Prenat Cardio 2021; 11(1): 13–17
DOI: https://doi.org/10.5114/pcard.2021.108431
Received: 6.03.2021
Accepted: 13.07.2021

Prenatal Cardiology
www.termedia.pl/Journal/Prenatal_Cardiology

Research paper

Epidemiological profile of paediatric outpatients followed in a tertiary paediatric cardiology centre in Brazil

Carlos César Assef[1], Eliane Lucas[1,2], Fernanda Maria Correia Ferreira Lemos[1], Diogo Pinotti[1], Rafael Pimentel Correia[1], Nathalie Jeanne Magioli Bravo-Valenzuela[3], Livia Maria de Andrade Sacramento[1], Larissa Vieira da Conceição[1], Maurício Amir de Azevedo[1], Edward Araujo Júnior[4]

[1]Service of Paediatric Cardiology, Bonsucesso Hospital, Rio de Janeiro, Brazil
[2]Discipline of Paediatrics, Faculty of Medicine of Teresópolis (UNIFESO), Teresopólis, Brazil
[3]Discipline of Paediatrics (Paediatric Cardiology), Faculty of Medicine, Federal University of Rio de Janeiro (UFRJ), Rio de Janeiro, Brazil
[4]Department of Obstetrics, Paulista School of Medicine, Federal University of São Paulo (EPM-UNIFESP), São Paulo, Brazil

Abstract

Introduction: Our aim was to describe the epidemiological profile of patients with congenital heart disease (CHD) followed up in the paediatric cardiology outpatient clinic at a tertiary hospital in the city of Rio de Janeiro (RJ), Brazil.
Material and methods: Data were collected from 4050 medical records from April 1987 to January 2020, and the prevalence of CHD was calculated based on the type and sex in this cohort. The data were expressed as percentages. Patients aged > 18 years at the time of initial diagnosis and those with acquired heart disease were excluded.
Results: Male sex comprised approximately 58% of the total CHD carriers. Among the left-right shunt CHDs, ventricular septal defect was the most prevalent (33%), followed by atrial septal defect (18%), and then patent ductus arteriosus (10.5%). Among the cyanogenic CHDs, tetralogy of Fallot was the most frequent (approximately 5% of all CHDs).
Conclusions: The study showed the prevalence of the main CHD in a tertiary referral hospital. We emphasize the importance of knowledge of the epidemiological profile of CHDs in the training of professionals involved in the diagnosis and appropriate treatment of these anomalies.

Key words: epidemiological profile, congenital heart disease, prevalence, tertiary service.

Corresponding author:
Prof. Edward Araujo Júnior, PhD
Rua Belchior de Azevedo, 156 apto. 111 Torre Vitoria
São Paulo-SP, Brazil
CEP 05089-030
tel./fax: +55-11-37965944
e-mail: araujojred@terra.com.br

Introduction

Congenital heart diseases (CHD) are the most common congenital malformations, and the literature shows that their incidence ranges from 0.5% to 2% of live births, depending on the population studied [1-3]. In premature babies, CHD can be twice as frequent [4]. Environmental factors such as exposure to teratogenic agents (e.g. alcohol and air pollutants), maternal diseases, and genetic disorders are related to CHD [5-10]. Some authors have proposed the use of folates, even in association with inositol, to prevent CHD due to the possible epigenetic effect acting on Wnt signalling and, therefore, on cell differentiation of the embryo [11, 12].

The World Health Organization (WHO) estimates that there are currently approximately 130 million children with CHD worldwide [13]. In Brazil, the Brazilian Institute of Geography and Statistics (IBGE) from 2010 showed approximately 3.2 million live births per year, with 1 in 100 presenting with CHD, amounting to about 32,000 new cases of CHD per year [14, 15]. This incidence rate only highlights the importance of knowledge of the epidemiological profile and early diagnosis of

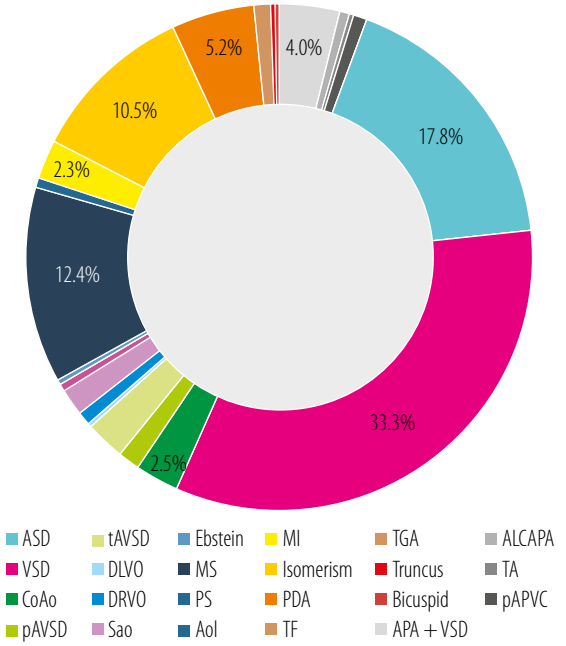

Figure 1. Distribution of types of congenital heart diseases in numbers and percentages of the cases

Abbreviations – see Table 1.

CHD for its therapeutic management. Technological advances in recent decades have allowed the prenatal diagnosis of most CHDs through fetal echocardiography, with a reduction of morbidity and infant mortality.

A major dilemma for the Brazilian public health system is how to provide adequate care for CHD patients. Therefore, this study aims to describe the epidemiological profile of CHDs in a tertiary paediatric cardiology service in the city of Rio de Janeiro (RJ), Brazil.

Material and methods

This was a retrospective cross-sectional study in which 4050 medical records, from April 1987 to January 2020, of patients with CHD at the Paediatric Cardiology Outpatient Clinic of Hospital Bonsucesso, Rio de Janeiro (RJ), Brazil were analysed.

Variables such as type of CHD and gender were obtained through a review of these medical records, and descriptive statistics was used for the analysis. Cases identified in the nursery or neonatal intensive care unit (ICU) were excluded, such as those with patent ductus arteriosus and critical duct-dependent pathologies such as hypoplastic left heart syndrome and pulmonary atresia with intact interventricular septum. Patent ductus arteriosus in children under 3 months of life and patent foramen ovale were not included in the statistical analysis. Patients aged > 18 years at the time of initial diagnosis and with acquired heart diseases such as rheumatic fever, Kawasaki disease, and cardiomyopathies were also excluded.

Data were tabulated in an Excel 2010 spreadsheet (Microsoft Corp., Redmond, WA, USA) and analysed using descriptive statistics as mean and standard deviation (SD), as well as percentages.

Results

Of the 4050 medical records reviewed, 4013 were included in the final statistical analysis with a male dominance (58%). The most prevalent CHDs (33%) were ventricular septal defects, followed by atrial septal defects (18%), and then patent ductus arteriosus (10.5%). Among the cyanotic CHDs, tetralogy of Fallot was the most frequent (about 5%), followed by transposition of the great arteries (1.3%), tricuspid atresia (0.7%), Ebstein's anomaly (0.6%), and pulmonary atresia with ventricular septal defect (0.6%). Rare CHDs occurred in the following proportions: truncus arteriosus, 10 cases (0.24%); congenitally corrected transposition of great arteries, 7 cases (0.09%); and atrial isomerism and total anomalous drainage of pulmonary veins, 3 cases each (0.07%). The very rare CHD such as ectopia cordis and conjoined twins with single heart were identified in only one case each. Figures 1 and 2 illustrate the epidemiological profile of the types of CHD in this cohort of patients.

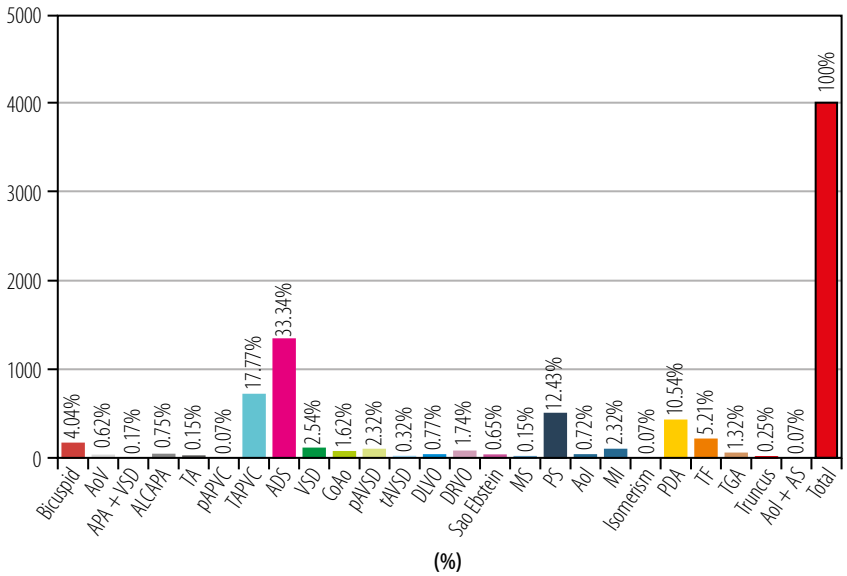

Figure 2. Distribution of the main types of congenital heart disease in the sample

Abbreviations – see Table 1.

Table 1 shows the distribution of patients according to the different types of CHD.

Discussion

Several studies have shown that the prevalence of CHD has been increasing globally in recent years. Left-right shunt CHD has always been the most frequent among all types of cardiac malformations and is among the most prevalent, especially since 2010 [3, 16-18]. Antagonistically, CHD with obstruction to systemic flow has been showing a global reduction in prevalence since 1995, probably due to the increase of prenatal diagnosis and possibility of pregnancy termination in several countries [3, 18]. However, the prevalence of different types of CHD may vary regionally.

Recently, a meta-analysis that included 160 studies on the prevalence of CHD between 1970 and 2017 observed that the most prevalent CHDs were as follows: ventricular septal defect, atrial septal defect, and patent ductus arteriosus (57.9%). In this study, Liu et al. [3] concluded that there was a 10% increase every 5 years in the prevalence of CHD and interestingly a 90% increase of less complex CHDs such as ventricular septal defect, atrial septal defect, and patent ductus arteriosus. Among these 3 CHDs, atrial septal defect was the CHD with the highest increase in prevalence between 2010 and 2017. In our study, conducted between 1987 and 2020, the 3 most prevalent CHDs were the same as those found in the study of Liu et al. [3] and with similar prevalence (61.5% vs. 57.9%). Similarly, we were not able to identify in detail the various types of VSD because the cases were catalogued by the international classification of diseases.

In a South African study conducted by Namuyonga et al. [18], interventricular communication, interatrial communication, and canal persistence were also the most common types of CHD. Similarly to our study, interventricular communication was the most prevalent (33% vs. 27.2%); however, in the study of Namuyonga et al. [18] it was possible to differentiate the types of interventricular communication, with perimembranous being the most frequent (76%). However, in the study by Namuyonga et al. the persistence of ductus arteriosus was much less frequent than in our series (10.5% vs. 22%), perhaps due to the deficit in the vaccination program for rubella and the higher prevalence of prematurity in Uganda. Another important factor is that our study did not include patent ductus arteriosus during the first 3 months of life.

As previously mentioned, a reduction has been observed in the prevalence of CHD with left ventricular outflow obstruction [3, 19]. Accordingly, in our study, the prevalence of CHD with obstruction to systemic flow was low. Among left ventricle outflow tract obstruction CHD, aortic coarctation was the most frequent (2.5% of all cases of CHD), followed by aortic stenosis (1.7%). However, the cases of patients seen in our service with obstructive lesions of the left ventricle, especially hypoplastic left heart syndrome, may be underestimated because patients in the neonatal ICU and nursery were excluded, which is a limitation of our study.

Among the cyanogenic CHDs, in our series, tetralogy of Fallot (about 5%) was the most common, similarly to the literature [17, 18, 20, 21]. The CHD of low prevalence was the truncus arteriosus (0.24%) and total anomalous pulmonary vein drainage (0.07%). However, truncus arteriosus presented a higher prevalence in the study of Namuyonga et al. [18] than in the literature in general, perhaps due to the lower age range of their cases.

In our specialized CHD service, ventricular septal defect was found to be the most prevalent type among all CHDs. Moreover, the most frequent CHDs were atrial septal defect and patent ductus arteriosus. Among the cyanogenic CHDs, the most common was the tetralogy of Fallot. The prevalence

Table 1. Distribution of the patients regarding the different types of congenital heart disease

Type of CHD	Number of the cases	Percentage of the cases (%)
Bicuspid AoV	162	4.04
PA + VSD	25	0.62
ALCAPA	7	0.17
TA	30	0.75
pATPVC	6	0.15
TAPVC	3	0.07
ASD	713	17.77
VSD	1338	33.34
CoAo	102	2.54
pAVSD	65	1.62
tAVSD	93	2.32
DLVOT	13	0.32
DRVOT	31	0.77
AoS	70	1.74
Ebstein	26	0.65
MS	6	0.15
PS	499	12.43
AoI	29	0.72
MI	93	2.32
Isomerism	3	0.07
PDA	423	10.54
TF	209	5.21
TGA	53	1.32
Truncus	10	0.25
AoI + AS	3	0.07
Total	4013	100.00

CHD – congenital heart disease, bicuspid AoV – bicuspid aortic valve, PA + VSD – pulmonary atresia + VSD, ALCAPA – anomalous origin of the left coronary from pulmonary artery, TA – tricuspid atresia, pATPVC – partial anomalous pulmonary veins connection, TAPVC – total anomalous pulmonary veins connection, ASD – atrial septal defect, VSD – ventricular septal defect, CoAo – coarctation of aorta, pAVSD – partial atrioventricular septal defect, tAVSD – total atrioventricular septal defect, DLVOT – left ventricle double outflow tract, DRVOT – right ventricle double outflow tract, AoS – aortic stenosis, Ebstein – Ebstein anomaly, MS – mitral stenosis, PS – pulmonary stenosis, AoI – aortic insufficiency, MI – mitral insufficiency, PDA – patent ductus arteriosus, TF – tetralogy of Fallot, TGA – transposition of great arteries, truncus – truncus arteriosus.

of the types of CHD found in our series is in accordance with several studies published on the global epidemiological profile of CHD and in several regions such as Africa, China, and India [3, 18, 19, 22].

Another study conducted in the Brazilian population by Miyague et al. [23] also showed a profile of the most prevalent CHD very similar to our study, i.e. ventricular septal defect (30%), atrial septal defect (19%), and patent ductus arteriosus (17%). Also, these authors found tetralogy of Fallot to be the most common cyanogenic CHD. Some differences found were the higher percentage of patent ductus arteriosus and higher incidence of coarctation of the aorta and transposition of the great arteries. The inclusion of patients from the neonatal period and the period of the study (1995-1997) is related to the epidemiological profile Miyague's study [23].

Several studies on CHD have demonstrated that they may be associated with genetic diseases, including trisomy 21 and deletion of trisomy 22 [24]. In our series, the association of trisomy 21 with atrioventricular defect was the most frequent. In the study by Namuyonga et al. trisomy 21 was also the most prevalent chromosomal anomaly [18]. These authors also observed an association of genetic syndromes with conotruncal anomalies such as tetralogy of Fallot and truncus arteriosus [18].

Regarding gender, we found a higher prevalence of CHD in males, which disagrees with most studies that show female dominance or similarity in the distribution of CHD in both sexes [18, 23]. The non-inclusion of the neonatal ICU and nursery may be related to our results.

Among the limitations, the cases presented do not correspond to the total number of patients seen in the study period because patients admitted to the neonatal ICU were not included. Difficulties were encountered in obtaining information from older cases, making it difficult to detail some CHD regarding the type of interventricular communication.

In summary, in our series, ventricular septal defect was the most frequent CHD. Among the left-to-right shunt CHDs, atrial septal defect and patent ductus arteriosus were the most prevalent. Regarding the cyanogenic CHD, the most common was the tetralogy of Fallot. We highlight, as an important point of the study, the knowledge of the epidemiological profile of the prevalent CHD in the paediatric population in a tertiary hospital. We emphasize the importance of knowledge of the epidemiological profile of CHD in the training of professionals involved in the diagnosis and appropriate treatment of CHD.

Conflict of interest

The authors declare no conflict of interest.

REFERENCES

1. Calzolari E, Garani G, Cocchi G, Magnani C, Rivieri F, Neville A, et al. Congenital heart defects: 15 years of experience of the Emilia-Romagna Registry. Eur J Epidemiol 2003; 18: 773-780.
2. Bouma BJ, Mulder BJ. Changing landscape of congenital heart disease. Circ Res 2017; 120: 908-922.
3. Liu Y, Chen S, Zühlke L, Black GC, Choy MK, Li N, et al. Global birth prevalence of congenital heart defects 1970-2017: updated systematic review and meta-analysis of 260 studies. Int J Epidemiol 2019; 48: 455-463.
4. Tanner K, Sabrine N, Wren C. Cardiovascular malformations among preterm infants. Pediatrics 2005; 116: e833-e838.
5. Serrano M, Han M, Brinez P, Linask KK. Fetal alcohol syndrome: cardiac birth defects in mice and prevention with folate. Am J Obstet Gynecol 2010; 203: 75.e7-75.e15.
6. Linask KK, Han M, Bravo-Valenzuela NJM. Changes in vitelline and utero-placental hemodynamics: Implications for cardiovascular development. Front Physiol 2014; 5: 390.
7. Caputo C, Wood E, Jabbour L. Impact of fetal alcohol exposure on body systems: A systematic review. Birth Defects Res C Embryo Today 2016; 108: 174-180.
8. Zhang B, Liang S, Zhao J, Qian Z, Bassig BA, Yang R, et al. Maternal exposure to air pollutant PM 2.5 and PM10 during pregnancy and risk of congenital heart defects. J Expo Sci Environ Epidemiol 2016; 26: 422-427.
9. Basu M, Garg V. Maternal hyperglycemia and fetal cardiac development: Clinical impact and underlying mechanisms. Birth Defects Res 2018; 110: 1504-1516.
10. Zaidi S, Brueckner M. Genetics and genomics of congenital heart disease. Circ Res 2017; 120: 923-940.
11. Linask KK, Huhta J. Folate protection from congenital heart defects linked with canonical Wnt signaling and epigenetics. Curr Opin Pediatr 2010; 22: 561-566.
12. Ford SM, Pedersen CJ, Ford MR, Kim JW, Karunamuni GH, McPheeters MT, et al. Folic acid prevents functional and structural heart defects induced by prenatal ethanol exposure. Am J Physiol Heart Circ Physiol 2021; 320: H1313-H1320.
13. World Health Organization. Congenital Anomalies. Available from: https://www.who.int/health-topics/congenital-anomalies#tab=tab_1/. Accessed on 2021-02-28.
14. IBGE. Censo Demográfico. Available from: https://sidra.ibge.gov.br/tabela/3148#resultado. Accessed on 2021-02-28.
15. Hoffman JI, Kaplan S. The incidence of congenital heart disease. J Am Coll Cardiol 2000; 39: 1890-1900.
16. Botto LD. Epidemiology and prevention of congenital heart didease. In: Allen HD, Shaddy RE, Penny DJ, Feltes TF, Cetta F (eds.). Moss & Adams' Heart Disease in Infants, Children and Adolescents, Including the Fetus and Young Adult. 8th ed. Lippincott Willians & Wolters Kluwer. Philadelphia 2016; 577-616.
17. van der Linde D, Konings EE, Slager MA, Witsenburg M, Helbing WA, Takkenberg JJ, et al. Birth prevalence of congenital heart disease worldwide: a systematic review and meta-analysis. J Am Coll Cardiol 2011; 58: 2241-2247.
18. Namuyonga J, Lubega S, Aliku T, Omagino J, Sable C, Lwabi P. Pattern of congenital heart disease among children presenting to the Uganda Heart Institute, Mulago Hospital: a 7-year review. Afr Health Sci 2020; 20: 745-752.
19. Qu Y, Liu X, Zhuang J, Chen G, Mai J, Guo X, et al. Incidence of congenital heart disease: the 9-year experience of the Guangdong Registry of Congenital Heart Disease, China. PLoS One 2016; 11: e0159257.
20. Parker SE, Mai CT, Canfield MA, Rickard R, Wang Y, Meyer RE, et al. Updated national birth prevalence estimates for selected birth defects in the United States, 2004-2006. Birth Defects Res A Clin Mol Teratol 2010; 88: 1008-1016.
21. Smitha R, Karat SC, Narayanappa D, Krishnamurthy B, Prasanth SN, Ramachandra NB. Prevalence of congenital heart diseases in Mysore. Indian J Human Genet 2016; 12: 11-16.
22. Naik S, Irshad M, Kachroo A, Ahmad M. A study of prevalence and pattern of congenital heart disease at Sopore, Kashmir, North India. Int J Contemp Pediatr 2019; 6: 275-279.

23. Miyague NI, Cardoso SM, Meyer F, Ultramari FT, Araújo FH, Rozkowisk I, et al. Epidemiological study of congenital heart defects in children and adolescents: analysis of 4,538 cases. Arq Bras Cardiol 2003; 80: 274-278.
24. Correia P. Genetics and congenital heart disease. In: Araujo Júnior E, Bravo-Valenzuela NJ, Peixoto AB (eds.). Pediatric Cardiology part 1. 1st ed. Bentham Science Publishers. Singapore 2020; 459-477.

Division of work:
Carlos César Assef (ORCID: 0000-0001-6661-1494): collection and assembly of data
Eliane Lucas (ORCID: 0000-0002-5945-8660): research concept and design
Fernanda Maria Lemos (ORCID: 0000-0001-5583-2363): collection and assembly of data, data analysis and interpretation
Diogo Pinotti (ORCID: 0000-0002-8398-2411): data analysis and interpretation
Rafael Pimentel Correia (ORCID: 0000-0002-9597-3563): collection and assembly of data, data analysis and interpretation
Nathalie Jeanne Bravo-Valenzuela (ORCID: 0000-0003-1491-4877): writing the article
Livia Maria Sacramento (ORCID: 0000-0002-3644-2906): critical revision of the article
Larissa Vieira da Conceição (ORCID: 0000-0003-0220-099X): critical revision of the article
Maurício Amir Azevedo (ORCID: 0000-0003-2324-2427): critical revision of the article
Edward Araujo Júnior (ORCID: 0000-0002-6145-2532): critical revision of the article