

Fetal cardiac abnormalities identified prior to 14 weeks' gestation

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ABSTRACT

Objective An increasing number of patients are presenting at early gestational age as being at high risk for congenital heart disease, as a result of ultrasound screening by nuchal translucency. The feasibility and accuracy of fetal echocardiography was assessed in a series of pregnancies studied before 14 weeks' gestation.

Methods Echocardiography was attempted in 478 fetuses of crown–rump length 40.0–85.0 mm (median, 60.3 mm) with increased nuchal translucency, suspected abnormalities on routine scan or a family history of heart defect. The findings were related to results of autopsy, karyotyping, later scans and postnatal follow-up.

Results Satisfactory images were obtained transabdominally in 402/478 (84.1%) and transvaginally in a further 13 patients. Cardiac defects were confidently identified in 60 fetuses and abnormalities of uncertain significance (isolated ventricular or great artery disproportion, or tricuspid regurgitation) were observed in a further 49. Defects were suspected in an additional 20 fetuses, and 286 were passed as normal. The karyotype was subsequently demonstrated to be abnormal in 70/286 (24.5%) fetuses with normal echocardiograms, and in 94/129 (72.9%) with abnormal or suspicious cardiac findings. Validation of the scan findings was possible in 241 fetuses. Normal heart structure was confirmed in 204 fetuses, and previously unsuspected cardiac abnormalities revealed in nine. Heart defects were verified in 28 fetuses, but five of these had important additional findings. There were false positive findings in three fetuses.

Conclusions Fetal echocardiography is feasible prior to 14 weeks' gestation. Cardiac defects, when present, may be identified or suspected in the majority of cases. In the risk group studied, heart defects were frequently a manifestation of chromosomal abnormality.

INTRODUCTION

Screening for chromosomal abnormalities by nuchal translucency thickness (NT)¹ also identifies fetuses at increased risk of other major anomalies, including cardiac defects^{2–4}, and encourages the early diagnosis of major anomalies such as anencephaly, holoprosencephaly, exomphalos and megacystis⁵, even in the presence of normal NT. Attendance for the scan at 10–14 weeks' gestation is also an opportunity to ascertain any family history of genetic syndromes or anomalies that warrant more detailed ultrasound assessment during the pregnancy. Satisfactory preliminary examination of the fetal heart is feasible considerably earlier than an examination at 18–20 weeks that was usual during the 1980s and early 1990s. Some institutions have predominantly used a transvaginal approach^{6–9} and others a transabdominal approach^{10–12} with similar success. It has been our own recent practice to offer specialist fetal echocardiography to patients at high risk of fetal cardiac defects at the time of their 10–14-week scan, as well as subsequently, and we report our experience here.

METHODS

Women attending a tertiary referral center for fetal medicine and fetal cardiology between June 1998 and June 2001 with a fetal crown–rump length (CRL) in the range 38–84 mm (10–13⁺6 weeks' gestation) were included in the study. They were attending either for a routine 10–14-week scan or for reassessment following suspicious findings in referring centers. Cardiac examination was carried out in high-risk fetuses by a fetal cardiologist (I.C.H.) and/or gynecologist with particular experience in fetal echocardiography (N.Z.). Risk factors for a cardiac defect were increased NT (NT ≥ 4 mm), a first-degree relative with a significant congenital heart defect and suspicion of a cardiac or extracardiac abnormality on the 10–14-week scan. Occasionally a full cardiac examination was performed at the parents' request

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or when abnormal ductus venosus flow was demonstrated in addition to an increased NT of < 4 mm. Fetal echocardiography was generally performed prior to chorionic villus sampling, where that was indicated, and karyotype results were not available in any case at the time of the cardiac study. Parents were counseled in advance about the potential limitations of the scan and the possibility of equivocal findings or technical failure.

Echocardiography was performed transabdominally using an Acuson Aspen ultrasound machine (Acuson Inc., Mountain View, CA, USA) with a 7-MHz convex transducer (C7). The four-chamber view and outflow tracts were identified on cross-sectional imaging with color flow mapping used as an adjunct to confirm identification. Pulsed wave Doppler traces of both atrioventricular and arterial valves were obtained where possible. Images were stored digitally and on videotape. In all cases, care was taken to minimize ultrasound exposure of the fetus, especially in Doppler modes. In some cases, where transabdominal echocardiography was a technical failure or provided incomplete information, transvaginal scanning was offered in addition.

Abnormal or suspicious findings were recorded and discussed with the parents immediately. During the early part of the study, the results of the cardiac scan rarely determined critical aspects of clinical management. Subsequently, as confidence in the reliability of findings grew, greater weight was placed on the results, including determining decisions to proceed to invasive testing or termination of the pregnancy.

The utility of the early cardiac scan was assessed according to the ability to visualize the major cardiac structures during the scan and by comparison of scan results with outcome data. For the purposes of the analysis, a study in which the four-chamber view and great artery size and relationships could be assessed was considered satisfactory. The scan findings were classified as normal, demonstrating definite cardiac abnormality, being suspicious of abnormality or as demonstrating abnormal features of uncertain significance. The latter category included isolated atrioventricular valve regurgitation and ventricular and great artery disproportion, which may be manifestations of an underlying defect, but are also compatible with a normal outcome.

The outcome data included autopsy results following termination or miscarriage, fetal karyotype results, findings on follow-up fetal echocardiography and reports of postnatal outcome. Autopsy was the preferred means of verification in non-continuing pregnancies, but was available in only a minority of cases. Termination of pregnancy was often performed at an early stage following rapid diagnosis of the common aneuploidies by a polymerase chain reaction method¹³, usually by destructive surgical methods and away from our own unit. Fetal heart tissue was generally retrieved by personal attendance at the termination of a fetal cardiac morphologist (A.C.C.) and painstaking examination of the entire products immediately following suction termination.

RESULTS

Fetal echocardiography was performed in 478 fetuses of CRL 40.0–84.0 mm (median, 60.3 mm, equivalent to 12 weeks and

2 days). Indications were increased NT or abnormal ductus venosus flow in 399, family history in 46, extracardiac abnormality in 25 and suspected structural cardiac abnormality in eight.

Satisfactory images were obtained in 415/478 fetuses overall, including 13 of 18 that had transvaginal scanning because transabdominal scanning failed. Typically, echocardiography took 15–20 min, although longer scanning was sometimes necessary to confirm and detail abnormal findings.

Definite cardiac abnormalities were identified in 60 fetuses, and suspected in a further 20. An additional 49 fetuses had abnormal cardiac findings of uncertain significance. The latter group consisted of 21 fetuses with apparently isolated tricuspid valve regurgitation and 28 with varying degrees of isolated ventricular and great artery disproportion. Pericardial effusion was a frequent finding in fetuses with increased NT and was not evaluated in this study. The fetal heart was passed as normal in 286 fetuses.

Outcome

For the 415 fetuses with technically adequate scans, pregnancy ended in termination in 196 fetuses and spontaneous intrauterine death (IUD) in 15. There were 198 continuing or completed pregnancies. No early follow-up information could be obtained in six pregnancies, five with normal scans and one with a diagnosis of hypoplastic left heart where the mother left the country.

Types of defect, verification and outcome

Verification following normal early fetal echocardiography is summarized in Figure 1. For ease of description, definite abnormalities were grouped in mutually exclusive categories as atrioventricular septal defect (AVSD), ventricular septal defect (VSD), hypoplastic left heart syndrome and other abnormalities (Figures 2–4 and Table 1). Apparently isolated tricuspid valve regurgitation and disproportion are presented separately in Figures 5 and 6.

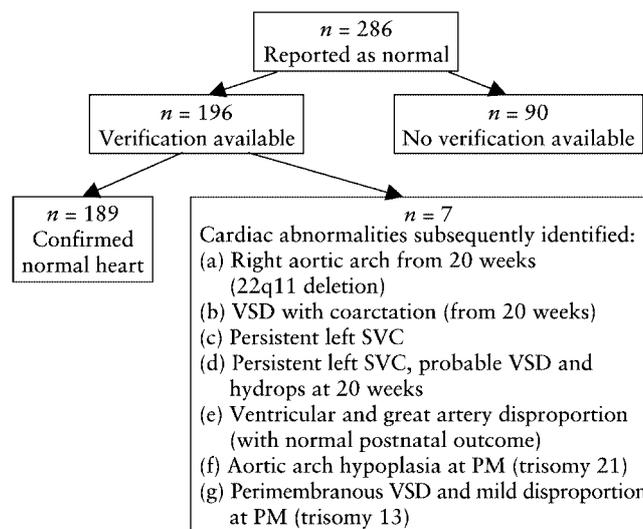


Figure 1 Flow chart summarizing the findings in 286 fetuses passed as normal on early fetal echocardiography. VSD, Ventricular septal defect; SVC, superior vena cava; PM, postmortem.

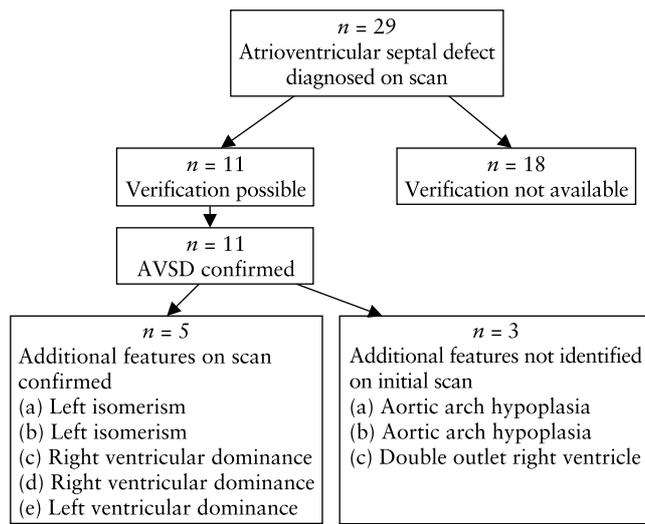


Figure 2 Flow chart summarizing the findings in 29 fetuses in which a definite diagnosis of atrioventricular septal defect was made on early fetal echocardiography. AVSD, Atrioventricular septal defect.

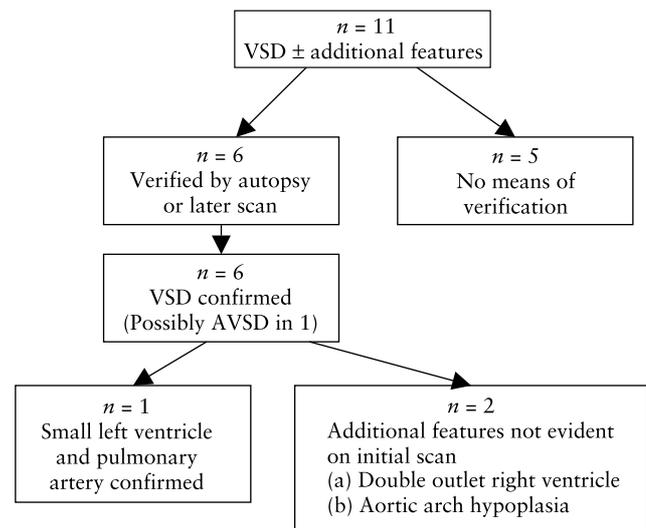


Figure 4 Flow chart summarizing the findings in 11 fetuses in which ventricular septal defect was a principal finding at early fetal echocardiography. VSD, ventricular septal defect; AVSD, atrioventricular septal defect.

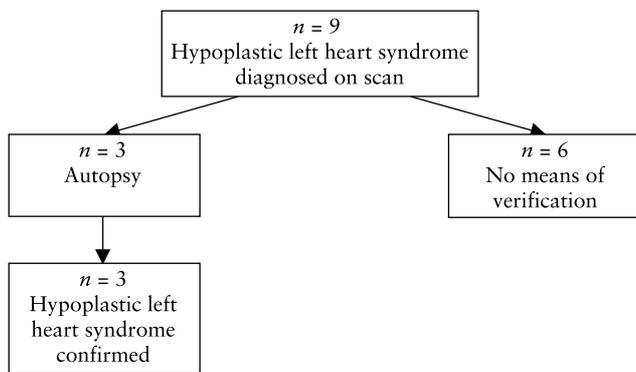


Figure 3 Flow chart summarizing the findings in nine fetuses in which a diagnosis of hypoplastic left heart syndrome was made on early fetal echocardiography.

Autopsy confirmation

Autopsy of the fetal heart was performed following termination of pregnancy or spontaneous IUD in 32 of the fetuses scanned, 28 of which had had technically satisfactory scans. In these 28, a cardiac abnormality was suspected on

echocardiography in 24 and the scan was considered normal in four. Autopsy ascertained normal heart structure in two fetuses with normal early echocardiography and, in the other two, demonstrated VSD and arch hypoplasia, and VSD alone, respectively. Cardiac abnormalities were confirmed in all 24 with abnormal or suspicious echocardiographic findings, but identified additional aortic arch hypoplasia in four of them. Autopsy demonstrated abnormalities in all four cases in which the initial echocardiography had been deemed a technical failure.

Confirmation in pregnancies without autopsy

In the absence of autopsy, verification of the early echocardiographic findings was dependent on echocardiography later in the pregnancy or on postnatal reports, one or both of which were available in 213/415 fetuses with technically satisfactory initial cardiac scans. There remained 174 fetuses with no means of verifying the cardiac scan diagnosis, 134 of which had chromosomal abnormalities. Of the 174, 85 fetuses had abnormal cardiac scan findings. There was a chromosomal abnormality in 69 of these 85 and the type of

Table 1 Outcome and features of 11 fetuses with abnormalities not classified in the flow charts

Scan diagnosis	Karyotype	Confirmation	Comments
Ebstein's anomaly	Unknown	Autopsy	As described (Figure 9)
Ebstein's anomaly/tricuspid dysplasia	Normal	Autopsy	Ebstein's anomaly at autopsy
Tricuspid valve dysplasia	Triploidy	None	Termination of pregnancy
Tricuspid atresia, concordant great arteries	Normal	Autopsy	As described (Figure 10)
Ectopia cordis	Normal	None	Termination of pregnancy
Common arterial trunk/pulmonary atresia VSD	22q11 del	Autopsy	Common arterial trunk. Truncal valve regurgitation on scan
Single ventricle, undetermined morphology	Unknown	None	Intrauterine death, twin pregnancy
Dilated poorly functioning RV	Unknown	None	Intrauterine death
Pulmonary atresia intact septum with dilated LV	Triploidy	Autopsy	Only part specimen
'Cardiomyopathy'	Normal	Later scan	Intrauterine deaths in both of monozygotic twins
Left isomerism	Normal	Postnatal scan	Azygous continuation, Right-sided stomach, normal intracardiac anatomy

VSD, ventricular septal defect; RV, right ventricle; LV, left ventricle.

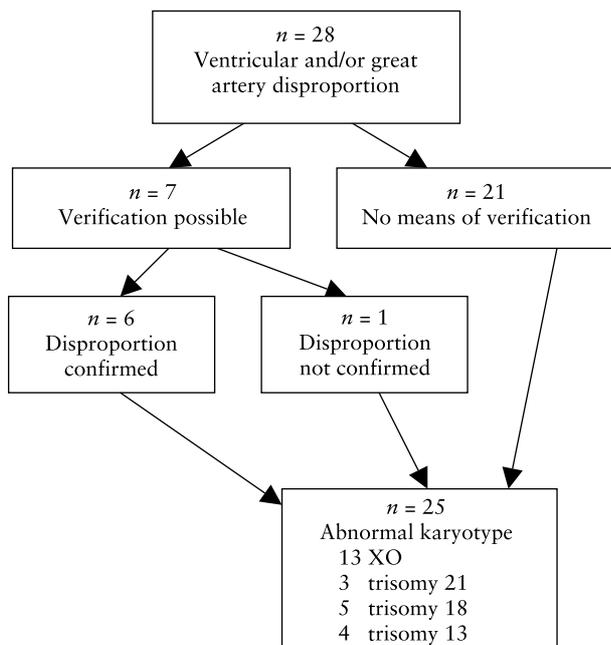


Figure 5 Flow chart summarizing the findings in 28 fetuses with ventricular and/or great artery disproportion at early fetal echocardiography.

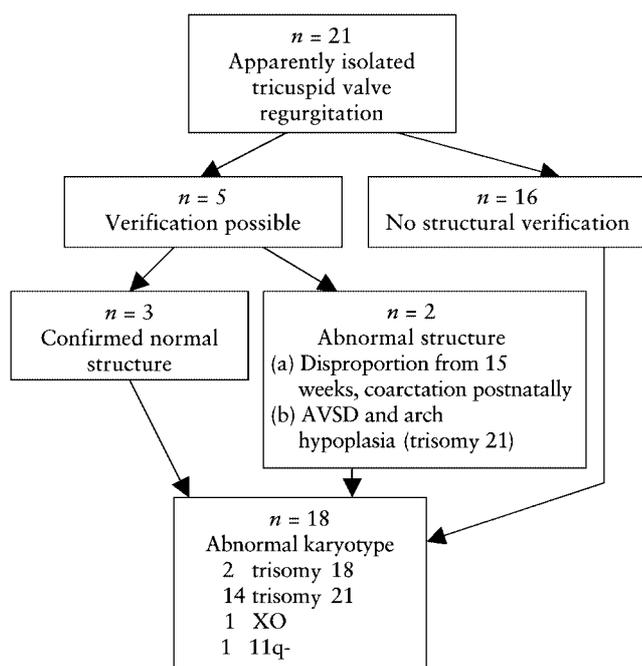


Figure 6 Flow chart summarizing the findings in 21 fetuses with tricuspid regurgitation, but no structural abnormality evident at early fetal echocardiography. AVSD, Atrioventricular septal defect.

cardiac abnormality was consistent with the type of chromosomal abnormality demonstrated in all cases.

Diagnostic accuracy

Normal initial echocardiogram

For the 196 fetuses with both a normal initial echocardiogram and a means of verification, a normal heart was confirmed in

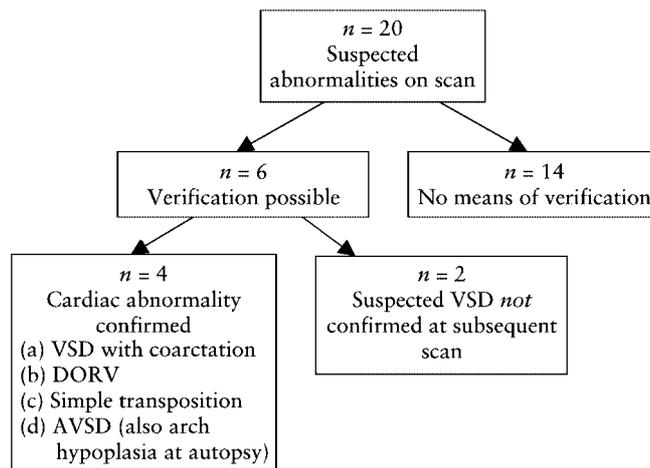


Figure 7 Flow chart summarizing the findings in 20 fetuses in which cardiac defects were suspected, rather than definitely diagnosed on early fetal echocardiography. VSD, ventricular septal defect; DORV, double outlet right ventricle; AVSD, atrioventricular septal defect.

189 (96%) (Figure 1). The seven heart defects overlooked (Figure 1) were types generally challenging for prenatal diagnosis.

Definite cardiac abnormality

Verification was possible in 34 fetuses with definite structural cardiac abnormalities (including disproportion) on initial fetal echocardiography. The principal abnormality was confirmed in 33 (97%) of these but significant other features were overlooked in six (18%) of them (Figures 2–5). The only false positive error concerned a fetus with Turner’s syndrome and apparent ventricular disproportion, not evident at subsequent scans prior to IUD (Figure 5).

Isolated tricuspid regurgitation

Verification was possible in only 5/21 hearts with normal structure but tricuspid regurgitation (Figure 6). Three were confirmed normal but, in one, disproportion became evident and coarctation was demonstrated after birth, while another had AVSD demonstrated at autopsy.

Suspected cardiac abnormality

Verification was possible in only six fetuses where findings were considered suspicious of, rather than diagnostic of, a cardiac abnormality (Figure 7). The suspected abnormality was confirmed in four of these (66%) but two suspected VSDs were not evident at subsequent scans.

Association of abnormal findings with referral indication

No cardiac abnormality was identified amongst 46 fetuses where the referral indication was a family history of congenital heart disease. Of the 33 fetuses undergoing echocardiography because of a suspicion of cardiac or extracardiac abnormality, 17 (52%) had abnormal hearts. Included in these are eight fetuses referred because of suspected cardiac

Table 2 Findings on fetal echocardiography in relation to the karyotype

Findings on fetal echocardiography	n	Abnormal karyotype					Others
		All (%)*	Trisomy 21	Trisomy 18	Trisomy 13	XO	
Confident diagnosis							
Normal heart	286	70/222 (32)	47	9	4	3	7
AVSD	29	18/26 (69)	14	1	2	—	1
HLH	9	5/7 (71)	—	1	—	4	—
VSD	11	8/10 (80)	2	5	1	—	—
Disproportion	28	25/27 (92)	3	5	4	13	—
Isolated TR	21	18/19 (95)	14	2	—	1	1
Others	11	3/7 (43)	—	—	—	—	3
Suspected diagnosis							
AVSD	8	8/8 (100)	4	2	1	—	1
VSD	7	5/7 (71)	1	3	—	—	1
Others	5	4/5 (80)	—	2	—	1	1
Technical failure	63	29/57 (51)	7	12	2	3	5

AVSD, Atrioventricular septal defect; HLH, hypoplastic left heart syndrome; VSD, ventricular septal defect; isolated TR, tricuspid valve regurgitation with apparently normal heart structure. *The denominator here excludes fetuses with unknown chromosome status.

abnormality, all of which were confirmed as abnormal. Cardiac defects were suspected or identified in 110 of 393 (28%) fetuses referred for increased NT, 106 of them in fetuses with NT \geq 4 mm. Of the 110, 14 (3.5% of 393) occurred with a normal karyotype and 10 with unknown karyotype. The incidence of heart defects suspected or identified on early scan in those seen because of increased NT with a subsequent normal fetal karyotype was 7.8% (14/179).

Association of abnormal findings with chromosomal abnormalities

Karyotyping was performed in 395 fetuses overall, including 17 in which the primary indication was findings on fetal echocardiography (11 with isolated cardiac findings and six cases with multiple malformations). The frequency of karyotype abnormalities in association with echocardiographic findings is summarized in Table 2. Fetuses in which the only abnormal cardiac findings were ventricular and/or great artery disproportion and those with apparently isolated tricuspid valve regurgitation had unexpectedly high incidences of chromosomal abnormality of 92% and 95%, respectively.

Apparently isolated cardiac abnormalities

Eighteen pregnancies with normal or unknown karyotype were terminated primarily because of a cardiac defect diagnosed on early cardiac scan. Seven of these fetuses had autopsy and an additional four had a second fetal echocardiogram at 13–16 weeks' gestation prior to the termination, but in seven there was no means of verifying the early cardiac scan diagnosis. Heart defects in these 18 fetuses included complex AVSD (six cases, one with complete heart block), isomerism with related complex intracardiac anatomy (four cases, two with complete heart block), hypoplastic left heart syndrome (three cases), Ebstein's anomaly (two cases), double outlet right ventricle (one case), tricuspid atresia (one case) and common arterial trunk (one case). Examples of isolated cardiac defects are shown in Figures 8–10.

Technical limitations and difficulties

An inability to obtain images of diagnostic quality transabdominally was attributed to maternal obesity, a persistently unfavorable fetal position or a retroverted uterus. Body mass index was significantly greater for women in whom transabdominal fetal echocardiography was a technical failure compared with those in whom it was successful (26.0 vs. 23.6 kg/m²; $P < 0.0001$, Student's *t*-test). Fetal size also influenced success of transabdominal echocardiography. The mean CRL was 61.4 mm in successful scans and 56.3 mm in unsuccessful scans ($P < 0.0001$, Student's *t*-test). Correspondingly, 25/251 (10.0%) of scans at CRL \geq 60 mm were technical failures compared with 51/224 (22.8%) at CRL $<$ 60 mm.

DISCUSSION

This study shows that by targeting particular high-risk groups for early specialist echocardiography, such as those with high levels of increased NT or those with appearances suspicious for abnormality, a high yield of cardiac defects can be identified. Within this high-risk group, early fetal echocardiography was effective in identifying major abnormalities. With a cautious approach to interpretation, a serious false positive diagnosis appears to have been avoided. We acknowledge, however, that autopsy in every terminated pregnancy would have been necessary to be certain of this and that incomplete verification is the main limitation of the study. It is evident from the autopsy results available that important associated features such as aortic arch hypoplasia could not always be detected on early fetal echocardiography. In the context of some types of heart defect (e.g. AVSD), such findings may markedly alter the outlook and the inability to exclude them would need to be taken into account in any prognosis given. In contrast, in some conditions (e.g. the hypoplastic left heart syndrome, or left isomerism with AVSD and complete heart block), unrecognized additional features would have less impact on the prognosis.

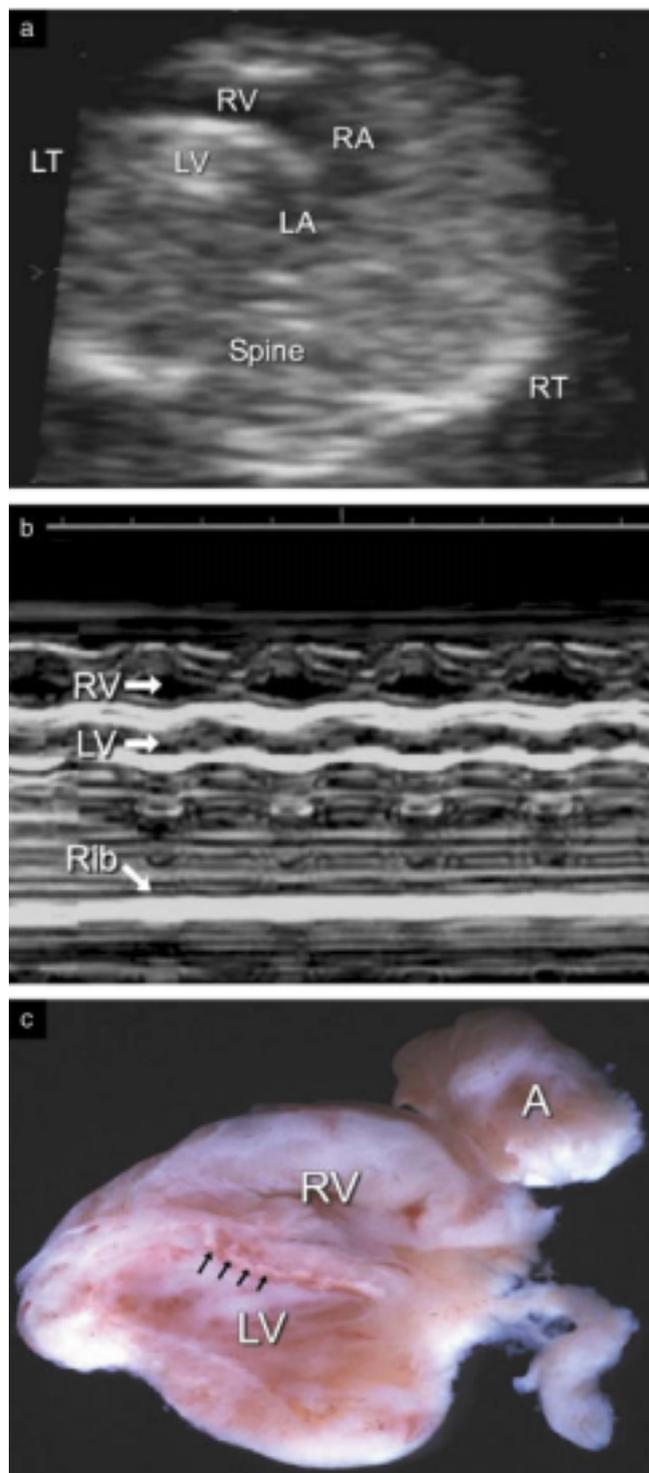


Figure 8 Aortic atresia diagnosed in a fetus with crown–rump length 72.4 mm (13+1 weeks). (a) Four-chamber view on fetal echocardiography. (b) M-mode trace through both ventricles. Brightness within the left ventricular myocardium and complete lack of contraction of the left ventricle were striking features. (c) Pathological specimen demonstrating gross endocardial fibroelastosis of the left ventricle and macroscopic calcification within the myocardium (arrows). LT, left; RV, right ventricle; LV, left ventricle; RA, right atrium; LA, left atrium; RT, right; A, atrium.

Care should be taken in extrapolating the results of this study to a population where the initial index of suspicion is not so high. Findings such as ventricular disproportion and isolated tricuspid regurgitation are recognized to be compatible with

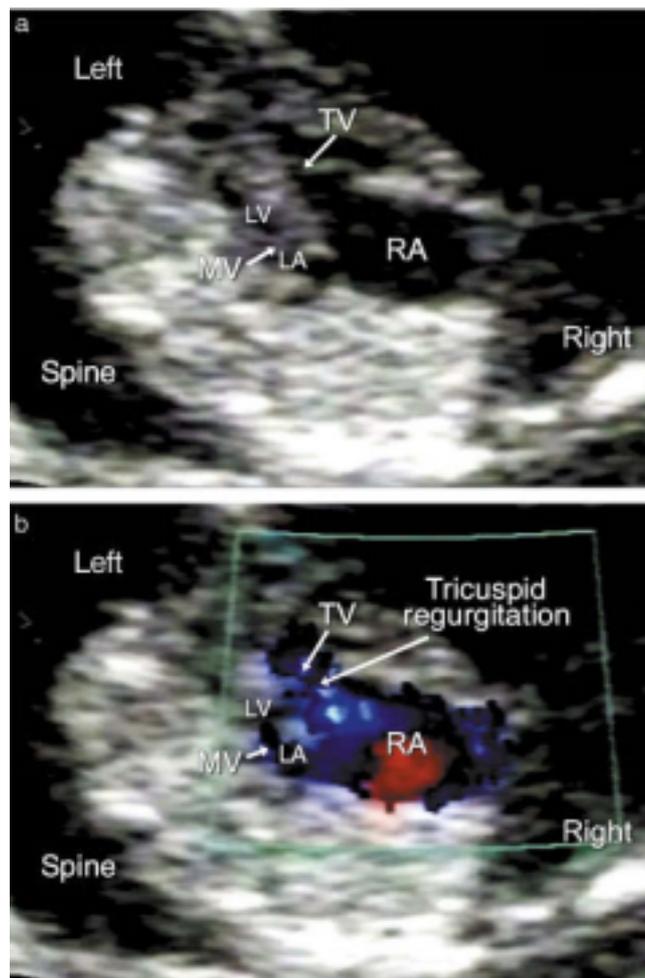


Figure 9 Ebstein's anomaly of the tricuspid valve in a fetus with crown–rump length 66.7 mm (12+6 weeks). (a) Four-chamber view demonstrating gross dilation of the right atrium and displacement of the tricuspid valve towards the apex of the right ventricle. (b) Color flow map in a similar view. The tricuspid regurgitant jet originates from deep within the anatomical right ventricle, corresponding to the displaced valve. TV, tricuspid valve; LV, left ventricle; MV, mitral valve; LA, left atrium; RA, right atrium.

a normal outcome at later gestations. Their frequent association with an abnormal outcome in our study may be a reflection of the high-risk nature of the population studied and preferential detection of more severe examples on technically demanding scans as well as the gestational age. Previous reported series of early fetal echocardiography, although including some examples at 12 weeks' gestation or earlier, have in the main included fetuses at a more advanced gestation than those in this study. The especially high-risk nature of the group of fetuses we have studied, and correspondingly high incidence of abnormalities seen, has facilitated a relatively rapid acquisition of experience in the interpretation of abnormal scans. Infrequent exposure of operators to abnormalities has been postulated as an important reason for disappointing results of screening of low risk populations by early fetal echocardiography¹⁴. Autopsy following termination of pregnancy for chromosomal abnormalities permitted pathological correlation of abnormal scan findings, such as VSDs that would not in themselves be indications for termination. Pathological and postnatal correlation of

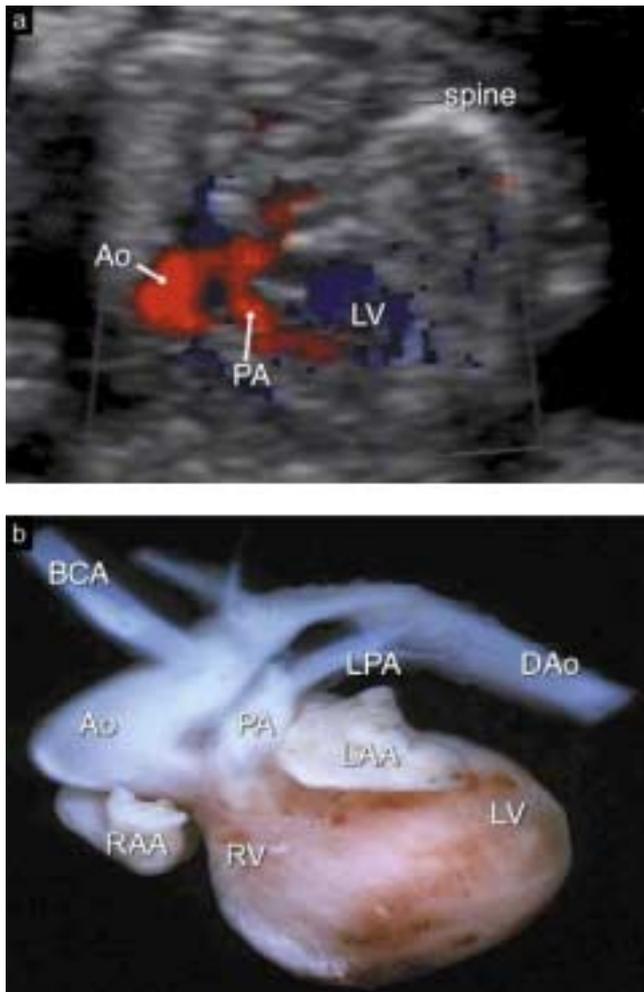


Figure 10 Tricuspid atresia with concordant ventriculoarterial connection in a fetus with crown–rump length 76.3 mm (13+3 weeks). (a) Flow map of the great vessels. Forward flow in a small pulmonary artery, normally related to the larger aorta, is demonstrated. (b) Pathological specimen in similar orientation for comparison. Ao, aorta; PA, pulmonary artery; LV, left ventricle; BCA, brachiocephalic artery; LPA, left pulmonary artery; DAo, descending aorta; RAA, right atrial appendage; LAA, left atrial appendage; RV, right ventricle.

fetal echocardiographic findings has been an essential part of the advancement of fetal cardiology generally and is especially important in those areas where ultrasound diagnosis is most challenging. Despite the considerable technical and logistic difficulties, we advocate that pathological confirmation be given high priority whenever early diagnosis is attempted.

Transabdominal vs. transvaginal echocardiography

Transabdominal fetal echocardiography predominated in this study because our early experience showed it to be effective and because of the limited availability of an operator skilled at cardiac scanning transvaginally, the importance of which has recently been emphasized¹⁴. The major potential advantage of transvaginal echocardiography is higher resolution, but there may be less flexibility in obtaining different scanning planes, especially when the uterus is still within the pelvis and restricts external manipulation. However, the study makes no attempt to formally compare the relative

merits of transvaginal and transabdominal scanning for early fetal echocardiography.

Indications for early fetal echocardiography

Fetal echocardiography becomes more consistently successful at a later gestational age, even within the range studied, and therefore early scanning does require justification. An advantage of early cardiac scanning is that it can be combined at the same visit with NT assessment (currently at 11+1 to 13+6 weeks). Furthermore, if termination of pregnancy is decided upon because of findings on the cardiac scan, then this is more likely to be achieved safely by surgical means than at later gestations, a method frequently preferred by the patient. However, whilst early fetal echocardiography can successfully identify major defects, it is not currently sufficiently accurate to dispense with follow-up fetal echocardiography in later pregnancy and therefore implies additional resource input. Valid indications for early fetal echocardiography are open to question.

No cardiac defects were identified in the 28 pregnancies in which a family history of a heart defect was the main indication for fetal echocardiography, a finding consistent with the recurrence rate of 2–3% for siblings¹⁵. The value of fetal echocardiography for someone with a family history lies mainly in the reassurance that it gives rather than a high yield of abnormalities revealed. Reassurance from a normal early scan is bound to be qualified but may help in the early resolution of the extreme anxiety which dominates those families with previously affected pregnancies.

The high levels of increased NT affecting the largest subgroup of fetuses scanned largely explain the high incidence of chromosomal abnormalities and of cardiac defects in the study group as a whole. Increased NT identifies fetuses at high risk of cardiac defects in association with chromosomal abnormality, as well as those with isolated cardiac defects. The diagnosis of a heart defect was often overshadowed by the subsequent diagnosis of a chromosomal abnormality with an even more devastating impact on the predicted outcome for the fetus. Often, the nature of the cardiac defect would allow prediction of a likely abnormal karyotype result in advance, and prepare the parents for further bad news. However, because of the rapid diagnostic techniques used, the delay between the chorionic villus sampling being performed and a result being available was usually only a few days. Nevertheless, cardiac defects without chromosomal abnormality were also frequently identified in this group with increased NT and, in these, the early cardiac diagnosis did impact significantly on the management.

Cardiac abnormalities suspected at the general 11–14-week scan were always confirmed as important abnormalities on subsequent echocardiography. Although detailed evaluation of the heart was often technically demanding and time consuming, it was also the case that some major cardiac abnormalities were surprisingly obvious on the scan; for example, those shown in Figures 8 and 9. An attempt to look at the fetal heart as part of the routine 10–14-week scan, with early echocardiography in those with suspicious findings, therefore does seem justified.

Association with chromosomal abnormality

Strong associations of VSD and of AVSD with the major trisomies, as shown in Table 2, are to be expected, especially given the predominance of fetuses with increased NT in the series.

Both the frequency and the severity of left heart disease occurring in fetuses with Turner syndrome in this series is greater than that identified in Turner syndrome postnatally¹⁶. Turner syndrome presenting prenatally is associated with a very high spontaneous loss rate. We postulate that severe genetic variants of Turner syndrome, associated with intrauterine lethality, generally have more frequent and severe heart disease than those less severe variants that are more likely to survive the pregnancy.

Both isolated tricuspid regurgitation and disproportion were frequently associated with trisomies. Apparently isolated atrioventricular valve regurgitation in association with trisomy may have been due to subtle examples of AVSD not recognized on the scan (demonstrated in one fetus), or to polyvalvar dysplasia associated with trisomy 18¹⁷. However atrioventricular valve regurgitation was identified in four fetuses where normal heart structure was confirmed at later scans. Moreover, in each of these fetuses, valve regurgitation was reduced or abolished at follow-up fetal echocardiography. Tricuspid regurgitation has been proposed as a second-trimester marker for trisomy 21^{18,19}. Similarly, disproportion in the second-trimester fetus is a marker for chromosomal abnormality^{18,19}; and our own data indicate a similar association earlier in pregnancy. Our preliminary data suggest that these features may be strong indicators of chromosomal abnormality at 10–14 weeks' gestation, although it remains uncertain to what extent the close relationship between adverse outcome and these findings is a manifestation of the early gestational age alone and to what extent it reflects the particularly high-risk nature of the fetuses studied.

CONCLUSIONS

Fetal echocardiography performed at the same visit as NT assessment can identify major heart defects in a very high-risk group with reasonable accuracy, often using transabdominal scanning alone. In this study, a high incidence of chromosomal abnormality in association with many heart defects limited the impact of the cardiac scan result on the overall clinical management in many patients. However, in some patients, the cardiac scan did provide vital information at an earlier stage in the pregnancy than would previously have been possible. The concentrated experience obtained from performing and interpreting many abnormal scans in this high-risk group was invaluable for the relatively few cases where confident diagnosis of the heart defect was critical to management.

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