

Pediatric Truncus Arteriosus and Transposition of the Great Arteries (TGA) — Causes and Survival Rate

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Truncus arteriosus is characterized by the presence of a common arterial trunk arising from both ventricles while transposition of the great arteries is a cyanotic congenital heart defect that is characterized by a pulmonary artery arising from the left ventricle and an aorta arising from the right ventricle. Patients with these two conditions are usually cyanotic and are usually diagnosed within the first few days of life. Echocardiography is the diagnostic modality of choice for the confirmation of the diagnosis in both conditions. Primary surgical repair of truncus arteriosus consists of committing the common arterial trunk to the left ventricle, while transposition of the great arteries is repaired by an arterial switch procedure. Both conditions should be repaired as early as possible, preferably within the neonatal period.



Overview

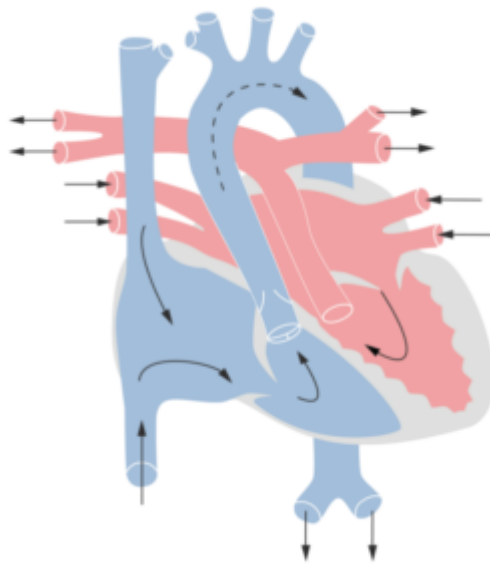
Truncus arteriosus (TA) is a rare form of **congenital heart defects** that is characterized by a **single arterial trunk** that arises from the right and left ventricles with a **single semilunar valve**, also known as a truncal valve. From this common arterial trunk, the

pulmonary arteries and coronary arteries arise.

Transposition of the great arteries (TGA) is considered as the most common form of **cyanotic heart disease** in the neonatal period. Patients with transposition of the great arteries have the aorta arising from the right ventricle and the pulmonary artery arising from the left ventricle.

Epidemiology of Truncus Arteriosus and Transposition of the Great Arteries

While TA is considered as a very rare congenital heart defect, TGA is relatively commonly diagnosed in cyanotic neonates. TA is responsible for approximately 1 % of all forms of congenital heart defects in liveborn infants. The incidence of TA is estimated to be around 5 to 15 per 100,000 live births. The incidence of TA in **stillbirths and aborted fetuses** with known cardiovascular anomalies is five times higher compared to the incidence of the condition in live births with **congenital heart disease**.



Transposition of the great vessels

Most patients with TA present during the **first days of life** while the remainder of the patients is diagnosed prenatally. TA is equally reported in **both sexes** without any major differences in incidence between different races.

Mortality within one year of untreated TA is 100 % while the mortality rate after early complete surgical correction of the anomaly goes down to 10 % in the initial postoperative period. The **20-year survival rate** of corrected TA is approximately 80 %.

The incidence of TGA in children with congenital heart diseases is around 7 %, which gives an annual incidence of TGA of 20 to 30 per 100,000 live births. The condition is more commonly seen in the **offspring of diabetic mothers**. Up to 90 % of the cases are an **isolated lesion** without any other congenital malformations **or associated with any syndromes**.

TGA appears to be **slightly more common in boys**. The most common age for presentation is during the early neonatal period. If surgically corrected, the 25-year survival rate can be as high as 97 % without any major morbidity.

The mortality rate within the first year of untreated TGA can be as high as 90 %. Patients with the delayed repair of the lesion might develop **congestive heart failure**, **cardiac arrhythmias**, and **irreversible pulmonary vascular obstructive disease**.

Pathophysiology of Truncus Arteriosus and Transposition of the Great Arteries

TA can be classified into **four different types**.

- **TA type 1** is characterized by the origin of a pulmonary trunk from the left side of the common trunk which then branches into the left and right pulmonary arteries.
- **TA type 2** is characterized by separate origins of the right and left pulmonary arteries from the common trunk.
- **TA type 3** is characterized by the presence of two pulmonary arteries that arise far from each other from the common trunk. Usually, one of the pulmonary arteries arises from the site of the ductus arteriosus.
- **TA type 4** is no longer considered a TA. Instead, it is now classified as pulmonary atresia with a **ventricular septal defect**.

Because both ventricles in TA give rise to a common trunk, it is understandable that deoxygenated blood and oxygenated blood are going to be mixed. Therefore, patients with TA are **cyanotic** and have **a systemic ventricular overload**. Because of the lower pulmonary resistance, patients usually have **pulmonary over circulation** which can predispose them to **pulmonary vascular disease** and **pulmonary arterial hypertension**.

The exact etiology of TGA is unknown. **TGA is not compatible with survival** unless mixing of oxygenated and deoxygenated blood happens at some level. In TGA, the oxygenated blood from the pulmonary circulation drains into the left atrium but instead of being pumped to the systemic circulation, the blood is pumped to the pulmonary artery which is connected to the left ventricle.

On the other hand, the deoxygenated blood that is drained into the right atrium is pumped back into the systemic circulation without passing through the pulmonary circulation to get oxygenated. Therefore, the only way for the blood to be partially oxygenated before being pumped into the systemic circulation is for **mixing** to happen at some anatomical point.

The most common anatomical sites for mixing are at the atrial level through an **atrial septal defect**, at the ventricular level through a **ventricular septal defect** or through a **patent ductus arteriosus**. Patients usually have **hypoxemia** and are **cyanotic**.

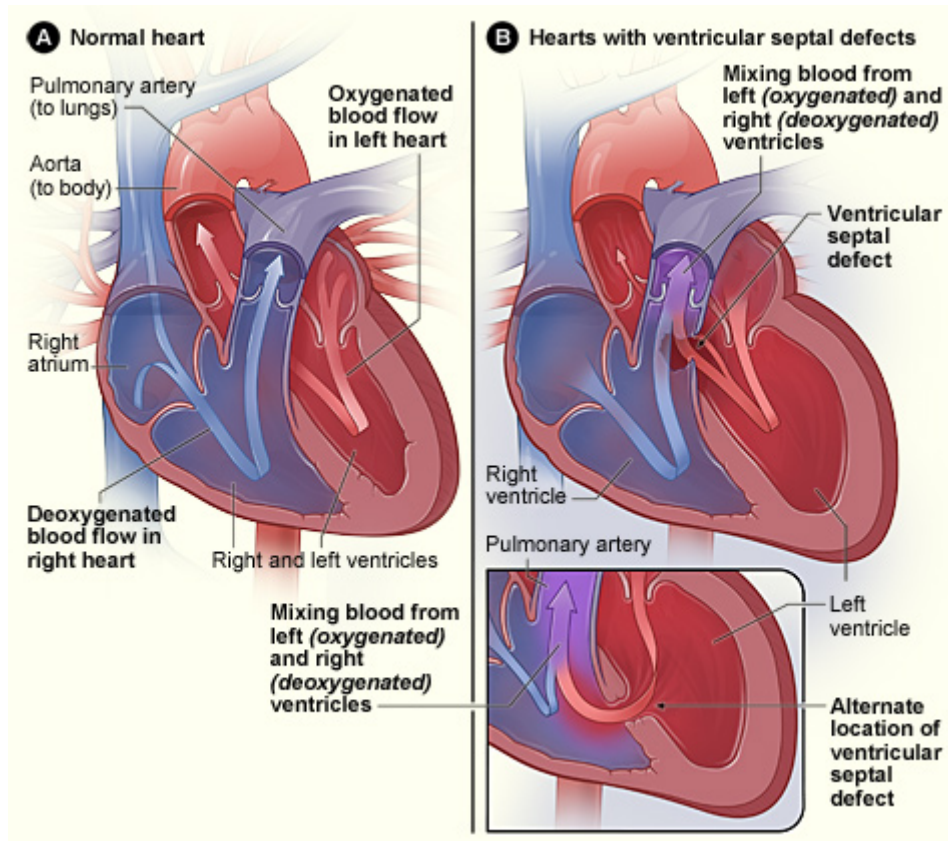
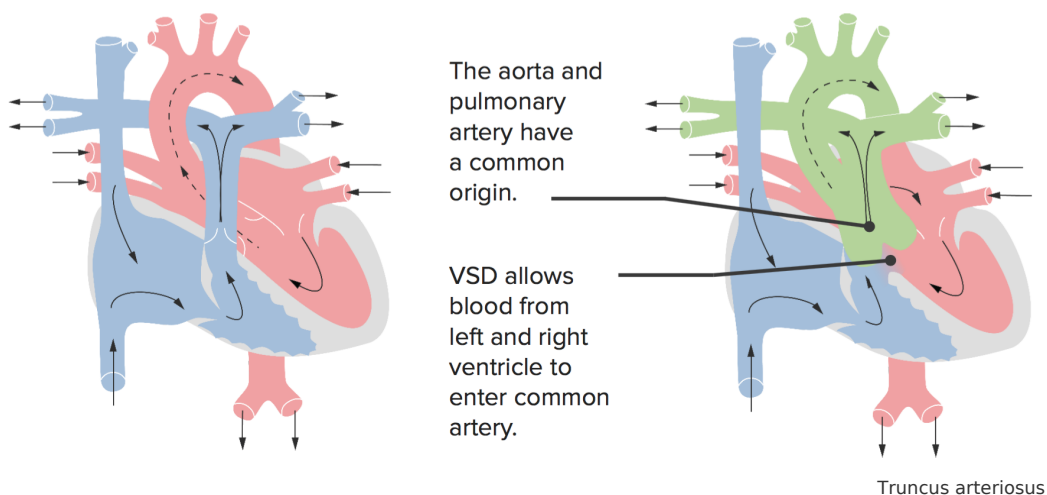


Figure A shows the structure and blood flow in the interior of a normal heart. Figure B shows two common locations for a ventricular septal defect. The defect allows oxygen-rich blood from the left ventricle to mix with oxygen-poor blood in the right ventricle.

Clinical Presentation of Truncus Arteriosus and Transposition of the Great Arteries

Patients with TA can be diagnosed prenatally with **fetal ultrasound**. Those diagnosed in the first few days of life usually present with **poor feeding, excessive sweating, tachypnea**, and **cyanosis**. Patients might also present with symptoms and signs suggestive of congestive heart failures such as **pulmonary congestion, liver enlargement**, and **weak peripheral pulses**.



Patients with TGA usually present within the **first few hours of life** with **cyanosis**. Patients with TGA with an intact ventricular septum usually are diagnosed within the first day of life because of cyanosis. Those with a large ventricular septal defect are usually diagnosed within the first 3 to 6 weeks of life when they develop **congestive heart failure**.

Patients with TGA with a **ventricular septal defect and left ventricular outflow tract obstruction** are usually diagnosed at birth because of severe cyanosis. **Hepatomegaly** may be present in patients presenting with congestive heart failure.

Diagnostic Workup for Truncus Arteriosus and Transposition of the Great Arteries

Patients with TA should undergo **arterial blood gases assessment** to determine the degree of **hypoxemia** and **acidosis**. Additionally, **serum calcium levels** should be determined in this group of patients because of the association between TA and **DiGeorge syndrome** which is characterized by **hypoparathyroidism** and **hypocalcemia**.

Electrocardiograms are usually not very helpful in patients with TA. Patients with TA usually have **pulmonary overcirculation** and **increased pulmonary vascular markings** on a **chest x-ray**.

The diagnosis of TA is confirmed by **echocardiography** which shows the presence of a **common trunk** arising from the right and left ventricles. The presence of other congenital heart defects, the type of the TA, and the presence of signs of congestive heart failure such as ventricular dilatation should be assessed on echocardiography.

Magnetic resonance imaging is usually not indicated in patients with TA because echocardiography is usually sufficient in confirming the diagnosis.

In TGA and TA, a **hyperoxia test** might be useful in the differentiation between **cyanotic heart disease** and **primary pulmonary disease** behind **hypoxemia**. Patients with pulmonary disease usually have a partial pressure of oxygen level that is above 150 mm Hg after the administration of 100 % oxygen for 10 minutes.

Echocardiography is the imaging modality of choice for the diagnosis of TGA and cardiac catheterization is no longer recommended as a first-line diagnostic modality. Patients with TGA might have a **normal chest x-ray**.

Patients with TGA with an intact ventricular septum might have an **egg on a string appearance of the heart** on their chest x-ray. Those who have a large **ventricular septal defect** usually have **increased pulmonary vascular markings** and **cardiomegaly**. When echocardiography is performed, the **anatomical location of the mixing** should be determined.

Patients with TGA are at an increased risk of developing **cardiac arrhythmias** even after arterial switch repair. The most likely cause of ventricular arrhythmias in this cohort of patients is **ventricular fibrosis** which can be assessed by **Gadolinium enhancement cardiovascular magnetic resonance imaging**.

Treatment of Truncus Arteriosus and Transposition of the Great Arteries

Patients with TA who present with signs and symptoms suggestive of congestive heart failure should be treated with **digoxin** and **diuretics**. **Surgical repair** should be offered as soon as possible.

The currently recommended surgical treatment of TA consists of the **closure of the ventricular septal defect**, committing the common arterial trunk to the left ventricle, and the reconstruction of the pulmonary artery and the right ventricular outflow tract. It is recommended to perform a **complete primary repair** instead of a staged procedure or a palliative procedure.

Patients with TGA are dependent on the mixing of blood at the previously mentioned anatomical sites. To improve mixing, the continuous administration of **prostaglandin E1** is recommended to maintain the patency of the ductus arteriosus. The second step in the management of TGA is to perform a **cardiac catheterization procedure** in patients with an intact ventricular septum to create an atrial septum defect by a **balloon atrial septostomy**.

The definitive treatment of TGA should be offered as soon as possible and is surgical. Patients with an intact ventricular septum should undergo an **arterial switch procedure** in which the arteries are switched to have the normal relationship with the corresponding ventricle, i.e. the pulmonary artery to the right ventricle and the aorta to the left ventricle. This procedure should be performed **before 4 weeks of age**.

Patients with TGA and a ventricular septal defect should undergo an **arterial switch procedure in addition to the primary closure of the ventricular septal defect**. Again, the procedure should be performed as early as possible, otherwise, the left ventricle will not be able to sustain the systemic circulation pressure.

References

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