Antenatal diagnosis of fetal heart disease

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Antenatal diagnosis of congenital heart disease is most commonly made at the routine 20-week anomaly scan. Not all abnormalities can be detected by prenatal ultrasound but detection can be improved by obtaining outlet views and by the use of colour Doppler. This article provides an overview of the uses and limitations of fetal echocardiography.

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The antenatal diagnosis of congenital heart disease is most commonly made at the routine 20-week anomaly scan. The four-chamber view is traditionally used. The average antenatal detection rate of major congenital heart disease in the UK is 23% (Bull, 1999). It has been suggested that this rate could be improved by adding visualization of the ventricular outflow tracts to the four-chamber view (Hunter et al, 2000; Carvalho et al, 2002).

LESIONS DETECTABLE ON FOUR-CHAMBER VIEW

The four-chamber view of the heart locates the cardiac apex to the left and on the same side as the stomach. It allows assessment that the cardiothoracic ratio is less than 50% and the two atria and two ventricles are of equal size. The atrioventricular valves are identified, with the tricuspid valve inserting a little lower in the interventricular septum than the mitral valve.

This view will recognize fetuses with a disproportion between the atria or ventricles such as left or right ventricular hypoplasia, or marked right atrial enlargement resulting from severe tricuspid regurgitation associated with Ebstein's anomaly of the tricuspid valve or pulmonary atresia with an intact ventricular septum. It will also show atrioventricular septal defects (*Figure 1*), very large ventricular septal defects, cardiomyopathy and intracardiac lesions such as rhabdomyomas (*Table 1*).

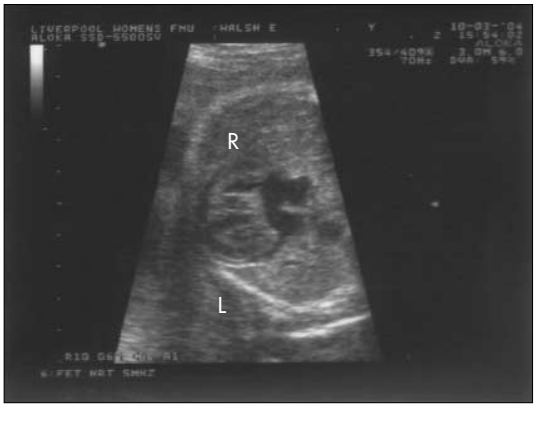
TABLE 1.
Common cardiac abnormalities detected on four-chamber view

Abnormality	Comment
Hypoplastic left heart syndrome	
Atrioventricular septal defect (AVSD)	80% of complete AVSDs are associated with Down syndrome as are 40–50% of partial AVSDs
Large ventricular septal defects	Usually associated with other cardiac or extracardiac abnormalities, particularly trisomy
Hypoplastic right ventricle	Usually seen in association with tricuspid atresia or pulmonary atresia with intact ventricular septum
Right atrial dilatation	Caused by Ebstein's anomaly of the tricuspid valve or pulmonary atresia with intact ventricular septum
Right ventricular dilatation	Occurring with coarctation of the aorta or severe tricuspid regurgitation
Left ventricular dilatation	Found in some cases of critical aortic stenosis
Double inlet left ventricle	Where both atrioventricular valves enter the left ventricle. The right ventricle is usually hypoplastic
Dilated cardiomyopathy	Dilated heart with poor ventricular function and atrioventricular valve regurgitation
Hypertrophic cardiomyopathy	May be associated with Noonan's or other syndromes. Sometimes familial.
Rhabdomyomata	Appear as discrete masses in either or both ventricles. Commonly associated with tuberous sclerosis

LESIONS NOT DETECTABLE ON FOUR-CHAMBER VIEW

Cardiac lesions which include abnormalities of the great vessels such as pulmonary atresia with ventricular septal defect, tetralogy of Fallot, transposition of great arteries, double outlet right ventricle, common arterial trunk and some ven-

Figure 1. A complete atrioventricular septal defect. The common valve is closed. The atrial defect affects septum primum.



tricular septal defects will not be visible in the four-chamber view (*Table 2*).

These can be detected when views of the ventricular septum below the great vessels and ventriculoarterial connections are obtained (*Figures 2* and *3*).

LESIONS THAT MAY NOT BE DETECTABLE ANTENATALLY

In some conditions such as pulmonary or aortic stenosis and coarctation of the aorta, a discrepancy in chamber or great vessel size increases with advancing gestation. However, mild degrees of stenosis may never be apparent antenatally. Total anomalous pulmonary venous drainage is difficult to detect antenatally as only about 7% of the circulation passes through the lungs and little flow is detected in the pulmonary veins before birth. The detection of small ventricular septal defects and minor valve abnormalities depends on the resolution of the ultrasound equipment and the skill of the fetal echocardiographers. Conditions such as atrial septal defect and patent arterial duct only become apparent postnatally, as these features are normal in the fetus.

INDICATIONS FOR SPECIALIST FETAL ECHOCARDIOGRAPHY

There are important maternal and fetal factors that increase the risk of congenital heart disease and indicate the need for specialist fetal echocardiography (*Table 3*). The incidence of congenital heart disease in some of these conditions is low. The main benefit of fetal echocardiography is parental reassurance. Further scans may be required for some progressive conditions, particularly left heart obstructive lesions.

MANAGEMENT OF AN ANTENATALLY DIAGNOSED CARDIAC LESION

A detailed examination of the rest of the fetus should be undertaken if a fetal cardiac abnormality is found, as 25% of children with a congenital heart defect have an associated malformation, often as part of a multiple malformation syndrome (Lacro, 2000). Studies have shown that as many as 20% of all infants with congenital cardiovascular malformations have chromosomal abnormalities (Harris et al, 2003). Most of these infants have Down syndrome. Cardiac malformations are found in 40–50% of individuals with Down syndrome. Extracardiac malformations and chromosomal abnormalities may adversely affect the cardiac outlook. Fetal karyotype can be performed to exclude aneuploidy (trisomy 13, 18 or 21). Deletions of chro-

TABLE 2.
Common cardiac abnormalities not detected on four-chamber view

Abnormality	Comment
Transposition of the great vessels	
Tetralogy of Fallot	
Pulmonary atresia with ventricular septal defect	
Common arterial trunk	
Ventricular septal defect situated below the aortic or pulmonary valve	
Double outlet right ventricle	Where both the aortic and pulmonary valves arise from the same (usually right) ventricle
Coarctation or interruption of the aortic arch	

Figure 2. Short axis view of pulmonary artery (PA) and duct. The pulmonary trunk divides into the left PA (arrow) and the duct. The aorta is in cross section in the centre of the image.

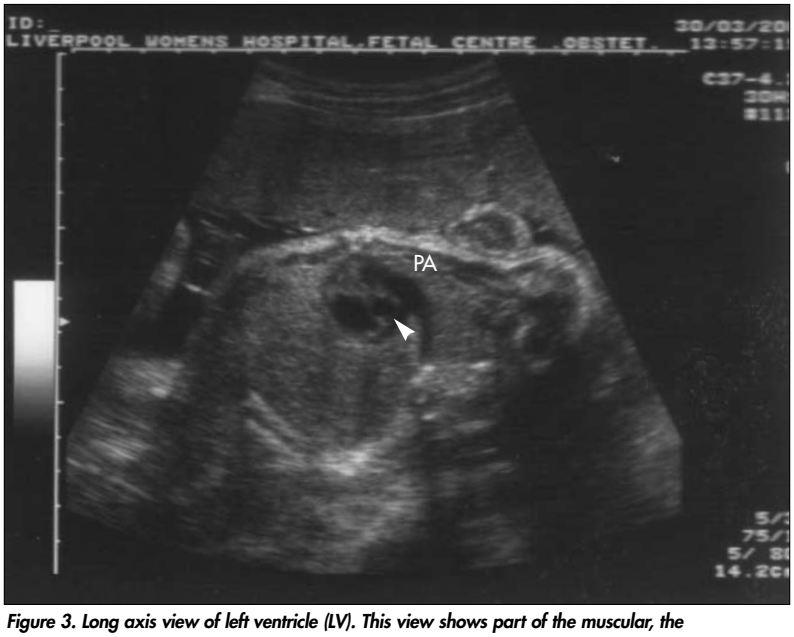
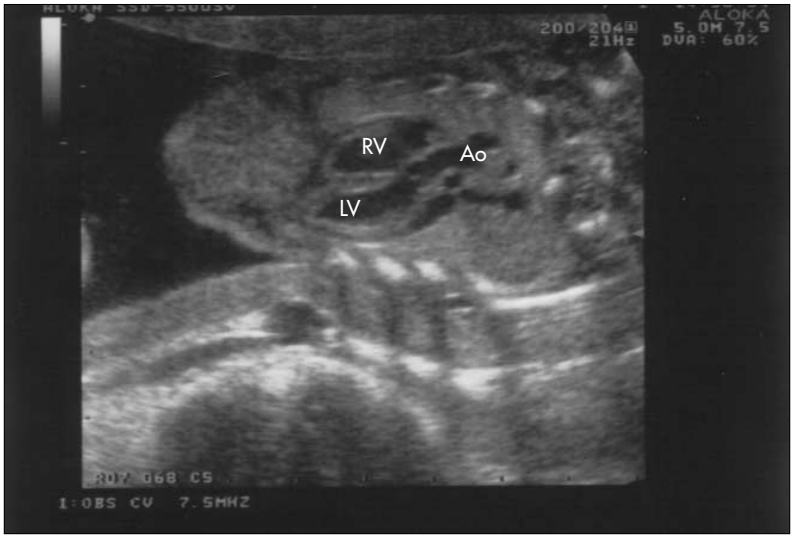


Figure 3. Long axis view of left ventricle (LV). This view shows part of the muscular, the perimembranous and outlet portions of the interventricular septum. Ao = aorta; RV = right ventricle.



mosome 22 can also be detected by fluorescence in-situ hybridization (FISH) analysis and should always be requested in the presence of significant fetal heart disease. Conotruncal malformations such as type B interruption of the aortic arch, truncus arteriosus and tetralogy of Fallot are overrepresented in individuals with 22q11 syndrome.

When fetal heart block is detected, measurement of the maternal auto-antibodies (anti-Ro and anti-La) should be performed. It is likely that the heart is structurally normal if these are present but a 30% recurrence risk will be indicated if maternal antibodies are present.

Antenatal detection of major congenital heart disease allows parents time to consider their options. Some families choose to terminate the pregnancy. The termination rate is reported as being between 30 and 60% in various studies (Fesslova et al, 1999; Brick and Allan, 2002). If most of the severely affected fetuses are terminated, survival rates in the remainder are likely to be higher. Termination rates have fallen as the results of surgery have improved. This is particularly the case when a diagnosis of hypoplastic left heart syndrome is made, as the results of surgery have improved markedly over the last 10 years. Stillbirths occur in approximately 10% of fetuses with congenital heart disease (Fesslova et al, 1999).

Prenatal diagnosis of congenital heart disease helps to prepare those parents who continue the pregnancy. It also facilitates appropriate postnatal management and avoids haemodynamic deterioration in those infants with duct-dependent cardiac lesions. The effects of this may be reflected in a reduction in the severity of late neurodevelopmental problems, but this is yet to be proved.

Fetal diagnosis also has implications for perinatal management. Most fetuses with congenital heart disease can be delivered according to normal obstetric parameters, the exceptions being some fetal arrhythmias where intrapartum monitoring may be compromised. Fetuses with known transposition of the great arteries should be delivered close to a paediatric cardiac centre to allow urgent atrial septostomy if the atrial septum is very restrictive. In most centres in the UK, fetuses with duct-dependent lesions are delivered in tertiary neonatal or cardiac centres.

TABLE 3.
Indications for specialist fetal echocardiography

Maternal and familial	Mother, partner or previous child with congenital or inherited heart disease
	Parental consanguinity
	Mother or partner with 22q11 deletion
	Maternal insulin-dependent diabetes
	Mother taking drugs such as lithium, sodium valproate or alcohol
	Tuberous sclerosis in first-degree relative
Fetal	Increased fetal nuchal translucency (>3.5 mm)
	Other congenital abnormality, e.g. exomphalos, congenital diaphragmatic hernia, tracheo-oesophageal fistula, bowel obstruction
	Chromosomal abnormality
	Multiple congenital abnormalities
	Cardiac anomaly detected on routine ultrasound scan, i.e. abnormal four-chamber view
	Fetal bradyarrhythmia or tachyarrhythmia
	Multiple monochorionic pregnancy

OUTCOME OF ANTENATALLY DIAGNOSED CONGENITAL HEART DISEASE

Antenatally diagnosed lesions are usually at the severe end of the spectrum of congenital heart disease, and these infants also have a much higher incidence of chromosomal and extracardiac abnormalities. Most deaths in infants with antenatally diagnosed congenital heart disease occur in the neonatal period, but there is a further small risk of death during infancy and beyond.

There is conflicting evidence about whether antenatal diagnosis of duct-dependent lesions significantly improves survival (Sullivan, 2002). Some studies have indicated a better outcome in those lesions suitable for a biventricular repair (Copel et al, 1997). Mortality in transposition of the great arteries was found to be significantly lower when diagnosed antenatally (Bonnet et al, 1999). A small study of coarctation diagnosed antenatall found a reduced morbidity and improved survival (Franklin et al, 2002). The prognosis of antenatally diagnosed hypoplastic left heart syndrome remains unclear. Brackley et al (2000) found that infants with antenatally diagnosed hypoplastic left heart syndrome had lower survival than those diagnosed postnatally. Tworetzky et al (2001) found a better survival and reduced morbidity in an antenatally diagnosed group. Andrews et al (2001) found similar survival rates in both groups.

FACTORS AFFECTING PROGNOSIS

Trisomy 13 and 18 are usually fatal early in the neonatal period. Fetal hydrops in association with congenital heart disease or cardiomyopathy is a poor prognostic indicator. Multiple fetal

abnormalities or haemodynamically significant arrhythmias associated with congenital heart disease carry a worse prognosis. Infants with Down syndrome, a cardiac lesion and growth retardation have only a 60% 1-year survival (Wessels et al, 2003). If intrauterine death is included this reduces to 40%. Complete heart block with an atrial rate of less than 100 per minute or a ventricular rate of less than 45 per minute carries a poor prognosis (Gardiner, 2001).

THE FUTURE

Antenatal ultrasonographic diagnosis of significant congenital heart disease is possible in around 50% of cases using two-dimensional ultrasound and by incorporating cardiac scanning into the second trimester fetal anomaly scans. Adding visualization of the ventricular outflow tracts to assessment of the four-chamber view and the use of colour flow Doppler improves the detection rate (Carvalho et al, 2002). More recently three-dimensional imaging has been shown to be particularly useful in the diagnosis of fetal heart disease. It may become an important complement to conventional fetal echocardiography. Currently, however, the imaging quality depends entirely on the skill of the operator. More development of this technology is required before it becomes routine practice. HM

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KEY POINTS

- The four-chamber view is traditionally used.
- Outlet views improve detection rates.
- Duct-dependent lesions require tertiary referral centre management.