Ultrasonographic Evaluation of the Fetal Heart

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Abstract

Congenital heart diseases (CHD) are the most common congenital anomalies, and most cases occur in the low-risk population. The majority of babies with congenital heart disease are born to mothers with no identifiable high-risk factors and so will not be detected unless there is widespread screening of the low-risk population. Congenital heart disease is eight times more common than trisomy 21 and four times more common than neural tube defects, the two conditions for which universal screening programs are in place. But, most of the countries in Europe do not perform routine fetal cardiac screening. Improvement of the technology, especially the improvement of the resolution of new ultrasound machines and gain of experience, is now possible to detect most forms of major congenital abnormalities during pregnancy.

Cardiac abnormalities are commonly overlooked during routine obstetric evaluation. There are vast variations in results between centers within the same country as well as between different countries despite using the same screening recommendations. Detection rates of congenital cardiac disease in low risk populations range from 8 to 48% (1). Total detection rate of congenital heart disease is 27.7% in EUROTUS Study and 25% in EUROSCAN Study (1,2). These numbers are under the general sensitivity rates of anomaly screening.

Detection rates depend on the two important factors. One of them is the experience of the sonographer and the other is whether the examination of the 4-Chamber View or Outflow...
Tracts and Aortic and Ductal Arc images have been performed or not. In general practice, high risk pregnancies are usually referred to tertiary centers for evaluation of the fetal heart. Risk factors for congenital heart defects originate both from the mother and the fetus. Maternal risk factors are positive family history of congenital heart diseases (CHD); maternal diabetes and exposure to teratogens in early pregnancy. Fetal high-risk factors include the detection of an extra-cardiac fetal anomaly on ultrasound, fetal arrhythmias, particularly complete heart block, non-immune fetal hydrops and increased nuchal translucency (NT) in the first trimester. However, out of a total of 2758 cases, 80% were referred to the fetal cardiology unit at London Guy’s Hospital as a result of screening of the low risk population (3). So in order to detect a large number of cases of congenital heart abnormality, there must be a system of screening the fetal heart in low the risk population.

There has been much controversy about whether ultrasound screening is of benefit. The conclusions of the RADIUS Study suggest that obstetric ultrasound screening did not improve perinatal mortality and morbidity and was not cost effective (4). But the Helsinki Ultrasound Trial demonstrated that in patients screened by ultrasound, there was a significant decrease in perinatal mortality and increased rate of detection of malformations and a decrease in total health care costs (5). The aim of this review is to make a critical assessment of the screening methods and of the importance of the screening of low risk populations in the first versus the second trimester. Diagnostic ultrasound modalities such as 4D ultrasound and STIC are beyond the scope of this review.

Importance of screening
Congenital heart disease is a common condition with a prevalence of 8 per 1000 live births (6). They are responsible for most infant and childhood deaths. The prenatal diagnosis may also carry a poor prognosis depending on the type of the malformation. Intrauterine fetal death occurs in 20 to 30% of the cases. Overall, 25 to 45% of the fetuses with congenital heart disease have other malformations and up to 35% of heart defects are associated with fetal aneuploidy (7-10). These associated findings can be important in counseling families about the chance of postnatal survival. According to the Eurocat Study, prevalence of trisomy 21 and open neural tube defects were 19.5 and 22.6 per 10000 birth respectively (11). Even congenital heart disease is four times more common than trisomy 21 and neural tube defects, but most of the countries in Europe do not perform routine screening. American Institute of Ultrasound in Medicine (AIUM) and American College of Obstetricians and Gynecologists (ACOG) recommend that the 4-chamber view be part of the routine prenatal sonography in the second and third trimesters (12,13). The International Society of Ultrasound in Obstetrics and Gynecology have published a guideline for performing the “basic” and “extended basic” ultrasound examinations. These guidelines can be used to evaluate the low risk fetuses that are examined as a part of routine screening (14).

Routine fetal cardiac screening; first trimester vs second trimester
Early prenatal diagnosis of the major defects is important for counselling patients about pregnancy options, therapeutic interventions, changes in obstetric care and alternative plans for delivery. For this reason fetal cardiac anatomy should be screened in detail during the first trimester by transvaginal or transabdominal probes.

There may be two types of screening strategy. One strategy is to screen patients identified to be at risk. Most cardiac malformations are missed if screening is done only on patients with risk factors. The other strategy is to screen the low risk population since more than half of prenatally detected cardiac defects are found in low risk population (3). The most predictive factor for detection of cardiac defects is the abnormal cardiac ultrasonography. So it is most important to know the normal cardiac examination in the first trimester and also in the second trimester.

In many countries, ultrasound examination is performed in the second trimester to look for congenital malformations as well as congenital cardiac defects. First, in France, the 4-chamber view of the heart is integrated into the routine obstetric scan performed between 18 and 22 weeks of gestation (15). Subsequently prenatal screening for congenital heart disease was introduced in the United Kingdom at south-east Thames region in 1988. In a 2.5-year period, 77% of all cardiac abnormalities were detected in 10 obstetrics centers. In the studies that were carried between 1990 and 2000, the sensitivity of 4-chamber view ranged between 30 and 52% in low risk and unselected populations (16). In Turkey, there is no universal screening strategy for detection of cardiac defects as well as of other malformations. Only the patients with previous malformation history are referred to first trimester cardiac screening. But low risk patients are recommended to be screened at 18 to 22 weeks.

The first trimester screening
The first trimester ultrasound examination has been an established method for several years to detect many chromosomal and structural abnormalities (17,18). Despite a few reports on the possibilities of visualizing and examining the fetal heart at this early gestational age, first trimester screening of cardiac defects in the late first trimester remains a rarely applied method (19-21). Major difficulty for early cardiac screening is misdiagnosis. In that case most of the parents will choose the option of termination of pregnancy. For that reason, the cardiac malformations, suspected in the first trimester should be confirmed at second trimester examination in case of suspicion.

In the first trimester; Haak et al. performed serially weekly transvaginal ultrasound examination in 85 singleton
pregnancies in 2002 (22). They studied the various cardiac plans such as the 4-chamber view, long axis of aorta and the pulmonary tract. Fetal heart was examined in 20% of the cases at 11 weeks but successful examination was obtained in 92% of the cases at 13-14th weeks. For this reason, the best time in the first trimester to perform transvaginal echocardiography is at 13+0 to 13+6 weeks of gestation. In transabdominal ultrasonography, successful examinations were obtained in 90% of the cases with crown-rump length (CRL) >60 mm. Body Mass Index (BMI) is also an important factor for successful examination. Mean maternal BMI is less than 24 kg per squaremeter in successful studies, in contrast; generally in patients with BMI over 26, examinations may be difficult and may result in technical failures (23).

In the first trimester scanning, whether performed via transvaginal or transabdominal probes, it is important to define what is normal at this stage of pregnancy. Normal means to see the 4-chamber view and to demonstrate the right sided and left sided symmetry. This can be obtained by visualization of the inflow and outflow tracts in B-mode and with colorflow mapping (Figure 1). In addition, if we demonstrate the septoaortic continuity, most major structural defects can be excluded (Figure 2). By additional use of color flow mapping to assess the intracardiac flow velocities, we can detect the valvular lesions such as aortic and pulmonary stenosis. But, these lesions may be mild in the first trimester. We can search for ductus venosus flow and the tricuspid regurgitation especially after the 12 to 13th weeks of gestation (Figure 3).

The two most common indications for scanning the fetal heart in first trimester are previous family history of cardiac defects and finding an increased NT thickness. Main goals of early scanning are reassurance of normality and option of early stage termination. If normality of situs and cardiac connections, atrioventricular junctions, right and left sided symmetry and septoaortic continuity can be demonstrated reliably, most of the major cardiac defects can be excluded. On the other hand, septal defects and developmental lesions may be missed, thus second trimester scans are needed for confirmation of normality (Table 1). The use of color flow mapping to evaluate intracardiac flow velocities will increase the detection rate of valvular lesions such as aortic and pulmonary stenosis (24). The use of power Doppler also facilitates visualization of the ventricular septum.

If the 4-chamber view of the fetus is asymmetrical, it should be considered abnormal and it is important to confirm the cardiac position accurately. If there is a mediastinal shift to the right with left sided apex, this is an early finding in cases of diaphragmatic hernia. If there is right-left asymmetry in cases of Turner syndrome, the diagnosis of coarctation of the aorta can be suspected (24).
As to whether the first trimester screening should be performed transabdominally or transvaginally, this decision on the route of examination depends upon the gestational week and resolution of the ultrasound machine. Most of the centers prefer transabdominal scan after 13 or 14 weeks. After 14 weeks of pregnancy two approaches are comparably successful. The only thing that can be seen at such an early stage is the symmetry of the 4-chamber view and cross over of the great arteries. We believe that from the patients’ perspective, transabdominal scan is more acceptable especially in our populations as most of the patients have the fear of abortion due to transvaginal examination despite its not being true. The goals of early scanning are the reassurance of normality and the termination of pregnancy at earlier gestation in case of a major cardiac abnormality.

Increased NT is known to be associated with an increased risk of aneuploidy, particularly trisomy 21, and recent studies have also identified increased NT as a nonspecific marker for various genetic syndromes and structural anomalies, to include congenital heart disease.

**Is NT measurement a useful screening tool?**

To find an answer to this question a multicenter study has been designed in Sweden (25). NT measurements were made in 16 383 euploid fetuses. Out of 127 cases of confirmed congenital heart defects, 55 were defined as major defects, of which 52 were isolated defects, meaning there were no other concomitant structural defects or chromosomal aberrations. Also, 72 were defined as minor cardiac defects. The sensitivity and the false positive rate for NT $\geq 95^{th}$ percentile were 13.5% and 2.6%, respectively; while for NT$>3.0$ mm, these values were 9.6%, 0.8%, respectively, and positive likelihood ratio was 12, meaning that the risk is increased 12 times when the NT measurement is over 3 mm (25).

According to these results, NT measurement is a poor screening method for isolated major cardiac defects. It appears that increased NT in the first trimester is associated with an increased risk of major fetal cardiac abnormalities in a high risk population. The role of NT screening in low risk populations is uncertain (26). Therefore a method with a much higher detection rate is needed. However women carrying fetuses with increased NT should be offered to fetal echocardiography.

**Is the first trimester scanning accurate?**

In a recent systematic review carried out at Birmingham in 2006, there were a total of 1243 mothers who had been given an ultrasound scan for major congenital heart disease in the first trimester. Four studies used a transabdominal approach, four studies used a transvaginal approach and two studies used both methods. The transabdominal approach had a higher sensitivity than the transvaginal route. Pooled sensitivity rate was 96% for transabdominal scans and 62% for transvaginal scans. Studies performed before the year 2000, had a lower sensitivity rate of 56%. In the studies carried out after 2000 the sensitivity rate was 92% for major heart defects such as atresia of the great arteries or valves or hypoplastic left heart syndrome (27).

Some cardiac abnormalities such as valvular stenosis or cardiac hypertrophy may develop later in pregnancy. Therefore, second trimester follow up should be performed for low risk patients. In Turkey, only the high risk population is referred to the second trimester cardiac screening. However, to achieve a high detection rates, high risk patients should be referred for first trimester screening and the low risk patients should be referred for universal screening at 18 to 22 weeks.

**Second trimester screening**

In 4-chamber view at 18 to 22 weeks of pregnancy, attention must be paid to the following features (28):
1. The heart occupies a third of the thorax. The apex points out of the left anterior thorax.
2. There are two atria of approximately equal size.
3. There are two ventricles of approximately equal size and thickness. Both ventricles show equal contraction (Figure 4 and 5).
4. The atrial and ventricular septa meet the two atriointeroventricular valves at the crux. Because of the septal leaflet of the tricuspid valve inserts slightly lower in the ventricular septum than the mitral valve, crux of the heart forms an offset cross.
5. The two atriointeroventricular valves (mitral and tricuspid) are observed to open equally.
6. The foramen ovale defect occupies the middle third of the atrial septum. Flap valve can be seen flickering in the left atrium.
7. The interventricular septum should appear intact.
8. The aorta arises from the left ventricle and the aortic outflow tract view also shows the upper ventricular septum is intact (Figure 6).
9. And finally, the pulmonary venous connections to the back of the left atrium should be identified.

The most common abnormalities detected on 4-chamber view examination are hypoplastic left heart syndrome and

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**Table 1. Cardiac lesions that may be missed in the first trimester**

<table>
<thead>
<tr>
<th>Developmental lesions</th>
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<tbody>
<tr>
<td>Mild aortic/pulmonary stenosis</td>
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<tr>
<td>Mild mitral/tricuspid valve abnormalities</td>
</tr>
<tr>
<td>Coarctation of the aorta</td>
</tr>
<tr>
<td>Rhabdomyomas</td>
</tr>
<tr>
<td>Cardiomyopathies</td>
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</tbody>
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<table>
<thead>
<tr>
<th>Septal defects and others</th>
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<tbody>
<tr>
<td>Ventricular septal defects</td>
</tr>
<tr>
<td>Atrioventricula septal defects</td>
</tr>
<tr>
<td>Tetralogy of Fallot with normal size pulmonary arteries</td>
</tr>
<tr>
<td>Abnormalities of pulmonary venous return</td>
</tr>
</tbody>
</table>
atrioventricular septal defects. At the left side, one can see the small left ventricle when compared with the right ventricle. And in the AV septal defects, instead of the windmill appearance there is a straight line across from side to side. Therefore leaflets are not offset.

But certain defects are easily missed on the 4-chamber view such as ventricular septal defects, atrial septal defects, coarctation of the aorta, tetralogy of Fallot, transposition of the great arteries. To detect such abnormalities aortic and pulmonary outflow tracts should be included in the screening procedure. During the examination of the outflow tracts the following should be noted:

1. Two arterial valves should always be seen.
2. To open out the left ventricular outflow tract, slight transducer angulation between the right shoulder and left hip should be performed.
3. The pulmonary artery arises from the right ventricle. It
is crossing over just anterior and cranial to the aorta (Figure 7).

4. In the transvers section, this view is known as the “three vessel view”. It shows the pulmonary artery in a long axis projections, with the aorta and superior vena cava lying to the right of it. The pulmonary artery is the largest vessel, the superior vena cava is the smallest one, and the aorta is in between them (Figure 8).

If the detailed 4-chamber view alone is scanned, flaw detection rate will be 40 to 50%. Adding the assessment of the ventricular outflow tract will increase the detection rate to 60 to 80%. All these assessments will take extra 5 minutes in normal cases. The inclusion of the examination of the outflow tracts will further increase the detection rate of cardiac abnormalities by 20% to 30% (28). The most common abnormalities detected on outflow tract examination are conotruncal anomalies such as transposition of the great arteries and tetralogy of Fallot (Figure 9).

In a recent study, importance of the examination of area behind the heart (ABTH) has been denoted. Under normal conditions, the only major vessel that can be observed behind the heart is the descending aorta. Descending aorta is positioned on the left side of the spine and on the same side as the cardiac apex (Figure 1 and 5). At least 10% of the fetal cardiac defects might be associated with abnormal findings in the ABTH (29). Berg et al. (29), from Bonn, Germany, examined in 2003-2005 retrospectively the congenital heart defects detected in 393 fetuses and 4666 fetal echocardiograms including 220 cases of CHD, between 2006 and 2007 prospectively. They found that 69 fetuses had abnormalities of the ABTH. In 28 fetuses, two equally sized vessels ran behind the heart. Of these, 26 had an interrupted vena cava with azygous continuation and two had total anomalous infracardiac venous connection. In 41 fetuses, only one vessel was visualized, but the descending aorta was positioned contralateral to the cardiac apex. Of these, 29 had levocardia with right descending aorta and 12 had dextrocardia with left descending aorta.

Different informations can be obtained from each part of the 4-chamber view. Objective structured screening of the fetal heart is more important than evaluating the four-chamber view as “normal” or “abnormal”. For this reason, we have to use a checklist for the fetal heart screen (Table 2). By using this approach the maximum amount of information will be obtained and recorded, even if the 4-chamber view cannot be completed.

To obtain a 4-chamber view we have to use some tools listed in Table 3. However, early or late gestational age, lack of amniotic fluid, fetal position, and maternal habitus make it impossible to obtain a complete 4-chamber view (30). For the best performance, higher frequency probes should be used. This is necessary for improved image resolution.

Table 2. A checklist for the fetal heart screen

<table>
<thead>
<tr>
<th>Four-chamber view</th>
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<tbody>
<tr>
<td>Aortic outflow tract</td>
<td>√</td>
</tr>
<tr>
<td>Pulmonary artery outflow tract</td>
<td>√</td>
</tr>
<tr>
<td>Four-chamber view components (When cannot obtain complete 4-chamber view)</td>
<td></td>
</tr>
<tr>
<td>Normal axis</td>
<td>√</td>
</tr>
<tr>
<td>Ventricles equal</td>
<td>√</td>
</tr>
<tr>
<td>Atria equal</td>
<td>√</td>
</tr>
<tr>
<td>Crux normal</td>
<td>√</td>
</tr>
<tr>
<td>Septum intact</td>
<td>√</td>
</tr>
<tr>
<td>Descending aorta is on the left side of vertebral column (ABTH)</td>
<td>√</td>
</tr>
<tr>
<td>Three vessel view</td>
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Table 3. Hints for the best performance to obtain a 4-chamber view

| Highest frequency probes should be used: 7-14 MHz as often as possible |
| Sector transducer |
| Harmonic imaging |
| Sepia coloring |
| Waiting until fetal lie improves |
| Rescheduling for another time within one week |
| With obese patients or when fetus is larger; rescheduling examination within two weeks |

Figure 9. Transposition of the great arteries with ventricular septal defect. LV: left ventricle, RV: right ventricle, Ao: aorta, PA: pulmonary artery, VSD: ventricular septal defect.
Conclusion

We can conclude that:
1. Despite the wide spread use of ultrasonography, so far, only 15 to 30% of infants with congenital heart defects are identified prenatally.
2. Congenital heart disease is eight times more common than trisomy 21 and four times more common than neural tube defects.
3. Screening strategies should be developed for the low risk population.
4. First trimester echocardiography can be performed at the 13-14th weeks successfully. TV or TA scans can be performed with 8 to 14 MHz probes. Symmetry of the 4-chamber view and visualization of outflow tracts and aortic arch are important images for screening of CHD.
5. Visualization of normal cardiac anatomy in the late first trimester can reduce the anxiety of parents.
6. Second trimester follow up should be performed for low risk patients. Because some cardiac abnormalities such as valvular stenosis or cardiac hypertrophy may develop later in pregnancy. Especially in Turkey, only high risk patients can be referred for 1st trimester screening, whereas low risk patients should also be referred for universal screening at the 18-22nd weeks.
7. In experienced hands, fetal echocardiography is a reliable tool for prenatal detection of cardiac defects. All patients should receive honest and accurate counselling about what can and cannot be identified at the examination.

References