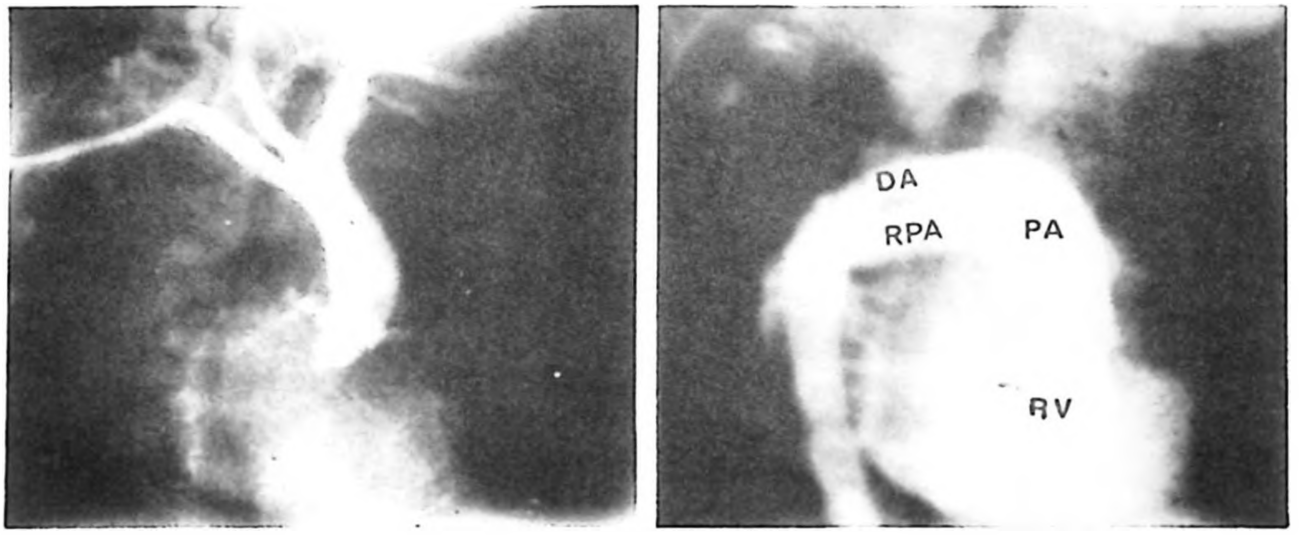

Figure 2

(a) Aortography of the patient in 45° right anterior oblique projection showing the interruption distal to the right subclavian artery.
(b) Right ventriculography in 45° right anterior oblique projection showing the filling of the descending aorta through the ductus arteriosus.

superior venae cavae were demonstrated. The right superior vena cava was connected to the coronary sinus (Figure 3).

With these findings, it was decided that the aortic interruption should be repaired and pulmonary banding performed. The patient was referred to the surgical department but the family refused to let the child be operated on and she was discharged from the hospital one and a half months after admission.

Discussion

IAA usually results in severe congestive heart failure and death in early infancy[1,3,4]. If the ductus remains widely patent, the blood flow to the descending aorta is provided by a right-to-left shunt through the ductus arteriosus. In the present case, the ductus arteriosus is widely patent and the pulses distal to the interruption have been well maintained. In addition, pulmonary vascular resistance is sufficiently high to ensure adequate right-to-left shunting through the ductus arteriosus.

However, a more interesting aspect of this case is the combination of IAA with other abnormalities such as dextrocardia, two superior venae cavae and

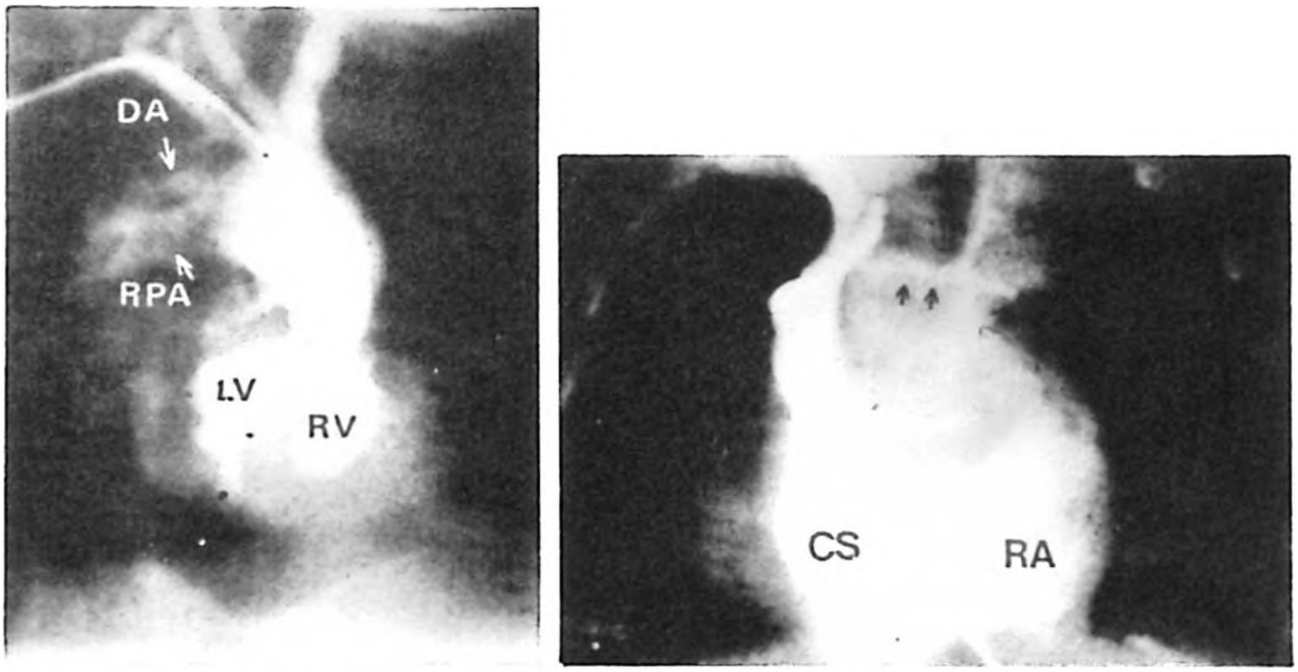

Figure 3

(a) Left ventriculography in 45° right anterior oblique projection and (b) Right superior venography in frontal projection (Arrows indicate the communication between the two superior venae cavae).

Sprengel's anomaly associated with hemivertebrae and segmentation anomalies of the ribs (Table II). As far as we know, there is no case in the literature with this combination although a few cases of dextrocardia associated with aortic coarctation have been reported[5,6]. However, there was no skeletal malformation in these patients and aortic interruption was not so pronounced.

Morphogenesis of IAA has been widely concluded to be due to reduced blood flow through the ascending aorta *in utero*[3,7]. Conal maldevelopment resulting in displacement of the crista supraventricularis into the left ventricle has been implicated as the main factor. Other lesions implicated have included mitral valve obstruction, truncus arteriosus with preferential flow to the pulmonary circulation and hypoplasia of the ventricle which gives rise to the aorta[7,8].

In our case, there was no evidence of mitral valve obstruction nor was there a truncus arteriosus or left ventricular hypoplasia. Aortic interruption can be explained by reduced blood flow into the ascending aorta due to the large left-to-right shunting through the ventricular septal defect. The type of the interruption can be defined as a mirror-image of type A.

Van Praagh et al[3] concluded that type A interruption occurs after the left subclavian artery has ascended to its definitive level, that is, after the seventh embryonic week. The leftward displacement of the parietal band resulting in a subpulmonic ventricular septal defect which is the basic pathology responsible for the IAA, occurs during the sixth week of gestation[9].

TABLE II

Possible Embryonic Basis and Occurrence Periods of Malformations

Malformation	Embryonic basis	Period
Dextrocardia	– Intrinsic faulty development	4th week
Right PDA	– Persistence of the right 6th aortic arch	5th "
Right aortic arch	– Persistence of the right 4th aortic arch and right dorsal aorta instead of the left ones	6th "
IAA Type-A		
VSD	– Failure of completion of IVS	6th "
Aortic interruption	– Reduced blood flow into the ascending aorta	8th "
Sprengel's anomaly	– Failure of the scapula to descend	7-8th "
Costal anomalies	– Segmentation anomalies of ribs	6-8th "
Hemivertebrae	– Failure of condrification of vertebral bodies	8th "
Persistent right superior vena cava	– Failure of regression of the right anterior cardinal vein in mirror-image dextrocardia	8th onward

PDA : Persistent ductus arteriosus
VSD : Ventricular septal defect
IVS : Interventricular septum

Sprengel's anomaly is secondary to the failure of the scapula to descend to its normal position, and is sometimes associated with condrification defects of vertebral bodies (hemivertebrae) and segmentation anomalies of the ribs. As shown in Table II, embryonic limb rotation and the descent of the scapula to its normal position, like the descent of the subclavian arteries, occurs during the seventh and eighth weeks of gestation[10]. Moreover, segmentation of the ribs,

condrification of vertebral bodies and regression of the left superior vena cava (the right in mirror-image dextrocardia) also occur around this stage of embryonic life[10].

However, dextrocardia and situs inversus are believed to be secondary to an intrinsic faulty development of the heart and viscera during the early single tube stage, that is, during the fourth embryonic week[9]. In mirror-image dextrocardia, involution of the left sixth aortic arch, in other words, persistence of the right ductus arteriosus takes place during the fifth week of gestation[3,9,10]. Involutions of the left fourth aortic arch and left dorsal aorta, which are responsible for a right-sided aortic arch, occur within the sixth embryonic week like completion of the interventricular septation[9].

In the present case, it is clear that all of these malformations occurred between the fourth and eighth weeks of gestation probably in an uninterrupted succession. Unfortunately, it is not possible for us to talk about the true etiology of this combination since neither the history of the pregnancy, nor the laboratory investigations performed on the mother and the child provided any information about a special noxious effect on the embryonic development.

Summary

A rare case of interrupted aortic arch (IAA) associated with mirror-image dextrocardia and Sprengel's anomaly of the scapula is briefly described. Additional cardiac anomalies include a ventricular septal defect, a large persistent ductus arteriosus and bilateral superior venae cavae. As far as we know this is the first such case to be reported in the literature.

REFERENCES

1. Fyler DC, Buckley LP, Hellenbrand WE, Cohn HE. Report of the New England regional infant cardiac program. *Pediatrics* 65 (Suppl):375, 1980.
2. Celoria GC, Patton RB. Congenital absence of the aortc arch. *Amer Heart J* 58:407, 1959.
3. Van Praagh R, Bernhard WF, Rosenthal A, et al. Interrupted aortic arch: surgical treatment. *Am J Cardiol* 27:200, 1971.
4. Collins-Nakai RL, Dick M, Parisi-Buckley L, et al. Interrupted aortic arch in infancy. *J Pediatr* 88:959, 1976.
5. Zelikovsky A, Vidne B, Levy MJ. Mirror-image dextrocardia with situs inversus and coarctation of the aorta. *Chest* 66:297, 1974.
6. Kamau P, Miles V, Toews W, et al. Surgical repair of coarctation of the aorta in infants less than six months of age: including the question of pulmonary artery banding. *J Thorac Cardiovasc Surg* 81:171, 1981.

7. Moore GW, Hutchins GM. Association of interrupted aortic arch with malformations producing reduced blood flow to the fourth aortic arches. *Am J Cardiol* 42:467, 1978.
8. Cottrell AJ, Holden MP, Hunter S. Interrupted aortic arch type A associated with congenitally corrected transposition of great arteries and ventricular septal defect: successful direct aortic anastomosis and pulmonary artery banding in an infant. *Br Heart J* 46:671, 1981.
9. Wilson JG. Embryology of the heart and great vessels. In Luisada AA (ed). *Development and Structure of the Cardiovascular System*. New York: McGraw Hill, 1961, pp 1-19.
10. Williams PL, Warwick R. *Gray's Anatomy: Embryology*. New York: Churchill Livingstone, 1980, pp 140-153.