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Editorial

# Congenital heart disease

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Congenital heart disease (CHD) is the most common cause of major congenital anomalies, and is the group of malformations that contributes the most for perinatal mortality [1]. It represents an important health care issue and knowing its incidence and risk factors helps developing public and private care policies and clinical protocols. In this paper, researchers published a retrospective analysis from a tertiary hospital in Portugal regarding CHD. In this editorial, we analyze their findings and compare with other publications.

CHD are serious and common conditions that have a significant impact on morbidity, mortality and healthcare costs in both children and adults [2]. Several studies have aimed to determine its prevalence among life births and different methods have been applied in this purpose.

Linde et al separated the 8 most common subtypes of CHD: ventricular septal defects (VSD), atrial septal defects (ASD), pulmonary stenosis (PS), patent ductus arteriosus (PDA), tetralogy of Fallot (TOF), aortic coarctation (CoAo), transpositions of great arteries (TGA) and aortic stenosis (AoS). Their study population was of 24,091,867 separated in 114 studies and the prevalence found have increased from 0,6 per 1000 live births to 9,1 per live births from 1930 to 2010. Marked heterogeneity between global regions was noted, probably due to differential availability of diagnostic technology.

This systematic review had a valuable impact, however, since then the literature have expanded significantly and new studies have been published. Liu et al proposed an expansion in diagnosis groups and hypothesized that a larger dataset including less common and typically more severe types of CHD would be possible to discern important prevalence trends [3].

In this meta-analysis, 260 studies were included with a sum of 130,758,851 live births and in their findings, the prevalence of CHD was 8,224 per 1000, number slightly higher then this present article.

It is important to highlight that the majority of patients from this database were from studies published post-2010, representing 79% of the total population. They mentioned that previous analyses had indicated an apparent plateau in global CHD prevalence between 1995 and 2009, however, by comparison with these years there was a ~10% increase in CHD prevalence from 2010 to 2017. VDS, ASD and PDA (the mild lesions) were the three most frequent types of CHD. In total, these mild lesions contributed 57,9% of all CHD. Changes in the prevalence of VSD, ASD and PDA together explained 93,4% of the increase in prevalence of total CHD after 2010.

Two trends in birth prevalence of CHD appeared in this study: 1) a progressive increase in the estimated prevalence of right ventricular outflow tract obstruction and 2) a decrease by approximately one-third in the estimated prevalence of left ventricular outflow tract obstruction from 1990 onwards (which includes hypoplastic left heart syndrome). This reduction is probably due to higher availability of prenatal echocardiography and the choice of pregnancy termination in such cases.

The present article have proposed to categorize CHD into 4 different groups: left to right shunt lesions, cyanotic, non-cyanotic obstructive and miscellaneous (diseases that did not fit into any of the three prior categories). As a result, the prevalence of CHD was 6 per 1000 live births, result that is similar then other studies. When divided into the 4 groups, 71% were left to right shunt lesions, followed by 16% of cyanotic, 11% of non-cyanotic obstructive and 3% miscellaneous. The mortality related to CHD was 3,4% (n = 10), of which nine belonged to th cyanotic group and one was classified as miscellaneous (a left ventricular noncompactation diagnosis). This high mortality frequency in cyanotic cases may be due the unorthodox distribution of diseases into the 4 categories chosen. The cyanotic group included complete atrioventricular canal defect (usually classified as a left to right shunt CHD) and hypoplastic left heart syndrome, a non-cyanotic obstructive CHD. Together, they represent one third of the cyanotic deaths described.

There are several ways to classify CHD [4]. A pathophysiology classification, namely, a classification based upon the clinical consequences of structural defects impairing the physiology of blood circulation was described by Thiene and Frescura and include:

1. CHD with increased pulmonary blood flow (septal defects without pulmonary obstruction and left to right shunt)
2. CHD with decreased pulmonary flow (septal defect with pulmonary obstruction and right to left shunt)
3. CHD with obstruction to blood progression and no septal defects (no shunt)
4. CHD so severe as to be incompatible with postnatal blood circulation
5. CHD silent until adult age

This classification would separate the cyanotic group of this article and obtain different mortality results.

This article authors describe a positive association between complete atrioventricular defect, atrial septal defect and left ventricular noncompactation with extra-cardiac malformations. They did not describe which extra-cardiac malformation they looked into.

Egbe et al studied the prevalence of congenital anomalies in newborns with congenital heart disease diagnosis and categorized the congenital anomaly groups: total non-cardiac congenital anomalies, genetic syndromes, non-syndromic congenital anomalies and multiple organ-system congenital anomalies [5]. Their data showed that newborns with CHD frequently have associated extra-cardiac congenital anomalies. This association is true for both syndromic malformation and non-syndromic congenital anomalies.

Craniofacial, respiratory and genitourinary malformations were associated. Contrary to some published data, they did not find any association between CHD diagnosis and gastrointestinal and limb anomalies. Genetic syndromes were strongly associated with CHD diagnoses and this association was most significant with septal defects, endocardial cushion defect, pulmonary valve disease, tricuspid valve disease, aortic valve disease, truncus arteriosus and aortic arch anomalies.

Rosa et al described extra-cardiac malformations most frequently reported among patients with congenital heart diseases. The authors separated into groups depending on the system involved: central nervous system, craniofacial, eyes, respiratory, digestive, musculoskeletal, genitourinary and spleen anomalies [6].

An important aspect highlighted by this article was the incidence of 24% prenatal diagnosis of all CHD cases. This points out to the low number of prenatal diagnosis even in high-income per capita countries and the need for such studies in order to establish proper screening policies. Hagemann et al submitted 3980 fetus of low obstetric and cardiological risk pregnancies to prenatal echocardiography and found a 2,5% prevalence of CHD in Brazil, an upper middle income country [7]. These patients would probably remain without diagnosis until complications arise.

Even when fetal echocardiography is unavailable, pulse oximetry around 24 hours of life is a low cost screening tool that can affect directly on child mortality due to CHD. A JAMA original investigation in 2017 showed a strong association between pulse oximetry screening and decrease in infant cardiac deaths between 2007 and 2013 compared to US states without screening policies [8].

We hope that in the future not only pulse oximetry but also fetal echocardiography be available in a satisfactory frequency in order to prevent cardiac congenital disease deaths due to late or no diagnosis. To obtain this goal, prevalence studies such as this are crucial.

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