An Observational Study of Frequency and Profile of Congenital Heart Diseases in Neonates Born in a Tertiary Care Hospital

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ABSTRACT:

Background and Objective: Congenital heart disease (CHD) is the most common congenital anomaly and a major cause of neonatal morbidity and mortality. Early identification through systematic clinical evaluation and pulse oximetry screening facilitates timely intervention and improves outcomes. This study assessed the frequency, clinical profile, diagnostic findings, management, and outcomes of CHD among neonates born at a tertiary care hospital.

Methods: This prospective observational study was conducted over 12 months in a tertiary care center. A total of 165 inborn neonates with structurally confirmed CHD on two-dimensional echocardiography were enrolled. All neonates underwent detailed clinical examination, pulse oximetry screening, and echocardiographic evaluation when indicated. Clinical characteristics, echocardiographic findings, management, and outcomes were analyzed using descriptive statistics.

Results: Among 8,534 live births, 165 neonates were diagnosed with CHD, yielding an overall prevalence of **1.9%**. The prevalence was higher among NICU admissions (5.8%) than postnatal ward neonates (0.7%). Most cases (84.2%) were diagnosed during the first week of life. Tachypnea (58.7%) was the commonest presenting symptom, while cardiac murmur (89.7%) was the predominant clinical sign. Prenatal diagnosis was achieved in only 9.1% of cases. Pulse oximetry screening identified CHD in 60% of neonates with positive screening results. Acyanotic CHD accounted for 82.5% of cases, with ventricular septal defect (24.8%) being the commonest lesion, followed by patent ductus arteriosus (21.8%) and

atrial septal defect (17.5%).

Conclusions: Congenital heart disease was diagnosed in 1.9% of neonates, with most cases identified during the first week of life. Acyanotic lesions predominated, particularly ventricular septal defects.

Keywords: Congenital heart disease; Neonate; Pulse oximetry screening; Echocardiography; Critical congenital heart disease.

Introduction

Congenital heart disease (CHD) comprises structural abnormalities of the heart or great vessels present at birth and represents the most common congenital anomaly, accounting for nearly one-third of all birth defects.[1] In India, the estimated birth prevalence of CHD is approximately 9 per 1,000 live births, resulting in nearly 240,000 affected newborns annually. Congenital cardiovascular malformations remain one of the leading causes of infant mortality during the first year of life.[2] Advances in prenatal imaging, neonatal care, and pediatric cardiac surgery have substantially improved survival; however, timely diagnosis remains essential to ensure early intervention and better clinical outcomes.

Prenatal ultrasonography has become an integral component of fetal assessment and enables the antenatal detection of many cardiac anomalies. Studies from India have demonstrated that early prenatal identification of CHD is an important step in reducing neonatal morbidity and mortality.[3,4] Nevertheless, antenatal screening and routine postnatal clinical examination alone fail to identify a considerable proportion of affected newborns. More than half of neonates with CHD may remain undiagnosed on clinical examination because many lesions are asymptomatic or present with subtle findings during the immediate neonatal period.

Although echocardiography is the gold standard for diagnosing CHD, it is resource-intensive and impractical as a universal screening tool for all newborns. Delayed diagnosis of critical congenital heart disease (CrCHD) may result in heart failure, circulatory collapse, severe hypoxemia, or even death. Therefore, reliable, inexpensive, and easily applicable screening methods are required to facilitate early detection before the onset of life-threatening complications. [5]

Pulse oximetry has emerged as a simple, non-invasive, cost-effective, and feasible screening modality for the early identification of CrCHD. It detects hypoxemia associated with many critical cardiac lesions before clinically apparent cyanosis develops. Neonates with CHD commonly present with feeding difficulty, tachypnea, cyanosis, congestive heart failure, or cardiovascular collapse, although presentations may vary depending on the underlying defect. The evaluation of suspected CHD includes careful clinical examination, preductal and postductal oxygen saturation measurement, four-limb blood pressure assessment, chest radiography, and confirmation by echocardiography with Doppler imaging. [6]

There is limited regional data regarding the clinical spectrum and outcomes of CHD in neonates. Therefore, the present study was undertaken to evaluate the clinical presentation, age at diagnosis, examination findings, diagnostic modalities, and outcomes, including mortality, among neonates diagnosed with congenital heart disease.

Materials and Methods

This prospective observational study was conducted over a period of 12 months at a tertiary care center. Consecutive sampling was employed to recruit eligible participants. The sample size was calculated using Yamane's formula based on an expected CHD prevalence of 10.7%, 95% confidence level, and 5% margin of error, yielding a minimum sample size of 147. A total of 165 inborn neonates fulfilling the eligibility criteria were included in the study.

All inborn neonates diagnosed with structural congenital heart disease (CHD) on two-dimensional (2D) echocardiography and whose parents or guardians provided written informed consent were included. Outborn babies and neonates whose parents or guardians declined consent were excluded.

After obtaining informed consent, all enrolled neonates underwent a detailed general and cardiovascular examination. Clinical assessment included evaluation for cyanosis, visible precordial pulsations, apical impulse, peripheral pulses, blood pressure, precordial heave or thrill, heart sounds, and the presence of systolic or diastolic murmurs.

Pulse oximetry screening (POxS) was performed after 24 hours of life or immediately before discharge if discharge occurred earlier. Oxygen saturation was measured in the right hand (preductal) and either foot (postductal). A screening result was considered negative when oxygen saturation was ≥95% in both limbs with a preductal-postductal difference of ≤3%. A result was considered positive when saturation was ≤89% in either limb. Neonates with saturation between 90% and 94% or a preductal-postductal difference of ≥4% underwent repeat screening after one hour. Persistent abnormal results were considered positive, and these neonates underwent further evaluation including chest radiography, blood pressure measurement, and 2D echocardiography.

Two-dimensional echocardiography was performed by a pediatric cardiologist in neonates with abnormal pulse oximetry screening, cyanosis (SpO_2 <94% on room air), unexplained respiratory distress, pathological murmurs, abnormal heart sounds, abnormal blood pressure, differential peripheral pulses, abnormal chest radiographs, prolonged ventilatory support, maternal gestational diabetes, syndromic features associated with CHD (e.g., Down syndrome), or those fulfilling Nadas' criteria.

Echocardiographic findings were reviewed to confirm structural CHD and classify lesions as cyanotic or acyanotic. Neonates with confirmed structural congenital heart disease were included in the final analysis and managed according to the recommendations of the pediatric cardiologist.

RESULTS

Table 1. Baseline Characteristics of Neonates with Congenital Heart Disease (n = 165)

Variable	n (%)
Total live births screened	8,534
Neonates with CHD	165 (1.9)
CHD among NICU admissions (n=1900)	112 (5.8)
CHD among postnatal ward neonates (n=6634)	53 (0.7)
Diagnosed during first week	139 (84.2)
Male sex	89 (53.9)
Female sex	75 (45.5)
Preterm	82 (49.7)
Term	83 (50.3)
Birth weight 1.5–2.49 kg	83 (50.3)
LSCS delivery	100 (60.6)
Mean age at diagnosis	1.17 ± 0.41 weeks

A total of 8,534 neonates were screened during the 12-month study period, including 1,900 NICU admissions and 6,634 postnatal ward neonates. Structural congenital heart disease (CHD) was diagnosed in 165 neonates, giving an overall prevalence of 1.9%. The prevalence was higher among NICU admissions (5.8%) compared to postnatal ward neonates (0.7%). The majority of

CHD cases (84.2%) were diagnosed during the first week of life (mean age: 1.17 ± 0.41 weeks). There was a slight male predominance (53.9%). Preterm (49.7%) and term (50.3%) neonates were almost equally represented. Half of the neonates (50.3%) had a birth weight between 1.5–2.49 kg, while 60.6% were delivered by lower segment cesarean section.

Table 2. Clinical Presentation and Associated Risk Factors

Variable	n (%)
Presenting symptoms	
Tachypnea	97 (58.7)
Cyanosis	26 (15.7)
Feeding difficulty	15 (9.0)
Asymptomatic	61 (36.9)
Maternal risk factors	
No maternal risk factor	128 (77.5)
Pregnancy-induced hypertension	17 (10.3)
Gestational diabetes mellitus	9 (5.4)
Hypothyroidism	7 (4.2)
Prenatal diagnosis by ANC scan	15 (9.1)

The most common presenting symptom was tachypnea (58.7%), followed by cyanosis (15.7%) and feeding difficulty (9.0%); however, 36.9% of neonates were asymptomatic at presentation. Maternal risk factors were absent in 77.5% of cases. Among identified maternal conditions, pregnancy-induced hypertension (10.3%), gestational diabetes mellitus (5.4%), and hypothyroidism (4.2%) were the most frequent. Prenatal diagnosis was achieved in only 9.1% of cases. Prematurity (49.6%) was the commonest associated comorbidity, followed by respiratory distress syndrome (18.1%) and sepsis (6.6%).

Table 3. Clinical Examination Findings and Investigations

Variable	n (%)
Murmur	148 (89.7)
Tachycardia	97 (58.7)
Tachypnea	97 (58.7)
Abnormal heart sounds	19 (11.5)
Congestive heart failure	12 (7.3)
Oxygen requirement	104 (63.0)
Cardiomegaly on chest X-ray	34 (20.6)
Pulmonary plethora	30 (18.2)

Variable	n (%)
Right axis deviation on ECG	6 (3.6)

On clinical examination, cardiac murmur was the most frequent finding (89.7%), followed by tachycardia (58.7%) and tachypnea (58.7%). Abnormal heart sounds were detected in 11.5% and congestive heart failure in 7.3% of neonates. Pulse oximetry screening was performed in 6,639 postnatal ward neonates; five newborns screened positive, of whom three (60%) were confirmed to have CHD on echocardiography. Oxygen therapy was required in 63% of patients. Cardiomegaly (20.6%) and pulmonary plethora (18.2%) were the most common chest radiographic abnormalities, while right axis deviation (3.6%) was the predominant electrocardiographic finding.

Table 4. Distribution of Congenital Heart Disease

Type of CHD	n (%)
Acyanotic CHD	136 (82.5)
Cyanotic CHD	29 (17.5)

Table 5. Most Common Echocardiographic Diagnoses

Lesion	n (%)
Ventricular septal defect (VSD)	41 (24.8)
Patent ductus arteriosus (PDA)	36 (21.8)
Atrial septal defect (ASD)	29 (17.5)
d-Transposition of great arteries (d-TGA)	5 (3.0)
Tetralogy of Fallot (TOF)	4 (2.4)
Truncus arteriosus	3 (1.8)
TAPVC	3 (1.8)
Other lesions	44 (26.7)

Table 4 and 5 Echocardiography demonstrated that acyanotic CHD (82.5%) was considerably more common than cyanotic CHD (17.5%). Ventricular septal defect (24.8%) was the most frequent lesion, followed by patent ductus arteriosus (21.8%) and atrial septal defect (17.5%). Among cyanotic lesions, d-transposition of the great arteries (17.2%) and tetralogy of Fallot (13.7%) were the most common.

Table 6. Management According to Type of Congenital Heart Disease

Management	Acyanotic CHD (n=136)	Cyanotic CHD (n=29)
Medical management	47 (34.5)	29 (100)
Surgical intervention	2 (1.4)	26 (89.6)
No intervention	91 (66.9)	0

Table 7. Clinical Outcomes According to Type of Congenital Heart Disease

Outcome	Overall n (%)	Acyanotic CHD n (%)	Cyanotic CHD n (%)
Discharged	128 (77.6)	124 (91.1)	1 (3.4)
Referred	19 (11.5)	1 (0.7)	18 (62.0)
LAMA	7 (4.2)	5 (3.6)	2 (6.8)
Death	14 (8.5)	6 (4.4)	8 (27.5)

Table 6 and 7- Regarding management, 34.5% of acyanotic CHD cases required medical treatment, while only 1.4% underwent surgery. In contrast, all cyanotic CHD patients required medical management and 89.6% required surgical intervention. Overall, 77.6% of neonates were discharged, 11.5% were referred to higher centers, 4.2% left against medical advice, and 8.5% died. Mortality was substantially higher in cyanotic CHD (27.5%) than in acyanotic CHD (4.4%), whereas discharge rates were higher among acyanotic CHD patients (91.1% vs. 3.4%).

Discussion

The present study evaluated the clinical profile, spectrum, diagnosis, management, and outcomes of congenital heart disease (CHD) in 165 neonates diagnosed at a tertiary care center over one year. The overall prevalence of CHD was 1.9% among all live births, with a substantially higher frequency among NICU admissions (5.8%) than postnatal ward neonates (0.7%). This higher NICU prevalence is comparable to previous neonatal studies by Shima et al. and Ekici et al., who reported CHD frequencies ranging from 3–4.5% among NICU admissions. The increased detection in NICU reflects the higher proportion of symptomatic and high-risk neonates requiring intensive care.[7,8]

Early diagnosis was a major finding of this study, with 84.2% of neonates diagnosed during the first week of life (mean age 1.17 ± 0.41 weeks). Nearly all cyanotic CHDs were recognized during the neonatal period, emphasizing the effectiveness of systematic neonatal screening and vigilant cardiovascular examination. The earlier identification of acyanotic lesions in the present study may be attributed to routine screening protocols and prompt echocardiographic evaluation of abnormal clinical findings.

Gestational age distribution was almost equal between term and preterm infants, whereas nearly three-fourths of affected neonates were of low birth weight. Similar observations have been reported by Seon Young Cho et al. and Amroliwala et al., supporting the established association between low birth weight and congenital cardiac anomalies. The comparable representation of term and preterm neonates in the present study is likely due to inclusion of both NICU and postnatal ward populations rather than NICU patients alone. [9,10]

Maternal risk factors were identified in approximately one-fifth of cases, with pregnancy-induced hypertension, gestational diabetes mellitus, and hypothyroidism being the most common. However, most mothers (77.5%) had no identifiable risk factors, emphasizing that CHD frequently occurs even in apparently low-risk pregnancies and reinforcing the need for universal newborn screening. Prenatal diagnosis was achieved in only 9.1% of neonates, indicating that despite advances in fetal ultrasonography, antenatal detection remains suboptimal in routine clinical practice.

Tachypnea was the most frequent presenting symptom, followed by cyanosis and feeding difficulty, while more than one-third of neonates remained asymptomatic. These observations agree with previous studies in which respiratory distress was the predominant initial manifestation of neonatal CHD. Importantly, the substantial proportion of asymptomatic infants highlights the limited sensitivity of clinical examination alone and underscores the importance of structured screening strategies. Cardiac murmur was the most common examination finding (89.7%), followed by tachycardia and tachypnea, emphasizing that careful cardiovascular examination remains the cornerstone for early recognition of congenital heart disease.

Pulse oximetry screening demonstrated good diagnostic utility in the present study. Among 6,639 newborns screened, five tested positive and three were confirmed to have CHD on echocardiography, yielding a positive predictive value of 60%. These findings are comparable with

previously published studies reporting detection rates between 65% and 72%. [11,12] The results support current recommendations advocating pulse oximetry as a simple, inexpensive, non-invasive, and effective screening tool for early detection of critical congenital heart disease before clinical deterioration occurs.

Consistent with previous epidemiological reports, acyanotic CHD predominated (82.5%), whereas cyanotic lesions accounted for 17.5% of cases. Ventricular septal defect was the commonest lesion, followed by patent ductus arteriosus and atrial septal defect. The relatively high frequency of PDA may reflect inclusion of all structurally significant PDAs, particularly among premature neonates, whereas some earlier studies excluded small PDAs. [13,14] Among cyanotic lesions, d-transposition of the great arteries was the leading diagnosis, followed by tetralogy of Fallot. This differs from studies including older infants and children, where tetralogy of Fallot predominates, suggesting that d-transposition is more likely to present during the immediate neonatal period because of its early hemodynamic consequences.

Management requirements reflected disease severity. Approximately one-third of acyanotic CHD cases required medical therapy, while only a small proportion underwent surgery. In contrast, all neonates with cyanotic CHD required medical stabilization, and nearly 90% subsequently required surgical intervention or referral to specialized cardiac centers. These findings emphasize the importance of prompt diagnosis and timely referral for definitive management of critical congenital cardiac lesions.

CONCLUSION

The frequency of congenital heart disease (CHD) in this tertiary care hospital was 165 (1.9%) out of 8534 registered neonates. 5.8% of the neonates admitted in the NICU were diagnosed with CHD. Majority of the cases were diagnosed with in 1st week of life .Male predominance was seen in this study. The proportion of CHD was more among low birth weight neonates . The most common presenting compliant was fast breathing followed by cyanosis and feeding difficulty Most of the patients diagnosed with CHD were born through LSCS. 22.5% of the neonates with CHD had significant maternal history. 22.8% patients were born through consanguineous marriage ,7.8% of the patients had family history of CHD. The majority of the neonates didn't show any extra cardiac anomalies, but most of the neonates in the study had associated co morbidities. The most common clinical examination finding among our patients was murmur, followed by tachycardia and tachypnoea. A significant number of patients had positive maternal history, consanguinity and h/o CHD in the siblings .60% of the patients who showed fail on POxS were diagnosed with CHD by 2D ECHO. Acyanotic CHD was more common than Cyanotic CHD . The most common type among Acyanotic group was VSD followed by PDA and ASD in this study .Among Cyanotic group most common type was dTGA followed by TOF and truncous arteriosus, TAPVC. Most of the neonates diagnosed with acyanotic CHD didn't require any intervention. Among cyanotic variety most of the patients required surgical and medical management.

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