

SPECTRUM OF VENTRICULAR SEPTAL DEFECTS IN PATIENTS WITH CONGENITAL HEART DISEASE

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Contribution

All the authors contributed significantly to the research that resulted in the submitted manuscript.

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ABSTRACT

Objective: To study the frequency and spectrum (echocardiographic picture) of congenital ventricular septal defect (VSD) among patients attending Paediatric Cardiology Department for suspected congenital heart disease (CHD).

Methodology: This descriptive study was conducted in Pediatric Cardiology Department Lady Reading Hospital Peshawar, from 01 September 2012 to 31 August 2013. All patient with suspected CHD was subjected to transthoracic echocardiographic examination. The frequency and spectrum of VSD was determine in these patients.

Results: A total of 2342 patients with suspected CHD were studied. Among them 708(30.2%) were having isolated congenital VSD. Males were 433(61%) and females were 275(39%). Mean age was 42.32 ± 40.64 months (range: 1 day to 30 years). Patients below 2 years were 54%. Out of this 708 patients, 447(63.1%) were of perimembranous type, 169(23.86%) were having muscular VSD, 48(6.78%) were Doubly committed subarterial type, and 44(6.21%) were having multiple VSDs. Two hundred and thirty-three (32.9%) of total VSD patients were have already developed complications including severe pulmonary artery hypertension (PAH) in 157(22.17%) patients, aortic cusp prolapse in 50(7.06%) cases including 30(6.86%) with perimembranous and 20(41.6%) with doubly committed subarterial type of VSD, AR in 40(5.64%) of Aortic cusp prolapse, Acquired right and left ventricular outflow tract obstruction in 10(1.4%) and 05(0.70%) respectively, 04(0.56%) patients had Echo evidence of infective endocarditis and 2 had already developed Eisenmenger syndrome with severe PAH.

Conclusion: VSD account for about 30% of the congenital cardiac defects. About one third of them had already developed complications at the time of diagnosis.

Key Words: Ventricular Septal Defect (VSD), Perimembranous VSD, Muscular VSD, Atrioventricular Canal Defect, Doubly Committed Subarterial VSD, Aortic Cusp Prolapsed (AVP)

INTRODUCTION

Congenital heart disease (CHD) is a significant cause of morbidity and mortality amongst infants and children.¹ Knowledge of the local spectrum of CHD provides the foundation for the rational allocation of healthcare resources. It has been reported that CHD spectra differ according to geographical location.^{2,3} The current study aims to determine the spectrum i.e the echocardiographic picture of VSD encountered at a referral hospital in Khyber Puktoonkhwa; with a view to providing data that may contribute to the rational allocation of resources to Paediatric Cardiac Care locally.

Ventricular septal defect (VSD) is a developmental defect of the interventricular septum.⁴ Isolated ventricular septal defect occurs in approximately 2-6 of every 1000 live births and accounts for more than 15-20% of all congenital heart diseases.⁵ VSD are classified by Soto et al, into perimembranous, Muscular and doubly committed subarterial (DCSA) types.⁶ Perimembranous ventricular septal defects are the most common type of VSD associated with aneurysms of the septal leaflet of the tricuspid valve, which can partially or completely close the defect.^{7,8} Muscular VSD tend to close spontaneously earlier than perimembranous VSD.⁹ The natural history has a wide spectrum, ranging from spontaneous closure to congestive cardiac failure to death.¹⁰ Atrioventricular septal defects (AVSDs) are anatomic defects that arise from faulty development of the embryonic endocardial cushions.^{11,12} Spontaneous closure frequently occurs in children, usually by age of 2 years.⁷ The natural history of VSD is characterized by many complications. Of special interest is prolapse of the aortic valve cusp, which classically occurs with doubly committed subarterial and less commonly with perimembranous outlet type.¹³ Secondary aortic insufficiency, is associated with prolapse of aortic valve cusps. Aortic regurgitation is due to a poorly supported right coronary cusp combined with the Venturi effect produced by the ventricular septal defect jet, resulting in cusp prolapse.¹⁴ Perimembranous outlet VSD are also associated with infundibular hypertrophy, and right ventricular outflow tract obstruction can progress in severity, which most often needs surgical intervention.¹⁵ Discrete fibrous subaortic stenosis is occasionally seen in association with ventricular septal defect more frequently with perimembranous VSD than other forms.¹⁶ Infective endocarditis is a rare complication of VSD and due to the possibility of this grieves complication, some cardiologists are of the opinion that all VSD whether haemodynamically significant or not should be closed.¹⁷ Pattern of VSD and associated complications are already known in the literature but very little is known in over local setup, so we conducted this study to know the frequency and pattern of VSD in our population.

METHODOLOGY

This descriptive cross-sectional study was conducted in Department of pediatric Cardiology, Lady Reading Hospital, Peshawar, from 01 September 2012 to 31 August 2013. A total of 2342 patients were included by consecutive sampling, who presented to echocardiographic department for cardiac evaluation for suspected congenital heart disease. The diagnosis was primarily made on echocardiography. Size, number and exact location of the defect as well as magnitude of shunt were identified by 2-D and Doppler echocardiography. Pulmonary artery pressure was estimated by PA pressure method. Aortic valve prolapse and aortic regurgitation was also noted.

Severity of aortic regurgitation was assessed by using parameters like left ventricular end diastolic and systolic dimensions, Doppler flow velocity measurement and assessment of length, width and area of regurgitant jet. All echocardiograms were performed by trained paediatric cardiologists using latest Xario Toshiba diagnostic ultrasound system model SSA-660A echocardiographic machine. Patients with VSD as a part of other congenital cardiac anomalies were excluded from the study. All echocardiography reports were reviewed from hospital record. VSD were classified as perimembranous, doubly committed subarterial, muscular and multiple VSD. Functionally VSDs were categorized into small, moderate and large VSDs. Small VSD was defined as a doppler CW gradient across VSD > 60 mmHg, with no LV dilation and absence of severe pulmonary hypertension. Moderate VSD was defined as Doppler CW gradient across VSD 30-60 mmHg and LV dilation with out severe pulmonary hypertension. Large VSD was a Doppler CW gradient across VSD <30mmHg while LV dilation may or may not be present, with presence of severe pulmonary hypertension or Eisenmenger syndrome.

Data was entered and processed using SPSS version 20. Data including patient name, gender, age, types of VSD, functional status of VSD, presence of other associated cardiac lesions, pulmonary arterial hypertension, Right coronary cusp prolapse with associated aortic regurgitation, left or right ventricular outflow obstruction, surgically or device closure of VSD, infective endocarditis and development of Eisenmenger syndrome were entered by a specially designed proforma.

RESULTS

A total of 2342 patients with suspected congenital heart disease were included in the study. Among them 708(30.2%) were having isolated congenital VSD. Males were 433(61%) and females were 275(39%). Mean age was 42.32±40.64 months (range: 1 day to 30 years). Patients below 2 years were 54%. Of 708 patients, 447(63.7%) were

Table 1: Types and Size of VSD

Types	Small VSD	Moderate VSD	Large VSD	Total Cases
Perimembranous	100(14.1%)	77(10.88%)	270(38.13%)	447(63.1%)
Muscular	39(5.5%)	40(5.65%)	90(12.71%)	169(23.86%)
Doubly committed subarterial				48(6.78%)
Multiple				44(6.21%)
Total				708

of perimembranous type of which 100(14.1%) were small, 77(10.88%) moderate while 270(38.13%) were of large perimembranous type. 169(23.86%) were having muscular VSD including 39(5.5%) small muscular VSD, 40(5.65%) moderate muscular type and 90(12.71%) large muscular type, 48(6.78%) were Doubly committed subarterial type, and 44 (6.21%) were having multiple VSD as shown in Table 1. Complications were noted in 233(32.9%) of total cases. Severe pulmonary hypertension was the most common complication associated with large VSD and it was noted in 157(22.17%) patients while 2 had already developed Eisenmenger syndrome. The most common complication seen with small and moderate VSD was aortic cusp prolapse and aortic regurgitation followed by right and left ventricular outflow tract obstruction as described in Table 2. of them Mild degree of aortic regurgitation was seen in 23(3.25%), moderate AR in 13(1.8%) while severe AR was present in only 4(0.56%) patients. This complication was observed more frequently with doubly committed type of VSD.

DISCUSSION

Pediatric Cardiology, Govt Lady Reading Hospital Peshawar, a tertiary referral center for paediatric cardiac patients in Khyber Pakhtoonkhwa catering to a population of almost 20 million. Catchment area extends top arts of Punjab and Gilgit/Baltistan. Isolated Ventricular Septal Defect was found in 32.4% of patients with congenital heart diseases analyzed during the study period. Similarly study done by Ahmad et al, in our province a few years back showed ventricular septal defect in 29% of the 144 patients with congenital heart disease.¹⁸ Kazmi et al, reported VSD incidence only in about 11% of congenital heart disease patients.¹⁹ The commonest type was perimembranous VSD in our study i.e 61.6%. The second in order of frequency were muscular VSD (22.45%) and Doubly committed subarterial type accounted for 6.78% of the total. Similar pattern of VSD is also found in Western literature, where the largest group of VSD consists of perimembranous type, muscular and doubly committed subarterial type in decreasing order of frequency.²⁰ Aziz et al, documented the frequency of perimembranous VSD as 92%

of total VSD, doubly committed subarterial type were 7% and the least common were muscular i.e. 1.7% as opposed to our study which shows 61% of perimembranous, 22% of muscular and about 6.8% of doubly committed subarterial type.²¹ However, in this study, the largest group of patients were older than one year (68% of patients), and muscular VSD was found mostly in younger patients as found in our study which included 54% of patients below 2 years of age, as most of the small muscular VSD closes in 1st year of life which may be responsible for the very less frequency of muscular VSD in their study.²⁰ Similar results were reported by Kazmi et al & Sadiq et al.^{19,22} Aortic valve prolapse was present in 6.7% of total patients which is similar to our study.²² Brauner et al, Lue et al, and Ando et al, reported aortic cusp prolapse in about 5% with isolated congenital VSD, 12%, 16% cases, respectively.^{23,13,24}

Doubly committed subarterial type VSD is associated more frequently with progressive development of aortic cusp prolapsed and aortic regurgitation than other types of congenital VSDs similarly our study showed higher incidence of aortic cusp prolapse and aortic regurgitation with Doubly committed subarterial type VSD than perimembranous outlet VSD. About, 6.8% of perimembranous outlet VSD had aortic cusp prolapsed in our study, which is in contrast to Kazmi et al, that reported 10.4% of perimembranous outlet VSD having prolapsed aortic cusp.¹⁹ Somanath et al, found aortic regurgitation more frequently with perimembranous VSD as compared to other types of congenital VSD (84% Perimembranous vs. 16% doubly committed subarterial VSD).²⁵

Table 2: Complications Associated with VSD

Types	Cases n (%)
AVP	50(7.06%) • 30(6.86%) in perimembranous VSD • 20(41.6%) in doubly committed subarterial VSD
AVP with AR	40(5.64%) • 23(5.26%) in perimembranous VSD • 17(35.4%) in doubly committed subarterial VSD
Severe PAH	157(22.17%)
Eisenmenger Syndrome	2(0.28%)
Left Outflow Obstruction	5(0.7%)
Right Outflow Obstruction	10(1.4%)
Infective Endocarditis	4(0.56%)

Glenn et al, in their study found that 5.8% patients of VSD developed infundibular stenosis but in our study right ventricular outflow tract obstruction was found only in 1.4% of cases.²⁶ VSD is found to be the most common acyanotic CHD (30%) in our study. Worldwide, VSD is the most common acyanotic CHD accounting for 25-30% of all CHD.²⁷ This may be explained by the difference in genetic make up and ethnicity.^{27,28} The male and female distribution was same as in our study i.e about 60% and 40% respectively.²⁸

Sharmin et al, found the frequency of VSD as 42% compare to our study showing 30%.²⁹ They also found that 80% of patients with VSD have cardiomegaly on chest X Ray and about 30% developed heart failure symptoms in their study.^{3,29} Ibrahim et al, found VSD as the most frequent congenital heart defect in about 34% of CHD.³⁰ In a systemic review of about 114 different studies from different region across the globe which resulted in a total study population of 24,091,867 live births in which CHD was identified in 164,396 individuals. VSD frequency was 34%, the highest was among Asian i.e 9.1 per 1000 live birth followed by Europe (8.2 per 1000 live births), while the least frequency of CHD was found in Africa. This met analysis support our study frequency of VSD.³¹ The vast majority of VSDs (roughly 70%) are located in the area of the membranous septum and they are defined as being perimembranous or subarterial. With a relative high incidence in Asian countries, the association of the subarterial type VSD with aortic valve prolapse (AVP), and mainly right coronary cusp prolapse, and aortic regurgitation (AR) has been shown. According to the previous reports, the incidence of AVP in the sub arterial VSD was 36% to 79% which matches our study where AVP is found in about 41% of patients with doubly committed VSD.^{32,33}

Thanarant et al, reported the percentage of perimembranous, subpulmonic, muscular, inlet and multiple types as 70.3%, 19.4%, 5.6%, 3.1% and 1.6%, respectively. while in our study the perimembranous VSD was about 62% , muscular VSD is 22%, multiple and doubly committed VSD in about 7 and 6% respectively.³⁹ This may be due to racial and geographic variation in incidence of different VSD. The incidence of AVP in subpulmonic VSD was 87.1% compared to 16.4% in perimembranous VSD in their study, AR in sub pulmonic VSD was 37.1% compared to 5.3% in perimembranous VSD.⁸ In another study by Layangool et al, the percentages of perimembranous and subpulmonic VSD was 74.8%, 17.5% respectively, similar to the reports from the Eastern countries(17.5%-30.9%) compared to the reports from the Western countries (3.3%-6.9%).^{13,35,37} Our study shows prevalence of doubly committed sub arterial VSD similar to Western i.e about 7% while in contrast to Eastern studies.

Layangool et al, reported the incidence rate of AVP and AR in

subpulmonic VSD patients were 87% and 37% respectively. while in most study from western world subpulmonic VSD is associated with 20.8%-57.2% of AVP and 14%-37.4% of AR except from Tohyama study who also found the high prevalence rate of AVP (80%) and AR (42%) at 5 year of age.^{13,35,38-41} AVP and AR can be diagnosed in infants less than one year of age as some previous reports.^{41,42} The overall incidence rate of AVP and AR in perimembranous VSD in previous reports is 8.8%-14% and 6-6.8% respectively, which is in accordance to our study.^{13,42}

A large VSD is associated with severe pulmonary hypertension and exposes the patient to risk of developing pulmonary vascular disease. This is the major indication of surgery in patients with large VSD. Severe pulmonary hypertension was noted in 21.34% cases and it was seen almost exclusively with a large VSD.

CONCLUSION

Ventricular septal defect which account for about 30% of the congenital cardiac defect is associated with significant complications including pulmonary artery hypertension, Aortic cusp prolapse, aortic regurgitation and infective endocarditis causing morbidity and mortality, so it requires special attention to be diagnosed and treated early.

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