

Newborns with congenital heart diseases: epidemiological data from a single reference center in Brazil

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Summary

Objective: to describe the epidemiological data of the population born with the diagnosis of Congenital Heart Disease (CHD); to compare diagnoses made using fetal echocardiography with the findings from postnatal echocardiography or anatomopathological examination of the heart; and to evaluate mortality among newborns that underwent surgical treatment.

Methods: this was a cohort study with information gathered from the medical records of the pregnant women and their newborns diagnosed with CHD during the fetal or postnatal periods, between January 2008 and December 2012. Means, standard deviations and maximum and minimum values were calculated for the quantitative variables. Relative and absolute values were calculated for the qualitative variables. The heart

malformations were categorized in four groups: complex lesions, significant lesions, minor lesions and others.

Results: we detected postnatal incidence of CHD of 1.9% at our service. The mean maternal age was 28.3 years and 10 (21.3%) of the pregnant women were ≥ 35 years old. The mean gestational age at the time of performing the fetal echocardiogram was 27.8 weeks. Mean gestational age at delivery was 38 weeks, and the mean weight of the newborns was 2,644.5 grams. Regarding the diagnosis of CHD, there were: 23 complex lesions (39%); 15 significant lesions (26%); 10 minor lesions (17%); 4 other lesions (7%) and 6 normal anatomies (10%). The diagnosis of CHD made on the fetus and postnatally coincided in 77.6% of the cases. A total of 27 patients (60%) underwent surgery, and the outcome was neonatal death in five cases.

Conclusion: we detected postnatal incidence of CHD of 1.9%, and it was more common among older pregnant women and with late detection in the intrauterine period. Complex heart diseases predominated, thus making it difficult to have a good result regarding neonatal mortality rates.

Key words: congenital heart diseases, prenatal diagnosis, newborn, echocardiography.

Introduction

Congenital heart diseases (CHDs) are the most common congenital fetal malformations and are responsible for a high rate of child mortality and morbidity. The prenatal incidence of CHD ranges from 2.4 to 52% (1-6). This huge variability is due to the policies adopted in different countries. Detection of CHD is much greater in regions where it is mandatory to perform at least one fetal echocardiogram during the pregnancy (7). The postnatal incidence is around 0.3 to 1.2% of live births (8, 9). This decrease between the prenatal and postnatal rates can be explained by the fact that many countries allow termination of pregnancy in cases of congenital malformation (7). According to the Mortality Information System of the Brazilian Ministry of Health, the second greatest cause of early and late neonatal death in the southeastern region are congenital malformations, accounting for 18.6 and 21.1%, respectively (10). In the light of this situation, the importance of early diagnosis of congenital malformations can be seen, especially with regard to CHDs, which are the most com-

mon congenital malformations (11). This early detection enables appropriate family counseling, programmed intrauterine intervention (when necessary) and planned delivery assisted by a specialized multidisciplinary team (1, 11-13).

In addition to an adequate screening program for CHDs during the fetal period, knowledge of the epidemiological data of this population needs to be promoted within a hospital structure, so as to draw up a plan of action that optimizes good results in the service. Thus, the objective of the present study was to describe the epidemiological data of the population born at our service with a diagnosis of CHD between January 2008 and December 2012. Moreover, we sought to compare the diagnoses made using fetal echocardiography with the findings from postnatal echocardiography or anatomopathological examination of the heart (gold standard) and to evaluate mortality among the newborns that underwent surgical treatment.

Methods

This was a historical cohort study with information gathered from the medical records of pregnant women and their newborns diagnosed with CHD, during either the fetal or the postnatal period. This investigation was carried out by the Fetal Cardiology Sector of the Department of Obstetrics, in conjunction with the Discipline of Neonatology of the Department of Pediatrics, Federal University of São Paulo (UNIFESP). The present study was approved by the Ethics Committee of UNIFESP.

The inclusion criteria were that the subjects should be fetuses and newborns diagnosed with CHD that were born at our service between January 2008 and December 2012. The exclusion criteria were situations in which adequate data-gathering from the fetal or postnatal echocardiogram was impossible, and diagnoses of persistent arterial canal.

The quantitative variables evaluated were: maternal age, gestational age at the time of fetus' CHD diagno-

sis, parity, gestational age at the time of birth, birth weight and Apgar at the first and fifth minutes. The means, standard deviations and maximum and minimum values were calculated for these variables. The qualitative variables evaluated were the indication to produce a fetal echocardiogram, karyotype analysis, presence of associated extracardiac malformations, type of delivery, sex of the newborn, diagnosis of CHD, newborns that underwent surgical treatment and neonatal death.

The CHDs were categorized in four major groups: complex lesions, significant lesions, minor lesions and others, in accordance with the classifications of Hunter et al. (14) and Wren et al. (15) (Tab. 1). Relative and absolute values were calculated to compare the types of CHD found through pre and postnatal echocardiography.

Results

During the five-year period, 4,835 children were born at our service, of which 94 were considered to have CHD during either the prenatal or the postnatal period. This corresponded to a postnatal incidence of CHD of 1.9% in our service, ranging from 1.3% to 2.5% according to the year studied. From these newborns diagnosed with CHD, 46 (48.9%) were excluded from the analysis due to flaws in recording the data relating to the diagnosis of CHD.

The mean maternal age was 28.3 years, and 10 (21.3%) of the pregnant women were ≥ 35 years of age. The mean gestational age at the time of producing the fetal echocardiogram was 27.8 weeks. The means for the number of gestations, parity and abortions were 2.4, 1 and 16, respectively. The indication for producing a fetal echocardiogram was because of maternal causes in 16 patients (33%), fetal causes in 29 patients (60%) and family causes in 3 patients (6%). In 50% of the fetuses, there was a suspicion of associated extracardiac malformation during the intrauterine period, although karyotype was only col-

Table 1. Classification system for congenital heart diseases used, according to the complexity of the heart anatomical abnormalities.

Classification	Congenital heart diseases
Complex	Heterotaxy or atrial isomerism, atresia or severe hypoplasia of a valve or chamber (hypoplastic left heart syndrome, pulmonary atresias, tricuspid atresias, aortic atresia, mitral atresia, Ebstein's anomaly), abnormalities of the valve inlet or outlet (complete atrioventricular septal defect, truncus arteriosus, double inlet left or right ventricle, double outlet left or right ventricle congenitally corrected transposition of the great arteries).
Significant	Transposition of the great vessels, tetralogy of Fallot, large ventricular septal defect, coarctation of the aorta, aortopulmonary window, critical aortic or pulmonary stenosis, partial atrioventricular septal defect, total anomalous pulmonary venous connection, tricuspid valve dysplasia (no Ebstein's anomaly).
Minor	Small ventricular septal defect, less severe aortic or pulmonary stenosis.
Others	Dysrhythmias, cardiomyopathies, secondary dextrocardia/levocardia, pulmonary sequestration, restrictive ductus arteriosus.

*This classification was adapted from Hunter et al. (14) and Wren et al. (15).

lected for nine patients. In only 31% of the cases delivery took place vaginally. The mean gestational age at birth was 38 weeks, the mean weight of the newborns was 2,644.5 grams and the Apgar score at the first minute was 6 and at the fifth minute 8. Female newborns represented 51.7% of our sample (Tab. 2). Regarding the diagnosis of CHD, taking either the postnatal echocardiogram or the anatomopathological examination as gold standards, there were 23 complex lesions (39%), 15 significant lesions (26%), 10 minor lesions (17%), 4 other lesions (7%) and 6 normal anatomies (10%) (Tab. 3).

Six cases were considered to have normal hearts in the neonatal period, but had been considered to be abnormal in the records from the fetal period. These cases consisted of two fetuses diagnosed with sus-

pected cardiomegaly, one with restrictive ductus arteriosus, one with suspected coarctation of the aorta, one with a small interventricular communication and one with persistent left vena cava drainage into the coronary sinus. Only one newborn presented an altered postnatal echocardiogram with a normal fetal echocardiogram. The diagnosis in this case was an interventricular communication with no need for surgical treatment (Tab. 3).

The diagnosis of CHD made on the fetus and postnatally coincided in 77.6% of the cases, considering the classification used in the present study. Among the patients diagnosed with CHD, 16 cases led to death within the first year of follow-up, while 12 newborns survived. A total of 27 patients (60%) underwent surgery, among whom five evolved to neonatal death,

Table 2. Characteristics of the population studied (n = 58 newborns).

Variables	
Maternal age in years (mean / min-max)	28.3 (14 - 48)
≥ 35 years – N (%)	10 (21.3%)
Gestational age of the fetal echocardiogram in weeks (mean / min-max)	27.8 (20 - 34.5)
Gestational history (mean / min-max)	
Gestation	2.4 (1 - 5)
Parity	1 (0 - 3)
Abortion	0.3 (0 - 3)
Indication for a fetal heart examination – N (%)	
Maternal	16 (33%)
Fetal	29 (60%)
Family	3 (6.2%)
Presence of fetal malformation – N (%)	29 (50%)
Karyotype – N	9
Sex of the newborn	
Female – N (%)	30 (51.7%)
Gestational age at birth (mean / min-max)	38 (32.8 - 41.4)
Type of birth	
Normal – N (%)	18 (31%)
Birth weight in grams (mean / min-max)	2644.5 (1310 - 3700)
Apgar (mean / min-max)	
1st minute	6.5 (1 - 9)
5th minute	8 (1 - 10)

SD – standard deviation, N – Absolute number, (%) – Relative number, min – minimum, max – maximum.

Table 3. Diagnosis of congenital heart diseases during the fetal and neonatal periods, classified according to the degree of complexity of the clinical and/or surgical treatment.

Congenital heart disease classified according to severity – N (%)	Prenatal echocardiogram (n = 58)	Postnatal echocardiogram (n = 58)
Complex heart disease	23 (39%)	23 (39%)
Significant heart disease	17 (29%)	15 (26%)
Minor heart disease	10 (17%)	10 (17%)
Other heart diseases	7 (12%)	4 (7%)
Normal anatomy	1 (2%)	6 (10%)

N – Absolute number, (%) – Relative number.

and one of these was a case of early death, with a diagnosis of critical pulmonary valve stenosis, which was classified as a significant lesion.

The Figure 1 presents four cases of prenatal echocardiogram diagnosis of CHD: double outlet right ventricle, tetralogy of Fallot, atrioventricular septum defect, and pulmonary stenosis.

Discussion

This retrospective study described the sample of CHD diagnoses at a tertiary-level service between January 2008 and December 2012. Our aim was to find information about the population of children with CHDs diagnosed at this service so as to improve our structure for specialized treatment of these patients.

The postnatal incidence of CHDs at the service was 1.9%, i.e. slightly higher than the data from the worldwide literature, which ranges from 0.3 to 1.2%. This corroborates the importance of better planning for early detection of these diseases and better-structured hospital support (8, 9).

Approximately 20% of the pregnant women who presented fetuses with CHD were more advanced in age, and this has been reported in several studies on CHDs (16-18). Also, we observed that although the fetal diagnosis of CHD should be made between the 18th and 24th weeks, it is still performed at a later stage of pregnancy, at approximately the 28th week (19-21). This is because these pregnant women are referred late to primary services, thereby delaying the diagnosis and, consequently, adequate specialized prenatal follow-up and delivery management by a multidisciplinary team.

Among the patients with an intrauterine diagnosis of CHD, 60% underwent echocardiography during the gestational period because of the fetal risk factor. This

was in accordance with recommendations in the current literature, thus demonstrating the extent to which fetal causes are associated with CHDs (3, 6, 22, 23). There are still some difficulties in analyzing the karyotype, probably because some pregnant women refuse to undergo sample collection for this examination, and also because of financial difficulties, since the analysis for this examination is only carried out in private laboratories, while the vast majority of our patients are of a lower socioeconomic level.

The birth weight of the patients with CHD maintained a mean of more than 2,500 grams, with no relationship with low birth weight due to the presence of CHDs, as reported in some studies. This was probably due to the fact that, in the present assessment, patients diagnosed with persistent ductus arteriosus were excluded (24).

Among the diagnoses of CHD, 65% of the cases presented complex and significant lesions. The explanation for this is that our service is at tertiary level, which creates selection bias. This also reinforces the idea that stronger fetal screening should be implemented so that the multidisciplinary team that will follow-up the pregnant woman and her newborn can become better prepared. Among the fetal echocardiograms performed, only one did not detect a heart abnormality that was considered to be a small lesions (small interventricular communication). Moreover, the pre and postnatal diagnoses coincided in 77.6% of the cases, respecting the classification used in the present study. This may demonstrate the efficiency of the fetal screening and diagnosis services. However, these numbers need to be improved, and performing this examination on the fetus earlier needs to be promoted (19-21).

The mortality rates are still high, even among those that underwent a surgical procedure. However, the degree of complexity of the CHD at our service was

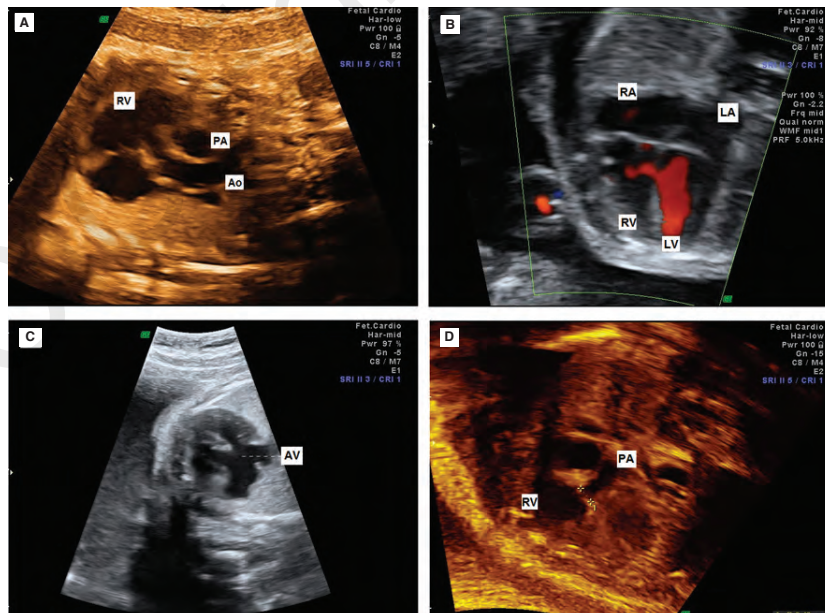


Figure 1. Prenatal echocardiogram diagnosis of congenital heart diseases: (a) Double outlet right ventricle; (b) Tetralogy of Fallot; (c) Atrioventricular septum defect; (d) Pulmonary stenosis. RA: right atrium; LA: left atrium; RV: right ventricle; LV: left ventricle; AV: atrioventricular septum; PA: pulmonary artery; Ao: Aorta.

very high, which makes it difficult to have a good postnatal result in these cases. Nevertheless, a constant search for improvements in the organization of hospital services is always a positive predictive factor for decreasing the morbidity and mortality of newborns with CHD.

As a limitation of the present study, we observed that nearly 50% of the newborns diagnosed with CHD were excluded from the sample due to flaws in obtaining data from the medical records, especially with regard to pre and postnatal echocardiographic reports. At our service, we still use a paper-based reporting system, which would explain this high rate of flaws. We believe that with the computerization of our reporting system, we will in the future be able to improve data organization, thereby making patient follow-up easier, as well as making it easier to conduct future research.

In summary, we detected postnatal incidence of CHDs of 1.9% at our service, and they were more common among pregnant women of more advanced age and with late detection during the intrauterine period. Complex heart diseases predominated, which made it difficult to have good results regarding neonatal mortality rates.

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