Truncus Arteriosus Associated with Mitral Atresia and a Hypoplastic Left Ventricle

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SUMMARY. The association of truncus arteriosus with congenital left heart abnormalities is rare. We describe a case of truncus arteriosus associated with mitral atresia and a hypoplastic left ventricle. In 11 other patients diagnosed with truncus arteriosus, none had left heart hypoplasia. Despite the truncal root being primarily or entirely committed to the right ventricle, more right than left heart abnormalities have been found. This is in contrast to double-outlet right ventricle in which left heart abnormalities are more often associated.

KEY WORDS: Truncus arteriosus — Hypoplastic left heart syndrome — Mitral atresia — Echocardiography

Persistent truncus arteriosus is an uncommon congenital cardiovascular malformation comprising 1–2% of congenital cardiac abnormalities. The association of truncus arteriosus with any kind of "single ventricle" is extremely rare and even rarer is the association of truncus arteriosus with a hypoplastic left heart. In truncus arteriosus, a single great artery arises from the heart. The coronary arteries, as well as the pulmonary and systemic arteries must originate from the single great artery. In many of the early descriptions of truncus arteriosus associated with a "single ventricle," the origin of the coronary arteries was not described. Therefore, it is likely that many of those hearts represented aortic atresia, especially if mitral atresia was also present, instead of truncus arteriosus. We describe an unusual patient, who had both truncus arteriosus and mitral atresia with a hypoplastic left ventricle.

Case Report

A 1-day-old female infant was transported to our hospital because of poor feeding, cyanosis, and tachypnea. She was 38 weeks gestation with Apgar scores of 8 and 9 at 1 and 5 min, respectively, and a birth weight of 2.1 kg.

On physical examination she had low set ears and thin lips but no other craniofacial abnormalities. Heart rate was 120, respiratory rate 40, and blood pressure 72/41 in all extremities. On cardiovascular examination there was a hyperdynamic precordium, a quadruple rhythm, a 2/6 systolic ejection murmur at the left sternal border, and a 1/6 soft diastolic murmur along the left sternal border. There was an atresia with a rectovaginal fistula. No other abnormalities were noted.

Chest x-ray showed mild cardiomegaly with increased pulmonary vascular markings. Electrocardiogram showed an axis of +110° and right ventricular hypertrophy. Capillary blood gas showed a pH of 7.36, a Pco2 of 39, and bicarbonate of 21. On 30% O2 oximetry showed the saturation to be 90%. A chromosomal analysis and abdominal ultrasound were normal.

The echocardiographic evaluation revealed situs solitus of the atria with atrioventricular concordance. There was mitral atresia with a hypoplastic left ventricle (left ventricular end-diastolic dimension = 7 mm; normal, 11–16 mm) and small left atrium (left atrium = 6 mm; normal, 6–10 mm). The right heart structures were enlarged but otherwise appeared normal. There was a ventricular septal defect (Fig. 1) and a patent foramen ovale. There was a single, large great vessel arising entirely from the right ventricle (Figs. 1B and 2). The single semilunar valve was a quadricuspid valve with thickened leaflets and severely insufficient (Fig. 3). The right coronary artery arose from the right-anterior cusp and the left coronary artery from the left-posterior cusp. The pulmonary arteries arose separately but close to each other, the right posteriorly and the left posterolaterally (Fig. 4). The aortic arch and descending aorta were normal. No patent ductus arteriosus was seen. There was a left superior vena cava, which drained into the coronary sinus.

At 7 days of age she developed episodes of supraventricular tachycardia, as well as increasing congestive heart failure. She was started on digitalis and furosemide. Given her complex intracardiac anatomy which would have required truncal valve replacement and pulmonary artery banding in a heart with only a functional right ventricle, she was felt to be too high risk for surgical intervention. She continued to be treated medically but died at 15 days of age. Autopsy was refused.

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Fig. 1. (A) Apical four-chamber view showing mitral atresia (arrow) with a hypoplastic left ventricle (LV) and a ventricular septal defect (arrowhead). (B) Parasternal long-axis view showing the truncus arteriosus (TA) arising from the right ventricle (RV), the hypoplastic left ventricle (LV), and the ventricular septal defect (arrowhead). CS, coronary sinus; LA, left atrium; RA, right atrium.

Fig. 2. Apical two-chamber view showing truncus arteriosus (TA) arising from right ventricle (RV) and a pulmonary artery (PA) arising from the truncal root and coursing posteriorly.

Other Cases

Eleven patients with truncus arteriosus have been diagnosed using two-dimensional echocardiography at our institution since 1983. In all 11 patients the degree of truncal commitment to the ventricular chambers could be determined. In none of the patients was the truncus arteriosus committed to the left ventricle. In seven patients there was mild commitment to the right ventricle (50-69%), in three there was moderate commitment (70-90%), and in one the truncus arteriosus was entirely committed to the right ventricle. In none of these patients was there hypoplasia of the left heart.

Discussion

Truncus arteriosus is characterized by a single arterial trunk from which the coronary, pulmonary, and systemic arteries originate. A review of the literature indicates that only a few cases have been associated with "single ventricle" anatomy. In most case reports of a "single ventricle" associated with truncus arteriosus, there has been either a common atrioventricular value [5, 8, 9, 14, 18] or an abnormality or atresia of the tricuspid valve [2-6, 9]. There have been associated mitral valve abnormalities reported [4, 9, 10, 16, 18], but in only two cases, where mitral atresia was associated with truncus arteriosus, have the coronary artery origins been described [1, 13]. Without identifying the coronary artery origins, the diagnosis of aortic atresia cannot be eliminated. The presence of a quadricuspid valve does not eliminate aortic atresia, since such a valve has been described in aortic atresia [9], as well as in tetralogy of Fallot [17]. Quadricuspid aortic valves have also been described in otherwise normal hearts though the valves are usually functionally tricuspid [7].

The truncal root commonly arises mostly or entirely from the right ventricle [4, 11, 16]. During the embryologic development of the heart, the primitive truncus arises from the bulbus cordis which eventually comprises much of the definitive right ventricle. An arrest in the normal aorticopulmonary septation of the truncus arteriosus could be associated with an arrest in the rotation and shifting of the truncus arteriosus toward the primitive ventricle leaving it related mostly or entirely to the right ventricle. An arrest early in development could also explain its association with a common atroventricular valve and tricuspid valve abnormalities.

Given that altered fetal cardiac blood flow patterns have been hypothesized to cause certain cardiac abnormalities, then one might expect more left heart and mitral valve abnormalities to be associated with truncus arteriosus where the truncal root arises from the right ventricle. If abnormal fetal cardiac hemodynamics can cause cardiac abnormalities, then an incidence of mitral valve disease similar to that seen in double-outlet right ventricle could be expected (12.5%) [15]. However, only one of our 12 cases (8%) had a left heart abnormality. Despite the low incidence of mitral valve disease in truncus arteriosus, there is a high incidence of interrupted