CONGENITAL HEART DISEASE

Improving the effectiveness of routine prenatal screening for major congenital heart defects

J S Carvalho, E Mavrides, E A Shinebourne, S Campbell, B Thilaganathan

Objective: To evaluate the effectiveness of adding outlet views to the four chamber view in routine prenatal ultrasound screening for major congenital heart defects (CHD) as performed by trained sonographers, and to compare the procedure with current practice.

Design and setting: Prospective observational study at a London teaching hospital.

Participants and methods: 9277 women booked at a single institution (80% had first trimester nuchal translucency measurement) due to have routine fetal cardiac screening using the four chamber and outlet tract views at >18 weeks of gestation.

Main outcome measure: Identification of major CHD in chromosomally normal and abnormal pregnancies antenatally or postnatally.

Results: There were 40 abnormalities (4.3/1000), of which 30 were chromosomally normal (3.3/1000). The overall antenatal detection rate was 75% (95% confidence interval (CI) 59% to 87%) and 70% (95% CI 51% to 85%) for euploid pregnancies. Abnormal cardiac views accounted for 70% of all prenatal diagnoses, 30% of which were made at ≤18 weeks. The sensitivity of cardiac views during the first scan at >18 weeks was 52%. Of all patients undergoing nuchal translucency screening, 34 had major CHD, nine with increased nuchal translucency (26.5%). Factors influencing the results of this screening programme were training and audit of operators, adequate equipment for antenatal examination, ease of access, and low threshold for referral to specialised fetal echocardiography.

Conclusion: Adding ventricular outlet views to the four chamber assessment of the heart at routine fetal anomaly scans at >18 weeks is the most effective technique to detect CHD prenatally. The success of such a programme depends on an infrastructure committed to continuous in house training of obstetric ultrasonographers coupled with feedback from specialised fetal cardiologists, as well as adequate resource allocation to obstetric hospitals involved with antenatal screening.

Congenital heart disease accounts for the majority of deaths from congenital defects in childhood, being six times more common than chromosomal abnormalities and four times more common than neural tube defects. Prenatal detection of specific cardiac anomalies such as complete transposition of the great arteries and hypoplastic left heart syndrome has been shown to improve neonatal morbidity and surgical outcome. The overall prevalence of congenital heart defects (CHD) is estimated at 8/1000 live births. Defects usually classified as major or critical are those that are lethal or require intervention in infancy or on long term follow up. The estimated prevalence of such major abnormalities is 4/1000 live births.

Historically, detailed fetal echocardiography was performed because of a positive family history for CHD, but cardiac scanning has gradually been incorporated into routine ultrasound screening programmes without strong evidence to support its implementation. Over the years, reported sensitivities for major CHD by examination of the four chamber view have ranged from 5–60%.

At the 18–23 week ultrasound scan, as part of the routine structural survey, the four chamber view of the fetal heart was examined. With the inception of fetal cardiology services within the unit, a comprehensive training programme overseen by the fetal cardiologist was instituted for all sonographers working in obstetric ultrasound.

The training was focused on two main principles. The first was that operators optimised the machine settings to obtain routine prenatal screening to obtain these views and showed an increase in detection rates. We have further developed such a programme with the aim of assessing the effectiveness of adding outlet views to the four chamber view in routine 18–23 week anomaly scans obtained by ultrasonographers.

PATIENTS AND METHODS

This was a prospective observational study of an unselected obstetric population in whom routine anomaly scans are performed at 18–23 weeks’ gestation. Between January 1997 and August 1999 all women booked for antenatal care at our institution and subsequently delivered within our unit were included in the study. High risk referrals from other hospitals were excluded to eliminate bias.

Two full time and four part time sonographers working only in obstetric ultrasound performed routine ultrasound scans. The obstetric ultrasound service was situated within a unit that provided fetal medicine and echocardiography services. High resolution ultrasound equipment with a cineloop facility (ATL 3000/5000, Letchworth, UK and Acuson XP10, Uxbridge, UK) was used. In the majority of patients, nuchal translucency thickness was measured in the first trimester for screening of aneuploidy, as previously described. At the 18–23 week ultrasound scan, as part of the routine structural survey, the four chamber view of the fetal heart was examined. With the inception of fetal cardiology services within the unit, a comprehensive training programme overseen by the fetal cardiologist was instituted for all sonographers working in obstetric ultrasound.

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satisfactory cardiac images. The second was the use of a checklist for detailed assessment of the four chamber view. In addition, all sonographers were taught how to assess the outflow tracts and were encouraged to extend the echocardiographic views to include the great arteries in all patients. The time allocated for the entire fetal anomaly scan was 20 minutes per patient, with most sonographers spending an average of 2–3 minutes (maximum 5–6 minutes) to assess the fetal heart. Whenever an abnormality was detected or suspected at this level, patients were referred for a detailed cardiac scan by the fetal cardiologist. The training was complemented by two policies within the department to ensure ease of access to fetal cardiology. Firstly, in addition to those thought to be possibly or frankly abnormal, all cases with unsatisfactory views of the fetal heart were referred to the fetal cardiologist. Whenever possible the sonographer would observe the detailed cardiac scan to compare the findings with his or her own scan. Additionally, the sonographers were provided with a detailed explanation of the abnormalities encountered and were subsequently given feedback regarding postnatal outcome or postmortem results in terminated pregnancies.

Referral for echocardiography

Prenatal
Any deviation from the normal appearance of the fetal heart or an unsatisfactory (inability to establish normal anatomy) cardiac view was considered an indication for more detailed fetal echocardiography at any stage of pregnancy. In addition, all pregnancies considered to be at high risk of fetal cardiac problems were referred for detailed fetal echocardiography at 20–23 weeks.

Postnatal
Echocardiograms were routinely recorded from neonates with signs of heart failure or cyanosis; when a heart murmur was associated with abnormal chest radiograph or ECG; or when a murmur persisted for more than six weeks in asymptomatic children. Additionally, all neonates in whom a cardiac anomaly was diagnosed in the antenatal period had postnatal echocardiography to confirm the diagnosis.

Classification of CHD
Cardiac defects were classified as major when they were potentially lethal or were severe enough to warrant termination of pregnancy, required surgery or interventional catheterisation in infancy, or were likely to need treatment on long term follow up. Patent ductus arteriosus, atrial septal defects (ostium secundum), small and restrictive ventricular septal defects, and valvar pulmonary stenosis with Doppler gradients ≤ 35 mm Hg were classified as minor for the purposes of this study.

Data collection
Patient demographic, ultrasound, and pregnancy outcome data were recorded prospectively on a computer database. Chromosomal abnormalities were identified by reviewing all abnormal fetal and infant karyotypes from the regional cytogenetic laboratory database. In pregnancies with fetal cardiac malformations that resulted in intrauterine death or where the parents opted for termination of pregnancy, detailed pathological examination was requested to confirm the antenatal diagnosis. All neonatal and infant echocardiograms performed during the study period and up until April 2000 were reviewed to identify cardiac defects not diagnosed in the prenatal period. All cases of major CHD diagnosed in our institution are referred to a single cardiac centre. Neonatal, obstetric, and cardiothoracic teams met weekly to exchange information regarding recently diagnosed cardiac anomalies. The cardiothoracic database of the tertiary cardiac centre was also reviewed to identify patients from our unit undergoing long term follow up, interventional cardiac catheterisation, or surgery.

RESULTS
During the study period 9277 women booked for antenatal care delivered within the unit. The mean maternal age of the study population was 27 years (range 15–44 years). First trimester nuchal translucency was measured in 7384 (79.6%) pregnancies.

Referrals
Referrals for echocardiography were made in 890 (9.6%) pregnancies and 517 children. Figure 1 shows the indications for fetal echocardiography by indication (percentage of referred cases; open bars) and subsequent antenatal detection of major congenital heart defects (percentage of detected cases; solid bars).
for fetal echocardiography. During the study period, the most common reason for referral was the presence of intracardiac echogenic foci (n = 235, 26.4%) and the least frequent reason was the finding of abnormal cardiac views (n = 53, 5.9%). The number of referrals for echocardiography fell from 26 per month (9/100 anomaly scans) at the beginning of the study to 23 per month (8/100 anomaly scans) at the end of the study.

Major CHD
Major defects of the heart and great arteries were identified in 40 pregnancies (4.3/1000 pregnancies) of which 30 were chromosomally normal (3.3/1000 pregnancies). Thirty of these cases (75%) were diagnosed antenatally (table 1 and 10 postnatally (table 2). CHD were diagnosed in 70% of all prenatal diagnoses (16/31) and 52% of all cases seen who had an anomaly scan after 18 weeks (16/31). Nine cases were diagnosed before the routine 18–23 week scan, seven of which were associated with increased nuchal translucency. Overall, the nuchal thickness was increased in 27% of cases of CHD (9/34) and accounted for 15% (4/26) of chromosomally normal pregnancies where the mother underwent a scan at 12–14 weeks.

During the study period, 44 chromosomally abnormal pregnancies were diagnosed, of which 10 were diagnosed as having major CHD. In seven the defect was minor or absent. Twenty seven pregnancies were terminated before 14 weeks, before a fetal anomaly scan or fetal echocardiography was performed. A chromosomal abnormality was diagnosed because of increased nuchal translucency measurement in 32, because of multiple markers on anomaly scan in nine, and postnatally in the remaining three. Table 2 shows the indications for echocardiography in all cases diagnosed postnatally.

**DISCUSSION**
The present study has shown that routine prenatal screening for major CHD can be highly effective in a low risk population. The 75% detection rate of the current study is the highest reported with routine ultrasound screening, where ascertainment of the prevalence of CHD was thorough, including complete data on postnatal follow up. Although the data in large
Prevalence of CHD and case ascertainment

The overall prevalence for major CHD in this study was 4.3/1000 pregnancies (3.3/1000 in chromosomally normal pregnancies). This figure is consistent with previous population reports on prevalence, indicating that ascertainment of fetal and infant CHD was rigorous in this study.

Table 2 Major cardiac defects: Postnatal diagnoses

<table>
<thead>
<tr>
<th>Case number</th>
<th>Karyotype</th>
<th>Indication for echocardiography</th>
<th>Cardiac defect</th>
<th>Outcome</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Normal</td>
<td>Persistent cardiac murmur at 6 week follow up</td>
<td>VSD</td>
<td>Live birth, surgery</td>
</tr>
<tr>
<td>2</td>
<td>Normal</td>
<td>Suspected VSD on antenatal scan</td>
<td>VSD</td>
<td>Live birth, surgery</td>
</tr>
<tr>
<td>3</td>
<td>Normal</td>
<td>Central cyanosis and cardiac murmur</td>
<td>Pulmonary stenosis</td>
<td>Live birth, balloon dilatation</td>
</tr>
<tr>
<td>4</td>
<td>Normal</td>
<td>Persistent cardiac murmur at 6 week follow up</td>
<td>Pulmonary stenosis</td>
<td>Live birth, balloon dilatation</td>
</tr>
<tr>
<td>5</td>
<td>Normal</td>
<td>Central cyanosis and cardiac murmur</td>
<td>Tetralogy of Fallot</td>
<td>Live birth, surgery</td>
</tr>
<tr>
<td>6</td>
<td>Normal</td>
<td>Tachypnoea and poor feeding</td>
<td>Coarctation of the aorta, VSD, subaortic stenosis</td>
<td>Live birth, surgery</td>
</tr>
<tr>
<td>7</td>
<td>Normal</td>
<td>Neonatal collapse</td>
<td>Coarctation of the aorta and bicuspid aortic valve</td>
<td>Live birth, surgery</td>
</tr>
<tr>
<td>8</td>
<td>Normal</td>
<td>Central cyanosis</td>
<td>Transposition of the great arteries</td>
<td>Live birth, surgery</td>
</tr>
<tr>
<td>9</td>
<td>Normal</td>
<td>Failure to thrive, cardiomegaly on chest radiograph</td>
<td>Hypertrophic cardiomyopathy (LVOTO)</td>
<td>Live birth, long term follow up</td>
</tr>
<tr>
<td>10</td>
<td>T21</td>
<td>Postnatal diagnosis of T21 and cardiac murmur</td>
<td>VSD</td>
<td>Live birth, surgery</td>
</tr>
</tbody>
</table>

LVOTO, left ventricular outflow tract obstruction.

Table 3 Number of cases of major CHD antenatally diagnosed at different gestational ages and the total antenatal detection rate

<table>
<thead>
<tr>
<th>Time of diagnosis (weeks)</th>
<th>Chromosomally normal (n=30)</th>
<th>Chromosomally abnormal (n=10)</th>
<th>Total pregnancies (n=40)</th>
</tr>
</thead>
<tbody>
<tr>
<td>&lt;18</td>
<td>6</td>
<td>3*</td>
<td>9</td>
</tr>
<tr>
<td>20–24</td>
<td>11</td>
<td>4</td>
<td>15</td>
</tr>
<tr>
<td>24–40</td>
<td>4</td>
<td>2</td>
<td>6</td>
</tr>
<tr>
<td>Total antenatal detection rate (%)</td>
<td>70</td>
<td>90</td>
<td>75</td>
</tr>
<tr>
<td>95% confidence interval</td>
<td>51% to 85%</td>
<td>56% to 100%</td>
<td>59% to 87%</td>
</tr>
</tbody>
</table>

*The majority of aneuploid pregnancies were terminated before full echocardiography.

Extended views and operator training

Only two previous studies have evaluated the influence of sonographer training within the context of a routine screening programme. Tegnander and colleagues specifically investigated the value of operator training in a single unit with good long term follow up from a comprehensive child health programme. They reported an increase in the sensitivity from 18% to 26% with operator training to obtain a four chamber view. More recently, Hunter and colleagues carried out a four year prospective multicentre study to evaluate the effect of operator training on sensitivity of CHD screening. The authors reported an improvement in the detection rate for major CHD from 17% to 36% after a two year training period to enable sonographers to visualise the four chamber and outflow tract views. These data suggest that operator training and routine visualisation of the outflow tracts alone are unlikely to explain the sensitivity of 75% for CHD in this study.

Indications for referral for detailed fetal echocardiography

Approximately 20% of referrals were made because of an abnormal or unsatisfactory cardiac view at the routine fetal anomaly scan. Nonetheless, this resulted in the detection of 95% of major CHD. Although maternal diabetes and a family history of CHD also accounted for 20% of referrals, only one case (3.3%) of major CHD was detected from these scans. The most common reason for referral (26%) was the detection of isolated intracardiac echogenic foci, as dictated by the established protocol within the unit at the time of the study. The finding that the prevalence of major CHD in these fetuses appears to be no higher than in the rest of the population has now led to cessation of this practice. As fetuses with isolated intracardiac echogenic foci are no longer referred, approximately 5% of pregnancies now receive a detailed cardiac scan. It is important to note that increased nuchal translucency remains an important indication for fetal echocardiography, with 31% of major CHD diagnosed in this group. Although the sensitivity of this marker is lower than originally predicted, increased nuchal translucency above the 99th centile should mandate referral for detailed fetal echocardiography.
Examination time and access to echocardiography

The reasons for the relatively high detection rate in this study deserve further analysis. Constraints placed on health service resources in the UK dictate that most routine fetal ultrasound examinations be performed in a limited amount of time and sometimes with inadequate equipment. In this study, sonographers were scheduled 20 minutes per fetal anomaly scan and they used ultrasound machines of a good standard, optimised for fetal cardiac examination.

The ease of access and the high frequency of referral in this study reflect the availability of fetal echocardiography within the same unit. The low threshold for fetal echocardiography and subsequent direct feedback enables sonographers gradually to build up their skills and confidence in detecting cardiac abnormalities. The sensitivity for CHD may have been relatively lower in the study carried out by Hunter and colleagues because it was based on data from 16 centres covering a large geographic area. The latter features mitigate the obvious value of fetal cardiology services.

Conclusions

Our data indicate that routine antenatal assessment of the four chambers and great vessels between 18 and 23 weeks is effective in the prenatal detection of major CHD. The sonographer performing the routine anomaly scan remains the major contributor to prenatal detection of major CHD. Proper equipment to allow optimisation of cardiac views, adequate examination time, and ease of access to tertiary level fetal cardiology services obviously influence the extent to which such a service is successful. While the need for improved antenatal detection of major CHD is known, the adoption of a screening programme similar to ours has significant resource implications for health care providers. Even a slight increase in scanning time may be more difficult to implement than training experienced sonographers to assess the ventricular outflow tracts and the four chamber view competently. Importantly, the success of this screening programme cannot be divorced from the ease of access and relatively high referral for detailed fetal echocardiography. The limited availability of such tertiary fetal cardiology specialists and services nationally is also likely to affect the success of other antenatal CHD screening programmes.

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