What is tetralogy of Fallot?
Tetralogy of Fallot (ToF) is a complex heart defect. If you have ToF, you are born with four different heart problems:

- A hole in the wall between your heart’s main pumping chambers (ventricular septal defect or VSD)
- A valve between your heart and lungs that is too narrow (pulmonary stenosis or PS)
- A right heart chamber with walls that are too thick (right ventricular hypertrophy)
- A major blood vessel (aorta) that is misplaced or moved (overriding aorta)

These problems cause the blood entering and leaving your heart to mix. Blood that is low in oxygen (“blue”) mixes with blood that is oxygen-rich (“red”). Babies born with ToF sometimes are called “blue babies” because the lack of oxygen can makes them look blue (cyanotic).

How common is ToF?
ToF is the most common cyanotic (blue) heart defect. About 5 out of every 10,000 babies are born with ToF.

How does ToF occur?
ToF starts very early in pregnancy. The heart starts out as a simple tube. It then divides into two equal sides. In ToF the heart divides unevenly. As a result, the right side is smaller than the left side. This mistake causes all four problems to develop as the heart grows. In 1888, the French physician Étienne-Louis Arthur Fallot recognized that these four heart problems often happened together. He named it “tetralogy of Fallot” after the Latin word “tetra” meaning “four.”

What causes ToF?
We do not know enough about what causes ToF and other heart defects. ACHA actively advocates for more research to help learn more about why ToF and other heart defects happen. Scientists are currently studying what role genes and environment may play in causing ToF. To learn more about CHD research, visit http://1.usa.gov/11v0deJ.

How old are the oldest people living with ToF?
The great news about ToF is that many people born with ToF do very well over time. Many are now in their 60s, 70s and even 80s. The first surgery for ToF, called a Blalock-Taussig shunt (B-T shunt), was done in 1944. This was followed by the development of open heart surgery to repair the defects in 1954. The B-T shunt helps babies born with ToF get enough oxygen to their bodies. Then the baby can have open heart surgery to repair the defects. Many people who had B-T shunts as children are still alive today.

If I had my ToF repaired as a child, am I at risk for new heart problems?
Most adults with ToF had a “complete ToF repair” in childhood. In this repair, surgeons close the hole in the heart and open up the narrow heart valve to the lungs (the pulmonary valve). While most people who undergo this repair feel well and have few problems in childhood, this does not mean that their hearts are fixed permanently. As people born with ToF age, new heart problems can develop. As adults, it’s important that they stay in the care of an adult congenital heart specialist.

What is the most common heart problem in adults with repaired ToF?
The most common problem for adults with ToF is a leaky valve between the heart and lungs (pulmonary regurgitation or PR). As part of the ToF repair, surgeons usually stretch out the valve between the heart and lungs (pulmonary valve). This almost always makes the valve leak. As people age, this leaking can get worse.

RA – Right Atrium
RV – Right Ventricle
LA – Left Atrium
LV – Left Ventricle
SVC – Superior Vena Cava
IVC – Inferior Vena Cava
MPA – Main Pulmonary Artery
Ao – Aorta
TV – Tricuspid Valve
MV – Mitral Valve
PV – Pulmonary Valve
AoV – Aortic Valve

Image courtesy of Centers for Disease Control and Prevention, National Center on Birth Defects and Developmental Disabilities
What health problems can be caused by a leaky valve?
A leaky valve can cause a number of serious problems. A leaky pulmonary valve can cause the right side of your heart to get bigger, which affects its pumping strength. This can lead to a loss of function in your heart and put you into heart failure. A leaky valve also increases your risk of serious rhythm problems, especially in the lower chambers of your heart (ventricular tachycardia or VT). VT is a dangerously fast heartbeat and can cause sudden death if not treated. Other rhythm problems in the upper chambers of the heart are common too. In some people with ToF, the aortic and tricuspid valves may also leak (for more details, see below).

How would I know if I have a problem with a leaky valve?
A leaky valve can be a sneaky valve! There are often few or no symptoms to alert you. The first sign can often be rhythm problems, like an unusually fast heartbeat or feeling your heart “flutter.” If your valve has been leaking a lot and for a long time, you may feel tired. It is important to see your ACHD cardiologist regularly to know how and when to treat a leaky valve.

How do doctors find and treat a leaky valve?
Your ACHD care team has a number of tools to look at your heart. Your doctor may want you to have an echocardiogram (echo), a scan that uses sound waves to look at your heart. You will also have an electrocardiogram (ECG or EKG), a test that records your heart's electrical activity. You may also need to have an exercise stress test and/or a cardiac MRI. Some patients' doctors may recommend having a heart catheterization. Together these tests can help your doctor decide if you need a pulmonary valve replacement (PVR) to replace your leaky pulmonary valve with a new valve.

There are two main kinds of valves—tissue valves and mechanical valves. Tissue valves can come from human cadavers (homograft valves) or from pig or cow heart tissue (heterograft valves). Mechanical valves are not used very often for PVR. A person with a mechanical valve will need to be on medicines to prevent blood clots since there is a higher risk of developing dangerous clots. Each kind of valve has its own pros and cons. Talk with your doctor about what kind of valve is best for you heart. Newer valves, such as the Melody or Edwards SAPIEN valves, can be placed with a catheter going from the leg into the heart. Right now there are strict standards for which patients can have a PVR done by catheter.

If I need a new valve, when should it be replaced?
The right time to replace a pulmonary valve will vary from one person to the next. Your ACHD heart doctor will decide when the right time is based on your symptoms and on the results of heart tests. Since valve replacements do not last forever, your doctor might want to wait as long as possible (without hurting your heart) to replace a valve. About 20%-30% of patients will need to have another PVR within ten years.

What other health problems might I be at risk for?
If you are an adult with repaired ToF, you may be at risk for an enlarged aorta. This can cause your aortic valve to leak and require an aortic root replacement. This is a surgery to replace the stretched-out part of your aorta and/or aortic valve. Less than 10% of people with ToF may have a leaking tricuspid valve, the valve between your right atrium (receiving chamber) and your right ventricle (pumping chamber). This is known as tricuspid regurgitation or TR and can only be repaired through surgery. Other possible health problems include heartbeats that are too fast or too slow (you can learn more in the educational sheet, “The Heart and Its Electrical System,” on the ACHA website). You may also have leaking around the area that closed the ventricular septal defect (VSD), and you may be at risk for lung problems or problems with the heart muscle. Talk to your ACHD heart doctor about what you need to look for. It is important to see your ACHD cardiologist regularly so that any changes in your heart can be detected early. Early detection can help prevent permanent damage.

Can women with ToF have children?
Many women with ToF can have children successfully. If you are a woman with ToF and are thinking about becoming pregnant you should first talk with your ACHD cardiologist. You should also talk with an obstetrician who specializes in higher-risk pregnancies. Your ACHD care team and your obstetrician should work together. Many women can safely go through pregnancy and delivery; however, some women may need to have a valve replacement or adjust medication first. Your doctors can develop a plan to care for and protect your heart during your pregnancy and delivery.

If I have ToF, do I have a higher chance of having a baby with a heart defect or other health problems?
Yes. Most people with ToF will have a baby without any health problems. But people with heart defects do have a slightly higher chance of having a baby with a heart defect or other health problem. One genetic problem called 22q11.2 deletion syndrome has been linked to ToF. If you have ToF and are thinking about having a baby, you should be screened for this syndrome.

Do I need special care for my ToF as an adult?
Yes. The American Heart Association and the American College of Cardiology classify ToF as moderately complex heart disease. This means that you should get your care at a special ACHD center and see your ACHD cardiologist regularly, at least every two years. Regular check-ups can help detect any problems early on and prevent long-term damage to the heart.

ACHA partnered with Naser Ammash, MD, on this document. Dr. Ammash is a professor of medicine at the College of Medicine at several Mayo Clinic sites. He is an expert specializing in the care of adults with congenital heart disease.