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RESEARCH ARTICLE

ROLE OF FETAL ECHOCARDIOGRAPHY IN THE DETECTION OF CONGENITAL HEART DISEASE- A HOSPITAL BASED STUDY

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ABSTRACT

Background and Objectives: Congenital heart disease is a leading cause of infant morbidity and mortality from birth defects with an estimated incidence of 6-12 per 1000 live births. The objectives of the study are to determine the prevalence of congenital cardiac anomalies in and around Cuddalore district, to ascertain the importance of four chamber view and Outflow tract views in the evaluation of congenital cardiac anomalies, to evaluate the efficacy and reliability of fetal echocardiography as a diagnostic tool in the detection of congenital heart disease and **t**o follow up and evaluate the outcome of fetuses diagnosed with congenital heart disease.

Materials and Methods: Fetal echocardiography was done as a hospital based prospective study in randomly selected 500 antenatal women referred for routine antenatal ultrasound for a period of 2 years.

Results: The prevalence of congenital heart disease by fetal echocardiography was 2.2%.Out of the 11 cardiac anomalies, 7 cases of structural cardiac anomalies were identified and 4 were non structural defects. We did not find any rhythm disorder in our study. Endocardial cushion defect was the most common cardiac anomaly in our study followed by echogenic cardiac focus. Majority of the antenatal women with cardiac anomalies were in the gestational age of 28-32 weeks. Majority of the antenatal women with cardiac anomalies did not have any high risk factors and extracardiac anomaly. Out of 11 cases with cardiac anomalies, 6 were terminated, 3 cases were delivered by vaginal delivery and 2 cases underwent LSCS. 2 cases were found to be normal and 3 cases were confirmed with cardiac anomaly on postnatal echocardiography. Additionally 3 cases were found to have cardiac anomaly on postnatal echocardiography. The sensitivity of Fetal echocardiography for cardiac anomalies was 75%, specificity was 99.59%, the positive predictive value was 81.8%, and the negative predictive value was 99.38% with an accuracy rate of 99%.

Conclusion: Routine fetal cardiac ultrasound using four chamber and outflow-tract views enables the detection and characterization of most of the cardiac anomalies. With moderate sensitivity and high specificity, Fetal echocardiography is a reliable prenatal diagnostic tool for cardiovascular problems with high accuracy. Timely diagnosis of cardiac anomalies allows the family to choose whether to continue pregnancy or terminate it. Since most cardiac anomalies occur without any risk factor, routine prenatal ultrasound almost misses an isolated cardiac anomaly, hence routine fetal echo in all antenatal cases must be done to rule out Congenital heart disease.

INTRODUCTION

Fetal echocardiography is defined as a detailed sonographic evaluation that is used to identify and characterize fetal cardiac anomalies prenatally[1]. Congenital heart disease is a leading cause of infant morbidity and mortality from birth defects with an estimated incidence of 6-12 per 1000 live births[2-3]

Prenatal diagnosis of congenital heart disease (CHD) has been shown to have a significant effect on prenatal and postnatal management and outcomes. In addition to the potential medical benefits, fetal diagnosis allows for valuable parental counseling, which allows families to make informed decisions regarding the pregnancy, and to prepare emotionally for the birth of the child with significant CHD. Accurate prenatal diagnosis can also lead to additional testing of the fetus, including genetic evaluation and other anatomic imaging, which can yield valuable information in overall assessment of the fetus[5]. Early knowledge of CHD also allows further

monitoring, testing for known associated non-cardiac structural and chromosomal anomalies and parental counselling about pregnancy management options including termination.

With advances in ultrasound technology and high-resolution imaging, fetal echocardiography can now be performed in the late first trimester and early second trimester of pregnancy around 18-22 weeks. Accurate prenatal diagnosis offers potential clinical benefit with regard to infant outcome. Prenatal detection accuracy have varied widely for CHD. Some of this variation can be attributed to examiner experience, maternal obesity, transducer frequency, abdominal scars, gestational age, amniotic fluid volume, and fetal position[6].

Various methods of antenatal ultrasound assessment of fetal heart are currently available. The 'four-chamber view' is a basic assessment which allows a general examination of the main structure of the heart and the atrioventricular junctions. 'Basic fetal echocardiography' is a more extensive antenatal ultrasound examination of the fetal heart and its associated structures, through additional assessment of the ventricular outflow tracts. 'Extended fetal echocardiography' involves two-dimensional scanning of the heart and its associated structures which is supplemented by spectral and colour-flow Doppler to assess blood flow within the heart[7].With this background, our study was conducted to evaluate the role of fetal echocardiography to detect congenital heart disease.

MATERIALS AND METHODS

The study included 500 antenatal women referred for routine antenatal ultrasound from the Department of Obstetrics and Gynecology during October 2014 till September 2016 who meet the inclusion criteria. The cases were selected randomly. The study was conducted in Department of Radiodiagnosis, Rajah Muthiah Medical College & Hospital, Annamalainagar, Chidambaram. All cases were done with Real time Grey Scale ultrasound by using Siemens Syngo Antares and Siemens Acuson X300 Ultrasound machine done with 2-6 Mhz convex probe and Philips Envisor machine with 5Mhz convex probe. Fetal echocardiography was performed in fetal echo settings.

Study design

A Hospital Based Prospective Study of Antenatal Women.

Selection criteria

Inclusion criteria- Antenatal women of gestational age from 18 weeks to 40 weeks of gestation referred for routine antenatal ultrasound with special emphasis on high risk cases like congenital heart disease in mother, family history of congenital heart disease, previous child with Congenital heart defects, Maternal Diabetes Mellitus, Mother on anti epileptic drugs, Maternal Collagen diseases, Increased nuchal Translucency in Early trimester ultrasound, polyhydramnios, Oligohydramios, IUGR, fetus with extracardiac anomalies etc.. All fetuses with or without known congenital anomalies are included in this study.

Exclusion Criteria- Antenatal women less than 18 weeks of gestational age due to lack of proper visualization of the fetal heart.

Procedure of study

A detailed clinical history is obtained. After obtaining informed written consent, obstetric ultrasound is done followed by detailed fetal echocardiographic study in fetal echo settings. If fetal heart is not properly visualized due to baby's position, mother is made to wait till satisfactory image is obtained.

- A detailed fetal echocardiographic study includes the following[1,16]

Situs and general aspects

- Fetal laterality (identify right and left sides of fetus)
- Stomach and heart on left
- Heart occupies a third of thoracic area
- Majority of heart in left chest
- Cardiac axis (apex) points to left by 45 +/-20
- Four chambers present
- Regular cardiac rhythm
- No pericardial effusion

Four Chamber View

- *Atrial chambers* - Two atria, approximately equal in size, Foramen ovale flap in left atrium, Atrial septum primum present (near to crux), Pulmonary veins entering left atrium.
- *Ventricular chambers* - Two ventricles, approximately equal in size, No ventricular wall hypertrophy, Moderator band at right ventricular apex, Ventricular septum intact (apex to crux).
- *Atrioventricular junction and valves* - Intact cardiac crux, Two atrioventricular valves open and move freely, Differential off setting: tricuspid valve leaflet inserts on ventricular septum closer to cardiac apex than does mitral valve.

Left Ventricular Outflow Tract
Right Ventricular Outflow Tract
Three Vessel View and Three Vessel Trachea View
Ductal and Aortic Arch View
M-Mode and colour Doppler if necessary

All the cases with and without cardiac anomaly were followed up postnatally with echocardiogram to confirm the presence of cardiac anomaly.

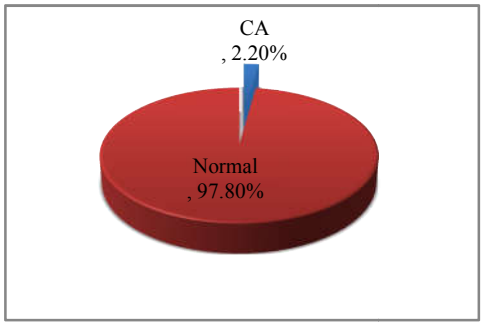
RESULTS AND DISCUSSION

Fetal echocardiography was performed in 500 antenatal cases and cardiac anomaly was found positive in 11 cases. Data was analysed and results were tabulated.

Majority of the antenatal women in the age group of 26 - 30 years had cardiac anomalies. Majority of the antenatal women with cardiac anomalies were in the gestational age of 28-32 weeks. Majority of cardiac anomalies were seen in multiparous women when compared with primigravida.

Prevalence of Cardiac Anomaly in Study Population

The incidence and prevalence of congenital heart diseases vary according to the investigation methods[13,17], i.e. from four to eleven cases per 1000 live borns [Allan L.D., 2000; Buskens E. et al., 1996; McGahan J.P., 1991; Stumpfen I. et al., 1996]. In our study, we found the prevalence of congenital heart disease by fetal echocardiography was 2.2%.



Distribution of Cardiac Anomalies

Endocardial cushion defect was the most common cardiac anomaly in our study found in 3 cases, followed by echogenic cardiac focus in 2 cases.

Cardiac Anomaly	No. of patients	Percentage
Echogenic cardiac focus	2	0.4
Endocardial cushion defect	3	0.6
AS with MR	1	0.2
Tetralogy of fallot	1	0.2
Ventricular septal defect	1	0.2
Hypoplastic left heart syndrome	1	0.2
Cardiomegaly	1	0.2
Coarctation of aorta	1	0.2

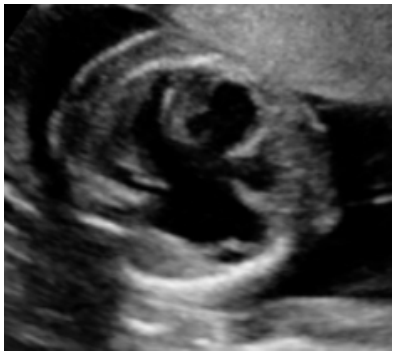


Fig.1 Showing Endocardial Cushion Defect As Defect In Crux.

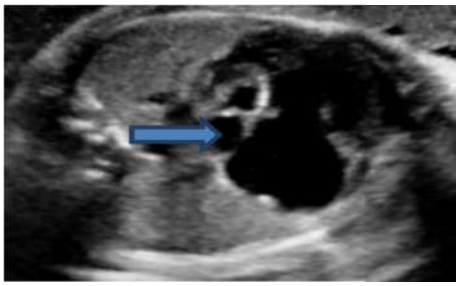


Fig.2 Shows Hypoplastic Left Heart Syndrome. Arrow Shows Small Left Ventricle and Left Atrium.

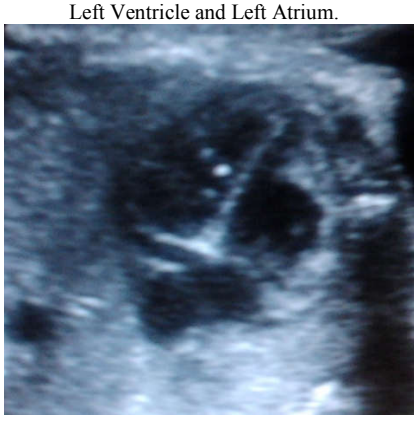


Fig.3 Showing Echogenic Focus in Left Ventricle

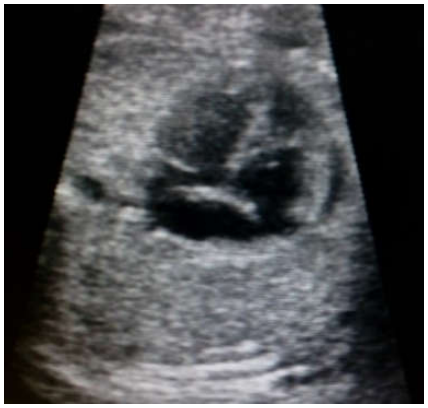


Fig. 4 Shows Membranous Ventricular Septal Defect

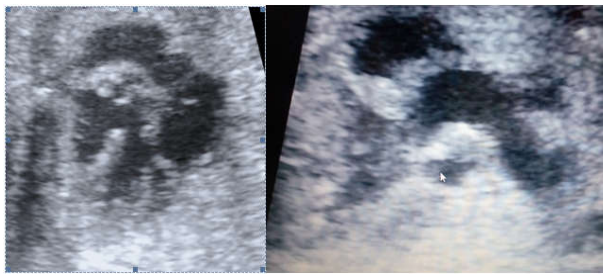


Fig.5 Tetralogy of Fallot. It Shows Membranous VSD with over Riding of Aorta and Pulmonary Stenosis (arrow)

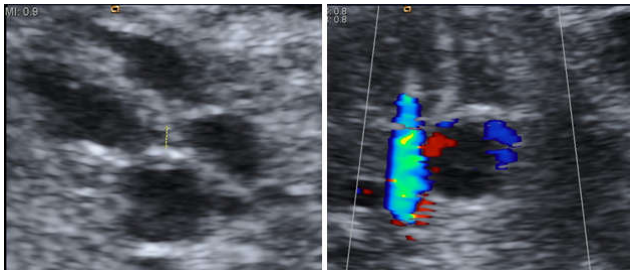


Fig.6 Aortic Stenosis with Mitral Regurgitation. There is Narrowed Lvot With Regurgitation of Flow Across Mitral Valve In Colour Doppler.

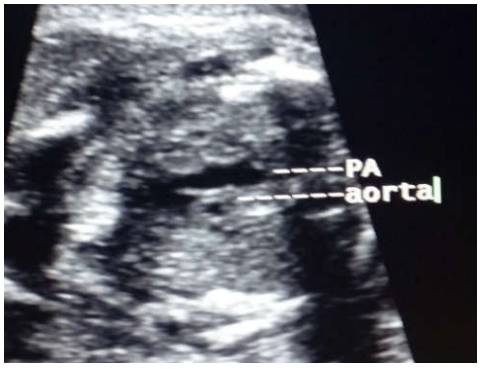


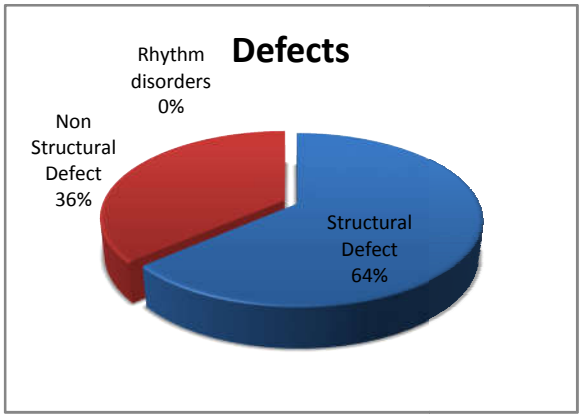
Fig.7 Coarctation of Aorta. It Shows Small Left Venticle on Four Chamber View. Three Vessel View Shows Narrowing Of Aorta with Left Sided SVC.

Distribution of cardiac defects

Vita Zidere[18] in his study found structural heart defects in 13.3%, non structural defects in 14.5% and heart rhythm disorders in 12.1%.

The common structural heart diseases are Atrioventricular septal defect, Aortic stenosis, Double outlet right ventricle, Ebstein's anomaly, Hypoplastic left heart syndrome, Tetralogy of fallot, Tricuspid atresia, Tricuspid valve dysplasia, VSD, Tricuspid and pulmonary valve stenosis, TGA, COA, Anomalous coronary arteries , Pulmonary stenosis and Truncus arteriosus coiminis. The Non structural heart pathologies includes Aortic regurgitation, aortic stenosis, Hypertrophic cardiomyopathy, Dilation cardiomyopathy, Left sided SVC, Mitral regurgitation, Tricuspid regurgitation, Situs inversus and Echogenic cardiac focus. Heart rhythm disorders includes premature contractions, Sustained bradycardia, Tachycardia and AV block.

In our study, 7 cases of structural cardiac anomalies were identified and 4 were non structural defects. We did not find any rhythm disorder in our study.



High risk factor distribution in relation to cardiac anomaly:

High risk history	Cardiac Anomaly				Total		P value
	No		Yes				
	N	%	N	%	N	%	
CHD in mother	1	0.20	1	9.09	2	0.4	0.000
GDM	13	2.66	1	9.09	14	2.8	0.201
IUGR	5	1.02	2	18.18	7	1.4	0.000
Mother on AED	4	0.82	0	0.00	4	0.8	0.763
Oligohydramnios	22	4.50	3	27.27	25	5	0.001
PIH	7	1.43	0	0.00	7	1.4	0.689
Polyhydramnios	5	1.02	0	0.00	5	1	0.736
Twins	1	0.20	0	0.00	1	0.2	0.881
No risk factor	432	88.34	4	36.36	436	87.2	0.000

Nuruddin Badruddin Mohammed et al[9] and Ingrid Stumpflen et al[12] studied that majority of pregnancies in fetus with cardiac anomaly are at no increased risk and routine prenatal ultrasound almost misses an isolated cardiac anomaly, hence routine fetal echo must be done to rule out Congenital heart disease.

In our study, out of 11 cardiac anomaly cases, 4 cases(36.6%) did not have any high risk factor followed by Oligohydramnios in 3 cases(27.27%)Mother with congenital heart disease, IUGR and Oligohydramnios were the risk factors found to have significant difference ($p<0.01$)between patients with and without cardiac anomaly. Patients without any risk factors was also found to have statistically significant difference ($p<0.01$).

Extracardiac Anomaly Distribution In Relation To Cardiac Anomaly

Extracardiac Anomaly	Cardiac Anomaly				Total		P value
	No		Yes				
	N	%	N	%	N	%	
Anencephaly	2	0.41	0	0.00	2	0.4	0.832
CTEV	1	0.20	1	9.09	2	0.4	0.000
Cystic Hygroma	1	0.20	0	0.00	1	0.2	0.881
Dandy Walker Anomaly	0	0.00	1	9.09	1	0.2	0.000
Renal Pyelectasis	0	0.00	2	18.18	2	0.4	0.000
Diaphragmatic Hernia	1	0.20	0	0.00	1	0.2	0.881
Fetal Ascites	1	0.20	1	9.09	2	0.4	0.000
Hydrops Fetalis	2	0.41	0	0.00	2	0.4	0.832
Posterior Urethral Valve	1	0.20	0	0.00	1	0.2	0.881
Single Umblical Artery	1	0.20	2	18.18	3	0.6	0.000
Spina Bifida	1	0.20	0	0.00	1	0.2	0.881
Noextracardiac anomaly	478	97.75	7	63.64	485	97	0.000

Rosana Cardosa M. Rosa et al[8] found out that extra cardiac anomalies are frequent among patients with CHD and these patients present with increased risk of morbidity and mortality. In our study, 7 cases with cardiac anomalies did not have any extracardiac anomaly and are statistically significant ($p<0.01$). Most common extracardiac anomalies associated with the presence of cardiac anomaly are single umblical artery and renal pyelectasis(18.18% each). Extracardiac anomalies including single umblical artery, renal pyelectasis, CTEV, Dandy Walker anomaly and fetal ascites are found to have statistically significant difference between patients with and without cardiac anomaly ($p<0.01$).

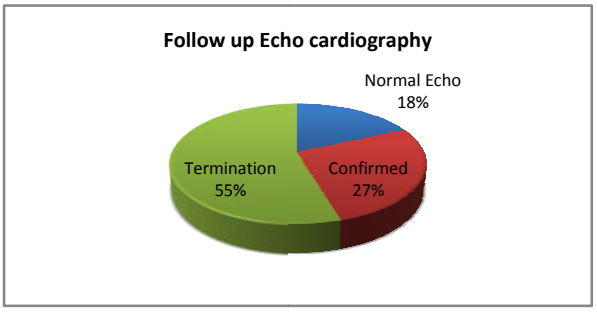
Mode of Delivery

In our study, 6 cases with cardiac anomalies were terminated, 3 cases were delivered by normal vaginal delivery and 2 cases underwent LSCS.

Postnatal Follow UP Echocardiography

Bakiler et al[10] in his fetal echo study of 197 women found 10 cases of CHD, whereas postnatal echo and post mortem examination revealed abnormalities in 21 patients.

In our study, out of 11 cases with cardiac anomalies, 6 cases underwent termination of pregnancy. Cardiac anomaly was confirmed in 3 cases and was found to be normal in 2 cases with follow up postnatal echocardiography. Additional 3 cases of cardiac anomaly was found in patients with normal fetal echocardiography in postnatal echocardiography.



Test Characteristics

Bakiler et al[10] found the sensitivity and specificity of fetal echo were 42 and 98% respectively. PPV was 90% and NPV was 93%. Ozkutlu. S et al[11] in his study found the sensitivity and specificity of fetal echo was 93.3% and 100% respectively.

In our study, we had 3 false negatives and 2 false positives. The sensitivity of Fetal echocardiography for cardiac anomalies was found to be 75%, specificity was found to be

99.59%, the positive predictive value was found to be 81.8%, and the negative predictive value was 99.38% with an accuracy rate of 99%.

Once an accurate diagnosis of prenatal CHD is made, the condition and its implications must be conveyed to the family with prenatal counselling. The aims of prenatal counselling are providing an accurate diagnosis of the malformation, providing a clear and truthful picture of prognosis, outlining management and treatment options that are available, and helping parents reach decisions concerning the form of management that is best for them[19,20].

CONCLUSION

The prevalence of congenital heart disease by fetal echocardiography was 2.2% in our study. A four-chamber view of the heart is sufficient enough to diagnose many cardiac anomalies. However, anomalies involving the outflow tracts of the aorta and pulmonary artery are not visualized on this view. By adding the base view of the heart, the outflow tracts can be visualized, thus increasing not only the sensitivity of detection of cardiac anomalies but also the accuracy of diagnosis. Routine fetal cardiac ultrasound using four chamber and outflow-tract views enables the detection and characterization of most of the cardiac anomalies. A further comprehensive evaluation can be performed with fetal echocardiography, particularly in high-risk pregnancies and extracardiac anomalies. Doppler imaging is used in the evaluation of vascular and valvular lesions. M-mode is used to detect any cardiac arrhythmias.

With moderate sensitivity and high specificity, Fetal echocardiography is a reliable prenatal diagnostic tool for cardiovascular problems with high accuracy and has an impact on the management at prenatal, natal and postnatal period. Timely diagnostics of fetal heart malformations allows the family to choose whether to continue pregnancy or terminate it. In our study - if heart diseases are diagnosed timely, the family decides not to continue pregnancy. In certain cases, patients can be referred to paediatric cardiologist for appropriate fetal medical therapy or fetal interventions. By identifying infants prior to birth, these high risk patients can be delivered in a center where they have ready access to a pediatric cardiologist and cardiovascular surgeons and hopefully a better outcome. Thus ,the families are better prepared and know more of what can be expected once the infant is born.

Majority of pregnancies in fetus with cardiac anomaly are at no increased risk and routine prenatal ultrasound almost misses an isolated cardiac anomaly, hence routine fetal echo in all antenatal cases must be done to rule out Congenital heart disease.

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