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Evaluation of first-trimester ultrasound screening strategy for fetal congenital heart disease

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KEYWORDS: congenital heart disease; fetus; first trimester; prenatal ultrasonography

ABSTRACT

Objective *To assess the performance of a standardized first-trimester ultrasound screening strategy for fetal congenital heart disease (CHD).*

Methods *This was a large retrospective study involving 74 839 consecutive mixed-risk pregnancies (77 396 fetuses). Routine ultrasound scans at 11 + 0 to 13 + 6 weeks' gestation were performed in a single center from January 2015 to June 2023. All fetuses were examined using a predefined standardized ultrasound scanning strategy with adjustment of imaging parameters, which included assessment of the fetal heart. The ultrasound results (e.g. extracardiac congenital malformations), ultrasound markers (e.g. nuchal translucency thickening, reversed a-wave in the ductus venosus and tricuspid regurgitation), follow-up, genetic tests and diagnostic results were recorded and analyzed.*

Results *In total, there were 831 cases of CHD, with an incidence of 1.07% (831/77 396). In the first-trimester scan, 590 fetuses were diagnosed with CHD, but four were confirmed as normal in later examinations. In addition, 245 cases were missed. The detection rate was 70.52%, with a sensitivity, specificity, false-positive rate and false-negative rate of 70.52%, 99.99%, 0.01% and 29.48%, respectively. In fetuses with negative ultrasound markers and no extracardiac malformations, the detection rate of CHD was 45.79% (185/404). There were 281 cases that underwent karyotyping and chromosomal microarray (245 fetuses) or whole-exome sequencing (36 fetuses). In total, 38.79% (109/281) had a positive genetic test result. There were 273/831 CHD cases associated with extracardiac malformations. The abnormal image patterns and abnormal features of each view in the scanning strategy were summarized.*

Conclusions *Ultrasound screening for fetal CHD in the first trimester of pregnancy enables earlier prenatal diagnosis and consultation. The standardized ultrasound screening strategy used in this study had a high detection rate for fetal CHD in the first trimester. Our proposed fetal heart screening strategy shows promising effectiveness for early diagnosis of CHD and we recommend its use. It is important to note, however, that first-trimester ultrasound screening for fetal CHD should not replace fetal echocardiography in the second trimester.* © *2025 International Society of Ultrasound in Obstetrics and Gynecology.*

INTRODUCTION

Congenital heart disease (CHD) describes a subset of fetal morphological malformations, with an incidence of 1% of all live births[1]. However, the incidence of CHD in the first trimester is higher, particularly in fetuses with ultrasound markers (such as nuchal translucency (NT) thickening, reversed a-wave in the ductus venosus and tricuspid regurgitation), extracardiac abnormalities, fetal edema and chromosomal abnormalities. Fetuses with multiple malformations combined with CHD often die *in utero* before 18 weeks' gestation, so the actual incidence of CHD in the first trimester may be five times higher than that in newborns[2–5]. Fetal CHD screening is recommended at 18–24 weeks' gestation[6,7]. For high-risk pregnancies, screening has been brought forward to the first trimester. The early detection of CHD is of utmost importance, not only for parental consultation, but also for best practice/physician decision-making. Unfortunately, up to 50% of major CHD cases are still misdiagnosed[8–10]. In recent years, first-trimester ultrasound screening for CHD in low-risk pregnancies has been carried out in some

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high-income countries or specialized medical centers, achieving satisfactory results[11,12]. In this study, we aimed to assess the feasibility and efficiency of a standardized first-trimester ultrasound screening strategy with suggested imaging parameters for the diagnosis of fetal CHD.

METHODS

Study population

This was a retrospective single-center study including consecutive mixed-risk pregnancies that underwent routine first-trimester (11 + 0 to 13 + 6 weeks' gestation) ultrasound scans at the Maternal and Child Healthcare Hospital of Guangxi Zhuang Autonomous Region, Nanning, China. The data collection period spanned from January 2015 to June 2023, and included low- and high-risk pregnancies and external hospital referrals. All pregnant women participating in the study provided written informed consent and the study was approved by the institutional review board of the Maternal and Child Health Hospital of Guangxi Zhuang Autonomous Region (File No. 20194).

Ultrasound examination

All ultrasound scans were conducted by certified physicians, who had obtained The Fetal Medicine Foundation NT certification. A Canon Aplio 500 (Canon Medical Systems, Otawara, Japan) ultrasound machine, equipped with a P-674 (3.0–10.0-MHz) transabdominal probe, and a GE Voluson E10 (GE Healthcare, Zipf, Austria) machine, equipped with a C1-5 (2.0–5.0-MHz) transabdominal probe, were used. Mechanical and thermal index settings were both adjusted to < 1.0.

All fetuses underwent screening using our standard first-trimester fetal ultrasound protocol[13]. In the fetal heart screen, the following views were obtained: (1) the axial view of the fetal upper abdomen, (2) the four-chamber view (4-CV) on color Doppler and (3) the three-vessel-and-trachea view (3-VT) on color Doppler. We evaluated the apical direction in the 4-CV on color Doppler instead of the cardiac axis. Adjustment of imaging parameters was essential to obtain high-quality images; in order to balance the blood flow filling and spatial resolution, and to acquire the best image, the ultrasound incidence angle should be adjusted appropriately. For example, the oblique 4-CV, which is between the apical and parasternal views, was usually optimal.

When CHD was suspected, the following views and morphology were examined as closely as possible: (1) 4-CV on grayscale ultrasound; (2) 3-VT on grayscale ultrasound; (3) left ventricular outflow tract (LVOT) view; (4) right ventricular outflow tract (RVOT) view; (5) long-axis view of the aortic arch on color Doppler; (6) sagittal plane bicaval view on color Doppler and (7) pulmonary vein examination. A senior physician (S.Y. or G.Q.) then conducted a re-examination to confirm the diagnosis.

The basic imaging parameter settings for each machine are listed in Table 1. Further adjustment was made based on the gravidas' condition when obtaining images. To better observe the tiny heart morphology, the following settings were crucial for acquiring high-quality images: preset the depth to 8–10 cm; range of fan scanning should be slightly larger than the chest by 5–10° and zoom-in on the fetal thorax so that the structure is centered and occupies more than two-thirds of the image. For color Doppler, the preset speed was 19–24 cm/s and the frame frequency of grayscale and color/power Doppler should be greater than 50 and 25 frames/s, respectively. In addition, attention should be given to adjusting the interface and ultrasound incidence angle for morphology that requires close observation.

Genetic testing, follow-up and pregnancy outcome

Prenatal consultation and genetic tests were recommended in fetuses with suspected CHD. For women who decided to continue the pregnancy and deliver, repeat ultrasound examinations were conducted in the mid-trimester and postpartum. Pathological examination was conducted in cases of termination of pregnancy (TOP), miscarriage or stillbirth. Fetuses with normal ultrasound results at 11 + 0 to 13 + 6 weeks' gestation underwent mid-trimester screening at 20–24 weeks, and the outcome of these pregnancies was followed up.

In liveborn cases with suspicion of CHD, the final diagnosis of CHD was based on surgical results or the last fetal/neonatal echocardiography examination. For pregnancy termination, miscarriage or stillbirth, the final diagnosis was based on the pathological examination or the last echocardiography examination (if the pathological examination was declined).

Statistical analysis

Continuous variables were expressed as mean ± SD. The detection rate was expressed as a percentage and was

Table 1 Basic imaging parameter settings for first-trimester fetal congenital heart disease screening, according to ultrasound machine used

Parameter	*Canon Aplio 500*	*GE Voluson E10*
Acoustic output (%)	< 90	< 90
Scan range/angle (%)	45	45
Depth (cm)	10	10
Brightness frame rate (fps)	> 50	> 50
Brightness dynamic range*	55	—
Dynamic contrast†	—	7
Brightness gain	80	6
Zoom	1.2	1.4
Color/power frequency (MHz)	3.8	Middle
Color PRF (kHz)	7.8	2.0
Color/power frame rate (fps)	> 25	> 25
Color/power color gain	35	−2.0
Color/power scale (cm/s)	19	24

*Canon Aplio 500 terminology. †GE Voluson E10 terminology. fps, frames per second; PRF, pulse repetition frequency.

calculated as the number of CHD fetuses detected by ultrasound examination divided by the total number of CHD fetuses. Increased NT was defined as a value greater than the 95th percentile in normal fetuses at the same gestational age. The rate of positive genetic test results was computed. The sensitivity, specificity, false-positive rate and false-negative rate were calculated. Statistical analyses were performed using the Statistical Package for the Social Sciences 25.0 software (IBM, Armonk, NY, USA).

RESULTS

Study population

A total of 74 839 women who underwent first-trimester ultrasound scanning were included in the study. The average maternal age, weight and body mass index at scanning were 30.5 ± 4.5 (range, 15–51) years, 53.4 ± 8.0 (range, 35–86) kg and 21.3 ± 2.9 (range, 15.1–36.7) kg/m^2, respectively. Of these pregnancies, 72 365 were singleton, 2399 were twin, 67 were triplet and 8 were quadruplet, totaling 77 396 fetuses.

In this study, ultrasound markers were positive in 3447 fetuses. Morphological malformations were found in 1597 cases, including 831 cases with CHD (at least one type of CHD). Among these cases of CHD, 771 were singleton and 60 were from multiple gestations.

In our mixed-risk pregnancies, the incidence of CHD was 1.11% (831/74 839), and CHD accounted for 52.04% (831/1597) of all fetal morphological malformations. Among fetuses with multiple malformations and those with ultrasound markers present, 17.09% (273/1597) and 10.18% (351/3447) were diagnosed with CHD, respectively (Table S1). Isolated CHD accounted for 48.62% (404/831) fetuses and the detection rate was 45.79% (185/404) (Table S2).

Genetic testing, follow-up and pregnancy outcome

There were 281 cases that underwent karyotyping and chromosomal microarray (245 fetuses) or whole-exome sequencing (36 fetuses). In total, 38.79% (109/281) had a positive genetic test result, including 16 cases of trisomy 21, 23 cases of trisomy 18, 22 cases of trisomy 13, 21 cases of 45X, three cases of triploid, one case of trisomy 7, one case of trisomy 22, one case of 47XXX, seven cases of 22q11.2 deletion and 14 cases of other pathogenic microdeletions, microduplications or gene abnormalities.

Among the 831 fetuses with CHD, there were 13 cases of intrauterine death or miscarriage, 36 underwent selective reduction, 549 underwent TOP, and 233 were delivered spontaneously (including two postnatal deaths before 1 year of age). All 36 cases of selective reduction and 438/549 cases of TOP were terminated before 18 weeks' gestation. Moreover, 451 fetuses underwent repeat ultrasound examinations in the second trimester. A pathological examination was performed in 79 fetuses after TOP. Neonatal echocardiography was performed in 235 cases. There were 507 fetuses that underwent repeated ultrasound examination or cardiac pathology examination. In total, 328 fetuses with severe malformations were directly reduced or underwent TOP without repeated examination, and the parents declined cardiac pathology examination.

Diagnostic efficiency

On first-trimester ultrasound examination, 590 fetuses were diagnosed with CHD, four of which were confirmed to be normal in later examinations. In addition, a missed diagnosis was encountered in 245 cases. Based on these data, the detection rate of fetal CHD in the first trimester was 70.52% (586/831), with a sensitivity, specificity, false-positive rate and false-negative rate of 70.52%, 99.99%, 0.01% and 29.48%, respectively. In fetuses with negative ultrasound markers and no extracardiac malformations, the detection rate of CHD was 45.79% (185/404). The specific CHD diagnoses in these populations are shown in Tables S1 and S2.

Ultrasound image patterns and diagnostic features

Standard first-trimester ultrasound screening involved the following. (1) Axial view of the fetal upper abdomen on grayscale and color Doppler ultrasound: in this view, observation focused on the location of the stomach and the relationship between the abdominal aorta and inferior vena cava. Usually, the stomach is located in the upper left abdomen. Location of the stomach on the right, the midline or in the chest, absent inferior vena cava, or the juxtaposition of the abdominal aorta and inferior vena cava were abnormal manifestations (Figure 1 and Videoclips S1–S3). (2) 4-CV on color Doppler: in this view, abnormal manifestations included abnormal cardiac position, blood flow from the atrium to the ventricle, abnormal ratio of the left and right blood flow width, Y-shaped blood flow, moderate or severe tricuspid regurgitation, mitral regurgitation and common atrioventricular regurgitation (Figure 2). (3) 3-VT on color Doppler: abnormal manifestations in this view included abnormal blood flow width ratio, single arterial blood flow, abnormal blood flow direction or morphology and detection of superior vena cava blood flow (Figure 3).

Although many major CHD have abnormal manifestations in the three views mentioned above, in order to confirm the diagnosis of suspicion of CHD, second-trimester echocardiography was required to provide more detailed information. However, acquisition of the additional views in the first trimester was feasible, though often challenging.

(1) 4-CV on grayscale: obtaining the 4-CV on grayscale was more challenging than on color Doppler (Figure S1a). However, it may be considered as supplementary, showing more detail and confirming the diagnosis. The parasternal 4-CV had the strongest interface reflection at the ventricular wall and interventricular septum interface, and at this position, the clearest image was acquired (Figure S1b). In this view, abnormal atrioventricular

septum, single ventricle, significantly abnormal proportion of ventricle width and enhanced echogenicity of ventricular endocardium were the abnormal manifestations observed (Figure S1c–j). (2) 3-VT on grayscale: in this view, when the spine was at 6 o'clock, the interface reflection was the weakest as the angle between the acoustic beam and the aortic arch and pulmonary artery was the smallest, and this view on grayscale was unclear (Figure S2a). The grayscale image was usually apparent when the spine was at 3 or 9 o'clock, as the angle between the acoustic beam and great vessels was the largest (Figure S2b). The single artery, abnormal proportion of large artery width and increased blood vessel numbers may be considered as abnormal manifestations in this view (Figure S2c–h). (3) LVOT view: when abnormalities were found in the 3-VT on color Doppler, further scanning of the LVOT was necessary (Figure S3a–c). Similarly, increasing the interface reflection between the acoustic beam and the ventricular wall, interventricular septum and large artery wall was recommended to improve the quality of grayscale images. In this view, conus arteriosus malformations could be differentiated further by evaluating the relationship between the aorta and pulmonary artery (Figure S3d,e). (4) RVOT view: this view was acquired by deflecting towards the cranial side from the LVOT view. The grayscale imaging of the parasternal RVOT view provided better interface reflection than the level short-axis view of the great vessels (Figure S4a,b). Pulmonary artery stenosis, reversed flow, severe regurgitation, the parallel origin of the aorta and pulmonary artery from the right ventricle were considered abnormal manifestations (Figure S4c–g). (5) Long-axis view of the aortic arch on color Doppler: a normal aortic arch on grayscale ultrasound imaging was often difficult to observe. Moreover, the views of blood flow through the aortic arch and the ductus arteriosus overlap one another on color Doppler (Figure S5a,b). Based on the spatial positional relationship, the blood flow of the aortic arch on the cranial side and the blood flow of the ductus arteriosus on the caudal side could be roughly distinguished. Reversed blood flow in the aortic arch and the ductus arteriosus were abnormal manifestations, and the type of CHD may be inferred based on the position of reversed blood flow (Figure S5c,d). (6) Sagittal plane bicaval view on color Doppler: grayscale imaging in the bicaval view was challenging, but color Doppler showed a satisfactory result (Figure S6a). Interruption of the inferior vena cava and complete filling of the azygos or semi azygos vein connected to the superior vena cava were abnormal manifestations (Figure S6b). (7) Pulmonary vein: color Doppler imaging of the pulmonary vein in fetuses at 11 + 0 to 13 + 6 weeks' gestation was challenging, but was improved by use of low-speed blood flow imaging technology (Superb Microvascular Imaging silhouette mode) (Figure S7a). However, diagnosing isolated anomalous pulmonary

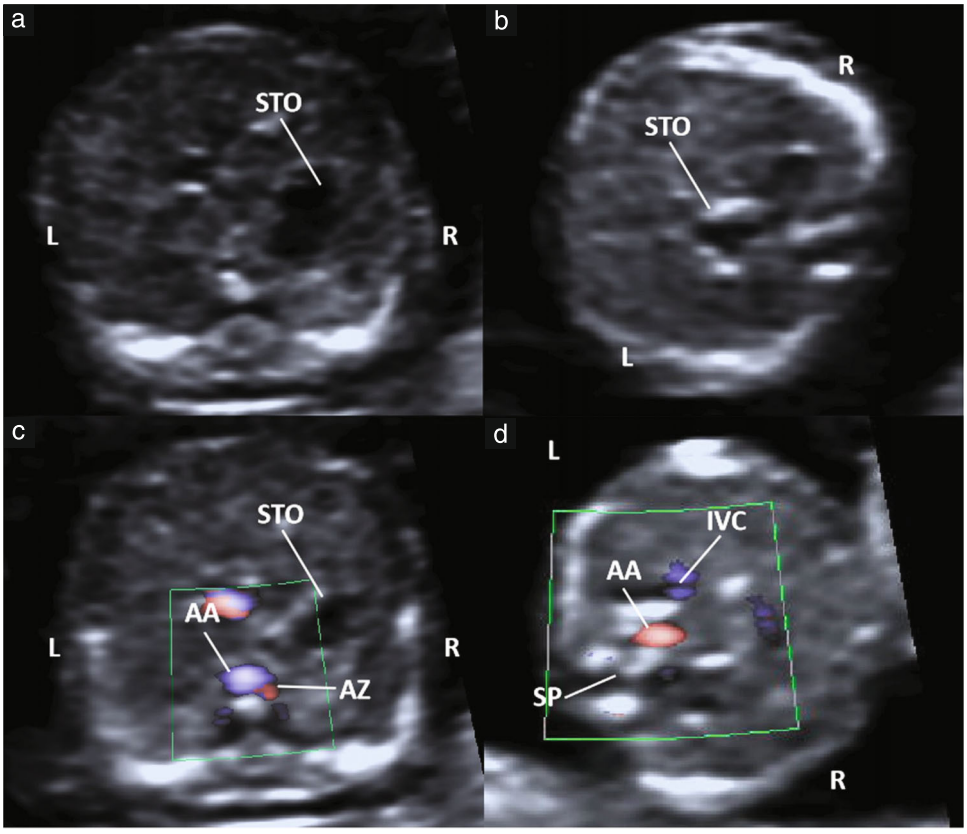

Figure 1 Grayscale and color Doppler ultrasound images in first trimester, showing: (a) fetal stomach (STO) on right; (b) fetal STO on midline; (c) inferior vena cava disconnection and azygos vein (AZ) dilation; (d) abdominal aorta (AA) and inferior vena cava, both located on left side of spine (SP). IVC, inferior vena cava; L, left; R, right.

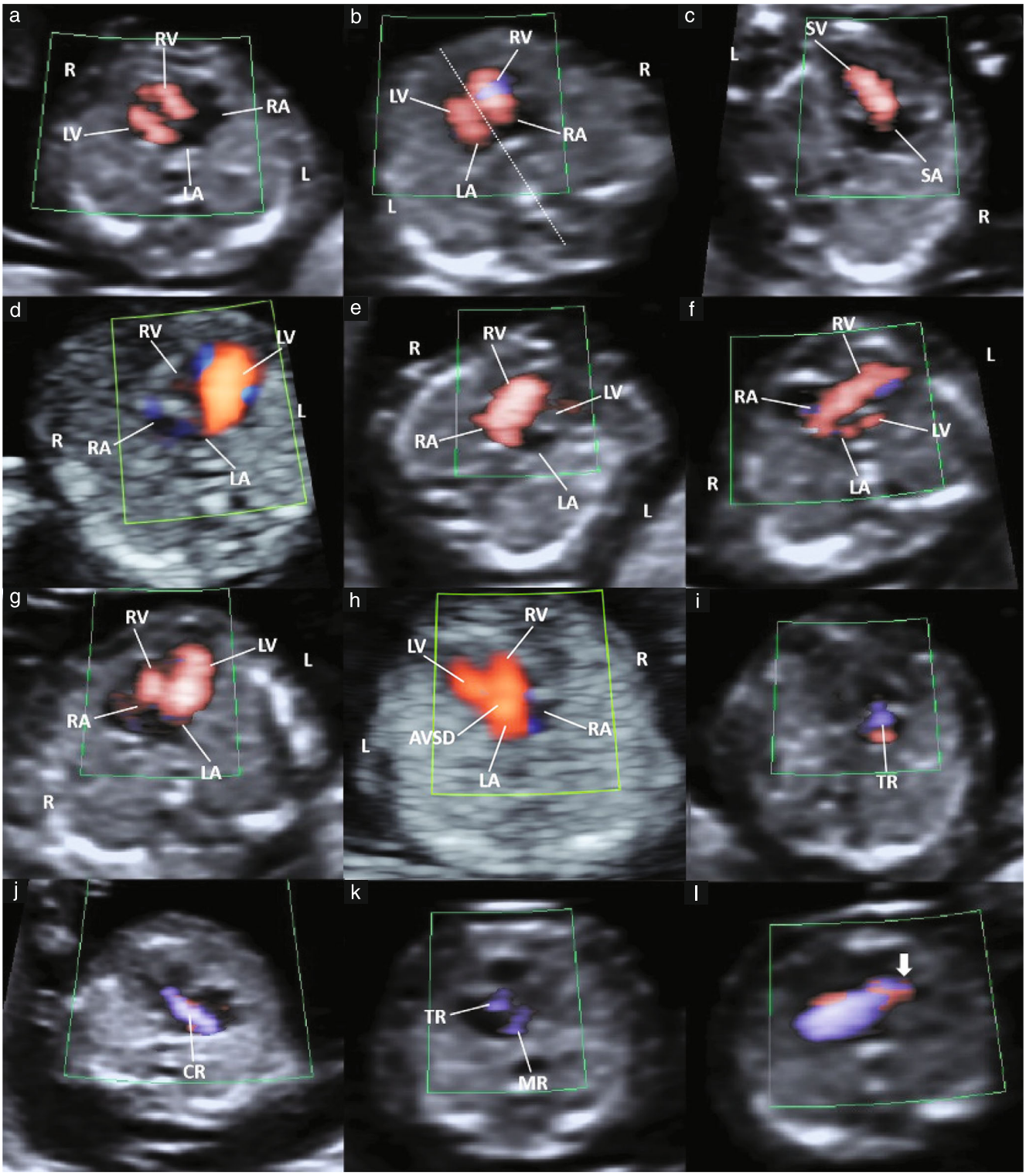

Figure 2 Four-chamber view on color Doppler imaging in first trimester, showing various types of congenital heart disease: (a) mirror-image dextrocardia; (b) mesocardia; (c) single ventricle (SV) and single atrium (SA), with blood flow passing through center of heart; (d) tricuspid valve atresia with only left-sided blood flow seen; (e) mitral valve atresia with only right-sided blood flow seen; (f) narrowed diastolic flow from left atrium (LA) into left ventricle (LV); (g) narrowed diastolic flow from right atrium (RA) into right ventricle (RV); (h) complete atrioventricular septal defect (AVSD) with Y-shaped blood flow; (i) moderate tricuspid regurgitation (TR); (j) common atrioventricular regurgitation (CR); (k) moderate mitral regurgitation (MR) and TR; and (l) origin of TR shifted significantly downward (arrow). L, left; R, right.

venous drainage was almost impossible in the first trimester[12]. Right atrial isomerism syndrome was often associated with anomalous pulmonary venous drainage. The common pulmonary vein is shown in Figure S7b–d.

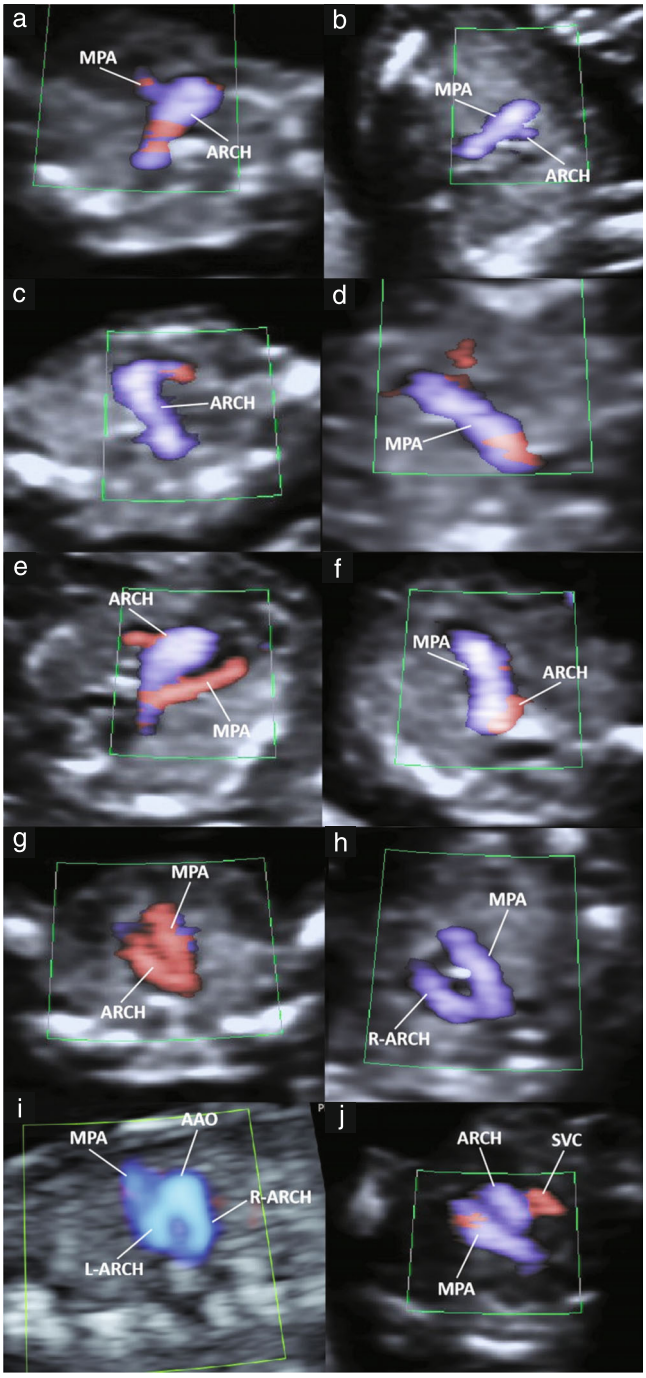

Figure 3 Three-vessel-and-trachea views on color Doppler imaging in first trimester, showing various types of congenital heart disease: (a) narrowed main pulmonary artery (MPA) blood flow; (b) narrowed aortic blood flow; (c) only aortic blood flow observed; (d) only MPA blood flow observed; (e) MPA stenosis with reversed blood flow; (f) aortic reversed blood flow; (g) MPA and aortic reversed blood flow; (h) U-shaped blood flow pattern; (i) O-shaped blood flow pattern; (j) superior vena cava (SVC) blood flow. AAO, ascending aorta; ARCH, aortic arch; L-ARCH, left aortic arch; R-ARCH, right aortic arch.

DISCUSSION

Main findings

In the past, the assessment of cardiac anatomy was not considered part of the routine ultrasound assessment in the first trimester[14]. In 2020, the American Institute of Ultrasound in Medicine (AIUM) guidelines first recommended using color Doppler 4-CV and 3-VT to screen for CHD[15]. In 2023, the International Society of Ultrasound in Obstetrics and Gynecology (ISUOG) updated their guidelines, which recommended that the color Doppler and grayscale scanning of fetal 4-CV and 3-VT were the best practice protocol[16]. It was believed that adding these views could significantly improve the detection rate of CHD[15,16]. Screening for fetal CHD in low-risk pregnancies during the first trimester will inevitably become a routine procedure in the future[17]. Multiview scanning can achieve relatively accurate diagnoses for suspected CHD at 11 + 0 to 13 + 6 weeks' gestation[18–24]. In this study, we conducted standardized first-trimester ultrasound screening for fetal CHD in China and confirmed further diagnoses using multiview scanning. The standardized ultrasound screening strategy used in this study had a high detection rate for fetal CHD in the first trimester, thus enabling earlier prenatal diagnosis and consultation.

Comparison with the literature

First-trimester ultrasound markers, such as NT thickening, reversed a-wave in the ductus venosus and tricuspid regurgitation, are not only used to assess the risk of aneuploidy but are also closely associated with CHD. Minnella *et al.*[25] reviewed 93 209 fetuses and showed that 55.5% of major CHDs were accompanied by ultrasound markers. In that study, the incidence rates of NT thickening, venous catheter a-wave inversion and tricuspid regurgitation were 36.5%, 28.9% and 27.5%, respectively. These rates were higher than our results in terms of ultrasound markers being positive (42.24%; 351/831) and NT thickening (33.94%; 282/831). The reason may be that we included all, not just major, CHD. The results also demonstrate that there was a lower incidence of positive ultrasound markers for those with mild CHD *vs* those with major CHD.

We identified more CHD cases associated with ultrasound markers than with extracardiac malformations (351 *vs* 273). In this study, CHD accounted for 52.04% (831/1597) of all fetal morphological malformations, and multiple malformations involving CHD accounted for 17.09% (273/1597), which is higher than the rate of fetuses with positive ultrasound markers of 10.18% (351/3447).

Transabdominal ultrasound was the method we applied in all cases, though transvaginal ultrasound may acquire higher-quality images, especially in the first trimester. In most cases, satisfactory heart images may be obtained through the transabdominal method using our strategy.

Low acceptance of transvaginal fetal screening is another reason we chose the transabdominal method. Severe obesity is rare in the population of Southwest China. In this study, the average weight of gravidas was 53.4 ± 8.0 kg. If the mother's weight is likely to affect image quality, fetal heart scanning may be delayed to around 14 weeks' gestation.

The CHD screening in the first trimester was based on three views, which may not necessarily secure a diagnosis. However, the echocardiographic views used in the second trimester may improve diagnostic accuracy. Nevertheless, many structures in those views are challenging to recognize because of the small size of the heart, especially on grayscale ultrasound imaging. Therefore, adjusting the angle between the ultrasound beam and interface is crucial.

Although the blood flows were not well recognized in the horizontal view, the maximum ultrasound reflection at the interface of the region of interest was beneficial for observation on grayscale ultrasound, especially for 4-CV, LVOT, RVOT and 3-VT. Information acquired through these views was decisive in confirming the diagnosis. The LVOT and RVOT views on grayscale ultrasound may be acquired by deflection towards the cranial side from the parasternal 4-CV. Aortic straddling, pulmonary artery stenosis and the parallel origin of arteries were vital pathological features critical for the differential diagnosis of isolated conus arteriosus anomalies and were easy to recognize. Low-speed blood flow imaging was beneficial for recognizing the systemic veins and pulmonary veins, which was helpful for the diagnosis of some particular types of CHD (e.g. pulmonary vein anomalous connection). Using the summarized abnormal cardiac views for major CHD described in this study may have contributed to our high diagnostic accuracy.

Many researchers have explored first-trimester fetal heart screening. In the research of Persico *et al.*[26], the detection rate of major CHD was up to 93.1% with well-trained physicians carrying out the examinations. In the meta-analysis of Karim *et al.*[27], only 53.1% of CHD cases were detected in low-risk pregnancies, but the rate increased to 70.4% in those at high risk. Factors influencing the detection rate included operator error, technical factors and maternal obesity. These factors had a more significant influence on low-risk pregnancies[28,29]. However, almost all studies were focused on major CHD rather than all CHD.

Most screening strategies are based on the 4-CV and 3-VT. Although these two view patterns have been reviewed extensively in the literature[30–32], detection rates of most CHD using these patterns were below 50%[13,33]. The main reasons may be that many critical cardiac views, such as LVOT, RVOT or long-axis view of the aortic arch on color Doppler or grayscale ultrasound, have yet to be validated. Following our scanning strategy and abnormal image patterns, the sensitivity and specificity for detection of all CHD in this study reached as high as 70.52% and 99.99%, respectively. Meanwhile, the overall detection rate for CHD on first-trimester ultrasound screening reached as high as 70.52%. For cases with major CHD, such as transposition of the great arteries, tetralogy of Fallot, atrioventricular septal defect, single ventricle, Ebstein's anomaly, tricuspid atresia, pulmonary atresia, hypoplastic left heart syndrome and double-outlet right ventricle, the detection rate (average, 91.00%) was much higher than that reported in the literature. For cases with mild CHD, such as double aortic arch and right aortic arch, the detection rate also reached more than 84.00%. In addition, focusing on ultrasound parameter adjustment may improve the diagnostic accuracy and the detection rate of rare CHD, such as systemic venous and pulmonary venous abnormalities.

Approximately 30% of chromosomal and microarray abnormalities are associated with CHD[34]. The incidence of genetic abnormalities in the first trimester was higher, as many fetuses with chromosomal abnormalities may die *in utero*. In this study, only 281/831 fetuses underwent genetic testing, with a positive detection rate of 38.79% (109/281). The actual positive detection rate of genetic abnormalities may be even higher, as many fetuses with multiple malformations underwent TOP rather than further prenatal diagnosis.

First-trimester screening is used to detect abnormalities earlier and provide more time for prenatal consultation and decision-making. In this study, 100% (36/36) of selective reductions and 79.78% (438/549) of pregnancy terminations were performed before 18 weeks' gestation, shifting the clinical decision-making time forward considerably, and thereby reducing complications caused by these procedures at advanced gestational age.

Strengths and limitations

Our proposed new first-trimester fetal heart screening strategy represents a major strength; the three standard views of the heart are efficient in screening for major complex CHD, and further multiview scanning of cases with suspected CHD, using our summarized abnormal image patterns and abnormal features of each view, may achieve a more precise diagnosis. This offers a relatively reliable foundation for clinical decision-making. The main limitations are those inherent to the retrospective nature of the study and the lack of generalizability, given that our data were derived from a single-center Chinese population. Although multiview scans are very helpful for confirmation of CHD types that are difficult to differentiate (e.g. single ventricle, hypoplastic left heart syndrome, hypoplastic right heart syndrome, mitral atresia and tricuspid atresia), more severe conditions, such as tetralogy of Fallot, pulmonary atresia with ventricular septal defect and persistent truncus arteriosus, may not be identified. Due to the decision to terminate the pregnancy in most cases of severe CHD, postnatal confirmation was not always possible. Additionally, follow-up was based on the last fetal echocardiography examination, which may have resulted in a missed diagnosis of mild CHD.

Conclusions

Ultrasound screening for fetal CHD in the first trimester of pregnancy enables earlier prenatal diagnosis and consultation. This provides more time for parents and doctors to make clinical decisions and deliver treatment. In this study, we proposed and applied a new first-trimester fetal heart ultrasound screening strategy for CHD. Adjustment of image parameters and scanning angles was important for visualization of fetal cardiac morphology. Although cases of mild CHD, such as ventricular septal defect, may be missed with this strategy, it had high accuracy for major CHD. It is important to note, however, that first-trimester ultrasound screening for CHD should not replace fetal echocardiography in the second trimester.

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SUPPORTING INFORMATION ON THE INTERNET

The following supporting information may be found in the online version of this article:

Figures S1–S7 Evaluation of congenital heart disease (CHD) on additional ultrasound views in first trimester: four-chamber views on grayscale ultrasound (Figure S1); three-vessel-and-trachea views on grayscale ultrasound (Figure S2); left ventricular outflow tract views on grayscale ultrasound and color

Doppler (Figure S3); right ventricular outflow tract views on grayscale ultrasound and color Doppler (Figure S4); long-axis view of aortic arch on grayscale ultrasound and color Doppler (Figure S5); sagittal plane bicaval view on color Doppler (Figure S6); fetal pulmonary vein assessment on grayscale ultrasound and Superb Microvascular Imaging (Figure S7).

Table S1 Congenital heart disease diagnosed during first-trimester ultrasound screening in fetuses with ultrasound markers or extracardiac malformations

Table S2 Congenital heart disease diagnosed during first-trimester ultrasound screening in fetuses with negative ultrasound markers and no extracardiac malformations

Videoclip S1 Grayscale ultrasound videoclip corresponding to Figure 1a, showing fetal stomach on right side.

Videoclip S2 Color Doppler videoclip corresponding to Figure 1c, showing inferior vena cava disconnection and azygos vein dilation.

Videoclip S3 Color Doppler videoclip corresponding to Figure 1d, showing abdominal aorta and inferior vena cava, both located on left (L) side of spine.