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Congenital Heart Diseases Detected by Fetal Echocardiography and the Prevalence: A Single Center Experience

Fetal Ekokardiyografi ile Tespit Edilen Doğumsal Kalp Hastalıkları ve Prevelansı: Tek Merkez Deneyimi

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Abstract

Aim: Fetal echocardiography is an effective screening tool for the detection of cardiac anomalies. The aim of this study is to evaluate risk groups of pregnant women, to determine congenital heart diseases in each group and to find the prevalence of cardiac anomalies in our region.

Material and Method: Our study included all pregnant women who were referred for fetal echocardiography between January 2023 and September 2024.

Results: A total of 387 pregnant women underwent fetal echocardiography of which 136 were low-risk group and 251 were high-risk group. The mean ages of pregnant women were 27.43±1.37 and 28.54±5.45 years, respectively. Among the high-risk group, maternal diabetes (13.17%), dysrhythmia (5.42%) and history of extracardiac anomaly in previous child or fetus (8.53%) were the most common reasons for referral. While ventricular septal defect was the most detected congenital heart disease in low risk group; hypoplastic left heart syndrome (1.20%) and pulmonary atresia/hypoplasia (1.20%) were the most detected cardiac anomalies in high-risk group. The prevalence of cardiac anomalies in the low-risk and high-risk pregnancies was found to be 2.94% and 6.37% respectively while the overall prevalence was 5.16%.

Conclusion: Fetal echocardiography is a very useful technique when performed by experienced individuals with sufficient time. Dedicated cardiac screening should be part of the routine anomaly scan. Also, detailed fetal echocardiography should be performed in all pregnant women who were in low or high risk groups.

Keywords: Congenital heart diseases, fetal echocardiography, referral risk group, prevalence

Öz

Amaç: Fetal ekokardiyografi, kardiyak anomalilerin tespiti için etkili bir tarama aracıdır. Bu çalışmanın amacı, gebe kadınların risk gruplarını değerlendirmek, her gruptaki konjenital kalp hastalıklarını belirlemek ve bölgemizde kardiyak anomalilerin prevalansını bulmaktır.

Gereç ve Yöntem: Çalışmamız Ocak 2023 ve Eylül 2024 tarihleri arasında fetal ekokardiyografi için sevk edilen tüm gebeleri içermektedir.

Bulgular: Toplam 387 gebe kadına fetal ekokardiyografi uygulandı, bunlardan 136'sı düşük risk grubunda ve 251'i yüksek risk grubundaydı. Gebelerin ortalama yaşları sırasıyla 27,43±1,37 ve 28,54±5,45 yıldı. Maternal diyabet (%13,17), disritmi (%5,42) ve ekstrakardiyak anomalisi olan önceki çocuk veya fetüs öyküsü (%8,53) yüksek risk grubunda en sık sevk nedenleriydi. Ventriküler septal defekt düşük risk grubunda en sık tespit edilen doğumsal kalp hastalığı iken; hipoplastik sol kalp sendromu (%1,20) ve pulmoner atrezi/hipoplazi (%1,20) yüksek risk grubunda en sık tespit edilen kardiyak anomalilerdi. Serimizde doğumsal kalp hastalıklarının prevalansı düşük riskli ve yüksek riskli gebeliklerde sırasıyla %2,94 ve %6,37 olarak bulunurken, genel prevalans %5,16 idi.

Sonuç: Fetal ekokardiyografi, yeterli zamana sahip deneyimli kişiler tarafından yapıldığında çok yararlı bir tekniktir. Özel kardiyak tarama rutin anomali taramasının bir parçası olmalıdır. Ayrıca, düşük veya yüksek risk grubunda olan tüm gebe kadınlara ayrıntılı fetal ekokardiyografi yapılmalıdır.

Anahtar Kelimeler: Doğumsal kalp hastalıkları, fetal ekokardiyografi, sevk risk grubu, prevelans

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INTRODUCTION

Fetal cardiology represents one of the most exciting and rapidly evolving areas in the field of fetal medicine. Fetal echocardiography is completely noninvasive and harmless and is the best tool in this direction.[1] Congenital heart disease (CHD) is the most common congenital anomaly found in humans. In various studies, the incidence of CHD varies between approximately 4-50/1000 live births, of which 2.5-3/1000 live births present seriously ill in the neonatal period or early infancy and require specialist cardiac care.[2] Despite considerable advances in medical care, CHD is associated with substantial morbidity and death.[3] Accurate prenatal diagnosis of the CHD subtype is important to enable better clinical decision making, including prenatal management, recommendations for termination of the pregnancy, postnatal management of the patient, and for early referral to pediatric cardiology and cardiovascular surgery centers.[2,3] Also, prenatal diagnosis has been suggested to impart survival advantage in the great arteries (TGA), hypoplastic left heart syndrome (HLHS), and coarctation of aorta (CoA).[4-6] Additionally, fetal therapy can be offered for specific defects such as critical aortic stenosis and critical pulmonary stenosis, and various rhythm disturbances. Also, early termination can be offered if indicated which can help in reducing the incidence of complex CHDs.[2,7]

The sensitivity and specificity of fetal echocardiography have increased over time. A study of meta-analysis about first-trimester echocardiography in diagnosing CHDs, showed a sensitivity of 85% and specificity of 99%.[8] However, two meta-analyses of whole gestation fetal echocardiography also included first trimester echocardiography in diagnosing CHDs showed lower sensitivity values of 63.1% and 60.3%, respectively.[9,10] On the other hand, in a recent meta-analyses study of Yu et al. demonstrated that first trimester echocardiography is effective in diagnosing the CHDs, as a pooled sensitivity of 75% and a specificity of 99.9%.[11]

Although fetal echocardiography is established screening tool for the detection of cardiac anomalies, its utility and awareness remains less in most areas of the world. There is no data from our local region about the usage and outcome of fetal echocardiography. In this study, the distribution of pregnant women who underwent fetal echocardiography in our tertiary hospital according to low and high-risk factors, the reasons for admission and CHDs detected in fetal echocardiography were evaluated.

MATERIAL AND METHOD

Study population

The study was a retrospective research and was obtained from our University Medical Research Ethics Committee (Date: 23.09.2024, Decision No: 10-2024/02, Number: E-11095095-050.04-216395). It was in accordance with the Declaration of Helsinki. Written informed consent was obtained from all patients for the use of data from medical records.

This study is a retrospective research which was performed between January 2023 and September 2024. A total of 387 pregnant women who were in the 18th to 36th week of gestation were included in our study. The patients consisted of pregnant women who were referred by obstetricians due to various reasons which were classified as high-risk and low-risk groups.[12] The study population was analyzed in detail in terms of pregnancy history, reasons for applying to our clinic, use of any medication, presence of chromosomal or fetal anomalies, number and characteristics of previous pregnancies, presence of congenital or acquired heart diseases and family history. Also, all pregnant women were grouped as either high-risk or low-risk pregnancies according to these parameters.

Fetal echocardiography

Fetal heart examinations were performed using Philips Affiniti 50 (Philips Healthcare, Andover, Netherlands) echocardiography device with 2.5-5 MHz transducers by the same echocardiographic scanner. All fetal echocardiographic examinations were performed using standard techniques that determined the fetal position and heart axis and provided Doppler and M-mode measurements.[13,14] In all the cases, four chamber view, outflow-tract views, three vessel view, aortic and ductal arch views were done. Fetal heart rate was noted and any arrhythmia was confirmed with M-mode imaging, color Doppler and pulse-wave Doppler were used whenever necessary. All the images were recorded.

RESULTS

A total of 387 pregnant women were included in the study. **Table 1** shows low-risk and high-risk pregnancy groups according to referral criteria. Thus, 136 (35.13%) pregnant women were in the low-risk group and 251 (64.87%) were in the high-risk group. The most common reasons for referral in the high-risk group were maternal diabetes (13.17%), dysrhythmia (5.42%) and a previous history of a child or fetus with extracardiac anomalies (8.53%). However, in the low-risk pregnancies, lack of good image of the fetal heart by ultrasonography was the major reason for referral (20.93%).

The demographic data were summarized in **Table 2**. The comparison of the pregnant women included in the study according to their risk status revealed p=0.06. In the risk groups the mean ages were 27.43±1.37 and 28.54±5.45 years, respectively and no statistically significant difference was found between the groups (p>0.05). The mean gestational week was 22.73±2.25 and 23.61±1.61 weeks in the study population, respectively. Similarly, no statistical difference was detected for this parameter between the groups (p>0.05). The percentages of primiparas in low and high-risk groups were 65.44% and 58.56, respectively. Additionally, 2 and 5 pregnancies were multiple in the low-risk and high-risk groups, respectively.

Table 1: Distribution of pregnant women according to low-risk and high-risk factors.

Risk groups and factors	N: 387	%
1. Low risk group		
Lack of good image of the fetal heart by ultrasound	81	20.93
Suspicion of CHD during 2nd trimester ultrasound	41	10.59
Self-referral	14	3.61
Total	136	35.13
2. High risk group		
2.a. Maternal factors		
Maternal diabetes	51	13.17
In vitro fertilization	18	4.65
Multiple pregnancy	15	3.87
Maternal use of medicine	11	2.84
Advanced maternal age	13	3.35
Maternal CHD	7	1.81
Maternal rheumatologic diseases	5	1.29
Maternal TORCH infections	1	0.25
2.b. Fetal factors		
Dysrhythmia	21	5.42
Polyhydramniosis, oligohydramniosis	13	3.35
Fetal extracardiac anomaly	12	3.10
Chromosomal anomaly	2	0.51
Increased nuchal translucency	3	0.77
2.c. Hereditary factors		
Previous child or fetus with CHD	21	5.42
Previous child or fetus with extracardiac anomaly	33	8.53
Familial CHD (excluding parents and siblings)	25	6.45
Total	251	64.87

CHD; congenital heart diseases, TORCH; Toxoplasma, O (others), Rubella, Cytomegalovirus, Herpes simplex virus.

Table 2: The demographic data of study population.

	Low risk group (N:136)	High risk group (N:251)	p value
Age (years)	27.43±1.37 (19-44)	28.54±5.45 (21-44)	>0.05
Gestational week (weeks)	22.73±2.25 (17-36 weeks)	23.61±1.61 (17-36 weeks)	>0.05
Primipara (N/%)	89/65.44	147/58.56	>0.05
Multipara (N/%)	45/34.56	99/41.44	>0.05
Multiple pregnancy (N/%)	2/1.47	5/1.99	>0.05

In the low-risk group; ventricular septal defect was detected in 4 cases (2.94%) (**Table 3**). However; ventricular septal defect was detected in 2 cases (0.79%), double outlet right ventricle in 2 cases (0.79%), hypoplastic left heart syndrome in 3 cases (1.20%), tricuspid atresia in 2 cases (0.79%), pulmonary atresia/hypoplasia in 3 cases (1.20%), tricuspid atresia and TGA in 1 case (0.40%), aortic coarctation/aortic arch hypoplasia in 1 case (0.40%), corrected TGA in 1 case (0.40%) and truncus arteriosus in 1 case (0.40%) were detected in the high-risk group (**Table 3**). So, the prevalence of CHD in low-risk and high-risk pregnancies were found as 2.94% and 6.37%, respectively. Also, we found the overall prevalence of CHD in our study population as 5.16%.

Table 3: Distribution of congenital heart diseases according to low-high risk groups

Congenital heart disease	Low risk group (N:136)	High risk group (N:251)
Ventricular septal defect	4 (2,94%)	2 (0.79%)
Double outlet right ventricle	-	2 (0.79%)
Hypoplastic left heart syndrome	-	3 (1.20%)
Tricuspid atresia	-	2 (0.79%)
Pulmonary atresia/hypoplasia		3(1.20%)
Tricuspid atresia and transposition of great arteries (TGA)	-	1 (0.40%)
Aortic coarctation/aortic arch hypoplasia	-	1 (0.40%)
Corrected TGA	-	1(0.40%)
Truncus arteriosus	-	1(0.40%)
Total	4(2.94%)	16(6.37%)

TGA; Transposition of great arteries

Rhythm disturbances were also detected in our study. By this way, premature atrial extra systoles were detected in 3 fetuses in low-risk group while premature ventricular beats were detected in 5 pregnant women in high-risk groups.

DISCUSSION

Today, fetal echocardiography has become increasingly used. The sensitivity of fetal echocardiography performed by experienced hands is also high.[9,10] With fetal echocardiography, not only simple CHDs but also complex CHDs and situs anomalies can be detected prenatally.[15] Also, fetal echocardiography helps to improve the pregnancy outcome of fetuses with selected CHD, and there is a clinical benefit with regard to infant outcomes.[13-16] The diagnosis of CHDs from fetal echocardiography is dependent on several factors including the skill of the operator, indications for referral, makeup of the population and the ultrasonographic skill of the referring obstetricians.

The reported incidence of CHD in different studies varies from 4 to 50/1000 live births,[2] with the generally the accepted incidence being 8/1000 live births.[17] Geographic and ethnic differences in the incidence of CHD subtypes have been reported.[18,19] The variations in particular congenital heart lesions could have a biological basis and may be related to different genetic/epigenetic susceptibility.[20,21]

The mean ages of our patients were 27.43±1.37 and 28.54±5.45 years in each risk groups, respectively. Also, these results are comparable to other studies.[22] The main indications for referral were lack of good image of the fetal heart by ultrasonography in low-risk group and maternal diabetes in high-risk group. There were also high prevalence of dysrhythmia (5.42%) and history of previous child or fetus with extracardiac anomaly (8.53%) in our high-risk population. These results were also similar with the studies which have shown fetal extracardiac anomaly

scan to be the most common indication for referral.[23,24] Our tertiary center is being the apex center of surrounding countries which especially gets referrals of high-risk pregnancies. Additionally, these results were the first reports of our tertiary center.

Various studies have reported different frequencies of CHD in pregnant women at different risk groups. In a study conducted by Özbarlas et al. in high- and low-risk pregnant women, the frequency of CHD was found to be 7.8% and 2.7%, respectively.[25] Also, in a recent study of Altın these frequencies were found as 15.2% in the high-risk group and 9% in the low-risk group (p=0.008).[26] Similarly, in our series, the prevalence of CHD was found to be 2.94% in low-risk pregnancies and 6.37% in high-risk pregnancies, while the overall prevalence was found to be 5.16%. On the other hand, in the studies conducted by Özkutlu et al.[27] and Özbarlas et al.[25] ventricular septal defect was the most frequently detected congenital heart disease. Similarly, in our study, ventricular septal defect was the most frequently detected CHD in both the high-risk and low-risk groups.

The fact that some heart anomalies are seen in the baby at a rate much higher than expected if the mother has a disease suggests the possibility of cytoplasmic (mitochondrial) transmission in these cases.[18,19] When the mother's metabolic diseases and the drugs she uses are considered as environmental factors for the fetus; diabetes mellitus, phenylketonuria, lupus, rubella, alcohol, lithium, amphetamines and antiepileptic drugs significantly increase the incidence of CHD.[13,16,25,27] There is also a relationship between chromosome structural disorders and congenital heart anomalies.[18,19]

In the study of Özkutlu et al., the risk factors such as maternal metabolic diseases, fetal arrhythmias and previous CHD in fetuses or children were found to be the most common risk factors in high-risk pregnant women.[27] In the study of Özbarlas et al., the most common risk factors were found to be maternal metabolic diseases, previous CHD in fetuses or children and non-cardiac fetal malformations.[25] In our study, the most common risk factors were diabetes due to maternal causes, dysryhtmia due to fetal causes and fetal anomalies in previous pregnancies due to hereditary causes. In addition, in our study, the most common reason for application in the low-risk group was found to be lack of good image of the fetal heart by ultrasound, in accordance with the literature. In the light of our results, we suggest that pregnancies with diabetes should be evaluated by fetal echocardiography regularly in certain gestational weeks.

Limitation

This study was limited by its retrospective nature at a single academic center and relatively small sample size. Also, information such as maternal and familial hereditary factors was lacking and was not evidence based.

CONCLUSION

Fetal echocardiography is a very useful technique when performed by experienced individuals with sufficient time. In the presence of CHD detected by fetal echocardiography, the family can be given the necessary counseling and precautions can be taken in advance for postnatal cardiac procedures. Fetal echocardiography should definitely be performed especially in high-risk pregnant women. However, the low prevalence of CHD in low-risk group, both in our study and in most studies in the literature, makes fetal echocardiography a priority for this group. However, it is necessary to properly identify selected cases in this patient group.

ETHICAL DECLARATIONS

Ethics Committee Approval: The study was obtained from Karamanoğlu Mehmetbey University Medical Research Ethics Committee (Date: 23.09.2024, Decision No: 10-2024/02, Number: E-11095095-050.04-216395).

Informed Consent: Signed written informed consent was taken from all participants.

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