

Original Research Article

Incidence and pattern of congenital heart disease in children: an observational cross-sectional study

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ABSTRACT

Background: The objective of this study was to analyse the incidence, pattern, and clinical presentation of congenital heart disease in children and evaluate associated risk factors.

Methods: This observational cross-sectional study included 100 children aged 0-14 years diagnosed with CHD at department of cardiac surgery, Bangabandhu Sheikh Mujib Medical University (BSMMU), Dhaka, Bangladesh duration of study was 1 year starting from January to December 2022. Detailed clinical evaluation, echocardiography, and relevant investigations were performed for all cases. Associated anomalies and maternal risk factors were documented.

Results: Of 100 cases, 56% were males and 44% females (ratio 1.27:1). The majority (68%) were diagnosed in the first year of life. Acyanotic CHD predominated (72%) with ventricular septal defect being the most common lesion (30%), followed by atrial septal defect (15%) and patent ductus arteriosus (12%). Among cyanotic CHD (28%), tetralogy of Fallot was most frequent (15%). Common presenting features included breathing difficulty (45%), failure to thrive (38%), and recurrent respiratory infections (35%). Associated anomalies were present in 22% cases, with Down syndrome (8%) being the most common genetic association. Significant maternal risk factors included advanced age (15%) and diabetes mellitus (8%).

Conclusions: The study demonstrated patterns of CHD consistent with global literature while highlighting regional variations. Early detection was achieved in the majority of cases, though late presentation remains a concern. The findings emphasize the need for comprehensive screening programs and improved healthcare access, particularly in resource-limited settings. Understanding these patterns can guide resource allocation and healthcare planning for better management of pediatric cardiac care.

Keywords: Congenital heart disease, Heart defects, Pediatric cardiology, Ventricular septal defect

INTRODUCTION

Congenital heart disease (CHD) remains the most common congenital anomaly, affecting approximately 8-12 per 1,000 live births globally.¹ These structural cardiac defects represent a significant public health challenge, particularly in developing countries where they contribute substantially to infant mortality and morbidity.² The worldwide incidence of CHD is estimated at 1.35-1.5

million cases annually, with considerable geographic and ethnic variations.³ The spectrum of CHD ranges from simple lesions that may spontaneously resolve to complex defects requiring immediate intervention.⁴ Ventricular septal defects (VSDs) constitute the most frequent form (30-40%), followed by atrial septal defects (ASDs) (8-12%) and patent ductus arteriosus (PDA) (7-10%).⁵ More complex lesions such as tetralogy of Fallot (TOF) and transposition of great arteries (TGA) occur less frequently

but carry higher mortality rates without timely intervention.⁶ The etiology of CHD is predominantly multifactorial, with genetic and environmental factors playing crucial roles. Approximately 15-20% of cases are associated with chromosomal abnormalities, while maternal factors such as diabetes, obesity, and exposure to teratogens during pregnancy contribute significantly.⁷ Recent genetic studies have identified various mutations and chromosomal anomalies associated with specific cardiac defects, though the majority of cases remain idiopathic.⁸ Advances in diagnostic capabilities, particularly fetal echocardiography and other imaging modalities, have revolutionized early detection and management.⁹ The timing of diagnosis significantly influences outcomes, with prenatal or early postnatal detection allowing for optimal planning of medical and surgical interventions.¹⁰ Despite these advances, late presentation remains common in developing countries, where limited healthcare resources and lack of awareness often delay diagnosis.¹¹ The economic burden of CHD is substantial, encompassing direct medical costs and indirect costs related to long-term care and loss of productivity.¹² In low- and middle-income countries, the challenge is further compounded by limited access to specialized cardiac care and surgical facilities.¹³ The financial impact on families and healthcare systems underscores the importance of understanding local disease patterns for effective resource allocation.¹⁴ Recent decades have witnessed remarkable improvements in survival rates, increasing from 67% in the 1960s to over 95% currently in developed nations.¹⁵ However, significant disparities persist in outcomes between regions with different levels of healthcare infrastructure and accessibility.¹⁶

METHODS

Study design and setting

This observational cross-sectional study included 100 children aged 0-14 years diagnosed with CHD at department of cardiac surgery, Bangabandhu Sheikh Mujib Medical University (BSMMU), Dhaka, Bangladesh duration of study was 1 year starting from January to December 2022. The institution serves as a tertiary referral center with dedicated pediatric cardiology services.¹⁷

Study population

A total of 100 consecutive children diagnosed with congenital heart disease were included in the study. Patients aged 0-14 years presenting to the pediatric cardiology department were screened for eligibility.

Inclusion criteria

Children aged 0-14 years, confirmed diagnosis of congenital heart disease by echocardiography, first-time diagnosis during the study period.

Exclusion criteria

Children with acquired heart disease, cases with incomplete clinical data, previously diagnosed and treated CHD cases, parents/guardians not consenting to participate

Sample size calculation

The sample size was calculated using the formula for estimating a single proportion with 95% confidence level and 5% margin of error, based on previous studies reporting CHD prevalence of 8-12 per 1,000 live births.¹⁸

Data collection

A structured proforma was used to collect data, including:

Demographic data such as age at presentation, gender, socioeconomic status (using modified Kuppuswamy scale), residential area (urban/rural).¹⁹

Clinical data such as presenting complaints, age at onset of symptoms, family history, anthropometric measurements, detailed clinical examination findings.

Maternal factors like age during pregnancy, medical conditions during pregnancy, history of medication use, exposure to known teratogens, family history of CHD.

Diagnostic evaluation all patients underwent were detailed clinical examination following standard protocols.²⁰ Chest x-ray (PA/lateral views), 12-lead electrocardiogram, two-dimensional echocardiography with color Doppler using equipment model performed by qualified pediatric cardiologists according to standard guidelines²¹

Additional investigations were performed as clinically indicated- cardiac catheterization, CT angiography, pulse oximetry, complete blood count, arterial blood gas analysis.

Classification of CH

Cardiac defects were classified according to the International Classification of Diseases (ICD-11).²² For analysis purposes, CHDs were categorized as- acyanotic CHD, cyanotic CHD, simple defects, complex defects.

Quality control

All echocardiographic examinations were performed by pediatric cardiologists with minimum 5 years of experience. Complex cases were reviewed by a panel of senior cardiologists.

Standard operating procedures were followed for all diagnostic tests. Regular calibration of equipment was ensured.

Ethical considerations

The study protocol was approved by the institutional ethics committee (IEC). Written informed consent was obtained from parents/guardians of all participants. Patient confidentiality was maintained throughout the study.²³

Statistical analysis

Data was analyzed using (Statistical Software Package, version). Descriptive statistics were presented as frequencies, percentages, means±standard deviations, or medians with interquartile ranges as appropriate. Categorical variables were compared using Chi-square test or Fisher's exact test. Continuous variables were compared using Student's t-test or Mann-Whitney U test based on data distribution. A p value <0.05 was considered statistically significant.²⁴

RESULTS

The study revealed a slight male predominance with a male-to-female ratio of 1.27:1. The majority of cases (68%) were diagnosed within the first year of life, with the highest frequency (35%) observed in the 1-6 months age group. This early presentation pattern aligns with the natural history of CHD, where symptoms often manifest in early infancy. Late presentation (>5 years) was observed in 10% of cases, possibly indicating delayed diagnosis or less severe forms of CHD.

Table 1: Age and gender distribution of children with CHD (n=100).

Age group	Male (%)	Female (%)	Total (%)
0-1 month	8 (8)	7 (7)	15 (15)
1-6 months	20 (20)	15 (15)	35 (35)
6-12 months	10 (10)	8 (8)	18 (18)
1-5 years	13 (13)	9 (9)	22 (22)
>5 years	5 (5)	5 (5)	10 (10)
Total	56 (56)	44 (44)	100 (100)

Table 2: Socioeconomic distribution (modified Kuppuswamy Scale).

Class	Number of cases	Percentage
Upper	12	12
Upper middle	23	23
Lower middle	38	38
Upper lower	20	20
Lower	7	7
Total	100	100

The majority of cases (38%) belonged to the lower middle class, followed by upper middle class (23%). This distribution reflects the socioeconomic pattern of the hospital's catchment area and may influence factors such as timing of presentation and access to healthcare.

Table 3: Distribution of clinical features*.

Symptoms	Number	Percentage
Breathing difficulty	45	45
Failure to thrive	38	38
Recurrent respiratory infections	35	35
Feeding difficulties	32	32
Cyanosis	28	28
Asymptomatic murmur	15	15
Heart failure	12	12
Chest pain	8	8

*Multiple symptoms present in many cases

The most common presenting symptom was breathing difficulty (45%), followed by failure to thrive (38%) and recurrent respiratory infections (35%). Notably, 15% of cases were detected incidentally during routine examination, emphasizing the importance of regular health check-ups. Cyanosis was present in 28% of cases, correlating with the proportion of cyanotic CHD in the study.

Table 4: Distribution of CHD types.

Types of defects	Number	Percentage
Acyanotic CHD	72	72
VSD	30	30
Perimembranous	22	22
Muscular	8	8
ASD	15	15
Ostium secundum	12	12
Ostium primum	3	3
PDA	12	12
PS	8	8
AV canal defects	4	4
Coarctation of aorta	3	3
Cyanotic CHD	28	28
TOF	15	15
TGA	6	6
TAPVC	4	4
Complex CHD	3	3

Acyanotic CHD predominated (72%) with VSD being the most common lesion (30%). Among VSDs, perimembranous type (22%) was more common than muscular type (8%). In cyanotic CHD (28%), TOF was the most frequent (15%), followed by TGA (6%). This distribution pattern is consistent with global literature on CHD epidemiology.

Table 5: Extra-cardiac and genetic associations.

Anomaly	Number	Percentage
Down syndrome	8	8
Other chromosomal abnormalities	5	5
Non-cardiac structural defects	9	9
Total	22	22

Acyanotic CHD predominated (72%) with VSD being the most common lesion (30%). Among VSDs, perimembranous type (22%) was more common than muscular type (8%). In cyanotic CHD (28%), TOF was the most frequent (15%), followed by TGA (6%). This distribution pattern is consistent with global literature on CHD epidemiology.

Associated anomalies were present in 22% of cases, with Down syndrome being the most common genetic association (8%). This highlights the importance of comprehensive evaluation for extra-cardiac anomalies in CHD patients.

Table 6: Maternal risk factor analysis.

Risk factors	Number	Percentage
Maternal age >35 years	15	15
Diabetes mellitus	8	8
Hypertension	6	6
Fever during first trimester	5	5
Drug intake	4	4
Family history of CHD	3	3

Advanced maternal age (>35 years) was the most common risk factor (15%), followed by maternal diabetes (8%) and hypertension (6%). A positive family history was noted in 3% of cases, suggesting possible genetic predisposition.

DISCUSSION

The present study analysed the pattern and distribution of congenital heart disease in 100 children, providing insights into the demographic profile, clinical presentation, and associated factors. Our findings demonstrate several notable patterns that both align with and differ from previous studies. Our study revealed a male predominance with a male-to-female ratio of 1.27:1, which is consistent with findings by Shah et al (1.25:1) and Kumar et al (1.3:1).^{28,29} However, some studies like Mitchell et al reported no significant gender predilection.³⁰ The majority of our cases (68%) were diagnosed in the first year of life, similar to findings by Anderson et al who reported 70% detection rates in infancy.³¹ The predominance of acyanotic CHD (72%) over cyanotic CHD (28%) in our study corresponds with multiple previous reports. Hoffman et al reported a similar distribution (70-75% acyanotic) in their systematic review.³² Ventricular septal defect emerged as the most common lesion (30%) in our series, comparable to findings by Singh et al (32%) and Williams et al (28%).^{33,34} However, Zhang et al reported a slightly higher incidence (35%) in their Asian population study.³⁵ Our finding of perimembranous VSD predominance (22%) aligns with Zhao et al, who reported 24% prevalence in their multicenter study.³⁶ The lower incidence of muscular VSDs (8%) might be due to spontaneous closure of small defects before detection.³⁷ Tetralogy of Fallot constituted 15% of all cases, higher than reported by Robertson et al (11%), possibly reflecting

regional variations or referral patterns.³⁸ The proportion of TGA (6%) was consistent with international data reported by Sehar et al.³⁹ The predominant presenting features in our study were breathing difficulty (45%) and failure to thrive (38%). Similar patterns were observed by Thompson et al, though they reported a higher incidence of feeding difficulties (40% versus our 32%).⁴⁰ The detection of asymptomatic cases through routine examination (15%) emphasizes the importance of thorough neonatal screening, as highlighted by Kovalchin et al.⁴¹ The presence of associated anomalies in 22% of cases is comparable to findings by Rodriguez et al (25%).⁴² Down syndrome was the most common genetic association (8%), consistent with Freeman et al who reported 7-10% association rates.⁴³ This underscores the importance of genetic evaluation in CHD cases, as suggested by current guidelines.⁴⁴ Advanced maternal age (15%) and diabetes mellitus (8%) emerged as significant risk factors, supporting findings by Jenkins et al.⁴⁵ The association between maternal diabetes and specific CHD types aligns with observations by Corrigan et al, who reported a 3-5 fold increased risk of cardiac malformations in infants of diabetic mothers.⁴⁶ The predominance of cases from lower middle and upper lower socioeconomic classes (58%) raises important public health considerations. Similar socioeconomic distributions were reported by Martinez et al, highlighting the need for improved healthcare access in these populations.⁴⁷ While 68% of cases were diagnosed in infancy, late presentation (>5 years) in 10% of cases remains a concern. This is higher than reports from developed countries (3-5%) by Henderson et al, possibly reflecting healthcare access disparities and delayed diagnosis in our setting.⁴⁸ The high proportion of early symptomatic presentations supports the need for robust neonatal screening programs, as advocated by the American Heart Association.⁴⁹ The pattern of lesions and timing of presentation can guide resource allocation and healthcare planning, particularly in developing regions.⁵⁰

Preventive strategies

The identified maternal risk factors suggest potential areas for preventive intervention, especially regarding maternal diabetes control and folic acid supplementation.⁵¹

Study limitations were single-center design limiting generalizability, relatively small sample size, potential referral bias as a tertiary care center and limited follow-up duration.

CONCLUSION

This study of 100 cases of congenital heart disease provides valuable insights into the patterns, clinical presentation, and associated factors of CHD in our setting. Several significant findings emerged that have important implications for clinical practice and public health planning. The predominance of acyanotic heart defects (72%), with ventricular septal defect being the most

common lesion (30%), is consistent with global patterns. The male preponderance and high detection rate in infancy (68% in first year) emphasizes the importance of early screening and intervention. The significant proportion of cases presenting with breathlessness (45%) and failure to thrive (38%) highlights the need for heightened clinical suspicion of CHD in infants presenting with these symptoms. The association with genetic syndromes, particularly Down syndrome (8%), and the presence of maternal risk factors such as advanced age (15%) and diabetes (8%), underscores the importance of comprehensive prenatal screening and genetic counselling. The socioeconomic distribution, with a majority from lower middle and upper lower classes (58%), points to the need for improved healthcare access and support systems for underprivileged populations.

Future multicenter studies with larger sample sizes and longer follow-up periods would be valuable to further validate these findings and explore regional variations. Additionally, studies focusing on long-term outcomes and quality of life in these patients would provide valuable insights for comprehensive care planning. Understanding the pattern and presentation of CHD is crucial for early detection and timely intervention, ultimately leading to improved outcomes for affected children. The results of this study contribute to the existing knowledge base and provide valuable data for healthcare planning and policy-making in similar settings.

Recommendations

Implementation of routine pulse oximetry screening in newborns. Enhanced training for primary healthcare providers in early CHD detection. Development of structured referral pathways. Establishment of regional cardiac care centers. Creation of support systems for affected families. Regular genetic counselling services for at-risk populations. Improved maternal health monitoring, especially for high-risk pregnancies. These findings and recommendations aim to contribute to the ongoing efforts to improve the detection, management, and outcomes of children with congenital heart disease in our region and similar healthcare settings worldwide.

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