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CONGENITAL HEART DISEASE – A PUBLIC HEALTH PROBLEM

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ABSTRACT

Introduction. A congenital heart disease is a defect in the structure of the heart or great vessels that is present at birth. Signs and symptoms depend on the specific type of defect. The cause of a congenital heart defect is often unknown but remain the most common congenital malformations in newborns and infants with a major role in early and late neonatal morbidity and mortality.

Material and methods. The study was carried out in the Department of Neonatology and Premature in the collaboration with the Department of Pediatric Cardiology, from the Clinical Emergency Hospital for Children Louis Țurcanu Timișoara, in a period of 3 years (2015- 2018). There were 70 patients included in the study, who presented congenital heart malformation diagnosed clinically, by ultrasound and Angio-CT.

Results. The incidence of cardiac malformations in our unit was 2.11%. The most common clinical signs encountered in patients with congenital heart disease were dyspnea associated with cyanosis, systolic murmur and oxygen saturation oscillations (SaO2). Associated with pregnancy, the presence of teratogenic factors with possible implication in congenital heart malformations was also detected. There were 12 cases involved genetic syndromes, Down's syndrome having the highest prevalence. Due to the complexity of the cardiac malformation in the studied group, 11 patients (15.71%) died.

Conclusions. Congenital cardiovascular malformations are a problem of public health. In the assessment of a newborn with cardiac pathology, there should be involved obstetricians, neonatologists, anesthetists, pediatric cardiologists, pediatric cardiovascular surgeons, geneticists, with the common goal of preventing, diagnosing, monitoring and treating congenital heart abnormalities.

Keywords: congenital heart malformation, newborn, echocardiography

INTRODUCTION

Congenital heart disease still remain the most common congenital malformations in newborns and infants, with a major role in early and late neonatal morbidity and mortality.

The cause often remains unknown, but important risk factors such as congenital infections, tobacco and alcohol consumption, obesity, parents related are found in pregnant women. Some genetic diseases such as Down syndrome, Turner syndrome and Marfan syndrome can be associated with heart defects.

The signs and symptoms depend on the type of malformation, the newborns can be asymptomatic, have minor and nonspecific symptoms or be severe, endangering life. In all cases, further investigations are needed to reach the diagnosis and determine the optimal course of action.

Major advances in medicine have led to improved survival rates, particularly through the use of echocardiography with Doppler color flow measurements. Computer tomography, MRI, angio-CT and angio-MRI have made it possible to diagnose asymptomatic, minor or even lesions without murmur.

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AIM

The authors proposed to analyze the incidence, evolution and complications of complex cardiac malformations as risk factors for neonatal mortality.

MATERIAL AND METHODS

The authors proposed to analyze the incidence, evolution and complications of complex cardiac malformations as risk factors for neonatal mortality. A retrospective study was conducted over a period of 3 years (01.01.2015-31.12.2018), on a batch of patients at the Neonatology – Premature Clinic of the "Louis Turcanu" Emergency Hospital for Children, Timisoara, which included 70 patients admitted with congenital heart disease diagnosis.

The target group consisted of hospitalized children aged between zero and 6 months who had clinical signs of congenital heart disease and specific imaging investigations for congenital heart disease. We mention that the congenital heart disease diagnosis has been established based on a collaboration protocol with the Department of Pediatric Cardiology.

The variables included for the analysis were gestational age, birth weight, sex, associated malformative syndromes, evolving complications (cardio-respiratory insufficiency, endocarditis, pulmonary hypertension, pulmonary hemorrhage, death).

Determining the frequency of congenital cardiovascular malformations in the child based on maternal age, teratogenic factors (maternal diseases, medication or drug use during pregnancy, viral infections).

RESULTS

During the 3 years out of 3,316 patients admitted to the department, 70 were diagnosed with congenital heart disease, the incidence of cardiac malformations in our unit being 2.11%.

The average age of mothers was 28.5 years. There were 8 cases over the age of 35 (11.42%). The average gestation age was 37 weeks. In the studied group 38 newborns were on term, 19 newborns were premature (GA 32-36 weeks), and 13 cases were within the term with intrauterine growth restriction. The average birth weight was 2455.6 grams. Gender distribution revealed a slight predominance of male gender: 57.2% boys versus 42.8% girls (Table 1).

Associated with pregnancy, the presence of teratogenic factors with possible implication in congenital heart malformations was also detected. In the studied group, 37 mothers (52.8%) presented chronic illness during the pregnancy: pre-existing hypertension in 17 cases, diabetes in 13 cases, hypothyroidism in 3 cases and epilepsy in 4 mothers. Additionally, tobacco consumption was found in 4 cases (5.7%) that gave birth to newborns with congenital heart disease (Table 2). Regarding the prenatal congenital heart disease diagnosis, the percentage was relatively low, 5.71% representing 4 cases.

TABLE 1. *Characteristics of the study group (n = 70 patients)*

Variables	
Age of mother in years (average / min-max)	28.5 (18-40)
Age of gestation in weeks	37 (32-40)
GA distribution – n (%)	
Newborns within term (AGA)	38 (54.28%)
Premature newborns	19 (27.15%)
Newborn infants with IGRU	13 (18.57%)
Weight at birth – grams (average / min-max)	2,455.6 (1,600-4,120)
Gender of newborn babies	
Male – n (%)	41 (57.2%)
Female – n (%)	29 (42.8%)

TABLE 2. *Possible teratogenic factors involved in congenital heart malformations etiology*

Chronic diseases associated with pregnancy in mothers (%)	37 (52.8%)
Pre-existing hypertension	17 (24.3%)
Diabetes	13 (18.5%)
Hypothyroidism	3 (4.3%)
Epilepsy	4 (5.7%)
Tobacco consumption in mothers (%)	4 (5.7%)

The most common clinical signs encountered in patients with congenital heart disease were dyspnea associated with cyanosis, systolic murmur and oxygen saturation oscillations (SaO2). Clinically cyanosis was present in 38 cases (54.28%), of which 32 cases were cyanogenic congenital heart disease, systolic murmur was reported in 56 of the patients (80%). The etiological differentiation of these symptoms (common in respiratory disorders) was primarily due to the hyperoxia and pulse oximetry tests. Pulse oximetry revealed in all cases of cyanogenic congenital heart disease values of SaO2 below 90%.

As the first imaging step, thoracic radiography was performed, which revealed in 30 cases (42.86%) cardiomegaly, 25 newborns (35.71%) showed changes of the aortic button and pulmonary vasculature and in 15 cases (21.43%) no radiographic changes were present.

Transthoracic echocardiography, being the most non-invasive exploration in identifying the morphological and functional data characteristic of each cardiac defect and allowing a correct diagnosis most often, was performed in all cases with suspicion of

congenital malformation and was repeated for periodic evaluations. The device used was the Philips CX50 with the S12-4 probe.

Following the imaging investigations (thoracic radiography, cardiac ultrasound, angio-CT, angio-MRI), the arterial channel persistence prevailed within 37.14% (26 cases), pulmonary artery stenosis 11.43% (8 cases) followed by Tetralogy of Fallot 5.71% (4 cases), total anomalous pulmonary venous drainage with coronary sinus drainage 5.71% (4 cases), 6 cases (8.57%) of atrioventricular canal full Rastelli type A. Other diagnosed congenital malformations were: transposition of the great arteries (Fig. 1), right ventricle with double exit path, type A Ebstein disease (Fig. 2) with tricuspid valve attached to septal and associated severe tricuspid regurgitation, severe aortic coarctation 3 cases, common arterial trunk, left ventricular hypoplasia, 1 case of Taussig-Bing anomaly, dextrocardia associated with ventricular septal defect, tricuspid valve atresia with ventricular septal defect and left branch peripheral stenosis (Table 3).

TABLE 3. *Congenital heart disease present in the study group (n = 70 patients)*

Congenital heart disease in patients (%)	70
Arterial canal persistence	26 (37.14%)
Pulmonary artery stenosis	8 (11.43%)
Transposition of the great arteries	6 (8.57%)
Atrioventricular canal full form Rastelli type A	6 (8.57%)
Fallot Tetralogy	4 (5.71%)
Total anomalous pulmonary venous drainage	4 (5.71%)
with coronary sinus drainage	4 (5.71%)
Right ventricle with double exit path	1 (1.42%)
Type A Ebstein disease	2 (2.85%)
Common arterial trunk	2 (2.85%)
Hypoplastic left heart syndrome	3 (4.28%)
Severe aortic coarctation	1 (1.42%)
Taussig-Bing anomaly	2 (2.85%)
Dextrocardia associated with ventricular septal defect	1 (1.42%)
Tricuspid valve atresia with ventricular septal defect and left branch peripheral stenosis	

In the studied group, 12 cases involved genetic syndromes, representing 17.14% of the total congenital heart disease, Down's syndrome having the highest prevalence. Cases were clinically diagnosed and confirmed by cytogenetic analysis.

Due to the complexity of the cardiac malformation in the studied group, 11 patients (15.71%) died, in the literature the percentage of deaths due to congenital malformations in the neonatal period being 21%. The rest of the cases developing severe complications (pulmonary hypertension, cardio-respiratory insufficiency, pulmonary haemorrhage) required prolonged hospitalization with increased consumption of human and material resources.

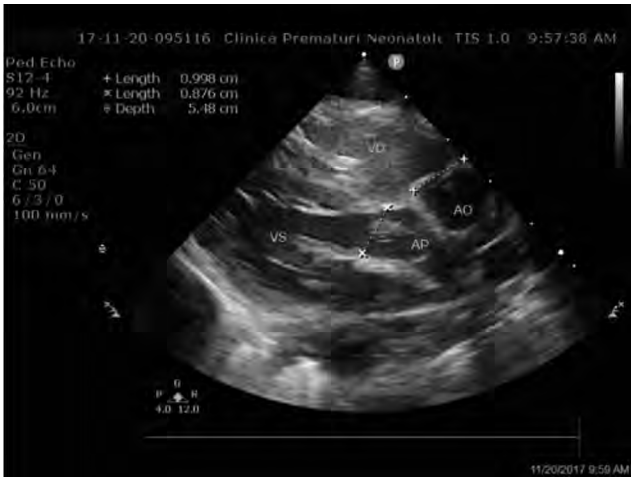

FIGURE 1. *Transposition of the great arteries*

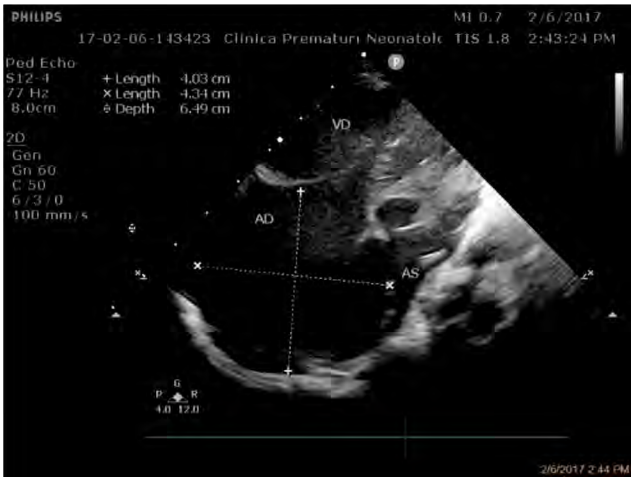

FIGURE 2. *Ebstein disease*

DISCUSSIONS

Congenital heart malformations are the most common cause of malformations present at birth. The congenital heart disease incidence in our unit was 2.11%, slightly higher than the literature with 0.3-1.2% [1]. In a study published in 2010 on congenital heart disease prevalence and perinatal mortality in Europe in the years 2000-2005, a prevalence of 8 cases per 1,000 births is reported [2].

The average age of mothers was 28.5 years and the percentage of mothers aged over 35 years was 11.42%. The association of a congenital heart disease with mothers over 35 years of age, described in the literature, was only sustained when cardiac disease was a feature of a syndrome.

Concerning the neonatal variables included for analysis: gestational age and birth weight, there was no significant correlations between them and the presence or absence of congenital heart defects. In the studied group predominantly were term newborns (AGA and IGRU) compared to preterm newborns. In a study published by Kirsty Tanner and colleagues on

the correlation between heart malformations and prematurity, they reported a 16% premature birth rate associated with congenital heart defects, the probability rate for a cardiac malformation during prematurity is of 2.4 (95% CI: 2.2-2.7) [3].

Gender distribution revealed a slight predominance of male sex: 57.2% boys versus 42.8% girls, data inconsistent with those in literature [4].

In the studied group of 37 mothers (52.8%) with chronic diseases during pregnancy, the highest weight had the ones with pre-existing hypertension in 17 cases (24.3%), followed by those with diabetes 13 cases (18.5%), hypothyroidism and epilepsy.

In our study, the incidence of newborn pregnancies in mothers with pre-existing hypertension and diabetes mellitus is slightly higher compared to literature with an incidence of 2.5 and 3.1-18% [5,6].

Regarding the prenatal diagnosis of CHD, the proportion was relatively low at 5.71% compared with studies in Europe where the incidence is 20.2% [2]. Most heart malformations can be diagnosed prenatally by fetal ultrasound. The main benefit of prenatal diagnosis of severe cardiac malformation is to schedule birth in a specialized center and perform surgery in a timely manner.

The presence of a systolic breath at routine examination may be evidence of a heart malformation. In the studied group, 80% of the patients had systolic breathing during the examination. According to the data, routine examination has a sensitivity of 44% and the presence of a blast has a positive predictive value of 54% [7]. For etiological differentiation of cyanosis and oxygen saturation oscillations (SaO2) pulsoximetry and hyperoxia test were used. Pulse oximetry is the main procedure for the indirect detection of hypoxemia since 1980. A meta-analysis of pulmonary screening in CHD patients reported a sensitivity of 76.5% (95% CI 67.7-83.5) and specificity of 99.9% (95% CI 99.7-99.9) [8].

In a 5-year study at the Emergency Institute for Cardiovascular Disease and Transplantation in Tg. Mures, the ACP incidence was 22.10%, in our study the incidence was higher [9].

Preterm newborns have an increased incidence of ACP due to a physiological abnormality rather than a structural anomaly, leading to an increase in the number of premature infants with ACP. Recent studies based on echocardiography reveal that in neonates the arterial canal is almost closed until the age of 7 days of life [10].

From the studied CHDs, pulmonary artery stenosis, large vessel transposition, atrioventricular canal had the same incidence as in the literature [11].

The association of heart malformations with genetic syndromes was lower compared to other studies, and the diagnostic methods were not widespread, some of the syndromes remaining undiagnosed.

In the group studied the association between CHD and genetic syndromes accounted for 17.14% of the total CHDs, Down's syndrome having the highest prevalence. A study in children diagnosed with trisomy 21 in Denmark reported an incidence of CHD in these children of 43% [12].

Due to the complexity of the cardiac malformation in the studied group, 11 patients (15.71%) died. A negative prognosis had the cases that developed complications: heart failure, pulmonary hypertension, pulmonary haemorrhage, and CHD in Down syndrome. Statistical data on the studied group confirm a relevant loss of newborns diagnosed with congenital heart malformations, especially complex disease forms, representing one of the main causes of perinatal and infantile death.

CONCLUSIONS

After confirming the diagnosis and initial stabilization of the patient, priority was to transport to a tertiary center for palliative or early corrective surgery.

CHD are a public health problem due to the frequency and costs of care, the patient requiring a multidisciplinary team with the common goal of preventing, diagnosing, monitoring and treating congenital heart abnormalities.

The newborn diagnosed with a congenital malformation needs a good understanding of the associated complications both in the short and long term, the diagnostic methods and the therapeutic behavior being in continuous development.

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