Case Reports

Isolated Diffuse Supravalvular Aortic Stenosis

with Severe Aortic Narrowing in a 41-Year-Old Man

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Isolated supravalvular aortic stenosis in adults is a rare form of left ventricular outflow tract obstruction. We describe a case in a 41-year-old man in whom the supravalvular aorta had narrowed to approximately the size of the left anterior descending coronary artery. The patient underwent aortic surgery with replacement of the ascending aorta and repair of supravalvular aortic stenosis with a pantaloon graft. A postoperative echocardiogram showed substantial improvement: the mean gradient across the aorta had fallen from 48 to 8 mmHg. Surgery is the definitive treatment in symptomatic patients with supravalvular aortic stenosis. ***(Tex Heart Inst J 2017;44(4):287-9)***

Isolated supravalvular aortic stenosis (SVAS) in adults is a rare form of left ventricular (LV) outflow tract obstruction.[1] The condition is usually associated with Williams syndrome, a neurodevelopmental genetic disorder. We describe a case of isolated diffuse SVAS in a middle-aged man who did not have Williams syndrome and in whom the supravalvular aorta had narrowed markedly.

Case Report

In August 2014, a 41-year-old man with a history of heart murmur presented with worsening shortness of breath on exertion, which had begun one month earlier. On auscultation, a grade 3/6 systolic ejection murmur at the right upper sternal border radiated to both carotid arteries; his S_2 was normal. The results of his physical examination were otherwise normal.

A transthoracic echocardiogram revealed a hypoplastic ascending aorta with SVAS, dilated coronary arteries, mild concentric LV hypertrophy, and a normal LV ejection fraction. It also revealed a long, tubular narrowing of the ascending aorta, starting at the sinotubular junction; a trileaflet aortic valve that opened normally; and trace aortic insufficiency. The proximal aortic root was 2.18 cm in diameter, and the sinotubular junction and tubular aorta were 1.4 cm in diameter (Fig. 1A). Continuous-wave Doppler echocardiography revealed a diffusely narrowed aorta with a peak pressure gradient of 97 mmHg (Fig. 1B), an estimated mean pressure gradient of 48 mmHg, and a peak velocity of 4.86 cm/s (Fig. 1C). Computed tomographic angiography (CTA) of the chest, performed to confirm the diagnosis of SVAS and to detect any other congenital heart anomalies, showed diffuse stenosis of the ascending aorta, especially in the supravalvular region (diameter, 1 cm) (Fig. 2). The CTA also showed aneurysmal dilation of the left coronary artery (1.2 cm), proximal left anterior descending coronary artery (1 cm), left circumflex coronary artery (1.1 cm), and proximal right coronary artery (0.8 cm). The aorta just above the aortic valve and the remainder of the ascending aorta were diffusely narrowed to a total length of 3.8 cm. There was mild prominence of the proximal left common carotid artery (1.1 cm) and the left subclavian artery (1.5 cm).

While under deep hypothermic circulatory arrest, the patient underwent replacement of the ascending aorta and repair of the SVAS with a HEMASHIELD PLATINUM® pantaloon graft (Maquet Cardiovascular, LLC; Wayne, NJ). A postoperative transthoracic echocardiogram (TTE) showed substantial improvement; the mean pressure gradient across the aorta had decreased from 48 to 8 mmHg (Fig. 3). At the patient's

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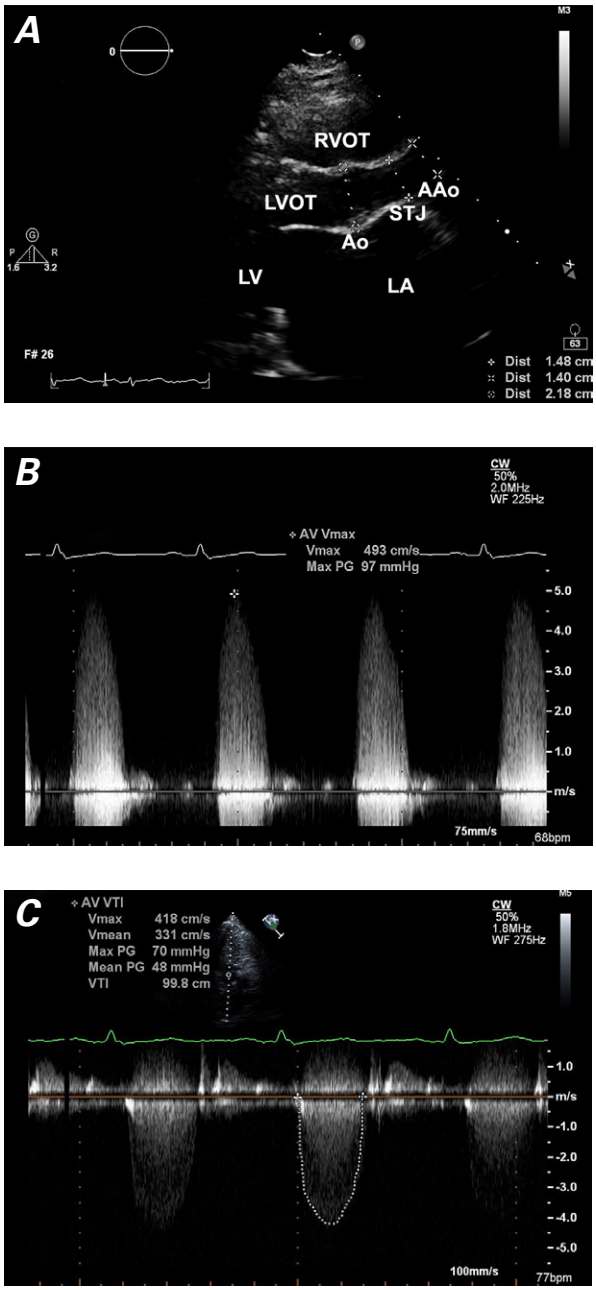

Fig. 1 *Transthoracic echocardiograms.* ***A****) Parasternal long-axis view shows the diameters of the proximal aortic root (Ao) and the diffusely narrowed ascending aorta. Continuous-wave Doppler mode (apical 3-chamber view) shows* ***B****) a peak pressure gradient of 97 mmHg through the diffusely narrowed aorta in the suprasternal view, and* ***C****) a mean pressure gradient of 48 mmHg across the diffusely narrowed aorta.*

AAo = ascending aorta; LA = left atrium; LV = left ventricle; LVOT = left ventricular outflow tract; RVOT = right ventricular outflow tract; STJ = sinotubular junction

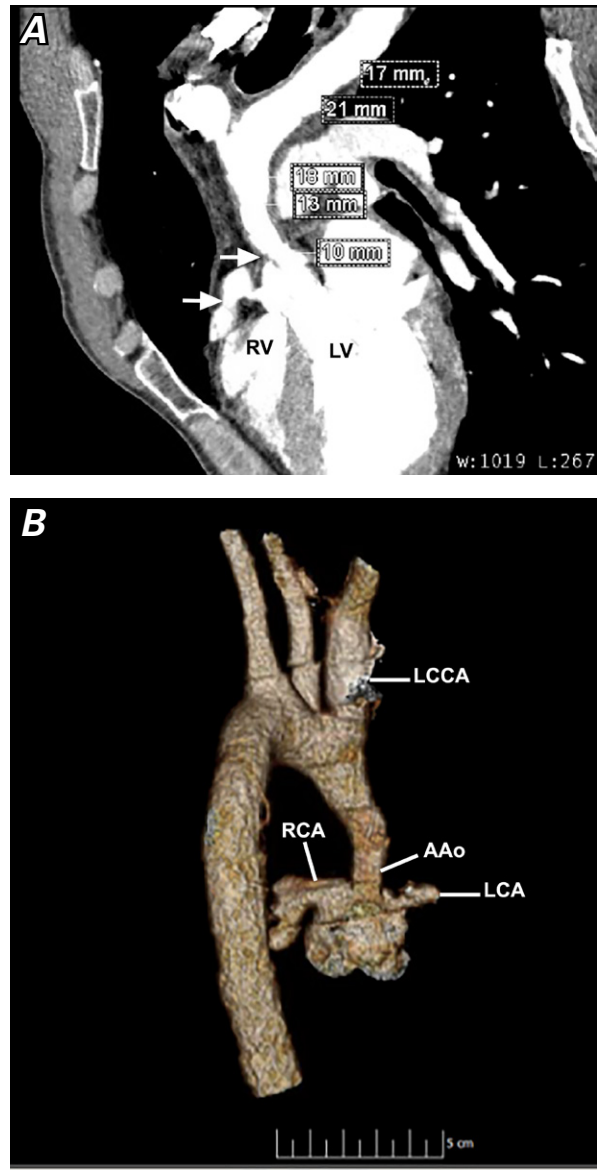

Fig. 2 *Computed tomographic angiograms.* ***A****) Multiplanar reconstruction image shows the diffusely stenosed ascending aorta (top arrow) and aneurysmal right coronary artery (bottom arrow).* ***B****) Volume-rendered reconstruction image shows diffuse stenosis of the ascending aorta (AAo) and the aneurysmal left coronary artery (LCA), right coronary artery (RCA), and left common carotid artery (LCCA).*

LV = left ventricle; RV = right ventricle

3-month follow-up examination, the findings on TTE were similar, with mild-to-moderate aortic regurgitation. As of February 2017, the patient was doing well and had a good prognosis.

Discussion

Supravalvular aortic stenosis is characterized by narrowing of the aorta, starting at the sinotubular junction and sometimes extending up to the ascending aorta, as in our patient. Rarely, the aortic arch and the peripheral arterial system are involved.[1,2] There are 3 types of SVAS: focal segmental (hourglass), which is the most typical; tubular (diffuse) hypoplasia, as seen in our pa-

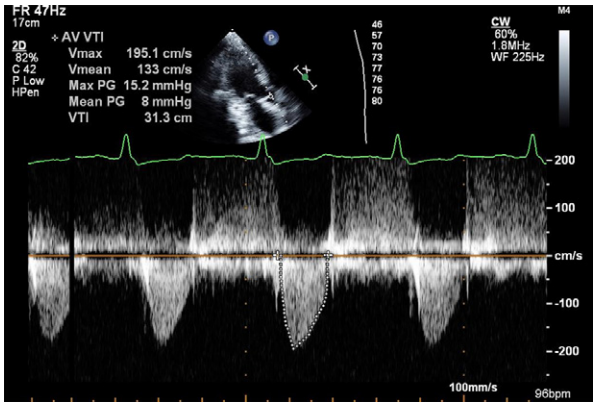

Fig. 3 *Postoperative transthoracic echocardiogram (apical 5-chamber view) with continuous-wave Doppler mode shows a substantial decrease in the mean gradient across the aorta.*

tient; and membranous, the least prevalent type.[3] In all types of SVAS, the coronary arteries usually originate proximal to the obstruction and are subjected to high systolic pressure and decreased diastolic pressure; as a result, individuals with SVAS are prone to coronary aneurysms and premature atherosclerosis.[2,3]

Supravalvular aortic stenosis is caused by a mutation in the elastin gene on chromosome 7q11.23. This mutation leads to a decrease in elastin fibers and elasticity, an increase in shear stress in the ascending aorta, collagen deposition, and smooth muscle cell hypertrophy.[1] In addition to Williams syndrome, SVAS is associated with bicuspid aortic valve,[3] coarctation of the aorta, and pulmonary artery stenosis. Less frequently, it is associated with hypoplasia of the entire aorta, or with stenosis of the renal artery, major aortic branches, or long-segment pulmonary artery.[2] Our patient had isolated SVAS, which is rare; only a few cases have been reported.[4-7] He did not have Williams syndrome, and he had no other conditions that predisposed him to SVAS.

Supravalvular aortic stenosis may manifest itself as angina, dyspnea, or syncope secondary to substantial outflow tract obstruction, or as ischemia from premature atherosclerosis of the coronary arteries or perfusion mismatch.[2,3] Transthoracic echocardiography can be used to visualize the narrowed aortic lumen and to estimate the severity of the gradient across the stenosis.[2,3] Magnetic resonance imaging and CTA enable noninvasive evaluation of the entire aorta and its branches.[2] We used CTA to determine whether our patient had congenital anomalies and to confirm the extent of aortic narrowing.

Surgical correction is the definitive treatment for SVAS patients who are symptomatic or who have a mean pressure gradient ≥50 mmHg or a peak gradient ≥70 mmHg. It is also appropriate for pregnant patients with a pressure gradient ≥30 mmHg and for patients with LV hypertrophy and low LV ejection fraction.[2,3] Patients presenting with ischemic symptoms need ostial débridement, dilation, or coronary artery bypass grafting.[2] Surgical correction of focal SVAS is associated with low mortality rates and good long-term results; in contrast, surgical correction of diffuse SVAS is challenging and associated with high mortality rates.[8]

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