

# FETAL ECHOCARDIOGRAPHY FOR RISKY PREGNANCIES IN SULAIMANI GOVERNORATE

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## ABSTRACT

### *Background*

Prenatal diagnosis of congenital heart disease is now well established for a wide range of cardiac anomalies. Diagnosis of congenital heart disease during fetal life not only identifies the cardiac lesion but may also lead to detection of associated abnormalities. This information allows a detailed discussion of the prognosis with parents. For continuing pregnancies, appropriate preparation can be made to optimize the postnatal outcome. Morbidity and mortality, following antenatal diagnosis, has been reported for coarctation of the aorta, hypoplastic left heart syndrome, and transposition of the great arteries.

### *Objective*

The aim of this prospective study is to evaluate our experience and results of fetal echo for high risk pregnancies in Sulaimani city.

### *Methods*

This is a prospective descriptive study conducted on 450 pregnant ladies referred for fetal echocardiography from obstetricians, ultrasonographers and (self-referral) from the 1<sup>st</sup> January 2013 to 1<sup>st</sup> February 2017. Echocardiography was done with a Siemens machine with a semicurved phased array probe at their first visit at 16-22 weeks in 330 ladies, at 22-28 weeks in 70 cases and at 31 weeks and above in 50 ladies. Also, 2<sup>nd</sup> visit echo was done for 60 patients for follow-up at 31-33 weeks gestation. All babies were checked post-labour for accuracy of the diagnoses.

### *Results*

Among 450 pregnant ladies, 98 cases were found to have abnormal findings, of which 27 were diagnosed as hypertrophic cardiomyopathy (just one persisted after 6 months post-labour), 25 cases as ASD (just 10 left true ASD post-labour), 14 diagnosed as tricuspid atresia and single ventricle physiology, 5 AVSD, 5 arrhythmia with hydrops, 5 with MV and TV abnormalities, 4 AS, 3 hypoplastic left heart syndrome, 3 PS, 2 L-TGA and one CoA. For cases who classify as low risk populations of defects, no action was undertaken, for high risk population, decision for termination was one of the options. 2 missed diagnoses were observed after labour, checking among anomalies observed on echo findings and they were of low risk cases, and no missed diagnosis was observed in major risk group diagnosed cases. Fetal echocardiography examination specificity was sensitivity as 97.9% and specificity as 99.4% which is within 95% CI, positive predictive value was 97.9%, negative predictive value was 99.4% in which both are within 95% CI.

### *Conclusion*

The present study showed that fetal echo has great value in detection of congenital heart disease, prognosis and outcome in high risk pregnancies. It is recommended to familiarize our obstetrician about this important diagnostic test. We should train our ultrasonographer and pediatric cardiologist to increase awareness about this diagnostic test (is it a screening test, or a diagnostic test). Also, as far as we know, this is the first paper written on this kind of diagnostic technique, so more studies need to be conducted in the future.

**Keywords:** *Fetal echo, Congenital heart disease, Gestational age*

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## INTRODUCTION

A wide range of congenital heart defects during in utero life can be diagnosed with a very high accuracy in specialist centers <sup>(1)</sup>.

In most parts of the world, the main routes of diagnosing congenital heart defects are, first, a suspicion of a defect in the heart during a routine ultrasound, or / and second, because specific risk factors made to referral to a cardiology unit for further evaluation. The risk factors for assessing by echocardiography [Table 1].

Detection of these cases depend on the sonographer and personnel experience. Some cardiac lesions, particularly those evident on a “four chamber view” of the fetal heart are more easily detected by the nonspecialist sonographer than others, for which more extended views of the outflow tracts are required for detection. For example, United Kingdom national data obtained between 1993 and 1995 reported a detection rate of 38% for atrioventricular septal defects and 66% for hypoplastic left heart (“four chamber” abnormalities) compared to 3% for transposition of the great arteries (outflow tract views required for detection) <sup>(2,3)</sup>. Congenital heart problems in which should be seen in different views, lesions which are considered very difficult to detect even in specialist hands, and those which cannot be detected during fetal life. [Table 2]

**Table 1. Summary of risk factors that should prompt detailed cardiac evaluation <sup>(2)</sup>.**

### Fetal factors

- Suspected cardiac abnormality on screening ultrasound
- Increased nuchal translucency thickness
- Fetal hydrops
- Fetal abnormality with known association of congenital heart disease
- e.g., exomphalos, diaphragmatic hernia
- Fetal arrhythmia
- Abnormal fetal karyotype, e.g., trisomy 21

### Maternal and familial risk factors

- Family history of congenital heart disease (CHD) in a first degree relative.
- Diabetes mellitus– Mothers who are established diabetics on treatment
- Mothers with a tendency toward diabetes that is solely related to pregnancy are not judged candidates for fetal echocardiography
- Mothers taking known teratogenic drugs, e.g., anticonvulsants, lithium

- Maternal anti-Ro or anti-La antibodies
- Mothers who have anti-Ro and / or La antibodies are candidates for fetal cardiology assessment, in view of the risk of developing fetal
- heart block. Mothers NOT having anti Ro or La antibodies are not candidates for fetal echocardiography
- Maternal infections, e.g., parvovirus, Coxsackie

**Table 2. Summary of the scope and limitations of fetal echo in the diagnosis of commonly occurring major cardiac malformations<sup>(2)</sup>.**

Examples of major lesions evident on “Four chamber views” of the fetal heart

- Hypoplastic left heart syndrome
- Severe coarctation of the aorta
- Critical aortic stenosis
- Tricuspid atresia
- Pulmonary atresia with intact ventricular septum
- Atrioventricular septal defect
- Double inlet ventricles

Examples of major lesions where the four chamber view of the heart is typically normal / near normal and for which views of the outflow tracts are required

- Transposition of the great arteries
- Tetralogy of Fallot + / - pulmonary atresia
- Common arterial trunk
- Some forms of coarctation of the aorta

Examples of lesions that are difficult to detect even in experienced hands

- Total anomalous pulmonary venous drainage
- Coarctation of the aorta (milder forms)
- Some types of ventricular septal defect
- Milder forms of aortic and pulmonary valve stenosis

Lesions that cannot be predicted from prenatal cardiac imaging

- Patent arterial duct
- Secundum atrial septal defects

### **Impact of fetal echocardiography on prevalence of congenital Heart disease**

When congenital heart disease is diagnosed during fetal life, the expectant parents should have a detailed discussion with a fetal cardiologist with regard to the

prognosis of the cardiac lesion, covering not only procedural risks, but also long-term mortality, morbidity, and quality of life. There should also be a discussion with regard to possible associations, including karyotypic abnormalities, noncardiac structural anomalies, and syndromes<sup>(2-5)</sup>, to have a full picture of the prognosis for their baby. Thus, effective management demands a close liaison between the cardiologist, fetal medicine specialist, genetics, and other relevant subspecialties. Depending on the severity of the cardiac lesion, the associated abnormalities, gestational age, and local policy and national guideline regarding termination for major structural abnormalities in the heart, one of the options open to parents may include termination of pregnancy. Termination mostly decided by parents mostly in hypoplastic left heart but less commonly with transposition of great arteries<sup>(2,3)</sup>.

Early detection was associated with a higher termination rate than diagnoses made later in gestation. Nuchal translucency screening involves measurement of a sonographically lucent area at the back of Nuchal translucency screening done at 11-14 wks gestation was introduced to identify fetuses at high risk for trisomy 21<sup>(6,7,8,9)</sup>.

#### **Impact of prenatal diagnosis on cardiac morbidity and mortality**

The vast majority of data on the impact of prenatal diagnosis on the morbidity and mortality of congenital heart disease is from developed countries. The data published to date will be reviewed briefly, but it should be emphasized that all the data comes from countries where the following requirements were met:

i.) Availability of appropriate prenatal investigations: If a cardiac lesion was diagnosed prenatally there was availability of relevant prenatal investigations, such as, fetal karyo typing and detailed ultrasound for noncardiac malformations.

ii.) High level delivery facilities and neonatal care was available: Following a prenatal diagnosis of congenital heart disease there would need to be a pattern of referral for delivery and treatment at a high level neonatal nursery/cardiac center if the outcome was to be optimized.

iii.) Postnatal surgical, interventional or medical treatment for the cardiac lesion in question was available without this being financially prohibitive.

iv.) Availability of long-term therapies: Prenatal diagnosis has an ascertainment bias for more severe forms of congenital heart disease. Cardiac lesions which will lead to single ventricle palliation are over represented in prenatal versus postnatal series. Any country or region instituting a prenatal screening program will have to consider which surgical options are available. Prenatal diagnosis of hypoplastic left heart was associated with better ventricular function, less tricuspid regurgitation, and a reduced requirement for inotropes and bicarbonate. Other data has confirmed better condition at presentation for infants who were diagnosed prenatally, but this did not lead to reduced overall mortality<sup>(10)</sup>. A further publication has reported a reduced incidence of abnormal neurological events related to prenatal diagnosis<sup>(11)</sup>.

#### **Aim of the study**

The aim of this prospective study is to evaluate our experience and results of fetal echo for high risk pregnancy in sulaimany and its impact on outcome of pregnancy.

#### **PATIENTS AND METHOD**

This is prospective descriptive study conducted on 450 pregnant ladies referred for fetal echocardiography from obstetricians, ultrasonographers and self referral routine check on their own requests from 1<sup>st</sup> January 2013 to the 1<sup>st</sup> February 2017.

Echocardiography done with Siemens machine with semicurved phased array probe with software specific for fetal echocardiography. Fetal echo done for the evaluation of abdominal situs and heart position within the chest, four chamber view, atrioventricular valve offsetting, ventricle filling with Color Doppler (CD), pulmonary artery-aorta crossing (X sign) with CD, forward flow and equal size at the confluence of the aortic arch and arterial ductus (V sign)<sup>(12,13)</sup>.

First visit fetal echo was done at 16-22 weeks in 330 ladies, at 22-28 weeks in 70 cases and at more than 31 weeks in 50 ladies. Then 2<sup>nd</sup> visit echo done for 60 patients for follow up at 31-33 weeks gestation. All babies after they born were checked for detail neonatal echocardiography and diagnosis were confirmed. They were informed of the limitations of the screening method and info med consent was obtained.

Patient included in the study were included in the risk factors as follows:

1. Arrhythmias
2. U/S abnormalities like structure abnormalities skeleton, head, kidneys, bowel, and others, Fetal hydrops, heart seems abnormal
3. IDM
4. Previous hx of child with CHD
5. CHD in the parents especially mother
6. Fetal distress
7. Abnormal amniocentesis

Limitation for fetal echocardiography

1. Some heart abnormalities are not detectable prenatally even with a detailed expert examination.
2. These tend to be minor defects, such as small holes in the heart, or mild valve abnormalities.
3. In addition, some cardiac defects do not become evident until after birth like PDA

Then the defects classify to major and minor in which the fetal cardiac defects are defined as major if they are either lethal or require surgery or interventional cardiac catheterization during the first year of postnatal life.

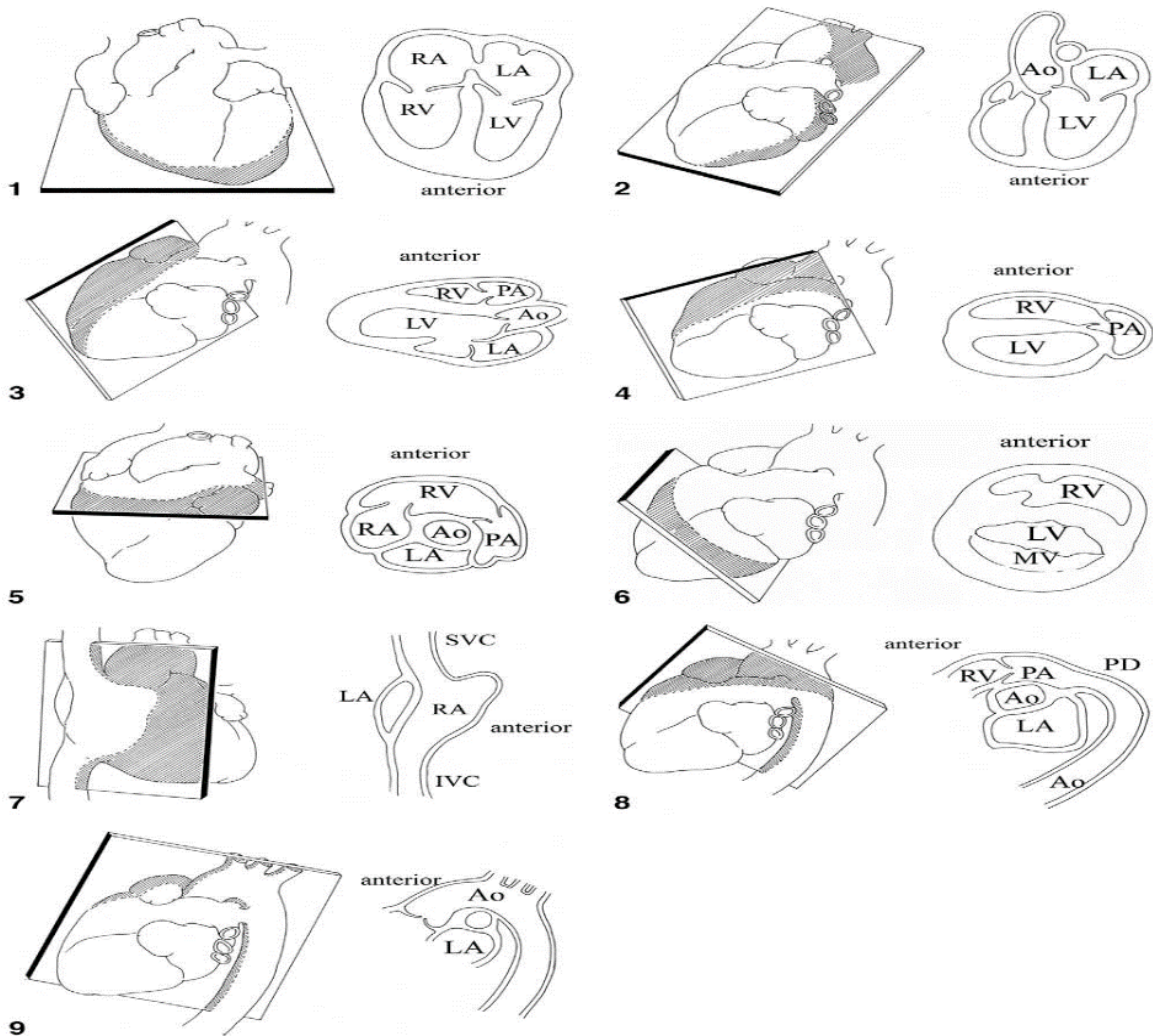


Figure 1. Shows standard different fetal echocardiography views <sup>(13)</sup>.

## RESULTS

During period of the study 450 pregnant ladies did fetal echocardiography for different reasons of which 180 cases (40%) of them got history of CHD in their children or the parent them self, 70 cases (38.8%) fetuses were evaluated for extra-cardiac anomalies evidenced by obstetric ultrasound screening, 30 cases (16.6%) for family history of genetic like diseases, 40 (22.2%) cases suspected to have CHD by the ultrasonographer, 60 (33.3%) cases with maternal disease and on medication.

Among total number of screened pregnant ladies, 98 cases were identified as an abnormal fetal echo findings, in which it subdivided according to the reason of doing echocardiography as follows 7 (7.1%) for family history of congenital heart diseases, 24 (24.48%) fetuses were evaluated for extra-cardiac anomalies evidenced by obstetric ultrasound screening, 17 (17.34%) for family history of genetic-linked diseases, 10 (10.2%) cases suspected to have CHD by the ultrasonographer, 35 (35.71%) cases with maternal disease or receive medications and another five (5.1%) patients came on routine check. (figure. 2).

Among the pregnant ladies with abnormal fetal echo findings 41 cases classified as major congenital heart defects in which they are subclassify to following as shows in (figure 3).

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As far as post natal detail echocardiography done for babies born in first month of life and followed up furthermore most of hypertrophic cardiomyopathic patient which account of 26 out of 27 cases did recover to normal myocardium in 3-6 months follow up and 10 out of 25 cases of ASD secundum cases persist after labour .

Among the postnatal echocardiographic follow ups we found two false +ve case diagnosis of CHD in minor group fetal finding and also another three false -ve case diagnosis of CHD in which 2 in minor group and one in major group.

So accordingly we calculate sensitivity as 97.9% and specificity as 99.4% which is within 95% CI (Confidence intervals for sensitivity and specificity are "exact" Clopper-Pearson confidence intervals).

Also positive predictive value was 97.9% , negative predictive value was 99.4% in which both within 95% CI. (Confidence intervals for the predictive values are the standard logit confidence intervals given by Mercaldo et al. 2007.)

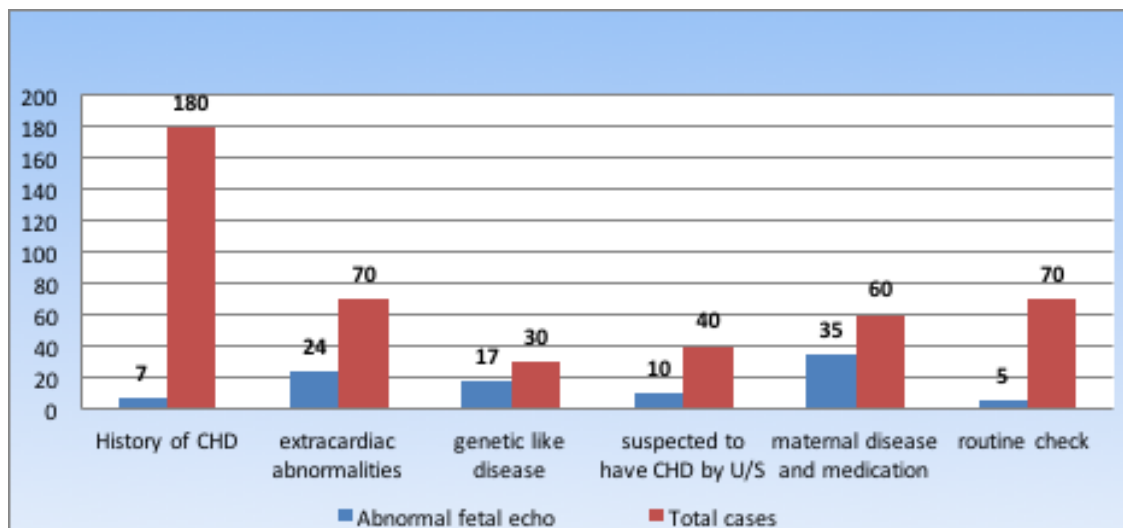


Figure 2. Reason for doing echo both abnormal and normal results groups.

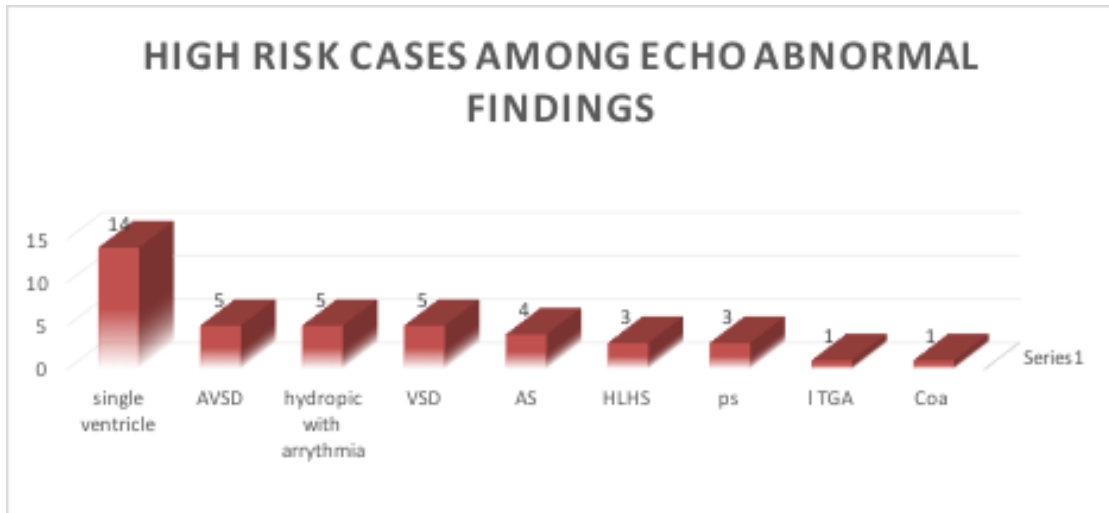


Figure 3. Classification of major anomalies according to the subtypes.

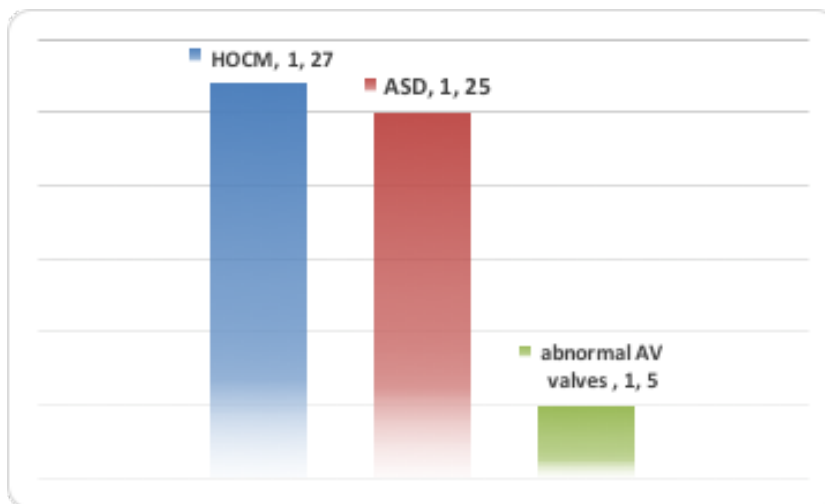


Figure 4. Abnormal echo finding classify as minor one.

## DISCUSSION

The most important issue is that the experience of specialist sonographer has important role in diagnosis and further management, future outcome of the fetus, as we moved in our study we found out easier and more confident in diagnosis even in early gestation.

As in other studies one of the major limitations for using fetal echocardiography as a screening tool is that it is operator-dependent, published data has confirmed the influence of operator training and experience <sup>(14, 15)</sup> on the ability to confirm normality of the cardiac connections.

If we compare the reason for doing echo in our study as shown in the (figure 1) in our results nearly same as they

found in a study done in USA , among the abnormal fetal echo cases, Extracardiac anomalies were noted in 36% and karyotypic abnormalities in 17% of patients <sup>(16)</sup>.

In another study in Austria they found 46 cases with congenital heart disease prenatally by echocardiography among 3085 cases, if compared to our result (98 among 450 cases), probably because of high selectivity cases we screened 5 among 2181 cases in the group with no risk factors its different from that we found (5 among 450 cases screened in our study), three among 540 cases in the group with maternal risk factors, and 28 among 365 cases in the group with sonographic abnormalities <sup>(17)</sup>.

Our study shows that targeting high-risk groups by fetal echocardiography, such as those with extracardiac abnormalities, history of CHD, karyotype abnormality and maternal disease, result for high sensitivity for cardiac defects detection rate which is constant with studies done in UK <sup>(18)</sup>.

In our records 41 (41.8%) cardiac defects classify as major and 57 (58.2%) classify as minor defects as compared to a study done in 2011 which shows among 100 cases with suspected cardiac defects included 54 (54%) in which the defect was classified as major and 46 (46%) in which it was classified as minor <sup>(12)</sup>.

The most common major cardiac defect in our study was single ventricle then secondly AVSD and minor one was hypertrophied ventricles specially LV but in study done in England shows the most common major anomaly was AVSD and minor one was disproportion of the ventricles and/or the great arteries <sup>(12)</sup>.

May be this finding explained by that we are not doing fetal echo as an early type scanning we did at more than 16 weeks and late time.

In another study done in USA, 39 patients classify as major and 30 as minor risk for CHD <sup>(16)</sup>. In our study we found 7 abnormal fetal echo of 180 scanned patients observed, in another study they found no cardiac defects in the 28 pregnancies in which a family history of a heart defect was the main indication for fetal echocardiography <sup>18</sup>, but the finding consistent with the recurrence rate of 2–3% for siblings <sup>(19)</sup>.

Our sample of TV regurgitation was not large number of observations that is why we can not compare to other studies for its association with congenital heart problems.

In our study we found 4 cases with TV regurgitation in which one found to have trisomies as they found in other studies regurgitation has been proposed as a second trimester marker for trisomy 21 <sup>20</sup>, <sup>(21)</sup>.

One of arrhythmia induced hydrops fetuses diagnosed as complete heart block and treated in utero by sympathomimetics and hydrops significantly decreased within 2 weeks, but unfortunately the mother could not tolerate the drug side effect so after self stopping medication the hydrops back again and end up with fetal intrauterine death.

In other studies the major congenital heart disease resulted in termination of pregnancy but in our study

termination only decided by the family in 4 cases the rest either end up in late pregnancies with intrauterine death or deliver with major defects and died in neonatal, only 4 still alive and did some kind of heart palliations or surgery.

In our study we calculate sensitivity as 97.9% and specificity as 99.4% which is within 95% CI, which nearly the same as compared to a study done Italy with sensitivity 85.7% and specificity 100% <sup>(22)</sup>.

### **Conclusion**

The present study showed that fetal echo has great value in detection of congenital heart disease, prognosis and outcome in high risk pregnancies.

Major congenital heart disease can be detected by high sensitivity in our study in which plan for pregnancy termination and continuation can depended.

When we have extracardiac ultrasound abnormalities, history of genetic like disease in the family fetal echo finding abnormalities are more likely.

It's recommended to familiarize our obstetrician about this important diagnostic test.

We should train our ultrasonographer and pediatric cardiologist to increase awareness about this diagnostic test. Also as far as we know we are the first doing fetal echo and this the first paper written on this kind of diagnostic technique, so more studies need to be conducted in the future.

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