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ORIGINAL ARTICLE

Pattern of congenital heart defects among neonates admitted to Bahawal Victoria Hospital.

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ABSTRACT… Objective: To determine the prevalence and pattern of CHDs among neonates admitted to a tertiary care hospital in Bahawalpur. **Study Design:** Analytical Cross-sectional study. **Period:** September 2025 to March 2026. **Setting:** Pediatric Medicine Department of Bahawal Victoria Hospital/Quaid-e-Azam Medical College, Bahawalpur. **Methods:** A total of 357 consecutively enrolled neonates (≥1000 g, ≥5-minute APGAR score≥5, gestational age up to 28 days) underwent standardized echocardiographic assessment by a consultant pediatric cardiologist. Demographic and clinical data were recorded, and CHDs were classified as acyanotic or cyanotic. Data were analyzed using SPSS 27.0. **Results:** Echocardiography identified congenital heart defects in 38 neonates, yielding 10.6% frequency. Acyanotic lesions predominated (81.6%), with ventricular septal defect being most common (47.4%). Family history showed a strong association with CHD ($p<0.001$), while lower five-minute APGAR scores demonstrated a modest correlation ($p=0.049$). Sex and birth weight were not significant predictors. Cardiac murmurs, cyanosis, tachypnea, and feeding difficulties were significantly more prevalent among affected neonates (all $p<0.001$), highlighting essential clinical screening indicators. **Conclusion:** This study demonstrates high frequency of congenital heart disease among neonates predominantly acyanotic and strongly associated with family history and clinical indicators such as murmurs and respiratory compromise.

Key words: Congenital Heart Defects, Echocardiography, Infant, Newborn, Prevalence, Pakistan.

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INTRODUCTION

Congenital heart defects (CHDs) are structural or functional anomalies of the heart or great vessels that occur in embryogenesis and can severely affect cardiovascular functioning.[1,2] The etiology of CHDs is multifactorial and generally complex, comprising genetic predisposition and environmental factors, including maternal infections, uncontrolled diabetes, or teratogenic exposure during pregnancy.[3,4] CHDs are divided into two groups: acyanotic and cyanotic. Acyanotic defects include ventricular septal defect (VSD), patent ductus arteriosus (PDA) and atrial septal defect (ASD), which are usually associated with left to right shunting, resulting in increased blood flow to the lungs without systemic oxygen desaturation. On the contrary, cyanotic defects, such as Tetralogy of Fallot (TOF) and transposition of the great arteries (TGA), are characterized by right to left shunting or the mixing of deoxygenated blood with the systemic circulation, resulting in a lack of oxygen saturation and clinical cyanosis.[5,6] International studies have shown the prevalence of congenital heart disease (CHD) to range between 8.6 and 9.38 per 1,000 live births.[7,8] In pediatric populations, research involving children from birth to 10 years revealed that 70.67% had acyanotic CHD, while 29.3% had cyanotic CHD.[9] Study of 624 children aged between birth and 10 years found that 87% had congenital heart disease and 29.3% had acquired heart disease. Out of the CHD cases, 73.1 per cent were acyanotic, and 26.9 per cent were cyanotic. The most frequent lesion was a ventricular septal defect (33%), then an atrial septal defect (14.9%), and finally a patent ductus arteriosus (13.1%). The most common cyanotic defects were tetralogy of Fallot (TOF) (10.1%) and transposition of the great arteries (TGA) (7.4%).[10]

The rationale of the current research was to generate local evidence based data about the prevalence of congenital heart defects among the neonatal population of South Punjab, a region with scarce healthcare facilities and poorly reported disease burden.

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Timely referral, intervention, and better long-term outcomes of CHDs can be achieved by early diagnosis of the condition during the neonatal period. The study helps address the existing knowledge gap and provides a basis for evidence-based clinical practice, effective resource allocation, and the development of locally suitable diagnostic and management guidelines.

METHODS

This analytical cross-sectional study was conducted in the Department of Pediatric Medicine, Bahawal Victoria Hospital, Bahawalpur, from September 2025 to March 2026, following approval from Institutional Review Board (Reference No. 2399/DME/QAMC Bahawalpur dated. 23-04-2024). The sample size of 357 was determined based on calculations that considered 95% confidence level and absolute precision of 1%, with an estimated prevalence of congenital heart disease of 9.38 per 1,000 (0.938%).[7] Neonates were included by using a non-probability consecutive sampling technique. The inclusion criteria included neonates of either gender, with a gestational age of at least 28 days, a ≥5 APGAR score at 5 minutes, and a birth weight of ≥1000 grams. The exclusion criteria were patients with a bicuspid aortic valve without aortic valve stenosis or mitral valve prolapse, patients with cardiac malposition without structural heart disease, preterm infants and neonates with acquired heart diseases.

Informed consent was taken from the parents or legal guardians of the neonates. Basic data, i.e., name, days of age, gestational age, gender, birth weight, current weight and any family history of congenital heart disease, were noted. An experienced consultant pediatric cardiologist of over five years performed echocardiographic assessments on a standardized protocol and with a suitable pediatric transducer to detect and categorize congenital heart defects based on the operational definitions. The diagnosis of defects was made as either acyanotic or cyanotic, based on the results of echocardiography and related clinical manifestations. Specific abnormalities, such as ASD, VSD, PDA, TOF, TGA, and TAPVR, were recorded according to standard protocols.

The data analysis was conducted using SPSS version 27.0. Numerical variables were reported in the form of mean ± standard deviation, whereas categorical variables were reported in frequencies and percentages. Associations were determined using the chi-square test, with a p-value of 0.05 considered statistically significant.

RESULTS

The mean age of participants was 18.4 ± 9.2 days, with a mean gestational age of 38.1 ± 1.8 weeks. Males comprised 55.5% (n = 198) of the sample, while females accounted for 44.5% (n = 159). Birth weight distribution indicated that 52.3% of neonates weighed ≥2500 g (mean: 2685 ± 512 g), 35.9% weighed 1500–2499 g, and 11.8% fell within the 1000–1499 g range. Mean current weight at enrollment was 2890 ± 498 g. The majority of participants (82.4%, n = 294) achieved a 5-minute APGAR score of 8–10, and 8.1% (n = 29) had a documented family history of congenital heart disease (Table-I).

Echocardiography confirmed structural cardiac anomalies in 38 neonates, yielding prevalence of 10.6%. Acyanotic lesions predominated, representing 81.6% (n = 31) of all diagnosed cases. Ventricular septal defect was the most frequently identified abnormality (47.4%, n = 18), followed by atrial septal defect (18.4%, n = 7) and patent ductus arteriosus (15.8%, n = 6). Cyanotic defects accounted for 18.4% (n = 7) of diagnoses, with tetralogy of Fallot observed in 3 neonates (7.9%), and transposition of the great arteries and total anomalous pulmonary venous return each identified in 2 cases (5.3% each) (Table-II).

Neonatal sex and birth weight category showed no statistically significant relationship with structural heart disease ($p = 0.764$ for both). In contrast, a positive family history of CHD demonstrated a strong association with defect diagnosis; 23.7% (n = 9) of affected neonates had an affected first- or second-degree relative, compared to 6.3% (n = 20) in the unaffected group ($p < 0.001$). Lower 5-minute APGAR scores (5–7) were modestly associated with CHD, occurring in 28.9% of diagnosed cases versus 16.3% of neonates with normal cardiac anatomy ($p = 0.049$) (Table-III).

A cardiac murmur was auscultated in 89.5% (n = 34) of neonates with confirmed CHD, compared to 12.9% (n = 41) of those without structural defects (p < 0.001). Cyanosis was documented in 31.6% (n = 12) of the CHD patients and 0.9% (n = 3) of the unaffected participants (p < 0.001). Signs of cardiopulmonary compromise, including tachypnea (>60 breaths/min), poor feeding, and failure to thrive, were significantly more prevalent among neonates with structural anomalies (55.3%, 47.4%, and 23.7%, respectively) than among those with normal echocardiograms (8.8%, 11.0%, and 3.8%, respectively; all p < 0.001). Conversely, 87.1% (n = 278) of neonates without CHD remained entirely asymptomatic at enrollment, whereas only 10.5% (n = 4) of those with confirmed defects lacked overt clinical signs (p < 0.001) (Table-IV).

DISCUSSION

This investigation identified a congenital heart disease frequency of 10.6% among neonates evaluated at a tertiary pediatric center in South Punjab, a figure that aligns with hospital-based surveillance data from comparable resource-constrained regions.

TABLE-I

Demographic and baseline clinical characteristics of enrolled neonates (n = 357)

Variable	Category	Frequency (n)	Percentage (%)	Mean ± SD (if applicable)
Age at enrollment (days)	—	—	—	18.4 ± 9.2
Gestational age at birth (weeks)	—	—	—	38.1 ± 1.8
Gender	Male	198	55.5	—
	Female	159	44.5	—
Birth weight (grams)	1000–1499	42	11.8	—
	1500–2499	128	35.9	—
	≥2500	187	52.3	—
	—	—	—	2685 ± 512
Current weight (grams)	—	—	—	2890 ± 498
5-min APGAR score	5–7	63	17.6	—
	8–10	294	82.4	—
Family history of CHD	Yes	29	8.1	—
	No	328	91.9	—

TABLE-II

Classification of diagnosed congenital heart defects by hemodynamic category (n = 38 CHD Cases)

Hemodynamic Category	Specific Defect	Frequency (n)	% of CHD Cases
Acyanotic CHD	Ventricular Septal Defect (VSD)	18	47.4%
	Atrial Septal Defect (ASD)	7	18.4%
	Patent Ductus Arteriosus (PDA)	6	15.8%
	Subtotal	31	81.6%
Cyanotic CHD	Tetralogy of Fallot (TOF)	3	7.9%
	Transposition of Great Arteries (TGA)	2	5.3%
	Total Anomalous Pulmonary Venous Return (TAPVR)	2	5.3%
	Subtotal	7	18.4%
Total CHD Cases		38	100%

TABLE-III

Association between Selected Risk Factors and Congenital Heart Disease Diagnosis

Risk Factor	Category	CHD Present (n=38)	CHD Absent (n=319)	P-Value
Gender	Male	22 (57.9%)	176 (55.2%)	0.764
	Female	16 (42.1%)	143 (44.8%)	
Birth Weight	<2500 g	19 (50.0%)	151 (47.3%)	0.764
	≥2500 g	19 (50.0%)	168 (52.7%)	
Family History of CHD	Yes	9 (23.7%)	20 (6.3%)	<0.001
	No	29 (76.3%)	299 (93.7%)	
5-min APGAR Score	5–7	11 (28.9%)	52 (16.3%)	0.049
	8–10	27 (71.1%)	267 (83.7%)	

TABLE-IV

Clinical presentation at enrollment among neonates with and without CHD

Clinical Feature	CHD Present (n=38)	CHD Absent (n=319)	P-Value
Cardiac murmur	34 (89.5%)	41 (12.9%)	<0.001
Cyanosis	12 (31.6%)	3 (0.9%)	<0.001
Tachypnea (>60/min)	21 (55.3%)	28 (8.8%)	<0.001
Poor feeding	18 (47.4%)	35 (11.0%)	<0.001
Failure to thrive	9 (23.7%)	12 (3.8%)	<0.001
Asymptomatic	4 (10.5%)	278 (87.1%)	<0.001

The elevated rate relative to community based estimates is consistent with referral bias, wherein neonates exhibiting clinical suspicion or perinatal complications are preferentially directed to specialized cardiology services.[11,12] Acyanotic defects predominated, comprising 81.6% of confirmed diagnoses, with ventricular septal defect representing nearly half of all lesions. This distribution mirrors contemporary pediatric cardiology registries across South Asia, where left-to-right shunts consistently constitute the majority of structural anomalies.[13] The proportion of cyanotic defects (18.4%) and the relative frequencies of tetralogy of Fallot, transposition of the great arteries, and total anomalous pulmonary venous return are similarly congruent with regional epidemiological patterns, reinforcing the stability of CHD phenotypic distribution across diverse healthcare settings.[14,15]

Family history emerged as the most robust predictor of structural cardiac anomalies, with affected neonates demonstrating a nearly fourfold higher likelihood of having a first- or second degree relative with CHD. This observation underscores the polygenic and heritable nature of cardiac morphogenesis and aligns with recent genetic epidemiology studies from Pakistan and neighboring countries that report familial clustering in approximately 20–25% of structural heart defect cases.[16,17] In contrast, neonatal sex and birth weight categories showed no statistically significant association with CHD diagnosis, supporting contemporary meta-analyses that indicate these perinatal variables do not independently predict structural malformations in term and late-preterm populations. The modest but significant association with lower 5-minute APGAR scores likely reflects transient hemodynamic compromise rather than a primary etiological driver, as impaired cardiac output in early life can delay physiological transition and depress neonatal responsiveness.[18] Clinically, the high sensitivity of cardiac murmurs (89.5%), tachypnea (55.3%), and feeding difficulties (47.4%) in our cohort reinforces established pediatric guidelines that emphasize systematic auscultation and symptom recognition during routine newborn assessments.[19,20] Notably, 87.1% of neonates without CHD remained entirely asymptomatic, whereas only 10.5% of diagnosed cases lacked overt clinical signs, highlighting the indispensable role of echocardiography in

detecting hemodynamically silent lesions before decompensation occurs.[21,22]

The principal strengths of this study include the application of a standardized echocardiographic protocol performed by a single experienced pediatric cardiologist, which minimizes inter-observer variability and enhances diagnostic reproducibility. Consecutive enrollment reduced selection bias, while comprehensive documentation of perinatal parameters and clinical phenotypes strengthened the validity of observed risk associations. Furthermore, the focus on an underserved geographic region contributes critical localized epidemiological data that has historically been underrepresented in national health registries. Several limitations warrant consideration. The single-center, hospital-based design restricts generalizability to community populations, as referral pathways likely over represent symptomatic or complex presentations. The cross-sectional framework precludes longitudinal tracking of spontaneous closure rates, hemodynamic progression, or long-term surgical outcomes. Additionally, the absence of routine chromosomal or molecular genetic testing limits the ability to correlate phenotypic findings with underlying syndromic or monogenic etiologies. Resource constraints also restricted access to advanced imaging modalities or fetal echocardiography correlation, which could have refined early diagnostic precision.

Future research should prioritize multicenter, population-based surveillance to establish accurate baseline prevalence rates across South Punjab and mitigate referral bias inherent in tertiary care cohorts. Integration of universal pulse oximetry screening alongside targeted echocardiography for high-risk neonates could improve early detection of critical cyanotic lesions before clinical deterioration. Developing standardized referral algorithms and expanding neonatal cardiology training programs in district hospitals would address current diagnostic delays and reduce preventable morbidity. Policymakers and regional health authorities should consider subsidizing genetic counseling and familial screening initiatives, given the strong predictive value of family history. Routine incorporation of structured cardiac assessment into neonatal discharge protocols, coupled with public health campaigns aimed at maternal infection control and glycemic management during pregnancy, would further reduce the regional burden of congenital heart disease.

CONCLUSION

This study demonstrates high frequency of congenital heart disease among neonates predominantly acyanotic and strongly associated with family history and clinical indicators such as murmurs and respiratory compromise. These findings highlight the critical need for standardized neonatal cardiac screening, targeted familial risk assessment and strengthened regional referral networks to optimize early diagnosis and long-term cardiovascular outcomes.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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AUTHORSHIP AND CONTRIBUTION DECLARATION	
1	**Zohaib Akhtar:** Conception of idea, study design, manuscript drafting.
2	**Malik Muhammad Naeem:** Data analysis, interpretation of data.
3	**Himayat Ullah:** Data analysis, manuscript drafting.
4	**Sana Rafiq:** Substantial contributions to data collection.
5	**Bushra Arshad:** Manuscript writing.
6	**Muhammad Soomair Akbar:** Data collection.