DOI: https://doi.org/10.54393/pjhs.v6i7.3362

PAKISTAN JOURNAL OF HEALTH SCIENCES
(LAHORE)
https://thejas.com.pk/index.php/pjhs
ISSN (E): 2790-9352, (P): 2790-9344
Volume 6, Issue 07 (July 2025)

Original Article

OPEN ACCESS

Frequency and Pattern of Congenital Heart Disease in Term Neonates Presenting in Respiratory Distress at Tertiary Care Hospital, Larkana

Zeeshan Ali Memon[1], Dilijan Baloch[1], Muhammad Fareed[1], Sohail Aman Jokhio[2], Sohail Ahmad Soomro[1], Zainab Ayoub[1] and Vijia Kumar Gemnani[2*]

[1]Department of Pediatrics, Children's Hospital, Chandka Medical College, Shaheed Mohtarma Benazir Bhutto Medical University, Larkana, Pakistan
[2]Faculty of Community Medicine and Public Health, Chandka Medical College, Shaheed Mohtarma Benazir Bhutto Medical University, Larkana, Pakistan

ARTICLE INFO

Keywords:
Neonate, Respiratory Distress, Neonatal Screening, Cyanotic Heart Disease

How to Cite:
Memon, Z. A., Baloch, D., Fareed, M., Jokhio, S. A., Soomro, S. A., Ayoub, Z., & Gemnani, V. K. (2025). Frequency and Pattern of Congenital Heart Disease in Term Neonates Presenting in Respiratory Distress at Tertiary Care Hospital, Larkana: Frequency and Pattern of Congenital Heart Disease in Term Neonates. Pakistan Journal of Health Sciences, 6(7), 254-258. https://doi.org/10.54393/pjhs.v6i7.3362

***Corresponding Author:**
Vijia Kumar Gemnani
Faculty of Community Medicine and Public Health, Chandka Medical College, Shaheed Mohtarma Benazir Bhutto Medical University, Larkana, Pakistan
gemnanivijay@yahoo.com

Received Date: 25th May, 2025
Revised Date: 20th July, 2025
Acceptance Date: 26th July, 2025
Published Date: 31st July, 2025

ABSTRACT

Congenital heart disease (CHD) is a leading cause of infant morbidity and mortality. This is particularly the case in resource-limited settings where diagnosis is commonly delayed. **Objectives:** To assess the frequency and pattern of CHD among term neonates who presented with respiratory distress at a tertiary care hospital in Larkana, Pakistan. **Methods:** This cross-sectional study was conducted at the Department of Pediatrics, Chandka Medical College, Shaheed Mohtarma Benazir Bhutto Medical University, Larkana, Pakistan, from December 1, 2024, to June 7, 2025. A total of 105 term neonates presenting with respiratory distress were enrolled. Each neonate underwent detailed clinical examination and echocardiographic evaluation to identify structural heart defects. Demographic and perinatal data were also collected to explore associations with different CHD types. Statistical analysis was performed using SPSS version 22.0. **Results:** CHD was diagnosed in 41.9% of the neonates. The most common defect was a ventricular septal defect (24.8%), followed by an atrial septal defect (15.2%) and patent ductus arteriosus (12.4%). Cyanotic defects were less frequent, with tetralogy of Fallot identified in 6.7% and transposition of the great arteries in 2.9%. No statistically significant associations were found between CHD and neonatal age, gender, or mode of delivery. **Conclusions:** A significant proportion of term neonates with respiratory distress had underlying CHD, with acyanotic lesions being more common. These findings support the incorporation of routine echocardiographic screening and timely specialist referral for symptomatic neonates, particularly in resource-constrained healthcare settings.

INTRODUCTION

Congenital heart disease (CHD) is a group of anomalies that cause morphological and functional issues in the heart or main blood arteries from birth. Globally, 240,000 newborns die within 28 days after birth from congenital disease, and 170,000 die aged between one month and five years [1, 2]. CHD can present in various ways, with some patients remaining asymptomatic until discovered in late infancy or early childhood. Other extremes include cyanosis, heart failure, or both at neonatal age [3]. Neurodevelopmental delay is a common condition in children with CHD, which affects 46% of them [4]. Hence, the full physical examination on a newborn infant, along with heart auscultation and immediate echocardiography, is the key feature in the early diagnosis and management, thereby decreasing the morbidity and mortality from delayed diagnosis [5]. The crucial hemodynamic alteration in the

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process after birth is the transformation from fetal circulation, in which the oxygenation takes place within the placenta, to the postnatal circulation, where the oxygenation takes place in the lungs [6]. In fetal circulation, the oxygen-rich blood from the placenta returns to the right atrium (RA) via the umbilical vein and the ductus venosus; the oxygen-rich blood then passes through the foramen ovale and is transferred into the left ventricle (LV), which sends the oxygenated blood towards the brain and the heart, both with great metabolic demands [7, 8]. Blood with lower oxygen levels from the superior vena cava (SVC), the portal veins, and the coronary sinus, along with that from the lower body through the inferior vena cava (IVC), is primarily channeled to the right ventricle (RV). The RV then pumps this blood into the descending aorta via the main pulmonary artery and the ductus arteriosus, ultimately reaching the placenta for oxygenation [9, 10]. Under the system, the central shunts (ductus arteriosus, ductus venosus, and foramen ovale) are important in the normal circulation in the fetus. At birth, oxygen is transferred in the lung bed, receiving independent blood from the right ventricle (RV), while the left ventricle (LV) will deliver the oxygenated blood into the systemic circulation. In the cycle process, these shunts are redundant and will close within the first hours or days of life [11, 12]. Subhani *et al.* identified the prevalence of congenital heart disease as follows: VSD at 37%, ASD at 12.3%, patent ductus arteriosus at 11.1%, tetralogy of Fallot at 6.2%, and transposition of the great arteries at 2.5% [13]. A more recent study showed 13% of live-born infants had severe CHDs and were released undiagnosed from the hospital at birth [14]. Respiratory distress is one of the most common clinical presentations in neonates with congenital heart disease, especially in those with left-to-right shunts or obstructive lesions. Failure to recognize cardiac causes early can delay life-saving interventions [15]. Consequently, general pediatricians need to understand that the early identification and prompt interventions to stabilize these patients, along with their transfer to a suitable cardiac center, are vital for enhancing outcomes in critically ill individuals. In Pakistan, especially in underserved regions like Larkana, limited access to pediatric cardiology services and diagnostic tools often results in delayed or missed diagnoses of CHD. Studying term neonates, who are typically expected to be low-risk, can uncover the hidden burden of CHD in this group, aiding in early detection and timely referral. Unfortunately, there is a dearth of national data on the incidence or prevalence of congenital heart disease (CHD) that might indicate the disease burden of congenital heart defects in our nation, even though sparse hospital-based research at the regional level has been conducted in attempting to show the prevalence of CHD. Additionally, there isn't a national registration system in place at the moment to determine the overall number of instances. The age at which these patients are first diagnosed remains unknown. Most studies have taken the age at which cases of CHD present at the study center as the age of initial diagnosis. The data gathered from this study will give us insight into the extent of the issue in our country. Ultimately, this will assist us in formulating preventive strategies through regular screening.

Congenital heart disease (CHD) is a significant cause of neonatal morbidity and mortality, particularly in developing countries where early diagnosis is often delayed. Respiratory distress is a common presenting symptom in neonates; however, its association with underlying structural heart defects is frequently overlooked in routine clinical evaluation. In Pakistan, especially in peripheral regions such as Larkana, there is limited local data describing the frequency and pattern of CHD among term neonates presenting with respiratory distress. This lack of region-specific evidence highlights the need for studies that can support early detection and guide timely referral and management strategies. This study aimed to assess the frequency and pattern of CHD among term neonates who presented with respiratory distress at a tertiary care hospital in Larkana, Pakistan.

METHODS

This cross-sectional study was conducted in the Department of Pediatrics, Chandka Medical College, Shaheed Mohtarma Benazir Bhutto Medical University, Larkana, Pakistan, from 1 December 2024 to 7 May 2025. Ethical approval was obtained from the Institutional Ethical Review Committee of the College of Physicians and Surgeons, Pakistan (Ref. No. CPSP/REU/PED/-2022-221-6986), and written informed consent was obtained from parents or guardians after explaining the study purpose, risks, and benefits. The sample size of 105 neonates was calculated using WHO software, based on a previously reported frequency of patent ductus arteriosus of 11.1% from a study at Pak Emirates Military Hospital, Rawalpindi, with a 95% confidence level and a 6% margin of error. Patients were recruited through non-probability consecutive sampling. Term neonates aged 0–14 days presenting with respiratory distress were eligible if they exhibited at least one of the following: respiratory rate ≥ 60 breaths/min, oxygen saturation ≤ 88%, or arterial blood gas showing pCO_2 ≥ 60 mmHg. Additional indicators included murmur, cyanosis with or without feeding difficulty, clinical features of congestive heart failure, or a pre- and post-ductal SpO_2 difference greater than 3%. Neonates were excluded if parental consent was not provided, or if they had congenital anomalies such as meningomyelocele or spina bifida, or clinical diagnoses of sepsis or necrotizing enterocolitis. A detailed history and clinical examination

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were performed by the principal investigator, and echocardiography was carried out by a pediatric cardiologist with more than 10 years of experience. Congenital heart diseases were diagnosed according to established echocardiographic criteria, including ventricular septal defect, atrial septal defect, patent ductus arteriosus, atrioventricular septal defect, tetralogy of Fallot, and transposition of the great arteries. All findings were recorded on a pre-designed data collection form. Data were analyzed using SPSS version 22.0, with descriptive statistics (frequencies, percentages) applied to categorical variables and associations between clinical variables and congenital heart disease assessed using the Chi-square test, considering a p-value<0.050 as statistically significant.

RESULTS

105 term neonates were enrolled who presented with respiratory distress at our tertiary care facility. Most infants were between 8 and 14 days old (55.2%), and the gender distribution was nearly equal, with 51.4% males and 48.6% females. The majority were delivered via normal vaginal delivery (79%), and 71.4% had a gestational age of more than 37 weeks. Maternal characteristics showed that 59% of mothers had a parity greater than three, and 65.7% had been pregnant more than three times. Roughly half of the families reported a monthly income of ≤75,000 PKR, and most mothers had either primary (50.5%) or secondary (25.7%) education, while a small proportion (3.8%) were illiterate, Table 1.

Table 1: Distribution of Baseline Characteristics among the Study Participants

Variables		N (%)
Neonatal Age	01 to 07 days	47 (44.8)
	08 to 14 days	58 (55.2)
Gender	Male	54 (51.4)
	Female	51 (48.6)
Mode of Delivery	Cesarian	22 (21)
	Normal vaginal delivery	83 (79)
Gestational Age	≤37 weeks	30 (28.6)
	>37 weeks	75 (71.4)
Parity	≤3	43 (41)
	>3	62 (59)
Gravida	≤3	36 (34.3)
	>3	69 (65.7)
Family Monthly Income	≤75000 per month	54 (51.4)
	>75000 per month	51 (486)
Educational Status	Illiterate	04 (3.8)
	Primary	53 (50.5)
	Secondary	27 (25.7)
	Higher	21 (20)

The study found that 24.8% of neonates were diagnosed with a ventricular septal defect, making it the most common congenital heart defect. Other common congenital heart defects include atrial septal defect (15.2%), patent ductus arteriosus (12.4%), tetralogy of Fallot (6.7%), and transposition of great arteries (2.9%). CHD was diagnosed in 44 neonates (41.9%). Among these, acyanotic defects were more common (Table 2)

Table 2: Frequencies of CHD Types

CHD Type	Acyanotic/Cyanotic	N (%)
Ventricular Septal Defect (VSD)	Acyanotic	26 (24.8)
Atrial Septal Defect (ASD)	Acyanotic	16 (15.2)
Patent Ductus Arteriosus (PDA)	Acyanotic	13 (12.4)
Tetralogy of Fallot (TOF)	Cyanotic	7 (6.7)
Transposition of the Great Arteries (TGA)	Cyanotic	3 (2.9)

None of the congenital heart defects (Ventricular Septal Defect (VSD)(p=0.230), Atrial Septal Defect (ASD)(p=0.310), Patent Ductus Arteriosus (PDA) (p=0.480), Tetralogy of Fallot (TOF) (p=0.140), Transposition of the Great Arteries (TGA) (p=0.250) showed a statistically significant difference in prevalence between the two neonatal age groups. The findings suggest that the distribution of these CHDs does not significantly vary between neonates aged 01–07 days and those aged 08–14 days (Table 3).

Table 3: Distribution of Congenital Heart Defects by Age Group in Neonates

Variables		Age		p-Value
		01 to 07 Days	08 to 14 Days	
Ventral Septal Defect	Yes	09 (19.1)	17 (29.3)	0.230
	No	38 (80.9)	41 (70.7)	
Atrial Septal Defect	Yes	09 (19.1)	07 (12.1)	0.310
	No	38 (80.9)	51 (87.9)	
Patent Ductus Arteriosus	Yes	07 (14.9)	06 (10.3)	0.480
	No	40 (85.1)	52 (89.7)	
Tetralogy of Fallot	Yes	05 (10.6)	02 (3.4)	0.140
	No	42 (89.4)	56 (96.6)	
Transposition of the Great Arteries	Yes	00 (00)	03 (5.2)	0.250
	No	47 (100)	55 (94.8)	

Across all types of congenital heart defects (Ventricular Septal Defect (p-value: 0.86), Atrial Septal Defect (p-value: 0.900), Patent Ductus Arteriosus (p-value=0.680), Tetralogy of Fallot (p-value: 0.630), Transposition of the Great Arteries (p-value=0.610), showing, no statistically significant differences were found between male and female neonates (all p-values>0.050). While the distribution patterns varied slightly across subgroups, the findings did not suggest any strong demographic predictors of specific heart defects within this cohort (Table 4).

DOI: https://doi.org/10.54393/pjhs.v6i7.3362

Table 4: Distribution of Congenital Heart Defects by Gender in Neonates

Variables		Gender		p-Value
		Male	Female	
Ventral Septal Defect	Yes	13 (24.1)	13 (25.5)	0.860
	No	41 (75.9)	38 (74.5)	
Atrial Septal Defect	Yes	08 (14.8)	08 (15.7)	0.900
	No	46 (85.2)	43 (84.3)	
Patent Ductus Arteriosus	Yes	06 (11.1)	07 (13.7)	0.680
	No	48 (88.9)	44 (86.3)	
Tetralogy of Fallot	Yes	03 (5.6)	04 (7.8)	0.630
	No	51 (94.4)	47 (92.2)	
Transposition of the Great Arteries	Yes	01 (1.9)	02 (3.9)	0.610
	No	53 (98.1)	49 (96.1)	

DISCUSSION

In the present study, neonates were admitted with respiratory distress at a referral center in Larkana. Ventricular septal defect (VSD) was the most common lesion, followed in decreasing frequency by atrial septal defect (ASD), patent ductus arteriosus (PDA), tetralogy of Fallot (TOF), and transposition of the great arteries (TGA). These findings are consistent with regional and international data. Sehar *et al.* reported VSD as the most frequently occurring acyanotic CHD, accounting for 27.5% of congenital defects at their center in Rawalpindi [16]. In our study, demographic characteristics such as age, gender, and mode of delivery did not show statistically significant associations with the type of CHD. Comparable findings were reported by previous studies, which observed that respiratory distress was the leading presenting feature, and demographic factors were not predictive of CHD type [17, 18]. In our cohort, respiratory distress was indeed the most frequent symptom, reinforcing that common neonatal presentations may mask underlying structural heart disease. This highlights the importance of routine echocardiographic evaluation in neonates with unexplained respiratory distress [16, 19]. Late diagnosis remains a persistent problem, particularly in cyanotic CHDs such as TOF and TGA. Previous studies highlighted the risks of delayed recognition, which contribute significantly to preventable morbidity and mortality due to missed opportunities for timely intervention [18, 20]. Although maternal and perinatal factors were not evaluated in detail in our study, other research has established their significance. Islam *et al.* reported maternal diabetes, hypertension, and antipyretic drug exposure as risk factors for CHD [17], while Dey *et al.* identified perinatal complications as predictors of poor outcomes in neonates with CHD [19]. Aryal *et al.* further emphasized the association of congenital birth defects with broader maternal health indicators in South Asia [21]. Collectively, these findings underscore gaps in early identification and referral pathways for CHD in neonatal populations. The spectrum of CHD observed in our study aligns with findings from Bangladesh and other South Asian cohorts, where acyanotic lesions such as VSD and ASD predominate [17, 19]. The higher burden of left-to-right shunt lesions suggests an urgent need for early detection to prevent long-term complications such as pulmonary hypertension and Eisenmenger syndrome [16, 20]. In Pakistan, pediatric cardiology services remain concentrated in major cities, limiting access for neonates in peripheral regions. Both Qazi *et al.* and Javed *et al.* emphasized the necessity of expanding pediatric cardiac services to secondary and district-level hospitals to facilitate earlier diagnosis and reduce the burden of delayed treatment [18, 20]. Taken together, our results highlight that while demographic factors may not predict CHD type, respiratory distress remains a key presenting feature that warrants prompt echocardiographic evaluation. Universal screening strategies, rather than selective risk-based approaches, may be more effective in ensuring early detection and timely management of CHD in resource-limited settings.

This study has certain limitations, including its single-center design and relatively small sample size, which may limit the generalizability of the findings. Additionally, the cross-sectional nature of the study does not allow assessment of long-term clinical outcomes or progression of congenital heart disease. Detailed evaluation of maternal risk factors and perinatal complications was also beyond the scope of this research. Future multicenter studies with larger sample sizes, comprehensive maternal and neonatal risk assessment, and longitudinal follow-up are recommended to better understand the burden and outcomes of CHD in neonates.

CONCLUSIONS

The study reveals a high prevalence of congenital heart disease in full-term neonates with respiratory distress, with ventricular septal defect being the most common diagnosis. It emphasizes the need for routine echocardiographic screening and improved referral systems to reduce morbidity and mortality in resource-limited healthcare settings.

Authors' Contribution

Conceptualization: ZA, ZAM
Methodology: ZAM, SAJ
Formal analysis: SAS, SAJ
Writing and Drafting: ZA, VKG, DB
Review and Editing: ZA, VKG, DB, SAS, SAJ, ZAM

All authors approved the final manuscript and take responsibility for the integrity of the work

DOI: https://doi.org/10.54393/pjhs.v6i7.3362

Conflicts of Interest

All the authors declare no conflict of interest.

Source of Funding

The author received no financial support for the research, authorship and/or publication of this article.

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