

## PRENATAL DIAGNOSIS OF CONGENITAL HEART DEFECTS IN HIGH RISK PREGNANCY

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### SUMMARY

180 females with high risk pregnancy were subjected to fetal echocardiographic examination including M Mode and 2 dimensional echo between 14 and 36 weeks of gestation. Eleven abnormalities were detected. Five complex and six simple. Two cases of brady-arrhythmia were noted both had congenital heart block - both with complex structural heart disease. One false positive diagnosis of ASD was made, one associated lesion L-transposition was missed. The incidence of positive fetal echocardiography is found to be 1 in 16 pregnancy - much higher than the incidence of congenital heart disease in live born. The fetal echocardiography is very helpful to the obstetrician in deciding future management of individual patient and also for genetic counselling.

### INTRODUCTION :

Congenital heart disease (CHD) is second only to neural tube defects in frequency of congenital abnormalities in the liveborn. The incidence of congenital heart defects in prenatal life is much higher than at birth, Allah et al 1981. Unfortunately it has not received enough attention so far. However, with advancement in quality of imaging, the M-Mode and two dimensional examination of fetal heart is done in realtime very frequently with fairly adequate yield, DeVore et al 1982 Kleinman et al 1980.

In this paper, our aim is to study high risk females with a variety of indications and find out

the incidence of congenital cardiac malformation. This helps a great deal in counselling the patient, and selecting an appropriate obstetric option.

### MATERIAL & METHOD :

We examined 180 females with the gestational age varying from 14 to 38 weeks. Serial examination were carried out where visibility was poor or where there was doubt of certain anomalies. The maternal, fetal and familial indications for the study are shown in Table I. The most common indication was extracardiac malformation on sonography of current fetus or previous sibs. The other indications included previous occurrence of congenital heart disease in siblings, parents or current fetus; a maternal disease known to affect the fetus such a diabetes,

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TABLE I

Indication for Fetal Echocardiography	
— Congenital Heart Disease (CHD)	44
— Maternal Diseases, Exposure To Infection or Drugs	14
— Late Maternal Age	11
— H/o Down's Syndrome	03
— Bad Obstetric History	32
— Abnormal Amniotic Fluid	05
— Hydrops fetalis	01
— Intra Uterine Growth Retardation (IUGR)	10
— Arrhythmia	04
— Extra Cadiac Defects	56

hyperthyroidism; maternal use of drugs and anesthetic exposure and also late maternal age. Bad obstetric history was a common indication. This included abortions, intrauterine deaths (IUD), preterm deliveries and multiple congenital defects in one or more than one pregnancy. The other reasons for referral included abnormal amniotic fluid volume, hydrops fetalis, intrauterine growth retardation (IUGR) and presence of arrhythmia. Routine fetal anomaly scan was performed in all the mothers in detail to determine placental position, fetal lie, fetal maturity and other routine parameters. The realtime examination was performed by 4 MHZ linear array and 3 MHZ Sector Scanner of Philips Model SDR 1550 XP. Linear transducer was used initially as it has constant line density throughout the view and moreover whole fetus could be imaged in one view. Once the echocardiographic window was identified further details of fetal heart were obtained by Sector.

The mother was made to sleep in supine

position. Examination took 45 - 50 minutes on an average.

The ideal time of examination is between 16-28 weeks. Before 16 weeks, the machines resolution does not permit identification of cardiac structures and in advanced stage, fetal ribs and vertebrae interfere with good examination. The examination is ideally performed when the fetus is lying on its back with spine in posterior position and when the fetal movements are not in excess. At times when anterior spine obstructed examination it could be well obtained from fundal position. Obesity in mother, and Oligohydramnios also hinders a good examination.

Fetal echocardiography means much more than routine four chamber view with which the Obstetrician is most familiar. It includes 4 conventional reference planes as per Allan et al 1984.

These are long axis view both at great vessel and ventricular level and subcostal four chamber view. In addition there are various unconventional views possible unlike in a post-natal echocardiography. The whole extent of the aorta from its root to descending aorta could be noted in one plane, the great systemic veins could be seen to enter right atrium in one plane.

**RESULT :**

Table II lists the abnormalities we detected in a group of 180 high risk females on prenatal echocardiography. A false positive diagnosis of atrial septal defect (ASD) was made in one case. This case is excluded from the table. In two other cases (no. 7 & 8) the delivery is awaited. In nine cases, the diagnosis is confirmed either on post-natal echocardiography or fetal autopsies. In two cases (number 1 & 7) congenital complete heart block was found, both had associated complex structural defects. One had VSD with L-transposition where the latter was missed and second one had common atrium, single atrioventricular valve (A-V valve) with single ventricle. There were six cases of simple perimembraneous VSD-in one of

**TABLE - II**  
**Cardiac Anomalies Detected Prenatally**

Indications	Gestations in weeks	Cardiac Defect on Fetal Echocardiography	Associated Extra cardiac defects	Outcome
1. Fetal Bradycardia in present pregnancy	28	Congenital Complete Heart (CCHB) with VSD	—	(CCHB with VSD with L - Transposition (LTGA)
2. History of Prolonged hormone ingestion	20	Double Outlet right vent. (DORV).	—	DORV on autopsy
3. Polycystic kidney and Polydactili in previous sib. Family history of imperforate anus and ambiguous genitalia	23	VSD	Oligohydramnios. Enlarged Kidneys. Absent Urinary Bladder, Microcephally, Polydactili.	VSD on Autopsy.
4. Previous sib with Down's Syndrome	30	VSD	—	VSD on Postnatal Echo.
5. Abnormality suspected on routine sonography	36	Ebstein's Malformation	—	Ebstein's Malformation with pulmonary artery stenosis on Postnatal echo.
6. 1) Previous sib with CHD (Truncus arterious Type I died.) 2) Intra Uterine Death (IUD)	17.3	VSD	Hypoplastic left kidney, absent, urinary bladder. Polyhydramnios.	Fetal P.M. done VSD found.



Table II (Contd.)

Indications	Gestations in weeks	Cardiac Defect on Fetal Echocardiography	Associated Extra cardiac defects	Outcome
7. Fetal bradycardia 80 - 90 BMP in present pregnancy	29.3	Congenital Complete Heart Block (83 BPM) Common Atrium Single Ventricle. Single Atrio-ventricular	—	Delivery awaited
8. 1) Spontaneous abortion 3 months 2) IUGR - 6 months 3) Anencephaly - 7 months	24	Tricuspid regurgitation with dilated right ventricle.	Oligohydramnios	Delivery awaited.
9. Achondroplasia in father Dwarfism in mother	24	Small closing VSD with aneurysm formation.	—	VSD with aneurysm of of membraneous septum on postnatal echo.
10. 1. Cynotic CHD died - 18 days 2. Birth asphyxia died - 8 days.	25	VSD	—	VSD confirmed on echo.
11. Oesophageal atresia	25.3	VSD	—	VSD on post natal echo.

them there was an attempt at aneurysm formation in fetal life. Two cases (5 & 8) had dilated right sided chambers one of them had tricuspid regurgitation and the other had Ebstein's malformation. One case had a double outlet right ventricle (DORV) with Subaortic VSD.

The long axis view is very useful in detecting complex malformations like Tetralogy of Fallot, DORV as in our case. Of course, even simple VSD's can be diagnosed in this view. The apical 4 chamber view is useful in detecting simple VSD's, Ebstein's malformation, single ventricle and many other conditions. It is useful as a routine screening view. The short axis view is important in diagnosing semilunar valve abnormalities, transposition of great arteries and the subcostal four chamber view is important for M-Mode measurements and also to diagnose ASD.

#### DISCUSSION :

The incidence of congenital heart defect is 6 - 8/1000 live births but the incidence of positive fetal echo is found to be very high. It varies from 1 in 7 to 1 in 125 pregnancies. Keith, 1967, Allan et al 1981. Our incidence is found to be 1 in 16 pregnancies. All the patients studied in this paper had high risk factors but even routine fetal echocardiography in normal pregnancies our own study has yielded two abnormalities-one arrhythmia and one structural defect, Rane and Purandare 1990. Many number of times, a child is born with CHD without a known risk factor and therefore the importance of routine screening. Incidence of extracardiac malformation with CHD is much higher in prenatal life as opposed to neonates and therefore in all cases of extracardiac malformation, fetal echocardiography should be carried out, Ferencz et al 1987.

In the group with congenital heart defects the recurrence risk is low - 2 - 4% if only one sib is affected. It increases to 10 - 14% if one of the parents or more than 1 sib is affected with CHD, Rose et al 1985. We had two patients with positive fetal echocardiography findings where

there was a history of CHD earlier. Even when the echocardiographic examination turns out to be normal, it is extremely helpful in relieving tensions of mothers who have given birth to neonate with CHD in their first pregnancies.

It is a well known fact that more severe the defect, better is its change of defection, Benacerraf et al 1987. Simple defects like VSD, coarctation semilunar valve stenosis could easily be missed. ASD and patent ductus arteriosus (PDA) are normally also patent in fetal circulation and therefore the difficulty in picking up these abnormalities. Also certain associated lesions could be missed in a complex malformation, Sunder et al 1986. Lesions like dextrocardia are difficult to diagnose unless the fetus is continuously imaged spatially.

The yield of positive fetal echocardiography in a high risk population is high. Our aim is to detect these anomalies as early as possible in gestation. With newer machines and vaginal probes this could be easily achieved. This helps the Obstetrician a great deal in deciding about the nature of labour, when to interfere and terminate a pregnancy in correctible or non-correctible lesions. In certain cases when the pregnancy is too advanced while the echocardiography information is obtained, necessary arrangement can be made to have the delivery at a well equipped tertiary Centre. In case of tachyarrhythmia, drugs could be administered to mother to prevent hydrops in fetus Kleiman et 1985. In bradyarrhythmias caesarean section could be avoided, Shenoi et al 1991. Thus fetal echocardiography is not only a great boon to parents who have had earlier trauma of having a malformed child, but also to Obstetrician, pediatrician, cardiologist and geneticists for better management and effective counselling.

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