

Congenital Heart Diseases in the Newborn: from the Pediatrician's Request to the Cardiologist's Evaluation

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Summary

Objective: To analyze the importance of symptoms as a reason for referral to pediatric cardiologists in the diagnosis of congenital heart diseases (CHD) in the newborn (NB).

Methods: Prospective study on live NB referred for cardiac evaluation, with performance of electrocardiogram, chest radiography and echocardiography. Cardiology consultation was requested by means of a multiple-choice form including signs and symptoms suggestive of CHD. Patent ductus arteriosus (PDA) without clinical and/or hemodynamic consequences was not considered a heart disease.

Results: From 1999 to 2002, 358 out of 3716 NB were studied, and 49 cases of CHD and 128 of PDA were found. The prevalence of CHD was 13.2:1000 NB. The main reason for referral to the cardiologist was heart murmur in 256 (72%) NB, of which 39 (15%) had CHD, and in 91% of the 128 cases of PDA. In 14 (4%) NB, the reason for referral was cyanosis, and eight of these patients (57%) had a CHD. Heart failure was the reason for referral in 37 (10%) NB, of whom 17 (46%) had CHD. Arrhythmia, associated congenital malformations, or chromosome disorders were the reasons for referral in 14% of the cases.

Conclusion: The main reason for referral was detection of a heart murmur on cardiac auscultation. Although cyanosis and heart failure were uncommon reasons for referral, their presence indicated a high probability of the diagnosis of heart disease. Pediatric screening plays a key role in this diagnosis. (Arq Bras Cardiol 2007; 89(1) : 6-10)

Key words: Congenital heart diseases, echocardiography, newborn.

Introduction

Congenital heart defects have varying presentations, from defects that progress asymptotically to those with significant symptoms and high mortality rates. Prevalence analyses show different, sometimes very conflicting results depending on the age of the population studied. Conflicting results also result from factors such as the inclusion of defects that may go completely unnoticed on physical examination like the presence of a bicuspid aortic valve, or from the classification of ductus arteriosus sometimes as a defect, sometimes as a persistence that can still be considered physiological.

The objective of this study was to determine the importance of newborns' symptoms in the diagnosis of congenital heart diseases.

Methods

This prospective study started in August 1999, in live births from the university hospital maternity ward. Pediatricians responsible for newborn care were trained and instructed to

request an assessment from pediatric cardiologists whenever a congenital heart disease was suspected, that is, whenever a heart murmur or clinical conditions such as persistent dyspnea or cyanosis were detected. The request for referral was made by filling out a multiple-choice form that included all signs and symptoms that could result from a congenital heart disease.

After chest radiography and electrocardiography were performed, the newborns underwent an echocardiographic study with a commercially available instrument; the result of this study was used as a parameter for the diagnosis of cardiac malformation. The protocol was duly approved by the hospital's Research Ethics Committee. Whenever a heart disease with significant hemodynamic or clinical consequences was detected, the newborns were referred for hemodynamic study and/or heart surgery after the defect was confirmed.

With the purpose of analyzing the importance of pediatricians' referral for cardiac evaluation, physical examination and echocardiographic study were performed by a cardiologist in other 400 newborns without indication for referral and randomly selected.

Results

A total of 3716 live births were recorded in the study period extending from August 1999 to July 2002. Referral was requested and full cardiac evaluation was performed in 358

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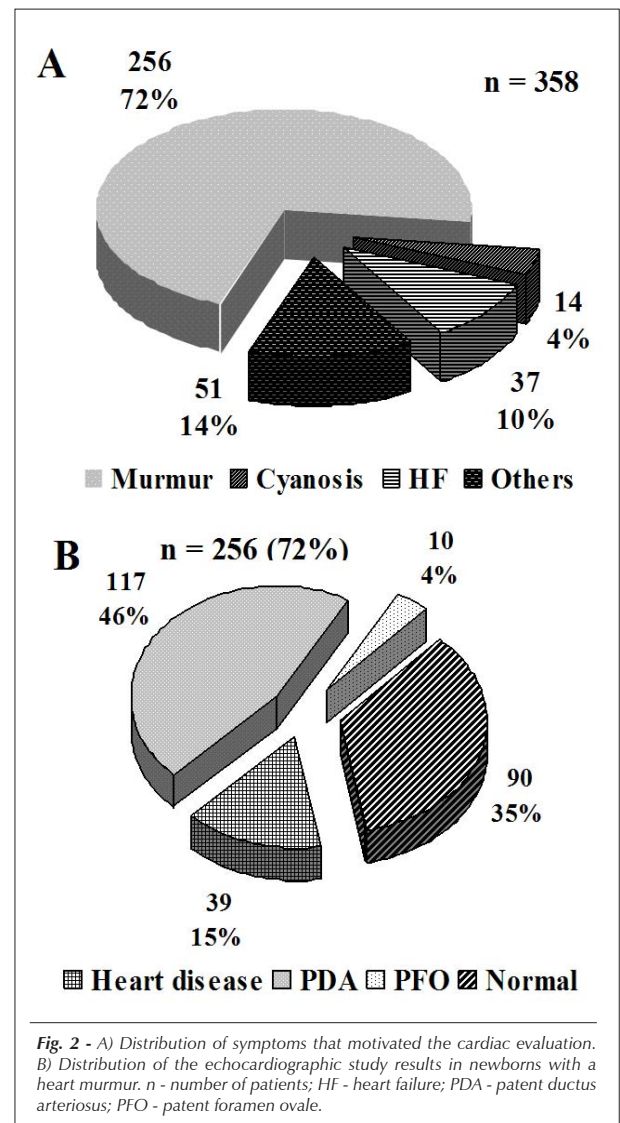
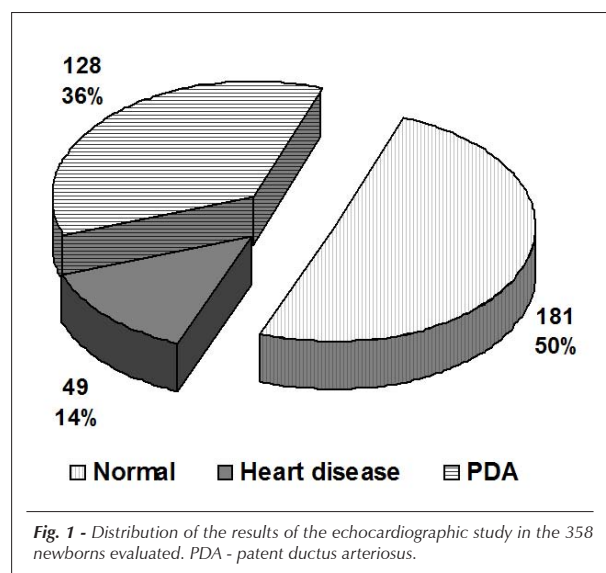
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(9.6%) of them. The echocardiographic study was normal in 181 (50%), diagnosed patent ductus arteriosus in 128 (36%) and congenital heart disease in 49 (14%) (Figure 1). Since the study was conducted in newborns within their first few hours or days of life, and some of them were low-birth weight or premature, patent ductus arteriosus was not considered a malformation when detected alone and without clinical and/or hemodynamic consequences.

During the outpatient follow-up, however, eight (6%) of these infants persisted with the ductus arteriosus and some were awaiting surgical treatment.

The main reason for referral was detection of a heart murmur on cardiac auscultation in 256 out of the 358 newborns evaluated (72%). Of these, 39 (15%) had congenital heart diseases: 19 cases of ventricular septal defect, seven cases of atrial septal defect, three cases of pulmonary stenosis alone, two cases of double inlet right ventricle, four cases of atrioventricular septal defect, three of tetralogy of Fallot, and one case of Taussig-Bing complex (double outlet right ventricle with subpulmonary defect and vessels of the base originating latero-laterally). Detection of a heart murmur on cardiac auscultation was also the main reason for referral in 117 out of 128 (91%) cases of patent ductus arteriosus. In 90 of the 256 (35%) newborns referred because of detection of a heart murmur on auscultation, the echocardiographic study was normal, and in 10 (4%) an intermittent flow through the atrial sept compatible with patent foramen ovale was demonstrated (Figure 2). Persistent cyanosis was the reason for referral in 14 (4%) of the 358 newborns evaluated. Of these, eight (57%) had a congenital heart disease. In six of the eight (75%) cases of heart disease a heart murmur was also present on cardiac auscultation, and in four (50%) there were signs of heart failure (double inlet right ventricle, transposition of the great arteries, and Taussig-Bing complex). In the six cases (43%) not presenting a cardiac malformation, the diagnosis of pulmonary disease was made further along (Figures 2A, 3A). Among the 358 patients evaluated, heart failure was the reason for referral in 37 (10%), 17 (46%) of

whom had congenital heart diseases (Figures 2A, 3B). In 29 of the 37 cases (78%) a heart murmur was also present on auscultation (seven cases of ventricular septal defect, four cases of complete atrioventricular septal defect, two cases of double inlet right ventricle, and one case of Taussig-Bing complex, the three latter ones with associated cyanosis, and 15 with patent ductus arteriosus with hemodynamic consequences) and were, therefore, included in the group in which murmur was the main reason for referral. The cases in which no heart murmur had been detected were: aortic arch interruption, complete transposition of the great arteries, and truncus arteriosus. The 15 cases of patent ductus arteriosus with heart failure accounted for 12% of the cases of ductus arteriosus and 41% of the requests because of heart failure. If congenital heart disease is considered because of the presence of early hemodynamic consequences, the sum with the other 17 cases represents 86% of the cases of heart disease with this type of presentation. In the 10 newborns with no heart diseases, another evaluation was performed, and other



causes such as sepsis and respiratory distress syndrome of the newborn were established for the symptoms.

In 51 (14%) newborns, the reason for referral was suspected arrhythmia, chromosome disorders or the presence of associated congenital malformations; no morphological alteration was found when the reason for referral was arrhythmia. One case of atrioventricular septal defect and four cases of atrial septal defect were recorded among newborns referred because of chromosome disorders, and one case of biventricular hypertrophy was recorded with suspected Noonan Syndrome, thus accounting for 12% of the congenital heart diseases found.

None of the 400 newborns evaluated by the cardiologist without being referred had cyanosis or heart failure, and a very faint heart murmur had been auscultated in eight of them (2%). The echocardiographic study was normal in all these newborns.

The prevalence of congenital heart disease was 13.2:1000 live births. The malformations found were ventricular septal defect (19 cases, 38.8%), atrial septal defect (11 cases, 22.5%), complete atrioventricular septal defect (5 cases, 10.2%), Down Syndrome (one case), pulmonary valve stenosis alone (3 cases, 6.2%), tetralogy of Fallot (3 cases, 6.2%), and double inlet right ventricle (2 cases, 4.1%), followed by type-B aortic

arch interruption, complete transposition of the great arteries, type-IV *truncus arteriosus* or pulmonary atresia with ventricular septal defect, Taussig-Bing complex associated with severe coarctation of the aorta, and pulmonary atresia with intact septum and significant biventricular hypertrophy - one case of each (2.0%), respectively. In four cases diagnosed with ventricular septal defect there was a small associated atrial septal defect, and in one case of atrial septal defect a mild pulmonary valve stenosis was associated (peak systolic gradient of 30 mmHg) (Table 1).

Discussion

According to Mitchell et al's¹ definition, congenital heart disease is a gross structural malformation of the heart or great intrathoracic vessels with a real or potential functional importance. Therefore this definition excludes anomalies such as bicuspid aortic valve without valve dysfunction, mitral valve prolapse, persistent left superior vena cava, anomalous origin of the left subclavian artery, mild valve regurgitation, and functional alterations without a structural component. This definition was adopted in this study, and cases of patent ductus arteriosus, an anomaly that could still be considered functional in the first few hours of life when this study was conducted, were also excluded.

Results from different studies show that the incidence of congenital heart diseases ranges from 4/1000 to 50/1000 live births². This wide variation shows how difficult it is to obtain data both at the moment of study sample selection and when establishing epidemiologic definitions such as

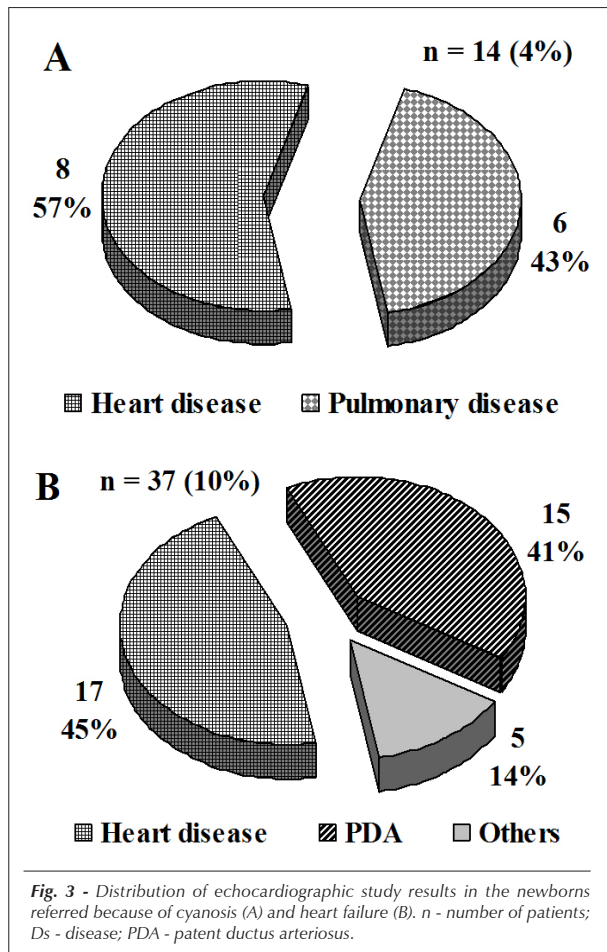


Tabela 1 - Distribution of the frequencies of the congenital heart diseases observed

Malformation	n	Frequency (%)
Ventricular septal defect	19	38.8
Atrial septal defect	11	22.5
Complete atrioventricular septal defect	5	10.2
Pulmonary valve stenosis alone	3	6.2
Tetralogy of Fallot	3	6.2
Single ventricle (double inlet right ventricle)	2	4.1
Type-B aortic arch interruption	1	2.0
Complete transposition of the great arteries	1	2.0
Type-IV <i>truncus arteriosus</i>	1	2.0
Taussig-Bing complex + coarctation of the aorta	1	2.0
Pulmonar atresia	1	2.0
Significant biventricular hypertrophy	1	2.0
Total	49	100

incidence and prevalence³.

The selection of the population in which the study is conducted is also a source of significant differences in epidemiological calculations. Thus, if on one hand 20% of the children with congenital heart diseases are estimated to die within their first year of life⁴, and, therefore, studies after this age could underestimate the real prevalence of congenital heart diseases, on the other hand approximately 30% of congenital heart diseases are believed to be unrecognized in the first few weeks of life⁵, a factor that could also underestimate the real prevalence of this condition in newborns.

The real importance of echocardiographic studies in the diagnosis of congenital heart diseases is unquestionable⁶. Early studies on incidence showed rates of approximately 8:1000 to 9:1000 live births^{1,7,8}, which may have increased mildly after echocardiographic studies became available, and as malformations without evident hemodynamic consequences were recognized.

In Brazil, Guitti⁹ demonstrated a congenital heart disease prevalence of approximately 5.5:1000 live births. In this important study, although only 18% of the patients were younger than one month of age when data were analyzed, frequencies of 44.2% or 74.8% (if cases of ventricular septal defect were included) of malformations were observed that could have been diagnosed in the nursery in the first few weeks of life (excluding the cases of L-transposition of the great arteries, aortic and/or pulmonary valve stenosis, atrial septal defect, and the group classified as "others"). Therefore, further studies on this age range are necessary. Similar results were reported by Miyague et al¹⁰, with a higher frequency of congenital heart disease in neonates and infants.

In the present study, the prevalence of congenital heart disease was of approximately 13.2:1000 live births, which could initially implicate an increase in relation to the studies previously mentioned^{1,7,8}. Several factors may have contributed to this result, but most likely it resulted to a great extent from the training provided to the professionals requesting the referral. In this study, normal results were reported in 90 patients referred to echocardiographic study because of the detection of a heart murmur on auscultation, although the cardiologist had confirmed the presence of a low-grade systolic murmur in all cases. In some of these newborns, flow acceleration in the beginning of the pulmonary branches was observed, more frequently in the left pulmonary artery, which is a common cause of innocent murmur in newborns, especially preterm ones.

Another important factor to be considered is the possibility of diagnosing, in this age range, severe malformations that lead to early death or also defects that could close spontaneously, such as ventricular septal defects, although none of the patients had had this outcome in the subsequent follow-up. According to Hoffman and Kaplan², previous studies may have found incidences of 4:1000 to 5:1000 live births as a result of the almost exclusive diagnosis of more severe malformations, mainly when echocardiographic studies were not available and many cardiologists were reluctant to make patients with mild lesions undergo catheterization: this group included those patients with a murmur suggestive of small ventricular

or atrial septal defects¹¹.

Currently, fetal echocardiography is an important method routinely used for the diagnosis of cardiac malformations. However, the diagnosis of some heart diseases such as small ventricular and atrial septal defects and also patent ductus arteriosus, as well as some cases of coarctation of the aorta may be missed. Several studies show that congenital heart defects are more likely to be diagnosed when echocardiography is requested because of fetal heart failure than because of maternal factors^{2,12-14}.

No cases of heterotaxy were observed in this study, and all cases of ventricular septal defect were referred for echocardiographic study because of the detection of a heart murmur on auscultation, unlike in atrial septal defects, in which only seven (64%) were referred because of this reason. Although some studies use a higher than 5-mm-diameter defect with dilation of the right chambers as a parameter to establish this diagnosis¹⁵, all cases with continuous left atrial to right atrial flow were classified as atrial septal defects. Regarding the presence of heart failure, four out of the five (80%) patients diagnosed with complete atrioventricular septal defect and those with aortic arch interruption, transposition of the great arteries, and truncus arteriosus were referred for this reason. Cyanosis was the reason for referral in all cases of a double-inlet right ventricle, transposition of the great arteries, tetralogy of Fallot, Taussig-Bing complex, and pulmonary atresia with intact septum.

Flow acceleration through the aortic valve was observed in some cases, without exceeding the criterion of 160 cm/s to indicate an evident stenosis¹⁵. Although the criterion of flow velocity higher than 120 cm/s had been used to define pulmonary stenosis, all cases observed and defined as stenosis in this study had gradients higher than 25 mmHg.

One case of significant biventricular hypertrophy with predominance of the right ventricle was included in this study. Even in the lack of a positive family history or associated cardiac malformations, the diagnosis of Noonan Syndrome was later confirmed by genetic evaluation.

Ventricular septal defect is the most frequent defect in most of the studies conducted to date^{2,5,7-9,11}, with mild variations of frequency in the different reports. The frequency of 38.8% observed in the present study is similar to that obtained in some of the previous studies (34.7%⁷, 30.5%¹⁰, 41.6%¹¹ and 39%¹⁴) and different from the 28.5% found by Guitti⁹, probably because of the different age range of the patients studied.

Conclusion

This study demonstrated a high prevalence of congenital heart diseases when data from live births of a general tertiary care hospital were analyzed, which certainly will call attention to the need to implement services for the treatment of these newborns or for the creation of a local referral center for this purpose.

We should point out the importance of the pediatrician in identifying early manifestations of congenital heart diseases, as well as in detecting heart murmurs that could go unnoticed on the physical examination of newborns.

Despite the high prevalence found, the present study

could have excluded malformations that do not lead either to alterations on cardiac auscultation or to symptoms in the first few hours of life. However, after hospital discharge, no infant was reported to have attended follow-up visits because of an unrecognized heart disease in the neonatal period; it should be considered that these newborns, even the healthy ones, have at least one outpatient follow-up visit.

The main reason for referral was the early detection of a heart murmur at the physical examination of newborns. Although infrequent as a reason for referral, the presence of cyanosis and heart failure indicated a high probability of the

diagnosis of a congenital heart disease. Pediatric screening plays a key role in this diagnosis.

Potential Conflict of Interest

No potential conflict of interest relevant to this article was reported.

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