CARDIAC DISEASES AND THE IMPORTANCE OF FETAL ECOCARDIOGRAPHY: A BIBLIOGRAPHIC REVIEW

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ABSTRACT

The aim of this study is through a literature review to describe the importance of fetal echocardiography in screening for fetal heart disease. Since the first report of fetal echocardiography in 1972 by Winsberg, several advances in ultrasound technology have occurred, allowing for a detailed assessment of the cardiac anatomy in the fetus. It is essential to remember that more than 90% of cardiac malformations occur in fetuses without any risk factor. Therefore, targeted population screening, during routine prenatal ultrasound, through systematic observation of the fetal heart, combined with basic knowledge on the part of the operator of its normal characteristics, is the only way for the diagnosis of heart disease congenital diseases can be expanded in terms of primary care to the population. The detailed anatomical knowledge of the cardiac structures, combined with the precise interpretation of echocardiographic images taken during fetal development, allow the early diagnosis of particular forms of congenital disease that have serious pathophysiological consequences. Prenatal diagnosis helps in planning the optimal management of the baby with the choice of a tertiary care center for stabilization and early initiation of therapy.

KEYWORDS: HEART DISEASE, ECHOCARDIOGRAPHY, FETAL

INTRODUCTION

Congenital heart diseases (CHD) are among the most common malformations in human fetuses and considered the most frequent. Due to their poor prognosis, they significantly contribute to infant mortality, accounting for about 10% of infant deaths and half of deaths due to congenital malformations[1].

It is essential to remember that more than 90% of cardiac malformations occur in fetuses without any risk factor. Therefore, targeted population tracking, during routine prenatal ultrasound, through systematic observation of the fetal heart, combined with a basic knowledge, by the operator, of its normal characteristics, is the only way to expand the diagnosis of congenital heart disease, in terms of primary care to the population[2].

According to the American College of Cardiology, the main indications for fetal echocardiography are fetal cardiac 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, karyotype fetal anomaly and other abnormalities of the fetal system. For fetuses with suspected congenital heart disease or extra-cardiac abnormality detected in the second trimester fetal anatomy scan is adviced[3,4].

One of the main goals of prenatal diagnosis is the detection of severe CHD, whose prognosis depends, in most cases, on the planning of delivery in a specialized reference center[5].

Therefore, the aim of this study is to describe the importance of fetal echocardiography in the screening of fetal heart disease through a literature review.

2 HEART DISEASES AND THE IMPORTANCE OF FETAL ECHOCARDIOGRAPHY

2.1 INCIDENCE AND IMPORTANCE OF DETECTION

Congenital heart disease (CHD) is a leading cause of mortality in children, with an estimated prevalence of about 4-13 per 1000 live births. Given the increased risk of morbidity and mortality in babies with CHD, prenatal diagnosis is essential to help plan peripartum management4. The incidence of congenital heart disease in deaths is 10 times more frequent during abortions 22 to 42%6. In fetal life, this incidence is estimated to be up to five times higher, and this difference is justified by fetal deaths[5].

Approximately 50% of cases present early hemody-

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namic repercussions, requiring interventional treatment or surgical correction in the first year of life. The association with extracardiac malformations can be observed in up to 50% of those affected, further increasing pre- and postoperative morbidity and mortality[5].

Prenatal diagnosis of congenital heart disease (CHD) has been shown to have a significant effect on prenatal and postnatal care. In addition to the potential medical benefits, fetal diagnosis allows counseling for parents, in which well-informed families can make decisions regarding pregnancy, and prepare emotionally for the delivery of a child with significant CHD. Accurate prenatal diagnosis can also lead to additional tests of the fetus, which can yield valuable information in the general assessment of the fetus[7].

The performance of early first trimester ultrasound and the greater use of fetal echocardiography in recent years have contributed to an increase in the intrauterine diagnosis of congenital heart diseases, with consequent better perinatal evolution of them. However, fetal echocardiography is not yet universally available in our country, with a greater concentration of professionals with training in Fetal Cardiology in the South and Southeast regions, and a more restricted number in the North, Northeast and Midwest regions where the availability of this tool is mainly restricted to capital cities and is not readily available in the Unified Health System (SUS)[5].

As Congenital Heart Disease (CHD) is the most common serious birth defect and the least diagnosed by routine obstetric ultrasound, the challenge of recent years has been to discover an early screening method for fetal heart disease, since most fetuses with heart disease are born to mothers that do not have the classic indications for fetal echocardiography[5].

Although fetal echocardiography, traditionally indicated for high-risk pregnant women, is quite accurate, 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 ultrasound[5].

Given this scenario, some advocate that fetal echocardiography should be indicated for all pregnant women, since, in experienced hands, it can detect close to 100% of all cardiac anomalies in fetal life, being considered the gold standard for fetal cardiac diagnosis. Prenatal diagnosis significantly reduced pre- and postoperative mortality[5].

The most recent literature shows sensitivity around 13.5% for detecting cardiac anomalies, and the measurement of NT ≥ 3.5 mm should be considered an indication for fetal echocardiography. Doppler flowmetric analysis of the fetal cardiovascular system is also applied to screen for fetal heart disease, which may or may not be associated with chromosomal disorders. Several studies advocate that the altered flow of the ductus venosus, that is, the appearance of the reverse wave during atrial contraction (A wave) in fetuses with TN ≥ 3.5 mm increases the probability of CHD by three times, while a normal flow pattern reduces the risk of heart disease by half[5].

Although several studies have evaluated the effectiveness of congenital heart disease screening and the accuracy of fetal echocardiography, information on the use of specific risk factors to refer patients for fetal echocardiography is limited, especially for cases where cardiac findings on prenatal ultrasound are normal. This low detection rate may reflect the inability of traditional risk factors to identify the majority of patients at risk, as well as the limitations of ultrasound to detect cardiac anomalies in the prenatal period[3].

However, most pregnant women whose children are born with congenital heart disease have no known risk and, therefore, may not undergo high-risk obstetric consultations, with ultrasound professionals being responsible for referring those who present with ultrasound suspicion of fetal heart disease[8].

2.2 THE FETAL ECOCARDIOGRAPHIC STUDY

Until a few years ago, the only aspect of fetal cardiovascular physiology that could be routinely monitored was heart rate. With the introduction of M-mode, two-dimensional echocardiography (ECHO) and Doppler techniques (especially color flow mapping) in the study of the fetus, accurate description of the intracardiac anatomy, sequential analysis of the chambers and, finally, the recognition of malformations, disturbances of cardiac function and rhythm in the prenatal period[9].

Initially, fetal echocardiography included only the four-chamber view (basic cardiac echocardiographic examination [BCEE]) of the heart. Then, outflow tract view (OTV) and three-vessel tracheal view (3VTV) were added to increase the accuracy of fetal echocardiography. More recently, ECEE (extended cardiac echocardiographic examination), which included visualizing the 4 chambers, the right ventricular outflow tract, the left ventricular outflow tract, and the main pulmonary artery and its branches, was used as a specific protocol to identify some minimal defects and provide more details about suspected fetal heart[10].

The combined sensitivity of prenatal echocardiographic diagnosis of fetal heart disease in the first trimester, second trimester, and third trimester was 60.3%, 60.9%, and 77.4%, respectively[10].

Two-dimensional ECHO can visualize fetal heartbeats as early as the 6th week of gestation, but an adequate structural analysis is usually only feasible after the 16th week. At this gestational age, the fetal heart is still very small and often a complete study is not possible. The ideal time for the visualization of the fetal heart extends from the 18th to the 24th gestational week, when a large volume of amniotic fluid surrounds the conceptus. In the 3rd trimester, the fetal spine is often anterior and the ribs are more calcified, "shading" the cardiac area and making it difficult

to properly assess the fetal heart in this gestational phase[9].

Fetal cardiac examination is optimally performed between 18 and 22 weeks of menstrual age. Some anomalies can be identified during the end of the first and beginning of the second trimester of pregnancy, especially when an increase in the nape of the neck and translucency is identified11. From 18 weeks onwards, all cardiac structures can be analyzed by echocardiography. This is the initial gestational age at which the echocardiogram should be performed. The best images, however, are obtained between 24 and 28 weeks, when the heart is already larger, the fetus is still moving well, and the bones are not a significant barrier to ultrasound. It is noteworthy that the early assessment of the heart can be performed either via the transvaginal or transabdominal route (after the 14th week)[5].

Fetal cardiac examination consists of a systematic and careful study of the atrial situs and of the venoatrial, atrioventricular and ventricle-arterial connections. It also involves an adequate assessment of the myocardial walls, atrial and ventricular septa, in addition to the aortic and ductal arches[9].

It is essential that the fetal cardiologist have knowledge of basic ultrasound concepts, particularly with regard to fetal status and position. Before starting the assessment of the heart, the presentation of the fetus must be determined, identifying its right and left sides. The main marker on the left side of the fetus is the stomach. In situs inversus or ambiguous situations, it may be misplaced and cannot be used as a marker on the left fetal side[5].

In order to determine the cardiac anatomy and connections, four sections of the heart are needed, in general: four chambers, longitudinal of the left ventricle (LV), arch of the ductus arteriosus or ductal and aortic arch[9].

The basic cardiac screening exam is based on a four-chamber view of the fetal heart. This view should not be confused with a simple chamber count, as it involves a careful assessment of specific criteria (Figure 1)[11].

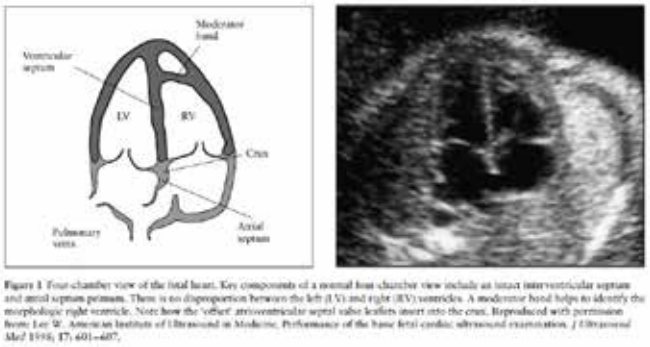

Figure 1- Four Chambers of the fetal heart. ISUOG, 2006

The most easily obtained cut is the four-chamber one (fig.1). The descending aorta lies between the spine and the left atrium (LA). The atrial cavities have similar dimensions and the membrane of the foramen ovale can be seen, moving towards the LA and the Eustachian valve inside the right atrium (RA) [9].

Among the technical factors, it is important to start the examination by adjusting the equipment for the best possible image resolution, which involves choosing the transducer with the highest frequency, gray scale with better contrast between the interfaces and the use of harmonics. The scanning angle must be sufficient to delimit the cardiac area under study and the depth and zoom adjusted so that the heart image fills 1\3 to 1\2 of the equipment screen. Visualization of cardiac structures using cineloop (frame-by-frame image) is also a resource used in the assessment of the fetal heart, an organ in constant movement whose frequency varies between 120 and 160 beats per minute[12].

A normal heart is usually no larger than a third of the chest area. Some views may reveal a small hypoechoic border around the fetal heart that may be mistaken for pericardial effusion. An isolated finding of this type usually represents a normal variation[11].

It is also possible to assess the morphology and dynamics of the atrioventricular valves, as well as the atrioventricular connection. The two ventricles have similar dimensions, but the contractile geometry shows a dominance of the right ventricle (RV). The differentiation between the ventricles can be made by trabeculation (coarser in the RV), the presence of a moderating band in the RV, the more distal insertion of the tricuspid valve, and the well-defined appearance of the papillary muscles in the LV. Wall thicknesses are similar[9].

Failure to obtain a normal four-chamber projection during obstetric ultrasound is an absolute indication to perform a fetal echocardiogram by a specialist in Fetal Cardiology. Although the four-chamber projection has great value in tracking normality by evidencing the proportional size between the cardiac chambers, as it does not pass through the aorta and pulmonary artery, it does not define cardiopathies of the transposition type of the great arteries, tetralogy of Fallot (T4F), common arterial trunk, among others. Tables 1.1 and 1.2 show the cardiopathies commonly associated with normal and abnormal four-chamber projection, respectively[5].

Tetralogia de Fallot
Transposição das grandes artérias
Tronco arterioso comum
Anomalias do arco aórtico
Estenose valvares aórtica e pulmonar leves
Comunicações interventriculares perimembranosas

Table 1 - Heart diseases commonly associated with normal four-chamber position. Source: PEDRA et al. 2019[5]

Atresias das valvas mitral e aórtica
Atresias das valvas tricúspide e pulmonar
Anomalia de Ebstein/ displasia da valva tricúspide
Defeito do septo atrioventricular
Comunicações interventriculares grandes
Ventrículo único
Estenoses das valvas aórtica e pulmonar graves
Coartação da aorta
Drenagem anômala total de veias pulmonares
Cardiomiopatias
Tumores cardíacos

Table 2 - Heart diseases commonly associated with abnormal four-chamber position. Source: PEDRA, et al. 2019[5].

The best image of the heart is obtained from the abdomen by sliding the transducer slightly towards the chest. Although it is also possible to obtain images through the chest or back, as the fetal lungs are filled with fluid and do not offer a barrier to the passage of ultrasound, these images are of lower quality, especially at the end of pregnancy, when the ossification of the ribs and spine represents an important barrier to the passage of ultrasound[5].

In this situation, to improve image quality, it is often necessary to ask the pregnant woman to be in the left or right lateral decubitus position. Difficulties can occur in the presence of polyhydramnios and maternal obesity. Polyhydramnios is a situation that can make it difficult or even impossible to perform echocardiography, as the fetus is positioned further away from the transducer and tends to move a lot, making it difficult to take measurements and position the Doppler volume sample to obtain the usual tracings. In this situation, the fetus can be brought closer to the transducer, placing the pregnant woman in a position of four supports on her knees and elbows[5].

Maternal obesity also hinders the technical quality of echocardiography and, often, obtaining a cardiac image is only possible by using a more vigorous compression of the transducer, or by using low-frequency sectorial transducers, such as those used for echocardiography of adults[5].

Fetal echocardiography performed in the third trimester follows the same principles as the examination performed in the second trimester. Some adjustments can be made if there is a more exacerbated acoustic shadow characteristic of this period. The use of images (chroma) can improve the contrast between the more evident structures and facilitate the identification of structures[12].

Heart rate and regular rhythm should be confirmed. The normal rate ranges from 120 to 160 beats per minute. Mild bradycardia is temporarily seen in normal second trimester fetuses. Corrected bradycardia, especially heart rates that remain below 110 beats per minute, require timely evaluation for possible heart block. Repetitive heart rate slowdowns during the third trimester can be caused by fetal distress. Occasional skip beats are not usually associated with an increased risk of structural fetal heart disease. However, this finding may occur with clinically significant heart rate or rhythm disturbances as an indication for fetal echocardiography[11].

Mild tachycardia (>160 beats per minute) can occur as a normal variant during fetal movement. Persistent tachycardia, however, should be evaluated for possible fetal distress or more severe tachydysrhythmias[11].

The heart is normally shifted about 45± 20◦ (2 standard deviations (SD)) to the left side of the fetus (Figure 2). Special attention must be paid to the cardiac axis and position because they can be easily assessed even if the four-chamber view is not visualized satisfactorily. Abnormalities of situs should be suspected when the fetal heart and/or stomach is/is not found on the left side as well. Abnormal axis increases the risk of a cardiac malformation, especially involving the outflow tracts. This finding may be associated with a chromosomal anomaly[11].

Some hearts are abnormally displaced from their usual position in the left anterior central chest. The abnormal cardiac position may be caused by a diaphragmatic hernia or a space-occupying lesion such as a cystic adenomatoid malformation. Positional abnormalities can also be secondary to fetal lung agenesis-hypoplasia. Both atrial chambers normally appear similar in size and the foramen ovale flap should open into the left atrium. Pulmonary veins can often be seen entering the left atrium. However, their identification should not be considered a mandatory part of a basic cardiac screening exam. The lower border of the atrial septal tissue, called the septum primum, must be present[11].

Both ventricles should also be similar in size, with no evidence of thick walls. Although mild ventricular disproportion can occur as a normal variant, hypoplastic left heart syndrome and coarctation of the aorta are important causes of this disparity. The ventricular septum should be carefully examined for cardiac wall defects from the apex to the crux. Septal wall defects can be difficult to detect when the transducer insonation angle is directly parallel to the ventricular wall. In these circumstances, a defect may be falsely suspected because of the acoustic "fall" artifact. Small septal defects (1–2 mm) can be very difficult to confirm if the ultrasound imaging system does not provide a sufficient degree of lateral resolution, especially if fetal size and position are unfavorable[11].

Two distinct atrioventricular valves (in the right side, tricuspid and in the left side, mitral) should be seen to open separately and freely. The septal leaflet of the tricuspid valve is inserted into the septum closer to the apex when

compared to the mitral valve (ie. normal displacement). Abnormal atrioventricular valve alignment may be a key ultrasound finding for cardiac anomalies, such as atrioventricular septal defect[11].

In the context of Fetal Cardiology, antenatal diagnosis allows the monitoring of pathologies, with the potential for intrauterine hemodynamic decompensation, in addition to helping to schedule cases whose heart diseases will present hemodynamic repercussions in the immediate neonatal period[5].

FINAL CONSIDERATIONS

Detailed anatomical knowledge of cardiac structures, combined with accurate interpretation of echocardiographic images taken during fetal development, allow early diagnosis of particular forms of congenital disease that have serious pathophysiological consequences. Prenatal diagnosis helps in planning the optimal management of the baby with choosing a tertiary care center for stabilization and early initiation of therapy.

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