Received: 27 April 2023 | Revised: 13 May 2023 | Accepted: 27 May 2023

DOI: 10.1002/pd.6391

REVIEW

PRENATAL DIAGNOSIS WILEY

Congenital heart anomalies in the first trimester: From screening to diagnosis

Wawrzyniec Rieder[1,2] | Isabelle Eperon[2] | Simon Meagher[3,4,5]

[1]Department of Obstetrics and Gynecology, Lausanne University Hospital, Lausanne, Switzerland

[2]Dianecho, Geneva, Switzerland

[3]Department of Perinatal Medicine, Mercy Hospital for Women, Heidelberg, Victoria, Australia

[4]Department of Obstetrics and Gynecology, University of Melbourne, Parkville, Victoria, Australia

[5]Monash Ultrasound for Women, Melbourne, Victoria, Australia

Correspondence
Wawrzyniec Rieder, Department of Obstetrics and Gynecology, Centre Hospitalier Universitaire Vaudois (CHUV), Lausanne 1011, Switzerland.
Email: wawrzy.rieder@gmail.com

Abstract

Congenital heart defects occur in approximately 1% of liveborn children and represent the most common form of congenital malformation. Due to the small size and complexity of the heart structures, prenatal diagnosis is most often made in the second trimester of pregnancy. Early diagnosis however offers significant advantages regarding the timing of further investigations, prenatal counseling, and access to management options. In the last decade, advances in antenatal imaging have improved the detection of cardiac malformations with increasing emphasis on earlier pregnancy screening and diagnosis. We aim to summarize current "state of the art" imaging of the fetal heart in the first trimester.

Key points

What's already known about this topic?

- Congenital heart defects are the most common form of congenital malformation.
- Antenatal detection of major congenital heart defects is possible in the first trimester.

What does this study add?

- We provide a comprehensive review of the first trimester cardiac assessment in line with updated international guidelines.
- We offer a simple and efficient visual guide to first trimester normal and abnormal heart views.

1 | INTRODUCTION

The use of ultrasound in pregnancy monitoring is constantly evolving. For several decades, ultrasound has been employed in the first trimester for assessing the nuchal translucency (NT) for aneuploidy screening between 11 and 14 weeks of gestation. In many countries, the first trimester scan has evolved into a routine anatomical assessment in early pregnancy that continues to be recommended alongside cell-free DNA based aneuploidy screening.[1] In parallel, technological innovations have allowed for better visualization of smaller and complex structures such as the fetal heart.[2,3]

Congenital heart diseases (CHDs) affect 4–9/1000 children at birth, are the most common congenital malformations and remain a major cause of infant mortality and morbidity worldwide.[4] Cardiac examination became a routine part of the fetal anatomical assessment in the mid-trimester over 20 years ago.[5] There is now growing evidence regarding our ability to identify cardiac defects in the earlier stages of pregnancy.[6] Most recent guidelines recognize the benefit of an early diagnosis as well as the need to improve screening for heart anomalies by extending the first trimester cardiac examination beyond the assessment of heart rate and position.[7] In this article, we review the technical and practical aspects of the early cardiac scans and describe early sonographic findings and patterns for some of the most common or severe congenital cardiac malformations.

2 | WHY SCAN ALL HEARTS IN THE FIRST TRIMESTER?

The embryological development of the heart represents a complex multiple-step process that is influenced by various genetic and molecular mechanisms. Adverse events that occur at any stage of this process may result in a congenital heart defect.[8] The anatomy of the heart is fully completed by 8–10 weeks of pregnancy and cardiac anomalies are already present by 11–14 weeks.[9] The majority of CHDs occur in low-risk groups, yet current practice commonly limits the first trimester cardiac examination to patients with specific risk factors. Such specialized ultrasounds are performed by an expert sonographer/sonologist and usually reserved to tertiary referral centers. In the absence of an early targeted fetal echocardiogram, the diagnosis of cardiac malformations is typically made in the second trimester.[10]

Prenatal screening for CHDs represents a cost-effective strategy, although no studies addressed this question specifically regarding the first trimester scan.[11] However, an early diagnosis of fetal anomalies presents several beneficial aspects.[12] It provides the parents with valuable time to integrate the information and benefit from a well-informed decision-making process. It also allows health professionals to counsel the expecting parents and schedule further prenatal investigations without the pressure of time-sensitive considerations such as gestational-age limitations on access to termination of pregnancy (TOP). Genetic abnormalities are observed in up to one third of prenatally diagnosed CHDs.[13] Given the time necessary to obtain the results, it is important to offer the possibility of genetic tests as early as possible. Jicinska et al. showed that an early diagnosis of cardiac anomalies modifies the spectrum of CHDs that can be encountered in the second or third trimester and greatly impacts the final outcomes of the pregnancy.[14] Certain complex CHDs have a poor prognosis, which increases the rates of TOP.[15,16] In such circumstances, early intervention is medically safer, offers more privacy and presents important psychological benefits for patients.[17]

Standard mid-trimester cardiac anatomical sections may be difficult to obtain during the 11–14 weeks scan. The examination technique must be adapted in accordance with the stage of pregnancy and the examination technique requires adequate training and experience. While the first trimester heart examination can be performed in just a few minutes, it is often perceived as an overly time-consuming procedure. This may be particularly relevant in challenging technical conditions such as a retroverted uterus, suboptimal fetal position, or increased maternal body mass index (BMI). Finally, many CHDs present with a broad spectrum of severity. Prognostic indicators may not be evident in the first trimester (e.g. diminution of pulmonary artery caliber in Tetralogy of Fallot [TOF]), and as such, accurate prognostication may prove challenging. Medical professionals may wish to postpone parental counseling to a later stage of pregnancy when more definitive counseling can be provided. Arguments in favor and against early screening for CHDs are summarized in Table 1.

3 | HOW TO SCAN THE HEART IN THE FIRST TRIMESTER?

Appropriate training, including a systematic approach to examining the fetal heart, is key in the first trimester cardiac assessment. Indeed, when a detailed cardiac examination is being performed by the revised International Society of Ultrasound in Obstetrics and Gynecology (ISUOG) first trimester guidelines (2023) advocate routine evaluation of the 4-chamber view of heart and outflow tracts with both 2D and color Doppler examination.[7]

Technical aspects are important when considering the examination of the fetal heart between 11 and 14 weeks and machine

TABLE 1 Arguments in favor and against early screening for congenital heart disease.

In favor	Against
Indirect sonographic markers (cardiac axis deviation, nuchal translucency, tricuspid valve and ductus venosus Doppler changes) are present between 11 and 14 weeks and provide a unique opportunity which may lead to early cardiac diagnosis	Different ultrasound technique from second trimester scan and need for specific operator training and expertise
Cardiac anomalies are already present and detectable in >50% of cases during the 11–14 weeks scan	Difficulties in achieving a diagnosis given small size of cardiac structures
More time for prenatal counselingMore time to arrange further prenatal investigations	Reluctance of medical professionals to accept the early anatomical screening and diagnosis, Limitations in prognostication, given the variable natural history of certain cardiac lesions
More time for parents to process information and make decisions	Little evidence of cost benefit
Earlier and safer access termination of pregnancy	

settings should be set accordingly. Table 2 presents our suggested settings for both grayscale and color Doppler imaging.

During this narrow time frame, the heart measures approximately 8–10 mm from the apex to base and 7–9 mm from side to side at the level of the atrio-ventricular (AV) annuli while outflow tracts measure 1–2 mm.[18,19] This small size provides an imaging challenge and thus the correct transducer frequency selection and adequate magnification are of paramount importance.

The overall success rate at obtaining correct cardiac views in the first trimester increases progressively with advancing gestational age and is the highest after 13 week.[20–22] Hutchinson et al. reported that a 4 chambered heart is visualized in 52% and 80% of fetuses at 8 and 10 weeks of gestation respectively. The identification of outflow tracts seems more challenging. By week 10, both the aortic and ductal arches are seen in 29% on gray scale imaging, and in 58% with the addition of color Doppler.[22] The ability to successfully recognize cardiac structures by gestational week is summarized in Table 3. Targeting appropriate timing for the first trimester evaluation of the fetal heart plays a crucial role in improving detection rates. We suggest optimizing detection rates by planning the first trimester ultrasound as close as possible to 13 weeks of pregnancy.

Although highly dependent on fetal position, the first trimester examination offers some technical advantages over the second trimester. Increased movements enhance the chances for the fetus to position itself appropriately for a cardiac assessment during the time of the examination and a relative absence of shadowing that result from low ossification of the bones allows to obtain good quality images even with interposed bony structures such fetal spine. In addition, considering a very active fetus, the Cineloop acts as a valuable tool during a heart examination in the first trimester.

3.1 | Transabdominal or transvaginal?

Appropriate images can be obtained with an abdominal approach alone in most cases which in the context of screening presents the benefits of a shorter scanning time and patient's comfort. A transvaginal approach has the advantage of higher image quality but remains dependent on fetal position and requires the sonographer to be familiar with trans-vaginal imaging. It is of particular value in patients with a large BMI as it provides a satisfactory examination in more than 90% of patients.[23–28] It is authors belief that a transvaginal approach should be encouraged in all cases of suspected cardiac pathology or if sufficient image quality cannot be obtained transabdominally.

3.2 | Color Doppler

Color Doppler examination permits the evaluation of not only the anatomical dimensions of the ventricles and great arteries but also the dynamics of blood flow, which are so relevant in the first trimester cardiac diagnosis. Color Doppler examination overcomes some of the limitations of gray, especially in regard to the examination of the outflow tracts at the level of the 3-vessels tracheal view

TABLE 2 Suggested machine settings for the first trimester heart examination.

Gray scale	- Narrow the window width to the size of the fetal thorax - Magnify the image (fetal thorax should cover >50% of the image) - Increase dynamic contrast and gain. - Use of highest frequency possible (ideally 9 MHz trans-abdominal, 5–9 MHz or 6–12 MHz trans-vaginal)
Color Doppler	- Optimize gray-scale image first - Size of the box should include the entire heart - Pulse rate frequency between 20 and 30 cm/s - Ultrasound power (usually between 90% and 95%) - Filter levels (mid to high) and color gain (the image should display a well-defined blue and/or red flow, without overlapping onto neighboring structures and without aliasing)

TABLE 3 Visualization of selected cardiac structures according to gestational age.

Week of gestation	Four chambers	Cardiac axis	Outflow tracts		Crossing of great arteries		Aortic arch	
	2D	2D	2D	Color Doppler	2D	Color Doppler	2D	Color Doppler
10	80%	84%	24%	64%	24%	64%	29%	60%
11	98%	97%	72%	91%	69%	91%	66%	81%
12	100%	100%	76%	94%	75%	95%	75%	96%
13	100%	100%	97%	97%	94%	94%	94%	90%

Source: Adapted from Hutchinson et al.[22]

(3VT).[29,30] Indeed, routine use of Doppler during a first trimester cardiac assessment improves the detection rates of cardiac anomalies, and for this reason, it has been incorporated in the revised ISUOG guidelines 2023.[6,7]

New Doppler technologies optimized for low velocity vasculature are now available on ultrasound machines. This improves Doppler spatial resolution and allows for a better visualization of slow flow rates in tiny vessels such as pulmonary veins, especially in early stages of pregnancy.[31,32]

3.3 | Spatiotemporal image correlation 4D sonography

Spatiotemporal image Correlation (STIC) 4D sonography is a potentially valuable tool for first trimester fetal heart assessment. It has the advantage of reducing operator dependency and allows for off-line storage of images that can be reviewed later by other specialist examiners.[33–36] Use of STIC volumes in the first trimester has shown reproducibility and inter-observer agreement.[37] Turan et al. demonstrated excellent diagnostic accuracy with a sensitivity of 91% and specificity of 100% in the diagnosis of complex CHD using a standardized approach to STIC, tomographic ultrasound imaging and Color Doppler Stored volumes.[38] However, there is a need of a high level of operator expertise and access to specialist technology, which limits the incorporation of this technique into routine clinical practice.

3.4 | Safety of first trimester fetal heart ultrasound

The safety of ultrasound in pregnancy is an important matter that has been previously addressed.[39] In the first trimester, concerns are particularly related to the highly focused energy delivered during Doppler ultrasound examination. The performance of modern machines makes it possible to limit the bioeffect by keeping the tissue Thermal Index and Mechanical Index below the recommended thresholds of 1.0.[40,41] Therefore, it appears safe to use ultrasound and Doppler for cardiac examination after 11 weeks of pregnancy if the ALARA ("As Low as Reasonably Achievable") principles are followed.

4 | HOW ACCURATE IS AN EARLY CARDIAC EXAMINATION?

The precise estimation of CHD detection rates is challenging due to the significant heterogeneity in study design, target population, gestational age, center expertise, and local protocols. Performance is higher following a referral scan due to greater operator experience, rigorous protocols, and an increased prevalence of anomalies in a high-risk population (e.g. presence of an increased nuchal translucency or previous CHD). The risk of cardiac defects is six-fold higher in case of a NT above the 99th percentile, one of the strongest indirect markers of CHDs in the first trimester.[42–47]

Detection rates are poorer while screening in an unselected (low prevalence) population, and when the examination is carried out by staff with limited training. In a recent meta-analysis of over 300,000 pregnancies, Karim et al. reported an overall first-trimester sensitivity of 56% in an unselected population and 68% in high-risk groups, with a comparable positive predictive value of 94%.[6]

4.1 | Counseling after a suspected cardiac anomaly in first trimester

Gestational age at the time of scanning plays another important role. Although several authors have reported cardiac anomalies before 11 weeks of gestation, great caution should be taken before making a diagnosis at such an early stage of pregnancy.[48] Screening performance is influenced by the size of the fetus with better detection after 13 weeks of gestation. Changes in the morphology and function of an early heart are to be taken into consideration.[49] While most studies specifically report on detection rates, limited data exist regarding false positive results and the risk of error. Ebrashy et al. reported 53 cases of mismatching results (out of 3240 examinations) between first- and second-trimester ultrasounds, which included 33 unconfirmed cardiac malformations.[20] In fact, the false-positive rate is particularly difficult to estimate because of the limited number of postmortem confirmations if a TOP is performed in the first trimester.

Sonographers experience and time allowed for a heart examination are among the most important factors that influence the ability to appropriately assess cardiac structures in the first trimester.[26,50] Botelli et al. identified human error as the primary cause of false negative diagnoses.[51] The lack of operator's adaptational skills (rather circumstantial aspects) mainly impacts correct appreciation of CHDs in the second trimester and it is likely that such factors similarly affect the early detection rate.[23] The frequency of successful examinations grows with the number of scans performed and a learning curve of 180 ultrasound appears necessary to appropriately visualize the heart in most cases.[26]

Great caution should be taken following early identification of a CHD as the negative impact of a false positive diagnosis cannot be underestimated. The consequences of a false negative result are of lesser consequence as patients usually benefit from a second ultrasound in the mid-trimester. Several pathologies can remain undetectable in the first trimester (e.g. hypoplastic left ventricle) and it is therefore important to inform patients of the limitations of an early examination and highlight the importance of subsequent anatomical ultrasound assessment. It is also essential to establish clear local referral pathways for all patients with a suspected fetal cardiac abnormalities. Confirmation of the diagnosis and counseling by a specialist in fetal medicine or pediatric cardiologist are of paramount importance. Given the limitations in service provision, the anxiety associated with early diagnosis and the impact of a false positive diagnosis, practitioners may be reluctant to screen for CHD so early in the pregnancy. Such a paradigm shift requires education about the benefits of first trimester cardiac scanning and the importance of appropriate training among medical professionals when interpreting early images and providing parental counseling. A multi-disciplinary approach, including fetal medicine specialists, pediatric cardiologist and cardiac surgeons, geneticists and neonatologists should be promoted during prenatal care.

4.2 | What should the standard screening views be at 11–13 weeks?

The degree of detail sought during anatomical assessment of the heart strongly impacts its performance. When a routine assessment is limited to the 4-chamber view (4CHv) a high proportion of major cardiac anomalies are missed.[52] Failure to adopt a systematic examination of the cardiac outflow tract results in low antenatal detection rates. More recently, it has been shown that color Doppler visualization of cardiac chambers, transverse aortic and ductal arch is achievable in over 80% of cases.[25,29] Indeed, the use of standardized protocols appears to significantly impact the performance of the first trimester scan.[2,28,53] In a recent meta-analysis, Karim et al. showed that the sensitivity of ultrasound increases progressively from 32% (if only 4CH are examined), to 56% (4CH and outflow tracts), to >80% if both structures are assessed on gray-scale and color Doppler.[6] Most recent guidelines now encourage the examination of 4-chambers and outflow tracts with color Doppler in routine clinical practice.[7,54]

5 | CLINICAL GUIDELINES ON FIRST TRIMESTER ULTRASOUND

The recently published update of the ISUOG guidelines for the first trimester describe 2 levels of screening, acknowledging that successful detection of CHDs depends in part upon local resources and sonographer's experience (Table 4).[7] The “minimum requirements” only include the assessment of the heart in the 4CHv to confirm its intra-thoracic position and the presence of regular heart rhythm. While this approach is primarily intended to establish fetal viability, it does not allow for either a dedicated assessment of normal cardiac anatomy or detection of cardiac malformations. On the other hand, the “best practice” recommendations intend for a more comprehensive examination of the heart with the aim of detecting CHDs and include an evaluation of different cross-sectional planes of the heart in combination with pulsed and color Doppler ultrasound. Normal aspects of the fetal heart are summarized in Figure 1.

5.1 | 4 Chamber view

After 11 weeks of gestation, a standard 4 chamber view (4CHv) can be completed on grayscale ultrasound and color Doppler in 98% and 86% respectively.[22,25,55] Heart rate is usually faster in the early stages of pregnancy and ranges between 140 and 170 beats per minute at the 11–14 weeks scan. Fetal arrhythmias are extremely rare in the first trimester. Cardiac situs is easier to observe trans-abdominally as the spatial orientation may be more difficult to address on trans-vaginal approach. Situs solitus refers to a normal arrangement of vessels and organs with the stomach and heart positioned on the left. The fetal heart in the first trimester usually occupies about one third of the thorax. Dilated or hypertrophic cardiomyopathies are typically evolving pathologies and are not seen in early pregnancy. Therefore, cardio-thoracic disproportion in the first trimester is much more likely to be due to a small thorax, as seen in some lethal forms of skeletal dysplasia.

The normal heart is positioned at an angle of 45° (+/−15°). An abnormal axis presents mostly as a left deviation. In the first trimester, axis deviation is seen in up to 74% of fetuses with CHD and

TABLE 4 “Minimum requirements” and “best practice” for heart examination (adapted from 2023 updated ISUOG guidelines for 11–14-week ultrasound scan).

Minimum requirements	
4 chambers view (axial)	- Heart inside chest - Regular rhythm
Best practice	
4 chambers view: gray scale (axial)	- Heart activity, regular heart rhythm - Situs: Intra-thoracic - Cardiac axis: 30°–60°, pointing to the left - Size: One third of thoracic space - Presence of 2 distinct ventricles, - Relative symmetry of right and left atria and ventricles
4 chamber: Pulsed-wave and color Doppler (axial)	- Presence of 2 distinct ventricles - Diastolic flow from the right and left atria into the right and left ventricles, respectively - Absence of tricuspid regurgitation
3 vessel trachea view: color Doppler	- Direction of blood flow in the aorta and pulmonary artery - Size of great vessels - Great vessels pointing to the left side
Ductus venosus: Pulsed-wave and color Doppler	- Antegrade ductus venosus a-wave

Abbreviation: ISUOG, International Society of Ultrasound in Obstetrics and Gynecology.

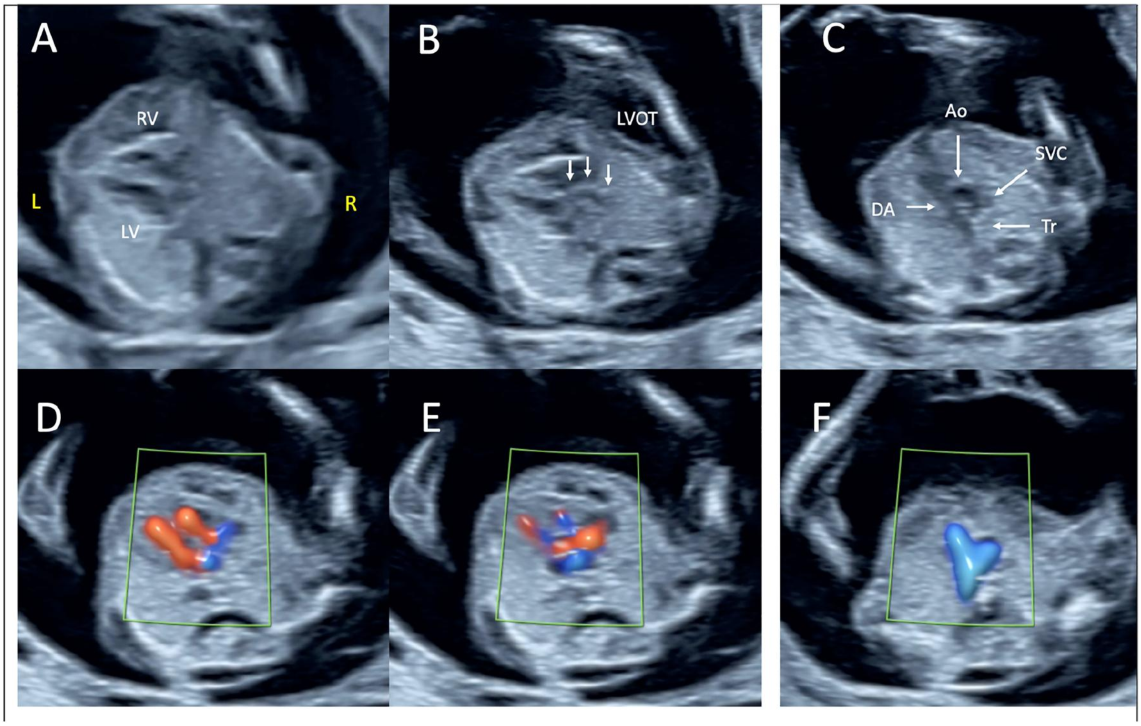

FIGURE 1 First trimester ultrasound aspect of a normal heart. 4CHv: heart located slightly left with apex pointing to the left at an angle of 45° (+/−15°), occupies one third of the thorax, symmetrical size of atria and ventricles, presence of the interventricular septum, insertion of AV valves (crux of the heart) (A); Passage of flow through AV valves and a parallel, symmetrical ventricular filling, absence of tricuspid regurgitation (D); LVOT (aorta) pointing towards the right on gray scale and color Doppler (B and E); 3VTv: Ductal arch, aortic arch and superior vena cava (C) and V-shape configuration on the isthmus, pointing left to the trachea (F). 3VTv, three vessels trachea view; 4CHv, four chamber view; AV, atrio-ventricular; LVOT, left ventricular outflow tract. [Colour figure can be viewed at wileyonlinelibrary.com]

exceeds 90% for some of the more complex cardiac anomalies.[56–58] Although the cardiac axis deviation has not been studied as extensively as NT, ductus venosus (DV), or tricuspid regurgitation (TR), it appears promising as a strong screening tool for CHDs. Its identification during the first trimester ultrasound should prompt a dedicated fetal echocardiography.

Assessment of the 4CHv as such does not differ from the examination performed in the second trimester, except for the smaller size of the individual cardiac structures. Grayscale imaging displays the symmetrical size of the cardiac ventricles, the presence of the interventricular septum, the level of insertion of the mitral and tricuspid valves (crux of the heart) as well as their mobility. Color Doppler shows the passage of flow across the AV valves and a parallel, symmetrical filling of both ventricles (see Figure 1).

Tricuspid regurgitation can be observed in the presence of retrograde flow and confirmed on pulsed waved Doppler by positioning a 2–3 mm sample across the tricuspid valve with a maximum angle of 30°. Under physiological conditions, the peak systolic velocity should not exceed 60 cm/s and the jet should last less than half of the systole. TR is present in a high proportion of fetuses with aneuploidy but in chromosomally normal fetuses the presence of TR is associated with an eight-fold increased risk of CHD.[59,60] Almost one third of fetuses with a major CHD presented with TR during the 11–14 weeks ultrasound, compared to less than 2% of those without a heart defect.[47,61]

5.2 | Outflow tracts

Due to their small size, outflow tracts are difficult to examine during the 11–14 weeks scan, though under good sonographic conditions, similar images to the mid-trimester examination can be obtained. All aspects such as the connections to the heart chambers, the size, position and crossing of the great vessels are detectable at the end of the first trimester (specifically with the use of color Doppler mapping). Outflow tracts can be visualized by progressively tilting the ultrasound probe from the 4-chambers towards the fetal head. Given the small size of the heart structures, the movements should be of minimal amplitude. The aorta originates from the left (posterior) ventricle and points towards the right hemithorax. It is crossed by the main pulmonary artery, which emerges from the right (anterior) ventricle that points towards the left hemithorax. The branching of pulmonary arteries can be brough into view with lowering of the pulse rate frequency and adequate color Doppler gain settings. Finally, the three-vessel trachea (3VT) view is seen even more caudally and displays, from left to right, the ductal arch, transverse aortic arch and superior vena cava. Both the Ductal and aortic arches can be seen on both gray-scale and color Doppler. Both vessels are of similar size and cross the mediastinum left to the trachea before joining at the level of the isthmus, forming a typical V-shaped configuration (see Figure 1).

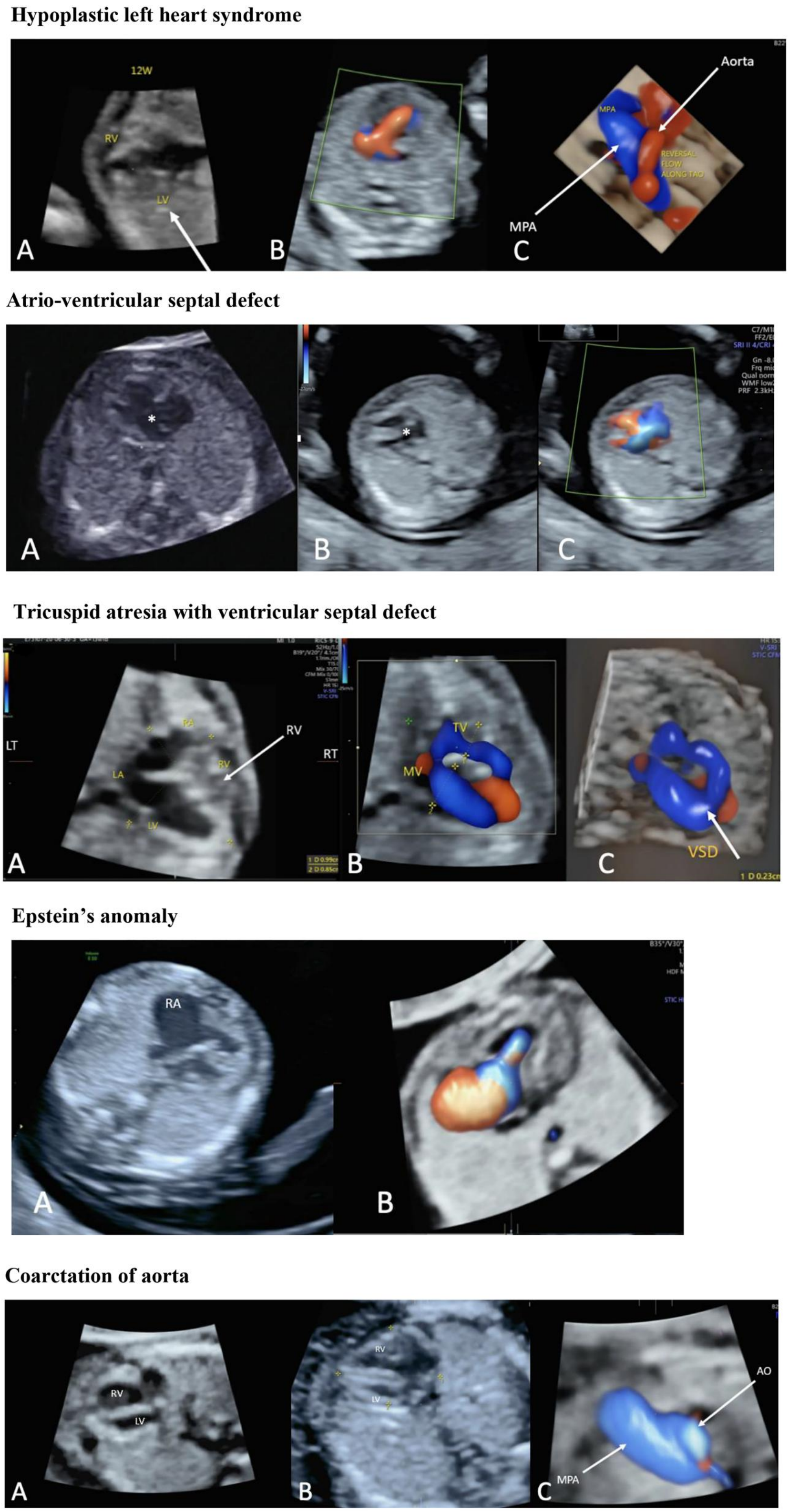

FIGURE 2 First trimester appearance of selected cardiac anomalies identified on the 4-chamber view. Hypoplastic left heart syndrome: Underdeveloped left ventricle, the aspect of an univentricular heart on 4CHv (A); Absent filling of left ventricle on color Doppler (B); Large MPA and ductal arch, hypoplastic aortic arch with reverse flow on Color Doppler (C). Atrio-ventricular septal defect: Large defect in the centre of the heart and absent crux (*) (A, B); Single channel of blood filling both ventricles through a common AV valve; common AV valve regurgitation on Color Doppler (C). Tricuspid atresia with ventricular septal defect: Small, hypoplastic RV (level of ventricular disproportion depends on size of the VSD which is always present) (A); 4CHv Color Doppler: the appearance of a univentricular heart with absent flow through the tricuspid valve; (C) 3VTv: small pulmonary artery with anterograde flow. Epstein's anomaly: Enlarged right atrium visible on 4CHv, the level on tricuspid valve insertion may be difficult to establish during the first trimester (A); tricuspid valve regurgitation with turbulent flow within the enlarged right atrium on color Doppler (B). Coarctation of aorta: Asymmetrical aspect of 4CHv with a small left ventricle (A, B); Hypoplastic aortic arch and forward flow on 3VTv with color Doppler (C). 3VTv, three vessels trachea view; 4CHv, Four Chamber view; AV, atrio-ventricular; MPA, main pulmonary artery; RV, right ventricle; VSD, ventricular septal defect. [Colour figure can be viewed at wileyonlinelibrary.com]

5.3 | Ductus venosus

Given the strong association between an abnormal Doppler flow in the DV and cardiac malformations, it is relevant to include DV assessment in the first trimester cardiac scan. A normal DV waveform is biphasic and constantly positive. Abnormal flow is present when the a-wave, which corresponds to the atrial contraction, displays an increased pulsatility index or a retrograde flow. It is seen in 1/3 of euploid fetuses with a major CHD.[62]

Tetralogy of Fallot

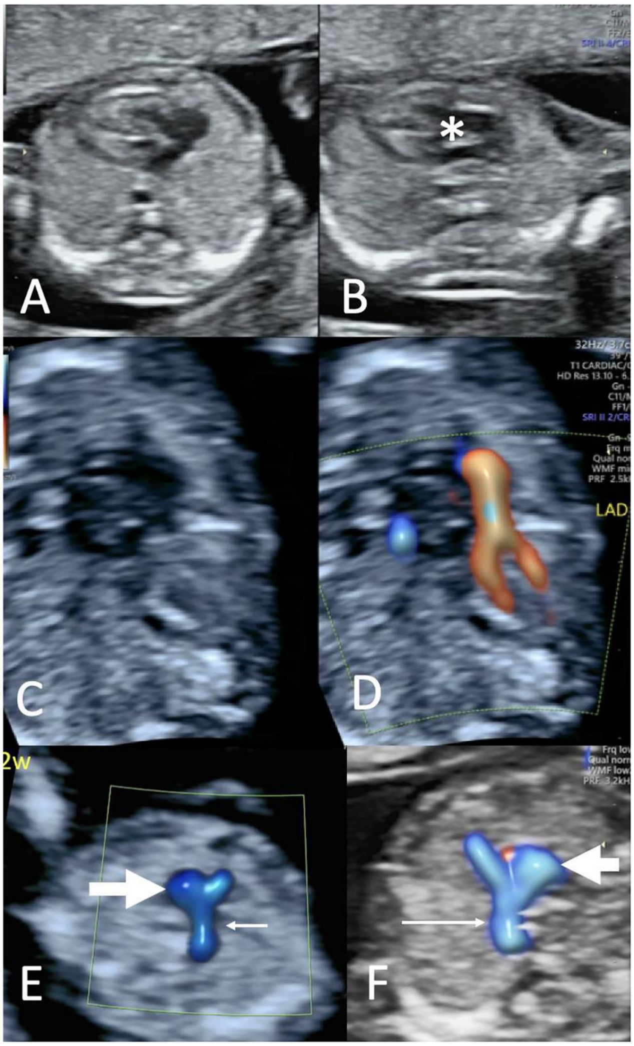

FIGURE 3 First trimester aspect of selected cardiac anomalies identified on an abnormal 3 vessel trachea view and likely normal 4 chamber view. Tetralogy of Fallot: Cardiac axis deviation on 4CHv (A); Aorta over-riding the interventricular septum (B, C); aortic filling arising from both left and right ventricle (D); Aorta (thin arrow) and ductal arch with preserved anterograde flow on color Doppler and abnormal aspect of the isthmus ("Y" sign) (E, F). D-Transposition of great arteries: Two instead of three vessels seen on 3VTv with reverse curvature of the right outflow tract (aorta = thick arrow) displaying a typical "boomerang sign" (A, C, D) and normal 4CH view (B); Parasagittal view of the heart: aorta (thick arrow arising from the RV and main pulmonary artery from the left ventricle (E). Common arterial trunk: Type 1 common arterial trunk displayed on color Doppler and 3D renders (A, B); 3D render of complex common arterial trunk arising from the RV (C). Abnormal positioning of the great vessels: Right aortic arch: Ductal arch (thick arrow) on the left and transverse aortic arch (thin arrow) on the right of the trachea (*) on gray scale (A) and color Doppler (B); "U-shaped" appearance of 3VTview (C, D); Double aortic arch: Ductal arch (thick arrow), division of the aortic arch (thin arrows) into two, trachea in the middle (*) (E, F): 3VTv, three vessels trachea view; 4CHv, four Chambers view. [Colour figure can be viewed at wileyonlinelibrary.com]

D-Transposition of great arteries

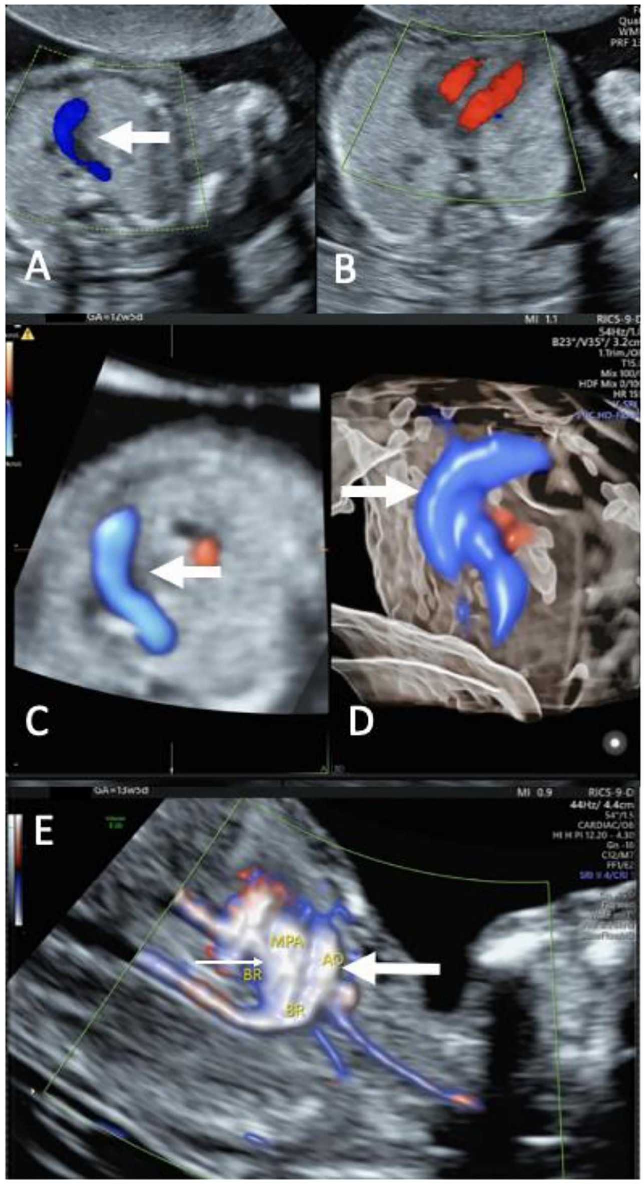

FIGURE 3 (Continued)

5.4 | Beyond screening

A dedicated fetal echocardiography is performed on selected patients based on the presence of risk factors or an abnormal finding during a first trimester screening ultrasound. It aims not only to confirm the presence of a CHD when suspected but also to differentiate the type of defect and, if possible, to specify its prognosis. It is expected during such an expert ultrasound assessment that all cardiac and vascular structures are meticulously examined in greyscale and color Doppler, in a similar way to the second or third trimester.[5]

Common arterial trunk

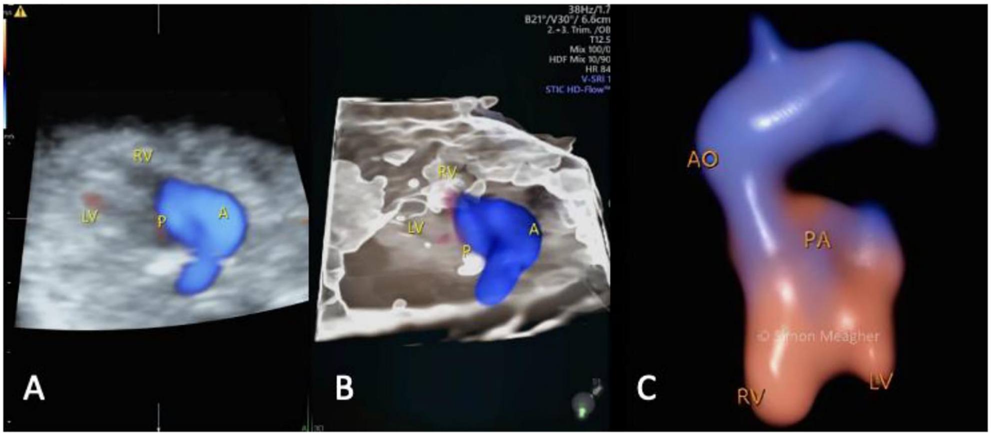

FIGURE 3 (Continued)

6 | HOW TO RECOGNIZE A HEART ANOMALY IN THE FIRST TRIMESTER

One third of CHDs are considered severe (major) and significantly impact perinatal mortality and morbidity. Identification of cardiac malformation may be divided into different groups: always detectable, potentially detectable and never detectable in the first trimester.[63] In experienced hands conditions such as tricuspid or pulmonary atresia, hypoplastic left heart syndrome and AV septal defect (AVSD) have reported detection rates of over 90% during the first trimester.[2,6,53] Not surprisingly, those highly detectable anomalies have a similar appearance in the first trimester to the mid-trimester scan or have very distinct ultrasound features mostly identified on the 4CHv of the heart.[64,65] On the other hand, the identification of CHDs, such as cono-truncal anomalies, remains challenging at this stage of pregnancy. Some major anomalies such as pulmonary and aortic stenosis may seldom, if ever, be detected during the first trimester given their evolving nature and late phenotypical presentation. Minor anomalies such as small ventricular septal defects are also challenging given their small size in association with prominent fetal activity present frequently so early in the pregnancy.[2]

It is relevant to mention that some anomalies present with a varied spectrum of severity. In TOF for instance, outcomes are related to the caliber and growth velocity of the right outflow tract. These factors, however, have only been shown to be predictive in later stages of pregnancy as progressive narrowing of the pulmonary artery seen in 25% of cases.[66] At the most severe end of the spectrum, pulmonary artery may be atretic (pulmonary atresia with intact septum) and blood flow thought the ductus arteriosus display a retrograde flow while a very large pulmonary artery should rise the suspicion of TOF with an absent pulmonary valve. Other pathologies, such as a common arterial trunk, present as a heterogeneous group of complex malformations and whose exact nature may be difficult to define in the first trimester. It is beyond the scope of this article to describe all anomalies. Figures 2 and 3 summarize a selection of the few most typical anomalies whose diagnosis in the first trimester may have relevance for the further course of the pregnancy.

7 | CONCLUSION

For the majority of fetal cardiac abnormalities, the sonographic appearances at 11–14 weeks are not different from the well-recognized landmarks of malformation at the mid-trimester. In many respects, the challenges over recent decades have been related to the small cardiac size at this early gestation. However, these limitations have been overcome in recent years with the advances in 2D and color Doppler technologies. With adequate training and strict protocols, the reported detection rates of cardiac malformation during the first trimester now approximate those at the mid trimester. However, it is of crucial importance that in this evolving field of fetal medicine that checks, and balances are in place, particularly in the setting where TOP is being considered and where the final arbiter a post-mortem is not available. Strick audit of pregnancy outcomes and the involvement of fetal or pediatric cardiologists is key in achieving a necessary standard of practice with the aim of maintaining false positive and false negative detection rates in line with those reported for the second trimester. The recent ISUOG guidelines reflect this doctrine and outline the importance of recognizing the varied skill set amongst practitioners in this field and have suggested the stratification of imaging at 11–14 weeks into a basic assessment or a more detailed fetal anatomical review. This is a significant step forward and with these new guidelines the evaluation of the 4-chamber view of the heart and three vessel tracheal view with both 2D and color Doppler examination now paves the way for improving detection rates early in the pregnancy. Patients and their families can avail themselves of the many clinical advantages that come with this shift in screening for

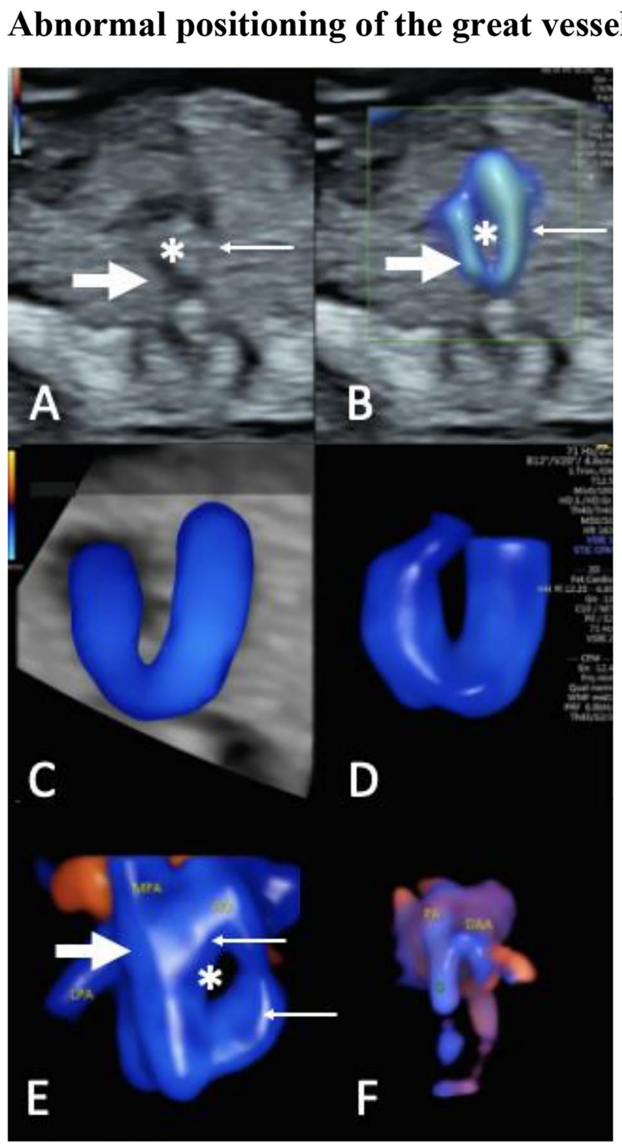

FIGURE 3 (Continued)

fetal malformations so early in the pregnancy and benefit from the reassurance that may be provided to the majority where the fetal anatomy including the cardiac assessment is deemed normal.

ACKNOWLEDGMENTS

The authors thank prof. Lisa Hui for her careful review of the article.

Open access funding provided by Universite de Lausanne.

CONFLICT OF INTEREST STATEMENT

All authors declare that they have no conflicts of interest.

DATA AVAILABILITY STATEMENT

Not applicable.

ORCID

Wawrzyniec Rieder https://orcid.org/0000-0002-1762-8609

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How to cite this article: Rieder W, Eperon I, Meagher S. Congenital heart anomalies in the first trimester: from screening to diagnosis. *Prenat Diagn*. 2023;43(7):889-900. https://doi.org/10.1002/pd.6391