A systematic review of the accuracy of first-trimester ultrasound examination for detecting major congenital heart disease


CRD summary
This review found that a positive ultrasound scan is accurate for diagnosing major congenital heart disease (CHD) in the first trimester and that a negative scan has reasonable accuracy for ruling out CHD. The conclusions regarding a positive scan appear reliable, whereas those regarding a negative scan should be interpreted with caution given the variability between the studies.

Authors' objectives
To determine the accuracy of ultrasound screening for major congenital heart disease (CHD) in the first trimester.

Searching
MEDLINE and EMBASE were searched from inception to July 2005; the search terms were reported. The Cochrane Library, National Research Register and Research Findings Electronic Register were also searched. Selected specialist journals were handsearched, reference lists of primary studies and relevant reviews were screened, and experts in the area were contacted.

Study selection
Study designs of evaluations included in the review
Studies that included at least 10 women were eligible for inclusion.

Specific interventions included in the review
Studies that assessed transvaginal or transabdominal ultrasound to detect structural abnormalities of the heart were eligible for inclusion. Studies in which the ultrasound technique was not fully described, was only four-chamber view, or was not specifically directed at identifying CHD were excluded.

Reference standard test against which the new test was compared
Studies that included verification by mid-trimester scan (four-chamber view plus outflow tracts), postnatal echocardiography or post-mortem were eligible for inclusion. Studies that did not include a reference standard were excluded.

Participants included in the review
Studies of women in the first trimester of pregnancy (<14 weeks) were eligible for inclusion. Studies in which women were examined after 13 + 6 weeks' gestation were excluded. Most studies included high-risk mothers with a previous history of a child with cardiac disease or increased nuchal translucency thickness.

Outcomes assessed in the review
The studies had to report sufficient data to construct a 2x2 table of test performance. All studies used a diagnostic cohort design. The included studies were both prospective and retrospective in design.

How were decisions on the relevance of primary studies made?
Three reviewers assessed the studies for relevance; it was unclear whether this was done independently.

Assessment of study quality
Studies were assessed for methodological quality using the following seven items: patient spectrum, prospective design, consecutive recruitment, adequate description of the technique, adequate reference standard, blinding of the results, and over 90% verification. Studies that fulfilled four or more items were considered to be of a high quality. It was unclear
how many reviewers performed the quality assessment.

Data extraction
Data were extracted as 2x2 tables of test performance. Data were only extracted for foetuses of less than 14 weeks' gestation. Authors were contacted for further information if necessary. Sensitivity and specificity, together with 95% confidence intervals (CIs), were calculated for each study. If any of the 2x2 table cells contained 0 cells, 0.5 was added to all cells of that table. It was unclear how many reviewers performed the data extraction.

Methods of synthesis
How were the studies combined?
The sensitivity and specificity were pooled separately for transvaginal and transabdominal ultrasound. These pooled values were then combined to give an overall pooled value. The pooled measures were used to estimate pooled positive and negative likelihood ratios. The Fagan nomogram was used to transform pre-test probability to post-test probability using the pooled likelihood ratios.

How were differences between studies investigated?
Heterogeneity was assessed using the chi-squared test. A subgroup analysis was carried out for high-quality studies.

Results of the review
Ten studies (1,243 mothers) were included.

Eight studies assessed a high-risk population. Eight studies used a prospective design of which seven enrolled consecutive patients. Only 4 studies used an appropriate reference standard (postnatal or postmortem assessment). No studies reported blinding. Nine studies verified the diagnosis in over 90% of foetuses.

Transabdominal ultrasound (4 studies).

Sensitivity was 25% in 1 study and 100% in 2 studies; it could not be calculated in the remaining study as no CHD was detected. Specificity was 100% in all studies.

Transvaginal ultrasound (4 studies).

The sensitivity ranged from 33 to 86% and the specificity from 96 to 100%; there was no statistical evidence of heterogeneity (p=0.347 and p=0.120, respectively). The pooled sensitivity was 62% (95% CI: 44, 78) and the pooled specificity was 99% (95% CI: 98, 100).

Transabdominal and transvaginal ultrasound combined (2 studies).

The sensitivity was 78% in 1 study and 89% in the other. Specificity was 100% in both studies.

Authors' conclusions
The accuracy of a positive ultrasound scan for diagnosing major CHD in the first trimester was very high. A negative ultrasound scan has reasonable accuracy for ruling out a diagnosis of CHD.

CRD commentary
This review addressed a focused question that was supported by clearly defined inclusion criteria. The literature search appeared adequate with attempts made to locate both published and unpublished data and no apparent language restrictions imposed. Three independent reviewers selected the studies, but it was unclear whether appropriate steps were taken to minimise bias in other review processes such as extracting the data and assessing study validity. A detailed quality assessment was conducted and considered in the analysis.
The methods used to pool the data were appropriate but given the heterogeneity in the results, especially in terms of sensitivity, the analysis based on these pooled values and the estimates of the post-test probability of disease should be interpreted with great caution. In particular, for the analysis of transabdominal ultrasound, there was an extreme outlier and potential reasons for this were not explored. Given the differences in sensitivity between transabdominal and transvaginal ultrasound it does not appear valid to combine these measures. The authors' conclusions that a positive scan can be used to rule in the diagnosis appear reliable. However, given the heterogeneity in estimates of sensitivity their conclusions regarding the ability of a negative scan to rule out CHD should be interpreted with great caution.

**Implications of the review for practice and research**
The authors did not state any implications for practice or research.

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