Transposition of the Great Arteries

Description and Epidemiology

Transposition of the Great Arteries (TGA) is a cyanotic congenital heart defect in which the aorta arises from the right ventricle and the pulmonary artery arises from the left ventricle, creating two parallel circuits. The first circuit sends deoxygenated systemic venous blood to the right atrium and then back to systemic circulation through the right ventricle and aorta. The second circuit sends oxygenated pulmonary venous blood to the left atrium, which then goes back to pulmonary circulation via the left ventricle and pulmonary artery.

The prevalence of TGA is between 2.3-4.7 per 10,000 births. It accounts for about 3 percent of all congenital heart disease.

A video clip describing TGA:
http://www.youtube.com/watch?v=ZY11g3VZGVI&feature=player_embedded

Anatomy

The most common form of TGA is Dextro-TGA (D-TGA), in which the right ventricle is to the right of the left ventricle and the aorta anterior to and to the right of the pulmonary artery.
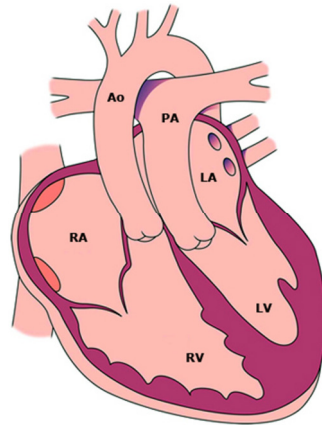

Diagram of D-TGA. Ao: aorta; LA: left atrium; LV: left ventricle; PA: pulmonary artery; RA: right atrium; RV: right ventricle *Multimedia Library of Congenital Heart Disease, Children's Hospital, Boston, editor Robert Geggel MD, www.childrenshospital.org/mml/cvp.*

The presence of D-TGA and no other cardiac abnormalities is referred to as *simple TGA*. The presence of other cardiac abnormalities in addition to D-TGA is referred to as *complex TGA*.

- About 50% of patients with TGA also have a ventricular septum defect (VSD).

- Patients with TGA and VSD are more likely to have other defects like pulmonary stenosis or atresia, overriding or straddling of an atrio-ventricular valve, or coarctation of the aorta.

- Obstruction of the left ventricular out flow tract is present in 25% of patients. This can be dynamic, where increased blood volume in the right ventricle causes bowing of the interventricular septum, or anatomical, involving pulmonary stenosis or atresia.

- There is abnormal coronary artery anatomy in one third of patients with D-TGA. Variations include vessels with an abnormal epicardial course, multiple coronary ostia (roots) arising from the same sinus of Valsalva, or an intramural coronary artery passing between the two great vessels. It is important to delineate coronary artery anatomy before surgical intervention because it can impact the surgical approach and procedure.

Levo-Transposition of the Great Arteries (L-TGA), occasionally referred to as "congenitally corrected-TGA," describes an anatomical abnormality in which the morphological left ventricle is on the right side of the heart. Deoxygenated systemic blood enters the right atrium, goes through the morphologic left ventricle, and enters the pulmonary circulation via the pulmonary artery. Oxygenated blood from the pulmonary circulation then enters the left atrium, goes through the morphologic right ventricle and enters systemic circulation via the aorta.
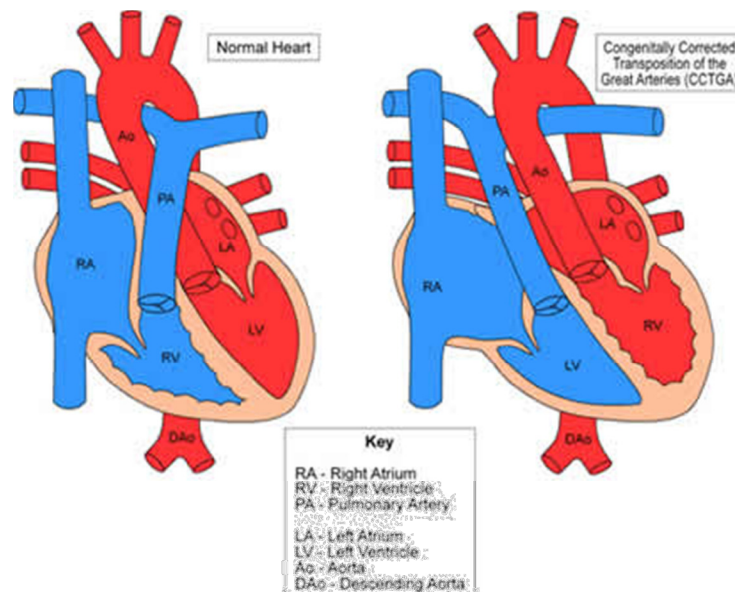

Diagram of L-TGA. *Heart Center, Nationwide Children's Hospital, Columbus, Ohio.* *http://www.nationwidechildrens.org/congenitally-corrected-transposition-great-vessels*

Although L-TGA frequently presents with other anatomical cardiac defects, individuals with simple L-TGA are generally asymptomatic and do not require

surgical intervention. Since each ventricle is designed to handle different blood volumes, the right ventricle may eventually hypertrophy, leading to symptoms of dyspnea and fatigue later in life. *From this point, this article will only discuss D-TGA, which will be referred to as TGA.*

Embryology, Pathogenesis and Genetics

Although many of the specific developmental aspects of TGA have not been elucidated, TGA is thought to arise from abnormal growth and development of the bilateral subarterial conus. Normally, the subpulmonic and subaortic conus are present in the first month of gestation above the right ventricle. The subaortic conus is resorbed between days 30-34 of gestation, allowing the aortic valve to migrate to its normal position above the left ventricle. The subpulmonic conus persists, allowing the pulmonic valve to stay in its position above the right ventricle.

In TGA, the subpulmonic conus is resorbed instead of the subaortic conus. This causes the pulmonary valve to migrate to a position above the left ventricle, while the aortic valve is forced anteriorly and associates with the right ventricle.

TGA, unlike many other congenital heart defects, is not associated with any one common genetic abnormality. TGA rarely occurs in other members of the family and the incidence in siblings is no higher than that of the general population. Less than 10% of cases of TGA have associated to non-cardiac anomalies, which is less frequent than other CHD defects. As a result, genetic testing and family screening are not warranted for children with TGA.

Physiology

TGA is well tolerated by the fetus in-utero. Oxygen-rich blood enters through the umbilical vein and passes from the right atrium to the left atrium through the fossa ovalis. From the left heart circulation, oxygenated blood then flows into the pulmonary artery and enters the systemic circulation via the ductus arteriosis (DA). The high pressure of the pulmonary capillary beds allows a right to left shunt across the ductus into the descending aorta. Since most blood is not pumped through the ascending aorta, oxygenation of the head and neck can be impaired even in utero.

After delivery, the degree of cyanosis and stability of the infant depends on the volume of blood mixing between the two parallel circulations. There must be some delivery of oxygenated blood to systemic circulation for compatibility with life. Mixing occurs most efficiently at the level of the atria across the fossa ovalis due to the lower pressure gradient between the two chambers; the large pressure gradients present at VSDs and the DA limit mixing between the two circulations.

Clinical Presentation and Diagnosis

TGA is one of the more difficult cardiac abnormalities to diagnose by fetal ultrasound. As a result, most cases are diagnosed after delivery. The typical ultrasound screening focuses on a four-chamber view of the heart, so TGA is often missed due to an absence of ventricular size discrepancy. Accuracy is improved by visualization of the great arteries and evaluation of whether they cross normally or not.

Most patients with TGA present within the first 30 days of life. The most commonly observed symptoms on physical exam are cyanosis, tachypnea, and cardiac murmur.
- The degree of cyanosis depends on the volume of mixing between the two circulations: patients with a medium to large-size VSD will typically be less cyanotic than those with an intact septum or small VSD. Cyanosis is not affected by exertion or supplemental oxygen.

- Most patients will have a respiratory rate greater than 60, but usually will not show other signs of respiratory distress like flaring, grunting or retractions. Patients with a large VSD can present with signs of respiratory distress by the 3rd or 4th week due to heart failure.

- Murmurs are only present in patients with a VSD or outflow tract obstruction. A VSD will present with a holosystolic murmur, while an outflow tract obstruction may have a systolic ejection murmur at the upper left sternal border.

- Up to 5% of patients will have diminished femoral pulses secondary to coarctation or interruption of the aortic arch.

Postnatal diagnosis is based on clinical suspicion of cyanotic congenital heart disease and is confirmed with echocardiography (Echo). Electrocardiography (ECG) is typically normal. Chest x-rays (CXR) can show a normal cardiac silhouette, although the classic CXR presentation features a heart with an "egg on a string" appearance.

Most patients who do not receive treatment die within the first year of life.

Treatment

Initial management of TGA focuses on stabilization of cardiac and pulmonary function. Patients with suspected or confirmed TGA are started on an IV infusion of prostaglandin E1 (alprostadil) to maintain a patent DA and may undergo Balloon Atrial Septostomy (BAS). Side effects of prostaglandin include apnea and hypotension secondary to vasodilation, which can be managed with intubation and volume expansion with normal saline.

BAS uses cardiac balloon catheterization via the umbilical vein or femoral artery. A balloon is inflated in the left atrium across the atrial septum. It is then vigorously pulled across the septum at least once until circulatory mixing is adequate for systemic oxygenation.

A video clip illustrating BAS: http://www.youtube.com/watch?v=hio-h3YRDwA&feature=player_embedded

Most patients are referred for surgical between day 3-5 of life. Delay of surgical treatment after 30 days of life can result in myocardial deconditioning. Patients with TGA undergo Arterial Switch Operation (ASO). Patients with TGA and a small VSD undergo ASO and VSD repair.

- ASO: both great arteries are transected and translocated to the opposite root. The coronary arteries are replanted at the neo-aortic root.
  Video clip illustrating ASO:
  http://www.youtube.com/watch?v=gYEo5z0hajM&feature=player_embedded

Follow-Up

Patients must be followed by a cardiologist with expertise in congenital heart disease for the rest of their lives. They must assess for arrhythmias, coronary artery insufficiency or stenosis, neo-aortic root dilation or regurgitation, pulmonary stenosis, and ventricular function. Patients with ASO may be more likely to develop atherosclerotic disease in the coronary arteries, so their cholesterol and triglyceride levels should be monitored.

Prognosis

Survival rates for patients with TGA following surgical correction are excellent, greater than 90% have 20-year survival. Since the ASO procedure was developed in the 1980s, existing survival data for the procedure does not extend much farther than 20 years.

Patients who have undergone ASO and have no residual defects, have a normal exercise tests, normal ventricular function and no evidence of ventricular tachyarrhythmias have no exercise restrictions and can participate in competitive sports. Most patients have slightly reduced exercise capacity, but it does not normally restrict daily activity.

However, case studies indicate that survivors after ASO are more likely to show neurologic impairment, most likely due to perioperative processes like hypoxemia, acidosis and hemodynamic instability.

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