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Original Research Paper

Role Of Extended Fetal Echocardiography in Diagnosing Tetralogy of Fallot in Southern India

Medical Science

KEYWORDS : TOF , Pulmonary stenosis, septo aortic discontinuity, LVOT 'Y sign

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ABSTRACT

Aim: To elicit the key points in diagnosing the most common cyanotic prenatal congenital heart disease - Tetralogy of Fallot (TOF) by extended fetal echocardiography and to increase the rate of prenatal detection of Tetralogy of Fallot in a low-risk population.

Methods and Material: Our study is a prospective study done at our Fetal Medicine Research centre. During our study we screened about 15000 cases for prenatal congenital heart diseases in low and high- risk pregnancies between 20 and 22 weeks gestation by Extended Fetal echocardiographic examinations over a period of two and half years. Extended Fetal echocardiography includes the four chamber view and visualization of the left ventricular outflow tract(LVOT), the right ventricular outflow tract(RVOT), the main pulmonary artery and its branches by applying M- mode, Doppler and colour doppler techniques. We analyzed the abnormalities seen in various sonographic views.

Results: Out of 15000 cases screened by Extended Fetal Echocardiography, 8 cases were diagnosed as typical Tetralogy of Fallot (Absent pulmonary valve syndrome, Pulmonary Atresia , Ventricular septal defect with TOF were excluded) in low- risk mothers. The average age of the women was 28.25 (range 20 - 37) years. The mean gestational age at the first routine screening was 20.63 (20 -24) weeks; In our study, the specificity of ultrasound in detecting the typical TOF was 100% and also it had a good positive predictive value of 100%. There were no false-positive diagnoses (sensitivity 63.06% to 100.00% at 95% confidence interval, specificity 100%).

Conclusions : The main outcome of our Extended Fetal Echo is the detection of all prenatal typical tetralogy of Fallot cases during the second trimester scan(Level 2 scan) by the presence of septo aortic discontinuity , Dilated overriding of aorta and Y sign in Doppler in LVOT view irrespective of low or high- risk pregnancies. We emphasize the value of Extended fetal echocardiography in the prenatal diagnosis of typical tetralogy of Fallot which can improve the neonatal outcomes.

Introduction:

Background:

Tetralogy of Fallot(TOF) is the most common prenatal congenital cyanotic heart disease accounts for about 1/3600 live births and about 7% to 10% of all congenital heart disease and is the most common cause of cyanosis in the neonatal period [1,2]. Typical Tetralogy of Fallot (TOF) may be missed in Four chamber view. An abnormal Four chamber view is rarely seen in typical TOF cases . In the typical form of TOF the fetal echocardiography is often characterized by a normal four-chamber view, a subaortic Ventricular septal defect (VSD), Dilated over riding of aorta seen in an LVOT view, Pulmonary stenosis and an increased Aorta/ Pulmonary artery ratio, minimal antegrade flow in the main pulmonary artery and the aortic flow originated from right and left ventricle. All pregnant women with low- risk factors , suspected fetal cardiac anomalies and with increased risk factors should be analyzed by extended fetal echo during the level 2 scan (18 – 22 weeks scan). It has been very important to identify the side of the aortic arch also, which is quite easier in the prenatal period in three vessel view . This will be useful if surgery is planned postnatally. Extracardiac abnormalities like brain and renal anomalies may also be found in fetuses with TOF. These extracardiac defects are associated with chromosomal abnormalities. When TOF is diagnosed, serial follow up scans at 5 – 6 weeks intervals are adviced to know about the size of the pulmonary arteries and to reassess the direction of main pulmonary artery and ductus flow and to evaluate if seen .

Subjects and Methods:

Our study is a prospective study done at our Fetal Medicine Research centre in southern India. During our study we screened about 15000 cases for prenatal congenital heart diseases in low and high risk pregnancies between 20 and 24 weeks gestation by Extended Fetal echocardiographic examinations over a period of two and half years from June 2013 to December 2015. We examined the fetal heart in various imaging planes with continuous sweeping of the transducer from situs plane to Arch views, and routinely obtained the following six basic fundamental views: the transverse view of the upper abdomen, the four-chamber view, the three vessel view , the left ventricular outflow tract view, the right ventricular outflow tract view and the aortic arch view.

Results:

Out of 15000 cases screened by Extended Fetal Echocardiography, 8 cases were diagnosed as typical Tetralogy of Fallot (Absent pulmonary valve syndrome, Pulmonary Atresia ,VSD with TOF were excluded) in low risk mothers. The average age of the women was 28.25 (range 20 - 37) years(Table 1). The mean gestational age at the first routine screening was 20.62 (20 -24) weeks (Table 1). Four chamber view was obtained in all of the fetuses(100%) ; the extended fetal heart examination was completed in 14760 fetuses at the first sitting during the anomaly scan. Six cases of typical TOFs were diagnosed in routine anomaly scan screening at first sitting . Due to maternal obesity and Fetal position, this outflow tract views were postponed after 4 -7 days in the remaining 240 fetuses. During second sitting, two cases of TOF were diagnosed. In our study, the specificity of ultrasound in detecting the typical TOF was 100% and also it had a good positive predictive value of 100%. There were no false-positive diagnoses (sensitivity 63.06% to 100.00% at 95% confidence interval, specificity 100%). Only one case was associated with multiple extracardiac anomalies. Remaining 7 cases were not associated with any syndromes/extracardiac anomalies. The Extended Fetal Echo abnormalities are listed in Table 2 and shown in Figures (1 – 4). The four-chamber view was normal in 8 cases. The Left ventricular outflow tract and three-vessel views were abnormal in all 8 fetuses. The septo aortic continuity was

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lost and abnormal in all 8 cases. 3 cases of TOF show the right aortic arch.In all TOF cases ascending aorta is dilated and over riding the VSD.A Small pulmonary artery with antegrade or forward flow could be noticed in all cases. So the Aorta/MPA ratio were over 1.1. In ventricular systole blood from right ventricle and left ventricle forms the classical 'Y'sign in LVOT view.

Discussion:

Tetralogy of Fallot (TOF) is a common congenital cyanotic heart disease characterized by four distinct anatomical abnormalities: ventricular septal defect, overriding of the aorta, right ventricular outflow obstruction and right ventricular hypertrophy(RVH). RVH is uncommon in the fetal period. There are a number of different subtypes of tetralogy of Fallot. But we analyzed only the typical TOF cases.Prevalence of TOF in fetuses is much lower than that found in infants[3,4] which indicates that TOF may be missed or least diagnosed Congenital heart diseases during anomaly screening[5,6,7]. With an improvement in ultrasound equipment and operator experiences, as well as detailed dedicated attention to the segmental evaluation of the cardiac structures in all planes , it is possible to get an accurate diagnosis of typical tetralogy of Fallot. Of late, fetal echocardiography has become very much an important tool for the diagnosis of TOF and for the precise demonstration of Intracardiac fetal anatomy in developing countries. The inclusion of Extended fetal echocardiography in routine anomaly scan has a pivot role on detection of congenital heart disease since a larger proportion of prenatally detectable cases occur in a low-risk population. Four-chamber view detects major congenital heart disease, but it has some limitations in detecting anomalies involving the ventricular outflow tracts and/or great arteries, and therefore of tetralogy of Fallot[8,9-17]. This weakness can be compensated for if the examination is extended to include the ventricular outflow tract views[8,11-17]. Our Extended Fetal Echo study detected 8 cases of typical tetralogy of Fallot cases during the second trimester scan(Level 2 scan) by the presence of septo aortic discontinuity , Dilated overriding of aorta and Y sign in Doppler in LVOT view. Wiechec et al study showed "Y sign" at the level of three vessel and trachea view: a new sensitive early marker of tetralogy of Fallot[18] . TOF associated anatomical variations have different outcomes. Prenatal diagnosis of TOF aids parental counselling and enables delivery to be planned at a pediatric cardiac centre with timely initiation of proper management and treatment , which will improve treatment outcome. With an advancement of high tech ultrasound equipment , operator experience and detailed dedicated approach to the segmental evaluation of the prenatal cardiac structures, it is possible to obtain an accurate diagnosis of tetralogy of Fallot . In conclusion, prenatal ultrasound screening paves the ways for the diagnosis of a large proportion of severe cardiac defects .If ultrasound is performed only on indication, very few cases are diagnosed. When comparing countries with the same screening recommendations we found a large difference in prenatal detection rate. Prenatal diagnosis of TOF is useful for parental counselling and enables delivery to be planned at a good tertiary pediatric cardiac centre with early initiation of proper treatment , which will further improve the management outcome.

Conclusion :

Extended fetal echo should be done in all fetal anomaly scan (level 2 scan) irrespective of low and high risk pregnancies. If Fetal Echo is performed only in high risk cases, only very few cases are diagnosed with congenital heart disease. Of late, In countries like India particularly in southern India , there is a resurgence in detection rate of prenatal congenital heart diseases. Efforts should be made to accurately diagnose typical TOF prenatally by Extended fetal echocardiography. The main outcome of our Extended Fetal Echo is a detection of all prenatal typical TOF cases during the second trimester scan(Level 2 scan) by the presence of septo aortic discontinuity ,Dilated overriding of aorta and the Y sign in colour Doppler in LVOT view.

Table1 : Age and Gestational age of Prenatal mothers

Age	Gestational Age
33	20
22	21
28	21
20	22
31	20
26	21
37	20
29	20
28.25	20.62

Table 2 Extended Fetal Echocardiography views

Cardiac axis	4 C view	LVOT view	Septo aortic continuity	Y sign	3 V V	Arch	Size of aorta	Dilated, Overriding aorta	Size of MPA	Ao /MPA ratio
levo	N	Abn	Abn	Y	Abn	Rt	Enlarged 6.9mm	Y	Small 2 mm	3.45
normal	N	Abn	Abn	Y	Abn	Lt	Enlarged 7mm	Y	Small 2.2 mm	3.18
levo	N	Abn	Abn	Y	Abn	Lt	Enlarged 5.9 mm	Y	Small 1.9 mm	3.10
levo	N	Abn	Abn	Y	Abn	Rt	Enlarged 6.2mm	Y	Small 2.7 mm	2.24
normal	N	Abn	Abn	Y	Abn	Lt	Enlarged 7.2mm	Y	Small 2.2 mm	3.27
levo	N	Abn	Abn	Y	Abn	Rt	Enlarged 7.5mm	Y	Small 2.7 mm	2.77
normal	N	Abn	Abn	Y	Abn	Lt	Enlarged 6.6mm	Y	Small 2mm	3.33
levo	N	Abn	Abn	Y	Abn	Lt	Enlarged 7.4mm	Y	Small 2.2 mm	1.94

Levo- Extreme left axis deviation . N – Normal , Abn – Abnormal, Rt – Right aortic arch, Lt – Left aortic arch, Y – Yes, 4 C view – 4 Chamber view ,3VV-3vessel view , MPA-Main Pulmonary artery, Ao- Ascending Aorta.

Figure 1 : AORTA/MPA RATIO

Figure 2 &3 : OVERRIDING OF AORTA (colour doppler)

Figure 4 : 'Y' SIGN