



## Prenatal diagnosis of congenital fetal heart abnormalities and clinical analysis\*

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**Abstract:** Objective: To study the value of detecting fetal congenital heart disease (CHD) using the five transverse planes technique of fetal echocardiography. Methods: Nine hundred and eighty-two high-risk pregnancies for fetal CHD were included in this study, the fetal heart was scanned with the five transverse planes technique of fetal echocardiography described by Yagel, autopsy was conducted when pregnancy was terminated. Blood from fetal heart was collected for fetal chromosome analysis. A close follow-up was given for normal fetal heart pregnancies and neonatal echocardiography was performed to check the accuracy of prenatal diagnosis. Results: (1) Forty-six cases (4.68%) were found to have fetal heart abnormalities in this study, 69.56% of them were diagnosed by single four-chamber view, another 30.43% fetal CHD were found by combining other views; (2) Forty-one parents of prenatal fetuses with CHD chose to terminate pregnancy, thirty-two of them gave consent to conduct autopsy, 93.75% of which yielded unanimous conclusion between prenatal fetal echocardiography and autopsy; (3) Thirty-two of 46 cases underwent fetal chromosome analysis, 8 cases (25%) were found to have abnormal chromosome; (4) Five cases were found to have right ventricle and atrium a little bigger than those on the left side, with the unequal condition being the same after birth, but there were no clinical manifestations and they are healthy for the time being; (5) Nine hundred and thirty-six cases were not found with abnormality in this study, but one case was diagnosed with ventricular septal defect after birth, one case was diagnosed with patent ductus arteriosus, one case had atrial septal defect after birth. Conclusions: (1) The detected CHD rate was 4.68% by screening fetal heart with five transverse planes according to Yagel's description of high risk population basis for CHD. The coinciding rate of prenatal diagnosis and autopsy was 93.75%; (2) The sensitivity of detecting fetal heart abnormality is 92%, the specificity is 99.6% using the five transverse planes technique of fetal echocardiography; (3) Fetuses with mild or moderate disproportion of right and left side in the heart are potentially healthy babies.

**Key words:** Fetus, Heart, Abnormality, Prenatal, Diagnosis

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### INTRODUCTION

Congenital heart disease (CHD) is the most common congenital disorder resulting in significant prenatal and postnatal morbidity and mortality (Gillum, 1994; Gembruch, 1997) and is believed to be a multifactorial disorder arising from the combined effect of genetic predisposition and environmental factors. Prenatal diagnosis of CHD is very important for both fetal prognosis and decreasing economic burden of family and society. It was demonstrated

recently that fetal echocardiography is a valuable tool for prenatal diagnosis of CHD. We scanned 982 high risk pregnancies for fetal CHD from 18 to 40 gestational weeks in order to investigate the value of detecting CHD with simplified and streamlined five transverse planes technique of fetal echocardiography described by Yagel *et al.*(2001).

### MATERIALS AND METHODS

#### Patients

Nine hundred and eighty-two pregnant women at

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Second Affiliated Hospital of China Medical University with high risk for fetal CHD were included in the study during the year from 2001 to 2003. Among them, 21 women had family history of CHD, 324 women had delivered fetuses with different malformations, 83 women were more than 35 years old, 49 cases were complicated with diabetes, 78 pregnancies were complicated with abnormal amniotic fluid, 32 fetuses suffered from fetal growth restriction, 62 fetuses had been exposed to teratogen, 107 fetuses were found to have extra-cardinal malformations, 216 cases were fetal arrhythmia, 18 cases were suspected CHD after routine ultrasonography. Gestational weeks were from 18 weeks to 41 weeks, maternal age was 21 to 42 years.

### Machine

Fetal echocardiography was carried out with GE VIVID7 Ultrasound Doppler machine produced by America GE Company, the transducer frequency was 3.5 MHz or 5 MHz.

### Examination methods

Fetal heart examination was performed with the woman in supine position. Fetal echocardiography was performed with five heart transverse planes according to Yagel *et al.* (2001)'s description. The simplified and streamlined five transverse planes were as follows: the first and most caudal plane is a transverse view of the upper abdomen: moving cephalad. The next is the traditional four-chamber view. The third is the plane commonly termed the five-chamber view, in which the aortic root is visualized. The fourth transverse view reveals the bifurcation of the pulmonary arteries. The fifth is the three vessels and trachea plane to reveal the main pulmonary trunk in direct communication with the ductus arteriosus (Yoo *et al.*, 1999).

### Parents were informed of the cardiac diagnosis and prognosis

Subsequent counseling regarding the pregnancy was given by obstetrician and pediatric cardiologist together. The decision was made by parents after counseling, the option of termination of pregnancy was supported for severe CHD cases and the autopsy was conducted to confirm the prenatal diagnosis; for normal heart and mild CHD fetuses, a close follow-up

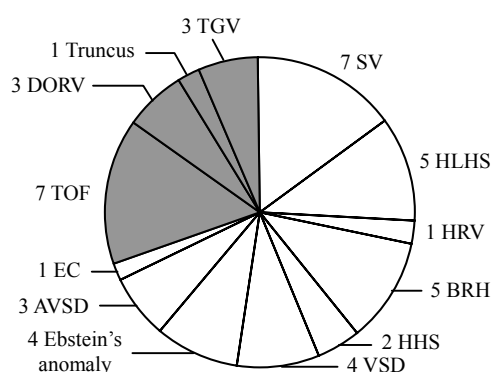
was given until one year after delivery, and neonatal echocardiography was performed to check the accuracy of prenatal diagnosis.

### Chromosome analysis

Blood from umbilical artery or from fetal heart was collected for chromosomal analysis. The strip level of analysis for chromosome was about 400.

## RESULTS

Forty-six cases (4.68%) were found to be fetal heart malformations, among them, eight cases (8/324, 2.47%) had poor obstetrical history (delivered an abnormal fetus), two cases (2/21, 9.53%) had family history of CHD, five cases (5/216, 2.31%) were fetal arrhythmia. The types of CHD are shown in Fig.1. Among these 46 cases with CHD, 32 of them (69.56%) were found by single four-chamber view; another 14 cases (30.43%) were diagnosed by combining five transverse planes. CHD was found by combining five transverse planes which were tetralogy of Fallot (TOF), aorta overriding the interventricular septum, ventricular septal defect and mildly stenotic to atretic pulmonary artery are key points for TOF, this finding can be demonstrated by the third to fifth transverse planes); common arterial truncus (a single artery overriding the ventricular septum and the origin of the



**Fig.1 Types of congenital heart disease in 46 cases**

AVSD: Atrial ventricular septal defect; BRH: Bigger right heart; DORV: Double outlet right ventricle; EC: Ectopia cordis; HHS: Heart hypoplastic syndrome; HRV: Hypoplastic right ventricle; HLHS: Hypoplastic left heart syndrome; SV: Single ventricle; Truncus: Common arterial truncus; TOF: Tetralogy of Fallot; TGV: Transposition of great vessel; VSD: Ventricular septal defect. Grey color denotes CHD detected by combining five transverse planes; White color part denotes detected with only four-chamber view

pulmonary arteries from the truncus are key points, this finding can be demonstrated by the third to fifth transverse planes); Double outlet right ventricle (the aorta and the pulmonary artery arising from the right ventricle are the key points, although the findings may closely resemble TOF or transposition of great vessel with ventricular septal defect, and can be demonstrated by the third to fifth transverse planes); Transposition of great vessel (the absence of the normal anatomic criss-crossing and the parallel presence of the aorta and pulmonary arteries are key points, and can be demonstrated by the third and fourth transverse plane).

All parents of fetuses with fetal CHD except for 5 cases with mild large right ventricle and right atrium decided to terminate pregnancy. Autopsies were conducted in 32 cases, 93.75% (30/32) of which led to unanimous conclusion between prenatal echocardiography and autopsy; one case of truncus arteriosus initially diagnosed as Tetralogy of Fallot, another case was double-outlet right ventricle but was diagnosed as transposition of the great vessels.

The 5 cases with moderately large right ventricle and right atrium (The ratio of left to right was about 1:1.4~1:1.6) found at 25, 26, 28, 31, 34 gestational weeks were followed up closely, they are from 3 months to 1 year old now; echocardiography after birth yielded almost the same result prenatally and there were no clinical manifestations for the time being.

Among 46 cases with CHD, 32 were subjected to chromosomal analysis, and eight cases (25%) were found to have abnormal chromosomes, three of which were 18 trisomy, four cases were 21 trisomy, one case was 13 trisomy.

Among 216 cases with fetal arrhythmia, fetal CHD were found in five cases (2.31%). The types of fetal arrhythmia were as follows: 41 cases were frequent premature atrial beats, 12 cases were frequent premature ventricular beats, 9 cases were complete heart block, 9 cases were bradycardia, 94 cases were supraventricular tachycardia, 54 cases were ventricular tachycardia, 3 cases were atrial flutter, 1 case was atrial fibrillation.

Nine hundred and thirty-six cases were found to be without abnormality prenatally in this study, but one case was diagnosed as small ventricular septal defect (VSD,  $\Phi=5$  mm) after birth, one case was

patent ductus arteriosus, one case was atrial septal defect (ASD) after birth.

## DISCUSSION

Our results showed that the detected rate of CHD was 4.68% if the fetal heart is screened with five transverse planes according to Yagel's description on the high risk population basis for CHD, 93.75% of them led to unanimous conclusion between prenatal ultrasound and autopsy. Although two cases did not lead to an unanimous conclusion between prenatal ultrasonography and autopsy, actually they were not false diagnose in view of the clinical significance; they were complicated with great vessel abnormalities of similar image, which made it difficult to make exact diagnosis prenatally.

Our results showed that the sensitivity of detecting CHD with single four-chamber view was 64%, the sensitivity increased to 92% if other transverse planes were combined. One case with small VSD was missed prenatally in our study, probably because the ovale foramen and ductus arteriosus are open prenatally, which cause the same pressure in the right ventricle and the left ventricle, therefore there is no shunt through the VSD prenatally and the small defect is beyond the ultrasound resolving power (Benacerraf and Sanders, 1990). For cases of patent ductus arteriosus and secondary ASD, it was impossible to make diagnosis prenatally since the ovale foramen and ductus arteriosus are open during pregnancy, and will close naturally after birth. There will be patent ductus arteriosus and secondary ASD if they do not close after birth. The above factors limit the sensitivity and specificity of fetal echocardiography. However, the sensitivity of detecting fetal heart abnormality is 92%, and the specificity is 99.6% using simplified and streamlined five transverse planes in our study.

Benacerraf *et al.* (1987) reported that the recurrence risk of fetal CHD was 4% if parents delivered a fetus with CHD before, and the risk of having a fetus with CHD was 4% if the gravida patient had a family history of CHD. Our result showed that the recurrence risk of fetal CHD was 2.47% if parents delivered a fetus with CHD before, and the risk of having a fetus with CHD was 9.53% if the patient had a family history of CHD. The CHD risk due to abnormal obstet-

rical conditions and family history was 21.74%.

The fetal heart's right side is dominant during pregnancy, the ratio of right to left side being about 1:1.0~1:1.3 (Hung *et al.*, 1991). An obvious disproportion of right and left side usually denotes an abnormal heart anatomy, such as hypoplastic left heart syndrome, hypoplastic right ventricle, coarctation of the aorta, aortic stenosis and premature closure of ductus arteriosus, etc. Five cases had light or moderate disproportion, but no other abnormalities were found, a close follow-up given, all of them showed that they were healthy for the time being. Therefore, good prognosis is possible for mild or moderate disproportion of fetal heart without other abnormalities.

Our results showed that 25% of fetus with CHD had abnormal chromosome. Abnormal chromosome is a major hereditary factor and 50% of fetus with abnormal chromosome had CHD (Ferencz *et al.*, 1989), therefore a routine fetal heart screening is strongly suggested for high risk population with chromosomal abnormalities.

Fetal arrhythmia is common during pregnancy, its incidence is about 1%, with approximately 10% being caused by CHD (Kleinman and Nehgme, 2004; Allan, 1994). Our result showed that 2.31% of fetal arrhythmia was caused by CHD.

CHD is a multifactorial and complicated disorder; the etiology is unclear and the prognosis is poor, so prenatal diagnosis of CHD is important. Our result suggested that the simplified and streamlined five

transverse planes method is an effective fetal echocardiography for finding CHD and is of benefit for clinical management.

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