

# FETAL ECHOCARDIOGRAPHY: MOST COMMON FINDINGS

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## ABSTRACT

**INTRODUCTION:** Congenital cardiopathies or congenital heart disease (CHD) has a prevalence of about 0.8% of live births, being responsible for about 40% of perinatal deaths. Prenatal diagnosis of CHD can be performed using echocardiography, as this test has a sensitivity of 43% to 85% for visualization of the four cardiac chambers.

**OBJECTIVE:** To define the most frequent findings of abnormalities among the heart diseases observed on fetal echocardiography, to establish the most frequent maternal age group in fetal echocardiography exams and to establish the frequency of the altered findings.

**METHODS:** Retrospective, cross-sectional study that evaluated the importance of echocardiography for the early diagnosis of cardiac pathology in fetuses, as well as its prevalence in the results of 1701 reports of electronic medical records of patients who underwent fetal echocardiography at Clínica Fértil, in Goiânia, Goiás, between 01/01/2015 to 12/31/2019. The variables analyzed were the altered findings found, the maternal age and the frequency of each finding.

**RESULTS:** The age group with the highest incidence of changes was 18 to 34 years old, the frequency of changes found was 8.3% and the most frequent change was interventricular communication and cardiomegaly, both with 16.2%.

**CONCLUSION:** The frequency of altered findings according to the proposed study was 8.3%, the maternal age group with the largest number of changes on fetal echocardiographic examination was between 18 and 34 years old. The most frequent finding was without abnormalities. As for the most frequent findings of abnormalities, interventricular communication and cardiomegaly, were the most common findings.

**KEYWORDS:** FETAL ECHOCARDIOGRAPHY, CONGENITAL HEART DISEASE, PRENATAL.

## INTRODUCTION

Fetal echocardiography is a method of excellence and high accuracy for the diagnosis of cardiac and circulatory abnormalities in the fetus<sup>1</sup>. This exam has a sensitivity of 43% to 85% for the visualization of the four cardiac chambers<sup>2</sup>. Given the increased risk of morbidity and mortality in babies with congenital heart disease (CHD), an accurate prenatal diagnosis is essential to help plan peripartum management, as it improves survival after surgery and neurological outcomes<sup>3</sup>.

According to the American College of Cardiology, the main indications for fetal echocardiography are fetal heart abnormalities or arrhythmia detected by routine prenatal ultrasound, family history of congenital heart disease, maternal diabetes or systemic lupus erythematosus, fetal exposure to a teratogen, altered fetal karyotype and other abnormalities of the fetal system<sup>4</sup>. An additional indication for this procedure is in those fetuses with suspected coronary disease or extracardiac abnormality detected at the time of scanning the fetal anatomy of the second trimester<sup>3</sup>. Despite the recognition of these risk factors, only 15 to 30% of cardiac

defects are detected before birth<sup>4</sup>.

The prenatal diagnosis of congenital heart disease (CHD) has shown to have a significant effect on prenatal and postnatal management and outcomes. In addition to the potential medical benefits, fetal diagnosis allows valuable advice to parents, which allows families to make informed decisions regarding pregnancy and to prepare emotionally for the birth of the child with significant CHD<sup>5</sup>.

Referral to fetal echocardiography typically occurs between 18 and 22 weeks of gestational age. In addition, with the wide availability and practice of nuchal translucency (NT) measures, which generally occur between 11 and 14 weeks of gestational age, the demand for early fetal cardiac images has increased, but it is not a standard practice<sup>5</sup>.

The main cardiopathies are: congenital malformations of the cardiac chambers and connections, congenital malformations of cardiac septum, congenital malformations of the lungs and tricuspid valves, congenital malformations of the aorta and mitral valves, congenital malformations of the great arteries, congenital malformations of the great veins<sup>3</sup>.

Congenital heart disease occurs in nine out of 1,000 live

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births. Around 25% of cases are severe heart diseases that require intervention in the first year of life. Newborns with congenital heart disease represent a high-risk group due to high mortality and morbidity. Early diagnosis and immediate initiation of treatment minimizes the risk of the child's hemodynamic deterioration, even preventing other organs from being injured, the most important of which being the central nervous system<sup>6</sup>.

Therefore, the objective of the present study is to evaluate the most frequent cardiopathies and the importance of fetal echocardiography in the screening of human heart diseases.

**METHODS**

This is a cross-sectional, descriptive, retrospective study conducted at Clínica Fértil, in the city of Goiânia - GO.

The observed universe consists of patients seen at the clinic for screening fetal ultrasound with a sample for convenience according to the demand, established from January 2015 to December 2019. Inclusion criteria were pregnant women with indication for fetal ultrasound. Exclusion criteria were pregnant women with other ultrasound indications.

For data collection, reports of the conclusion and observation of the fetal echocardiography exams were used, as well as the maternal age, located and saved in the Ultra System 3.8.1 program.

The data were analyzed with the aid of the statistical package SPSS, (26.0). The normality of the data was tested using the Kolmogorov-Smirnov test. The prevalence of heart disease according to the age group and period of the study was performed using absolute frequency (n) and relative frequency (%) using Pearson's chi-square test. The prevalence of the type of heart disease was based on the cumulative relative frequency. Pearson's correlation was used to verify the relationship between the number of heart diseases and the age of the patients. The level of significance adopted was 5% (p <0.05).

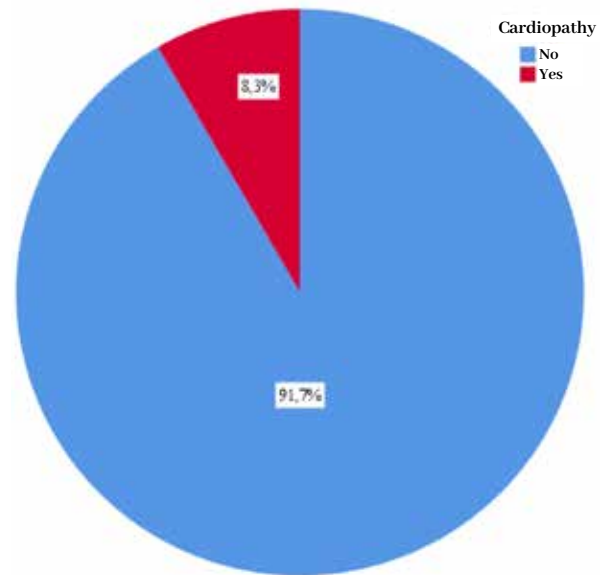
No patient identification was performed and the only variables analyzed were age and echocardiogram report.

Due to the large number of cardiopathies found, it was chosen to categorize "others", exams with a frequency of onset less than four.

As for ethical aspects, it is highlighted that the research will be based on Resolution n. 466/2012, and the rights of those involved are ensured, being approved by the Ethics Committee by the number 4,196,514.

**RESULTS**

In a period of five years, 1701 fetal echocardiography exams were analyzed in search of the most frequent findings. There was an absence of heart disease in 91.7% of the exams. Heart diseases were found in 8.3% of the exams. Most of the results found were without altered findings (Graph 1).



Graph 1- Relative frequency of the prevalence of fetal heart disease in the population studied.

Among the age groups analyzed, the range with the highest incidence of changes was 18 to 34 years, representing 74.2% of cases. In absolute numbers, the age group that most performed the exam was pregnant women between 18 and 34 years of age. There was no statistical difference in the relationship between the finding of heart disease and age, and with the distribution per year of the exam (Table 2).

	Cardiopathy n (%)		Total n = 1701	p*
	No	Yes		
<b>Age Group</b>				
< 18	12 (0,8)	3 (2,1)	15 (0,9)	
18 a 34	1159 (74,3)	104 (73,2)	1263 (74,2)	0,26
≥ 35	389 (24,9)	35 (24,6)	424 (24,9)	
<b>Year</b>				
2015	238 (15,3)	32 (22,5)	270 (15,9)	
2016	306 (19,6)	26 (18,3)	332 (19,5)	
2017	289 (18,5)	33 (23,2)	322 (18,9)	0,06
2018	429 (27,5)	30 (21,1)	459 (27,0)	
2019	297 (19,1)	21 (14,8)	318 (18,7)	

\* Pearson's chi-square; n = absolute frequency; % = relative frequency

Table 2. Description of the prevalence of heart disease according to the age group and period of the study.

The average age group in the study was 30.33, with a standard deviation of 5.84 years (Figure 2).

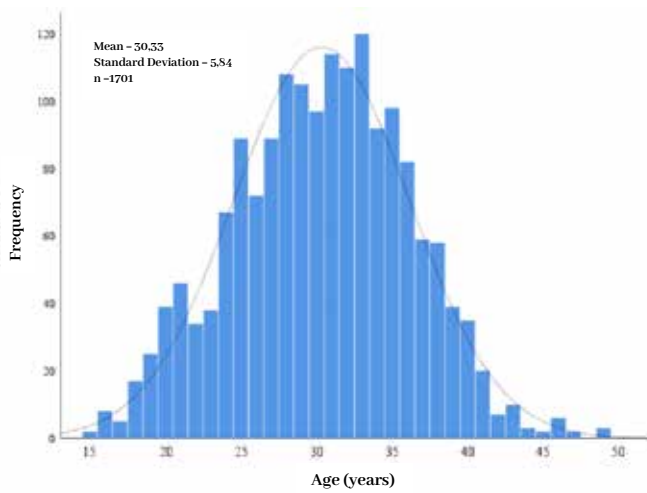


Figure 2. Histogram graph showing the age distribution of the patients

Trend analysis was carried out to assess whether there was a trend in the reduction in the finding of heart diseases over the years. A negative trend was found for the reduction of heart diseases, with  $p = 0.30$ . Therefore, it cannot be said that there has been a decrease in the prevalence of heart disease over the years (Figure 3).

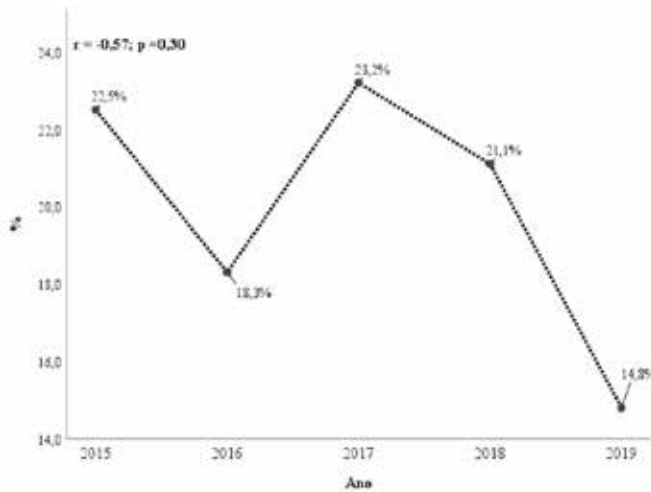


Figure 3. Bar graph showing the prevalence of fetal heart diseases in the period from Jan 2015 to Dec 2020.

In the group of fetuses with anomalies, the most frequent cardiopathies were interventricular communication (16.2%) and cardiomegaly (16.2%), followed by pericardial effusion (10.6%), golf ball (8.5%), hypoplastic left heart syndrome (6.3%), endocardial cushion (6.3%), dilation of the pulmo-

nary trunk (5.6%), congestive heart failure (4.9%), interatrial communication (4.2%) and tetralogy of fallot, transposition of the great vessels and single atrium (2.8%). "Others" were considered cardiac anomalies found at a frequency lower than four exams (Table 3).

Cardiac Anomalies	N	%
Interatrial communication	6	4,2
Interventricular communication	23	16,2
Tetralogy of Fallot	4	2,8
Transposition of the great vessels	4	2,8
Single atrium	4	2,8
Cardiomegaly	23	16,2
Endocardial cushion	9	6,3
Pericardial effusion	15	10,6
Dilatation of the pulmonary trunk	8	5,6
Golf Ball	12	8,5
Hypoplastic left heart syndrome	9	6,3
Congestive heart failure	7	4,9
Others	82	57,7

n = absolute frequency; % = relative frequency

Table 3. Distribution of cases of fetal heart disease according to the type of cardiopathy (n = 142).

Of the women who had heart disease, a positive correlation was observed in the cumulative number of cardiopathies and age. The greater the age, the greater the number of cardiopathies in cumulative terms, with  $p = 0.02$ , and  $r = 0.19$  (Figure 4).

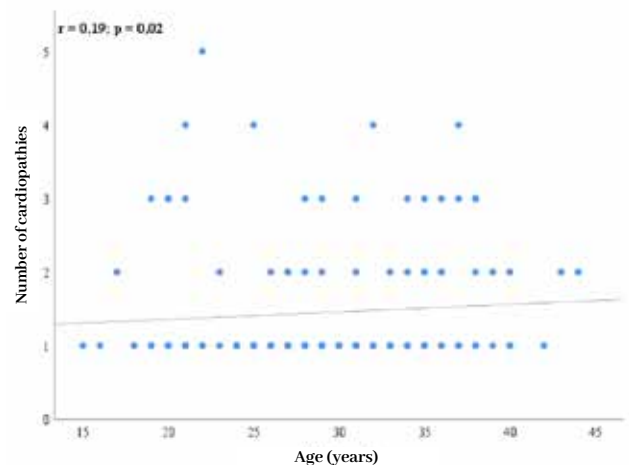


Figure 4. Scatterplot showing Pearson's correlation of age with the number of fetal cardiopathies.

## DISCUSSION

Congenital cardiopathies or congenital heart disease (CHD) are among the most common congenital anomalies, with a prevalence of about 0.8% of live births<sup>2</sup>. Satomi et al. also report that since this statistic does not include abortions or stillbirths, it can be inferred that the actual number of fetuses with heart defects is almost five times higher than what has been reported<sup>7</sup>.

Due to their poor prognosis, they contribute significantly to infant mortality, becoming responsible for about 10% of infant deaths and half of deaths due to congenital malformation<sup>8</sup>.

Such diseases are significantly associated with perinatal morbidity and mortality, accounting for about 40% of perinatal deaths. The intrauterine diagnosis of cardiac alterations allows the doctor to have information related to the characteristics of the disease, its evolution, therapeutic possibilities and prognosis; in addition to recurrence for future pregnancies.

In this study, it was possible to analyze the importance of echocardiography in the diagnosis of congenital heart diseases. The prevalence of heart disease was observed in 8.3% of pregnancies in the tests performed, that is, of the 1701 tests evaluated in the last five years 142 diagnosed some heart disease. Bahtiyar and Copel are emphatic in saying that despite their high prevalence, CHD is not identified as it should be during prenatal care<sup>9</sup>. The early diagnosis of CHD remains low, compared to the diagnosis of other types of congenital structural malformations, since echocardiography is commonly indicated for patients with high-risk pregnancies<sup>10,11</sup>.

Fetal echocardiography is traditionally indicated for high-risk pregnant women, but most newborns with heart disease are still born undiagnosed in all parts of the world. This is because many cases of congenital heart disease occur in low-risk groups and are not detected by screening at the time of prenatal ultrasonography<sup>12</sup>.

The prenatal diagnosis of CHD can be performed by means of echocardiography, since this exam has a sensitivity of 43% to 85% for the visualization of the four cardiac chambers<sup>2,7,11</sup>.

Nayak et al report that fetal echocardiography is an exam that requires time and requires experienced examiners<sup>10</sup>. In this study, all exams were performed by the same examiner, which hides the selection bias. In addition, there was no access to the indication of the exam; they are patients who arrived due to spontaneous demand. However, despite recognizing the importance of this test and that many heart diseases exist in low-risk pregnancies, there is no formal indication in the literature about the same being indicated for all patients, therefore, doctors indicate this test when there is a known risk factor. Therefore, it is believed that the patients in the study had some risk factor for the examination to have been indicated and because of this, they found the rate of heart diseases similar to that of the current literature.

When analyzing maternal age compared to the findings, the maternal indications for performing fetal echocardiography should be highlighted: family history of congenital heart disease, metabolic disorders (diabetes, thyroid disease), exposure to teratogens, exposure to prostaglandin synthase inhibitors (ibuprofen, salicylic acid), rubella infection, autoimmune disease (SLE, Sjogren), family hereditary disorders (Ellisvan Creveld, Marfan) and in vitro fertilization. There are no specific prenatal markers to identify the fetus with congenital heart disease. The increase in nuchal translucency between 10 and 13 weeks of gestation has been associated with an increased risk of congenital heart disease.

Early diagnosis helps both in prenatal and postnatal management and in counseling for parents; as well as decreasing morbidity and mortality rates, since it allows treatments to be implemented early<sup>7</sup>.

According to Mogra et al, major cardiac abnormalities are potentially lethal or require surgical treatment in the first year of life and affect about four born between 1000<sup>13</sup>. Holland, Myers and Woods claim that prenatal diagnosis reduced the risk of death before planned cardiac surgery in relation to patients with postnatal diagnosis<sup>14</sup>. Additional studies and efforts to improve the prenatal diagnosis of congenital heart diseases should be considered. For this reason, the importance of this study, because through it, in a five-year study in a selected population, a rate of 8.1% has already been found, probably if this test was indicated for the population in general, the prevalence found would be different. It would increase.

The fetal echocardiography exam is a high-cost exam, which requires an experienced examiner and is not present and accessible to the general population.

Contrary to age logic, which relates more advanced ages to the involvement of congenital disorders (higher prevalence of metabolic disorders, in vitro fertilization, greater exposure to teratogens), the study presents the age group over 35 years with the lowest incidence of changes, 24.9% compared to the age group from 18 to 34 years old with 74.5%. The average age found was 30.33 years, with a standard deviation of 5.84 years. Perhaps this finding occurred due to the greater number of tests performed in this age group, with 1263 tests performed between 18 and 34 years old and 424 over 35 years old. The age group with the lowest prevalence of CHD were those under 18 years old, yet, also with a smaller number of patients,  $n = 15$ .

In this study, regarding the age group, there was no statistically significant difference in the relationship between maternal age and heart diseases, with  $p = 0.26$ . However, when comparing maternal age and cumulative heart diseases, an  $r = 0.19$  was observed, that is, the higher the maternal age, the greater the prevalence of more than one heart disease in the same fetus, this evaluation being statistically significant with  $p = 0.02$ .

There was also a tendency of reduction of heart disease in this study, with respect to the last five years, with  $r = -0.57$ .



However,  $p = 0.30$ , showing no statistical significance. This data also differed from the current literature, in which the tendency is to increase the number of cardiopathy findings, due to the improvement of ultrasound devices and greater access to the population to them.

With regard to the types of CHD's and their frequencies in the populations studied, Wei et al show that the five most frequent defects in their study are: single ventricle (15.9%, 31/195), atrioventricular septal defect (12, 3%, 24/195), interventricular communication (IVC) (11.8%, 23/195), tetralogy of Fallot (10.8%, 21/195), and double outlet right ventricle (8.2% , 16/195). They also state that the IVC represented the largest proportion (24.4%, 77/316) of ventricular malformations. The total proportion of obstructive lesions in this group was much higher for the right side than for the left side of the heart (18.4% (58/316) vs 9.5% (30/316), respectively)<sup>15</sup>.

In this study, it was observed that the frequency of normal findings was 91.7% and that of altered 8.3%. Of these, interventricular communication and cardiomegaly were responsible for the highest prevalence, both with 16.2%. Followed by pericardial effusion with 10.6%, golf ball with 8.5%, endocardial cushion and hypoplastic left heart syndrome both with 6.3%, congestive heart failure with 4.9%, interatrial communication with 4.2% and tetralogy of Fallot, transposition of the great vessels of the base and a single atrium with 2.8%. Therefore, this study contradicted the findings of previous studies.

Comparing the results obtained by this study with the results presented by Hagemann and Zielinsky, it is possible to notice some considerations to be made: 1) "Golf ball" was not the most frequent alteration, as was observed in this study; 2) Pericardial effusion and chamber hypoplasia are considerable findings<sup>8</sup>.

All studies were emphatic in demonstrating that early echocardiography should be implemented as a routine in all prenatal care, even in patients who are not part of the group considered at risk as well as the importance of recognizing such cardiopathies in the prenatal period, so that intrauterine interventions can be performed or delivery in a specialized referral center for better survival and reduction of perinatal mortality.

Therefore, this study contributed to demonstrate that heart diseases are still present at a high rate in our country, and that if the fetal echocardiography exam was inserted universally, the prevalence would probably be higher. And also due to the observation that congenital heart diseases are unrelated to older age, being more prevalent at an average of 30.33 years, the age group with the highest number of pregnancies.

## CONCLUSION

The results of this work allow us to conclude that:

- The maternal age group with the largest number of abnormalities on fetal echocardiographic examination was

between 18 and 34 years old.

- It can be concluded that the frequency of abnormal findings according to the proposed study was 8.3%.
- As for the most frequent findings of abnormalities, interventricular communication and cardiomegaly were the most frequently found alterations, with 16.2% each, in the examination findings.

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