TYPES OF CARDIAC HEART DEFECTS IN INFANTS

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Abstract: *Congenital heart defects (CHD) continue to have a great preponderance in pediatric cardiovascular pathology, and making an accurate diagnosis and early application of the treatment represent constant goals of modern cardiology. The aim of the study is to analyze the clinical profile of children with CHD and to evaluate the use of echocardiographic examination in diagnosing CHD. There were 81 infants with CHD whose data were recorded in a "CHD file". Later, they were recorded in an EXCEL database, used to extract material for the study. These 81 infants represent 47.9% of the children with CHD, hospitalized during the four years of study. In the most dynamic years of the study, the number of newly diagnosed cases was significantly higher compared with other years, through the "active diagnosis", with echocardiographic examination upon every suspicion of CHD. Cardiovascular complications occurred in ⅓ of children before the age of one year old.*

Cuvinte cheie: *malformaţii cardiace congenitale, sugar, ecocardiografie*

Rezumat: *Malformaţiile cardiace congenitale (MCC) deţin ponderea patologiei cardiovasculare pediatrice, iar realizarea cu acurateţe a diagnosticului acestora şi aplicarea precoce şi nuanţată a terapiei reprezintă deziderate constante ale cardiologiei moderne. Scopul studiului a fost analiza profilului clinic al copilului cu MCC la vârsta de adresare de sugar şi evaluarea utilităţii examenului ecocardiografic în diagnosticul MCC. Lotul a cuprins 81 de sugari cu MCC ale căror date au fost notate, într-o „Fişă MCC". Ulterior, acestea au fost înregistrate într-o bază de date în programul EXCEL, de unde au fost extrase aspectele semnificative ale studiului. Cei 81 de sugari reprezintă 47,9% din copiii cu MCC internaţi pe parcursul celor 4 ani de studiu. În anii cei mai dinamici ai studiului, numărul de cazuri nou-diagnosticate a fost net mai mare comparativ cu ceilalţi ani, prin „diagnosticarea activă", cu efectuarea ecocardiografiei în momentul oricărei suspiciuni de MCC. Complicaţiile cardiovasculare au apărut la ⅓ din copii, înaintea vârstei de un an.*

INTRODUCTION

Congenital heart diseases (CHD) are still a challenge for the medical world because of the frequency with which this pathology is common in pediatrics and, also because of the problems faced by the medical practitioner in the management of these cases. These malformations continue to have a high presence in the cardiovascular pathology and represent a major cause of mortality in childhood.

In cardiovascular pathology, there are several reasons why early infancy is the most delicate period in human life: in principle, 70% of CHD cases can be diagnosed before the age of one month and 90% of cases up to one year (1); it is the time when the pediatric cardiologist will decide for each malformation case what type of therapeutic procedure is suitable and which of these cases requires total or partial surgical intervention; some surgeries could lead to transient/irreversible complications; for instance physical growth and psychomotor development of the future adult could be affected due to the fact that physical and cognitive development are fastest during infancy; however, statistics show that one in ten children who die during the first year of life has not been diagnosed with heart disease.(2)

PURPOSE

The purpose of this study is to analyze the clinical profile of children with congenital heart defect in infancy age, depending on the type and severity of the heart defect, highlighting the most frequent clinical features for diagnosis of CHD, their risk of developing complications, the cardiac defects that had undergone interventional procedure or surgical correction, association of other congenital anomalies, and the use of echocardiography in the diagnose of CHD.

METHODS

In this study, 81 infants (30 days old - 12 months old) with CHD, were included, hospitalized in the Pediatric Clinics I and II of the Emergency County Hospital of Craiova, between January 2007 and December 2010.

In this group, neonates with CHD were not included, being the subject of a separate study, due to the particularities found in this period of childhood.

As a doctor, I have done a "CHD file", where I retrospectively and prospectively noted the history, clinical examination, biological and radiological findings, echocardiography, medical/interventional/surgical treatment and case evolution. 58 infants had radiological examination of the chest and 79 infants had echocardiographic examination.

The registration of the subjects' data in the Excel program has led to the original database, from which the material of this study was extracted.

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RESULTS

The study group is part of a larger study that included 169 children (0 -16 years old) diagnosed with CHD, hospitalized in the mentioned period. The 81 infants represent 47.9% of the children with CHD hospitalized during the four years of study and besides the group of infants, they represent 61.99% (figure no. 1). Area of origin: 34 infants (41.98%) came from the rural area and 47 infants (58.02%) from the urban area. The gender distribution: there was a slight difference in favour of males, 51 of the children (62.96%) were male and 30 (37.04%) were females. 8 infants had family history of CHD (9.88%). In 18 cases (22.22%), there were associated malformations of other organs or systems, malformations that were not diagnosed as a syndrome, and 10 cases (12.22%) had genetic syndromes: 7 cases with Down syndrome (70%), 1 case with Proteus syndrome, 1 case with Potter syndrome and 1 case with Werdnig Hoffmann syndrome (figure no. 2). Of seven infants with Down syndrome only two were diagnosed with atrioventricular canal (CAV), and the rest of them with atrial septal defect (ASD) and/ ventricular septal defect (VSD). Of these 28 infants (34.56%) with multiple malformation syndromes, only 4 had a family history of CHD, and only one had surgical correction of the cardiac defect.

Figure no. 1. Distribution of cases according to the year of diagnosing the patients

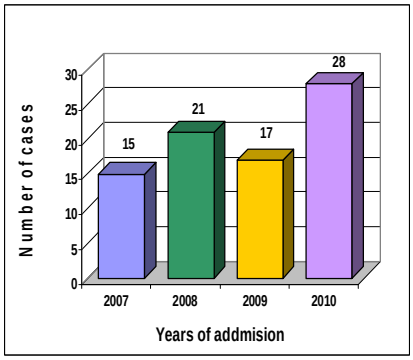
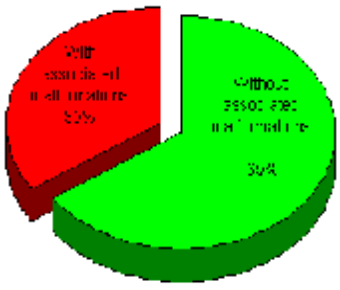


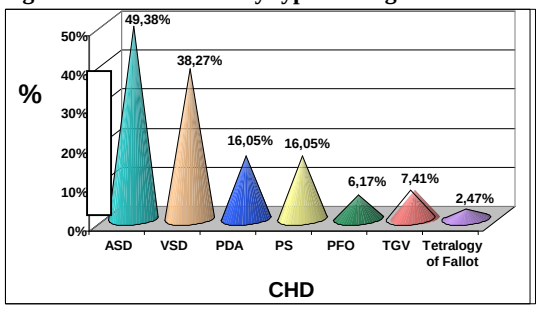
Figure no. 2. Distribution of cases according to the presence of malformations in other organs



21 infants (25.92%) were underweight at birth. Three of these children were with patent ductus arteriosus (PDA) (17.64%), and 10 infants (47.61%) had associated malformations of other organs and systems. The age at diagnosis of CHD: 17 infants (20.98%) up to one month, 79.01% by the age of 4 months and 91.39% by the age of 8 months. One case of CHD was diagnosed prenatally. 17 infants were diagnosed early in the newborn period: 7 had malformations classified as cardiovascular emergencies in the newborn (4 with transposition of the great vessels (TGV), 2 with tricuspid atresia (TA) and 1 with coarctation of the aorta - six of them were operated), 3 infants had AVC. Other 5 infants had multiple malformation syndromes. Complications of CHD: 7 infants developed heart failure (HF) (3 infants of those who had surgery) and 4 infants developed pulmonary hypertension (PH) (2 infants of those who had surgery).

Seven infants were diagnosed later in life, after 8 months old: 2 of these infants were diagnosed with Down syndrome, and one was diagnosed with tetralogy of Fallot. In time, they developed PH and CF and one of them died. No infant had surgery. The incidence of congenital heart defects was as follows: ASD 49.38%, VSD 38.27%, PDA 16.05%, pulmonary artery stenosis (PS), 16.05%, TGV 7.41%, CAV 6.17%, patent foramen ovale (PFO) 6.17%, tetralogy of Fallot 2.47%, TA 2.47%, partial anomalous pulmonary venous drainage (PAPVD) 1.23% and other rare malformations (figure no. 3).

Figure no. 3. Incidence by type of congenital heart defects



Clinically, cyanosis was mentioned in 13 infants (16.05%), and systolic murmur was present in 66 of the infants (81.48%). From all 13 cases that had been reported, only 8 were with cyanotic CHD. Echocardiographic examination was performed in 79 infants (97.53%). Radiography of the chest was performed in 58 infants (71.60%): in 31 cases (53.44%) changes were present and in 15 cases (25.86%) X-ray was normal.

11 infants died: 9 of them had echocardiographic examination and 5 of them had X-ray - 2 infants died shortly after admission, and in these 2 cases, the diagnosis of CHD was made by the pathological examination. The echocardiographic findings were confirmed by the pathological examination in all cases, and the radiological examination was normal in 4 infants. During the time, infants with CHD also had been diagnosed with other pathologies with high incidence: protein-energy malnutrition 44 cases (54.32%), rickets 40 cases (49.38%) (without taking into account infants younger than 3 months), iron deficiency anemia 54 cases (66.67%), upper respiratory tract infections 64 cases (79.01%), pneumonia 59 cases (72.84%), gastrointestinal pathology 17 cases (20.99%). In our study group, there were 9 infants (11.11%) that had surgery, 7 of them up to 4 months and 2 of them after one year of age (figure no. 4).

None of the infants that had surgery is listed among the dead children, but six of them had developed CF or PH. Heart failure was present in 28 infants with CHD (34.57%). 4 of these infants were a month old at the diagnosis of CF, 12 of them were between 1-4 months old, 4 - between 4-8 months old, 3 infants – between 8 - 12 months old, and 5 between 1 year - 3 years old (figure no. 5). 12 cases (14.81%) were complicated with pulmonary hypertension.

In the study group, there were 11 deaths. All have occurred under the age of one year old (6 between 1-4 months old, 3 between 4-8 months old and 2 between 8-12 months old).

Figure no. 4. Distribution of cases according to the surgical treatment

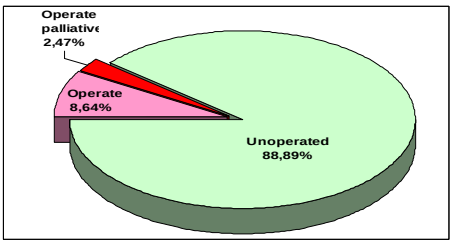
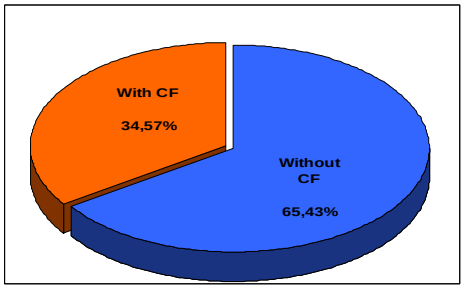


Figure no. 5. Distribution of cases according to the complications with IC



DISCUSSIONS

Congenital heart defects have a high priority in public health illnesses chart for detection, diagnosis, treatment and surgery, representing the most common congenital malformations present at birth with an impact on childhood morbidity and mortality. They are responsible for 6-10% of the total of 20-40% of infant deaths caused by congenital malformations.(3) Currently, in Romania, there are no records of a National Register of CHD in the pediatric population (0-18 years old) and therefore, we cannot assess objectively the extent and the impact of this pathology morbidity and mortality indicators in real time.(4) Among children with CHD, infants are a very important segment. We all know that the real chance of survival of the patients with CHD is the application of surgical treatment at the right time, so that all factors that could adversely affect their evolution may not have time to intervene. After solving the neonatal cardiovascular emergencies, the management of infant cases depends on what and how we treat them in other periods of childhood.

From the data processed in our study, 81 infants with CHD represent a significant percentage (47.9%), since they represent only one year of the child development, compared to other periods in which several years are grouped and new cases diagnosed in the respective periods were added. 2008 and 2010 were the most dynamic years in our study, this fact being reflected in the number of new cases diagnosed.

When considering the living environment, which is an important factor in population access to health services, we did not find any significant differences between the urban and rural population, and the percentages from our study are similar to those established by other studies in the country.(4)

Family history of CHD had a higher percentage (9.88%) compared to other studies where it represented only 1.6% and 3.77%.(4) These cases were associated with a lower age at diagnosis, in two thirds of cases being less than two months.

The difference noticed in the gender distribution is not consistent with the literature data that give a ratio of M/F = 1:1 (5, 6), in our study, males representing 62.96%.

Infants born with weight deficit and CHD require special attention.

Recovery from this deficit is very difficult (or not) compared to babies who do not have CHD. They often associate in evolution chronic protein-energy malnutrition and they require additional pre-and postoperative care. Another important aspect of weight at birth is the fact that 47.61% had also other organs and systems malformations, which increases the complexity of these cases. The percentage of premature infants who had presented PAC approaches the percentage established by other studies.(7)

Incidence by the type of heart disease differs from other statistics:(6,8) ASD has met the highest percentage and not VSD, and while both were above the rates found in other studies, PAC, PS, and TGV have also presented a higher percentage and AVC, TA and PAPVD approached the data found in literature.

Regarding the age at which they were diagnosed, we can say that there are still deficiencies in the primary care network and also in terms of public awareness. Only one infant was diagnosed antenatally and from 7 infants diagnosed after the age of 10 months, at least 3 could have been diagnosed much earlier, 2 being diagnosed with Down syndrome and one presenting cyanotic CHD. The consequences of the late diagnosis and a poor medical addressability appear immediately: 5 developed pulmonary hypertension and/cardiac failure and one died. None of them were operated.

CHD association with genetic syndromes or other organ and system abnormalities was lower than in other studies; probably many of them remaining undiagnosed yet, diagnostic methods not being widely available. A similar percentage with literature data we noted in the reporting of Down syndrome to other genetic syndromes diagnosed (4,5), and 2 infants of 6 children with AVC had also Down syndrome (33.33%). Surgical correction in infants with multiple malformations is difficult / impossible; in our study only one of the 28 children with multiple malformations received surgical treatment.

Because of the clinical heterogeneity of CHD, from the asymptomatic CHD - detected incidentally - to the complex CHD, with noisy symptoms, there is still concern for determining the clinical signs and symptoms that once pointed to a child must be followed by cardiology consultation and echocardiography. In our study, cyanosis was observed in 72.72% of cases with cyanotic CHD, 2 of 3 cases of cyanotic CHD that had no evidence of cyanosis had undergone surgical correction. Infants with non-cyanotic CHD that presented cyanosis at clinical examination showed CF / PH. Systolic murmur was present at clinical examination in 81.48% of cases. Even if it is considered that innocent murmurs are more common than the pathological ones, 10:1, simple evaluation by a non-cardiologist is insufficient to distinguish the innocent murmurs from the pathological ones.(1) Thus, the detection of a heart murmur at physical examination of a child should be followed by a cardiology consultation.

The radiography of the chest had a limited value in paraclinical diagnosis because of several reasons: many parents have refused this investigation because it is irradiating, only in 53.44% of cases cardiopulmonary changes were found, half of the cases being accompanied by radiological changes (especially in cases that are well tolerated and without important hemodynamic response), and in case of dead infants, the percentage of those infants where X-ray signalled a cardiopulmonary change decreased to 20%. Echocardiography is the method of diagnosis in all age groups; diagnostic sensitivity is superior to other investigations, regardless the child's age, the severity and complexity of the case. Using this investigation we can make a proper selection of patients for invasive

explorations, we can diagnose PH and quantify its severity, and not least to monitor cases.

Frequently associated pathologies in these children were: acute upper respiratory tract infections, pneumonia, iron deficiency anemia, rickets and chronic protein-energy malnutrition. This creates a vicious cycle from which benefits only infant morbidity and mortality.

After the diagnosis is determined, it must be established the optimal time for the surgical or interventional treatment, corrective / palliative, depending on the lesion, which can only be followed by the improvement of symptoms, normalization of the parameters of weight and growth and the neuro-psychological development, improving prognosis of these patients on long term. The percentage of children who received surgical or interventional treatment was of 11.11%, well below the national average, probably contributing to this situation the lack of a centre for pediatric cardiovascular surgery in the region. Another important aspect is that all children who have received surgical treatment are from the urban area.

Heart failure has installed in more than one third of children and, in most cases, before the age of one year. Pulmonary hypertension, a redoubtable complication of CHD, was reported in 12 cases. All 5 patients diagnosed with AVC are among the 12 cases with PH, the other cases being with ASD, VSD, PCA and complex CHD. In 10 cases, the complication occurred before the age of one year, including here also 4 cases with AVC (at 2 months old, 3 months old, 4 months old and 11 months old), and in the 5^{th} case, PH was diagnosed at 1 year and 4 months old. In most cases, there is a very small period of time (1-2 months) between the age of diagnosis of the heart malformation and the age when PH was diagnosed.

Death occurred in a number of 11 children, but without establishing a direct relationship between death and CHD as the cause of death, some of them being "dead carriers of CHD". 5 of the deceased had genetic syndromes and two of them had malformations in other organs. Seven of those who died had neurological impairment (of various causes). No dead child had cyanotic CHD, none of them had been operated and only one presented PH. As in the cases with complications of congenital heart defects with PH, the interval between the age at which cardiac malformations were diagnosed and death was low and the age of death in 4 cases coincided with the diagnosis of cardiac malformation.

CONCLUSIONS

1. Infants with CHD who were taken into study represented a high percentage of the children with CHD (47.9%), with a ratio M/F = 2:1, and 9.88% had a family history of CHD.
2. The incidence of the type of congenital heart diseases was different from other statistics, ASD has met the highest percentage and not VSD, and both were above the rates found in other studies. PAC, PS, and TGV also showed higher percentages and AVC, TA and PAPVD were close to the data found in literature.
3. Regarding the age of diagnosis, we can say that there are still deficiencies in primary health care network and awareness of the population. Only one infant was diagnosed antenatally.
4. The presence of malformations in other organs and systems (22.22%) and genetic syndromes (12.22%) was lower than in other studies, probably many of them still remain undiagnosed, the diagnostic methods not being widely available. A similar percentage with the one in literature data we had noted in the reporting of Down syndrome to other genetic syndromes.
5. Complications of congenital heart defects occurred in infancy, shortly after the baby was born offering almost no time to be discovered in earlier stage.
6. In the most dynamic years of the study, the number of newly diagnosed cases was significantly higher compared with other years, through the "active diagnosis", echocardiographic examination in the moment of any suspicion of CHD, without delaying the investigation for a period of time, so echocardiography examination could confirm the clinical diagnosis, and, by creating a closer link between the clinics of pediatrics and cardiology.
7. The early diagnosis and the application of surgical treatment in time are important, from infancy management of malformative pathology point of view that depends very much on the subsequent development of the children with CHD.
8. National programmes are needed for these children, which aim to establish a network of pediatric cardiology at national level and also, to widely inform the population about the treatment possibilities.

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