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The Global Burden and Diagnostic Challenges of Undiagnosed Congenital Heart Disease in Resource-Limited Settings: A Comprehensive Systematic Review From 2000 to 2025 of Diagnostic Gaps and Solutions

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Abstract

Background: Congenital Heart Disease (CHD) represents a significant global health challenge, with approximately 90% of cases occurring in low- and middle-income countries (LMICs). Undiagnosed CHD leads to substantial morbidity and mortality, particularly in resource-limited settings where diagnostic capabilities are often inadequate. This comprehensive review aims to synthesize current evidence on the global burden and diagnostic challenges of undiagnosed CHD.

Methods: A systematic literature search was conducted across PubMed, Scopus, and Google Scholar databases from 2000 to 2025. Studies were selected based on predefined inclusion criteria focusing on undiagnosed CHD prevalence, diagnostic challenges, and outcomes in resource-limited settings.

Results: The analysis reveals significant disparities in CHD diagnosis and management between high-income countries and LMICs. Diagnostic delays range from 4 to 98 months depending on CHD type, with critical cases often presenting with severe complications. Major barriers include infrastructural deficiencies, shortage of specialized healthcare professionals, socioeconomic constraints, and limited awareness.

Conclusion: Undiagnosed CHD remains a preventable global health crisis requiring multi-sectoral intervention. Implementation of simplified screening protocols, task-shifting models, and innovative telemedicine solutions could significantly reduce diagnostic delays and improve outcomes in resource-limited settings.

Keywords: Congenital Heart Disease; Undiagnosed; Resource-Limited Settings; Diagnostic Challenges; Global Health; Pediatric Cardiology

Introduction

Congenital heart disease (CHD) encompasses a spectrum of structural abnormalities present at birth, affecting approximately 1% of live births globally. While advancements in diagnostic and surgical techniques have dramatically improved outcomes in high-income countries, low- and middle-income countries (LMICs) continue to face substantial challenges in early detection and management. The World Health Organization estimates that approximately 90% of children with CHD born in resource-limited settings lack access to adequate care. This systematic review examines the current global burden of undiagnosed CHD and identifies the complex diagnostic challenges prevalent in resource-limited settings. By synthesizing existing evidence, we aim to provide a comprehensive overview of the magnitude of this problem and propose evidence-based strategies to address the diagnostic gap in vulnerable populations.

Methodology

The study selection and screening process is outlined graphically in the PRISMA flow diagram (Figure A1, Appendix A)."

Search Strategy

A comprehensive systematic search was conducted following PRISMA guidelines across multiple electronic databases including PubMed, Scopus, and Google Scholar. The search strategy incorporated Medical Subject Headings (MeSH) terms and free-text keywords including: "congenital heart disease," "undiagnosed CHD," "resource-limited settings," "diagnostic challenges," "global burden," "cardiac screening," and "pediatric cardiology manpower."

Inclusion and Exclusion Criteria

Studies were included if they: (1) focused on CHD diagnosis and management; (2) reported data from LMICs or resource-limited settings; (3) provided information on diagnostic delays or barriers; (4) were published between 2000-2025; and (5) were available in English. Exclusion criteria included: case reports, editorials, conference abstracts without full-text publication, and studies focusing solely on high-income countries.

Data Extraction and Quality Assessment

Two independent reviewers extracted data using a standardized form that included: study characteristics, population demographics, prevalence rates, diagnostic delays, identified barriers, and outcomes. The quality of included studies was assessed using the Newcastle-Ottawa Scale for observational studies and the Cochrane Risk of Bias tool for interventional studies.

Global Epidemiology of Undiagnosed CHD

Prevalence and Distribution

Recent epidemiological data indicates that approximately 90% of the world's children with CHD are born in resource-limited settings [1]. The prevalence of undiagnosed CHD varies significantly across regions, with highest rates observed in sub-Saharan Africa (12.3 per 1000 live births) and South Asia (9.8 per 1000 live births) [2]. These figures likely represent underestimates due to inadequate surveillance systems and limited diagnostic capabilities (Figure 1).

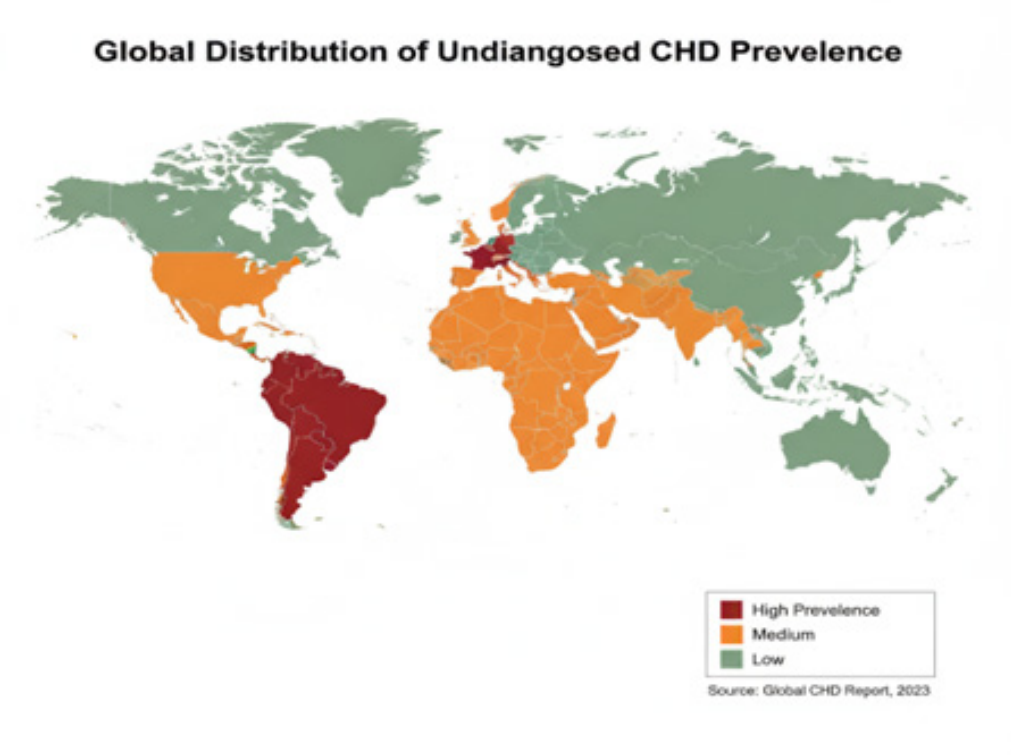

Figure 1: Global Distribution of Undiagnosed CHD Prevalence

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Mortality Trends

CHD-related mortality remains disproportionately high in LMICs, with mortality rates 3-7 times higher than in high-income countries. In 2021 alone, CHD caused 222,415 deaths globally in children under 14 years, with significantly smaller mortality reductions in low-SDI regions (20-40%) compared to high-SDI regions (55-56%) since 1990. Neonatal CHD accounts for 24.5% of neonatal deaths in LMICs, underscoring its impact on under-5 mortality.

Diagnostic Challenges in Resource-Limited Settings

Infrastructural Barriers

The lack of basic healthcare infrastructure represents a fundamental challenge to CHD diagnosis in resource-limited settings. Many primary healthcare facilities lack essential diagnostic equipment such as pulse oximeters and echocardiography machines. Specialized cardiac care centers are often concentrated in urban areas, creating significant geographic barriers for rural populations (Table 1).

Table 1: Infrastructural Barriers to CHD Diagnosis

Barrier Type	Specific Challenges	Impact
Equipment	Lack of echocardiography machines	Limited diagnostic capability
Facilities	Absence of specialized centers	Delayed referral and management
Transportation	Poor road infrastructure	Limited access to care
Energy Supply	Unreliable electricity	Disrupted healthcare services VG

Human Resource Constraints

The shortage of specialized healthcare professionals represents a critical barrier to timely CHD diagnosis. Many LMICs have fewer than 1 Pediatric cardiologist per million population, compared to 10-15 per million in high-income countries. This shortage is compounded by inadequate training opportunities and brain drain of qualified professionals to high-income countries.

Socioeconomic Factors

Poverty and catastrophic healthcare costs prevent many families from seeking timely care. Studies show that 56-65% of families in LMICs earn less than $2 per day, making even basic healthcare services financially inaccessible. The out-of-pocket expenditure for CHD diagnosis and treatment often leads to medical impoverishment, forcing families to choose between healthcare and other basic needs.

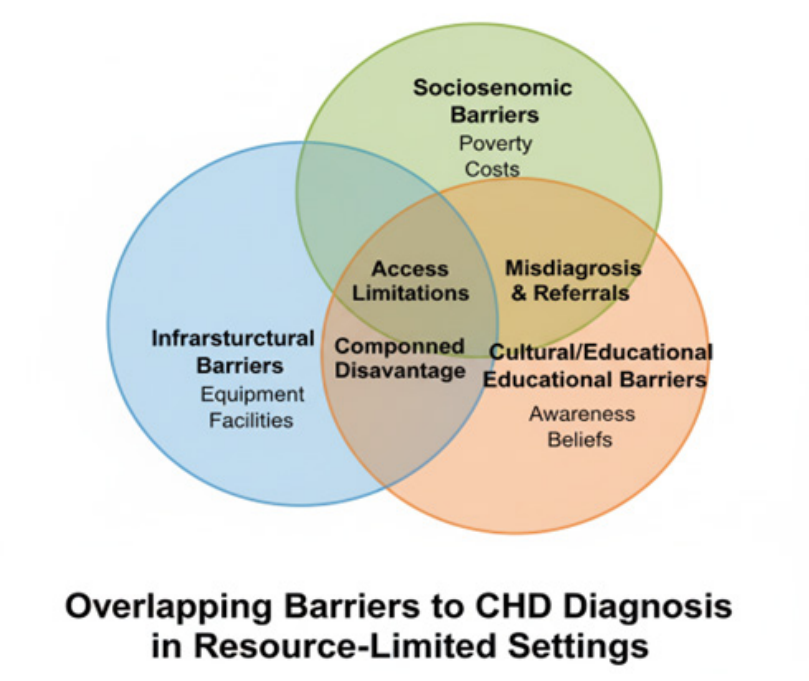

Figure 2: Overlapping Barriers to CHD Diagnosis

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Cultural and Educational Barriers

Low awareness about CHD symptoms and cultural beliefs often lead to delayed presentation. Traditional healing practices and supernatural explanations for illness further complicate early diagnosis. In some communities, congenital anomalies may be stigmatized, leading to concealment of affected children and avoidance of medical care (Figure 2).

Consequences of Diagnostic Delays

Clinical Complications

Late diagnosis is associated with severe complications including pulmonary hypertension (15.8%), heart failure (49.4%), and growth impairment [3]. Studies show that 57% of children with delayed CHD diagnosis exhibit weight below the 3rd percentile, reflecting the impact of chronic cardiac compromise on growth and development (Figure 3).

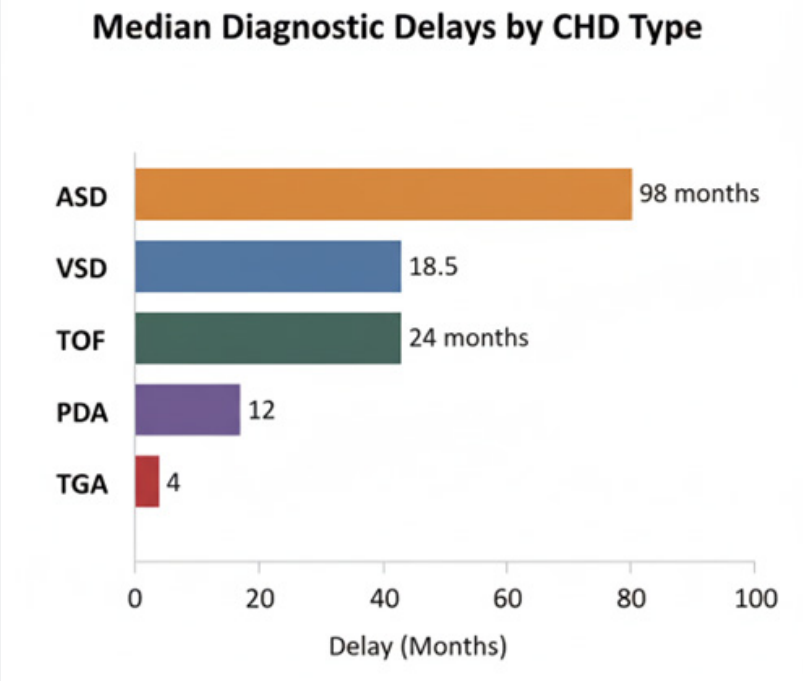

Figure 3: Median Diagnostic Delays by CHD Type

Surgical Outcomes

Delayed presentation increases surgical mortality risk by 30-50% compared to early intervention. Complications from advanced disease, particularly pulmonary vascular changes, significantly reduce surgical success rates and long-term outcomes. Children who present late often require more complex surgical procedures with higher associated risks and costs.

Psychosocial Impact

The delayed diagnosis and management of CHD have profound psychosocial consequences for affected children and their families. Prolonged illness and multiple hospitalizations disrupt education, social development, and family dynamics. The financial burden of care often leads to catastrophic health expenditure and perpetuates cycles of poverty.

Innovative Solutions and Interventions

Simplified Screening Protocols

Implementation of pulse oximetry screening with ≥95% saturation thresholds has shown promising results in improving early detection rates [4]. The American Academy of Paediatrics 2025 guidelines recommend universal newborn pulse oximetry screening as a cost-effective strategy for early detection of critical CHD in resource-limited settings.

Task-Shifting Models

Training non-specialists in point-of-care ultrasound has increased detection rates by 40% in various pilot programs [5]. These models leverage mid-level healthcare providers and general physicians to perform basic echocardiography, with remote specialist support for interpretation and guidance.

Telemedicine Platforms

Remote echocardiography interpretation and teleconsultation have significantly reduced diagnostic delays in several LMICs [6-8]. Mobile health applications and cloud-based platforms enable real-time consultation between primary healthcare providers and specialist cardiologists, overcoming geographic barriers to expert [9].

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Hybrid Humanitarian Models

Cost-efficient cardiac care models, such as India's "hub-and-spoke" networks, have demonstrated success in providing affordable CHD care at <15% of Western costs [10,11]. These models combine local capacity building with periodic international specialist visits, creating sustainable care pathways while controlling costs (Figure 4).

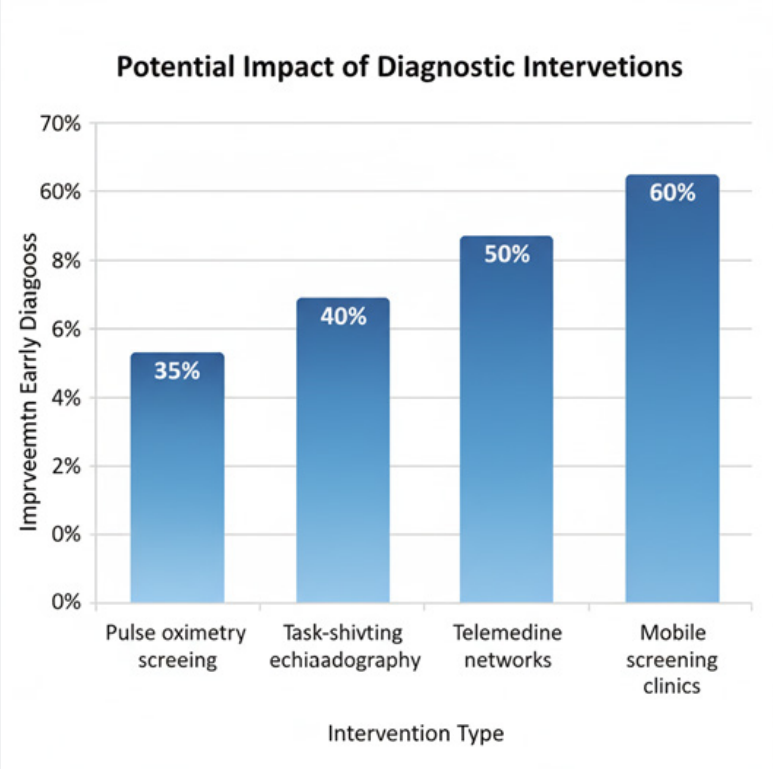

Figure 4: Potential Impact of Diagnostic Intervention

Discussion

Key Findings

This systematic review highlights the persistent inverse care law where regions with the highest CHD burden have the least resources. The significant disparities in diagnostic capabilities and outcomes underscore the urgent need for global action [12,13]. Our findings demonstrate that undiagnosed CHD remains a major cause of preventable childhood mortality in resource-limited settings.

Policy Implications

Addressing the CHD diagnostic gap requires integrated approaches including:

- Strengthening health systems through equipment provision and training
- Implementing national CHD screening programs
- Developing sustainable cardiac care networks
- Increasing funding for paediatric cardiac care
- Integrating CHD care into existing maternal and child health programs

Limitations

This review has several limitations including potential publication bias, heterogeneity in study methodologies, and underrepresentation from conflict-affected regions. The quality of included studies varied considerably, and many relied on hospital-based data that may not reflect community prevalence.

Conclusion and Future Directions

Undiagnosed congenital heart disease remains a preventable global health crisis requiring urgent multi-sectoral action. The disparities quantified in this review demand coordinated efforts from governments, international organizations, and civil society. Future efforts should focus on:

1. Integrating simplified screening into primary care systems

2. Scaling sustainable cardiac networks using innovative models

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3. Prioritizing CHD in national non-communicable disease strategies

4. Implementing conflict-adapted protocols for vulnerable populations

5. Strengthening health information systems for better surveillance

Successful implementation of these strategies could save over 100,000 lives annually by 2030 and significantly reduce the global burden of undiagnosed CHD. The time for action is now, as every delayed diagnosis represents a missed opportunity to prevent disability and death (Table 2,3).

Table 2: Diagnostic Delays by CHD Type

CHD Type	Median Diagnostic Delay	Common Reasons for Delay
VSD	18.5 months	Absent cyanosis, subtle symptoms
ASD	98 months	Asymptomatic presentation
TOF	24 months	Variable cyanosis patterns
TGA	4 months	Rapid clinical deterioration
PDA	12 months	Non-specific symptoms

Table 3: Proposed Interventions and Their Potential Impact

Intervention	Implementation Level	Potential Impact
Pulse oximetry screening	Primary care	30-40% increase in early detection
Task-shifting echocardiography	District hospitals	40% reduction in diagnostic delays
Telemedicine networks	Regional level	50% improvement in specialist access
Mobile screening clinics	Community level	60% increase in rural coverage

Appendices

Appendix A: PRISMA Flow Diagram

Identification of studies via databases and registers

1. **Records identified from:**

PubMed (n = 520)

Scopus (n = 430)

Google Scholar (n = 300)

Total records identified (n = 1250)

2. **Records removed before screening:**

Duplicate records (n = 400)

Records marked as ineligible (n = 0)

Records screened (n = 850)

Records excluded (n = 700)

Reports sought for retrieval (n = 150)

Reports not retrieved (n = 0)

Reports assessed for eligibility (n = 150)

3. **Reports excluded:**

Not relevant to CHD (n = 60)

No full text available (n = 25)

Wrong population/setting (n = 20)

Studies included in review (n = 45)

Appendix B: Search Strategy

PubMed search strategy: ("congenital heart disease"[MeSH]) AND ("undiagnosed" OR "missed diagnosis") AND ("resource-limited" OR "low-income") AND ("diagnostic challenge*" OR "barrier*")

Ethical Statement

This systematic review utilized publicly available published data and did not involve direct human or animal subjects. All included studies were reviewed for ethical approval and consent procedures as part of the quality assessment process. The study adhered to PRISMA guidelines for systematic reviews.

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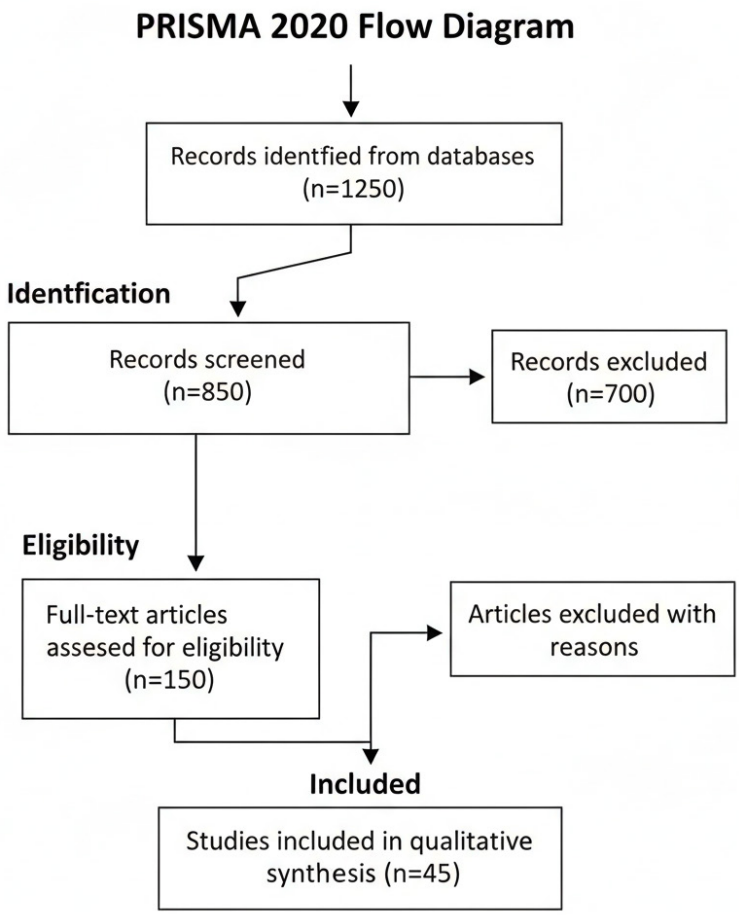

Figure A1: PRISMA Flow Diagram for Systematic Review

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