Fetal heart defects: Potential and pitfalls of first-trimester detection

Asma Khalil, Kypros H. Nicolaides

Keywords:
Congenital heart defects
Ductus venosus
Echocardiography
Fetal
First trimester
Nuchal translucency
Tricuspid regurgitation

1. Introduction

Congenital heart defects (CHDs) account for one-third of all congenital anomalies and are the leading cause of infant mortality due to birth defects. They are commonly associated with fetal aneuploidy and genetic syndromes. In the last 30 years extensive studies have reported the prenatal diagnosis of cardiac defects during the second trimester of pregnancy. However, in the last 15 years, with the shift in screening for aneuploidies to the first trimester, extensive research has concentrated on early screening and detection of CHDs. Although the primary aims of the early ultrasound scan, which takes place at 11–13 weeks of gestation, are dating of the pregnancy, detection of multiple pregnancies and screening for aneuploidies there is increasing emphasis on the early detection of major defects. The advantages of early detection of major fetal defects include the possibility of scheduling additional assessment well before the limits for legal termination, the option for an earlier and safer pregnancy termination, and, in cases with a normal scan, earlier reassurance that a major defect is unlikely to be present.

This article reviews the detection of major cardiac defects during the first trimester of pregnancy including description of the markers which could help identify the high-risk group requiring specialist fetal echocardiography and the techniques which could improve the detection of these defects.

2. Detection rate of congenital cardiac defects in the first trimester

The results of screening studies providing data on the prevalence of cardiac abnormalities and the proportion detected in the first-trimester scan are summarised in Table 1. In most of these studies, all abnormalities were classified by the authors as being major. Most studies included only euploid fetuses but four included fetuses with aneuploidies. The combined data on specific groups of cardiac abnormalities and their early detection in euploid fetuses from 14 studies that provided such details are presented in Table 2. The early detection rate for the most common cardiac abnormalities varied from around 51% for hypoplastic left heart to 16% for coarctation of the aorta, 18% for tetralogy of Fallot and transposition of the great arteries. The largest study, involving 44 859 singleton pregnancies undergoing a first-trimester ultrasound scan as part of routine screening for aneuploidies, reported that the detection rate of major CHDs was 34%. The study reported that this scan led to the diagnosis of around half of the cases of double outlet right ventricle, hypoplastic left heart and transposition of the great arteries, around one-third of the cases of atrio-ventricular septal defect, coarctation of the aorta, tetralogy of Fallot and pulmonary atresia.
but none of the cases of ventricular septal defect, Ebstein anomaly, aortic or pulmonary stenosis, tricuspid atria or cardiac tumours.

A recent review of the published series with more than 1000 cases from 1993 to 2008, which included data from 36 237 pregnancies generated by eight centres, suggests that the overall detection rate of major congenital anomalies at 11–13 weeks is 29% (95% confidence interval: 25–33). The pooled detection rate of cardiac defects was 17% (10–25%). The authors suggested that the detection rate could be improved if the ultrasound assessment at the first trimester follows well-delineated protocols.

### Table 2

<table>
<thead>
<tr>
<th>Cardiac abnormality</th>
<th>Screening study</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>Coarctation of the aorta</td>
<td>0/1</td>
<td>0/3</td>
</tr>
<tr>
<td>Tetralogy of Fallot</td>
<td>1/2</td>
<td>0/1</td>
</tr>
<tr>
<td>Hypoplastic left heart</td>
<td>1/2</td>
<td>0/2</td>
</tr>
<tr>
<td>Transposition of the great arteries</td>
<td>0/2</td>
<td>0/1</td>
</tr>
<tr>
<td>Atrioventricular septal defect</td>
<td>0/3</td>
<td>0/7</td>
</tr>
<tr>
<td>Pulmonary stenosis</td>
<td>0/1</td>
<td>0/1</td>
</tr>
<tr>
<td>Aortic stenosis</td>
<td>1/3</td>
<td>0/2</td>
</tr>
<tr>
<td>Tricuspid atresia</td>
<td>0/1</td>
<td>0/2</td>
</tr>
<tr>
<td>Ebstein’s anomaly</td>
<td>0/1</td>
<td>0/2</td>
</tr>
<tr>
<td>Double outlet right ventricle</td>
<td>0/2</td>
<td>0/1</td>
</tr>
<tr>
<td>Anomalous pulmonary venous return</td>
<td>0/1</td>
<td>0/1</td>
</tr>
<tr>
<td>Mitral atresia</td>
<td>0/1</td>
<td>0/1</td>
</tr>
<tr>
<td>Interrupted aortic arch</td>
<td>0/1</td>
<td>0/1</td>
</tr>
<tr>
<td>Pulmonary atresia</td>
<td>0/1</td>
<td>0/1</td>
</tr>
<tr>
<td>Double inlet left ventricle</td>
<td>0/1</td>
<td>0/1</td>
</tr>
<tr>
<td>Common truncus arteriosus</td>
<td>0/1</td>
<td>0/1</td>
</tr>
<tr>
<td>Ventricular septal defect</td>
<td>0/1</td>
<td>0/1</td>
</tr>
<tr>
<td>Total</td>
<td>1/3</td>
<td>3/12</td>
</tr>
</tbody>
</table>

Adapted from Syngelakis et al.11

has a more important role in the first trimester. A systematic approach should be used which includes assessment of the fetal position and orientation, examination of the four-chamber view to assess heart size, position, chamber sizes and the crux, assessment of the tricuspid valve and slow sweep upwards towards the head from the four-chamber plane in order to identify the great arteries (Figs. 1 and 2).32

3.1. Assessment of the fetal position and orientation

The position of the abdominal aorta and inferior vena cava at the level of the diaphragm may be clear enough to determine the atrial situs. The stomach and cardiac apex can always be identified.

3.2. Examination of the four-chamber view

This should be assessed in both apical and transverse views. Colour flow mapping should delineate the flow into both ventricles and gives an indication of the ventricular size.

3.3. Assessment of the tricuspid valve

The presence or absence of tricuspid regurgitation (TR) is determined by pulsed-wave Doppler during fetal quiescence. The presence of TR is best detected by colour flow mapping. If TR is seen on colour, a sample volume of 2.0–3.0 mm is positioned above the tricuspid valve in an apical four-chamber view such that the angle to the direction of flow is <20°. The colour Doppler will demonstrate the direction of the regurgitation jet, which may vary its direction within the right atrium. Tricuspid regurgitation is diagnosed if it was found during at least half of the systole and with a velocity of >80 cm/s, since aortic or pulmonary arterial blood flow at this gestation can produce a maximum velocity of 50 cm/s (Fig. 3). Examples of CHD associated with tricuspid regurgitation are atrioventricular septal defect, Ebstein’s anomaly, and pulmonary atresia with intact ventricular septum.

3.4. Slow sweep upwards towards the head from the four-chamber plane

The left outflow appears first in the heart with concordant ventriculo arterial connections and continues as the aorta, initially directed towards the right shoulder. At a slightly higher level, the pulmonary artery arises anteriorly from the right ventricle and passes almost directly posteriorly, in continuity with the arterial duct. Slightly higher still, the aortic arch is seen close to the right side of the arterial duct as the two converge to meet the descending aorta. Colour flow mapping is useful in delineating the great arteries.

![Fig. 1](image-url)

Fig. 1. (a) Ultrasound image at 12 weeks demonstrating a normal four-chamber view with equal ventricles and normal offsetting of the atrio-ventricular valves. (b) Colour Doppler demonstrating a normal four-chamber view with forward flow and equal filling of both ventricles. (c) Colour Doppler demonstrating crossing of the aorta and the pulmonary artery (X sign). (d) Colour Doppler demonstrating forward flow and equal size of the aortic arch and the ductus arteriosus at their confluence (V sign); LV, left ventricle; RV, right ventricle; LA, left atrium; RA, right atrium. Adapted from Persico et al.35
Failure to visualise the two great arteries should raise the suspicion of a cardiac abnormality. Identification of a single blood vessel may be associated with a diagnosis of a common arterial trunk, aortic atresia or severe coarctation, pulmonary atresia with intact ventricular septum or tetralogy of Fallot. If the normal ‘crossover’ relationship between the aorta and pulmonary artery is not seen, transposition of the great arteries should be excluded.

4. Improvement in first-trimester imaging of the fetal heart

Increasingly, a combination of transabdominal and transvaginal sonography using high-frequency transducers (4–8 and 5–9 MHz, respectively) is used for detailed assessment of the fetal heart in the first trimester. Two strategies have been proposed: the first uses the transvaginal approach only when transabdominal cardiac views are suboptimal, and the second uses both transabdominal and transvaginal accesses as a routine, as it is felt that they provide complementary information.

Factors affecting the ability to detect fetal heart defects at 11–13 weeks can be divided into operator-dependent skills and equipment limitations. The necessary operator skills include good scanning technique in the detailed first-trimester assessment, competence in transvaginal scanning, a high level of expertise in fetal echocardiography, and technical efficiency in image optimization. The ultrasound machine used for assessment of the fetal heart in the first trimester should include a wide spectrum of modern image enhancers, such as high harmonics, low compounding and high levels of speckle reduction algorithm. Harmonic imaging improves spatial resolution to enable visualization of smaller objects, and contrast resolution to demonstrate fine

Fig. 2. (a) Ultrasound image at 12 weeks showing the absence of atrio-ventricular valves offsetting in a case of atrio-ventricular septal defect. (b) Ultrasound image at 12 weeks showing disproportion. LV, left ventricle; RV, right ventricle. Adapted from Persico et al.35

Fig. 3. An apical four-chamber view of the heart at 12 weeks of gestation, with the pulsed wave Doppler positioned across the tricuspid valve. Tricuspid regurgitation is diagnosed if it is found during at least half of the systole and with a velocity of >80 cm/s.
differences in greyscale. Compounding enhances contrast resolution and allows for better tissue differentiation. von Kaisenberg et al.33 have shown that good reproducibility and improved cardiac imaging in the first trimester could be achieved using the combination of harmonics and compounding. Speckle reduction algorithm results in imaging based on speckle suppression, edge enhancement and feature preservation. Dynamic range control and rejection control should be optimised for the particular patient to produce better contrast imaging.

In the first trimester the fetal heart is only 6–10 mm across. It is therefore essential to utilise the maximum available high-definition zoom box which creates a field of up to 24 mm × 29 mm in the axial view. This maximises spatial and temporal resolution. Lombardi et al.34 demonstrated that high-quality first-trimester cardiac views could be obtained using linear probes with frequencies of 6 and 15 MHz. In a prospective study, transabdominal fetal heart examination was performed by a well-trained obstetrician using a 9 MHz linear transducer (9L, Acuson Sequoia 512, Imagegate, Siemens, Erlangen, Germany) at 11–13 weeks of gestation and successfully identified 93.1% of the 58 major cardiac defects.35

Recent studies have reported that the use of spatio-temporal image correlation (STIC) in the first trimester is feasible and is likely to improve the detection of CHD in expert hands.36–38 STIC combines the advantages of volume imaging with the application of fetal echocardiography, where all of the basic cardiac views can be assessed along with secondary views in motion in multiple viewing planes. However, the angle of acquisition needs to be set according to the size of the heart, and the time of capture should be as short as possible.

5. Pitfalls in first-trimester imaging of the fetal heart

Some cardiac abnormalities are not evident until later in pregnancy, such as cardiac tumours, complete heart block and cardiomyopathies.32 Some forms of CHD, such as aortic and pulmonary stenosis, can progress into more severe malformations with advancing gestation, so may not have been obvious at 11–13 weeks. These cardiac abnormalities often go undetected, even at the routine anomaly scan in the second trimester.

 Adequate examination of the fetal heart may be hampered by technical difficulties such as image resolution, limited clarity in relation to the size of the structures being examined and fetal movements. The precise gestation of the first-trimester scan is important. Early detailed examination of the fetal heart is technically more difficult at 11 compared to 13 weeks of gestation. The overall success rate in the assessment of the fetal heart has been reported to be 45% at 11 weeks and 90% at 13 weeks.39 Similarly, successful examination of the four-chamber view, great arteries and three-vessel view was reported in 20% of cases at 11 weeks increasing to 92% at 13 weeks.40 Another study reported successful examination of the four-chamber view, great arteries, ascending aorta and ductus arteriosus in 43%, 56% and 62% of cases at 11, 12 and 13 weeks, respectively.41

6. First-trimester markers of congenital cardiac defects

Measures to improve the detection of cardiac abnormalities include the appropriate training of sonographers, extra time allocated to the scan and inclusion of detailed examination of the heart in the protocol. However, effective diagnosis ultimately depends on the examination being carried out by an expert in fetal echocardiography, as demonstrated by the experience with the second-trimester scan in the last 30 years. Therefore the major challenge in routine scanning is to identify easily recognizable markers of the high-risk group that can then be referred to the expert. The sonographic markers that have been investigated in their relation to CHDs are increased nuchal translucency (NT), abnormal flow in the ductus venosus (DV) and tricuspid regurgitation (TR).

6.1. Increased nuchal translucency

Increased NT was first shown to be strongly associated with the risk of CHD in a screening study involving 29 154 singleton pregnancies with chromosomally normal fetuses at 10–14 weeks of gestation.12 The study included 50 cases with major CHD and reported that increased NT above the 95th centile could achieve a detection rate of 56%. The prevalence of CHD increased with increasing NT thickness (3% and 20% in cases with NT 3.5–4.4 mm and ≥5.5 mm respectively). In this study increased NT was observed with all types of CHD, but there was a stronger association with left-sided abnormalities, such as hypoplastic left heart and coarctation of the aorta.12

The observation of increased NT in fetuses with CHD is not fully explained. Proposed mechanisms include narrowing of the aortic isthmus accompanied by narrowing of the aortic valve and ascending aorta, leading to diversion of more blood to the head and neck, heart failure due to the potential strain that the abnormalities impose on cardiac function at a stage of pregnancy when a high proportion of cardiac output is normally diverted to the head and neck.42 The latter theory was supported by the finding of increased cardiac mRNA expression of atrial natriuretic peptide and brain natriuretic peptide in fetuses with increased NT.43 However, in another study, which included 83 fetuses with CHD, Simpson and Sharland44 performed a retrospective quantitative analysis of cardiac size and left ventricular ejection fraction in fetuses with ventricular septal defects or hypoplastic left heart syndrome. The authors demonstrated that all measurements of cardiothoracic ratio and left ventricular ejection fraction were within the normal range and there was no significant difference between fetuses with increased and those with normal NT.

Recent studies have confirmed the findings reported in the earlier study by Hyett et al.15 A study of 6921 fetuses reported that the prevalence of CHD increases with the degree of thickening of the NT (Fig. 4). The prevalence of CHD was about 0.5% in fetuses with NT < median; 1% for NT between median and 95th percentile, 2% for NT between 95th and 99th percentiles and increased to 3.5%,

![Fig. 4. Nuchal translucency thickness distribution in fetuses with normal karyotype and cardiac defects. M, median; 95th, 95th centile. Adapted from Atzei et al.45](image-url)
nosed in the second trimester or postnatally. Table 1 shows the data
diagnosed at 11 weeks compared with 15.4% of the 78 diag-
osed in the second trimester or postnatally. Table 1 shows the data
on NT screening performance in the studies reporting on the
effectiveness of the first-trimester scan in the diagnosis of
major CHD.

A pooled analysis of data from major fetal echocardiography
centres concluded that finding NT ≥3.5 mm may lead to an earlier
diagnosis of all major types of CHDs.46 Earlier studies have
demonstrated a significant association between increased NT and
left heart lesions and septal defects.12,21 However, the multicentre
study, based on 637 CHDs, did not replicate this finding.46 In a
meta-analysis, involving 58 492 pregnancies which aimed to assess
the screening performance of NT for major CHD, the detection rate
was poor (31% for a false-positive rate of 1%).47 However, the
studies varied in their de-

The NT cut-off recommended to use for referral for specialist
echocardiography varies according to the local set-up and
facilities, in particular the access to specialist fetal echocardiogra-
phy service. In the study by Atzei et al.45 we have demonstrated
that the prevalence of major CHD in fetuses with NT above the 99th
centile (>30 per 1000) is substantially higher than in patients with
a family history of cardiac defects and diabetes mellitus (about 20
per 1000), which are commonly used as indications for fetal
echocardiography.

Fetal NT >3.5 mm is found in about 1% of pregnancies. The risk
of major chromosomal abnormalities in such fetuses is very high
and increases from about 20% for NT of 4.0 mm to 33% for NT of
5.0 mm and 60% for NT of ≥5.5 mm.48 Consequently, our proposed
management would be first to offer the parents the option of fetal
karyotyping by chorionic villus sampling. The prevalence of major
fetal defects or fetal death in the chromosomally normal group
increases with NT thickness from about 10% for NT of 4.0 mm to 20%
for NT of 5.0 mm and 50% for NT of ≥5.5 mm.49 The next step
would be to carry out a detailed scan, including fetal echocardi-
ography at 14–16 weeks of gestation in the fetuses with normal
karyotype with increased NT.45

In fetuses with NT between the 95th and 99th centiles, the
prevalence of cardiac defects is about 2%, which is similar to that
found in patients with a family history of cardiac defects and dia-
abetes mellitus. The extent to which specialist fetal echocardiogra-
phy should be offered to these pregnancies, which constitute about
4% of the total population, depends on the availability of such
services.

6.2. Abnormal ductus venosus blood flow

Abnormal DV flow was initially reported in the second and third
termers in association with cardiac dysfunction associated with
structural heart defects, post-tachycardia cardiomyopathy and end-
stage fetal hypoxia or increased right ventricular afterload.50,51 In
hearts with markedly impaired diastolic function, atrial contraction
occurs against increased impedance to forward flow, resulting in a
transient flow reversal in the ductus venosus, which constitutes the
negative a-wave. However, DV flow reversal beyond the first
trimester has been noted mainly in situations where there were other
manifestations of cardiac dysfunction, such as fetal hydrops.

In the assessment of ductus venosus a right ventral mid-sagittal
view of the fetal trunk is obtained and colour flow mapping is used
to demonstrate the umbilical vein, ductus venosus and fetal heart. A
small pulsed Doppler sample (0.5–1.0 mm) is used to avoid
contamination from the adjacent veins and it is placed in the
yellowish aliasing area which is the portion immediately above the
umbilical sinus. The insonation angle should be <30°, the filter
should be set at a low frequency (50–70 Hz) to allow visualization of
the whole waveform and the sweep speed should be high (2–3 cm/
s) so that the waveforms are widely spread. Impedance to flow is
assessed by measuring the pulsatility index or by qualitative clas-
sification of the a-wave into positive, negative or reversed (Fig. 5).

The first study reporting on the association between abnormal
DV flow and CHDs in the first trimester demonstrated reversed or
absent flow during atrial contraction in 10 of 140 euploid fetuses. Major CHDs were present in six of the 10 with abnormal DV flow but in none of the 134 with normal flow. In a meta-analysis including seven studies (n = 50354) regardless of the NT status, nine studies (n = 2908) with increased NT and seven studies (n = 47610) with normal NT, the summary sensitivity and specificity of abnormal DV flow in the detection of CHDs were 50% and 93%, 83% and 80%, and 19% and 96% respectively. The corresponding positive likelihood ratio of the test was 8.1, 4.35 and 4.97 and the negative likelihood ratio was 0.52, 0.20 and 0.8, respectively.

Maiz et al. have studied the additive effect of DV velocimetry and reported that the risk of CHD, based on NT measurement, was three-fold higher or two-fold lower depending on the presence of reverse or positive a-wave, respectively (Fig. 6). A recent study assessed the best method of combining NT and DV Doppler in the detection of major CHD in euploid fetuses. The study included 37 fetuses with a major CHD and 12799 unaffected pregnancies. The authors demonstrated different detection rates depending on the cut-offs of NT or DV PI used (Table 3).

The mechanism of abnormal DV flow in fetuses with CHD is not clear. Similar to increased NT, the proposed theories include cardiac dysfunction. It was proposed that DV reversed flow corresponds to similar abnormalities described in inferior caval vein blood flow. However, the normal inferior caval vein blood flow is reversed, corresponding to atrial contraction, and abnormality will be manifest as a quantitative rather than a qualitative change. By contrast, in DV forward flow throughout the cardiac cycle is normal and any flow reversal is abnormal. Obstructive right heart defects have been reported to be more frequently associated with abnormal DV flow and normal NT, suggesting that the pathophysiology of the reversed a-wave could be attributed to a right heart overload and diastolic dysfunction.

6.3. Tricuspid regurgitation

Tricuspid regurgitation (TR) is a frequent finding in trisomic fetuses at 11–13 weeks of gestation and it is also observed in euploid fetuses with CHDs. The underlying mechanism for the association between CHD and TR, as well as increased NT and DV abnormal blood flow, has not been fully explored. The proposed theory, similar to that of NT and DV, is impairment in cardiac function that is manifested only during the first trimester because, at this gestation, the compliance of the fetal heart is low and cardiac afterload resulting from placental resistance is high.

The performance of TR in the detection of major CHDs was examined in a screening study at 11–13 weeks of gestation which included 85 cases with major CHDs and 40905 without CHDs. The prevalence of TR was ~1% in normal fetuses and in one-third of those with major CHD. The incidence of TR and DV reversed a-wave increased with NT thickness both in fetuses with and in those without major CHD. NT above the 95th centile, TR or DV reversed a-wave were observed in 35.3%, 32.9%, and 28.2% of the cases with major CHD, respectively, and in 4.8%, 1.3%, and 2.1% of those without CHD. Any one of the three markers was found in 57.6% of CHD cases and in 8% of those without.

![Fig. 6](image-url) Relationship between nuchal translucency (NT) thickness in chromosomally normal fetuses and risk of major cardiac defects. The a-priori NT-related risk (solid line) is multiplied by the positive and negative likelihood ratios for abnormal (upper dashed line) and normal (lower dashed line) A-waves in the ductus venous, respectively, to derive the adjusted risk. Adapted from Maiz et al.
examination was performed by a well-trained obstetrician using a high-frequency linear transducer in 886 cases. TR was reported in 16 (61.5%) euploid fetuses with CHD and in 62 (9.2%) of 670 in euploid fetuses with normal heart. Similar results have been reported in first-trimester study in 4445 pregnancies. There was a higher prevalence of TR in fetuses with CHD compared with normal fetuses (33% vs 1.7%). In the same study the corresponding values for NT above the 95th centile and abnormal DV flow were 38%, 5.6%, 22% and 3.1%, respectively.

Two approaches have been proposed for the use of the algorithm combining NT, DV Doppler and TR to estimate the patient-specific risk for major CHD. The first one is to define the risk cut-off that selects the patients requiring referral for specialist fetal echocardiography. The risk increases exponentially with NT thickness from 1 per 1000 in those with NT at or below the 95th centile to 7 per 1000 for NT between the 95th and 99th centile and 58 per 1000 for NT above the 99th centile. The risk is further increased if there is DV reversed a-wave, TR, or both and is decreased if flow in the DV and across the tricuspid valve is normal. The second approach is to define as high risk all cases with TR, DV reversed a-wave, or both, which constitute ~3% of the population and contain 48% of those with major cardiac defects. If cases with nuchal translucency above the 99th centile are also included, the screen-positive rate would increase to ~4% and the estimated detection rate would be 52%. If there are available resources for performing fetal echocardiography in 8% of the population, then the NT cut-off for defining the high-risk group could be reduced to the 95th percentile with an increase in the estimated detection rate to 58%.

### 6.4. Maternal serum markers and congenital cardiac defects

A case–control study of 68 cases of isolated fetal CHDs and 340 normal controls at 11–13 weeks of gestation reported lower maternal serum placental growth factor (PLGF) levels in CHD (0.80 vs 1.00 multiple of median). This decrease in PLGF was observed in conotruncal and valve defects but not in left heart defects. The decrease in serum PLGF was not related to impaired placental perfusion.

A case–control study of 306 cases of fetal CHDs and 1224 no-CHD controls reported abnormal second-trimester serum α-fetoprotein (AFP), human chorionic gonadotrophin (hCG) and unconjugated estriol (uE3) in the CHD group. Cases with critical CHDs were more than twice as likely to have AFP multiple of the median (MoM) >95th centile and/or an hCG and/or uE3 MoM ≤5th centile.

The value of first- and second-trimester maternal serum biochemical markers in screening for fetal CHDs remains to be determined.

### 7. Conclusion

First-trimester detection of CHD is feasible, but early detailed assessment of the fetal heart requires a high level of expertise in early anomaly scanning and fetal echocardiography. However, the detection of major CHDs at 11–13 weeks is influenced by their association with easily detectable markers and a policy decision as to the objectives of this scan and the allocation of resources necessary to achieve them. The use of transvaginal ultrasound and newer techniques are likely to improve the detection rate. However, the limitations of fetal echocardiography in the first trimester must be borne in mind, and resort to follow-up mid-gestational echocardiography should always be considered.
**Practice points**

- The detection rate of CHD at the first trimester is low and varies according to the type of the centre and the population studied.
- The detection rate varies according to the type of the cardiac abnormality, e.g. from around 51% for hypoplastic left heart to 18% for tetralogy of Fallot and transposition of the great arteries.
- The detection of major CHDs at 11–13 weeks could be improved if we use easily detectable markers for screening for CHD, e.g. nuchal translucency.
- The detection rate could be improved if the ultrasound assessment at the first trimester follows structured protocols.
- The detection rate of CHD could be improved by the use of transvaginal ultrasound and newer techniques.
- The limitations of fetal echocardiography in the first trimester must be borne in mind, and resort to follow-up mid-gestational echocardiography should always be considered.

**Research directions**

- The development of algorithms for the screening for CHD in the first trimester, using a combination of maternal and pregnancy characteristics, nuchal translucency, ductus venosus Doppler and tricuspid regurgitation.
- Prospective assessment of the routine implementation of tools, such as transvaginal ultrasound and STIC, for improving the detection rate of CHD.

**Conflict of interest statement**

None.

**Funding sources**

The study was supported by a grant from The Fetal Medicine Foundation (UK Charity No: 1037116).

**References**


