ANALYSIS OF THE CURRENT PROTOCOL FOR SCREENING AND EARLY DIAGNOSIS OF ACYANOGENIC CONGENITAL HEART DISEASE IN PEDIATRICS

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INTRODUCTION

Congenital malformations are developmental disorders present at birth that appear in the embryonic period, and this includes any structural, functional, or metabolic alteration that causes physical or mental abnormalities in the individual. These anomalies, when caused by anatomical defects of the heart or the associated great vessels, are called congenital heart diseases, classified as cyanogenic or acyanogenic due to the clinical signs presented and the cardiovascular structures that present alterations. We can identify correlations between possible factors associated with some genetic syndromes, such as Down's Syndrome, in addition to adverse maternal conditions during pregnancy, such as diabetes, hypertension, obesity, and use of licit and illicit substances, as well as family history. In childhood, there is evidence that the presence of congenital heart disease can affect physical and weight development, as well as motor, cognitive, and neurological functioning, represented by a higher incidence of academic difficulties, behavioral problems, speech delays, inattention, and hyperactivity in patients with such conditions. About 1.35 million babies are born with congenital heart disease each year worldwide. The comprehensive care of children with congenital heart disease in Brazil is one of the biggest challenges of the Brazilian Unified Health System (S.U.S.), represented by its high incidence of 8 to 10 cases per 1,000 live births, constituting the 3rd cause of neonatal death in the country. It is known through technological resources, such as echocardiography, that fetal mortality from congenital heart disease is also high, ranging from 15% to 25% of cases. Comprehensive pediatric care for infants with heart disease requires resources and infrastructure, and it is a real challenge to develop a successful and effective program in an environment with limited resources. Providing optimal care for all these patients is a daunting task and requires adequate planning at various levels of the healthcare system. Amid this scenario, the Federal Government together with the Ministry of Health launched the National Plan of Assistance to Children with Congenital Heart Disease, which aims to integrate, qualify, and expand the actions and services of assistance to children with congenital heart disease within the public health network. Among the guidelines of this plan, greater investment in technological resources, training and qualification of professionals, expansion of prenatal and neonatal diagnosis, and greater cardiovascular care, including early neonatal surgical interventions, are identified. Prenatal diagnosis of congenital heart defects is a complex process that involves a specialized multidisciplinary medical team and specific technological support. In this context, the role of fetal echocardiography stands out, which promotes the detection of anatomical cardiac changes, in addition to abnormal functioning, and, through sequential tests, allows to assess fetal hemodynamics and cardiovascular status from the time of diagnosis until delivery. The prenatal identification of these pathologies provides an early understanding of the fetal condition by parents and guardians, besides conferring numerous benefits such as accurate diagnosis, early referral to a trained health professional, birth planning within the conditions found, analysis of available treatments, and improved prognosis. According to the National Plan, the protocol for the diagnosis and treatment of these heart diseases must be done by performing some procedures listed below in the prenatal period, delivery, and neonatal period. Prenatal care must be performed according to the recommendations of the Ministry of Health, checking the maternal risk factors for the development of congenital heart disease.
diseases in the fetus and performing obstetric ultrasonography (USG) at an appropriate gestational week (between the 18th and 22nd week) to screen for heart diseases with a trained professional; if there is any change in the USG, the pregnant woman must be referred to a Reference Center in order to investigate the severity of the malformation through fetal echocardiography and make available, depending on the need, benefits and risks, a programmed intervention, addressing the malformation intrauterus, or after delivery. The birth must take place in a maternity hospital with a multiprofessional team trained for neonatal care, if necessary, and in a Reference Center. In the neonatal period, the newborn must be adequately received and cared for, a thorough physical examination must be performed, highlighting the presence of signs of respiratory distress and congestive heart failure, changes in cardiac auscultation and in the morphology and dynamics of the precordium, in addition to pulse oximetry (the little heart test) between 24 and 48 hours of life and before hospital discharge, with appropriate measurement. Subsequently, if there is an alteration in pulse oximetry, an echocardiogram with color flow mapping must be performed. Over the years, the prospects for children with heart disease have changed considerably due to technological and therapeutic advances. Early screening for such conditions is necessary in order to reduce neonatal morbidity and mortality, increase survival, and mitigate the suffering of the child with the disease.

RESULTS AND DISCUSSION

With the survey, 1297 medical records were analyzed, and 60 of them had the pre-established necessary criteria, representing a 5% inclusion rate. Among the 1237 medical records that were disapproved, 93% represented children aged 2 years or older on 06/30/2019. The other 7% were excluded for the following reasons: gestational age of less than 34 weeks, absence of Congenital Heart Disease, presence of Cyanogenic Congenital Heart Disease or other heart disease, lack of diagnosis and inadequate medical records. It is important to emphasize that the medical records were considered inadequate due to the fact that they belong to the Pediatric Cardiology sector and contain only the Little Tongue Test and/or the Little Ear Test, because they lack information or the information contained is not understandable.

Graph 1: Causes for disapproval of medical records.

The medical records that presented all the criteria for inclusion in the research project, when analyzed were divided into 3 groups of according to the characteristic of the diagnosis, namely: Prenatal Diagnosis, Neonatal Diagnosis and Late Postnatal Diagnosis.

Graph 2 proportion of diagnosis per period
PRENATAL DIAGNOSIS

Based on the analyzed medical records that fit the criteria for inclusion in the research project, only 3% had a pre-diagnosis acyanogenic congenital heart disease. Prenatal diagnosis is characterized by performing the Fetal echocardiogram during pregnancy, enabling the monitoring of fetal hemodynamics and cardiovascular status from the time of diagnosis until childbirth. There was no information in the medical records regarding prenatal care, such as possible changes in ultrasound scans or risk factors mothers, elements that would lead to the request of the fetal echocardiogram. However, in these records there is a description of childbirth in Centro of reference with adequate reception and care for the newborn. In addition, immediate changes were characterized in the physical examination, which showed signs of cardiovascular disease, such as: murmur and lower members. The diagnoses made during intrauterine life were confirmed in the neonatal period by echocardiography. In view of this, they were sent for trained professionals and for a specialized service center. It is observed that, although it is not possible to identify the factors directly related to the request for the fetal echocardiography exam, the performing this in the prenatal period brought clear benefits to patients, especially regarding childbirth, neonatal reception and specific care after the birth.

Graph 3: Proportion of heart diseases in prenatal diagnosis

NEONATAL DIAGNOSIS

Based on the analyzed medical records that fit the criteria for inclusion in the research project, 69% had a Neonatal diagnosis of Acyanogenic congenital heart disease. Since they were considered neonatal diagnoses all those performed after birth, however, before hospital discharge. Of these records, 66% had only one heart disease Congenital Acyanogenic:

Graph 4: Proportion of single heart disease in neonatal diagnosis.

The other 34% had two or more Congenital Heart Defects Acyanogenic:

Graph 5: Proportion of two or more heart diseases in neonatal diagnosis.

The diagnosis of these heart diseases was
performed before 48 hours of life in 24% of the patients and after 48 hours of life in 65% of them, being that in 11% of the records the time was not mentioned. Among those that presented the date of diagnosis, the mean number of days was 12.5. With regard to prenatal care, it was found that 54% of pregnant women did so in accordance with the recommendations of the Ministry of Health, having a number of consultations greater than 6, with the average number of consultations between them was 8. Despite a considerable rate of adherence to prenatal care, no information was obtained regarding ultrasound changes that would lead to a possible referral to carry out Fetal echocardiography during the prenatal period. The medical records of 41% of these patients did not contain information to regarding maternal risk factors for the development of heart diseases congenital. The other 59% had the following maternal morbidities: 42% were elderly pregnant women (over 35 years old), 38% had Gestational Diabetes Mellitus, 33% had Urinary Tract Infection during pregnancy, 21% had Systemic Arterial Hypertension, 21% have Hypothyroidism, 17% have Type 2 Diabetes Mellitus, 13% are obese, 13% had a specific hypertensive disease of pregnancy, had pre-eclampsia, 7% had a family history of heart disease congenital, 4% have Hyperthyroidism, 4% have Pneumonia in the pregnancy, 5% used tobacco during pregnancy and 2% used alcohol during pregnancy, these being important factors for referral pregnant women to perform a fetal echocardiogram. We observe that the maternal comorbidities were not sufficiently relevant factors to a meticulous investigation of possible alterations in the fetus, since in just 4% of the cases, which corresponds to one patient, the fetal echocardiogram was accomplished. The only fetal echocardiogram performed did not reveal any heart disease, but explained that this diagnosis could not be ruled out. Therefore, after birth and before 48 hours of the newborn’s life, an echocardiogram was performed that confirmed the diagnosis of Persistence of the Arterial Duct. This way, the importance of carrying out a fetal echocardiogram to obtain an early diagnosis. Regarding conditions at birth: 15% of patients required neonatal resuscitation, 12% required mechanical ventilation, 4% lacked a venous umbilical catheter, 2% used CPAP and 2% transfusion blood was performed. It is important to highlight that 41% of the patients who fit the category of Neonatal Diagnosis has mentioned hospitalization in its medical records in the neonatal Intensive Care Unit (ICU), and the average time hospital stay was 17 days. The prolonged length of hospital stay added to severity of the condition that many of these patients presented, may have contributed directly to the precocity of the diagnosis, since the hospitalized patients receive daily evaluations, a fact that optimizes the finding of characteristic signs and symptoms, in addition to greater ease in request for subsidiary examinations. Among the main comorbidities and signs and symptoms characterized during hospitalization at birth, stand out:

Graph 6: Main comorbidities found in newborns with neonatal diagnosis of acyanogenic congenital heart disease.
Graph 7: Other comorbidities found in newborns with a neonatal diagnosis of acyanogenic congenital heart disease.

Were also found, in low expressive values, the following comorbidities: urinary tract infection, neonatal asphyxia, acute respiratory failure, bronchiolitis, anemia, conjunctivitis, hypertriglyceridemia and thrombocytopenia. These, however, with less relevance for the search.

Graph 8: Main signs and symptoms found in newborns with neonatal diagnosis of acyanogenic congenital heart disease.

The following signs and symptoms were also found: tremor and cough. These, however, with lesser relevance to the research. Among the data observed in the graphs above, the predominance of patients who had pulmonary hypertension persistent, signs of respiratory distress, cyanosis, tiredness during feedings and murmur on cardiac auscultation, all related to congenital heart disease. Factors such as the presence of fetal extracardiac malformations and karyotype amended are also worth mentioning, as they advocate the request of the Neonatal echocardiogram. It is important to point out that some of the patients had more than one comorbidity and/or signs and symptoms. Only 12% of these patients were described in their medical records information about the little heart test (oximetry test of pulse), and 5% of them did not pass. Mean days to diagnosis of the cases that failed the exam was 7 days, noting, therefore, the relevance test for early diagnosis. All patients had a diagnosis of congenital heart disease acyanogenic confirmed by echocardiogram.

LATE POST CHRISTMAS DIAGNOSIS

Based on the analyzed medical records that fit the criteria for inclusion of the research project, 28% had Postnatal Diagnosis Late Acyanogenic Congenital Heart Disease. As the criterion used to designate them that way the diagnosis made after discharge from the maternity ward.

Graph 9: Proportion of heart diseases in late postnatal diagnosis.
Regarding prenatal care, there was little or no information in the analyzed medical records. Only 24% of those cited or briefly described the gestational follow-up. Thus, it is not known whether pregnant women received adequate care during the prenatal period, their risk factors, as well as the existence of morbidities and tests altered fetuses. Furthermore, there were no data related to a possible referral to perform a fetal echocardiogram or to centers of reference in Pediatric Cardiology. It must be noted that prenatal care is of great importance in the early diagnosis of heart diseases congenital. Know how to identify the main maternal risk factors, and use the technological tools available, it is essential to avoid the underdiagnosis and its consequences. As for delivery and birth conditions, only one patient was born in a specialized reference center and the reasons for not were included in their records. None of the patients required resuscitation and 18% were admitted to the Neonatal ICU, for an average of 5.3 days. Despite the high proportion of patients who were hospitalized in Intensive Care Unit, the verification of signs and symptoms were only performed after discharge from the maternity ward. An important fact to note is that 89% of patients who fit the Postnatal Diagnosis category Late presented specific symptomatology for congenital heart diseases, but did not receive specific investigations for such signs and symptoms. Among the clinical findings on examination, the following stand out:

### Graph 10: Main signs and symptoms found in newborns with late postnatal diagnosis of acyanogenic congenital heart disease.

In the graph above, patients who presented: murmur and hyperphonesis of the second sound on cardiac auscultation, signs of discomfort breathing, cyanosis and tiredness during breastfeeding, all related to congenital cardiopatics. It is important to point out that some of the patients showed more than one sign and symptom, and even so they were not investigated. Factors such as the presence of fetal extracardiac malformations, altered karyotype and oligoamnios are also worth mentioning, as they are advocates for requesting a neonatal echocardiogram. However, no there were indications of request or performance of the examination in the documents analyzed.

### Graph 11: Main comorbidities found in patients with late postnatal diagnosis of acyanogenic congenital heart disease.
Only 12% of patients were described in their medical records information about the pulse oximetry exam (little heart test), which they passed the exam. The importance of the examination in the neonatal screening of congenital heart diseases and the repercussions unfavorable effects of such a low rate of application, a factor that contributes directly to the increase in the number of patients discharged from maternity without the diagnosis and investigation of heart disease. On average, these patients received the diagnosis at 91.7 days of age. life, having reached the Children’s Cardiology service via referral by health professionals from Basic Units and Emergency Attendance, since they already had a clinical picture and repercussions systemic symptoms that may be related to the disease, such as: fainting, difficulty and tiredness during feedings and deficit in weight and height gain. All patients had a diagnosis of congenital heart disease acyanogenic confirmed by Echocardiogram in late phase and currently are being followed up by the Child Cardiologist.

CONCLUSION

The analysis of the records of the Children’s Cardiology sector of the Set Hospitalar de Sorocaba (CHS) from July 2018 to June 2019 showed predominance of diagnoses in the neonatal period (69%), to the detriment of prenatal (3%) and late postnatal (28%) periods, the latter also obviously expressive. The reasons for performing a fetal echocardiogram that generated the prenatal diagnosis are unknown. However, it is noted that the realization of this examination in the period in question provided clear benefits to patients, mainly regarding childbirth, neonatal reception, care after birth, follow-up with professionals qualified and better prognosis of the disease. In the neonatal period, it was observed that the main factors for the diagnostic conclusion were: the meticulous performance of the physical examination, the greatest period of hospitalization and the execution of the little heart test. The increase in time the patients remained hospitalized directly interfered with the early diagnosis due to daily monitoring of signs, symptoms and comorbidities presented by them, leading to the realization of the Echocardiogram and finding of heart disease. Multiple factors are related to the provoking causes of late postnatal diagnosis, with emphasis on non-performance Fetal Echocardiogram and no finding of specific signs and symptoms for congenital heart diseases, which were only observed after discharge from maternity. It is observed that these patients, at the time of diagnosis, already presented systemic repercussions and worse prognosis when compared to patients who obtained an early diagnosis. Therefore, there is a high rate of neonatal and prenatal diagnoses birth, corresponding to 72% of cases, of patients who used the service of Child Cardiology at Conjunto Hospitalar de Sorocaba. This index is extreme relevance as early diagnosis reduces the neonatal morbidity and mortality, increases survival, in addition to mitigating suffering of the child with the disease. However, there is still a need to increase this rate in order to minimize the repercussions for the patient generated by the late diagnosis, in addition to promoting an improvement in quality of life and reduction of public spending on health. The protocol of the National Assistance Plan for Children with Heart Disease Congenital, includes a thorough evaluation of the baby with heart disease from the period until its referral and postnatal follow-up, promoting better prognosis for children affected with such conditions. However, comprehensive pediatric care for infants with heart disease requires resources and infrastructure, being a real challenge to execute the proposals of the plan successfully.
and effectively in an environment with resources limited. Providing optimal care for all these patients is an important task and still requires improvement at various levels of the system of health.

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International Journal of Health Science ISSN 2764-0159 DOI 10.22533/at.ed.1593502307073