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# A Retrospective Analysis of Fetal Cardiovascular System Anomalies in a Tertiary Center

## Üçüncü Basamak Bir Merkezde Fetal Kardiyovasküler Sistem Anomalilerinin Retrospektif Analizi

İbrahim Ömeroğlu<sup>1</sup>, Hakan Gölbaşı<sup>1</sup>, Elif Uçar<sup>2</sup>, Barış Sever<sup>1</sup>, Halil Gürsoy Pala<sup>1</sup>

<sup>1</sup>University of Health Sciences Turkey, İzmir Tepecik Education and Research Hospital, Clinic of Perinatology, İzmir, Turkey

<sup>2</sup>Siverek State Hospital, Clinic of Obstetrics and Gynecology, Şanlıurfa, Turkey

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### Abstract

**Objective:** We shared the frequency and our experiences of cardiac anomalies detected during routine ultrasound scanning in pregnant women who applied to our perinatology clinic.

**Methods:** 288 pregnant women who were admitted to İzmir Tepecik Education and Research Hospital Perinatology Clinic between January 2017 and January 2020 due to high-risk pregnancy and who were found to have cardiac anomalies included. The data were retrospectively scanned from the hospital digital archive and patient files.

**Results:** The demographic data of the pregnant women included in the study such as age, gestational week, gravida, and parity and the anomalies detected categorized. The mean age of the pregnant women was calculated as 27.99 (16-46) and the mean week of gestation as 25.2 (14-40). While the average gravida of the pregnant women was 2.29 (1-8), the mean parity was 0.97 (0-7). The most common cardiovascular system (CVS) anomaly was septal defects with a rate of 29.5%. When septal defects were examined, the most common ventriculoseptal defect (62.3%) followed by an atrioventriculoseptal defect (27.05%) was found.

**Conclusion:** Three-dimensional ultrasonography has replaced two-dimensional ultrasonography with the developing technology and increasing physician experience. CVS anomalies can be detected safely with fetal echocardiography in three-dimensional ultrasonography. Early diagnosis and routine-detailed screening of pregnant women gain importance to determine the prognosis of detected structural anomalies and to provide healthy counseling service.

**Keywords:** Cardiac anomaly, septal defect, fallot tetralogy, echocardiography

### Öz

**Amaç:** Perinatoloji kliniğimize başvuran gebelerde rutin ultrasonografi taraması sırasında saptanan kardiyak anomalilerin sıklığını ve deneyimlerimizi paylaşmayı amaçladık.

**Yöntem:** Ocak 2017-Ocak 2020 tarihleri arasında İzmir Tepecik Eğitim ve Araştırma Hastanesi Perinatoloji Kliniği'ne yüksek riskli gebelik nedeniyle başvuran ve kalp anomalisi saptanan 288 gebe çalışmaya dahil edildi. Veriler hastane dijital arşivinden ve hasta dosyalarından geriye dönük olarak tarandı.



**Address for Correspondence/Yazışma Adresi:** İbrahim Ömeroğlu MD, University of Health Sciences Turkey, İzmir Tepecik Education and Research Hospital, Clinic of Perinatology, İzmir, Turkey

**Phone:** +90 506 854 34 70 **E-mail:** dribrahimomeroglu@gmail.com

**ORCID ID:** orcid.org/0000-0001-9200-0208

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## Öz

**Bulgular:** Çalışmaya alınan gebelerin yaş, gebelik haftası, gravida ve parite gibi demografik verileri ve saptanan anomaliler kategorize edildi. Gebelerin ortalama yaşı 27,99 (16-46) ve ortalama gebelik haftası 25,2 (14-40) olarak hesaplandı. Gebelerin gravida ortalaması 2,29 (1-8) iken, ortalama parite 0,97 (0-7) idi. En sık görülen kardiyovasküler sistem (KVS) anomalisi %29,5 ile septal defektlerdi. Septal defektler incelendiğinde en sık ventriküloseptal defekt (%62,3), ardından atriyoventriküloseptal defekt (%27,05) saptandı.

**Sonuç:** Üç boyutlu ultrasonografi, gelişen teknoloji ve artan hekim deneyimi ile iki boyutlu ultrasonografinin yerini almıştır. Üç boyutlu ultrasonografide fetal ekokardiyografi ile KVS anomalileri güvenle tespit edilebilir. Tespit edilen yapısal anomalilerin prognozunu belirlemek ve sağlıklı bir danışmanlık hizmeti verebilmek için gebelerin erken teşhisi ve rutin detaylı taramaları önem kazanmaktadır.

**Anahtar Kelimeler:** Kardiyak anomali, septal defekt, fallot tetralojisi, ekokardiyografi

## Introduction

Congenital malformations occur when fetal structures develop differently from normal in terms of shape, structure and function during morphogenesis<sup>(1)</sup>. Congenital anomalies are seen in approximately 3% of all newborns<sup>(2)</sup>. Cardiovascular system (CVS) anomalies are the most common congenital anomalies<sup>(3)</sup>. Cardiac anomalies are seen in 0.4-1.3% of live births<sup>(4)</sup>. This rate is higher in stillbirths and premature babies. One-third of perinatal deaths are due to cardiac anomaly<sup>(5)</sup>.

Most of the anomalies are detected in healthy pregnant women without any risk factors. Genetic factors, environmental factors, maternal diseases, alcohol, smoking, toxic drugs taken during pregnancy, radiation, and infections in the antenatal period lie in the background of the minority. There are types of cases whose treatment is available, as well as severe types that can result in death when not diagnosed. When it is associated with chromosomal abnormalities and genetic syndromes, it can significantly change the clinical outcomes of the affected fetus<sup>(6)</sup>. Therefore, early diagnosis of CVS anomalies and karyotype analysis is critical. According to the data collected by the World Health Organization between 1950 and 1994, 42% of child deaths were determined due to cardiac anomalies<sup>(7)</sup>.

It has been reported that cardiac anomalies were detected with a rate of 85.7% between the 11<sup>th</sup> and 14<sup>th</sup> week of gestation<sup>(8)</sup>. Since heart defects such as ventricular septal defects (VSD), ebstein's anomaly, cardiac tumors cannot be seen before the 14<sup>th</sup> gestational week, ideal cardiac scanning is performed between the 18<sup>th</sup> and 22<sup>nd</sup> gestational weeks. It is accepted that the evaluation of cardiac structures becomes difficult due to the occurrence of various factors such as oligohydramnios and the progression of ossification in later weeks. In this screening, it is an important point to evaluate the heart as a whole, to look at the four chambers

and outlets. Examination of the outflow tracts enables the detection of conotruncal anomalies such as tetralogy of fallot, transposition of the great artery, and truncus arteriosus<sup>(9-11)</sup>. The most common VSD is followed by ASD and PDA in the 2<sup>nd</sup> and 3<sup>rd</sup> place.

With the advancing technology and the increasing experience of clinicians, the rate of detection of cardiac anomalies has increased and a goal of antenatal care has been achieved. Detailed anamnesis of the pregnant woman should be taken. Risk factors should be determined, the fetal prognosis should be determined in ultrasonography, and the family should be informed.

The current study presents the frequency, type and distribution of CVS anomalies detected in our high-risk pregnant women evaluated in our perinatology council.

## Materials and Methods

The study included cases who applied to the Perinatology Clinic of İzmir Tepecik Education and Research Hospital between January 2017 and January 2020 because of high-risk pregnancy and who were found to have cardiac anomalies. The cases were reviewed retrospectively from hospital records and patient files.

## Statistical Analysis

Data analysis was performed using the Statistical Package for Social Sciences 20 programs. Variables were expressed in n (frequency) and percentage (%).

## Results

In our study, data of 81,163 pregnant women who applied to our perinatology outpatient clinic were analyzed. Among these patients, 288 pregnant women with CVS anomaly were detected. The demographic data of the pregnant women included in the study, such as age, gestational week,

gravida, and parity, are shown in Table 1. The mean age of the pregnant women included in our study was calculated as 27.99 (16-46), and the mean week of gestation as 25.2 (14-40). While the average gravida of pregnant women was 2.29 (1-8), the mean parity was 0.97 (0-7) (Table 1).

The most common CVS anomaly was septal defects with a rate of 29.5%. Subsequently, left vessel malformations (20.1%), conotruncal malformations (18.75%) and myocardial-pericardial malformations (17.7%) were detected (Graphic 1).

When septal defects were examined, ventriculoseptal defect was the most common (62.3%), followed by atrioventruloseptal defect (27.05%). It was observed that 58% of the cases with left heart malformations were hypoplastic left heart (HLHS) and 34% were aortic coarctation/interrupted aortic arch. It was observed that 31% of conotruncal malformations were double outlet right ventricle, 25% were tetralogy of fallot, 25% were great artery transposition, and 16% were truncus arteriosus. 49% of myocardial and pericardial anomaly cases are echogenic focus, 39% pericardial effusion, 5.8% hypertrophic cardiomyopathy, 5.8% dilated cardiomyopathy (Table 2).

## Discussion

The frequency of congenital anomalies in our country varies according to diet, environmental factors, socioeconomic level, familial factors, and teratogenic agents exposed during

pregnancy. While congenital anomalies are held responsible for approximately 10-12% of infant deaths, 50% of these are CVS anomalies<sup>(12)</sup>. Cardiac anomalies are also the most common undetected anomaly group. Detailed examination of the fetus has gained momentum in the last 30-40 years, and the vast majority of them have become detectable with today's technology and increasing physician experience. Early prenatal diagnosis of fetal anomaly is important in determining the group that can be treated after birth and deciding the pregnancies that should be terminated if necessary.

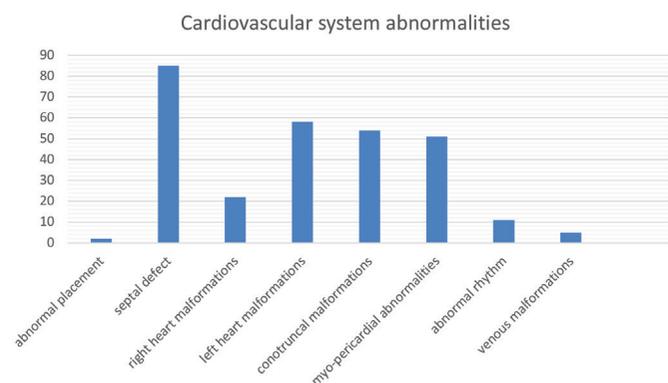
Due to regional factors and the fact that our hospital is a tertiary hospital, we have a large patient population, so the cases with CVS anomaly are 0.035% of all our cases. In the literature, this rate is 0.5% in the study by Bronshtein et al.<sup>(13)</sup> and Todros et al.<sup>(14)</sup> found CVS anomalies in 4.9% in their study. Perri et al.<sup>(15)</sup> found 2.7% in their study. Tutuş<sup>(16)</sup> found cardiac anomalies with a rate of 9.6% in their study. We think that the reason for the low rate in our study is the higher number of screened pregnant women compared to other studies.

In our study, while the average diagnosis weeks in terms of cardiac anomaly was 25.2, this mean was found to be 24.7±5.2 in the study of Binboğa<sup>(17)</sup> we observed that our study was compatible with the literature.

The most common CVS anomalies found in our study were VSD (53 patients, 18.4%), atrioventricular septal defect (AVSD) (23 patients, 7.9%), HLHS (34 patients, 11.8%) as we determined. In a study by Paladini et al.<sup>(18)</sup>, the most frequently detected cardiac pathologies were VSD (20.8%), HLHS (10.8%), and AVSD (10.5%). In the study by Hallioğlu et al.<sup>(19)</sup>, VSD (38.8%) and AVSD (11.9%) are the most common pathologies. In the study of Ozbarlas et al.<sup>(20)</sup>, it was observed that the most frequently detected pathologies were VSD (36.2%) and AVSD (14.1%). Persico et al.<sup>(21)</sup> found AVSD as the most common major anomaly. Chu et al.<sup>(22)</sup> grouped cardiac anomalies as low and high risk and found VSD most frequently in both groups. The results of our study are compatible with the literature, and we think that the reason for our lower rates compared to other studies is the high number of patients we screened and the fact that not all pregnant women who applied to our clinic were high-risk patients.

**Table 1. Demographic data**

	Minimum	Maximum	Mean
Age	16	46	27.99
Gravida	1	8	2.29
Parity	0	7	0.97
Pregnancy week	14	40	25.2



**Graphic 1.** Distribution of cardiovascular system anomalies detected

## Study Limitations

Our study included some limitations. One of the biggest deficiencies of our study is that we could not compare

**Table 2. Detailed distribution of CVS anomalies in our study**

Abnormal placement	Situs inversus	1
	Heterotaxy syndrome	1
Septal defects	VSD	53
	AVSD	23
	Foramen ovale aneurysm	1
	Hypoplastic right heart	8
Right heart malformations	Ebstein anomaly	8
	Tricuspid atresia	10
	Tricuspid dysplasia	4
Left heart malformations	Hypoplastic left heart	34
	Aortic coarctation/intermittent aortic arch	20
	Aortic stenosis	4
Conotruncal malformations	Tetralogy of fallot	14
	Bat	14
	Truncus arteriosus	9
	Double outlet right ventricle	17
Myocardial and pericardial abnormalities	Echogenic focus	25
	Hypertrophic kmp	3
	Dilate kmp	3
	Pericardial effusion	20
Abnormal rhythm	Rhythm disorders	4
	Tachyarrhythmias	2
	Bradyarrhythmias	5
Venous malformations	Total pulmonary venous return anomaly	5

CVS: Cardiovascular system, VSD: Ventricular septal defects, AVSD: Atrioventricular septal defect

the anomalies we detected with the postpartum results. Although patients with suspected anomalies are called to echocardiography (ECHO) by the pediatric cardiologist for the postpartum period, our postpartum records are insufficient due to the patients not coming for postpartum control and fetal deaths. The refusal of the families for whom the termination of pregnancy recommended due to fetal major cardiac defects, and the inability to perform a postmortem examination in families who accepted termination limited the effectiveness of the study.

## Conclusion

While cardiac anomalies cannot be easily detected even in today's conditions, this ratio is tried to be reduced to minimum values with the developing technology and additional diagnostic tests. Despite these, about 40% of major anomalies cannot be detected in ultrasonography. It is thought that 3D ultrasonography is more beneficial than two-dimensional ultrasonography because large vessel anomalies and venous system can be evaluated in different

planes. Fetal ECHO is a reliable method for CVS anomalies. When a structural anomaly is detected, counseling with a perinatologist and geneticist should be provided to the pregnant woman in a timely and impartial manner, in a manner that respects the socioeconomic level, culture and beliefs of the family. This is necessary not only for termination of cases with poor prognosis, but also for a better delivery, postnatal care and prognosis of ongoing pregnancies.

## Ethics

**Ethics Committee Approval:** This study was approved by the İzmir Tepecik Education and Research Hospital Ethics Committee (20201/02-02, date: 22.02.2021).

**Informed Consent:** Retrospective study.

**Peer-review:** Externally peer-reviewed.

## Authorship Contributions

Concept: İ.Ö., HG., E.U., B.S., H.G.P., Design: İ.Ö., HG., E.U., B.S., H.G.P., Data Collection or Processing: İ.Ö., HG., E.U., B.S.,

H.G.P., Analysis or Interpretation: İ.Ö., HG., E.U., B.S., H.G.P., Literature Search: İ.Ö., HG., E.U., B.S., H.G.P., Writing: İ.Ö., HG., E.U., B.S., H.G.P.

**Conflict of Interest:** No conflict of interest was declared by the authors.

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