Knowledge update on routine midtrimester ultrasound screening for heart defects
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September 2012

OBJECTIVE: to get a sense of the summary of knowledge relating to screening 18-20 weeks in terms of heart defects and their detection rates

SOURCES SEARCHED: Medline (OvidSP), Embase, and the Cochrane Library.

DATES OF SEARCH: January 2003 – September 2012

SEARCH STRATEGY:

1. exp Heart Defects, Congenital/ (112448)
2. ((heart or cardiac) adj (defect$ or anomal$ or malformation$ or abnormalit$)).tw. (17738)
3. coarct$.tw. (7460)
4. (double outlet right ventricle or DORV).tw. (1250)
5. (double outlet adj2 ventricle).tw. (1296)
6. endocardial cushion defect.tw. (295)
7. ((left ventric$ outflow adj2 obstruct$) or LVOT$).tw. (2017)
8. ((interrupt$ adj3 aort$ heart) or IAA).tw. (3612)
9. (hypoplastic left heart or HLHS).tw. (3738)
10. ((mitral or aorti$) adj (atresia or stenosis)).tw. (16729)
11. PVOD.tw. (117)
12. Eisenmenger$ syndrome.tw. (663)
13. ((transposition adj3 great arter$) or TGA).tw. (7623)
14. (univentric$ heart or UVH).tw. (494)
15. single ventric$.tw. (2202)
16. (anomalous pulmonary adj2 drainage).tw. (639)
17. (anomalous pulmonary venous adj (return or connection)).tw. (1445)
18. (TAPVD or TAPVR or TAPVC or PAPVD or PAPVR).tw. (367)
19. (ventricular septal defect or VSD).tw. (9072)
20. (((atrioventricular or ventricular) adj septal defect) or VSD or AVSD).tw. (9720)
21. (pulmonary adj2 (atresia or stenosis)).tw. (6708)
22. (tricuspid adj2 stenosis).tw. (576)
23. tetralogy of fallot.tw. (5858)
24. patent ductus arteriosus.tw. (5667)
25. atrial septal defect.tw. (5952)
26. patent foramen ovale.tw. (3023)
27. ventricular septal defect.tw. (7900)
28. branch pulmonary artery.tw. (193)
29. 1 or 2 or 3 or 4 or 5 or 6 or 7 or 8 or 9 or 10 or 11 or 12 or 13 or 14 or 15 or 16 or 17 or 18 or 19 or 20 or 21 or 22 or 23 or 24 or 25 or 26 or 27 or 28 (156046)
30. exp Prenatal Diagnosis/ (56396)
31. 29 and 30 (3626)
32. (screen$3 or detect$3 or test or tests or testing).tw. (2754727)
33. Mass Screening/ (75851)
34. 32 or 33 (2771937)
35. (midtrimester or 2nd trimester or second trimester).tw. (10883)
36. Pregnancy Trimester, Second/ (11734)
37. 35 or 36 (17994)
38. 29 and 34 and 37 (266)
39. Ultrasonography, Prenatal/ (22568)
40. (ultrasonogra$ or ultrasound or scan).tw. (279634)
41. 39 or 40 (290210)
42. 38 and 41 (225)
43. limit 42 to yr="2003 -Current" (131)

Similar searches were also carried out in Embase, and the Cochrane Library.

All searches carried out on 10 September 2012

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Inclusions and exclusions

The above search strategies retrieved 285 references in total. After duplicate references were removed a total of 201 potentially relevant references were left. The title and abstracts of the remaining citations were scanned for relevance to routine midtrimester ultrasound screening for heart defects with the following exclusions and inclusions

**Exclusions**
- Screening in the first trimester (unless compared with second trimester screening)
- Echocardiography (unless compared with second trimester screening or in relation to referral for echocardiography after screening)

**Inclusions**
- Second trimester ultrasound screening for heart defects.

35 references were deemed to be relevant and are classified into the categories below. There will inevitably be some overlap between categories.

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<td>Accuracy</td>
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These references are listed below. Abstracts have been provided where possible.

**Accuracy (11)**


Background: Prenatal detection rate of conotruncal heart defects have remained significantly below 50%. We have recently established a concept of 3D/4D Automated Multiplanar Imaging (AMI) as well as developed formulas for the automated retrieval of six diagnostic cardiac planes out of a 3D/STIC volume of the fetal chest in the 2<sup>nd</sup> trimester of pregnancy. Reliability of this software was tested on volumes obtained on fetuses with normal cardiac anatomy. The aim of this study is to assess potential clinical applicability AMI in prenatal detection of conotruncal heart defects. Methods: Detailed 2D echocardiography performed in 25 fetuses with normal cardiac anatomy and 24 fetuses with conotruncal heart anomalies between 18 and 23 weeks of gestation by trained sonographer. In addition 3D/STIC volumes of the fetal chest were acquired at the level of the 4-chamber view. Only one volume dataset per case was included in the study. The initial scan was interpreted and reported by based on the 2D images. The volume data sets were independently reviewed offline using AMI software by pediatric cardiologist with experience in fetal heart assessment. The diagnostic value, image quality as well as time for acquisition and reading of AMI was evaluated and compared with original 2D report. Prenatal diagnosis was confirmed in all cases by postnatal echocardiography, angiography, operative findings, or autopsy.

Results: A summary of the results is provided in the table. (Table presented)

Conclusion: The developed software reported by based on the 2D images. The volume data sets were independently reviewed offline using AMI by trained sonographer. In addition 3D/STIC volumes of the fetal chest were acquired at the level of the 4-chamber view. Only one volume dataset per case was included in the study. The initial scan was interpreted and reported by based on the 2D images. The volume data sets were independently reviewed offline using AMI software by pediatric cardiologist with experience in fetal heart assessment. The diagnostic value, image quality as well as time for acquisition and reading of AMI was evaluated and compared with original 2D report. Prenatal diagnosis was confirmed in all cases by postnatal echocardiography, angiography, operative findings, or autopsy.

Results: A summary of the results is provided in the table. (Table presented)

Conclusion: The developed software demonstrated an excellent display of the diagnostic landmarks of conotruncal defects with appropriate quality of images in most cases. This should help improve the detection of these heart anomalies in future. Automated sonography also has the potential for improving the efficiency of ultrasound imaging by reducing the time needed to complete an ultrasound examination, thereby resulting in increased throughput of ultrasound laboratories.


OBJECTIVE: We sought to evaluate the influence of maternal body mass index (BMI) on sonographic detection employing data from the FaSTER trial. METHOD: Unselected singleton pregnancies underwent detailed genetic sonogram to evaluate for structural fetal anomalies and soft markers for aneuploidy. BMI (kg/m(2)) were calculated from reported initial visit values. Sensitivity, specificity, false positive and false negative rates (FPR and FNRR), likelihood ratio, detection rates, and a missed diagnosis rate (MDR) were calculated. RESULTS: Eight thousand five hundred and fifty-five patients with complete BMI information had detailed genetic sonography. A lower sensitivity with an elevated FNR and MDR was observed in obese women for multiple aneuploid markers (e.g. > or =2 markers 32% sensitivity with 68% FNR among BMI < 25 vs 22% and 78% among BMI > 30). Similarly, the detection rate for cardiac anomalies among women at BMI < 25 was higher (21.6%) at a significantly lower FPR (78.4%; 95% CI 77.3-79.5%) in comparison to obese women (8.3% with FPR 91.7%; 95% CI 90.1-93.2%). In a logistic regression model, maternal obesity significantly decreased the likelihood of sonographic detection of common anomalies (adjusted OR 0.7; 95% CI 0.6-0.9; p = 0.001). CONCLUSION: The performance of second trimester genetic sonography is influenced by obesity, with a significantly higher MDR for multiple minor markers and lower likelihood for detecting common anomalies. Copyright (c) 2009 John Wiley & Sons, Ltd.


OBJECTIVE: Routine ultrasound screening in the second trimester detects 40-70% of major congenital anomalies. We examined the effect of increasing BMI on the detection rate and accuracy of prenatally diagnosed congenital anomalies. STUDY DESIGN: Retrospective chart review. Neonates with discharge diagnosis of any congenital anomalies between 2003 and 2008 were included. Maternal charts were reviewed for gestational age at ultrasound, ultrasound diagnosis, and maternal BMI. Subjects were classified into BMI categories by CDC criteria: underweight and normal (control): 18.5-24.9 kg/m(2), overweight: 25.0-29.9 kg/m(2), and obese: 30 kg/m(2) and above. Detection rate and how well the ultrasound diagnosis correlated to the discharge diagnosis was compared between categories. RESULTS: 223 neonates were included; 26 neonates had more than one anomaly. Anomalies were classified into cardiac, central nervous system (CNS), and other (urology, GI, chest, skeletal, face). Detection rate was lower in the overweight (70.8%, p = 0.0267) and obese (71.8%, p = 0.0218) groups, when compared to controls (90%). Decrease persisted when only the non-referred population was included (59%, p = 0.0181 for overweight, 55.3%, p = 0.0038 for obese). For cardiac anomalies, the detection rate was significantly decreased in the obese group (41.2%, p = 0.0266), and approached significance in the overweight group (44%, p =
Correlation between ultrasound and discharge diagnosis was decreased with increasing BMI: 90%, 69%, and 70% in the control, overweight, and obese groups (p = 0.0180 and p=0.0136). Correlation for cardiac abnormalities was also decreased (p=0.0329, overweight and p=0.188, obese). No difference was noted in the CNS group. CONCLUSION: Increasing BMI significantly decreases the detection rate of fetal anomalies during ultrasound evaluation, when compared to women with underweight or normal BMI. The detection of cardiac anomalies is the most affected by this decrease. Correlation between ultrasound and discharge diagnosis is also negatively affected by increasing BMI, mostly with cardiac anomalies.


OBJECTIVE: To assess whether medically qualified sonologists with low-to-intermediate scanning experience are able to detect major abnormalities of the outflow tracts by reviewing the A-plane of cardiac volume datasets acquired with spatiotemporal image correlation (STIC). METHODS: Fourteen sonologists of low-to-intermediate scanning experience were recruited among residents and colleagues involved in the screening ultrasound clinic at our referral center. Basic criteria for selection were: ability to perform the 20-week anomaly scan and to assess the four-chamber view, inability to perform extended cardiac screening (outflows); willingness to participate in the study. These sonologists attended a 2-hour lesson on: a) how the outflow tract views can be abnormal, and b) how to use a laptop and the dedicated software to review cardiac volumes in the A-plane only. After this briefing, each of them, independently, reviewed 26 preselected volumes at a workstation (from 16 normal fetuses and 10 with outflow tract abnormalities), without knowing how many of them were normal. After reviewing each volume, the sonologist was asked to define the outflow tract views as normal or abnormal and, if willing, to hypothesize the anomaly. The sequence of cases was changed for each participant. The time allotted for review of the volumes was 1 hour (about 2 min per case). RESULTS: Of the 364 diagnoses from review of the volumes, 116 (31.9%) were true positives, 195 (53.6%) were true negatives, 29 (8.0%) were false positives and 24 (6.6%) were false negatives. The sensitivity, specificity and positive and negative predictive values were 83%, 87%, 80% and 89%, respectively. Individual diagnostic accuracy ranged from 66 to 100% (median, 85.5%) and individual detection rate from 50 to 100% (median, 85%). The detection rate per single congenital heart disease ranged from 50% (for TGA with intact ventricular septum) to 100% (for DORV, DORV with pulmonary atresia and TGA with ventricular septal defect). There was no correlation between detection rate and alignment of the four-chamber view with the ultrasound beam (apical vs. transverse). CONCLUSIONS: In this preliminary study, we have demonstrated that sonologists with low-to-intermediate experience of anomaly ultrasound screening in the second trimester and no experience of insonating the outflow tracts were able to identify outflow tract abnormalities by reviewing the A-plane of cardiac volume datasets, after detailed briefing. (c) 2008 ISUOG.


OBJECTIVES: Congenital heart disease is associated with high mortality and morbidity rates, being the most life-threatening defect in the first month of postnatal life and accounting for approximately half of all childhood mortality from birth defects. Despite this, the prenatal detection rate for congenital outflow tract abnormalities is the most affected by this decrease. Correlation between ultrasound imaging is relatively low. The aim of this study was to establish a nomogram for the pulmonary artery/aorta (PA/IO) ratio measured in the three-vessel view plane. This ratio was investigated as a simple screening tool for congenital cardiac outflow tract abnormalities. METHODS: The study was a prospective evaluation of 966 singleton fetuses at 16-24 weeks of gestation and 46 fetuses with congenital cardiac outflow tract abnormalities. The diameters of the pulmonary artery and aorta were measured in the three-vessel view. The PA/IO ratio was calculated and a nomogram was constructed. The mean and 95% CI for the ratio were defined and the PA/IO ratios of cases with outflow tract abnormalities were plotted against the values for normal fetuses. RESULTS: The gestational age of the normal group ranged from 16 to 24 weeks, with a median of 19 weeks. The pulmonary artery diameter measured between 2.1 and 4.93 mm (mean, 3.3 mm) and the aorta measured between 2.1 and 5.2 mm (mean, 2.93 mm). The mean (SD) PA/IO ratio was 1.16 (0.18) (95% CI, 0.87-1.58; range 0.61-1.86; median, 1.14). For fetuses with outflow tract abnormalities, the median gestation was 19 weeks and 37/43 (86%) had a PA/IO ratio outside the 95% CI. CONCLUSIONS: The PA/IO ratio derived from measurements in the three-vessel view plane can be used as an initial screening tool for outflow tract anomalies and may have a sensitivity of up to 86%, with a 5% false-positive rate. Copyright 2007 ISUOG. Published by John Wiley & Sons, Ltd.


OBJECTIVES: To compare fetal heart evaluation done through two-dimensional (2DUS) and three-dimensional ultrasonography (3DUS) as to optimal plane imaging, image quality, and time needed to perform the examination.

METHODS: Prospective study involving 12 normal pregnant women, with gestational ages ranging from 22 to 26 weeks, scanned with a VOLUSON 730 with a convex 4.0-7.0 MHz transducer, in both two- and three-dimensional modes. In each case, three basic view planes were obtained: four-chambers view, right, and left ventricular outflow tracts. Each view was subjectively evaluated by three different examiners as to image quality, and graded from 0 (minimum) to 4 (maximum) cross-marks (+). The sum of all grades obtained for each case was used to classify the quality of the exam as unsatisfactory (0 to 1+), poor (2 to 4+), regular (5 to 7+), and good (8 to 12+). The time taken to obtain the views was recorded for each case, starting with the acquisition of the first view on the 2D exam and with the identification and opening of the volume blocks on the three-dimensional software.

RESULTS: The number of three-dimensional blocks with good, regular, poor, and unsatisfactory grades were, respectively, 6, 15, 9, and 10. The average in cross-marks of the cases graded good in each group without the worst result for each plane was 8. 2DUS was superior to 3DUS regarding the quality of the images obtained by the three pattern view planes and the average time to obtain high quality view planes was longer for 3DUS when compared to 2DUS. 2DUS offered better quality images and in less time than 3DUS. CONCLUSIONS: Three-dimensional ultrasound is an advancement in fetal heart evaluation; however two-dimensional ultrasound remains the best screening method in diagnosing cardiac malformations, due to the good quality of its images and the lesser time needed to perform the exam.


OBJECTIVES: To determine whether training and experience in performing ultrasound examinations are factors that influence the prenatal detection of congenital heart defects (CHDs) in a non-selected population, in order to evaluate and improve the current training program. METHODS: All pregnant women who received a routine second-trimester ultrasound scan by a sonographer/midwife and delivered at our hospital between February 1991 and December 2001 were registered prospectively. Less experienced sonographer/midwives who had performed between 200 and 2000 routine examinations were compared with experienced sonographer/midwives who had carried out more than 2000 examinations. During the first 5 years of the study the heart structures obtained were registered in detail. RESULTS: Of 29,035 fetuses, 35/82 (43%) major CHDs were prenatally detected at the routine examination. The experienced sonographer/midwives obtained both the four-chamber view and the great arteries in 75%; the figure for the less experienced sonographer/midwives was 36% (P < 0.001). The differences in detecting major heart defects were 22/42 (52%) and 13/40 (32.5%), isolated CHDs 8/18 (44%) and 6/22 (27%) and CHDs with associated malformations 14/24 (58%) and 7/18 (39%), respectively. In both groups some CHDs with an abnormal four-chamber view were missed, although the experienced sonographer/midwives recognized significantly more of the abnormal views than did the less experienced sonographer/midwives (P = 0.002). CONCLUSIONS: Experience has a significant impact on the examination of the fetal heart and the prenatal detection rate of major CHDs. To avoid a relatively long learning curve, ultrasound education needs to intensify the teaching of the basic four-chamber view. The great arteries should be included after additional training. Those basic views of the fetal heart must be mastered before new views and advanced technology are added to the fetal heart examination. 2006 ISUOG. Published by John Wiley & Sons, Ltd.


OBJECTIVE: To test the efficacy of a recently introduced ultrasonic scanning plane (three vessel and trachea view - 3VTV- plus color flow mapping -3VTCV) on a low-risk population for detection of congenital heart disease (CHD).

PATIENTS AND SETTING: Antenatal clinic dealing with local low-risk population. All antenatal patients having a second trimester scan in a 1 year period. All patients had a 3VT plus 3VTC views added to routine four chamber view. Postnatal examinations were performed according to standard hospital protocol. RESULTS: 2847 patients were examined. The plane was achievable in all 23 fetuses with CHD detected, three false negative (aortic coarctation) and two false positive. Sensitivity of the examination was 88.5%, as high as more sophisticated and difficult targeted cardiac scanning. The extra time necessary to perform the test was minimal. CONCLUSION: 3VTV and 3VTCV were satisfactory used as imaging planes in a busy antenatal clinic in a low-risk population. They could
be easily added to the four chamber view as routine screening for CHD and increase the detection rate to 90%.


BACKGROUND: Foetal echocardiography has become a diagnostic method to detect foetal congenital heart disease with high probability. However, it is not only time consuming and but also difficult to visualize outflow tract of foetus early in the second trimester of pregnancy, even for an experienced obstetric ultrasonographer. Recently, many methods for screening foetal cardiac anomalies were explored, but much more work is needed to develop an effective and suitable screening method. The aim of this study was to investigate the clinical significance of utilising the ductus venosus (DV) Doppler examination and the four-chamber view of heart to screen for foetal cardiac malformation in early second trimester of pregnancy. METHODS: Heart and DV of 401 consecutive foetuses in early second trimester (12(+1)-17(+6) weeks) in high risk pregnancies were examined with Acuson 128 xp/10 or Sequoia 512 ultrasound diagnostic systems. Absent or reversed flow during atrial contraction (A-wave) in the DV was defined as sufficiently abnormal to screen for foetal cardiac malformations. The foetal echocardiographic diagnosis was confirmed by postnatal echocardiography (or postmortem). The sensitivities of screening tests were compared among the three methods: DV Doppler examination, four-chamber view alone, and the combination of both techniques. RESULTS: Satisfactory examinations were obtained in 383/401 foetuses (95%). Thirty foetuses with cardiac abnormalities were confirmed by neonatal echocardiography (or postmortem). The sensitivity of DV Doppler examination or four-chamber view alone is 63% (19/30) and 60% (18/30), respectively. The sensitivity of combining information, DV Doppler flow waveform and four-chamber view, to screen for foetal cardiac malformation is 83% (25/30) and significantly better than that of either DV Doppler flow waveform or four chamber view alone (P < 0.05). CONCLUSION: Doppler flow waveform of DV can be used to screen for foetal cardiac malformation early in the second trimester. Combining information from Doppler flow waveform of DV and four-chamber view will improve the overall sensitivity of the screening.


Background. To assess the sensitivity for detecting fetal congenital anomalies by a routine ultrasound examination program at midtrimester performed in an unselected population by midwives and specialists in obstetrics and gynecology. Methods. Six hundred seventy-six of the pregnancies had the midtrimester ultrasound examinations performed outside the county. Three hundred seventeen of the women had midtrimester ultrasound examinations performed in the county, but delivered outside the county. A total of 18 181 pregnancies were eligible for the study. Results. Altogether there were 267 fetuses and newborns with anomalies, which gives a prevalence of 1.5%. One hundred three of the 267 anomalies were detected at the midtrimester ultrasound examination, yielding a sensitivity of 39.0%. There were 11 false positives and 163 remained undiagnosed (false negatives), which gives a specificity of 99.9% and a positive predictive value of 90.4%. The sensitivity for detecting anomalies ranged from 74.4 to 8.3% according to the organ system of the fetus. Conclusions. Our study shows that midtrimester routine ultrasound examination in district hospitals can achieve a detection rate of congenital anomalies comparable with tertiary centers. One-stage ultrasound examination at midtrimester gives acceptable results concerning congenital anomalies with few false-positive results. Acta Obstet Gynecol Scand 2005.


OBJECTIVE: The purpose of this study was to determine whether a repeated antenatal ultrasound examination improves fetal cardiac visualization for the obese and nonobese population. METHODS: A computerized ultrasound database (October 1999-June 2003) was used to identify singleton pregnancies undergoing repeated prenatal ultrasound examinations because of initial suboptimal ultrasonographic visualization (SUV) of the 4-chamber view, outflow tracts, or both. Women with maternal diabetes, abnormal maternal serum screening results, or known fetal anomalies at the initial examination were excluded. Patients were classified by maternal body mass index (BMI): less than 30 kg/m^2^ (nonobese), 30 to 34.9 kg/m^2^ (class I obesity), 35 to 39.9 kg/m^2^ (class II obesity), and 40 kg/m^2^ or greater (morbid obesity). The association between maternal BMI and SUV of the fetal heart was analyzed. RESULTS: Three hundred seventy-two patients were abstracted from the database. The median gestational age was 19.0 weeks at the initial visit (range, 18.0-21.9 weeks) and 21.4 weeks at the second visit (range, 18.9-23.9 weeks). The median BMI was 32.6 kg/m^2^ (range, 16.4-58.7 kg/m^2^). Sixty-three percent of patients were obese (BMI >or=30). Cardiac anatomy continued to have SUV in 11% of the women. The
rate of SUV was associated with the obesity class (1.5% for nonobese, 12% for obesity I, 17% for obesity II, and 20% for morbid obesity; P < .0001). A cardiac anomaly was found in 1 of 372 repeated examinations (arteriovenous canal defect) for a patient with BMI of 24.8 kg/m2. CONCLUSIONS: Repeated ultrasound examination for SUV of the fetal heart at a later gestational age dramatically reduces SUV. However, obese patients continue to have much higher rates of persistent SUV.

Detection (11)


   Objectives. To investigate the detection rate of major fetal heart defects in a low-risk population implementing routine use of color Doppler. Material and Methods. In a prospective observational study, all women undergoing fetal heart scanning (including 6781 routine examinations in the second trimester) during a three-year period were included. First a gray-scale scanning was performed including assessment of the four-chamber view and the great vessels. Thereafter three cross-sectional planes through the fetal thorax were assessed with color Doppler. Results. Thirty-nine fetuses had major heart defects, and 26 (67%) were prenatally detected. In 9/26 (35%) of cases the main ultrasound finding was related to the use of color Doppler. The survival rate of live born children was 91%. Conclusions. Routine use of color Doppler in fetal heart scanning in a low-risk population may be helpful in the detection of major heart defects; however, still severe malformations were missed prenatally.


   Objective: To observe the clinical value of combined ultrasound screening for abnormal fetuses during the first and second trimesters. Methods: A total of 2844 pregnant women (3135 fetuses) underwent ultrasound scan between 11-13<sup>+</sup> weeks of gestation. The thickness of fetal nuchal translucency (NT) was measured, and fetal major anatomy structures were observed specially on cross-sectional view of the fetal brain. In 18-24 weeks of gestation, 2865 fetuses in the same group accepted multi-section ultrasound screening test for fetal systematic examination, and fetal growth situation was followed in 32 weeks of gestation. All the neonates were followed up. Results: In the first trimester, there were 153 abnormal fetuses among 3135 fetuses (4.88%), including NT >=3.0 mm in 20 fetuses, fetal hydrops in 25, dead fetus in uterus in 75, acrania in 9, umbilical hernia in 2, megabladder in 2, limb abnormalities in 2, multiple malformations in 5, conjoined twins in 1 and single umbilical artery in 12 fetuses. In the second trimester of the same group, there were 66 abnormal fetuses identified among 2865 fetuses (2.30%), including 6 fetal edema, 13 general nervous system abnormalities, 3 facial deformity, 11 cardiac defect, 1 diaphragmatic hernia, 2 umbilical hernia, 1 limb abnormalities, 9 urinary system abnormalities, 1 multiple malformations, 5 fetal growth restriction and 14 abnormal umbilical cord. Conclusion: Ultrasound screening in the first trimester can effectively detect the abnormal fetuses in some degree, but the second trimester screening is still necessary.


   INTRODUCTION: Second-trimester fetal screening for congenital heart defects (CHD) included in routine obstetric care provides relevant information for decision making. The aim of this study was to describe the clinical practice of prenatal detection of CHD in terms of the process and results. METHODS: The characteristics and results of ultrasound screening for major CHD were documented using data provided by hospitals for a national survey in Spain over the period of 2004-2006. Sixty-seven percent of eligible centers (56/83), covering 36% of total births nationwide, responded to the survey; 33 of these returned complete data regarding the screening results. RESULTS: The number of major CHD occurring in the centers which provided data with results of screening was 1,060. The overall prenatal detection rate of major CHD was 65.7% (95% CI 57.8-74.7), but the detection rate in the routine second-trimester scan was 52.6% (95% CI 45.6-60.8). In 61% of these cases the parents chose to terminate the pregnancy. Two independent predictors of increased detection by center were identified: first, the uniformity and systematic character of the examination of the heart showing at least the 4-chamber view and outflow tracts (prevalence ratio 1.3, 95% CI 1.0-1.8) and second, the local availability of specialists in fetal echocardiography (prevalence ratio 1.4, 95% CI 1.1-1.9). CONCLUSIONS: The detection of major CHD in the first half of pregnancy has an important impact on parental decision making. The prenatal screening program for CHD should be globally strengthened in terms of qualifications and methodological approaches. To improve its performance locally, close collaboration with fetal heart specialists should be promoted. Copyright Copyright 2010 S. Karger AG, Basel.

OBJECTIVE: Most fetuses with congenital heart disease (CHD) occur in women that are not at increased risk, and since it is impractical to perform detailed fetal echocardiography on everyone, detection of CHD relies mainly on routine second trimester fetal anatomic surveys. We therefore attempted to improve the detection rate of CHD at the time of routine second trimester obstetrical sonography in low-risk patients. METHODS: This was a retrospective review of an 18-month period in which color Doppler was added to the standard grey scale evaluation of the fetal heart at the time of our routine second trimester anatomic surveys that we performed on fetuses at low risk for CHD. Cases in which CHD was suspected were reviewed with special attention to those in which abnormalities on color Doppler were the primary finding. RESULTS: CHD was suspected in 17 of 1,766 (1%) routine fetal anatomic surveys that we performed between 16 and 22 weeks. There were 13 cases with findings on grey scale, and 4 cases (24%) that relied on findings with color Doppler, as the grey scale evaluation was normal or near normal. Of these 4 cases, 3 had critical pulmonic stenosis requiring balloon valvuloplasty shortly after birth; the fourth case had a mildly dysplastic pulmonic valve that did not require intervention in the immediate newborn period. CONCLUSIONS: The addition of color Doppler evaluation of the fetal heart to routine obstetrical sonographic structural surveys in low-risk patients aids in the detection of pulmonic stenosis. Copyright Copyright 2010 S. Karger AG, Basel.


OBJECTIVE: To assess and compare the sensitivity for detecting fetal anomalies and chromosomal aberrations by routine ultrasound examination performed in the second trimester with results from an examination performed at 11-14 weeks gestation. DESIGN: Observational study. SETTING: Five centers in the southeast region of Sweden. POPULATION: A total of 21,189 unselected pregnant women. METHODS: The scan was performed at one center in the first trimester and at the remaining four centers in the second trimester. Outcome measures resulting from first trimester scanning were compared with those from the second trimester scanning. MAIN OUTCOME MEASURES: Detection rates of fetal structural anomalies and chromosomal aberrations. Results. At the first trimester scan 13% of all anomalies were detected, and at the second trimester scan 29% were detected. Lethal anomalies were detected at a high level at both times: 88% in the first, 92% in the second. The percentage of chromosomal aberrations discovered at the early scan was 71%, in the later 42%. The percentage of heart malformations detected was surprisingly low. CONCLUSION: The results showed the advantages of the later scan in discovering anomalies of the heart, urinary tract and CNS, and of the early scan in discovering chromosomal aberrations. Lethal malformations were detected at a high level in both groups, but detection of heart malformations needs improvement.


The purpose of this study was to determine if acquiring real-time sweeps of the fetal heart would be a more effective method of identifying normal cardiac structures compared with using static images during routine second-trimester obstetric sonograms. Subjects were scanned using three different techniques. The static image acquisition (protocol A) included three images of the fetal heart. Protocol B used two gray-scale sweeps through the fetal heart. Protocol C acquired three color loops of the fetal heart. The sweeps demonstrated a complete normal cardiac assessment in 71% of studies, compared with the static image and color Doppler techniques that completed a normal cardiac assessment in only 39% of studies, respectively. The real-time technique detected four chambers, the left ventricular outflow tract (LVOT), the right ventricular outflow tract (RVOT), the LVOT/RVOT crossover, and size and axis of the heart with a greater frequency than the static images and color loops in all cases. In addition, the real-time technique was able to demonstrate the pulmonary veins in 56% of cases compared with 3.6% for static images. The color Doppler acquisition demonstrated blood flow through the atrial-ventricular and semilunar valves in 86% of cases.


OBJECTIVE: To assess prenatal heart disease screening program by ultrasound. METHODS: A total of 11,544 second-trimester screening scans were performed before 24 weeks’ gestation on 11,410 women between

OBJECTIVE: To evaluate the outcome of screening for structural malformations in twins and the outcome of screening for twin-twin transfusion syndrome (TTTS) among monochorionic twins through a number of ultrasound scans from 12 weeks' gestation. METHODS: Enrolled into this prospective multicenter observational study were women with twin pregnancies diagnosed before 14 + 6 gestational weeks. The monochorionic pregnancies were scanned every second week until 23 weeks in order to rule out early TTTS. All pregnancies had an anomaly scan in week 19 and fetal echocardiography in week 21 that was performed by specialists in fetal echocardiography. Zygosity was determined by DNA analysis in all twin pairs with the same sex. RESULTS: Among the 495 pregnancies the prenatal detection rate for severe structural abnormalities including chromosomal aneuploidies was 83% by the combination of a first-trimester nuchal translucency scan and the anomaly scan in week 19. The incidence of severe structural abnormalities was 2.6% and two-thirds of these anomalies were cardiac. There was no significant difference between the incidence in monozygotic and dizygotic twins, nor between twins conceived naturally or those conceived by assisted reproduction. The incidence of TTTS was 23% from 12 weeks until delivery, and all those monochorionic twin pregnancies that miscarried had signs of TTTS. CONCLUSION: Twin pregnancies have an increased risk of congenital malformations and one out of four monochorionic pregnancies develops TTTS. Ultrasound screening to assess chorionicity and follow-up of monochorionic pregnancies to detect signs of TTTS, as well as malformation screening, are therefore essential in the antenatal care of twin pregnancies. Copyright (c) 2007 ISUOG.


OBJECTIVE: To compare the rate of prenatal diagnosis of heart malformations between two policies of screening for heart malformations. DESIGN: Randomised controlled trial. SETTING: Six university hospitals, two district general hospitals. SAMPLE: A total of 39 572 unselected pregnancies randomised to either policy. METHODS: The 12-week policy implied one routine scan at 12 weeks including measurement of nuchal translucency (NT), and the 18-week policy implied one routine scan at 18 weeks. Fetal anatomy was scrutinised using the same check-list in both groups, and in both groups, indications for fetal echocardiography were ultrasound findings of any fetal anomaly, including abnormal four-chamber view, or other risk factors for heart malformation. In the 12-week scan group, NT >or=3.5 mm was also an indication for fetal echocardiography. MAIN OUTCOME MEASURE: Prenatal diagnosis of major congenital heart malformation. RESULTS: In the 12-week scan group, 7 (11%) of 61 major heart malformations were prenatally diagnosed versus 9 (15%) of 60 in the 18-week scan group (P= 0.60). In four (6.6%) women in the 12-week scan group, the routine scan was the starting point for investigations resulting in a prenatal diagnosis versus in 9 (15%) women in the 18-week scan group (P=0.15). The diagnosis was made <or=22 weeks in 5% (3/61) of the cases in the 12-week scan group versus in 15% (9/60) in the 18-week scan group (P=0.08). CONCLUSIONS: The prenatal detection rate of major heart malformations was low with both policies. The 18-week scan policy seemed to be superior to the 12-week scan policy, although the differences in prenatal detection rates were not statistically significant.

OBJECTIVE: The purpose of this study was to determine whether Doppler velocimetry of the ductus venosus (DV) predicts adverse perinatal outcome in congenital heart disease (CHD). METHODS: We conducted a retrospective cohort study of all pregnant women undergoing fetal echocardiography for CHD in a single perinatal center during a 2-year period. We compared outcomes for fetuses having a diagnosis of CHD in the second trimester and abnormal DV Doppler velocimetric findings with those having CHD and normal DV Doppler findings. Karyotype, gestational age at delivery, fetal loss rate, and rate of termination were assessed. The referral value for an abnormal DV pulsatility index was above the 95th percentile for gestational age. Statistical analysis included the t test, Fisher exact test, and chi(2) test. RESULTS: The incidence of CHD in our population was 7%. There were 98 patients with CHD; of those, 31 had DV measurement. A total of 9 patients had an abnormal DV. Three of this group (33%) had intrauterine fetal death or perinatal death. In patients with CHD and normal DV measurements, 83% had living children versus 33% in the group with an abnormal DV (P < .05). There was no statistically significant difference in the rate of aneuploidy between the normal DV (15%) and abnormal DV (20%) groups (P = .65). The mean gestational age at delivery was similar between the normal (37.63 weeks) and abnormal (38.33 weeks) DV groups (P = .71). There was no difference in the rate of pregnancy termination. CONCLUSIONS: Abnormal second-trimester DV measurements are predictive of adverse perinatal outcome in patients with CHD, independent of karyotype or gestational age at delivery. This information may have a role in the counseling of parents with CHD.


OBJECTIVE: To evaluate fetal heart anatomy in the late first and in the early second trimester. MATERIALS AND METHODS: The study included 75 fetuses between 11 and 19 weeks' gestation. Exams were performed using transabdominal or transvaginal probes. RESULTS: The proportion of cases successfully visualized to all cases in particular week of pregnancy was following: 11 week--0/1 (0%), 12--1/1 (100%), 13 week--1/1 (100%), 14--18/23 (79%), 15--6/9 (67%), 16--23/27 (86%), 17--4/5 (80%), 18--6/6 (100%), 19--2/2 (100%). In 11 weeks' gestation two ventricles and two atrias could be imaged. Anatomy was seen from 12 weeks' gestation with transvaginal and from 13 weeks' gestation with transabdominal transducers. Two tricuspid regurgitations were detected. The interventricular septum was the most difficult structure to visualize in all cases. The grey scale alone was not sufficient for accurate examination of the heart and it was necessary to use colour Doppler to confirm normal forward flow to both ventricles and to identify outflow tracts. CONCLUSIONS: 1. Successful visualization of the heart in early pregnancy is possible from 12 weeks' gestation with transvaginal and from 13 weeks' gestation with transabdominal probe. 2. Fetal echocardiography between first and second trimester should lead to better understanding of fetal hemodynamics in normal and abnormal fetuses and help to introduce new therapeutic treatment in some cases.

Reviews (8)


Fetal heart is one of the most difficult organs to examine during second trimester fetal ultrasound. Conventional two-dimensional screening ultrasound using a four-chamber view alone allows detection of 5%-60% of congenital cardiac defects. If ventricular outflows are included in routine screening protocol, the detection rate of major cardiac disease is significantly increased to 85,5%-90%. Therefore ISUOG (International Society of Ultrasound Obstetrics and Gynecology) state that the left and right ventricular outflows should be included in the "Optimal Examination" of the fetus. Another important issue is that fetal echocardiography when combined with first and/or second trimester screening for trisomy 21 may increase the detection rate to almost 94%. This may be advantageous for patients who desire a highest sensitivity before considering invasive testing.


One of the major roles of ultrasound in pregnancy is the detection of structural abnormalities. Ultrasound screening has become an accepted part of antenatal practice. However there is a wide variety in the practice between different centers. The majority of fetal anomalies are diagnosed by ultrasound in the second trimester. However, a number of abnormalities are amenable to diagnosis as early as 11-14 weeks gestation. National guidelines have been adopted in an attempt to standardize the practice in the UK. Prenatal detection rate is higher in countries with a national screening program. Prenatal ultrasound is a screening test and will have false
negatives and positives. Structural abnormalities can occur as isolated events, as part of a genetic syndrome or as a result of a chromosomal abnormality. When an abnormality is detected prenatally, a multidisciplinary approach is necessary to optimize the outcome. It is important to provide appropriate information to the prospective parents but remain non-judgmental with their decision. 2009 Elsevier Ltd. All rights reserved.

The authors have evaluated data from the international literature regarding the frequency of antenatally screened out cardiac congenital anomalies. Literary data confirm the fact that the screening of congenital cardiac anomalies is highly influenced by the fact whether the country in question has its own regular routine antenatal screening programme or not and whether this screening is applicable to only selected cases or low-risk cases, too. It is an accepted fact, that with a routine second trimester fetal 4 chamber cardiac screening 50% chances are there to detect cardiac and great vessel anomalies. Efficacy of the screening could further be increased by a combined examination of the four heart chambers as well as their outflow tracts, including fetal echocardiography in selected high-risk cases.

Echocardiography allows the prenatal diagnosis of most congenital cardiac malformations. Although the indications for this specialized ultrasound technique are multiple, suspicion of congenital heart disease at the second trimester ultrasonographic screening has become the most frequent indication. Specially trained pediatric cardiologists can confirm the diagnosis or rule it out. If the diagnosis is confirmed, then a fetal karyotype should be proposed in most cases and thorough examination of extracardiac organs, should be performed. If the prognosis is poor, French law authorizes termination of pregnancy. If the pregnancy goes on, a multidisciplinary team should organize the perinatal care according to expected neonatal difficulties.

In many countries, ultrasound examination is used in the second trimester to look for congenital malformations as part of routine prenatal care. While tertiary centres scanning high-risk pregnancies have reported a high degree of accuracy in the detection of congenital heart disease, many studies have shown that cardiac abnormalities are commonly overlooked during routine obstetric evaluation and there still remains a huge variation between centres. The majority of babies with congenital heart disease are born to mothers with no identifiable high-risk factors and so will not be detected unless there is widespread screening of the low-risk population. It is feasible to achieve widespread screening for fetal congenital heart disease in low-risk groups, but this does need commitment and effort from those performing the scans and those teaching them how to examine the heart. Staff performing routine obstetric ultrasound scans should learn a simple technique for examining the fetal heart and to use this in all patients. Links to a tertiary centre can provide support for checking scans of concern as well as for providing training and for obtaining feedback. In addition, an audit system needs to be established in each centre to trace false-positive and false-negative cases as well as to confirm true positives and true negatives. 2004 John Wiley & Sons, Ltd. [References: 42]

PURPOSE OF REVIEW: The purpose of this review is to describe several of the most relevant and exciting recent advances in the field of fetal cardiology. RECENT FINDINGS: First, the prenatal detection of congenital heart disease has improved, and continues to improve, with the increasingly widespread incorporation of the four-chamber view and outflow tracts into the routine screening fetal ultrasound evaluation. Second, increasingly sophisticated computer processing systems and improvements in imaging technology have enabled the development of automated three-dimensional ultrasound imaging systems that promise to revolutionize both the prenatal detection and diagnosis of congenital heart disease. Conventional two-dimensional imaging approaches may soon become obsolete. Third, there has been an increasing ability to intervene successfully prenatally not only for fetal arrhythmias and heart failure, but also for some forms of structural heart disease. In some cases of left or right ventricular outflow tract obstruction, early intervention during the second trimester may prevent the development of ventricular hypoplasia. Finally, several recent studies suggest that prenatal diagnosis may improve neonatal outcome for fetuses with congenital heart disease. The growing ability to intervene prenatally has the potential to improve neonatal outcome still further. SUMMARY: These critical and exciting developments in fetal
cardiology promise to increase fetal echocardiography's clinical impact dramatically during the years to come.

[References: 64]


Referral to echocardiography (5)


OBJECTIVE: Fetal echocardiography (FE) is considered for fetal, maternal or hereditary reasons in pregnant with suspect of intrauterine heart disease (IUHD). However, in few studies it was reported that most of the fetuses with IUHD are in the low-risk group (suspicion of IUHD during 2nd trimester ultrasound, lack of good vision of the heart, self-referral). Our aim is to examine retrospectively the reasons for referral of pregnant, the results of FE, distribution of pregnant having fetuses with IUHD according to low- and high-risk factors and to evaluate reliability of FE. METHODS: Our study group consisted of 1395 fetuses and 1370 pregnant underwent FE between 1999 and 2006. These cases included self-referred women and the pregnants having previous child or family history of cardiac anomaly or referred by obstetricians. The prevalence of IUHDs in low- and high-risk pregnancies was compared by Chi-Square test. RESULTS: The low risk group included 453 patients and the remaining 917 women were in the high-risk group. Intrauterine heart diseases were detected in 152 (10.9%) of 1395 fetuses. The prevalence of IUHDs was 19% in the low-risk group and 7% in the high-risk group. Of the 152 fetuses 56.6% were in the low-risk group and 43.4% were in the high-risk group. The sensitivity of FE for diagnose of IUHDs was 97%, the specificity was 100%. CONCLUSION: Fetal echocardiography is highly reliable method for diagnosing of IUHDs. The most IUHDs occur in the low-risk group.


OBJECTIVE: To investigate the application of "Guidelines for performing fetal cardiac scan", issued by the International Society of Ultrasound in Obstetrics and Gynecology in 2006, in prenatal screening of fetal congenital heart disease (CHD). METHOD: Totally, 5000 singleton pregnancies presented at the Maternal-Fetal Medical Center of the Affiliated Drum Tower Hospital of Nanjing University Medical School from September 2006 to July 2007, for prenatal screening were included in this study, with the median maternal age of 28 (range, 18-approximately 48) and the median gestation of 27 (range, 18-approximately 40) weeks. Ultrasound screenings were performed on each fetal heart according to "Guidelines for performing fetal cardiac scan" via the four-chamber and outflow tracts & three-vessel views and fetal echocardiographies were further conducted for suspected cases. Once congenital heart disease was confirmed, amniocentesis or cordocentesis was suggested for fetal karyotyping for ongoing pregnancies and autopsy was performed when the pregnancy was terminated after formal consent. Born babies were followed up at 2 approximately 6 months of age using echocardiography. RESULT: The four-chamber views were successfully obtained in 97.64% (4882/5000) of all the pregnancies, among which the left ventricular and right ventricular outflow tracts and three-vessel views were obtained in 87.69% (4281/4882), 82.51% (4028/4882) and 96.29% (4701/4882), respectively. Higher successful rate was found in the second trimester than the third trimester in obtaining the standard views (P < 0.05). Finally, 73 (1.50%) among the 4882 cases were diagnosed as CHD. Fifty of them were diagnosed prenatally (24 cases in the second trimester and 26 cases in the third trimester) and 23 were missed and 1 misdiagnosed by prenatal ultrasound. Eighteen cases were found with extracardiac malformations. Autopsy was performed in 19 CHD which diagnosed prenatally, and all autopsy reports were consistent with ultrasound foundings. Twelve babies received postnatal echocardiography among which 11 were unanimous, and 1 baby diagnosed as tricuspid insufficiency prenatally was confirmed normal after birth. Abnormal karyotype was found in 7 out of the 23 who had karyotyping performed. Altogether, 28 cases were diagnosed by four chamber view only and 50 cases by combining other views, giving the sensitivity, specificity, false negative rate and false positive rate of 69% (50/73), 99.98% (4808/4809), 0.48% (23/4831) and 2% (1/51), respectively. CONCLUSION: The "Guidelines for performing fetal cardiac scan" is practical and easy to abide by. The optimal time for fetal cardiac examination is at 18 approximately 27 weeks of gestation. Four-chamber view together with the outflow tracts and three-vessel views
examination can detect 69% of CHD in utero.

 OBJECTIVES: This study was conducted to evaluate the referral indications for fetal echocardiography (FE) in a tertiary center and to determine which indications were significantly associated with prenatal detection of congenital heart disease (CHD). METHODS: The medical records of 1425 consecutive women who underwent second- and third-trimester FE at the Ultrasound Center of Beijing Obstetrics and Gynecology Hospital from March 2003 to December 2007 were reviewed. Referral indications, FE diagnoses, and pregnancy outcomes were collected. Univariate and multivariate logistic regression analyses were performed to identify those referral indications associated with prenatal detection of CHD. RESULTS: In 126 patients (8.8%), CHD was detected prenatally and confirmed postnatally. Logistic regression analysis showed that abnormal cardiac views and extracardiac malformation findings (especially a single umbilical artery) on second-trimester ultrasound screening were found to have significantly more CHD (P < .001). The adjusted odds ratios were 15.2 (95% confidence interval, 9.85-23.45) and 6.78 (95% confidence interval, 2.38-19.27), respectively. CONCLUSIONS: Abnormal cardiac views and extracardiac malformation findings on second-trimester ultrasound screening were significantly associated with prenatal detection of CHD.

 OBJECTIVE: Fetal echocardiography accurately detects congenital cardiac anomalies, but it is costly, time-consuming, and requires highly-skilled operators. Our aim was to define those patients for whom fetal echocardiography is justified. METHODS: The files of 1696 consecutive patients who underwent second- to third-trimester fetal echocardiography at our tertiary center between 1997 and 1999 were reviewed for reason for referral, echocardiography diagnosis, and pregnancy outcome. RESULTS: The patients were categorized by reason for referral into high- and low-risk groups. The high-risk group included 662 patients (39%) with fetal risk factors, 178 (10.5%) with maternal risk factors and 279 (16.5%) with poor obstetric history. The remaining 577 women (34%) were considered low-risk population. These included 282 self-referred women (due to maternal anxiety) who served as control group, 78 women who were referred because of a suspected cardiac malformation on routine second-trimester ultrasound, and 213 women who were referred because of failure to view the heart on second-trimester ultrasound. In 46 women, cardiac anomalies (2.7%) were detected prenatally and confirmed postnatally; most of them (41/46, 89%) were in the low-risk population. Abnormal cardiac findings on second-trimester ultrasound and a diagnosis of a single umbilical artery made the most significant contribution to the detection of cardiac abnormalities (p < 0.001 and p = 0.02, respectively). CONCLUSIONS: Most fetal cardiac malformations occur in the low-risk population. Abnormal view of the fetal heart on routine second-trimester screening is highly predictive of congenital cardiac anomalies.

 PURPOSE: The present study was conducted to evaluate the indications for fetal echocardiography in a tertiary-care obstetric sonography practice and to determine the incidences of confirmed congenital heart disease for each primary indication. METHODS: A retrospective analysis of all pregnant women referred to a pediatric cardiology unit for fetal echocardiography by the tertiary-care sonography unit over a 2-year period was performed. The primary indications for referral for fetal echocardiography were obtained from the sonographers’ reports. Outcome data were extracted from the fetal echocardiograms, postnatal echocardiograms or pathology and autopsy reports, and patient medical records. RESULTS: Of 6,002 pregnant women who had undergone prenatal sonographic examination during the study period, 275 (4.6%) had been subsequently referred for fetal echocardiography. The most common primary indication for referral had been abnormal cardiac findings on the prenatal sonographic examination, which had been present in 64 (23.3%) of the 275 cases. In 44 (69%) of those 64 cases, congenital heart disease had been confirmed. Among the 211 patients who had had normal cardiac findings on prenatal sonography but had been referred for fetal echocardiography owing to other primary indications, congenital heart disease had been confirmed in only 7 cases (3.3%). CONCLUSIONS: An abnormal cardiac finding during prenatal sonographic examination is a common primary indication for fetal echocardiography and is more useful for identifying congenital heart disease than are other risk factors. Careful routine cardiac screening during routine prenatal sonographic examination may facilitate further investigation and treatment. Copyright 2004 Wiley Periodicals, Inc.