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Original Article PEDIATRICS

Fetal echocardiography: A nine-year experience at a single tertiary center

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ABSTRACT

OBJECTIVE: Fetal echocardiography is a vital diagnostic tool used during pregnancy to evaluate the structure and function of the fetal heart. It allows for the early detection of congenital heart diseases (CHDs) and fetal arrhythmias, enabling timely prenatal counseling, perinatal planning, and therapeutic interventions when necessary. While routine obstetric ultrasounds may detect basic structural anomalies, fetal echocardiography provides more detailed anatomical and functional information, particularly in high-risk pregnancies. This study presents a nine-year experience of fetal echocardiography at a tertiary center, focusing on diagnostic accuracy, clinical contribution, and postnatal outcomes.

METHODS: We retrospectively analyzed 405 fetuses who underwent fetal echocardiography between March 2016 and March 2025. All evaluations were performed by an experienced pediatric cardiologist using standard imaging protocols. Fetal cardiac findings and postnatal confirmation data were assessed.

RESULTS: Major structural cardiac anomalies were identified in 48 fetuses (11.9%), with the most common being Tetralogy of Fallot and Transposition of the Great Arteries. Minor findings such as echogenic intracardiac focus, premature atrial contractions, and small ventricular septal defects (VSDs) were seen in 104 cases (25.7%). Significant arrhythmias, including supraventricular tachycardia and complete heart block, were detected in 14 cases (3.5%).

CONCLUSION: Fetal echocardiography is a reliable, non-invasive method for the early and accurate diagnosis of fetal cardiac anomalies. It facilitates appropriate prenatal referral and postnatal care planning, ultimately improving neonatal outcomes.

Keywords: Congenital heart disease; fetal arrhythmia; fetal echocardiography.

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Congenital heart diseases (CHDs) are observed in approximately 1% of all live births and are the most common cause of death in the early neonatal period [1, 2]. Timely diagnosis is essential for planning the appropriate medical and/or surgical interventions, and fetal echocardiography plays a central role in the early detection of these conditions [3].

Fetal echocardiography is primarily recommended for pregnancies classified as high-risk due to fetal, maternal, or genetic indications. However, it may also be performed in low-risk pregnancies based on various clinical concerns [4, 5]. Although the optimal time for fetal echocardiography is between 18 and 22 weeks of gestation, it can be performed earlier or later when necessary [4]. The transition from fetal to neonatal circulation is a critical period for postnatal adaptation. Prenatal diagnosis of CHDs is crucial to prevent hemodynamic instability during this transitional phase and to facilitate the planning of specialized postnatal care [6]. Furthermore, timely diagnosis aids in making critical decisions regarding the timing, location, and mode of delivery.

Also, fetal arrhythmias can occur even in fetuses with structurally normal hearts. While most fetal arrhythmias are benign, persistent arrhythmias and conduction blocks

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can lead to decreased cardiac output, resulting in non-immune hydrops fetalis and fetal loss. Early detection and appropriate management of fetal arrhythmias are essential for preventing intrauterine and neonatal mortality [7].

The aim of this study is to emphasize the role of fetal echocardiography in detecting CHDs and major fetal arrhythmias, and its role in guiding prenatal counseling and perinatal management.

MATERIAL AND METHODS

Study Design and Data Collection

This retrospective study was conducted using fetal echocardiographic evaluations performed between March 2016 and March 2025 in the Pediatric Cardiology Outpatient Clinic. All fetal echocardiographic assessments included in the study were performed and reported by the same pediatric cardiologist (Oyku Tosun, the author). The data were obtained from clinical records and fetal echocardiography reports and were analyzed retrospectively. Cardiac anomalies detected during fetal echocardiography were categorized based on the anticipated need for postnatal management. Cardiac anomalies that were likely to require urgent intervention or treatment during the early postnatal period were defined as complex CHDs. In contrast, anomalies that were unlikely to necessitate immediate postnatal intervention and required only routine follow-up were defined as minor CHDs.

The following variables were reviewed from the medical records at the time of referral:

- Gestational age at the time of examination
- Maternal age
- Gravida, parity, and history of abortion
- Presence of multiple pregnancy
- Use of assisted reproductive technologies (ART)
- Referral indication for fetal echocardiography

As this was a retrospective study based on medical records, informed consent was not required. All patient data were handled in accordance with Helsinki Declaration.

Inclusion and Exclusion Criteria

Inclusion Criteria

Patients who underwent detailed fetal echocardiographic evaluation in the Pediatric Cardiology Outpatient Clinic and had examinations that were fully reported by the same physician.

Highlight key points

- Complex congenital heart disease was detected in 11.9% of fetuses, with all diagnoses confirmed postnatally by the same pediatric cardiologist.
- Tetralogy of Fallot and D-transposition of the great arteries were the most frequently identified complex congenital heart diseases.
- Minor cardiac findings were common (25.7%), predominantly echogenic intracardiac focus, premature atrial contractions, and small ventricular septal defects.
- Significant fetal arrhythmias occurred in 3.5% of cases; timely transplacental and/or direct fetal therapy restored sinus rhythm and resolved hydrops when present.
- Abnormal cardiac findings on second-trimester ultrasound independently constituted the leading referral indication for complex congenital heart disease detection.

Exclusion Criteria

- Patients who had fetal echocardiography performed only in other departments and had no reports documented by the author.
- Patients with incomplete or missing detailed reports for any reason.
- Cases in which postnatal diagnosis could not be confirmed due to loss to follow-up.
- Patients in whom reports indicated that adequate imaging quality could not be performed.

Fetal Echocardiography Procedure

All examinations were carried out by the same pediatric cardiologist using a Samsung H60 Ultrasound System (Samsung Healthcare Systems) with a convex transducer. All examinations were performed in accordance with the American Heart Association (AHA) fetal echocardiography guidelines [8]. The patient was placed in the supine position, with the upper body slightly elevated to optimize the imaging quality. The fetal position and left/right and anterior/posterior orientation were first identified in order to determine the abdominal situs, cardiac position and axis. Standard imaging planes, including the four-chamber view, outflow tracts, great arteries, three-vessel and trachea view, aortic arch, and ductal arch view were assessed in all cases. Relevant findings from these standard views were recorded for each case. Color Doppler and pulsed-wave (PW) Doppler imaging were used to assess blood flow velocities and direction, and obtained data were documented. Fetal heart rate was assessed and recorded using both M-mode imaging and PW Doppler techniques. The

Table 1. Demographic characteristics of the study population (n=405)

Characteristic	Value
Mean maternal age (years)	29.80±5.87
Mean gestational age (weeks)	22±1.6
Nulliparous patients (%)	56.8
Multiparous patients (%)	43.2
Twin pregnancies (%)	5.9
Pregnancies via ART (%)	8.9

ART: Assisted reproductive technologies.

presence and type of any arrhythmia detected during the examination were also documented.

Only examinations that resulted in optimal imaging quality, without limitations caused by maternal body habitus or fetal position and movement, were recorded and included in the analysis.

Statistical Analysis

Descriptive statistical analyses were performed for all variables included in the study. The frequency and percentage distributions of categorical variables were analyzed using the SPSS software program (version 21.0. Armonk, New York: IBM Corp.).

RESULTS

Study Population

A total of 427 pregnant women who underwent fetal echocardiographic evaluation between March 2016 and March 2025 were retrospectively reviewed. Of these, 10 patients were excluded due to suboptimal image quality, and 12 patients were excluded due to the lack of postnatal confirmation of findings. The final study population consisted of 405 patients who met the inclusion criteria.

Demographic Characteristics

The mean maternal age was 29.80±5.87 years (range: 16–48 years), and the mean gestational age at the time of examination was 22±1.6 weeks. Among the included patients, 230 (56.8%) were nulliparous and 175 (43.2%) were multiparous. There were 24 twin pregnancies (5.9%), and 36 pregnancies (8.9%) were conceived using assisted reproductive technologies (ART) (Table 1).

Table 2. Referral indications for fetal echocardiography

Referral indication	Number of patients (n)	Percentage (%)
Abnormal cardiac findings on second-trimester USG	80	19.8
Suboptimal cardiac imaging on routine screening	60	14.8
Self-referral	53	13.1
Fetal arrhythmia	50	12.3
GDM	40	9.9
ART pregnancy	36	8.9
Previous child with CHD	22	5.4
Multiple gestation	24	5.9
Pregestational DM	20	4.9
Advanced maternal age	20	4.9
Total	405	100

ART: Assisted reproductive technologies; CHD: Congenital heart disease; DM: Diabetes mellitus; GDM: Gestational diabetes mellitus; USG: Ultrasonography.

Referral Indications

Referral indications were recorded for all patients. The most common referral reason was abnormal cardiac findings on second-trimester ultrasound (19.8%), followed by suboptimal cardiac visualization on routine screening (14.8%) and self-referral (13.1%) (Table 2).

Fetal Echocardiographic Findings

Among the 405 patients who underwent fetal echocardiographic evaluation, 239 fetuses (59.0%) had normal cardiac anatomy and rhythm. In 48 fetuses (11.9%), complex CHD was detected and all diagnoses were postnatally confirmed by the same pediatric cardiologist. Among fetuses diagnosed with complex CHDs, the most common referral indication was the detection of abnormal cardiac findings during routine second-trimester ultrasound screening (80%). The most frequently identified anomalies were Tetralogy of Fallot (TOF) and D-Transposition of the Great Arteries (D-TGA), each observed in 9 cases (18.8%). Other notable diagnoses included double outlet right ventricle (DORV), complete atrioventricular septal defect (AVSD), Ebstein anomaly, truncus arteriosus (TA), hypoplastic left heart syndrome (HLHS), and pulmonary atresia with intact ventricular septum (PA-IVS). Detailed distribution of the complex fetal cardiac CHDs is pre-

Table 3. Distribution of fetal complex congenital heart diseases

Diagnosis	Number of cases (n)	Percentage (%)
BLV	1	2.1
DORV	5	10.4
TOF	9	18.8
TOF-APVs	1	2.1
cAVSD	3	6.3
D-TGA	9	18.8
Ebstein's anomaly	3	6.3
Critical PS	2	4.2
DCRV	1	2.1
TA	2	4.2
HLHS	2	4.2
DAA/VR	1	2.1
IVS-PA	3	6.3
CMP	2	4.2
Taussig-Bing anomaly	3	6.3
Severe CoA	1	2.1
IAA	1	2.1
Total	48	100

APVs: Absent pulmonary valve syndrome; BLV: Borderline left ventricle; cAVSD: Complete atrioventricular septal defect; CMP: Cardiomyopathy; CoA: Coarctation of the Aorta; PS: Pulmonary stenosis; DAA/VR: Double aortic arch / vascular ring; DCRV: Double-Chambered right ventricle; DORV: Double outlet right ventricle; D-TGA: Transposition of the great arteries; HLHS: Hypoplastic left heart syndrome; IAA: Interrupted aortic arch; TA: Truncus arteriosus; TOF: Tetralogy of fallot.

Table 4. Distribution of fetal minor heart diseases

Cardiac finding	Number of patients (n)	Percentage (%)
EIF	40	38.5
PAC	30	28.8
VSD	21	20.2
-Small perimembranous VSD	11	—
-Small muscular VSD	10	—
AS/BAV	5	4.8
C-TGA	2	1.9
Left atrial isomerism (isolated)	1	1.0
Left pulmonary artery sling	2	1.9
Isolated right aortic arch	2	1.9
PAPVR+ASD	1	1.0
Total	104	100

EIF: Echogenic intracardiac focus; PAC: Premature atrial contractions; VSD: Ventricular septal defect; AS/BAV: Mild aortic stenosis/ bicuspid aortic valve; C-TGA: Corrected transposition of the great arteries; PAPVR+ASD: Partial anomalous pulmonary venous return+ atrial septal defect.

sented in Table 3. For all fetuses diagnosed with complex CHD, families were counseled in detail regarding postnatal prognosis and the expected clinical course. In cases where urgent postnatal intervention or surgical treatment was expected, prenatal consultations were arranged with appropriate tertiary care centers. Delivery planning was coordinated through a multidisciplinary approach to ensure optimal perinatal and postnatal management.

Among the 405 patients who underwent fetal echocardiographic evaluation, 104 (25.7%) had minor cardiac anomalies, including benign rhythm disorders and mild structural anomalies. All diagnoses were postnatally confirmed by the same pediatric cardiologist through standardized echocardiographic and electrocardiographic evaluations. The distribution of these findings is presented in Table 4. The most frequently observed minor cardiac finding was isolated echogenic intracardiac focus (EIF) (38.5%), followed by premature atrial contractions (PAC) (28.8%) which disappeared in the third trimester. The most commonly detected mild structural congenital heart defect was ventricular septal defect (VSD) (20.2%). Among the 24 fetuses diagnosed with VSD, 11 had small perimembranous defects and 10 had small muscular defects requiring postnatal follow-up.

Among the 405 fetuses, significant rhythm disturbances were identified in 14 cases (3.5%) (Table 5). Sustained tachyarrhythmia was diagnosed in 10 of these 14 fetuses. Hydrops fetalis was present in five of these fetuses, atrial fibro-flutter (AF) was detected in one, while the remaining four had long ventriculoatrial (VA) tachyarrhythmia. All cases with hydrops were treated with a combination of direct fetal therapy and transplacental antiarrhythmic treatment. Sinus rhythm was successfully restored in all, and hydrops regressed. In the other five fetuses without hydrops, short VA tachyarrhythmia was observed and sinus rhythm was successfully achieved with transplacental therapy alone in all.

Complete atrioventricular block (CAVB) accompanied by fetal bradycardia was detected in three of 14 fetuses. Maternal serologic evaluation revealed the presence of anti-Ro/SSA and anti-La/SSB antibodies. To prevent the progression to cardiac failure and the de-

Table 5. Summary of significant fetal arrhythmias and management

Type of arrhythmia	Number of cases (n)	Hydrops fetalis present	Treatment approach	Sinus rhythm achieved
Sustained tachyarrhythmia (total)	10	5	Direct fetal+ transplacental therapy	Yes
— AF	1	Yes	Direct fetal+ transplacental therapy	Yes
— Long VA tachycardia	4	Yes	Direct fetal+ transplacental therapy	Yes
— Short VA tachycardia	5	No	Transplacental therapy only	Yes
CAVB + bradycardia	3	No	Maternal treatment; postnatal pacemaker implanted	CAVB
2:1 AVB+ suspected LOTS	1	No	Transplacental therapy; Long QT confirmed postnatally	LQTs

AF: Atrial flutter; AV: Atrioventricular; AVB: Antrioventricular block; CAVB: Complete atrioventricular block; LQTS: Long QT syndrome; VA: Ventriculoatrial.

velopment of hydrops fetalis transplacental treatment, including corticosteroids and/or β-agonists, was administered in all cases. Postnatally, all 3 infants required permanent pacemaker implantation and were referred to a specialized arrhythmia center.

One fetus referred due to bradycardia was found to have a 2:1 atrioventricular block (AVB). Based on these findings, long QT syndrome (LQTS) was suspected, and close prenatal follow-up was initiated. Genetic counseling was provided, and LQTS was also diagnosed in the mother. The diagnosis was confirmed postnatally by electrocardiography (ECG), which revealed a prolonged QT interval of 650 milliseconds.

DISCUSSION

In this retrospective study, we evaluated the diagnostic efficacy and clinical impact of fetal echocardiography in detecting CHDs and major arrhythmias.

In line with previous studies [9, 10], our findings indicate that the most common reason for referral to fetal echocardiographic evaluatin was the detection of abnormal cardiac findings during the second-trimester detailed ultrasonography ultrasoun (19.8%), followed by suboptimal cardiac visualization during routine obstetric ultrasound (14.8%). The high referral rate based on detailed ultrasound findings demonstrate the importance of systematic cardiac screening protocols and adequate training of sonographers. Furthermore, the considerable proportion of referrals due to suboptimal imaging suggests the ongoing need for improved imaging conditions and techniques, particularly in low-resource settings or in cases with maternal or fetal factors limiting visualization.

Among 405 fetuses, complex CHDs were identified in 11.9%, which is comparable to rates reported by Makkar et al. [11] (15%) in a single center study. It is slightly higher than those observed in the previous studies reported by Alp et al. [12] and Ozbarlas et al. [13], where the CHDs detection rate was approximately 5.6% and 7.8%. respectively. This discrepancy may be attributable to differences in referral criteria, as our study group included a high proportion of patients referred due to abnormal ultrasound findings (19.8%) or suboptimal cardiac visualization (14.8%).

In our study, the most common complex CHDs were TOF and D-TGA, each identified in 2.2% of cases. Similar results were reported in the systematic review and meta-analysis by Van der Linde et al. [14], where TOF was the most frequent complex CHD, followed by D-TGA. Early and accurate prenatal diagnosis of these conditions is essential for appropriate counseling, postnatal planning, and timely surgical intervention.

Minor cardiac findings, including isolated VSD and EIF, were present in 25.7% of cases in our study. This is consistent with prior literature, where rates of incidental or minor findings have ranged from 20% to 30% depending on population characteristics and imaging protocols. The clinical significance of these findings varies; while EIF is typically considered a benign variant, isolated small VSDs require follow-up for potential hemodynamic relevance [15–17].

In previous studies, fetal arrhythmias have been reported to occur in 1–3% of pregnancies and to account for approximately 20% of referrals for fetal cardiological evaluation [18, 19]. Consistent with the literature, fetal arrhythmias constituted 12.3% of the indications for fetal echocardiographic evaluation in our study.

Benign PACs were observed in 28.8% of our cases, consistent with previously reported rates [20, 21].

Notably, we identified significant arrhythmias in 3.5% of fetuses, including sustained tachyarrhythmias with hydrops fetalis present in 5 cases. These findings are higher than the previous studies [22, 23]. In cases of persistent fetal tachyarrhythmia, heart failure is an unavoidable consequence of hydrops fetalis and is associated with a high mortality rate [24]. Therefore, timely restoration of a normal fetal heart rate through appropriate treatment is crucial for fetal survival and the achievement of term delivery. The favorable response to transplacental antiarrhythmic therapy in our cohort, including complete rhythm normalization and hydrops resolution, underscores the importance of early detection and specialized management, as also emphasized by Demirci et al. [25].

Additionally, we identified one case of LQTS, which was confirmed postnatally. Similarly, in the case series by Zidere et al. [26], LQTS was suspected in four fetuses based on the presence of fetal bradycardia. In our case, suspicion was also raised due to bradycardia, and the concurrent finding of second-degree AV block further supported the diagnosis. Although rare, such cases are increasingly recognized through advanced fetal rhythm assessment and highlight the importance of early detection for appropriate neonatal management and genetic counseling.

In our study, CAVB was detected in three fetuses, all of whom tested positive for maternal anti-Ro/SSA and anti-La/SSB antibodies, indicating an immune-mediated etiology. Transplacental treatment, including corticosteroids and/or β-agonists, was administered in all cases. Similarly, in the study by Jaeggi et al. [27] involving 37 fetuses with CAVB, combined transplacental therapy was applied and shown to significantly reduce both fetal morbidity and mortality [26]. These findings, in line with our results, indicate the importance of early detection and effective transplacental management to improve fetal survival and reduce complications associated with this potentially life-threatening condition.

Limitations

The retrospective and single-center nature of this study limits its generalizability to the broader population. Additionaly, our institution serves as a referral center for high-risk pregnancies, which may have resulted in a higher observed prevalence of cardiac anomalies compared to the general obstetric population. Future prospective, multicenter studies with larger cohorts are needed to validate these findings and provide more generalizable insights into the impact and outcomes of fetal echocardiography.

Conclusion

When performed under appropriate conditions by experienced specialists, fetal echocardiography plays a critical role in the diagnosis of CHDs and significant fetal arrhythmias. Detecting congenital heart anomalies during the fetal period not only enables effective multidisciplinary planning for postnatal care but also allows early counseling of families regarding prognosis and management options. Additionally, the early detection of life-threatening arrhythmias facilitates early initiation of transplacental or direct fetal therapy, markedly improving perinatal outcomes and survival rates. These findings underscore the importance of fetal echocardiography in optimizing outcomes for both the fetus and the family. Our study reinforces the high diagnostic utility of fetal echocardiography in both structural and rhythm-related cardiac anomalies. The results further support the integration of fetal cardiology services into perinatal care, particularly in tertiary referral centers.

Ethics Committee Approval: The Istanbul Medeniyet University Non-Interventional Clinical Research Ethics Committee granted approval for this study (date: 22.04.2025, number: 2025/07-17).

Informed Consent: Written informed consents were obtained from patients who participated in this study.

Conflict of Interest: The author declare that there is no conflict of interest.

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