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Clinical and epidemiological characteristics of pregnant women and their fetuses with congenital heart disease diagnosed by fetal echocardiography

Nicole Lerner¹, Maria de Fatima M. Leite², Carla Verona², Michael Barmpas², José Paulo Pereira Júnior², Heron Werner³

1. Universidade Federal do Estado do Rio de Janeiro, Brazil. 2. Fundação Oswaldo Cruz, Rio de Janeiro, Brazil.
3. Biodesing Lab Dasa/PUC-Rio, Rio de Janeiro, Brazil.

Introduction

Congenital heart defects (CHDs) are the most common congenital anomalies, with high morbidity and mortality. Fetal echocardiogram is a fundamental tool in perinatal care by allowing early diagnosis, timely counseling, referral, planning of delivery and postnatal interventions. Identifying maternal and fetal characteristics associated with CHDs enhance screening strategies and improve clinical outcomes. Our aim is to describe clinical and epidemiological characteristics of pregnant women, and their fetuses diagnosed with CHD in a high-complexity referral center.

Methods

Retrospective cross-sectional study conducted at Instituto Nacional de Saúde da Mulher, da Criança e do Adolescente Fernandes Figueira (IFF/Fiocruz) in Rio de Janeiro, Brazil on medical records, from Sep/2020 to Dec/2023. Data on exam date, prior diagnoses, indication, maternal age and final diagnosis were collected and analyzed.

Results

A total of 817 fetal echocardiograms from 696 pregnancies were analyzed. CHDs were identified in 195 cases (28.02%). Mean maternal age in CHD group was 30.57 ± 7.93 years, compared to 29.96 ± 7.84 years in non-CHD pregnancies, with no significant difference ($p > 0.05$). The main indication for fetal echocardiogram was abnormal obstetric ultrasound (39.08%), followed by advanced maternal age (14.94%) and morphology scan with CHD (11.93%). Cardiac defects were confirmed in 74.70% cases with morphology scan suspicion, 28.31% with abnormal obstetric ultrasound, and 5.77% with advanced maternal age. The most common CHDs found are show in Table 1. Among fetuses with CHD, 55.90% were isolated, while 44.10% had at least one additional malformation. The most prevalent being diaphragmatic hernia (8.21%), followed by omphalocele (5.64%) and Down syndrome (3.08%).

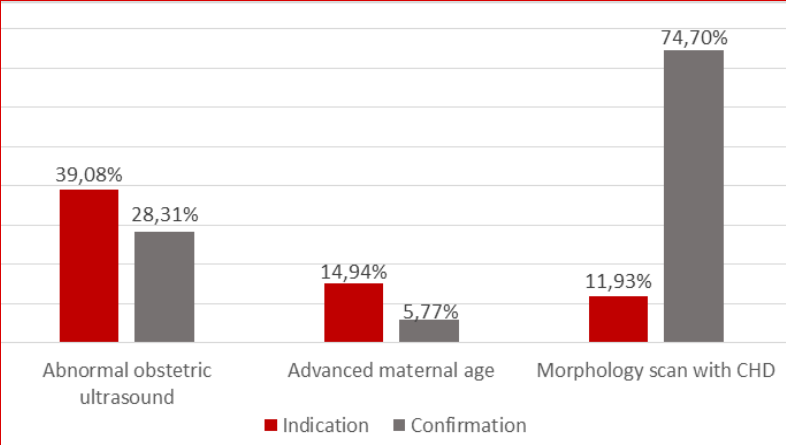


Figure 1: Percentage of echocardiography indications and confirmation rates

CHD	n	%
Ventricular septal defect	33	16,92%
Suspected heart disease	19	9,74%
Hypoplastic left heart syndrome	13	6,67%
Ductal constriction	10	5,13%
Total atrioventricular septal defect (TAVSD)	9	4,62%
Tetralogy of Fallot	9	4,62%
Isomeric syndrome	5	2,56%
Tetralogy of Fallot + TAVSD	5	2,56%
Fetal arrhythmia	4	2,05%
Anomalous subclavian artery	4	2,05%
Tricuspid atresia	4	2,05%

Table 1: Most prevalent CHD

Conclusion

The study highlights the importance of abnormal ultrasound findings to the diagnosis of fetal CHD, emphasizing its importance in early suspicion and timely referral for fetal echocardiography.