Successes and shortcomings of fetal echocardiography

Helena M Gardiner

While it is possible to perform balloon valvuloplasty of a critically stenosed pulmonary valve in the fetus safely, the majority of serious heart conditions remain undiagnosed until after birth. This article addresses the successes and shortcomings of fetal cardiology.

Fetal cardiology is an expanding field as cardiologists work in conjunction with fetal medicine units and obstetricians to improve detection of fetuses with cardiac defects before birth, the subsequent counselling of the family and the management of these cases.

Technically challenging fetal therapies such as ultrasound-guided pulmonary valvuloplasty have recently been shown to be possible (G Tulzer, unpublished data, 2000; HM Gardiner, unpublished data, 2001). Laser photocoagulation of placental anastomoses has an established role in the treatment of circulatory imbalance resulting from unbalanced placental transfusion in identical twins in the twin–twin transfusion syndrome (Hecher et al, 1999). The treatment of fetal tachycardia has long been successful in the majority of cases and the diagnosis of duct-dependent lesions before birth, particularly of transposition of the great arteries, has been shown to save lives (Bonnet et al, 1998).

However, while almost all women in the UK have a detailed scan during pregnancy (usually at about 20 weeks), and the detection of renal and skeletal problems is high, the diagnosis of cardiac defects was found to be only 23% overall in the UK (Bull, 1999). This figure encompasses a wide variation with some centres, mostly those in the South-East and North-East regions, having detection rates above 70%, while some regions approach zero.

This article examines the successes of fetal diagnosis and therapy and discusses potential solutions to the problems that impede continuing progress.

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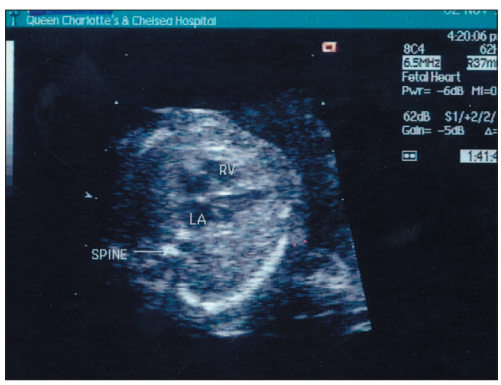
FETAL CARDIOLOGY: THE BENEFITS OF EARLY DETECTION

Early examination of the fetal heart was pioneered using M-mode echocardiography and then cross-sectional images (Allan et al, 1982). Technological advances have enabled the rapid advances in image quality such that it is now possible to examine a very small heart at 12–14 weeks using transabdominal or transvaginal ultrasound (*Figure 1*).

For the vast majority of women, the first comprehensive scan they receive is at 20 weeks' gestation when major problems such as absent valves or chambers, or large septal defects, can be detected. The advantages of early detection are not simply those associated with the possibility of termination of pregnancy for an affected fetus as emphasized in the past (Allan et al, 1991b), because many of the severe cardiac conditions could be legally terminated after 24 weeks gestation. Improvements in cardiac surgery have resulted in improved survival for most cardiac conditions, including the very severe ones. There is now evidence of improved survival and quality of life associated with antenatal diagnosis, par-

Figure 1. Four-chamber view of the heart of a 14-week-old fetus showing that four cardiac chambers are present and normally connected and that there is symmetry between right and left. LA= left atrium; RV = right ventricle.



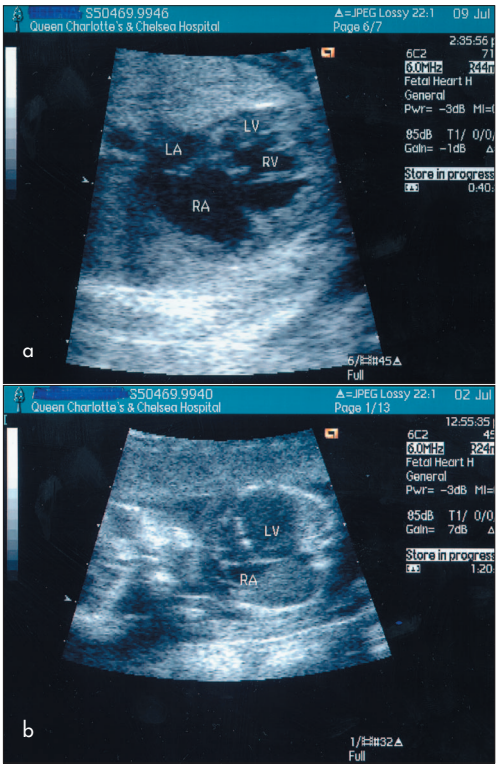


Figure 2. a. Four-chamber view of a fetus at 20 weeks showing a small left ventricle (LV) as a result of mitral and aortic atresia diagnostic of hypoplastic left heart syndrome. b. The left ventricle is not always small and may become dilated, thin walled and fibrous. RA = right atrium.

ticularly in the fetus with a circulation that depends on patency of the arterial duct (Bonnet et al, 1998; Tworetzky et al, 2001), but in unselected series this has yet to be demonstrated where there is either a hypoplastic left or right side of the heart (Brackley et al, 2000) (*Figures 2* and *3*).

While no formal studies have shown that parents benefit from knowledge of their baby's heart defect before birth, health professionals comment that they usually approach the perinatal period and cardiac intensive care better informed and more able to understand the complex issues they face.

The time available before delivery is valuable as it is important to realize that about 25% of fetuses with cardiac effects also have chromosomal or other structural abnormalities (Allan et al, 1994). Thus an antenatal diagnosis allows health professionals to consult each other (e.g. geneticists, paediatric surgeons, paediatric haematologists), to investigate the fetus further both by ultrasound and by offering invasive testing as appropriate, and to explore the possibility that the parents may have related medical or chromosomal problems. This period allows the team to counsel the parents as new information becomes available.

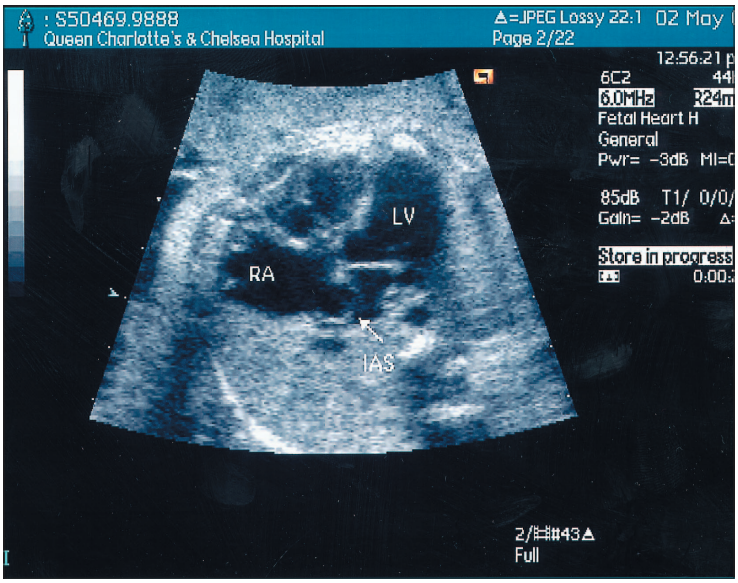


Figure 3. Four-chamber view of a heart with pulmonary atresia and intact ventricular septum. The right ventricle may be very small or potentially adequate for a future biventricular repair as in this case. There is marked bowing of the interatrial septum (IAS) because the right-sided pressures are supra-systemic with severe tricuspid regurgitation. LV = left ventricle; RA = right atrium.

Discussions before birth will also include the timing, mode and place of delivery, depending on the constellation of problems anticipated. Those with additional abnormalities such as gastroschisis or exomphalos (*Figure 4*) will need to be delivered by elective caesarean section in a hospital with paediatric surgery close at hand, while those with balanced defects that are not

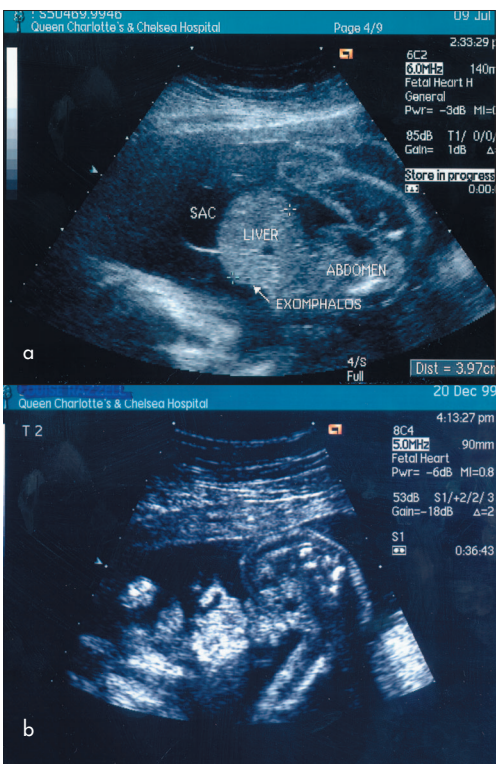


Figure 4. Extracardiac anomalies and chromosomal problems are common (a) with exomphalos, shown as a skin-covered herniation from the abdominal wall, but rare with (b) gastroschisis when loops of bowel float freely in the amniotic fluid.

duct dependent, such as atrioventricular septal defects, can be delivered in their local hospital and be seen in the outpatient department within the first days or weeks of life.

SHORTCOMINGS OF CURRENT SCREENING AND POSSIBLE SOLUTIONS

Despite the technical advances in imaging, these diagnostic benefits have not been passed on to the vast majority of women who still give birth to a baby with a major cardiac malformation unexpectedly.

What are the problems that have resulted in this poor overall detection rate? There are problems in some obstetric ultrasound units with quality of equipment, lack of continuing training and feedback of abnormal cases to the primary screeners. The heart is also perceived to be particularly difficult to scan.

One remedy for these problems is to establish an ongoing programme of training and support, funded centrally. Training programmes have been run that have shown a sustained improvement in diagnostic rate (Sharland and Allan, 1992; Hunter et al, 2000). The establishment of an ongoing programme would undoubtedly improve current detection rates to 70% or more.

The primary screeners, usually the obstetric sonographers, receive adequate theoretical instruction during their training in ultrasound examination of the heart but for many departments the goalposts have moved. It is now less acceptable for a hospital department to scan only the four-chamber view, which may be normal even in major heart defects such as tetralogy of Fallot and transposition of the great arteries (*Figures 5* and *6*).

Teasing out the precise diagnosis may be time consuming and is not as important as detecting that there is a problem and referring it to a fetal/perinatal cardiologist. However, it is more satisfying for the sonographer or obstetrician to be able to describe the conditions in more detail, and training programmes are available that correlate the morphology of the heart with ultrasound findings (shortcourses.nhli@ic.ac.uk).

THE SUCCESSES: DIAGNOSIS

Fetal medicine physicians and obstetricians have pioneered the study of Doppler of the fetal circulation. It is possible to monitor the wellbeing of

Figure 5. a. Four-chamber views are often normal but extension of the scan to include examination of the outflow tracts (b) reveals the diagnosis of tetralogy of Fallot with an overriding aorta (AO).

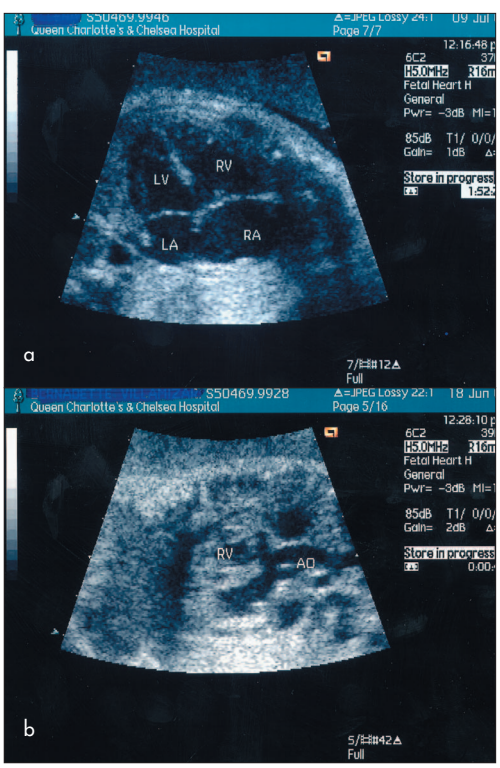
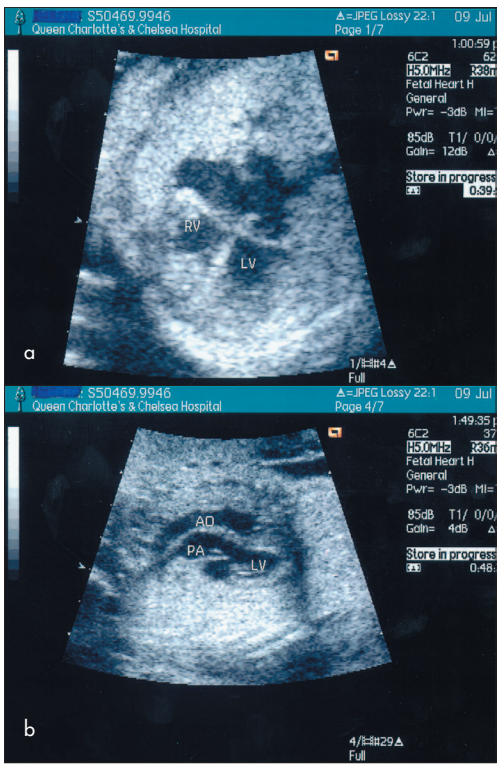


Figure 6. a. A normal four-chamber view may be seen in transposition of the great arteries only revealed (b) by demonstrating the aorta (AO) and pulmonary artery (PA) running in a parallel fashion from the heart.



the fetus using Doppler waveform patterns of the umbilical and middle cerebral arteries and the umbilical vein in conjunction with biophysical profiles and measures of fetal growth and amniotic fluid volume (*Figure 7*).

Early scanning in the first trimester has taught us much about the maturation of the fetal circulation. Many of the waveforms seen in the first trimester are characterized by marked reversal of flow in the venous system (coincident with atrial contraction) and reduced or absent flow in diastole in the arterial tree (*Figure 8a*). With improving ventricular compliance there is an increase in diastolic flow in the arteries (*Figure 8b*) and a reduction in the proportion of reversed flow in the venous system (*Figure 9*).

The heart is fully septated by 7 weeks and absent valves, arteries or chambers can be detected on early scans at 12–14 weeks. However, the more reliable views are at about 20 weeks, at the time of the anomaly scan. Diagnoses that tend to be progressive are more difficult to be sure of at an early scan. A basic tenet of cardiac development is that form follows flow and progressive stenosis of semilunar valves may lead to hypoplasia of the supporting ventricle by 20 weeks of gestation to produce hypoplastic left heart syndrome or pulmonary atresia with intact ventricular septum (*Figures 2* and *3*). If one scans only in the second trimester one usually only sees the end result of this process. Occasionally, however, this happens later in gestation with progressive reduction of forward flow through the mitral or tricuspid valve and the associated semilunar valve.

Figure 7. Umbilical cord Doppler shows abnormalities of flow with absent end diastolic flow in the umbilical artery (UA) and pulsations of the umbilical vein (UV).

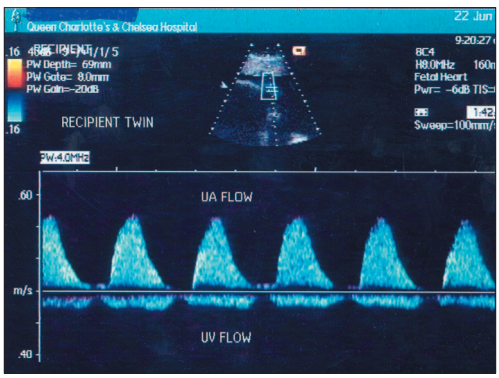


Figure 8. a. Early Doppler (12 weeks) in the fetal descending aorta (AoD) shows absent end-diastolic flow because of high distal impedance as a result of high placental resistance. As successful trophoblastic invasion of the placental spiral arteries occurs the impedance falls and diastolic flow is apparent in the arterial tree (b) as seen in this normal umbilical cord Doppler tracing.

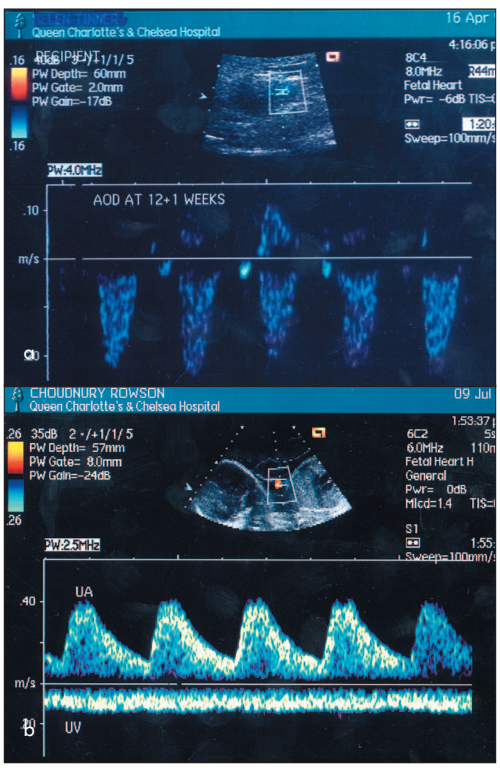
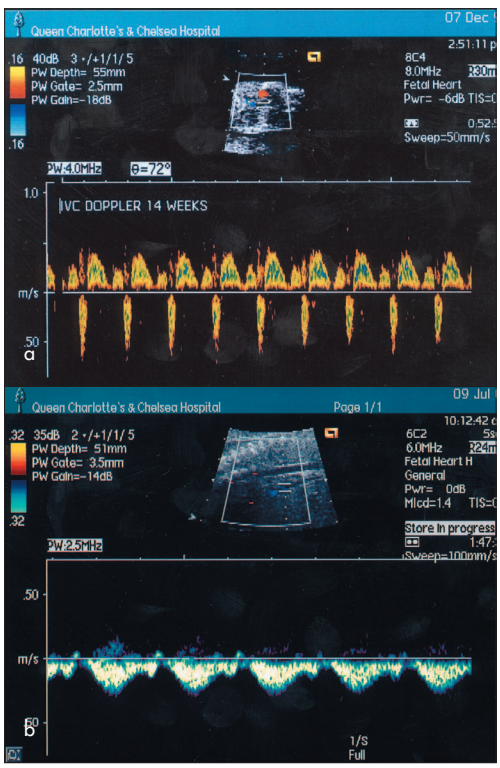


Figure 9. a. Similarly there is pronounced reversal of flow with atrial contraction in the venous system of the first trimester fetus that (b) resolves with maturity as ventricular filling improves.



THE SUCCESSES: FETAL THERAPY

The treatment of fetal tachycardia is generally successful with transplacental therapy. Maternal digoxin administration is generally safe and understood and slows the fetal ventricular rate thus improving output and preventing or reversing fetal hydrops. There are other therapeutic options such as flecainide, verapamil and sotalol and the choice of drug depends on practitioner familiarity and preference, rather than evidence-based decisions, as there are no large trials to guide management (Allan et al, 1991b). The treatment of fetal bradycardia is more difficult. Complete heart block may result in fetal demise if the rate is below 50. Sympathomimetic drugs have been used with some limited success (Groves et al, 1996), and steroids have been given anecdotally to women with anti-Ro and anti-La antibodies. Intrauterine pacing has been tried, but without much success in the human to date (Walsh et al, 1994).

Intrauterine balloon valvuloplasty of stenosed aortic valves has been attempted in the past with some technical successes but generally a poor outcome because of the severity of the diseased left ventricle (Kohl et al, 2000). However, there have now been two recent successful pulmonary valvuloplasties with improvement of right ventricular filling and resolution of circulatory dysfunction and impending hydrops allowing the fetuses to be delivered at or near term in good condition (G Tulzer, unpublished data, 2000; HM Gardiner, unpublished data, 2001) (*Figure 10*). As techniques improve these procedures may be more widely implemented and perhaps may change the natural history of ventricular hypoplasia and allow these infants to achieve a biventricular repair after birth.

KEY POINTS

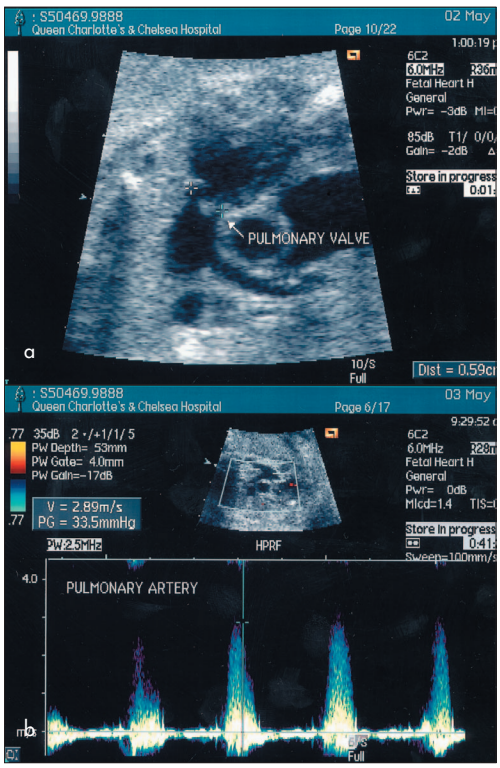
- Fetal cardiology has both diagnostic and therapeutic roles.
- Obstetric ultrasound screening for heart defects is not consistent across the UK and would benefit from additional training programmes.
- Congenital heart disease affects 6–8 per 1000 live births with at least 4 per 1000 being detectable before birth.
- Fetal vascular programme has been demonstrated in identical twins that have experienced differing haemodynamic stresses in utero.
- Fetal arrhythmias can be treated successful by giving drugs to the mother or directly to the fetus.
- Fetal balloon valvuloplasty has been performed safely with benefits to the fetus.

CONCLUSIONS

Fetal cardiology is now rightly regarded as a subspeciality in its own right. It is not the same as paediatric cardiology as the spectrum of disease differs from postnatal series and fetal physiology differs from that of the child. The potential for growth and remodelling of the heart and vessels of the fetus is extensive and may provide an opportunity for pathophysiological studies and perhaps successful early intervention. Fetal programming of various organ systems has been proposed as a mechanism for later adult disease, including the cardiovascular system (Barker, 1999), and there is now a vast body of supportive evidence.

Observations of fetal pathophysiology and its later consequences are available, but do not generally as yet have long-term follow-up. However, early reports seem to confirm areas of the Barker hypothesis; it has been shown that differing haemodynamic stresses experienced by the fetus in utero influence arterial stiffness in infancy (Cheung et al, 2000). These findings may be permanent and, if so, it might be predicted that timely fetal interventions performed in a safe and controlled manner may be of more benefit to the individual than anything that can be achieved after birth. HM

Figure 10. a. The thickened pulmonary valve in membranous pulmonary atresia and (b) Doppler profile of flow across it after fetal valvotomy.



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