Echocardiographic Features of Supravalvular Aortic Stenosis

By James L. Bolen, M.D., Richard L. Popp, M.D., and James W. French, M.D.

SUMMARY
A method for the echocardiographic detection of supravalvular aortic stenosis (SVAS) is described and the findings in a series of patients are presented. When compared to angiography, the echo tended to underestimate the severity of the supravalvular aortic obstruction. However, echocardiography appears to be a valuable, noninvasive method for detecting SVAS.

DETECTION of supravalvular aortic stenosis (SVAS) and differentiation from valvular or subvalvular obstruction remains a significant clinical problem. Although the clinical features of obstruction at the three levels have been well described,1-8 and SVAS is frequently associated with a syndrome including mental retardation and elfin faces,9 typical features may be absent or inconstant and the identification, differentiation of types, and assessment of the severity of left ventricular outflow obstruction may be difficult. Echocardiography has significantly enhanced the noninvasive assessment of left ventricular outflow obstruction. Echocardiographic features of valvular aortic stenosis,9 fixed subvalvular aortic stenosis,10,11 and muscular subaortic stenosis12,13 have been reported. The echocardiographic features of SVAS have received little attention, with the exception of one case report.14 We describe the echocardiographic features of six cases of SVAS seen within the past two years. A reliable technique for recording SVAS is outlined and comparison with radiographic and hemodynamic assessment of narrowing is made.

Methods

Echocardiography

Echocardiography was performed using a Smith-Kline Ekoline 20A ultrasonoscope and 2.25 MHz transducers of either ½ or ¼ inch diameter, depending on the size of the patient. Echocardiograms were recorded on an IREX 101 or Honeywell 1856 strip chart recorder. In five patients studied, diagnosis of SVAS was confirmed at catheterization and in one case there was a strong familial predisposition and clinical signs of SVAS. Twelve children without aortic or aortic valvular abnormalities were studied as controls. Patients were studied in the supine position using 15-30° of left lateral decubitus rotation. The transducer was placed in the second or third left intercostal space adjacent to the sternum, such that the aorta was recorded at the level of the aortic valve when the transducer was perpendicular to the chest wall or pointing slightly medially and cephalad (fig. 1). The supravalvular area was examined by angling the transducer superiorly and somewhat medially. To minimize recording a "false positive" sweep caused by visualizing the aorta off-center, a minor axis sweep, similar to the T-scan described for left ventricular examination,15 was utilized at the level of the ascending aorta to locate the largest aortic diameter. The same scan technique was applied at the level of the aortic leaflets for aortic root dimensions. As the echo beam sweeps up the ascending aorta cephalad, the pulmonic valve could frequently be seen if the sweep were too lateral (figs. 1, 2). The proximal portion of the right pulmonary artery may be seen behind the ascending aorta as the sweep is carried further cephalad (figs. 1-3). Examination is usually limited by loss of visualization of the posterior aortic wall.

Measurements

The aortic width was measured at the level of the aortic valve and at the level of minimal echocardiographic dimension in the ascending aorta. The diameter was determined at the point of maximum posterior excursion of the aorta (occurring 0.06 to 0.10 sec after the R wave), from the innermost posterior aortic echo to the outermost echo of the anterior aortic wall.16 Figure 2 shows a sweep of a normal aorta and notes where the aortic dimensions were measured. Measurement was made in this way to avoid signal amplification artifacts.

Results

Patient Data

Patient 1. This eight-year-old girl was the oldest of four sisters, all of whom had SVAS. Patients 2 and 3 are her living sisters (a sister one year younger died at another institution following surgery for correction of her SVAS). Catheterization pressures included: left ventricle 250/10; supravalvular chamber 250/70;
Figure 1

Echocardiographic technique for examination of the ascending aorta. The transducer is placed on the chest wall in the second or third left interspace adjacent to the manubrium; generally aortic root and valve can be recorded when the transducer is perpendicular to the chest wall (position 1). The supravalvular area is visualized by angling the transducer cephalad and medially in a sweep. If the transducer beam strays too far laterally, the anterior pulmonic valve can be seen (position 2). As the transducer is directed even more superiorly, the right pulmonary artery posterior to the aorta is seen (position 3).

ascending aorta 100/60. Systolic gradients of 60 mm Hg were present between main pulmonary artery and both right and left pulmonary arteries. Angiography (fig. 3) showed severe supravalvular aortic stenosis 3 cm in length with proximal and distal aortic dilatation and narrowing of the right coronary artery and left common carotid arteries at their origins. At echocardiography, there was significant supravalvular narrowing which was present as far superiorly as the aorta could be scanned (table 1, fig. 4). The patient underwent surgery during which a dacron patch was utilized to enlarge the stenotic area from an internal diameter of about 7 mm to 16 mm. Repeat echocardiogram revealed a persistent supravalvular narrowing and the area of patching was seen (fig. 5).

Patient 2. The six-year-old sister of Patient 1 was catheterized at age four years at another institution

Table 1

Echocardiographic Data on the Aorta in 12 Normal Subjects

<table>
<thead>
<tr>
<th></th>
<th>Age (yr.)</th>
<th>Aortic echo diameter (cm)</th>
<th>% Echo change</th>
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<tr>
<td></td>
<td>AA (cm)</td>
<td>AA (cm)</td>
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<tr>
<td>DM</td>
<td>2</td>
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</tr>
<tr>
<td>SH</td>
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<td>1.4</td>
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<tr>
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<td>5</td>
<td>1.9</td>
<td>1.9</td>
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<td>WT</td>
<td>7</td>
<td>1.9</td>
<td>1.8</td>
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<td>SP</td>
<td>7</td>
<td>2.2</td>
<td>1.9</td>
</tr>
<tr>
<td>LH</td>
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<td>2.4</td>
</tr>
<tr>
<td>JW</td>
<td>10</td>
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</tr>
<tr>
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<td>11</td>
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<td>CE</td>
<td>12</td>
<td>2.4</td>
<td>2.1</td>
</tr>
<tr>
<td>BM</td>
<td>13</td>
<td>2.3</td>
<td>2.3</td>
</tr>
<tr>
<td>RH</td>
<td>15</td>
<td>2.8</td>
<td>2.6</td>
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<td>2.08</td>
<td></td>
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<tr>
<td>SEM</td>
<td>±0.11</td>
<td>±0.10</td>
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</table>

Abbreviations: AA = diameter in ascending aorta; AV = diameter at aortic valve; SEM = standard error of the mean.

Figure 2

Sweep of a normal aorta. Aortic root and portions of the aortic valve leaflets are seen at the left. The aortic diameter is measured from the most anterior echoes of the anterior and posterior aortic wall. As the scan moves superiorly up the aorta, the left atrium is lost. The ascending aortic width is again measured at this point. In normals, the aortic widths at both levels are almost identical (table 1). Paper speed 50 mm/sec. Abbreviations: LA = left atrium; AA = ascending aorta; Ao = aorta; AR = aortic root; AV = aortic valve.

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and a left ventricular angiogram showed moderate SVAS which extended several centimeters above the aortic valve. Echocardiography revealed supravalvular narrowing as in Patient 1 (table 1, fig. 6).

Patient 3. A two-and-one-half-year-old girl and the youngest sibling of Patients 1 and 2 was asymptomatic, but a harsh ejection murmur radiating into the neck and ECG evidence of mild left ventricular hypertrophy were present. Catheterization has not been performed. Echocardiogram showed supravalvular narrowing similar to that seen in her sisters (table 1).

Patient 4. An 18-year-old girl was catheterized at age 11 and found to have SVAS with a systolic gradient of 150 mm Hg. She, like Patient 1, had a dacron patch placed to widen the area of obstruction at that time. She underwent repeat left-sided catheterization seven years after operation. This revealed a residual 50 mm Hg supravalvular gradient and moderate aortic regurgitation. Angiography revealed an area of supravalvular aortic narrowing (fig. 6). Echocardiography revealed supravalvular narrowing (fig. 7); the area above the constriction could not be resolved echocardiographically.

Patient 5. A seven-and-one-half-year-old girl had evidence of SVAS syndrome. Catheterization data revealed significant stenosis of the proximal right and left pulmonary arteries. No left ventricular, aortic or intra-aortic gradients were detectable. Angiography, however, showed discrete SVAS of mild degree. Echocardiography confirmed the supravalvular narrowing (table 1).

Patient 6. A nine-and-one-half-year-old male also manifested the triad of mental retardation, abnormal facial characteristics, and SVAS. He underwent cardiac catheterization at four years; a supravalvular gradient of 33 mm Hg was recorded. There was no evidence of other cardiac abnormalities, including peripheral pulmonic stenosis. He has remained

Figure 3

Supravalvular angiogram in Patient 1. Significant supravalvular narrowing extends over a 3–4 cm area (white arrows). Proximal and distal dilation of the aorta is present. Narrowing of the origin of the right coronary (open arrow) and left common carotid (black arrow) arteries is also visible.

Figure 4

Preoperative echocardiogram of Patient 1. The ascending aorta is scanned from left to right. The narrowing in the supravalvular aortic segment (SAS) is indicated by the length of the arrows. Paper speed 25 mm/sec. RPA = right pulmonary artery; LA = left atrium.
asymptomatic. Echocardiography showed characteristic supravalvular narrowing.

Echocardiographic Analysis

The echocardiographic data are summarized in tables 1 and 2. In normals, the echocardiographic aortic diameter was fairly constant from the level of aortic leaflets to the arch. The mean diameter of the aorta at the level of the aortic valve leaflets was 2.17 ± 0.11 cm (mean ± SEM). The diameter in the ascending aorta was 2.08 ± 0.10 cm. The difference in mean diameters was not significant when tested with Student's t-test. The percentage change ranged from +11% to -14% when comparing ascending aortic diameter to aortic diameter at the level of the leaflets. In each patient with SVAS, a decrease in aortic dimension was recorded in the ascending aorta compared with the level of the aortic leaflets. The mean aortic diameter at the level of the leaflets was 2.03 ± 0.16 cm and the diameter at the level of minimum aortic dimension was 1.48 ± 0.11 cm. This degree of narrowing is significant at the P < 0.001 level. The narrowing ranged from 19 to 35% of the dimension at the level of the valve. The degree of narrowing of the aorta was within 10% on repeated sweeps.

Radiologic Analysis

The values are included in table 2. As the magnification factors were not recorded, the actual aortic diameters cannot be calculated; hence the measurements are expressed as a fraction of the diameter measured at the aortic leaflets (sinuses). Two instances of hypoplastic aortic segments were noted (Cases 1 and 2) and three of segmental or hourglass narrowing (Cases 4, 5, and 6). The radiologic narrowing ranged from 42 to 63% in the five patients studied.

Discussion

We have found echocardiography to be quite accurate in detecting SVAS in this initial group of patients. The echocardiographic techniques used gave reproducible results when carefully done. There was

Figure 5
Postoperative study of Patient 1. Sweep of the ascending aorta from left to right, paper speed 50 mm/sec. Residual evidence of the supravalvar narrowing remains on the right (SAS) as an inferior echo-free segment in the aortic lumen. The heavy superior echoes now seen in this area and absent from the preoperative study (fig. 4) probably represent reverberating echoes from the Dacron patch used to widen the lumen.

Figure 6
Supravalvar angiogram in Patient 4 with prior surgery for supravalvar aortic stenosis. Residual focal or hourglass supravalvar narrowing is present (arrows). Thickening of the aortic valve leaflets and moderate aortic regurgitation are present.
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Figure 7
Echocardiogram from Patient 4, paper speed 50 mm/sec. Though the echograms are mounted separately, they are from a continuous sweep. Supravalvular narrowing is evident in the right panel representing the supravalvular aortic segment (SAS).

clear echocardiographic supravalvular narrowing in each of the six patients examined. Conversely, in 12 children with normal aortas, echocardiographic change was found to be absent or minimal. Thus, echocardiography appears to be a reliable, noninvasive technique for documenting or excluding SVAS. The difficulties inherent in clinically differentiating supravalvular from valvular or subvalvular aortic stenosis make this technique especially valuable.

In contrast to the previous report of a single case of localized SVAS, we were not able to document reconstitution of the aortic diameter above the area of narrowing, and therefore we were unable to differentiate instances of hourglass narrowing from the more diffuse hypoplastic variety. This was due to our inability to resolve posterior aortic wall above the area of narrowing.

In vivo, the ascending aorta's maximum diameter is at the level of the sinuses of Valsalva. This is seen radiologically in the normal aorta and in our patients comparing the radiologic sinus diameter to that of the aorta above the supravalvular narrowing (table 1). As one angles the sound beam cephalad from the sinuses to sweep the ascending aorta, the beam strikes the

Table 2
Echocardiographic, Radiologic, and Hemodynamic Data in Six Patients with Supravalvular Aortic Stenosis

<table>
<thead>
<tr>
<th>Patient</th>
<th>Age (yr)</th>
<th>Aortic echo diameter (cm)</th>
<th>% Echo change</th>
<th>Radiologic diameters (arbitrary units)</th>
<th>Ao Above SAS</th>
<th>Estimated length of SAS</th>
<th>Supravalvular systolic gradient (mm Hg)</th>
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<tr>
<td>DH</td>
<td>8</td>
<td>2.3 1.5</td>
<td>-35</td>
<td>Sinus 1.0 SAS 0.37 % Angio change 63.58</td>
<td>3 cm</td>
<td>150</td>
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<tr>
<td>JH</td>
<td>6</td>
<td>1.6 1.2</td>
<td>-25</td>
<td>ND 1.0 SAS 0.50 % Angio change 50.76</td>
<td>3-4 cm</td>
<td>ND</td>
<td></td>
</tr>
<tr>
<td>AH</td>
<td>2.5</td>
<td>2.0 1.5</td>
<td>-25</td>
<td>ND 1.0 SAS 0.40 % Angio change 60.66</td>
<td>ND</td>
<td>ND</td>
<td>50</td>
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<tr>
<td>SR</td>
<td>8</td>
<td>2.6 2.0</td>
<td>-23</td>
<td>1.0 1.0 SAS 0.40 % Angio change 60.66</td>
<td>discrete hourglass</td>
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<tr>
<td>TC</td>
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<tr>
<td>CM</td>
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<td>discrete hourglass</td>
<td>discrete</td>
<td>33</td>
</tr>
</tbody>
</table>

Abbreviations: Ao = aorta; AV = diameter at aortic valve; ND = no data; SAS = minimum diameter in supravalvular aortic segment; Sinus = diameter at level of aortic sinuses.
aorta increasingly obliquely and the apparent internal diameter is enlarged (fig. 1). In the normal, the opposing effects of the anatomic narrowing of the aorta above the sinuses and the increasing obliqueness of the angle of the beam striking the aorta apparently counteract one another and the measured aortic diameter appears to change little as the sound beam moves up the ascending aorta.

In patients with SVAS, these same factors are present and tend to diminish the apparent severity of narrowing as judged by radiography. In addition, with the sometimes rather striking dilatation of the sinuses of Valsalva in SVAS, there is greater opportunity for the echo beam to strike obliquely or off-center and hence produce a falsely narrow diameter at this level. Thus, echocardiography may underestimate the aortic width at the level of the sinuses and overestimate it in the ascending aorta. However, patients with the most severe degree of narrowing as judged by radiography and pressure gradients tended to have the greatest echocardiographic narrowing. Examination of the "quantitative" figures indicates that echocardiography produced a nonlinear underestimation of the severity of the narrowing.

Although the effect of an increasingly oblique sound beam sweeping up the aorta might be expected to obscure an anatomic discrete obstruction, especially of mild degree, the technique was reliable in our two cases (5 and 6) where the obstruction was hemodynamically mild and fairly discrete anatomically. This series did not include any cases of the discrete membranous type of SVAS. Therefore, we can only speculate that in the membranous type, echocardiography may be valuable in identifying the proximal aortic dilatation which occurs with significant obstruction. The membrane may not be identified, although visualization of a discrete subaortic membrane has been reported.10

Acknowledgment

We would like to acknowledge the assistance of Ms. J. Welty in manuscript preparation.

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Circulation, Volume 52, November 1975
Echocardiographic features of supravalvular aortic stenosis.
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*Circulation*. 1975;52:817-822
doi: 10.1161/01.CIR.52.5.817
*Circulation* is published by the American Heart Association, 7272 Greenville Avenue, Dallas, TX 75231
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Print ISSN: 0009-7322. Online ISSN: 1524-4539

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