Prenatal Diagnosis of Congenital Heart Disease by Fetal Echo

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Abstract

**Background:** Infant mortality rate in sick neonatal unit is on an increase. By subjecting antenatal mothers to fetal echo, congenital heart disease (CHD) of the fetuses can be screened earlier and the labor and further management of the fetus can be planned in tertiary care centre in coordination with cardiothoracic surgeons.

**Aim of the Study:** This study aims to screen for CHD in utero by 4-chamber view. To pick up the major cardiac anomalies earlier and to counsel the parents accordingly. This will have indirect improvement in both maternal and fetal outcome and finally to reduce infant mortality rate.

**Materials and Methods:** Antenatal mothers referred for fetal cardiac evaluation were subjected for transabdominal fetal echo evaluation. Gross anomalies which can be detected by 4-chamber views, aortic level short axis views, were arrived at such as ventricular septal defect (VSD), tetralogy of Fallot (TOF), coarctation, and aortic stenosis Wipro GE Logiq 5 Echo machine with transducer frequency between 3.5 and 5 MHz.

**Observations and Results:** Of the 450 maternal cases studied, 87 (20.4%) were high maternal risk cases and 3 (0.6%) had high-risk fetal factors. Of the above screened fetuses, 4 (0.9%) had echogenic focus in left ventricle, 1 (0.2%) had VSD, 1 (0.2%) had TOF, and 1 (0.2%) had bradycardia.

**Conclusions:** Fetal echo evaluation is possible as a routine screening. High-risk maternal and fetal cases should undergo fetal echo evaluation. 4-chamber and 5-chamber views at least should be done to diagnose VSD/TOF and other major CHD. Incidence of fetal anomaly is 0.4/1000 according to our study and correlates with published data. Diagnosing CHD in fetal life will have its impact on fetal outcome and improvement in quality of care, both for the mother and the newborn. It gives mental satisfaction for the parent to know their fetal heart condition in utero itself. Early intervention after birth will prevent future complications of CHDs.

**Key words:** Anomalies, Congenital heart disease, Fetal echocardiogram antenatal

INTRODUCTION

Prenatal detection is essential for improving perinatal outcomes of neonates with critical congenital heart disease (CHD). Comprehensive evaluation of the fetal heart includes evaluation of the situs, sagittal and transverse plane imaging, evaluation of the fetal cardiac rate and rhythm and Doppler assessment.

**Period of Study**

The study period was from August 2015 to February 2016 – 6 months.

**Institute of Study**

This study was at the Department of Cardiology, Thoothukudi Medical College Hospital, Thoothukudi.

**MATERIALS AND METHODS**

Antenatal mothers referred for fetal cardiac evaluation we subjected for transabdominal fetal echo evaluation. Gross anomalies which can be detected by 4-chamber views, aortic level short axis views, were arrived at such as ventricular septal defect (VSD), tetralogy of Fallot (TOF), coarctation, and aortic stenosis Wipro GE Logiq
Fetal cardiac evaluation was done in the following steps:

- Situs\(^5\)
- Cardiothoracic ratio (CTR)\(^5\)
- Axis\(^5\)
- 4-chamber view\(^6\)
- 5-chamber view\(^6\)
- OT views\(^6\)
- Real-time evaluation of valves\(^5\)
- Real-time evaluation of interventricular septum (IVS) and interatrial septa\(^5\)
- M mode\(^6\)
- Color flow mode and Doppler.\(^6\)

**Inclusion Criteria**

Antenatal mothers 13–24 weeks were included in the study. Viable fetuses were only screened. Twins are also included. High-risk mothers of having fetal cardiac anomalies were also included. All age mothers were included.\(^7\)

**Exclusion Criteria**

Already diagnosed cases of fetal cardiac anomalies were excluded. Intrauterine device cases were included. Those not willing to give consent were also excluded.

**RESULTS**

Comorbid illness and maternal risk factors – Table 1. Fetal risk factors – Table 2. Transabdominal views done – Table 3. Cardiac abnormalities detected – Table 4.

**DISCUSSION**

Detecting fetal cardiac anomaly is most challenging because of moving fetus.\(^8\) Locating fetal heart and getting standard views are also very difficult. Foramen ovale and ductus are seen normally;\(^9\) hence, autism spectrum disorder (ASD) and pathological demand avoidance (PDA) diagnosis are possible only after delivery.\(^9\) Patent foramen ovale will persist even after 1 week.\(^10\) as shown in Figure 1. PDA will close in a day after birth.\(^10\) Fetal circulation and streaming could be visualized by serial Doppler echo. Color flow mapping and pulse Doppler imaging in M mode can be used for the assessment of ventricular function.\(^11\) Doppler assessment across mitral inflow is shown in Figure 2. IVS defects can be diagnosed in utero.\(^11\) Valvular abnormalities can be diagnosed easily. Risk factors for having cardiac anomaly are elderly, diabetic mothers and those with CHDs and those on drugs such as antiepileptics and antipsychotics, and warfarin, alcoholic mothers can have fetal anomalies.\(^12\) Those who had TORCH infections during the first trimester, those who are exposed to irradiation and teratogenic agents are also at risk of fetal anomalies and extracardiac anomalies.\(^13\)

SLE mothers have fetal risks of hydrops, genetic abnormalities, IUGR, polyhydramnios, increased nuchal translucency, and absent nasal bone.\(^14\)
Preferred 4-chamber view as screening tool helps in diagnosing at least one-third of cases of CHD. Split of data of gestational age of fetus was not collected since some had unknown LMP. It is obtained from the obstetrician’s record.

Preferred duration of pregnancy is after 16 weeks in our study. We included AN mother in the second trimester. Term and near term will have poor echo window due to the reflection of ultrasound waves by bones of grown-up fetus. Incidence of GDM and DM is increasing and they have 5 times more chance of having cardiac structural abnormalities. Hence, in our study, special attention was given to diabetic mothers.

Among the total 450 mothers screened, almost all had situs solitus, levocardia. One had muscular VSD. One had TOF. Four had echogenic foci in LV. One had fetal bradycardia without any structural heart disease [rate around 90/mt]. There was no follow-up in that case. No tachycardia except for VPC in one case while recording M mode across the LV cavity level in long axis view. There were no subaortic, aortic, and supravalvular stenosis. No coarctation case was diagnosed. Rhabdomyoma can be diagnosed. In our study, pericardium was normal; no mass and no vegetation were seen. One mother had twin gestation. Both had normal pericardium structure and activity. Thin sheet of pericardial effusion may be a normal finding in fetal echo.

Average time taken for fetal echo was 10–20 mts. 4 chamber, 5 chamber, and aortic views can be seen without much difficulty in almost all cases. Other views were little difficult since fetal movements were exclusive.

The epidemiological data are similar to published series.

Fetal cardiac intervention is possible in advanced centers.

Follow-up and advice given to the parent of CHDs, i.e., VSD and TOF cases. 2D and color flow across VSD is shown in Figures 3 and 4, respectively. TOF was shown in Figures 5 and 6. After delivery, echo was done to confirm the diagnosis. TOF case expired in the postnatal period due to fatal cyanotic spell. A case of Muscular Ventricular defect diagnosed antenatally was delivered and the child is under follow up at higher Centre and planned for device closure later.

There was no PHT in the VSD case.

Neonatal echo screening was done for sick neonates and underweight babies. Larger ASDs were diagnosed without

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**Table 4: Cardiac abnormalities detected**

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>Count</th>
</tr>
</thead>
<tbody>
<tr>
<td>Echogenic focus in left ventricle</td>
<td>4</td>
</tr>
<tr>
<td>Ventricular septal defect</td>
<td>1</td>
</tr>
<tr>
<td>TOF</td>
<td>1</td>
</tr>
<tr>
<td>Transposition of great arteries</td>
<td>-</td>
</tr>
<tr>
<td>Coarctation of aorta</td>
<td>-</td>
</tr>
<tr>
<td>Single ventricle</td>
<td>-</td>
</tr>
<tr>
<td>Double outlet right ventricle</td>
<td>-</td>
</tr>
<tr>
<td>Aortic stenosis</td>
<td>-</td>
</tr>
<tr>
<td>Pulmonary stenosis</td>
<td>-</td>
</tr>
<tr>
<td>Atrioventricular canal defects</td>
<td>-</td>
</tr>
<tr>
<td>Malposition</td>
<td>-</td>
</tr>
<tr>
<td>Bradycardia</td>
<td>1</td>
</tr>
<tr>
<td>Other defects</td>
<td>-</td>
</tr>
</tbody>
</table>

TOF: Tetralogy of Fallot

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**Figure 1:** Fetal echocardiogram showing normal apical 4-chamber view with color flow showing foramen ovale

**Figure 2:** Fetal echocardiogram showing Doppler across mitral valve showing A wave dominance than E wave

Prenatal diagnosis depends on the experience of examiner, obesity of the mother, frequency of transducer, abdominal conditions, gestational age of fetus, and fetal position.
any difficulties and were advised follow-up at 30 days of life.

Tiny PDAs were found in the neonatal period. They were also advised to come for follow-up at 30 days.

Separate neonatal echo registry was made in the SNN ward and it is not discussed here.

**Limitations of the Study**
1. Small sample.
2. Not all standard views were possible in every case.
3. Observer variation.
4. Cardiac malpositions may be missed.

**CONCLUSION**

Fetal echo evaluation is possible as a routine screening. High-risk maternal and fetal cases should undergo fetal echo evaluation. 4-chamber and 5-chamber views at least should be done to diagnose VSD/TOF and other major CHDs. Incidence of fetal anomaly is 0.4/1000 according to our study and correlates with published data.\textsuperscript{19} Diagnosing CHD in fetal life will have its impact on fetal outcome and improvement in quality of care, both for the mother and the newborn. It gives mental satisfaction for the parent to know their fetal heart condition \textit{in utero} itself. Early intervention after birth will prevent future complications of CHDs.

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REFERENCES


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