

## **ATRIOVENTRICULAR SEPTAL DEFECT**

### **What is an atrioventricular septal defect?**

An atrioventricular septal defect (AVSD) is a congenital heart defect. It is also referred to as a atrioventricular (or AV) canal or an endocardial cushion defect. One area of the heart does not develop correctly, and this leads to several issues. The wall between the two bottom pumping chambers (ventricles), or ventricular septum, does not form correctly, leaving a hole, or ventricular septal defect (VSD). There is also a hole between the two top collecting chambers (atria), called an atrial septal defect (ASD). There are normally two valves which divide the atria from the ventricles, allowing blood to go from the atria into the ventricles, called a tricuspid and a mitral valve. In AVSDs there is one large valve (common AV valve) that divides the atria and the ventricles. In most cases of AVSDs, both ventricles are equal in size, and we call these “balanced AVSDs”. If one ventricle is larger than the other, these defects may be called “unbalanced AVSDs”. (These result in different symptoms and require a different approach, and are not discussed here.)

### **What causes AVSDs?**

AVSDs are common in Down syndrome, and can occur with other genetic syndromes. However, this heart defect can develop in otherwise normal people. In those cases, we don’t know what causes the heart to form abnormally.

### **How is an AVSD diagnosed?**

AVSDs are usually diagnosed with an echocardiogram an ultrasound of the heart. First trimester screening is a very useful screening tool to identify patients who might have a higher risk for this condition. If your first trimester screening indicates that your baby is likely to have this condition, a fetal echocardiography will be done later in pregnancy to confirm. If you don’t have first trimester screening, routine targeted ultrasound can diagnose AVSD. Sometimes AVSDs are not be detected until after birth. These defects may be picked up after a diagnosis of Down syndrome. Sometimes, the baby may have a murmur (abnormal heart sound) or other abnormality that indicates the problem.

### **How will your pregnancy be managed?**

If your baby is diagnosed with AVSD, a high-risk obstetrician from Center for Advanced Fetal Care will participate in your obstetric care. Overall care should be transferred to a specialized center, where multidisciplinary care is available. The team should include a perinatologist, fetal and pediatric cardiologists, a genetic counselor, a neonatologist and a pediatric cardiac surgeon. Fetal well-being will be followed closely by fetal ultrasound and nonstress tests. Towards the end of pregnancy, visits may be as often as two to three times a week. If there is no specific maternal or fetal reason for a C-section, vaginal delivery is often possible. Induction of labor is often scheduled for pregnancies affected with AVSD to make sure that all of the team members are available at the time of delivery

### **Why do AVSDs make babies sick?**

Before birth, babies are not generally affected by this heart defect. The pressures on the right and left sides of the heart are similar, and the blood flows almost normally.

After birth there are changes in the baby which lead to new patterns of blood flow. The pressures are high in the lungs before birth and start to decrease right after birth and through the first few months. Since there is a large VSD, blood from the right and left ventricles can go through the hole to either side of the heart. Blood will follow the path of least resistance (to the lower pressure). As the pressure in the lungs drops, more and more blood will flow across the VSD (from left to right) and into the lungs. The heart will continue to pump enough blood so that the body gets what it needs, but it will have to pump extra blood to compensate for the blood going to the lungs.

The heart will deal with this extra work by enlarging and pumping more blood with each beat. The process is inefficient, sending blood with oxygen back to the lungs where it came from, and this makes the heart have to work very hard. The heart is able to deal with this extra work for a while, but eventually it will lead to babies developing symptoms. Babies will start to breathe harder and faster, they will have trouble feeding, and they will not be able to gain weight.

### **What can I expect after my baby is born?**

Most babies do very well right after birth. If the baby does not have any other medical problems, like Down syndrome, the baby should be able to stay with you after birth. An echocardiogram will be done to confirm the diagnosis. The baby would be discharged with you and come back for follow-up appointments. If there are other issues, or if there are any problems after birth, the baby will go to the neonatal intensive care unit. The baby would be stabilized and an echocardiogram would be done. The care that the baby needed would depend on the other problems.

### **What is the treatment/surgery for AVSDs?**

Babies with this heart defect may start to have symptoms of heart failure (due to the extra work the heart is doing) when they are 1-2 months old. There are medications that can help to decrease these symptoms. All babies with AVSDs will require surgery. The timing of the surgery depends on the symptoms that the baby develops and whether or not the medications help the symptoms. Almost all babies will have their surgery by 6 months of age, although some may need it sooner.

The surgeon will repair the AVSD by closing both holes with a patch and dividing the common AV valve into two separate parts. Babies are usually in the hospital for 5 to 10 days after surgery.

### **What other procedures or follow up will my baby need?**

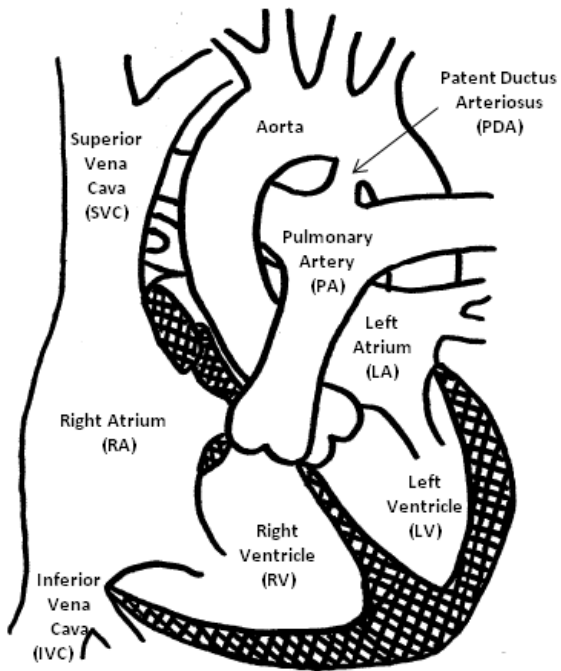
The surgery is very effective at closing the holes and dividing the valve into two. However, the valve can never be made 100% normal. Patients with AVSDs need lifelong follow-up with a cardiologist. The surgeon tries to repair the valves so that they don't leak, but they still open easily. Despite the best results, the valve may have leakage (regurgitation) or obstruction (stenosis). A mild amount of regurgitation or stenosis does not usually cause problems, but if these become more severe, some children need another surgery to repair or replace the valve. Around 10% of patients will need a repeat surgery on the valve.

### **What is the long term prognosis for AVSDs?**

The long term outlook for AVSD is very good. The survival from the surgery is very high. Children are usually able to participate in normal activities, including sports. The patients with AVSD who develop valve problems sometimes need medications.

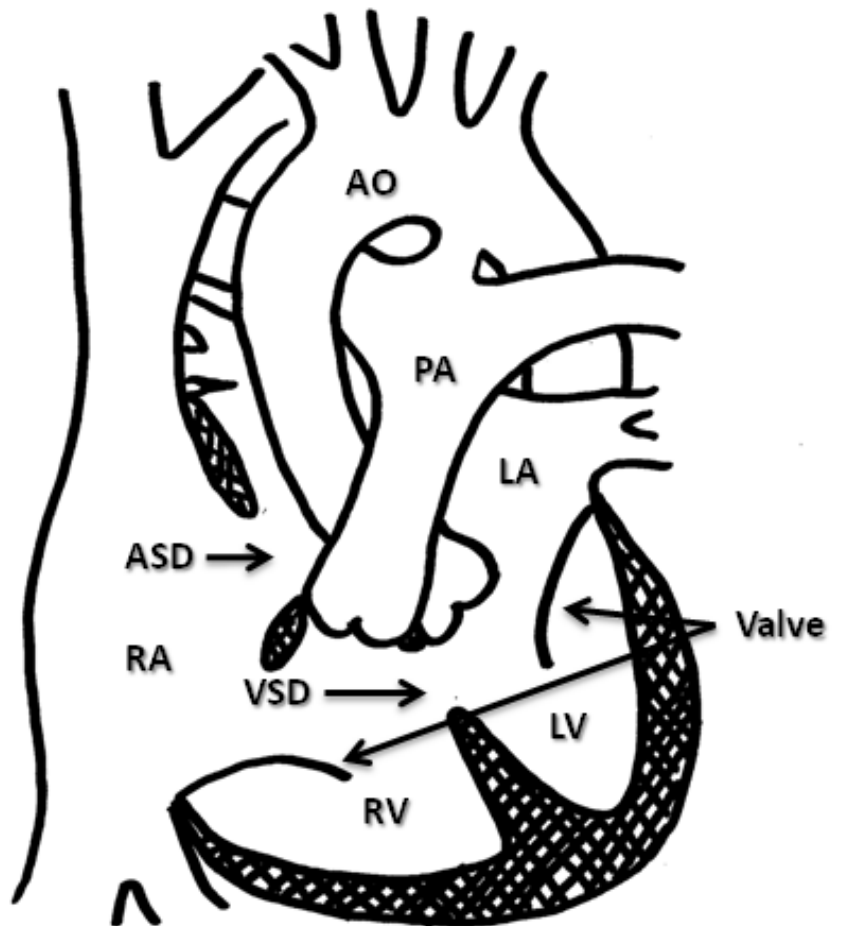
### **If I have had one baby with an AVSD, are my future pregnancies more likely to have an AVSD?**

If your baby's AVSD is related to a chromosome abnormality (like Down syndrome) or a genetic syndrome, a genetic counselor can tell you what the chances are that a future pregnancy would have the same condition. Studies show that when an AVSD is not associated with a genetic problem, the chance that future children will have any heart defect is about 2.5%. In future pregnancies, nuchal translucency ultrasound (at the end of the first trimester), targeted anatomy ultrasound (between 18-20 weeks) and fetal echocardiography are recommended.

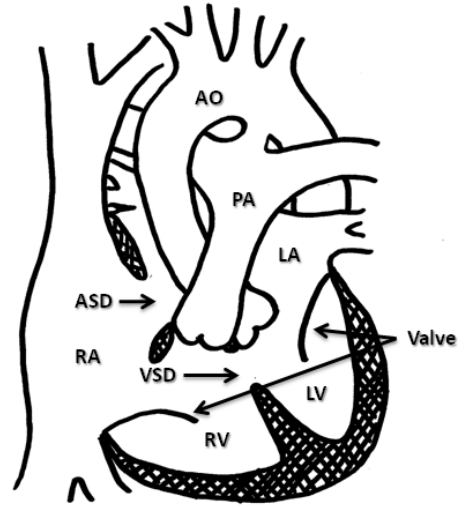


## Normal Newborn Heart

## Atrioventricular Septal Defect (AVSD)



## Before Surgery



## After surgery

