

## ABNORMAL CARDIAC FINDINGS IN PRENATAL SONOGRAPHIC EXAMINATION: AN IMPORTANT INDICATION FOR FETAL ECHOCARDIOGRAPHY?

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**Aim:** The present study was conducted to evaluate the most common indications for fetal echocardiography in a tertiary-care fetal echocardiography practice.

**Methods:** A retrospective analysis (July 2005 to July 2006) of all pregnant women referred by obstetricians or fetomaternal sonographers to a pediatric cardiology unit for fetal echocardiography over a 1-year period was performed. The study was performed at a large university hospital in the western region of Saudi Arabia. The primary indications for referral for fetal echocardiography were obtained from the obstetric referral forms. Outcome data were extracted from the fetal echocardiograms, postnatal echocardiograms and/or patient medical records.

**Results:** Sixty pregnant women had been referred for fetal echocardiography. The most common primary indication for referral had been abnormal cardiac findings on the prenatal sonographic examination, which had been present in 21 (35%) of the 60 cases. In 19 (90%) of those 21 cases, congenital heart disease (CHD) had been confirmed prenatally and postnatally. Among the 39 patients who had had normal cardiac findings on prenatal sonography but had been referred for fetal echocardiography owing to other primary indications, CHD had been confirmed in only 8 cases (20%). All 19 of 21 patients in whom fetal echocardiography had shown CHD had completed their pregnancies.

**Conclusions.** Our study shows that a common primary indication for fetal echocardiography during prenatal sonographic examination is an abnormal cardiac finding, which is more useful for identifying congenital heart disease than are other risk factors. Detailed routine fetal echocardiography during routine prenatal ultrasound may result in further investigation and treatment.

**Keywords:** prenatal ultrasonography; fetal echocardiography; congenital heart disease

FETAL ECHOCARDIOGRAPHY HAS BEEN ACCEPTED as a prenatal noninvasive diagnostic tool for cardiovascular diseases in fetuses for more than three decades. Cardiac abnormalities are frequently not detected by routine ultrasound screening examinations. Although detailed fetal echocardiography is more sensitive in detection of congenital heart disease, it is used only for high-risk cases. The primary indications for fetal echocardiography, according to the American College of Cardiology,<sup>1</sup> are fetal heart abnormalities or fetal arrhythmia detected by routine prenatal sonography, a family history of congenital heart

disease, maternal diabetes or systemic lupus erythematosus, fetal exposure to a teratogen, fetal karyotype abnormality, and other fetal system abnormalities. Despite the recognition of these risk factors, only 15–30% of cardiac defects are detected before birth.<sup>2,3</sup> This low detection rate may reflect the inability of traditional risk factors to identify most of the patients at risk, as well as the limitations of sonography to detect cardiac anomalies prenatally. To our knowledge there are limited studies to evaluate the accuracy of referral for fetal echocardiography while referring to specific risk factors, especially for cases in which the cardiac findings on prenatal sonography are normal.

There are limited data about the common indications for fetal echocardiography in Saudi Arabia. The purpose of this retrospective study was to review the primary indications for fetal echocardiography among pregnant women who underwent prenatal sonographic examination over a 1-year period at a tertiary-care fetal

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echocardiography practice and try to determine the ability of each primary indication to identify congenital heart disease.

### Patients and Methods

For this retrospective analysis, the medical records of all pregnant patients who had, during a 1-year study period (July 2005 to July 2006), undergone a routine complete prenatal sonographic examination during the second or third trimester at a prenatal diagnostic center at a large university hospital in the western region of Saudi Arabia were reviewed.

At the time of routine prenatal sonography, performed by the referring obstetrician or fetomaternal specialist, patients had been screened for risk factors for congenital heart disease. As a standard practice during the study period, pregnant patients in whom the fetal heart had appeared abnormal on sonography or in whom other maternal, fetal, genetic, or familial risk factors for congenital heart disease had been identified, despite normal-appearing cardiac sonograms, were referred to the pediatric cardiology unit for fetal echocardiography. The cardiac sonograms were performed by a single pediatric cardiologist (RB) with experience in perinatal cardiology. After the initial fetal echocardiogram, most patients were rescanned at 4-6 weekly intervals. However, if there was a risk of fetal hydrops or fetal demise, patients were rescanned more frequently mostly at a 2 weekly intervals.

As a standard practice during the study period, pregnant patients in whom the fetal heart had appeared abnormal on sonography or in whom other maternal, fetal, genetic, or familial risk factors for congenital heart disease had been identified despite normal-appearing cardiac sonograms were referred to the pediatric cardiology unit for fetal echocardiography. This examination had been performed using a HP Sonos 7500 machine (Philips Medical Systems,) with a 4-6- MHz curved-array transducer). For this analysis, the author reviewed all prenatal data for all patients referred to the pediatric cardiology unit during the study period. For all patients who had been referred for fetal echocardiography, follow-up imaging and other records were available. The primary reason for the referral for fetal echocardiography was recorded. A fetal cardiac defect was considered to have been

detected prenatally if it had been diagnosed before delivery. The ability of each primary indication for fetal echocardiography to predict congenital heart disease was calculated on the basis of the number of cases confirmed by postnatal echocardiography. Post-mortum pathologic evaluation was not possible for religious considerations.

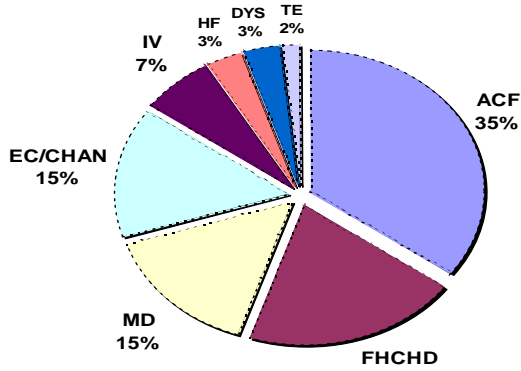
In this report, the calculated values are expressed as percentages, and the gestational ages of the fetuses at the time of diagnosis are reported as means  $\pm$  standard deviations. All newborns had postnatal echocardiograms in the first week of life.

### Results

During the study period, the prenatal diagnostic center performed 1001 complete prenatal sonographic examinations in 801 consecutive patients in the second or third trimester of pregnancy. Of these 801 patients, 60 (7.4%) had been referred for fetal echocardiography. The mean maternal age and the mean gestational age at the time of the initial prenatal echocardiographic examination were  $26 \pm 3.1$  years and  $29. \pm 7.0$  weeks, respectively. All the 60 patients who were referred for fetal echocardiography had completed their pregnancies; 56 babies were delivered at term and 4 babies were born prematurely. There were 2 (3.3%) neonatal deaths. The 2 neonates had hypoplastic left heart syndrome and no surgical option was possible at the time of delivery. There was no data to evaluate the CHD with highest postnatal survival due to incomplete follow-up of all patients who had undergone prenatal sonography. The primary indications for fetal echocardiography are shown in Figure 1.

Abnormal cardiac findings on prenatal sonography accounted for the highest proportion of the most common primary indication for referral cases of congenital heart disease (35% of patients). The next most common indication for referral was a positive family history of congenital heart disease in 12 (20%) or conditions that had arisen during gestation, such as maternal diabetes in 9 (15%); extracardiac fetal anomalies or chromosomal anomalies in 9 (15%), inadequate visualization of the fetal heart in 4 (6.6%), hydrops fetalis in 2 (3.3%), dysrhythmia in 2 (3.3%) and teratogen exposure in 1 (1.7 %). Of the 9 fetuses with chromosomal anomalies or extracrdiac anomalies there were 3 fetuses with trisomy 21, one with





Abbreviation: ACF=Abnormal cardiac findings on screening sonography, FHCHD=Family history of CHD, MD=Maternal diabetes, EC/CHAN=Extracardiac fetal anomalies/Chromosomal abnormality, IV=inadequate visualization, HF=hydrops fetalis, DYS=Dysrhythmia, TE=Teratogen exposure

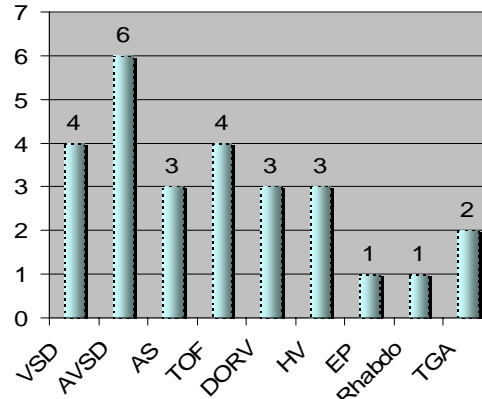
**Figure 1.** Indications for referral for fetal echocardiography (n=60)

trisomy 13, 2 had renal anomalies, and 3 had central nervous system anomalies.

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The cardiac malformations diagnosed prenatally are shown in Figure 2. The most common congenital heart disease diagnosed prenatally was defect of the atrioventricular canal (6 patients). The presence of defect of the atrioventricular canal in 6 of 28 (21%) patients could possibly be explained by the presence of 4 fetuses with chromosomal anomalies, namely trisomy 21 (3 patients) and one patient with trisomy 13.

In the 39 (65%) patients who had been referred for other primary indications and had normal cardiac findings on screening prenatal sonography,



VSD=ventricular septal defect, AVSD=atrioventricular septal defect, AS= aortic stenosis, TOF=tetralogy of Fallot, DORV=double outlet right ventricle, HV=hypoplastic ventricle, EP=Ebstein anomaly, Rhabdo=rhabdomyoma, TGA=transposition of great arteries

**Figure 2.** Cardiac malformations diagnosed prenatally (n=27)

in 8 (20.5%) of these fetuses, congenital heart disease had been suspected and confirmed postnatally on the basis of the findings of fetal echocardiography. In one fetus CHD had been diagnosed postnatally (small muscular ventricular septal defect) after being overlooked on both prenatal sonography and fetal echocardiography. In 21 (35%) of the 60 patients referred for abnormal cardiac findings on prenatal sonography, congenital heart disease was diagnosed prenatally and confirmed postnatally in 19 patients (90%). Thus in fetuses with normal cardiac findings on prenatal sonography but referred for other indications, the incidence of congenital heart disease was lower (23%) than the incidence of CHD in fetuses with abnormal cardiac findings on prenatal echocardiography (90%).

In total, cardiac malformations had been confirmed by postnatal echocardiography in 28 (46%) of the 60 infants delivered after completed pregnancies.

### Discussion

Cardiac abnormalities are frequently not detected by routine ultrasound screening examinations.



Although detailed fetal echocardiography is more sensitive in detection of congenital heart disease, it is used only for high-risk cases. An abnormal cardiac finding on routine prenatal sonography has become a common reason for referring patients for fetal echocardiography.<sup>4, 5</sup> Traditional risk factors for congenital heart disease, such as maternal diabetes, a family history of cardiac malformations, and teratogen exposure, identify relatively few cases of congenital heart disease prenatally.<sup>2,6</sup> In a retrospective analysis by Cooper and colleagues,<sup>2</sup> 4% of fetal echocardiographic examinations had been performed because a cardiac defect had been suspected on the basis of screening prenatal sonography, whose rate of detection of congenital heart disease was 68%. In other studies, 4–5% of echocardiographic examinations were performed because prenatal sonographic findings had suggested a cardiac abnormality, with congenital heart disease confirmed in 38–50% of these cases.<sup>7, 8,9</sup> Reports on prenatal sonography cite a wide range of rates of detection of congenital heart disease, from less than 10% to greater than 90%.<sup>3,8,10,11,12.</sup>

In the present study, 35% of echocardiographic examinations had been performed because of abnormal cardiac findings on routine prenatal sonography, whose detection rate was 90%. Although this rate of detection of cardiac abnormalities is higher than the rates reported in the previously published studies, the high rate of detection might reflect improvements in sonographic imaging since the earlier studies or more thorough screening techniques. Also, all scans were performed by a single operator with relative experience in the field of fetal echocardiography. Although the results of the present study might not be applicable to the general population, they demonstrate the importance of visualizing the fetal heart during routine prenatal sonographic examination. Reported results may also depend on whether they are obtained at a community or a tertiary-level center, whether the study is retrospective or prospective, whether the patients are at high or low risk, and the nature and duration of follow-up.<sup>13</sup> Although screening patients for traditional risk factors for congenital heart disease remains warranted, it has been reported that most cardiac defects detected prenatally occur in cases of low risk pregnancy.<sup>14</sup>

The current study is limited by the small number of referrals for fetal echocardiography, incomplete

follow-up of all patients who had undergone prenatal sonography and by the fact that only short-term follow-up data on those infants who had had normal fetal echocardiographic findings were available. Although a small ventricular septal defect had been detected postnatally in 1 infant who had had normal findings on fetal echocardiography, other minor lesions likely would have been identified if follow-up data beyond the perinatal period had been available. However, minor abnormalities such as atrial and ventricular septal defects remain difficult to diagnose prenatally and are often of little clinical significance. In the present study, congenital heart disease had been confirmed in 90% of cases with abnormal cardiac findings on prenatal sonography, compared with 23% for all other risk factors combined. However, because only the primary indication for fetal echocardiography was recorded in each case, any additional risk factors for congenital heart disease that had been present in patients with abnormal cardiac imaging may have been overlooked.

In cases of an affected first-degree relative, the ability of fetal echocardiography to show congenital heart disease might have been greater than the present results suggest. In a study of the effects of various risk factors to identify congenital heart disease, Buskens and coworkers<sup>15</sup> found that the relative risk for congenital heart disease varied depending on the specific nature of the family history of this disorder.

Despite the limitations of the present study, knowing the overall value of fetal echocardiography for various indications is clinically important in the setting of normal cardiac results on prenatal sonograms obtained in a tertiary-care center.

Universal screening for congenital heart disease at the time of routine prenatal sonographic examination might be the most effective approach to improving the rate of prenatal detection of serious cardiac anomalies. The American College of Obstetricians and Gynecologists<sup>16</sup> and the American Institute of Ultrasound in Medicine<sup>17</sup> recommend that the 4-chamber view be evaluated during routine prenatal sonographic examinations performed in the second and third trimesters. It is estimated that with appropriate training and experience, sonographers evaluating the 4-chamber view of the fetal heart can identify 40–50% of cases of congenital heart disease and that adding views of the ventricular outflow tracts can increase the detection rate to 60–80%.<sup>18.</sup>



To date there is no universal protocol for echocardiographic screening for evidence of congenital heart disease in the midtrimester. The timing of referral in the present study reflects this, where the mean gestational age was 29 weeks.

We believe that selective screening for fetal echocardiography remains the standard method of prenatal diagnosis. Adoption of universal screening of the fetal heart during routine prenatal sonographic examination has the greatest potential to improve the rate of prenatal detection of major cardiac defects.

### References

- Cheitlin MD, Armstrong WF, Aurigemma GP, et al. ACC/AHA/ASE 2003 guideline update for the clinical application of echocardiography—summary article: a report of the American College of Cardiology/American Heart Association Task Force on Practice Guidelines(ACC/AHA/ASE Committee to Update the 1997 Guidelines for the Clinical Application of Echocardiography). *J Am Coll Cardiol* 2003;42:954.
- Cooper MJ, Enderlein MA, Dyson DC, et al. Fetal echocardiography: retrospective review of clinical experience and an evaluation of indications. *Obstet Gynecol* 1995;86:577.
- Grandjean H, Larroque D, Levi S. The performance of routine ultrasonographic screening of pregnancies in the Eurofetus Study. *Am J Obstet Gynecol* 1999;181:446.
- Allan LD, Sharland GK, Milburn A, et al. Prospective diagnosis of 1,006 consecutive cases of congenital heart disease in the fetus. *J Am Coll Cardiol* 1994; 23:1452.
- Davis GK, Farquhar CM, Allan LD, et al. Structural cardiac abnormalities in the fetus: reliability of prenatal diagnosis and outcome. *Br J Obstet Gynaecol* 1990;97:27.
- Roodpeyma S, Kamali Z, Afshar F, et al. Risk factors in congenital heart disease. *Clin Pediatr (Phila)* 2002;41:653.
- Wheller JJ, Reiss R, Allen HD. Clinical experience with fetal echocardiography. *Am J Dis Child* 1990; 144:49.
- Copel JA, Pilu G, Green J, et al. Fetal echocardiographic screening for congenital heart disease: the importance of the four-chamber view. *Am J Obstet*
- Smythe JF, Copel JA, Kleinman CS. Outcome of prenatally detected cardiac malformations. *Am J Cardiol* 1992;69:1471.
- Ferencz C, Rubin JD, McCarter RJ, et al. Congenital heart disease: prevalence at livebirth. The Baltimore-Washington Infant Study. *Am J Epidemiol* 1985; 121:31.
- Buskens E, Grobbee DE, Frohn-Mulder IME, et al. Efficacy of routine fetal ultrasound screening for congenital heart disease in normal pregnancy. *Circulation* 1996;94:67.
- Skeels M, Taylor D, Park J, et al. Test characteristics of a level I or II fetal ultrasound in detecting structural heart disease. *Pediatr Cardiol* 2002;23:594.
- Wong SF, Chan FY, Cincotta RB, et al. Factors influencing the prenatal detection of structural congenital heart diseases. *Ultrasound Obstet Gynecol* 2003;21:19.
- Achiron R, Glaser J, Gelernter I, et al. Extended fetal echocardiographic examination for detecting cardiac malformations in low-risk pregnancies. *BMJ* 1992;304:671.
- Buskens E, Stewart PA, Hess J, et al. Efficacy of fetal echocardiography and yield by risk category. *Obstet Gynecol* 1996;87:423.
- American College of Obstetricians and Gynecologists. *Ultrasonography in pregnancy* (Technical Bulletin No. 187). Washington, DC: ACOG; 1993.
- American Institute of Ultrasound in Medicine. *Guidelines for the performance of the antepartum obstetrical ultrasound examination*. *J Ultrasound Med* 1991;10:576.
- Small M, Copel JA. Indications for fetal echocardiography. *Pediatr Cardiol.* 2004 May-Jun;25(3):210-22.

