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IMPORTANCE OF THE DIAGNOSIS OF CONGENITAL HEART DISEASES DURING PRENATAL

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Abstract: INTRODUCTION: Heart diseases form the most prevalent group of congenital anomalies at birth. They are considered the main cause of morbidity, mortality and disability associated with congenital malformations. Currently, the diagnosis of congenital heart diseases can be performed through morphological ultrasound, fetal echocardiography and genetic tests, during prenatal care. This way, prenatal diagnosis is extremely important, since it allows a better follow-up of the pregnancy, with adequate guidelines and planning, in the search for the reduction of morbidity and mortality. The objective of this study is to review the literature and interpret the data on prenatal diagnosis of congenital heart diseases in order to reaffirm the importance of this still in the gestational phase. **METHODOLOGY:** This is an integrative literature review regarding the following hypothesis that guides the research problem: How important is prenatal diagnosis of congenital heart disease in prenatal care? Publications from the last 4 years in Portuguese, English or Spanish were included, provided that their approach contributed to the thesis under analysis. After the selection and collection of articles, a summary and discussion of the results was carried out based on their interpretation and data synthesis. **RESULTS:**In the present integrative review, 14 articles met the previously established inclusion criteria. Regarding the type of research design of the articles evaluated, the sample showed: six literature reviews, two epidemiological studies and other case reports. **CONCLUSIONS:** In the last 4 years, all studies have positively reinforced the importance of instilling the diagnosis of congenital heart disease in the level of prenatal care. This strategy aims to better elucidate cardiological treatment of conceptuses with heart disease and, therefore, may represent the difference between life and death in the perinatal period,

as well as impacting the reduction of costs for the health system.

Keywords: Congenital heart diseases; Fetal Mortality; Prenatal Diagnosis.

INTRODUCTION

Heart diseases form the most prevalent group of congenital anomalies at birth. They are considered the main cause of morbidity, mortality and disability associated with congenital malformations. As a consequence, it causes detriment to the quality of life of its patients and increases health costs (PINTO et al. 2018).

This congenital malformation can occur by association of genetic and non-genetic factors. This implies the importance of evaluating personal and family history in order to identify risk factors in the preconception and prenatal periods. The result is early care and intervention and better neonatal outcomes (LIU et al. 2015).

As risk factors for the development of congenital heart diseases, there are family history (first-degree relatives) and Motherly and fetal conditions (MORHY et al. 2020). With emphasis on pregnancies in women aged between 20 and 29 years, the high rate of late abortion in previous pregnancies and teratogenic exposures, such as the use of antibiotics (PINTO et al. 2018). It is also associated with difficult-to-control Motherly phenylketonuria, Motherly anti-RO and anti-LA antibodies (SSA/SSB), Motherly ingestion of Angiotensin Converting Enzyme Inhibitors (ACEI), retinoic acid and non-steroidal anti-inflammatory drugs. in the third trimester (PEDRA et al. 2019). Another important risk factor is Motherly diabetes mellitus, both pre-gestational and that diagnosed in the first trimester,

These factors increase the incidence of this malformation so that approximately half of the cases have early hemodynamic repercussions, requiring interventional treatment or corrective surgery in the first year of life (FRANKLIN et al. 2021). In Brazil, it is estimated that the average need for cardiovascular surgery in congenital is around 23,000 procedures/year, considering in this estimate, in addition to new births with congenital heart disease, cases of reinterventions (JESUS et al. 2018).

Currently, the diagnosis of congenital heart diseases can be performed through morphological ultrasound, fetal echocardiography and genetic tests, during prenatal care. Ultrasonography behaves as a triage bridge for echocardiography, considering that patients who have alterations in the ultrasound are referred for a more specific examination of the affected area. Thus, fetal echocardiography is performed from the end of the first trimester to the end of pregnancy and is currently considered the main diagnostic test - the frequency of this test may depend on the management, severity of the lesion, signs of heart failure and disease progression mechanisms (MORHY et al., 2020). Furthermore, genetic tests are a relevant tool to detect chromosomal anomalies associated with CC's,

In this sense, prenatal diagnosis is extremely important, as it allows a better follow-up of the pregnancy, with adequate guidelines and planning, in the search for a reduction in morbidity and mortality. Therefore, measures such as referral of the pregnant woman to a tertiary care facility with a trained neonatal team, treatment solutions or improvement of the prognosis, adequate family monitoring and guidance, measurement of the risk of recurrence in future pregnancies and delivery planning, contribute to the reduction of this indicator of health (DOS SANTOS et. al., 2021).

Therefore, the objective of this study is to review the literature and interpret the data

about the prenatal diagnosis of congenital heart diseases in order to reaffirm the importance of this still in the gestational phase.

METHODOLOGY

This is an integrative literature review regarding the following hypothesis that guides the research problem: How important is the prenatal diagnosis of congenital heart diseases in prenatal care? From this, search strategies were used in the following databases: National Center for Biotechnology Information and National Library of Medicine - (PUBMED), Latin American Literature on Health Sciences - (LILACS), Scientific Electronic Library Online - (SciELO), UpToDate and Google Scholar.

Publications from the last 4 years in Portuguese, English or Spanish were included, provided that their approach contributed to the thesis under analysis. In this scenario, the search involved combinations between the following keywords: Congenital Heart Diseases; Fetal Mortality; Prenatal Diagnosis, indexed to the Health Sciences Descriptors (DECS).

After the selection and collection of articles, a summary and discussion of the results was carried out based on their interpretation and data synthesis.

RESULTS

article name	Authors and year of publication	Intervention studied	Results	Recommendations/ Conclusions
Implications of prenatal diagnosis of congenital heart disease on fetal mortality: literature review	DOS SANTOS et. al., 2021	Review the literature and interpret the data on the prenatal diagnosis of congenital heart diseases (CHDs), in order to complete a synthesis on the subject and see what its implications are for fetal mortality rates.	Prenatal diagnosis of cardiac anomalies, compared to postnatal diagnosis, showed a better survival rate after surgical correction of CHDs. Studies have shown a high sensitivity and high specificity of the diagnosis performed by means of fetal echocardiography, however, the infeasibility of screening by this exam was highlighted, given the Brazilian socioeconomic context. This way, morphological ultrasound has become the best option and the combination of ultrasound with genetic testing in the prenatal period has demonstrated good efficacy and accuracy for the diagnosis of heart defects associated with fetal chromosomal anomalies.	The benefits of identifying and treating CC's early are undeniable, and there must be qualification and investment in public health for the early detection of this diagnosis.
Early diagnosis of congenital heart diseases: an integrative review	PEACOCK et. al., 2017	To analyze the early diagnosis of congenital heart diseases.	The most frequently used and relevant diagnostic methods are fetal ultrasound (nuchal translucency) and fetal echocardiography. Ultrasound screening in pregnant women is the best way to detect malformations in the first trimester, and fetal echocardiography is an important method used for a more accurate analysis of heart diseases.	The importance of early diagnosis for an adequate treatment, prevention of diseases and sequelae, providing the opportunity to improve the prognosis and allowing a prolonged survival to the health of the child, with emphasis on the participation of other professionals for the quality of the final result obtained.

Importance of early diagnosis of congenital heart diseases: an integrative review	LINHARES et. al., 2021	Build a solid and uniform knowledge for the accomplishment of a quality medical practice, through theoretical basis and critical analysis on the importance of early diagnosis of congenital heart diseases.	It can be inferred that the best way of early identification of cardiac anomalies is through prenatal ultrasound screening and, for a more accurate and confirmatory diagnosis, fetal echocardiography.	The relevance of early diagnosis is confirmed in order to provide the patient with adequate treatment, quality of life and reduction of morbidities, highlighting the need to insert tests such as ultrasound and fetal echocardiography in prenatal screening programs.
Congenital heart diseases in the neonatal period - retrospective study	BRANCHES, 2019	To assess the prevalence and characterize newborns with congenital heart disease, with pre- or postnatal diagnosis, in a tertiary-level maternity hospital.	The analyzed sample consisted of 143 newborns, with a prevalence of congenital heart disease of 9.5/1000 live births and mortality due to heart disease of 2.1%. The most frequent heart disease was IVC (53.5%), followed by ASD (16.8%). Critical congenital heart diseases represented 20% of the sample and 67.7% of these had a prenatal diagnosis, the most frequent being TGA. Phenotypic alterations or major extracardiac malformative anomalies were identified in 46 patients, with a specific etiological diagnosis in 13 cases (9.8%).	It would be important to design a study with a prospective record of the various environmental risk factors, as well as the existence of cardiac pathology in parents or family members, so that it is possible to intervene in the prevention of the appearance of heart diseases or lead to its early diagnosis, in order to establish appropriate therapy and surveillance, as well as their comorbidities, when present.
Genetic Testing and Pregnancy Outcome Analysis of 362 Fetuses with Congenital Heart Disease Identified by Prenatal Ultrasound	LUO et. al., 2018	To determine the type and frequency of chromosomal abnormalities in fetuses with CHD and to analyze the pregnancy outcomes of fetuses with cardiac abnormalities caused by different genetic factors.	Of the 362 fetuses, 220 had isolated coronary artery disease and 142 had coronary artery disease with an extracardiac anomaly. Among these 362 fetuses, 140 were identified with a genetic cause, including 111 cases with aneuploidy, 10 cases with abnormal chromosomal structure by karyotyping and 19 cases with pathogenic or probably pathogenic copy number variations (CNVs) by CMA. The detection rate is approximately 38.7%. Only one (identified as trisomy 18 syndrome) in 140 positive cases resulted in perinatal death, with the rest being induced. The remaining 222 cases were negative for both genetic tests, and of these, 56 resulted in induced labor and 77 had natural deliveries or cesarean sections. The pregnancy outcome of the remaining 89 cases was uncertain.	Karyotyping and CMA are effective and accurate prenatal genetic techniques for identifying fetal chromosomal anomalies associated with heart defects, and this can help clinicians perform appropriate genetic counseling regarding the etiology and outcome of congenital heart defects.

Evolution of mortality from congenital heart diseases in Brazil – an ecological study	BRAGA et. al., 2017	To analyze the trend of mortality from cardiac malformations in Brazil, observing age, sex and geographic region in the period from 2008 to 2013.	The standardized mortality coefficients from cardiac malformations in males and females, aged 0 to 4 years, by geographic region in Brazil in the period from 2008 to 2013, show, in general, a fluctuation over the long period studied, with a decrease observed in the last year (2013), with the exception of the female population of the Midwest, whose rate, although it fluctuated, ended up being equal in 2008 and 2013. The Midwest region was the one with the highest mortality rates during the period studied. The standardized mortality rates for congenital heart diseases were lower compared to 2008 for both sexes, with the lowest rate observed in females in the North region in 2013.	Although they fluctuate, infant mortality rates resulting from congenital heart diseases have shown a tendency to decrease due to better prenatal conditions and the possibility of access to early diagnosis and treatment.
Congenital heart diseases: a literature review	OLIVEIRA, 2018	To identify, in the face of the scientific literature, how health care is provided to NB and children with congenital heart disease.	For the early identification of heart diseases, an adequate assessment is important to analyze signs and symptoms of the pathologies in any period, whether neonatal or pediatric. Regarding the immediate treatment of patients with heart disease, even with the government programs free prenatal care, neonatal screening, and ordinances for the treatment of heart diseases, a large number of patients still cannot access treatment or immediate care and end up with complications. or death, this is due to the high rate of heart disease, the high cost of treatment faced by the government, and the scarce number of specialized professionals.	The results of this study demonstrate that the prevalence and presentation of congenital heart diseases in our country are similar to what is found in developed countries, alerting to the need for the health system to prepare to diagnose and treat these patients earlier, reducing economic expenses, possible sequelae and emotional exhaustion of those affected and their families. We can consider that congenital anomalies are already the second cause of mortality in the first year of life in our country and that their relative importance tends to grow, making this challenge even more important to be faced.
The reality of congenital heart disease in Brazil: literature review	AMORIM et. al., 2021	Assess the CC scenario in Brazil.	The analyzed articles point out that carrying out continuing health education can contribute to a better quality of life and understanding of the patient and/or caregiver in the face of the limitations of CC. The factors that showed a reduction in morbidity and mortality were: early diagnosis; performing screening and/or imaging tests; and multidisciplinary follow-up. The national context reflects the underreporting of the incidence of CC, due to possible difficulties in its diagnosis. It is worth mentioning that the patient's prognosis varies according to the structure of the service, access to complementary exams, specialized medical knowledge and early treatment: clinical, surgical or endovascular.	The need to disseminate pre- and postnatal screening, easy access to diagnostic tests, adequate treatment, continuing education of the health team and caregivers, as well as carrying out epidemiological studies that contribute to an effective strategic planning in public health.

Prenatal diagnosis of double aortic arch	NORONHA et. al., 2021	The authors describe a case of prenatal diagnosis ofDouble Aortic Arch (DAA).	The diagnosis of AAD can be made through the 3VT projection described by Yagel et al. Early surgical repair of AAD has been reported to eliminate symptoms in over 70% of cases, although airflow limitation may persist due to residual tracheal stenosis.	Although challenging, the prenatal diagnosis of AAD allows for timely characterization of the vascular ring and facilitates the planning of surgical intervention before or shortly after the development of symptoms. Although symptoms may not resolve immediately, early separation of the AAD is crucial to prevent long-term sequelae of tracheobronchial compression and feeding difficulties.
Intrauterine diagnosis of complete atrioventricular septal disease: case report	STEPS et. al., 2018	The authors describe a case of prenatal diagnosis ofcomplete atrioventricular septal disease.	Heart rhythm analysis allows for a more complete and detailed assessment. Ventricular hypoplasia and atrioventricular septal defects are the most frequent defects detected prenatally through the 4-chamber section. The prenatal diagnosis of atrioventricular septal defect provides a complete and early repair in relation to cases diagnosed only in the postnatal period.	Routine obstetric ultrasound has one of its main functions the identification of congenital heart diseases, as the identification of this pathology indicates the need for a prompt approach soon after birth. Identification in the prenatal period reduces the chances of perinatal mortality and morbidity.
Public Policies: the importance of effective applicability for early detection of congenital heart disease	HAPPY et. al., 2021	To assess whether effectively implemented public policies interfere with the early detection of congenital heart disease.	It was noticed that 43.14% of the pregnant women went to six or more prenatal consultations, as recommended. In 60.78% of newborns with CC, the diagnosis occurred only after birth. There were reports from mothers about not having a multidisciplinary team during prenatal care.	With trained health professionals and the performance of the multidisciplinary team, it is possible to notice suggestive signs of CC, promote qualified care and detect CC early. Considering material and human resources, it is possible to start treatment at the service of origin or refer, including under treatment away from home (DT), recommended by the Ministry of Health.
The impact of the advent of Fetal Medicine on the diagnosis, treatment and prognosis of corrected transposition of the great arteries (TCGA) – a case report	VITORINO et. al., 2022	To highlight the advent of fetal medicine in the diagnosis, treatment and prognosis of TCGA.	It was observed that the pathology presented is a rare congenital heart disease, and its prenatal diagnosis can be quite difficult, especially as in the case described, in which there are no other associated abnormalities that are more easily detected. Due to these factors, one can perceive the relevance and peculiarity of the case. Furthermore, CTGA in the absence of major cardiac anomalies is believed to have a good outcome, especially in the presence of a prenatal diagnosis, as in a case series comparing the impact of prenatal versus postnatal diagnosis of CTGA those with prenatal diagnosis had greater intervention-free survival.	The advent of fetal medicine is of paramount importance for prenatal diagnosis, since the moment of diagnosis directly interferes with survival without interventions.

Intrauterine surgeries: alternative intervention for congenital heart disease	BARCELOS, 2018	To demonstrate the evolution of intrauterine surgery for cardiac correction, based on the evaluation of the benefits to the fetus and aiming at minimal and acceptable risks for the mother.	There is an annual increase of about 5,800 cases of adults with congenital heart disease requiring outpatient medical follow-up in the country. The difficulty for patients to reach specialized centers (due to the insufficient number of centers or the geographic distribution), the diagnosis not made by the assistant physician or the lack of adequate management can encourage the increase in the number of adults with congenital heart disease. These data further fuel the need for investment in prenatal diagnoses and treatments performed according to pre-established criteria.	Monitoring the evolution of intrauterine surgery is a challenge to be faced, and its performance in Brazil is still premature. However, with the refinement of cardiac puncture techniques and the qualification of professionals, their results are promising.

In the present integrative review, 14 articles met the previously established inclusion criteria. Regarding the type of research design of the articles evaluated, the sample showed: six literature reviews, two epidemiological studies and other case reports. Regarding the objective of this review, the articles expressed all positive results for the importance of diagnosing congenital heart diseases during prenatal care.

DISCUSSION

The description in the literature of early diagnosis of congenital heart diseases is still incipient. As these are resources that are still in the process of incorporating accessibility, such as exams and procedures in fetal medicine, few recent studies provide subsidies for impact estimation. Despite this, it is possible to infer the change in the prognosis of newborns in the face of a favorable scenario for quality prenatal care.

There is no controversy regarding the high prevalence of fetal cardiac abnormalities in pregnancies at increased risk, whether due to maternal or fetal factors (family history of heart disease, exposure to teratogens, exposure to drugs that cause fetal distress, maternal metabolic disorders, older age than 35 years, viral infections, collagenosis, polyhydramnios, oligohydramnios, increased nuchal translucency, extracardiac anomalies, cardiac arrhythmias, non-immune fetal hydrops, restricted intrauterine growth, and suspected cardiac anomaly on previous ultrasound examination) (BACALTCHUK et. al., 2001).

Since the incidence of cardiac malformations in the population is around 1%, it is understood why fetal echocardiography must not be performed indiscriminately in pregnant women outside the risk group. However, there are numerous advantages in avoiding or recognizing fetal cardiac abnormalities. In cases of normal outcome, peace of mind is returned to the parents. In cases of cardiac abnormality, it is possible to plan the delivery in a hospital that has the necessary support to assist the newborn with heart disease. In cases of fetal arrhythmias and heart failure, treatment through drugs is a reality and, in the future, intrauterine surgical correction of malformations will probably become possible (LOPES et al., 1990).

CONCLUSIONS

In the last 4 years, all studies have positively reinforced the importance of instilling the diagnosis of congenital heart disease in the level of prenatal care. This strategy aims to

better elucidate cardiological treatment of conceptuses with heart disease and, therefore, may represent the difference between life and death in the perinatal period, as well as impacting the reduction of costs for the health system.

For these reasons, it is necessary to encourage new research in the area and the development of effective programs to finance routine ultrasound examinations by the Unified Health System for pregnant women, in addition to raising awareness among the medical community, sonographers and the population itself about this reality.

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