PROFILE OF CONGENITAL HEART DISEASE AS DIAGNOSED BY FETAL ECHOCARDIOGRAPHY, A TERTIARY CARE EXPERIENCE
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ABSTRACT: BACKGROUND: The incidence of congenital heart disease is 0.8 in 1000 live births. Fetal echo cardiography offers a chance to detect most hemodynamically significant congenital heart disease in early pregnancy, so that their management prenatally, at birth and postnatally can be planned better. OBJECTIVES: To analyze the profile of congenital heart disease as diagnosed by fetal echocardiography, in pregnant women referred to a tertiary care centre. MATERIAL AND METHODS: The study design is retrospective, observational study. A total number of 583 fetal echo studies were performed at our Centre from January 1st 2014 to June 30th 2015. All studies were performed by a single operator. The main indications for the referral were: inability to visualize a clear four chamber view on obstetric scan, echogenic focus in LV cavity, maternal diabetes, family history of congenital heart disease, and maternal request. Statistical analysis was performed using statistical package for social sciences SPSS: CHICAGO, 3L VSA program. All the women with abnormal FE studies, except ones with complex CHDS were instructed to bring the new borns for 2D echo within 1 – 2 days after birth. RESULTS: Overall, significant CHD was found in 5.4% of FE studies (32 out of 583 cases). The most common indications for referral were: inability to visualize a clear four chamber view on obstetric scan, echogenic focus in LV cavity, maternal diabetes, family history of congenital heart disease, and maternal request. The risk factors with highest yield of CHD were poly hydroamnios and maternal diabetes. The commonest lesion found was ostium secundum ASD. There was a significant correlation between the presence of echogenic focus prenatally and small to moderate sized ostium secundum ASD after birth. CONCLUSION: Our data suggests that the risk factors with highest yield of CHD are polyhydromnios and meternal diabetes. Presence of echogenic focus in the LV in FE is associated with increased frequency of ostium secundum ASD postnataally and may be an indication for postnatal 2D echo screening.

INTRODUCTION: Congenital heart disease is an important cause of infant mortality in the first year of life, with an estimated incidence of 6 to 8 per 1000 live births.¹ For some cardiac lesions prenatal diagnosis imparts a survival advantage and helps the parental intention to treat decision making process.² In addition, a pre natal diagnosis of CHD provides the opportunity to discuss the findings with parents, plan the obstetric management and perinatal care.³ But CHDS are the most commonly missed malformations on the obstetric scan, including the anomaly scan, i.e. TIFFA scan. FE has a sensitivity of 78% and specificity of 99.9%, when performed by an expert.⁴

The purpose of the present study was to analyze the risk factor profile of the pregnant women referred for fetal echo and to analyze the profile of the CHDS, as diagnosed by FE and to analyze which risk factors were associated with highest yield of CHD, and to provide follow up when available.
MATERIAL AND METHODS: The study was a retrospective analysis and included the data of 583 FE studies performed at our centre between January 2014 and June 2015. The studies were performed mostly between 20 to 22 weeks of gestation, by a single operator.

Fetal 2D, M mode and Doppler echocardiography including color flow imaging was performed with IE 33 echo machine (Philips), using 1 to 5 MHz and 3 to 8 MHz Probes.

The FE studies were performed in accordance with American institute of ultra sound in medicine (AIUM) practice guidelines for the performance of fetal echo cardiography.

Statistical analysis was performed using statistical package for social sciences SPSS: CHICAGO, IL USA program, version 15. Continuous data were expressed as mean + standard deviation.

Women with diagnosis of complex CHD on FE were advised close prenatal monitoring and in hospital delivery, with neonatologist and pediatric cardiologist in attendance. All the other women with diagnosed CHD and women with LV echogenic focus on FE were advised to bring the new borns for 2D echo within 1 to 2 days of delivery.

RESULTS: No total numbers of cases was 583. The average maternal age was 23 +/- 5(mean + standard deviation). The mean gestational age at the time of examination was 20 +/- 2 weeks. No differences were found between the maternal age and gestational age in women with and without significant CHD in their fetuses.

Overall, out of the 583 studies, 551 studies (94.7%) were normal. 32 FE studies (5.31%) showed CHD.

The common indications for the referral and the percentage of normal FE studies in each group, the percentage of abnormal studies in each group are presented in table – I

| Total Number Of Cases | Normal FE Study (percentage of total cases) | Abnormal FE Study (Percentage of Total Cases) |
|-----------------------|-------------------------------------------|-------------------------------------------|
| Unsatisfactory 4C view on TIFFA | 227 | 218(96.1%) | 9(3.9%) |
| Maternal request | 205 | 203(99.1%) | 2(0.9%) |
| Maternal diabetes | 54 | 42(77.7%) | 12(22.3%) |
| Echogenic focus | 39 | 37(94.88%) | 2(5.12%) |
| Maternal age>30yrs | 16 | 15(93.75%) | 1(6.3%) |
| Non cardiac anomalies | 9 | 8(88.8%) | 1(1.2%) |
| Single umbilical artery | 7 | 7(100%) | 0 |
| Increased nuchal translucency | 5 | 5(100%) | 0 |
| polyhydroamnios | 4 | 3(75%) | 1(25%) |
| Maternal infection in 1st trimester | 2 | 2(100%) | 0 |
| Family history of CHD | 13 | 12(92.3%) | 1(7.7%) |

Table 1: The referral groups and percentage of normal and abnormal FE studies in each group
Largest group referred for an FE study was the inability to visualize a satisfactory 4C view on TFFA scan (227 cases), but this was associated with CHD in only 3.9% cases (9 out of 227 studies). Out of the 9 abnormal scans 2 cases were ostium secundum ASDs, 3 cases were subaortic VSDs, 1 was TOF, 1 was situs inversus totalis. One case had HLHS, One case had Type I truncus arteriosus.

Of all the referral groups, the highest incidence of CHD was in polyhydroamnios (75%, 3 out of 4 FE studies) and maternal diabetes. (22.3%, 12 out of 54 FE studies).

Of the four studies with polyhydromnios, two cases had Ebstein’s anomaly and one had DTGA, in a fetus of a diabetic mother.

Totally 54 FE studies were in diabetic women. 42 of them (77.7%) were normal studies, 12 were abnormal with ostium secundum ASD (2), subaortic VSD (2), hypertrophic cardiomyopathy (2), DTGA (4) AVSD (1), DoRV with AVSD (1).

Totally 39 patients were referred with echogenic focus in the fetal LV cavity. But only two of them were abnormal (5.12%). One was subaortic VSD and another one was an OSASD. All the 39 new borns were subjected to 2D ECHO in the first two days after birth. Nine out of them had small to moderate sized OS ASD (23%).

Noncardiac anomalies, single umbilical artery (SUA), increased nuchal thickness and family history of CHD were not associated with significant CHD in our patient population.

| Types of CHD                          | No. of Abnormal Studies (N=32) |
|--------------------------------------|--------------------------------|
| OS ASD                               | 8(25%)                         |
| Subaortic VSD                        | 7(21.8%)                       |
| AVSD                                 | 3(9.3%)                        |
| DTGA                                 | 4(12.5%)                       |
| Ebstein’s Anomaly                    | 2(6.25%)                       |
| Muscular VSD                         | 1(3.1%)                        |
| DORV with AVSD                       | 1(3.1%)                        |
| CTGV                                 | 1(3.1%)                        |
| Tetrology of Fallot                  | 1(3.1%)                        |
| DORV, DTG with Subaortic VSD         | 1(3.1%)                        |
| Hypoplastic Left Heart Syndrome      | 1(3.1%)                        |
| Situs Inversus Totalis               | 1(3.1%)                        |
| Type 1 Truncus Arteriosus            | 1(3.1%)                        |

Table 2: Profile of congenital heart diseases on Fetal Echocardiography

**DISCUSSION:** Cardiac development occurs in the first 6 to 7 weeks of development. A structural heart defect usually develops during this period. CHD during gestation may further progress as the pregnancy advances and it may result in other anomalies such as fetal hydrops. Some CHDs may become apparent only in neonatal period or later. i.e. small OS ASD, valvar PS.

In our study, we reviewed 583 FE studies and analysed the indications and their yield in the detection of CHD. All examinations were done transabdominally, by a single operator, thus
eliminating inter observer variability. AIUM practice guidelines were followed for performing the FE studies. Overall, our data showed that CHD was detected in 5.3% of cases referred to us. The rate is higher than the rate reported by Baroom et al who reported a 3.4% incidence of CHD, but is lower than the rate reported by Reuven Sharony et al, who reported 5.8% incidence of CHD in FE studies.

Common maternal indications for FE study are maternal metabolic diseases, (Diabetes, phenylketonuria) autoimmune diseases with antibodies (anti Ro ssA/ anti La ssB), teratogen exposure such as lithium and retinoids, familial inherited disorders (ex: 22q11.2 deletion syndrome) and invitro fertilization.

The fetal indications are abnormal cardiac screening examination, 1st degree relatives with CHD, abnormal fetal heart rate or rhythm, fetal hydrops, extra cardiac anomalies, increased nuchal translucency, single umbilical artery (SUA), and monochorionic twins.

In our study, the highest number of referrals were because of an unsatisfactory 4C view on TIIFFA scan. But this group revealed CHD in only 9 pts. (3.9%). FE studies done because of maternal request were a large number in our study, but was not associated with significant CHD. The reason for this low yield in these groups may be due to high index of suspicion and easy accessibility to FE study.

The patient groups with highest yield of CHD were polyhydroamnios (75% cases had CHD) and maternal diabetes. (22.3% cases). It is in accordance with other studies which reveal high incidence of CHD in these groups.

Echogenic focus within the left ventricle (LV) cavity is thought to relate to increased mineralization of the tips of the papillary muscles within the LV. By itself it is not thought to increase incidence of CHD. But its presence may increase the odds of Down’s syndrome complicating the pregnancy by a factor of 5 to 6. In our study there was no significant association of echogenic focus and CHD. Interestingly, on follow up after birth, 9 out of the 39 newborns had small to moderate sized OS ASDs.

Single umbilical artery is associated with increased incidence of CHD. Increased nuchal fold thickness in the 1st trimester of pregnancy is associated with a high incidence of CHD, Trisomy 21 or Turner’s syndrome. In our study, neither of these factors, nor other extra cardiac abnormalities were associated with significant CHD.

In summary, our data suggests that polyhydroamnios and maternal diabetes are valuable indications for FE and are associated with detection of significant CHD. Unsatisfactory 4C view on TIIFFA is a low yield indication for FE. Presence of echogenic focus was associated with increased incidence of OS ASD postnatally and may be an indication for follow up 2D Echo of the newborn. The limitations of the study are the relatively small number of the patients. Also, the operator was not blind to the referral indications.

ABBREVIATION:

CHD: Congenital heart disease.
FE: Fetal echo.
4C: 4chamber.
AIUM: American institute of ultra sound in medicine (AIUM).
DTGA: D Transposition of Great Arteries.
AVSD: Atrioventricular Septal Defect.
TIFFA: Targeted imaging for fetal anomalies.
OS ASD: Ostium secundum atrial septal defect.
VSD: Ventricular septal defect.

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