

## Repetitive Delay in Diagnosis of Ventricular Septal Defect

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### Abstract

**Objective:** Although ventricular septal defect (VSD) is the most common congenital heart disease, it is usually diagnosed late. The presentation of the disease is variable; sometimes it is so quiet and silent that might even improve and heal spontaneously, and in some certain cases if the appropriate, on time and early treatment is not done, this would lead to irreparable complications and mortality even in the early life period. This study reviews the diagnostic process, treatment and follow-up of the patients. It is hoped that the results of the present study be used to improve the patients' condition.

**Methods:** This was a cross-sectional study done on 145 patients with VSD during 54 months in Isfahan. The disease was identified through color Doppler echocardiogram, cardiac catheterization and angiography if necessary. The required data were collected at the time of definite diagnosis.

**Findings:** Mean age at initial and definite diagnosis of the disease was 17 months and 44 months, respectively. Heart murmur led to initial diagnosis in 85% of the cases. In 27.5% VSD was associated with other cardiac anomalies. Pulmonary artery hypertension existed in 16.5% of the cases. Fifty nine surgeries were performed on 40 patients.

**Conclusion:** In routine physical examination of the infants, the probability of heart disease should be considered; conducting echocardiogram in suspected cases would lead to early diagnosis and eventually timely treatment. Appropriate follow-up of the patients will provide optimal care and treatment at proper time.

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**Key Words:** Ventricular Septal Defect; VSD; Diagnosis; Congenital Heart Disease

### Introduction

Ventricular septal defect (VSD) is the most common congenital heart disease which can occur isolated or be associated with other cardiac abnormalities. It includes about 20% of the cases

of congenital heart disease and is observed more common in neonatal period. VSD is sometimes so quiet and silent that heals spontaneously and in some cases, if on time and early medical-surgical treatment is not implemented, this would lead to irreparable complications and mortality even in

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the early life period<sup>[1-7]</sup>. Although it is a congenital disease, it will be diagnosed neither at birth nor at early ages of life, and in some cases at initial diagnosis there is severe pulmonary artery hypertension which affects morbidity and mortality<sup>[8]</sup>. We conducted this study to evaluate the process of diagnosis, treatment and follow-up of our community patients, so that results of this study can be used for improving the patients' condition.

## Subjects and Methods

This cross-sectional study was conducted on 145 patients with VSD who had been referred to the researcher during 54 months. Diagnosis of the disease was based on clinical findings and confirmed by Color Doppler echocardiogram, cardiac catheterization and angiography if necessary.

## Findings

Studying 145 cases of VSD indicated that although the mean age at initial diagnosis was 17 months ( $16.77 \pm 13.70$ ), the mean age at definite diagnosis was 44 months ( $43.58 \pm 29.06$ ). Heart murmur led to the initial diagnosis in 123 (85%) cases. An isolated VSD existed in 105 (72.5%) cases and the rest had additional cardiac abnormalities. Table 1

indicates the type and number of additional cardiac anomalies in line with other findings. Pulmonary arterial hypertension (more than half of the systemic pressure) was observed in 24 (16.5%) cases from which 8 cases had an isolated VSD, and in 16 cases VSD was associated with other cardiac anomalies. Fifty nine surgeries were performed on 40 patients. Out of 34 VSD surgeries, in 15 cases an isolated VSD was found, and 19 cases had VSD associated with other cardiac abnormalities.

Twenty five cases of VSD associated with other cardiac anomalies were operated. The mean age of patients operated on isolated VSD was 45 ( $45.47 \pm 22.26$ ) months, and that of VSD associated with additional cardiac abnormalities was 37 ( $37.26 \pm 23.61$ ) months; and mean age at which associated cardiac abnormalities were operated was 10 ( $10.36 \pm 3.17$ ) months.

## Discussion

The most important finding in this study was delayed diagnosis of the disease. Diagnosis of VSD at the age of 44 months is too late and this might lead to irreparable complications. Although always and everywhere one talks about increased complications due to delayed diagnosis, the delay observed in current study is too significant. It would be appropriate that 88% and 96% of the patients be diagnosed during the first and fourth year of life, respectively<sup>[9]</sup>. The study of Baspinar in Turkey indicated that the age of diagnosis of congenital heart diseases was 2.2 years, he also

**Table 1:** Different types of VSD and additional cardiac anomalies, pulmonary arterial hypertension (PAH) and the number and age at surgery in the studied patients

Type of VSD	No (%)	PAH No. (%)	Surgery No. (%)	Mean (SD) Age at surgery (mo)
Isolated VSD	105 (72.5)	8 (7.6)	15 (14)	45 (22)
VSD with additional cardiac anomalies	40 (27.5)	16 (40)	19 (47.5)	37 (14)
Patent Ductus Arteriosus (PDA)	20 (50)	11 (55)	19 (95)	10 (3)
PDA with aortic coarctation	6 (15)	5 (83)	5 (83)	10 (3)
Atrial septal defect	5 (12.5)	-	1 (20)	44 (29)
Pulmonary artery stenosis	9 (22.5)	-	-	-
Bacterial endocarditis	1 (0.7)	-	-	-
Mortality	2 (1.4)	-	-	-

concluded that it requires urgent measures in terms of organization of early diagnosis and proper management<sup>[10]</sup>. The study of Brown 1992-2002 in Britain indicated that delayed diagnosis even in the neonates worsens the postoperative condition and outcomes of the surgery<sup>[11]</sup>. Sabramanyan in Oman showed that 70% of the congenital heart diseases have been diagnosed until the age of one year <sup>[12]</sup>. Mehta in United States indicated that probably all VSDs have been diagnosed before the first year of life <sup>[3]</sup>; and ultimately, Grech in Malta showed that although 97% of the heart disease cases have been diagnosed before age of four, the age of diagnosis is decreasing progressively <sup>[13]</sup>.

The interesting finding we faced with in this study was the 27-month interval between the initial and definite diagnosis. This repetitive delay or the interval between the initial or probable diagnosis and the definite diagnosis of the disease absolutely does not exist anywhere or had not been reported ever. For the patients of our study, the possibility of heart disease has been raised averagely at the age of 17 months, while the definite diagnosis was established at the age of 44 months. Certainly, if definite diagnosis was established and as a result of it appropriate treatment occurred at the age of initial diagnosis, not only the symptoms and complications would be less, but also the mean age of diagnosis in our patients had not been far different from that in other studies <sup>[10-13]</sup>.

In 85% of the cases, heart murmur led to possibility of the disease. Therefore, according to the physiopathology of the murmur in VSD, undoubtedly the heart murmur has existed before the mean age of the initial diagnosis and even after the age of 1 month <sup>[14]</sup>.

This long delay in the initial diagnosis either can be due to lack of any examination of the patient or in routine examinations, heart murmur and heart disease have been ignored. Therefore, if in some cases, the first reason is true, there should be some measures and interventions to examine all neonates and infants periodically and regularly.

However, because all the patients of our study had been frequently examined before the diagnosis of heart disease due to various reasons, it can be said with certainty that at the time of routine examination of the infants and children,

existence of disease has been ignored due to lack of necessary attention to the heart. It is worth to mention that Massin and Dessy in Belgium in their article titled as "delayed recognition of congenital heart disease" concluded that in all cases of late diagnosis, clinical cardiac findings were present that should have alerted the physician on the possible presence of underlying heart disease<sup>[14]</sup>.

Significant pulmonary arterial hypertension was observed in 16.5% of the cases. The increase in pulmonary artery pressure rate has a direct relation with mortality of this disease. Delayed diagnosis in the studied patients had an effective role in high pulmonary artery pressure. Bendriss in Morocco also concluded that surgically closing the VSD is the best way to avoid irreversible pulmonary arterial hypertension; the mean age of the patients in this study was 2.25 years <sup>[8]</sup>. In a study in Karachi it was concluded that low age could not increase the morbidity rate after VSD repair and there was no excuse of delaying in repair of VSD <sup>[5]</sup>. Other studies also confirmed this subject <sup>[6]</sup>.

The results of this study indicated that in 27.5% of cases VSD is associated with other cardiac abnormalities, which increased the severity of signs and symptoms of the disease and affected type and time of the treatment. Glen et al on 1488 VSD patients in Britain showed that 22% of the cases were associated with additional cardiac abnormalities and they suggested that this should be considered in the follow-up of the patients and the proper decision should be made<sup>[2]</sup>. The pulmonary arterial hypertension of our patients who had additional cardiac abnormalities was 5 times higher than in patients with isolated VSD; therefore, recognizing additional cardiac abnormalities and correcting them must be done as soon as possible to achieve better treatment results. Previous studies also confirmed this subject <sup>[2,7,8,15]</sup>.

The mean age of 10 months in the surgery of additional cardiac abnormalities of VSD our study indicates that earlier diagnosis of the patients will lead to better therapeutic results in lower ages.

Pulmonary arterial hypertension was observed in 55% of the cases of VSD with patent ductus arteriosus; while when aortic coarctation and patent ductus arteriosus were associated with VSD, 83% of them had pulmonary arterial

hypertension. In this regard, the importance of clinical follow-up and taking notice of the additional cardiac anomalies is emphasized more than ever<sup>[2,8]</sup>.

Pulmonary artery stenosis was one of the other common additional cardiac anomalies.

Pulmonary arterial hypertension was not observed in these cases and delayed surgery could be performed in a more appropriate condition. In the study of Glen in Britain, the prevalence of pulmonary artery stenosis with VSD was 8.5%<sup>[2]</sup>, in the present study it was 6.2%.

Although bacterial endocarditis was seen only in one case which unfortunately led to death, with earlier diagnosis of heart disease in the early age of life, endocarditis will be prevented<sup>[13,14,16,17]</sup>.

In terms of mortality, there were two fatal outcomes. One occurred after the surgery and the other was the result of subsequently acquired bacterial endocarditis, which indicated that if the disease had been diagnosed earlier and the required preventions and treatments specified, the condition of the patient would be different. Other studies also confirm this subject<sup>[8,11,14]</sup>.

## Conclusion

Heart examination should be considered during the routine and daily physical examinations of the infants in order to diagnose the possible heart disease. Conducting echocardiogram and cardiac consultation in the suspected cases would lead to earlier diagnosis and ultimately treatment of the heart disease. Considering the possibility of associated cardiac abnormalities and also pulmonary arterial hypertension would provide optimal care and treatment services at proper time.

**Conflict of Interest:** None

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