Screening for congenital heart disease in 2011

By Helena Gardiner
Reader and Director of Perinatal Cardiology, Imperial College, London

Importance of CHD
Congenital heart disease (CHD) is both important and common and still accounts for 10% of infant deaths and nearly half of all deaths from malformations in developed countries. The majority of babies born with CHD have no prenatal identifiable risk factors – the average risk of a pregnancy being affected by CHD is about 1% overall with important CHD (defined as that requiring surgery or catheter intervention in the first year following delivery) occurring in 3.4-4 per 1000 women screened in the second trimester. The commonest time to detect cardiac defects is at the fetal anomaly screening scan at 20 weeks of gestation, although some advocate a first trimester anomaly screen. The UK Fetal Anomaly Screening Programme (FASP) has for many years set standards of detection for conditions that are less prevalent in the population than CHD and arguably are not associated with higher morbidity (figure 1). Certain pregnancies, for example those where either parent has CHD or pre-existing diabetes have a higher risk, usually 3-5%, but this does not imply they should be screened by fetal cardiologists outside the second trimester screening process, provided the local screeners are competent in imaging the five transverse views of the fetal heart.

Current detection of important CHD
The first comprehensive report on the antenatal detection rate of CHD in babies requiring surgery or interventional catheterisation for major heart defects in the first year following delivery in the UK was produced on behalf of the British Paediatric Cardiac Association (now the British Congenital Cardiac Association) in 1999. At the time of data collection (1993-5), screening protocols recommended the four-chamber view only, with very few obstetric departments looking routinely at the outflow tracts. The report described antenatal detection of major CHD ranged from as little as 0% up to 71% in areas where there had been active teaching programmes. Although the average detection rate of 23.5% is very low for a screening service, the report confirmed that teaching programmes could produce a sustained improvement in the antenatal detection of CHD. However, effective training programmes have not been established in the UK until this year, even though reports have highlighted the importance of including the outflow tracts and, more latterly, the three vessel and tracheal views to detect important duct dependent lesions of the aortic arch.

Reducing neonatal morbidity and mortality from duct dependent lesions
Babies are discharged home, often at 6 or 24 hours after delivery when the arterial duct is still patent and supplying adequate flow to the systemic or pulmonary circulation of a baby with important valvular or arch obstruction (figure 2). Neonatal screening of CHD is notoriously difficult in this period and studies confirm that at least half of all babies with major CHD are sent home without a diagnosis and in about 15% it is life-threatening. Substantial morbidity may result following neonatal collapse and the introduction of routine pulse oximetry screening and greater awareness that the arterial duct may take up to three weeks to close after delivery may help to remind health professionals in the community to check for cardiac disease in the sick newborn. However, as a funded and almost universal antenatal screening programme exists in the UK, it makes sense to optimise ascertainment of major CHD at this time.

Best practice in screening
Protocol
The heart lies horizontally in the fetal thorax and so five transverse planes through the fetal abdomen and thorax provide a series of views similar to those seen on magnetic resonance imaging, namely abdominal situs, the four-chamber view, the great artery crossovers and the three-vessel and tracheal view comprising the transverse aortic and ductal arches, superior caval vein and trachea. At each level the cardiac structures are assessed for symmetry, the characteristic features of left- or right-sided morphology, the cardiac connections and the presence of septal defects. Using a transabdominal approach at 18-20 weeks of gestation, these five transverse scanning planes are sufficient to suspect CHD and should be feasible in most pregnancies. There may be difficulties in multi-fetal pregnancies because of overcrowding, and the three-vessel and tracheal view, which is obtained high in the fetal chest, may not always be seen if the fetus is in a flexed position. Pulmonary veins can only be confidently examined by the concomitant use of colour Doppler flow which is not generally included in screening programmes. However, more obstetric units are routinely including colour in the four-chamber view where pulmonary veins can be identified entering the left atrium in the normal heart.

Image storage and review
Storage of appropriate digital clips is helpful for training and is a medico-legal necessity for the cardiologist working in the UK. It is desirable to meet regularly with other members of the screening and referral pathway (the referring units or fetal medicine department) to discuss the outcome of cases referred with a suspected cardiac anomaly and those that may have been missed at screening. For this to be a useful exercise, one must be able to review studies and identify whether the lesion was captured, but not recognised, at screening or whether inadequate views were obtained. A well organised review process involving members of the local screening pathway should improve ascertainment and provide “on the job” continuing education of staff. Storage of appropriate clips is also helpful for remote diagnosis, for example by a telelink to a local cardiac centre. This may be useful in triage of suspected cases of CHD from remote centres and may be the most cost-effective way of managing the increased caseload of women with a slightly increased risk, such as diabetic women, while the local screeners gain confidence in examining the heart locally. This will enable tertiary centres to eventually receive only cases of suspected abnormality for fetal cardiology review. Studies have been published evaluating the utility and cost of this practice.

Local and central audit
Local audit is essential to monitor the progress of a department. Knowing what has been missed is an important part of the training process. This may be difficult to achieve without additional resources to link the newborn babies with heart defects managed in the cardiac units to the findings at the anomaly scan, which is often performed in a hospital remote to this centre. National audit is vital to monitor training initiatives. The Central Cardiac Audit Database (CCAD) was established in 2000 following the Bristol inquiry into excess deaths occurring during congenital heart disease surgery; these data are verified and open to public scrutiny and all centres performing congenital heart surgery contribute. Latterly, this database has included a field indicating whether a baby attending for surgery or interventional catheterisation had an antenatal diagnosis. http://www.ccad.org.uk/002/congenital.nsf/vwContent/Antenatal%20Diagnosis

http://www.radmagazine.co.uk/37/408, 31-32

Image usage
http://www.radmagazine.co.uk/37/408, 31-32

http://www.radmagazine.co.uk/37/408, 31-32

http://www.radmagazine.co.uk/37/408, 31-32

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This information is now available across the UK for each PCT (figure 3) and although is not the same as the antenatal detection rate of CHD (as it does not take fetal loss into account, nor those with arrhythmias or mild heart disease) it does document the proportion of babies born with major heart disease detected antenatally, by PCT. Disappointingly, the proportion of those detected antenatally, between 2004-2008, appears similar in proportion and geographical distribution to the data collected in the UK between 1995-96. However, data available for 2009 show an encouraging rise in some areas, such as Wales and Northern Ireland, where an active sonographer training programme has been put in place. It is hoped that the current FASP initiative will result in a more widespread improvement in ascertainment.

Patient pathway following a cardiac diagnosis

The Tiny Tickers charity ran a workshop, convened by Royal College of Obetetrics and Gynaecology in 2008, during which a multidisciplinary panel devised an integrated care pathway for the pregnant woman following the suspicion of fetal CHD at screening. The panel produced guidelines and a pathway (Prenatal congenital heart disease: A new beginning) that may be downloaded from the TinyTickers.org website and adapted for local use. http://www.tinytickers.org/index.php?Itemid=70

There was multispeciality consensus on management of the pregnant woman with suspected fetal CHD and discussion highlighted several areas where improvement in current practice was desirable including:

i) Co-location of the screening and tertiary teams is very rare. The workshop felt it was best practice for the cardiologist to work within the fetal medicine unit. This joint approach provides the patient with a one-stop-shop approach, the opportunity for joint counselling and improved communication.

ii) Communication between the screening and management parts of the pathway was generally poor so that local teams had no idea of their true performance.

iii) Audit requires an integrated local approach (ideally including a congenital malformations registry) with easy access to a central national database – ideally CCAD. Any initiative, such as the FASP guidelines and training programme, should be supported by a mechanism so it is able to assess what it has achieved.

iv) Continuing professional education had suffered due to health care cutbacks and some of the unequal detection rates across the UK could be attributed to lack of access to on-going training once a sonographer is established in post. A positive approach by department superintendent and managers was also thought vital to successful screening.

Congenital heart champions

In response to this latter observation and the desire to audit the FASP initiative, Tiny Tickers has called for interested health professionals to become local “Congenital Heart Champions” ideally one in every obstetric ultrasound department. Details of this are on the website. The local Champion should ideally:

1. Show personal proficiency in detecting CHD
2. Champion best practice and continued improvement in screening for CHD
3. Identify issues that may prevent good detection rates
4. Co-ordinate simple, verified audit of referrals to – and feedback from – cardiac referral centres
5. Inspire change and improvement, with their support and enthusiasm.

Conclusions

Although it has been difficult to prove benefit to the fetus from an early cardiac diagnosis in the past, and paradoxically many series have reported worse outcomes, fetuses with isolated CHD have survival rates similar to those reported in individuals diagnosed with CHD in postnatal life. More latterly, studies of outcome in fetuses with duct-dependent lesions such as simple transposition of the great arteries or coarctation of the aorta have shown reduced morbidity if detected antenatally.11

There is the potential to give babies with heart defects a better start in life and a better future, but this will require considerable cross-speciality collaboration and a team approach that is appropriately funded to meet these new challenges.

Conflict of interest: Helena Gardiner is a co-founder and medical advisor to Tiny Tickers charity (UK registered charity 1078114).

References


FIGURE 1

Estimated live birth prevalence of malformations screened for in the Fetal Anomaly Screening Programme protocols. The graph shows estimated live birth prevalence per 1000 live births by maternal age. Congenital heart disease is substantially more prevalent than all other malformations, including Down syndrome.
FIGURE 2
Ultrasound images of isolated coarctation of the aorta where disproportion at four chamber view (with left sided structures smaller than right) may alert the screener but inclusion of the complete views of the heart will help to confirm the diagnosis. a) disproportion at the three vessel and tracheal view showing hypoplasia of the transverse aortic arch and reversal of flow at the isthmus b) colour flow of a sagittal view of the aortic arch revealing tortuosity, hypoplasia and a coarctation shelf at the isthmus and c) a postnatal CT angio confirming the diagnosis of coarctation of the aorta.

FIGURE 3
Percentage of babies with major congenital heart disease attending the UK congenital heart disease surgical units with an antenatal diagnosis, by PCT.