Screening for congenital heart disease prior to fetal surgery
Cribado de cardiopatías congénitas previo a la cirugía fetal

Enrique Gil Guevara[1,2,a], Isabel Valdez Guerra[3,b], Jairo Muñoz Acosta[1,4,c]

ABSTRACT

Congenital cardiopathies are the most frequent malformations and represent a high neonatal morbidity and mortality rate, especially in those neonates without timely prenatal diagnosis. They appear as the first cause of infant death associated with congenital malformations. Advances in technology and the standardization of ultrasound diagnostic protocols have allowed increasingly accurate identification of congenital heart diseases. However, this has not been sufficient and there continues to be a high rate of neonates with congenital heart disease undiagnosed in prenatal surveillance. On the other hand, with the advent of fetal surgery, it is necessary to identify the presence of any major congenital heart disease that could cloud the prognosis associated with the underlying fetal pathology that made intrauterine intervention necessary. Considering the above, both parents and medical professionals need to be made aware of the importance of screening for congenital heart disease, especially in those fetuses that are going to undergo prenatal surgery. There is currently a high rate of errors in congenital heart disease screening, which causes anxiety in parents. This may be due to inadequate imaging technique, lack of standardization in the study protocol based on clinical practice guidelines, or lack of adequate training to perform this type of study. This article focuses on the most common errors in screening for congenital heart disease during second trimester morphological ultrasound, based on clinical practice guidelines for fetal echocardiography.
Key words: Fetal heart, Heart defects, congenital, Prenatal diagnosis; Echocardiography, Surgery, fetal

RESUMEN

Las cardiopatías congénitas son las malformaciones más frecuentes y significan una alta tasa morbimortalidad neonatal, sobre todo en aquellos neonatos sin diagnóstico prenatal oportuno. Representan la primera causa de muerte infantil asociada a malformaciones congénitas. El avance de la tecnología y la estandarización de los protocolos de diagnóstico ecográfico han permitido identificar cada vez con más precisión las cardiopatías congénitas. Sin embargo, esto no ha sido suficiente y sigue habiendo una alta tasa de neonatos con cardiopatías congénitas no diagnosticadas en la vigilancia prenatal. Por otro lado, con el advenimiento de la cirugía fetal se hace necesario identificar la presencia de alguna cardiopatía congénita mayor que pudiera ensombrecer el pronóstico asociado con la patología fetal de fondo que hizo necesaria la intervención intrauterina. Considerando lo anterior, se necesita concientizar tanto a los padres como a los profesionales médicos sobre la importancia del cribado de cardiopatías congénitas, especialmente en aquellos fetos que van a ser sometidos a cirugía prenatal. Actualmente existe una alta tasa de errores en el tamizaje de cardiopatías congénitas, lo que ocasiona ansiedad en los padres. Esto puede deberse a una técnica inadecuada en la toma de imágenes, falta de estandarización en el protocolo de estudio basado en las guías practicas clínicas, o bien la falta de entrenamiento adecuado para realizar este tipo de estudios. El presente artículo se enfoca en los errores más comunes en el tamizaje de cardiopatías congénitas durante la ecografía morfológica del segundo trimestre, tomando como base las guías practicas clínicas de ecocardiografía fetal.
Palabras clave. Corazón fetal, Cardiopatías congénitas Diagnóstico prenatal, Ecocardiografía, Cirugía fetal

INTRODUCTION

Congenital heart disease (CHD) is the most common intrauterine malformation, with an incidence of 8 per 1,000 live births[1]. One in four of these infants has critical congenital heart disease and requires early intervention or surgery during the first year of life[2]. Between 10% and 20% of congenital heart diseases are related to genetic and chromosomal alterations, maternal diseases, exposure to teratogens and familial recurrence[3]. In 1994, the World Health Organization attributed

30% of neonatal deaths to congenital heart disease[4]. Many risk factors related to congenital heart disease have been identified; however, 90% of heart malformations occur in low-risk patients[5].

Late diagnosis of critical heart disease, which occurs in 20%-30% of cases, causes significant infant morbidity and mortality[6,7]. Therefore, early detection is crucial to improve the survival of these children.

Prenatal diagnosis of congenital heart disease by echocardiographic screening has improved clinical outcomes, allowing adequate prenatal planning and timely interventions of the most important structural congenital heart diseases[8]. Likewise, fetal echocardiography is mandatory in any patient whose fetus is to undergo prenatal surgery, since the presence of major or critical congenital heart disease contraindicates fetal surgery, whereas minor or mild congenital heart disease does not contraindicate intrauterine intervention. Therefore, the identification of fetal congenital heart disease and its classification determines the course of fetal surgery, as well as the overall prognosis.

Although fetal cardiac evaluation can currently be performed from the first trimester ultrasound, the optimal gestational age for congenital heart disease screening is in the second trimester morphological ultrasound between 18 and 22 weeks of gestation[9] since the fetal cardiac anatomy can be well visualized at this stage of pregnancy. In addition, a complete fetal anatomical study can be performed and allows for further evaluation if necessary (advanced fetal echocardiography, genetic studies, among others).

The standard obstetric ultrasound examination includes evaluation of the four chambers and ventricular outflow tracts of the fetal heart. The inclusion of views of the outflow tracts increases the probability of identifying conotruncal anomalies, such as tetralogy of Fallot, transposition of the great arteries, double outlet right ventricle and common truncus arteriosus.

This examination has been systematized and standardized in clinical practice guidelines for ultrasound screening of the fetal heart by different organizations such as the International Society for Ultrasound in Obstetrics and Gynecology (ISUOG), the American Institute of Ultrasound in Medicine (AIUM), the American College of Radiology, the American College of Obstetricians and Gynecologists, and the Society for Maternal-Fetal Medicine (SMFM)[10-12].

Most of the data in the published literature describe the success of ultrasound for screening of congenital heart disease by prenatal diagnosis. In contrast, there are far fewer publications on errors, false-positive or false-negative interpretations. Therefore, the aim of this article is to address the possible reasons for diagnostic errors in each ultrasound slice and how to avoid them, knowing the transcendence of the presence of these cardiopathies in the context of a possible fetal surgery.

METHODOLOGY

An analytical cross-sectional time series study was performed in which recorded data were obtained from guidelines for the performance of fetal echocardiography and from the training process of fetal medicine physicians.

In the article we systematically describe the steps that are performed to generate an efficient fetal cardiac evaluation before deciding to perform fetal surgery.

FETAL ECHOCARDIOGRAPHY

Fetal echocardiography begins in the axial section of the abdomen at the level of the gastric chamber.

The first step is to determine the fetal presentation and situation. Precising which is the left and right side of the fetus is the basis for fetal anatomical orientation.

Fetal cardiac imaging should always be performed at the highest possible transducer frequency to maximize image resolution and at the highest possible frame rate, preferably > 50 hertz (Hz), due to the rapid motion of the heart which normally beats between 110 and 160 times per minute.

The standard four-chamber view can be obtained in 95-98 percent of second-trimester pregnancies[13-15].

Four-chamber view

The 4-chamber view is the most important section in the ultrasound examination of the fetal heart. Knowledge of the anatomy of the fetal thorax and heart, as well as knowledge of ultrasound dynamics, is fundamental for the clinical interpretation of the image. One should always start from the cut-off points recommended by ISUOG to obtain a perfect four-chamber cut-off, since recognizing the normality of this image excludes almost 60% of complex malformations of the fetal heart[16].

One of the most frequent errors occurs at the beginning of the visualization of the cardiac cross, fundamental at the moment of evaluating the differential insertion of the atrioventricular valves (valvular off-setting). Precisely at this level, when the four-chamber view is apical, the artifact caused by the fetal sternum could give the impression of a defect of the interventricular septum in its muscular portion, leading to a diagnostic error. Therefore, the evaluation of the interventricular septum should always be performed in a view of the interventricular septum with the ultrasound beam incident perpendicularly to the septum (Figure 1).

The use of color Doppler is a point of good practice and should be applied with proper optimization of the pulse repetition frequency (PRF) and color Doppler gain to avoid inadequate interpretations (Figure 2).

The visualization of conspicuous structures must be carefully evaluated, such as the presence of atypical structures in the thorax (among them the fetal stomach), so it is necessary to obtain a four-chamber section where only one costal arch is visualized on each side[17] (Figure 3).

Another common error is to consider as pathological the presence of mild and transient bradycardia, which is characteristic in healthy fetuses in the second trimester. Similarly, the existence of occasional skipped beats that spontaneously resolve corresponds to a normal condition of the maturation of the fetal sympathetic and parasympathetic systems[18].

Section of outflow tracts

Implementing the section of the outflow tracts gives a sensitivity of up to 80% in the screening for congenital heart disease in the second trimester. Despite advances in prenatal diagnosis of congenital heart disease, the greatest error occurs in conotruncal heart disease[19].

Inadequate visualization or insonation in the outflow tracts can lead to errors in screening for congenital heart disease. This can be avoided by always starting from the four-chamber view

Figure 1. Image A shows a 4-chamber view of a fetus with posterior dorsum in cephalic presentation with the heart in apical position. The artifact caused by the sternum and the interventricular septum itself give the impression of a perimembranous defect. Image B shows the same fetus in four-chamber view with dorsum to the right and insonation perpendicular to the interventricular septum, showing the integrity of the septum.
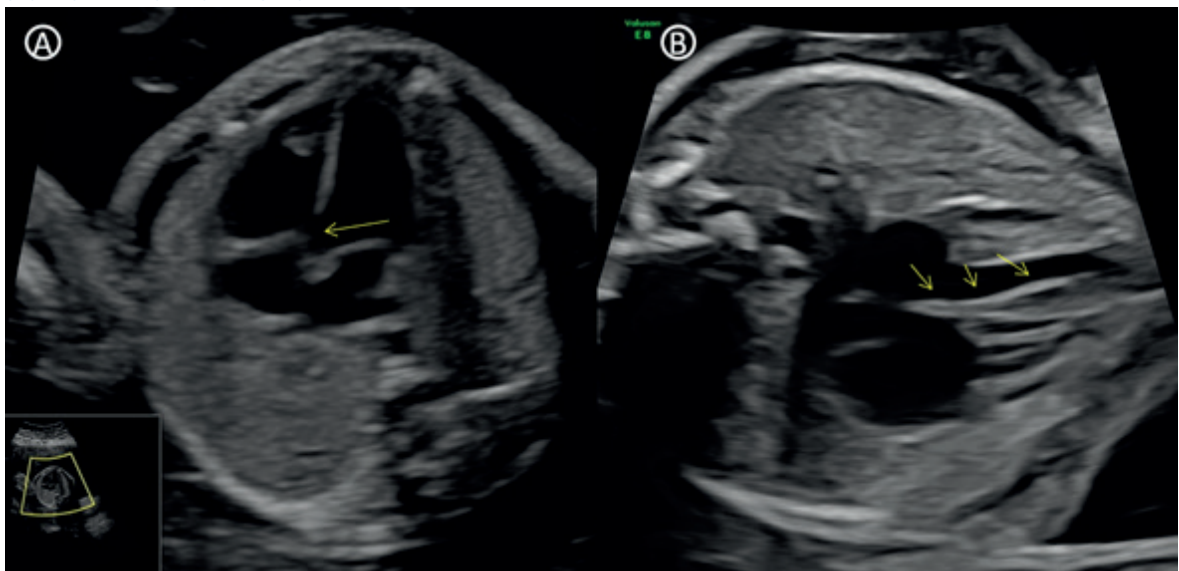
FIGURE 2. IMAGE A SHOWS INADEQUATE MANAGEMENT OF COLOR DOPPLER OPTIMIZATION. A LOW PRF AND INCREASED GAIN LEAD TO A LOW-QUALITY IMAGE AND IN THIS CASE THE IMPOSSIBILITY OF DIFFERENTIATING BETWEEN AN INTACT SEPTUM OR AN INTERVENTRICULAR DEFECT. IMAGE B SHOWS AN ADEQUATE OPTIMIZATION OF COLOR DOPPLER WITH HIGH PRF AND LOW GAIN, WHICH ALLOWS ADEQUATE DISCRIMINATION OF THE TWO FLOW CHANNELS IN THE CARDIAC CAVITIES.
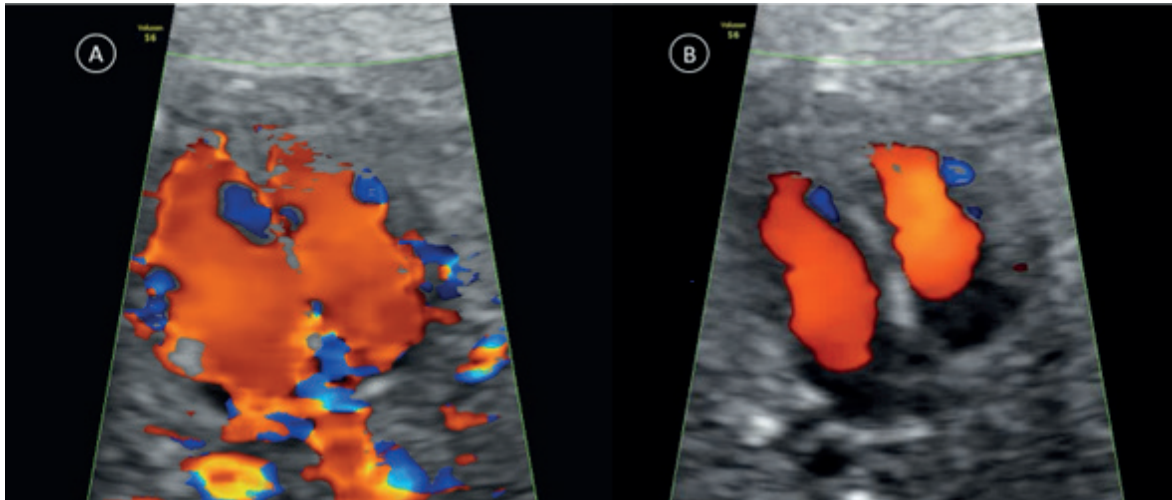

FIGURE 3. IMAGE A SHOWS AN INADEQUATE TECHNIQUE OF THE FOUR-CHAMBER VIEW. THE SECTION IS OBLIQUE, SHOWING THE GASTRIC CHAMBER (E) AND THE HEART (C) IN THE SAME PLANE, EVEN WITH ASYMMETRIC CARDIAC CAVITIES, AND THE CARDIAC AXIS DISPLACED, WHICH COULD LEAD TO AN INADEQUATE DIAGNOSIS OF CONGENITAL DIAPHRAGMATIC HERNIA. IMAGE B SHOWS THE SAME FETUS WITH AN ADEQUATE FOUR-CHAMBER VIEW SHOWING ANATOMICAL NORMALITY, FOLLOWING THE REFERENCE POINTS RECOMMENDED BY CLINICAL PRACTICE GUIDELINES.
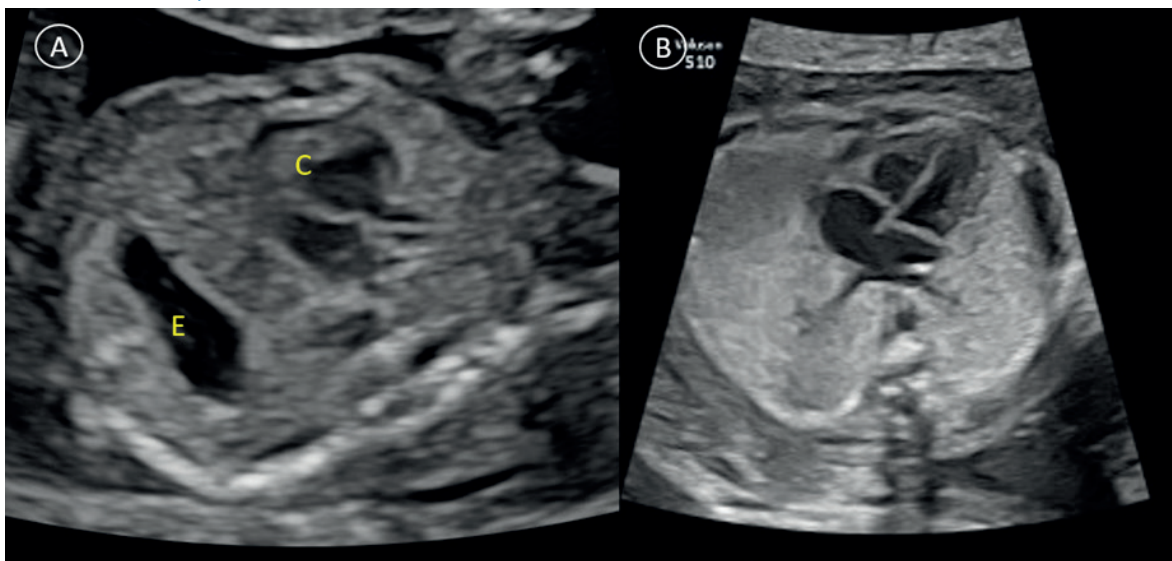
to the outflow tracts. The main mistake when locating the sections of the great vessels is to go directly to look for them without having a correct 4-chamber view, since this always represents the starting point to evaluate the outflow tracts[20](Figure 4).

Obtaining an optimal 4-chamber plane and then performing a transversal sweep in cephalic direction with gentle displacement and rocking movements will allow us to obtain the outflow tract sections. We can also perform a rotational movement towards the fetal right shoulder (left ventricular outflow tract) and from this point perform an angulation movement in cephalic direction (right ventricular outflow tract).

A frequent error occurs when applying this rotational technique in an apical cut of 4 chambers, so it is recommended to perform it from a subcostal section with the ultrasound beam perpendicular to the interventricular septum[21].

The fetal position is of vital importance at the moment of achieving the correct cuts. Therefore, it is essential to avoid the mistake of post-

Figure 4. Image A shows an inadequate cut of the left outflow tract. Image B shows the same slice in which the fetus is seen in coronal view. This technique may give the false impression of a perimembranous defect, which is even measured in image C. Image D shows the same fetus with an adequate cut of the aortic outflow tract in which the integrity of the long axis of the interventricular septum is confirmed. The error in images A, B and C is avoided by always taking the four-chamber section as the starting point for addressing the outflow tracts of the great vessels.
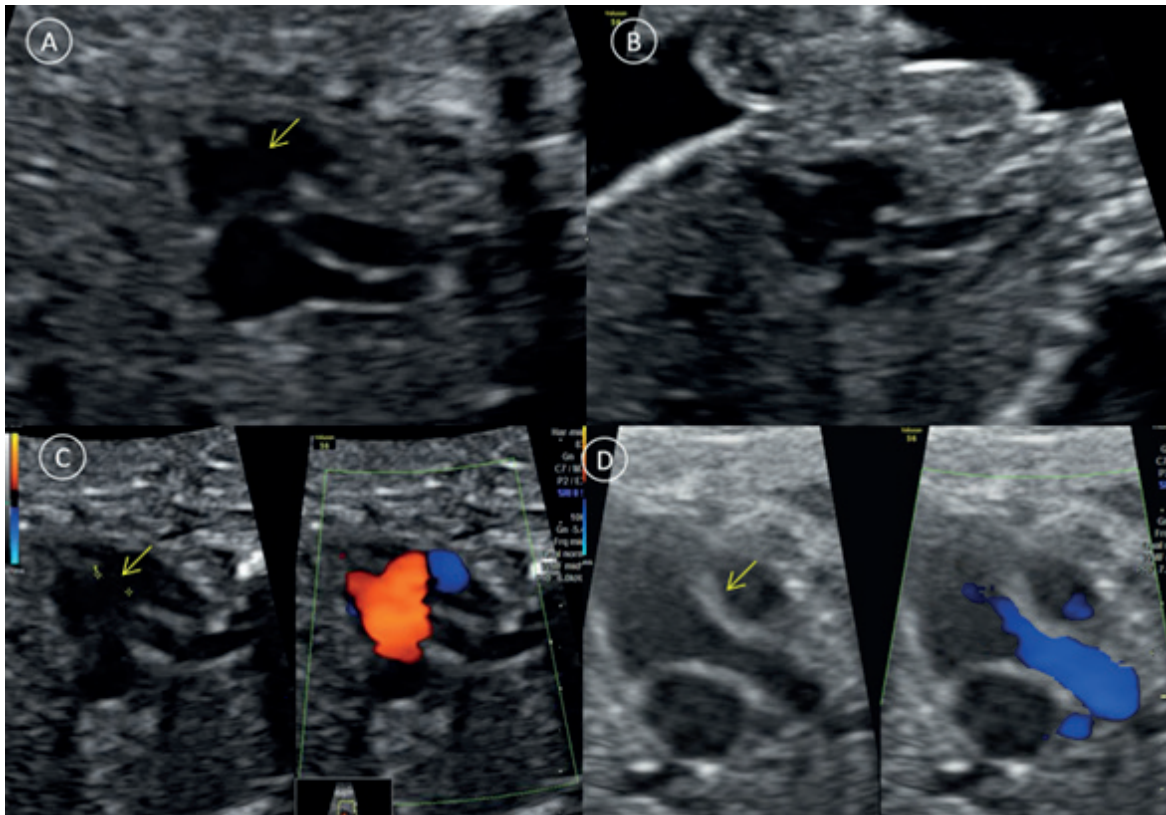
poning the evaluation of the outflow tracts if a favorable fetal position is observed at the time of starting the fetal exploration.

THREE-VESSEL TRACHEAL SECTION

The sections corresponding to the 3 vessels (ascending aorta, pulmonary artery and superior vena cava) and their relationship with the trachea (3-vessel trachea) were described by Yoo et al. and Yagel et al, respectively[22].

In these sections we will evaluate the relationship of the vessels to each other in terms of their size and their relation to the airway (trachea). Again, it is of utmost importance to visualize the 4-chamber slice in a satisfactory manner and then perform a transversal sweep in cephalic direction up to the 3-vessel and 3-vessel trachea section. Finally, our screening for congenital heart disease would be incomplete if we do not complement our examination with the application of color Doppler. It is an indispensable tool, and it would be a mistake to do without it, especially in obese patients where cardiac anatomy becomes difficult to evaluate.

We emphasize the importance of the basics of obstetric ultrasound. The determination of the fetal presentation and situation to determine the left and right side of the fetus is a pillar in every fetal ultrasound examination.

Regarding the three-vessel tracheal section, in a general and panoramic visualization we will have to see the disposition of the vessels in order not to make mistakes. Thus, from left to right, from the largest vessel to the smallest, the pulmonary artery is located, then the aorta artery and finally the superior vena cava. The most common error when using color Doppler is to inappropriately insonate the 'V' sign given by the arrangement of the aorta and pulmonary artery. Therefore, to avoid this error, the insonation of the area of interest at the level of the three tracheal vessels should always be in an anteroposterior or posteroanterior direction in relation to the fetal spine (Figure 5).

CONCLUSIONS

During the last decade, fetal medicine has advanced by leaps and bounds, thanks to advances in technology and training programs carried out by major fetal medicine organizations.

Ultrasonographic detection of congenital heart disease at different stages of gestation is often difficult because the fetal heart is a small organ, it is constantly moving, and the fetus is not always in the best position to evaluate it.

To be able to achieve a level of accuracy in this study requires highly trained physicians with extensive training in obstetric ultrasound, as well as in the diagnosis of congenital malformations and with knowledge of pathophysiology and malformations of the fetal heart.

Good equipment technology is also required. In order to assess fetal cardiovascular anatomy and function, a high-resolution two-dimensional ultrasound system is needed, optimized M-mode, integrated with pulsed Doppler, continuous Doppler and color Doppler systems, and with real-time analysis commonly used in pediatric cardiology.

Finally, fetal echocardiography is mandatory in all patients whose fetus is to undergo fetal surgery, since the presence of major congenital heart disease contraindicates fetal surgery, while minor congenital heart disease does not contraindicate intrauterine intervention.

REFERENCES

1. Van der Linde D, Konings EE, Slager MA, Witsenburg M, Helbing WA, Takkenberg JJ, Roos-Hesselink JW. Birth prevalence of congenital heart disease worldwide: a systematic review and meta-analysis. J Am Coll Cardiol. 2011 Nov 15;58(21):2241-7. doi: 10.1016/j.jacc.2011.08.025

2. Mahle WT, Newburger JW, Matherne GP, Smith FC, Hoke TR, Koppel R, Gidding SS, Beekman RH 3rd, Grosse SD; American Heart Association Congenital Heart Defects Committee of the Council on Cardiovascular Disease in the Young, Council on Cardiovascular Nursing, and Interdisciplinary Council on Quality of Care and Outcomes Research; American Academy of Pediatrics Section on Cardiology And Cardiac Surgery; Committee On Fetus And Newborn. Role of pulse oximetry in examining newborns for congenital heart disease: a scientific statement from the AHA and AAP. Pediatrics. 2009 Aug;124(2):823-36. doi: 10.1542/peds.2009-1397

3. Buskens E, Grobee DE, Frohn MI, Wladimiroff JW, Hess J. Aspects of the aetiology of congenital heart disease. Eur Heart J 1995;16:584-7.

4. Bishop KC, Kuller JA, Boyd BK, Rhee EH, Miller S, Barker P. Ultrasound Examination of the Fetal Heart. Obstet Gynecol Survey. 2017;72(1):54–61.

5. Rosano A, Botto LD, Botting B, Mastroiacovo P. Infant mortality and congenital anomalies from 1950 to 1994: an international perspective. J Epidemiol Community Health. 2000;54:660–666. doi: 10.1136/jech.54.9.660

6. Liberman RF, Getz KD, Lin AE, Higgins CA, Sekhavat S, Markenson GR, Anderka M. Delayed diagnosis of critical congenital heart defects: trends and associated factors. Pediatrics. 2014 Aug;134(2):e373-81. doi: 10.1542/peds.2013-3949

7. Eckersley L, Sadler L, Parry E, Finucane K, Gentles TL. Timing of diagnosis affects mortality in critical congenital heart disease. Arch Dis Child. 2016 Jun;101(6):516-20. doi: 10.1136/archdischild-2014-307691

8. Fixler DE, Xu P, Nembhard WN, Ethen MK, Canfield MA. Age at referral and mortality from critical congenital heart disease. Pediatrics. 2014 Jul;134(1):e98-105. doi: 10.1542/peds.2013-2895

FIGURE 5. IMAGE A SHOWS A CUT OF THREE VESSELS WITH INADEQUATE INSONATION OF COLOR DOPPLER GIVING THE IMPRESSION OF REVERSE FLOW AT THE LEVEL OF THE AORTA. IMAGE B DENOTES AN ADEQUATE TECHNIQUE WITH THE FETUS IN POSTERIOR DORSUM AND INSONATION OF THE ULTRASOUND BEAM IN AN ANTEROPOSTERIOR DIRECTION IN RELATION TO THE FETAL SPINE, CORROBORATING THE NORMAL FLOW DIRECTION.
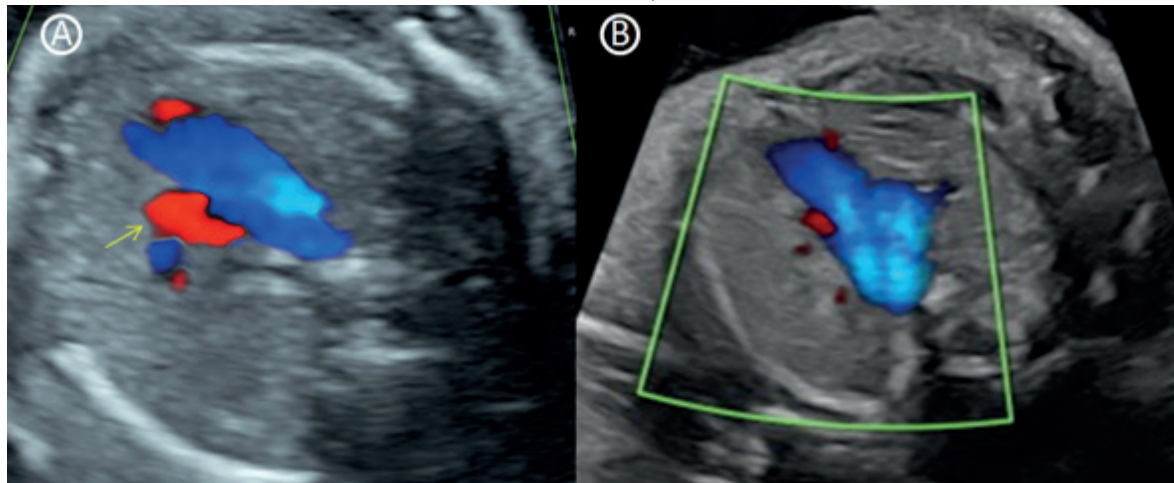
9. Mozumdar N, Rowland J, Pan S, Rajagopal H, Geiger MK, Srivastava S, Stern KWD. Diagnostic Accuracy of Fetal Echocardiography in Congenital Heart Disease. J Am Soc Echocardiogr. 2020 Nov;33(11):1384-90. doi: 10.1016/j.echo.2020.06.017

10. Donofrio MT, Moon-Grady AJ, Hornberger LK, Copel JA, Sklansky MS, Abuhamad A, Cuneo BF, Huhta JC, Jonas RA, Krishnan A, Lacey S, Lee W, Michelfelder EC Sr, Rempel GR, Silverman NH, Spray TL, Strasburger JF, Tworetzky W, Rychik J; American Heart Association Adults With Congenital Heart Disease Joint Committee of the Council on Cardiovascular Disease in the Young and Council on Clinical Cardiology, Council on Cardiovascular Surgery and Anesthesia, and Council on Cardiovascular and Stroke Nursing. Diagnosis and treatment of fetal cardiac disease: a scientific statement from the American Heart Association. Diagnosis and treatment of fetal cardiac disease: a scientific statement from the American Heart Association. Circulation. 2014 May 27;129(21):2183-242. doi: 10.1161/01.cir.0000437597

11. International Society of Ultrasound in Obstetrics and Gynecology, Carvalho JS, Allan LD, Chaoui R, Copel JA, DeVore GR, Hecher K, Lee W, Munoz H, Paladini D, Tutschek B, Yagel S. ISUOG Practice Guidelines (updated): sonographic screening examination of the fetal heart. Ultrasound Obstet Gynecol. 2013 Mar;41(3):348-59. doi: 10.1002/uog.12403

12. AIUM-ACR-ACOG-SMFM-SRU Practice Parameter for the Performance of Standard Diagnostic Obstetric Ultrasound Examinations. J Ultrasound Med. 2018 Nov;37(11):E13-E24. doi: 10.1002/jum.14831

13. Carvalho JS, Mavrides E, Shinebourne EA, Campbell S, Thilaganathan B. Improving the effectiveness of routine prenatal screening for major congenital heart defects. Heart. 2002 Oct;88(4):387-91. doi: 10.1136/heart.88.4.387

14. Tegnander E, Eik-Nes SH, Linker DT. Incorporating the four-chamber view of the fetal heart into the second-trimester routine fetal examination. Ultrasound Obstet Gynecol. 1994 Jan 1;4(1):24-8. doi: 10.1046/j.1469-0705.1994.04010024.x

15. Shipp TD, Bromley B, Hornberger LK, Nadel A, Benacerraf BR. Levorotation of the fetal cardiac axis: a clue for the presence of congenital heart disease. Obstet Gynecol. 1995 Jan;85(1):97-102. doi: 10.1016/0029-7844(94)00328-b

16. Del Bianco A, Russo S, Lacerenza N, Rinaldi M, Rinaldi G, Nappi L, Greco P. Four chamber view plus three-vessel and trachea view for a complete evaluation of the fetal heart during the second trimester. J Perinat Med. 2006;34(4):309-12. doi: 10.1515/JPM.2006.059

17. ACOG Practice Bulletin No. 106: Intrapartum fetal heart rate monitoring: nomenclature, interpretation, and general management principles. Obstet Gynecol 2009;114:192–202.

18. Forbus GA, Atz AM, Shirali GS. Implications and limitations of an abnormal fetal echocardiogram. Am J Cardiol. 2004 Sep 1;94(5):688-9. doi: 10.1016/j.amjcard.2004.05.047

19. Vettraino IM, Lee W, Bronsteen RA, Comstock CH. Sonographic evaluation of the ventricular cardiac outflow tracts. J Ultrasound Med. 2005 Apr;24(4):566. doi: 10.7863/jum.2005.24.4.566

20. DeVore GR. The aortic and pulmonary outflow tract screening examination in the human fetus. J Ultrasound Med. 1992 Jul;11(7):345-8. doi: 10.7863/jum.1992.11.7.345

21. Yoo SJ, Lee YH, Kim ES, Ryu HM, Kim MY, Choi HK, Cho KS, Kim A. Three-vessel view of the fetal upper mediastinum: an easy means of detecting abnormalities of the ventricular outflow tracts and great arteries during obstetric screening. Ultrasound Obstet Gynecol. 1997 Mar;9(3):173-82. doi: 10.1046/j.1469-0705.1997.09030173.x

22. Chaoui R, McEwing R. Three cross-sectional planes for fetal color Doppler echocardiography. Ultrasound Obstet Gynecol. 2003 Jan;21(1):81-93. doi: 10.1002/uog.5