Original Article

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DETECTION OF CONGENITAL HEART DISEASE BY FETAL ECHOCARDIOGRAPHY

Afshan Fayyaz, *Syed Muhammad Imran Majeed, **Waqar Ahmed

Army Medical College Rawalpindi, Armed Forces Institute of Cardiology Rawalpindi*, AFIC and Kulsoom International Hospital Rawalpindi**

Objective: The objective of the study was to determine the sensitivity, specificity, accuracy and predictive value of fetal echocardiography in our set up using postnatal echocardiography as gold standard.

Study Design: Validation study.

Place and Duration of study: This is an ongoing study in the Radiology department of CMH Rawalpindi and Armed Forces Institute of Cardiology (AFIC) Rawalpindi and the data collected from January 2007 to Jan 2012 is presented.

Patients and Methods: Two hundred eighty seven patients reported for fetal echocardiography. Two hundred twenty nine patients were subsequently included in the study. These included patients of all ages who reported to the Radiology department of CMH Rawalpindi for fetal echocardiography. Fetal echo was done on Toshiba Aplio with 3.5 MHz probe having Doppler facility. Post natal evaluation was done by a pediatric cardiologist.

Results: There were 207 (90.4%) true negative cases, 15 (6.6%) true positive, 2 (0.9%) false positive and 6 (2.2%) false negative cases. The sensitivity, specificity, positive and negative predictive values were 75%, 99%, 88%, 97% respectively.

Conclusion: Fetal echocardiography has high specificity, negative predictive values and accuracy and cases diagnosed as normal can reassure the parents about the normal cardiac status of the fetus.

Keywords: Fetal Echocardiography, Fetal Anomalies, Post Natal Evaluation, Specificity, Accuracy.

INTRODUCTION

Congenital heart disease is the most common congenital anomaly affecting 8 of 1000 live births[1,2]. It is 6 times more common than chromosomal anomalies and 4 times more common than neural tube defects[3]. Fetal echocardiography serves as a diagnostic tool of relief for concerned parents who have a previously affected child. On the other hand, a fetus affected by congenital cardiac problem diagnosed in utero can have a delivery planned close to a cardiac care center where steps can be taken to avertthe adverse outcome of the neonate as it is believed that at least 50% of congenital cardiac defects are amenable to surgery[4]. Sensitivity and specificity of fetal echocardiography are claimed to be close to 100% in some studies. To our knowledge, no study has been published in local literature regarding sensitivity, specificity, accuracy and predictive value of fetal echocardiography.

Correspondence: Dr. Afshan Fayyaz, Consultant and Professor of Radiology, Army Medical College Rawalpindi.
Email: afshan_fayyaz@hotmail.com
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PATIENTS AND METHODS

This validation study was carried out in the Radiology department of CMH Rawalpindi and AFIC Rawalpindi from January 2007 to January 2012. All patients referred for fetal echocardiography were entertained. There was no fetal age constraint for fetal echocardiography. Fetal echocardiography was done by a single radiologist trained for the job. The examination was done on Toshiba Aplio with 3.5 MHz probe and Doppler facility. Pediatric cardiologist was involved in difficult cases. All structural cardiac defects were included in the study. These included determination of fetal situs, septal defects, anomalous arterial and venous connections, chamber abnormalities as single atrium/ventricle, hypoplastic left or right heart, valvular abnormalities, rhythm abnormalities and pericardial effusion either alone or in association with hydrops fetalis. Patent ductus arteriosus and atrial septal

defects were considered post natal diagnosis and neonates with these defects diagnosed postnatally were considered true negatives.

Post natal evaluation of the fetus was done by neonatal echocardiography and clinical evaluation by a pediatric cardiologist. A telephonic contact was maintained with all patients. Fetal echocardiographic examination began with exclusion of extra cardiac anomalies. A detailed anomaly scan was carried out. Fetal stomach bubble was identified and fetal situs was determined by identifying left and right sides of the fetus. The fetal cardiac evaluation included standard 4 chamber view. This excluded anomalies as single ventricle, hypoplastic left heart, ventricular septal defect, Ebstein anomaly and single atrium. The transducer was then rotated towards the base of the heart and anomalies as transposition of the great arteries, truncus arteriosus and over riding aorta were excluded. Pulmonary veins were evaluated by Doppler ultrasound examination and identification of a vein on right and left side behind the left atrium was considered sufficient for exclusion of total anomalous pulmonary venous drainage. IVC and SVC connections were seen in longitudinal axis of the fetus. Fetal data was fed on SPSS version 15. Pre natal evaluation of fetus was correlated with post natal diagnosis. Sensitivity, specificity, positive and negative predictive values and accuracy were calculated. ASD and PDA are post natal diagnoses and neonates with these conditions diagnosed postnatally were included in true negative cases

RESULTS

Two hundred eighty seven patients were evaluated by fetal echocardiography during this period. Out of these, 39 were lost to follow up. There were 11 neonatal deaths, 6IUDs and 2 still births. Two hundred twenty nine patients were subsequently included. Mean age of the mother at the time of fetal echocardiography was 28.21 ± 4.5 years with minimum age of 19 and maximum age of 42 years. Mean time of fetal ultrasound examination was 29.27 ± 4.752 weeks of pregnancy.

Description of fetal echo cardio graphy and post natal ecocardiography is given in table 1.

Sensitivity of fetal echo in our study is 95%, specificity is 99%, positive predictive value is 88%, negative predictive value is 98% while accuracy is 97%. Area under the curve is 0.884 ($p < 0.001$) fig 1.

The false negative cases were a case of situs inversus which went urecognized due to failure on the part of fetal echocardiographer to identify right and left sides of the fetus, 2 cases of small VSDs, which required medical management postnatally and a case of low ejection fraction. This went unrecognized as ejection fraction was not determined routinely during fetal echocardiography. A case of pulmonary stenosis was missed on fetal echocardiography although we gave indirect evidence of congenital heart disease to the concerned parents but as the accurate diagnosis was not reached this case was included as false negative case in our study.

False positive cases were a case of small pulmonary artery which was normal postnatally, and a case of small VSD which was normal postnatally.True positive cases are given in table 2.

Two cases of hypoplastic left heart syndrome died after birth. One case of congenital complete heart block was associated with complete AVSD. This child died after birth. A case of total anomalous pulmonary venous drainage died before surgery could be planned. Two cases of congenital complete heart block were associated with ascites and pericardial effusion and died after birth. The other 2 cases were managed medically.

Twelve (5.2%) fetuses died after birth. Out of these, 2 fetuses had cardiac anomalies of complete AVSD with bradycardia and TGA on fetal echocardiography. Extra cardiac abnormalities were seen in 5 of these fetuses. Two had diaphragmatic hernia, 1 had hydrocephalus, 1 had skeletal dysplasia, 1 had hydrops fetalis. Five fetuses were normal on fetal ultrasound and died of causes unrelated to the heart. These children could not have a post

Table-1: Description of fetal and postnatal echocardiography.

	Post natal echocardiography	
Fetal echocardiography	Abnormal	Normal
Abnormal	True positive 15 (6.6%)	False positive 2 (0. 9%)
Normal	False negative 5(2.6%)	True negative 207 (90.4%)

Table-2: Description of true positive cases (n=15).

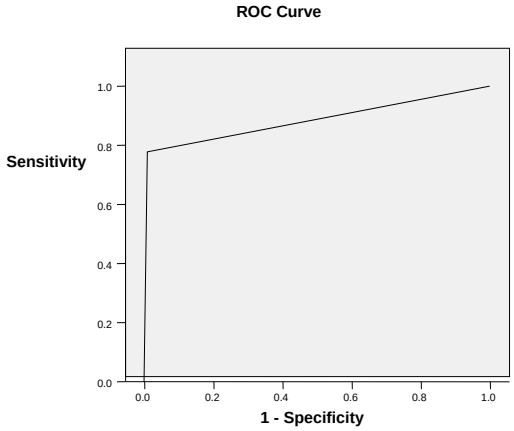
True positive cases	Number of cases
Ventricular septal defects	2 (13.3%)
Hypoplastic left heart syndrome	2 (13.3%)
Atrio ventricular septal defects	2 (13.3%)
Complete congenital heart block	3 (20%)
Double outlet left ventricle	1 (6.7%)
Pericardial effusion	3 (20%)
Total anomalous pulmonary venous drainage	1 (6.7%)
Complete AVSD with congenital complete heart block	1 (6.7%)

natal cardiac evaluation and were excluded from the study.

DISCUSSION

Fetal echocardiography is an important tool in the pre-natal fetal evaluation as congenital heart disease is the commonest congenital anomaly with a high post natal morbidity and mortality. It accounts for approximately 20% of neonatal deaths and 50% of infant deaths and is seen four to five times more frequently in stillbirths[1,5]. Prenatal diagnosis of congenital cardiac anomaly may entail delivery close to a cardiac care unit with facilities to handle the child. Late diagnosis postnatally carries a much greater morbidity than a fetus diagnosed prenatally with subsequent therapy instituted during the neonatal period[6-8]. As this examination carries a high sensitivity and specificity, parents with a previously affected child with congenital heart disease can carry on the pregnancy comfortably if the fetal echo examination is normal.

Before a fetal echocardiographic examination is carried out, fetal sites should be determined. The fetal orientation with respect to maternal abdomen is seen and the right and left side of the fetus is determined. The relationship of the fetal heart to the stomach bubble is seen. In cases of dextrocardia, the stomach bubble lies opposite to the cardiac axis. We missed a case of situs inversus in our study



which is a false negative case in our study.

The fetal cardiac examination begins with four chamber view of the heart. This view is taken in fetal cross section (fig 2). The cardiac chambers are evaluated in this view. The two ventricles should be of equal size. The two atria should also be equal. The interventricular septum should be intact. The interatrial septum should have the foramen ovale flap opening towards the left side. Pulmonary veins should be seen opening into the left atrium. Following the four chamber view, the base view of the fetal heart should be taken (fig 3). The normal crossing of the vessels should be seen. The pulmonary artery should be seen entirely. It should be seen to divide into the right pulmonary artery and the ductus arteriosus

Figure-1: ROC curve for fetal echocardiography using postnatal echo as gold standard (n= 229).

which is seen continuing into the descending aorta. The aorta is seen posteriorly. The anterior wall of the aorta is continuous with the interventricular septum and the posterior wall of the aorta is continuous with the mitral annulus. Due to unique fetal dynamics, the size of the pulmonary artery is larger than the aorta. Rotating the transducer further towards the fetal head shows the aortic arch with the arch vessels arising from it. The descending aorta is seen in the long axis of the fetal spine. IVC and SVC connections are identified in this view along the long axis of the fetal spine.

Fetal echocardiography yields best results when done in the second trimester of pregnancy[9]. At this stage, the liquor volume is good and fetal size is appropriate for better visualization of fetal cardiac structures . Now studies are being conducted for a fetal echo examination in the first trimester. These have shown a high accuracy when done early in pregnancy before 16 weeks[10]. In our study, fetal echocardiography was done on fetuses of all ages as patients were entertained as and when they came to the examination.

Total anomalous pulmonary venous drainage poses a challenge for the echocardiographer. Indirect signs as right predominance with dilated coronary sinus or SVC do not have significance as indirect markers of the abnormality and if this condition is clinically suspected should be sought on fetal echocardiography[11]. In our study, there were 2 cases of right predominance. One was a case of TAPVR. The other case was normal postnatally.

The importance of fetal echocardiography lies in the prenatal detection as well as post natal planning of the fetus. The delivery of fetus with significant cardiac defects detected prenatally can be planned close to a tertiary cardiac care center so that the neonate can benefit from immediate cardiac care. As congenital heart disease is the commonest condition to affect the newborn, with a mortality as great as 40% in the neonatal period[12]this condition requires diagnosis prenatally with subsequent post natal planning. According to Sharland, congenital cardiac

Figure-2: Fetal heart in cross section showing the four cardiac chambers.

Figure-3: Base view of the heart showing normal crossing of the vessels.

malformations are a cause of neonatal and intrauterine deaths in 30%[13].

Fetal echocardiography has been found to have varying sensitivity and specificity in different studies. Berghella V et al achieved a sensitivity of 95% and accuracy of 87%[14]. According to Bennasar M et al in their study a sensitivity of 90.9%, specificity of 96.2%, positive predictive value of 83.3%, negative predictive value of 98.1% and accuracy of 95.3% was achieved[10]. A specificity of 100% was seen in a systematic review by P Randall et al[15]. Nuruddin et al obtained a sensitivity of 85.4% for the detection of congenital heart disease, and a specificity of 99.9% to rule out such anomalies. Positive and negative predictive rates were 87.7% and 99.9%[16].

In our study, we achieved a specificity of 99% and a negative predictive value of 98% and accuracy of 96%.

CONCLUSION

Fetal echocardiography has high specificity, negative predictive value and accuracy and parents can be reassured of the normal cardiac status of the fetus when fetal echo is normal.

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