Echocardiographic Diagnosis Alone for the Complete Repair of Major Congenital Heart Defects

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Objectives. The study was done to determine the diagnostic accuracy of echocardiography alone in the preoperative diagnosis of children with major congenital heart defects undergoing primary complete repair.

Background. Although echocardiography is well established as the first-line imaging technique for the diagnosis of all forms of congenital heart disease, most institutions continue to perform cardiac catheterization prior to complete repair of more complex defects.

Methods. To determine the diagnostic accuracy of echocardiography alone and echocardiography plus catheterization, we reviewed the records of 503 children with major congenital heart defects who underwent primary complete repair at our institution between July 1992 and June 1997. We included children with transposition of the great arteries, tetralogy of Fallot, double-chamber right ventricle, interrupted aortic arch, aortic coarctation, atrioventricular septal defect, truncus arteriosus, aortopulmonary septal defect, and totally anomalous pulmonary venous return. We excluded children with less complex defects such as isolated shunt lesions, as well as those with the most complex defects that would require surgical palliation (e.g., functional univentricular heart). We defined major errors as those that increased the surgical risk and minor errors as those that did not. Errors in diagnosis were determined at surgery.

Results. Eighty-two percent of children (412 of 503) underwent surgery after preoperative diagnosis by echocardiography alone. There were 9 major (2%) and 10 minor errors in the echocardiography alone group and 7 major and 5 minor errors in the echocardiography plus catheterization group. The most common type of error was misidentification of coronary artery anatomy in patients with transposition of the great arteries. No error in either group resulted in surgical morbidity or mortality.

Conclusions. This study suggests that echocardiography alone is an accurate tool for the preoperative diagnosis of major congenital heart defects in most children undergoing primary complete repair, and may obviate the need for routine diagnostic catheterization.

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Echocardiography is accepted as the first-line imaging modality for diagnosis of both simple and complex congenital heart defects. In addition, echocardiography provides sufficient preoperative anatomic and physiologic information for repair of defects such as patent ductus arteriosus, and atrial and ventricular septal defects (1–4). For this reason, cardiac catheterization is typically no longer performed prior to repair in young patients with these defects, but has been reserved for patients with more complex congenital heart defects, to complement the diagnostic information obtained by echocardiography.

There is controversy, however, over whether cardiac catheterization is routinely necessary in patients with all major defects (2,3,5–17). Although certain complex defects such as pulmonary atresia with aortopulmonary collaterals invariably require preoperative catheterization, echocardiography alone may be sufficient to obtain the anatomic and physiologic information needed to guide surgical repair in patients with most major congenital heart defects. Indeed, there has been a trend at specialized congenital heart defect centers to reserve preoperative diagnostic catheterization for the most complex defects, owing in part to increasing confidence in echocardiography. Since the late 1980s, a number of reports have highlighted the changing trends in the approach to preoperative diagnosis of congenital heart defects (5–19). These reports, which have mainly focused on simple defects such as isolated shunt lesions, functional single ventricle defects requiring palliative surgery, and a small number of patients with more complex defects, have demonstrated that echocardiography can be utilized safely in select patients. Although the practice of primarily repairing patients with major congenital heart defects without preoperative catheterization is becoming increasingly common at many centers, both the diagnostic accuracy and the safety of this approach have not been established in a large consecutive series.

Thus, to determine the accuracy of echocardiography alone,
we undertook the present retrospective study, evaluating the accuracy of preoperative diagnosis by echocardiography alone and its impact on surgical management and outcome in a select group of patients undergoing primary complete repair of major congenital heart defects. In addition, we determined the preoperative diagnostic accuracy in patients with these same defects who also underwent cardiac catheterization.

Patients and Methods

Patients. We reviewed the records of 503 consecutive patients who underwent primary complete repair of nine major congenital heart defects during the 5-year period from July 1992 through June 1997 (Table 1). We excluded patients who had previously undergone palliative or corrective surgery, patients with primary diagnoses other than those listed above, and patients with complex functional single-ventricle defects that included one or more of the above defects. Patients with tetralogy of Fallot and pulmonary atresia, for example, were excluded because the anatomy and physiology of the pulmonary blood supply can be adequately defined for surgical intervention only by angiography. Patients with unbalanced atrioventricular septal defect, for example, were included only if they underwent complete primary biventricular repair (20).

All patients had undergone comprehensive preoperative evaluation, including physical examination, 12-lead electrocardiogram (ECG), and cross-sectional echocardiography with spectral and color-flow Doppler, as well as contrast echocardiography when indicated. In patients referred for surgery by cardiologists outside our institution, the outside echocardiogram was reviewed for adequacy at a weekly combined pediatric cardiology and cardiothoracic surgery preoperative conference. Echocardiograms were repeated at our institution if additional preoperative information needed to be obtained. In patients who had also undergone cardiac catheterization, the catheterization was performed either before or after referral to our institution, and was for either diagnostic or combined interventional and diagnostic purposes. The decision to perform catheterization was made either by the patient’s referring cardiologist, the attending cardiologist, or if requested by the surgical team. Thus, indications for catheterization were variable, depending on the preferences of the referring physician. In patients who underwent catheterization, discrepancies between echocardiographic and catheterization diagnoses were reported to the surgeon preoperatively.

The indications for catheterization are summarized in Table 2. The primary indication was interventional in 11 patients and diagnostic in 80. In patients with transposition of the great arteries who required a balloon atrial septostomy, the decision to perform the procedure in the catheterization suite instead of at the bedside under echocardiographic guidance was based on physician preference or difficulty obtaining vascular access at the bedside.
Table 3. Associated Defects in 76 Patients With a Combination of Defects

<table>
<thead>
<tr>
<th>Primary Defect</th>
<th>Associated Defects</th>
<th>No. of Patients</th>
</tr>
</thead>
<tbody>
<tr>
<td>Transposition of the great arteries</td>
<td>Interrupted aortic arch</td>
<td>6</td>
</tr>
<tr>
<td></td>
<td>Coarctation of the aorta</td>
<td>9</td>
</tr>
<tr>
<td></td>
<td>Aortopulmonary septal defect</td>
<td>1</td>
</tr>
<tr>
<td></td>
<td>PAPVR</td>
<td>1</td>
</tr>
<tr>
<td></td>
<td>Multiple VSDs</td>
<td>1</td>
</tr>
<tr>
<td>Tetralogy of Fallot</td>
<td>Discontinuous pulmonary arteries</td>
<td>4</td>
</tr>
<tr>
<td></td>
<td>Double chamber right ventricle</td>
<td>4</td>
</tr>
<tr>
<td></td>
<td>Atrioventricular septal defect</td>
<td>6</td>
</tr>
<tr>
<td></td>
<td>Absent pulmonary valve</td>
<td>5</td>
</tr>
<tr>
<td></td>
<td>Multiple VSDs</td>
<td>1</td>
</tr>
<tr>
<td></td>
<td>Straddling tricuspid valve</td>
<td>1</td>
</tr>
<tr>
<td>Truncus arteriosus</td>
<td>Interrupted aortic arch</td>
<td>5</td>
</tr>
<tr>
<td>Aortopulmonary septal defect</td>
<td>Interrupted aortic arch</td>
<td>1</td>
</tr>
<tr>
<td>Atrioventricular septal defect</td>
<td></td>
<td>1</td>
</tr>
<tr>
<td>Coarctation of the aorta</td>
<td></td>
<td>1</td>
</tr>
<tr>
<td>Multiple VSDs</td>
<td></td>
<td>7</td>
</tr>
<tr>
<td>Pulmonary stenosis</td>
<td></td>
<td>1</td>
</tr>
<tr>
<td>LSVC to left atrium</td>
<td></td>
<td>1</td>
</tr>
<tr>
<td>Cor triatriatum</td>
<td></td>
<td>1</td>
</tr>
<tr>
<td>Branch pulmonary arterial stenosis</td>
<td></td>
<td>1</td>
</tr>
<tr>
<td>Coarctation of the aorta</td>
<td></td>
<td>1</td>
</tr>
<tr>
<td>Multiple VSDs</td>
<td></td>
<td>3</td>
</tr>
<tr>
<td>Shone's complex</td>
<td></td>
<td>7</td>
</tr>
<tr>
<td>PAPVR</td>
<td></td>
<td>1</td>
</tr>
<tr>
<td>Interrupted aortic arch</td>
<td></td>
<td>1</td>
</tr>
<tr>
<td>Double aortic arch</td>
<td></td>
<td>1</td>
</tr>
</tbody>
</table>

Patients with more than one primary defect are listed only once. Because some patients had more than one associated lesion, numbers do not total to 76. LSVC = left superior vena cava; PAPVR = partially anomalous pulmonary venous return; VSD = ventricular septal defect.

For the purposes of tabulation and analysis, we analyzed the 503 patients in two groups: those who underwent preoperative echocardiography alone and those who had preoperative cardiac catheterization in addition to echocardiography. In addition, we considered patients to have either a single defect or a combination of defects (Table 3). We defined patients with a combination of defects as those in which there were two or more separate defects (not including a single-ventricle lesion, atrial or ventricular septal defect, or patent ductus arteriosus) that would each, on its own, require surgical repair. All patients in whom two of the major defects (Table 1) were present were considered to have a combination of defects (e.g., transposition of the great arteries with coarctation, or tetralogy of Fallot with atrioventricular septal defect). When two of the primary defects were present, we classified them according to the more severe defect, as listed above and in Table 1.

During the 5-year study period, 503 patients underwent complete primary repair of the nine complex congenital heart defects included in this study; the surgical mortality within 30 days of operation was 3% (n = 16). Of these 503 patients, 412 (82%) underwent surgery with echocardiography alone as the definitive preoperative diagnostic test (echo group) and the remaining 91 (18%) had both echocardiography and catheterization (echo+cath group). The numbers of patients in each group and with each diagnosis are summarized in Table 1. A combination of defects was present in 76 patients (15%; Table 3), 57 of whom (75%) underwent surgery after echocardiography alone. This was a significantly lower percentage than among patients with single defects (57/76, 75% vs. 355/427, 83%; p = 0.04).

The primary outcome measure was the accuracy of preoperative diagnosis. Diagnostic accuracy was assessed by comparing the preoperative diagnosis made by echocardiography and/or catheterization and the diagnosis made at surgery. Errors and surgical diagnoses were identified by reviewing operative reports and records from division morbidity and mortality conferences. In addition, diagnostic errors are recorded in our echocardiography database once they are discovered, and we used this database to identify and cross-reference errors. Errors were categorized retrospectively as major or minor by an author (the surgeon who performed the operations, F.L.H.) who was blinded to patient identity and the preoperative diagnostic imaging modalities used. Errors were defined as major if they necessitated a change in the operative procedure or affected (favorably or adversely) the surgical risk or outcome, and minor if they did not. (For example, we determined the severity—major versus minor—of errors in the identification of coronary anatomy based on the surgical implications. Single coronaries called normal or normal coronaries called single were considered to be more significant errors than were errors of omission or commission regarding a circumflex off the right.) In the echo+cath group, diagnostic accuracy was tabulated according to the combined echocardiographic and catheterization diagnosis.

**Data analysis.** Data are expressed as median and range or mean ± SD. The χ² analysis was used to compare dichotomous variables. Nonparametric analysis of ordinal variables was conducted with the Wilcoxon signed-rank test. An independent-samples t test was used for simple comparison of means between groups, and general factorial analysis of variance was used to compare continuous variables for more than two groups. SPSS for Windows, version 6.01 (SPSS Inc., Chicago, Illinois), was used to perform statistical calculations.

**Results**

Diagnostic errors were identified at the time of surgery by the opening surgeon in a total of 31 out of 503 patients (6%). In the echocardiography-alone group, diagnostic accuracy was 95%. Diagnostic errors were made in 19 of the 412 echo group patients (5%), 8 of them (2%) major and 11 minor (3%). Errors were made in 12 of the 91 echo+cath patients (13%), 7
of them (8%) major and 5 minor (5%). Errors are listed in Table 4. Among the echo group patients, the diagnostic error rate was significantly greater in those with a combination of defects than among those with single defects (7/57, 12% vs. 13/355, 4%; \( p < 0.05 \)). None of the errors in diagnosis led to complications or affected surgical outcome adversely.

No instances arose in which catheterization corrected an echocardiographic error in patients with either a single defect or a combination of defects. There were two patients in whom echocardiography at our institution corrected an erroneous diagnosis made on the basis of catheterization prior to referral to our institution. These diagnoses were both in patients with transposition of the great arteries and normal coronary arteries; in each, the erroneous diagnosis made at catheterization was single coronary artery. Echocardiography group patients (median age 45 days, range 1 day to 30 years) were significantly younger than echocardiography + cath group patients (median age 135 days, range 2 days to 37 years; \( p < 0.001 \)), and patients with a combination of lesions were significantly younger than were patients with single lesions (8 ± 22 months vs. 15 ± 44 months; \( p = 0.03 \)). Among the 91 patients in the echocardiography + cath group, the catheterization was performed at our institution in 43 patients and prior to referral in the remaining 48.

### Discussion

**Results and implications of the study.** In the present study, we have shown that the majority of a large group of patients with major congenital heart defects can undergo primary complete repair safely after echocardiography alone as the definitive preoperative diagnostic modality. The rate of diagnostic errors in these patients was very low (5%), and no error affected patient outcome adversely. Most of the major errors related to coronary artery anatomy in patients who had transposition of the great arteries. This anatomy can be readily recognized at surgery.

Patients with six of the nine defects studied were catheterized \( \leq 20\% \) of the time, and there were no instances of an erroneous echocardiographic diagnosis being corrected at
catheterization. Catheterization was performed more frequently in patients with a combination of defects than in those with single defects, and the rate of echocardiographic errors was higher in patients with a combination of defects. However, none of these errors in diagnosis led to an adverse surgical outcome. Therefore, we believe that surgery after echocardiography alone is safe for primary complete repair in patients with the complex congenital heart defects examined in this study.

Of the remaining three defects, in which catheterization was performed in 23% to 38% of patients, only those with transposition of the great arteries were neonates. Among patients with the remaining two defects (double-chamber right ventricle and tetralogy of Fallot), those who underwent both echocardiography and catheterization were generally older than the patients who underwent echocardiography alone (median age at surgery of echocardiography+cath patients, other than transposition of the great arteries, was 10 months). The indications for catheterization in these older patients were age-related physiologic changes such as increased pulmonary vascular resistance and impaired ventricular function. Because it is increasingly uncommon for patients with defects included in this study to undergo repair beyond early infancy, catheterization for these age-related indications should be necessary less frequently.

Although none of the major errors in this study led to operative complications, some were potentially very significant. Of these, the single most common type of diagnostic error in both the echocardiography-alone patients and the echo+cath patients was incorrect definition of the proximal coronary artery anatomy in patients with transposition of the great arteries, several of whom had intramural or single-coronary arteries. Because the coronary artery anatomy is identified at the time of surgery, and because the arterial switch procedure is performed in patients with transposition and all forms of complex coronary anatomy at our institution, these errors did not alter the surgical treatment. At institutions where an arterial switch is performed selectively according to coronary arterial anatomy, however, these errors might have had grave implications (21–27). It is important to acknowledge that some major diagnostic errors may be inevitable, such as misdiagnosis of an intramural coronary artery, which appears equally difficult to diagnose by echocardiography or angiography, despite reports on its diagnosis by echocardiography (24).

Other defects in which coronary arterial anatomy may complicate repair are tetralogy of Fallot and truncus arteriosus (19,28,29). In our study, diagnostic errors regarding the coronary arteries were made in both of these defects. However, as with transposition, intraoperative evaluation of the coronary arteries is imperative and sufficient in tetralogy and truncus, and complications did not result from these errors.

The only other major error made on more than one occasion was the failure to diagnose a double-orifice in the left-sided component of the common atrioventricular valve in an atrioventricular septal defect, which was committed equally by echocardiography and catheterization. Nevertheless, echocardiography is probably the imaging modality of choice for this particular abnormality (18).

Although there were diagnostic errors in both groups of patients, the risk-benefit and cost-benefit ratios appear to favor echocardiography without catheterization for the defects included in this study. Catheterization did provide some additional diagnostic information in this study. However, the risks and cost of catheterization are markedly greater than those of echocardiography, and the additional diagnostic information and the incremental benefit provided by catheterization was found to be minimal in this study. We have not performed a formal cost analysis in this study, but we estimate that there would be substantial cost savings by avoiding routine diagnostic catheterization. At our institution, the cost of a diagnostic catheterization to a privately insured patient is approximately $9,000, whereas the cost of an echocardiogram is approximately $1,200.

Echocardiography is noninvasive, relatively inexpensive, portable, and easily repeatable, and it has no known side effects or complications. In contrast, cardiac catheterization is invasive, more expensive, and not portable or easily repeatable, and it has complications such as radiation exposure, pulse loss, the need for possible blood transfusion, and a small risk of mortality (30).

Study limitations. Three limitations of this study must be considered. Regarding study design, we only included defects that we believe are amenable to complete primary repair after diagnosis by echocardiography alone. These are defects in which preoperative anatomical rather than hemodynamic information is crucial. Defects excluded by these criteria are those that require staged palliative surgery, and invariably require precise preoperative hemodynamic data only obtained at catheterization.

Another limitation of this study is its retrospective nature, precluding the patients from being randomized to the two groups of preoperative diagnostic modalities evaluated. However, the use of catheterization varies considerably between the referring cardiologists. Thus, the range of indications for catheterization within each group of defects is variable, in some cases reflecting physicians’ different levels of comfort with echocardiography for evaluating such features as the coronary arteries, central pulmonary arteries, and the aortic arch. Over half of the catheterization procedures in the echo+cath patients were performed before the patient was referred for surgery, and may not have been performed in other institutions. Therefore, we did not attempt to evaluate whether each catheterization was indicated, which is a complex question that cannot be assessed adequately in a retrospective study. We acknowledge that there remain a number of indications for diagnostic catheterization in patients with the defects studied in this report, and that every patient with congenital heart disease who requires surgery should be assessed individually to decide whether catheterization is necessary.

Finally, perhaps the major limitation of this study is that it may not be possible to generalize our results to other institutions. The practices and results analyzed in this report were
those of a pediatric cardiac service based at a regional referral center, where a large volume of both common and unusual heart defects are seen. Such a setting facilitates the development of proficiency at diagnosing and operating on complex congenital defects. Moreover, the surgeons at our institution are comfortable operating on the basis of echocardiographic diagnosis alone, which is not the case everywhere. This may have significant implications for patients with defects such as tetralogy of Fallot, which some may opt to palliate in certain cases, or transposition of the great arteries, in which coronary artery variations may preclude some surgeons from performing an arterial switch. However, the established trend supported by our data should lead to greater reliance on noninvasive assessment and a movement away from routine diagnostic catheterization.

Conclusions. In this era of advanced imaging technology and steadily improving surgical techniques, there is a continual effort to provide specialized medical care within a context of increasingly stringent economic limitations. We should always question whether additional costly tests provide essential information, and the most important factor in the equation should always be the effect on the patient. In this review of our experience over 5 years, we have shown that coordinated efforts by cardiologists and surgeons can lead to a reduction in diagnostic procedures and their associated morbidity and cost, with no adverse effect on diagnostic accuracy or patient outcome. On the basis of these findings, we conclude that catheterization adds little to the findings of echocardiography and outcome. On the basis of these findings, we conclude that coordinated efforts by cardiologists and surgeons can lead to a reduction in diagnostic procedures and their associated morbidity and cost, with no adverse effect on diagnostic accuracy or patient outcome. On the basis of these findings, we conclude that catheterization adds little to the findings of echocardiography and outcome. On the basis of these findings, we conclude that coordinated efforts by cardiologists and surgeons can lead to a reduction in diagnostic procedures and their associated morbidity and cost, with no adverse effect on diagnostic accuracy or patient outcome. On the basis of these findings, we conclude that catheterization adds little to the findings of echocardiography and outcome. On the basis of these findings, we conclude that coordinated efforts by cardiologists and surgeons can lead to a reduction in diagnostic procedures and their associated morbidity and cost, with no adverse effect on diagnostic accuracy or patient outcome.

We would like to express our thanks to Julian LE. Hofman, MD, and Kirsten E. Fleischmann, MD, MPH, for their expert review of the manuscript.

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