Whether coarctation or interruption (IAA) of the aorta, ostensibly similar in morphology (and management), result from the same or different developmental errors can be inferred by examining the pattern of associated anomalies. Among the most common associated lesions, especially in IAA, is ventricular septal defect (VSD). Although muscular and perimembranous VSDs are the most common in aortic coarctation, the prevalence of various VSD morphologies in IAA has not been examined in as much detail. As part of the recent prospective multiinstitutional study of IAA conducted by the Congenital Heart Surgeons Society, 53 echocardiographic studies were reviewed; 42 of 45 patients with type B IAA had VSDs involving maldevelopment of the outflow region. In type A IAA, a significantly lower percentage (4/8) had this kind of VSD. Therefore the mechanism of development of type B IAA is likely to be different from that of type A IAA. (J Am Soc Echocardiogr 1996;9:199-201.)

Whether coarctation or interruption (IAA) of the aorta, ostensibly similar in morphology (and management), result from the same or different developmental errors can be inferred by examining the pattern of associated anomalies. Among the most common associated lesions, especially in IAA, is ventricular septal defect (VSD). Although muscular and perimembranous VSDs are the most common in aortic coarctation, the prevalence of various VSD morphologies in IAA has not been examined in as much detail. In fact, Freedom et al.² stated that there is no obvious relation between the position of the ventricular septal defect and the type of aortic interruption. Postmortem series³ are always deal with biased populations; thus a large cohort examined antemortem would help clarify the issue. A recent prospective multiinstitutional study conducted by the Congenital Heart Surgeons Society⁴ afforded us the opportunity to examine the distribution of VSD morphologies in a large number of relatively unselected patients with IAA.

Although ascertaining VSD morphology by angiography is usually successful, distinguishing between malalignment and membranous VSDs is difficult without clear visualization of the aortic outflow. In most infants with large VSDs, dye injected into the left ventricle predominantly opacifies the pulmonary arterial circulation, hampering angiographic visualization of the aortic outflow. Because the magnitude of left-to-right shunting does not affect ultrasonic characterization of the aortic outflow region, echocardiography offers some advantages in assessing the anatomic types of VSD encountered in patients with IAA.

MATERIAL AND METHODS

The Congenital Heart Surgeons Society prospectively evaluated the outcome of 174 infants seen within the first 30 days of life with IAA and VSD who underwent repair at 30 institutions during a 5-year period.⁵ Echocardiograms for 75 of these cases were available for review. Patients with truncus arteriosus were then excluded; of the remainder, 53 patients with segmental anatomy (i.e., situs solitus, ventricular loop, and solitus great arteries) were entered into this study.

A single observer (A.J.C.) reviewed all 53 echocardiographic studies to analyze the anatomic type of VSD. The types were (1) malalignment; (2) malalignment, with infundibular septal hypoplasia (or absence); (3) infundibular septal hypoplasia (or absence), without obvious malalignment; (4) membranous; (5) muscular; and (6) atrioventricular canal type. No attempt was made to subdivide muscular VSDs further into posterior, anterior, mid, and apical.

Infundibular septum was defined echocardiographically as the structure lying below the level of the arterial valves that septated the outflow region. If the two arterial
Figure 1  Subcostal long-axial oblique view. Malalignment of infundibular septum (arrow-head) in relation to plane of trabecular (muscular) septum (sept). Note that anteriormost margin of aortic (ao) root is posterior to plane of trabecular septum. alpm, Anterolateral papillary muscle; LV, left ventricle; A, anterior; R, rightward; S, superior.

Table 1  Anatomic types of VSD in IAA

<table>
<thead>
<tr>
<th>Anatomic type of VSD</th>
<th>Type A IAA (n)</th>
<th>Type B IAA (n)</th>
<th>Total (n)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Malalignment</td>
<td>4</td>
<td>30</td>
<td>34</td>
</tr>
<tr>
<td>Malalignment with infundibular septal hypoplasia (or absence)</td>
<td>0</td>
<td>5</td>
<td>5</td>
</tr>
<tr>
<td>Infundibular septal hypoplasia (or absence)</td>
<td>0</td>
<td>7</td>
<td>7</td>
</tr>
<tr>
<td>Membranous</td>
<td>2</td>
<td>3</td>
<td>5</td>
</tr>
<tr>
<td>Muscular</td>
<td>2</td>
<td>0</td>
<td>2</td>
</tr>
<tr>
<td>Total</td>
<td>8</td>
<td>45</td>
<td>53</td>
</tr>
</tbody>
</table>

It is important not to let VSD size cloud the issue of whether a VSD is malalignment type or membranous. Many malalignment VSDs can appear less than 4 mm in the vertical direction when imaged in the long-axial oblique view or sagittal view. Furthermore, it is important to remember that a membranous VSD must be contiguous to the aortic valve cusps.

RESULTS

Type B IAA accounted for 45 (85%) of the 53 patients. Forty-two (93%) of 45 patients had VSDs involving maldevelopment of the outflow region (Table 1). The most common anatomic type was malalignment (Figure 1). Not a single patient had a muscular VSD. Type A IAA was seen in eight (15%) of the 53 patients. Four patients had malalignment VSDs. The percentage of VSDs involving maldevelopment of the outflow region (50%) was significantly less (p = 0.04, Fisher’s exact test) in type A versus type B IAA.

DISCUSSION

Although the development of the outflow tract of the heart appears to be linked to the formation of the aortic arches, the reason behind this linkage is still unknown. Three possible models come to mind.
First, abnormal septation of the outflow tract modifies hemodynamics in utero and this in turn alters the formation of the aortic arches. Second, genetic alteration leads to abnormal formation of both outflow tract and aortic arches. Third, some aortic arch anomalies could be a result of hemodynamic alteration induced by outflow tract anomalies, whereas other aortic arch anomalies are a result of genetic defects that also affect outflow tract development.

Our study, together with other data, is most consistent with the latter two models. There is strong evidence that DiGeorge and Shprintzen (velocardiofacial) syndromes, part of the Catch-22 phenotype, result from deletions of 22q11, a particular region of chromosome 22. Patients with DiGeorge syndrome with IAA always have type B. If both types of IAA were the result of hemodynamic alteration such as subaortic stenosis, patients with DiGeorge syndrome would be expected to have both types of IAA. Thus in patients with type B IAA and VSD, it is likely that genetic alteration causes abnormal formation of both the outflow tract and the aortic arches.

If both type B and type A IAA were caused by the same mechanism, one would expect a similar distribution of associated lesions. Our data on the distribution of anatomic types of VSD are consistent with the hypothesis that the mechanism of development of type B IAA is likely to be different from that of type A IAA. Thus either a genetic alteration other than deletion of 22q11 is responsible for type A IAA or an in utero hemodynamic alteration (as has been postulated for coarctation) is responsible for type A IAA.

In a previous postmortem series, the prevalence of VSD involving the outflow tract in the two types of IAA was not as disparate (6/12 in type A versus 15/20 in type B). The difference between their study and ours is likely to arise from differences in selection and sample size. Our results appear to be more similar to the report of Van Mierop and Kutsche that 17 (81%) of 21 postmortem cases of type B IAA were a “malalignment VSD,” although they were designating as “malalignment (alone)” cases we would designate “malignment with infundibular septal hypoplasia.”

Finally, our study is limited by the fact that the accuracy of echocardiography compared with the reference standard of autopsy as a way of distinguishing this particular type of malalignment VSD has not been tested; however, others have reported high agreement rates (91%) between echocardiography and angiography for malalignment VSDs taken as a group.

We thank Dr. John W. Kirklin for allowing us access to the Congenital Heart Surgeons Society database.

REFERENCES