



ACCURACY OF PRENATAL DETECTION OF CONGENITAL HEART ANOMALIES USING ULTRASONOGRAPHY: A SYSTEMATIC REVIEW

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ABSTRACT

Ultrasound examinations in the second trimester for detection of congenital malformations are now part of pregnancy care in most developed countries. Major heart defects can be diagnosed before birth by sonographic assessment of the four-chamber view. This review is aimed to evaluate the evidence published on the accuracy of prenatal detection of congenital cardiac anomalies using ultrasonography. A web-based search was conducted in MEDLINE database and eligible studies were identified and then screened against inclusion criteria such as detection of congenital heart anomalies and reporting of ultrasonography accuracy. The full texts were retrieved for eligible studies and secondary in-depth screening were conducted for the study against inclusion criteria. Data were extracted from our studies regarding study's characteristics, type of heart anomalies and level of accuracy. The data were synthesized and discussed with qualitative approach. The electronic search resulted in 145 eligible studies. After screening of titles and abstracts of these articles, irrelevant and duplicated studies were excluded and finally full-texts of 13 articles were retrieved. Overall sample size was ranged between 31 to 4172 with gestational age ranged between 11 weeks to 41 weeks. Overall accuracy of ultrasonography in the prenatal detection of heart anomalies was ranged between 81% to 98.4%. Ultrasonography has fair to high accuracy in prenatal detection of heart anomalies. The variation depends on factors such as technology, experience of the operator, and type of the anomaly. The findings of the included studies showed an acceptable accuracy of ultrasonography in detection of heart anomalies either in high or low risk groups.

KEYWORDS: *Screening, Prenatal, Ultrasound, Diagnostic, Cardiac anomalies*



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INTRODUCTION

Congenital heart diseases (CHD), with a prevalence of 8/1000 live births are considered the commonest severe congenital anomalies.¹ Heart anomalies are among major abnormalities that are often missed in prenatal sonographic examinations,² and the congenital undetected cardiac disease increases the hazard of early neonatal death.³ As technology and skill improve, more fetal malformations are being recognized by ultrasound.⁴ Ultrasound examinations in the second trimester for detection of congenital malformations are now part of pregnancy care in most European countries.⁴ Major heart defects can be diagnosed before birth by sonographic assessment of the four-chamber view.⁵ But general screening of low-risk populations shows a detection rate as low as 5–6%.⁶ Others have reported prenatal detection rates of between 14 and 45% by general screening.³ Routine investigation or exclusion of congenital heart deformity through transabdominal ultrasound is possible as early as the 18th week of pregnancy.⁷ Heart anomalies commonly affect children born for families without history of heart disease. Thus, the routine prenatal screening can improve the identification of such serious life-threatening anomalies. The suspicion of CHD in the fetus should be raised at the screening procedure and a more detailed diagnosis can be performed later after referral for fetal echocardiography.⁴ However, fetal echocardiography is a procedure which takes a long time and requires many two-dimensional cross-sectional views of the heart is considered as time-consuming procedure which requires many views of the heart⁸ in addition to Doppler.⁹ There are specific factors which increase a mother's risk of carrying a baby with congenital heart disease have been identified: family history, coexisting mother's disease (eg, diabetes mellitus) maternal age, and infection of rubella. In those cases, complete fetal echocardiography is typically performed as part of a sonographic examination between 18th and 22nd (or 24) weeks' gestation.⁸ In addition, the risk of a fetal cardiac anomaly is significantly higher when a routine antenatal ultrasound examination shows abnormal four-room or some abnormal cardiac abnormalities (the organ abnormality is significantly higher when a routine prenatal ultrasound examination shows an abnormal view or certain abnormalities outside the heart (organ malformations, intrauterine growth retardation, amniotic fluid excess or deficiency, fetal arrhythmias). Thus, fetal echocardiographic exam is also indicated.¹⁰ Echocardiography for the fetus is not regarded as a part of the routine

screening program before pregnancy, but is reserved for cases at high risk of congenital cardiac disease, and it needs highly experienced skilled investigators in fetal cardiology.³ This review is aimed to evaluate the evidence published on the accuracy of prenatal detection of congenital cardiac anomalies using ultrasonography.

METHODS

A web-based search was conducted in MEDLINE using search terms such as (prenatal OR fetal OR embryonic) AND (ultrasound OR sonography OR ultrasonography) AND (heart OR cardiac) AND (accuracy). The filters applied for the search was date from March 2008-March 2018, human studies, and English language. A relevant study was an original study that assessed the accuracy of ultrasonography in prenatal detection of congenital heart anomalies. The inclusion criteria were an observational pro- or retrospective study, study group either routine or high-risk group of mothers at any gestational age, any type of congenital heart anomalies. The intervention studied should be the diagnostic ultrasonography with transabdominal or transvaginal techniques. The outcome assessed was the accuracy represented in percentage of valid prenatal diagnosis in comparison to postnatal clinical findings. Data were collected for characteristics of included study such as study design, sample size, mean age of mothers, mean gestational age, indications of ultrasonography screening, and type of ultrasonography. Moreover, data were extracted for assessed outcomes included types of anomalies detected and accuracy of detection in reference to postpartum findings. The extracted data were discussed in a qualitative approach and presented in summary of finding Table.

RESULTS

The electronic search resulted in 145 relevant studies. After screening of titles and abstracts of these articles, irrelevant and duplicated studies were excluded and finally eligible article were 13 articles for which full-texts were retrieved. Furthermore, after reading of full texts, six studies were excluded, three of them were because of inconsistent outcome, and other articles because they were reviews. Thus, seven articles were included in this review. Overall sample size was ranged between 31¹¹ to 4172¹². With gestational age ranged between 11 weeks¹³ to 41 weeks¹⁴. Mothers risk factors was reported in two studies, in

the first study women chose to have CVS (chorionic villus sampling) after risk assessment by a combination of maternal age, fetal nuchal thickness, assessment of the nasal bone, blood flow in the ductus venosus or flow across the tricuspid valve, and maternal serum-free β -human chorionic gonadotropin and pregnancy-associated plasma protein-A¹³. While in the second study all women were smokers¹² as demonstrated in Table 1. The ultrasound was routinely indicated in only one study¹⁵, while it was done for high risk group in four studies.¹¹⁻¹⁴ Indication for ultrasound screening was not reported in two studies. Trans abdominal technique was used in six of included studies^{11-14, 16-17}, while trans vaginal ultrasound was used in only one study¹⁵. Types of anomalies detected were varying among included studies. Aneuploidy was detected by Karadzov et al., 2013. Other anomalies detected include septal shunts or transvalvular regurgitation/aliasing double-crossing of the great arteries¹⁵, conotruncal anomalies, right heart anomalies, left heart anomalies, complex congenital heart defect and other anomalies.¹⁴ Also hypoplastic right ventricle VSD, pulmonary atresia, overriding aorta, truncus arteriosus HLHS, hypoplastic left heart syndrome were detected by Zhu et al. 2009¹⁷. Overall accuracy was ranged between 81%¹¹ to 98.4%¹⁵. Detection rate also was calculated in one of the included studies, it was ranged between 82% to 88%¹². The highest accuracy was detected in a study done by Bennasar et al. it was a prospective study includes 69 pregnant women with gestational age ranged between 11 to 15 weeks, without any type of risk factors for CHD recruited from pregnant women. The scan was routine with transvaginal approach, color Doppler also was used. Anomalies detected include septal shunts or transvalvular

regurgitation/aliasing and double-crossing of the great arteries with accuracy equals 98.4%¹⁵. While the lowest accuracy was seen in a retrospective study conducted by Gómez et al 2016. The study was done on 31 fetuses, with gestational age between 11 and 14 weeks. Ultrasound was done transabdominally, this study was done to distinction between truncus arteriosus communis (CAT) and pulmonary atresia with ventricular septal defect (PA-VSD) and to describe the association with extracardiac and chromosomal anomalies with overall diagnostic accuracy ranged between 81% to 93.5%¹¹. The anomalies detected was varying among the studies. Two of included studies measure the accuracy for each anomaly in addition to overall accuracy. The first study was a retrospective study done by Zhu et al., on 113 pregnancies, with mean gestational age 26.8 weeks. Trans abdominal echocardiography with Doppler ultrasound was used. the accuracy of prenatal diagnosis was 86%. And it was varying among anomalies, which include conotruncal malformations, septal defects, valve abnormalities, and univentricular hearts with accuracy 77%, 96%, 90% and 83% respectively¹⁷. In the second study the overall accuracy was 82.1%, while the accuracy for each anomaly was 93.4% for septal defect, valvular anomaly and biventricular heart (71%) venous return anomaly (69.2%), aortic arch anomaly (75%), conotruncal anomaly (87.2%), hypoplastic right heart syndrome (88.2%), hypoplastic left heart syndrome (91.5%), other univentricular heart defect (90.8), complex defects with atrial isomerism (75%), miscellaneous (86%). This study was a multicenter cohort study done on 708 pregnant women, with mean gestational age 23 weeks, transabdominal echocardiography technique was used.¹⁶

Table 1
Summary of the outcomes reported by the included studies

Study	Study design	Sample size	Gestational age (mean, range)	Mothers risk factors	Type of ultrasound technique	Indication for ultrasound screening (routine or high-risk group)	Types of anomalies detected	Accuracy in detection in reference to postpartum findings
(Bennasar et al., 2009) ¹⁵	A prospective study	69 pregnant women	From 11 to 15 weeks	No risk factors reported	Transvaginal approach + color Doppler US	Routine screening	Septal shunts or transvalvular regurgitation/aliasing	98.4%
(Karadzov-Orlic et al., 2012) ¹²	A prospective study	4172 singleton pregnancies	Mean=12.2 weeks (range 11 to 13 weeks)	Smoker women	Transabdominal ultrasound examination	High-risk group	Aneuploidy	General incidence rate= 0.87% to 0.92. Trisomy 21=88%, Trisomy 18=85% Trisomy 13=82%
(Bennasar et al., 2010) ¹⁴	A prospective study	363 pregnant women	14 - 41 weeks of gestation	Non-reported	Ultrasound examinations with Voluson 730 Expert machine (GE Medical Systems, Milwaukee, WI, USA) and the 4D-STIC volumes were acquired transabdominally with a 4–8-MHz transducer	High risk group	Septal defects, conotruncal anomalies, right heart anomalies, left heart anomalies, complex congenital heart defect	The overall accuracy was 91.6%
(Persico et al., 2011) ¹³	A prospective study	886	11–13 weeks	Maternal age, blood flow in the ductus venosus or flow across the tricuspid valve, maternal serum-free β -human chorionic gonadotropin, pregnancy-associated plasma protein-A	Transabdominally using a 9-MHz linear transducer	High-risk group	Septal defects, conotruncal anomalies, right heart anomalies, left heart anomalies, complex congenital heart defect	93.1%

(Gómez et al., 2016) ¹¹	A retrospective study	31 fetuses	11-14 weeks	Non-reported	Transabdominal US	High-risk group	Distinction between truncus arteriosus communis (CAT) and pulmonary atresia with ventricular septal defect (PA-VSD)	The overall diagnostic accuracy was 81% to 93.5%
(Zhu et al., 2009) ¹⁷	A retrospective study	113 pregnancies	Mean= 26.8 weeks	Non-reported	Transabdominal echocardiography + Doppler US	Non-reported	Hypoplastic right ventricle VSD, pulmonary atresia, overriding aorta, AV septal defect, coarctation of aorta, truncus arteriosus, hypoplastic left heart syndrome	The overall accuracy was 86% Prenatal diagnosis was accurate (77%) with conotruncal malformations, (96%) with septal defects, (90%) with valve abnormalities, (83%) with univentricular hearts.
(van Velzen et al., 2016) ¹⁶	A multicenter cohort study	708	Mean= 23 weeks± 36 days.	Non-reported	Transabdominal echocardiography	Non-reported	Septal defect, valvular anomaly, biventricular heart, venous return anomaly, aortic arch anomaly, conotruncal anomaly, hypoplastic right heart syndrome, hypoplastic left heart syndrome, complex defects with atrial isomerism.	Overall accuracy= 82.1%, accuracy for septal defect =93.4, for valvular anomaly, biventricular heart =71%, for venous return anomaly=69.2, for aortic arch anomaly =75%, conotruncal anomaly = 87.2%, hypoplastic right heart syndrome =88.2%, hypoplastic left heart syndrome =91.5%, complex defects with atrial isomerism=75%

DISCUSSION

Congenital heart diseases (CHD) are the commonest severe congenital abnormalities, which are the leading cause of mortality of infants during their first year of life and of babyhood disability.¹⁸ The incidence of CHD is 1.1% in fetuses¹⁹, while it is 0.2–0.4% in live born²⁰. Thus, effective population-based diagnosis before birth necessitates improved ways for determining the high-risk group for referring to specialists and developed standards of examination in those who undergoing routine screening.¹³ The indication for ultrasound scan was routine in only one study¹⁵, while it was done for high risk group in four studies¹¹⁻¹⁴, echocardiography regards the first modality to evaluate the accuracy of diagnosis in CHD¹⁴. Trans abdominal technique was used in six of included studies^{11-14, 16, 17}, while trans vaginal ultrasound was used in only one study¹⁵. Although in (TVS) transvaginal scanning using high frequency transducers with a small distance between the fetus and the ultrasound source provides a high resolution to fetal organs under examination, this technique depends on the fetal position and has less flexibility in terms of ability to examine various scanning planes.¹³ Types of anomalies detected were varying among included studies, conotruncal anomalies were a type of anomalies which was detected by Zhu et al. 2009¹⁷, Prenatal detection of conotruncal anomalies has greatly improved in the last years²¹. Overall accuracy was ranged between 81%¹¹ to 98.4%¹⁵. Fetal echocardiography has improved significantly in recent years, but its diagnostic accuracy varies in different centers.²² Low level of accuracy can be referred to lack of experience, experience of the sonographer is of the greatest importance to more accurate diagnosis. Experience, as in any field, is obtained by continuous, repeated study in the specific field.²³ In

addition, diagnosis of congenital heart disease before birth is highly accurate when it is done by experienced hands. Some kinds of CHD remain difficult to rule out or diagnose in the fetus. The pitfalls related to some types of CHD should be taken into consideration when making a definitive finding and prognosis. Accuracy information and fetal echocardiography limitations can be used in counseling before and during pregnancy. The study designs included in this review are an observational cohort designs because congenital anomalies are mostly studied by these approaches. The evaluation of the methodological quality of observational study is still a controversial issue. The quality of reporting in the included study was generally good. Despite, in this review, no quality assessment was conducted for the included studies.

CONCLUSION

Ultrasonography has fair to high accuracy in prenatal detection of heart anomalies. The variation depends on factors such as technology, experience of the operator, and type of the anomaly. The findings of all included studies were consistent and revealed an acceptable accuracy of ultrasonography in detection of heart anomalies whatever the risk was among examined group.

AUTHORS CONTRIBUTION STATEMENT

Maimsh and Algorashi planned for this study from the proposal phase to the writing of the manuscript.

CONFLICTS OF INTEREST

Conflict of interest declared none.

REFERENCES

- Hoffman JIE, Christianson R. Congenital heart disease in a cohort of 19,502 births with long-term follow-up. *Am.J.Cardiol.*1978;42(4):641-7. DOI:10.1016/0002-9149(78)90635-5
- Hohlfeld P, Daffos F, Costa J-M, Thulliez P, Forestier F, Vidaud M. Prenatal Diagnosis of Congenital Toxoplasmosis with a Polymerase-Chain-Reaction Test on Amniotic Fluid. *N Engl J Med.* 1994;331(11):695–9. DOI:10.1056/nejm199409153311102
- Stümpflen I, Stümpflen A, Wimmer M, Bernaschek G. Effect of detailed fetal echocardiography as part of routine prenatal ultrasonographic screening on detection of congenital heart disease. *Lancet.* 1996;348(9031):854–7. DOI:10.1016/s0140-6736(96)04069-x
- Garne E, Stoll C, Clementi M. Evaluation of prenatal diagnosis of congenital heart diseases by ultrasound: experience from 20 European registries. *Ultrasound Obstet Gynecol.* 2001;17(5):386–91. DOI:10.1046/j.1469-0705.2001.00385.x

5. Fernandez CO, Ramaciotti C, Martin LB, Twickler DM. The Four-Chamber View and Its Sensitivity in Detecting Congenital Heart Defects. *Cardiology*. 1998;90(3):202–6. DOI:10.1159/000006844
6. Queisser-Luft A, Stopfkuchen H, Stolz G, Schlaefer K, Merz E. Prenatal diagnosis of major malformations: quality control of routine ultrasound examinations based on a five-year study of 20 248 newborn fetuses and infants. *Prenat Diagn*. 1998;18(6):567–76. DOI:10.1002/(sici)1097-0223(199806)18:6%3C567::aid-pd302%3E3.0.co
7. Sahn DJ, Lange LW, Allen HD, Goldberg SJ, Anderson C, Giles H. Quantitative real-time cross-sectional echocardiography in the developing normal human fetus and newborn. *Circulation*. 1980;62(3):588–97. DOI:10.1161/01.cir.62.3.588
8. Fyfe DA. Fetal echocardiography: An atlas edited by Kathryn L. Reed, Caroline F. Anderson, Lewis Shenker Alan R. Liss, Inc., New York (1988) 146 pages, illustrated, ISBN: 0-84514250-X. *Clin Cardiol*. 1988;11(11):804. DOI:10.1002/clc.4960111116
9. Reed KL, Meijboom EJ, Sahn DJ, Scagnelli SA, Valdes-Cruz LM, Shenker L. Cardiac Doppler flow velocities in human fetuses. *Circulation*. 1986;73(1):41–6. DOI:10.1161/01.cir.73.1.41
10. Ferencz C, Rubin JD, McCarter RJ, Boughman JA, Wilson PD, Brenner JI. Cardiac and noncardiac malformations: Observations in a population-based study. *Teratology*. 1987;35(3):367–78. DOI:10.1002/tera.1420350311
11. Gomez O, Soveral I, Bennasar M, Crispi F, Masoller N, Marimon E. Accuracy of Fetal Echocardiography in the Differential Diagnosis between Truncus Arteriosus and Pulmonary Atresia with Ventricular Septal Defect. *Fetal Diagn Ther*. 2015;39(2):90–9. DOI:10.1159/000433430
12. Karadzov-Orlic N, Egic A, Milovanovic Z, Marinkovic M, Damjanovic-Pazin B, Lukic R. Improved diagnostic accuracy by using secondary ultrasound markers in the first-trimester screening for trisomies 21, 18 and 13 and Turner syndrome. *Prenat Diagn*. 2012;32(7):638–43. DOI:10.1002/pd.3873
13. Persico N, Moratalla J, Lombardi CM, Zidere V, Allan L, Nicolaides KH. Fetal echocardiography at 11-13 weeks by transabdominal high-frequency ultrasound. *Ultrasound Obstet Gynecol*. 2011;37(3):296–301. DOI:10.1002/uog.8934
14. Bennasar M, Martínez JM, Gómez O, Bartrons J, Olivella A, Puerto B. Accuracy of four-dimensional spatiotemporal image correlation echocardiography in the prenatal diagnosis of congenital heart defects. *Ultrasound Obstet Gynecol*. 2010;36(4):458–64. DOI:10.1002/uog.7720
15. Bennasar M, Martínez JM, Olivella A, del Río M, Gómez O, Figueras F. Feasibility and accuracy of fetal echocardiography using four-dimensional spatiotemporal image correlation technology before 16 weeks' gestation. *Ultrasound Obstet Gynecol* 2009;33(6):645–51. DOI:10.1002/uog.6374
16. van Velzen CL, Clur SA, Rijlaarsdam MEB, Pajkrt E, Bax CJ, Hruda J. Prenatal diagnosis of congenital heart defects: accuracy and discrepancies in a multicenter cohort. *Ultrasound Obstet Gynecol*. 2016;47(5):616–22. DOI:10.1002/uog.15742
17. Zhu R, Li L, Chen R, Shi L, Niu J WX. Accuracy of prenatal diagnosis of congenital heart defects by fetal echocardiography: a 7-year experience in a Chinese tertiary obstetric center. *Zhonghua xin xue guan bing za zhi*. 2009;37(4):343–6. Available from: <https://europepmc.org/abstract/med/19791471>
18. Abu-Harb M, Hey E, Wren C. Death in infancy from unrecognised congenital heart disease. *Arch Dis Child*. 1994;71(1):3–7. DOI:10.1136/adc.71.1.3
19. Tegnander E, Eik-Nes SH, Johansen OJ, Linker DT. Prenatal detection of heart defects at the routine fetal examination at 18 weeks in a non-selected population. *Ultrasound Obstet Gynecol*. 1995;5(6):372–80. DOI:10.1046/j.1469-0705.1995.05060372.x
20. Ferencz C, Rubin Jd, McCarter Rj, Brenner Ji, Neill Ca, Perry Lw, Et Al. Congenital Heart Disease: Prevalence At Livebirth. *Am J Epidemiol*. 1985;121(1):31–6. DOI:10.1093/oxfordjournals.aje.a113979
21. Swanson TM, Selamet Tierney ES, Tworetzky W, Pigula F, McElhinney DB. Truncus Arteriosus: Diagnostic Accuracy, Outcomes, and Impact of Prenatal Diagnosis. *Pediatr Cardiol*. 2008;30(3):256–61. DOI:10.1007/s00246-008-9328-7
22. Buskens E. Efficacy of Fetal Echocardiography and Yield by Risk Category. *Obstet Gynecol*. 1996;87(3):423–8. DOI:10.1016/0029-7844(95)00439-4
23. Berghella V, Pagotto L, Kaufman M, Huhta JC, Wapner RJ. Accuracy of Prenatal Diagnosis of Congenital Heart Defects. *Fetal Diagn Ther*. 2001;16(6):407–12. DOI: 10.1159/000053949