Incidence of Congenital Heart Defects in a Public Maternity Hospital in the Beginning of the National Congenital Heart Defects Program

Incidencia de cardiopatías congénitas en una maternidad pública en los inicios del Programa Nacional de Cardiopatías Congénitas

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ABSTRACT

Introduction: Congenital heart defects (CHD) are among the most common congenital diseases with great impact in infant morbidity and mortality; however, their real incidence in our country is unknown. The Ministry of Health created the National Congenital Heart Defects Program to contribute to the reduction of infant mortality for this cause. Therefore, determining the incidence and the relative frequency of CHD at the Hospital Materno Infantil Ramón Sardá, a public tertiary institution specialized in high risk pregnancies and congenital defects would provide information to the Program.

Objectives: 1) To determine the incidence and relative frequency of CHD of life birth (LB) at the Hospital Materno Infantil Ramón Sardá in the beginning of the PNCC program. 2) To evaluate the association with other congenital defects. 3) To analyze the distribution of CHD by sex.

Methods: The study has a descriptive, observational and retrospective design. The digital clinical records of 1,161 LB with confirmed diagnosis of CHD from the Section of Pediatric Cardiology, Hospital Materno Infantil Ramón Sardá, between January 1, 1998 and December 31, 2011, evaluated during hospitalization and follow-up at the outpatient clinic, were analyzed. Congenital heart defects were classified based on their severity according to the meta-analysis published by Hoffman and Kaplan in USA assessing their incidence and relative frequency.

Categorical data are presented as number of absolute cases and percentage.

Results: From a total of 92,725 LB during the study period, CHD were present in 1,161 of cases (12.5/1000). After excluding small muscular ventricular septal defects, the incidence decreased to 7/1000 LB. The incidence of moderate and severe CHD was 44% (3 to 3.5/1000).

Conclusion: Despite being a tertiary center for high risk pregnancy and congenital defects, the incidence of CHD determined at the Hospital Materno Infantil Ramón Sardá is similar to the one published worldwide.

Key words: Newborn - Congenital Heart Defects - Incidence.
INTRODUCTION

Congenital heart defects (CHD) are the most common congenital diseases with great impact in neonatal and pediatric morbidity and mortality.

The real incidence of CHD in our country is unknown. The available data comes from foreign publications, which estimated that the incidence of CHD is 8 per 1,000 live births (LB), ranging from 2 to 50 per 1,000 LB. If this number is related with the birth rate (approximately 750,000 per year) in our country, about 6,100 infants are born with a congenital heart defect. Of these, 70% need surgery: 50% are high-complexity defects and 25% should be operated on within the first 28 days of life.

In 2010, The National Ministry of Health created The National Congenital Heart Defects Program (Programa Nacional de Cardiopatías Congénitas, PNCC) and the National Registry of Congenital Heart Defects to reduce infant mortality related with these heart diseases.

This study was performed between January 1, 1998 and December 31, 2011, to evaluate the still unknown incidence and relative frequency of CHD in LB at the Hospital Materno Infantil Ramón Sardá (HMRIS), a public tertiary care center specialized in high-risk pregnancies and congenital defects, with the aim of providing information to optimize health care programs.

METHODS

Population

Hospital Materno Infantil Ramón Sardá is a public tertiary care center specialized in high risk pregnancies with prenatal diagnosis of congenital defects, taking care of women attending the outpatient clinic and those nationwide referred to this hospital.

A retrospective observational analysis of 1,161 digital clinical LB records with confirmed diagnosis of CHD obtained between January 1, 1998 and December 31, 2011 from the HMRIS Section of Pediatric Cardiology, was performed. The infants had been evaluated during hospitalization and in subsequent follow-up at the outpatient clinic.

Stillbirths were excluded due to lack of data. Asymptomatic patent ductus arteriosus (PDA) in premature LB and full term LB < 1 month, cardiomyopathies including those in infants of diabetic mothers, patent foramen ovale < 5 mm, heart rhythm disturbances, temporary pulmonary branch stenosis and cardiac neoplasms were also excluded.

A consecutive sample was obtained from the clinical record data of all the LB with a diagnosis of CHD, confirmed by a pediatric cardiologist with clinical examination, electrocardiogram, chest x-ray and color-Doppler echocardiography.

The incidence and the relative frequency of CHD were analyzed. The incidence was calculated from the total number of cases with CHD recorded at the HMRIS per total number of LB during the period 1998-2011. The relative frequency was calculated as the percentage of each CHD per the total number of cases studied.

Categorical data are presented as number of absolute cases and percentage.

Congenital heart defects were classified according to their clinical presentation in mild, moderate and severe based on the meta-analysis published by Hoffman and Kaplan in the USA:

1) Mild: this is the group with the highest number of cases. These patients do not have symptoms, may have a soft murmur, and some of them present a spontaneous resolution of the defect. The inclusion of these defects has influence on the incidence of CHD. These defects include PDA, ventricular septal defect (VSD), atrial septal defect (ASD) and mild pulmonary stenosis (PS).

2) Moderate: many of these patients do not have symptoms and the diagnosis arises during the consultation with a pediatric cardiologist due to suspected heart disease. These patients need specialized care, yet less intensive than severe defects: pulmonary stenosis (S), ventricular septal defect associated with other malformations and coarctation of the aorta (COAO).

3) Severe: all the patients are seriously ill during the neonatal period or early infancy, and many of them can die...
before the cardiologic diagnosis. They include: hypoplastic left heart syndrome (HLHS), pulmonary atresia (PA), tricuspid valve atresia (TVA), complete transposition of the great vessels (CTGV), total anomalous pulmonary venous return (TAPR), tetralogy of Fallot (TOF), single ventricle (SV), troncus arteriosus (TA), double outlet right ventricle (DORV), Ebstein’s disease, severe aortic stenosis (AS) and complete atrioventricular canal (CAVC).

Their association with extracardiac congenital defects or chromosome abnormalities, distribution by sex and family history of CHD were recorded.

Extracardiac congenital malformations were recorded in the database as major or minor; the most common chromosome associated abnormalities and the CHD associated were also recorded. Chromosome abnormalities included trisomy 21 and Edwards, Patau, Di George and Turner syndromes.

All the patients with CHD were evaluated by the genetic specialist.

**Ethical considerations**

The protocol was reviewed and approved by the Ethical Board of each institution, excluding the informed consent form as no sensitive data or clinical follow-up were required (in accordance to the Habeas Data Act 23,326 on Protection of Personal Data.

**RESULTS**

Of a total of 92,725 LB between January 1, 1998 and December 31, 2001, 1,161 had CHD. The annual incidence ranged from 7.2 per 1,000 LB to 15.7 per 1,000 LB, representing an incidence of 12.5 per 1,000 LB (Figure 1).

When the diagnosis was made, LB age ranged from 2 hours to 28 days, with a median of 14 days; gestational age was 38 weeks or more in 96% of the cases, and birth weight ranged between 1.6 g and 4.6 kg (median 2.8 kg).

The number of CHD in the period 1998-2011 remained stable.

Figure 1 shows that the total number of CHD diagnosed increased twofold during the studied period because of small muscular VSD early diagnosis.

When small muscular VSD which present spontaneous closure before the first year of life were excluded, the incidence decreased to 7 per 1,000 LB with a range of 4.03 to 8.33 per 1,000 LB (Figure 2).

Fifty-two percent of CHD that were diagnosed during the study period corresponded to small muscular VSD which close spontaneously before the first year of age (Figure 1).

According to the CHD classification, 58% were mild (636 muscular VSD and perimembranous VSD, 33 PS and 4 PDA). The remaining 42% were moderate or severe, and would require surgery at some point of their lives.

The relative frequency of CHD per year is detailed in Table 1.

Among mild CHD, septal VSD was the most common (73.13%), followed, in order of frequency, by PS with 4.39% and ASD with 3.36%. The most frequent moderate and severe CHD were COAO (2.93%), CAVC (2.58%), TOF (2.33%) y and HLHS (2.07%) (see Table 1).

Male sex was slightly more frequent (52%), but this
difference was not statistically significant (Figure 3).

Fourteen percent of LB with CHD presented major or minor congenital malformations or chromosome abnormalities.

Congenital defects included omphalocele (VSD, PDA, CAVC), diaphragmatic hernia (HLHS), hydrocephalus (VSD), esophageal atresia (VSD, PDA), and VACTER association (VSD, PDA).

Congenital heart defects were present in the following chromosome abnormalities: Down syndrome (PDA, VSD, ASD, CAVC), Edwards syndrome 70% (VSD, PDA, DORV), Patau syndrome 60% (VSD, PDA, ASD, CAVC), total anomalous pulmonary venous return. DORV: Double outlet right ventricle. CCTGV: Congenitally corrected transposition of the great vessels.

One hundred and two cases of Down syndrome were confirmed during the analyzed period (1%) and 46 of them (45%) presented CHD. The most common defects were PDA (16/46), VSD (14/46) and ASD (8/16), followed by CAVC (6/46) and TOF (2/46).

### Table 1. Relative frequency of congenital heart defects per year

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### DISCUSSION

The progress in neonatology and the higher survival rates in LB with congenital defects produce a greater impact in morbidity and mortality rates in newborns and infants. Congenital heart defects are associated with more than 50% of neonatal and pediatric morbidity and mortality. (1, 2)

Congenital heart defects are defined as structural abnormalities of the heart or great vessels due to altered embryonic development of the heart between the third and tenth week of pregnancy. (3) They represent a heterogeneous group of disorders caused by chromosome abnormalities, Mendelian disorders, exposure to teratogenic agents and unknown etiologies. It is traditionally accepted that more than 90% of CHD are due to multifactorial polygenic inheritance. (4, 5)

Several studies about the incidence of CHD evidence great variability among them. There are
different classifications and systems of CHD registry.

In a meta-analysis of 62 studies published in the United States from 1955 to 2002 about the incidence of CHD, Hoffman and Kaplan determined a variability of 4 to 50 per 1,000 LB, with moderate to severe CHD in approximately 6 per 1,000 LB. They concluded that the difference of the reports depended mainly on the number of trivial or minor heart malformations included, on the moment the patient underwent cardiological diagnosis, technological advances and the ability of the operator. (6)

In recent population studies performed in Europe, the incidence of CHD ranges between 3.5 and 13.7 per 1,000 LB and in South America from 5 to 8 per 1,000 LB, the latter being the most widely reported in series published in our setting. (7-9)

The incidence of mild CHD, as small muscular VSD, has increased over the last years, and that of the most severe defects has remained unchanged. (10-13)

The real incidence of CHD in our country is unknown. The available data comes from foreign publications, which estimated that the incidence in Argentina is 8 per 1,000 LB. (14) If this number is associated to the birth rate (approximately 750,000 per year according to data of the Health Statistics Administration), about 6,100 infants may be born with a congenital heart defect.

Hospital Materno Infantil Ramón Sardá is, within the current health care system, a tertiary care center specialized in high-risk pregnancies with prenatal diagnosis of congenital defects and one of the public maternity hospitals with the highest number of deliveries. Yet, despite the information obtained from this hospital may be overestimated, the incidence of CHD was similar to the figures known in our setting.

Interestingly, in our population, 52% of CHD correspond to small muscular VSD which close spontaneously before the first year of age, probably because newborns are evaluated in the immediate neonatal period, the diagnosis is more accurate, the echocardiography system is more sophisticated and we have incorporated new professionals to the staff of cardiology. Small muscular VSD are the most prevalent defects in all the studies.

Among mild CHD, the relative frequency of ASD and PDA is relatively lower than the one found in other studies performed in our country. (15, 16) On the other hand, the incidence of moderate and severe CHD in our population was similar to that of other series, 4 per 1,000 LB, 2-3 per 1,000 of which required prostaglandins. Complete atrioventricular canal is the most common of these defects, followed by TOF and HLHS.

This is relevant because all the strategies aimed at reducing infant mortality related with CHD are focused on medical and surgical care in this group.

According to the information about CHD and family history, the risk of recurrence with one child affected is 3%, 10% if the mother is affected and 2% if the father has a CHD. In our population, we only found two cases with family history.

Congenital heart defects are the most common congenital diseases with great impact in pediatric morbidity and mortality.

A review reported that the real incidence of congenital defects, considering those presenting later in life as malformations of the central nervous system, may be around 5%. Minor abnormalities occur in approximately 10% of newborns. About 2-3% of LB have at least one major abnormality that is apparent at birth. Some recent studies report a higher incidence of some congenital defects or trisomies.

In 2008, the Latin American Collaborative Study of Congenital Malformations (Estudio Colaborativo Latino Americano de Malformaciones Congénitas) examined congenital defects in Latin America and reported that 12% of CHD form part of a genetic syndrome or a congenital abnormality. This percentage is similar to the 14% of CHD associated with other congenital malformations or chromosome abnormalities in our population.

The major congenital abnormalities or trisomies most frequently associated with CHD are omphalocele, gastroschisis, diaphragmatic hernia, esophageal atresia, hydrocephalus, Down syndrome, Edwards syndrome, Patau syndrome and Turner syndrome.

Four percent of CHD present Down syndrome. The
frequency of CHD in Down syndrome in our institution is within the expected range (45%) and is coincident with that mentioned in the international literature (40-60%).

The most common CHD associated with Down syndrome are PDA, VDS and ASD, as opposed to those reported by Anglo-Saxon and European countries, where ASD, particularly CAVC, are the most common.

As in other publications, we did not find significant differences by sex in CHD.

The significant advances of ultrasound techniques achieved by the end of the seventies, and the joint work of the specialists in Pediatric Cardiology and Fetal Medicine, have made fetal echocardiography a routine study in specialized centers (17), allowing the diagnosis of CHD before birth and thus improving neonatal morbidity but not mortality. The intrauterine diagnosis of CHD is extremely valuable, as it offers the couple cardiovascular genetic counseling before birth in order to provide information about the characteristics of the disease, its outcome, the therapeutic possibilities, the prognosis and the risk of recurrence in future pregnancies.

Once the prenatal diagnosis has been made, the couple might choose to end the pregnancy in those countries where abortion is legal, or to program the birth in a tertiary care center with cardiovascular facilities for the baby in order to provide immediate medical care.

The option to end pregnancy as a result of a fetal diagnosis of severe heart disease has altered the prevalence of some severe CHD. Ultrasound can be performed to all pregnant women, with 15% to 21% reduction in the prevalence of CHD (depending on the severity of the condition, the sensitivity and the frequency of pregnancy termination).

Along with the better diagnoses made by pediatric ultrasound and obstetric ultrasound, it may be expected that fetal echocardiography will have a substantial impact on the future epidemiology of the newborn with CHD. (18)

For the last 10 years, the HMIRS holds a multidisciplinary team of professionals to solve the demand of high-risk pregnancies with prenatal diagnosis of congenital defects and thus the information obtained might be underestimated.

We consider that our study is important because it has the largest population studied in our setting, performed in one of the public maternity hospitals with the highest number of deliveries per year. The information here provided might collaborate with the PNCC to establish the distribution of the necessary resources in order to optimize the results in CHD care more accurately.

We believe it necessary to keep on strengthening the early and prenatal diagnoses of CHD in our country.

Conflict of interest
None declared.

See authors’ conflicts of interest forms in the web / Supplementary Material.

REFERENCES