ORIGINAL ARTICLE

SPECTRUM OF CONGENITAL HEART DISEASES IN EARLY CHILDHOOD (1–60 MONTHS): INSIGHTS FROM A SUB-HIMALAYAN TERTIARY CARE CENTRE

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ABSTRACT

Background: Congenital heart disease (CHD) is a leading cause of birth defect-related mortality, with delayed diagnosis common in resource-limited regions. In India, gaps in CHD detection and care persist, especially in remote areas like Himachal Pradesh. This study evaluates the spectrum and outcomes of CHD in early childhood at a Sub-Himalayan tertiary care centre.

Objective: To study the spectrum and outcomes of congenital heart disease (CHD) in children aged 1 month to 5 years in a tertiary hospital of sub-Himalayan region.

Methods: This prospective observational study was conducted over a period of one year, from December 2023 to December 2024. Children aged 1–59 months with echocardiographically confirmed CHD were enrolled. Clinical details and outcomes were recorded during follow-up.

Results: Seventy children (19.2per 1000) were diagnosed with CHD. Among them, 67% were infants, with a female preponderance (male: female ratio 0.9:1). Acyanotic lesions comprised 81%, most commonly ventricular septal defect (34%), atrial septal defect (28%), and patent ductus arteriosus (15%). Tetralogy of Fallot (14%) was the main cyanotic lesion. Congestive heart failure occurred in 35 children. Ten underwent surgery, and four deaths were recorded.

Conclusion: CHD is a significant burden in early childhood, with prevalence of 19.2/1000 recorded in this cohort. It contributed to substantial morbidity (50%) and mortality (5.7%).

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Introduction

Congenital heart disease(CHD) is a structural or functional abnormality of the heart that develops before birth, though it may not be detected until later in life. It is the second most common congenital birth defect with a global prevalence of 22.8%, accounting for 46.7% birth defect-related deaths.[1] The birth prevalence of CHD has increased over the last 15 years, reaching 9.4 per 1,000 live births.[2] In India, approximately 2 lakh newborns are born with CHD each year, and about 20% of them have critical defects that need prompt medical or surgical treatment. CHDs are often missed during the newborn period because of late clinical presentation, as cardiovascular physiology changes with time. One in four CHDs is classified as critical congenital heart disease (CCHD), which may remain asymptomatic and be diagnosed incidentally after birth. Delayed diagnosis can lead to significant morbidity and mortality, impacting both families and society.

The current healthcare infrastructure for managing CHD in India remains highly inadequate, with most centers located in the southern region of India.[3] Himachal Pradesh, a northern state in India with challenging terrain and remote villages, faces significant challenges in healthcare delivery. In rural regions of Himachal Pradesh, the prevalence of congenital heart disease among individuals under 18 years of age was observed to be 12.95 per 1,000 population, with variations in prevalence noted across different altitudes.[4]

Government initiatives, such as the Rashtriya Bal Swasthya Karyakram (RBSK), aim to improve CHD management by screening and treating children. Early diagnosis and timely intervention can improve life expectancy and reduce hospital visits for complications, such as recurrent respiratory infections and congestive heart failure. Determining the prevalence and case burden of CHD is crucial for guiding health care policies. However, data on prevalence, morbidity, and mortality related to CHD in Himachal Pradesh are still scarce, emphasizing the importance of in-depth research. With this in mind, the study was designed at a tertiary care center that serves patients from six neighboring districts. The study focused on a thorough evaluation of congenital heart disease (CHD), covering its clinical presentations, related complications, and mortality trends. It emphasized how different types and outcomes of CHD are influenced by timely diagnosis and appropriate management, ultimately affecting survival and quality of life.

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Objective: To study the spectrum and outcomes of congenital heart disease (CHD) in children aged 1 month to 5 years in a tertiary hospital of sub- Himalayan region.

Methodology

This prospective observational study was conducted at a tertiary care center over a period of 1 year, following approval from the Institutional Ethics Committee and Research Board. Informed written consent was obtained from the parents before enrollment. Children between 1 month and 5 years of age who presented with fast breathing, grunting, central cyanosis, or poor weight gain were considered as suspected cases of congenital heart disease (CHD). A detailed clinical history was obtained, followed by a comprehensive physical examination. The diagnosis of CHD was confirmed using two-dimensional echocardiography (2D-ECHO), after which the children were included in the study. All patients received emergency care initially, followed by routine management based on individual needs. Cardiologist consultations were conducted within our institute, and referrals to pediatric cardiology centers were arranged when necessary. Each patient was registered with the DEIC, and follow-up was maintained throughout the study period.

StatisticalAnalysis

Data were collected on a prescribed proforma and entered into a spreadsheet, and analyzed using SPSS software. The data was divided into continuous and categorical variables. Categorical variables, such as types of congenital heart disease and clinical features, were summarized as frequencies and percentages. Continuous variables, such as age at presentation, were expressed as the mean for normally distributed data. The relative proportions of acyanotic and cyanotic CHD were compared descriptively. A P value <0.05 was considered statistically significant wherever applicable.

Results

During the study period, a total of 3,648 children within the specified age range were admitted. Among these, 85 children were clinically suspected to have congenital heart disease (CHD), however, echocardiographic confirmation could only be obtained in 70 cases (19.2 per 1,000). So, 15 children were excluded from the final analysis.

Of the 70 confirmed cases, 67% were infants, and a slight female predominance was observed (M: F ratio 0.9:1). Demographic profile of patients with congenital heart disease is shown in Table 1. A majority of the cohort (61%) belonged to the upper and lower middle socioeconomic strata. The predominant presenting complaints were cough and dyspnea, reported in 73% and 61% of cases. On cardiovascular examination, a murmur was detected in 91% (n=64) of the children. The most frequently associated comorbidity was anemia affecting 60% of children, followed by congestive cardiac failure (CCF) in 50%, pneumonia (36%), and failure to thrive (24%). Notably, Down syndrome was diagnosed in 3 patients (4.3%).The clinical presentation and associated comorbidities are shown in Table 2.

Acyanotic CHD was the predominant form, observed in 81% of the cohort. Ventricular septal defect (VSD) was the most common lesion, comprising 34% of all CHD cases, followed by atrial septal defect (ASD, 28%) and patent ductus arteriosus (PDA, 15%). Tetralogy of Fallot (TOF) accounted for 14% of the cases and was the leading cyanotic CHD identified. The types of congenital heart disease observed in the study are presented in Table 3.

Table 1: Demographic Profile of CHD Patients

Parameter	Male (n=34)	Female (n=36)	Total (n=70)	Percentage (%)
Age Group				
1 month-1 year	25	22	47	67.1%
1-2 years	4	6	10	14.3%
2-3 years	2	2	4	5.7%
3-4 years	2	1	3	4.3%
4-5 years	1	5	6	8.6%

Table 2: Clinical Presentation and Associated Comorbidities in Children with Congenital Heart Disease

Variable	Number (n=70)	Percentage(%)
Clinical presentation		
Cough	51	73%
Dyspnea	43	61%
Poor feeding	14	20%
Cyanosis	8	11%
Comorbidities		
Anemia	42	60%
Congestive cardiac failure	35	50%
Pneumonia	25	36%
Malnutrition	17	24%
Down Syndrome	3	4.2%

Table 3: Pattern of Congenital Heart Diseases

Acyanotic heart disease	57	81.2%
VSD	24	34.2%
ASD	20	28.5%
PDA	11	15.7%
COA	1	1.4%
AVCD	1	1.4%
Cyanotic heart disease	13	18.4%
TOF	10	14.2%
TGA	1	1.4%
TR	1	1.4%
DORV	1	1.4%

VSD, ventricular septal defect; ASD, atrial septal defect; PDA, patent ductus arteriosus; COA, coarctation of aorta; AVCD, atrioventricular canal defect; TOF, tetralogy of Fallot; TGA, transposition of great arteries; TR, tricuspid regurgitation; DORV, double outlet right ventricle

Intensive care unit (ICU) admission was required in 27% (n=19) of the children. Among infants, 50% had a mean hospital stay of 10 days. All patients received medical management, and 10 children (14%) underwent surgical interventions. These included VSD closure in 5 patients (7%), PDA ligation in 4 (6%), and a palliative procedure in one child (1.4%). There were four deaths (5.7%) during the study period, three of whom were infants with acyanotic CHD.

Discussion

This study assessed the clinical profile, comorbidities, and outcomes of CHD in children aged 1 month to 5 years. Only about 20% of CHDs present in the neonatal period, and up to 80% may be missed by pulse oximetry done at birth. In this study, 41 (58.5%) children had peripheral oxygen saturation (SpO_2) values below 95%. Acyanotic lesions typically manifest at 3–6 weeks with a fall in pulmonary vascular resistance.[5] Jat et al. found that 55% of acyanotic CHD cases were first identified during pneumonia episodes.[6] In our study, 33 children (41.4%) had a history of recurrent pneumonia, and 25 children (35.7%) had pneumonia at the time of enrollment. Other clinical presentations observed in our patients included anemia in 42 (60%), congestive cardiac failure (CCF) in 35 children (50%) and failure to thrive (FTT) in 17 children (24%) as shown in Table 2.

With declining infant deaths from infections, CHD's relative contribution to infant mortality is rising. Delayed detection in our region is driven by low awareness, overlapping symptoms with common illnesses, poor access to echocardiography, and few cardiologists in peripheral centers.

Of 3,648 admissions, CHD was confirmed in 70 patients (19.2/1,000). Bhardwaj et al. reported 12.95/1,000 in a community survey.[4] Our higher prevalence reflects referral bias as a tertiary center. Other studies reported wide variation: Kapoor and Gupta 26.4/1,000,[7] Abqari et al. 8.79/1,000,[8] Rama Kumari et al. 10.4/1,000,[9] and neonatal rates of 17.8–24.6/1,000 in Southwest Asia.[10] Kiran B et al. reported only 1.9/1,000 in a rural South Indian center, possibly due to exclusion of preterm infants and access/financial constraints.[11]

Most patients (67%) were under one year, consistent with Indian studies showing 45%–70% in this age group[8,11,12,13] and Sherzad AG's Afghanistan series (86.4% under one year).[13] We observed a slight female preponderance (M:F 0.9:1), similar to Bhardwaj et al. and Kiran et al.,[4,11] although many reports describe male predominance, including ratios up to 2.6:1.[7,8,10,13]

Respiratory symptoms (cough, dyspnea) and murmurs dominated presentations (73% and 91%, respectively), aligned with certain earlier studies.[7,10,13] However other studies noted cyanosis or sudden deterioration as initial signs.[3,7,8,10,14,15]

Acyanotic CHD accounted for 81%, with VSD (34%) being the most common, followed by ASD (28%) and PDA (15%). TOF (14%) was the leading cyanotic lesion (Table 3). These distributions mirror many studies where VSD predominates,[3,7,8,12] though some found ASD[10] or PDA[13] as commonest; Singh et al. reported TGA as the most frequent cyanotic lesion.[13]

Comorbidities were frequent: CCF in 50%, anemia in 60%, and failure to thrive in 24%. Meena et al. reported malnutrition in 52.6%,[11] and similarly Saxena A emphasized undernutrition as a major challenge.[3] Down syndrome was present in 4.3%, comparable to Hasan et al. (3.9%),[10] while Singh et al. found it in 8.3%.[13]

ICU care was needed in 27%, and over half of the infants had a mean stay of 10 days. Sixty-one percent belonged to lower-middle or upper-lower socioeconomic strata, implying significant financial strain. In-hospital mortality was 5.7%, lower than Singh et al. (22.4%)—likely due to exclusion of neonates and shorter follow-up—and comparable to Hasan et al. (6.5%)[10] Eckersley L highlighted worse outcomes with delayed diagnosis.[15]

Surgical correction was done in 14% (mostly acyanotic lesions); one patient had palliative surgery. No patient received definitive surgery for critical CHD, underscoring gaps in specialized care. As Saxena et al. noted, India suffers from a shortage and unequal distribution of trained cardiac specialists and infrastructure, especially in northern regions, necessitating stronger central and state commitment.[3] Notably, 60% of surgeries were supported under the RBSK scheme, reflecting the importance of public programs in improving access.

The relatively short follow-up period represents a key limitation, as it precludes evaluation of long-term outcomes and therapeutic impact. The age range was also limited to children below five years, potentially missing cases with late-onset or milder presentations. Future research should include extended follow-up and a broader age group to reflect the full clinical spectrum of CHD.

Conclusion

We found a significant prevalence (19.2/1000 in the 1month to 5-year age group) of CHD. Neonates and pediatric patients with critical CHD are struggling to survive due to lack early diagnosis and specialized cardiac care centers for management. National programs like RBSK and Ayushman Bharat hold the potential for addressing healthcare challenges.

Compliance with Ethical Standards
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