EPIDEMIOLOGICAL DATA OF CONGENITAL HEART DISEASE IN OUR CENTER

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Abstract

Background: Congenital heart defects (CHD) are among the most common birth defects and are the leading cause of birth defect-related deaths. The incidence of congenital heart disease in live newborns varies from 4.1/1000 to 12.3/1000 (1.5). The incidence of CHD in underdeveloped countries is unknown, but the distribution of different lesions is fairly similar to those in developed countries. This study is the first report that aims to present the epidemiological data of congenital heart disease in our Pediatric Cardiology Unit as the single tertiary care pediatric cardiac center in Albania.

Methods: Were analyzed retrospectively the data-base of the outpatient clinic of our service between December 2010 and December 2011 who were referred for evaluation by the Pediatric community. All patients were evaluated with physical examination, electrocardiogram and echocardiography.

Results: Were analyzed 1878 cases: 887 cases (48%) males and 991 cases (52%) females, ages 1 day-15years (mean age 3.8years ± 3.7). Among 1878 patients examined, 1325 cases (70.6%) had a normal heart, and 553 cases (29.4%) were diagnosed with CHD, and 198 of these cases were diagnosed for the first time. Ventricular Septal Defect (VSD) was the most common lesion with 49 cases (25%), followed by Patent Ductus Arteriosus (PDA) with 19 cases (9.7%), ASD atrial septal defect with 17cases (8.5%). Tetralogy of Fallot is the most common cyanotic heart disease with 17 cases (9%) and complex heart defect in about 8-9% of cases. Maximum number of children with heart disease were diagnosed between 0-3 years of age, where only in the neonatal period were diagnosed 72 patients out of 104.

Conclusion: CHD constitute the largest number of congenital defects in children. Distribution of specific lesions and sex distribution was similar to findings from other parts of the world. It is important to create Albania Infant Cardiac Registry to have an accurate picture of the prevalence and incidence of CDH in our country.

Key words: congenital heart disease, epidemiology, Infant Cardiac Registry.

Introduction

Congenital heart disease is a category of heart diseases that includes abnormalities in cardiovascular structures that occur before birth, while the fetus is developing in the uterus.

"1 million infants are born each year with heart defects". This figure seems extraordinarily high. Estimates of the incidence of congenital heart disease in live newborns vary from 4.1/1000 to 12.3/1000 (4,6). Observed differences may also be of

genetic, environmental, socioeconomical, or ethnic origin, and there needs to be further investigation to tailor the management of this global health problem (1,4,6).

Heart defects are among the most common birth defects and are the leading cause of birth defect-related deaths. For more than a half of these babies, the condition is a minor problem which either doesn't need any treatment, or can be successfully corrected with surgery after they are born.

Signs and symptoms are related to the type and severity of the heart defect. Symptoms frequently present early in life, but it is possible for some CHDs to go undetected throughout life. Some children have no signs while others may exhibit shortness of breath, cyanosis, syncope, heart murmur, under-developing of limbs and muscles, poor feeding or growth, or respiratory infections. Congenital heart defects cause abnormal heart structure resulting in production of certain sounds called heart murmur. These can sometimes be detected by auscultation; however, not all heart murmurs are caused by congenital heart defects.

Congenital heart disease may be diagnosed before birth, right after birth, during childhood, or not until adulthood. It is possible to have a defect and no symptoms at all. The advent of echocardiography with Doppler color flow measurements has made it possible to diagnose lesions that are asymptomatic, minor, and even without murmurs. Given these differences, there does not appear to have been a significant increase in the incidence of CHD over the last 20–30 years. The incidence of CHD in underdeveloped countries is not known, but the distribution of different lesions is fairly similar to those in developed countries except perhaps for fewer with aortic stenosis and coarctation of the aorta (5).

How Is Congenital Heart Disease Diagnosed?

Congenital heart disease is often first detected when your doctor hears an abnormal heart sound or heart murmur when listening to your heart. Depending on the type of murmur your doctor hears, he or she may order further testing such as:

- Echocardiogram or transesophageal echocardiogram (TEE).
- Cardiac catheterization.
- Chest X-ray.
- Electrocardiogram (ECG).
- MRI.

Aim

To give the epidemiological data of congenital heart disease in patients referred to the outpatient clinic of our service pediatric cardiology as the single tertiary care pediatric cardiac center in Albania.

Methods

Were analyzed retrospectively the data-base of the outpatient clinic of our service between December 2010 and December 2011 who were referred for evaluation by the Pediatric community. All patients were evaluated with physical examination, electrocardiogram and echocardiography.

The investigation of choice is echocardiography with Doppler color flow:

- It is non-invasive.
- It can elucidate both anatomy and flow and will give some indication of the underlying abnormality.
- The advent of echocardiography with Doppler color flow measurements has made it possible to diagnose lesions that are asymptomatic, minor, and even without murmurs.

Results

Were analysed 1878 cases: 887 males (48%) and 991 females (52%), ages 1 day-15 years (mean age 3.8years ± 3.7). The reason for referral was:

- A heart murmur or signs and symptoms of suspected CHD in 1551 cases (82.6%): innocent murmur, tachycardia, tachypnea, hepatomegalia, cianosis, week pulses etc.
- A genetic syndrome: Down syndrome, Turner syndrome, Marfan syndrome 107 cases (5.7%).
- Parent with cardiopathics or suspected familiar recurrence in 94 cases (5%).
- Symptoms of arrhythmias in 70 cases (3.7%) tachycardia supraventricular, BAV of different degrees or other rare disorders of rhythm.

- Prematurity in 56 cases (3%).

Among 1878 patients examined, 1325 cases (70.6%) had a normal heart, and 553 cases (29.4%) were diagnosed with CHD, and 198 of these cases were diagnosed for the first time. Ventricular septal defect (VSD) was the most common lesion with 49 cases (25%), followed by atrial septal defect (ASD) with 17cases (8.5%) and patent ductus arteriosus (PDA) with 19 cases (9.7%), coarctation of the aorta (CoA) with16 cases (8.1%), pulmonary stenosis (SAP) with 18 cases (9%), aortic stenosis (StAo) with 16 cases (7%), endocardial cushion defect (ECD) with 6 cases (3%). Tetralogy of Fallot (TF) is the most common cyanotic heart disease with 17 cases (9%), transposition of the great arteries(TGA) with 6 cases (3%), persistent truncus arteriosus (TAC) with 1 case (0.5%), single ventricle (VU) with 2 cases (1%), Ebstein's anomaly with 2 cases (1%), double–outlet right ventricle (DORV) with 3 cases (1.5%), hypoplastic left heart syndrome (HLSL) with 2 cases (1%) etc. (Figure nr.1). Maximum number of children with heart disease were diagnosed between 0-3 years of age, where only in the neonatal period were diagnosed 72 patients out of 104. From 198 patients: 105 cases (53%) female and 93 cases (47%) male. In ASD and PDA there is a predominance of females about 2:1, but in aortic stenosis and coarctation of aorta there is a predominance of male about 3:1.

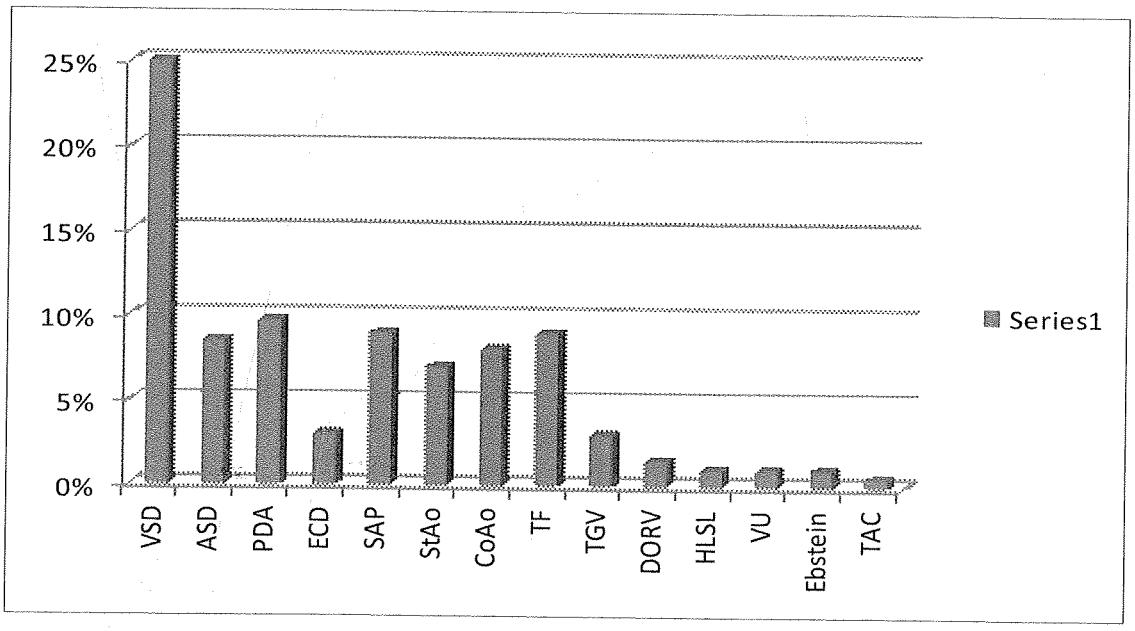

Figure nr.1. Form distribution of CHD

From patients referred to a genetic syndrome resulted: in 21 patients with Down syndrome 4 cases were with EDC and 2 with EDC+TF. In 5 patients with Turner syndrome 2 cases resulted with CoAo.

When the reason for referral was parent, sisters, brothers with cardiopathie or suspected familiar recurrence 4 cases (2%) of CHD were attributed to CHD family story.

From 56 cases with prematurity in 25 resulted the PDA, on the first day of life, but after treatment with oral or venosus ibuprofen had a shutdown in 18 cases on the third day after treatment. Ductus arteriosus remained open from this contingent only in 7 cases.

Discussion

It is very difficult to determine the true incidence of CHD in Albania, because we must declare all new cases with CDH from all examiners in the Infant Cardiac Registry, which does not exist in Albania. Absence of this register risk was the same case examined by different examiners referred to as two cases. Although the Pediatric Cardiology Unit

is the only tertiary center in Albania, for various reasons, not all the cases are presented here. It is important to note that in Albania is almost natural incidence of CHD, due to the lack of specialized structures for performing cardiac fetal examination. In our study we can present the distribution of CHD in the cases submitted for examination.

Previous studies documented the prevalence of CHD among infants using surveillance registries (1,4,10). Reported rates varied from 4 to 10 per 1000 live births. For severe CHD, the New England Infant Cardiac Registry reported a prevalence of 1.5 per 1000. Referral-based participation may result in overascertainment of severe disease and underascertainment of mild lesions more likely to become manifest after the first year of life. Not surprisingly, the population-based prevalence of CHD in children in this study is higher than previously found. For severe CHD, which is expected to become manifest in the first year of life regardless of the ascertainment method, our results for children are in line with the findings of others.

In our study *the ventricular septal defect* is the most common defects (25%) of CDH. This fact is in correlate with the others studies. (5,9). The total incidence of CHD was related to the relative frequency of ventricular septal defects (VSDs), the most common type of CHD. The incidences of individual major forms of CHD were determined from many studies. The incidence of CHD depends primarily on the number of small VSDs included in the series, and this number in turn depends upon how early the diagnosis is made. The incidence of moderate and severe forms of CHD is about 6/1,000 live births (19/1,000 live births if the potentially serious bicuspid aortic valve is included), and of all forms increases to 75/1,000 live births if tiny muscular VSDs present at birth and other trivial lesions are included. Given the causes of variation, there is no evidence for differences of incidence in different countries or times (1,8).

Patent ductus arteriosus is another common lesion, the incidence of which varies with the age at the time of study and the gestational age of the subject. In the term infant the ductus arteriosus is almost always closed by four to seven days after birth (5,12). Preterm infants have an increased incidence of PDA based on abnormal physiology rather than on a structural abnormality. With improvement in oxygenation, the pulmonary vascular resistance falls rapidly, but the dustus remains patent because its responsiveness to oxygen is immature in premature newborns (5,9).

From 56 cases with prematurity in 25 of them resulted the large PDA, on the first day of life. These patients were treatment with oral or venosus ibuprofen had a shutdown in 18 cases on the third day after treatment. Ductus arteriosus remained open from this contingent only in 7 cases, who were treated surgical closure.

Atrial septal defect in our study makes up 8.5% of the cases. Because an ASD is usually asymptomatic and has murmurs that are often soft, these defects frequently do not lead to early diagnosis or referral (11,12).

This is why many of these subjects present in adult life so the incidence in childhood usually underestimates the true incidence of the lesion (5). Overall, sex distribution was similar, more females than males were affected. In previous studies (9) we find the same ratio, but in Saudi Arabia the males are more affected than females (8). The prevalence of ASDs, when all types are considered together, it is higher in female individuals than in male individuals. The female-to-male ratio in our study is approximately 2:1.

Atrioventricular septal defects (AVSDs) (endocardial cushion defects, common atrioventricular canal) have an incidence that varies with the age of the involved mothers. We have found in our study 3% of cases, of which 2% in Down syndrome. Trisomy 21 (Down syndrome) is much more common in mothers more than 34 years old, and AVSDs are much more frequent in those with trisomy 21 than with normal chromosomes (5).

Thus, the proportion of older mothers in a series greatly increases the incidence of AVSDs. Because therapeutic abortions may be performed if trisomy 21 is discovered early in pregnancy, the incidence of AVSD at term is likely to decrease in future years, in our country.

Severe CHD was defined as tetralogy of Fallot, truncus arteriosus, transposition complexes, endocardial cushion defects, univentricular heart and hypoplastic left heart syndrome. In previous studies it is determined the prevalence of severe and other CHD lesions (3). The prevalence was 4.09 per 1000 children in the year 2000 for all CHD and 9% for those with severe lesions. High number (18%) of severe CHD in our study is explained by the fact that our country is almost a natural incidence of this defect due to lack of specialized structures for fetal cardiac examination.

In our study the proportion of females in the CHD population was higher than males 53% versus 47% ($P<0.0001$). Same authors (1) have found than in those with severe disease, the prevalence was higher in female adults (0.41 per 1000 versus 0.35 per 1000; $P=0.0001$) but not in female children. Shunt lesions (ASD, VSD, PDA, and AVCD) were more common in females (9.95 per 1000 versus 7.92 per 1000; $P<0.0001$). Transposition complexes and coarctation were more common in males (0.31 per 1000 versus 0.22 per 1000, $P=0.0002$; 0.30 per 1000 versus 0.19 per 1000, $P<0.0001$, respectively).

Down syndrome constitutes the majority of chromosomal defects. EUROCAT study (7) estimate the contribution of Down syndrome with CHD to the total pediatric CHD case load to vary from 3% to 4% (Italy, France, and Switzerland) to 15% to 19% (Ireland and Malta), assuming an average nonchromosomal CHD LB prevalence of 6.5 per 1000 for all countries. Twelve percent of CHD cases in the EUROCAT series were chromosomal, the majority Down syndrome. Variation in total prevalence of chromosomal CHD cases can be explained by large differences in the maternal age profile of European populations. Approximately half of live-born children with Down syndrome are usually considered to have a cardiac defect (9), 45% in our data set. Most frequent defect of Down syndrome is EDC (31%) following by VSD (18%) and ASD (15%). We have found 2 cases with Down syndrome with a rare cardiopathie EDC+TF.

In our study 2% of CHD were attributed to CHD family story. Same authors have found that only 2.2% of heart defect cases in the population (4.2% after the exclusion of chromosomal aberrations) were attributed to CHD family history in first-degree relatives. Family history of any CHD among first-degree relatives accounted for a small proportion of CHD cases in the population (2).

The present study has a number of limitations. Although we have a single tertiary care pediatric cardiac center in Albania, for various reasons, not all cases of CDH presented here, so this information is not completely representative of the Albanian population. Another limitation relates to information recorded on the age at diagnosis: The data set includes whether a congenital anomaly was diagnosed in postnatal age at first detection, but if the neonate is multiply malformed it does not specify which anomaly was first diagnosed.

Conclusion

Cardiac heart disease constitute the largest number of congenital defects in children. Distribution of specific lesions and sex distribution was similar to findings from other parts of the world. Their diagnose must be made as early as possible during intrauterine period, which remains a challenge before us. It is important to create Albania Infant Cardiac Registry to have an accurate picture of the prevalence and incidence of CDH in our country.

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