Left Ventricular Apical Aneurysm as a Consequence of Diffuse Type Congenital Nonfamilial Supravalvular Aortic Stenosis in a 30-Year-old Female

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SUMMARY

Congenital nonfamilial supravalvular aortic stenosis (SVAS) is relatively rare, its diffuse type being the least common. We present a 30-year-old woman with diffuse SVAS complicated with left ventricular apical aneurysm. We believe that subtle left ventricular myocardial ischemia or infarction and long-lasting severe pressure overload to the apical chamber caused LV apical aneurysm in our case. Acquired LV apical aneurysm secondary to supravalvular aortic stenosis, in the absence of atherosclerotic coronary artery disease and hypertrophic obstructive cardiomyopathy, has not been described before. (Int Heart J 2005; 46: 153-159)

Key words: Supravalvular aortic stenosis, Left ventricular aneurysm

SUPRAVALVULAR aortic stenosis (SVAS) is a congenital obstruction of the ascending aorta caused by a narrowing in the lumen immediately at the tip of the origin of the coronary vessels adjacent to the aortic valve. SVAS is the least common form of aortic obstruction and affects both sexes equally. The three forms of clinical presentation are nonfamilial, sporadic cases with normal facies and intelligence, autosomal dominant cases with normal facies and intelligence, and Williams syndrome with abnormal facial appearance and mental retardation.1-4)

Aortic supravalvular stenosis may be due to the presence of a discrete fibrous membrane, an hour glass type narrowing, or a diffuse narrowing.5,6) The preoperative functional class and the presence of diffuse SVAS and associated congenital defects are important determinants of death.7)

We present an adult woman with the rare diffuse type of SVAS and review the literature.

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CASE REPORT

A 30-year-old woman was referred for diagnostic cardiac catheterization in order to be evaluated for recurrent syncopal episodes, exertional dyspnea, and palpitations without anginal pain. Two of the three syncopal episodes occurred during running and the other during bathing. The patient had initially been diagnosed as having idiopathic hypertrophic subaortic stenosis, although there was no family history. She had been well until the onset of these symptoms 1 year earlier.

Physical examination disclosed a prominent and displaced apical impulse and harsh midsystolic murmur radiating to the right and left carotid arteries. The murmur did not vary appreciably with maneuvers. There was evidence of left ventricular (LV) hypertrophy and strain on a surface ECG. Only one ECG recording was taken just after her first syncopal episode, which showed severe sinus bradycardia. The only rhythm disturbance documented in our center was frequent episodes of multifocal ventricular arrhythmia. Radiographically, the sinuses of Valsalva were dilated and the ascending aorta and aortic arch appeared small. On echocardiography, the aortic vessel diameter was 2 cm at the level of the sinuses of Valsalva; the aortic wall appeared thickened and densely echogenic. There were three aortic cusps. The aorta was not well visualized after the sinotubular junction because of a poor echocardiographic window. Interventricular septum thickness was 21 mm and the posterior wall was 19.8 mm. There was systolic anterior motion of the mitral valve and minimal mitral insufficiency with increased left atrial diameter. Continuous wave Doppler echocardiography confirmed stenosis with a peak gradient of 90 mmHg across the aortic root. Echocardiography failed to show the left ventricular apical aneurysm. Pulsed- and continuous-wave Doppler flow velocities were normal in the parasternal short axis view. Transesophageal echocardiography was not contributory.

In magnetic resonance angiography, the aortic root measured 20 mm and decreased to 6-7 mm just after sinotubular junction. The aortic arch diameter was 10 mm. The diameters of the descending aorta, brachiocephalic artery, left subclavian artery, and left common carotid artery were within normal ranges (Figure 1). There were no pathologic signal intensities on the walls of these arteries. Pulmonary arterial and venous structures were normal. Severe LV wall hypertrophy was present.

Cardiac catheterization and coronary angiography were performed afterwards. A pigtail catheter was placed in the LV cavity; left ventricular peak systolic pressure was 280 mmHg and LV end-diastolic pressure was 19 mmHg. At the aortic level, peak systolic pressure dropped to 118 mmHg, thus creating a pressure gradient of 162 mmHg (Figure 2). LV cavity pressure did not change on recurrent pressure tracings. Aortic pressure was the same at different levels of the aorta. Left ventriculography in the right anterior oblique projection demonstrated systolic cavity obliteration and apical aneurysm (Figure 3). The length of the
Figure 1. Magnetic resonance angiogram of the hypoplastic ascending aorta and normal sized great arteries.

Figure 2. Left ventricle to aorta pressure tracing shows a gradient of 162 mmHg.

aneurysm was 20 mm and the width was 10 mm. Aortography performed at a 60° left anterior oblique projection showed diffuse narrowing of the ascending aorta, a characteristic and striking sign of the diffuse type of SVAS. Diffuse hypoplasia started at the level of the sinotubular junction and dilated coronary arteries were visualized in this projection (Figure 4). The stenosed segment after the sinotubu-
The sino-tubular junction measured 22 mm long and 7.3 mm wide; the aortic arch diameter was 9.5 mm. The diameters of the brachiocephalic, left subclavian, and left common carotid arteries were normal. The aorta widened to 19 mm after the left subclavian artery and the abdominal arteries were normal. There were no stenosed pulmonary artery segments on selective pulmonary angiography. The left and right coronary arteries were severely ectatic but not stenotic; the right coronary artery measured 9-10 mm (Figure 5). Aortic insufficiency was ruled out with aortic root injection. As the patient did not accept operative risks, she was prescribed metoprolol (200 mg/day) and aspirin (300 mg/day) and scheduled for a regular bimonthly follow-up.

Figure 3. The systolic (A) and diastolic (B) phases of left ventriculography show the apical aneurysm and systolic cavity obstruction. Arrows show the apical aneurysm.

Figure 4. Aortogram showing the diffuse hypoplasia of the aorta starting from the sinotubular junction and extending to the aortic arch. Arrows show the stenotic ascending aorta.
SVAS is unique among left ventricular outflow tract obstructions. High systolic pressure at the level of the coronary artery orifices leads to dilatation and tortuosity and thus makes the patient susceptible to sudden cardiac death before and after repair. The coronary ostia may be obstructed by the overhanging thick sinus rims as well as bound-down aortic cusps. This more often occurs in the left sinuses, but can also occur in the right.8)

Patients with supravalvular aortic obstruction are subject to the risks of unexpected sudden death, endocarditis, and myocardial infarction. Some of these have been discovered at autopsy.9) Dissection of the ascending aorta, severe coronary arterial disease, and microfocal myocardial fibrosis were shown in a patient with Williams-Beuren syndrome. Pathological findings in seven autopsy cases implicated two anatomic abnormalities that predisposed individuals with Williams-Beuren syndrome to sudden death: coronary artery stenosis and severe biventricular outflow tract obstruction. The mechanisms for sudden death for both anatomic subgroups include myocardial ischemia, decreased output, and arrhythmias. Ambulatory ECG recordings showed malignant ventricular arrhythmia in sudden death cases with aortic stenosis.10-12)

The principal mechanisms of sudden death in aortic stenosis appear to be: (1) activation of left ventricular baroreceptors which cause reflex bradycardia and cardiac arrhythmias, or (2) arrhythmia as a complication of left ventricular hypertrophy. Myocardial ischemia has been anatomically proven and may be due to diastolic compression of intramural coronary arteries. Aortic dissection occurs with increased frequency in patients with a bicuspid aortic valve. Heart block can result from calcification of the bundle of His in aortic stenosis of any cause;13) however, our case did not display any sign of heart block.

**Figure 5.** Selective right (A) and left (B) coronary angiography showing extremely widened coronary arteries. Arrows show the ectatic proximal left coronary artery.
Hypertrophic obstructive cardiomyopathy is one of the more common causes of acquired LV aneurysm in the absence of atherosclerotic coronary artery disease. Wiggle, et al described a patient who died of intractable ventricular arrhythmias who was earlier demonstrated by angiography and hemodynamic recordings to have had midventricular obstruction at the level of the papillary muscles. The apex of the LV was a site of extensive myocardial infarction and aneurysm formation, yet the coronary arteries exhibited no significant luminal narrowing.

As observed in our patient too, the question is again raised whether dynamic, long-standing intracavitary pressure overload might exert a deleterious effect on the LV myocardium and cause formation of an aneurysm. Transmural myocardial infarction, extensive at times, occurs quite commonly in IHSS in the absence of significant coronary atherosclerosis. We speculate that the described pathologies in SVAS (such as subtle myocardial ischemia, microfocal myocardial fibrosis and infarction) when combined with long-lasting severe pressure overload may have led to aneurysm formation in our case.

In the series of Harikrishnan, et al, the SVAS rate among cases with congenital heart disease was 0.12%. All of the patients had the discrete type of SVAS, except one who had the diffuse variety (6.66%) and was 14 years old. In the series of Sharma, et al, the rate of the diffuse variety was 15%. Brown, et al reported that 28 of 101 patients had the diffuse variety of SVAS, while Braunstein, et al diagnosed 2 of 13 patients with the diffuse variety.

The hour-glass type of obstruction is the most common. It is important to remember that the diffuse type of stenosis has been described as the type that more commonly results in death. The discrete type of SVAS is more amenable to surgery than the diffuse type. Surgical treatment options for the diffuse form of SVAS are less well defined. It may become necessary to replace or widen the entire hypoplastic aorta with an appropriate prosthesis. The operative results obtained by Brown, et al in diffuse SVAS were encouraging, while those of Braunstein, et al in their 2 diffuse type cases were excellent.

To the best of our knowledge, LV apical aneurysm secondary to SVAS has not been reported before. LV aneurysm-related complications such as sudden death and arrhythmia may complicate the clinical status of SVAS and may be one of the causes of sudden death in these patients.

In conclusion, congenital nonfamilial diffuse SVAS is a complex and uncommon disease. It is much more uncommon in adults with congenital heart disease. We think that long-lasting severe systolic pressure overload and myocardial pathologies may have caused the LV apical aneurysm in our patient. Aneurysm-related complications may be one of the causes of sudden death in patients with diffuse SVAS.
REFERENCES