

Pulmonary Atresia with Ventricular Septal Defect

Patient Information Series – What you should know, what you should ask.

What is Pulmonary Atresia with Ventricular Septal Defect (PA-VSD)?

Pulmonary Atresia with Ventricular Septal Defect (PA-VSD) is a complex congenital heart defect (CHD). It occurs in less than 1 in 10,000 fetuses. Our hearts pump blood out through two Great Arteries, the Aorta, which carries blood rich in oxygen from the left pumping chamber of the heart, called the left ventricle, to the body and brain, and the Pulmonary Artery, which carries oxygen poor blood from the right pumping chamber, or right ventricle, to the lungs. Blood flow from the heart ventricles into the great arteries is controlled by one-way doors, called valves- the aortic valve and the pulmonary valve. In pulmonary atresia, the valve between the right ventricle and the pulmonary artery does not develop properly.

The right and left ventricles of the heart are separated by a wall of muscle, the ventricular septum. A hole can sometimes occur in this septum: this is known as a ventricular septal defect (VSD). VSD's are quite common, in fact they are the most common form of heart defects at birth. PA-VSD is diagnosed when pulmonary atresia and VSD occur together. In this case, the aortic valve will attach to the heart ventricles such that it straddles the VSD; this is called an Overriding Aorta. This combination of factors causes many changes in fetal heart anatomy and function. In particular, the course of blood flow and the delivery of oxygen-rich and oxygen-poor blood, and the further growth of the heart ventricles and other blood vessels, are affected. Each case of PA- VSD is unique, depending on these and many other developments. For example, how far the aorta overrides the VSD may vary. In some cases of PA-VSD, the aorta does not override the VSD. Both pulmonary atresia and VSD can occur separately from each other, and these defects can occur with other types of CHD. Your caregiver will refer you to specialists in fetal cardiology (a maternal-fetal medicine specialist or a pediatric cardiologist) to diagnose as accurately as possible your fetus's individual anatomic structure and monitor the progress of the defect during the course of your pregnancy.

How does a PA-VSD happen?

There appears to be a genetic component in PA-VSD, however in many cases no specific cause can be found. It may occur in genetic conditions such as DiGeorge syndrome (a change in a small portion of the genetic material called chromosome 22) or Trisomy 21 (also known as Down Syndrome), or others. There is an increased risk of PA-VSD in siblings of a previous child with this defect, and in the children of individuals with a heart condition called Tetralogy of Fallot.

Should I have more tests done?

Your caregiver may refer you for genetic counseling and genetic testing. This may include tests like amniocentesis or chorionic villus sampling (CVS) to check for chromosomal abnormalities and other genetic diagnostic tests such as chromosomal microarray testing or whole exome sequencing. Such testing can provide essential information regarding your individual case. In addition, your caregiver may refer you to specialists in fetal cardiac disease, such as a maternal-fetal medicine specialist and/or a pediatric cardiologist. They can monitor your baby's progress with fetal echocardiography: ultrasound scans focused on the fetal heart and cardiovascular system. You will likely be offered tests to look for some of the known genetic changes that can cause PA-VSD. Which test is offered depends on many factors.

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What are the things to watch for during my pregnancy?

Your caregiver will probably order serial ultrasound scans, to watch carefully how the heart defects are developing, how they are affecting your baby's growth and well-being, and to monitor for any signs of distress. You may be asked to track your baby's movements through the day.

What does it mean for my baby after it is born?

In PA-VSD, blood flow from the right ventricle to the pulmonary artery is blocked, so blood is not pumped to the lungs, and a mixture of oxygen-rich and oxygen-poor blood flows out through the VSD to the aorta. The ductus arteriosus is a small vessel that carries blood between the aorta and the main pulmonary artery during fetal life. Normally, after delivery the ductus arteriosus closes. In PA-VSD, the infant is treated with a medication called prostaglandins to prevent closure of the ductus arteriosus, to keep blood flowing to the lungs by going around the blocked pulmonary valve, until surgery can be performed.

PA-VSD is treated with cardiac surgery with a goal of creating a channel between the right ventricle and the pulmonary artery and closing the VSD. The surgical approach is individualised according to the infant's specific anatomy and needs. Your baby will be evaluated with a heart ultrasound and other imaging tests as required, such as chest X-ray, to assist the surgeons in devising the optimal management plan. PA-VSD is usually managed with a series of surgeries, to correct the anatomical defects gradually and allow the heart, lungs and blood vessels to adapt.

Will it happen again?

It appears that there is a genetic component to PA-VSD. As mentioned, if a child suffered from PA-VSD, their siblings will be at a greater risk than the general population. This also depends on whether a genetic cause was found to explain the PA-VSD. If no genetic reason was found for the problem, the risk of this happening again is less than 5%. So, it is not necessary that your other children will also be affected. Your caregiver will likely order early targeted fetal scanning to rule out fetal heart defects in subsequent pregnancies.

What other questions should I ask?

- Does this look like a severe PA-VSD?
- How often will I have ultrasound examinations done?
- Where should I deliver?
- Where will the baby receive the best care after it is born?

Pulmonary Arterial Hypertension with Ventricular Septal Defect

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