FETAL CARDIAC MALFORMATIONS

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The underlying problem

Congenital heart disease affects 6–8 per 1000 live births, at least half of which should be detectable before birth because of their serious nature.

However, if examination of the fetal heart is confined to traditional high risk groups only about 20% of babies born with congenital heart disease will be identified.
Therefore the focus must be to examine all pregnancies for cardiac malformations.
The opportunity to do so is present in some centers at 11–14 weeks when nuchal fold thickness is measured to assess the risk of chromosomal defects in the fetus.

However, most pregnant women are assessed for the first time in detail by sonographers at about 20 weeks in their local obstetric ultrasound department.
Transvaginal scanning of the heart is favored in some centers as part of the first trimester scan at 11–14 weeks, and impressive views can be obtained because of the close proximity of the probe to the fetal heart when the fetal lie is Favorable.

Transabdominal fetal cardiac scanning is more commonly performed.
Diagnostic views can be obtained at 14 weeks using modern probes with limits of resolution of about 50 µm in the axial plane at 6 MHz and less than 100 µm in the lateral plane, at normal obstetric scanning depths.
A curvilinear probe with a cardiac package incorporating fast frame rates is ideal, although standard cardiac probes may also be used which have the advantage of continuous wave Doppler available to interrogate high velocity atrioventricular valve regurgitation or severe semilunar valve stenosis.
Color flow mapping and Doppler may be helpful adjuncts to early diagnosis but their use should be limited in the first trimester to avoid any possible damage to the developing fetus.
NEW IMPROVEMENTS IN IMAGING

Two dimensional imaging is still the most commonly used imaging modality in fetal echocardiography.

Most probes in use have very fast frame rates, achieved by parallel processing where the transducer transmits one line and receives two, resulting in a doubling of previous rates.
More advanced techniques such as three dimensional rendering and spatial compounding may be of use in the future, but currently are used in more general imaging settings and as research tools.
COLOUR DOPPLER IMAGING

Color flow mapping and particularly power Doppler are helpful in imaging flow in the very small heart.

The advantages of color flow mapping are that it allows assessment of valve opening and of symmetry in ventricular filling, in cases where visualization using the grey scale alone, even with harmonic imaging, is difficult.

It may highlight areas of turbulence or of important atrioventricular valve regurgitation that provide important clues to major structural defects during first trimester scanning.
Fetal cardiac function

An assessment of fetal cardiac function can be made using traditional M mode to provide information on wall thickness and ventricular shortening fraction, but it is somewhat crude.

Fetal long axis function may provide additional insight into endocardial function, most usefully in the detection of early ischemic changes before the development of sonographically detectable endocardial fibroelastosis (EFE).
Long axis recordings show the importance of atrial function in the fetal heart, and Doppler tissue imaging (DTI) provides a transmyocardial tissue velocity profile and permits assessment of radial and longitudinal strain rate, but the timing of cardiac events cannot be verified with a concurrent ECG recording and currently depends on surrogates such as mitral valve closure signals.
Left ventricular DTI has shown increased duration of early diastole and reduced endocardial velocities in children with obstructive lesions (such as aortic stenosis) before obvious abnormalities of function are apparent.
In the future these methods may be helpful in determining the timing of interventions such as either early delivery of the fetus, or of in utero interventions such as fetal aortic balloon valvuloplasty, before irreversible damage to the endocardium occurs.

Congestive cardiac failure is seen in the fetus during tachyarrhythmias caused by the rise in systemic venous pressure and reduced ventricular filling time, but may also be seen secondary to viral myocarditis and restrictions to flow—for example, at the foramen oval level.
Associated findings:

- may include an increased cardiothoracic ratio,
- severe atrioventricular valvar regurgitation,
- reversal of flow in the systemic veins at end diastole,
- the development of effusions or of fetal hydrops.

A “heart failure score” has been developed using this profile and methods such as the Tei index, which is afterload independent, may prove useful in serial assessment of the fetus with hydrops.
It is clear from these methods that fetal cardiac function is not assessed from measures of intracardiac function alone but by Doppler assessment of the fetal circulation, which provides a more complete evaluation of the effects of cardiac preload and afterload on fetal wellbeing.
The salient features of the fetal circulation that differentiate it from the postnatal cardiovascular system and make Doppler such a useful tool are:

- the arterial and venous ducts,
- the foramen oval,
- the aortic isthmus,

all of which close or differ functionally after birth.
With increased pulmonary venous return after birth, left atrial pressure rises and the oval foramen becomes functionally closed, or if it remains patent flow reverses to become a left to right shunt. In addition, flow through the aortic isthmus increases with a concomitant increase in size. An understanding of the role of these structures in the fetus helps with functional assessment and in serial assessment of the fetus with structural heart defects.
Malformations
Schematic Diagram of Fetal Circulation

- HEAD AND FORELIMBS
- CRANIAL VENA CAVA
- LUNGS
- AORTA
- ABDOMINAL AORTA
- REST OF BODY
- LIVER
- PLACENTA

Legend:
- **highly oxygenated blood**
- **mixed blood**
- **poorly oxygenated blood**

UV = umbilical vein
LV = left ventricle
UA = umbilical artery
LA = left atrium
DV = ductus venosus
RA = right atrium
DA = ductus arteriosus
FO = foramen ovale
PA = pulmonary artery
(A) A full examination of the fetal heart may be obtained by five transverse sections through the abdomen and chest of the fetus.
The first section shows abdominal situs (B) with the aorta (Ao) to the left of the spine and the inferior caval vein (IVC) anterior and to the right. The normal fetal stomach (St) and heart lie on the left side.
The second section (C) illustrates the four chambers of the heart with the left atrium (LA) in front of the spine and the right ventricle (RV) just below the sternum.
The third cut (D) shows the aorta arising centrally in the heart from the left ventricle (LV)
The fourth the pulmonary trunk (PV) arising from the anteriorly placed right ventricle and crossing to the fetal left over the ascending aorta (E)
The fifth section shows the anteriorly positioned ductal arch (D) and the transverse aortic arch (Ao) to be of equal size traversing back to the fetal spine (F).
A normal variant “three vessel” view is shown with a right sided aortic arch and persistent left superior caval vein (LSVC). The trachea (T) can be seen lying between the aortic (Ao) and ductal (D) arches (G).
Additional Views
Coronal view of the inferior bridging leaflet of the common valve in an unbalanced atrioventricular septal defect
Sagittal views of the superior (SVC) and inferior caval (IVC) veins draining to the right atrium (RA)
the aortic arch
Coarctation of the aorta may manifest as early ventricular disproportion with persistent enlargement of the right ventricle (RV)
1. Lesions that **may be** associated with an abnormal four-chamber view

   a. **At the venous-atrial junction**
      - total anomalous pulmonary venous drainage*
   b. **At the atrioventricular junction**
      - mitral atresia
      - tricuspid atresia
      - atrioventricular septal defect
      - Ebstein’s anomaly/tricuspid valve dysplasia

*Although TAPVD is potentially detectable prenatally, it is a very difficult diagnosis to make in-utero.
tricuspid atresia
hypoplastic left heart syndrome may be diagnosed from these five screening views
c. At the ventriculo-arterial junction
   – aortic atresia
   – pulmonary atresia with intract interventricular septum
   – critical aortic stenosis
   – coarctation of the aorta

d. Other
   – ventricular septal defects
   – cardiomyopathy
PA-Valve
pulmonary atresia with intact ventricular septum
2. Lesions that may be associated with a normal four chamber view

- transposition of the great arteries
- double outlet right ventricle
- tetralogy of Fallot
- pulmonary atresia with a ventricular septal defect
- common arterial trunk
- absent pulmonary valve syndrome
Children with Tetralogy of Fallot exhibit bluish skin during episodes of crying or feeding.

“Tet spell”
tetralogy of Fallot
transposition of the great arteries may be missed on a four chamber view
tetralogy of Fallot may be missed on a four chamber view
3. Lesions likely to be overlooked pre-natally

- persistent arterial duct
- secundum atrial septal defect
- milder forms obstructive lesion of great arteries (milder forms aortic stenosis, pulmonary stenosis and coarctation of the aorta)
- some forms ventricular septal defects
Three-dimensional (3D) echocardiographic analysis of congenital heart disease in the fetus: comparison with cross-sectional (2D) fetal echocardiography

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**Conclusions** Three-dimensional imaging of fetal heart disease is feasible for a wide range of lesions, and may provide additional information of clinical value in a small number of cases when compared with 2D imaging.
PA with intact septum
Figure 3  Surface-rendered three-dimensional ultrasound images of a fetus at 29 weeks' gestation demonstrating gross aortic dysplasia of the tricuspid valve and right atrial dilatation. In (a) the phased 3D four-chamber view is seen in systolic frame. Both atrioventricular valves are displayed in a three-dimensional cross section from an atrial en face view during diastole in (b). FO, foramen ovale; IVS, interventricular septum; LA, left atrium; MV, mitral valve; RA, right atrium; RV, right ventricle; TV, tricuspid valve.
Figure 4  Reformatted three-dimensional sonographic long-axis view of tetralogy of Fallot in a fetus of 26 weeks demonstrating the aorta overriding a large defect of the outlet ventricular septum (a). A similar section through the area of override shows the biventricular origin of the aorta at a foreshortened systolic frame (b). AA, ascending aorta; IVS, interventricular septum; LA, left atrium; LV, left ventricle; MV, mitral valve; RV, right ventricle; VSD, ventricular septal defect.
Figure 5  Three-dimensional surface reconstruction of a four-chamber view from a fetus of 22 weeks' gestation with a muscular inlet ventricular septal defect (VSD). DAO, descending aorta; IVS, interventricular septum; LA, left atrium; LV, left ventricle; pul. vein, pulmonary vein; RA, right atrium; RV, right ventricle.
Figure 6  This three-dimensional posterior cross section of a 26-week fetal heart simulates removal of the anterior wall of the heart. The atrioventricular septal defect is clearly visualized together with the inferior bridging leaflet (IBL). IVS, interventricular septum; LV, left ventricle; RV, right ventricle; VSD, ventricular septal defect.
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