Chapter 1

Introductory Chapter: Congenital Heart Disease

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Additional information is available at the end of the chapter

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1. Introduction

Birth defects result in abnormal physiology, often with detrimental consequences, be it physical, developmental or intellectual disability. The resulting phenotype can range from mild impairment to severe to near incompatible with life. In most extreme cases, the foetus is incompatible with life and is spontaneously aborted *prepartum*, at much distress to the parents. However, all forms are of great personal pain to the families involved.

Birth defects are broadly categorised as either structural, affecting the 'shape' of the body, or 'functional' affecting the functionality of an organ or body system, in this case, the heart and circulation. Congenital heart disease (CHD) is a non-specific medical term for a range of defects present at the time of birth that affect the normal physiology of the heart and associated circulatory system [1]. CHD can arise from a combination of genetic and environmental causes. Recent advances in molecular testing techniques aid in diagnosis of these conditions; however, this creates ethical considerations with respect to sustainability and continuity of life with such conditions, especially when treatment options to alleviate debilitating symptoms may not be available.

1.1. Epidemiology of CHD

CHD is one of the most common types of birth defects. About 28% of major congenital abnormalities are a result of a cardiac pathology [2]. These can be broadly categorised as those that result in cyanosis and those that do not (**Figure 1**). CHD occurs in approximately 1% of live births and affects up to 9 in every 1000 babies born in the United Kingdom (REF); however, the reported prevalence differs considerably globally.

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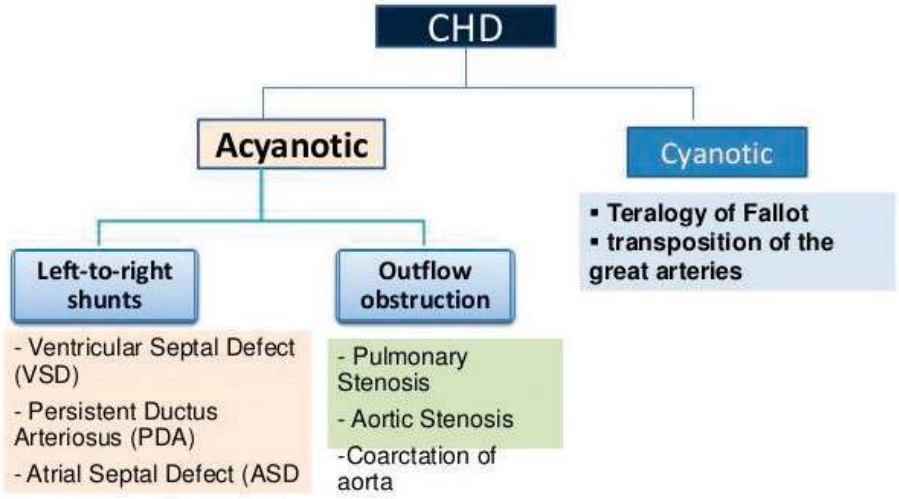

Figure 1. Broad classification of congenital heart disease.

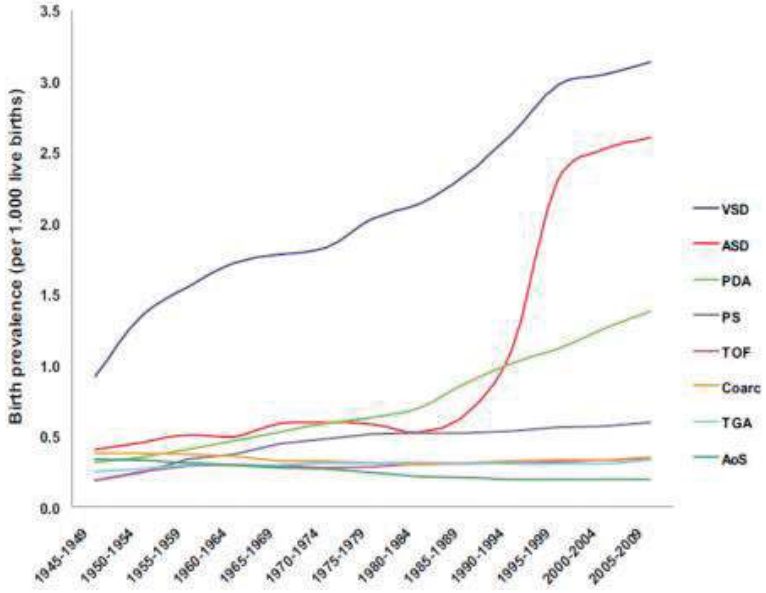

Figure 2. Prevalence of CHD between 1945 and 2009 according to defect classification. AoS, aortic stenosis; ASD, atrial septal defect; Coarc, coarctation of the aorta; PDA, patent ductus arteriosus; PS, pulmonary stenosis; TGA, transposition of the great arteries; TOF, tetralogy of Fallot; VSD, ventricular septal defect (source: [5]).

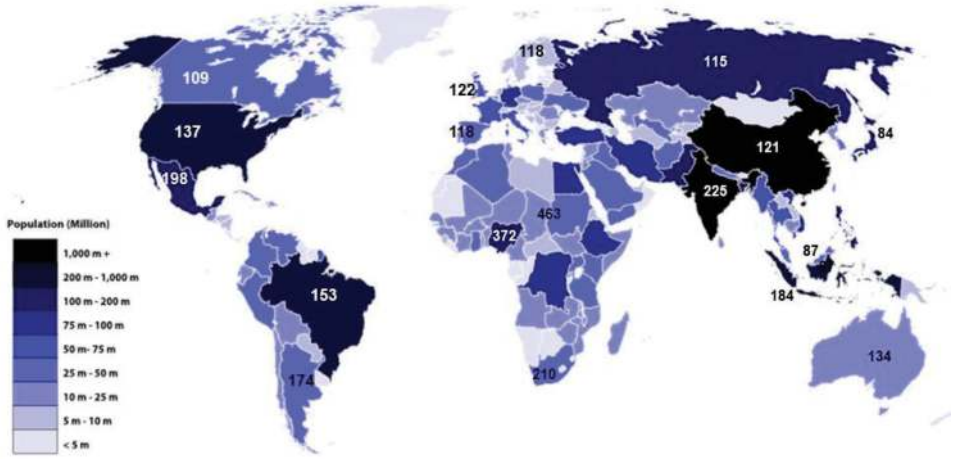

Figure 3. Global distribution of congenital heart disease in birth numbers per million population.

It was estimated that 48.9 million people had a congenital heart abnormality in 2015 [3] and CHD was responsible for 223,000 deaths in 2010 [4]. The incidence of birth defects has increased over time (**Figure 2**) partly due to the increased rates of fertility but also in part due to significant improvements in diagnostic modalities along with concomitant advances in anaesthesia and surgical intervention. The largest increase in incidence is in those born with atrial septal or ventricular septal defects.

The prevalence of CHD in the global population is illustrated in **Figure 3**. The incidence of CHD is similar between countries; however, those with higher rates of fertility have a disproportionately higher number of birth defects. Infant survival is poorest in developing countries often as a direct result of lack of medical care due to socioeconomic status. Future trends would suggest as socioeconomic conditions improve in such countries, an expected reduction in infant CHD mortality will follow.

2. Conclusion

In conclusion there is documented evidence of increasing prevalence of CHD births over the last century with a worldwide estimate of 9:1000 live births. This equates to 1.35 million annual CHD births, which has a major impact on healthcare and socioeconomic systems. The health burden falls mainly on those countries with low economic prospects, high fertility rates and poor access to advanced diagnostics and treatment options. Global healthcare monitoring and strategies to improve diagnosis and care in these areas are warranted.

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