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Prenatal Diagnosis of Congenital Heart Disease: Clinical Experience and Analysis

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Abstract

Over a five-year period, we reviewed 19 fetuses who were prenatally diagnosed with congenital heart disease, including hemodynamically significant arrhythmias. Five of them had fetal tachyarrhythmias, and 14 had structural heart disease. The outcomes were: six intrauterine deaths, five neonatal deaths, and three infant surgeries. Six of the fetuses had chromosomal abnormalities, four had extracardiac anomalies, and two had hydrops fetalis. Of the 96 neonates with congenital heart disease found during the study period, the overall detection rate was 20%; 16% of the neonates with structural cardiac defects and 83% of the neonates with arrhythmias. Some of the complex cardiac defects with normal fetal four-chamber view were difficult to detect prenatally. During the course of the pregnancy, 37% of the fetuses with prenatally diagnosed congenital heart disease were found to have intrauterine growth retardation, and 26% were found to have an abnormal amniotic fluid volume. In view of our findings, a comprehensive screening system should be more frequently considered in order to improve both detection rate and perinatal management.
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Key words: congenital heart disease, prenatal diagnosis

Introduction

Congenital heart disease is the most common fetal anomaly, which has a significant impact on perinatal mortality and morbidity. In addition, most infants with cardiac malformations are born to mothers with a low-risk pregnancy. Therefore, prenatal detection of congenital heart disease is largely dependent on routine obstetrical ultrasonographic scanning. In recent years, the ultrasonographic imaging systems for the fetal heart have markedly improved and provide a high degree of diagnostic accuracy in the detection of congenital heart disease, even at an early gestational age[1]. The addition of pulsed and color flow Doppler to the modality provides additional imaging information[2,3]. We present our experience with the prenatal diagnosis of major congenital heart disease including hemodynamically significant fetal arrhythmias in our institution, and analyze its clinical impact on a perinatal medical practice.

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Patients and Methods

From April 1999 through April 2004, antenatal records from obstetrical ultrasonographic examinations and postnatal diagnostic records for cardiovascular abnormalities from our neonatal intensive care unit were retrospectively reviewed. During the five-year study period, we had 9,152 live births, and 52 stillbirths and/or intrauterine fetal deaths at our institution. To evaluate the efficacy of prenatal diagnosis of congenital heart disease, including hemodynamically significant arrhythmias, on fetal and neonatal clinical course, we analyzed the diagnosis, outcome, and obstetrical factors of the fetuses. We also reviewed all neonates with major congenital heart disease during the same study period, regardless of whether the condition was prenatally diagnosed; problems with prenatal diagnosis are also presented.

In this study, congenital heart disease signifies major structural malformation of the heart and/or the great vessels which requires surgery or catheter intervention within the first six months of life, and hemodynamically significant arrhythmias with structural normal heart.

Delayed closure of a ductus arteriosus in a premature infant and ventricular septal defects of the muscular type were excluded from the study. At our maternity hospital, women attending for routine prenatal care are offered two ultrasonographic examinations, at 20～24 weeks and at 28～32 weeks of gestation by specially trained obstetrician or pediatric cardiologist.

Results

During the study period, 19 fetuses were diagnosed with congenital heart disease; 14 had structural anomalies and five had hemodynamically significant fetal arrhythmias. A ventricular septal defect (VSD) was the most frequently observed form of defect (nine cases), following by two cases of a double outlet of the right ventricle (DORV), one case of tetralogy of Fallot (TOF), and one case of transposition of the great arteries (TGA) with DORV and Ebstein's anomaly.

Precise evaluation for fetal arrhythmia was done with M-mode echocardiography. Four of the patients were diagnosed with supraventricular tachycardias and one was diagnosed with atrial flutter with 2 : 1 atrioventricular conduction (**Table 1**). We found a total of 96 affected neonates during the study period; 90 had structural heart disease and six had hemodynamically significant arrhythmias. Therefore, the overall detection rate was 20% (19/96); 16% (14/90) of the neonates with structural anomalies, and 83% (5/6) of the neonates with arrhythmias (**Table 2**). Of the 19 prenatally diagnosed fetuses, six suffered a fetal demise, five were neonatal deaths, and corrective surgery was performed in three infants (**Table 3**).

Table 1 Classification of the fetal diagnosis

- Structural anomaly: 14
  - VSD (9)
  - DORV (2)
  - TGA + DORV (1)
  - TOF (1)
  - Ebstein's anomaly (1)
- Hemodynamically significant arrhythmias: 5
  - Supraventricular tachycardia (4)
  - Atrial flutter with 2 : 1 conduction (1)

＊ VSD: Ventricular Septal Defect DORV: Double Outlet of Right Ventricle TGA: Transposition of the Great Arteries TOF: Tetoralogy of Fallot

Table 2 Prenatal detection rate of congenital heart disease

	Number of the patients	prenatal diagnosis	detection rate
Structural anomaly	90	14	16%
Hemodynamically significant arrhythmias	6	5	83%
	96	19	20%

All five patients with fetal arrhythmias were successfully treated with transplacental anti-arrhythmic agents. Chromosomal abnormalities were found in six patients; three were trisomy-18, one was a trisomy-21, one was a trisomy-13, and one was a 4p-syndrome. We found three cases of omphalocele, one case of esophageal atresia with tracheoesophageal fistula, and two cases of hydrops fetalis. Seven fetuses had intrauterine growth retardation of unknown etiology, and five fetuses had an abnormal amniotic fluid volume. Of the 90 neonates with structural congenital heart disease, 36 infants were transported from other hospitals, and had not been prenatally diagnosed. Of the remaining 54 infants born in our hospital, 46 were not prenatally diagnosed with a cardiac defect. Aside from a simple VSD, the diagnoses not made in utero were four cases of coarctation of the aorta (CoA) and TOF, three cases of TGA, two cases of totally anomalous pulmonary venous return (TAPVR), one case of atrioventricular septal defect (AVSD), and one case of pulmonary atresia with an intact ventricular septum (PA/IVS) (**Table 4**). There was no false positive case in prenatal diagnosis of congenital heart disease. The pregnancy weeks at prenatal diagnosis was 27.4±7.2 (16～38) in total patients; 25.4±7.1 (16～38) weeks of gestation in case of structural heart disease and 33.2±2.7 (31～37) weeks of gestation in hemodynamically significant arrhythmias.

Table 3 Outcomes

- Corrective surgery 3 (VSD 2, TOF 1)
- Medically treated 5 (SVT 4, AF 1)
- Fetal demise 6 (VSD 4, DORV 2)
- Neonatal death 5 (VSD 3, Ebstein 1, DORV+TGA 1)

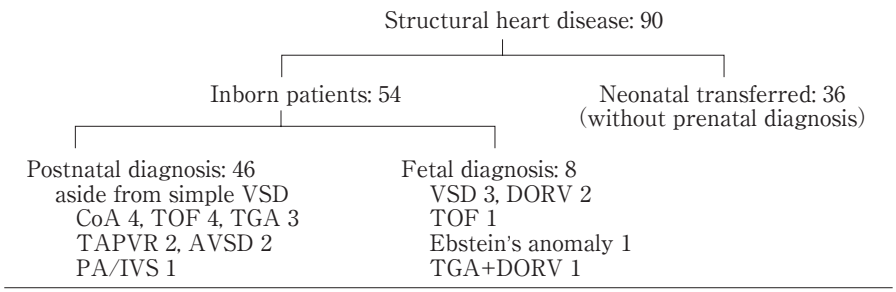
＊VSD: Ventricular Septal Defect
SVT: Supraventricular Tachycardia
DORV: DoubleOutlet of Right Ventricle
AF: Atrial Flutter
TGA: Transposition of the Great Arteries
TOF: Tetoralogy of Fallot

Discussion

Congenital heart disease is the most common severe congenital abnormality with an incidence of about 8 in 1,000 live births[4,5]. Half of these are minor and easily corrected by surgery; however, the remainder account for more than half of the deaths from congenital abnormalities in childhood[6]. This condition is life-threatening and requires intervention in the early neonatal period[7]. Antenatal detection of cardiac defects would facilitate pregnancy counseling and provide more time to develop an appropriate therapeutic strategy.

Early maternal or neonatal transportation to a perinatal tertiary care center with pediatric cardiac facilities ensures optimum conditions for cardiac intervention. Fetal echocardiography has become widely accepted as a useful technology for detecting congenital heart disease in an obstetrical practice. However, with the widespread use of fetal echocardiograms, it has been found that a different

Table 4 Diagnosis of structural heart disease



＊ VSD: Ventricular Septal Defect TGA: Transposition of the Great Arteries CoA: Coarctation of the Aorta TAPVR: Total Anomalous of Pulmonary venous Return TOF: Tetralogy of Fallot AVSD: Atrioventricular Septal Defect DORV: Double Outlet of Right Ventricle PA/IVS : Pulmonary Atresia with Intact ventricular Septum

spectrum of diseases is observed in prenatal life than that found in fetuses who survive to infancy[8]. Fetuses diagnosed in utero frequently have more severe forms of cardiac defects than those diagnosed postnatally; furthermore, those diagnosed in utero have a higher incidence of associated extracardiac lesions or chromosomal abnormalities. In addition, a significant number of affected fetuses spontaneously terminate as an intrauterine demise[9]. Therefore, the overall outcome for fetuses with congenital structural cardiac abnormalities is unfavorable[8], and this situation was confirmed in our series; six fetuses died in utero and five died in the neonatal period.

Among our 19 prenatally diagnosed patients, six had chromosomal abnormalities, four had extracardiac anomalies, and two were hydropic with an intra-uterine demise. Conversely, all five cases of fetal hemodynamically-significant arrhythmias were treated successfully with maternally-administrated digoxin. Because protocols for transplacental anti-arrhythmic treatment are well established, particularly for tachyarrhythmias[10], prenatal diagnosis of fetal arrhythmias facilitates the achievement of a good outcome.

The reported detection rates for fetal cardiovascular abnormalities varies significantly. In our study, the overall detection rate was 20% of all neonates with congenital cardiac abnormalities; 16% of the neonates with structural heart disease and 83% of the neonates with hemodynamically significant arrhythmias. The four-chamber view of the fetal heart is commonly imaged during routine screening, because it does not require specialized skills and is easily obtained. This technique has been found to be an effective method for detecting several of the severe cardiac malformations in utero. Specifically, several cardiac abnormalities are readily visualized with a normal fetal four-chamber view, such as CoA, TOF, TGA and TAPVR. In our series, four cases of CoA, four cases of TOF, three cases of TGA, and two cases of TAPVR could not be diagnosed prenatally; these conditions are difficult to identify with only a fetal four-chamber view. To improve the detection rate, scanning of the great vessels' outflow tracts in addition to the four-chamber view has been proposed[16]. However, despite the fact that the vast majority of fetuses with severe congenital heart disease occur in low-risk pregnancies, it is currently controversial to routinely apply this study because it, requires significant time and skill. To address this dilemma, the clinician should take into account any risk factors of congenital heart disease, such as a family history of congenital heart disease, maternal diabetes, exposure to teratogens in early pregnancy, the detection of an extracardiac fetal anomaly, and severe intrauterine growth retardation.

Related obstetrical factors, which were found in this study, were intrauterine growth retardation of unknown origin (37%), and an abnormal amniotic fluid volume (26%). Fetal echocardiography provides additional information about the development of the heart in the presence of structural or functional disease; thus, it contributes to a better understanding of the natural history of congenital heart disease. Although only a small percentage of fetuses with congenital heart disease can be prenatally detected, there is evidence that fetuses with some types of defects experience decreased morbidity and mortality when a prenatal diagnosis is made, particularly in cases of ductus-dependent complex cardiac lesions[17]. In the future, cost-and time-effective screening procedures should be included in educational programs for sonographers because the initial detection of major fetal malformations is almost completely dependent upon routine obstetric ultrasonographic scanning.

References

1. Allan LD, Chita SK, Sharland GK, Fagg NLK, Anderson RH, Crawford DC: The accuracy of fetal echocardiographiy in the diagnosis of congenital heart disease. Int J Cardiol 1989; 25: 279–288.
2. Allan LD, Chita SK, Al-Ghazahi W, Crawford DC, Tynan MJ: Doppler echocardiographic evaluation of the normal human fetal heart. Br Heart J 1987; 57: 528–533.
3. Sharland GK, Chita SK, Allan LD: The use of colour Doppler in fetal echocardiography. Int J Cardiol 1990; 28: 229–236.
4. Mitchell SC, Korones SB, Berendes HW: Congenital heart disease in 56,109 births: Incidence and natural history. Circulation 1971; 43: 323–332.
5. Ferencz C, Rubin JD, McCarter RJ, Brenner JI, Neill

CA, Perry LW, Hepner SI, Downing JW: Congenital heart disease: Prevalence at livebirth. The Baltimore-Washington infant study. Am J Epidemiol 1985; 121: 31–36.
6. Hoffman JIE, Christianson R: Congenital heart disease in a cohort of 19,502 births with long-term follow-up. Am J Cardiol 1978; 42: 641.
7. Tegnander E, Eik-Nes SH, Johansen OJ, Linker DT: Prenatal detection of heart defects at the routine fetal examination at 18 weeks in a non-selected population. Ultrasound Obstet Gynecol 1995; 5: 372–380.
8. Allan LD, Sharland GK, Miburn A, Lockhart SM, Groves AM, Anderson RH, Cook AC, Fagg NL: Prospective diagnosis of 1,006 consecutive cases of congenital heart disease in the fetus. J Am Coll Cardiol 1994; 23: 1452–1458.
9. Sharland GK, Lockhart SM, Chita SK, Allan LD: Factors influencing the outcome of congenital heart disease detected prenatally. Arch Dis Child 1990; 64: 284–287.
10. Simpson JM, Sharland GK: Fetal tachycardias: management and outcome of 127 consecutive cases. Heart 1998; 79: 576–581.
11. Chitty LS, Hunt GH, Moore J, Lobb MO: Effectiveness of routine ultrasonography in detecting fetal abnormalities in a low risk population. BMJ 1991; 303: 165–169.
12. Luck CL: Value of routine ultrasound scannning at 19 weeks: a four year study of 8849 deliveries. BMJ 1992; 304: 1474–1478.
13. Shirley IM, Bottomley F, Robinson VP: Routine radiographer scannning for fetal abnormalities in an unselected low risk population. Br J Radiol 1992; 65: 564–569.
14. Crane JP, LeFevere ML, Winborn RC, Evans JK, Ewigman BG, Bain RP, Frigoletto FD, McNellis D: A randomized trial of prenatal ultrasonographic screening: Impact on the detection, management and outcome of anomalous fetuses. Am J Obste Gynecol 1994; 171: 392–399.
15. Levi S, Hyjazi Y, Schaaps JP, Defoort P, Coulon R, Buekens P: Sensitivity and specificity of routine antenatal screening for congenital anomalies by ultrasound: the Belgian multicentric study. Ultrasound Obste Gynecol 1991; 5: 366–371.
16. Yagel S, Cohen SM, Achiron R: Examination of the fetal heart by five short-axis views: a proposed screening method for comprehensive cardiac evaluation. Ultrasound Ob Gy 2001; 17: 367–369.
17. Davis GK, Farquhar CM, Allan LD, Crawford DC, Chapman MG: Structural cardiac abnormalities in the fetus: reliability of prenatal diagnosis and outcome. Br J Obstet Gynecol 1990; 97: 27–31.

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