

The Adolescent and Adult Congenital Heart Disease Program

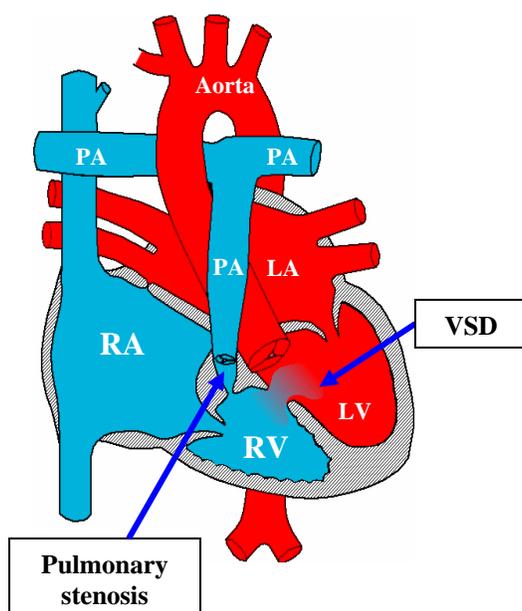
The Heart Center at Nationwide Children's Hospital
& The Ohio State University

Tetralogy of Fallot (TOF)

Tetralogy of Fallot is a congenital heart defect that is made up of 4 problems and results in not enough blood flow to the lungs:

- **Ventricular septal defect (VSD):** A hole between the 2 bottom pumping chambers of the heart (ventricles)
- **Pulmonary stenosis:** Narrowing of the arteries that supply blood to the lungs
- **Overriding of the aorta:** The aorta (major artery that supplies oxygen blood to the body) normally comes off of the left ventricle. In TOF, it sits over both the ventricles and “straddles” the VSD.
- **Right ventricular hypertrophy:** Increased thickness of the walls of the right pumping chamber (ventricle)

In the normal heart, the heart is divided into four chambers, separating the blue (low-oxygen) blood on the right side of the heart from the red (with oxygen) blood on the left side of the heart. In TOF, this combination of heart defects leads to reduced blood flow to the lungs as well as mixing of red and blue blood across a “hole in the heart” or VSD. This allows blue blood to pump out to the body, resulting in low blood oxygen levels. This is seen as blueness of the skin, lips, fingernails and tongue (often called a “blue baby”) and may also result in poor feeding, growth problems, and activity intolerance.



Blue (low-oxygen) blood returns from the body and should be pumped from the right ventricle (RV) to the pulmonary arteries (PAs) and out to the lungs.

Red (oxygenated) blood returns from the lungs and is pumped from the left ventricle (LV) to the aorta and out to the body.

In Tetralogy of Fallot, there is a narrowing of the pulmonary arteries (can be below, at, or above the valve) as well as a ventricular septal defect (VSD). Blue arrows show this narrowing and the VSD, which allows mixing of red and blue blood.

KEY

RA- Right Atrium
RV- Right Ventricle
PA- Pulmonary Artery

LA- Left Atrium
LV- Left Ventricle
Ao - Aorta

The severity of abnormalities in patients with TOF varies. For example, some patients have severe pulmonary narrowing whereas others have mild pulmonary narrowing. The most severe form results in little or no blood flow to the lungs (pulmonary atresia). Patients with pulmonary atresia have pulmonary arteries that are very under-developed and require early surgery in order to survive.

DIAGNOSIS OF TOF

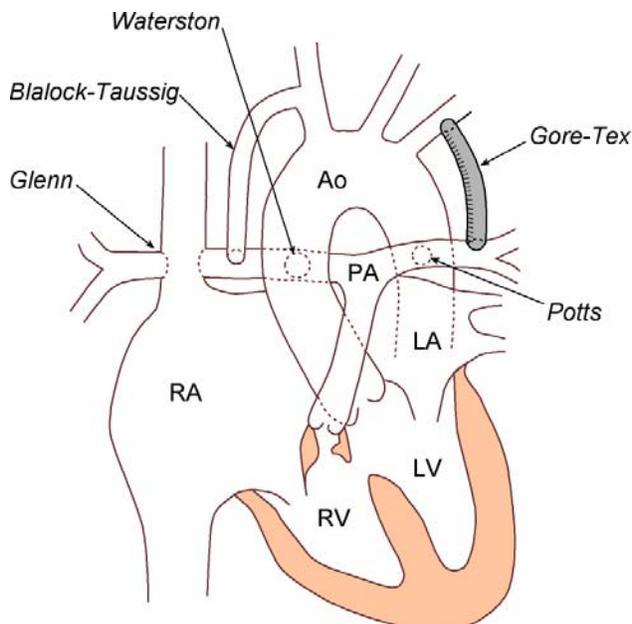
TOF is usually diagnosed in infancy or early childhood, however patients with mild abnormalities (“pink” Tets) may be diagnosed later, as adolescents or adults. Typically, because there is not enough blood flow to the lungs, patients often require surgery early in life. Findings on the history and physical examination prompt expert cardiac evaluation and guide the workup. Tetralogy of Fallot is usually diagnosed by echocardiogram (ultrasound pictures of the heart). An electrocardiogram (EKG) assesses the electrical activity of the heart. Cardiac catheterization angiography (cardiac cath), cardiac MRI (magnetic resonance imaging), and cardiac CT (computerized tomography scan) also identify the typical findings in TOF. These imaging modalities are also useful in identifying long-term complications in TOF patients.

ASSOCIATED DEFECTS

Additional congenital heart problems can occur in patients with TOF. These may include atrial septal defect (ASD), patent foramen ovale (PFO), right sided aortic arch, and coronary artery anomalies. 22q11 deletion is a specific chromosomal disorder that is present in 15% of patients with TOF. 22q11 deletion may also include facial, palate, thymus, calcium regulation and behavior abnormalities.

EARLY TREATMENT

Surgeries are performed to supply more blood to the lungs and are called “palliations”, because they are not typically considered to be a long-term repair. The placement of shunts bypasses the pulmonary narrowing, however the pulmonary stenosis and VSD remain until completely repaired. The use of early shunt procedures provided blood to the lungs by using the higher pressure arterial supply (aorta) to the lower pressure lung arteries. Over time, increased pressure and blood flow to the lungs can result in pulmonary vascular disease and high blood pressure in the lungs (pulmonary hypertension). Many adults underwent Blalock-Taussig, Waterston or Potts shunt palliations in childhood.



A classic Blalock-Taussig shunt (BT shunt) connects the right or left subclavian artery directly to the left or right pulmonary artery.

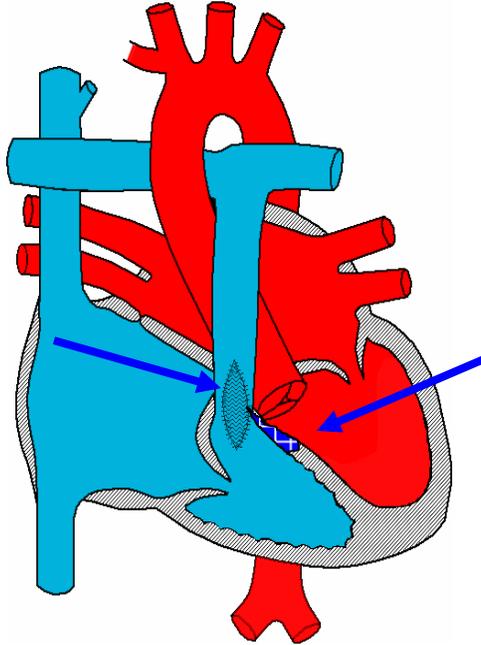
A modified BT shunt consists of a Gore-Tex graft that then connects the subclavian artery to the pulmonary artery. The advantage to a modified BT shunt is having more controlled blood flow.

A Waterston shunt connects the ascending aorta to the main or right pulmonary artery.

The Potts shunt connects the descending aorta to the pulmonary artery.

COMPLETE SURGICAL REPAIR

“Complete” repair consists of closure of the VSD, correction of aortic override, and alleviation of right ventricular outflow tract (RVOT) obstruction. The RVOT refers to the area where blood flows from the right ventricle and out to the main pulmonary artery. If there is narrowing or obstruction of the RVOT, it is resected. Often times, the valve was “torn or stretched open” or removed and a patch was placed across the RVOT or across the valve to alleviate the narrowed area. Patients were often left with a non-functioning pulmonary valve which leaks.



Patch closure of VSD and patch opening of pulmonary stenosis

SURGICAL REPAIRS IN TOF

Pulmonary valvotomy	Opening-up of a stenotic pulmonary valve	Augments blood flow to the lungs
Infundibular resection	Removal of muscular tissue in below the pulmonary valve	Augments blood flow to the lungs
RVOT patch	Patch placed across the RVOT	Augments blood flow to the lungs
Transannular patch	Patch placed across the narrowed pulmonary valve	Disrupts the pulmonary valve; may result in severe pulmonary valve leaking
Pulmonary valve replacement	Human or porcine (pig) tissue valve is used to replace the original pulmonary valve	Used in adolescents and adults to treat pulmonary valve leaking (regurgitation)
Extracardiac conduit	Conduit (or tube) connecting the right ventricle to the pulmonary artery	Treatment of pulmonary atresia
Angioplasty or patch augmentation	Repair of pulmonary arteries	Treatment for hypoplastic (small) main pulmonary artery or stenosis of central pulmonary arteries

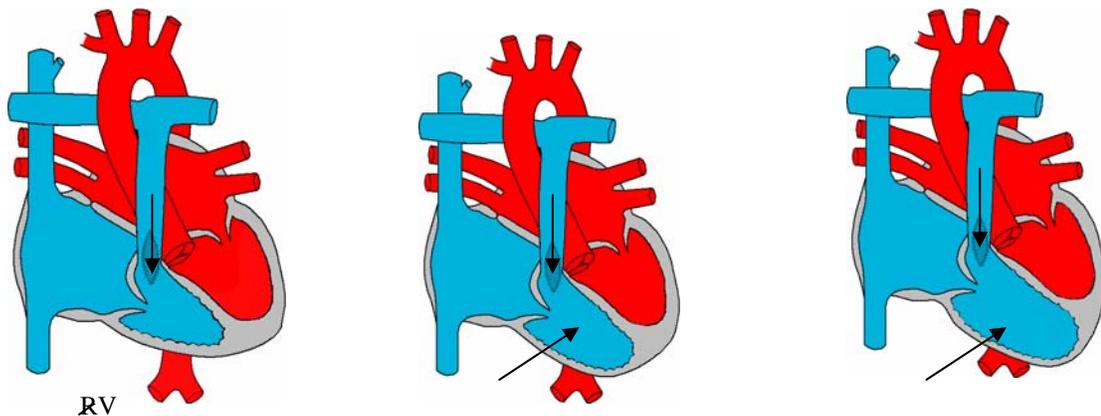
RVOT-right ventricular outflow tract

LONG TERM COMPLICATIONS

Previously a routine technique, making a large incision in the right ventricle (ventriculotomy) and using a large transannular patch to repair TOF, have been abandoned. Extensive incisions, patches, and suturing (stitches) lead to the formation of scar tissue in the right ventricle, which is associated with weakness of heart muscle and heart conduction (electrical) system issues. Currently, the goal of surgical techniques is to minimize the extent incision (or cutting) of right ventricle and to maintain the integrity and competence of the pulmonary valve.

Despite surgical procedures to augment blood flow to the lungs as well as complete repair operation, some patients may develop long-term complications:

- **Right ventricular hypertrophy (RVH):** Obstruction (RVOT or pulmonary stenosis) forces the right ventricle to work harder to pump blood through the narrowed area. Over time, the muscle of the right ventricle may become thick, bulky, and not pump efficiently.
- **Right ventricular enlargement (RVE):** More commonly, RVE results from chronic leaking of blood from the pulmonary valve, back into the right ventricle. This extra blood volume causes enlargement (stretching) of the right ventricle, resulting in a boggy ventricle that does not pump efficiently. This may lead to heart failure, abnormal heart rhythms, and the tricuspid valve leaflets may lose their ability to close properly, resulting in tricuspid valve leaking as well.

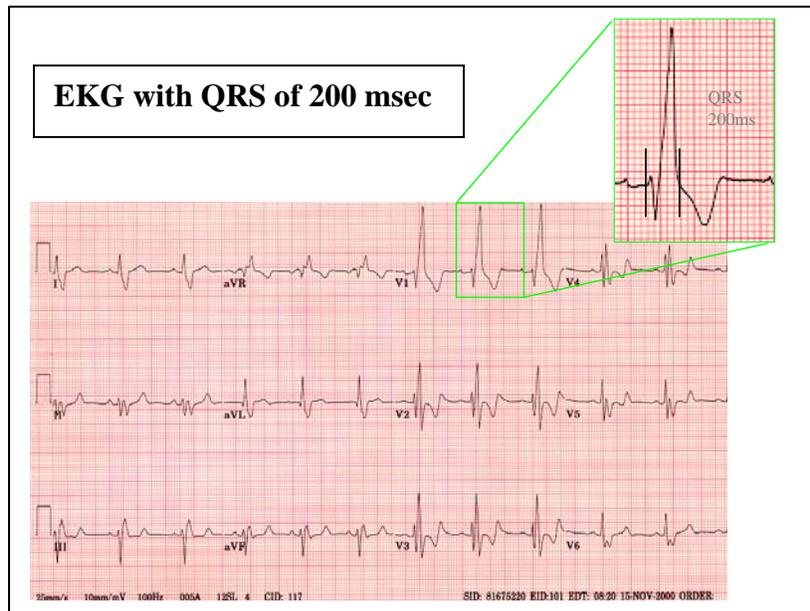


Right Ventricular Enlargement Over Time

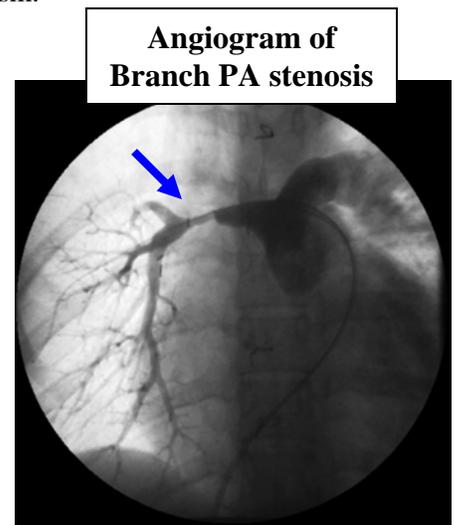
- **Right ventricular failure:** Chronic high-pressure and volume loading on the right ventricle eventually interferes with its ability to pump adequately. Further deterioration in pump function results in right ventricular failure. Symptoms associated with right ventricular failure include exercise intolerance, visible pulsations in the jugular veins, enlargement of the liver (sometimes associated with a feeling full after a small amount of food intake), weight gain due to fluid accumulation, and swelling in the hands, feet or legs.

- Ventricular arrhythmias:** The combination of RVH, RVE, right ventricular dysfunction and previous surgical incisions can all place TOF patients at substantially increased risk of heart rhythm problems. Patients can develop slowing of the heart's conduction (electrical) system abnormalities called right bundle branch block, abnormal heart rhythms from the upper cardiac chambers (atrial arrhythmias) called atrial fibrillation or atrial flutter, or life-threatening heart rhythms from the bottom cardiac chambers (ventricular arrhythmias) called ventricular tachycardia or fibrillation. Therefore, it is very important to routinely monitor for heart rhythm problems. Information from multiple sources indicates that the risk for sudden cardiac death in patients with repaired TOF is 100 times greater than in the general population.

There have been a number of risk factors identified for sudden cardiac death and repaired TOF. An EKG finding of a widened QRS >180 msec that correlates with an enlarged RV has been one of the most common risk factors identified. Therefore, this concept brings together the known enlargement of the RV, due to severe pulmonary insufficiency, leading to the EKG changes and sudden cardiac death.



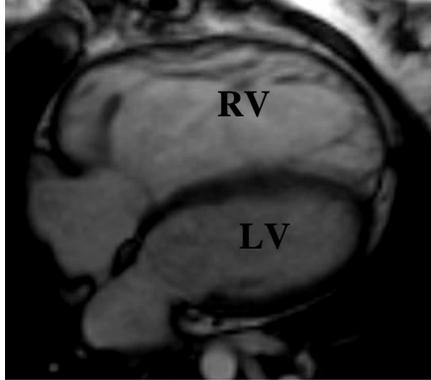
- Aortic regurgitation and aortic dilatation:** Aortic valve regurgitation (leaking) occurs due to damage of the aortic valve with VSD repair. The aortic tissue is thought to be abnormal in patients with TOF and may account for aortic enlargement/aneurysm.
- Branch Pulmonary Artery Stenosis:** The right and/or left pulmonary arteries may become narrowed, therefore reducing blood flow to the lungs. This may occur as part of TOF from birth, or as a result of surgical shunts that were placed. This narrowing also creates excess pressure and work on the right ventricle.



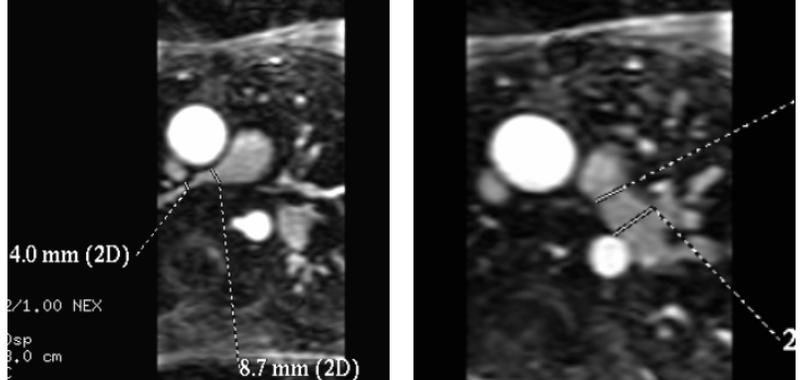
DIAGNOSTIC TESTING IN ADULTS WITH TOF

Physical examination, EKG, and echocardiography provide useful information about right ventricular size and function, valve function, pulmonary artery anatomy, and aortic size in TOF patients. Additional imaging modalities are also useful in identifying long-term complications in TOF patients.

- **Cardiac MRI:** Provides the most accurate measure of right ventricular size and function. Cardiac CT is helpful in patients who are not candidates for MRI due to the presence of pacemaker or internal cardioverter-defibrillator (ICD), claustrophobia or other reasons.

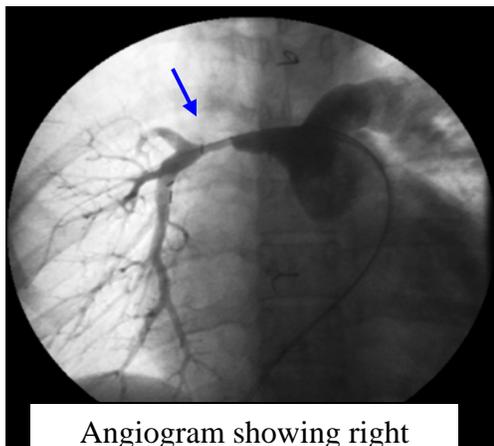


MRI showing RV size (enlargement) and RV function

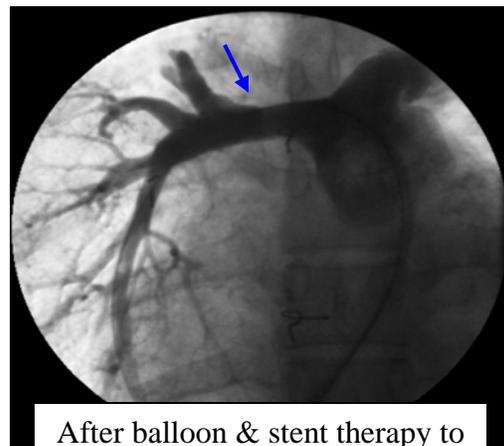


MRI showing pulmonary arteries and stenosis

- **Exercise stress testing:** Provides objective information about a patient's functional capability, exercise tolerance and development of exercise-induced heart rhythm problems. Despite feeling well, some patients with TOF may have significant exercise limitations.
- **Ambulatory EKG monitoring:** This is helpful in assessing the heart for conduction system abnormalities or arrhythmias. **Holter monitoring** records each heart beat in a 24 hour period. A 30-day event monitor is worn to help determine the cardiac rhythm during symptoms such as skipped beats, palpitations, heart racing, dizziness, or fainting.
- **Cardiac catheterization:** Directly measures pressures in the heart, identifies abnormalities involving the right ventricle, pulmonary valve, pulmonary arteries, left ventricle and aorta. Cardiac catheterization also provides an opportunity for an intervention to open up narrowed pulmonary arteries, stenotic pulmonary valve, or right ventricular outflow tract.

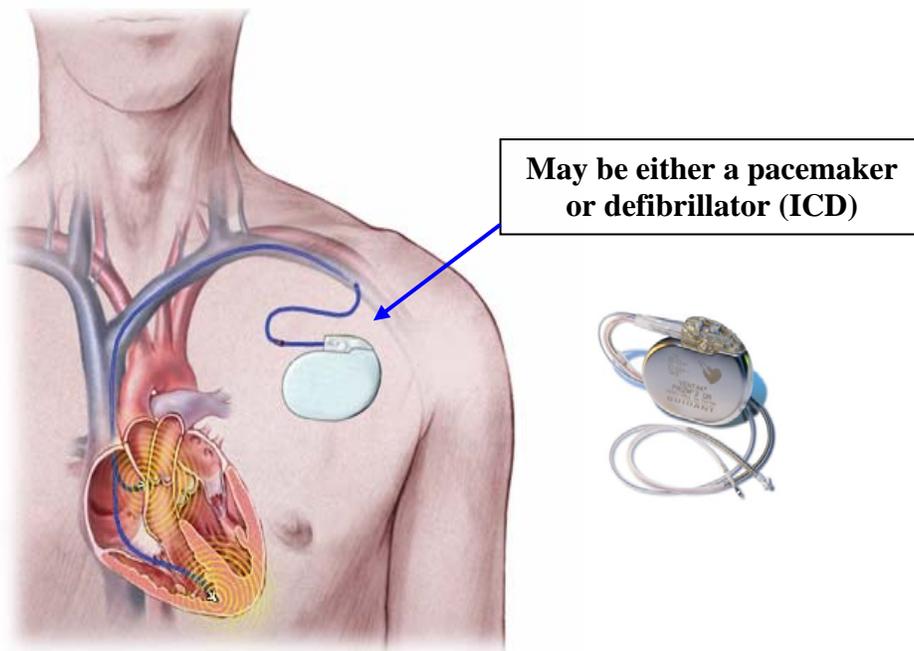


Angiogram showing right pulmonary artery stenosis



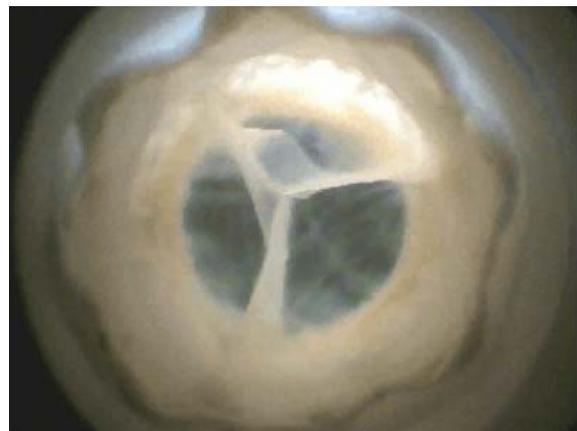
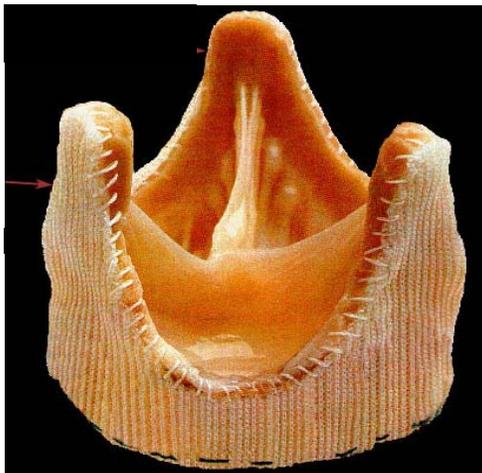
After balloon & stent therapy to expand the area of narrowing

Electrophysiologic study (EPS): An invasive test similar to a heart catheterization and evaluates the electrical system of the heart. The test is able to detect abnormal rhythms from either the upper chambers (atria) or lower chambers (ventricles). In some instances, the EP physician may decide to “map” the rhythm problem to find out where it is coming from in the heart. By doing so, the abnormal heart rhythm may be “ablated” or short circuited to prevent it from recurring. This may be done in the EP lab or the operating room, if open heart surgery is pending. The EP lab is the also the place where an ICD or intracardiac defibrillator or pacemaker will be placed. In many instances, the cardiac catheterization and the EPS will be done at the same time.



PULMONARY VALVE REPLACEMENT

With severe pulmonary valve insufficiency and an enlarged RV, in most cases a pulmonary valve replacement (PVR) will be recommended. With PVR, we would expect the RV to reduce in size, and possibly improve pumping function. There is some data to suggest that PVR also reduces the chance for sudden cardiac death. When PVR is combined with ablation (or short circuiting) dangerous heart rhythms, such as ventricular tachycardia, the chance of developing a life threatening arrhythmia is significantly reduced. Usually, a tissue valve is recommended which does not require the use of a blood thinner.



KEEP YOURSELF HEALTHY:

- Maintain routine cardiac follow-up
- Notify your cardiologist of concerning symptoms
- Take medications as they are prescribed (including antibiotics for dental procedures, tattoos, & piercings if indicated for your heart defect)
- Maintain a healthy weight
- Eat a healthy diet (low-sodium, low-fat)
- Avoid all tobacco products
- Participate in regular aerobic exercise (walking, swimming, jogging, or biking) for at least 30 minutes, most days of the week. Most patients should avoid isometric activities, or those to the point of grunting or straining (chin-ups, push-ups, heavy lifting) as these activities cause added strain to the heart
- Avoid dietary or medication sources of stimulants (caffeine in soda or chocolate, ephedrine/pseudoephedrine-containing decongestant cold medications)
- If you are female, it is important to plan pregnancy with your cardiologist to ensure optimal cardiac status, prior to becoming pregnant