

### What is Shone syndrome?

Shone syndrome is a collection of eight left-sided obstructive heart lesions. These affect blood flow to and from the left ventricle, or lower left heart chamber.

Shone syndrome was identified by Dr. John Shone in 1953. He described four lesions. Now, eight lesions are considered part of Shone syndrome. A person must have at least three of these lesions to be diagnosed. Of the eight lesions, supra mitral valve, parachute mitral valve, subaortic stenosis, and coarctation of the aorta were the first four described.

Because so many different defects are involved, individuals present in varying ways, with a wide range of combinations of defects, symptoms, and issues. The number of lesions does not necessarily determine the severity of the disease.

### What are the eight lesions in Shone syndrome?

**Cor Triatriatum.** Cor triatriatum occurs in less than one percent of all congenital heart defects (CHD). It occurs when the pulmonary veins, which drain blood from the lungs into the left side of the heart, fail to connect normally to the left atrium. As a result, there is a membrane in the left atrium. It divides the left upper heart chamber into two parts and obstructs blood flow into the left ventricle. The severity of this obstruction varies widely. The more severe the obstruction, the earlier it may present. On the other hand, those with milder obstruction may not present until late adulthood.

In children with severe obstruction, the membrane may prevent enough blood from getting into the left side of the heart. This can lead to fluid backing up in the lungs. This is known as pulmonary edema. If the obstruction is less severe, it is still important. Over time, high blood pressure or pulmonary arterial hypertension can develop in the lungs. Patients with cor triatriatum can also develop a fast heart beat or arrhythmia.

Mild forms of cor triatriatum cause few or no symptoms in early life. For these patients, the problems often occur later in life. They include arrhythmias, blood clots, pulmonary edema, and pulmonary hypertension.

Surgery to remove the membrane is generally successful. Recurrence is rare.

**Supramitral Ring.** The mitral valve connects the left atrium and the left ventricle. Mitral valve disease happens when this valve is either too narrow or becomes leaky. Valves that are too narrow obstruct the flow of blood. Valves that leak allow blood to flow back into the left atrium. Mitral valve disease is rare, accounting for less than one percent of CHD.

A supramitral ring is a fibrous membrane that surrounds and rests on top of the annulus or base of the valve. The membrane looks like an orange peel—thick and fibrous. It can be peeled off of the annulus. This narrows the opening of the valve and results in obstruction.

It has a variable presentation, ranging from mild to severe. Surgical resection is the treatment of choice and it is generally successful. The recurrence rate is fairly high, especially in young children. For this reason, surgery in children is not recommended unless it is causing problems. Catheter based techniques (balloon dilation) are sometimes temporarily successful. However, the obstruction usually returns.

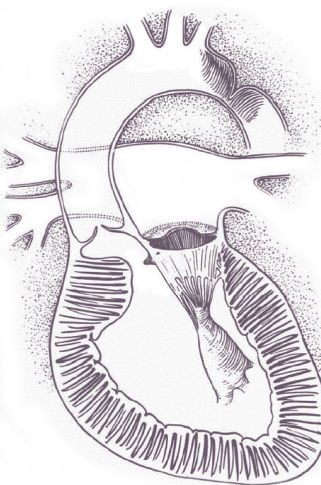
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**Parachute mitral valve.** In a normal heart, fibrous strands, called chordae are attached to the papillary muscles around the mitral valve. They help the valve open and close. In a heart with a parachute mitral valve, there is only a single papillary muscle. The way it attaches to the valve looks like a parachute. A parachute mitral valve often does not cause problems. Infrequently, it can cause stenosis (narrowing) or regurgitation (leaking) of the valve.

**Subaortic stenosis.** Subaortic stenosis is a common defect in patients with Shone syndrome. It occurs when a membrane or muscular thickening forms under the aortic valve. When this happens, it causes narrowing or obstruction of the left ventricular outflow tract. The left ventricle then has to work harder to pump blood out of the chamber and into the aorta.



Subaortic stenosis tends to cause symptoms earlier than aortic valve stenosis. Earlier treatment may help save the aortic valve and thus avoid valve replacement.

Image courtesy of Shone, JD, Sellers, RD, Anderson, RC, Adams, P, Lillehei, CW, Edwards, J. The developmental complex of "parachute mitral valve," supra-valvular mitral ring of left atrium, subaortic stenosis and coarctation of aorta. American Journal of Cardiology. 1963;11:714.

The treatment of choice is surgery. It cannot be repaired in the cath lab.

In children, the membrane often regrows, sometimes rapidly. This is less common in adults. Subaortic stenosis in adults can remain stable for many years.

For a more detailed description of subaortic stenosis, please refer to Adult Congenital Heart Association's (ACHA) Q&A on Subaortic stenosis.

### **Bicuspid Aortic Valve and Small Aortic Valve Annulus.**

Bicuspid aortic valve and small aortic valve annulus often occur together in Shone Syndrome. A normal aortic valve is tricuspid. This means it has three leaflets or moving parts. A bicuspid valve has only two leaflets. The leaflet motion is restricted and the opening of the valve is small as well. Together these cause an obstruction in the flow of blood through the valve. Patients with this defect may require either valve repair or replacement. A Konno procedure is often done at the same time to open up or enlarge the valve.

**Coarctation of the Aorta.** Coarctation of the aorta is a narrowing of the aorta at the level of the ductus arteriosus. The ductus arteriosus is a blood vessel found in the fetus that allows blood to bypass the lungs. The ductus arteriosus is oxygen sensitive. At or shortly after birth, it closes. In coarctation of the aorta, the ductal tissue surrounds the aorta. As it closes, cinches the aorta like a noose.

The presentation is variable. If found at birth, it is generally more severe. If the infant is not getting adequate blood to the lower extremities, immediate surgical repair is needed. This is the treatment of choice in the infant and young child. Adults diagnosed later in life often present with a milder form. Often these patients will present with high blood pressure that is difficult to control and diagnostic findings on examination.

In teens and adults with coarctation of the aorta, balloon angioplasty and the placement of a stent is the treatment of choice. This is also true in adults who have a re-narrowing of the aorta after initial repair. The balloon opens the narrowing and the stent keeps the aorta open. Success rates for this procedure are very high.

It is not uncommon for people who had coarctation of the aorta repaired as infants to have residual narrowing as an adult. It is not entirely clear whether this is a regrowth of the obstruction or tissue that was not removed at the original surgery.

In some patients, aneurysms (weakened blood vessels) can form at the site of repair. Another complication is chronic and difficult to control high blood pressure. After repair, even if your blood pressure is normal at rest, it can still be elevated with exercise. Your doctor may want to check your blood pressure on the treadmill. Anyone with chronic high blood pressure who had a repair should be evaluated to see if the problem has returned. The lesion probably increases the risk for early coronary artery disease.

For a more detailed description of coarctation of the aorta, please refer to ACHA's Q&A on coarctation of the aorta.

**Hypoplastic (stiff) left ventricle.** Hypoplastic (small) left ventricles are often stiff. They are often missed, or underappreciated, in patients with left sided heart lesions. Left ventricular stiffness is one of the biggest causes of morbidity in Shone syndrome and is not easily surgically managed.

A stiff ventricle is able to pump blood well. However, when it relaxes to fill with blood for the next beat, it sits at a higher filling pressure than normal. This can cause fluid to back up into the lungs (pulmonary edema), pulmonary arterial hypertension (high blood pressure in the lungs), and fast heart beats (arrhythmias).

There is no surgery to fix this problem, rather it is treated with medication. Such medications include diuretics and ACE inhibitors. Diuretics help remove fluid from the body. ACE inhibitors relax the muscles and take pressure off the heart. It is very important to monitor these conditions closely as they can lead to a number of long term problems. Your ACHD doctor will want to monitor this carefully.

**Small Aortic Arch.** The aorta is the main blood vessel that carries blood from the heart to the body's organs and tissues. A small or hypoplastic aorta can be found in people with coarctation of the aorta. People with Shone Syndrome can also have a small or hypoplastic aortic arch. This results in chronic high blood pressure. This means that the left ventricle pumps at a higher resistance.

### **Can women with Shone syndrome have children?**

If you have Shone syndrome, it is very important that you talk to your ACHD heart doctor before you get pregnant. There are many factors to consider to determine if pregnancy would be safe for you.

### **What kind of test will my doctor do during my visit?**

Regular imaging is a key part of your follow up. This includes:

- Echocardiograms every 1-2 years
- Cardiac MRI or CT every 3-5 years if you have ever had surgery or a procedure on your aorta
- Regular exercise testing on the treadmill to look at exercise capacity and blood pressure response to exercise
- Cardiac Catheterization to evaluate severity of the lesions.

You should talk to your adult congenital heart specialist about what kind of testing is right for you and your condition.

### **What kind of heart care is recommended for adults with Shone syndrome?**

Experts recommend that anyone with Shone syndrome have regular follow up in an adult congenital heart program. Care is highly individualized. Your ACHD heart doctor will recommend what type of tests need to be done and how often you need to be seen.

For her work on this article, ACHA thanks Alison Meadows, MD, PhD, Director, ACHD Program, Northern California Kaiser Permanente.