Adult Congenital Heart Disease: Scope of the Problem

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KEYWORDS

• Adult • Congenital/epidemiology • Prevalence • Health services • Quality of care

KEY POINTS

- Birth prevalence of congenital heart disease (CHD) can be modified by many factors, including prenatal care, pregnancy termination and prevention, and changing sex distribution of the adult CHD population.
- As a result of decreasing mortality and increasing survival in all forms of CHD, the median age of patients has increased and adults now compose two-thirds of patients with CHD.
- Disease burden and resulting health services utilization increase significantly across the life span of the CHD population compared with the general population.
- Bridging the gap between policy and quality of care can be improved by referral to specialized adult CHD centers and planning delivery of specialized services that are commensurate with population needs, program accreditation criteria, and certified training of designated workforce.

INTRODUCTION

Congenital heart disease (CHD) lesions occur during embryonic development and consist of abnormal formations of the heart walls, valves or blood vessels. The adult congenital heart disease (ACHD) population is one of the fastest growing populations in cardiology. Contributing factors includes the improvement in CHD prenatal detection and treatment, novel surgical and interventional procedures, and the improvement in the organization of care. Echocardiography in clinical practice improves the ability to diagnose asymptomatic patients and patients with only mild lesions. These strides have resulted in rapidly changing demographics of those born with congenital lesions. Previously almost exclusively in the domain of pediatric cardiology, care delivery needs to be continuous across pediatric and adult health care systems.

This article is divided in 3 parts. The authors first review the epidemiology of CHD summarizing the impact of the changing epidemiology on the demographics of the CHD population. Then they review the impact of changing demographics on this population's disease burden across the life span and the resulting increase in health services utilization (HSU). Finally, they examine the progress in how as a field we are beginning to bridge the gap between policy and quality in order to improve outcomes in this patient population.

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PREVALENCE OF CONGENITAL HEART DISEASE ACROSS THE LIFE SPAN

Incidence and Birth Prevalence of Congenital Heart Disease

The product of CHD incidence and survival determines the prevalence of CHD at all ages. Thus a clear understanding of the determinants of the incidence of CHD is important in understanding the challenges of measurement using empirical data. The exact incidence of CHD cannot be accurately measured because it would require tracking the number of new cases of CHD from conception in utero. Thus, the best proxy available to estimate the incidence of new cases of CHD born each year is *birth prevalence*.[1] Reported birth prevalence rates of CHD vary widely depending on the lesions included and geographic world region where they were measured. In the United States, data from the Centers for Disease Control and Prevention (CDC) from 1998 to 2005 identified using the Metropolitan Atlanta Congenital Defects Program, an overall prevalence of 8.14 per 1000 births in 398,140 births of which 3240 had CHD. The most common CHDs were muscular ventricular septal defect, perimembranous ventricular septal defect, and secundum atrial septal defect.[2] The prevalence of tetralogy of Fallot, the most common cyanotic CHD, was twice that of transposition of the great arteries. The European Surveillance of Congenital Anomalies (EUROCAT) database is a population-based surveillance system for CHD based on more than 16 European countries.[3] These registry data were based on cases including live births, late fetal death/stillbirths, as well as terminations of pregnancy for fetal anomaly. The reported total CHD prevalence based on 26,598 cases of CHD was 8.0 per 1000 births (ranging across the countries between 5.36 and 15.32 per 1000 births), and live birth prevalence was 7.2 per 1000 births.[3]

A worldwide overview was provided in a recent systematic review of birth prevalence for the 8 most common CHD lesions through 2010.[4] The reported birth prevalence of CHD increased to 9.1 per 1000 live births after 1995.[4] The birth prevalence among different geographic areas and World Bank income groups was statistically significant. The reported total CHD prevalence was higher in Asia (9.3 per 1000 live births) compared with all other continents, including Africa. High-income countries consistently had higher CHD birth prevalence (8.0 per 1000 live births) compared with lower-middle-income countries (6.9 per 1000 live births).[4] These variations likely reflect variations in infant health surveillance systems and the availability of early diagnostic tools. There were also significant geographic changes among the prevalence of the CHD subtypes. Asia reported more pulmonary outflow obstructions and lower rates of transposition of the great arteries at birth prevalence compared with the other continents.[4]

Factors that can modify prevalence of CHD at birth are numerous. Prenatal care and pregnancy prevention and termination impact both measures and biologically mediated pathways of birth prevalence rates of CHD. In reports from the EUROCAT registry, perinatal mortality was 0.25 per 1000 live births. Pregnancy termination for fetal anomaly after prenatal diagnosis varied widely ranging between less than 0.3 and 1.1 per 1000 births.[3] Other factors, including mandatory folate supplementation during pregnancy, may also modify birth rates of CHD, in particular decreasing the birth rate of severe CHD. This finding has been observed in Quebec and in other jurisdictions.[5]

Sex Differences in Congenital Heart Disease Distribution and Outcomes

Sex differences in the incidence of CHD are well described. Of the most common CHD lesions, atrial septal defects have a higher frequency in females, whereas conotruncal anomalies, such as transposition of the great arteries, are more common in males.[6] Consistent with the finding that female babies are less likely to have severe CHD, a large US population study, showed that more male children underwent CHD surgery and had high-risk procedures, although female infants who had high-risk procedures were at higher risk of death. In Canada in more than 45,000 adults with CHD, women accounted for 57% of patients, a proportion that was significantly higher than the predominance of females observed in the general population.[7] Using death registry data in 11,040 adults in the United States, a study from the CDC showed lower mortality rates in women with CHD compared with men.[8] Potential causes of a shift in demographics toward a predominance of females in the ACHD population include milder lesions in females and/or differences in outcomes in adults. A protective effect of sex was demonstrated on in-hospital mortality in women between 18 and 45 years of age.[9] In a large European survey of adults with CHD, men were more likely to die of CHD than women. Although women had a 33% higher risk for pulmonary hypertension, they also had a 47% lower risk for endocarditis, a 55% lower risk of cardioverter-defibrillator implant, and a lower