Epidemiological Characteristics of Congenital Heart Diseases in Londrina, Paraná South Brazil

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Objective – *To determine the prevalence and other epidemiological characteristics of congenital heart diseases.*

Methods – *A retrospective population based study of children who were born in Londrina, from January '89 to December '98 (80,262 live births). Diagnoses were confirmed through autopsy, surgery, catheterization, or echocardiography.*

Results – *A total of 441 patients was as certained what corresponds to a prevalence of 5.494:1,000 live births. Ventricular septal defect was the commonest lesion. A small number of transpositions of the great vessels and of left ventricular hypoplasia was observed. A high propation of ventricular septal defect (28.3%) and atrioventricular septal defects (8.1%) occurred. Fifty-one (11.35%) affected children had syndromic diseases and 52 (12.01%) children had nonsyndromic anomalies.*

Conclusion – *The prevalence of congenital heart diseases in Londrina is in accordance with that of other regions of the globe. This prevalence also may reflect the reality in the southern region of Brazil, because population characteristics are very similar in the 3 southernmost Brazilian states.*

Keywords: congenital heart diseases, epidemiology, Brazil.

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"O Solomon! What wisdom is needed for that physician who deals with a child and a "pre-existing condition."
Ron Louie, MD

Based on the pioneering studies by Abbott in the '20s, a large number of articles about the prevalence of congenital heart diseases in North America and Europe have already been published. In 1967, Caddell [1] pointed out that the actual incidence and distribution of congenital heart diseases in tropical and subtropical regions were not known. The prevalence of congenital heart diseases in Latin America remains unknown, because no information has been published about this population (source: MEDLINE, LILACS).

Londrina is the second most important city of Paraná State and the third greatest city in the southern region of the country. Its population is 426,607 inhabitants, 90% of whom live in the urban area. The major causes of death in the first year of life are perinatal diseases (47%), congenital anomalies (21%), and respiratory and infectious diseases (18%). Infant mortality is 14.48:1,000 live births, which, even though high, is far below the Brazilian rate, which is 40.0:1,000 live births (Brazilian Demographic Census, 1996).

Approximately 3 out 4 inhabitants of Londrina live in the peripheral region of the city, in extreme poverty or in lower middle class conditions. In regard to the more indigent population, which lives mainly in the rural area, several factors account for the few visits that occur to the pediatric services, where the precocious detection of cardiovascular anomalies may cause a significant difference in the survival of children with lesions that may be treated, but that are lethal if not diagnosed and treated in time [2].

Methods

This study comprised 441 children born in Londrina (rural and urban areas) from January '89 to December '98. The precise determination of the live-born population was performed through official demographic data (*Fundação Instituto Brasileiro de Geografia e Estatística* – Brazilian Institute of Geography and Statistics).

Data collected included the following: 1) all children with congenital heart disease born in public hospitals or in the hospital network accredited to SUS (*Sistema Único de Saúde* – Integrated Health System); 2) children suspected of having congenital heart disease, referred from the basic health units to the pediatric cardiology ambulatory service. All diagnoses were confirmed, corrected, or complemented, by echocardiography, catheterization, surgical procedures, or autopsy. The cases of complete atrioventricular block were diagnosed by electrocardiography and echocardiography to exclude concomitant structural defects.

Congenital heart disease has been defined as a structural anomaly of the heart or the great vessels, which has a real or potential functional significance. This definition differs from the classical definition by Mitchell et al [3] that includes only the gross malformations. Children with persistence of ductus arteriosus were only admitted if older than 10 days (normal weight at birth) or if older than 3 months when gestational age below 37 weeks. The cases of pulmonary atresia with ventricular septal defect were classified as tetralogy of Fallot.

As a considerable number of patients had more than one heart defect, a hierarchic system of classification was adopted, which allowed the inclusion of each patient in only one diagnostic category [4].

Those patients suspected of having genetic or chromosomal disorders underwent a genetic study, except those with a conclusive phenotype. In those cases with concomitant syndrome and nonsyndromic extracardiac anomaly, the most severe disease prevailed for effect of classification.

Results

Most of the patients had the diagnosis established in the first year of life (Table I).

The calculated prevalence of congenital heart diseases was 5.494:1,000 live births. Lesions distribution and corresponding frequencies are shown in Table II.

Seventy (17%) patients had 2 or more heart defects, which were classified according to an order of hierarchical importance, which takes the severity of the lesions into consideration. The defects most commonly found in association with other defects were ventricular septal defect, pulmonary stenosis, and atrioventricular canal.

Table I – Distribution of the patients according to age

Age	n	%
<1 month	81	18
1 – 3 months	102	23
4 – 6 months	69	16
7 – 12 months	97	22
1 year	55	12
2 – 4 years	24	5
5 – 9 years	13	3
Total	441	100

Table II – Diagnostic frequencies according to the hierarchical classification

Diagnoses	n	%	n:1000
Heterotaxia	15	3.4	0.186
Single ventricle	12	2.7	0.149
Left ventricular hypoplasia	8	1.8	0.099
Tricuspid atresia	7	1.6	0.087
Truncus arteriosus	2	0.5	0.024
Right ventricular double inlet	5	1.1	0.062
D-Transposition of the great vessels	8	1.8	0.099
L-Transposition of the great vessels	3	0.7	0.037
Atrioventricular canal	36	8.1	0.448
Total anomalous drainage of the pulmonary veins	6	1.4	0.074
Pulmonary atresia	5	1.1	0.062
Tetralogy of Fallot	33	7.5	0.411
Coarctation of the aorta	17	3.8	0.211
Ventricular septal defect	125	28.3	1.557
Aortic stenosis	19	4.3	0.236
Pulmonary stenosis	41	9.3	0.510
Atrial septal defect	34	7,7	0.423
Persistence of ductus arteriosus	26	5.9	0.323
Others (*)	39	8.8	0.485
Total	441	100.0	5.494

(*) Bicuspid aortic valve: 15; 3.4; 0.186 - Mitral valve prolapse with regurgitation: 6; 1.4; 0.074 – Idiopathic dilation of the pulmonary artery: 3; 0.7; 0.037 – Ebstein's anomaly: 3; 0.7; 0.037 – Complete atrioventricular block: 2; 0.5; 0.024 – Mitral valve prolapse associated with dilation of the aortic root: 2; 0.5; 0.024 (right pulmonary artery agenesis, right ventricle isolated anomalous band, pulmonary valve agenesis, double aortic arch, parachute mitral valve, congenital mitral regurgitation, aortic-pulmonary window, right ventricular hypoplasia: 1; 0.25; 0.012 for each anomaly).

Under the diagnosis of heterotaxia, 16 underlying defects were detected, which if considered within their own anatomical categories, some mild alterations in number, percentage and prevalence would have been observed (single ventricle: 15, 3.4%, 0.186; tricuspid atresia: 8, 1.8%, 0.099; truncus arteriosus: 3, 0.7%, 0.037; D-transposition of the great vessels: 10, 2.26%, 0.124; L-transposition of the great vessels: 4, 0.9%, 0.049; atrioventricular canal: 39, 8.8%, 0.485; pulmonary atresia: 8, 1.8%, 0.099; tetralogy of Fallot: 34, 7.7%, 0.423; ventricular septal defect: 126, 28.5%, 1.569).

Noncardiac anomalies were detected in 104 patients (24%): syndromic diseases in 51 (Table III) and nonsyndromic anomalies in 53 patients (Table IV).

The tendency to not perform palliative surgeries but to perform corrective ones preceded the onset of this study. Nevertheless, 52 (11.8%) patients underwent such procedures, because of the high risk that corrective surgeries represented to them at the time of the diagnosis. Percutaneous procedures were performed in 29 (6.6%) patients and corrective surgeries in 154 (34.9%) patients. The total mortality rate was 10% (deaths after palliative procedures: 7; deaths after percutaneous procedures: 2; deaths after corrective surgeries: 14).

Fourteen defects evolved to spontaneous resolution, all confirmed by echocardiography (11 ventricular septal defects and 3 atrial septal defects).

Out of the 441 children studied, 346 (78%) were completely followed up during the study period. In 35 cases,

Table III – Association between syndromic disorders and diagnostic categories

Syndrome	n	%	Diagnostic category	n	%
Trisomy 21	28	55	Atrioventricular canal	17	33
			Ventricular septal defect	9	18
			Persistence of ductus arteriosus	2	4
Congenital rubella	10	20	Ventricular septal defect	1	2
			Pulmonary stenosis	3	6
			Atrial septal defect	1	2
			Persistence of ductus arteriosus	3	6
			Others	2	4
Marfan	2	4	MVP + dilation of the aortic root	2	4
Trisomy13	1	2	Atrioventricular canal	1	2
Beckwith-Wiedeman	1	2	Pulmonary stenosis	1	2
Pierre Robin	1	2	Tetralogy of Fallot	1	2
Cornelia de Lange	1	2	Ventricular septal defect	1	2
Holt-Oram	1	2	Atrial septal defect	1	2
Coffin-Lowry	1	2	Atrial septal defect	1	2
Not defined	2	4	Aortic stenosis	2	4
CHARGE association	2	4	Tricuspid atresia	1	2
			Ventricular septal defect	1	2
VACTERL association	1	2	Single ventricle	1	2
Total	51	100		51	100

MVP- mitral valve prolapse.

Table IV - Extracardiac anomalies and diagnostic categories

Diagnostic category	Extracardiac anomalies				
	n	%	mild	moderate	severe
Heterotaxia	-	-	1	1	2
Single ventricle	-	-	1	1	2
Tricuspid atresia	1	1	-	2	4
RV double outflow tract	-	1	1	2	4
Transposition of the great vessels	1	3	-	4	8
Atrioventricular canal	1	4	2	7	13
Pulmonary atresia	2	-	-	2	4
Tetralogy of Fallot	3	1	1	5	9
Coarctation of the aorta	-	1	-	1	2
Ventricular septal defect	6	8	2	16	30
Aortic stenosis	1	-	-	1	2
Pulmonary stenosis	1	1	-	2	4
Atrial septal defect	1	1	-	2	4
Others	5	2	-	7	13
Total	21	23	9	53	100

RV – right ventricle

contact with the respective families was not possible. Twenty-six children withdrew during he follow-up because of psychosocial or cultural factors or both of these. Of those children not returning for follow-up, those who underwent corrective surgeries predominated. Therefore, late surgical mortality could not be determined.

Discussion

Not all congenital heart defects are evident at early childhood. Therefore, studies of prevalence in children above one year of age may obviously detect a greater number of cases. On the other hand, the set of heart diseases seen in admission units of any large tertiary hospital may not represent the population as a whole.

Recognition and diagnosis of congenital heart diseases may occur among children who were born or who lived, or both, in modern urban centers or close to them, where a special interest in the problem usually exists. In addition, these centers depend on resources necessary for complete diagnosis and appropriate treatment. The epidemiological study that most probably provides reliable results about the real incidence of congenital heart diseases is the one that comprises a great number of births and in which the incidence of heart diseases is determined based on official population data [5].

In the last 40 years, several studies about the estimated index of congenital heart diseases in population groups were carried out, as were studies about the distribution of specific lesions. During this long period, significant diagnostic and therapeutical developments occurred. Echocardiography brought a notable improvement in diagnostic ac-

curacy, which previously was only obtained through invasive methods. Due to their own experience, Tubman et al [6] concluded that Doppler echocardiography should be considered the gold standard for detecting congenital heart diseases.

The relation between the number of children with congenital heart disease and that of the live-born infants was 5.494:1,000. The mean obtained based on 8 significant studies performed in the last 2 decades in different regions with diverse ethnicities was 6.450:1,000 (Table V).

If this mean could be applied to the reality of Londrina, 77 cases of congenital heart disease would not have been detected among the 80,262 live-born infants in the period studied. Such increase would correspond to 0.956:1,000 live-born infants. Considering that 20% of the children with congenital heart disease die in the first year of life and that 5% of the remaining die every year in the 4 subsequent years [15], we may determine that at least 53 children with unrecognized congenital heart diseases would be living in the community. The studies shown in Table V were selected after the echocardiographic assessment had been included as an essential criterion for the diagnosis of congenital heart diseases. The indices obtained, however, varied from 4.50 to 8.80 per 1,000 live-born infants. Methodological differences, mainly the comprehensiveness of the diagnostic approach and the strictness in the criteria of case admission, may explain these significant discrepancies.

Ferencz et al [10] have admitted that very restrictive criteria may constitute biases when evaluating the real prevalence of congenital heart diseases. Hoffman and Christianson [16] and also Lorenzo et al [17] have defended a similar concept based on a study carried out in Spain in a population of 38,674 students, with ages ranging from 4 to 12 years, all of them without a previous diagnosis of congenital heart disease. Those authors have detected an incidence of congenital heart disease of 2.3:1,000 and have cited several studies carried out in other regions with similar results.

Our study comprised all children with congenital heart disease born in Londrina from 1989 to 1998. The adoption of this criterion allowed the inclusion in the study of 92 (21%) patients older than one year. If the admission criterion had been more strict, considering only the children under one year of age, the prevalence would have been reduced to 4.348:1,000 live-born infants, which is very similar to the mean value of 4.185:1,000 obtained considering data from 3 of the most important studies carried out in North America, which adopted this criterion [4,10,11]. Therefore, our methodology is in accordance with the one that Ferencz et al [10] pointed out in the Baltimore-Washington Infant Study, which states that the systematic assessment of cohort studies is of great value for establishing the prevalence of congenital heart diseases using an admission criterion that includes children at school age. On the other hand, if only the congenital heart diseases diagnosed through invasive methods in children in the first year of life (222 cases) had been considered in our case series, the prevalence would have been 2.765:1,000, which is very close to the index reported in the Report of the New England Infant Cardiac Program [4].

Table V - Prevalence (n: 1.000 live births) in different regions in the last 20 years

Authors			n:1000
Abu-Harb M, et al. [7]	England	1985-1990	4.70
Alexis C, Ishmael RG [8]	Barbados	1988-1990	5.50
Bower C, Ramsay JM [9]	Australia	1984-1993	7.65
Ferencz C, et al. [10]	United States	1981-1984	4.50
Grabitz RG, et al. [11]	Canada	1981-1984	5.54
Greh V [12]	Malta	1990-1994	8.80
Klimentova T, et al. [13]	Slovakia	1981-1987	7.81
Schoetzan A, et al. [14]	Germany	1984-1991	7.13
Mean			6.45

In our study, the frequencies obtained for transposition of the great vessels and left ventricular hypoplasia were significantly smaller than the ones expected, probably because of not diagnosing all cases. If the actual prevalence of these defects was equal to that obtained in other studies, 8 cases of transposition of the great vessels and 7 of left ventricular hypoplasia would not have been detected. Adopting this hypothesis, it would be possible to admit that such patients, considering the severity of the lesions, would have died a few hours or days after birth, before the diagnosis could be made. Abu-Harb et al [7] have shown that the diagnosis of congenital heart disease may pass unnoticed in 30% of infants during the first weeks of life. According to data gathered by these authors, about 200 children die annually in Great Britain due to unrecognized congenital heart disease. They conclude that the occurrence of these cases may be even greater in countries where access to more specialized services is more difficult. When the deaths resulting from unrecognized congenital heart diseases are not considered, a low prevalence of complex heart diseases is obtained, which masks the actual scenario. Therefore, it is necessary to assure a more efficient diagnostic approach through training programs. These programs, according to the same authors, should involve neonatal and primary health care staff, aiming to increase the recognition of congenital heart diseases in newborn and small infants, making the early treatment and reduction of infant mortality possible. The current trend, however, is directed toward intrauterine diagnosis using fetal echocardiography. This method, however, is still unavailable to the population of most of our urban centers, because of its costs and the lack of skilled technicians.

Congenital cardiovascular malformations are frequently associated with other anomalies. According to Goodship et al [18], the chromosomal defect that most commonly accompanies congenital heart disease is the 21trisomy.

Mitchell et al [3] have found in their series that 30.1% of all children with congenital heart disease also had extracardiac malformations, with a high mortality rate, calling attention to Down's syndrome that was present in 14% of their case series. Similar data have been reported by other authors [4,6,9,19].

In our study, except for 3 patients (2 with the Marfan's syndrome and 1 with the Coffin-Lowry syndrome), the extracardiac anomalies were detected in the first year of life.

The most frequent condition was the trisomy 21, which was detected in 28 patients, corresponding to 54.9% of the syndromic patients and to 8.02% of all patients with congenital heart disease. The second most frequent syndrome was congenital rubella, affecting 10 patients. The vaccine against rubella has been available in the public health system of Paraná state since 1996, but it was only in 1998 that the first state program of vaccination for women in childbearing age was carried out.

Nonsyndromic extracardiac anomalies, similar to the criterion used by Fyler [4], were arbitrarily classified as follows: a) severe – those representing a significant threat to life and that can only be treated in a supportive way; b) moderate – responsible for a variable degree of physical incapacity and that can be treated in a partial corrective way; c) mild – with a small repercussion in the well-being of the patients, without the risk of death and that can be completely corrected. The combination of any extracardiac anomaly with congenital heart disease, however, may interfere in a significant way in the prognosis of both diseases and, therefore, the adopted classification has only a didactic merit.

Spontaneous occlusion of some congenital defects has already been reported [4, 20].

In our case series, the spontaneous occlusion of the defect occurred in 11 patients with ventricular septal defect and in 3 with atrial septal defect. No patient with persistence of ductus arteriosus had a spontaneous occlusion of the defect, and, therefore, all patients with that type of defect were systematically referred for surgical repair. As our study has a longitudinal characteristic and the follow-up of patients with atrial or ventricular septal defect is mainly clinical most of the time, we expect to detect the occurrence of other spontaneous occlusions in the following years.

Special attention was directed at those patients not returning to follow-up. Many patients belonging to the most needy layers of the population come from families living on the periphery of the city and working in the rural area. These families move from one place to the other according to the availability of work. Most of them have a low level of education, lacking appropriate understanding of the real condition of the patient, as Allen et al [2] have already reported.

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