

# Prevalence of Fetal Heart Disease in Pregnant Women Referred to a Specialized Service Due to Cardiac Abnormalities on Morphological Ultrasound

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## Abstract

**Background:** Congenital Heart Disease (CHD) is a malformation of the heart and/or great vessels and is the third leading cause of neonatal death. It can be detected in utero by Morphological Ultrasound/Morphology Scan (MUS) and confirmed by Fetal Echocardiography (FE).

**Objective:** This study aims to determine whether heart disease identified by MUS in pregnant women at a maternal referral center in a southern Brazilian city is confirmed by FE.

**Methods:** This observational, descriptive, and retrospective study was conducted from March 2019 to January 2023. It focused on pregnant women in the public health system, treated through the Unified Health System (SUS) at a maternal referral center in a southern Brazilian city, who had cardiac abnormalities detected on MUS and confirmed by FE.

**Results:** Among the 37 pregnant women with suspected cardiac abnormalities on MUS, 15 cases were confirmed by FE, yielding a diagnostic agreement rate of 21.62% (n = 8) between the two exams. Ventricular Septal Defect (VSD) was the most prevalent heart defect, accounting for 20% of isolated cases. Of the 15 cases with cardiac alterations on FE, two were transferred to a specialized hospital and five died.

**Conclusion:** Fetal cardiac abnormalities were confirmed in fewer than half of the women referred following MUS, with concordance between exams in only eight cases. Continuous training in MUS reduces costs and inconvenience. Professionals qualified in FE are essential for the early diagnosis of CHD, improving care, and reducing infant mortality.

**Keywords:** Congenital Heart Defects; Echocardiography; Prenatal Ultrasonography.

## Introduction

Congenital Heart Diseases (CHD) are malformations of the heart and major vessels that develop during fetal growth. According to the Brazilian Ministry of Health, CHDs are a leading cause of infant mortality and rank third in neonatal mortality. The neonatal cardiology department of the Brazilian Society of Pediatrics estimates that 1 to 2 in every 100 live newborns have some critical CHD.<sup>1</sup> These are classified into two groups: cyanotic and acyanotic, the former caused by a mixture of poorly oxygenated and poorly oxygenated blood, and the latter by deviations in the pulmonary circulation.<sup>2</sup>

Severe malformations can be identified prenatally through MUS and confirmed by Fetal Echocardiography (FE). Therefore, parents must be aware of prenatal care. Qualified professionals are essential for proper diagnosis, as well as a healthcare

structure for birth, providing quality care for these newborns with heart disease.<sup>3,4</sup> Given this scenario, the study proposes to evaluate CHDs confirmed by FE in fetuses of pregnant women with suspected fetal heart malformation, identified in morphological exams (Central Illustration).

## Methods

This is an observational, descriptive, retrospective study from March 2019 to January 2023, totaling 47 months, authorized under opinion 6,164,759 by the Research Ethics Committee. The analyses were conducted with patients from the public health system, treated through the Brazilian Unified Health System (SUS), in a city in southern Brazil. The inclusion criteria were: pregnant women whose fetuses had some suspicion of cardiac malformation on MUS, during the study period, and underwent FE with a pediatric cardiologist, at a maternal reference center, the device used was a Mindray DC-40 Ultrasound with a 3C5A convex transducer. The examination results of pregnant women assisted by SUS in the study region were consolidated at this unit and evaluated by the same pediatric cardiologist over 47 months. Pregnant women who, despite having an indication, did not attend the maternal referral center, and those whose morphological exams lacked any mention of possible CHD or had incomplete medical records, were excluded.

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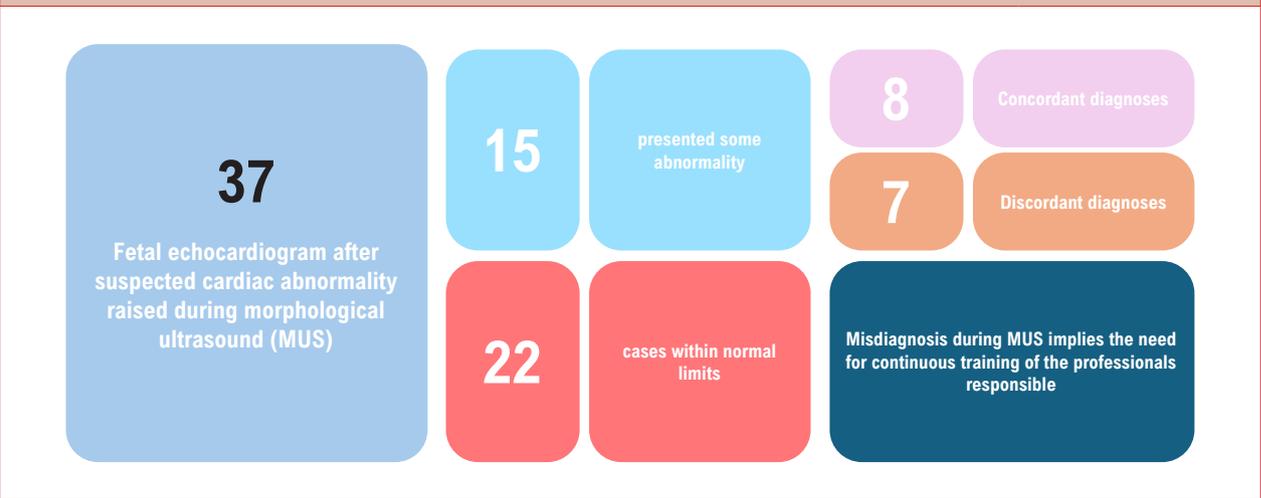
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The study analyzed several factors: cardiac abnormality diagnosis on MUS and FE, maternal comorbidities, baby's sex, and outcomes for those with CHD confirmed by FE. The data were organized in a Microsoft Excel spreadsheet and analyzed through descriptive statistics, using absolute and relative frequencies.

## Results

The study included 37 cases of pregnant women who met the inclusion criteria and underwent MUS exams between March 2019 and January 2023, showing indications of cardiac abnormalities. The diagnostic findings were categorized according to the 2019 Brazilian Guideline for Fetal Cardiology into two main groups based on intrauterine impact: heart diseases without fetal hemodynamic repercussions and heart diseases with fetal hemodynamic repercussions. In addition, within these two groups, there was a subdivision into structural and functional heart diseases.<sup>4</sup>

Among structural heart diseases without fetal hemodynamic impact, the disproportion of the cardiac chambers was the most common anomaly, accounting for 18.91% of cases (n = 7). Next came disproportion of the great vessels and Ventricular Septal Defect (VSD), each with 13.51% (n = 5). Other findings included Atrioventricular Septal Defect (AVSD) in 8.11% (n = 3), transposition of the great vessels in 5.40% (n = 2), and VSD with hypoplasia of the left ventricle, as well as a double outlet of the right ventricle with pulmonary stenosis, each representing 2.70% (n = 1). With regard to heart diseases without functional fetal hemodynamic repercussions, only one case of tricuspid reflux was identified, also representing 2.7% (n = 1) (Table 1).

In the group of structural heart diseases with fetal hemodynamic effects, transposition of the great vessels was observed in 5.40% of cases (n = 2), while pulmonary valve

stenosis (PVS) and pericardial effusion with right chamber enlargement each accounted for 2.70% (n = 1). Functional heart diseases with fetal hemodynamic impact were most commonly associated with arrhythmias, making up 16.22% (n = 6). In addition, in 10.81% of cases (n = 4), the report indicated the impossibility of visualizing the cardiac structures (Table 1).

When comparing the MUS reports (n = 37) with the FE results (n = 37) per year, it was found that, from 2019 to 2023, only 40.54% (n = 15) confirmed some cardiac abnormality, while 59.46% (n = 22) presented results within normal limits.

Regarding abnormality confirmation on FE (n = 15), 21.62% (n = 8) of cases matched the MUS diagnosis as per the pediatric cardiologist's report in the FE. However, in 18.92% (n = 7) of cases, the reports did not fully agree between the two evaluations.

When analyzing the FE sample that presented some cardiac abnormality, identified by the pediatric cardiologist (n = 15), and classifying them according to the groups established in the 2019 Brazilian Guideline for Fetal Cardiology, the following results for these abnormalities (Figure 1) were observed: in structural heart diseases without fetal hemodynamic repercussions, VSD appeared in 20% of cases (n = 3), VSD associated with malaligned aorta in 13.33% (n = 2), and the other findings, each representing 6.67% (n = 1), included AVSD, Tetralogy of Fallot (T4F), AVSD with Transposition of the Great Arteries (TGA), AVSD with Double Outlet Right Ventricle (DORV), T4F with pulmonary valve agenesis, and Coarctation of the Aorta (CoAo).<sup>4</sup>

In the group of functional heart diseases without fetal hemodynamic repercussions, extrasystoles were identified in 13.33% (n = 2) of cases in the FE, being classified as arrhythmias in the MUS (Figure 1).

Structural heart diseases with fetal hemodynamic repercussions included cases of PVS and TGA, each

**Table 1 – Changes in MUS in pregnant women treated from March 2019 to January 2023 at a maternal referral center in a city in southern Brazil**

Structural CHD without fetal hemodynamic repercussions	
MUS Report	Number of cases
Chamber Disproportion	7
Vessel Disproportion	5
VSD	5
AVSD	3
Vessel Transposition	2
VSD with Left Ventricular Hypoplasia	1
DORV with Pulmonary Stenosis	1

Functional CHD without fetal hemodynamic repercussions	
MUS Report	Number of cases
Tricuspid Reflux	1

Structural CHD with fetal hemodynamic repercussions	
MUS Report	Number of cases
Vessel Transposition	2
Valve Stenosis Pulmonary	1
Pericardial Effusion with Enlargement of the Right Chambers	1

Functional CHD with fetal hemodynamic repercussions	
MUS Report	Number of cases
Arrhythmias	6

CHD: Congenital Heart Disease; MUS: Morphological Ultrasound; VSD: Ventricular Septal Defect; AVSD: Atrioventricular Septal Defect; DORV: Double Outlet Right Ventricle

representing 6.67% (n = 1) of cases (Figure 1). Though MUS identified cardiac alterations, including arrhythmias classified as functional heart diseases with fetal hemodynamic impact, these findings were not confirmed by FE.

Among the 15 babies with a CHD diagnosis on FE, 40% (n = 6) were female, 20% (n = 3) were male, and for the remaining 40%, the sex was not recorded.

In this study, six deaths occurred among the 37 cases analyzed, with five of these having an FE report indicating possible CHD. The causes of death associated with CHD were: two cases of Edwards Syndrome and one case of Down Syndrome. Another death was due to a hypoxemic crisis, and one miscarriage occurred with suspected cardiac alteration associated with chromosomal abnormalities, intrauterine growth retardation, and Golf Ball. One additional death was reported where the echocardiogram was within normal limits, but the death was attributed to a different fetal malformation, with no further details provided on the type of malformation.

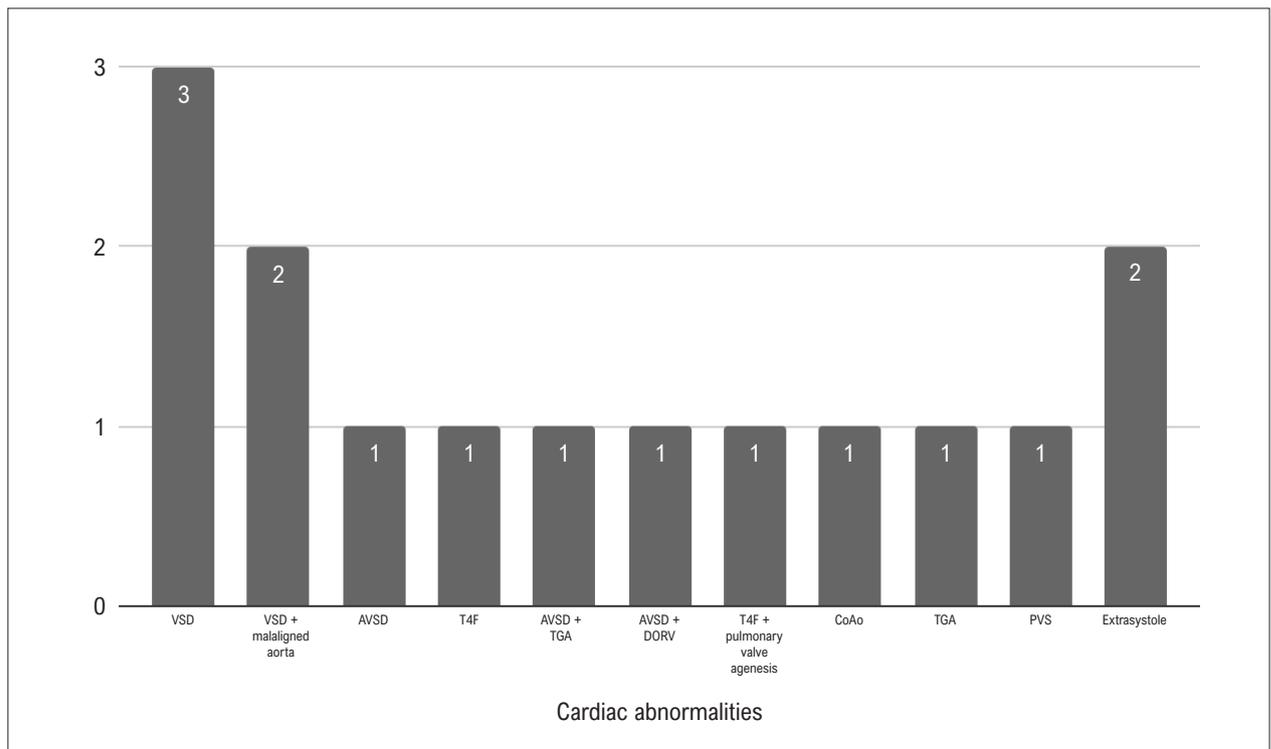
Other outcomes included that among the 40.54% (n = 15) of cases with cardiac alterations on FE, five babies were discharged from the hospital with their mothers without major complications. In two cases, transfer to the referral hospital for pediatric cardiac surgery was necessary. In three cases, no further information was obtained about the babies, as the birth did not occur in the local hospital.

## Discussion

From 2019 to 2023, at a maternal referral center in southern Brazil, 37 pregnant women monitored on an outpatient basis underwent an FE after the MUS indicated a possible cardiac alteration. Of these, 15 pregnant women had the diagnosis of cardiac alteration confirmed by the FE, and 21.62% (n = 8) of the cases presented the same diagnosis as the MUS, representing less than half of the sample with the same diagnoses in both exams.

CHDs are the most common anomalies at birth and are one of the main causes of death in early childhood. According to the World Health Organization (WHO), approximately 130 million children worldwide have some type of CHD. The American Heart Association indicates an incidence of one case for every hundred births, resulting in 1.35 million cases annually. In Brazil, Ministry of Health data show that CHD affects ten in every thousand births, totaling about 29,000 new cases annually, with around 6% of affected children dying before reaching one year of age. Moreover, this condition can be responsible for up to 30% of deaths in the neonatal period in more severe cases.<sup>5</sup>

Early diagnosis is critical, and in Brazil, it is done through prenatal screening, which includes MUS and FE. Recognizing the importance of this exam, the Brazilian Federation of Gynecology and Obstetrics Associations (FEBRASGO) developed a protocol to standardize procedures and guide



**Figure 1** – Structural and functional changes, with or without intrauterine fetal hemodynamic repercussions, found in fetal. VSD: Ventricular Septal Defect; AVSD: Atrioventricular Septal Defect; T4F: Tetralogy of Fallot; TGA: Transposition of the Great Arteries; DORV: Double Outlet Right Ventricle; CoAo: Coarctation of the Aorta; PVS: Valvular Pulmonary Stenosis. Source: Own authorship

Brazilian ultrasound technicians. Among the aspects to be evaluated, cardiac anatomy stands out due to the frequency of cardiac malformations. The basic examination of the fetal heart should include views of the four chambers, the left and right outflow tracts, the three vessels, the aortic arch, and the ductus arteriosus.<sup>6</sup> To emphasize the importance of prenatal screening, a study analyzing data from 20 databases in 12 European countries found that 25% of registered cases of cardiac malformations were diagnosed during pregnancy.<sup>7</sup> Likewise, the observational, cross-sectional study by the Instituto de Medicina Integral Professor Figueira (IMIP), with 457 high-risk pregnant women, indicated that prenatal MUS diagnosed 289 patients (63.2%) with cardiac anomalies, of which 257 (56.2%) were confirmed after delivery.<sup>8</sup>

Comparative studies conducted at the Centro de Referral Perinatal Oriente and the Cardiovascular Center of the Hospital Luiz Calvo Makenna in Santiago, Chile, showed a significantly higher agreement, of 87%, between MUS and FE.<sup>9</sup> In this study, the agreement in the exams was 21.62% (n = 8). This highlights the importance of training professionals and equipping them with the right tools for MUS to reduce maternal anxiety related to false-positive exams and lower public health costs.

Despite a significant false-positive rate, the method remains valuable since ultrasound's effectiveness in identifying malformations depends on the skill and expertise of the technician, as well as maternal and fetal conditions.

In the RADIUS (Routine Antenatal Diagnostic Imaging with Ultrasound) study, anomaly detection was nearly three times more effective when the technician had higher qualifications and training. Similarly, other European studies indicate a significant difference in sensitivity to detect anomalies between exams performed in tertiary centers and specialized centers.<sup>10</sup> Furthermore, there were few studies correlating MUS with CHD in the national and international literature, requiring more data, especially from Brazilian literature, to understand the reality of this aspect in the current Brazilian context in order to aim for improvements in public health.

In this study, the qualifications of the ultrasound technicians conducting MUS and the condition of the equipment used are unknown. For this reason, FEBRASGO emphasizes the importance of standardizing the exam to try to minimize discrepancies and improve the technical level.<sup>6</sup> In addition, it is important to highlight that these data refer exclusively to pregnant women monitored on an outpatient basis by the maternal referral center. It should be considered that during the study's 43 months, other pregnant women in the municipality who received prenatal care may have had MUS-detected changes that were not reported, did not receive adequate follow-up, or were managed in services outside of SUS.

In order to increase the number of diagnoses of CHD during pregnancy, the Brazilian Federal Government enacted Law No. 14598 on June 14, 2023, which includes the following

items in the care provided to pregnant women in the public health system:<sup>11</sup>

According to Art. 1 of the public health regulation, subject to budgetary constraints, the care protocol for pregnant women will include the following procedures as per the regulation:

I - fetal echocardiogram in the prenatal care of pregnant women;

In the study by Pinheiro et al.,<sup>12</sup> FE exams performed during pregnancy showed a precision of 97.7%, specificity of 88.9%, and accuracy of 93% in detecting heart disease.<sup>12</sup> Therefore, the benefit of this exam for the diagnosis of heart changes during pregnancy is indisputable, as well as its positive impact on prognosis. On the other hand, Lopes et al.<sup>13</sup> points out that the indiscriminate performance of this exam in pregnant women outside the risk group is not recommended, since the incidence of heart malformations in this group is around 1%. Moreover, it is a high-cost exam, not available in all regions of Brazil, and whose reliability depends on the quality of the equipment and the experience of the ultrasound specialist.<sup>4,13</sup>

However, the Department of CHDs and Pediatric Cardiology of the Brazilian Society of Cardiology emphasizes that adding this exam would greatly increase fetal heart condition detection, allowing for more detailed evaluations by obstetrics specialists. This would bring numerous advantages, including the ability to rule out or confirm fetal heart abnormalities. By reassuring parents and attending physicians in cases of normal results and cardiac abnormalities, it is possible to plan the birth in a hospital with the necessary support for the newborn, thereby improving the survival rate.<sup>14</sup>

During the period of this study, the Live Birth Information System (SINASC), of the Brazilian Ministry of Health, registered 12,184 cases of congenital malformations of the circulatory system (ICD - Q20 to Q28) throughout Brazil. Of this total, 4.29% (n = 523) occurred in the state of Santa Catarina and 0.44% (n = 53) in the region of Foz do Rio Itajaí.<sup>15</sup> Among the malformations of the circulatory system, the prevalent heart disease in the FE findings of this study was VSD (ICD-Q21.0), found in 20% (n = 3) of these cases. The article by Franklin (2021) cites a study carried out with 1,221 pregnant women in the city of Rio de Janeiro, in which it was also observed that some of the main alterations found were VSD (6.39%), septal hypertrophy (3.35%), and AVSD (1.14%).<sup>16</sup>

In the same period, SINASC reported 1,021 cases of VSD throughout the national territory, with 2.35% (n = 24) in the state of Santa Catarina and 0.2% (n = 2) in the region of Foz do Rio Itajaí.<sup>15</sup> Notably, only in the municipality of Itajaí, three pregnant women followed up on an outpatient basis were diagnosed with VSD, while SINASC recorded only two cases in the entire Vale do Itajaí. This small discrepancy can be explained by the lack of monitoring and/or underreporting by the municipalities in the region since the existence of the three cases was monitored and confirmed in this study.

According to Pedra et al.,<sup>4</sup> cardiac alterations in VSD are classified as structural fetal heart diseases without intrauterine hemodynamic repercussions and do not require immediate neonatal care.<sup>4</sup> This definition was confirmed in this study, in which two of the three babies diagnosed with VSD were born in the local referral hospital and were discharged with their mothers without the need for special care for CHD. The third baby with VSD died, but the cause of death was associated with prematurity, intrauterine growth restriction (IUGR), and chromosomal abnormalities, not VSD.

The other cardiac pathologies found, such as AVSD, T4F, TGA, DORVD, and CoAo, had a lower incidence of 6.67% (n = 1) each. In the study by Pinheiro et al.,<sup>12</sup> with a sample of 96 pregnant women, TGA appeared in 11.5% of cases, T4F in 9.4%, and AVSD in 7.3%, while DORV and CoAo were not observed. In contrast, the study by et al.<sup>17</sup> identified CoAo, VSD, and T4F as the predominant anatomical defects, differing from the findings of the present study and the literature.

Cardiac abnormalities were more common among female babies (40%, or six cases) than male (21.43%, or three cases), at a ratio of 2:1. It is important to note that we had a small sample. This result is inconsistent with the article by Zancanaro et al.,<sup>18</sup> which presents a predominance of males with cardiac alteration, in a ratio of 1.5:1, and with the study by Wu et al.,<sup>19</sup> which also indicates male predominance, in a ratio of 1.22:1. Of the babies for whom information regarding sex was not obtained, 75% (n = 6) had some cardiac alteration. The inability to determine the sex was due to fetal death before birth or delivery in a hospital outside the municipality's referral center.

Six deaths were identified among the 37 cases analyzed. Of these, five had FE reports indicating possible CHD. Considering the 15 pregnant women with a cardiac alteration report, these five represent 40% of the sample with a tragic outcome. The causes of death associated with CHD include two deaths from Edwards Syndrome and one case of Down Syndrome. Another death was due to a hypoxemic crisis, and one miscarriage occurred with suspected cardiac alteration associated with chromosomal disorders, IUGR, and Golf Ball. One death also occurred despite a normal echocardiogram, attributed to a different fetal malformation, though details of the malformation were unavailable. The research by Lopes et al.,<sup>13</sup> which followed 52 cases of newborns with CHD, had 17 deaths. The main cause was a cardiogenic shock in 41.1% (n = 7) of cases, followed by sepsis (17.6%) in three newborns with RVSD and the impossibility of treatment due to heart disease (17.6%), and ill-defined causes also representing 17.6% (n = 3).<sup>13</sup>

According to Bruns et al.,<sup>20</sup> the incidence of CHD in fetal deaths varies from 0.5% to 39.5%, depending on gestational age. Early deaths are mainly associated with complex CHD, and 35% of infant deaths are related to CHD, highlighting its relevance in neonatal and infant mortality. CHD mortality rate and its diagnosis are a global concern, especially in underdeveloped countries, where there is a shortage of trained professionals and structural difficulties in health services. Additionally, the concentration of pediatric cardiologists in large urban centers results in many

undiagnosed newborns. In this context, other professionals must acquire skills for adequate screening.<sup>13,20</sup>

CHD has a major social and economic impact, representing the highest mortality rate and the highest costs among congenital malformations for health services. Therefore, early diagnosis in utero is critical for effective planning, treatment, and prognosis, and for emotionally preparing the family. Despite advancements in the field, like the rollout of FE for all pregnant women, this is not yet a reality across Brazil, where resource disparities between states persist, and a lack of diagnosis and treatment options for these babies remains.<sup>9</sup>

In addition, the early diagnosis of CHDs has a significant impact on obtaining more accurate epidemiological data. Newborns with CHD may die in the neonatal period without a confirmed diagnosis, adding to the gap in accurate knowledge of heart diseases in the country. "In Brazil, for example, it has already been demonstrated that, according to notifications in the Mortality Information System (SIM), the mortality rate due to CHD exceeds the prevalence of these anomalies at birth, according to data from Sinasc".<sup>21</sup>

This study faced some limitations, starting with the small sample, which may compromise the generalization of the results. However, it should be noted that it reflects all mothers monitored only by the SUS in this region, reflecting the reality in this context. Monitoring the babies also represented a significant challenge, due to the lack of information in the medical records and the fact that some had been transferred to other hospitals. Despite these challenges, the study highlights the importance of promoting specific initiatives to encourage healthcare teams to improve their MUS and FE exams, as well as assessing this situation in other regions, given the limited available literature on the topic.

The discrepancies between recorded cardiac malformations in Brazilian national databases and study findings suggest a need for improvements in the notification and monitoring systems for pregnant women. Broad, equitable inclusion of FE can improve both diagnosis and treatment and also yield more accurate epidemiological data, which would help in formulating more effective health policies. In this context, the study reinforces the importance of early diagnosis and thorough screening for CHD, demonstrating that detailed, standardized examinations can save lives and significantly improve outcomes for many children.

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## Conclusion

Less than half of the MUS examinations indicating possible fetal heart disease had their findings confirmed by FE reports. Only a quarter of the cases showed agreement between the two examination reports. VSD was the most frequently identified CHD. Additionally, notable discrepancies were found between SINASC data and this study's findings, likely due to monitoring and/or underreporting issues by local municipalities.

The high infant mortality rate from CHD relative to the number of diagnoses points to the need for better screening methods and improved case identification. Investing in MUS training, increasing the number of FE specialists, standardizing procedures, and upgrading prenatal care equipment are urgent public health needs. The recent legislation that includes FE in the public prenatal care protocol is a major advancement, though access to this exam remains uneven across Brazil.

## Author Contributions

Conception and design of the research and acquisition of data: Michael AP, Soares MF, Witkowski SM; analysis and interpretation of the data and statistical analysis: Michael AP, Witkowski SM; writing of the manuscript: Michael AP, Witkowski SM; critical revision of the manuscript for intellectual content: Rodrigues MOD, Foresti RA, Witkowski SM.

## Potential Conflict of Interest

No potential conflict of interest relevant to this article was reported.

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## Study Association

This study is not associated with any thesis or dissertation work.

## Ethics Approval and Consent to Participate

Trabalhos experimentais envolvendo animais: This study was approved by the Ethics Committee on Animal Experiments of Universidade do Vale do Itajaí under the protocol number 6.164.759.

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