Infundibular Ventricular Septal Defect, Aneurysm of the Sinus of Valsalva, and Bicuspid Aortic Valve in a Caucasian Male

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We report the case of a Caucasian man with the unusual combination of an infundibular ventricular septal defect (VSD), an aneurysm of the sinus of Valsalva communicating with the right ventricle, and a bicuspid aortic valve. When aortic valve regurgitation associated with mild right aortic cusp prolapse appeared during follow-up examination, the patient, although totally asymptomatic, underwent surgical repair of the aortic sinus and closure of the VSD. (J Am Soc Echocardiogr 2005;18:268-71.)

During cardiac evaluation for a systolic murmur in an asymptomatic young Caucasian man, an unusual complex congenital anomaly became apparent in the left ventricular outflow tract: the triad of an infundibular ventricular septal defect (VSD), an an—eurysm of the sinus of Valsalva communicating with the right ventricle, and a bicuspid aortic valve. When aortic valve regurgitation associated with mild right aortic cusp prolapse appeared during follow-up examination, the patient underwent surgical repair of the aortic sinus and closure of the VSD.

Here we report this case with an unusual combination of congenital cardiac defects. We discuss the history of these lesions and their hemodynamic interactions, and address the question of early surgical intervention in a totally asymptomatic patient.

Case report

Our patient, a Caucasian man, was initially seen at our center in 1993 at age 21 years. He had first been told that he had a cardiac murmur in early childhood. His develop—ment and growth had been normal, and he had remained asymptomatic. On physical examination, he appeared well and healthy with normal peripheral pulses and no clinical signs of cardiac failure. His blood pressure was 125/70 mm Hg. The cardiac impulse was normal. Auscultation revealed an ejection click and a localized 2-3/6 systolic murmur at the 2nd and 3rd left intercostal spaces. The second heart sound was normal, and no diastolic murmur was audible using all clinical maneuvers. Chest x-ray showed a heart of normal size, and electrocardiography findings were within normal limits.

The initial echocardiographic and Doppler study revealed the following:

• A small subarterial infundibular VSD with a small left-to-right shunt (Figure 1).

• An aneurysm of the right coronary sinus of Valsalva with a defect shunting into the right ventricular outflow tract (Figure 2).
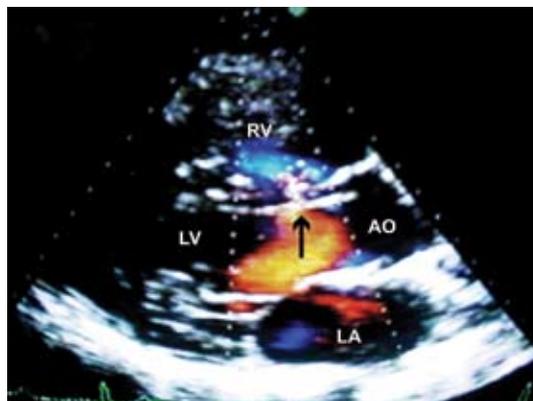
• A bicuspid aortic valve (Figure 3).

Figure 1 Transthoracic left parasternal color Doppler echocardiogram, longitudinal axis view, systolic phase. The black arrow points to the jet of an infundibular ventricular septal defect. AO, aorta; LA, left atrium; LV, left ventricle; RV, right ventricle.
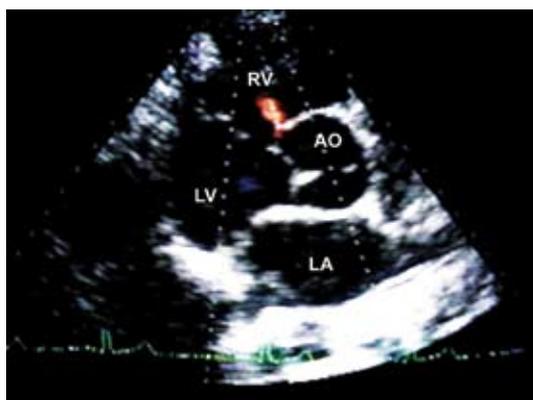

Figure 2 Transthoracic left parasternal color Doppler echocardiogram, intermediate longitudinal and short-axis view, diastolic phase. The white arrow points to the jet of blood from the aorta to the right ventricular outflow through a sinus of Valsalva defect. AO, aorta; LA, left atrium; LV, left ventricle; RV, right ventricle.

Figure 3 Transthoracic left parasternal 2-dimensional echocardiogram, short-axis plane, cross-section of the aor—tic valve, diastolic phase. A straight line delineates the commissure between the 2 aortic cusps. av, aortic valve; LA, left atrium; RA, right atrium; RV, right ventricle.

The patient was observed with yearly Doppler echocardiographic examinations, and the foregoing findings were consistently present. After 3 years, mild aortic valvular regurgitation was detected for the first time. Aortic valve regurgitation was not confirmed on cardiac catheterization. The cardiac catheterization and angiography also failed to detect the sinus of Valsalva shunt. Doppler studies repeatedly revealed aortic valve regurgitation described as trivial to mild, associated with mild right aortic cusp prolapse.

Despite the total lack of symptoms, the patient was referred for surgical intervention because of concern regarding the natural history of aortic cusp prolapse and his other cardiac defects. We decided to surgically repair the lesions to prevent further progression of aortic valve regurgitation and progression of the shunt through the coronary sinus perforation.

At surgery, the infundibular VSD was closed through the pulmonary artery and the sinus of Valsalva was covered with a right ventricular outflow Dacron patch. The defect in the sinus was also closed directly through the aortic valve.

Because the bicuspid aortic valve is likely to progress and surgery often is not curative, the patient has been instructed to observe bacterial endocarditis prophylaxis and has been followed with yearly clinical and echocardiographic examinations. He has thus far remained asymp—tomatic with trivial aortic regurgitation.

Discussion

Ventricular Septal Defect

The most common type of VSD (approximately 80% of cases) in Caucasians is membranous VSD. The second most common type comprises those exclusively contained within the muscular septum. The third and least common type is a VSD located superiorly in the infundibular septum roofed by the conjoined rings of the aortic and pulmonary valves (hence subarterial).[1] This infundibular defect generally accounts for 5%-7% of all defects seen at surgery or autopsy. An exception occurs in Japan and other Asian countries, where the percentage is much higher, approximately 30%.[2] This latter type of VSD was the type seen in our case. This type is com—monly associated with aortic sinus herniation, which most commonly affects the right or occasion—ally the noncoronary sinus and aortic cusp, resulting in progressive aortic regurgitation.[3]

Inadequate leaflet support is exacerbated by the highh-velocity jet of a protracted left-to-right shunt.[4] Commonly, the aortic sinus protrudes through the VSD, causing a decrease in the left-to-right shunt while contributing to additional aortic regurgitation.[5] Aortic regurgitation with VSD typically deveops years after birth and progresses over time.[6] More commonly, associated aortic regurgitation is insidious and progressive rather than sudden and severe, unless infective endocarditis supervenes.

Congenital Defects of the Sinus of Valsalva

Between 90% and 95% of congenital aneurysms of the

sinus of Valsalva originate in the right and the noncoronary aortic sinus and project into the right ventricle or the right atrium.[7] The developmental fault is a weakness at the junction of the aortic media and the annulus fibrosus.[8] These malformations can present acutely if the sinus of Valsalva ruptures, causing sudden severe aortic regurgitation with sudden hemodynamic overload of the heart. Smaller, "subacute" perforations are better tolerated and may even go unnoticed. As a rule, until the era of echocardiography, congenital aortic sinus aneurysms went unrecognized until they ruptured.[9]

Deformity of the aortic cusps and associated aortic valve regurgitation in conjunction with a sinus of Valsalva aneurysm is common.[10] Aortic regurgitation can also be caused by protrusion of the sinus of Valsalva into the left ventricular outflow and interference with the aortic valve cusps.[11] Conversely, the relatively rare perforation of the sinus into the left ventricle can result in aortic regurgitation predominately through the ruptured sinus rather than through the aortic valve. Other complications include atrioventricular conduction disturbances, coronary artery compression and myocardial ischemia, and infective endocarditis.[7]

Bicuspid Aortic Valve

A bicuspid valve is the most frequent congenital anomaly affecting the aortic valve and the most prevalent gross morphological congenital abnormality of the heart or great arteries.[12] A common characteristic is progressive degeneration accompanied by gradual fibrous thickening and calcification, which can render the valve immobile and stenotic or regurgitant.[13] Approximately 1/3 of bicuspid aortic valves function normally throughout life, 1/3 develop predominant aortic stenosis, and 1/3 develop combined stenosis and regurgitation.

Because the congenital bicuspid valve is most com—monly functionally normal at birth, it very often goes undetected. Development of a murmur usually occurs with subsequent thickening and calcification.[13] The incidence of infective endocarditis is low in infants and young children, but increases with age.[14]

Our Case

Our patient had 3 congenital cardiac lesions in the ventricular outflow. These lesions had gone undetected until he was 21 years old, because the only physical finding was a localized high-frequency VSD murmur. The bicuspid aortic valve and the sinus of Valsalva aneurysm were clinically silent. Doppler echocardiography easily delineated all 3 lesions as well as the aortic valve regurgitation that developed 3 years later. The failure of angiography to detect the sinus of Valsalva shunt could be due to the view plane selected for the angiogram. Because the cardiac catheterization was done in an adult catheterization facility, no oximetric studies were performed. With regard to the failure of angiography to detect aortic valve regurgitation, a possible explanation is the intermittency and variability of the aortic cusp prolapse. In this respect, the superiority of Doppler echocardiography using multiple plains of view and many cardiac cycles is readily apparent.

Each of these 3 malformations alone could account for the development of progressive aortic valve regurgitation. At

the same time, there existed the risk of a gradual or abrupt progression of the small perforation of the coronary sinus aneurysm. Each lesion also carried the risk of infective endocarditis.

The development of aortic regurgitation as a result of cusp prolapse is an indication for surgical closure of an infundibular VSD.[15,16] Considering the aforementioned risks, when aortic valvular regurgitation with cusp prolapse appeared during follow-up examinations, surgical closure of the VSD and repair of the aortic sinus aneurysm even in the absence of symptoms was considered clinically prudent.

Conclusions

Multiple congenital cardiovascular malformations frequently occur. In the presence of multiple lesions, the auscultatory findings may be masked or unobtrusive. As a result, such malformations remain undetected. Today, echocardiography/Doppler provides the best delineation of nearly all congenital lesions and provides information comparable to cardiac catheterization and angiography. Certain lesions that are clinically and even hemodynamically mild may require surgical repair. Medical and/or surgical management should be guided by the known natural history and cumulative risk imposed by the interaction of the cardiac lesions.

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