

DOI: <https://dx.doi.org/10.18203/2320-1770.ijrcog20221291>

Original Research Article

## Observational study to determine the proportion of types of congenital heart diseases in high-risk pregnancies with detailed prenatal echocardiography

Balakrishna\*, Ramanand Gehlot, Kirti Chaturvedy, Dalpat Singh Rajpurohit,  
Manvika Chandel, Nauratmal Kumawat, Viram Singh Rathod,  
Deepak Pareek, Rughnath Patel, Pradeesh Sheoran, Divyangi Mishra

Department of Radio-diagnosis, S. N. Medical College, Jodhpur, Rajasthan, India

**Received:** 22 March 2022

**Revised:** 13 April 2022

**Accepted:** 14 April 2022

**\*Correspondence:**

Dr. Balakrishna,

E-mail: balakrishnask9@gmail.com

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### ABSTRACT

**Background:** Specific factors that increases a mother's risk of carrying a fetus with CHD, with the use of extended views in fetal echocardiography in addition to basic standard views there is increased detection of CHDs. Purpose of study current study was to investigate the role of fetal echocardiography in prenatal diagnosis of the congenital heart diseases in high-risk pregnancies between 18-36 weeks of gestational age and to determine the proportion of occurrences of different types of CHDs in and high-risk group.

**Methods:** Observational study with consecutive sampling 880 subjects. Study done in department of radiodiagnosis, SNMC, Jodhpur, Rajasthan

**Results:** With use of basic standard views, extended views and 3D views total 27 cases were detected out of all 880 subjects, which is significant statistically. Many CHDs which can be missed by the standard views, can be more efficiently detected with use of extended views in the fetal echocardiography.

**Conclusions:** Most common type of CHD was found to be VSD and hypoplastic left heart syndrome.

**Keywords:** High-risk pregnancy, Fetal echocardiography, Extended views, Congenital heart diseases

### INTRODUCTION

Congenital heart disease is most frequently occurring congenital disorder, responsible for 28% of all congenital birth defects. The birth prevalence of CHD is 8-12/1000 live births in India.<sup>1</sup> CHD birth prevalence worldwide and over time is suggested to vary differences may also be of genetic, environmental, socioeconomic, or ethnic origin. Intrapartum/intranatally fetal echocardiography is best & most effective modality to assess the heart & to detect any valvular, myocardial, vascular anomalies. Fetal echocardiography is effective because of its availability, good safety profile, reproducible, pain less

and relatively low cost. Real time evaluation of the anatomical & hemodynamic relationships of intracardiac lesions.<sup>2,3</sup> Specific factors that increases a mother's risk of carrying a fetus with CHD, maternal age older than 35years, coexisting maternal disease, teratogen exposure. In Such cases detailed fetal echocardiography is commonly done as a part of sonographic screening between 18 -22 weeks of gestation.<sup>4,5</sup> A family history of congenital heart defects among first degree relatives increases the risk to 4% to 12%. In addition, patients with diabetes, phenylketonuria & other noncardiac abnormalities are at increased risk for CHDs.

A systematic approach in fetal echocardiography for screening of CHDs involving the basic views can miss out outlet obstructions and upon including extended views incidence of CHDs increased.<sup>6-9</sup> With the use of systematic approach for screening CHDs and recent advances in three dimensional views have shown their strong hold in effective diagnosis in prenatal diagnosis.

### ***Aim and objectives***

Aim of the current investigation was to study the role of fetal echocardiography in prenatal diagnosis of the types of congenital heart diseases in high-risk pregnancies between 18-36 weeks of gestational age. Objectives of the study were taking high risk group pregnancies as cases in between 18-36 weeks of gestational age and to determine the proportion of occurrences of different types of CHDs in and high-risk group.

## **METHODS**

### ***Study design, location, duration and location***

Current study is a cross sectional observational study performed at department of of radio-diagnosis, DR S. N. medical college, Jodhpur Rajasthan. Study was done from July 2021 to December 2021.

### ***Inclusion criteria***

Inclusion criteria for current study were; all pregnant mothers in second trimester between 18 to 36 weeks by LMP who are at high risk for congenital heart disease in fetus which includes questionable anomaly seen on outside ultrasonography, which has to be confirmed in the departmental sonography, uncontrolled diabetes-which involves uncontrolled blood sugar levels with high HBA1c values on medical and insulin for treatment, gestational Diabetes mellitus-diagnosed by 2-hour oral GTT test, maternal drug exposure- Teratogenic such as Isotretinoin, Indomethacin, lithium. Anti-epileptics such as phenytoin, antibiotics-doxycycline, tetracycline, fetal heart arrhythmia-diagnosed on routine sonography on M-mode, history of structural anomaly associated with congenital heart disease in mother, Previous child with heart defect, history of previous child born with anomaly, history of extracardiac anomaly at present scan and collagen vascular disease- previously diagnosed by blood, urine tests for hormonal levels, biopsy for microscopic study, viral infection in early pregnancy; CMV virus, Rubella, Herpes virus, history of first degree relative with congenital heart defect, family history of CHDs, maternal medical diseases such as-thyroid disease, autoimmune diseases and h/o radiation exposure to more than 50 mGy.

### ***Exclusion criteria***

Exclusion criteria for current study were; pregnant women with previous normal ultrasonography anomaly scan performed in our hospital within a period of 1 month, pregnant women with twins/triplets/multiple fetal pregnancy.

### ***Sampling technique and sample size estimation***

Consecutive sampling was employed in the current study. Sample size will be calculated at 95% confidence interval to verify an expected 23% proportion of CHDs among high-risk group pregnancies in between 18-36 weeks of gestational age and taking 10% absolute allowable error. Sample size was calculated using the formula for sample size for estimation of proportion;

$$n = \frac{(Z_{1-\alpha/2})^2 P (1 - P)}{E^2}$$

Where,  $Z_{\alpha/2}$ =standard normal deviate for 95% confidence interval (taken as 1.96), p=expected proportion of CHDs among high-risk group pregnancies in between 18-36 weeks of gestational age (taken as 2.3% as reported by Nayak et al.<sup>19</sup> E=absolute allowable error (taken as 1%). Sample size was calculated to be to be minimum 863 subjects of high-risk group pregnancies in between 18-36 weeks of gestational age.

### ***Procedure***

Patient personal details & a detailed history of the patient is noted. Radiological imaging: Imaging was done with equipment of Philips, Affiniti 70G USG machine. A routine obstetric sonography is done for determination of the gestational accurate age using obstetric scan parameters. Once the adequate gestational age is established a dedicated fetal echocardiography is performed and data is entered in the following data as described below in table 4 (Obstetric data). In high risk group a detailed extended fetal echocardiography study is performed as described in (Table 3). Statistical analysis will be done with the data available.

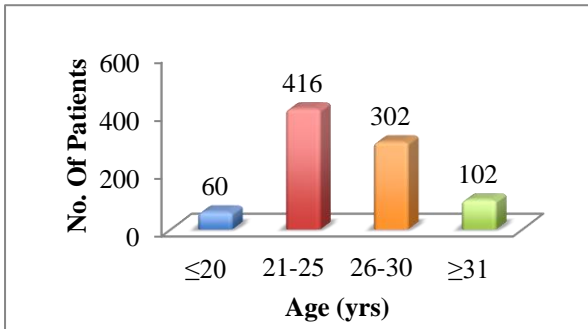
## **RESULTS**

The study included 880 subjects involving the high risk pregnancies for congenital heart disease as per inclusion criteria of this study. They underwent fetal echocardiography between May 2021 to December 2021. There were varied age groups among the subjects who underwent fetal echocardiography. The age of study subjects ranged from less than 20 years i.e., from 18 years to 40 years. Maximum incidence was seen in the age group of 21 to 25 years. 60 patients were below 20 years, 416 between 21 -25 years, 320 were between 26-30 years and 102 were between 31 to 40 years. In our study, fetal echocardiography was performed between 18

to 36 weeks of gestational age the distribution of cases seen as follows.

**Table 1: Age distribution among the high-risk pregnant subjects.**

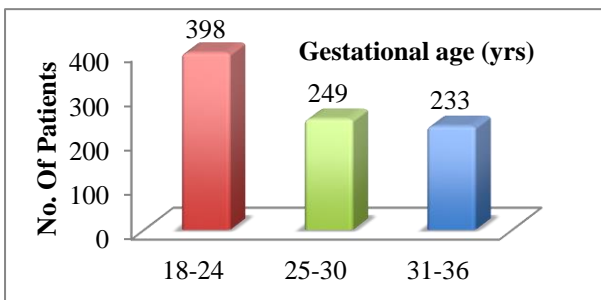
Age (yrs)	N	%
≤20	60	6.82
21-25	416	47.27
26-30	302	34.32
≥31	102	11.59
<b>Total</b>	<b>880</b>	<b>100.00</b>



**Figure 1: Age distribution among the high-risk pregnant subjects.**

**Table 2: Distribution of cases in gestational weeks.**

Gestational age (weeks)	N	%
18-24	398	45.23
25-30	249	28.30
31-36	233	26.48
<b>Total</b>	<b>880</b>	<b>100.00</b>

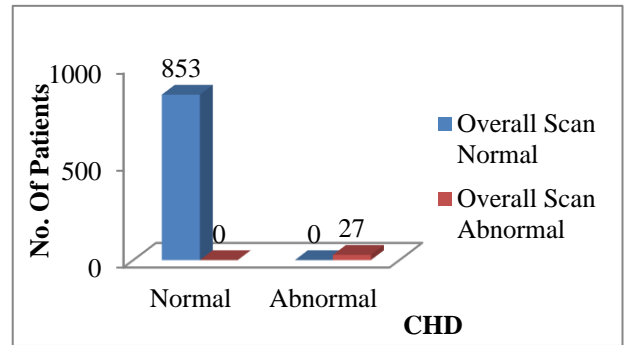


**Figure 2: Distribution of cases in gestational weeks.**

There were 398 fetuses which were between 18-24 weeks of gestational age (GAs) & 233 fetuses were in the range of 31 to 36 weeks of GA. Indicating that most fetuses between 18 to 24 weeks of GA & fetal echo were performed in this GA were highest in number. Over all scan of the all 880 cases who underwent fetal echocardiography involving all the views mentioned above showed 27 abnormal cases & 853 cases with normal views with no abnormality is shown in (Table 3).

**Table 3: Overall scan among the 880 study subjects showing total normal/abnormal cases.**

CHD	Overall scan				Total	
	Normal		Abnormal		N	%
	N	%	N	%		
<b>Normal</b>	853	100	0	0	853	96.93
<b>Abnormal</b>	0	0	27	100	27	3.16
<b>Total</b>	<b>853</b>	<b>100</b>	<b>27</b>	<b>100</b>	<b>880</b>	<b>100</b>



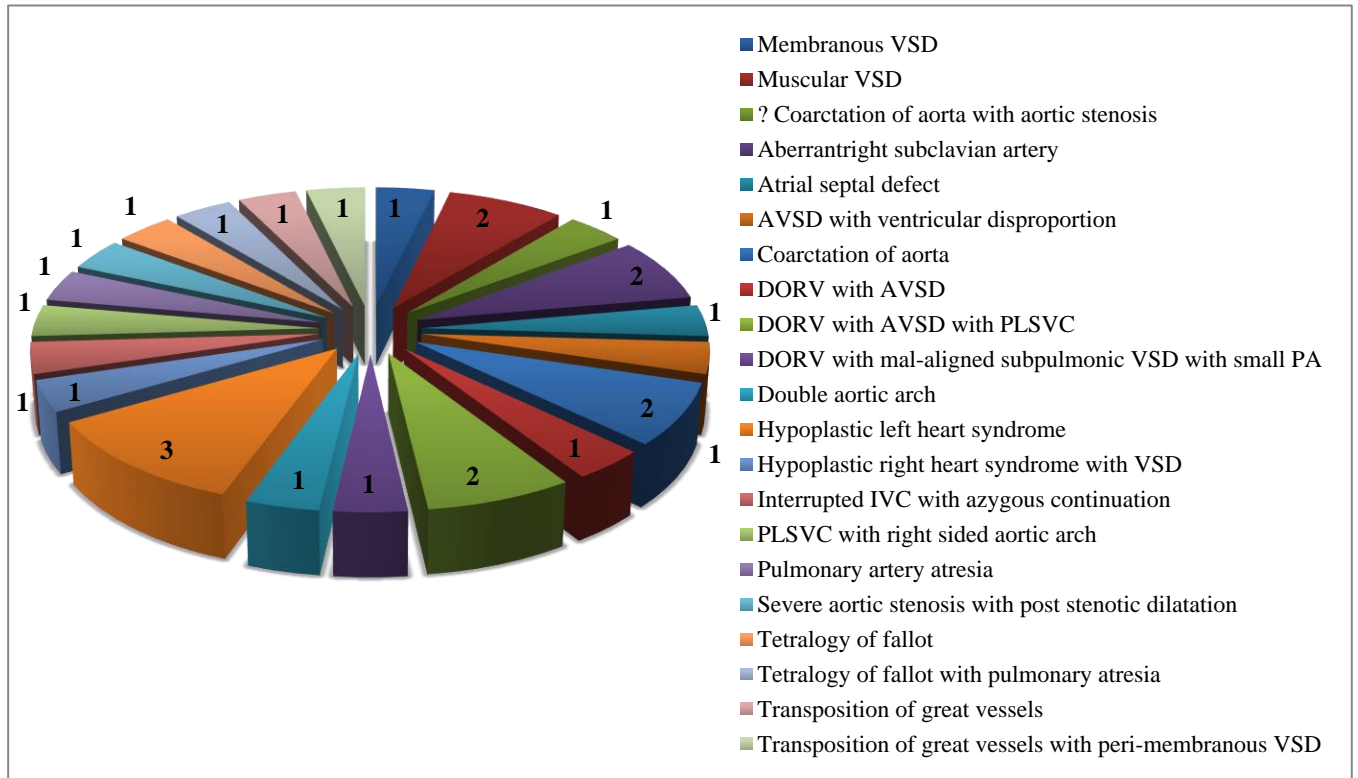
**Figure 3: Overall scan among the 880 study subjects showing total normal/abnormal cases.**

**Table 4: Proportion of different types of CHDs among study subjects.**

Type of CHD	N	%
Membranous VSD	1	3.70
Muscular VSD	2	7.41
Coarctation of aorta with aortic stenosis	1	3.70
Aberrant right subclavian artery	2	7.41
Atrial septal defect	1	3.70
AVSD with ventricular disproportion	1	3.70
Coarctation of aorta	2	7.41
DORV with AVSD	1	3.70
DORV with AVSD with PLSVC	2	3.70
DORV with mal-aligned subpulmonic VSD with small PA	1	3.70
Double aortic arch	1	3.70
Hypoplastic left heart syndrome	3	11.11
Hypoplastic right heart syndrome with VSD	1	3.70
Interrupted IVC with azygous continuation	1	3.70
PLSVC with right sided aortic arch	1	3.70
Pulmonary artery atresia	1	3.70
Severe aortic stenosis with post stenotic dilatation	1	3.70
Tetralogy of fallot	1	3.70
Tetralogy of fallot with pulmonary atresia	1	3.70
Transposition of great vessels	1	3.70
Transposition of great vessels with peri-membranous VSD	1	3.70
<b>Total</b>	<b>27</b>	<b>100</b>

The result was found significant at  $p < 0.05$ . Single cardiac defects are most common type of the fetal CHDs i.e. 16 cases. 11 cases out of 27 had multiple cardiac defects. Details of proportion of different types of the CHDs is shown in results. There were 3 cases of hypoplastic left heart syndrome and 3 cases of DORV in association with other cardiac defects. 3 cases of VSD, 3 cases of Coarctation of aorta. 2 cases of tetralogy of fallot, and 2

cases of transposition of great vessels, Aberrant subclavian artery. Singles cases of ASD, AVSD with ventricular size discordance, double aortic arch, pulmonary atresia, severe AS with post-stenotic dilatation, PLSVC with right sided aortic arch. Most common single cardiac defect being VSD and HLHS. Most common complex cardiac defect was DORV.



**Figure 4: Proportion of different types of CHDs among study subjects.**

## DISCUSSION

Congenital heart diseases (CHD) are suggested to be associated with advanced maternal age in different ethnicities and geographical locations. Hashim et al.<sup>10</sup> They assessed the association between maternal age and congenital heart diseases for different age groups during the period from 2016 to 2018. Found an association between maternal age and the type of CHD was found. Mothers who are 35 years old or younger are more likely to have a baby with atrial septal defects, while babies of mothers who are older than 35 year presented mostly with ventricular septal defects and patent ductus arteriosus. However, in our study it was different from Hashim et al.<sup>10</sup> There were 2 positive cases (7.4%) of CHDs were detected in 30-40 years out of 102 cases (11.5%) screened in this age group. 25 positive Cases (92.5%) of CHDs were detected in the age group of 20-30 years out of 718 cases (81.5%) screened. 60 cases were below <20 years had no detectable CHDs. Most common type of

CHD in our study was VSD amongst 20 -30 years (Table 1).

According to Volpe et al most forms of heart defects can be diagnosed early in pregnancy, some may develop and become apparent only later in gestation.<sup>11</sup> Persico et al performed fetal echo at 11-13 weeks, it was concluded that using high-resolution ultrasound equipment can assess the fetal heart at 11-13 weeks with a high degree of accuracy.<sup>12</sup> Simpsom et al did a respective analysis at 12-15 weeks of GA in a population at high risk for CHDs.<sup>13</sup> Our study was comparable to all above studies, hence fetuses between 18 to 36 weeks were considered & found that second trimester was ideal time for fetal echocardiography (Table 2). Nayak et al in their study showed that proportion of CHDs among the high risk pregnancy is 2.3%.<sup>14</sup> In current study with use of extended views proportion of CHDs was higher i.e., 2.7% in comparison to the study done by Krishnanand nayak et al. Solidifying the notion that use of extended views decreases false negative cases (Table 3). Out of all the 880 high risk pregnant screened, most common type of

CHD was found to be VSD and hypoplastic left heart syndrome (Table 4). This result was comparable to study done by Bromley B et al.<sup>15</sup>

### Limitations

Current study our objective was mainly to study the improved detection of the congenital heart diseases using extended views & three-dimensional views in addition to the basic views in fetal echocardiography, it was confirmed and statistically found that in fact additional use of extended views and 3D views in association with basic views improved detection of congenital heart diseases. However, The concern in current study was lack of follow up post-nataly.

### CONCLUSION

Fetal echocardiography can effectively identify abnormal hearts and has enhanced prenatal detection of CHD. In our study objective was mainly to study the improved detection of the congenital heart diseases using extended views and three-dimensional views in addition to the basic views in fetal echocardiography, it was confirmed and statistically found that in-fact additional use of extended views and 3D views in association with basic views improved detection of congenital heart diseases. In current study with the use of basic standard views, extended views and 3D views total 27 cases were detected out of all 880 subjects, which is significant statistically. Many CHDs which can be missed by the standard views can be more efficiently detected by use of extended views in the fetal echocardiography. In current study most common type of CHD was found to be VSD and hypoplastic left heart syndrome. Hence educating the women in reproductive age group regarding GDM and other risk factors for CHDs can significantly decrease the incidence of the CHDs. Early detection of CHDs with extended views helps in management of high-risk pregnancy and significantly reduced the burden of morbidity amongst both mother and infants to be born. In the Indian scenario the concern is the late referrals, lack of follow-up, and financial difficulties, all of which conspire against the chance of the fetus with heart disease getting appropriate treatment.

*Funding: No funding sources*

*Conflict of interest: None declared*

*Ethical approval: The study was approved by the Institutional Ethics Committee*

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**Cite this article as:** Balakrishna, Gehlot R, Chaturvedy K, Rajpurohit DS, Chandel M, Kumawat N, et al. Observational study to determine the proportion of types of congenital heart diseases in high-risk pregnancies with detailed prenatal echocardiography. *Int J Reprod Contracept Obstet Gynecol* 2022;11:1545-50.