Original Article

Clinical Profile of Congenital Heart Diseases in Children Above 1 Month of Age in Tertiary Health Care Settings

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Abstract

Background: Congenital heart disease (CHD) is the most common congenital anomaly and a major contributor to pediatric morbidity and mortality, particularly in developing countries. Delayed diagnosis beyond the neonatal period is common due to limited antenatal screening and awareness. The present study was undertaken to evaluate the clinical profile of CHD in children above one month of age and to assess their outcomes, including utilization of government health schemes. **Material and Methods:** This hospital-based cross-sectional observational study was conducted over one year in a tertiary care center in Indore, India. A total of 100 children aged >1 month to 16 years with clinically diagnosed CHD were included using consecutive sampling. Detailed clinical evaluation, echocardiography, and relevant investigations were performed. Outcomes, complications, and utilization of government health schemes were assessed. **Results:** The majority of cases presented in infancy (78%) with male predominance (59%). Acyanotic CHD was more common (66%), with ventricular septal defect being the most frequent lesion, while Tetralogy of Fallot predominated among cyanotic CHD. Respiratory symptoms such as fast breathing (82%) and cough (70%) were the most common presentations. Malnutrition was prevalent in 79% of children. Pneumonia (55%) and congestive cardiac failure (26%) were the major complications. Most patients (85%) were registered under RBSK/Ayushman schemes. Mortality was 25%, higher in cyanotic CHD (32.4%) compared to acyanotic CHD (21.2%). **Conclusion:** CHD beyond the neonatal period remains a major health concern with significant morbidity and mortality. Early diagnosis, improved antenatal screening, nutritional support, and strengthening referral systems and pediatric cardiac services are essential to improve outcomes.

Keywords: Congenital heart disease, Acyanotic CHD, Cyanotic CHD, Pediatric cardiology.

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Introduction

Congenital heart disease (CHD) comprises a heterogeneous group of structural abnormalities of the heart and great vessels that arise due to defects in cardiac embryogenesis. These defects may involve the cardiac septa, valves, or major vascular connections and are broadly classified into acyanotic and cyanotic types based on the presence of central cyanosis. Among these, ventricular septal defect (VSD) and atrial septal defect (ASD) are the most common acyanotic lesions, whereas cyanotic defects such as Tetralogy of Fallot (TOF) are associated with significant hypoxemia and altered hemodynamics.[1,2] The clinical spectrum of CHD is wide, ranging from asymptomatic murmurs detected incidentally to severe presentations with congestive heart failure and life-threatening cyanotic spells, underscoring the importance of early diagnosis and timely intervention.

Globally, CHD is the most common congenital anomaly, with an estimated prevalence of 8–10 per 1000 live births.[3,4] In India, the burden is particularly high due to large birth cohorts and limited prenatal and early neonatal screening in many regions. It is estimated that over 200,000 children are born annually with CHD, contributing significantly to pediatric morbidity and mortality.[4,5] CHD accounts for nearly one-fourth of all congenital malformations and constitutes a major proportion of pediatric hospital admissions in tertiary care centers.[5-8] Despite advances in diagnostic modalities such as echocardiography, a substantial number of cases remain undiagnosed during the neonatal period and present later due to subtle clinical features, delayed referrals, or lack of awareness.[7,9]

Children presenting after the first month of life often exhibit symptoms such as feeding difficulties, failure to thrive, recurrent respiratory infections, tachypnea, and, in severe cases, cyanosis.[7,10] Delayed diagnosis may result in complications including pulmonary hypertension, congestive cardiac failure, and impaired growth, which adversely affect clinical outcomes.[7,9] Echocardiography plays a pivotal role in confirming the diagnosis, delineating the anatomical defect, and

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guiding management strategies, thereby forming the cornerstone of CHD evaluation.[1,10,11]

In developing countries, access to specialized pediatric cardiac care remains uneven, posing challenges in timely management and follow-up. Government-supported health initiatives such as Rashtriya Bal Swasthya Karyakram (RBSK) and Ayushman Bharat have been implemented to enhance early detection, facilitate referral through Regional Early Intervention Centres (REIC), and improve access to definitive treatment.[12] Evaluating the clinical profile of CHD in children above one month of age in tertiary care settings, along with assessing their outcomes under these national programs, is therefore essential to understand disease patterns, identify gaps in care, and improve overall pediatric cardiac health services.

Materials and Methods

After obtaining approval from the Institutional Ethics Committee, this hospital-based cross-sectional observational study was conducted among children with congenital heart disease admitted to the pediatric wards and PICU of M.Y. Hospital and Chacha Nehru Hospital, Indore, over a period of one year. Written informed consent was obtained from parents or guardians, and confidentiality and voluntary participation were ensured.

The sample size was calculated using a prevalence-based formula (expected prevalence 0.8%, 95% confidence interval, 2% margin of error), yielding a minimum of 75 participants. To improve statistical reliability, a total of 100 children were included using consecutive sampling.

Inclusion Criteria: Children aged more than 1 month up to 16 years who were clinically diagnosed with congenital heart disease during hospital admission were included in the study.

Exclusion Criteria: Children who had undergone prior surgical correction for congenital heart disease or had incomplete clinical or investigational records were excluded from the study.

Methodology: After obtaining informed consent, eligible children were enrolled consecutively during admission. A detailed history including demographic profile and maternal risk factors such as maternal age, consanguinity, diabetes, drug exposure, antenatal history, and family history of CHD was recorded using a predesigned semi-structured proforma. A comprehensive clinical examination was performed, noting features such as cyanosis, murmur, tachypnea, tachycardia, hepatomegaly, edema, and growth parameters.

All patients underwent relevant investigations including pulse oximetry, electrocardiography (ECG), chest X-ray, and two-dimensional echocardiography with color Doppler for confirmation and classification of CHD. Based on echocardiographic findings, patients were categorized into acyanotic and cyanotic CHD groups. Syndromic associations were also documented where present .

Outcome Assessment: Patients were followed up periodically during hospitalization and, where feasible, through monthly follow-up visits. Outcomes assessed included clinical status, type of management (medical or surgical), and registration under government health schemes such as Rashtriya Bal Swasthya Karyakram (RBSK) and Ayushman Bharat. Progress and complications, if any, were documented during follow-up

Statistical Analysis: Data were entered into Microsoft Excel and analyzed using JAMOVI statistical software. Continuous variables were expressed as mean ± standard deviation, while categorical variables were presented as frequencies and percentages. Comparative analyses between groups (cyanotic vs acyanotic, syndromic vs non-syndromic) were performed using appropriate statistical tests such as Chi-square test, unpaired t-test, or ANOVA. A p-value of less than 0.05 was considered statistically significant.

Results

The present study included 100 children diagnosed with congenital heart disease (CHD). The majority of cases were identified during infancy (78%), followed by children aged 1–5 years (19%) and 5–16 years (3%). There was a male predominance (59%). Most patients belonged to the upper-lower socioeconomic class (49%), followed by lower-middle class (36%), indicating a higher burden of CHD among economically weaker sections.

Table 1: Demographic Profile of Study Population (N = 100)

Variable	Category	Frequency (n)	Percentage (%)
Age Group	1 month–1 year	78	78.0
	1–5 years	19	19.0
	5–16 years	3	3.0
Gender	Male	59	59.0
	Female	41	41.0
Socioeconomic Status	Lower	10	10.0
	Upper Lower	49	49.0
	Lower Middle	36	36.0
	Upper Middle	5	5.0

Table 2: Maternal and Perinatal Risk Factors (N = 100)

Variable	Category	Frequency (n)	Percentage (%)
Folic Acid Intake	Yes	77	77.0
	No	23	23.0
Target Scan	Done	25	25.0
	Not Done	75	75.0
Consanguinity	No	90	90.0
	Yes	10	10.0

Maternal risk factors	GDM	4	—
	PIH	2	—
	Hypothyroidism	2	—
	Overt DM	1	—
	Elderly gravida	9	—
NICU Admission	Yes	23	23.0
	No	77	77.0
Gestational Age	Term	69	69.0
	Preterm	31	31.0

Maternal and perinatal factors revealed that 77% of mothers had taken folic acid supplementation, while only 25% underwent antenatal target scan. Consanguinity was present in 10% of cases. A notable proportion (31%) of children were preterm, and 23% required NICU admission, reflecting early-life vulnerability among CHD patients. [Table 2]

Clinically, respiratory symptoms predominated, with fast breathing (82%), cough/cold (70%), and fever (63%) being the most common presenting complaints. Cyanosis was observed in 19% of cases. Anthropometric assessment revealed a high burden of malnutrition, with 79% of children below −2 SD. [Table 3]

Table 3: Clinical Presentation and Nutritional Status (N = 100)

Variable	Category	Frequency (n)	Percentage (%)
Symptoms	Fast breathing	82	82.0
	Cough/Cold	70	70.0
	Fever	63	63.0
	Difficulty breathing	18	18.0
	Cyanosis	19	19.0
	Refusal to feed	14	14.0
	Failure to thrive	8	8.0
Anthropometry (W/H)	−3 SD	34	34.0
	−2 SD	45	45.0
	−1 SD	18	18.0
	Normal	3	3.0

Regarding diagnosis, 65% of patients were previously diagnosed cases, while 30% were newly diagnosed and 5% were incidentally detected. Acyanotic CHD constituted the majority (66%), while cyanotic CHD accounted for 34%. [Table 4]

Table 4: Diagnosis Profile of Congenital Heart Disease (N = 100)

Variable	Category	Frequency (n)	Percentage (%)
Mode of Diagnosis	Previously diagnosed	65	65.0
	Newly diagnosed	30	30.0
	Incidentally diagnosed	5	5.0
Type of CHD	Acyanotic	66	66.0
	Cyanotic	34	34.0

Among acyanotic CHD, ventricular septal defect (24%) was the most common lesion, followed by endocardial cushion defect (18%) and atrial septal defect (13%). Among cyanotic CHD, Tetralogy of Fallot (16%) was the predominant lesion. [Table 5]

Table 5: Lesion-wise Distribution of CHD (N = 100)

Category	Diagnosis	Frequency (n)	Percentage (%)
Acyanotic CHD	VSD	24	24.0
	ASD	13	13.0
	Endocardial cushion defect	18	18.0
	PDA	4	4.0
	Suspected	7	7.0
Cyanotic CHD	TOF	16	16.0
	Complex CHD	6	6.0
	TGA	4	4.0
	TAPVC	2	2.0
	Suspected	6	6.0

Investigations revealed that moderate anemia (56%) was most common. Chest X-ray showed pulmonary plethora in 51% and oligemia in 28% of cases. Cardiomegaly (CT ratio >0.5) was present in 70% of patients. Pneumonia (55%) was the most common complication, followed by congestive cardiac failure (26%) and cyanotic spells (24%). In terms of healthcare utilization, 85% of patients were registered under RBSK/Ayushman schemes. Outcome analysis showed that

60% were referred for surgical intervention, 15% were discharged with follow-up advice, and 25% died. Mortality was higher in cyanotic CHD (32.4%) compared to acyanotic CHD (21.2%). Among those treated, most interventions were performed for VSD (46.2%), ASD (30.8%), and TOF (23.1%). [Table 6]

Table 6: Investigations, Complications, and Outcomes (N = 100)

Variable	Category	Frequency (n)	Percentage (%)
Hemoglobin	<7 g/dL	12	12.0
	7–9 g/dL	56	56.0
	>9 g/dL	32	32.0
Chest X-ray	Pulmonary plethora	51	51.0
	Oligemia	28	28.0
	Consolidation	21	21.0
CT Ratio	≤0.5	30	30.0
	>0.5	70	70.0
Complications	Pneumonia	55	55.0
	CCF	26	26.0
	Cyanotic spells	24	24.0
	Severe pneumonia	22	22.0
	FTT	18	18.0
	VAP	7	7.0
	Brain abscess	1	1.0
Registration Status	RBSK	85	85.0
	Ayushman	2	2.0
	Not registered	13	13.0
Outcome	Referred for surgery	60	60.0
	Discharged	15	15.0
	Death	25	25.0
Outcome by CHD	Acyanotic mortality	14/66	21.2
	Cyanotic mortality	11/34	32.4

Discussion

The present study evaluated the clinical profile, risk factors, and outcomes of congenital heart disease (CHD) in children above one month of age presenting to a tertiary care center. The findings highlight that CHD predominantly manifests early in life, with 78% of cases identified within the first year. This observation is consistent with studies by Sani MU et al. and Patra U et al., where the majority of cases presented in early childhood, emphasizing that clinically significant CHD becomes evident soon after birth.[13,14] Early presentation is often related to hemodynamically significant lesions that produce symptoms such as respiratory distress and feeding difficulties.

A male predominance (59%) was observed in the present study, which aligns with findings reported by Zahid SB et al. and Roy K et al., suggesting a consistent trend across different populations.[15,16] However, some studies have reported near-equal gender distribution, indicating that while male preponderance is common, it is not universal.[17] This variation may be attributed to genetic factors as well as sociocultural influences affecting healthcare-seeking behavior.

Socioeconomic analysis revealed that nearly 85% of patients belonged to lower socioeconomic strata, highlighting the disproportionate burden of CHD in economically weaker populations. Similar findings have been reported in developing countries, where limited access to healthcare, inadequate antenatal services, and delayed diagnosis contribute to increased morbidity.[16,18] The present study reinforces the role of social determinants in influencing disease burden and outcomes.

A significant gap in antenatal screening was observed, with only 25% of mothers undergoing target scans during pregnancy. This is consistent with Rashid U et al., who reported delayed diagnosis due to inadequate prenatal screening facilities.[18] Despite 77% of mothers receiving folic acid supplementation, CHD occurrence remained substantial, supporting the concept that CHD has a multifactorial etiology and is not entirely preventable through nutritional interventions alone.[3,9] These findings emphasize the need to strengthen antenatal screening programs, particularly fetal echocardiography in high-risk pregnancies.

Clinically, respiratory symptoms were the predominant mode of presentation, with fast breathing (82%), cough/cold (70%), and fever (63%) being the most common complaints. These findings are comparable to Roy K et al. and Chelo D et al., who reported respiratory distress and recurrent infections as key presenting features.[16,19] Cyanosis was present in 19% of cases, corresponding to cyanotic CHD, and remains a hallmark of these lesions.[20] The overlap of cardiac and respiratory symptoms may lead to delayed or missed diagnosis, particularly in resource-limited settings.

Malnutrition was a significant finding in this study, with nearly 79% of children falling below −2 SD. Similar observations have been reported by Roy K et al. and Song L et al., highlighting the impact of CHD on growth and nutritional status.[16,21] The pathophysiology is multifactorial, involving increased metabolic demand, feeding difficulties, and recurrent infections. These findings underscore the importance of integrating nutritional support into CHD management.

In terms of disease pattern, acyanotic CHD was more common (66%) than cyanotic CHD (34%), consistent with studies by Wanni KA et al. and Patra U et al.[14,18] Ventricular septal defect (24%) was the most common lesion, aligning with global data

identifying VSD as the predominant congenital cardiac defect.[16,22] Among cyanotic CHD, Tetralogy of Fallot (16%) was the most frequent lesion, which is in agreement with multiple studies.[15,20] The higher proportion of cyanotic CHD in this study may reflect referral bias to tertiary care centers where more severe cases are managed.

The study also demonstrated a high burden of complications, with pneumonia (55%) and congestive cardiac failure (26%) being the most common. These findings are consistent with previous studies highlighting increased susceptibility to infections and heart failure in CHD patients.[15,17] Cyanotic spells and rare complications such as brain abscess further reflect the severity of disease in certain subsets. The presence of moderate anemia in a significant proportion of patients may have further aggravated cardiac workload and clinical outcomes.

Outcome analysis revealed that 60% of patients were referred for surgical intervention, while mortality was observed in 25% of cases. Mortality was higher in cyanotic CHD (32.4%) compared to acyanotic CHD (21.2%), indicating poorer prognosis in cyanotic lesions. Similar trends have been reported in previous studies, where cyanotic CHD is associated with higher morbidity and mortality due to chronic hypoxemia and complex anatomical defects.[20] The relatively high mortality rate in this study may be attributed to delayed presentation, high complication rates, and limited access to timely surgical intervention.

An important finding of the study was the substantial utilization of government healthcare schemes, with 85% of patients registered under RBSK and a small proportion under Ayushman Bharat. This highlights the critical role of public health programs in facilitating access to diagnosis and treatment, particularly for economically disadvantaged populations. However, the presence of unregistered cases indicates the need for improved awareness and outreach.

Overall, the present study indicates that congenital heart disease in children is marked by early presentation, predominance of acyanotic lesions, substantial respiratory morbidity, a high burden of malnutrition, and significant mortality, particularly in cyanotic cases. However, the findings are constrained by a single-center design, limited sample size, and absence of long-term follow-up. Referral bias may have resulted in a higher proportion of severe cases. Additionally, incomplete antenatal data, lack of genetic evaluation, and restricted access to advanced diagnostic and surgical facilities may have influenced outcomes. Enhancing antenatal screening, ensuring early diagnosis, improving nutritional support, and expanding pediatric cardiac services are crucial for better outcomes.

Conclusion

Congenital heart disease beyond the neonatal period remains a major cause of morbidity and mortality, especially in resource-limited settings. It predominantly presents in infancy, with acyanotic lesions being more common, though cyanotic CHD carries higher mortality. Respiratory symptoms and malnutrition reflect delayed diagnosis and chronic burden. Programs like RBSK have improved early detection and referral. However, gaps in antenatal screening and access to surgical care persist. Strengthening early diagnosis, referral systems, and pediatric cardiac services is essential. An integrated approach combining clinical vigilance and public health initiatives is crucial to improving outcomes in affected children.

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Conflicts of interest

There are no conflicts of interest.

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