Diagnosis of Ventricular Septal Defect
by Pulsed Doppler Echocardiography

Sensitivity, Specificity and Limitations

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SUMMARY The M-mode echocardiographic findings of ventricular septal defect (VSD) are nonspecific. A specific pulsed Doppler echocardiographic (PDE) diagnosis of VSD can be made by following the turbulent VSD jet through the septum. To assess the sensitivity, specificity and limitations of PDE diagnosis of VSD, 105 children undergoing cardiac catheterization were examined by PDE. These children had a variety of cardiac defects, and a PDE diagnosis of VSD was made in 46/51 (90%) who had VSD proven at catheterization. There was one false positive PDE diagnosis of VSD, for a specificity of 98%. Factors influencing the ability to diagnose VSD by PDE include the location of the defect, level of pulmonary vascular resistance and direction of blood flow through the VSD. The presence of additional defects did not interfere with PDE diagnosis of VSD. The PDE detection of additional defects may identify situations where M-mode echocardiographic estimation of dimensions may not be indicative of the size of the VSD shunt.

THE M-MODE ECHO findings of ventricular septal defect (VSD) are usually suggestive unless there is significant aortic overriding of the VSD. The severity of the defect is usually estimated indirectly on the basis of shunt-induced alterations in size or function of cardiac chambers. Pulsed Doppler echocardiography (PDE), by its ability to determine direction and quality (smooth vs rough) of blood flow at given sites, allows specific detection of the jet of the VSD, and detection of associated defects that may alter the reliability of using echocardiographic dimensions to assess the magnitude of shunt at ventricular level.

Materials and Methods

To assess the sensitivity, specificity and limitations of the PDE diagnosis of VSD, we reviewed PDE experience over a series of 105 infants and children having adequate PDE exams and undergoing cardiac catheterization. The 105 children (65 male, 43 female, age 1 day to 13 years) had a variety of cardiac defects, including 51 with VSD. In 25 of the 51, the VSD was associated with only an interatrial communication, usually a patent foramen ovale; the VSD was a component of tetralogy of Fallot in seven, endocardial cushion defect in six and truncus arteriosus in five. It was associated with transposition of the great arteries (TGA) in three and patent ductus arteriosus (PDA) without characteristic ductal murmur, in three. The M-mode and PDE exams, performed during the hospitalization for catheterization, were performed with a previously described research prototype PDE unit, or its commercially available counterpart (3 and 5 MHz Echo, 3 or 5 MHz Doppler). The examinations were complete, with standard M-mode assessment of vessel, valve, chamber position and measurements, and PDE assessment of blood flow throughout each cardiac chamber and great vessel, as well as proximal and distal to each valve. Cardiac dimensions were compared with established normal values. The diagnosis of VSD by PDE was made by following the turbulent VSD jet through the septum (fig. 1). The presence of turbulent flow by PDE on either side of the septum is not specific for VSD, since such turbulence may be due to subvalvular stenosis; only by following flow through the septum is one sure that a VSD is present. The position in which the septum was traversed was recorded as high, apical or near the atroventricular (AV) valve plane, and compared to the angiographic or operative visualization of shunt site.

Flow abnormalities characteristic of additional defects were 1) patent ductus arteriosus: turbulent diastolic flow in pulmonary artery, directed toward precordial transducer (fig. 2); 2) atrial septal defect (ASD): turbulent right atrial flow traced to position of atrial septum and just across it (fig. 3); 3) AV valve insufficiency: turbulent systolic jet posterior to respective valve; and 4) semilunar valve insufficiency: turbulent diastolic flow beneath respective valve in respective outflow tract (fig. 1).

Results

The results of PDE diagnosis of VSD are summarized in table 1. The turbulent VSD jet could be followed through the septum in 46 of the 51 children with proven VSD, a sensitivity of 90%. There was one false positive diagnosis of VSD, in a child whose catheterization showed only the presence of a left ventricular-to-right atrial shunt, yielding a specificity of 98%. Five patients had catheterization-proven VSD, but the turbulent jet could not be followed through the septum. Four of these five had large defects with pulmonary hypertension (pulmonary resistance, 710 to 1,066 dyne-sec-cm⁻²), and while right ventricular
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Figure 1. Pulsed Doppler echocardiographic (PDE) recording following turbulent blood flow through the ventricular septum. At the top is a condensed M-mode echocardiogram, showing the right ventricle (RV), left ventricle (LV) and septum (S). SV indicates position of sample volume in which position flow is sampled. ECG = electrocardiogram, and FLOW = PDE spectral flow record. On the right, the SV is in RV and during systole, the flow record is comprised of scattered dots, representative of the multiple directions and velocities of the rough systolic flow in the RV. Note that in diastole, the flow record from RV is comprised of dots closely clustered into a clean wave form, indicative of smooth diastolic flow in the RV. When the SV is positioned in the septum, the flow record reveals presence of harsh systolic flow (scattered dots in systole), as the turbulent VSD jet is followed into the septum. As the SV is moved to the LV, harsh systolic flow is seen just at the left ventricular margin of the septum near the arrow. The left ventricular systolic flow is not rough, but diastolic flow is quite rough. This patient also had aortic insufficiency, with PDE recording of rough diastolic flow in the left ventricular outflow tract.

outflow tract flow was rough, no discrete jet could be followed. One child had a tiny apical VSD (visualized on left ventricular angiocardiogram) and turbulent flow detected by PDE in the right ventricular apex, but not through the septum. The remaining VSD jet, in another child with TGA, moderate pulmonary hypertension, and a small ventricular shunt from right ventricle to left ventricle, could only be followed up to, but not

Figure 2. Pulsed Doppler echocardiographic findings in patent ductus arteriosus. In panel A, PA = pulmonary artery, AO = aorta, and 1 and 2 are precordial transducer positions. The hatched tear-drop represents the position of the sample volume, being just distal to pulmonic valve in 1, and deep in pulmonary artery in 2. In panel B, representative flow records are shown, obtained from precordial positions 1 and 2. On the left, the sample volume (SV) is in position 1, distal to pulmonic valve (PV), and the flow record is of smooth systolic flow directed away from the transducer. On the right, the SV is at position 2, deep in pulmonary artery and diastolic ductal flow directed toward the transducer, and is shown in the flow record.
through, the septum. All of these five false negatives had PDE flow patterns suggestive of VSDs, but no firm PDE diagnosis of VSD could be proven without following the jet through the septum.

The majority (41 of 51) of the VSDs were high, as judged by angiocardiography or surgical visualization. The high VSD position had been correctly identified by PDE in those who had PDE diagnosis of VSD. Two VSDs were apical, and one of these could not be proven by PDE because we could not follow the jet through the septum. The six endocardial cushion defect VSDs were correctly located by PDE, as was one non-endocardial cushion defect VSD located posterior to the tricuspid valve.

Additional flow abnormalities were discovered in 39 patients. These PDE findings included ASD in 25, tricuspid insufficiency (TR) in five, mitral insufficiency (MR) in five, PDA in three and aortic insufficiency in one. Some patients had more than one additional flow abnormality, and often, the additional flow abnormality had not been clinically manifest (three with PDA did not have a ductal murmur, three of eight with MR did not have a MR murmur detected, and none of the children with TR had a TR murmur detected).

The left atrial (LA) dimension exceeded established normal values in most patients with pulmonary-to-systemic flow ratios of greater than 2:1. In nine, however, the LA dimension was not abnormal, in spite of a significant VSD shunt, measured at catheterization (table 2). All nine had PDE findings of an interatrial communication and catheterization evidence of significant atrial step-up without a significant interatrial pressure gradient.

**Discussion**

Factors influencing the ability to diagnose VSD by PDE include the location of the defect and position of the septum, the level of pulmonary resistance and the direction of blood flow through the VSD. Since the majority of VSDs are high or membranous, and have left-to-right shunts, the usual procedure for their detection involves examination of the right ventricular side of the septum or the septum itself, beginning high under the pulmonary valve, and proceeding toward the apex. Examples of normal ventricular flows are shown in figure 4. When a VSD is present, flow throughout the right ventricular (RV) outflow tract may be rough, but usually an area of maximal turbulence is found and can be traced into and through the septum by angulating the transducer and controlling the depth of the sample volume (fig. 1). When the septum is not in the usual position, such as with gross enlargement of one ventricle and resultant displacement and/or rotation of the septal plane with septal plane nearly parallel to Doppler beam, or in situations that may also have reversed direction of

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**TABLE 1. Pulsed Doppler Echocardiographic Detection of Ventricular Septal Defect**

<table>
<thead>
<tr>
<th>Type</th>
<th>Cath/Angio VSD</th>
<th>No VSD</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pulsed</td>
<td>VSD 46</td>
<td>1 TP</td>
</tr>
<tr>
<td>Doppler</td>
<td></td>
<td>1 LV-RA</td>
</tr>
<tr>
<td>Echo</td>
<td>No 5 FN</td>
<td>53 TN</td>
</tr>
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</table>

**FIGURE 3. Pulsed Doppler echocardiographic findings in atrial septal defect (ASD). Panel A shows position of right atrium (RA) and left atrium (LA), separated by the atrial septum. The respective atrioventricular valves are hatched. In transducer position A, the sample volume is in the LA, and in position B, in the RA. Panel B shows a flow record obtained from a child with proven ASD. The sample volume (SV) is posterior to the tricuspid valve (TV), and a very scattered flow record of rough flow is seen, corresponding to position B in panel A. The transducer is angled to position the SV in the LA, sweeping across position of the atrial septum while flow record displays scattered, rough flow stemming from the ASD flow.**

Abbreviations: VSD = ventricular septal defect; TP = true positives; FP = false positives; LV = left ventricle; RA = right atrium; TGA = transposition of the great arteries; PHT = pulmonary hypertension; LOW = low muscular defect; FN = false negatives; TN = true negatives.
flow through VSD, as in TGA and VSD, the maneuvers for following VSD jet through the septum may be difficult or impossible. When the VSD is not high, the maximal turbulence is usually found in another portion of the right ventricle, and the septum can be traversed at that level. When the VSD is a component of a more complex defect (tetralogy of Fallot, truncus arteriosus or endocardial cushion defect), one receives additional clues from M-mode as to the location of the VSD. The one false positive in our series occurred in a child with left ventricular-to-right atrial (LV→RA) shunt. Her PDE exam was interpreted as representative of a VSD plus TR, a combination that may be difficult to distinguish from LV→RA shunt by angiocardiography as well. The diagnosis of VSD by PDE in patients with endocardial cushion defect may present special problems, since flow may be from left ventricle to left atrium by cleft mitral valve, from left ventricle to right atrium, or from left ventricle to right ventricle underneath the valve plane. In all six patients with endocardial cushion defects, ventricular level shunts were present by PDE and angio. We have had some difficulty determining the presence or absence of VSD in other patients with endocardial cushion defect, primarily because of difficulties in establishing the level of AV valve plane with abnormal mitral attachment. The presence of VSD in endocardial cushion defect may be difficult to prove by LV angiocardiography as well.

Since the turbulence of the VSD jet facilitates its being followed through the septum, diminution of turbulence, such as would be expected with increased pulmonary resistance, may make a VSD jet more difficult to follow. Of the VSDs whose jets could not be followed through the septum, four of the five were associated with elevated pulmonary resistance, and one of these (TGA/VSD) had a net shunt from right ventricle to left ventricle. When the VSD shunt is right to left, the maximal turbulence is found in the LV outflow tract or at the aortic valve, and can be followed back through septum to RV outflow tract. The degree of turbulence of right-to-left VSD shunts seems considerably less than with those shunting left to right, so the jet is more difficult to follow.

The presence of shunts at other than ventricular level did not interfere with our ability to diagnose the VSD, since atrial, ventricular, and great vessel level shunts each have characteristic flow patterns that can be localized to the respective level (figs. 1–3). The PDE detection of other shunts (ASD, PDA) may be useful, since the shunts may be clinically silent. Likewise, MR or TR may be detected by PDE, while auscultatory findings may be masked by the VSD murmur. These additional "silent" defects may be of hemodynamic importance, and their detection by PDE is important in order to correctly interpret echocardiographic dimensions. For example, in a child with VSD and associated PDA and/or MR, both the VSD and associated defects may be contributing to LA enlargement. If the associated defects are silent, as occurred in several patients in our series, one may erroneously conclude that the degree of LA enlargement seen on M-mode echo is due only to VSD, when in fact the LA enlargement is the result of multiple defects.

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**TABLE 2. Nine Patients with Large Ventricular Septal Defect, but Without Large Left Atrial Dimension**

<table>
<thead>
<tr>
<th>Age (Months)</th>
<th>Surface Area (m²)</th>
<th>Weight (kg)</th>
<th>Qp/Qs</th>
<th>LA Dimension (mm)</th>
<th>Top Normal LA Dimension (mm)*</th>
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<tbody>
<tr>
<td>2.5</td>
<td>—</td>
<td>2.7</td>
<td>4.7/1</td>
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</tr>
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<td>6.6</td>
<td>2.2/1</td>
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<td>18</td>
</tr>
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<td>0.5</td>
<td>11</td>
<td>3/1</td>
<td>17</td>
<td>20</td>
</tr>
</tbody>
</table>

*Normal values for those less than 0.3 m² are from reference 5; those 0.3 m² or larger are from reference 6.

| Abbreviations: Qp/Qs = pulmonary-to-systemic flow ratio; LA = left atrial. |

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**FIGURE 4. Normal right and left ventricular pulsed Doppler echocardiographic flow patterns. RV = right ventricle; LV = left ventricle; SV = sample volume; IVS = ventricular septum. The SV is being moved from the RV (left side of figure) across the septum (center) to the LV (right side of figure). With SV positioned in RV, or LV, the flow record is seen to be comprised of closely organized non-scattered dots, indicative of smooth flow. Contrast to flow record in figure 1. No discernible flow signal is found with SV in the septum.**
Just as loudness of murmur need not correlate with shunt size, degree of turbulence of the VSD jet need not correlate with shunt size. The breadth of the VSD jet may provide a clue as to the VSD size, with some of the smallest very turbulent and difficult to follow through the septum. Some of the anatomically larger defects had only mild turbulence, but covered a broad area of the septum. We continue to use LA and LV dimensions and LV contractility as indicators of the magnitude of left-to-right VSD shunt when PDE reveals no additional defect. All of our patients with pulmonary-to-systemic flow ratios greater than 2:1 had large LA dimension, LV dimension or increased VCF. In nine, however, decompression had apparently occurred at atrial level, since LA dimensions were normal (table 2). These nine had PDE evidence of left-to-right atrial shunt, so LA dimension was not used as an index of shunt size. It seems reasonable to conclude that if the left atrium or left ventricle are large in a patient with VSD, the likelihood of a significant shunt at ventricular or great vessel level is great. If pulmonary arterial flow is rough in systole and normal in diastole, a PDA is not likely, and the shunt is likely only at the ventricular level. In patients with VSD and small or normal LA dimension, and with no PDE evidence of atrial shunt, it is likely that the VSD shunt is small. If an interatrial communication is present by PDE, caution is in order, as our nine patients with normal LA dimension and interatrial communication all had sizable VSD shunts, showing that M-mode echo determination of LA size in patients with VSD may not always reflect VSD shunt size.

References
Diagnosis of ventricular septal defect by pulsed Doppler echocardiography. Sensitivity, specificity and limitations.
J G Stevenson, I Kawabori, T Dooley and W G Guntheroth

*Circulation*. 1978;58:322-326
doi: 10.1161/01.CIR.58.2.322
*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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