

# Congenital Cardiac Anomalies

## Prenatal Readings Versus Neonatal Outcomes

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### Abbreviations

VSD, ventricular septal defect

**Objective**—The purpose of this study was to determine the variation between prenatal and postnatal diagnosis of congenital cardiac lesions diagnosed by both fetal center primary physicians and fetal pediatric cardiologists at a single tertiary referral center in the United States and evaluate why cases were misdiagnosed.

**Methods**—A retrospective review of all cardiac abnormalities identified prenatally by level II sonography at a tertiary referral fetal center between January 2006 and December 2008 was performed to include any patient with a fetal cardiac abnormality and with a documented autopsy or neonatal follow-up. Congenital heart disease diagnoses were classified as correct, incorrect, or incorrect but within the same spectrum of disease. Cases of correct diagnosis by primary physicians and pediatric cardiologists were compared.

**Results**—Sixty patients with fetal heart abnormalities were identified among 8894 patients who had level II sonography. The combined detection rate for fetal heart abnormalities for both primary physicians and pediatric cardiologists together was 81.7%. The detection rates of congenital heart disease were not statistically different between primary physicians and pediatric cardiologists: 77.9% (46 of 59) versus 85.0% (34 of 40;  $P = .3$ ). The most common cardiac abnormalities misdiagnosed in our study population included pulmonic stenosis, ventricular septal defect, myxoma, truncus arteriosus, and coarctation of the aorta.

**Conclusions**—Congenital heart disease is misdiagnosed in tertiary care centers by both pediatric cardiologists and fetal imaging specialists. We believe that this occurrence is related to multiple factors, including evolution of congenital heart disease, maternal body habitus, associated congenital anomalies, decreased amniotic fluid volume, gestational age at evaluation, imaging techniques, and, most importantly, the experience of the sonographer.

**Key Words**—congenital heart disease; fetal echocardiography; fetal heart; prenatal detection

Screening for congenital heart disease continues to be a challenge in prenatal diagnosis. Major congenital heart disease has an incidence of 2 to 3 per 1000 live births.<sup>1,2</sup> It is the most common major birth defect and accounts for 40% of perinatal deaths<sup>3,4</sup> and more than 20% of deaths within the first month of life.<sup>5</sup> Despite improvements in imaging techniques and training, the overall detection rate of congenital heart disease remains low. In the literature, prenatal detection rates vary across the world from 16% to 65%.<sup>6–9</sup> The Routine Antenatal Diagnostic Imaging with Ultrasound study reported a poor detection rate even at tertiary referral centers, as low as 16%.<sup>6</sup> Garne et al<sup>7</sup> identified 2454 cases of congenital heart disease from 20 European registries with an overall

prenatal detection rate of 25%. Carvalho et al<sup>10</sup> reported a detection rate for major congenital heart disease of 75% at a single tertiary institution. Oggè et al<sup>8</sup> reported one of the higher rates of 65% using 4-chamber and outflow tract screening views from 15 obstetric units in Piedmont, Italy.

Several studies have suggested that prenatal detection of congenital heart disease can improve neonatal neurologic outcomes and postoperative survival.<sup>11–16</sup> Unfortunately, providers continue to misdiagnose major and minor congenital heart disease despite the improved imaging techniques and attention to different views of the fetal heart. The objective of this study was to determine the variation between prenatal diagnosis (by sonography) and postnatal diagnosis (neonatal echocardiography, cardiac catheterization, or autopsy) of congenital cardiac lesions diagnosed by both fetal center primary physicians and fetal pediatric cardiologists at a single tertiary referral center in the United States, and evaluate why cases were misdiagnosed.

## Materials and Methods

We conducted a retrospective review of all patients undergoing prenatal level II sonography at a tertiary referral center between January 2006 and December 2008 who met the following criteria: (1) a cardiac abnormality was suspected on prenatal sonography, or (2) a neonatal outcome revealed congenital heart disease. Patients were included with sonograms at any gestational age and with a documented autopsy or neonatal follow-up, including patients who were referred for a possible cardiac abnormality but were healthy at delivery. Approval for this study was obtained from the Institutional Review Board at the University of California San Diego Medical Center. Because this facility is a tertiary referral center, postnatal follow-up was not available on all cases; therefore, only cases with prenatal and postnatal correlative testing performed were included. Neonatal follow-up data included immediate neonatal data obtained by neonatal examination, echocardiography, cardiac catheterization, or autopsy from the institution of delivery.

All fetal sonographic and echocardiographic examinations performed in our unit for a patient included in this study were reviewed by 3 authors (N.T., D.L., and D.H.P.) to ensure that the sonographic report corroborated with the images and reported prenatal diagnosis. All reported prenatal diagnoses seemed appropriate on the basis of the images reviewed for all cases. In addition, an attempt was made to determine why misdiagnoses were made. In general, if a major cardiac anomaly was diagnosed by level II sonography, the patient was referred for fetal echocardiography;

however, not every patient with a fetal cardiac anomaly was evaluated by fetal echocardiography, often because of advanced gestational age or aneuploidy in patients electing pregnancy termination. Our fetal sonography unit is staffed by a total of 10 radiologists and perinatologists referred to as primary physicians (primary fetal physicians). All fetal echocardiographic examinations included in this study were performed and interpreted by 2 pediatric cardiologists with greater than 17 years of experience. These examinations are typically performed at 20 to 22 weeks' gestation or at any gestational age at which a lesion is identified by any provider. All fetuses scanned by both primary physicians and pediatric cardiologists were initially scanned by primary physicians.

Fetal cardiac anomalies were defined as any major or minor cardiac anomaly associated with cardiac size, structure, axis, or effusions. The cardiac examination included in level II sonographic examinations by primary physicians included 4-chamber and right and left outflow tract views, including cine loops as well as still images; color and pulsed Doppler imaging was used when abnormalities were suspected at the discretion of the primary physicians. Fetal echocardiography was performed using gray scale, color Doppler, and pulsed Doppler techniques. Diagnoses were classified as correct, incorrect, or incorrect but within the same spectrum of disease. In the final analysis, the diagnosis was classified as correct or incorrect (cases that were misdiagnosed). The third category aforementioned was classified in the "correct" category because these patients still were classified as having a congenital heart abnormality and received the appropriate follow-up or workup, including fetal echocardiography and neonatal evaluation.

Statistical analysis was performed using means with standard deviations for normally distributed data, and the proportion of patients with the correct diagnosis when the disease was present was calculated for this cohort for primary physicians, pediatric cardiologists, and primary physicians + pediatric cardiologists. The  $\chi^2$  test was used to compare the overall proportion of correct diagnoses between primary physicians and pediatric cardiologists and the McNemar test was used to compare correct diagnosis for cases reviewed by both primary physicians and pediatric cardiologists. SPSS version 14 statistical software (SPSS Inc, Chicago, IL) was used to conduct data analysis. Statistical significance was defined as  $P < .05$ .

## Results

Sixty patients who met the above inclusion criteria were identified among 8894 patients examined by level II sonography during the study period. The mean maternal age at

sonographic evaluation was  $30 \pm 7$  years. The mean gestational age at the first sonographic examination was  $22.9 \pm 6.2$  weeks, and at the first echocardiographic examination, it was  $27.5 \pm 5.4$  weeks. The most common cardiac abnormalities identified in our study population included ventricular septal defect (VSD), atrioventricular canal defects, truncus arteriosus, tetralogy of Fallot, and coarctation of the aorta (Figure 1). Cases read by both primary physicians and pediatric cardiologists included VSD (8), atrioventricular canal defects (5), truncus arteriosus (5), pulmonic stenosis (3), coarctation of the aorta (3), tetralogy of Fallot (2), transposition of the great vessels (2), Ebstein anomaly (1), tricuspid valve atresia (1), double-outlet right ventricle (1), and others. Indications for primary physicians were as follows: suspected congenital heart disease (12), high risk for chromosomal anomalies (20), congenital anomalies other than congenital heart disease (9), low risk (5), type 2 diabetes (4), maternal history of a previous child with congenital heart disease (3), size less than dates (2), twin gestation (2), valproic acid exposure (1), and Rh isoimmunization (1). The indication for referral to a pediatric cardiologist was uniformly suspected congenital heart disease.

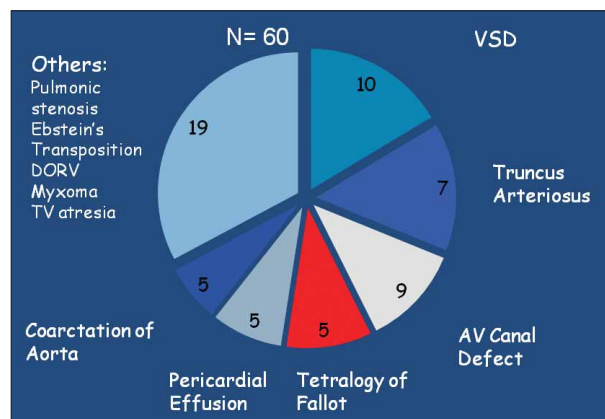
Overall, for our study population with outcome results, the combined detection rate for fetal heart abnormalities for both primary physicians and pediatric cardiologists together was 81.7% (Table 1). In this study cohort, 59 cases were read by primary physicians and 40 cases by pediatric cardiologists (because some patients underwent pregnancy termination or were scanned at a later gestational age, or some fetuses died in utero); therefore, there was some overlap. Only 39 cases were read by both primary

physicians and pediatric cardiologists; 1 patient had her prenatal sonography at an outside institution and was directly referred to a pediatric cardiologist without being scanned at our facility. Although the detection of heart disease was not statistically different between primary physicians and pediatric cardiologists, 77.9% (46 of 59) versus 85.0% (34 of 40;  $P = .3$ ), or when comparing cases reviewed by both primary physicians and pediatric cardiologists ( $P = .7$ ), perhaps this finding was related to our small study cohort and only 13 cases misdiagnosed. Primary physicians misdiagnosed 13 cases. The most common misdiagnosed cases included isolated VSD (5), pulmonic stenosis (3), and truncus arteriosus (2). Pediatric cardiologists misdiagnosed 6 cases, which included pulmonic stenosis (2), VSD (1), coarctation of the aorta (1), truncus arteriosus (1), and a normal heart that was called abnormal (Table 2). Four of the 6 cases misdiagnosed by pediatric cardiologists were complicated by other major fetal anomalies or morbid obesity.

All 60 cases were reviewed to ensure that the interpretations were accurate and correct. For the misdiagnosed cases, all images were reviewed for every sonographic examination performed during that pregnancy, and a reason for misdiagnosis was established. Most common reasons for difficult imaging or misdiagnosis included oligohydramnios or anhydramnios (3), congenital anomalies (3), cystic hygroma (2), hydrops (2), and maternal body habitus (2; Table 3). At least 1 of these factors, but more commonly more than 1 factor, were present in 9 of the 15 cases misdiagnosed; the other 6 included no identifiable reason (3), overcalls (called abnormal but found to be normal; 2), and 1 probable evolution of coarctation of the aorta.

Figures 2–5 represent images from the more common cases misdiagnosed by either primary physicians or pediatric cardiologists. Figure 2 represents 1 of the 5 cases of

**Figure 1.** Most common cardiac abnormalities identified in our study population. AV indicates atrioventricular; DORV, double-outlet right ventricle; TV atresia, tricuspid valve atresia; and VSD, ventricular septal defect.



**Table 1.** Accuracy of Diagnosis of Fetal Heart Abnormalities

Providers	Cases Correct, n (%) <sup>a</sup>	Cases Missed, n (%)
Primary physicians	46/59 (77.9)	13/59 (22)
Pediatric cardiologists	34/40 (85)	6/40 (15)
Primary physicians + cardiologists <sup>b</sup>	49/60 (81.7)	11/60 (18.3)

<sup>a</sup> $P = .3$  for comparison of correct diagnoses for primary physicians versus cardiologists.

<sup>b</sup>Total number of cases correct or missed by both primary physicians and pediatric cardiologists. Some cases were correctly identified by the other provider; therefore, these cases were not counted as missed cases because they received the proper neonatal medical care if either the primary physician or cardiologist diagnosed a cardiac abnormality prenatally.

**Table 2.** Misdiagnosed Cases

Cardiac Lesion	Misdiagnosed by Primary Physician, n	Misdiagnosed by Pediatric Cardiologist, n	Total, n
Pulmonic stenosis	3	2 <sup>a</sup>	2
Isolated VSD	5 <sup>b</sup>	1 <sup>a</sup>	4
Myxoma	1	0	1
Coarctation of aorta	1	1 <sup>c</sup>	1
Truncus arteriosus	2	1 <sup>a</sup>	2
Multiple echogenic tumors	0	1 <sup>d</sup>	1
Reported as thickened RV and pericardial effusion	1 <sup>e</sup>	0	0
Total	13	6	11

RV indicates right ventricle; and VSD, ventricular septal defect.

<sup>a</sup>Represents the same case missed by both the primary physician and cardiologist.

<sup>b</sup>One case was misdiagnosed by the primary physician but correctly identified by the cardiologist.

<sup>c</sup>This case was misdiagnosed by the cardiologist but correctly identified by the primary physician.

<sup>d</sup>This case was read only by the cardiologist and diagnosed as abnormal. Autopsy revealed a normal heart.

<sup>e</sup>This case was read by the primary physician as abnormal but was read as normal by the cardiologist. The neonate had normal postnatal echocardiographic findings.

**Table 3.** Reasons for Misdiagnosis

Case	Missed Diagnosis	Who Imaged	Who Misdiagnosed	GA at 1st Sonogram	GA at 1st Fetal Echo	Reason for Misdiagnosis
1	Pulmonic stenosis	Primary physician, cardiologist	Primary physician, cardiologist	18	23	Morbidly obese, congenital diaphragmatic hernia, possibly lesion evolved over time
2	Pulmonic stenosis	Primary physician, cardiologist	Primary physician, cardiologist	17	19	Cystic hygroma
3	Pulmonic stenosis	Primary physician, cardiologist	Primary physician missed, cardiologist correct	29	33	Cystic hygroma, hydrops
4	VSD	Primary physician, cardiologist	Primary physician missed, cardiologist correct	20	29	Color Doppler imaging not used
5	VSD	Primary physician	Primary physician	22	37	Anhydramnios
6	VSD	Primary physician, cardiologist	Primary physician, cardiologist	21		Unilateral multicystic dysplastic kidney
7	VSD	Primary physician	Primary physician	20		Anhydramnios
8	VSD	Primary physician	Primary physician	31		Color Doppler imaging not used
9	Myxoma	Primary physician	Primary physician	20		Dichorionic twins, lesion in twin with multiple anomalies
10	Called abnormal but normal	Primary physician, cardiologist	Primary physician	18	26	Unknown
11	Coarctation of aorta	Primary physician	Primary physician	17		Possibly evolved; normal heart views on 4 examinations until 33 wk GA
12	Coarctation of aorta	Primary physician, cardiologist	Cardiologist missed, primary physician correct	19	19	Pulmonary artery size > aorta on 2 primary physician examinations; cardiologist thought within normal limits
13	Truncus arteriosus	Primary physician, cardiologist	Primary physician, cardiologist	17	24	Morbidly obese
14	Truncus arteriosus	Primary physician	Primary physician	20		Anhydramnios, hydrops
15	Called abnormal but normal	Cardiologist	Cardiologist	Referred to cardiologist only	20, 21	2 cardiologists read as multiple intraventricular echogenic tumors; autopsy showed no tumors, but calcifications present

GA indicates gestational age; and VSD, ventricular septal defect.

isolated VSDs misdiagnosed. The 4-chamber images from the sonograms read by primary physicians appeared normal, but the 4-chamber views from the fetal echocardiograms from the same patient showed the defect. Figure 3 shows a normal-appearing right ventricular outflow tract view read by a primary physician; the pulmonic stenosis

**Figure 2.** Isolated ventricular septal defect missed by a primary physician at 20 weeks' gestation and seen by a pediatric cardiologist at 29 weeks. **A,** The 4-chamber view from the sonogram read by the primary physician appeared normal. **B,** The 4-chamber views from the fetal echocardiogram showed a 3-mm midmuscular ventricular septal defect.



defect was identified by a pediatric cardiologist on fetal echocardiography using color Doppler flow. Figure 4 represents an enlarged pulmonary artery on a right ventricular outflow tract and 3-vessel view that was identified by a primary physician but appeared normal on fetal echocardiography read by a pediatric cardiologist. Figure 5 shows an abnormal outflow tract view in one of our fetuses with truncus arteriosus. The primary physician recognized the abnormality but diagnosed the lesion as tetralogy of Fallot. This difference in diagnosis was categorized as “correct” because it fell into the same spectrum of disease.

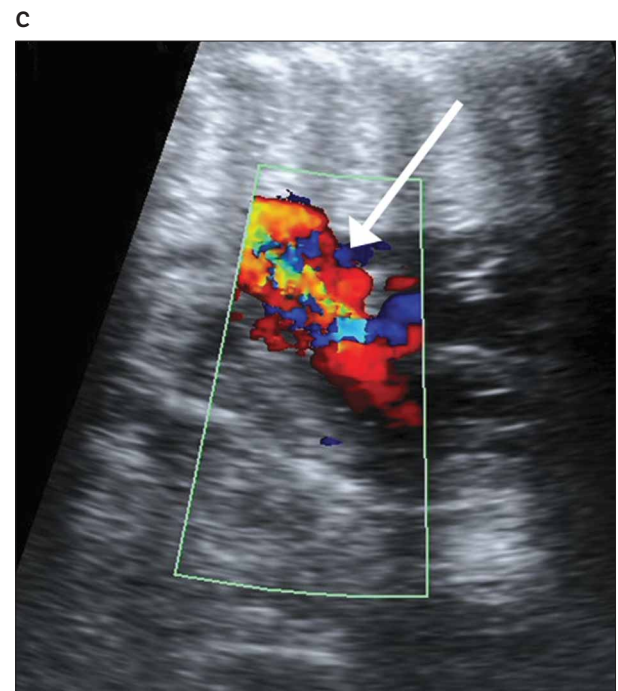
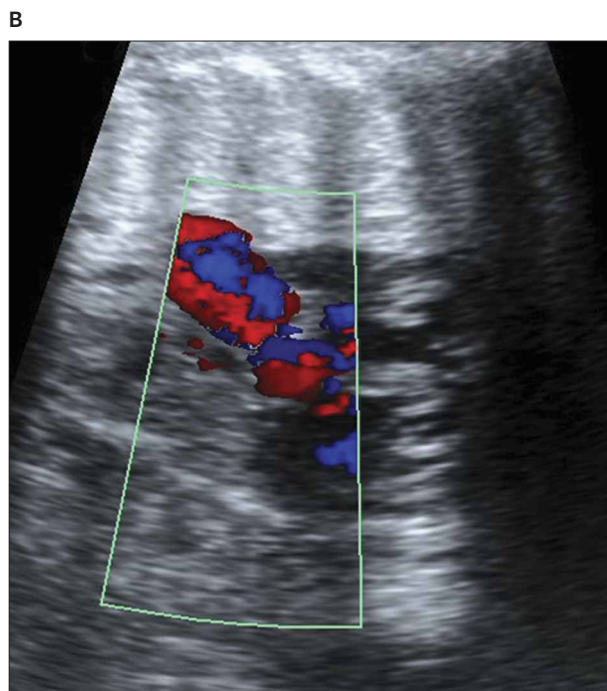
## Discussion

The importance of the role of fetal sonographers and imaging interpreters such as radiologists, perinatologists, and pediatric cardiologists is becoming more evident in the detection of congenital heart disease. Unfortunately, over the last 20 years, prenatal detection of congenital heart disease has not markedly improved despite better equipment, improved imaging techniques, and intense training for fetal sonographers. More recently, authors have suggested that detection rates for congenital heart disease remain low. Acherman et al<sup>17</sup> studied a population of 161 patients among 77,000 live births with congenital heart disease and determined a detection rate for fetal heart disease of 36% when evaluating both community screening and fetal echocardiography in Nevada.<sup>17</sup> McBrien et al<sup>18</sup> described a 27% antenatal congenital heart disease detection rate in northern Ireland in 272 patients with major congenital heart disease, and among those detected antenatally, there was a 26% neonatal mortality rate. Friedberg et al<sup>9</sup> reviewed data for 309 children who needed surgical or catheter intervention for congenital heart disease and determined that 36% had prenatal diagnosis. More recently, Sklansky et al<sup>19</sup> reported an overall 33% prenatal detection rate of congenital heart lesions in 65 infants among 200 prospectively undergoing cardiac surgery for major congenital heart disease in California. At tertiary care centers, the detection rates are higher, as described by Friedberg et al<sup>9</sup> (80%), Carvalho et al<sup>±</sup> (75%), and Wong et al<sup>20</sup> (61%), which is consistent with our overall detection rate of congenital heart disease of 81.7%. Although the detection of major and minor congenital heart disease was not statistically different between primary physicians and pediatric cardiologists in our study, existing data show that fetal echocardiography is superior in detecting congenital heart disease, for which the sensitivity and specificity of current 2-dimensional Doppler fetal echocardiography approach 80% to 100%.<sup>17,21</sup> The limitations of our study include a

small number of anomalies and misdiagnosed cases and incomplete follow-up data for some cases with detected fetal cardiac anomalies during our study period because we are a referral center.

Many authors have tried to identify ways to improve these detection rates for both community and tertiary care centers. Guidelines adopted by the American Institute of

Ultrasound in Medicine, American College of Obstetricians and Gynecologists, and American College of Radiology recommend obtaining the 4-chamber view while attempting the outflow tract views as part of the fetal cardiac screening examination.<sup>22</sup> Authors have studied whether adding different views, such as the right and left ventricular outflow tract views, 3-vessel view, and ductal and aortic arch views,



**Figure 3.** Pulmonic stenosis called normal by a primary physician at 29 weeks' gestation and seen by a pediatric cardiologist at 33 weeks, when it was identified. **A**, Normal-appearing right ventricular outflow tract (RVOT) read by the primary physician. **B**, Pulmonic stenosis was identified by the pediatric cardiologist on the fetal echocardiogram using color Doppler flow. The image shows narrowing of the color stream at the pulmonary valve. **C**, Second image from the cine video showing aliasing across the pulmonary valve region of stenosis. Arrow indicates the pulmonary valve region.

to the routine prenatal fetal heart survey improves detection of congenital heart disease. From their study, Sklansky et al<sup>19</sup> surmised that the potential sensitivity for detecting

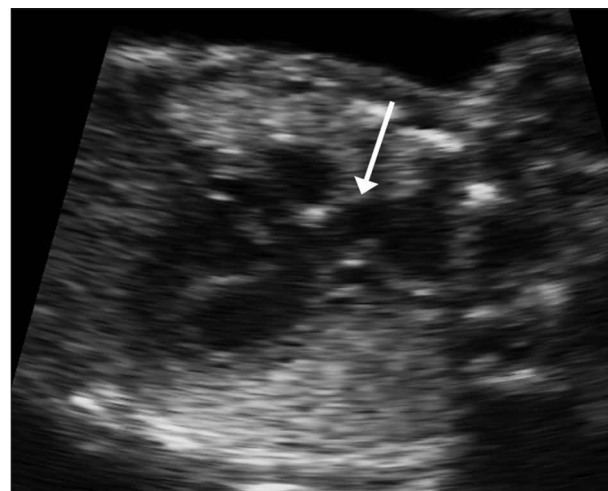
**Figure 4.** Coarctation of the aorta (AO) called abnormal by a primary physician at 18 weeks' gestation and seen by a pediatric cardiologist at 19 weeks, when it was called normal. **A**, Enlarged pulmonary artery (PA) on the right ventricular outflow tract view. **B**, Three-vessel view.



these cases was higher with outflow tract views when compared to the 4-chamber view (91% versus 63%;  $P < .001$ ). The low sensitivity of the 4-chamber views is consistent with previous reports suggesting that outflow tract views improve the overall accuracy of detecting congenital heart disease.<sup>8,10,23</sup> Wu et al<sup>24</sup> prospectively studied 8025 fetuses and determined that the sensitivity of the combination of the 4-chamber and 3-vessel views was 81.3%, and the specificity was 99.9%, which was higher than the sensitivity of the 4-chamber view alone (65.6%).

Wong et al<sup>20</sup> showed that independent variables affecting the detection rate of congenital heart disease included the complexity of the cardiac lesion, experience of the operator, and detection of chromosomal anomalies. Other factors influencing the accuracy of obtaining optimal heart views include gestational age (increasing age increases the ability to obtain these views), maternal adipose tissue thickness ( $>3$  cm), and previous lower abdominal surgery.<sup>25</sup> Consistent with these data, in reviewing the misdiagnosed cases (Table 3), we believe that increasing gestational age and maternal body habitus affect the ability to accurately diagnose congenital heart disease. In addition, we found that other factors affecting the accuracy of detecting congenital heart disease include the amount of amniotic fluid and abnormal fetal findings such as cystic hygromas, hydrops fetalis, and other coexisting congenital anomalies. At least 1 of these factors was present in 9 of the total 15 misdiagnosed cases of congenital heart disease in

**Figure 5.** Truncus arteriosus diagnosed at birth but diagnosed as tetralogy of Fallot by a primary physician at 22 weeks' gestation and a pediatric cardiologist at 23 weeks on fetal echocardiography. The abnormal outflow tract view shows the great vessel overriding the ventricular septum (arrow). This diagnosis was categorized as "correct" because it fell into the same spectrum of disease.



our cohort. Interestingly, 2 cases were called abnormal when postnatal evaluation or autopsy confirmed normal hearts. In 1 case in which right ventricular hypertrophy was overcalled, perhaps nomograms of ventricular size should have been consulted.<sup>26</sup> In the second case, 2 experienced fetal cardiologists (>17 years of experience) thought that multiple intraventricular echogenic masses represented myxomas and possibly tuberous sclerosis, which resulted in pregnancy termination. Perhaps as providers, we must accept the fact that even despite our best attempts at diagnosing congenital heart disease prenatally, we will overdiagnose and underdiagnose some lesions depending on other influencing factors involved in the case.

The importance of diagnosing congenital heart disease antenatally cannot be understated. Treatment of patients with severe congenital heart disease ideally begins before birth. Early and accurate prenatal diagnosis allows for the education and psychological preparation of the family (with counseling) and medical preparation for optimizing the health of the mother and infant after birth. In a study by Verheijen et al,<sup>5</sup> prenatal diagnosis of congenital heart disease minimized acidosis in patients with the disease and may be associated with an improved long-term outcome and prevention of cerebral damage. Tworetzky et al<sup>13</sup> showed more specifically that the prenatal diagnosis of hypoplastic left heart syndrome was associated with an improved preoperative clinical status and improved survival after the first stage of palliation in comparison with patients with a diagnosis after birth. This study was comparable to the study by Mahle et al,<sup>27</sup> which showed that prenatal diagnosis of hypoplastic left heart syndrome had a favorable impact on the treatment of patients and reduced early neurologic morbidity. They suggested that long-term neurologic outcomes could be improved by prenatal detection.

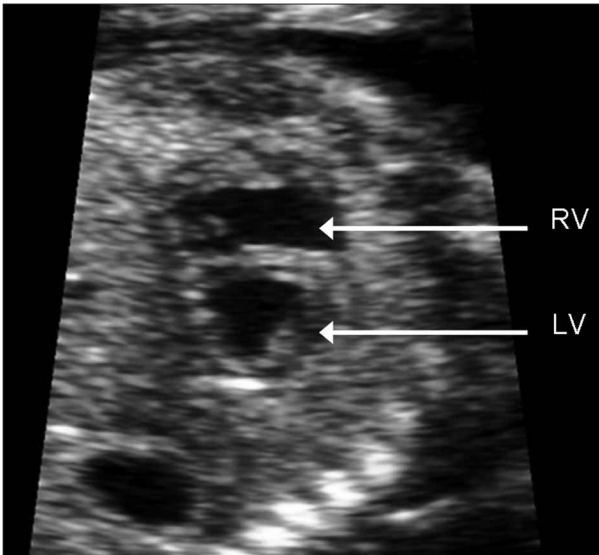
Congenital heart disease may progress and evolve during gestation as well as infancy. Although cardiac screening is typically done before 22 weeks, we suggest that the fetal heart should be evaluated on all follow-up sonographic examinations. Outflow tract obstructions such as pulmonic or aortic stenosis can progress slowly and be detectable only in the third trimester or at birth. These lesions typically are associated with a normal ventricular growth pattern because there has been a longer period of normal ventricular filling.<sup>28</sup> Anand and Mehta<sup>29</sup> reported that pulmonary stenosis can worsen in infancy, going from asymptomatic mild stenosis to major stenosis requiring intervention. Limitations in prenatal detection of pulmonic stenosis are the possibility of the late appearance of typical sonographic features, including right ventricular hypertrophy, a de-

crease in the right ventricular size, and poststenotic dilatation of the pulmonary artery.<sup>30</sup> Similarly, coarctation of the aorta may be associated with progression, and the type of coarctation identified prenatally is usually associated with preductal obstruction.<sup>31</sup> In the absence of a nonprogressive type of aortic arch hypoplasia, the ventricular size may or may not be discrepant. Therefore, these lesions may not be identified until the ductus arteriosus constricts.<sup>32</sup> In addition, Acharya et al<sup>33</sup> described using color Doppler echocardiography to better characterize congenital heart disease in the second and third trimesters because color disturbances may assist in detecting flow abnormalities. These evolving lesions may potentially be detected by using color flow Doppler imaging, assessing the cardiac chamber size and contractility throughout gestation.<sup>34,35,36</sup>

Technical aspects in obtaining fetal heart views play a major role in detecting congenital heart disease. Five isolated VSDs were misdiagnosed in our series. A VSD can be detected on sonography by using a combination of 2-dimensional and color flow Doppler imaging techniques.<sup>37</sup> First, a fetal echocardiographic preset may be helpful for assessment of the ventricular septum. Second, the ideal scan plane for evaluation of the septum is a short-axis view, which is obtained by rotating the transducer 90° from the 4-chamber view and angling the transducer along the long axis of the ventricles (Figure 6); this process eliminates the artifactual dropout frequently noted in the high membranous ventricular septum when it is orientated parallel to the ultrasound beam (Figure 7). Third, narrowing the field of view and decreasing the imaging depth will improve temporal and spatial resolution. Fourth, increasing the scanning frequency and magnifying the heart improves the overall image resolution. Fifth, areas of consistent dropout in the septum must be confirmed over multiple cardiac cycles. Sixth, a true VSD often has a bright specular reflection on either side of the defect. Seventh, small VSDs may require color Doppler imaging for detection. Specific features of color Doppler evaluation of the septum include the following: (1) use the short-axis view; (2) use a low-velocity setting ( $\approx 30$  cm/s) because pressures between the right and left ventricles are usually similar; (3) apply a small color box to improve temporal resolution and color sensitivity; and (4) angle the transducer back and forth along the long axis of the ventricles, and look for consistent color shunting between the ventricles; the direction of the flow is most frequently noted to be left to right but can also be bidirectional.

Pulmonic stenosis was missed in 3 cases, all of which had other congenital anomalies (Table 3). Pulmonic stenosis can best be detected on sonography by observing the morphologic characteristics of the valve and the size of

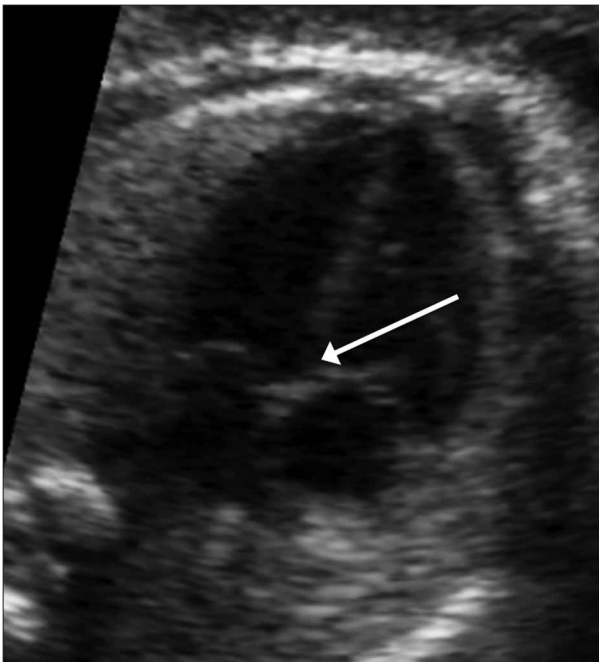




**Figure 6.** Short-axis view of the right ventricle (RV) and the left ventricle (LV) of a normal fetal heart at 19 weeks' gestation.

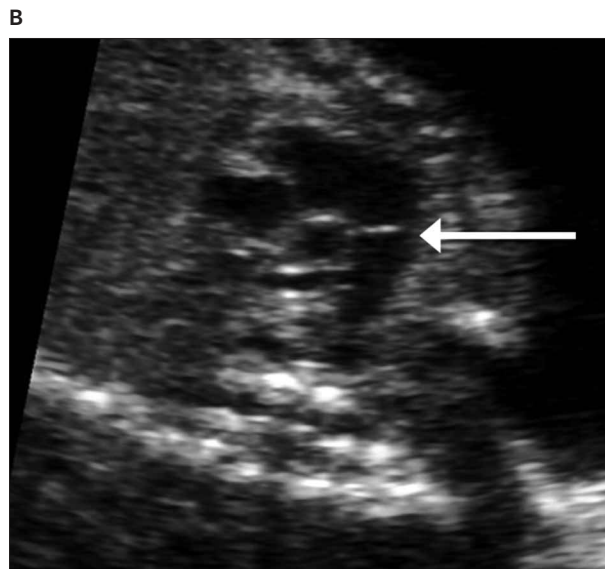
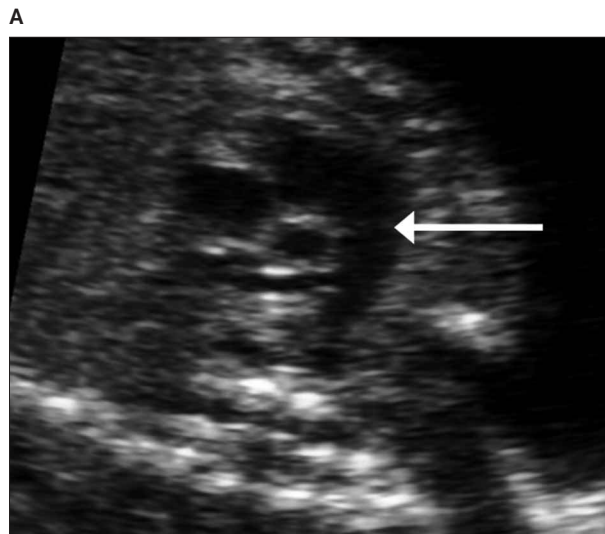
the valve annulus and also by interrogating the flow across the valve using pulsed wave Doppler, continuous wave Doppler, and color flow Doppler techniques.<sup>38</sup> Nadel<sup>38</sup> recently showed that color Doppler improves the detec-

**Figure 7.** Artifactual dropout of the ventricular septum simulating a ventricular septal defect. This appearance occurs when the septum is oriented parallel to the ultrasound beam (arrow).



tion of pulmonic stenosis, as seen in our study cohort. First, the pulmonary valve and main pulmonary artery are best evaluated in the right ventricular outflow tract view. The ideal plane to obtain this view is in a projection in which the main pulmonary artery is parallel to the ultrasound beam (Figure 8). The valve leaflets should be thin and move freely. Second, poststenotic dilatation of the main pulmonary artery may be seen. Third, pulsed and continuous wave Doppler imaging can be used to assess the velocity of the blood flow through the pulmonary valve.

**Figure 8.** Normal appearance of the pulmonary valve at 21 weeks' gestation. **A.** Main pulmonary artery during systole showing an open pulmonary valve (arrow). **B.** Main pulmonary artery during diastole showing a closed pulmonary valve (arrow).



Fourth, using color Doppler imaging, the scale should be set to a high-velocity setting ( $\approx 80\text{--}90$  cm/s), and color flow should be laminar across the valve. Even in moderate to severe pulmonary valve stenosis, the Doppler velocity across the valve may not be high (because of equalization of pressures between the aorta and the patent ductus arteriosus). Pulmonary valve stenosis should be considered when a velocity of greater than 150 to 200 cm/s is identified.<sup>39</sup>

Data from first-trimester sonographic evaluation of fetuses suggest that congenital heart disease can be identified in some cases early in gestation.<sup>40,41</sup> Identification at this stage often requires the use of color Doppler techniques. In addition, nuchal translucency screening in the first trimester may identify fetuses at higher risk for congenital heart disease, detecting approximately 23% to 31% of fetuses with congenital heart disease when the nuchal translucency measures above the 99th percentile.<sup>41–43</sup>

In conclusion, this study was done to show the success and reality of misdiagnoses not only by primary physicians but also by pediatric cardiologists. We cannot expect to find all anomalies because of the limitations of our patient population (anhydramnios, obesity, etc), and this circumstance is a clinical reality. We can, however, strive to improve, as all medical providers learn the limitations of our techniques. Primary providers in fetal imaging centers will continue to rely on the expertise of fetal echocardiographers to assist in congenital heart disease diagnosis, even though the difference in congenital heart disease diagnosis was not significantly different between primary physicians (78%) and pediatric cardiologists (85%) in our cohort. Our study shows that this finding is related to multiple factors, including evolution of congenital heart disease, maternal body habitus, associated congenital anomalies, decreased amniotic fluid volume, gestational age at evaluation, imaging techniques, and, most importantly, the experience of the sonographer. To improve the detection of congenital heart disease and ultimately improve neonatal outcomes, we suggest the following: (1) additional views should be added to the cardiac screening examination, eg, 3-vessel view and arch views; (2) color Doppler and/or pulsed Doppler imaging should be used for evaluation of the septum, valvular flow, and ventricular size; (3) screening cardiac examinations should be attempted on all initial and follow-up sonographic examinations with cine loops as well as still images; and (4) continuous educational training in fetal heart evaluation must be emphasized. Despite all our efforts, we will continue to misdiagnose some cases of congenital heart disease because of many varied factors that affect optimal fetal heart assessment.

## References

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