ABSTRACT: BACKGROUND: Structural abnormalities of the heart and great vessels are fairly common congenital abnormalities with incidence of 8 in 1000 live births. With the advent of real time scanners fetal cardia can atomy can be analyze dechocardiographically. The earlier diagnosis will make an impact on clinical management of fetus with congenital heart disease. It helps intimely triage and optimal management of specific congenital heart disease either structural, functional or arrhythmia. OBJECTIVES: This study was conducted to note the spectrum of congenital heart diseases detected on fetal echo in pregnant mothers referred with high risk for CHD sand to assess the outcome of prenatally detected congenital heart diseases. MATERIALS AND METHODS: The study is aprospective descriptive study conducted in a tertiary care pediatric hospital in Mumbai over period of one year. Pregnant mothers were referred for fetal echo, where pregnancy was considered as high risk for CHDs due to maternal, fetalfactorsorabnormallevel 1 scan. Fetal echowas performed by a trained pediatric cardiologistat 18 to 20 week of gestation using HP sonos 2000 echocardiographicmachinewith3/3.5 Hztransducer. Cardiac lesionsandoutcome of pregnancy was noted by postnatal follow-up of patients. RESULTS: A total of 170 patients underwent fetal echo, 13 patients have not delivered and 48 were lost to follow-up. Fetal echo was normal in 130(76.4%) and abnormalities were detected in 40(23.5%). Structural anomalies were seen in 24(14.1%), arrhythmia in 5(2.9%) and functional abnormalities in 11(6.4%). On outcome analysis 84 (77.1%) arealive, IUD /terminationof pregnancyoccurred in 18(16.5%), neonatal death in 6(5.5%), infant death in 1 (0.9%). CONCLUSIONS: All ranges of CHDs can be diagnosed by fetal echocardiography. Outcome of prenataletly detected complex congenital heart disease is poor: nonetheless earlier detection provides an opportunity for early interventions and optimizes intranatal and postnatal care. KEY WORDS: fetal echo, outcome, congenital heart diseases.

INTRODUCTION: Structural abnormalities of the heart and great vessels are fairly common congenital abnormalities with incidence of 8 in 1000 live births.¹ Ultrasound for the prenatal diagnosis of congenital heart disease is now a well- accepted component of perinatal medicine. Fetal cardiac anatomy can be analyze dechocardiographically, with the advent of real-time scanners usingestablished principles of sequentialsegmental structural analysis.² Earlier diagnosis of Congenital Heart disease in utero will make a significant positive impact on clinical outcome of patients in terms of choosing the place/institution and route of delivery when the fetal Echo has recognized a serious Cardiac abnormality in the fetus. We also would have the ability to provide genetic counselingand monitor the Cardiac arrhythmias during transplacental treatment. In the near future, acquisition of such information may even facilitate in utero Cardiac intervention as apart of fetal therapy³-⁵.
With termination of pregnancy being one of the options provided to parents of fetus with severe congenital heart disease detected prenatally, the postnatal incidence of severe congenital heart disease like hypoplastic left heart syndrome and pulmonary atresia with intact ventricular septum may be declining, as demonstrated in some studies from select areas. Thus it is important to recognize that prenatal diagnosis can have a very profound effect on the incidence of congenital heart disease in the community.

The prenatal diagnosis of congenital heart disease aids formulation of management protocols for prenatally diagnosed congenital heart disease. Newer therapeutic approaches evolving for fetal therapy has allowed a new sub-specialty to develop, that of Fetal Cardiology.

Despite the advances in the west there are only few centers in India doing fetal echo and management of fetus thereof. No database or registry exists in India to give an actual insight into the problem of congenital heart disease and their prenatal diagnosis.

Hence a study was conducted in our center to note the spectrum of congenital heart diseases in the fetus and to note the outcome, both antenatal and postnatal.

OBJECTIVES:
1. To note the spectrum of congenital heart disease detected on fetal echocardiography in mothers referred with high risk for congenital heart disease.
2. To assess the outcome of prenatally detected congenital heart disease and to note the fetal and neonatal survival.

MATERIALS AND METHODS: The study is a prospective study conducted in tertiary level multi-specialty twin hospitals with pediatric and maternity facilities in the city of Mumbai. Mothers were referred consecutively, where pregnancy was considered as high risk for congenital heart disease. The standard risk factors were gestational, familial or fetal.

A total of 170 antenatal mothers were referred for fetal echo, of whom 48 were lost to follow up and 13 have not delivered.

Study period was 1 year.

INCLUSION CRITERIA WERE:
1. Maternal metabolic risk factors
   - Diabetes
   - Phenylketonuria
   - Homocystinuria
2. Exposure to Cardiac teratogens.
   - Lithium Carbonate
   - Folate antagonist
   - Alcohol
   - Anticonvulsants
   - Maternal infections
3. Maternal or paternal congenital heart disease.
FETAL FACTORS:
- Abnormal situs
- Non-immune Hydrops.
- Chromosomal abnormalities.
  - Fetal Extra Cardiac abnormalities.
  - Increased first trimester Nuchal translucency
  - Fetal arrhythmia.
- Besides the above those mothers referred with abnormal level I scans were included.

Education regarding the importance of obtaining 4-chamberview on routine obstetric scan in screening for congenital heart disease was given to sonographers by the fetal echocardiographer.

Fetal Cardiac Scanning was done by fetal echocardiographer using HP sonos 2000 echocardiographic Machine, 2/2.5 Hz or 3/3.5 Hz frequency transducer. Video recordings for further reference were also made.

TIMINGOF FETAL ECHO: The first echo examination was electively done at 18-20 week of gestation or whenever pregnant mother was referred.

All mothers had undergone level I USG Scan in obstetric unit, performed by Sonographer. Screening for Congenital heart disease was done by 4 CH/ 5– Chamber view, which is a part of the basic obstetric ultrasound examination.

DATAENTRY: A detailed register and video recordings were maintained noting the patient details, reason for referral, echo findings, obstetric ultrasound findings and any extracardiac anomaly.

POSTNATAL FOLLOWUP: Subsequently patient follow-up was done to note pregnancy outcome, mode and place of delivery; post natal scans were done to confirm the findings on fetal echo. Any intervention done in the pregnancy or neonatal period was noted.

Immediate postnatal scans were done in all complex lesions or when they were symptomatic. In simple lesions postnatal outcome was noted and scans were done electively before discharge. In patients with normal scans outcome was noted telephonically and postnatal scans done electively whenever possible.

STATISTICALANALYSIS: The significance of difference between the proportions was tested statistically by applying ‘Z’ test and ‘P’ value was derived. The 95% confidence interval of proportions was derived by calculating the standard error of proportions.

RESULTS: Distribution of mothers according to gestational age at which the fetal echo was done. 48.7% of mothers were between 21-28 wks. followed by 24.7% in 29-32 weeks.

<table>
<thead>
<tr>
<th>Gest. Age</th>
<th>n = 170</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>16-20 week</td>
<td>19</td>
<td>11.1</td>
</tr>
<tr>
<td>21-24 week</td>
<td>45</td>
<td>26.4</td>
</tr>
<tr>
<td>25-28 week</td>
<td>38</td>
<td>22.3</td>
</tr>
<tr>
<td>29-32 week</td>
<td>42</td>
<td>24.7</td>
</tr>
<tr>
<td>33-36 week</td>
<td>19</td>
<td>11.1</td>
</tr>
<tr>
<td>&gt; 37 week</td>
<td>7</td>
<td>4.1</td>
</tr>
</tbody>
</table>

Table 1
37.5% of referrals were before 24 weeks of gestation and rest 62.5% were after 24 weeks when termination cannot be undertaken safely even if a complex CHD is detected.

<table>
<thead>
<tr>
<th>Antenatal USG</th>
<th>n</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Abnormal 4-CH View</td>
<td>41</td>
<td>24.1</td>
</tr>
<tr>
<td>Hydrops</td>
<td>11</td>
<td>6.4</td>
</tr>
<tr>
<td>Extracardiac Anomalies</td>
<td>15</td>
<td>8.8</td>
</tr>
<tr>
<td>Arrhythmia</td>
<td>11</td>
<td>6.4</td>
</tr>
<tr>
<td>Polyhydramnios</td>
<td>5</td>
<td>2.9</td>
</tr>
<tr>
<td>Others</td>
<td>2</td>
<td>1.2</td>
</tr>
</tbody>
</table>

Table 2: Diagnosis on Antenatal USG on Level I performed by Sonologist

**Extracardiac anomalies detected were**
- Single umbilical artery 3
- Congenital diaphragmatic hernia 3
- Renal anomalies 4
- Duodenal atresia 1
- Cystic hygroma 1
- Dandy walker cyst 1
- Sacral meningomyelocele 1
- Gastrochisis 1

Others include 1 patient with absent diastolic flow in umbilical arteries and 1 patient with high resistance to flow in uterine arteries.

Table – 3: Reasons for referral for fetal echo. Previous child with CHD and abnormal level I scan were predominant reasons for referral with 30% and 24.1%.

<table>
<thead>
<tr>
<th>Reason for referral</th>
<th>n = 170</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Previous child with CHD</td>
<td>51</td>
<td>30</td>
</tr>
<tr>
<td>Abnormal 4-CH view</td>
<td>41</td>
<td>24.1</td>
</tr>
<tr>
<td>Gestational diabetes</td>
<td>21</td>
<td>12.3</td>
</tr>
<tr>
<td>Extracardiac anomalies</td>
<td>15</td>
<td>8.8</td>
</tr>
<tr>
<td>Arrhythmia</td>
<td>10</td>
<td>5.9</td>
</tr>
<tr>
<td>Hydrops</td>
<td>11</td>
<td>6.4</td>
</tr>
<tr>
<td>Others</td>
<td>38</td>
<td>22.4</td>
</tr>
</tbody>
</table>

Table 3

In 17 patients (10%) there was more than one reason for referral. Others category include maternal factors like Advanced maternal age, Bad obstetric history, mother with ASD, maternal SLE, MVP & polyhydramnios.
Fetal echo

<table>
<thead>
<tr>
<th>Condition</th>
<th>n</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Normal</td>
<td>135</td>
<td>79.4</td>
</tr>
<tr>
<td>Large Atrial septal defect.</td>
<td>2</td>
<td>1.2</td>
</tr>
<tr>
<td>Ventricular septal defect.</td>
<td>7</td>
<td>4.1</td>
</tr>
<tr>
<td>AV Canal defect</td>
<td>5</td>
<td>2.9</td>
</tr>
<tr>
<td>Tetralogy of Fallot</td>
<td>3</td>
<td>1.8</td>
</tr>
<tr>
<td>Hypoplastic left heart syndrome.</td>
<td>1</td>
<td>0.6</td>
</tr>
<tr>
<td>Ebsteins anomaly</td>
<td>1</td>
<td>0.6</td>
</tr>
<tr>
<td>Tricuspid atresia</td>
<td>5</td>
<td>2.9</td>
</tr>
<tr>
<td>Arrhythmia</td>
<td>5</td>
<td>2.9</td>
</tr>
<tr>
<td>Others</td>
<td>11</td>
<td>6.5</td>
</tr>
</tbody>
</table>

Table 4: Cardiac findings on fetal ECHO

Others category include functional abnormalities,
- RA/ RV overload with TR: 3
- Ventricular hypertrophy: 2
- Cardiomegaly in: 3
- Others: 3
- Echogenic mass on LV
- Mild PR with dilated PA
- Pericardial effusion.

Outcome

<table>
<thead>
<tr>
<th>Outcome</th>
<th>n=109</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Alive</td>
<td>84</td>
<td>77.1</td>
</tr>
<tr>
<td>Intra uterine death or Termination of pregnancy.</td>
<td>18</td>
<td>16.5</td>
</tr>
<tr>
<td>Neonatal death</td>
<td>6</td>
<td>5.5</td>
</tr>
<tr>
<td>Infant Death</td>
<td>1</td>
<td>0.9</td>
</tr>
</tbody>
</table>

Table 5: Outcome of all the cases who underwent FETAL ECHO

<table>
<thead>
<tr>
<th>CHD</th>
<th>N=35</th>
<th>Top/IUFD</th>
<th>NND</th>
<th>Inf. Death</th>
<th>Alive</th>
<th>LTFU</th>
<th>Not Delivered</th>
</tr>
</thead>
<tbody>
<tr>
<td>AV canal defect</td>
<td>5(14.3)</td>
<td>3</td>
<td>1</td>
<td>1</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Tetralogy of fallot</td>
<td>3(8.6)</td>
<td>1</td>
<td>1</td>
<td>1</td>
<td>1</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Hypoplastic left heart syndrome</td>
<td>1(2.9)</td>
<td>1</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Ebsteins Anomaly</td>
<td>1(2.9)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Tricuspid atresia</td>
<td>5(14.3)</td>
<td>2</td>
<td>1</td>
<td>2</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Arrhythmias</td>
<td>5(14.3)</td>
<td>2</td>
<td>1</td>
<td>1</td>
<td>1</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Dilated cardio-myopathy</td>
<td>1(2.9)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Others **</td>
<td>18(51.4)</td>
<td>3</td>
<td></td>
<td>7</td>
<td>7</td>
<td>1</td>
<td></td>
</tr>
</tbody>
</table>

Table 6: outcome in various diagnostic categories

* One NND of the patient with Ebsteins anomaly was due to Klebsiella sepsis and was not related to the cardiac condition.
** Others category include simple lesions – ASD/VSD, functional abnormalities found like RA/RV overload, cardiomegaly, chamber hypertrophy, pericardial effusion, Echogenic mass on Left ventricle and mild PR with dilated PA.

**DISCUSSION:** Prenatal detection of congenital heart disease has become possible now with the evolution of fetal echocardiography. The reliability and accuracy of this procedure has been confirmed and is now becoming essential part of pediatric cardiology practice.10-12

The total mortality of live born babies with Congenital Heart disease is around 17% in the first year and about 1/3 of infant deaths from congenital malformations are related to cardiovascular anomalies. These account for a significant proportion of deaths, hence improving detection rates for these problems is important13

Fetal cardiac anatomy is analyzed echocardiographically using established principles of sequential segmental structural analysis. It is a unique non-invasive method for imaging, easy to perform with adequate training and with the advent of high resolution machines it is possible for earlier detection of congenital heart disease in the fetus. It has been suggested that prenatal detection of cardiac malformation not only aids in parental counseling, but also ensures optimal care for the infant at, and following delivery. In addition, with the earlier diagnosis of complex congenital heart disease such as Hypoplastic left heart syndrome and termination of pregnancy as parental choice the incidence of detecting these lesions postnatally is decreasing as has been demonstrated in some studies14-17.

Out of 170 fetal Echo performed 35 scans were abnormal and 135 were normal. This amounts to an incidence of 20.6% of abnormalities detected among referred patients.

The overall positivity rate in a study by Allan Brick was 25% conducted in 1999 in New York.18

**EXTRACARDIAC ANOMALIES:** 15 patients (8.8%) among who underwent Fetal ECHO had extracardiac anomalies. Congenital diaphragmatic hernia in 3, renal anomalies in 4, single umbilical artery in 3, duodenal atresia, Cystic hygroma, Dandy-walker Cyst, Sacral meningomyelocele and Gastrochisis in 1 each. 5 patients (33%) had associated cardiac abnormality. The association of extra cardiac anomaly with congenital heart disease reported in Literature was 32% by Smythe et al in Yale fetal series and 16% by Allan L.D. et al.

The higher incidence of extracardiac anomalies associated with congenital heart disease in present study could be due to the fact that study was conducted in a referral institute where all high-risk mothers were referred for Level II malformation scan, and if an extracardiac anomaly was detected they were referred for Fetal Echo to rule out associated congenital heart disease.

**CARDIOVASCULAR LESIONS DETECTED ANTENATALLY:** There were a total of 35 abnormal scans detected among 170 fetal Echo performed. (20.6%)

24 were structural heart disease, 9 had functional abnormalities and 5 had arrhythmia. In 3 patients there was more than one category of abnormalities AV Canal + Complete heart block in 2, Perimembranous-VSD + Complete heart block in 1.
Higher incidence of VSD and TA was found as compared to other series, and relatively lower incidence of HLHS. On analyzing the post natal scans 3 with detected small VSDs were normal. The lower incidence of HLHS could be due to late referrals in present study as there is spontaneous abortion in 1st and 2nd trimester in pregnancies with HLHS. This needs to be analyzed in more detail with further analysis of 1st and 2nd trimester abortions.

Functional abnormalities which were detected include 3 patients with RA/ RV overload, Ventricular hypertrophy in 2, cardiomegaly in 3 and Echogenic mass on LV, pericardial effusion, mild PR with dilated PA in 1 each. The functional abnormalities were noted in mothers with complicated pregnancies and placental insufficiency. Postnatal scans were normal in most of them except 1 patient with dilated cardiomyopathy with LV dysfunction who had early neonatal death.

The analysis of AV dysfunction could be the key to fetal cardiac dynamics. If a dysfunction is found careful periodic antenatal monitoring of fetal wellbeing needs to be done.

Unexplained RA/RV pressure overload with TR was noted without any hemodynamically important Cardiac anomaly by Sharland G.K. et al which was suspected to be due to mild coarctation of Aorta or a developing future ASD. 2 patients with cardiomegaly in present study had normal LV function and 1 patient with dilated cardiomyopathy had LV dysfunction and was suspected to have fetal myocarditis.

Arrhythmia was found in 5 patients out of 35 abnormal scans (14%) which are higher as compared to the earlier studies. Italian series (Fesslova et al) reported arrhythmia in 4.8% of cases (41/850 cases). In the Yale series by Smythe arrhythmia was found in 2.3% of cases. The reported incidence of arrhythmias in literature 1-10%\(^1^9\). In the present study arrhythmia associated with structural heart disease was also included hence relatively higher incidence was found compared to other series.

One patient in which fetus had Hypoplastic left heart syndrome did not survive till postnatally when P.G. infusion was planned. Though the outcome is poor in all the centers in India for want of expertise and resources, this category of patients is thought to benefit from prenatal diagnosis, considering the opportunity of a planned birth in qualified centers and reduction of diagnostic delay after birth.

In Italian (Fesslova) series termination rate was 29%\(^2^0\) and 55% in a study by Allan L.D. et al. This decision is strongly related to gestational age at diagnosis, which was late in present study with a mean of 28 weeks of gestation when first fetal Echo was performed.
Among the continuing pregnancies there was a 45% survival in Italian (Fesslova) Series, 41% series by Allan L.D. el al and 38% in series by Smythe et al.

In the present study 34% survival was noted following prenatal detection of CHD. Among the 9 survivors are 4 patients with Ventricular septal defect, 1 patient with OS- ASD, 2 with Tricuspid atresia, 1 with Tetralogy of fallot and 1 patient with Tachyarrhythmia.

CONCLUSIONS:

1. All ranges of congenital heart diseases can be easily diagnosed prenatally by fetal echocardiography.
2. Outcome of a prenatally detected complex congenital heart disease is poor; nonetheless earlier detection provides an opportunity for early interventions if available and helps in optimizing intranatal and postnatal care.
3. Fetal echo is highly sensitive and specific in detecting the complex congenital heart diseases.

Thus it is imperative that all mothers irrespective of risk factors for congenital heart disease should be screened during routine antenatal obstetric scan. Training the sonographers for identifying the 4 or 5 chamber view would be not only cost effective but also improve the yield of detection.

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