FETAL ECHOCARDIOGRAPHY: A STUDY OF CLINICAL OUTCOME

Rajanish K.V[1], S.S. Prabhu[2]

HOW TO CITE THIS ARTICLE:
Rajanish K.V, S.S. Prabhu. "Fetal Echocardiography: A Study of Clinical Outcome". Journal of Evolution of Medical and Dental Sciences 2014; Vol. 3, Issue 01, January 06; Page: 68-76.

ABSTRACT: BACKGROUND: Structural abnormalities of the heart and great vessels are fairly common congenital lab normalities with the incidenceof8 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 orarrhythmia. **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 analysis84 (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 prenatally 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 therapy[3-5.]

With termination of pregnancy being one of the options provided to parents of fetus with severe congenital Heart diseasedetected 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[6, 7]. 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[8].

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 ofFetal Cardiology[9].

Despitethe advances in thewest there are only fewcenters in India doingfetal echo andmanagement of fetus thereof.No database or registry exists in India to give an actualinsight into the problem of congenitalHeart disease andtheir prenataldiagnosis.

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 inmothers referred with high risk for congenitalheart disease.
2. To assess the outcome of prenatally detected congenital heart disease and to note the fetal and neonatal survival.

MATERIALSANDMETHODS: 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 standardrisk factors were gestational, familial or fetal.

A total of170antenatalmothers were referred for fetal echo, of whom48 were lost to follow up and13 have not delivered.

Studyperiod was 1 year.

INCLUSION CRITERIA WERE:

i. Maternal metabolic risk factors
   Diabetes
   Phenylketonuria
   Homocystinuria
ii. Exposure to Cardiac teratogens.
   Lithium Carbonate
   Folate antagonist
   Alcohol
   Anticonvulsants
   Maternalinfections
iii. Maternal or paternal congenital heart disease.
iv. Congenital heart disease in previous sibling.

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 wasdone 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.Insimple 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 togestational age at which the fetal echo was done. 48.7% ofmothers were between 21-28 wks. followed by 24.7% in 29-32 weeks.

Gest. Age	n = 170	Percentage
16-20 week	19	11.1
21-24 week	45	26.4
25-28 week	38	22.3
29-32 week	42	24.7
33-36 week	19	11.1
> 37 week	7	4.1

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.

Antenatal USG	n	%
Abnormal 4-CH View	41	24.1
Hydrops	11	6.4
Extracardiac Anomalies	15	8.8
Arrhythmia	11	6.4
Polyhydramnios	5	2.9
Others	2	1.2

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
Gastroschisis 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. Previouschildwith CHD and abnormal level I scan were predominant reasons for referral with 30%and 24.1%.

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

Table 3

In 17 patients (10%) therewas 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	n= 170	%
Normal	135	79.4
Large Atrial septal defect.	2	1.2
Ventricular septal defect.	7	4.1
AV Canal defect	5	2.9
Tetralogy of Fallot	3	1.8
Hypoplastic left heart syndrome.	1	0.6
Ebsteins anomaly	1	0.6
Tricuspid atresia	5	2.9
Arrhythmia	5	2.9
Others	11	6.5

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.

Out come	n=109	%
Alive	84	77.1
Intra uterine death or Termination of pregnancy.	18	16.5
Neonatal death.	6	5.5
Infant Death	1	0.9

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

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

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 important[13]

Fetal cardiac anatomy is analyzedechocardiographically using established principles of sequential segmental structural analysis.It is a unique non-invasivemethod 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 cardiacmalformation 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 suchas Hypoplastic left heart syndrome and termination of pregnancy as parental choice the incidence of detecting these lesions postnatallyis decreasing as has been demonstrated in some studies[14-17].

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 Gastroschisis 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 Echocardiographyperformed. (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 blockin 2, Perimembranous-VSD + Complete heart block in 1.

CHD	Present study	Allan et al	Fesslova	Smythe
AV canal defect	14.2	17.5	16	22
Tetralogy of fallot.	8.6	3	7.8	8.2
Hypoplastic left heart syndrome	2.9	16	14	13
Ebsteins anomaly	2.9	7	16	4.1
Tricuspid atresia	14.3	4	4	2.3
Ventricular septal defect	20	5	12	8.8
Large Atrial septal defect	5.7	-	3.6	1.2

Percentages of malformations found in present study in comparison to major fetal series reported.

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 smallVSDs 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 detailwith 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 alwhich 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 cardiomyopathyhad LV dysfunction and wassuspected 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%[19]. In the present study arrhythmia associatedwith 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%[20] 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 withTetralogy of fallot and 1 patient with Tachyarrhythmia.

CONCLUSIONS:

1. All ranges of congenital heart diseases can be easilydiagnosedprenatally byfetal 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 intranatalandpostnatal 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 screenedduring routine antenatal obstetric scan.Training thesonographers foridentifying the 4 or 5chamber view wouldbe notonlycosteffectivebut also improve the yield ofdetection.

REFERENCES:

1. Hernandeg – Diag S et al.Risk of birth defects during pregnancy N.Engl J. Med 2000; 343: 1608-1614.
2. Outcome of prenatally Diagnosed congenital heart disease an update. Brick D.H., Allan L.D.; Pediatric Cardiology 23: 449-453; 2002.
3. Achiron R, Glaser J. Gelernter I, et al. Extended fetal echocardiographic Examination for detecting Cardiac malformation in low risk pregnancies. BMJ 1992; 304: 671-674.
4. OttWj. The accuracy of antenatal fetal echocardiography screening in high – and low-risk patients. Am J ObstetGynecol 1995; 72: 1741-1749.
5. Sharland G. changing impact of fetal diagnosis of congenital Heart disease. Arch. Dis. Child 1997; 77: F1- F3.
6. Eronen M. outcome of fetuses with heart disease diagnosed in utero. Arch Dis. Child 1997: 77: F41-F46.
7. Hoffman JI. Congenital heart disease: incidence and inheritance ped. Clin North Am. 1990;37: 25-43.
8. Sharland G.K., Lockhart SM, Chitask et al. Force influencing the outcome of congenital heart disease detected prenatally. Arch. Dis. Child 1990; 65: 284-287.
9. Buskens E, Grobbe DE, Frohm- Mulder I M et al. Efficacy of routine fetal ultrasound screening for Cong. Heart disease in normal pregnancy. Circulation 1996;94: 67-72.
10. Shi C, Song, Li Y et al. value of four chamber view of the fetal echocardiography for prenatal diagnosis of congenital heart disease. Prenatal Diagnosis 1997 October (10) 901 – 6.
11. Bull C. Current and potential impact of fetal diagnosis on prevalence and spectrum of serious congenital heart disease at term in the UK. Lancet 1999: 354: 1242-1247.
12. Burn J. Brennan p, Little J, et al.Recurrence risks in offspring of adults with major heart defects: results from first cohort of British collaborative study. Lancet 1998: 351: 311-316.

13. Payne RM, Johnson MC et al, Toward a molecular understand of congenital Heart disease. Circulation 1995; 91: 494-504.
14. Gelb BD. Genetic basis of Syndromes associated with Cong.Heart disease Curropincardiol. 2001: 16: 188-194.
15. Gold Muntg E. The Epidemiology and genetics of congenital Heart Disease Clin. perinatal 2001; 28: 1-10.
16. Smythe JF et al. Outcome of prenatally detected Cardiac malformation Am J. cardiol 1992: 69: 1471-4
17. Michels V V, Riccardi VM. Congenital Heart defects. in principles and practices of medical Genetics, 2nd Ed. Churchill livingstone; 1990: 1207-237.
18. Allan L. D. et al.Prospective diagnosis of 1, 006 consecutive cases of congenital Heart disease in the fetus. J. Am. Coll. Cardiol 1994; 23: 1452-8.
19. Montana E et al. Trends and outcomes after prenatal diagnosis of congenital cardiac malformation by fetal echocardiography in a well-defined birth population, Atlanta, Georgia, 1990-1994. JACC 1996; 28: 1805-9.
20. FesslovaV,et al.Evolution and longterm outcome in cases with fetal diagnosis of congenital heart disease :Italian multicentre study.Heart .1999 nov;82(5):594-9

AUTHORS:

1. Rajanish K.V.
2. S.S. Prabhu

PARTICULARS OF CONTRIBUTORS:

1. Associate Professor, Department of Paediatrics, RRMCH, Bangalore.
2. Professor and HOD, Department of Paediatrics, B.J. Wadia Hospital for Children, Parel, Mumbai.

NAME ADDRESS EMAIL ID OF THE CORRESPONDING AUTHOR:

Dr.Rajanish K.V.,
#42, Anagha, 5th Block,
34th Main, BhavaniHsg Soc. Layout,
Banagirinagara, BSK 3rd Stage,
Bangalore – 560085.
Email- rajanish.kv@gmail.com

Date of Submission: 14/12/2013.
Date of Peer Review: 16/12/2013.
Date of Acceptance: 20/12/2013.
Date of Publishing: 01/01/2014