

## SPORADIC SUPRAVALVULAR STENOSIS IN A YOUNG MAN

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**Supravalvular aortic stenosis (SVAS) is a form of congenital left ventricular outflow tract (LVOT) obstruction that occurs as a localized or diffuse narrowing of the ascending aorta beyond the superior margin of the sinuses of Valsalva. SVAS is a rare progressive congenital heart defect with a higher risk of sudden cardiac death. Our patient was a 30-year-old man referred by his GP for an incidental finding of heart murmur. Echocardiography showed a hypertrophic left ventricle with an increased pressure gradient. CT and MRI angiography and eventual arteriography confirmed the diagnosis by showing a narrowing of the aorta at the sinotubular junction with significant accelerated flow. This stenosis was eventually surgically corrected with no complications.**

**Key-word:** Aorta, stenosis or obstruction.

Supravalvular aortic stenosis (SVAS) is an uncommon but well characterized congenital narrowing of the ascending aorta above the level of the coronary arteries. It can be a familial disorder, can occur sporadically, or be associated with Williams syndrome (WS) which is a neurodevelopmental disorder affecting connective tissue and the central nervous system. Sudden cardiac death has a higher prevalence in patients with congenital supravalvular aortic stenosis.

### Case presentation

A 30-year-old man was referred by his GP for an incidental finding of a heart murmur. Physical examination showed a mid-systolic murmur without further abnormalities.

ECG showed sinus rhythm with signs of inferior and lateral strain. Genetic evaluation was normal.

Transthoracic echocardiography showed a hypertrophic left ventricle with a septal thickness of 20 mm and accelerated flow above the aortic valve with a pressure gradient of 89mmHg. Supravalvular aortic stenosis was suspected.

Further cardiac MRI angiography confirmed a concentric hypertrophic left ventricle (Fig. 1) without signs of delayed enhancement (Fig. 2). MRI angiography showed a supravalvular aortic stenosis (Fig. 3) with accelerated flow above the aortic valve (Fig. 4).

Conventional angiography confirmed the presence of an aortic stenosis at the sinotubular junction with a maximum diameter of 18 mm and normal coronaries (Fig. 5). No

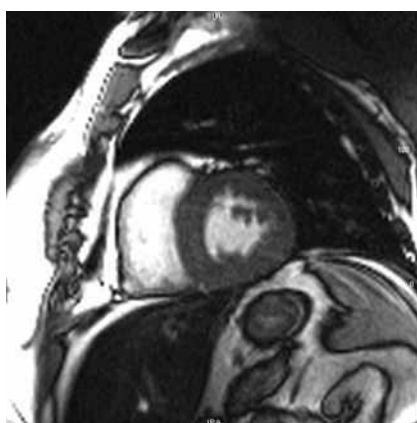


Fig. 1. — Short axis view showing concentric left ventricular hypertrophy.



Fig. 2. — Image after contrast administration: absence of delayed enhancement.

other vascular abnormalities were found. The stenotic lesion was surgically corrected (Fig. 6).

### Discussion

Supravalvular aortic stenosis is the rarest lesion of the left ventricular outflow tract obstruction abnormalities (LVOTOs). The majority of cases of congenital SVAS are associated with Williams-Beuren syndrome (WS). WS has an incidence of approximately 1:20,000 live births (1).

Sudden death occurs at a rate higher than in the general population, both in patients with congenital supravalvular aortic stenosis associated with WS and nonsyndromic SVAS.

WBS was first described in 1961 as a combination of stenoses of the large- and medium-sized arteries combined with facial dysmorphic

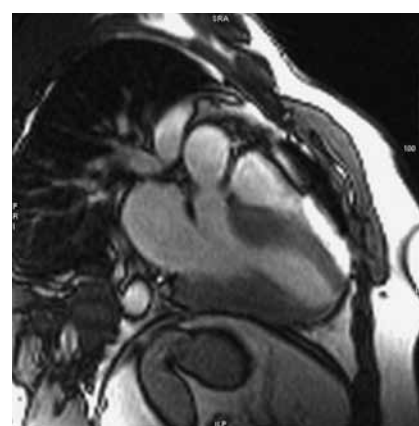


Fig. 3. — Three chamber view showing supravalvular aortic stenosis.

signs, short stature, failure to thrive and mild to moderate mental retardation. Associated features include transient hypercalcaemia in infancy, small teeth, joint stiffness, scoliosis, sensorineural hearing loss, hyperreflexia, problems of visio-spatial processing and anxiety. The syndrome is caused by heterozygous microdeletion of the 'WBS critical

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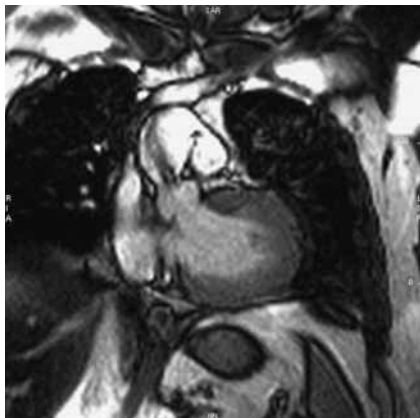


Fig. 4. — Left ventricular outlet view showing turbulent flow at the level of stenosis.

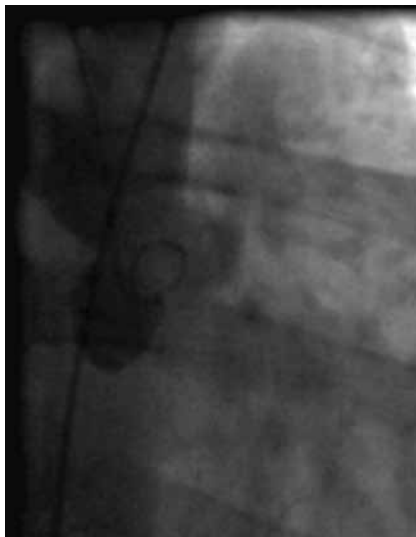


Fig. 5. — Confirmation of supraaortic aortic stenosis at angiography.



Fig. 6. — Correction of the stenosis after surgery.

region' on the long arm of chromosome 7 (2).

The vascular features of congenital SVAS are due to an elastin arteriopathy. A large quantity of elastin is normally present in the media of the great vessels, whereas smooth muscle and collagen are the primary components of smaller arteries. Smooth muscle cells from patients with isolated SVAS produce only 50% of the elastin produced by normal cells, whereas smooth muscle cells from patients with WS produce only 15% of the normal quantity of elastin (3). The reduced net deposition of arterial wall elastin leads to increased proliferation of arterial wall smooth muscle cells resulting in multilayer thickening of the media of large arteries and subsequent development of obstructive hyperplastic intimal lesions. As a result, a characteristic hourglass narrowing of the aorta develops at the sinotubular junction. In approximately 30% of cases, there is diffuse tubular narrowing of the ascending aorta, often extending to the arch and the origin of the brachiocephalic vessels (4).

In approximately 40% of WS patients, there is severe pulmonary stenosis and right ventricular pressure overload on top of the LV pressure overload and hypertrophy (5). The aortic valve may also be involved in SVAS.

The elastin arteriopathy may involve the coronary arteries in a diffuse pattern whereas a thickened aortic wall can directly narrow the coronary ostia (6). There may also be stenosis in the renal and mesenteric arteries.

The diagnosis of SVAS can be made by multiple imaging modalities.

CT and MR imaging allow visualization of the entire aorta and are the modalities of choice to demonstrate the extent of the SVAS (7). If an ECG-gated technique is used, associated findings such as left ventricular myocardial hypertrophy and BAV can be depicted. Cardiac magnetic resonance imaging and computer tomography are capable of showing ventricular hypertrophy, wall motion abnormalities, myocardial tissue characteristics, associated vascular anomalies. In addition they are able to diagnose obstructive coronary disease.

Transthoracic Doppler echocardiography is useful in deriving peak instantaneous and mean pressure gradients. However, the full extent of the ascending aorta is difficult to visualize in adults with transthoracic echocardiography; transesophageal echocardiography is superior. Echocardiography is useful in assessing ventricular hypertrophy, ventricular outflow tract gradients, and wall motion abnormalities but is an insensitive method for evaluating coronary blood flow.

Cardiac catheterization with coronary and aortic angiography remains the "gold standard" for delineation of aortic leaflet tethering and assessment of coronary artery lumen calibre (1). Obviously, cardiac catheterization carries its own risks in these patients.

Surgical correction of the narrowing is indicated in symptomatic patients (ie, those with angina, dyspnea, syncope) or those with a mean

pressure gradient of greater than or equal to 50 mm Hg. Balloon angioplasty is not useful. Patients with SVAS and Williams syndrome show regression of stenosis without intervention, and overall survival after treatment is excellent (94% at 10 years) (7).

## Conclusion

SVAS is a rare progressive congenital heart defect with a higher risk of sudden cardiac death than in the general population, emphasizing the importance of early diagnosis. Identifying those patients with congenital SVAS at risk for myocardial ischemia is challenging, and each patient has to be monitored closely.

Cardiac imaging modalities, including multislice computed tomography and magnetic resonance imaging, show increasing promise as noninvasive imaging modalities to detect SVAS. These noninvasive modalities also can correctly identify other cardiac, aortic and pulmonary vascular abnormalities in the same session.

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