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

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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.
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.

Material and methods

This was a retrospective cross-sectional study in which 4050 medical records, from April 1987 to January 2020, of patients...
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 cataloged 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 ducus 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**

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

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

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