Antenatal Detection of Heart Defects is Important and Achievable

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Congenital heart disease occurs in about 1 in 125 live births, approximately half of which require urgent evaluation and management. Cardiovascular malformations account for 10% of infant deaths and nearly half of all deaths from all malformations. They are, therefore, both important and common, but are still frequently missed in primary screening programmes worldwide. The UK average detection rate is 23% with wide postcode variation. The reasons for missing defects are difficult to ascertain. However, with 'hands-on' training of obstetric sonographers in their own unit, detection levels may increase to 70–80%. With ongoing support these levels are sustainable. This 'hands-on' training uses a systematic protocol, the five transverse views, which allows the majority of defects to be detected at the routine 20-week scan without increasing routine examination time.

Key Points

- Congenital heart disease (CHD) occurs in 6–8 per 1000 live births
- Cardiovascular malformations account for 10% of infant deaths and nearly half of all deaths from malformations
- Almost 4800 pregnancies were affected by 'serious' CHD in 1994, but only 23% were detected before birth with wide postcode variation, ranging from 0 to 71%
- Effective on-site training of obstetric sonographers, increases detection at the 20-week scan to about 80% without increasing routine examination time
- Prenatal detection of CHD is important to allow choice, discover co-morbidities and plan for a safer perinatal period for babies with duct-dependent lesions
- Prenatal detection of CHD is cost-effective as it rationalizes the use of emergencies services and allows better planning of healthcare delivery

Introduction

Congenital heart disease (CHD) occurs in 6–8 per 1000 live births. Cardiovascular malformations account for 10% of infant deaths and nearly half of all deaths from malformations. They are, therefore, both important and common, but still frequently missed in screening programmes worldwide.

Major congenital heart defects have been classified in practical terms as those requiring treatment by surgery or catheter intervention during the first year of life. Important cardiac defects include those with absent valves, chambers or arteries, and may be identified by disproportion at mid-trimester. Moreover, they are often ductal dependent and, therefore, babies are at risk of collapse and death once the duct closes. This group of babies are most likely to benefit from an antenatal diagnosis.

Fetal Cardiac Screening 'Myths'

Screening programmes should find important cardiac defects in 4/1000 pregnant women. Several factors may contribute to the failure to reach this detection level. The obstetrics screeners may feel that it is too difficult to detect heart defects because they have received insufficient training, have inadequate equipment or too little time. They may feel CHD is easier to detect after delivery and that if nothing can be done during pregnancy, why worry the family with the diagnosis earlier than necessary?

Neonatal Detection of CHD

Congenital heart defects are difficult to detect during the routine clinical examination of the newborn. Several studies have been published demonstrating that of those neonates with heart defects, 55% had a normal neonatal examination (i.e. were ' missed'), and this 45% detection was similar in both large and small centres.[1]

About one-third of those found to have abnormal clinical signs were not referred appropriately for investigation or to a cardiologist, but brought back for a later clinical review in a paediatric clinic. In this region, overall 82% of babies with CHD were sent home without a diagnosis being made. This is likely to be representative of most regions.

What Happens to Babies with CHD?

The overall outcome for babies with CHD is improving. A proportion will have multiple congenital abnormalities or chromosomal defects, but overall about 83% survive. However, those with duct-dependent lesions are in danger of collapse and death if their heart disease is unrecognized. In another report from the Northern region, 10% died within 24 hours because of ductal closure and one-third of those dying in infancy died from unrecognized CHD.[2] Neonates with hypoplastic left heart syndrome and pulmonary atresia, or with very poor myocardial function will die early, while those with malformations of the arch, such as interrupted aortic arch and coarctation of the aorta and aortic stenosis more often present with collapse or death after the first week.

Antenatal Detection

Reports of the efficacy of antenatal detection vary widely depending on the centre and obstetric practice (whether

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private office examination or nationwide screening).[3,4] A multicentre study was instituted by the British Paediatric Cardiac Association to determine the efficacy of screening for CHD in the UK. There were almost 4800 pregnancies affected by 'serious' CHD in 1994, but overall only 23% of those born with CHD were detected before birth. Postcode analysis revealed a widespread of results, ranging from 0 to 71%. The consequences of a missed diagnosis included increased morbidity and mortality due to delayed postnatal diagnosis and an increased, avoidable use of emergency services. There were 15 postcodes with above average detection and these were areas with a history of active teaching programmes.

Technical Reasons Restricting Success at Screening

Most obstetric ultrasound departments now have good ultrasound equipment and Trusts recognize the importance of including a rolling programme of renewal into their budgets. However, sometimes imaging is poor. Fetal and maternal factors contribute to this, and are difficult to overcome. Training in examination of the heart can give examiners confidence to recognize normality. Referral to a more experienced examiner, such as a perinatal cardiologist, is justified if these structures are not identified. Examination of the details of cases that reach the law courts often reveals a trail of re-examinations within the department, rather than referral to a specialist centre.

Training Views: The Five Transverse Planes

There is a simple and systematic method of examining the fetal heart that is easy to teach and to incorporate into the level 2 anomaly scan.[5,6] Teaching materials (posters and CDs) and hands-on training, subject to resources, are available from echo charity uk (www.echocharity.org.uk) to help sonographers, obstetricians and paediatric cardiologists in training.

The views comprise examining the way the fetus is lying to establish the left and right of the fetus, and then examine the relative position of the aorta and inferior caval vein in the abdomen (abdominal situs) as this gives important information about possible duplication of right- or left-sided structures in the fetus (isomerism). The planes within the heart can be examined by sweeping from the 'four-chamber' to the aortic root (sometimes called a five-chamber view) and then towards the fetal head to the pulmonary artery that leads into the arterial duct, from there, moving cephalad to the 'three-vessel view', where the transverse aortic arch, the ductal arch, the superior caval vein and trachea can be seen (Fig. 1).

During this sweep the most important features to look for are symmetry, if the scan is at about 20 weeks. Abnormalities of cardiac size, pericardial or pleural effusions, an irregular rhythm and abnormal location of the heart within the chest may be detected.

A more detailed examination of the four chambers of the heart is required to check whether the chambers are morphologically right- or left-sided structures. There are special features of each chamber that characterize its right- or left-sidedness; the ventricles are defined by their inlet valves so the right ventricle has a tri-leaflet, tricuspid valve with septal attachments that is usually displaced more towards the apex of the ventricle than the bi-leaflet 'fish mouth' mitral valve, thus forming the normal 'offset'. The atriums are defined sonographically by the shape of their appendages (a hooked little finger on the left and a broad triangular one on the right) and by the presence of the coronary sinus running behind the left atrium (Fig. 2a,b).

Outcome of Cardiac Training in Obstetric Screening Programmes

Detection rates can improve if sonographers are supported in their screening programmes with appropriate training and feedback, and given the opportunity to see malformations. However, not all sonographers work near congenital heart disease centres and so opportunities are limited for some. This is where training aids, such as CDs that show abnormalities and training courses, are helpful.

We audited the outcomes of 34,332 consecutive pregnant women who underwent routine anomaly scanning between 20 and 24 weeks gestational age in our obstetric unit over an 8-year period. They were screened by sonographers with different degrees of training experience in ultrasound, but the four-chamber view was assessed in all. During the first 3 years (1997–2000) the ventricular outflow tracts were assessed only by sonographers trained in these views (approximately 50%), but by 2003 the five transverse views (abdominal situs, four chambers, both outflow tracts and three-vessel view) of the fetal heart were examined by all sonographers.

One-hundred-and-twenty cases of major CHD occurred (4/1000 screened women) of which 96 (80%) were detected prenatally. The detection rate and missed cases (24/120) for the different types of CHD are illustrated in the Table 1. Year by year, detection improved from 50% in 1997 to above 80% by 2000 and has been sustained at this level for 5 years.

Does Fetal Cardiac Scanning Increase Screening Time?

Initially, extra time is required for sonographers to become practiced at performing the five transverse views. However, with practice, we found that sonographers became more confident in determining fetal lie, establishing situs and then performing a sweep up the fetal chest. With increasing confidence a full examination can be performed without increasing the overall time.

Why are Some Lesions Still Missed?

In order to improve detection, it is important to understand why some lesions are 'missed' at screening. The cases missed in our department included those detectable at the 'four-chamber' view, such as VSD and AVSD, as well as those that usually require examination of the outflow tracts such as simple transposition of the great arteries, Tetralogy of Fallot and double outlet right ventricle. There were no easily identifiable reasons for missing those detectable on four-chamber views, but comments recorded by the sonographers included difficult fetal lie and maternal habitus. As in all departments staff turnover was variable from year-to-year. Cases characterized by disproportion such as coarctation of the aorta were usually referred for review, but progressive lesions such as isolated aortic and pulmonary valvar stenosis were more difficult to detect at 20 weeks as the signs may be subtle or absent.

In Bull's UK review, only about 38% of atrioventricular septal defects AVSD were detected, and this has recently been confirmed by others.[7] This is a lesion that usually has a very characteristic four-chamber appearance (Fig. 3) and is of

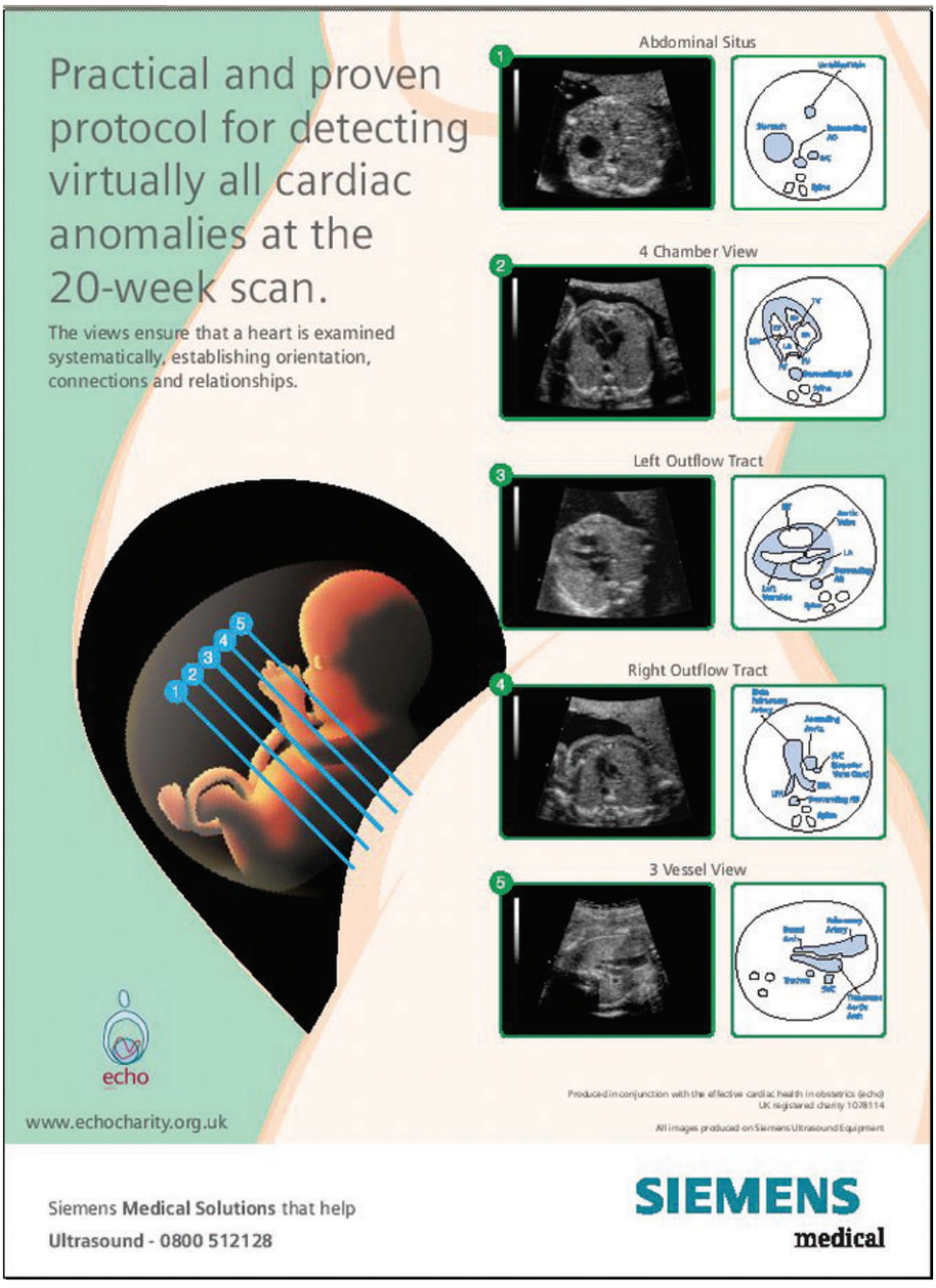
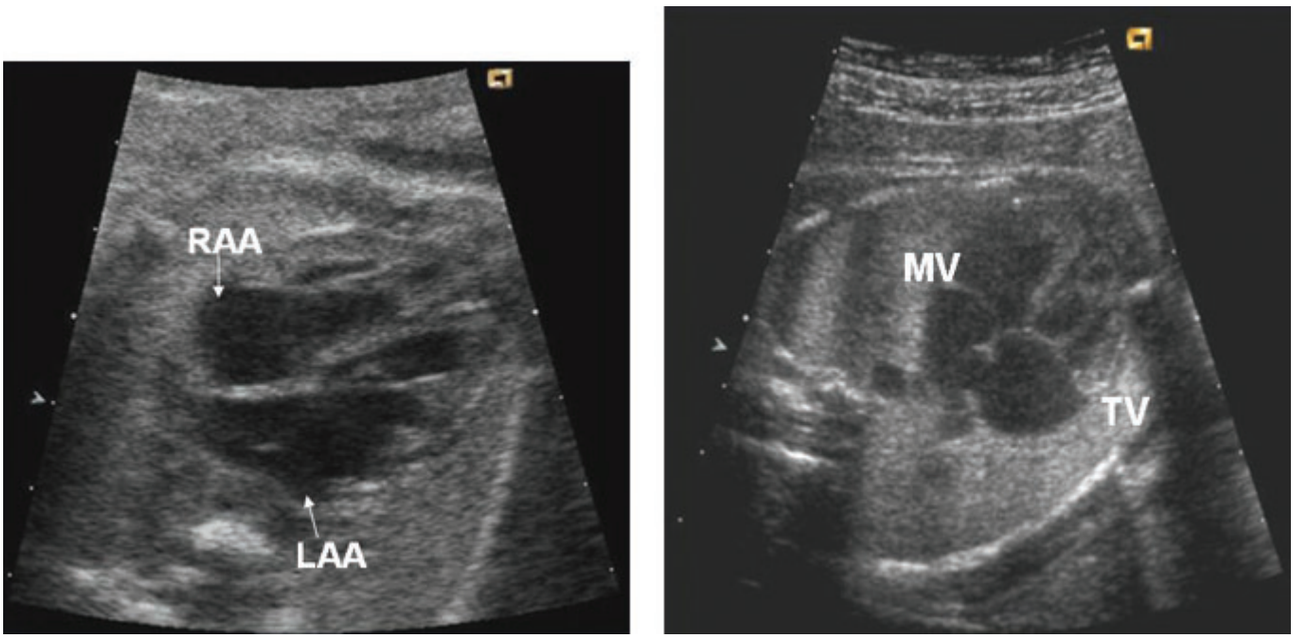
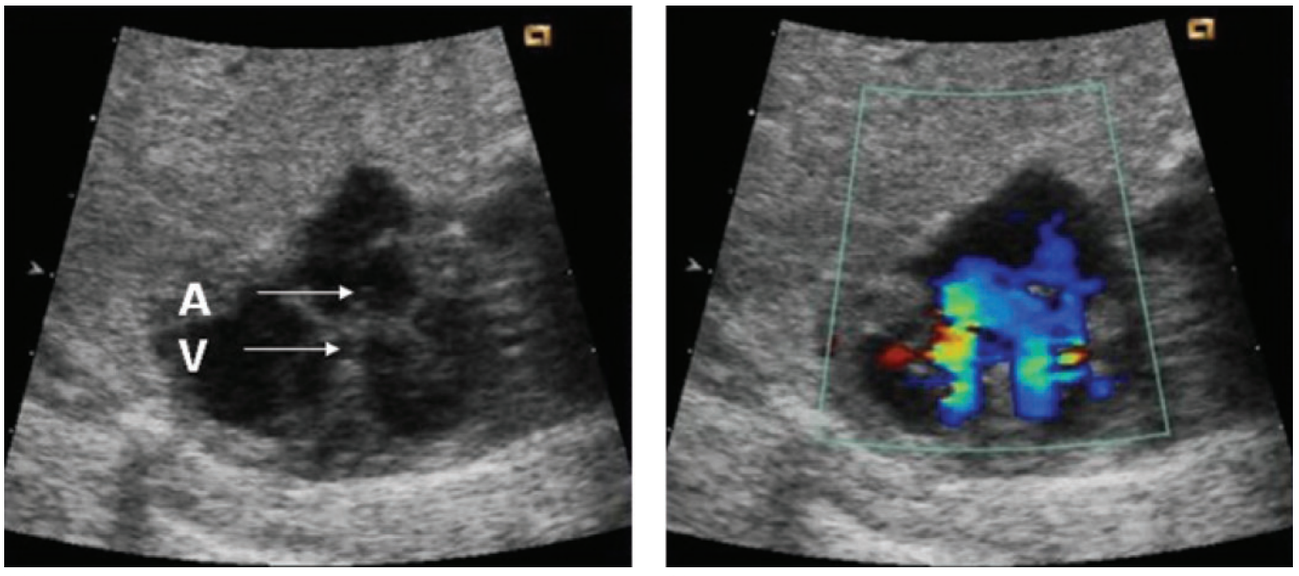


Figure 1. Poster produced to show the 'five transverse view protocol'. These five views through the fetal chest and abdomen show abdominal situs, the four chambers and great arterial crossover up to the transverse aortic and ductal arches, the 'three-vessel view'.



(a) (b)

Figure 2. (a) Characteristic morphological features of the atriums with hooked left atrial appendage (LAA) and a broad triangular right appendage (RAA); (b) characteristic morphological features of the ventricles include a tri-leaflet, tricuspid valve (TV) in the right ventricle displaced more towards the apex than the bi-leaflet 'fish mouth' mitral valve (MV), thus forming the normal 'off-set'.



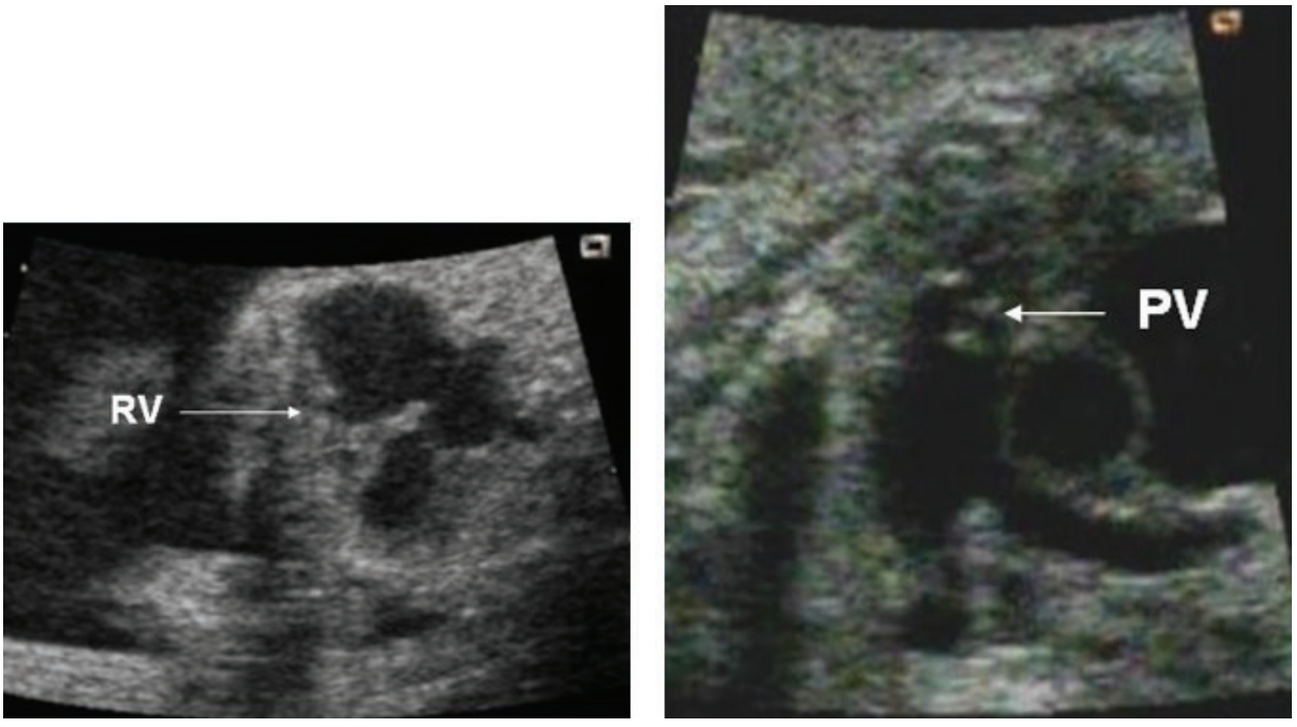
(a) (b)

Figure 3. (a) Four chamber view of an atrioventricular septal defect (AVSD) showing a common junction and valve. (b) Colour flow demonstrates shunting across the moderate atrial (A) and ventricular (V) components of the defect.

Table 1. Detection rate and missed cases (24/120) for the different types of CHD

Congenital heart defect	Detection rate % (number)	Missed cases (% of total detected)
Tetralogy of Fallot	75% (18/24)	6/24 (25%)
Transposition of great arteries	60% (9/15)	6/15 (40%)
Ventricular septal defects	78·5% (22/28)	6/28 (21·5%)
Atrioventricular septal defects	61·5% (8/13)	5/13 (38·5%)
Double outlet ventricle	92% (11/12)	1/12 (8%)
Hypoplastic heart	100% (10/10)	0 (0%)
Other*	100% (18/18)	0 (0%)

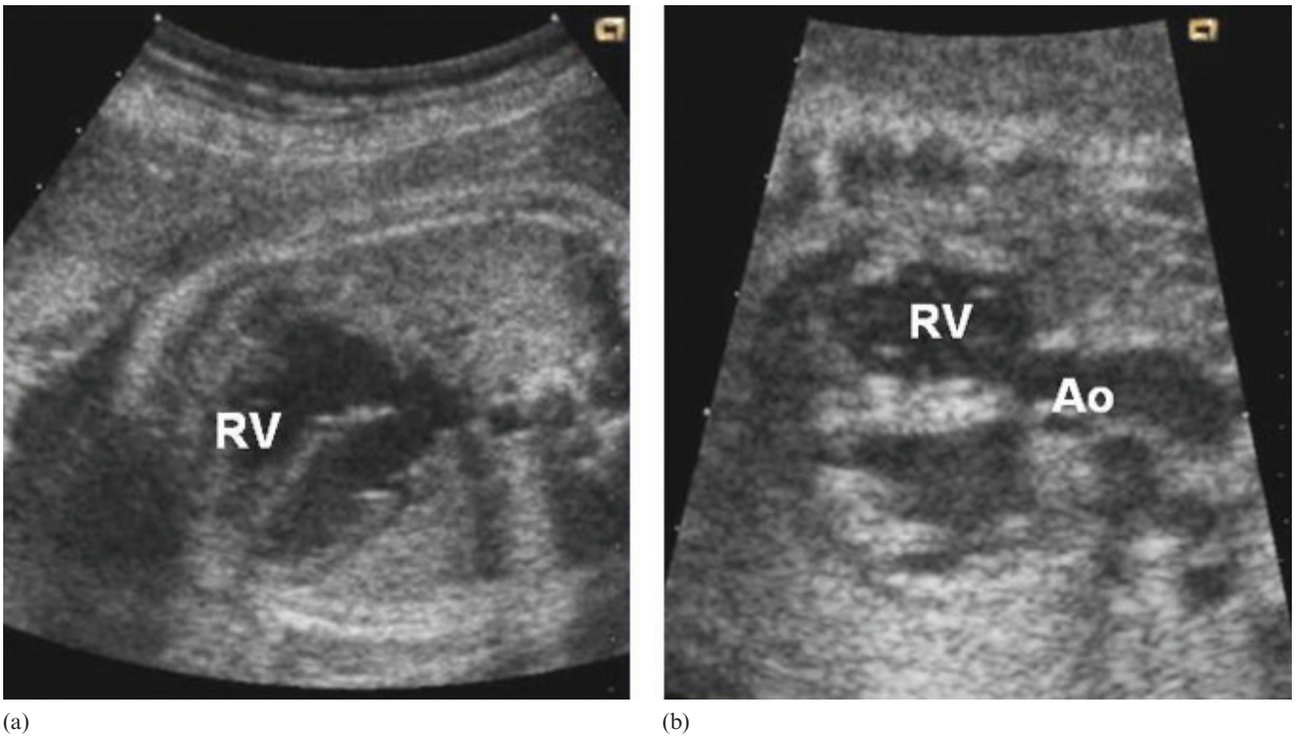
*Other includes: coarctation of aorta, pulmonary stenosis/atresia, truncus arteriosus, tricuspid atresia.



(a) (b)

Figure 4. Four chamber view of pulmonary atresia with intact ventricular septum (PAIVS) showing disproportion with right-sided enlargement and hypertrophy of the right ventricle (RV). The pulmonary trunk may be of good size and the pulmonary valve (PV) show membranous atresia that is opened by balloon valvuloplasty.

tremendous importance because of its association with trisomy 21, yet antenatal detection remains surprisingly low. Another lesion presenting with a very abnormal four-chamber appearance is pulmonary atresia with intact ventricular septum (PAIVS) and just over half (55%) were detected overall in the UK (Fig. 4). This is a duct-dependent lesion and antenatal detection allows optimization of perinatal care for these babies. Lesions that require examination of the outflow tracts were more rarely detected in Bull's study and included Tetralogy of Fallot (TOF) with only 11% antenatal detection (Fig. 5) and simple Transposition of the Great Arteries where $< 3\%$ were detected.



(a) (b)

Figure 5. (a) Four chamber view in Tetralogy of Fallot (TOF) may appear normal, but there may be left axis deviation and a perimembranous ventricular septal defect; (b) the third transverse plane at the level of the aortic root (Ao) demonstrates the large outlet ventricular septal defect with aortic override.

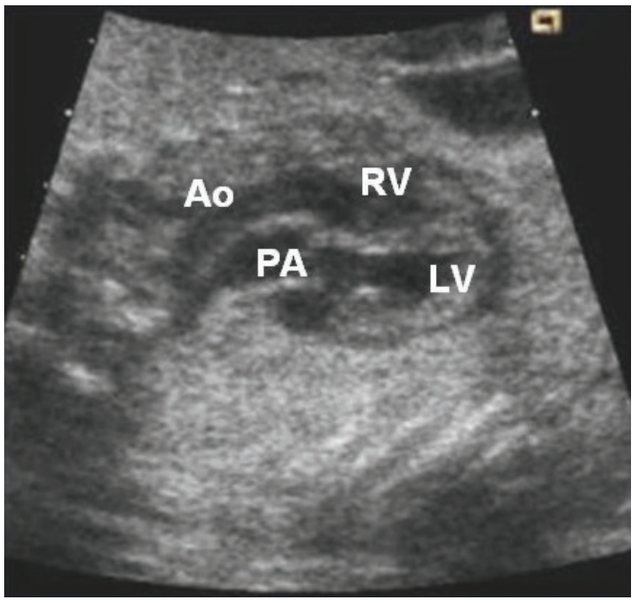


Figure 6. The four chamber view may be normal in simple Transposition of the Great Arteries (TGA), but the third and fourth transverse planes demonstrate a bifurcating artery (pulmonary artery, PA) arising from the left ventricle (LV) and an anterior aorta (Ao) arising from the right ventricle (RV).

Ongoing Support to Sustain Increased Detection Levels

We have found that an effective 'hands-on' training programme includes follow-up visits, to reinforce training, highlight areas of uncertainty and support sonographers as they gain experience and confidence. This is particularly important when there is a high turnover of staff and many work part-time.

In addition, it is important to establish an audit with feedback from local neonatologists and cardiologists, so that referred cases can be reviewed and missed cases can be identified so lessons can be learnt. Care should be taken to establish a culture of learning, rather than one of blame.

Fetal Therapy

Fetal therapy is well established for the treatment of arrhythmias and may be of benefit in fetuses with progressive valvar stenosis, e.g. to prevent progression of aortic stenosis to HLHS, and to minimize secondary myocardial damage in aortic and pulmonary atresia, thus permitting a biventricular repair after delivery. Intervention to open the inter-atrial septum to prevent hydrops and optimize the pulmonary circulation may be of use in simple transposition of the great arteries and HLHS. There have been encouraging reports and longer-term outcomes are awaited in these individuals.[8–10]

Summary

Prenatal detection of CHD is important because it widens parental choices, alerts the obstetrician to the possible existence of co-morbidities, such as chromosomal defects, and allows for a safer perinatal period for the baby with a duct-dependent lesion. With effective on-site training of obstetric sonographers, the majority of cardiac defects can be detected at the routine 20-week scan using five transverse views without increasing routine examination time.

References

1. Wren C, Richmond S, Donaldson L. Presentation of congenital heart disease in infancy: implications for routine examination. Arch Dis Child Fetal Neonatal Ed 1999;80:F49–F53.
2. Abu-Harb M, Wyllie J, Hey E, Richmond S, Wren C. Presentation of obstructive left heart malformations in infancy. Arch Dis Child Fetal Neonatal Ed 1994;71(3):F179–F183.
3. Bull C. Current and potential impact of fetal diagnosis on prevalence and spectrum of serious congenital heart disease at term in the UK. British Paediatric Cardiac Association. Lancet 1999;354:1242–1247.
4. Stumpflen I, Stumpflen A, Wimmer M, Bernaschek G. Effect of detailed fetal echocardiography as part of routine prenatal ultrasonographic screening on detection of congenital heart disease. Lancet 1996;348:854–857.
5. Yagel S, Cohen SM, Achiron R. Examination of the fetal heart by five short-axis views: a proposed screening method for comprehensive cardiac evaluation. Ultrasound Obstet Gynecol 2001;17(5):367–369.
6. Gardiner HM. Fetal Echocardiography: 20 years of progress. Heart 2001;86 Suppl 2:II12–II22.
7. ter Heide H, Thomson JD, Wharton GA, Gibbs JL. Poor sensitivity of routine fetal anomaly ultrasound screening for antenatal detection of atrioventricular septal defect. Heart 2004;90:916–917.
8. Tulzer G, Arzt W, Franklin RC, Loughna PV, Mair R, Gardiner HM. Fetal pulmonary valvuloplasty for critical pulmonary stenosis or atresia with intact septum. Lancet 2002;360:1567–1568.
9. Tworetzky W, Wilkins-Haug L, Jennings RW, *et al.* Balloon dilation of severe aortic stenosis in the fetus: potential for prevention of hypoplastic left heart syndrome: candidate selection, technique, and results of successful intervention. Circulation 2004;110:2125–2131.
10. Marshall AC, van der Velde ME, Tworetzky W, *et al.* Creation of an atrial septal defect *in utero* for fetuses with hypoplastic left heart syndrome and intact or highly restrictive atrial septum. Circulation 2004;11:253–258.

The CD to accompany this article is generously sponsored by the echo charity UK (the tiny tickers charity) and further details can be found at www.echocharity.org.uk or by email at echocharity@yahoo.com. Telephone enquiries should be made to Ian Averiss on 07714235079.