**Approach to Infant with Critical Congenital Heart Disease: Clinical Pearls**

**Introduction:**

- The most common congenital disorder in newborns is congenital heart disease (CHD) affecting approximately 1% of live births.
- We will focus on cyanotic congenital heart diseases, which are lesions that allow circulation of deoxygenated blood in the systemic circulation.
- The common cyanotic congenital heart diseases are often referred to as the 5 T's: Truncus Arteriosus, Transposition of the Great Arteries, Tricuspid Atresia, Tetralogy of Fallot, and Total Anomalous Venous Pulmonary Return.
- All of these conditions require surgical correction, but a more detailed discussion of surgical management is out of the scope of this talk.

**Truncus Arteriosus**:

Anatomy/Physiology:

- Truncus arteriosus results from the lack of septation of the embryologic aorticopulmonary trunk, leaving one great artery as outflow from the heart.
- Blood flows from the left and right ventricle through a common semilunar valve and out through the truncal artery which gives rise to the aorta and pulmonary arteries. The systemic and pulmonary venous blood flow are mixed.
- The oxygenation status, and therefore, level of cyanosis, of the patient depends on the relative balance of systemic and pulmonary blood flow. As pulmonary vascular resistance (PVR) decreases in the first weeks of life, blood flows preferentially into the pulmonary artery instead of the aorta. This left-to-right shunting increases and the patient can rapidly develop heart failure.

Clinical Presentation

- Truncus arteriosus presents as mild or moderate cyanosis in the first days of life or heart failure in the first weeks of life.
- On physical exam, the most common finding on auscultation is a loud and single second heart sound (from closure of the single aorto-pulmonary valve) and a prominent ejection click at the apex or left sternal border.
- Peripheral pulses are bounding and pulse pressure is often increased (as seen in aortic regurgitation, due to regurgitation through the common aorto-pulmonary valve).
- As the child progresses to heart failure, chest x-ray shows cardiac enlargement and increased pulmonary vascular markings.

**Transposition of the Great Arteries:**

Anatomy/Physiology:

- Transposition of the Great Arteries is caused by failure of rotation of the aorta and pulmonary artery during embryological development of the heart.
- This leaves the aorta attached to the right ventricle and the pulmonary artery attached to the left ventricle, creating two parallel circuits.
- Oxygenated blood travels from the lungs through the pulmonary veins to the left atrium, left ventricle and then back through the pulmonary artery to the lungs. Deoxygenated blood travels from the body through the superior and inferior vena cava to the right atrium, right ventricle and then back through the aorta to the body
- The classic form of TGA is dextro-TGA (as opposed to levo- or L looped-TGA, which is congenitally correct TGA)
- About 25% of patients with D-TGA also have a VSD

Clinical Presentation and Management

- Most infants present immediately with cyanosis. The degree of cyanosis is dependent on the presence and size of communications between the two parallel circulations that allow mixing, such as a patent ductus arteriosus, atrial septal defect, or ventricular septal defect.
- The condition is most often diagnosed by prenatal or postnatal echocardiography.
- Chest x-ray classically shows an “egg on a string appearance” due to the great arteries forming a narrower vascular pedicle when transposed.
- The mainstays of initial treatment include prostaglandins to maintain PDA patency and balloon atrial septostomy to establish an ASD if it is not already present.
- The definitive treatment is an arterial switch operation, performed in the first week of life, where the transposed vessels and coronary arteries are dissected away from their valves and switched, restoring a normal circulation.

**Tricuspid Atresia**:
Anatomy/Physiology:
- Tricuspid valve atresia results from congenital agenesis or absence of the tricuspid valve, leaving no communication between the right atrium and right ventricle.
- Blood exits the right atrium through the foramen ovale to the left atrium. Mixing of deoxygenated systemic blood with oxygenated pulmonary venous return in the left atrium results in cyanosis.
- The amount of pulmonary blood flow and degree of cyanosis are dependent on the presence and size of a ventricular septal defect as well as the anatomy of the great arteries.
- In patients without a VSD or with significant pulmonary stenosis, a patent ductus arteriosus is the only source of pulmonary blood flow

Clinical Presentation and Management
- Tricuspid atresia presents with central cyanosis and a single second heart sound.
- If the patient has a VSD or PS, a murmur at the left lower sternal border may also present.
- Chest x-ray shows lack of the smooth convexity of the right heart border due to the lack of filling in the right ventricle.
- Initial medical management focuses on general cardiorespiratory support and administration of prostaglandins to provide a source of pulmonary blood blow by maintaining a patent ductus arteriosus (especially in patients without a VSD or with significant pulmonary stenosis).

**Tetralogy of Fallot:**
Anatomy/Physiology:
- Tetralogy of Fallot is the most common cyanotic congenital heart disease.
- The condition is defined by four major features: pulmonary artery stenosis or atresia, ventricular septal defect, overriding aorta, and right ventricular hypertrophy.
- The degree of cyanosis and other symptoms are dependent on the degree of right ventricular outflow tract obstruction.
- The greater the right ventricular outflow obstruction, the less pulmonary blood flow, and the more flow through the VSD from the right to the left. These changes lead to more deoxygenated blood in the systemic circulation and greater cyanosis.

Clinical Presentation and Management
- Tetralogy of Fallot is most commonly diagnosed prenatally by fetal echocardiogram. However, it can present postnatally with cyanosis or an asymptomatic murmur.
- Infants with unrepaired Tetralogy of Fallot are at risk for “Tet spells,” where agitation or other factors can cause an acute increase in right ventricular outflow tract obstruction or PVR and profound cyanosis.
- Management of “Tet spells” aims to decrease PVR and increase SVR while prolonging diastole. You can decrease PVR with calming (or morphine to calm agitation) and oxygen, increase SVR

with knees to chest maneuver or phenylephrine, increase preload with fluid, and decrease HR with beta blockade (increase diastolic filling time). Older children with unrepaired Tetralogy of Fallot will sometimes squat down or bring their legs to their chest to increase systemic resistance and decrease the right to left shunting through the VSD, thus increasing pulmonary blood flow and improving oxygen saturation.

- On physical exam, the second heart sound is usually single because of a soft pulmonic component. A crescendo-decrescendo murmur with a harsh systolic ejection quality along the left mid to upper sternal border is due primarily to right ventricular outflow obstruction.
- Classic CXR shows a boot-shaped heart

**Total Anomalous Pulmonary Venous Return:**

Anatomy/Physiology:

- Total Anomalous Pulmonary Venous Return (TAPVR) is caused by failure of the four pulmonary veins to connect to the left atrium.
- There are a number of different anatomic variants. In the most common variant, the pulmonary veins convene in a confluent chamber posterior to the left atrium and drain into the superior vena cava.
- This anatomy leads to mixing of the oxygenated pulmonary venous return and deoxygenated systemic venous return to the right atrium. The increased flow to the right heart leads to dilation of the atrium and ventricle.
- The partially oxygenated blood is shunted from right-to-left through an atrial septal defect or less commonly through a patent ductus arteriosus resulting in cyanosis and early right sided heart failure.

Clinical Presentation and Management

- On physical exam, patients with TAPVR have a fixed split second heart sound due to right ventricular volume overload
- There is often a systolic ejection murmur due to increased volume of flow across the pulmonary valve.
- Classic chest X-ray findings include a “snowman” or “figure 8” sign.

References


1. Altman, Carolyn A. “Identifying Newborns with Critical Congenital Heart Disease.” *UpToDate*, 21 Nov. 2017, www.uptodate.com/contents/identifying-newborns-with-critical-congenital-heart-disease.
2. Balasubramanian, Sowmya and Tacy, Theresa. “Tricuspid valve (TV) atresia.” *UpToDate,* 24 Feb 2016, https://www.uptodate.com/contents/tricuspid-valve-tv-atresia.
3. Doyle, Thomas and Kavanaugh-McHugh, Ann. “Management and outcome of tetralogy of Fallot.” *UptoDate,* 15 Feb 2018, https://www.uptodate.com/contents/management-and-outcome-of-tetralogy-of-fallot.
4. Doyle, Thomas and Kavanaugh-McHugh, Ann. “Pathophysiology, clinical features, and diagnosis of tetralogy of Fallot.” *UptoDate,* 15 Feb 2018, https://www.uptodate.com/contents/pathophysiology-clinical-features-and-diagnosis-of-tetralogy-of-fallot.
5. Fulton, David R and Kane, David A. “Management and outcome of D-transposition of the great arteries.” *UpToDate*, 1 Mar 2018, https://www.uptodate.com/contents/management-and-outcome-of-d-transposition-of-the-great-arteries.

6. Fulton, David R and Kane, David A. “Pathophysiology, clinical manifestations, and diagnosis of D-transposition of the great arteries.” *UpToDate*, 1 Mar 2018, https://www.uptodate.com/contents/pathophysiology-clinical-manifestations-and-diagnosis-of-d-transposition-of-the-great-arteries.
7. Fulton, David R and Soriano, Brian. “Total anomalous pulmonary venous connection.” *UpToDate*, 10 Nov 2017, https://www.uptodate.com/contents/total-anomalous-pulmonary-venous-connection.
8. Fulton, David R and Soriano, Brian. “Truncus Arteriosus.” *UpToDate*, 12 Jul 2016, https://www.uptodate.com/contents/truncus-arteriosus.
9. Shetty, Aditya. “Snowman Sign (Total Anomalous Pulmonary Venous Return) | Radiology Reference Article.” *Radiopaedia.org,* 2018, radiopaedia.org/articles/snowman-sign-total-anomalous-pulmonary-venous-return-1.
10. van Roekens CN, Zuckerberg AL. Emergency management of hypercyanotic crises in tetralogy of Fallot. Ann Emerg Med 1995;25:256–8.