24 Supravalvar Aortic Stenosis

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Supravalvar aortic stenosis (SVAS) is an LVOT lesion that poses unique challenges and can be part of a more diffuse arteriopathy syndrome.

Pathophysiology and Clinical Presentation

SVAS is the characteristic congenital heart lesion of elastin deficiency, secondary to deletion of one copy of the ELN gene as part of Williams syndrome or to mutations in ELN (non-syndromic SVAS). Elastin deficiency primarily affects the large arteries resulting in medial thickening and luminal narrowing. SVAS is therefore often seen in the context of diffuse arteriopathy. Associated cardiovascular lesions are common and include valvar and supravalvar pulmonary stenosis, branch PA stenosis, aortic arch hypoplasia and coarctation, and mitral valve prolapse. SVAS is also commonly associated with coronary artery ostial stenosis, which may be secondary to tethering of the aortic valve leaflets at the narrow sinotubular junction. Anatomically, SVAS is classically categorized as membranous type, hourglass type, or diffuse hypoplasia; the later two are seen in elastin deficiency. From a surgical standpoint, SVAS is classified as isolated or as part of a diffuse arteriopathy.

Timing of presentation is dependent upon the severity of the obstruction and the presence of associated cardiovascular lesions. Most individuals with SVAS are asymptomatic at presentation and are evaluated due to findings on examination (e.g., dysmorphic features, heart murmur). In Williams syndrome, moderate to severe obstruction is more likely to progress, but the majority of individuals are stable over time. Individuals with Williams syndrome are less likely to require intervention compared to those with non-Williams SVAS (i.e., due to ELN mutations or other as-of-yet undefined causes); the reasons for this are not yet delineated.

Like other LVOT lesions, SVAS results in a harsh systolic ejection murmur with carotid radiation and a suprasternal notch thrill. One potentially distinguishing examination finding is a difference in BP (greater than approximately 18 mmHg) between the right and left arms, which is hypothesized to arise secondary to the Coanda effect (streaming of blood along a boundary wall).

Diagnosis

- **ECG.** May show LVH or BVH depending on the age of presentation and severity of the lesion.
- **CXR.** May suggest LVH or BVH.
- **Echocardiography.** Mainstay of diagnosis. Complete characterization includes 2D measurements of the sinotubular junction, color Doppler across the stenosis, and assessment of the maximal spectral Doppler velocity. The peak instantaneous gradient calculated from the maximal velocity overestimates the peak-to-peak gradient by cardiac catheterization; the mean gradient should therefore also be reported. The aortic valve annulus size and aortic morphology are also important

for surgical planning. Assessment for associated cardiovascular lesions is essential. Importantly, coronary ostial stenosis cannot be excluded by echocardiography.

- **CTA and MRI.** Primarily used as adjunct imaging modalities for assessment of the coronary arteries and detection of distal stenoses in diffuse arteriopathy. The abdomen is often included to assess for midaortic syndrome and stenosis of the aortic branches, particularly renal artery stenosis. Cross-sectional imaging is also useful to assess the branch PAs.
- **Genetic evaluation.** Should be performed at diagnosis, including family history and assessment of dysmorphic features and extracardiac anomalies, in order to direct genetic testing (i.e., chromosomal microarray to assess for Williams syndrome, elastin gene sequencing with deletion/duplication analysis to assess for nonsyndromic SVAS).

Indications / Timing of Intervention

The treatment of SVAS is surgical. Since the degree of stenosis may be progressive, especially in patients with moderate or severe SVAS, and there is commonly coronary involvement, patients are put forward for surgery earlier than patients with subvalvar or valvar AS. In general, a peak-to-peak gradient or a mean gradient of 50 mmHg is an indication for surgical intervention. Patients with lower gradients may benefit from surgical intervention if there is evidence of coronary ischemia at rest or during exercise. Isolated branch PS may be addressed by balloon angioplasty in the cardiac catheterization lab unless the stenosis is diffuse.

Anesthetic Considerations

Patients with SVAS are at high risk for myocardial ischemia, especially during induction of anesthesia. The hemodynamic goals are to avoid tachycardia and maintain adequate coronary perfusion pressure. It is imperative to have adequate preload, maintain SVR, and avoid hypotension. The combination of tachycardia and hypotension can lead to rapid hemodynamic deterioration in an already tenuous oxygen supply and demand situation. The combination of LVH and coronary obstruction puts patients at high risk for cardiac arrest, especially during induction, and make CPR less effective. For this reason, it is customary at TCH for the surgeon and the perfusionist to be available in the OR during induction.

Medications that increase heart rate (e.g., atropine, glycopyrrolate, ketamine), or decrease SVR (e.g., propofol, volatile anesthetics) should be used with caution. It is common to administer vasopressors during induction to keep an adequate SVR. It is important to note that the degree of SVAS does not correlate with the severity of coronary obstruction. Patients with mild-to-moderate SVAS can have significant coronary obstruction. Branch PA stenosis and RVOT obstruction can also lead to RVH and RV dysfunction. It is thus imperative to avoid increases in PVR with adequate ventilation and oxygenation.

After repair, patients with SVAS tend to do well. It is important to avoid wide swings

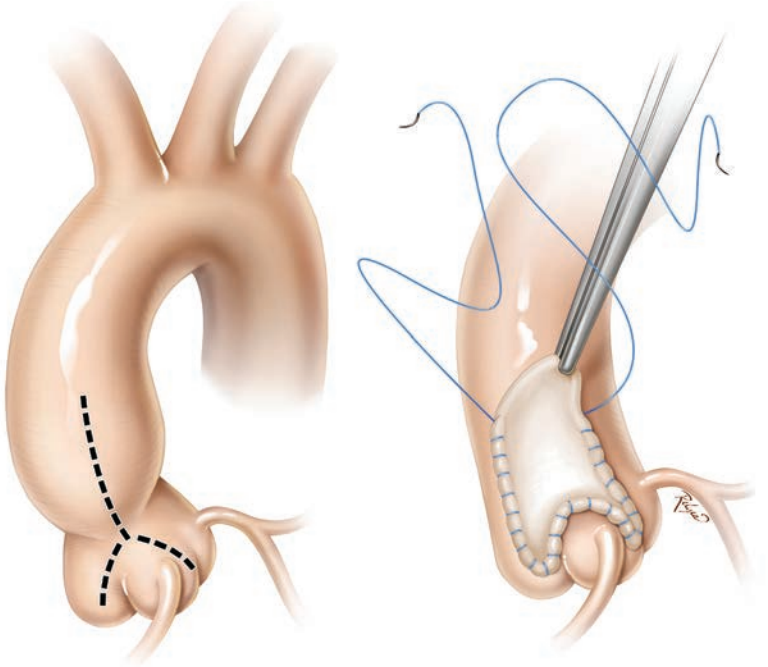

Figure 24-1. Surgical repair of SVAS with the Doty-patch technique. An inverted "Y" incision is created across the area of stenosis and an autologous pericardial pantaloon patch is used to enlarge the aortic root.

in BP. Other anesthetic considerations for patients with SVAS associated with Williams syndrome include hypertension and potential renal dysfunction.

Surgical Repair

The procedure may be performed using standard aorto-bicaval cannulation. However, the aorta in these patients tends to be thick and difficult to cannulate. A reasonable alternative is to suture a Gore-Tex® graft into the innominate artery for arterial cannulation. This also leaves more room on the ascending aorta to perform the repair. Myocardial protection in these patients may be challenging due to coronary obstruction and the degree of LVH.

The preferred repair at TCH is the Doty (pantaloon) patch aortoplasty (Figure 24-1). After cardioplegic arrest and venting of the left heart, a longitudinal incision is performed on the ascending aorta and carefully extended proximally up to the sinotubular junction, just above the right/non-coronary commissure. This is the usually the area where the thick fibrous ring creates the worst obstruction. The incision is then extended as a "Y" with one limb into the nadir of the right coronary cusp, between the right coronary and the left/right coronary commissure, and the other limb into the nadir of the non-coronary cusp. All dimensions of the "Y" incision are carefully measured,

in particular the length between the apex of the incision on the distal ascending aorta and the bifurcation site of the "Y" at the sinotubular junction. Leaving this length too long will allow the anterior aspect of the aortic valve to fall down and may create significant AI or torsion of the right coronary artery.

It is not unusual to have fibrous tissue involve the ostia of the coronary arteries, in particular the left coronary, thus creating a "hood". The fibrous tissue at the sinotubular junction is shaved and the fibrous hood excised from the coronary ostia. A pantaloon-type, short, and wide patch of glutaraldehyde-treated pericardium is then used to reconstruct the aortic root and ascending aorta, therefore enlarging the sinotubular junction.

In some patients, the extent of SVAS may be more diffuse and extend into the arch and brachiocephalic vessels. These patients will require more extensive and/or separate patch aortoplasties. If there is significant stenosis of the central branch PAs, it may be addressed at the time of surgery with homograft or autologous pericardial patches.

Postoperative Management

Postoperative management of patients with SVAS is focused on the following tenets:

- **Preservation of coronary perfusion pressure.** Preoperatively, the coronary arteries are perfused at higher diastolic pressures. If the coronaries are of small caliber and have been unroofed, systemic hypertension may not be necessary, but if they are small and there is a history of ventricular arrhythmias, a combination of epinephrine at doses not higher than 0.05 mcg/kg/min and low-dose vasopressin (no more than 0.03 U/kg/hr) may be helpful given the abnormal coronary wall elasticity.
- **Adequate ventricular performance.** RV or LV dysfunction may be present due to coronary insufficiency from RVH or LVH, and/or prolonged cross-clamp time from more extensive repairs. Given the degree of LVH and poor LV compliance, there will be more significant changes in ventricular filling pressures at small changes in volume. One should aim for the lowest filling pressures to achieve normal hemodynamics. Inotropes may be used, if needed, to increase BP and perfusion. Significant ventricular unloading with diuretics could easily precipitate hypotension. Diuresis should be conservative.
- **Identification and treatment of arrhythmias.** Ectopic atrial rhythms are infrequent but could be secondary to elevated ventricular filling pressures and more than moderate mitral or tricuspid valve regurgitation. Ventricular arrhythmias should increase suspicion for coronary ischemia and ventricular dysfunction.

Complications

Some potential complications after repair of supravalvar AS include:

- **Development of AI.** Failure to achieve an adequate geometry of the aortic root and ascending aorta during repair may distort the aortic valve and condition the development of AI. This complication should be noted and corrected in the OR.
- **Coronary insufficiency.** Adequate myocardial protection is imperative during

repair to avoid myocardial dysfunction. In addition, an inadequate repair may cause torsion of the coronary arteries.

Long-Term Follow-Up

Individuals with SVAS require lifelong follow-up as they may develop arterial stenoses in other locations or recurrence of SVAS following intervention. Additionally, they are at risk for hypertension secondary to stenotic lesions, vascular stiffness, or renal involvement. Hypertension can be difficult to manage and may require a multidisciplinary approach. Nonsyndromic SVAS secondary to ELN mutation is an autosomal dominant condition and therefore families should be counseled that an affected individual has a 50% chance of passing the trait to their offspring.