Antenatal diagnosis of congenital heart disease

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Introduction
Cardiac abnormalities are the commonest form of congenital malformation, with moderate to severe forms affecting approximately 0.6% of live births.¹ Not all forms of congenital heart disease (CHD) will be evident at birth or in the early neonatal period, with up to 25% of babies with severe forms of CHD being discharged from hospital undiagnosed.² Diagnosis of CHD in fetal life can be made with a high level of diagnostic accuracy in tertiary centres dealing with high-risk pregnancies.

However, most of CHD will occur in low-risk groups and will be detected only by screening at the time of obstetric anomaly scans. There are some lesions – like secundum type atrial septal defects and persistent arterial duct (PDA) – that cannot be predicted in fetal life as they are normal findings in fetuses. In addition, milder forms of obstructive lesions of the aorta and pulmonary artery can develop in later life, with no signs of obstruction in fetal life.

Although antenatal screening of CHD has been common practice in the UK for over 20 years, there remains large regional variation, with many obstetric units still using the standard four-chamber view. The Royal College of Obstetrics and Gynaecology (RCOG) and National Institute of Clinical Excellence (NICE, 2008) have recommended fetal echocardiography screening to include four-chamber cardiac and outflow tract views.³ The Fetal Anomaly Screening Programme (FASP) has developed a protocol for fetal cardiac screening as part of the National Ultrasound Standards for England, for national implementation in 2010,⁴ to ensure uniformity, improve standards and detection rates of major CHD in England, with a target detection rate of 50%.

Fetal cardiac screening
The general approach to fetal cardiac screening is to look for five key sonographic views, all of which can be obtained by cranial or caudal angulation of the ultrasound probe from the four-chamber view.

1. Normal situs view (see figure 1) – The stomach is normally to the left of the fetus.
2. Normal four-chamber view (see figure 2) – The left and right side should be equal size. The atriovenous (AV) valves should have slight offsetting with the right AV valve being more apically placed.
3. Normal outflow tract views (see figures 3-4) – The aorta should be connected to the LV, and the pulmonary artery from the RV and “crossing over” of the aorta and pulmonary artery.
4. Normal three-vessel view (see figure 5) – This is the most cranial view. The ductal arch should be the largest vessel, followed by a medium sized aortic arch and a smaller superior vena cava (SVC).

The goal of such screening is to ensure that normal structures are visualised, and any deviation from normal patterns are recognised and therefore referred to specialist units.

In recent years, technical advances using three-dimensional echocardiography to include the fetal heart as a “volume” study – spatiotemporal image correlation (STIC),⁵ has been used at some units. However, retrospective analysis takes time and is dependent on image quality. STIC volumes can be useful for screening centres which are geographically remote from specialist centres, but its role in “routine screening” for CHD has not been established.

Examples of CHD that can be suspected on abnormal four-chamber views (figures 6-7) and normal four-chamber but abnormal outflow tract views (figures 8-9) are shown.

In balanced complete AVSD (see figure 6), there is equal size ventricular chambers, but the right and left atrioventricular valves (AVV) lie at the same level. There can be a hole seen in the atria (atrial septal defect) or ventricular septum (ventricular septal defect).

In HLHS (see figure 7), the left ventricle (LV) is much smaller and may appear to be quite thick-walled. Occasionally there can also be a bright lining on the inner wall of the LV, indicating fibrosis/scarring.

In tetralogy of Fallot (see figure 8), there is normal looking four-chamber view. A ventricular septal defect (VSD) with an over-riding great vessel is seen as the probe is angled more cranially from the four-chamber view.

In transposition of great arteries (TGA) (see figure 9), the great arteries do not cross-over and look parallel.

Antenatal cardiac diagnosis and management
At fetal cardiac units, precise diagnosis can be made, with prognosis and management plan formulated and discussed with the families of the affected fetus. Support and information is provided, to guide the family, in deciding what is “right” for them (option of interruption of pregnancy, compassionate care or active management). Planned perinatal care and delivery with early transfer to tertiary paediatric cardiology/cardiac surgical centres for confirmatory diagnosis and definitive management, can be organised. Antenatal diagnosis of life-threatening forms of CHD may help improve survival and reduce morbidity.

In addition, for families with a previous affected child, confirmation of normality and providing reassurance to anxious parents may be invaluable.

Antenatal detection rates
Impact of antenatal diagnosis on birth prevalence
In the United Kingdom, national data regarding impact of antenatal diagnosis on birth prevalence (1993 -1995) showed that 50% of pregnancies affected by fetal CHD ended in termination of pregnancy, although overall detection rate of CHD (requiring intervention or surgery in infancy) was only 24%.⁶ It also found that there is wide geographical variation in detection rates. Termination rate was affected by gestational age at diagnosis, with higher termination rate at earlier detection. Further European data emphasised the importance of non-cardiac malformations and karyotypic abnormalities in prenatal detection rates and parental decision making.⁷ For transposition of great arteries, single ventricle, hypoplastic left heart, the percentage of late termination was 12-23%.⁸

Examples of three serious and complex CHD are discussed – Hypoplastic Left Heart Syndrome, Atrioventricular Septal Defect and Transposition of great arteries.

Hypoplastic left heart syndrome (HLH S)
HLH S describes a range of congenital heart lesions that have a small left ventricle and left-sided heart structures (small mitral valve, small aortic valve, small aortic arch) which is unable to support the systemic circulation. There can be a spectrum of severity of the lesion, with severe obstructive left heart lesions developing early resulting in
a diminutive left ventricle that is non-functional and severely scarred (endocardial fibroelastosis, EFE), while other less severe valve stenosis or arch lesions may evolve during later stages of gestation with left ventricles, which may look relatively balanced at routine 20-week scans.

In most units, some 60% of fetuses are detected antenatally, with over 50% termination rate. However, despite this, it has not resulted in improved outcome postnatally, due to more severe lesions and other co-morbid factors that are detected antenatally. For families who decide on surgical management postnatally, the results of three-staged palliative surgery, culminating in Fontan procedure, have steadily improved over time, with five-year survival now approaching 70%. These figures, however, do not quote survival risk for those with very poor functional outcome or gross neurological deficits, or the uncertain future options if the systemic right ventricle fails.

Atrioventricular septal defect (AVSD)
AVSD is a lesion with a common atrioventricular junction (seen as loss of normal offset of left and right atrioventricular valves). There can be an associated hole in the top chamber of the heart (atrial septal defect) and hole in the bottom pumping chamber (ventricular septal defect). Recent UK published data indicate that less than 30% of liveborn infants with AVSD were detected by routine obstetric antenatal ultrasound. There is poorer antenatal diagnosis rate for liveborn infants with trisomy 21 or those with isolated AVSD. The spectrum of AVSD in fetal life is usually more severe with associated complex cardiac lesions and chromosomal abnormalities (58% risk of aneuploidy – mainly trisomy 21).

Transposition of great arteries (TGA)
TGA is a structural cardiac abnormality where the aorta is connected to the right ventricle and pulmonary artery is connected to the left ventricle. In isolated TGA, there are no other intracardiac defects and this circulation is incompatible with life when the ductus arteriosus (PDA) and the foramen ovale (PFO) closes.

Isolated TGA is detectable on antenatal ultrasound scanning, with as high as 72.5% detection, if the screening scans include the outflow tract views17 which show parallel great arteries or failure of the great arteries to cross over, in an otherwise normal four-chamber view. In a recently published data there was increase in detection rate of isolated TGA from 0-25%, from 1993 to 2006,18 due to inclusion of outflow tract views. In the antenatal diagnosis group, all survived corrective surgery.

Conclusion
It is important to realise that most of the CHD detected antenatally are found in patients with low-risk. Therefore, with improvement of routine obstetric screening to include outflow tract views in addition to the four-chamber view, it is hoped that more of such CHD can be picked up. Once such lesions are diagnosed, then appropriate counselling and management can be instituted, with careful perinatal management of such fetuses, to ensure that the baby is born in optimal condition so that the best clinical outcome can be achieved.

References
5. UK National Screening Committee – Fetal Anomaly Screening Programme (FASP). www.fetalanomaly.screening.nhs.uk.
FIGURE 1
Normal situs view.

FIGURE 2
Normal four-chamber view (RV=right ventricle; LV=left ventricle).

FIGURE 3
Left ventricular outflow tract (LVOT).

FIGURE 4
Right ventricular outflow tract (RVOT).

FIGURE 5
Normal three-vessel view (SVC= superior vena cava; Ao=aorta; DA=ductal arch).

FIGURE 6
AVSD (common valve with loss of normal offset of atrioventricular valve).

FIGURE 7
HLHS (small left ventricle).

FIGURE 8
Tetralogy of Fallot: Ventricular septal defect (VSD) with over-riding aorta.

FIGURE 9
Parallel great arteries typically seen in Transposition of great arteries (TGA).