Isolated Ventricular Septal Defect

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Keywords

Ventricular Septal Defect (VSD) • Perimemberanous • Sub-arterial VSD Muscular VSD • Echocardiography

Definition

Ventricular septal defect (VSD), as the most common congenital heart disease (CHD) at birth [1], is found less in older infants and in the adults since most of small VSDs will close spontaneously over the time [2, 3].

Four anatomic types of VSDs have been described (with various nomenclature and synonyms) [2–6]:

- *Type 1:* Outlet supracristal, sub-arterial, subpulmonary, infundibular, supracristal, or conal VSD is located in the outflow tract of the right ventricle (RV), under the semilunar valves in conal or outlet septum, and has a variable prevalence ranging from approximately 6 % of defects in non-Asian patients to up to 33 % in Asian populations [4]. *Spontaneous closure in type 1 VSD is uncommon.* This type of VSD is often related to the progressive aortic regurgitation (AR) secondary to prolapse of the aortic valve cusps (usually right cusp).
- *Type 2:* Perimembranous, paramembranous, or conoventricular VSD constitutes almost 80 % of defects in the membranous septum. This defect might also extend into inlet, trabecular, or outlet septum. Perimembranous VSD is near to the tricuspid septal leaflet, which can frequently adhere to the defect and create a pouch or "aneurysm" in the ventricular septum. This pouch limits left-to-right shunting and may result in partial or complete closure of septal defect. This type of defect is also close to the aortic valve on the LV side of interventricular septum.
- *Type 3:* Inlet, atrioventricular (AV) canal, or AV-type VSD lies in the lower portion of RV, near to the tricuspid valve, and just inferior of

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AV valve apparatus. This type of VSD is typically seen in patients with Down syndrome.
- *Type 4:* Muscular or trabecular VSD is completely surrounded by muscle. It may have a central position (midmuscular) or an apical position or may be located at the margin of septum and RV free wall. This type of VSD is frequently multiple in number. Spontaneous closure is particularly common in this type, and although these lesions account for up to 15–20 % of VSDs in infancy, the incidence is much less in adults.

Associated Lesions

Although VSD is most often an isolated lesion, multiple defects can also be seen. Moreover, VSD is a common part of complex cardiac lesions such as tetralogy of Fallot (TOF) and transposition of great arteries (TGA). VSD also occurs in association with left-sided obstructive anomalies such as subvalvular aortic stenosis and coarctation of the aorta. Spontaneous closure of VSD most often occurs in muscular/trabecular type but is uncommon in perimembranous and outlet defects [7]. Although small defects can close spontaneously at any age, this most commonly happens during infancy [8, 9].

Pulmonary vascular resistance (PVR), size of the defect, LV and RV systolic/diastolic function, and presence of RV outflow tract obstruction (RVOTO) are the main determinants of the shunt direction and magnitude.

Clinical Presentation and Natural History

Possible scenarios for adults with an isolated VSD might contain:
- An asymptomatic patient with a systolic murmur
- Fever and bacteremia caused by infective endocarditis (IE)
- Progressive pulmonary vascular disease in patients with large VSD with originally large left-to-right shunting eventually resulting in cyanosis and exercise intolerance secondary to shunt reversal (Eisenmenger complex)

Clinical presentation of isolated VSDs is mainly determined by the defect size and PVR:
- Small VSDs (less than or nearly equal to 25 % of the aortic annulus diameter) have small left-to-right shunts, no LV volume overload, and no pulmonary artery hypertension (PAH) and present as systolic murmurs.
- Moderate VSDs (more than 25 % but less than 75 % of the aortic annulus diameter) have small to moderate left-to-right shunts, mild to moderate LV volume overload, and mild or no PAH. These patients may remain asymptomatic or experience symptoms of mild congestive heart failure. Symptoms generally subside with the medical management and with time as VSD diameter decreases in absolute terms or relative to the increasing size of the body.
- Large VSDs (greater than or equal to 75 % of the aortic diameter) are associated with a moderate to large left-to-right shunts, LV volume overload, and PAH. A history of congestive heart failure during infancy might be noted in most of adult patients with large VSDs.
- In patients with a small VSD, endocarditis (IE) may be complicated by pulmonary embolism or cerebral abscess.
- Postsurgical presentation consists of signs and symptoms associated with IE, aortic regurgitation, heart block, LV dysfunction, PAH, tricuspid regurgitation (TR), recurrent VSDs, and ventricular arrhythmias.

Clinical Examination

Clinically, VSD is characterized by a systolic murmur maximal at the left lower sternal border. A precordial thrill may also be palpable. The VSD murmur is “blowing” and “pansystolic” if the RV pressure is low. With higher RV pressures, the murmur becomes “shorter,” “softer,” and “lower pitched.” Small muscular VSDs are usually very high pitched and are heard only in early systole due to the closure of the defect by muscular contraction.

Diagnostic Workup

Clinical Examination

Clinically, VSD is characterized by a systolic murmur that is usually maximal at the left lower sternal border. A precordial thrill may also be palpable. The VSD murmur is blowing and pansystolic in the presence of a low pressure RV. With higher RV pressure, the murmur is shorter, softer, and lower pitched. Small, muscular VSDs are usually very high pitched and are heard only in early systole due to the closure of the defect by muscular contraction.

Electrocardiogram

The ECG may show biventricular hypertrophy or isolated RV hypertrophy in patients with a large VSD and significant PAH (Fig. 22.1).

Chest X-Ray

Patients with a small VSD have a normal chest x-ray. In the presence of significant left-to-right shunt, evidences of left atrial (LA) and LV enlargement and increased pulmonary vascular markings can be seen on CXR (Fig. 22.2). Patients with significant PAH do not demonstrate LV enlargement but will have a prominent pulmonary artery and diminished pulmonary vascular markings at the periphery of the lung.

Echocardiography

Echocardiography is the mainstay diagnostic technique used in both the diagnosis and assessment of disease severity. The large VSDs are easily detected by 2D echocardiography, as the small VSDs may be difficult to visualize. Most of the VSDs in adults have small flow; consequently a

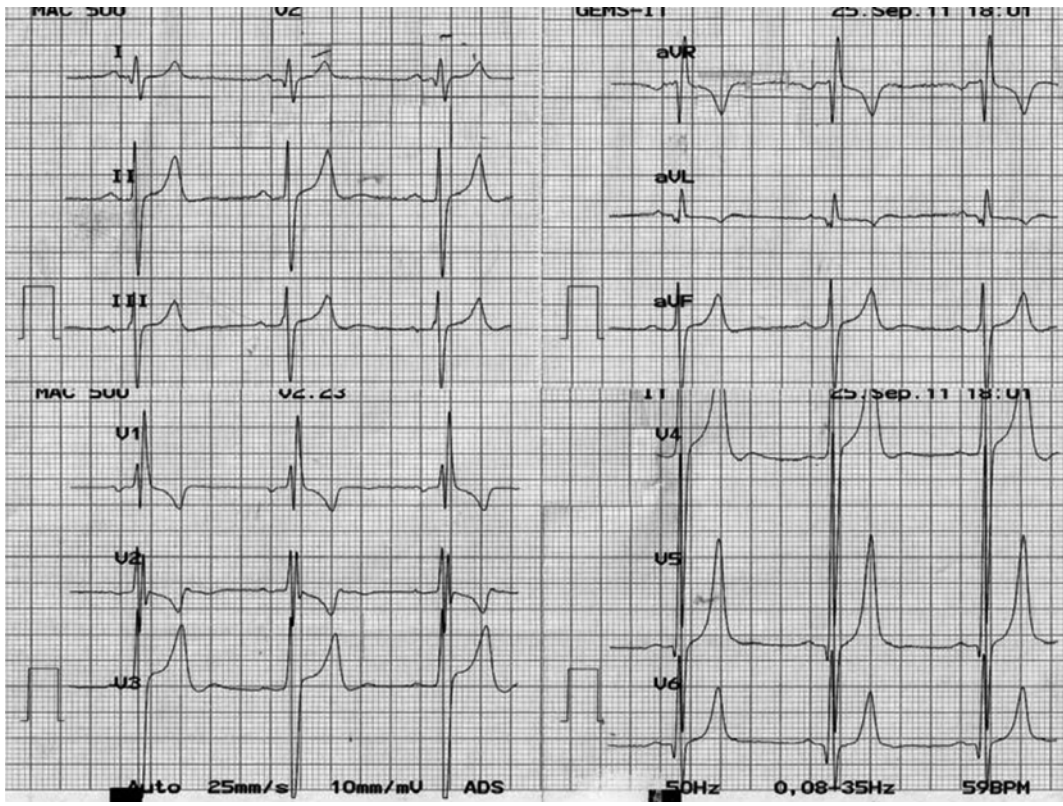

Fig. 22.1 Electrocardiography showing left atrium enlargement associated with biventricular hypertrophy

complete 2D and color flow imaging study besides the physical exam would be helpful.

Key findings on echocardiographic examination include the number of defects, location of defects, chamber size, ventricular function and the severity of LV volume overload, presence or absence of aortic valve prolapse and/or regurgitation especially in the case of outlet (supracristal) and high perimembranous VSDs, presence or absence of RV or LV outflow obstruction, and presence or absence of TR and estimated PAP.

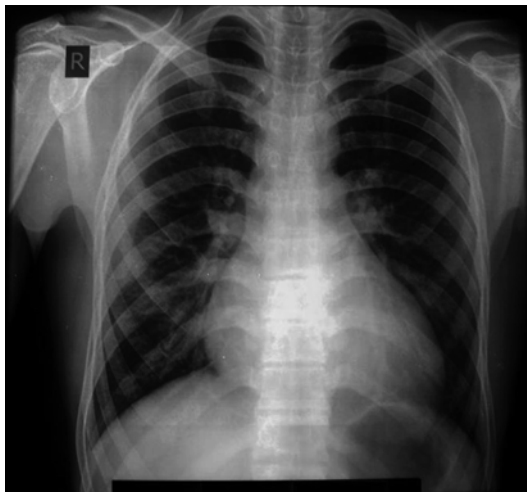

Fig. 22.2 Shunt vascularity with left-to-right shunt; left ventricular and left atrial enlargement are suggestive for ventricular septal defect (VSD)

VSDs that are categorized earlier will be described based on the adjacent structure:

A. *Perimembranous VSD* which is the most common type of VSD is seen *adjacent to the aortic valve and tricuspid valve*. Usually it is imaged in parasternal view (both long axis and short axis), apical 5-chamber view, and subcostal view (Fig. 22.3).
B. *Subarterial or doubly committed VSD* which is more common in Asian population is seen *adjacent to the both aortic valve and pulmonic valve* (*doubly committed VSD)* in the short-axis view (Fig. 22.4).
C. *Inlet-type VSD* usually is seen as a part of atrioventricular septal defect (AVSD) although it can be seen as isolated defect. It is *adjacent to both the mitral and tricuspid valves and best seen in the apical 4-chamber view* and short-axis view.
D. *Muscular-type VSD* which is surrounded by muscle is located anterior, posterior, apical, or midmuscular. Sometimes the muscular VSD has multiple defects known as "Swiss cheese" septum. Detection of muscular VSDs needs precise sweeping of the whole septum by 2D and color Doppler study (Fig. 22.5, Video 22.1).

Estimation of RV systolic pressure from TR jet, VSD jet, and/or septal configuration should also be performed.

Echocardiographic evaluation of VSD consists of the precise sweeping of the whole septum

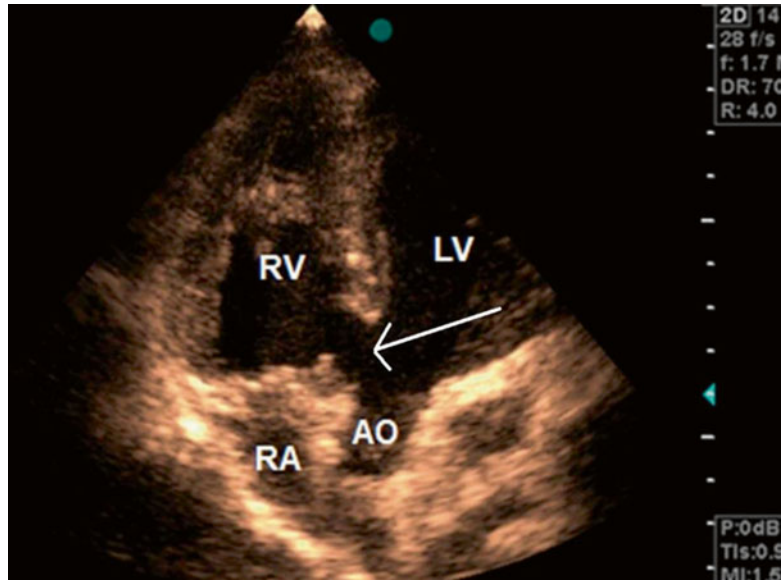

Fig. 22.3 Transthoracic echocardiography in five-chamber view showing large perimembranous VSD (*white arrow*). *RV* right ventricle, *LV* left ventricle, *RA* right atrium, *Ao* aorta

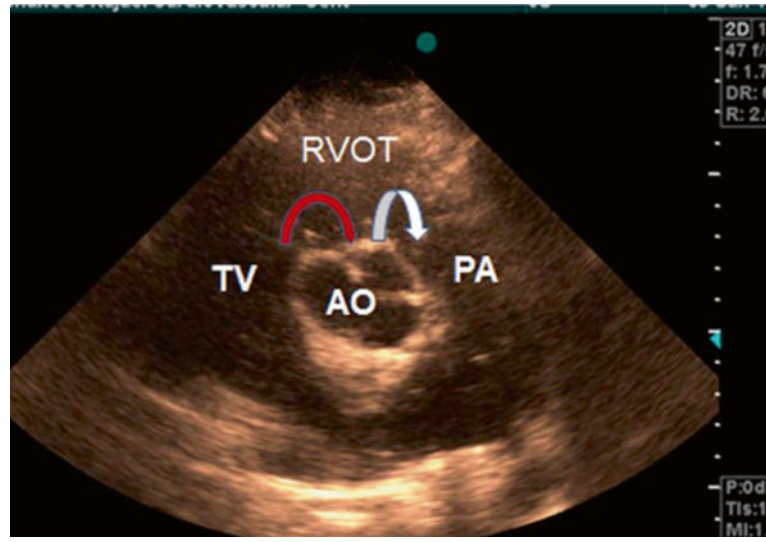

Fig. 22.4 *White arrow* demonstrating the subarterial location of the VSD and *red line* showing the location of the perimembranous VSD. *RVOT* right ventricular outflow tract, *TV* tricuspid valve, *AO* Aorta *PA* pulmonary artery

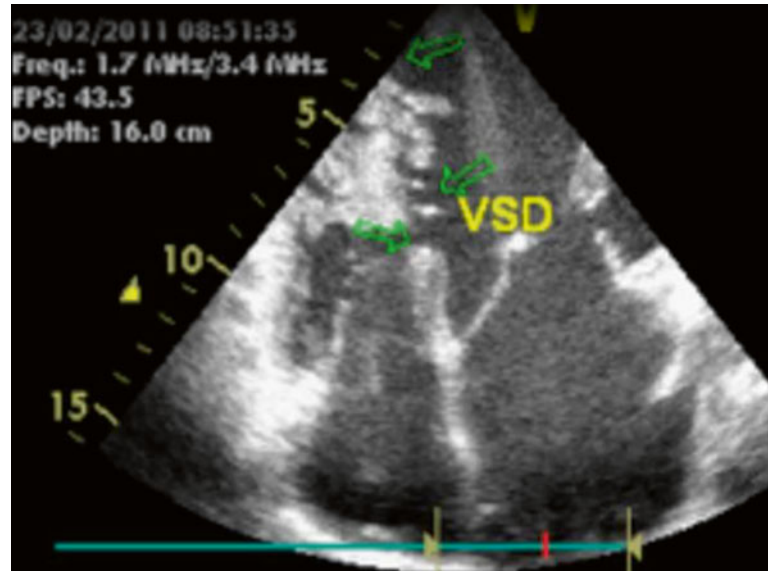

Fig. 22.5 Transthoracic echocardiography in 4-chamber view showing multiple midmuscular VSD (Swiss cheese septum) associated with left heart enlargement

by 2D and color flow imaging from the base to apex in multiple views.

Cardiac Magnetic Resonance

CMR may be useful, particularly for the following:

- Assessment of pulmonary artery, pulmonary venous, and aortic anatomy in the presence of coexisting lesions
- Assessment of LV volume overload and shunt quantification
- Assessment of the anatomy of unusual VSDs such as inlet or apical defects which are not well seen by echocardiography

Cardiac Catheterization

In patients with high PAP based on echocardiography data, cardiac catheterization is required to measure PVR. Moreover, ACC/AHA recommends to use cardiac catheterization for the assessment of the operability of adults with VSD and PAH (Class I, *Level of Evidence: C)* [10].

Cardiac catheterization provides the following data in adults with VSD:

- Quantification of shunting.
- Assessment of pulmonary pressures and resistance in patients with suspected PAH. Using

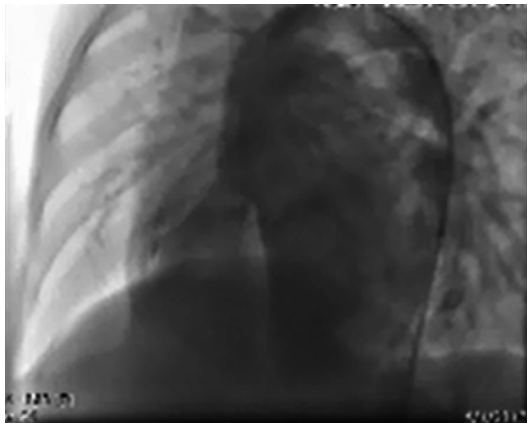

Fig. 22.6 LV injection showing subaortic VSD

pulmonary vasodilators, reversibility of PAH should be evaluated.
- Assessment of concomitant lesions such as aortic regurgitation and double-chambered right ventricle.
- Evaluation of VSD anatomy, particularly for patients in whom device closure is planned (Video 22.2) (Fig. 22.6).

Coincident opacification of the left and right ventricles indicates the presence of a VSD.

Management Strategies [10]

Medical Therapy

Adults with VSDs and severe PVD may benefit from pulmonary vasodilator therapy.

Surgical Ventricular Septal Defect Closure

In patients with a Qp/Qs (pulmonary to systemic blood flow ratio) of 2.0 or more and clinical evidence of LV volume overload, VSD should be closed surgically. Patients with a history of IE should also undergo surgical closure of the VSD. However, VSD closure is not recommended for patients with severe irreversible PAH.

Interventional Catheterization

Device closure may be considered in muscular VSDs, especially if the VSD is remote from the tricuspid valve and the aorta. Catheter device closure of VSD is performed in patients with residual defects following prior surgical closure, restrictive VSDs with a significant left-to-right shunt, traumatic VSDs, or iatrogenic VSDs after surgical replacement of the aortic valve. Restrictive VSDs in the adults may be closed in patients with a history of bacterial endocarditis or a hemodynamically significant left-to-right shunting (Qp/Qs >1.5:1). Percutaneous closure of VSD provides an alternative to surgery in patients with increased surgical risk factors, multiple previous cardiac surgeries, poorly accessible muscular VSDs, or "Swiss cheese"-type VSDs (Video 22.3).

Follow-up

Adults with VSD with residual heart failure, shunts, PAH, AR, or RVOT or LVOT obstruction should visit at least annually at an adult CHD center. In patients with a small residual VSD and no other lesions, 3- to 5-year intervals may be reasonable. Adults undergoing device closure of a VSD should be seen every 1–2 years depending on the location of the VSD. Continued follow-up is not required in adults with no residual VSD, no associated lesions, and normal pulmonary artery pressure.

Bifascicular block or transient trifascicular block after VSD closure increases the risk of development of complete heart block in later years. Thus, these patients should be followed up yearly by history, ECG, ambulatory monitoring, and/or exercise testing [10].

Additional Considerations

Pregnancy

Pregnancy in asymptomatic women with small VSDs, normal LV function, no PAH, and no associated lesions is generally well tolerated and not

accompanied by increased maternal mortality or cardiovascular risk for pregnancy. It is believed that although the increase in cardiac output during pregnancy may accentuate the left-to-right shunt, the decrease in peripheral resistance would counterbalance this effect. Pregnancy is contraindicated in patients with severe PVD (Eisenmenger physiology) since arrhythmias, ventricular dysfunction, and progression of PAH might occur in these patients leading to maternal and fetal mortality [10, 11].

Activity/Exercise

No activity restrictions are requisite for patients with small VSDs without associated lesions, pulmonary hypertension, significant arrhythmias, and normal left ventricular function and also after VSD closure. However, in the presence of PAH, activity is usually self-restricted, but patients should be advised to limit themselves to low-intensity recreational activity/sports and to avoid travel to altitudes above 5,000 ft. In long-distance air travel, dehydration should be prevented, and the need for supplemental oxygen must be consulted with an adult CHD specialist.

IE Prophylaxis

The European Society of Cardiology and also AHA/ACC recommend IE prophylaxis only for high-risk VSD patients.

References

1. Hoffman JI, Kaplan S. The incidence of congenital heart disease. J Am Coll Cardiol. 2002;39:1890–900.
2. Du ZD, Roguin N, Wu XJ. Spontaneous closure of muscular ventricular septal defect identified by echocardiography in neonates. Cardiol Young. 1998;8: 500–5. 277.
3. Kidd L, Driscoll DJ, Gersony WM, et al. Second natural history study of congenital heart defects. Results of treatment of patients with ventricular septal defects. Circulation. 1993;87:I38–51.
4. Graham Jr TP, Gutgesell HP. Ventricular septal defects. In: Emmanouilides GC, Riemenschneider TA, Allen HD, Gutgesell HP, editors. Moss and Adams heart disease in infants, children and adolescents. Baltimore: Williams & Wilkins; 1989. p. 724–46.
5. Prosad S. Ventricular septal defects. In: Gatzoulis MA, Webb GD, Daubeney PE, editors. Diagnosis and management of adult congenital heart disease. London: Churchill Livingstone; 2003. p. 171–8.
6. Jacobs JP, Burke RP, Quintessenza JA, Mavroudis C. Congenital heart surgery nomenclature and database project: ventricular septal defect. Ann Thorac Surg. 2000;69:S25–35.
7. Miyake T, Shinohara T, Fukuda T, Ikeoka M, Takemura T. Spontaneous closure of perimembranous ventricular septal defect after school age. Pediatr Int. 2008;50:632–5.
8. Neumayer U, Stone S, Somerville J. Small ventricular septal defects in adults. Eur Heart J. 1998;19: 1573–82.
9. Onat T, Ahunbay G, Batmaz G, Celebi A. The natural course of isolated ventricular septal defect during adolescence. Pediatr Cardiol. 1998;19:230–4.
10. Warnes CA, Williams RG, Bashore TM, et al. ACC/AHA 2008 guidelines for the management of adults with congenital heart disease: a report of the American College of Cardiology/American Heart Association Task Force on Practice Guidelines (Writing Committee to Develop Guidelines on the Management of Adults With Congenital Heart Disease). Developed in Collaboration With the American Society of Echocardiography, Heart Rhythm Society, International Society for Adult Congenital Heart Disease, Society for Cardiovascular Angiography and Interventions, and Society of Thoracic Surgeons. J Am Coll Cardiol. 2008;52(23):e143–263.
11. Baumgartner H, Bonhoeffer P, De Groot NM, et al. ESC Guidelines for the management of grown-up congenital heart disease (new version 2010). Task Force on the Management of Grown-up Congenital Heart Disease of the European Society of Cardiology (ESC); Association for European Paediatric Cardiology (AEPC); ESC Committee for Practice Guidelines (CPG). Eur Heart J. 2010;31(23):2915–57.