

Original Research Article

Prevalence, clinical characteristics, and growth implications of congenital heart disease in pediatric patients: a single-center retrospective study from India

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ABSTRACT

Background: Congenital heart disease (CHD) with structural defects of hearts and vessels is associated with significant mortality in pediatric population. Early timely intervention is critical. This study aims to examine the demographics and clinical profiles of pediatric population with CHD from India.

Methods: A retrospective observational study conducted at a district hospital in pediatric patients diagnosed with CHD. Basic demographics, clinical symptoms, diagnostic procedures and growth parameters were collated and analyzed. Analysis involved descriptive statistics and Chi-square test for proportions comparing different types of CHD.

Results: In this retrospective study of pediatric CHD cases (n=141) from India, acyanotic lesions predominated (87.2%), with ventricular and atrial septal defects (ASDs) most common. Over half were diagnosed within six months, though delays were notable in cyanotic CHD. Around one-third were asymptomatic at diagnosis, while others presented with classic symptoms like breathlessness and feeding difficulties. Over 90% showed growth below the 50th percentile, highlighting significant nutritional impact. Lesion type influenced timing and urgency of management.

Conclusions: These findings highlight the need for enhanced screening, early diagnosis, growth monitoring, and integrated nutritional support to optimize long-term outcomes in children with CHD.

Keywords: Congenital heart disease, Pediatric cardiology, Acyanotic CHD, Cyanotic CHD, Ventricular septal defect, Atrial septal defect, Growth parameters, India

INTRODUCTION

Congenital heart disease (CHD) is the most prevalent congenital anomaly globally, affecting approximately 8 to 10 per 1,000 live births.^{1,2} It encompasses a diverse group of structural defects involving the heart and great vessels that develop during fetal life and are present at birth. These defects can range from simple anomalies that may resolve spontaneously or require minimal intervention, to complex malformations that necessitate

early surgical correction and prolonged medical follow-up.

In the Indian context, the burden of CHD is particularly significant due to the country's high birth rate, with an estimated 25 million live births annually. Based on global prevalence rates, this translates to an estimated 200,000 to 250,000 new cases of CHD each year in India.³ Among these, approximately 20%-or 40,000 to 50,000 children-are born with critical CHD requiring surgical or catheter-based intervention within the first year of life. However,

due to unequal access to healthcare, limited availability of specialized pediatric cardiology services, and delayed diagnosis, only about 10 to 15 percent of affected children receive timely and appropriate treatment.⁴

CHD contributes significantly to infant and under-five mortality in India, particularly in rural and underserved regions.⁵ Complications arising from untreated or late-diagnosed CHD include congestive heart failure, pulmonary hypertension, cyanosis and chronic hypoxemia, arrhythmias, failure to thrive, and developmental delays. Children with complex or cyanotic defects are at higher risk of infective endocarditis and long-term neurodevelopmental impairments, especially when surgical interventions are delayed or postoperative care is inadequate.^{5,6} These outcomes highlight the pressing need for comprehensive epidemiological data to inform clinical and policy decision-making.

Despite growing awareness and advancements in diagnostic and therapeutic modalities, the absence of a nationwide CHD registry and limited region-specific data remain significant barriers to improving outcomes. Therefore, understanding the prevalence, clinical presentation, and complications of CHD in Indian children is critical for strengthening early detection strategies, optimizing resource allocation, and improving long-term health outcomes. This study aims to examine the demographic and clinical profiles of Indian CHD patients.

METHODS

Study design and population

This was an observational study conducted at Amrut Hrudayalaya and super speciality hospital Buldhana from December 2021 to November 2022. The study included pediatric patients (0-18 years) referred for 2D echo testing and diagnosed with CHD.

Ethical considerations

The study was approved by the institutional ethics review board at DUPMC Jalgaon (Ethical Approval Number: [IEC/DUPMCH/225/04]). Informed consent was taken from the patients or from their parents or guardians prior to the initiation of the study. All data were anonymized to ensure patient confidentiality.

Inclusion criteria

The inclusion criteria were as follows: confirmed diagnosis of CHD, age ≤ 18 years at the time of diagnosis, and complete clinical data available for analysis.

Exclusion criteria

Patients with acquired heart conditions or incomplete medical records were excluded from the study.

Outcome measures

Data for this study were retrospectively collected from hospital records, including patient demographics, clinical symptoms, diagnostic procedures, and growth parameters. The primary variables of interest included the type of CHD with patients classified into cyanotic and acyanotic groups based on their specific cardiac lesions. Age at diagnosis was also recorded, with patients categorized into three groups: less than six months, six months to five years, and older than five years. Clinical symptoms such as breathlessness, feeding difficulties, and asymptomatic presentation were extracted from patient medical records. In addition, the feasibility of prenatal diagnosis was assessed for all relevant cases. Growth parameters, including weight and height measurements, were obtained and compared to age-appropriate growth percentiles based on world health organization (WHO) standards to assess nutritional status and growth failure. Data were reviewed to ensure completeness and accuracy, and only those with complete clinical records were included in the analysis.

Data analysis and statistics

Descriptive statistics were used to summarize patient demographics, lesion types, and clinical characteristics. Continuous variables such as age and growth measurements were reported as means with standard deviations (SD), while categorical variables such as CHD type, symptoms, and age of diagnosis were expressed as percentages. Chi-square tests were used to compare the distribution of categorical variables between cyanotic and acyanotic groups. A $p < 0.05$ was considered statistically significant. Data analysis was performed using GraphPad Prism.

RESULTS

Of the total study population ($n=141$), 87.2% were diagnosed with acyanotic CHD, while 12.8% presented with cyanotic defects (Table 1). Early diagnosis was prevalent, with 54% of children being diagnosed before six months of age (Table 1). Notably, no gender-based differences were observed in the distribution of CHD types (Table 1). Prenatal diagnosis was confirmed in seven cases, all of which were acyanotic CHD. Approximately one-third of the children were asymptomatic at the time of diagnosis, while breathlessness and feeding difficulties were reported in 20% of the cohort, with these symptoms being more prevalent in the cyanotic subgroup (Table 1). Additionally, more than 90% of the study population were found to be below the 50th percentile for age-recommended height and weight, likely due to feeding difficulties and hemodynamic challenges associated with the condition (Figure 1). The distribution of lesion characteristics varied significantly between cyanotic and acyanotic types (Table 1 and 2).

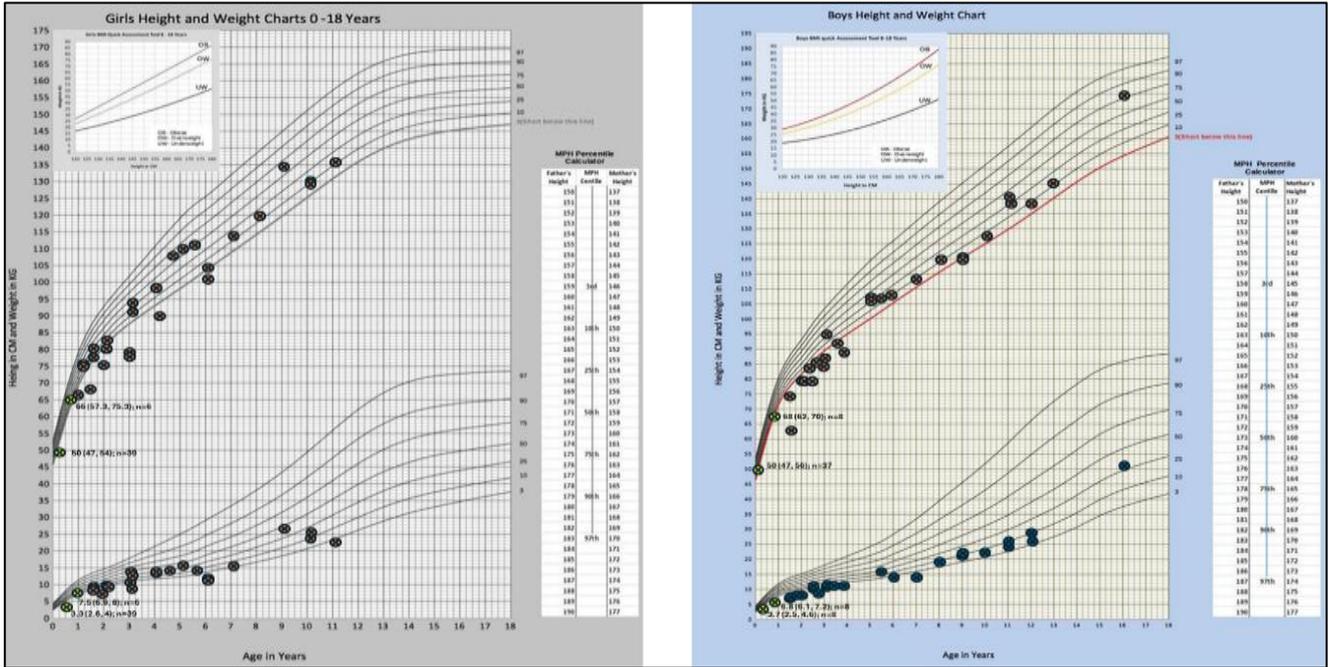


Figure 1: Height-weight chart of the study population.

*Legend: The curves represent the growth charts, showing the trend of age-recommended height and weight in girls (A) and boys (B). The top curves in both graphs represent height and the bottom curves represent the weight. The right Y-axis represents the percentile (from 3 to 97%) indicated at the end of each curve. The red filled dots indicates individual values and the yellow filled between the ages of 0-6 months and 6-12 months represent the median [IQR, n] values in that age group.

Table 1: Demographic and clinical characteristics.

Variables	Total population	Cyanotic	Acyanotic
N	141	18 (12.8)	123 (87.2)
Age (in years)			
0-6 months	76 (53.9)	10 (55.5)	66 (53.6)
7-12 months	14 (9.9)	3 (16.7)	11 (8.9)
1-2	11 (7.8)	0 (0)	11 (8.9)
2-5	18 (12.8)	0 (0)	18 (14.6)
>5	22 (15.6)	5 (27.8)	17 (13.8)
Gender			
Male	71 (50.3)	9 (50)	62 (50.4)
Female	70 (49.7)	9 (50)	61 (49.6)
Prenatal diagnosis	7/141 (5)	0/7 (0)	7/7 (100)
History of hospitalization	63 (44.7)	5 (27.8)	58 (47.2)
Cardiac involvement			
Possible	32 (22.7)	3 (16.7)	29 (23.6)
Non-cardiac	24 (17)	1 (5.6)	23 (18.7)
NA	85 (60.3)	14 (77.8)	71 (57.7)
Symptoms			
Asymptomatic	46 (32.6)	5 (27.8)	41 (33.3)
Breathlessness	26 (18.4)	6 (33.3)	20 (16.3)
Feeding difficulties	29 (20.6)	6 (33.3)	23 (18.7)
Recurrent respiratory infection	46 (32.6)	7 (38.9)	39 (31.7)
Murmur	134 (95)	17 (94.4)	117 (95.1)
Normal	5 (3.5)	0 (0)	5 (4.1)
Lesions			
OSASD	34 (24.1)	0 (0)	34 (27.6)
PDA	15 (10.6)	0 (0)	15 (12.2)

Continued.

Variables	Total population	Cyanotic	Acyanotic
VSD	32 (22.7)	0 (0)	32 (26)
TOF	7 (5)	7 (38.9)	0 (0)
COA	1 (0.7)	0 (0)	1 (0.8)
OSASD+VSD	11 (7.8)	1 (5.6)	10 (8.1)
Bicuspid AOV	3 (2.1)	0 (0)	3 (2.4)
OSASD+PDA	7 (5)	0 (0)	7 (5.7)
VSD+PDA	2 (1.4)	0 (0)	2 (1.6)
Others*	29 (20.6)	10 (55.6)	15 (12.2)

*The individual lesion phenotypes are presented in Table 2.

Table 2: List of uncommon complex lesions in the study population.

Variables	Cyanotic	Acyanotic
N	18	123
MVP AML prolapse	0	1 (0.8) 1/1 (100)
HCM + PS	0	1 (0.8) 1/1 (100)
OSASD + congenital MS	0	1 (0.8) 1/1 (100)
OSASD + PS	0	3 (2.5) 3/3 (100)
DTGA + DORV + VSD	1 (55.5) 1/1 (100)	0
VSD + OSASD + PDA	0	2 (1.6) 2/2 (100)
HOCM + LVH	0	1 (0.8) 1/1 (100)
VSD + DCRV	0	1 (0.8) 1/1 (100)
OSASD + bicuspid AOV	0	1 (0.8) 1/1 (100)
DTGA + VSD + ASD + PS	1 (55.5) 1/1 (100)	0
TAPVC + OSASD	1 (55.5) 1/1 (100)	0
VSD + PAH	1 (55.5) 1/2 (50)	1 (0.8) 1/2 (50)
OSASD + LPA stenosis	0	1 (0.8) 1/1 (100)
TOF + ASD	1 (55.5) 1/1 (100)	0
TOF + OSAVSD	1 (55.5) 1/1 (100)	0
VSD + PAH	1 (55.5) 1/1 (100)	0
VSD + OSASD + PAH	0	1 (0.8) 1/1 (100)
DTGA + OSASD + PS	1 (55.5) 1/1 (100)	0
VSD + PDA + PAH	0	1 (0.8) 1/1 (100)
TOF + DORV	1 (55.5) 1/1 (100)	0
VSD + ASD + PAH	0	2 (1.6) 2/2 (100)

Continued.

Variables	Cyanotic	Acyanotic
OSASD + LVH + HOCM	0	1 (0.8) 1/1 (100)
VSD + PFO	0	1 (0.8) 1/1 (100)
TAPVC + OSASD + PAH	1 (55.5) 1/1 (100%)	0

DISCUSSION

The results of this study provide valuable insights into the clinical characteristics, timing of diagnosis, and growth patterns of children with CHD in our cohort. A predominant proportion of the children were diagnosed early, yet a significant subset, particularly those with cyanotic defects, experienced delays in diagnosis, highlighting challenges in the timely detection of more complex lesions. In addition to the variations in clinical presentation and diagnosis, the study revealed concerning trends in growth impairment, with a substantial number of children falling below age-appropriate growth percentiles. These findings raise important questions about the underlying factors contributing to growth failure and the long-term health implications for children with CHD.

In this study, acyanotic CHD accounted for a substantial majority of cases (87.2%), with cyanotic CHD comprising 12.8% of the cohort. This distribution is consistent with prior epidemiological data, which indicates that lesions such as ventricular septal defects (VSDs) and ASDs are among the most common types of CHD globally.^{1,2} Notably, over half (54%) of the total study population were diagnosed within the first six months of life, aligning with the increasing awareness and availability of early pediatric screening programs. The age at admission with cyanotic lesions was observed to be 28% over five years of age, compared to only 14% in acyanotic cases. This may not reflect the age at diagnosis since many kids were already diagnosed before being referred to the current hospital. This may be attributed to the more variable or insidious clinical presentation of certain cyanotic defects and a lack of early access to echocardiographic evaluation in some settings.⁷ Despite the differences in timing of diagnosis, no gender-based variation was observed, which mirrors findings from other population-based studies suggesting no significant sex predilection in CHD prevalence.^{7,8}

Approximately one-third of patients in this cohort were asymptomatic at the time of diagnosis, reinforcing the need for routine cardiac evaluations in infants and young children to detect clinically silent but potentially serious heart defects. Among symptomatic patients, 20% reported breathlessness and feeding difficulties, which are hallmark signs of congestive heart failure and pulmonary over circulation in CHD.⁹ These symptoms were more frequently reported in cyanotic CHD (33.3%), consistent with the pathophysiology of right-to-left shunts and systemic desaturation that characterizes these conditions.

The heightened symptom burden in cyanotic lesions aligns with prior studies demonstrating increased morbidity in this subgroup, especially in the absence of early corrective surgery.⁷

Growth and nutritional impact

A striking finding in this study was that over 90% of the children were below the 50th percentile for age-recommended height and weight. Growth failure is a well-established complication of pediatric CHD and may occur even in the absence of overt clinical symptoms.^{9,10} Feeding difficulties, increased metabolic demands, and recurrent respiratory infections are common contributors to poor weight gain and stunted linear growth.⁹⁻¹¹ In cyanotic CHD, chronic hypoxemia impairs tissue oxygen delivery and can disrupt endocrine pathways critical for growth, including growth hormone secretion and insulin-like growth factor (IGF-1) activity.¹² Long-term nutritional deficits in these children can impact not only physical development but also cognitive outcomes, highlighting the need for early intervention, frequent growth monitoring, and integration of dietary support into cardiac care.^{6,10}

Lesion variability and management implications

The lesion characteristics differed notably between cyanotic and acyanotic groups, as shown in Table 1. This variation is clinically significant, as the type of lesion largely determines the need for intervention, timing of surgery, and overall prognosis. Acyanotic defects often follow a more stable clinical course and may be managed conservatively or scheduled for elective repair, while cyanotic defects usually demand urgent surgical attention to prevent irreversible physiological consequences.¹³ Prenatal diagnosis was confirmed in only seven cases, all of which were acyanotic. The low rate of antenatal detection may reflect limitations in screening, which is often influenced by the availability of trained professionals and equipment, as well as maternal health-seeking behavior.^{4,12} Expanding prenatal screening capacity could improve early diagnosis, parental counseling, and postnatal outcomes through planned delivery at tertiary care centers.¹⁴

Limitations

As a retrospective, single-center analysis, it depends on the accuracy and completeness of medical records and the findings may not be fully generalizable to other settings or populations. In addition, the relatively small number of

cyanotic CHD cases and the use of cross-sectional growth data without follow-up limit lesion-specific comparisons and do not allow assessment of long-term growth or clinical outcomes.

CONCLUSION

Strengthening systematic infant and early-childhood cardiac screening, together with timely referral, may reduce delayed presentation in children with congenital heart disease. Integrated nutritional surveillance may help address the high burden of growth impairment observed across lesion types, particularly in resource-limited settings.

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