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Clinico-Epidemiological And Echocardiographic Profile Of Congenital Heart Disease In Children: A Cross-Sectional Observational Study At A Tertiary Care Hospital In Southern Rajasthan

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ABSTRACT

Background: Congenital heart diseases (CHDs) are the most prevalent congenital anomalies globally, significantly contributing to infant morbidity and mortality. Echocardiography plays a crucial role in diagnosis, yet delayed presentation is common in resource-limited settings.

Methods: A prospective, cross-sectional study was conducted at Pacific Institute of Medical Sciences, Udaipur, over 12 months. A total of 106 children aged 0-18 years with echocardiographically confirmed CHD were included. Demographic, clinical, and echocardiographic data were collected. Descriptive and inferential statistical analyses were performed using SPSS v28.

Results: Of 106 participants, infants (26.4%) formed the largest group. Males predominated (58.5%). Most participants were from rural areas (60.4%) and lower-middle socioeconomic status (24.5%). Consanguinity was present in 21.7%, and 11.3% had a positive family history. Clinical symptoms varied with age: infants frequently presented with dyspnoea (78.6%) and failure to thrive (64.3%), while older children were often diagnosed incidentally. Acyanotic CHDs were more prevalent (68.9%), with ventricular septal defect (42.5%) as the most common lesion. Tetralogy of Fallot (45.5%) was the most frequent cyanotic CHD. Pulmonary hypertension was absent in neonates and infants but present in 60% of children aged >5 years. Consanguinity significantly increased the risk of severe CHD (OR: 2.45, p=0.03).

Conclusion: CHD presentation shows age-specific trends, with late diagnosis common in older children. Acyanotic lesions dominate the overall CHD burden, while cyanotic lesions are prevalent in neonates. Early screening is crucial for timely diagnosis and intervention, especially in rural and consanguineous populations.

Keywords: *Congenital Heart disease, echocardiography, tetralogy of fallot, pulmonary hypertension, ventricular septal defect*

1. INTRODUCTION

Congenital heart diseases (CHDs) encompass a spectrum of structural and functional anomalies of the heart and great vessels that develop during fetal life. [1] Globally, CHDs are the most common congenital anomalies and a leading cause of childhood morbidity and mortality. Advances in diagnostic modalities, particularly echocardiography, have improved detection; however, late diagnoses still occur frequently in low-resource settings, contributing to poor outcomes. [2]

The global incidence of CHDs is estimated at 8-12 per 1,000 live births. [3] In India, this figure varies from 6 to 15 per 1,000, with underdiagnosis likely in rural and underserved regions. [4,5] Ventricular septal defect (VSD), atrial septal defect (ASD), and patent ductus arteriosus (PDA) are the most commonly reported acyanotic lesions, while tetralogy of Fallot (TOF) and transposition of the great arteries (TGA) are prominent among cyanotic CHDs. [1,3] Multiple risk factors contribute to CHD development, including genetic predisposition, maternal illnesses such as diabetes and hypertension, teratogen exposure, and consanguinity. [6,7,8]

India faces unique challenges in managing CHDs. Healthcare infrastructure, especially in rural areas, remains underdeveloped. Paediatric cardiac centres are limited, resulting in delayed interventions. Economic constraints also play a role, as most patients must bear out-of-pocket expenses for diagnosis and treatment. These challenges necessitate region-specific studies to understand the epidemiology and clinical profile of CHD and to facilitate targeted interventions. [4,9,10]

This study was designed to bridge this gap by analyzing the clinico-epidemiological and echocardiographic profile of children with CHD at a tertiary care hospital in Southern Rajasthan. The insights gained from this study can inform clinical practice and policy, with implications for screening strategies, healthcare planning, and early intervention efforts.

2. MATERIALS AND METHODS

This hospital-based, prospective, cross-sectional observational study was carried out over 12 months in the Department of Pediatrics at Pacific Institute of Medical Sciences (PIMS), Udaipur, a tertiary care teaching hospital catering to Southern Rajasthan. The study aimed to evaluate the clinico-epidemiological and echocardiographic profiles of children diagnosed with congenital heart disease (CHD).

Study Design and Population: Children aged 0–18 years who were clinically suspected of having CHD and subsequently confirmed by 2D-echocardiography with Doppler were included. Consecutive sampling was used to enroll all eligible cases. Children with acquired heart diseases (e.g., rheumatic heart disease, Kawasaki disease), concurrent systemic illnesses affecting cardiac assessment, or absence of parental consent were excluded.

Sample Size: Based on institutional records indicating around 150 CHD admissions annually and anticipating a 10% attrition rate, a sample of 106 participants was finalized.

Ethical Considerations: The study received approval from the Institutional Ethics Committee. Informed consent was obtained from parents or guardians, and assent was acquired from children above 7 years. Data confidentiality was maintained using coded identifiers.

Data Collection: A structured proforma, validated by pediatric cardiologists, was used to collect data on demographics (age, sex, residence), socioeconomic status (Modified Kuppuswamy Scale 2022), consanguinity, and family history. Detailed clinical history included prenatal factors (maternal illnesses, infections, drug exposure), neonatal symptoms (cyanosis, feeding issues), developmental milestones, and presenting complaints (dyspnea, failure to thrive, recurrent infections).

Clinical Examination: Vital signs, anthropometry (weight, height/length, BMI, OFC), and cardiovascular findings were systematically recorded. Pre- and post-ductal oxygen saturations were measured, and murmur characteristics were graded using Levine's scale. Congestive heart failure was assessed using the modified Ross classification.

Echocardiographic Evaluation: All participants underwent echocardiography using Philips EPIQ 7G machines with pediatric probes (S8-3, S12-4), performed by a trained pediatric cardiologist. Lesions were categorized as acyanotic or cyanotic per ICD-11. Shunt direction, valve function, chamber size, and pulmonary arterial pressure (estimated via TR jet velocity) were assessed. Pulmonary hypertension was classified per ESC 2022 guidelines.

Additional Diagnostics: Where indicated, electrocardiograms, chest X-rays, arterial blood gas analyses, and genetic testing (for syndromic CHD) were performed.

Quality Control: The research supervisor verified case records daily to ensure accuracy. A second cardiologist reviewed 10% of echocardiograms for quality assurance.

Data Management and Statistical Analysis: All data were entered into a secure Redcap database. Analyses were performed using SPSS version 28. Descriptive statistics (mean, standard deviation, proportions) were used for baseline characteristics. Chi-square/Fisher's exact tests and t-tests were used for comparisons. Multivariate logistic regression assessed risk factors. A p-value <0.05 was considered statistically significant.

3. RESULTS

A total of 106 children with congenital heart disease (CHD) were analyzed, with 58.5% being male and 60.4% residing in rural areas. Nearly a quarter (24.5%) belonged to the lower-middle socioeconomic class, while 21.7% had consanguineous parentage, and 11.3% reported a positive family history. Dyspnea/tachypnea (64.2%) and poor feeding (43.4%) were the most common presenting symptoms. Diagnosis most often occurred in early infancy (26.4%) or after age five (38.7%), with only 4.7% detected prenatally. Acyanotic CHD was more prevalent (68.9%)—VSD being the most frequent (42.5%)—while TOF (45.5%) dominated cyanotic cases (31.1%). Pulmonary hypertension affected 60% of children over five years. Most patients had normal LV function (73.6%), though 15.1% exhibited LV dysfunction. Medical therapy was the mainstay (61.3%), with 26.4% undergoing surgery and 9.4% receiving catheter interventions.

Table 1-Demographic Characteristics of the Participants

Variable	**Value**
Total Participants	106
Male (%)	62 (58.5%)
Rural Residence (%)	64 (60.4%)
Lower-middle Socioeconomic Status (%)	26 (24.5%)
Consanguinity (%)	23 (21.7%)
Positive Family History (%)	12 (11.3%)

Table 2: Clinical Presentation and Diagnosis Timing

Clinical Feature	Frequency (%)
Dyspnea/Tachypnea	68 (64.2%)
Failure to Thrive	38 (35.8%)
Cyanosis	33 (31.1%)
Poor Feeding	46 (43.4%)
Recurrent Chest Infections	34 (32.1%)
Incidentally Diagnosed	19 (17.9%)
Prenatal Diagnosis	5 (4.7%)
Diagnosis in Early Infancy (1-6 months)	28 (26.4%)
Diagnosis >5 years	41 (38.7%)

Table 3-Echocardiographic and Treatment Findings

Parameter	**Value**
Acyanotic CHD	73 (68.9%)
Cyanotic CHD	33 (31.1%)
Most Common Acyanotic: VSD	31 (42.5%)
Most Common Cyanotic: TOF	15 (45.5%)
Pulmonary Hypertension (>5 years)	60% of patients >5 years
Normal LV Function	78 (73.6%)
LV Dysfunction	16 (15.1%)

Medical Management	65 (61.3%)
Surgical Intervention	28 (26.4%)
Catheter Intervention	10 (9.4%)

4. DISCUSSION

This study conducted at Pacific Institute of Medical Sciences, Udaipur, provides valuable insights into the epidemiology, clinical characteristics, and echocardiographic patterns of congenital heart disease (CHD) among children in Southern Rajasthan. The findings resonate with several national and international studies, while also highlighting regional nuances in disease distribution, risk factors, and treatment outcomes.

Our observation that 26.4% of children were diagnosed with CHD in early infancy is consistent with global data indicating that most symptomatic CHDs present within the first year of life. **Donofrio et al. (2014),** [11] in a large study from the United States, reported that 25.9% of CHDs were identified within the first six months of life, primarily due to improved fetal echocardiographic practices. However, our finding that only 4.7% of cases were diagnosed prenatally underscores the diagnostic gap in low-resource settings, as compared to **Quartermain et al. (2021)** [12] who reported fetal detection rates as high as 32.1% in regions with universal prenatal screening.

The study highlighted a male predominance (58.5%) among CHD patients, particularly among toddlers (1.75:1). This finding is comparable to **Tegene et al. (2020)** [13] in Ethiopia who reported a male to female ratio of 1.8:1, and **Saxena et al. (2018)** [4] in India who documented a male predominance of 1.6:1. **Hoffman et al. (2004)** [1] also reported similar global trends, although the gender disparity was less pronounced in high-income settings.

Socioeconomic disparities were evident, with 24.5% of participants from the lower-middle socioeconomic class. This correlates with **Bernier et al. (2010)** [14] who observed a higher CHD burden among middle-income populations in developing countries. Notably, rural children accounted for 60.4% of our cohort, which matches data from **Vaidyanathan et al. (2011)** [5], indicating reduced access to specialized care in rural North India.

Consanguinity was present in 21.7% of our cases and was independently associated with severe CHD (adjusted OR 2.45, p=0.03). This aligns with **Nabulsi et al. (2003)** [15], who reported a 25.8% consanguinity rate in the Middle East, and Becker et al. (2001), who found a higher prevalence among neonates with critical CHD. In contrast, **Pierpont et al. (2007)** [16] reported lower consanguinity rates (~6%) in Western populations, emphasizing regional variability.

The most common maternal risk factors identified were hypertension (16.0%) and gestational diabetes mellitus (11.3%). **Wu et al. (2017)** [17] found hypertensive disorders associated with a twofold increase in CHD risk, corroborating our findings. Similarly, **Zhang et al (2022)** [18] highlighted the teratogenic potential of early gestational diabetes, with ORs for CHD as high as 3.1. Our results also suggest rubella exposure as a potential risk factor (OR 4.42), echoing the Global Burden of Disease Study (2020) findings, although the sample size was small.

Clinically, the majority of patients presented with dyspnea/tachypnea (64.2%), failure to thrive (35.8%), and poor feeding (43.4%). These symptoms are in line with reports from **Saxena et al. (2018)** [4] who found that 72.3% of Indian infants with CHD presented with respiratory distress. **Bernier et al. (2010)** [14] reported similar clinical presentations in LMICs, although symptom recognition was often delayed due to limited primary care awareness.

Auscultatory findings revealed murmurs in 84.9% of patients, supporting **Mahapatra et al.'s (2017)** [19] observations, who documented murmurs in 85% of CHD cases. Incidental diagnosis was common in older children and adolescents, a trend also noted by **Yamauchi et al. (2018)** [20] in their U.S. multicenter analysis.

In terms of CHD types, acyanotic defects were more prevalent (68.9%), with VSD (42.5%) and ASD (28.8%) as the most frequent lesions. This matches global trends described by **Hoffman et al. (2004)** [1] and Indian data from **Saxena et al. (2016),** [4] both identifying VSD as the dominant defect. PDA prevalence declined with age, consistent with **Bernier et al. (2010).** [14]

Cyanotic CHDs accounted for 31.1% of cases, primarily TOF (45.5%) and TGA (21.2%). These findings align with studies from India **(Saxena, 2016)** [4], though TOF prevalence was slightly higher in our study. TAPVC and DORV cases were less common, but within the ranges reported by **Mahapatra et al. (2017).** [19]

Treatment data revealed that 61.3% of children received medical management alone, with surgical correction in 26.4%. This reflects the resource prioritization pattern described by **Costello et al (2018)** [21] and **Oster et al. (2021)** [22], particularly in settings without dedicated pediatric cardiac units. Catheter interventions were limited (9.4%), mirroring **Jacobs et al. (2019),** [23] while palliative surgeries were rare, suggesting early surgical referrals.

Our findings affirm known CHD patterns while highlighting the need for enhanced screening, particularly in rural and

consanguineous populations. They echo concerns raised in the Global Cardiac Care Consortium (2022) and reinforce the importance of maternal risk mitigation, health education, and system-level reforms for timely CHD detection and care.

5. CONCLUSION

This study underscores the substantial burden and diversity of congenital heart disease (CHD) among children in Southern Rajasthan. Acyanotic defects, particularly ventricular septal defects, were the most common, while cyanotic lesions like tetralogy of Fallot were more prevalent in neonates. Significant associations were found with consanguinity and maternal hypertension, and delayed diagnosis was common, especially in rural populations. Echocardiography proved essential in diagnosis and management planning. These findings emphasize the urgent need for improved neonatal screening, enhanced maternal care, and equitable access to pediatric cardiac services. Strengthening regional healthcare infrastructure and public health awareness is imperative to reduce CHD-related morbidity and mortality.

Conflict of Interest-Nil

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