Poor sensitivity of routine fetal anomaly ultrasound screening for antenatal detection of atrioventricular septal defect

H ter Heide, J D R Thomson, G A Wharton, J L Gibbs


PATIENTS AND METHODS

All patients with AVSD born between 1996 and 2001 within the catchment region of the tertiary centre were identified by a search of prospectively collected, comprehensive diagnostic information held in the department’s electronic congenital heart disease database.

Information relating to referrals for detailed fetal echocardiography was retrieved from the same database. Obstetric ultrasound screening was performed in referring hospitals according to unit specific protocols. A four chamber view of the heart was included as part of routine obstetric antenatal ultrasound. The antenatal diagnosis rate was worse for liveborn infants with trisomy 21 (12 of 49 (25%) v 15 of 43 (35%) chromosomally normal children) and for infants with AVSD without other structural heart disease (18 of 74 (24%) v 9 of 18 (50%) infants with associated structural heart disease).

Conclusion: Despite the potential ability of fetal ultrasound to detect AVSDs, the antenatal diagnosis rate is poor. This is particularly true for infants with trisomy 21 and is of importance when counselling parents with an apparently normal fetal ultrasound scan.
detected by routine antenatal obstetric ultrasound screening. All patients with antenatally diagnosed AVSDs had been referred for detailed fetal echocardiography. In one case where a four chamber view of the fetal heart could not be obtained at screening, specialist fetal echocardiography also failed to evaluate cardiac anatomy because of gross maternal obesity and an AVSD was diagnosed postnatally.

Nine of 18 (50%, 95% CI 4.8 to 13.2) infants with AVSD in association with complex congenital heart disease were identified by routine antenatal screening.

In the 49 liveborn infants with AVSD and Down’s syndrome the AVSD (and trisomy 21) was diagnosed antenatally in only 12 (24.5%, 95% CI 6.1 to 17.9). In the 43 children born with an AVSD and normal chromosomes the cardiac abnormality was detected antenatally in 15 (34.9%, 95% CI 8.8 to 21.1; not significant).

**DISCUSSION**

Antenatal diagnosis of AVSD is of particular clinical and medicolegal importance because of its strong association with trisomy 21. Despite multiple potential anomalies in babies with trisomy 21, non-cardiac sonographic markers (for example, renal pelvic dilatation) are often subtle and are frequently missed during screening. Therefore, the ability of fetal ultrasound to detect congenital heart disease accurately in this setting is particularly important. There is an understandable and widely held view that AVSD, particularly in the presence of a chromosomal anomaly, should not be missed at a screening scan. In low risk fetal populations, however, detection rates during screening for congenital heart disease as a whole are reported to vary from 13–92%. The antenatal detection rate for a consecutive series of liveborn patients with AVSD, a lesion potentially diagnosable on the standard four chamber view, has not been reported before. In our series 70% of AVSDs were diagnosed only after birth despite the majority of referring centres in our study having received basic on-site fetal echocardiography training. Although not significant, the rate of detection in babies with trisomy 21 was less than for chromosomally normal infants.

Once complex cases known to be associated with AVSDs in infants without trisomy were removed from the analysis the detection rate was similar at 25% and 23% for infants with and without trisomy 21, respectively.

It is beyond the scope of this paper to discuss detection rates for other congenital cardiac lesions; however, the echocardiographic hallmark of the AVSD is a common atroventricular valve, which even on a good four chamber view can appear superficially normal.

By implication AVSDs may be more difficult to detect at standard screening than lesions with more obvious structural pathology. Although training for obstetric screening units was almost universal in this series and this undoubtedly translated to a greater awareness of congenital heart disease, perhaps technical proficiency can be gained only by exposure to a large number of abnormal cases. For the average obstetric screening unit this is an unrealistic expectation. Effective screening for this important lesion may require a different strategy.

**Conclusion**

The antenatal detection rate for AVSD by standard ultrasound screening in the four chamber view is poor overall, with a trend towards being even poorer in the presence of trisomy 21.

**Authors’ affiliations**

H ter Heide, J D R Thomson, G A Wharton, J L Gibbs, Department of Paediatric Cardiology, Leeds General Infirmary, Leeds, UK

There were no conflicts of interest. All authors had full access to all the data in the study. The corresponding author had the final responsibility for the decision to submit for publication.

**REFERENCES**


**Table 1** Associated cardiac lesions

<table>
<thead>
<tr>
<th>Anatomy</th>
<th>Number (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Right atrial isomerism*</td>
<td>6 (33%)</td>
</tr>
<tr>
<td>Left atrial isomerism*</td>
<td>3 (17%)</td>
</tr>
<tr>
<td>Tetralogy of Fallot</td>
<td>3 (17%)</td>
</tr>
<tr>
<td>Pulmonary atresia</td>
<td>3 (17%)</td>
</tr>
<tr>
<td>Coarctation</td>
<td>1 (6%)</td>
</tr>
<tr>
<td>Double outlet right ventricle</td>
<td>1 (6%)</td>
</tr>
<tr>
<td>Muscular ventricular septal defect</td>
<td>1 (6%)</td>
</tr>
</tbody>
</table>

*All with complex intracardiac abnormalities.
Poor sensitivity of routine fetal anomaly ultrasound screening for antenatal detection of atrioventricular septal defect

H ter Heide, J D R Thomson, G A Wharton and J L Gibbs

Heart 2004 90: 916-917
doi: 10.1136/hrt.2003.018895

Updated information and services can be found at:
http://heart.bmj.com/content/90/8/916

These include:

References
This article cites 7 articles, 0 of which you can access for free at:
http://heart.bmj.com/content/90/8/916#BIBL

Email alerting service
Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

Topic Collections
Articles on similar topics can be found in the following collections

Clinical diagnostic tests (4779)
Congenital heart disease (762)
Drugs: cardiovascular system (8842)
Echocardiography (2127)
Epidemiology (3752)

Notes

To request permissions go to:
http://group.bmj.com/group/rights-licensing/permissions

To order reprints go to:
http://journals.bmj.com/cgi/reprintform

To subscribe to BMJ go to:
http://group.bmj.com/subscribe/