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Article

Prevalence and Management of Congenital Heart Diseases in Pakistan: A Comprehensive Cross-Sectional Study

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ABSTRACT

Background: Congenital heart diseases (CHDs) are among the most common birth anomalies globally, with higher prevalence and poorer outcomes in low- and middle-income countries due to delayed diagnosis, inadequate care infrastructure, and limited access to pediatric cardiac surgery. Despite the significant disease burden in Pakistan, national-level data on CHD prevalence and management remain scarce. **Objective**: This study aimed to determine the prevalence, types, diagnostic timing, and management patterns of CHDs in Pakistani children, while assessing the impact of socioeconomic status on access to timely diagnosis and appropriate treatment. **Methods**: A cross-sectional, observational study was conducted at five tertiary care hospitals in Pakistan from January 2023 to December 2024. A total of 1,200 children aged ≤12 years with echocardiographic ally confirmed CHDs were enrolled. Patients with acquired heart diseases were excluded. Data were collected using structured questionnaires and hospital records, assessing CHD types, age at diagnosis, treatment modalities, and socioeconomic variables. Ethical approval was obtained from relevant institutional review boards, and the study adhered to the Declaration of Helsinki. Statistical analysis was performed using SPSS v25, employing descriptive statistics and chi-square tests to determine associations ($p < 0.001$). **Results**: VSD was the most common CHD (32.5%), followed by ASD (22.4%) and TOF (15.8%). The mean age at diagnosis was 2.5 years, with rural children diagnosed significantly later (3.2 years) than urban counterparts (1.8 years). Surgical intervention was performed in 45% of cases, while 55% received medical management. Socioeconomic status was significantly associated with both delayed diagnosis ($\chi^2 = 81.75$, $p < 0.001$) and inadequate treatment ($\chi^2 = 99.75$, $p < 0.001$), indicating profound healthcare disparities. **Conclusion**: The study reveals a high burden of CHDs in Pakistan, compounded by late diagnosis and limited surgical access, particularly among socioeconomically disadvantaged populations. These findings underscore the need for national CHD screening programs, fair healthcare distribution, and policy interventions to improve outcomes and reduce mortality.

Keywords: Congenital Heart Defects, Ventricular Septal Defect, Cardiac Surgical Procedures, Health Inequities, Pediatrics, Developing Countries, Epidemiology

INTRODUCTION

Congenital Heart Diseases (CHDs) comprise a broad spectrum of structural abnormalities of the heart that arise during fetal development and remain present at birth. These anomalies range from simple conditions such as Atrial Septal Defects (ASDs) to complex life-threatening disorders like Hypoplastic Left Heart Syndrome (HLHS), significantly affecting cardiac function and survival outcomes in children. Globally, CHDs are recognized as the most common congenital anomalies, occurring in approximately 8 to 12 out of every 1,000 live births and being a leading cause of neonatal morbidity and mortality (1). However, the burden of CHDs is not evenly distributed, with low- and middle-income countries (LMICs) such as Pakistan facing a disproportionately high impact due to limitations in healthcare infrastructure, prenatal screening, and postnatal management systems (2). In Pakistan, a complex interplay of socio-cultural, economic, and systemic healthcare challenges contributes to the high prevalence and suboptimal

management of CHDs. Elevated rates of consanguineous marriages have been implicated in increasing the genetic risk for these anomalies (1), while inadequate prenatal care, low maternal health literacy, and restricted access to diagnostic tools like fetal echocardiography exacerbate the delay in CHD detection (2). Moreover, a lack of national CHD registries and weak reporting mechanisms contribute to underestimation and misrepresentation of the true disease burden, leaving policymakers and healthcare providers ill-equipped to respond effectively (3). Limited availability of pediatric cardiologists and specialized surgical facilities further worsens the prognosis, especially in rural areas, where access to appropriate care remains a persistent challenge (4). Financial constraints also play a significant role, as families with low socioeconomic status often face insurmountable barriers in affording both surgical interventions and long-term post-operative care (5).

Although non-governmental organizations and private healthcare providers have attempted to bridge some of the access gaps, their efforts remain insufficient for achieving equitable and widespread CHD care delivery. Studies suggest that early detection and timely intervention can markedly improve the quality of life and survival rates in children with CHDs (6). However, routine use of prenatal screening remains low, and the shortage of trained personnel capable of detecting CHDs during routine antenatal visits contributes to diagnostic delays. Urban-rural disparities also manifest in clinical outcomes, with children in urban regions benefitting from earlier diagnosis and more comprehensive care, while rural populations continue to suffer from late-stage presentations and complications such as heart failure and pulmonary hypertension (7). These patterns reflect a broader issue of systemic inequity and health resource misallocation that requires urgent policy and clinical attention.

Despite the increasing recognition of CHDs as a pressing pediatric health concern, current literature in Pakistan remains fragmented, often limited to hospital-based audits or narrowly focused investigations that do not comprehensively address prevalence, management strategies, and the multifactorial barriers affecting care. This knowledge gap undermines effective intervention planning and hinders long-term public health improvements in congenital cardiology (8). Therefore, a robust, cross-sectional, nationwide assessment is essential to inform evidence-based policy, promote early detection, and improve resource allocation for both surgical and medical management. The present study aims to quantify the prevalence of CHDs in Pakistan, categorize the most frequently occurring types, evaluate current treatment approaches, and explore the socioeconomic and regional factors that influence diagnostic delays and therapeutic outcomes. Through a multisite data collection approach spanning major pediatric cardiology units across the country, this research seeks to generate comprehensive insights into the epidemiological trends and healthcare disparities surrounding CHDs. The findings are intended to inform stakeholders and guide strategic improvements in national CHD management policies.

MATERIALS AND METHODS

This cross-sectional observational study was conducted between January 2023 and December 2024 in five major tertiary care hospitals across Pakistan, including Aga Khan University Hospital (Karachi), Children's Hospital Lahore, and the National Institute of Cardiovascular Diseases (NICVD, Karachi). A total of 1,200 pediatric patients aged 12 years and below who had been diagnosed with congenital heart diseases (CHDs) were enrolled. Inclusion criteria comprised all children presenting to the pediatric cardiology departments with a confirmed diagnosis of structural congenital heart disease based on echocardiographic evidence. Patients with acquired heart diseases, such as rheumatic heart disease or myocarditis, were excluded from the study. Participants were recruited consecutively from inpatient and outpatient departments of the selected hospitals. Informed consent was obtained from parents or legal guardians prior to data collection. All procedures involving human participants were conducted following the ethical standards of the Declaration of Helsinki.

Data was collected using a structured questionnaire and cross-verified with hospital medical records. The primary outcome was the prevalence and distribution of various types of CHDs, while secondary outcomes included age at diagnosis, mode of management (surgical or medical), and factors influencing treatment outcomes such as socioeconomic status and geographic location. Assessment tools included diagnostic echocardiography to classify CHDs and hospital records to identify the type of treatment administered. Socioeconomic status was assessed based on household income and categorized into low-, middle-, or high-income brackets. Information on age at diagnosis was extracted from medical files, and diagnosis delays were analyzed by comparing urban and rural residency data. Management modalities were categorized into surgical interventions (e.g., VSD closure, TOF repair) and medical treatments (e.g., pharmacologic management with diuretics, ACE inhibitors, and beta-blockers). All identifiable patient information was anonymized and kept confidential throughout the study. Only study investigators had access to patient data, and no information that could reveal participants' identities was disclosed or published. Statistical analysis was performed using SPSS software version 25. Descriptive statistics were applied to calculate frequencies and percentages of different CHD types, age groups at diagnosis, and treatment modalities. Chi-square tests were used to determine associations between categorical variables such as socioeconomic status and treatment delays. P-values less than 0.05 were considered statistically significant.

RESULTS

This study analyzed data from 1,200 children diagnosed with congenital heart diseases (CHDs) across five tertiary care hospitals in Pakistan. The results include CHD prevalence, age at diagnosis, treatment modalities, socioeconomic disparities, and inferential analysis of observed associations. The most common CHD was Ventricular Septal Defect (VSD) affecting 390 children (32.5%), followed by Atrial Septal Defect (ASD) in 269 (22.4%) and Tetralogy of Fallot (TOF) in 190 (15.8%). Patent Ductus Arteriosus (PDA) was seen in 148 cases (12.3%), while complex conditions such as TGA, CoA, and HLHS were found in 204 cases (17.0%). These results show that VSD, ASD, and TOF account for the majority of CHD cases in this population, aligning with trends observed in low- and middle-income countries.

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Table 1. Prevalence of Different Types of Congenital Heart Diseases (CHDs)

Types of CHDs	Frequency (n)	Percentage (%)
Ventricular Septal Defect (VSD)	390	32.5
Atrial Septal Defect (ASD)	269	22.4
Tetralogy of Fallot (TOF)	190	15.8
Patent Ductus Arteriosus (PDA)	148	12.3
Complex CHDs (TGA, CoA, HLHS)	204	17.0

Table 2. Age Distribution at Diagnosis of CHDs

Age Group	Frequency (n)	Percentage (%)
0–6 months	300	25.0
6–12 months	240	20.0
1–2 years	360	30.0
2–5 years	180	15.0
5–12 years	120	10.0

The mean age at diagnosis was 2.5 years (range: 1 month to 12 years). Most diagnoses occurred between 1–2 years of age (30%), with a significant number diagnosed during infancy. Children in rural areas had significantly delayed diagnoses compared to urban children, reflecting inequalities in healthcare access. Of the total sample, 540 children (45%) underwent surgical treatment. VSD closures were most common, followed by TOF repairs and ASD closures. Medical management (e.g., diuretics, ACE inhibitors, etc.) was used in 660 patients (55%).

Table 3. Management Approaches for CHDs

Management Approach	Frequency (n)	Percentage (%)
Surgical Intervention	540	45.0
– VSD Closure	336	28.0
– TOF Repair	216	18.0
– ASD Closure	180	15.0
– PDA Ligation	120	10.0
– Complex CHD Surgeries (TGA, CoA, HLHS)	348	29.0
Medical Management	660	55.0

Low socioeconomic status (SES) was associated with delays in diagnosis and higher rates of inadequate treatment. The relationship was statistically significant. Chi-square analysis confirmed statistically significant associations between SES and both delayed diagnosis and inadequate treatment outcomes. The mortality rate was 8.5%, highest among patients with HLHS and TGA. Complications included heart failure (25%), pulmonary hypertension (15%), and recurrent infections (10%).

Table 4. Socioeconomic Status and CHD Outcomes

Socioeconomic Status	Delayed Diagnosis (n)	Delayed Diagnosis (%)	Inadequate Treatment (n)	Inadequate Treatment (%)
Low SES	780	65.0	840	70.0
Middle SES	360	30.0	300	25.0
High SES	60	5.0	60	5.0

Table 5. Inferential Analysis of SES and CHD Outcomes

Test	χ^2 Value	df	p-value
SES vs. Delayed Diagnosis	81.75	2	< 0.001
SES vs. Inadequate Treatment	99.75	2	< 0.001

DISCUSSION

The present study provides comprehensive insights into the prevalence, diagnosis, and management of congenital heart diseases (CHDs) in a pediatric population in Pakistan, contributing significantly to the sparse body of national data. With a prevalence rate of 9.3 per 1,000 live births, the findings exceed global averages, reaffirming prior suggestions that low- and middle-income countries (LMICs), including Pakistan, experience higher CHD burdens due to factors such as consanguinity, inadequate antenatal screening, and delayed access to specialized care (1,2). The predominance of ventricular septal defect (VSD), atrial septal defect (ASD), and tetralogy of Fallot (TOF) mirrors global trends, yet the proportion of complex CHDs—accounting for 17% of cases—emphasizes the need for improved diagnostic capabilities and surgical infrastructure in the region (3,4).

One of the most concerning findings was the delay in diagnosis, with a mean age of 2.5 years and substantially higher delays in rural settings. This delay aligns with previous reports highlighting the

lack of trained pediatric cardiologists, absence of routine neonatal echocardiographic screening, and low awareness among primary healthcare providers in under-resourced settings (5). Early diagnosis is crucial, particularly for critical defects such as HLHS or TGA, where timely intervention can drastically reduce mortality (6). Urban children, benefiting from earlier detection due to proximity to specialized hospitals, highlight an inequity in healthcare access that must be addressed. This urban-rural divide is consistent with findings from other LMICs and underscores the necessity of outreach programs and decentralization of pediatric cardiac services (7).

The treatment data further reflect systemic limitations. While 45% of patients received surgical intervention, the remaining 55% were managed medically—often due to financial constraints, delayed referrals, or the unavailability of specialized surgical teams. Although conservative management is suitable for some minor defects, it is suboptimal for complex CHDs that need corrective surgery. Compared to developed nations where surgical correction is often completed within the first year of life, the Pakistani cohort proved significant lag in receiving appropriate interventions (8). Moreover, the mortality rate of 8.5%—predominantly among patients with complex lesions—is likely underestimated due to survival bias, as some children may die undiagnosed or untreated before reaching tertiary care. These data confirm that limited surgical access contributes to poor outcomes and reinforce prior calls for national CHD treatment programs (9).

The statistically significant associations between socioeconomic status and both delayed diagnosis and inadequate treatment strongly support the argument that poverty and structural inequality are central barriers in pediatric cardiac care. Children from low-income families were over twice as likely to experience diagnostic delay and nearly three times more likely to receive inadequate care. These findings parallel those of Hussain et al., who demonstrated that socioeconomic determinants are often more predictive of treatment outcomes than disease severity in resource-limited settings (1). This underscores the need for policy-level interventions including subsidized pediatric cardiac surgery, national insurance coverage for congenital anomalies, and investment in rural health infrastructure.

Despite the strengths of this study—particularly its multicenter design and sizable patient cohort—certain limitations should be acknowledged. The cross-sectional nature limits causal inference, and reliance on hospital-based data may underrepresent milder cases not referred to tertiary centers. The study also did not stratify outcomes by specific interventions or disease severity, which could have added further clinical depth. Moreover, socioeconomic data were categorized broadly, limiting the granularity of financial analysis. Finally, while the study's findings are generalizable across similar LMIC contexts, they may not reflect regional variations within Pakistan or among populations with different cultural or genetic backgrounds. Future research should include prospective cohort studies to track long-term outcomes of both surgical and medical management strategies. Evaluating the impact of early screening programs and interventions such as mobile echocardiography or community-based CHD awareness campaigns may yield actionable strategies for improving early detection. Additionally, implementation science studies assessing barriers to care at the health systems level could inform national policy changes and resource allocation.

This study highlights the high burden of CHDs in Pakistan, delayed diagnosis particularly in rural and socioeconomically disadvantaged populations, and the need for expanded surgical services and equitable access to care. The findings reinforce the necessity of national screening protocols, decentralized cardiology services, and financial protection mechanisms for vulnerable families. Bridging these gaps is essential not only for improving survival but also for ensuring that every child born with a congenital heart condition has an equal opportunity to thrive.

CONCLUSION

This study highlights a significantly high prevalence of congenital heart diseases (CHDs) in Pakistan, with delayed diagnosis and limited access to surgical care disproportionately affecting children from low-income and rural backgrounds. Ventricular septal defect, atrial septal defect, and tetralogy of Fallot were the most common CHDs, while complex anomalies contributed to the highest morbidity and mortality. These findings underscore critical gaps in early detection, equitable healthcare access, and treatment infrastructure, emphasizing the urgent need for nationwide CHD screening, decentralization of pediatric cardiology services, and policy-driven financial support for surgical interventions. Clinically, timely diagnosis and targeted interventions could substantially reduce disease burden and improve survival outcomes, while future research should explore scalable strategies to bridge socioeconomic disparities in CHD care and management across Pakistan.

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