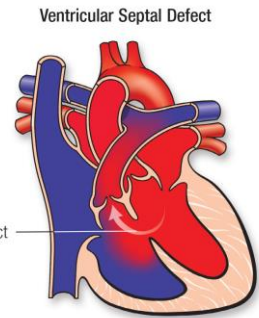


Ventricular Septal Defect (VSD)

Ventricular Septal Defect (VSD) is a common type of heart defect. It is a hole (or a number of holes) in the **ventricular septum**, which separates the left and right lower pumping chambers (ventricles) of the heart. When there is a hole (VSD), because of normally higher pressure in the left ventricle, blood is shunted (pushed) from the left ventricle into the right ventricle, potentially causing **pulmonary overflow** (increased blood flow in the vessels in the lungs) and **pulmonary hypertension** (high blood pressure in the lungs). The risk of pulmonary overflow and pulmonary hypertension depends on the size and location of the VSD. Small VSDs restrict the amount of blood shunting, leading to milder symptoms, and sometimes can close on their own. Large VSDs are non-restrictive, so blood flow to the lungs is increased. In this case, surgery in early infancy may be recommended to prevent the development of **pulmonary vascular disease**. Please **consult your cardiologist** for more information about your specific case.

VSDs can be classified into 4 types based on the location of the hole(s) on the septum. **Conoventricular VSD** (or perimembranous or subarterial), where the hole is located in the upper part of the septum just before the pulmonary valve and aortic valve, is the most common VSD in individuals with 22q11.2 deletion syndrome (22q11.2DS).

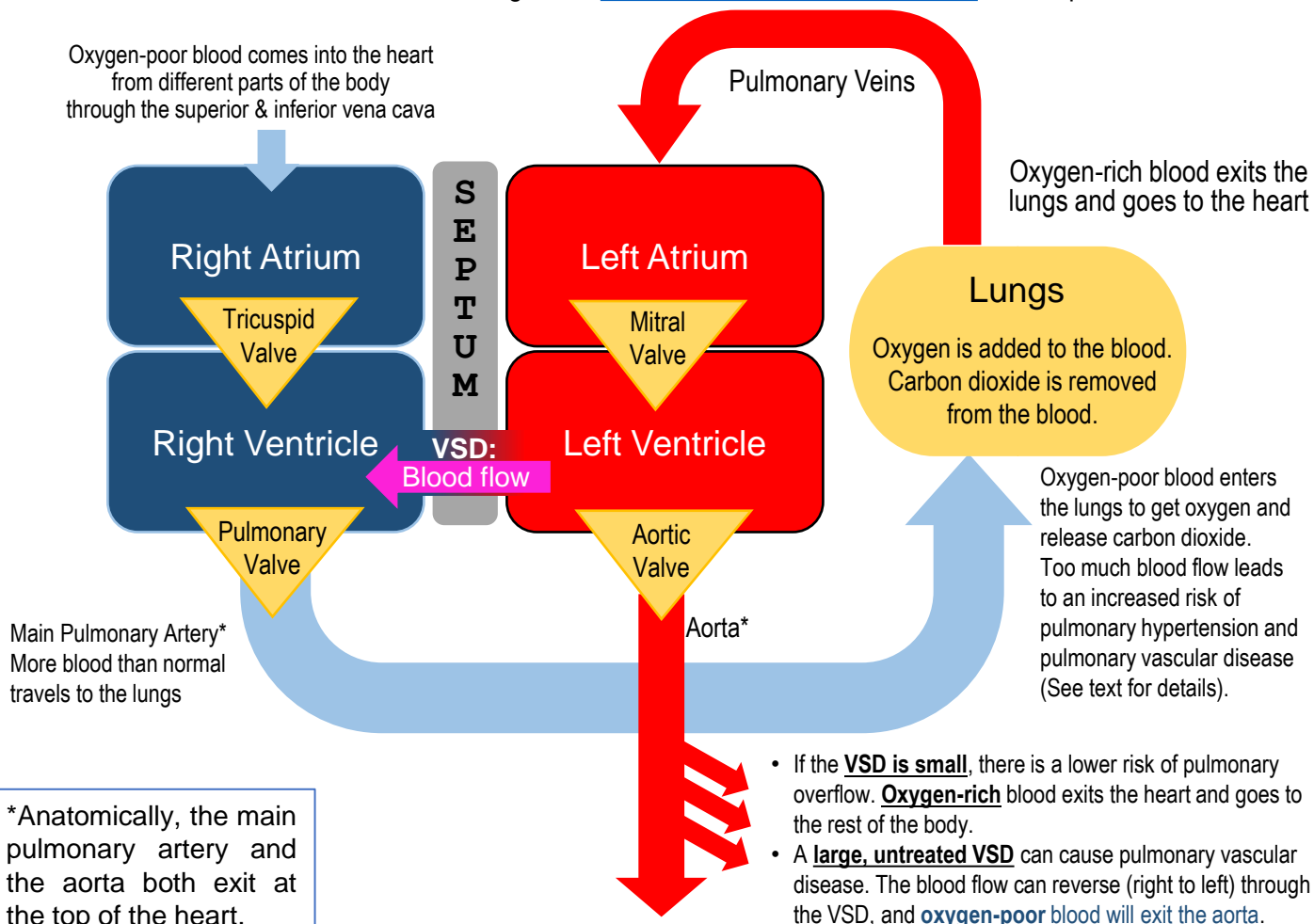


Please click on the image to see a larger version on the website of the [American Heart Association](http://www.heart.org)

A Schematic Diagram of the Heart with Ventricular Septal Defect (VSD)

Shown as if you are facing the patient; Not to scale

Please also see our schematic diagram of [The Heart and Normal Blood Flow](#) for comparison.



Ventricular Septal Defect (VSD) (continued)

Ventricular Septal Defect (VSD) and Individuals with 22q Differences

- Of all patients with **conoventricular VSD**, [5%](#) have 22q11.2 deletion syndrome (22q11.2DS).
 - Conoventricular VSD is a type of VSD where the hole is located in the upper portion of the septum just before the pulmonary valve and aortic valve
- [10 to 50%](#) of children with 22q11.2DS are born with conoventricular VSD.
- In a study, [3 out of 85](#) children with 22q11.2 duplication syndrome (22q11.2DupS) were born with VSD.
- VSD is one of the four features of **Tetralogy of Fallot (ToF)**, which happens in [20 to 45%](#) of children with 22q11.2DS and in a small proportion of those with 22q11.2DupS. For more information, please read our info sheet on [Tetralogy of Fallot](#).

Diagnosis for VSD

- VSD is usually diagnosed after birth, but the baby may not show symptoms if the hole is small.
- For babies where the VSD hole is large, they may have symptoms such as: difficulty breathing (dyspnea), irritability, sweating and tiredness while feeding, poor feeding and poor weight gain, and a higher susceptibility to respiratory infections.
- During a physical exam, the doctor may hear a “whoosh” sound (a murmur) of the heart. He/she may request an echocardiogram (an ultrasound of the heart) to check the severity of the VSD.

Treatment for VSD

- If the hole is small, the doctor will check the heart regularly. Sometimes the hole closes on its own.
- If the hole is large or does not close on its own, the child may need a surgery to patch it.
- Some children may need medications (e.g. diuretics to reduce the volume of excess blood reaching the lungs)
- Babies who are not gaining weight may need a high-calorie formula or even a feeding tube.

Ongoing care for VSD

1. **Follow-up with a cardiologist.** He/she may recommend **tests** to make sure the heart works well.
2. If the VSD is small or is closed with surgery, the individual may participate in **normal activities**. Please discuss exercise tolerance with your cardiologist.
3. Some people may need **endocarditis prophylaxis** – This means taking antibiotics before certain dental procedures to prevent bacteria from causing infections in the heart. Please discuss with your cardiologist.
4. Women planning on **getting pregnant should discuss their risks** with their healthcare providers.

For more info, please visit the websites of the [CDC](#) or the [American Heart Association](#).