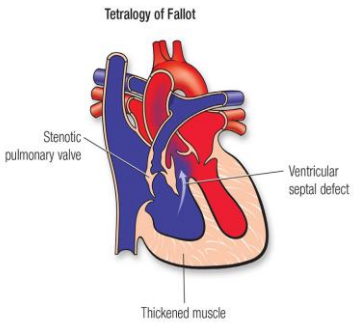
Tetralogy of Fallot (ToF)

Tetralogy of Fallot (ToF) is a **congenital heart defect** characterized by **4 features**:

1. **Malaligned ventricular septal defect**—a hole in the septum between the left and right lower pumping chambers (ventricles) of the heart.
2. **Pulmonary stenosis**—a narrowing of the pulmonary valve (which regulates the blood flow from the heart to the lungs) and the main pulmonary artery. Blood cannot be pumped easily to the lungs to receive more oxygen.
3. **Overriding aorta**—the aorta is positioned directly over the VSD and so gets the blood from both lower chambers instead of just the left one. Oxygen-poor and oxygen-rich blood gets mixed and pumped to the rest of the body.
4. **Right ventricular hypertrophy**—the muscle on the wall of the lower right chamber becomes thickened.

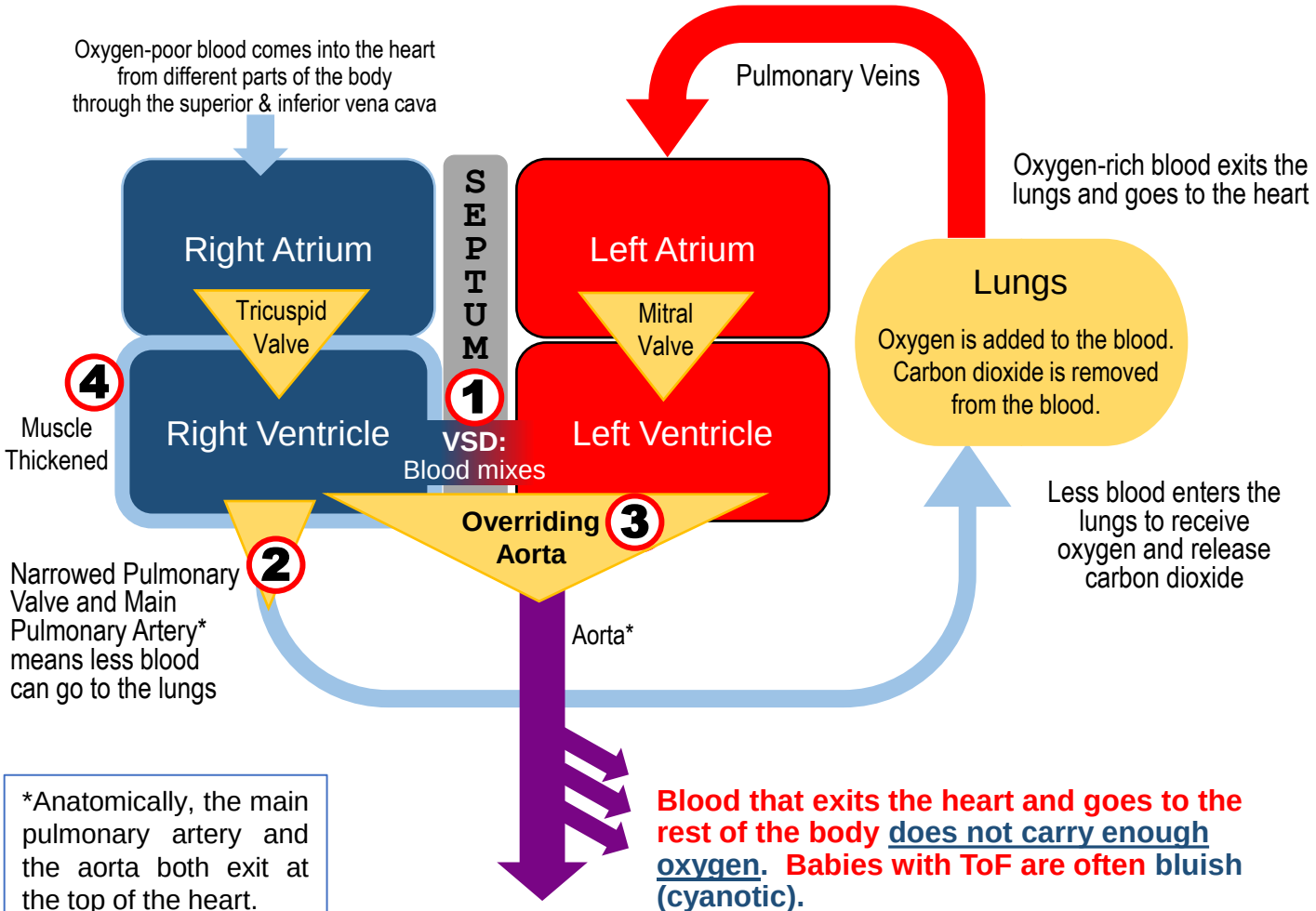


Please click on the image to see a larger version on the website of the American Heart Association

A Schematic Diagram of the Heart with Tetralogy of Fallot (ToF)

Shown as if you are facing the patient; Not to scale

Please also see our schematic diagram of The Heart and Normal Blood Flow for comparison.



*Anatomically, the main pulmonary artery and the aorta both exit at the top of the heart.

Blood that exits the heart and goes to the rest of the body does not carry enough oxygen. Babies with ToF are often bluish (cyanotic).

Tetralogy of Fallot (continued)

Tetralogy of Fallot (ToF) and Individuals with 22q Differences

- Of all patients with ToF, 10 to 15% have 22q11.2 deletion syndrome (22q11.2DS).
- 20 to 45% of children with 22q11.2DS are born with ToF.
  - In patients with 22q11.2DS, ToF frequently happen together with other specific heart defects.
- In a study, 2 out of 85 children with 22q11.2 duplication syndrome (22q11.2DupS) were born with ToF.
- **Pulmonary Atresia + VSD (PA + VSD)** is considered the extreme spectrum of Tetralogy of Fallot, in which the pulmonary valve does not form properly, and blood cannot go from the right ventricle of the heart to the lungs to receive more oxygen.
- **PA + VSD** occur in 10 to 25% of children with 22q11.2DS.

Diagnosis for ToF

- During pregnancy, ToF can be diagnosed using **ultrasound** and **fetal echocardiogram**.
- After birth, babies with ToF can look **bluish** when crying or feeding and have a heart murmur.
- Symptoms depend on the degree of right ventricular outflow tract obstruction (RVOTO) and the amount of shunting through the VSD.

Surgeries for ToF

1. A **temporary operation** (NOT an open-heart surgery) is usually performed during neonatal period, before the surgical complete repair.
   - A shunt (a small synthetic tube) is sewn between the aorta and the pulmonary artery to increase blood flow to the lungs. This shunt is closed when complete repair is done later.
2. A **complete repair** is usually performed during the first months of life.
   - The ventricular septal defect is closed with a patch.
   - Some of the thickened muscle below the pulmonary valve is removed. The pulmonary valve is repaired or removed, if needed.
   - Reconstruction of the right ventricular outflow tract is done with a patch.
   - In severe forms of ToF or in PA + VSD, a valved conduit is used to connect the right ventricle to the pulmonary arteries.

Ongoing care for ToF

1. Regular **follow-up with a cardiologist** is necessary. Ideally, the cardiologist should have special training in congenital heart defects.
2. The cardiologist may recommend **tests** for ongoing monitoring.
3. In some cases, **medications** and/or more **surgeries** are needed.
4. Even after surgeries, there may still be some obstruction or leak. Some people may need to **limit their activities**. Please discuss exercise tolerance with your cardiologist.
5. Some people with repaired ToF may have **disturbances to heart rhythms**, which may cause dizziness or fainting. The rhythm problems can potentially be managed using medications, surgeries, or a pacemaker if necessary.
6. Some people may need **endocarditis prophylaxis** – This means taking antibiotics before certain dental procedures to prevent bacteria from causing infections in the heart.
7. Pregnancy is not recommended for women who have unrepaired ToF. Women who have repaired ToF and are considering **getting pregnant should discuss the risks** with their healthcare providers.

For more info, please visit the websites of the CDC or the American Heart Association.

International 22q11.2 Foundation

The mission of the International 22q11.2 Foundation is to improve the quality of life for individuals affected by chromosome 22q11.2 differences through family and professional partnerships.
This information is brought to you by the Foundation for educational purposes only. It is not intended to be taken as medical advice. If you have concerns, please talk to your healthcare provider.

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