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

INTRODUCTION

Congenital heart disease (CHD) is the most common congenital disorder resulting in significant perinatal 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
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.