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Clinical and Echocardiographic Profile of Pediatric Patients with Ventricular Septal Defect

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ABSTRACT

Ventricular septal defect (VSD) is the most common congenital heart disease in children, contributing significantly to pediatric morbidity and mortality globally. In resource-limited settings like Pakistan, delayed diagnosis and limited access to pediatric cardiology services can exacerbate disease outcomes. **Objectives:** To evaluate the clinical presentation and echocardiographic profile of pediatric patients diagnosed with VSD at a tertiary care center in Pakistan. **Methods:** This retrospective, cross-sectional descriptive study was conducted at the Department of Pediatric Cardiology of the Peshawar Institute of Cardiology from July 1, 2025, to September 31, 2025. A total of 200 children under 18 years of age with echocardiographically confirmed VSD were included. Data on demographics, clinical symptoms, and physical examination findings were collected. Echocardiographic assessment was used to determine the size, type, and location of VSD, along with associated cardiac anomalies. **Results:** The median age at diagnosis was 9.4 months, with 78% diagnosed within the first year of life. The most common presenting feature was cardiac murmur (60%), followed by recurrent respiratory infections (49%), feeding problems (42.5%), and failure to thrive (38%). Perimembranous VSD was the most prevalent type (62%), followed by muscular (19%), inlet (10%), and outlet (9%) defects. Associated cardiac lesions included patent ductus arteriosus (18%), atrial septal defect (14%), aortic valve prolapse (11%), and pulmonary hypertension (24%). **Conclusions:** Early recognition and echocardiographic screening, especially in high-risk groups like children with Down syndrome, are essential to improving outcomes. These findings support the need for enhanced screening and resource allocation in pediatric cardiology services in Pakistan.

INTRODUCTION

Ventricular septal defect (VSD) is the most prevalent of the congenital heart diseases (CHDs), which continue to be a major cause of morbidity and mortality in infants and children around the world. The hallmark of VSD is aberrant communication between the left and right ventricles, which permits oxygen-poor blood in the right ventricle to mingle with oxygen-rich blood from the left. Increased pulmonary blood flow from this left-to-right shunting may cause congestive heart failure, pulmonary hypertension, and failure to thrive if treatment is delayed. With an estimated prevalence of 2.5 to 3.5 per 1,000 live births, VSD makes up approximately 20–30% of all congenital heart abnormalities worldwide [1, 2]. In order to avoid long-term consequences, moderate to large-sized defects frequently require medical or surgical intervention, even though many minor VSDs close on their own [3, 4]. Due to delayed diagnosis, restricted access to specialized pediatric cardiology care, and a lack of national screening methods, congenital heart disease presents a significant health burden in Pakistan. According to a recent multicenter study conducted in Pakistan, the majority of confirmed instances of congenital heart defects (CHDs) are VSD, with an incidence of 9–12 cases per 1,000 live births [5]. The need for early detection and therapy of congenital heart

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diseases, especially VSDs, has become more pressing due to the rapidly expanding pediatric population in Pakistan, where over 35% of the population is under the age of 15. The danger of consequences such as pulmonary vascular disease or irreversible pulmonary hypertension is raised since many cases in rural or resource-constrained areas go undiagnosed until symptoms worsen [6, 7]. The primary method for diagnosing and categorizing VSDs is still echocardiography. It evaluates shunt direction, chamber dilatation, and related anomalies in addition to assisting in determining the defect's existence, size, and location. Perimembranous, muscular, inlet, and outlet VSDs are among the morphological subtypes that have different risks of spontaneous closure, comorbidities such as aortic valve prolapse, and surgical techniques [8, 9]. Clinical decision-making requires prompt and precise echocardiographic examination, particularly in healthcare systems with limited resources, when postponed therapies may increase morbidity. Comprehensive national-level data examining the clinical and echocardiographic characteristics of children with VSD is lacking in Pakistan. The majority of published research is retrospective, small-scale, and frequently restricted to urban tertiary care facilities, which may not accurately reflect the pediatric community as a whole. Furthermore, many families put off seeking care until symptoms are more severe because of social stigma, ignorance, and financial limitations. This fact emphasizes the necessity of doing epidemiological research relevant to a certain region in order to inform the development of policies, the distribution of resources, and community-based screening initiatives [10]. By examining the clinical presentation, echocardiographic characteristics, and related cardiac abnormalities of a cohort of pediatric patients with VSD in a tertiary care facility in Pakistan, the current study seeks to close this knowledge gap. The study aims to enhance early detection and direct focused interventions by offering a thorough description of these cases. Clinicians can better predict difficulties, plan surgical procedures, and provide families with appropriate counselling by having a thorough understanding of the typical presenting signs, distribution of VSD types, and prevalence of concomitant lesions, including PDA and pulmonary hypertension.

This study aimed to evaluate the clinical presentation and echocardiographic profile of pediatric patients diagnosed with VSD at a tertiary care center in Pakistan.

METHODS

This was a retrospective, cross-sectional descriptive study conducted at the Department of Pediatric Cardiology of the Peshawar Institute of Cardiology, a specialized tertiary care facility, from July 1, 2025, to September 31, 2025, with data were collected from July 2023 to May 2024. The study comprised children under 18 years of age with echocardiographically validated VSD. The ethical approval was taken from the Institutional Review Board with ref no. IRC/25/203. Patients with a history of surgical or device closure of VSD or with intricate congenital heart conditions (e.g., tetralogy of Fallot, atrioventricular septal defect, transposition of the great arteries) were excluded to facilitate a concentrated analysis of isolated or predominantly VSD lesions. Based on the expected prevalence of pulmonary hypertension in children with VSD (approximately 24%, as reported by Hopper et al. [11]), a confidence level of 95%, and a margin of error of 6%, the minimum required sample size was calculated to be 200 using standard sample size formulas for proportions in descriptive studies. The calculation was performed using the formula: $n = Z^2 \times p \times (1 - p) / d^2$. where Z = 1.96 (for 95% confidence), p = 0.24, and d = 0.06. Informed consent was taken from each participant. This ensured adequate power to estimate the prevalence of key echocardiographic findings, including pulmonary hypertension, among children with VSD. A standardized proforma was used to collect data on patients' demographics (age, gender, and anthropometric measurements), clinical history (e.g., syndromic features, recurrent respiratory infections, hospitalizations), and specific physical examination findings. The study got the information from the hospital's electronic medical records and patient charts. Transthoracic echocardiography was conducted utilizing pediatric transducers. Defects were categorized based on dimensions (small: <3 mm, moderate: 3–6 mm, large: >6 mm) and anatomical location (perimembranous, muscular, inlet, outlet). Atrial septal defect (ASD), patent ductus arteriosus (PDA), aortic valve prolapses, and pulmonary hypertension (PH) were among the associated findings recorded. The study used SPSS version 25.0 to do the statistical analysis. Qualitative variables (e.g., gender, defect type, presence of pulmonary hypertension, and associated cardiac lesions) were summarized using frequencies and percentages. Quantitative variables (e.g., age and weight) were reported as mean ± standard deviation (SD) for normally distributed data, or as median and interquartile range (IQR) for non-normally distributed data. The Chi-square test or Fisher's exact test was utilized for categorical data when comparisons were pertinent. We used independent samples t-tests for continuous variables that were normally distributed, and Mann–Whitney U tests for continuous variables that were not normally distributed. A p-value of less than 0.05 was deemed statistically significant.

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RESULTS

A total of 200 children diagnosed with ventricular septal defect (VSD) were included in the study. The mean age was 11.6 ± 9.4 months (range 1 month to 17 years), with a male-to-female ratio of 1.27:1. Most patients (78%) were diagnosed during the first year of life. Approximately 38% of the children were underweight or exhibited growth parameters below the 5th percentile for their age. Clinical history revealed that 49% of patients experienced recurrent respiratory tract infections, 42.5% had feeding difficulties, and 38% presented with features of failure to thrive. Syndromic characteristics, primarily Down syndrome, were observed in 13% of cases. Hospitalizations due to cardiac or respiratory issues occurred in 37% of the cohort (Table 1).

Table 1: Clinical Presentation of Patients

Variables	Frequency (%)
Gender	
Male	112 (56%)
Female	88 (44%)
Age Distribution	
≤1Year	156 (78%)
>1Year	44 (22%)
Anthropometric Status	
Normal Growth	124 (62%)
Failure to Thrive / Underweight	76 (38%)
Clinical Presentation	
Cardiac Murmur	120 (60%)
Recurrent Respiratory Infections	98 (49%)
Feeding Difficulties	85 (42.5%)
Cyanosis	12 (6%)
Syndromic Features	
Down syndrome	26 (13%)
Hospitalizations	
Due to Cardiac or Respiratory Issues	74 (37%)

Comparative analysis showed that pulmonary hypertension was significantly more frequent in patients with large VSDs compared to those with small or moderate defects ($p<0.001$). It was also more common among patients with associated PDA ($p=0.02$). No statistically significant difference was found between male and female ($p=0.48$) or between syndromic and non-syndromic children ($p=0.31$). Perimembranous VSDs were the most common subtype (62%), followed by muscular (19%), inlet (10%), and outlet (9%) types, aligning with global patterns of VSD morphology in children. Among children with Down syndrome (n=26), inlet-type VSDs were common (53.8%), reflecting the known link between trisomy 21 and AVSD, and highlighting the need for targeted screening. Small VSDs were the most commonly observed, comprising 44% of the cases (n=88), followed by moderate-sized defects in 34% (n=68), and large VSDs in 22% (n=44) (Table 2).

Table 2: Echocardiographic Findings of Patients (VSD Type and Size) (n=200)

Variables	Frequency (%)
VSD Type	
Perimembranous	124 (62%)
Muscular	38 (19%)
Inlet	20 (10%)
Outlet	18 (9%)
VSD Size	
Small	88 (44%)
Moderate	68 (34%)
Large	44 (22%)

No significant association was observed between the type of VSD and the presence of pulmonary hypertension ($p=0.27$).

Most patients (93%, n=186) had a left-to-right shunt, while 5% (n=10) had bidirectional flow and 2% (n=4) showed right-to-left shunting (Table 3).

Table 3: Echocardiographic Findings of Patients (Shunt Direction) (n=200)

Shunt Direction	Frequency (%)
Left-to-right	186 (93%)
Bidirectional	10 (5%)
Right-to-left	4 (2%)

PDA was present in 18% of cases, ASD in 14%, and aortic valve prolapse in 11%, mainly linked to perimembranous VSDs. Pulmonary hypertension was found in 24% of patients, highlighting the need for early detection to prevent long-term complications (Table 4).

Table 4: Echocardiographic Findings of Patients (Associated Lesions) (n=200)

Associated lesions	Frequency (%)
PDA	36 (18%)
ASD	28 (14%)
Aortic Valve Prolapse	22 (11%)
Pulmonary Hypertension	48 (24%)

Bivariate analysis of associated cardiac lesions revealed that pulmonary hypertension (PH) was present in 48 (24%) of the 200 children. PH was significantly more frequent among patients with patent ductus arteriosus (PDA), with 18 of 36 children with PDA (50%) exhibiting PH ($p = 0.02$). Atrial septal defect (ASD) and aortic valve prolapse were present in 28 and 22 children, respectively, with PH observed in 12 (6%) and 10 (5%) cases; however, these associations were not statistically significant ($p > 0.05$). Among children without additional lesions, 8 (4%) had PH. The total number of children with PH aligns with the overall prevalence of 24%, reinforcing the importance of early detection and management of high-risk lesions such as PDA. The distribution of PH in relation to cardiac lesions is summarized (Table 5).

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Table 5: Association of Pulmonary Hypertension (PH) with Common Cardiac Lesions in Children with VSD (n=200)

Cardiac Lesion	PH Present, n (%)	PH Absent, n (%)	Total, n (%)	Statistical Test	p-value
Patent Ductus Arteriosus	18 (9%)	18 (9%)	36 (18%)	Chi-square	0.02
Atrial Septal Defect	12 (6%)	16 (8%)	28 (14%)	Chi-square	0.18
Aortic Valve Prolapse	10 (5%)	12 (6%)	22 (11%)	Chi-square	0.11
No Associated Lesion	8 (4%)	106 (53%)	114 (57%)	–	–
Total	48 (24%)	152 (76%)	200 (100%)	–	–

DISCUSSION

The most common congenital cardiac abnormality in children, ventricular septal defect (VSD), is evaluated clinically and echocardiographically in 200 pediatric patients. According to the demographic data, there is a small male predominance (56%), which is in line with findings from another study [1], which found that males had a slightly greater incidence of VSD. In line with findings of a study [11], where early detection within the first year was critical due to the severity of symptoms in hemodynamically significant abnormalities, the median age of diagnosis was 9.4 months, with 78% of cases being discovered in infancy. In 60% of patients, cardiac murmur was the most common initial presentation, highlighting its crucial function as the first clinical hint in the screening for congenital heart disease. Similar results were noted in a study, which found that 64% of patients with VSD had an audible murmur at presentation, which prompted an echocardiogram [12]. In VSDs, murmurs are usually pansystolic, produced by high-velocity turbulent flow through the septal defect, and are best audible near the left lower sternal border [13]. In 49% of our cohort, recurrent lower respiratory tract infections were documented. Pulmonary overcirculation due to a left-to-right shunt is a known consequence. There may be a connection between VSDs and pediatric respiratory morbidity, as seen in studies [14, 15], which both showed a high rate of respiratory infections. Poor weight gain and frequent hospital stays are also caused by the load of these illnesses. 42.5% of our patients have feeding issues, which is a common yet sometimes underappreciated symptom. Tachypnea and perspiration during feedings cause infants with VSD to consume more energy, which is reflected in these problems. This aligned with the previous research [16], which highlighted feeding issues as a precursor to congestive heart failure in newborns with large shunts. Out of the youngsters in this study, 38% were found to have failed to flourish. This finding is consistent with research [17], which discovered that growth retardation is common in children with large or moderate VSDs because of increased metabolic demands, recurrent infections, and chronic undernutrition. After a defect is closed or the shunt flow naturally decreases, these growth issues frequently go away. Cyanosis was detected in only 6% of instances, mostly in individuals with bidirectional or right-to-left shunts or severe VSDs worsened by pulmonary hypertension. Although cyanosis is uncommon in isolated VSD, it can indicate severe illness, Eisenmenger physiology, or other cardiac abnormalities. This is consistent with research [18], which found that patients with higher pulmonary vascular resistance were more likely to have cyanosis. Among our cohort, 13% had Down syndrome. Crucially, inlet-type VSDs were seen in over half (53.8%) of children with Down syndrome, supporting the established link between trisomy 21 and atrioventricular septal defects (AVSDs). Nearly 40–50% of children with Down syndrome have congenital heart disease, with AVSD being the most common kind that frequently affects the inlet septum [11, 19]. 62% of patients in our study had the peri-membranous type, which was the most common anatomical subtype. Muscular (19%), inlet (10%), and outlet (9%) types were next in line. This distribution reflects developments in epidemiology worldwide. Over 70% of all VSDs are caused by peri-membranous abnormalities [20]. Both the membrane septum's embryological vulnerability and its frequent inclusion in diagnostic echocardiographic planes are reflected in the high frequency of peri-membranous VSDs. The second most frequent type of VSD was muscular (19%). Despite being smaller and more prone to closure on their own, these abnormalities are occasionally underdiagnosed because of their faint Doppler signals, especially in newborns. According to research, up to 80% of muscle VSDs spontaneously close during the first two years of life [21]. On the other hand, our cohort's children with Down syndrome had a disproportionately high number of inlet-type VSDs, indicating the necessity for closer echocardiographic monitoring in these groups. Small VSDs accounted for the biggest percentage of defects (44%), followed by moderate (34%) and large (22%). This is consistent with research [22], which found that the greater usage of neonatal echocardiographic screening led to a higher frequency of tiny VSDs. The hemodynamic cost imposed by a defect's size has clinical implications; larger flaws are more likely to cause heart failure, pulmonary hypertension, and volume overload. As would be predicted in the absence of pulmonary vascular disease or elevated right-sided pressures, left-to-right shunting was the predominant flow pattern (93%). Only a small percentage of individuals showed right-to-left (2%) or bidirectional (5%) shunting. These examples probably indicate more severe pulmonary vascular alterations that could lead to Eisenmenger syndrome. Shunt reversal is a serious prognostic indicator that frequently necessitates early surgical or palliative

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intervention [23]. With 18% of patients having it, patent ductus arteriosus (PDA) was the most common related lesion. PDA and VSD together have the potential to greatly increase pulmonary over-circulation and worsen symptoms. Similar prevalence was reported in a study [24], which also underlined the importance of co-management of both disorders in order to avoid pulmonary hypertension. A trend towards multiple septal defects in certain children is suggested by the 14% of patients who had an atrial septal defect (ASD). Such combinations may result from common embryologic abnormalities in the atrioventricular septation and call for a customized interventional strategy [25]. Eleven percent of patients, primarily those with peri-membranous VSDs, had aortic valve prolapse. The fact that this issue is linked to aortic regurgitation makes it noteworthy. Up to 10–15% of children with peri-membranous VSDs experience aortic valve prolapse over time [26], highlighting the significance of serial echocardiographic surveillance. Twenty-four percent of patients had pulmonary hypertension, which is indicative of a significant burden of chronic volume overload and delayed diagnosis. Long-term results are greatly impacted by pulmonary hypertension since it can result in Eisenmenger physiology and inoperability. This prevalence is consistent with a study [27], which emphasized early intervention to mitigate pulmonary vascular remodeling in children with congenital left-to-right shunts.

CONCLUSIONS

This study provides an extensive analysis of the clinical and echocardiographic characteristics of pediatric patients with ventricular septal defect (VSD) at a tertiary care facility in Pakistan. Most patients were diagnosed within the first year of life, and the most common signs were a heart murmur, repeated respiratory infections, trouble eating, and not growing. Peri-membranous VSD was the most common subtype, followed by muscular, inlet, and outlet types. A significant number of patients had heart problems that were linked to their condition, such as patent ductus arteriosus, atrial septal defect, aortic valve prolapse, and pulmonary hypertension. Notably, pulmonary hypertension was present in nearly one-fourth of the cohort, underscoring the consequences of delayed diagnosis.

Authors Contribution

Conceptualization: RM
Methodology: AA, HDW
Formal analysis: AZ
Writing review and editing: HDW

All authors have read and agreed to the published version of the manuscript

Conflicts of Interest

All the authors declare no conflict of interest.

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