Indications of Fetal Echocardiography and Detection of Congenital Heart Disease Prenatally in Tertiary Care Hospital

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ABSTRACT

Background

Congenital Heart Diseases are common childhood congenital anomalies encountered in developed and developing countries. Due to the improved prenatal diagnostic modalities, the diagnosis of cardiac disease is increasing in newborns. Fetal echocardiography is considered a good and accurate diagnostic method for congenital heart disease.

Objective

To study the common indications of fetal echocardiography and the detection of congenital heart disease in Dhulikhel Hospital, Kathmandu University Hospital.

Method

In this hospital-based, retrospective study, we reviewed medical records of 324 fetuses who were indicated for fetal echocardiography at Dhulikhel Hospital over 24 months period of time (September 1, 2017 to August 31, 2019).

Result

The mean age of pregnant women at presentation for fetal echocardiography was 30.7 ± 4.7 years. The mean gestational age at diagnosis was 28.9 ± 4.9 weeks. Among all the pregnant women, 208 (65%) presented during the second trimester and 112 (35%) presented during the third trimester. Maternal indications for fetal echocardiography were seen in48.12% cases, whereas fetal indications were seen in 42.81% cases. The most common indication was for maternal gestational diabetes (30.31%). Among the 324fetal echocardiography results, 65.43% cases had normal findings, echogenic intra-cardiac foci were seen in 18.82% and significant abnormal findings were seen in 15.74% of cases. Among all women having maternal indication for echocardiography, 34 (22.07%) fetus had some form of abnormal findings, whereas only 17 (12.40%) fetus with fetal indication showed abnormal findings ($p < 0.05$).

Conclusion

Indication of fetal echocardiography is increasing and can be a good diagnostic tool to improve prognosis and outcome of a newborn. It has become widely used in pediatric cardiology. Raising awareness is necessary regarding the importance of fetal echocardiography in the management and outcome of a newborn with cardiac anomalies.

KEY WORDS

Congenital heart disease, Fetal echocardiography, Newborn

INTRODUCTION

Congenital heart disease (CHD) has emerged as a major public health problem and imposes an increasing burden on the health care system in Nepal. According to current scenario of heart diseases in Nepal on the basis of available studies, the prevalence of CHD is 1.3 per 1000 children.[1] CHD is one of the most common congenital anomalies, and approximately half of infant deaths are due to CHD.[2] Because early detection of CHD makes early medication, trans-catheter intervention, or even surgery possible in advanced centers, prenatal diagnosis is considered essential. Use of fetal echocardiography (FE) for prenatal diagnosis has increased since it was introduced in 1964.[3] Prenatal diagnosis with FE has improved the preoperative condition, morbidity, and mortality of patients with CHD. FE is now widely used in pediatric cardiology and perinatology and even for fetal cardiac intervention.[4] Many children die each year from CHD, while many more remain in desperate need of treatment in the developing world, CHD therefore significantly contribute to the economic burden on health care systems. Prenatal FE is the only means of detecting such defects, but little is known about the incidence and patterns of fetus with CHD encountered in Dhulikhel hospital.[5] As a tertiary referral center of the region, we aim to describe the common indications of FE and its usefulness in detecting cardiac abnormalities in fetal period presented in Dhulikhel hospital over a 24 months period of time (September 1, 2017 to August 31, 2019).

METHODS

This was a hospital-based, retrospective, cross-sectional, observational study conducted in department of pediatrics, Dhulikhel Hospital, Kathmandu University Hospital, Nepal. Dhulikhel hospital is the only tertiary care center in the surrounding area which is also a referral center in the region, it has a well-established pediatric unit treating children since birth, which can diagnose and manage pediatric cardiac cases and has facilities of FE.

We searched our database for FE performed at Dhulikhel Hospital from September 1, 2017 to August 31, 2019, and reviewed the medical records of these pregnant women and fetuses. Total of 324 FE were performed during this period.

The clinical data collected for the study included referral indications, gestational age, maternal age, referral personal and final diagnosis on echocardiography. Medical records for data collection were reviewed from FE record files. All detailed information of the fetus and their echocardiographic values were recorded.

This study was performed after obtaining approval of the Institutional Review Board of Kathmandu University School of Medical Sciences. The requirement for informed consent was waived because of the retrospective nature of the study.

Medical records of all pregnant women were reviewed who were indicated for FE in last 2 years at Dhulikhel Hospital. Gestational age was determined by last menstrual period or by ultrasound measurements of fetal biometrics. Gestational age was expressed in weeks. The first trimester was considered to last until the completion of 14 weeks. The second trimester lasted from the 15th week to the 28th completed week and the third trimester lasted from the 29th week to the 42nd completed week.[6] Maternal medical records were reviewed for genetic and other anatomic diagnoses of the fetus and indication for FE referral. Various referral indications included maternal CHD, family history of CHD, maternal diabetes, obstetrical scan suspicious for CHD, fetal arrhythmia, maternal cardiac disease, extra-cardiac congenital anomalies, and chromosomal anomaly. All FE was done by echocardiography machine (GE, Vivid 7) in department of pediatrics of Dhulikhel Hospital. GE 5S cardiac sector probe was used for recording FE. All the examination and echocardiography has been performed by a single examiner. FE was performed according to the American Society of Echocardiography guidelines.[7] In this study, two dimensional echocardiography, M mode echocardiography, color Doppler, and pulsed Doppler echocardiography were used. We evaluated cardiac position, situs, four-chamber view, five chamber view, short axis, ventricular out flow tract, aortic arch, rate, and rhythm. All pregnant women were informed to perform echocardiography of the baby after birth.

Statistical analysis was performed using the Statistical Package for Social Sciences (SPSS) software, version 20. Data analysis was done using both descriptive (mean, percentage and standard deviation) and inferential (χ^2 test) statistics. Linear regression was used to assess and describe the relationship between variables. Statistical test was considered significant at 0.05 level of confidence.

RESULTS

During the study period, total of 324 FE was performed in 320 pregnancies. There were 4 sets of twin pregnancy indicated for FE. The mean age of pregnant women presented for FE was 30.7 ± 4.7 years. (Range: 17 to 39 years). The mean gestational age on presentation was 28.9 ± 4.9 weeks (Range: 17 to 39 weeks). Among all the pregnant women, 208 (65%) presented during the second trimester and 112 (35%) presented during the third trimester. There were 6pregnant women who presented at term, just few days prior to delivery. No women presented before 17 weeks of gestation, as routine anomaly scan were normally performed between 18 to 22 weeks of gestation in the hospital.

Our results showed that 170 (53.12%) pregnant women were referred for FE by the gynecologist, whereas 121 (37.81%) women were referred by the radiologist after an anomaly scan, and 29 (9.06%) women came on their

own for screening purpose (fig. 1). Among all the referrals, 154 (48.12%) FE were performed as maternal indication (Table1), and 137 (42.81%) FE performed as fetal indication (Table2). The most common indication for FE was for maternal gestational diabetes (30.31%), followed by extra-cardiac anomaly (17.18%), echogenic intra-cardiac foci (13.43%), and bad obstetric history (8.12%).

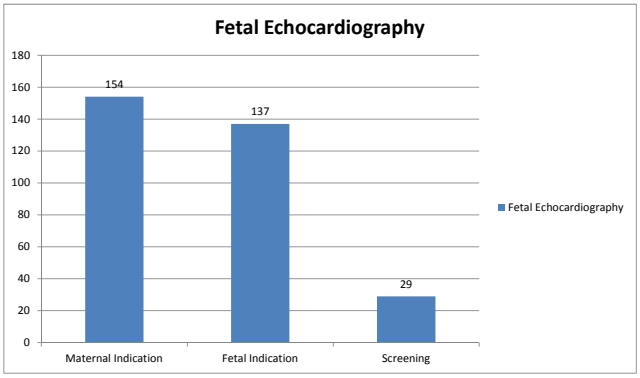

Figure 1. **Indications for fetal echocardiography**

Table 1. **Maternal indications for fetal echocardiography**

Maternal Indications	Number (%)
Gestational Diabetes Mellitus	97 (30.31)
Bad Obstetric History	26 (8.12)
Previous child with CHD	12 (3.75)
Hypothyroidism	10 (3.12)
Others	9 (2.81)

Others: eg, poor echo window, Trauma, IUGR, Oligohydromnios,

Table 2. **Fetal indications for fetal echocardiography**

Fetal Indications	Number (%)
Extra-cardiac anomaly	55 (17.18)
Echogenic intra-cardiac foci	43 (13.43)
Polyhydramnios	16 (5.0)
Anomaly scan suspicious of CHD	14 (4.37)
Fetal arrhythmia	5 (1.56)
Single umbilical artery	4 (1.25)

Among all the 324 fetuses, normal findings were seen in 212 (65.43%)FE whereas, echogenic intra-cardiac foci were seen in 61 (18.82%) cases and a total of 51 (15.74%) FE were reported to have significant abnormal findings. Out of 154 (48.12%) pregnant women having maternal indication for FE, 34 (22.07%) fetus had some form of abnormal findings, whereas only 17 (12.40%) fetus out of 137 (42.81%) women with fetal indication showed abnormal findings on FE ($p < 0.05$). The most common abnormal FE finding was isolated ventricle septal defect (18 cases), hypertrophic cardiomyopathy (16 cases), fetal arrhythmia (7 cases), suspected coarctation of aorta (2 cases), endocardial cushion defect (2 cases), Ebstein anomaly (2 cases) and truncus arteriosus (1 case) followed by other complex CHD (3 case) (Table 3). Postnatal Echocardiography was advised to all women having positive FE findings.

Table 3. **Findings in fetal echocardiography**

Echocardiographic findings	Numbers (%)
Echogenic intra-cardiac foci	61 (18.82)
Ventricle Septal Defect	18 (5.55)
Hypertrophic Cardiomyopathy	16 (4.93)
Fetal Arrhythmia	7 (2.16)
Suspected Coarctation of aorta	2 (0.61)
Endocardial Cushion Defect	2 (0.61)
Ebstein Anomaly	2 (0.61)
Truncus arteriosus	1 (0.30)
Heterotaxy syndrome with left isomerism with TGA with large ASD and VSD	1 (0.30)
DORV with TGA suspected Taussig-Bing anomaly	1 (0.30)
Heterotaxy syndrome with single ventricle with single AV valve with TGA	1 (0.30)

DISCUSSION

The incidence of CHD is about 6 in 1,000 live births and about 8 to 10 in 1,000 pregnancies.[5] CHDs are responsible for about 40% of perinatal deaths of which more than 20% of deaths occur in the first month of life.[5] According to a status report on CHD in India, 10% of the present infant mortality may be accounted for by CHD.[8] Therefore, CHDs significantly contribute to the economic burden on health care systems. In Nepal, as in many developing countries, perinatal programs are not fully established and epidemiological data on fetal disease are lacking. The rapid improvement made in other developed and developing countries in diagnostic options, surgical and interventional management of complex CHD have not been replicated in Nepal. Due to the limited antenatal screening for CHD, majority of CHD are not diagnosed before birth in developing countries. Early recognition of CHD will help to treat the child and if possible get corrective surgery done. Early detection has the potential to prevent progression of the disease. The detection of heart diseases early in childhood will surely lead to better treatment and reduction in the mortality and morbidity. CHD represents a large part of cardiovascular disease during the first years of life but little is known about the incidence and patterns of fetus with CHD encountered in Dhulikhel hospital. Therefore, this study aimed to assess the prevalence and patterns of fetal cardiac disease attending Dhulikhel hospital. We believe that this will help in the long term planning, care and follow up of children with CHD and to assess the indications, use, and yield of FE.

In addition to the potential medical benefits and timely interventions, 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.[9] Moreover, the noninvasive nature and wide availability makes it a good screening tool in evaluation of cardiac function and detection of congenital cardiac defects.[10] FE

has been established as the standard tool for diagnosing CHD in prenatal life.[11] Specificity and sensitivity of FE for the diagnosis of fetal heart defects were reported as 98% and 42%, respectively. The positive predictive value was found to be 90% and the negative predictive value was 93%.[12] In developing countries such as Nepal, the diagnostic and intervention facilities for CHD are very limited. Most of the females are unaware of the importance of FE, and they present late.

In this study, majority of pregnant women (65%) presented during the second trimester. Current guidelines suggest that FE be performed between 18 and 22 weeks gestation.[13] However, Becker and Wegner reported fetal echo between 11 weeks and 13 weeks.[6] Pike et al. found that the mean gestational age at presentation was between 18 weeks and 22 weeks.[9] In contrast with the aforementioned studies, our study showed that the mean gestational age on presentation was 28.9 ± 4.9 weeks (Range: 17 to 39 weeks). The reason for late presentation might be due to unavailability of FE in every hospitals and moreover the lack of awareness of proper timing of FE and their low educational status. When FE is considered necessary, it should be performed preferably between the first or early second trimester, as it is likely to provide a reasonable understanding of the fetal heart with a success rate of above 90% for visualizing the four-chamber and proximal outflow tracts in major referral centers.[6]

Our results showed that, 53.12% women were referred for FE by the gynecologist, whereas 37.81% of the women were referred by the radiologist after an anomaly scan and only 9.06% women came on their own for screening purpose, suggesting the lack of awareness in the general population regarding the importance of fetal detection and intervention in congenital cardiac anomalies. Interestingly, many studies shows that only about 10% of the CHDs are associated with obvious risk factors in pregnancy, such as extra-cardiac malformations, however most of the cardiac defects occur in the low-risk neonates without any risk factors in the pregnancy period.[14] Nearly 50% of the neonates with CHD do not have a definite risk factor. Most of the retrieved studies have suggested FE as a certain part of the prenatal screening performed in the second trimester. Accordingly, they recommended to implement this procedure for all pregnant women regardless of the presence of the risk factors.[13] Indications of FE primarily included parents with history of CHD, parents with a previous child with CHD, an anomaly scan with suspicion of CHD, fetal arrhythmia, extra-cardiac anomalies, chromosomal anomalies, presence of polyhydramnios or oligohydramnios, maternal diabetes, and history of maternal exposure to certain medications.[15] Among these, chromosomal anomalies and suspicious obstetrical scan are considered high yield indications, whereas family history of CHD and teratogen exposure are low yield indications.[16] Results of our study showed, the most common indication for FE was for maternal gestational diabetes (30.31%), followed by extra-cardiac anomaly (17.18%), echogenic intra-cardiac foci (13.43%) andbad obstetric history (8.12%). The distribution of indications can be expected to differ according to institute and nation. The study of Callan et al. has suggested that family history of CHD was the most common indication for referral followed by fetal dysrhythmia.[17] However, another study reported that the most common indication for FE was abnormal cardiac view on anomaly scan (which occurred in 50% of their cases), followed by fetal hydrops (30.8%), and polyhydramnios (25%). Moreover, a low percentage of referrals were associated with chromosomal defects and extra-cardiac anomalies.[6] Contrary to the aforementioned studies, Nair and Radhakrishnan described echogenic intra-cardiac foci as the most prevalent indication of referral.[18] Significant abnormal findings were found in 51 (15.74%) cases in our study. However, Pike et al. and Nair and Radhakrishnan reported a comparatively higher percentage of fetal cardiac anomalies, i.e., 22.5% and 19%, respectively.[9,18] Among all the pregnant women having maternal indication for FE, 22.07% fetus had some form of abnormal cardiac findings, whereas only 12.40% fetus among women with fetal indication for FE showed positive cardiac findings, which was statistically significant ($p < 0.05$). The detection rate of CHD was high in the group of patients referred due to maternal risk profile such as maternal diabetes, whereas low yield were seen in FE done for fetal indications, which is in contrary with other studies, however this may be secondary to the small sample size, as the number of those referred for these indications was limited. Similar studies from National Heart Centre of Nepal showed average gestational age of 25.6 weeks (range, 18 to 38 weeks) on presentation for FE. Thirty-eight (15.1%) pregnant women had abnormal fetal cardiac findings and the most common referral for fetal cardiac scan was also related to maternal indications (48.6%), which supports the results of our study.[19]

In general, isolated VSD is the most common CHD defect, and ASD is the second most common, whereas among cyanotic CHDs, TOF is considered most common.[4] It is expected that not all CHD will be identified, though we believe that with increasing experience and fetal cardiac expertise, improvements in technology, and creation of universal guidelines for fetal cardiac imaging, detection rates and accuracy will continue to improve.[9] In our study, isolated VSD (18 cases) was the most common cardiac defect seen in FE, and Hypertrophic cardiomyopathy (16 cases) was the second most common cardiac anomaly. This may be because we had many women (30.31%) with gestational diabetes mellitus indicated for FE which is known to have transient hypertrophic cardiomyopathy. In particular, secundum ASD, small VSD, and mild PS, which are common in CHDs, are very hard to identify in the fetal period; thus, most of them are diagnosed by postnatal echocardiography. In a study done in pediatric outpatient clinic at Dhulikhel hospital with a total of 218 pediatric

cardiac cases, 144 cases (66.05%) had CHD, among which majorities were diagnosed as isolated VSD in 36 cases (25%) and isolated ASD in 29 cases (20.13%) followed by PDA in 13 cases (9.02%), TOF in 10 cases (6.94%) and complex CHD were seen in 9 cases (6.25%).[20] This also supports our FE findings which had isolated VSD (18 cases) as the most common cardiac defect prenatally. The structural anomalies of moderate to complex CHDs can be detected much more easily by FE than those of simple CHDs. There are technical constraints to image resolution that become particularly important when imaging FE, and it is for this reason that septal defects may remain difficult to diagnosis with complete accuracy. FE has been widely used to assess fetal cardiac structure and function. The greatest challenge in performing a FE is the constant movement of the baby with the small size and dynamic nature of fetal heart. However, as lungs are not inflated and bones are not ossified early in gestation, it may be possible to image heart in planes otherwise not possible after birth. Later in gestation, the ossification of the spine and ribs may make examination difficult. Maternal obesity and oligo / polyhydramnios may produce technical limitations making it difficult to gather adequate information during FE.

There are several important limitations to this study. Since this is a single center, hospital based study with limited sample size it may not reflect the actual indication of FE and the prevalence of CHD in the community. There might have been number of abortions or fetal death before FE could be performed. Due to its retrospective nature, follow up and postnatal echocardiographic data were not available for the study. These limitations should be taken into consideration before replicating the study results in the general population.

CONCLUSION

Despite all the limitations we conclude that, FE is a well known diagnostic method for detecting prenatal CHD and has become widely used in pediatric cardiology. The general population must be made aware regarding the importance of prenatal diagnosis and timely management of cardiac anomalies for improved outcome. We assume the study findings will be helpful for indicating FE to improve the detection rate and timely interventions of CHDs in a developing country like Nepal.

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REFERENCES

1. Bahadur KC, Sharma D, Shrestha MP, Gurung S, Rajbhandari S, Malla R, et al. Prevalence of rheumatic and congenital heart disease in schoolchildren of Kathmandu valley in Nepal. *Indian heart journal*. 2003;55(6):615-8.
2. Zhang Y, Riehle-Colarusso T, Correa A, Li S, Feng X, Gindler J, et al. Observed prevalence of congenital heart defects from a surveillance study in China. *Journal of ultrasound in medicine : official journal of the American Institute of Ultrasound in Medicine*. 2011;30(7):989-95.
3. Friedman AH, Kleinman CS, Copel JA. Diagnosis of cardiac defects: where we've been, where we are and where we're going. *Prenatal diagnosis.* 2002;22(4):280-4.
4. Cha S, Kim GB, Kwon BS, Bae EJ, Noh CI, Lim HG, et al. Recent trends in indications of fetal echocardiography and postnatal outcomes in fetuses diagnosed as congenital heart disease. *Korean circulation journal*. 2012;42(12):839-44.
5. Nayak K, Chandra GSN, Shetty R, Narayan PK. Evaluation of fetal echocardiography as a routine antenatal screening tool for detection of congenital heart disease. *Cardiovascular diagnosis and therapy.* 2016;6(1):44-9.
6. Kanwal A, Sheikh AM, Saher T. Determining the factors causing delayed referral for fetal echocardiography at a tertiary care hospital. *Journal of the Saudi Heart Association*. 2018;30(3):205-10.
7. Rychik J, Ayres N, Cuneo B, Gotteiner N, Hornberger L, Spevak PJ, et al. American Society of Echocardiography guidelines and standards for performance of the fetal echocardiogram. *Journal of the American Society of Echocardiography : official publication of the American Society of Echocardiography.* 2004;17(7):803-10.
8. Kapoor R, Gupta S. Prevalence of congenital heart disease, Kanpur, India. *Indian pediatrics.* 2008;45(4):309-11.
9. Pike JI, Krishnan A, Donofrio MT. Early fetal echocardiography: congenital heart disease detection and diagnostic accuracy in the hands of an experienced fetal cardiology program. *Prenatal diagnosis.* 2014;34(8):790-6.
10. Garg S, Sharma P, Sharma D, Behera V, Durairaj M, Dhall A. Use of fetal echocardiography for characterization of fetal cardiac structure in women with normal pregnancies and gestational diabetes mellitus. *Journal of ultrasound in medicine : official journal of the American Institute of Ultrasound in Medicine.* 2014;33(8):1365-9.
11. American Institute of Ultrasound in M. AIUM practice guideline for the performance of fetal echocardiography. *Journal of ultrasound in medicine : official journal of the American Institute of Ultrasound in Medicine*. 2013;32(6):1067-82.
12. Bakiler AR, Ozer EA, Kanik A, Kanit H, Aktas FN. Accuracy of prenatal diagnosis of congenital heart disease with fetal echocardiography. *Fetal diagnosis and therapy.* 2007;22(4):241-4.
13. Donofrio MT, Moon-Grady AJ, Hornberger LK, Copel JA, Sklansky MS, Abuhamad A, et al. Diagnosis and treatment of fetal cardiac disease: a scientific statement from the American Heart Association. *Circulation*. 2014;129(21):2183-242.
14. Levi S, Zhang WH, Alexander S, et al. Short-term outcome of isolated and associated congenital heart defects in relation to antenatal ultrasound screening. *Ultrasound Obstet Gynecol*. 2003;21:532-538.
15. Abbott MB, Vlasses CH. Nelson textbook of pediatrics. *JAMA*. 2011;306:2387-8.
16. CarvalhoJ S, Mavrides E, Shinebourne EA, et al. Improving the effectiveness of routine prenatal screening for major congenital heart defects. *Heart.* 2002;88:387-91.

17. Callan NA, Maggio M, Steger S, Kan JS. Fetalechocardiography: indications for referral, prenatal diagnoses, and outcomes. *Am J Perinatol.* 1991;8:390-4.

18. Nair A, Radhakrishnan S. Evaluation of Referral Pattern for Fetal Echocardiography at a Tertiary Care Center in Northern India and Its Implications. *Journal of obstetrics and gynaecology of India*. 2016;66(Suppl 1):258-62.

19. Regmi S, Sharma D. Fetal Echocardiography: Short Term Profile from Shahid Gangalal National Heart Centre, Kathmandu, Nepal. *Nepal Hear J.* 2016;13(1):9-12.

20. Joshi A, Shrestha RPB, Shrestha PS, Dangol S, Shrestha NC, Poudyal P, Shrestha A. Pattern of Cardiac Diseases in Children Attended at Dhulikhel Hospital, Nepal. *Kathmandu Univ Med J.* 2016; 55(3): 239-43.