



Multimodal Approach (MRI and Ultrasonography) to the Diagnosis of Fetal Congenital Heart Diseases

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Abstract

Introduction: As the rate of congenital heart diseases (CHD) remains high, medical imaging specialists face a task of early diagnosis of CHD with minimal cost and burden to pregnant women and fetuses and need to verify the prenatal diagnosis in order to develop a strategy for managing pregnant women carrying a fetus with CHD.

Objective: To optimize diagnostic measures in fetuses with CHD by comparing fetal echocardiography and cardiac magnetic resonance imaging (MRI).

Materials and methods: We retrospectively evaluated findings from 35 fetal standard ultrasonography reports, 29 echocardiography reports, and 35 fetal autopsy reports (termination for medical reasons). We assessed 18 cases of CHD diagnosed by ultrasonography findings on the second screening; in 34% of the cases patients also underwent MRI at that time and a repeated procedure 30 weeks later. **Results:** When standard ultrasonography and an extended protocol with echocardiography were used together, diagnostic errors were 14.3%. In 85.7% of the fetuses, the findings of different imaging techniques fully coincided with the autopsy findings.

Conclusions: In this cohort of pregnant women, the second screening should include more examinations to verify the diagnosis of CHD. Based on the first screening findings (increased nuchal translucency thickness and ductus venosus pulsatility index) patients should be referred to an expert for the second screening. If necessary, to verify the diagnosis of CHD ultrasonography and MRI can be combined during the third screening (34-36 weeks) in order to plan postnatal management of the newborn. The proposed algorithm for fetal CHD diagnosis enables to minimize the likelihood of error and maintain continuity of care between obstetricians-gynecologists, ultrasonographers, radiologists, neonatologists, and cardiologists.

Keywords: congenital heart diseases, ultrasonography screening, magnetic resonance imaging

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Мультимодальный подход (МРТ и УЗИ) в диагностике врожденных пороков сердца плода

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Резюме

Введение: Неснижающаяся частота врожденных пороков сердца (ВПС) ставит перед врачами медицинской визуализации задачи по раннему диагностированию ВПС с минимальными затратами и нагрузкой на беременную и плод, а также необходимости верификации диагноза ВПС внутриутробно для выработки последующей тактики ведения беременных с ВПС у плода.

Цель исследования: Оптимизация диагностических мероприятий у плодов с ВПС путем сравнения эхокардиографии и магнитно-резонансной томографии (МРТ) сердца плода.

Методы: Ретроспективно оценены результаты 35 протоколов стандартного ультразвукового исследования (УЗИ) плода и 29 протоколов эхокардиографии (ЭхоКГ), а также 35 протоколов вскрытия плодов (прерывание по медицинским показаниям). Проведена оценка 18 случаев ВПС, диагностированных по данным УЗИ на 2-м скрининге, у 34% из которых проводилось также МРТ в эти сроки и в динамике после 30 недель.



Результаты: При сочетанном использовании стандартного УЗИ и расширенного протокола с выполнением ЭхоКГ неточность в диагностике составила 14,3%. Полное совпадение по данным проведенных исследований с использованием различных технологий визуализации плода с результатами вскрытия было у 85,7% плодов.

Заключение: При проведении второго скрининга необходимо расширить объем исследования у данной когорты беременных с целью верификации диагноза ВПС. Необходимо учитывать результаты первого акушерского скрининга (увеличение толщины воротникового пространства и величину пульсационного индекса венозного протока), направляя таких пациенток к эксперту для проведения второго скрининга. При необходимости с целью верификации диагноза ВПС сочетать УЗИ и МРТ при проведении третьего скрининга (34–36 недель) для планирования постнатального ведения новорожденного. Предложенный алгоритм диагностики ВПС у плода позволяет минимизировать вероятность ошибки, сохранить преемственность среди акушеров-гинекологов, врачей УЗИ, рентгенологов, неонатологов и кардиологов.

Ключевые слова: врожденные пороки сердца, ультразвуковой скрининг, магнитно-резонансная томография

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Introduction

Magnetic resonance imaging (MRI) is currently a rapidly evolving method of diagnosing congenital heart diseases (CHD) and assessing the cardiovascular system in the fetus. Although fetal echocardiography, introduced in the mid-1980s,¹ remains the primary technique, MRI offers a valuable complement. The advantages of echocardiography are safety, accessibility, and capability of diagnosing CHDs.² Nonetheless, it should be noted that limitations to echocardiography during pregnancy, such as oligohydramnios, fetal bone ossification, fetal position, as well as maternal overweight and obesity, have not been overcome yet.^{2,3} Some researchers also believe that it is sometimes impossible and/or difficult to assess such CHD as coarctation of the aorta using echocardiography due to difficulties associated with assessment of blood flow characteristics and valve regurgitation.⁴

Despite being the first-line diagnostic tool for fetal CHD, fetal echocardiography for cardiovascular abnormalities increasingly requires additional MRI not only to advance research but also to address a clinical need, primarily to improve diagnostic accuracy and detail structural disorders. First of all, this is due to the need for contractility monitoring and blood flow assessment over time. Certain limitations to MRI in pregnancy, particularly fetal movement, hinder real-time visualization. However, studies⁵ of the joint use of Doppler ultrasound cardiac gating and radial sampling with free breathing retrospective cine-MRI demonstrated an evident prospect of such approach, which increases the importance of fetal cardiac MRI in clinical practice. Advances in the diagnosis of fetal cardiovascular abnormalities are partly owing to MRI, which is now used both in prenatal and postnatal diagnosis of CHD, expanding our insights into the normal and pathologic physiology of circulation.

The study aimed to optimize diagnostic measures in fetuses with CHD by comparing fetal echocardiography and cardiac MRI.

Methods

We retrospectively evaluated findings from 35 fetal standard ultrasonography reports, 29 echocardiography

reports, and 35 fetal autopsy reports (termination for medical reasons). The diagnosis was verified by postnatal ultrasonography and computed tomography. We assessed 18 cases of CHD diagnosed during the second screening (19–21 weeks+6 days' gestation). In addition to ultrasonography, 34% of the patients underwent MRI at that time and after 30 weeks' gestation. The study included pregnant women who carried fetuses with CHD and were examined at perinatal centers of the Children's Regional Clinical Hospital and Regional Clinical Hospital No. 2 (Krasnodar, Russian Federation) between 2021 and 2024.

Inclusion criteria: pregnant women aged 18–45 years whose fetuses were diagnosed with CHD during the screening, newborns with CHD.

Exclusion criteria: pregnant women whose fetuses did not have CHD during the screening, newborns without CHD.

The statistical analysis was performed using STATISTICA 10 (Tibco, USA) and Microsoft Excel 2016 (Microsoft Corp, USA). In most cases, data was nonnormally distributed, so nonparametric methods were used. Along with the arithmetic mean and standard deviation, the data were described by the median and 25% and 75% interquartile ranges. We used a cross-tabulation method, contingency tables, to study the structure of the relationship between qualitative indicators. The nonparametric Mann-Whitney test, 2-tailed *t* test, and Wilcoxon signed rank test were used to assess the statistical significance of differences between the study groups. If $|R| \leq 0.25$, the correlation was weak; if $0.25 < |R| \leq 0.75$, the correlation was moderate, and if $|R| > 0.75$, the correlation was strong. All the tests used the conventionally accepted level of significance ($P = .05$).⁶

Results

We compared the findings of prenatal ultrasonography and echocardiography for fetal CHD with the final result (postnatal autopsy findings) and found diagnostic discrepancies in 5 cases (14.3%). The prenatal ultrasonography findings fully coincided with the autopsy findings in 30 (85.7%) fetuses. Of them, 14 (46.7%) were prenatally diagnosed with a ventricular septal defect (VSD)

alone or in combination with other CHD and/or congenital anomalies of the genitourinary tract, bones, and the nervous system. In 7 of 14 cases (50.0%), the ultrasonography diagnosis of VSD was confirmed by the autopsy findings. In 2 of 14 (14.3%) fetuses, there were diagnostic discrepancies between the echocardiography and autopsy findings. In one case, a VSD was combined with pulmonary atresia, and in another case, with a common arterial trunk.

Analysis of the 4-chamber view revealed the VSD (Figure 1).

The second most common confirmed CHD was hypoplastic left heart syndrome (HLHS) diagnosed by echocardiography in 10 fetuses (33.3%). In one case (10.0%), HLHS was combined with a complete atrioventricular canal defect.

The B-mode showed the decreased size of the left ventricle. Color flow Doppler revealed that there was no blood flow through the mitral valve, which is shown on the abnormal 4-chamber views (Figure 2A and Figure 2B).

The third most common diagnosis was coarctation of the aorta (3 [10.0%] fetuses). In the first case, coarctation of the aorta was combined with a VSD, and in the second case, with HLHS; in the third case, coarctation of the aorta occurred alone. An indirect sign of coarctation of the aorta is a decrease in the left ventricle size (Figure 3A) detected by the abnormal 4-chamber view, measurement of the ventricular cavity width. The aortic arch view showed an area of vessel narrowing in the isthmus region and subsequent poststenotic dilatation (Figure 3B).

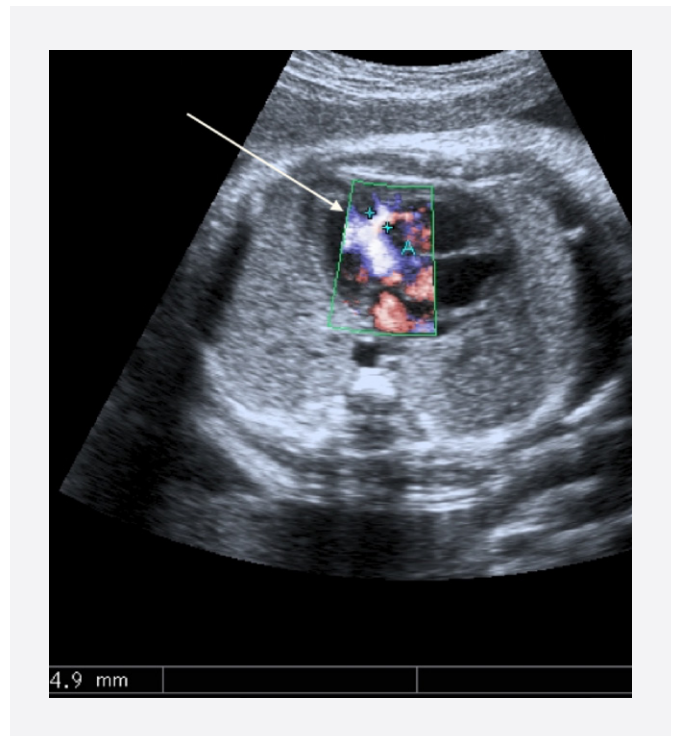
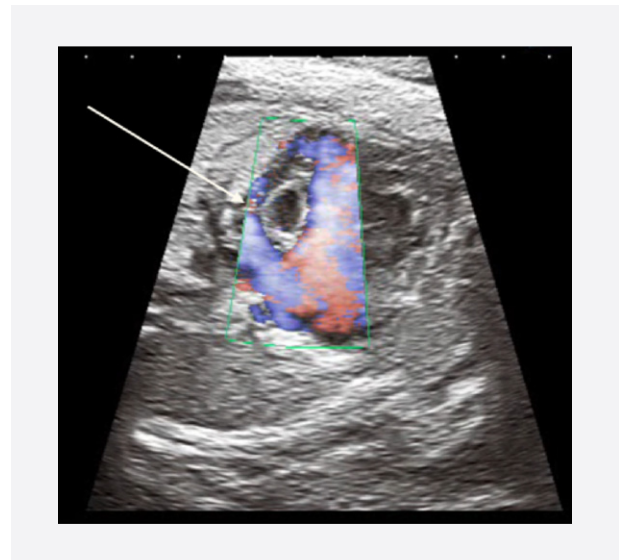


Figure 1. Ultrasonography of the fetal heart at 20 weeks' gestation (verified diagnosis). Anomalous 4-chamber view with color flow Doppler: ventricular septal defect (VSD) (4.9 mm) (arrow)

Рисунок 1. Ультразвуковое исследование (УЗИ) сердца плода на сроке 20 недель (диагноз верифицирован). Аномальный четырехкамерный срез в режиме цветового доплеровского картирования (ЦДК): дефект межжелудочковой перегородки (ДМЖП) – 4,9 мм (стрелка)



A. B-mode: decreased size of the left ventricle (arrow)
A. в B-режиме уменьшены размеры левого желудочка (стрелка)



B. Color flow Doppler: no blood flow through the mitral valve (arrow)
B. в режиме ЦДК отсутствует поток крови через митральный клапан (стрелка)

Figure 2. Ultrasonography of the fetal heart at 20 weeks' gestation (verified diagnosis). Anomalous 4-chamber views in hypoplastic left heart syndrome

Рисунок 2. УЗИ сердца плода на сроке 20 недель (диагноз верифицирован). Аномальные четырехкамерные срезы при синдроме гипоплазии левых отделов сердца

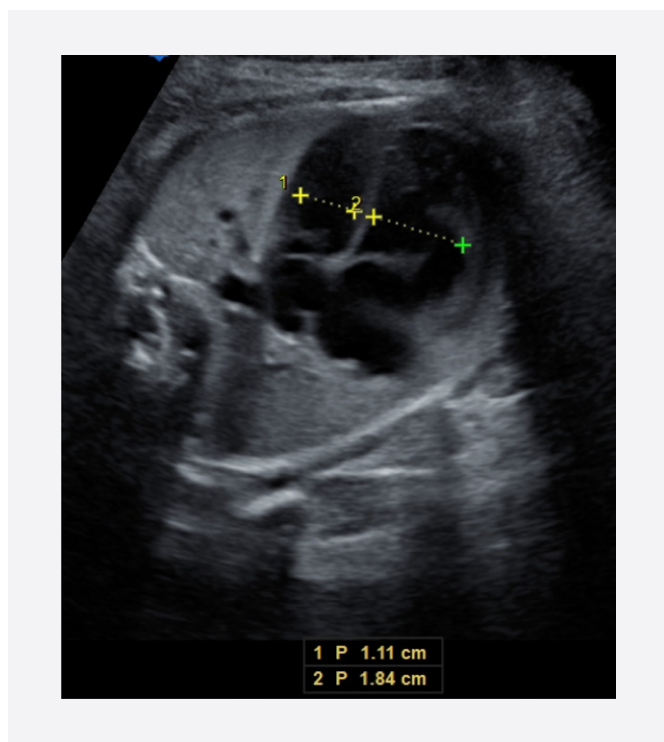


Figure 3A. Ultrasonography of the fetal heart at 20 weeks' gestation (verified diagnosis). Anomalous 4-chamber view, measurement of the ventricular cavity width, decrease in the left ventricle size (indirect sign of an aortic pathology)

Рисунок 3А. УЗИ сердца плода на сроке 20 недель (диагноз верифицирован). Аномальный четырехкамерный срез, измерение ширины полости желудочков, уменьшение размера левого желудочка (косвенный признак патологии аорты)

We performed postnatal computed tomography to confirm the diagnosis on day 60 (Figure 4).

In 3 cases (10.0%), a single ventricle was diagnosed by echocardiography and confirmed by the autopsy findings. Tetralogy of Fallot verified by echocardiography (3.3%) was confirmed during the fetal autopsy. In one case (3.3%), pulmonary atresia diagnosed by echocardiography was confirmed during the autopsy.

As we mentioned above complete diagnostic discrepancies were observed in 5 (14.3%) cases. In one case, a single ventricle diagnosed by echocardiography was not confirmed by the autopsy findings. The second case was related to the ultrasonography findings of an atrio-ventricular septal defect. Autopsy revealed an atrial septal defect (8 mm) and a VSD (8 mm), and one vessel located above the VSD (common arterial trunk, 9 mm in diameter) was found to arise from the heart. In the third case, a diagnosed interrupted aortic arch was not confirmed by the autopsy findings. The diagnosis of a single ventricle by ultrasonography and echocardiography was disproved, and autopsy revealed aortic atresia with an 8×7 mm VSD. We should note that the decision to terminate pregnancy in these cases was made due to established defects of the nervous and urinary systems, which were incompatible with life.

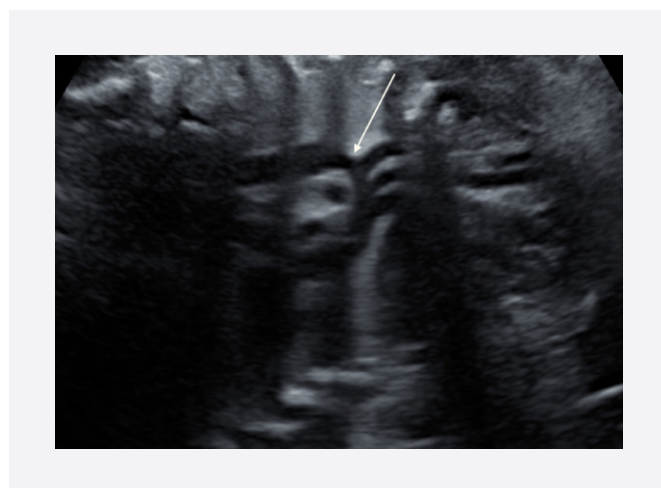


Figure 3B. Ultrasonography of the fetal heart at 20 weeks' gestation (verified diagnosis). Aortic arch view, area of vessel narrowing in the isthmus region with poststenotic dilatation (arrow)

Рисунок 3В. УЗИ сердца плода на сроке 20 недель (диагноз верифицирован). Срез через дугу аорты, участок сужения сосуда в области перешейка с последующим постстенотическим расширением (стрелка)

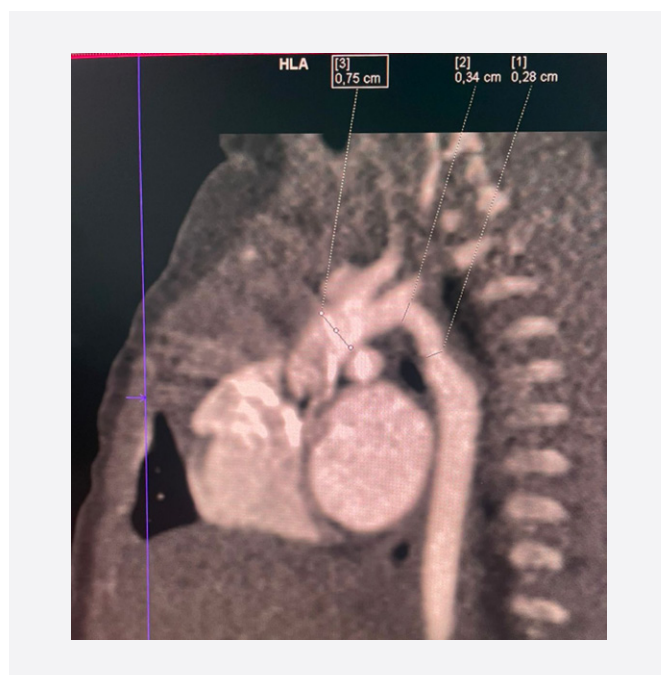


Figure 4. Coarctation of the aorta

Рисунок 4. Коарктация аорты

The fetal echocardiography findings in 5 pregnant women were evaluated. All of them, after the CHD diagnosis, continued their pregnancy owing to the possibility of surgical correction of the identified defects.

In one case, the echocardiography diagnosis of a muscular VSD was not confirmed postnatally. The postnatal diagnosis was transposition of the great arteries, patent ductus arteriosus, restrictive patent foramen ovale. This case indicates the need to expand methods of prenatal diagnosis of congenital cardiovascular abnormalities.

In another case, the fetal diagnosis of transposition of the great arteries was established. However, postnatal echocardiography revealed type 3-4 pulmonary atresia, major aortopulmonary collateral arteries, patent foramen ovale, chronic hypoxia.

We observed a case with the ultrasonography diagnosis of tetralogy of Fallot (Figure 5). It should be noted that the sonographic image with the 4-chamber view can be unchanged (Figure 5A), which justifies the need to use additional views.

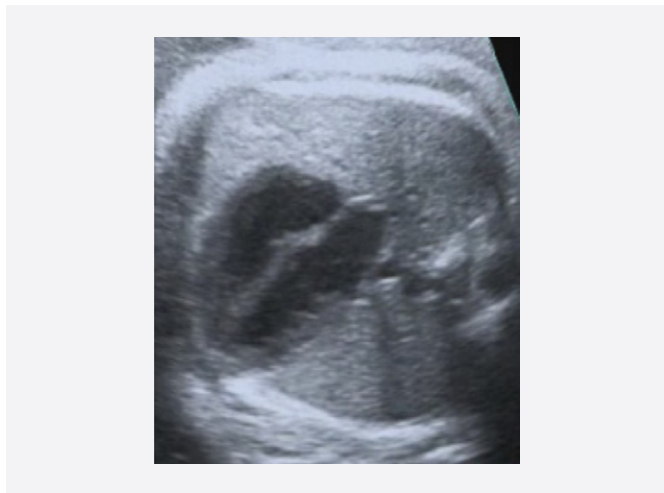


Figure 5A. Ultrasonography of the fetal heart at 20 weeks' gestation (verified diagnosis). In patients with tetralogy of Fallot, a sonographic image can show an unchanged 4-chamber view of the heart

Рисунок 5А. УЗИ сердца плода на сроке 20 недель (диагноз верифицирован). При тетраде Фалло возможно получение эхографического изображения неизмененного 4-х камерного среза сердца

During the imaging, all sonographic features of tetralogy of Fallot (Figure 5B) should be ruled out: an overriding aorta.

In the color flow Doppler mode, we registered the blood flow from the left and right ventricles into the dilated and displaced aorta (Figure 5C).

During the imaging, the pulmonary artery diameter was 2.4 mm, which is associated with 5% for this gestational age (Figure 5D).

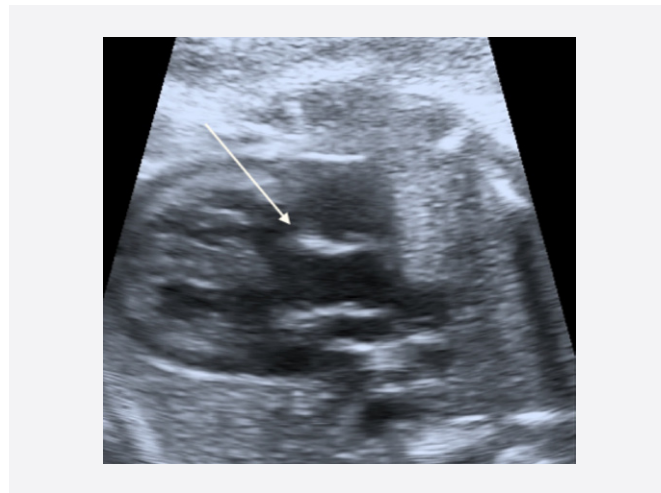


Figure 5B. Ultrasonography of the fetal heart at 20 weeks' gestation (verified diagnosis). The main sonographic sign of tetralogy of Fallot is an overriding aorta (arrow)

Рисунок 5В. УЗИ сердца плода на сроке 20 недель (диагноз верифицирован). Основной эхографический признак тетрады Фалло – выявление расширения аорты, располагающейся на межжелудочковой перегородке над дефектом (стрелка)

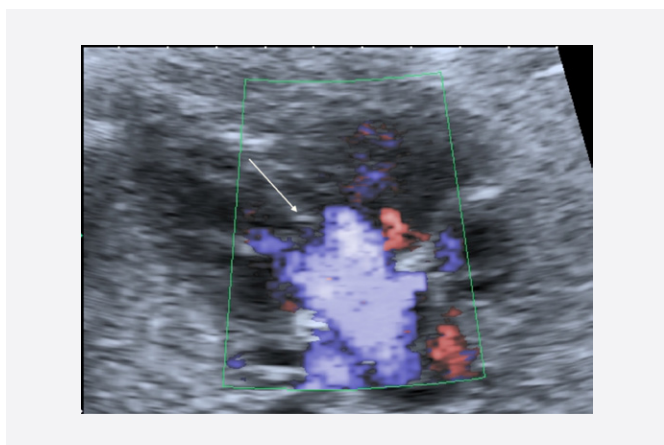


Figure 5C. Color flow Doppler: blood flow from the left and right ventricles into the dilated and displaced aorta (arrow)

Рисунок 5С. В режиме ЦДК регистрируется поток крови, поступающий из левого и правого желудочка в расширенную и смещенную аорту (стрелка)

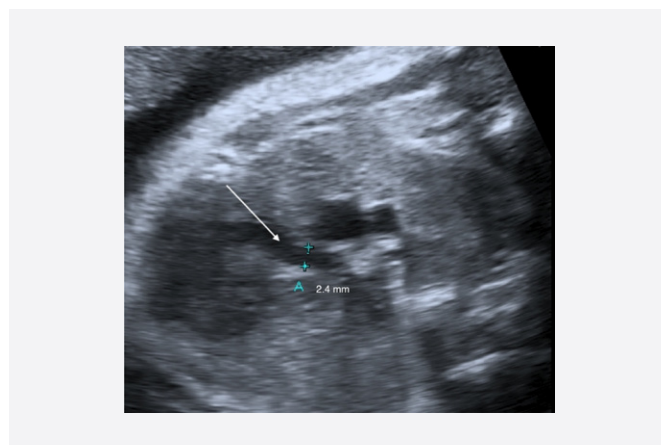


Figure 5D. Pulmonary artery diameter, 2.4 mm; less than 5% for this gestational age (arrow)

Рисунок 5D. Диаметр легочной артерии 2,4 мм, менее 5% для гестационного срока (стрелка)

Figure 5. Ultrasonography of the fetal heart at 32 weeks 4 days' gestation. Diagnosis: tetralogy of Fallot (verified diagnosis)

Рисунок 5. Ультразвуковое исследование сердца плода на сроке беременности 32 недели и 4 дня. Диагноз: тетрада Фалло (диагноз верифицирован)

The lack of prenatal diagnosis is often due to maternal overweight or obesity and hydramnios.

Analyzing this case, we should note that the difficulties in CHD visualization remained, and to overcome them we developed a comprehensive approach to the timely detection of fetal CHDs with subsequent referral to an appropriate perinatal center for delivery and surgical correction. Thus, we were convinced of the need to look for ultrasound markers during the second screening that should lay the foundation for additional methods of CHD diagnosis, including fetal MRI.

After this analysis, we came to the conclusion that additional MRI during pregnancy was needed to verify the CHD diagnosis. We report a case of

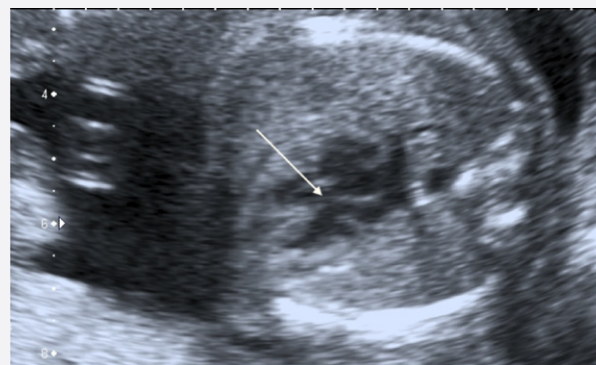
ultrasonography and MRI during the second screening (19–21 weeks + 6 days' gestation) and at 30 weeks 4 days' gestation. Ultrasonography during the second screening diagnosed a CHD at 21 weeks' gestation: double outlet right ventricle. Ultrasonography by an expert revealed a subaortic VSD (3.6 mm) at 22 weeks' gestation (Figure 6).

During the imaging, abnormal 4-chamber view of the heart and subaortic VSD (3.6 mm) were found. Both great arteries arose from the right ventricle. MRI performed at the same time revealed a double outlet right ventricle (Figure 7).

To develop a postnatal cardiovascular management strategy, both ultrasonography and MRI were performed



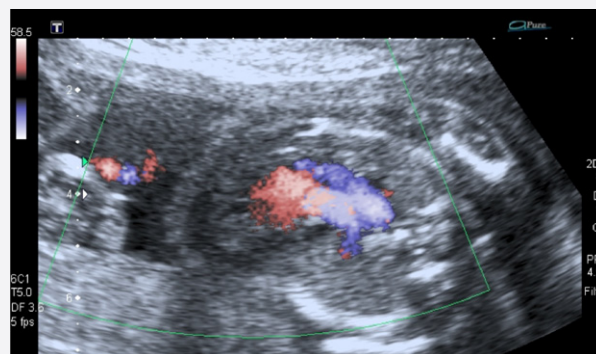
6A. Unchanged 4-chamber view
6A. Неизмененный 4х-камерный срез



6B. Subaortic ventricular septal defect (VSD) (3.6 mm)
6B. Подаортальный ДМЖП 3,6 мм

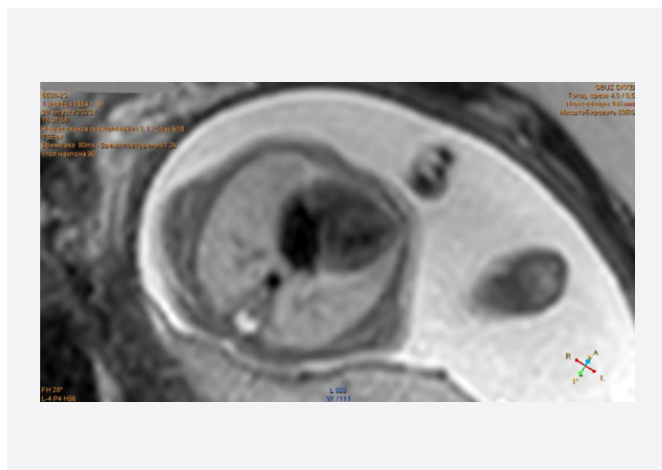


6C. Double outlet right ventricle
6C. Оба магистральных сосуда выходят из полости правого желудочка

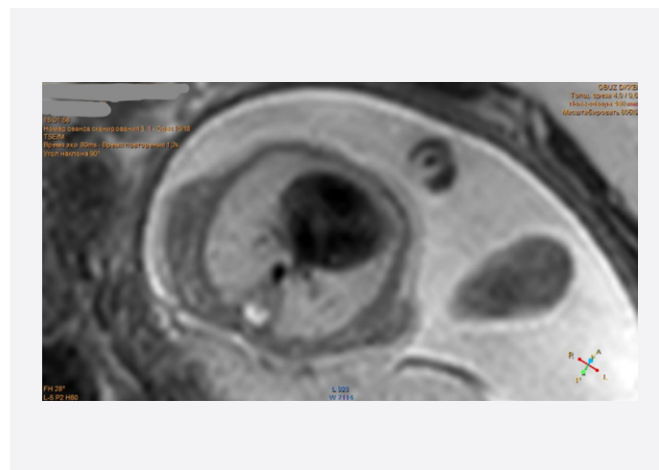


6D. Color flow Doppler: blood flow from the right ventricle into the great vessels
6D. В режиме ЦДК определяется поток крови, поступающий в магистральные сосуды из полости правого желудочка

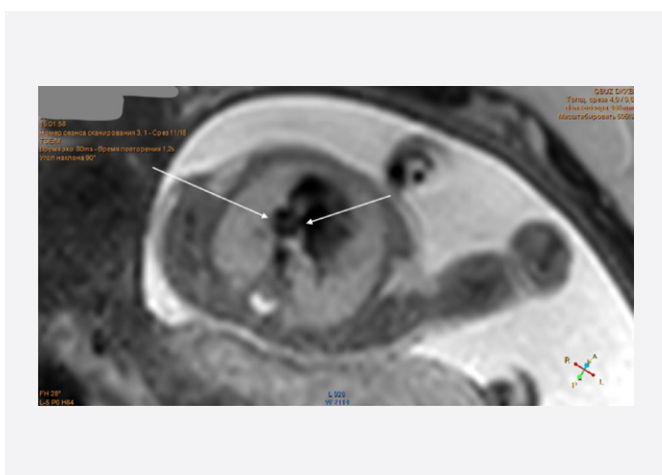
Figure 6. Ultrasonography of the fetal heart at 22 weeks 3 days' gestation (verified diagnosis)
Рисунок 6. УЗИ сердца плода на сроке 22 недели и 3 дня (диагноз верифицирован)



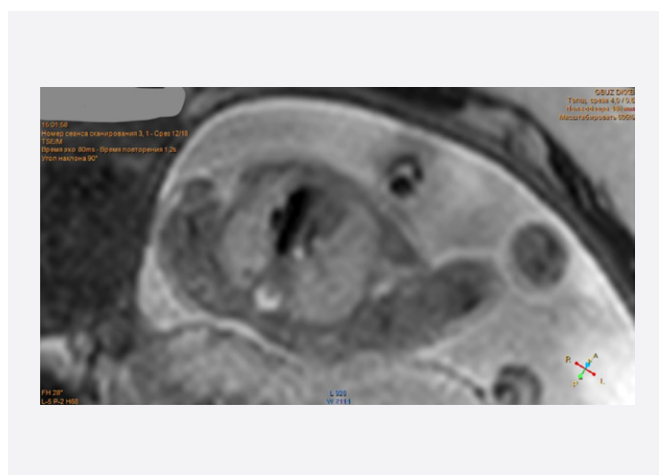
7A. 4-chamber view of the heart (MRI)
7A. 4-х камерный срез сердца (МРТ)



7B. 4-chamber view of the heart and descending aorta
7B. 4-х камерный срез сердца и нисходящая Ао



7C. Left arrow: aorta and pulmonary artery in cross-section;
right arrow: subaortic VSD
7C. Стрелкой слева указаны Ао и ЛА в поперечном сечении; справа стрелка подаортальный ДМЖП



7D. Aortic arch and superior vena cava
7D. Дуга Ао и ВПВ

Figure 7. Fetal MRI at 22 weeks' gestation: double outlet right ventricle; parallel arrangement of the great arteries (verified diagnosis)

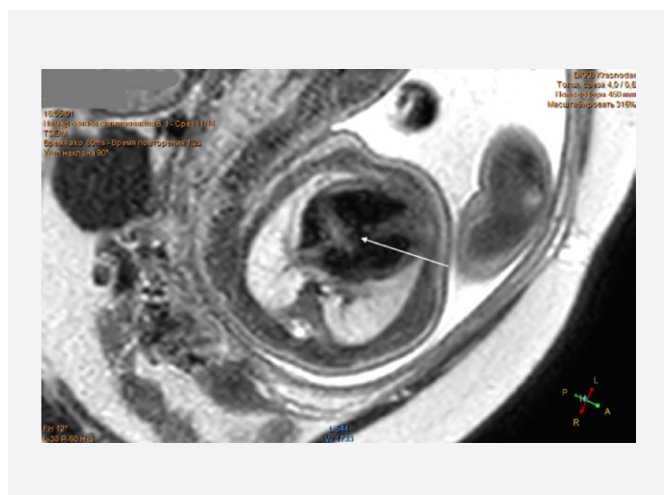
Рисунок 7. МРТ плода на сроке 22 недели: двойное отхождение магистральных сосудов; параллельный ход магистральных сосудов (диагноз верифицирован)

at 30 weeks 4 days' gestation. Repeated ultrasonography and MRI confirmed the diagnosis (Figure 8).

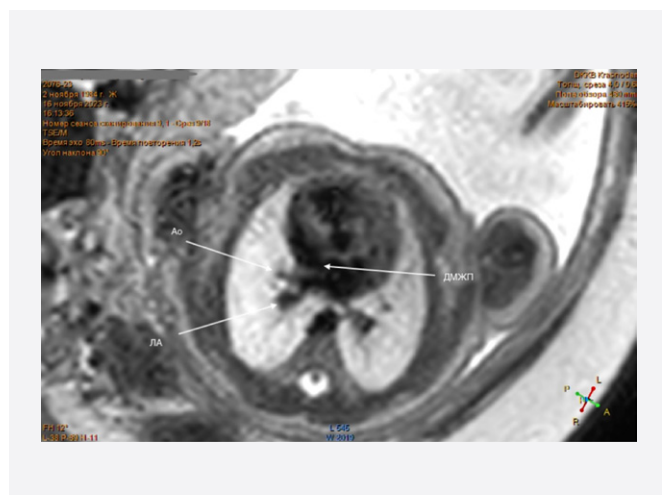
As shown above, ultrasonography performed by an expert at 22 weeks+3 days' gestation and at 30 weeks +4 days' gestation revealed that both great arteries arise from the right ventricle, which was confirmed by MRI. A subaortic VSD (3.6 mm) was described according to ultrasonography. A series of MRI scans showed signs of the ascending aorta origin from the right ventricle and the common pulmonary artery origin from the right ventricle (Figure 8).

As a result of the postnatal follow-up examination, the diagnosis was CHD, double outlet right ventricle with a subpulmonary VSD (Taussig-Bing syndrome),

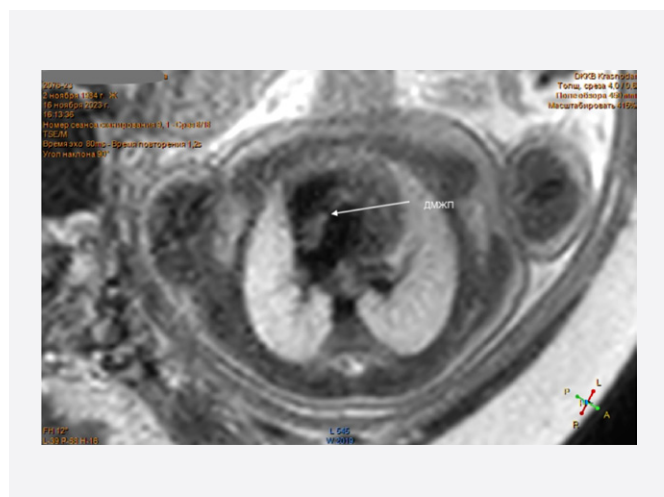
pulmonary artery stenosis, mitral valve anomaly, patent ductus arteriosus, chronic hypoxia. According to the echocardiography findings, left ventricle end-diastolic diameter was 20 mm; the walls were not thickened; ejection fraction was 69%; aorta, 13 mm; left atrium, 16 mm; right ventricle, 9 mm; right ventricular walls, 3 mm; the aortic valve was bicuspid; aortic valve velocity, 0.7 m/s; pulmonary valve, 11 mm; pulmonary valve velocity, 3.6-4.0 m/s (peak gradient, 56-64 mm Hg); VSD, 7 mm; the tricuspid valve chord and mitral valve were attached to the edge of the defect; the anterior mitral leaflet was split; grade 1 tricuspid regurgitation was observed; there was normal blood flow in the abdominal aorta with velocity of 1.5 m/s.



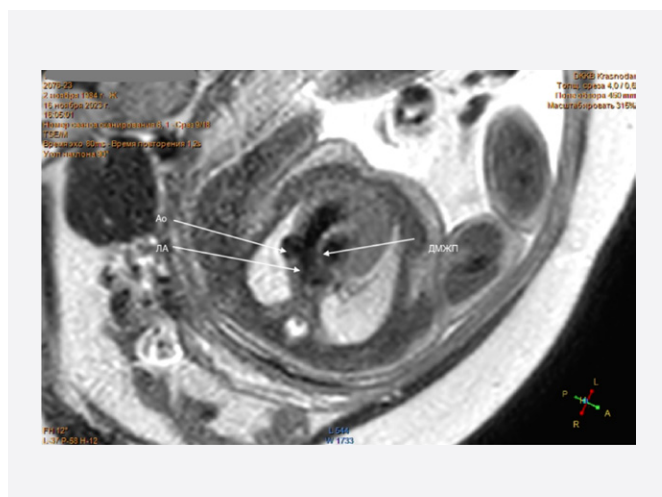
8A. 4-chamber view, subaortic VSD up to 5 mm
8A. 4-х камерный срез, ДМЖП подаортальный до 5 мм



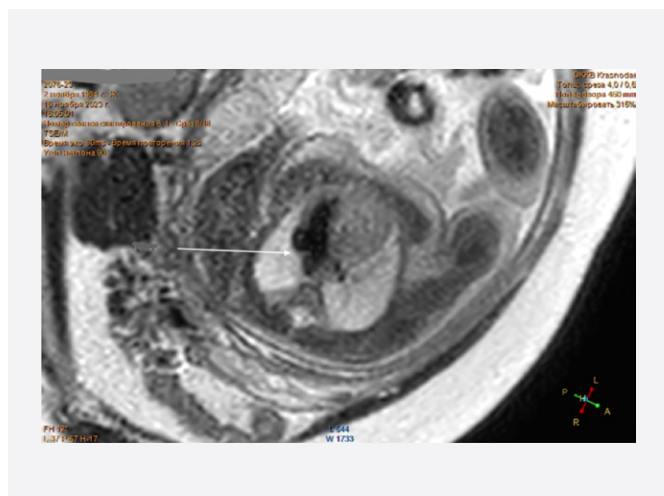
8B. Aorta, pulmonary artery, VSD
8B. Ao, ЛА, ДМЖП



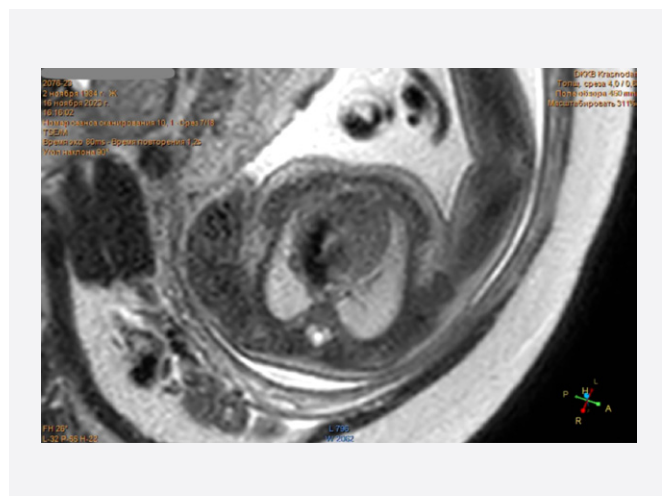
8C. VSD
8C. ДМЖП



8D. Aorta and pulmonary artery in cross-section, VSD
8D. Ao и ЛА в поперечном сечении, ДМЖП



8E. Level of the aortic arch and superior vena cava
8E. Уровень дуги Ао и ВПВ



8F. Level of the aortic arch
8F. Уровень дуги Ao

Figure 8. Fetal MRI at 30 weeks 4 days' gestation: double outlet right ventricle (verified diagnosis)
Рисунок 8. МРТ плода на сроке 30 недель и 4 дня: двойное отхождение магистральных сосудов (диагноз верифицирован)

Conclusions

In our earlier study,⁷ we studied the relationship between the nuchal translucency thickness and the ductus venosus pulsatility index, which are determined during the first screening (11-13 weeks+6 days' gestation) in fetuses with CHD. One of the early diagnostic criteria of a possible cardiovascular pathology was found to be nuchal translucency thickness of 3.68 ± 1.3 mm and ductus venosus pulsatility index of 1.098 ± 0.169 . We found a moderate correlation ($R=0.510$) between both indicators.

During the second screening, the scope of the examination in this cohort of pregnant women should be expanded to verify the CHD diagnosis. To do that and plan the post-natal management, ultrasonography and MRI can be combined during the third screening (34-36 weeks).

Compared with ultrasonography, fetal cardiac MRI is more informative when it comes to the diagnosis of a CHD combined with a pathology of the great arteries. During the study, we found limitations to fetal cardiac MRI during the second trimester of pregnancy that are linked to fetal behavioral responses (motor activity).

The development of a fetal heart imaging protocol aimed at the formation of risk groups among pregnant women who carry fetuses with CHDs is justified.

The proposed approach to the fetal CHD diagnosis enables to minimize the likelihood of error and maintain continuity of care between obstetricians-gynecologists, ultrasonographers, radiologists, neonatologists, and cardiologists.

Author contributions

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Acquisition, analysis, or interpretation of data: Pomortsev, Krivonosova, Goloseev

Manuscript drafting and revising: All authors

Final approval of the version to be published: Pomortsev, Karakhalis

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Конфликт интересов

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