Ventricular Septal Defect (VSD) is a heart condition that is prevalent in children, where the walls of the heart chambers do not close properly, leaving a hole anywhere in the ventricular septum. This leads to a variety of symptoms such as breathlessness and getting tired easily among others. VSD can be detected by a Physician with auscultation for specific signs specific to it. This diagnosis can be confirmed with electrocardiography, echocardiography, chest x-rays, MRI and cardiac catheterization. It can be managed conservatively, especially if the defect is very small, with larger holes requiring surgical interventions. New innovations such as catheter closure are reducing the risk involved with surgical interventions.
Typically, the wall between the heart chambers or septum closes prior to birth to prevent oxygen-carrying blood from interspersing with oxygen deficient blood. If the wall does not close appropriately and leaves a hole between the chambers, thus allowing blood to go through from the left side of the heart to the right, it is known as ventricular septal defect (VSD). It is a congenital defect that makes the heart work more rigorously than usual because instead of oxygen-nourished blood being distributed around the body, it is sent back to the lungs.

Ventricular septal defect is defined as the abnormal communication between the two ventricular chambers of the heart. These defects can be isolated or associated with other preexisting congenital heart malformations such as tetralogy of Fallot.

The most common type of ventricular septal defect (VSD) is perimembranous VSD. Perimembranous VSD is defined as the abnormal communication between the two ventricles through an opening within the left ventricle outflow tract below the aortic valve.

When the opening is located just under the pulmonic valve and is within the right ventricle outflow tract, the term supracristal VSD is used. Muscular VSDs occur within the muscular wall of the ventricular septum and are the second most common type of VSDs. They can be multiple. The last type of VSD is known as posterior or endocardial cushion VSD. The atrioventricular node might be involved with this type of VSDs.

**Epidemiology of Pediatric Ventricular Septal Defect**

Ventricular septal defects are considered the second most common type of congenital heart disease preceded by bicuspid aortic valve anomaly.

The estimated incidence of VSDs is approximately 2 to 7% of live births. The exact type of VSD is dependent on certain risk factors as well as the ethnicity of the patient. For instance, supracristal VSDs are far more common in Japan than in the United States.

Small VSDs may be asymptomatic and are known to undergo spontaneous closure within one year of life. Therefore, it is highly believed that the true incidence of VSDs at birth is
higher than what has been reported.

**VSDs have been found to be more common in females.** Small VSDs have an excellent prognosis without any significant increase in the mortality rate. On the other hand, moderate and large VSDs may be associated with significant morbidity if left untreated. Patients who develop significant pulmonary arterial hypertension might develop a reverse of their shunting into a right-to-left shunt. This phenomenon is known as **Eisenmenger’s syndrome** and is associated with significant morbidity and mortality. Patients with VSDs are also at risk of developing **infective endocarditis**.

**Etiology of Pediatric Ventricular Septal Defect**

All types of VSDs have been linked to maternal **phenylketonuria**. Maternal use of alcohol may increase the risk of muscular ventricular septal defects. The most important risk factor for VSDs is perhaps a family history of the disease. A family history of a VSD increases the risk in the future offspring by 3 times.

**Certain chromosonal abnormalities have been linked to an increased risk of VSDs** and include trisomy 13, trisomy 18, and trisomy 21. Despite the identification of these risk factors for VSDs, most cases remain of unknown etiology.

**Classification of Pediatric Ventricular Septal Defect**

The ventricular septum has two sides, a membranous part which is smaller, and a muscular area which is larger. The muscular part of the septum is further divided into three parts: an inlet, trabecular and outlet also called **infundibular septum**. The most useful clinical and surgical classification of VSDs depends on the location of the hole anywhere within the septum.

Any defect in the smaller membranous septum is termed as **perimembranous VSD**, and is the most prevalent. When there is a hole in the muscular septum, it is referred to as a muscular defect. This is further classified according to the exact location of the hole in the muscular septum.

If the hole is close to the arterial valves, it is called a sub arterial **infundibular defect**. This type of VSD usually leads to **aortic regurgitation** because the aortic valve collapses into the right ventricle. It is responsible for 5 to 7 % of VSDs in the West and approximately 30 % in the Middle East and Asia.

A **Coventricular Ventricular Septal Defect** develops when the ventricular septum beneath the pulmonary valves does not meet. The development of a hole adjacent to where blood passes through the tricuspid and mitral valves into the ventricles is known as inlet Ventricular Septal Defect and is responsible for 5 to 8 % of cases of VSD.

In situations where a person develops a myocardial infarction within the septum, it can lead to **acquired VSD**. However, these types are very rare and have poorer outcomes.

**Pathophysiology of Pediatric Ventricular Septal Defect**
The first pathologic consequence of a ventricular septal defect is the **formation of a left-to-right shunt**. This is associated with increased left ventricular volume overload due to increased pulmonary venous return to the left atrium. Pulmonary over circulation is commonly seen with moderate and large VSDs. Additionally, pulmonary arterial hypertension is a common complication.

The route and quantity of systolic flow through a VSD is either through the usual pathway (through a ventricle) or via the VSD and is dependent on the relative resistance offered by each path. If the VSD is big and offers no resistance, the resistance to pulmonary circulation is lower than that of the systemic circulation, leading to high systolic blood flow from left to right.

With a smaller VSD, higher resistance at the hole restricts shunting from left to right, even if pulmonary resistance is low. This increases Left Ventricle volume significantly as the body tries to increase cardiac output, leading to more pressure on the Left Atrium. This results in pulmonary venous congestion either at rest or when a person exerts energy.

Also, notwithstanding the size of the VSD, if the pulmonary resistance is more than that of the systemic, then the blood will shunt from right-to-left. A large VSD elevates systolic pressure at ventricles, atriums and pulmonary artery. This influences the occurrence of pulmonary vascular disease or **Eisenmenger's syndrome**.

**Pulmonary venous pressure** also increases in patients with VSDs due to the dilatation and hypertrophy of the left ventricle which is associated with elevated end-diastolic pressure. Additionally, patients with a moderate to large VSD are known to have pulmonary over circulation, elevated pulmonary arterial pressure, and elevated pulmonary capillary pressure. Elevated pulmonary capillary pressure can cause
pulmonary edema.

Cardiac output decreases in patients with VSDs because the blood escapes from the left ventricle to the right ventricle instead of going into the aorta.

Prolonged elevated pulmonary hypertension is associated with pulmonary arterial hypertrophy and persistent pulmonary vascular disease. This can elevate the pressure in the right ventricle to surpass that of the left ventricle and cause a reversal of the shunt.

Clinical Features of Pediatric Ventricular Septal Defect

Symptoms

Small VSDs rarely cause symptoms. If symptomatic, patients can complain of mild exertional dyspnea. Small VSDs are usually an incidental finding due to the hearing of a murmur during a routine physical examination.

Moderate VSDs can cause excessive sweating, difficulty with feeding, and tachypnea. Patients with moderate VSDs usually have problems with gaining weight unlike those with small VSDs who grow normally. These symptoms are usually obvious by the age of 3 months.

Larger VSDs tend to cause more severe symptoms and can present with congestive heart failure. The risk of recurrent pulmonary infections and infective endocarditis increases with larger VSDs.

Patients who develop Eisenmenger’s syndrome are usually asymptomatic at rest but develop severe dyspnea, chest pain or syncope during exertion.

The murmur of a VSD is usually a harsh holosystolic murmur that is better heard over the lower left sternal border. Cyanosis can be evident in patients who have a right-to-left shunt.

The symptoms of VSD are usually visible and can be diagnosed a few days, weeks or months after a child is born. Typically, the type of symptoms a child will manifest depends on the size of the VSD. In some cases, small holes close by themselves without the baby showing any signs of VSD. In some cases, a VSD may not be noticeable until adulthood. On the other hand, larger holes manifest the following symptoms:

- Shortness of breath, even without exertion
- Fast breathing
- Breathlessness during small activities like eating or crying
- Getting tired easily while feeding or playing
- Poor feeding habit and not gaining weight.

Signs

Signs of VSD may present at birth or may not appear until some time after birth. A physician can detect VSD by auscultation, with murmurs, which are basically labeled as holosystolic or pansystolic, heard during the process. The degree of the murmur is determined by the velocity of blood flow through the VSD, while the site of the hole determines the location of the murmur.
While minor VSDs are very loud with a thrill, larger VSDs have a constant sound all through the cardiac cycle, without a thrill. However, a child with Eisenmenger’s syndrome, a large defect, and a right-to-left shunt will not produce a VSD murmur. But they are usually **cyanotic** with club fingers and the second pulmonary heart sound is louder.

If the origin of the VSD is muscular, murmurs will be heard along the boundary of the lower-left sternum. For infundibular VSDs, the murmur will be heard at the boundary of the left upper sternum. Along with the usual murmur, a perimembranous VSD comes with a systolic click sound that mimics a tricuspid valve aneurysm.

The pulmonary element of the second heart sound is well defined. Larger defects that have no shunts but are accompanied by tricuspid regurgitation can be heard either at the left lower or right lower boundary of the sternum.

With the patient leaning forward in a sitting position, a decreasing diastolic murmur is heard at the boundary of the left sternum if there is aortic insufficiency.

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**Figure A** shows the structure and blood flow in the interior of a normal heart. **Figure B** shows two common locations for a ventricular septal defect. The defect allows oxygen-rich blood from the left ventricle to mix with oxygen-poor blood in the right ventricle.

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**Diagnostic Workup for Pediatric Ventricular Septal Defect**

Patients with significant pulmonary hypertension and a right-to-left shunt might develop **polycythemia** due to **hypoxemia**.
**Electrocardiography**

Electrocardiography is usually normal but might reveal left ventricular hypertrophy. Right ventricular hypertrophy can be seen in patients with a prolonged history of large VSDs.

Patients with a small defect typically have normal ECGs. However, if shunt increases, signs of an increase in left ventricular volume, along with its hypertrophy may be present. The left atrium may appear enlarged. If the pulmonary artery pressure is raised, then the following will be obvious on the ECG: right axis divergence, hypertrophy of the right ventricle and enlarged right atrium.

**Chest Radiography**

Imaging studies are needed for the diagnosis of VSDs. Chest x-rays might show a normal heart size with or without increased pulmonary vascular markings due to pulmonary over circulation.

**Small defects will not be obvious on a chest x-ray.** On the other hand, larger defects reveal various grades of an enlarged chamber, which is dependent on the output of the shunt. Increased pulmonary blood flow is evident, but if the patient has Eisenmenger syndrome or high pulmonary resistance, evidence of pulmonary vascularity is lost, along with tightening of the vessels. Furthermore, these patients do not show any enlargement of the right heart or dilation of the principal pulmonary artery.

**Echocardiography**

Echocardiography is the imaging modality of choice for the diagnosis of VSDs. Color flow Doppler echocardiography is useful in the identification of the flow and size of VSDs. Echocardiography is sensitive for all types of VSDs.
It is important to evaluate the size of the right and left ventricles, the right ventricular pressure, and the pulmonary arterial pressure on the echocardiography for risk stratification.

**High-Yield:** The subcostal approach is best for the visualization of a perimembranous VSD. Supracristal VSDs are best viewed on a parasternal long-axis view in echocardiography. Muscular VSDs are best viewed with color flow Doppler.

Echocardiography is a noninvasive instrument used to investigate VSDs that accurately differentiates between the structures of the heart along with any abnormalities. Echo (as it is simply called) will determine the presence of increased pressure in the pulmonary artery, an impediment to right ventricular flow, deficiency of the aortic valve, and deformation of the valve operation. If transthoracic echocardiography does not allow the production of quality images to determine the cardiac anatomy as outlined above, a transesophageal echo can be carried out. Shunts and other defects that are difficult to define with 2-D echocardiography are more accurate and can be visualized with 3-D imaging.

**Magnetic Resonance Imaging**

Magnetic resonance imaging is becoming common in the evaluation of previously mentioned parameters and in the identification of the level of the left-to-right shunt.

It is possible to use magnetic resonance imaging to define the type of VSDs in patients with accompanied complex lesions.

**Cardiac Catheterization**

Catheterization offers precise parameters for the resistance offered by pulmonary vasculature, pulmonary reactivity, and quantity of shunting. It can also help in determining the reaction of the above to pulmonary vasodilators, so as to guide management. The site of the VSD, as well as the amount and grade of aortic inadequacy, can be determined by angiography.

**Differential Diagnosis of Pediatric Ventricular Septal Defect**

**The following are the related general differential diagnosis:**

**Atrial septal defect**

It presents with a softer murmur that is heard in the middle or end of systole rather than holosystolic. The murmur is also towards the left parasternal region (which is higher up) and is caused by blood flow through the pulmonic valve.

The defect and location of the shunting are shown on an echocardiogram.

**Patent ductus arteriosus**

The murmur is heard at the base of the heart, throughout the systole and diastole. A defect will be visible at the ductus arteriosus on an echocardiogram.

**Mitral regurgitation**

Even though a holosystolic murmur is present, it is heard around the mitral area, which is
likely to be transmitted to the axilla or border of the lower-left sternum.

An echocardiogram will show no sign of VSDs but will reveal a backward movement of valve leaflets in the middle of systole.

**Tricuspid regurgitation**

A holosystolic murmur is heard at the lower part of the left parasternal area. A classic sign of tricuspid regurgitation is an upsurge in the intensity of the murmur during inhalation (Carvalho’s sign). An echocardiogram will show blood spewing out through the tricuspid valve.

**Pulmonic stenosis**

An ejection murmur during systole is heard at the top left parasternal boundary. A turbulent flow along with a gradient through the pulmonary valve is revealed by an echocardiogram.

**Tetralogy of Fallot**

Characteristically presents with a murmur and cyanosis. It usually will have been diagnosed while the baby is still in the womb through a fetal echocardiogram.

On examination, increased breathing and cyanosis is noted in severe cases. There is also a solitary second heart beat, an upsurge of impulse on the right ventricle and a strong murmur on systolic ejection prominent at the border of the left sternum.

Along with the VSD, an echocardiogram will show pulmonary stenosis, preponderant aorta, and hypertrophy of the right ventricle.

**Treatment of Pediatric Ventricular Septal Defect**

**Medical Management**

The symptoms of the VSD will determine medical therapy to be applied. While a small VSD does not require any intervention, average to large defects need different types of conservative and surgical management. In cases where congestive heart failure is present in infants, it is managed with diuretics and digoxin.

Adults who still have a defect are referred for surgery, and should continuously be monitored for any dysfunction of the aortic valve. Those with Eisenmenger syndrome require special care from trained professionals in specialized centers. Vasodilators are a necessary adjunct to conservative treatment of VSD and improve functionality.

**Surgical Management**

- The site of the VSD is the main indication for surgery.
- If the heart chambers are enlarged and impact on the grade of shunting, there may be need for surgical closure.
- Typically, a Qp: Qs of 1.5:1 to 2:1, or signs of an upsurge in resistance of pulmonary arterioles is pinned down for surgical closure.
- The detrimental effects of surgeries and bypass techniques have been reduced through technological advances
Catheter Closure

Innovations in catheter method and appliances have led to the advent of percutaneous closure of defects. While transesophageal echocardiography has traditionally been used, intracardiac echocardiography can presently be applied with accuracy and safety obtained through transesophageal echocardiography.

A nitinol implantable device for closing a muscular VSD (Ventricular Septal Defect). The centre is 4 mm across, and it's mounted on the delivery catheter.

The use of devices comes with risks and complications. This can range from total heart blockage to tricuspid stenosis and device failure. The use of less invasive techniques can provide treatment to those who cannot undergo bypass.

References


Centers for Disease Control and Prevention: Congenital Heart Defects (CHDs): Specific Heart Defects: Facts about Ventricular Septal Defect

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