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CONGENITAL HEART DISEASE: WORLDWIDE BURDEN AND REGIONAL PERSPECTIVES FROM SOUTH ASIA AND CENTRAL ASIA

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Abstract: Congenital heart disease (CHD) is the most frequent congenital malformation group and a major cause of infant illness and death across the world. Advances in imaging, cardiothoracic surgery, and intensive care have led to striking improvements in survival in high-income regions, resulting in a growing cohort of adults living with CHD. In contrast, outcomes in many low- and middle-income countries (LMICs)—including those in South Asia and Central Asia—remain suboptimal because of delayed diagnosis, scarce surgical capacity, and limited follow-up services. This review outlines current epidemiologic patterns of CHD, summarizes global trends, and examines region-specific issues and priorities in South Asia and Central Asia.

Keywords: Congenital, Heart, Disease, Burden, infant, South Asia, Central Asia

Introduction

Congenital heart disease refers to structural or functional abnormalities of the heart or great vessels that are present at birth, regardless of the timing of diagnosis[1]. The spectrum is broad: some defects are small and may close spontaneously, while others are complex lesions requiring staged interventions from early infancy.

Over recent decades, earlier detection, refined surgical and catheter-based techniques, and improved perioperative care have transformed CHD outcomes in high-income settings[2,3] . As more affected children reach adulthood, CHD has become a chronic condition rather than an invariably fatal one. At the same time, global population growth and uneven access to care mean that the total number of children and adults with CHD is increasing, especially in LMICs[4,5]. This article reviews epidemiologic data on CHD and considers global, South Asian, and Central Asian perspectives.

Classification

CHD can be categorized anatomically and physiologically[2]. Common lesion groups include:

• Septal defects: atrial septal defect (ASD), ventricular septal defect (VSD), atrioventricular septal defect (AVSD)

• Obstructive lesions: pulmonary stenosis, aortic stenosis, coarctation of the aorta

• Cyanotic defects: tetralogy of Fallot (TOF), transposition of the great arteries (TGA), truncus arteriosus, total anomalous pulmonary venous connection, tricuspid atresia

• Single-ventricle physiology: hypoplastic left heart syndrome and related complex variants. Multiple classification schemes exist (e.g. Mitchell, EUROCAT, ICD-based systems), and contemporary guidelines increasingly emphasize pathophysiology and long-term management needs rather than purely anatomic descriptions[3].

Global Epidemiology of Congenital Heart Disease

Birth prevalence

Recent systematic reviews indicate that global birth prevalence of CHD lies around 9–12 per 1,000 live births, with regional variation and clear effects of case-ascertainment methods[4].

• A large meta-analysis suggested a pooled prevalence of about 8.2 per 1,000 live births for moderate-to-severe CHD, higher when minor defects are included[6.]

• Apparent increases over time largely reflect better detection, especially of milder lesions. A cross studies, VSD, ASD, and patent ductus arteriosus (PDA) are generally the most frequently reported lesions, followed by pulmonary stenosis and TOF[7]. The distribution of specific defects varies by geography, diagnostic practices, and whether data derive from surgical, echocardiographic, or administrative sources.

Table 1. Approximate birth prevalence of CHD by region

Region / setting	Birth prevalence of CHD (per 1,000 live births)*
Global (all regions)[8]	~9–12/1,000 (all CHD); ~8.2/1,000 for moderate–severe CHD
High-income countries (N. America, W. Europe, parts of East Asia)[6,9]	Often 10–12/1,000 when including mild lesions
LMICs overall[9]	Frequently reported as 6–9/1,000, but true prevalence likely similar to HICs
South Asia (India, Pakistan, Bangladesh, Nepal, Sri Lanka, Bhutan, Maldives)[10,11]	Hospital-based estimates often 4–10/1,000 live births; population-based estimates sparse
Central Asia (Kazakhstan, Kyrgyzstan, Tajikistan, Turkmenistan, Uzbekistan)[15]	Where screening exists, reported ~8–10/1,000 in urban centres; likely lower in routine statistics

Table: Ranges are illustrative, based on meta-analyses and regional series; exact country-level estimates vary.

Mortality and overall burden

CHD is responsible for an estimated 260,000–300,000 deaths per year worldwide, most occurring in early life[5]. It is a leading cause of infant mortality linked to structural congenital anomalies. Global Burden of Disease (GBD) analyses show steep declines in CHD mortality in high-income settings, contrasted with more modest reductions in many LMICs. More than two-thirds of deaths from CHD are estimated to occur in LMICs[5].

Table 2. Approximate global and regional mortality burden from CHD

Region / income group	Annual CHD deaths (approx.)[5,8]	Share of global CHD deaths
Global total	~260,000–300,000 deaths/year	100%
High-income countries	Marked decline over recent decades; tens of thousands/year	Minority of global deaths
LMICs (all regions)	Majority of the 260,000–300,000 deaths	>66%
South Asia[4,5]	Large, poorly quantified share of global CHD deaths; some estimates suggest >25–30% of CHD mortality	Very high
Central Asia[10]	Smaller absolute number of deaths, but mortality proportionally higher than in HICs	Modest share, but important regionally

In high-income countries, advances in care have shifted the burden from early mortality to long-term morbidity among survivors; in many LMICs, substantial numbers of children still die without a diagnosis or before treatment becomes available[5,10].

Survival and the adult CHD population

Where comprehensive pediatric cardiac services exist, more than **85–90%** of newborns with CHD now survive into adulthood[9]. This has led to:

• A rapidly expanding adult CHD (ACHD) population, many of whom have repaired or palliated defects.

• New patterns of morbidity involving arrhythmias, heart failure, pulmonary hypertension, thromboembolism, and challenges related to pregnancy and employment[9].

Conversely, in many LMICs, survival gains are smaller, and the adult CHD population is smaller in absolute terms but significantly under- recognized and under- served[10].

Table 3. Approximate survival to adulthood among individuals born with CHD

Setting	Proportion of CHD births surviving to adulthood[12,13]
High- income countries	>85–90% in recent birth cohorts
LMICs overall	Substantially lower; varies widely by country and lesion complexity
South Asia	No robust population- level figures; survival highly centre- dependent
Central Asia	Intermediate survival between HICs and lowest- resource settings

Etiology and Risk Factors

Most CHD arises from multifactorial interactions between genes and environment[11]. Well- known risk factors include:

• Genetic and chromosomal anomalies: trisomy 21, Turner syndrome, 22q11.2 deletion, and numerous monogenic conditions.

• Maternal comorbidities: pregestational diabetes mellitus, phenylketonuria, obesity, and autoimmune diseases.

• Maternal infections and exposures: rubella, uncontrolled hyperglycemia, teratogenic drugs, alcohol, and possibly certain environmental toxins.

• Family history: higher recurrence risk in siblings and in offspring of affected individuals.

Nonetheless, a clear aetiologic explanation is identified in only a minority of cases[11]. Rapid advances in genomic technologies continue to uncover CHD- associated variants and pathways, but such tools are often unavailable in resource- constrained settings.

South Asia Perspective

South Asia (India, Pakistan, Bangladesh, Nepal, Sri Lanka, Bhutan, Maldives) accounts for a substantial proportion of global live births and therefore a high absolute number of children with CHD[12]. Even with prevalence similar to global averages, the regional burden is large.

Epidemiology and patterns of disease

Comprehensive population-based surveillance is limited, but available reports suggest birth prevalence estimates comparable to those elsewhere, with under-ascertainment likely[13,14]. Hospital-based series consistently show:

• Dominance of VSD, ASD, and PDA among diagnosed lesions, followed by TOF, pulmonary stenosis, and AVSD[14].

• Frequent presentation at advanced stages, such as severe pulmonary hypertension in longstanding left-to-right shunts.

CHD is recognized as an important contributor to infant and neonatal mortality from congenital anomalies, though exact figures are uncertain because of data gaps[15].

Table 4. Selected CHD epidemiology indicators in South Asia (illustrative)

Country	Reported CHD birth prevalence*	Most common lesions[12–14]
India[12,13]	Hospital-based: ~4–8/1,000 live births; population-based estimates scarce	VSD, ASD, PDA; TOF most common cyanotic lesion
Bangladesh[14]	Tertiary hospital series: CHD in ~9–12 per 1,000 hospital live births	VSD, ASD, PDA; TOF, PS, AVSD
Pakistan, Nepal, Sri Lanka	Limited population-level data; small studies suggest similar prevalence	Similar pattern: septal defects predominating; TOF common cyanotic lesion

**Values are approximate ranges from hospital and registry studies; true population prevalence likely closer to global estimates when mild lesions and undetected early deaths are considered.*

Table 5. CHD burden and services in Central Asia (indicative)

Country	CHD birth prevalence (where reported)*	Service organization
Kazakhstan	Limited data; local series suggest ~8–10/1,000 with systematic screening	National cardiac centres in major cities; some paediatric subspecialization
Kyrgyzstan, Tajikistan	Sparse data; presumed similar to global averages	Care concentrated in 1–2 main centres per country

Turkmenistan, Uzbekistan	Very limited published prevalence data	Historically adult-focused cardiac surgery centres now treating children

Where systematic screening has been conducted; otherwise inferred from global ranges.

Prevention, Screening, and Early Detection

Primary prevention

Long-Term Outcomes and Adult Congenital Heart Disease

As survival improves, ACHD has become an established subspecialty[2,3,9]. Long-term issues include:

• Residual or recurrent lesions and the need for re-interventions.

• Development of heart failure, arrhythmias, and pulmonary hypertension.

• Management of pregnancy and contraception in women with CHD.

• Education, employment, psychosocial well-being, and quality of life.

Guidelines from professional societies advocate life-long, lesion-specific follow-up in specialized ACHD centers[3] . In many parts of South and Central Asia, however, adult follow-up is often fragmented: patients may be lost to follow-up or managed in general cardiology clinics without dedicated ACHD expertise[15,16]. Structured transition programs from pediatric to adult care are still uncommon.

Conclusion

Congenital heart disease remains a major contributor to global child mortality and a growing cause of chronic cardiovascular morbidity. While many high-income countries now achieve excellent survival, substantial numbers of children in South Asia, Central Asia, and other LMICs still die without diagnosis or access to definitive treatment. At the same time, the rising population of survivors places new demands on health systems for life-long, specialized care.

Closing these gaps will require reliable data, expanded pediatric and adult CHD capacity, strengthened financial protection, and integration of CHD into broader maternal-child health and non-communicable disease agendas. With sustained commitment and collaboration, it is feasible to achieve major gains in survival and quality of life for children and adults with CHD in South Asia, Central Asia, and worldwide.

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