Absence of Pulmonic Valve Associated with Double-Outlet Right Ventricle


SUMMARY
The case of double-outlet right ventricle with absence of the pulmonic valve described illustrates well the problems inherent in the differentiation of this condition from Fallot’s tetralogy.

Additional Indexing Words:
Congenital heart disease Right ventricular obstruction Tetralogy of Fallot
Pulmonary arterial regurgitation

Absence of the pulmonic valve is a rare congenital lesion which is usually associated with tetralogy of Fallot. We have recently observed a patient in whom this anomaly coexisted with “double-outlet right ventricle.” This case is reported because a review of the literature has failed to reveal a similar case and because certain difficulties inherent in the differentiation of double-outlet right ventricle with right ventricular obstruction from tetralogy of Fallot are well illustrated by this case.

Report of Case
The patient, G.A., was the result of an uncomplicated pregnancy and delivery and weighed 2,840 g at birth. Cyanosis was noted at birth, and a heart murmur was first detected at age 2 days. The patient failed to gain, manifested intermittent cyanosis, and at age 3 weeks suffered an hypoxic spell. He was digitalized, maintained in oxygen, and at age 4 weeks was transferred to the Colorado General Hospital.

Physical examination at that time revealed a mildly cyanotic, poorly nourished male infant who was 49 cm long and weighed 2,640 g. The pulses were normal. There was a moderate right ventricular lift. A grade III/VI harsh systolic murmur was maximal in the third intercostal space at the left sternal border. The second heart sound was loud and single and was followed by a medium pitched decrescendo murmur along the middle and the lower left sternal border. The first heart sound was normal. The lungs were clear to auscultation. The liver and spleen were not enlarged and there was no edema.

The electrocardiogram (fig. 1) demonstrated right ventricular hypertrophy. A chest x-ray revealed an increased cardiothymic silhouette and normal to reduced pulmonary vascularity. The great vessels were not well defined by this study.

The patient was maintained on digoxin and at age 5 weeks underwent cardiac catheterization because of suspected tetralogy of Fallot with absence of the pulmonic valve. Blood oxygen saturations were 31% in the superior vena cava, 59% in the right atrium, 59% in the right ventricle, 63% in the main pulmonary artery, 61% in the left pulmonary artery, 82% in the left atrium, and 61% in the femoral artery. Pressures were 3 mm Hg in the right atrium with A and V waves of 7 and 3 mm Hg, respectively, 84/0-4 mm Hg in the right ventricle and 20/11 mm Hg in the left pulmonary artery.

Cineangiocardiology demonstrated findings which were considered consistent with the clinical diagnosis of tetralogy of Fallot with absent pulmonary valve. A large ventricular septal defect, a narrow pulmonic annulus, and a right aortic arch were noted. Anastomosis of the right pulmonary artery to the ascending aorta was scheduled.

Thoracotomy revealed that the aorta arose anteriorly from the right ventricle and the pulmonary artery appeared to originate from the posterior left ventricle. It was thought that there was...
complete transposition of the great vessels. The anticipated shunting procedure was not performed because of technical problems at the time of operation. The patient appeared to tolerate the thoracotomy well but died about 12 hours thereafter.

Postmortem examination revealed a slightly enlarged heart from which the great vessels arose as separate anatomic structures. The pulmonary artery was leftward and in the same coronal plane as the aorta. The main pulmonary artery was somewhat dilated. No ductus or ligamentum arteriosum was identified. The right and left coronary ostia were rotated about 90 degrees to the left of their normal positions. The distribution of the main coronary arteries was otherwise normal. The venae cavae entered a normal right atrium in the usual manner. There was a 9 by 3-mm atrial septal defect at the site of the fossa ovalis. The tricuspid valve appeared normal. There was a 9 by 4-mm ventricular septal defect inferior and posterior to a very prominent crista supraventricularis. The junction of the anterior leaflet of the mitral valve and the septal leaflet of the tricuspid valve formed the posterior wall of the ventricular septal defect. The aorta and pulmonary artery arose entirely from the right ventricle, anterior to the crista supraventricularis and ventricular septal defect. There was no continuity of the anterior mitral leaflet and the aortic valve. These anatomic relationships are illustrated in figure 2. In addition to these anomalies, the pulmonic valve was absent. There was some narrowing of the pulmonary annulus at the usual site of the pulmonic valve. A 1-mm circumferential rim of connective tissue protruded into the pulmonary arterial lumen at this point. This structure was grossly and microscopically identical to that previously described as absent pulmonary valve. The junction of the anterior leaflet of the mitral valve and the septal leaflet of the tricuspid valve formed the posterior wall of the ventricular septal defect. The aorta and pulmonary artery arose entirely from the right ventricle, anterior to the crista supraventricularis and ventricular septal defect. There was no continuity of the anterior mitral leaflet and the aortic valve. These anatomic relationships are illustrated in figure 2. In addition to these anomalies, the pulmonic valve was absent. There was some narrowing of the pulmonary annulus at the usual site of the pulmonic valve. A 1-mm circumferential rim of connective tissue protruded into the pulmonary arterial lumen at this point. This structure was grossly and microscopically identical to that previously described as absent pulmonary valve. Three somewhat thickened and verrucous-appearing aortic valve leaflets were present in their normal location. The pulmonary veins, left atrium, and mitral valve were all normal. The large ventricular septal defect was the only exit from the small left ventricle (fig. 3).
In the left ventricle, the atrial septal defect (ASD) and the large ventricular septal defect (VSD) are demonstrated. The ventricular septal defect was the only exit from the left ventricle.

Incised right ventricle (RV). Retraction of the free wall upward has displaced the aortic valve somewhat posteriorly over the ventricular septal defect. The aorta (A) and pulmonary artery (PA) arose anterior to the crista supraventricularis (CS). The narrow rim of connective tissue at the usual position of the pulmonary valve (single arrow) and the verrucous aortic valve (double arrows) are at the same horizontal level. The large ventricular septal defect is bounded anteriorly by the large crista supraventricularis (CS) and posteriorly by the junction of the tricuspid (TV) and mitral valves.

Discussion

Edwards and associates\(^4\) have emphasized the clinical, angiographic and anatomic similarities of “double-outlet right ventricle” with right ventricular outflow obstruction to tetralogy of Fallot. However, the marked difference in operative technique and surgical mortality makes it mandatory that these lesions be differentiated with certainty whenever possible.\(^5\)

There is no valid clinical means whereby double-outlet right ventricle with right ventricular outflow obstruction can be differentiated from tetralogy of Fallot. Mirowski and co-workers\(^6\) have reported a much higher incidence of first degree atroventricular block, right bundle-branch block, and left ventricular hypertrophy in the former than in the latter condition. Catheterization findings are similar in these two entities. Finding the pulmonic and aortic valves at the same coronal and cross-sectional level, a tongue-like filling defect separating the two outflow tracts, and the crista supraventricularis cephalad and more dorsal to the ventricular septal defect is reliable angiographic evidence for “double outflow right ventricle” and not tetralogy of Fallot.\(^7\)\(^8\) These findings may not always be ascertainable, however, as in this case. Indeed this anatomic differentiation may be difficult even when the heart is examined postmortem. Tetralogy of Fallot with extreme dextroposition of the aorta may differ from double-outlet right ventricle with pulmonic stenosis only in that the anterior mitral leaflet and aortic valve are not continuous in the latter.\(^4\)
Fallot's tetralogy was favored as the clinical diagnosis for our patient because of the pulmonic insufficiency murmur. This lesion has been noted most often in association with tetralogy of Fallot and has not been previously reported in association with double-outlet right ventricle. The electrocardiogram failed to demonstrate any of the features of "double-outlet right ventricle" with right ventricular outflow obstruction enumerated earlier. Angiographic differentiation of these two lesions was not possible in this case. The patient died following thoracotomy.

References