

Tetralogy of Fallot

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

What is a Tetralogy of Fallot?

Tetralogy of Fallot is a type of congenital heart defect. It gets its name from the fact that it is comprised of four (tetra- from the Greek word for four) anatomical defects in the heart occurring together. It was described by the French physician Étienne-Louis Arthur Fallot, after whom it is named. The four parts of tetralogy of Fallot (TOF) are: Pulmonary Stenosis, Ventricular Septal Defect (VSD), Overriding Aorta, and Right Ventricle Hypertrophy. It occurs in about 1 in 2500 to 1 in 3000 births.

The heart is made up of four chambers, two upper *atria* (right atrium and left atrium) and two lower, the right and left ventricle. The ventricles pump blood outward through two Great Vessels, the pulmonary artery from the right ventricle and the aorta from the left. This blood flow is modulated by two valves: the pulmonary valve and the aortic valve. In pulmonary stenosis, the pulmonary valve or the tissue just before it do not develop properly, such that blood cannot flow freely when the right ventricle contracts to pump blood toward the lungs. The opening is narrow and the valve leaflets may be thickened and stiff. The nature and degree of narrowing is highly variable, affecting how much blood is pumped to the lungs.

The right and left ventricles are separated by a septum, a wall of muscle. A ventricular septal defect (VSD) is a hole in this wall. VSD's are quite common, in fact they are the most common heart defect. In TOF, the aortic valve straddles (“overrides”) the VSD, allowing blood from both the right and left ventricles to flow outward through the “overriding aorta”. The degree of straddling, or how much of the VSD is covered by the aortic valve, varies among individuals with TOF. Right ventricle hypertrophy is thickening of the muscles of the right ventricle. This appears to develop as a response of the heart to the effects of the other defects, since it tends to progress over time.

How does a tetralogy of Fallot happen?

There may be a genetic component in TOF. It may be associated with chromosome 22 deletions and DiGeorge syndrome or with Trisomy 21 (Down syndrome). In many cases, the cause is not known.

How are chromosomes relevant to a TOF?

Chromosomes are where most of our genetic information is kept. We usually have 46 of them matched in pairs: 23 come from one parent and the other 23 come from the other parent. For example, people with Down syndrome have an extra chromosome number 21. People with DiGeorge syndrome have a change in the information in a fragment of chromosome number 22. Both of those conditions are seen in babies with a TOF: if such a change in information within the chromosomes occurs, it would be the cause for the TOF.

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Should I have more tests done?

Your caregiver may refer you to genetic counseling and genetic testing or discuss this with you, such as amniocentesis or chorionic villus sampling (CVS) with chromosomal microarray or whole exome sequencing, which can help determine if the TOF is part of a genetic syndrome.. This can provide information on the chromosomes and can provide crucial information about your baby, beyond his/her anatomy. If not already performed, a fetal echocardiography, a special ultrasound focusing on the heart and cardiovascular system of the baby, should be done to better understand the anatomy and function of the heart. A specialist such as a maternal-fetal medicine specialist or pediatric cardiologist usually performs fetal echocardiography. This can provide images of your baby's heart and blood vessels and inform your caregivers how well the cardiovascular system is working.

What are the things to watch for during the pregnancy?

Your caregiver will probably order serial ultrasound scans, to monitor your baby's growth and well-being, and to watch for any signs of distress. You may be asked to track your baby's movements throughout the day: your caregiver will advise you as the pregnancy progresses.

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

TOF can be repaired by open heart surgery. Success rates of these repair procedures have increased dramatically since their introduction in the 1950's and are now around 95%. Many individuals born with TOF have undergone surgical repair and grown to adulthood; many now have children of their own. Every fetus born with TOF is unique. The timing of repair will depend on your individual child's case, particularly on if oxygen poor blood is being carried from the lungs to the rest of the body. Also, the degree of stenosis, or narrowing, of the pulmonary outflow tract from the right ventricle to the lungs will affect how much blood is delivered to the lungs to receive oxygen. This can impact the baby's symptoms after delivery. The goal of surgery is to close the VSD so that only oxygenated blood from the left ventricle is pumped through the aorta to the body and brain, and to widen the pulmonary outflow tract so blood flows freely from the right ventricle to the pulmonary artery and from there to the lungs. Your baby's blood oxygen levels will be monitored and he/she will likely have additional ultrasound scanning and other imaging tests, such as chest X-ray, as required after delivery. The team of pediatric cardiologists and pediatric cardiothoracic surgeons will study your baby's anatomy to devise the optimal management and surgical plan.

Will it happen again?

It appears there is a genetic component in TOF and that siblings of an affected child are at a higher risk than the general population. If a genetic cause is found, the risk of this happening again will be dependent on the kind of genetic problem. Some genetic problems have a rarer chance of recurring whereas others can have a 1 in 2 chance of happening again. If there is no genetic cause, the risk of this happening again will be less than 1 in 10. Your caregiver will probably refer you to early fetal ultrasound scanning to rule out congenital heart disease, in your future pregnancies.

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What other questions should I ask?

- Does this look like a severe TOF?
- How often will I have ultrasound examinations done?
- Where should I deliver?
- Where will the baby receive the best care after it is born?
- Can I meet the team of doctors that will be assisting my baby when it is born, in advance of my delivery?
- Is there a support group in our area that we can talk to?

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