

TETRALOGY OF FALLOT

What is tetralogy of Fallot?

Tetralogy of Fallot (TOF) is a congenital heart defect. TOF is described as 4 abnormalities that occur together. One is that there is a large hole in the wall of muscle (septum) that divides the 2 bottom pumping chambers (ventricles) of the heart. That hole is called a ventricular septal defect (VSD). Because of the location of the VSD, two other abnormalities occur. One is that the large blood vessel which takes the blood to the body (aorta) is pulled forward, and “overrides” the ventricular septum so that it sits over both the left and right ventricles; this is called an overriding aorta. The other abnormality that happens is that the abnormally placed muscle causes obstruction to the blood flowing out of the right ventricle and across the pulmonary valve. The right ventricular outflow tract obstruction, can be due to obstruction below the valve, at the valve or in the pulmonary arteries as they deliver blood out to the lungs. The most extreme form of TOF is referred to as pulmonary atresia, because in those patients there is no functional pulmonary valve. The right ventricular outflow obstruction leads to thickening of the right ventricle, or right ventricular hypertrophy.

What causes TOF?

The exact cause of TOF is unknown. There are some genetic syndromes that can be seen with TOF, like Down syndrome or DiGeorge syndrome. However, most people with TOF do not have genetic syndromes. All babies with TOF will be checked before or after birth for genetic syndromes. TOF does not typically run in families, but congenital heart defects in general are slightly more common if there is a close relative with any kind of congenital heart defect.

How is TOF diagnosed?

TOF is usually diagnosed with an echocardiogram, an ultrasound of the heart. First trimester screening for chromosomal abnormalities is a good screening tool to identify patients who might be at an increased risk for cardiac defects. Severe TOF can be diagnosed as early as 12 weeks gestation. In general, targeted ultrasound at 18 -23 weeks is the best time to clearly identify TOF. In such cases an assessment of blood flow to the lungs (through the pulmonary artery) is an important part of the examination. Milder cases of TOF may not be diagnosed before birth. TOF may be diagnosed after birth due to a murmur (abnormal heart sound) or a decreased oxygen level.

How will your pregnancy be managed?

If your baby is diagnosed with TOF, a high-risk obstetrician 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 non-stress 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 high-risk obstetrician will participate in your obstetric care delivery is often possible. Induction of labor is often scheduled for pregnancies affected with single ventricle heart defects to make sure that all of the team members are available at the time of delivery.

Why does TOF make babies sick?

Before birth, babies are not generally affected by this heart defect. Because babies don't use their lungs to get oxygen before they are born, they don't need to send much blood to their lungs. After birth, the symptoms that the baby has will depend on how much obstruction there is to getting blood to the lungs. Since there is a large VSD, blood from the right and left ventricles can go to either side of the heart. Normally, there is higher pressure on the left side of the heart than on the right. This means that some of the blood will flow from the left ventricle, through the VSD and out to the lungs. In babies with TOF, the obstruction in the right ventricular outflow tract limits how much blood flows to the lungs. Over time, the obstruction gets worse. Once the obstruction becomes severe, the pressure will become higher on the right side than the left. At that point, the blood will flow through the VSD from the right ventricle to the left ventricle. Since the blood in the right ventricle is lower in oxygen, this will lead to lower oxygen levels in the body. When the oxygen level is lower, the baby may appear blue.

The worsening obstruction to blood flow out to the lungs is a gradual process and is faster in some babies than in others. Any baby with TOF can turn blue for short periods of time, especially when they are crying or pooping. Most of these blue spells last just a few minutes, but sometimes these spells are more severe and do not go away. This is rare, but may be an emergency.

What can I expect after my baby is born?

Most babies do very well right after birth. The amount of obstruction to blood flow to the lungs determines how babies do. Some are very blue right after they are born, and some may take a long time to become blue. The baby will need to go to the NICU for evaluation. An echocardiogram will be done shortly after birth. If the oxygen level is normal and the baby is breathing well, the baby will simply be monitored for a few days, and then be able to go home. If the oxygen level is low, the baby may need to get a medication called prostaglandins. This medication keeps a blood vessel called the patent ductus arteriosus (PDA) open. The PDA is a vessel that connects the pulmonary artery and aorta. It is present in all babies before birth and closes within the first few days of life. It can be used to supply extra blood to the lungs. If the baby needs the prostaglandins to keep the PDA open, the baby will require surgery before going home. Babies who have the most severe form of TOF with pulmonary atresia, will need prostaglandins and will have surgery before going home.

Because the care for children with congenital heart disease is complex, your baby will be cared for by a team of skilled clinicians. This starts before birth with maternal-fetal medicine and fetal cardiologists and continues with pediatric cardiologists and nurse practitioners, a pediatric cardiac surgeon, neonatal and pediatric intensive care physicians, pediatric anesthesiologists, pediatric cardiac operating room staff, pediatric nurses and many others.

What is the treatment/surgery for TOF?

The initial treatment and timing of surgery depend on how much blood flow there is to the lungs. Babies who don't have enough blood flow to their lungs right after birth will be placed on prostaglandins. After a few days they may need to have a surgery for placement of a shunt. A shunt is a small tube that connects the aorta to the pulmonary artery. It acts much like the PDA, and it provides extra blood flow to the lungs. Although it is not a final solution, it will give the baby time to grow and mature before a bigger surgery.

The surgery for TOF is usually done around 4-6 months, although it may happen earlier if the baby has low oxygen levels or frequent blue spells. During the surgery, the surgeon removes the obstruction to blood flow to the lungs. This is done by cutting out extra muscle tissue, enlarging narrow areas with a patch, and making the pulmonary valve open more easily. The VSD is also closed with a patch. In some cases where there is pulmonary atresia, or severe obstruction, the surgeon may not be able to remove the obstruction in the usual way. Instead, these patients will have a conduit placed. This is a tube that will bypass the pulmonary valve and connect the right ventricle to the pulmonary artery. Babies are typically in the hospital or about 5-10 days after the surgery.

What other procedures or follow up will my baby need?

Although the surgery will close the VSD and remove most of the obstruction to blood flow to the lungs, it does not make the heart 100% normal. Some people with TOF only need one surgery for their entire life, but there are complications that we will need to watch for throughout the child's life. Patients who have a conduit placed will develop obstruction to blood flow over time, since the child will grow but the conduit stays the same size. Those conduits need to be replaced periodically, usually a few times during childhood. When the surgeons remove the obstruction to blood flow to the lungs, they often have to sacrifice the pulmonary valve, which means that it will open, but not close well. This causes the valve to leak. Although this leakage does not cause immediate problems, over time it causes the right ventricle to enlarge and not squeeze as well. At some point, patients who have valve leakage will need a replacement valve.

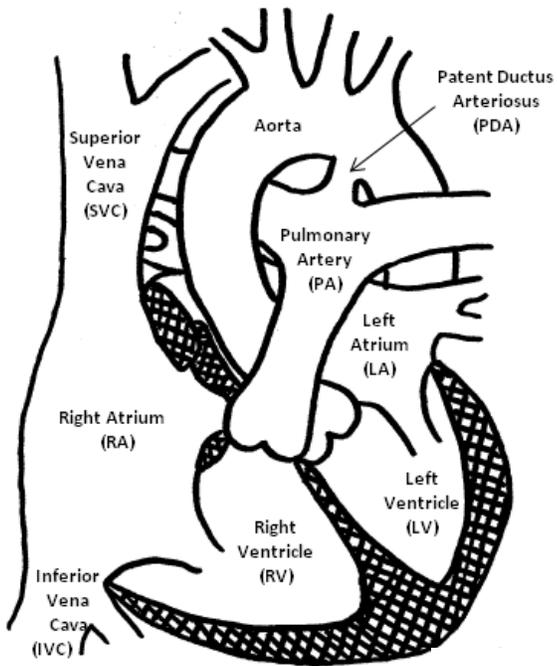
After surgery, there will be frequent visits with your cardiologist. After the first year, the appointments will become less frequent, eventually only yearly. At the follow-up visits, echocardiograms, electrocardiograms (EKGs-looking at the electrical activity of the heart), and other types of monitoring will be done. As children get older, they may have exercise stress tests, 24-hour heart monitors or cardiac MRIs.

What is the long term prognosis for TOF?

The long term outlook for TOF is very good. The survival from the surgery is very high. Children are able to participate in normal activities, including sports. Some patients with TOF will develop abnormal heart rhythms, sometimes needing medications or pacemakers. Patients may develop symptoms if there is obstruction to blood flow to the lungs or a lot of valve leakage, but in those situations surgery or catheterization procedures can relieve the problems.

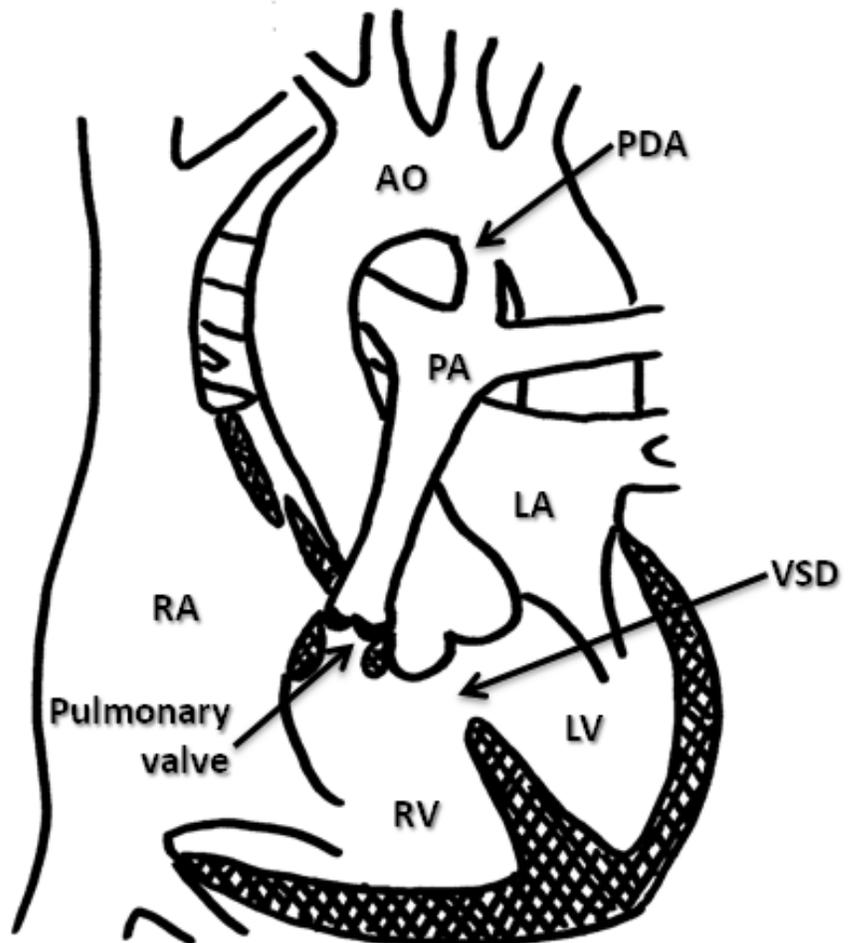
If I have one baby with TOF, are my future children more likely to have TOF?

If your baby's TOF is related to a chromosome abnormality 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 TOF is not associated with an underlying 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.

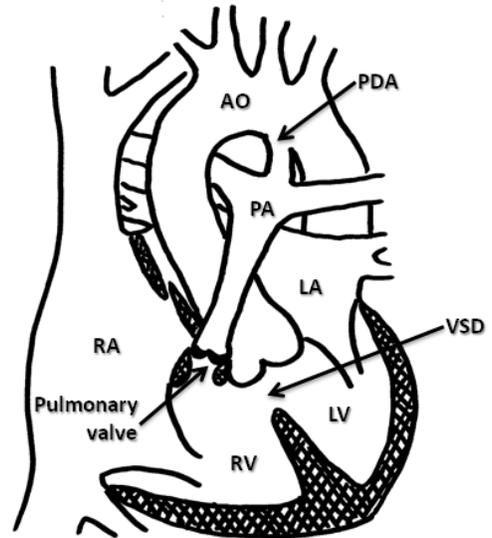


Normal Newborn Heart

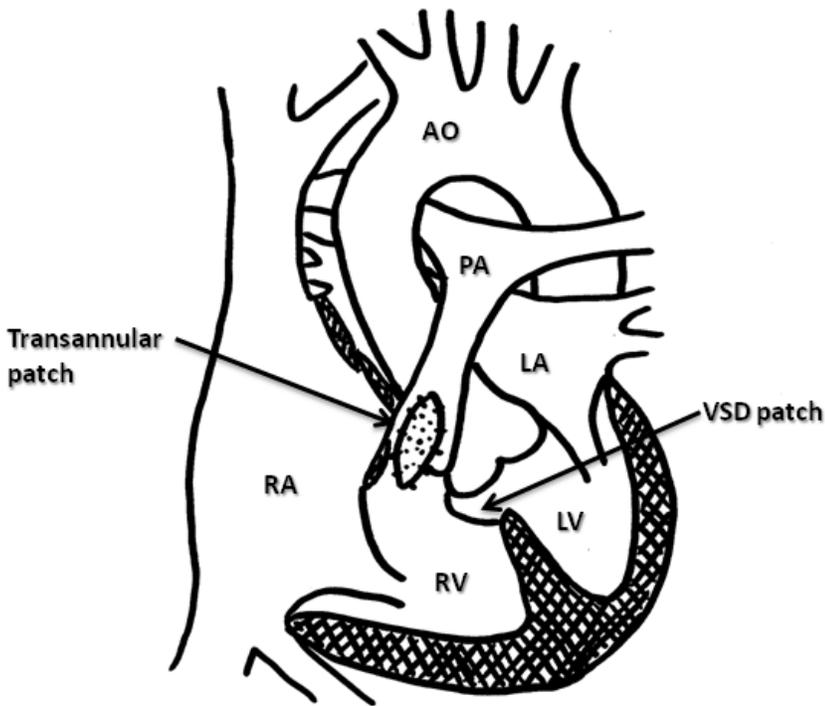
Tetralogy of Fallot (TOF)



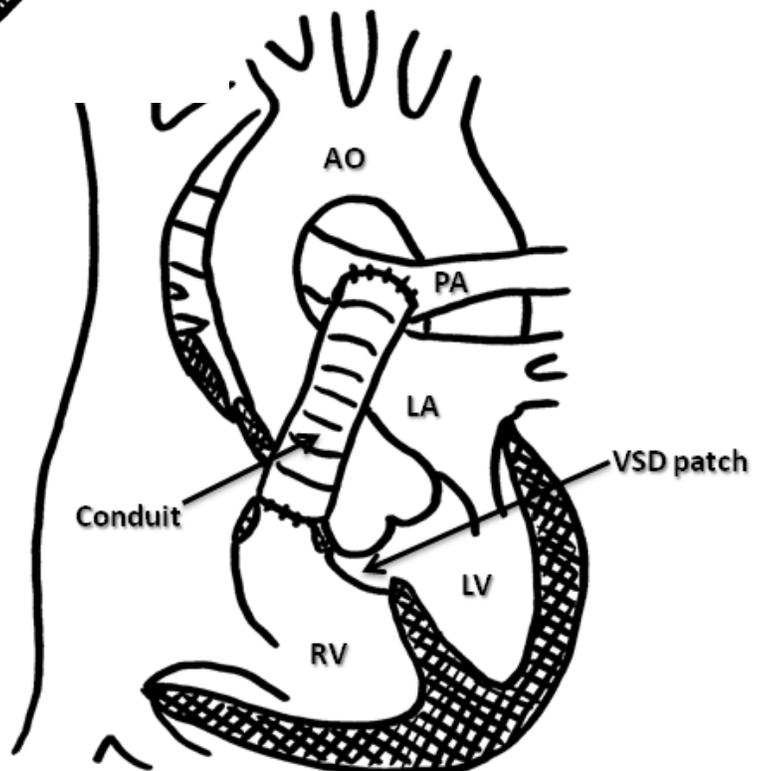
Before surgery



After surgery



Transannular patch repair



Rastelli repair