

Cyanotic heart disease

Summary

Cyanotic heart defects (CHDs) are congenital cardiac malformations that commonly affect the atrial walls, e.g., the right atrium (RA) or left atrium (LA); ventricular walls, e.g., the left ventricle (LV) or right ventricle (RV); heart valves; or large blood vessels. Common causes include genetic defects (e.g., DiGeorge syndrome), maternal conditions (e.g., diabetes), and spontaneous genetic mutations. Pathophysiologically, cyanotic heart defects are characterized by a right-to-left cardiac shunt, which leads to deoxygenated blood entering the systemic circulation. The resulting hypoxemia manifests clinically as cyanosis, which may occur as acute, life-threatening episodes. Other symptoms include failure to thrive, characteristic heart murmurs, and symptoms of heart failure. Diagnosis is confirmed through visualization of the defect on echocardiography. Further diagnostic findings include low oxygen saturation and characteristic x-ray findings. Heart defects requiring treatment are repaired via catheter procedures or surgery. Supportive medical therapy is required in patients with heart failure (e.g., diuretics, inotropic agents) or if surgery cannot be performed (e.g., prostaglandin). If untreated, most cyanotic heart defects are fatal within the first year of life.

FEEDBACK

A. General concepts

1. **Cyanosis** may be peripheral, central, or both. **Peripheral cyanosis** is usually caused by vasomotor instability or vasoconstriction as a result of cold temperature. **Central cyanosis**, especially apparent in the tongue and inner mucous membranes, may be attributable to both cardiac and noncardiac causes:
 - a. **Noncardiac causes of central cyanosis** include pulmonary disease, sepsis, hypoglycemia, polycythemia, and neuromuscular diseases that impair chest wall movement.
 - b. The **most common cardiac causes of central cyanosis** may be remembered using the mnemonic "5 T's": tetralogy of Fallot, transposition of the great arteries, tricuspid atresia, truncus arteriosus, and total anomalous pulmonary venous return.

2. Evaluation

- a. A thorough physical examination is essential.
- b. Other initial studies include pulse oximetry (in room air), complete blood count (CBC), arterial blood gas (ABG), ECG, and CXR. The degree of pulmonary blood flow seen on CXR may be useful in diagnosis (Table 8-3).
- c. The **100% oxygen challenge test** suggests cyanotic CHD when the PaO_2 fails to rise above 100–150 mm Hg despite the administration of 100% oxygen.
- d. **Echocardiogram** provides a definitive diagnosis.

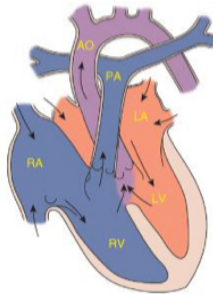
- **Cyanosis**, the visible sign of this shunt, occurs when 5 g/100 mL of reduced hemoglobin is present in systemic blood.
 - ♥ The patient's hemoglobin level determines the presentation of clinical cyanosis.
 - ♥ polycythemic patient appears cyanotic with a lower percentage of reduced hemoglobin.
 - ♥ patient with anemia requires a higher percentage of reduced hemoglobin for the recognition of cyanosis.
- The most common cyanotic congenital heart defects are the 5 "Ts":
 1. tetralogy of Fallot
 2. transposition of the great arteries
 3. tricuspid atresia
 4. truncus arteriosus
 5. total anomalous pulmonary venous return

Notes:

First: Tetralogy of Fallot:

ETIOLOGY AND EPIDEMIOLOGY

- The most common cyanotic congenital heart defect, 10% of all congenital heart defects .
- Anatomically, there are four structural defects:
 1. VSD
 2. pulmonary stenosis
 3. overriding aorta
 4. right ventricular hypertrophy
- Tetralogy of Fallot is believed to be due to abnormalities in the septation of the truncus arteriosus into the aorta and pulmonary arteries that occur early in gestation (3 - 4 weeks).
- The VSD is large
- the pulmonary stenosis is most commonly subvalvular or infundibular



Epidemiology

- Prevalence: 4-5/10,000 live births in the US^[15]
- The most common cyanotic CHD^[16]

Etiology

- Typically sporadic^[17]
- Associated with the following genetic disorders:
 - DiGeorge syndrome^{[18][19]}
 - Down syndrome^{[20][21]}
- Maternal exposures during pregnancy
 - Alcohol consumption
 - Phenylketonuria^{[22][23]}
 - Diabetes^{[24][25]}

Pathophysiology

- During fetal development, anterior and superior deviation of the **infundibular septum** → misaligned VSD with overriding aortic root and subsequent RVOTO
- Physiologic blood flow is determined by the severity of RVOTO.
 - A large VSD → equal pressures in the right and left ventricles → blood flow along the path of least resistance
 - Severe RVOTO → flow from RV to LV → desaturated blood entering the circulation via the aorta



CLINICAL MANIFESTATIONS

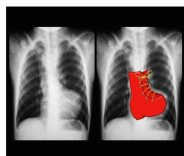
- The degree of cyanosis depends on the amount of pulmonary stenosis.
- Cyanosis develops within the first few years of life
- Infants initially may be acyanotic.
- First presentation may include poor feeding, fussiness, tachypnea, and agitation
- pulmonary stenosis murmur is the usual initial abnormal finding. Systolic ejection murmur .
- More cyanotic patients have greater obstruction and a softer murmur.
- An acyanotic patient with TOF (pink tet) has a long, loud, systolic murmur with a thrill along the RVOT.
- If the pulmonary stenosis is more severe, the amount of right-to-left shunting at the VSD increases and the patient becomes more cyanotic.
- single S₂ (Pulmonic valve closure not heard)
- May have a bulging left hemithorax+ Right ventricular predominance on palpation .
- right ventricular impulse at the left sternal border are typical findings (heave)
- hypoxic ("Tet") spells:
 - ♥ sudden, increased subpulmonic stenosis
 - ♥ they are usually progressive.
 - ♥ During a spell, the child typically becomes restless and agitated and may cry inconsolably.
 - ♥ Hyperpnea occurs with gradually increasing cyanosis and loss of the murmur.
 - ♥ In severe spells, prolonged unconsciousness and convulsions, hemiparesis, or death may occur.
- Independent of hypoxic spells, patients with tetralogy are at increased risk for cerebral thromboembolism and cerebral abscesses from their right-to-left intracardiac shunt.

Tetralogy of Fallot (hypercyanotic or "tet") spells are characterized by sudden cyanosis and decreased murmur intensity.

- Infants with mild TOF may be asymptomatic on initial examination.
- Mild to severe cyanosis, which depends on the severity of the RVOTO
 - Mild obstruction → more pronounced left-to-right shunt via VSD → little or mild cyanosis
 - Severe obstruction → more pronounced right-to-left shunt via VSD → severe cyanosis

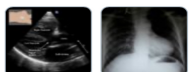
Diagnostics

- Prenatal diagnosis: Most cases are now diagnosed before birth due to improvements in fetal echocardiography.^[30]
- Imaging
 - Echocardiography (confirmatory test)
 - Detection of the main features of TOF
 - Quantification of right ventricular outflow tract pressure gradient
 - Supplementary cardiac catheterization may be performed.
 - Chest x-ray
 - Boot-shaped heart (due to upturned cardiac apex and RVH)
 - Normal or decreased pulmonary vascular markings
 - Concave pulmonary artery segment
- ECG
 - Right axis deviation
 - Prominent anterior R waves (V1-V2)
 - Prominent posterior S waves
 - Right atrial enlargement and RVH (P pulmonale)
- Pulse oximetry: ↓ SpO₂
- Hyperoxia test: helps to distinguish cardiac from pulmonary causes of cyanosis



TREATMENT

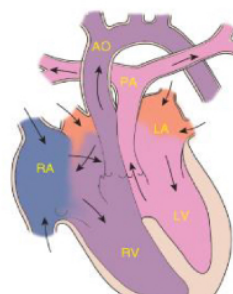
- The natural history of tetralogy of Fallot is progression of pulmonary stenosis and cyanosis.
- Treatment of hypoxic spells consists of:
 1. oxygen administration
 2. placing the child in the knee-chest position (to increase venous return),
 3. giving morphine sulfate^B (to relax the pulmonary infundibulum and for sedation)
 5. α-adrenergic agonist (phenylephrine) to create systemic vascular resistant.
 6. If spells are frequent, β-adrenergic antagonists (propranolol) ↓ muscular spasm.
- Complete surgical repair with closure of the VSD and removal or patching of the pulmonary stenosis can be performed in infancy.
- palliative shunt surgery between the subclavian artery and pulmonary artery is performed for complex forms of TOF with more complete repair at a later time.
- Subacute bacterial endocarditis prophylaxis is indicated.



2nd: Transposition of the great arteries:

ETIOLOGY AND EPIDEMIOLOGY

- Although **dextroposed transposition of the great arteries** represents only about 5% of congenital heart defects.
- **The most common cyanotic lesion to present in the newborn period.**
- In dextroposed transposition, the aorta arises from the right ventricle, anterior and to the right of the pulmonary artery, which arises from the left ventricle.
- This transposition results in desaturated blood returning to the right heart and being pumped back out to the body, while well-oxygenated blood returning from the lungs enters the left heart and is pumped back to the lungs.



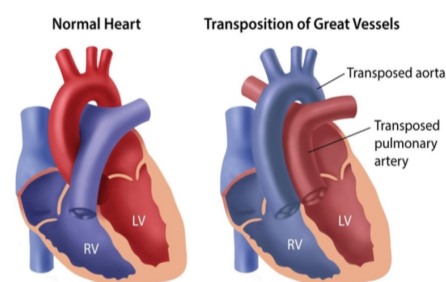
- Without mixing of the two circulations, death occurs quickly.
- Mixing can occur at the atrial (patent foramen ovale/ASD), ventricular (VSD), or great vessel (PDA) level.

Pathophysiology [42]

- Failed spiraling of the **aortopulmonary septum** → RV emptying into the aorta and LV into the pulmonary artery → complete isolation of the pulmonary and systemic circuits → ↓ oxygenated blood entering the systemic circulation
- Fatal, unless mixing occurs via an intracardiac shunt (e.g., PFO, VSD, ASD) or via an extracardiac connection (e.g., PDA)

CLINICAL MANIFESTATIONS

- A history of **cyanosis** is always present, although it may not be profound and depends on the amount of mixing.
- **Quiet tachypnea** and a **single S₂** are typically present.
- If the ventricular septum is intact, there may be no murmur.
- Children with transposition and a large VSD have improved intracardiac mixing.
- They may present with signs of CHF.
- The heart is hyperdynamic, with palpable left and right ventricular impulses.
- A loud VSD murmur is heard.

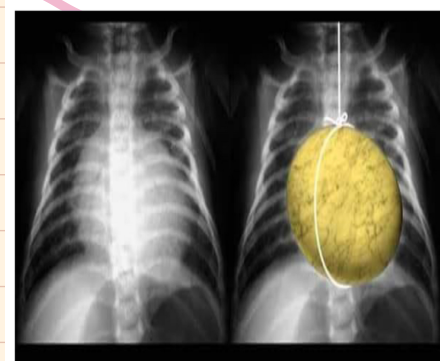


IMAGING STUDIES

- ECG findings typically include **right axis deviation** and **right ventricular hypertrophy**.
- The chest x-ray reveals **increased pulmonary vascularity**, and the cardiac shadow is classically **an egg on a string** created by the narrow superior mediastinum.
- Echocardiography shows the transposition of the great arteries, the sites and amount of mixing, and any associated lesions.

4. Management

- Neonates may require **initial management** with **PGE** to improve oxygen saturation by keeping the ductus patent. **Emergent balloon atrial septostomy** (Rashkind procedure) is an often life-saving procedure that increases the size of an ASD or PFO.
- Definitive repair** is the **arterial switch operation**, in which the great arteries are incised above their respective valves and implanted in the opposite root. The coronary arteries are attached to the original aorta, so the coronaries must also be incised and reimplanted.



Transposition Of Great Arteries.
(Egg on String Appearance)

3rd: Tricuspid atresia:

• Definition

- Absent or rudimentary tricuspid valve resulting in no blood flow between the RA and the RV
- Patient survival is only possible if there are interatrial and interventricular communications (see "Other cardiac defects" below). [56]

• Other cardiac defects associated with tricuspid valve atresia

- ASD
- VSD
- RV hypoplasia
- Other defects (e.g., pulmonary outflow obstruction, aortic stenosis, aortic coarctation)

ETIOLOGY AND EPIDEMIOLOGY

- accounts for 2% of all congenital heart defects
- The absence of the tricuspid valve results in a **hypoplastic right ventricle**.
- All systemic venous return must cross the atrial septum into the left atrium.
- The presence of a PDA or VSD also is necessary for pulmonary blood flow and survival.

2. **Pathophysiology.** Whether a VSD is also present plays a significant role in determining the clinical presentation.

- If no VSD is present and the **ventricular septum is intact, pulmonary atresia is also present**. For blood to flow to the lungs in this situation, a PDA must be present. As the PDA constricts after birth, visible cyanosis develops.
- If a **VSD is present**, blood flow from the left ventricle through the VSD and into the pulmonary artery (left-to-right shunt) may be adequate, facilitating acceptable systemic oxygen saturations (**Figure 8-8**).

3. **Clinical features.** Signs also depend on anatomy.

- Patients with an **intact ventricular septum and pulmonary atresia have no murmur and a single S₂**.
- Patients with a **VSD** have a VSD murmur (see **Table 8-2**).

IMAGING STUDIES

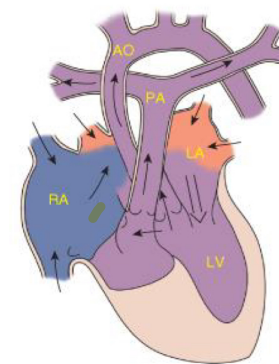
- The ECG shows **left ventricular hypertrophy** and a **superior QRS axis** (between 0 and -90 degrees).
- The chest x-ray reveals a normal or mildly enlarged cardiac silhouette with **decreased pulmonary blood flow**.
- Echocardiography shows the anatomy, associated lesions, and source of pulmonary blood flow.

TREATMENT

- Management initially depends on the **presence of a VSD** and the **amount of blood flow across the VSD to the lungs**.
- If **there is no VSD**, or it is small, **prostaglandin E₁** is used to maintain pulmonary blood flow until surgery.

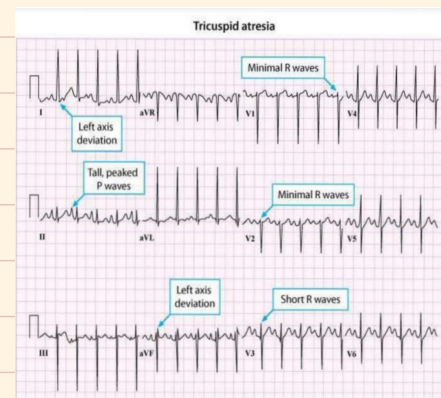
4. **Management.** Treatment includes staged surgery with eventual **Fontan** procedure at 3–6 years of age.

- A bidirectional **Glenn shunt** (SVC is anastomosed to the right pulmonary artery) is usually performed at 4–6 months of age.
- In the **Fontan procedure**, flow from the inferior vena cava is directed into the pulmonary arteries, usually by means of an extracardiac conduit.
- The **net result of these procedures is systemic venous return directed to the pulmonary artery**.



Clinical features

- **Central cyanosis** (occurs within days after birth)
- **Tachypnea**
- **Cardiac examination findings**
 - **Holosystolic murmur at the lower left sternal border**
 - **Single S₂**
 - **Jugular venous distention with a prominent A wave**
 - **Diminished peripheral pulses**



❖ N.B:

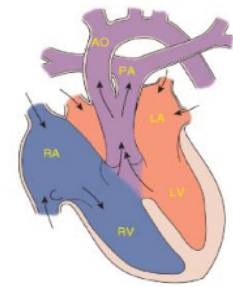
- **Left axis deviation on neonatal electrocardiogram is never normal.**
- **Tricuspid valve atresia is a cyanotic congenital heart defect characterized by left axis deviation on electrocardiogram and decreased pulmonary markings on chest radiograph due to hypoplasia of the right ventricle and pulmonary outflow tract.**

4th :truncus arteriosus:

- **Definition:** underdevelopment of the **aortopulmonary septum** → failure of the **truncus arteriosus** to divide into the aorta and **pulmonary trunk** → a single trunk that receives output from both ventricles
- 2. **Pathophysiology.** Because the aorta and pulmonary arteries are connected and the pulmonary arteries are well formed, excessive blood flows to the lungs and CHF commonly develops. Mixing of desaturated and saturated blood occurs within the truncus, and patients are commonly only *mildly desaturated and mildly cyanotic* (Figure 8-

ETIOLOGY AND EPIDEMIOLOGY

- Truncus arteriosus occurs in less than 1% of all cases of congenital heart disease.
- It results from the failure of septation of the truncus, which normally occurs during the first 3- 4 weeks of gestation.
- Anatomically a single arterial trunk arises from the heart with a large VSD immediately below the truncal valve.
- The pulmonary arteries arise from the single arterial trunk either as a single vessel that divides or individually from the arterial trunk to lungs.
- The failure of **neural crest cells** to migrate during the development of the cardiac outflow tract results in **incomplete AP septum** formation. [79]
- Associated with **DiGeorge syndrome** [80][81]



CLINICAL MANIFESTATIONS

- Varying degrees of **cyanosis** depend on the amount of pulmonary blood flow.
- If not diagnosed at birth, the infant may develop signs of **CHF** as pulmonary vascular resistance decreases.
- The signs include **tachypnea** and **cough**.
- Peripheral pulses are usually bounding as a result of the diastolic runoff into the pulmonary arteries.
- A **single S₂** is due to the single valve.
- There may be a systolic ejection click.
- there is often a **systolic murmur** at the left sternal border.

- Cyanosis
- Respiratory distress
- Failure to thrive
- Cardiac examination findings
 - Bounding peripheral pulses
 - Harsh systolic murmur at the lower left sternal border
 - Loud S₂

Physical examination findings

1. **Systolic ejection murmur** at the base from increased flow across the truncal valve and a **single S₂** caused by the presence of only one atrioventricular (AV) valve
2. **Diastolic murmur** of flow across the mitral valve at the apex as a result of excessive pulmonary blood flow that returns to the left atrium
3. **High-pitched diastolic murmur** at the base indicates regurgitation of the truncal valve.

IMAGING STUDIES

- ECG findings include **combined ventricular hypertrophy** and **cardiomegaly**.
- **Increased pulmonary blood flow** is usually seen on chest x-ray. Pulmonary arteries may appear displaced.
- Echocardiography defines the anatomy, including the VSD, truncal valve function, and origin of the pulmonary arteries.

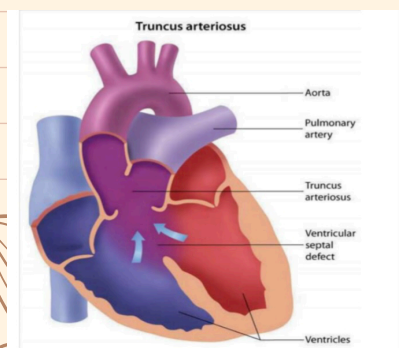
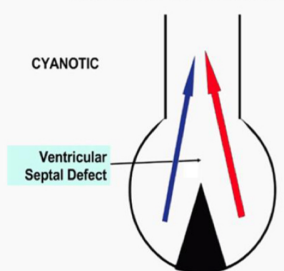
TREATMENT

- Medical management is usually needed and includes **anticongestive medications**.
- **Surgical repair** includes VSD closure and placement of a conduit between the right ventricle and pulmonary arteries.

Prognosis

- With surgical repair, the 30-year survival rate is ~ 70%. [85]

PERSISTENT TRUNCUS ARTERIOSUS



5th: Total anomalous pulmonary venous return:

1. **Definition.** TAPVR occurs when the pulmonary veins drain into the systemic venous side rather than into the left atrium. Sites of pulmonary venous drainage include supracardiac (into the right SVC or innominate vein), cardiac (into the right atrium or coronary sinus), and infracardiac (into the portal system and eventually into the inferior vena cava).
2. **Pathophysiology.** Systemic and pulmonary venous blood enters the right atrium and mixes together. As a result, this mixture of systemic and pulmonary venous blood is present in all four cardiac chambers (blood also passes across a PFO or ASD into the left side of the heart), pulmonary arteries, and aorta, resulting in desaturated systemic blood and visible cyanosis on examination.

ETIOLOGY AND EPIDEMIOLOGY

- Accounts for 1% of congenital heart disease.
- Disruption of the development of normal pulmonary venous drainage during the third week of gestation results in one of four abnormalities.
- Anatomically, all of the pulmonary veins fail to connect to the left atrium and return abnormally via the right heart.
- They may have **supracardiac, cardiac, infracardiac, or mixed drainage.**
- An atrial level communication is necessary for systemic cardiac output and survival.

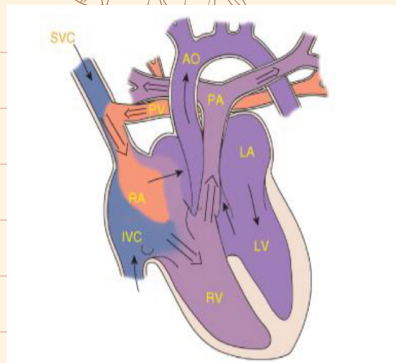
CLINICAL MANIFESTATIONS

- The most important determinant of presentation is the presence or absence of **obstruction to the pulmonary venous drainage.**
- Infants without obstruction have minimal cyanosis and may be asymptomatic.
- The pulmonary blood flow creates a **continuous murmur** and reenters the right atrium and right ventricle.
- There is a hyperactive **right ventricular impulse** with a **widely split S₂** (owing to increased volume ejected from the right ventricle)
- **systolic ejection murmur** at the left upper sternal border.
- Growth is relatively poor.
- Infants with **obstruction** present with cyanosis, marked tachypnea and dyspnea, and signs of right heart failure including hepatomegaly.
- The obstruction results in little, if any, increase in right ventricular volume.
- There may be no murmur or changes in S₂.

IMAGING STUDIES

- For infants without obstruction, the ECG is consistent with **right ventricular volume overload.**
- Cardiomegaly with increased pulmonary blood flow is seen on chest x-ray.
- Infants with obstructed veins have **right axis deviation** and **right ventricular hypertrophy** on ECG.
- On chest x-ray, the heart is normal or mildly enlarged with varying degrees of pulmonary edema that can appear similar to hyaline membrane disease or pneumonia.
- Echocardiography shows the volume-overloaded right heart, right-to-left atrial level shunting, and common pulmonary vein including site of drainage and degree of obstruction.

4. **Management.** Treatment is surgical repair shortly after diagnosis. The pulmonary veins are anastomosed to the back of the left atrium, and the PFO or ASD is closed.



TAPVR with and without Obstruction:

	Signs/symptoms	Diagnostic tests	Treatment
TAPVR with obstruction	• Early in life with respiratory distress and severe cyanosis • Severe pulmonary venous congestion and pulmonary hypertension with decreasing cardiac output and shock	• CXR shows pulmonary edema. • Echocardiography is definitive.	Surgery is the definitive choice for treatment.
TAPVR without obstruction	• Less likely to be severely symptomatic early • Age 1-2 years with right heart failure and tachypnea	• CXR shows snowman or figure 8 sign (large supracardiac shadow with an enlarged cardiac shadow). • Most accurate diagnostic test is an echocardiogram.	Surgical intervention to restore proper blood flow.



So, in general:

Cyanotic heart disease in newborns		
Diagnosis	Examination	X-ray findings
Transposition of the great vessels	• Single S2 • +/- VSD murmur	"Egg-on-a-string" heart (narrow mediastinum)
Tetralogy of Fallot	• Harsh pulmonic stenosis murmur • VSD murmur	"Boot-shaped" heart (right ventricular hypertrophy)
Tricuspid atresia	• Single S2 • VSD murmur	Minimal pulmonary blood flow
Truncus arteriosus	• Single S2 • Systolic ejection murmur (increased flow through truncal valve)	Increased pulmonary blood flow, edema
Total anomalous pulmonary venous return with obstruction	• Severe cyanosis • Respiratory distress	Pulmonary edema, "snowman" sign (enlarged supracardiac veins & SVC)

VSD = ventricular septal defect

Commonly associated conditions and risk factors of cyanotic congenital heart defects

	Most common associated conditions and risk factors
Tetralogy of Fallot	• DiGeorge syndrome • Down syndrome • Maternal risk factors ◦ Alcohol consumption ◦ Phenylketonuria ◦ Diabetes
Transposition of great vessels	• Maternal risk factors: diabetes
Tricuspid valve atresia	• Down syndrome
Ebstein anomaly	• Prenatal lithium exposure
Total anomalous pulmonary venous return	• Heterotaxy syndromes
Persistent truncus arteriosus	• DiGeorge syndrome
Hypoplastic left heart syndrome	• Trisomy 13 • Trisomy 18 • Turner syndrome • Jacobsen syndrome

By: Deema Muhanna

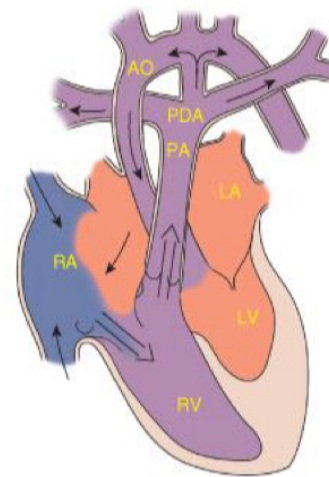
6th:hypoplastic It ventricular Syndrome

ETIOLOGY AND EPIDEMIOLOGY

- Hypoplastic left heart syndrome accounts for 1% of all congenital heart defects
- **the most common cause of death from cardiac defects in the first month of life.**
- Hypoplastic left heart syndrome occurs when there is failure of development of the mitral or aortic valve or the aortic arch.
- A small left ventricle that is unable to support normal systemic circulation is a central finding of hypoplastic left heart syndrome, regardless of etiology.
- Associated degrees of hypoplasia of the ascending aorta and aortic arch are present.
- Left-to-right shunting occurs at the atrial level.

CLINICAL MANIFESTATIONS

- After delivery, the infant is dependent on right-to-left shunting at the ductus arteriosus for systemic blood flow.
- As the ductus arteriosus constricts, the infant becomes critically ill with signs and symptoms of **CHF** from excessive pulmonary blood flow and obstruction of pulmonary venous return.
- Pulses are diffusely weak or absent.
- S₂ is single and loud.
- There is usually no heart murmur.
- Cyanosis may be minimal, but **low cardiac output** gives a grayish color to the cool, mottled skin.



IMAGING STUDIES

- ECG findings include **right ventricular hypertrophy** with **decreased left ventricular forces**.
- The chest x-ray reveals **cardiomegaly** (with right-sided enlargement) and pulmonary venous congestion or pulmonary edema.
- Echocardiography shows the small left heart, the degree of stenosis of the aortic and mitral valves, the hypoplastic ascending aorta, and the adequacy of left-to-right atrial flow.

TREATMENT

- Medical management includes **prostaglandin E₁** to open the ductus arteriosus, **correction of acidosis**, and ventilatory and blood pressure support as needed.
- **Surgical repair** is staged with the first surgery (Norwood procedure) done in the newborn period.
- Subsequent procedures create a systemic source for the pulmonary circulation (bidirectional Glenn and Fontan procedures), leaving the right ventricle to supply systemic circulation.
- There have been and continue to be many modifications to all three stages of the surgical repair.

7th: Ebstein Anomaly

Overview

- **Definition:** malformed tricuspid valve leaflets that are displaced into the right ventricle with subsequent tricuspid valve regurgitation and right heart enlargement (known as RV "**atrialization**")
- **Other cardiac defects associated with Ebstein anomaly**
 - Interatrial communication (e.g., PFO, ASD): seen in ~ 90% of patients ^[59]
 - **Conduction disorders** (e.g., Wolff-Parkinson-White syndrome, supraventricular tachycardia) ^[60]

Epidemiology

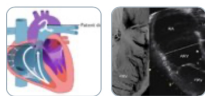
- Prevalence: 0.5–1/10,000 live births in the US ^[61]
- Accounts for < 1% of all CHD cases ^[16]

Etiology

- Multifactorial (most cases are sporadic) ^[62]
- Associated with prenatal **lithium** exposure ^{[63][64]}
- Isolated genetic defects (e.g., MYH7 which encodes β -myosin heavy chain)

Pathophysiology ^[59]

- **Incomplete delamination** (i.e., separation of valve tissue from the myocardium) during valve development can lead to one or more of the following: ^[62]
 - Incomplete closure of the right atrioventricular valve and reduction in RV volume → tricuspid regurgitation → **RA dilation** → right heart failure ^[62]
 - **RV atrialization** → reduced function of the RV → insufficient systolic contraction → functional pulmonary valve atresia → ↓ pulmonary flow → ↓ cardiac output → systemic hypoperfusion, dyspnea, and fatigue
 - Bulging of the large **sail-like anterior tricuspid leaflet** into the RV → RVOTO → ↑ right heart pressure → ↑ flow through the patent foramen ovale → right-to-left shunt → cyanosis

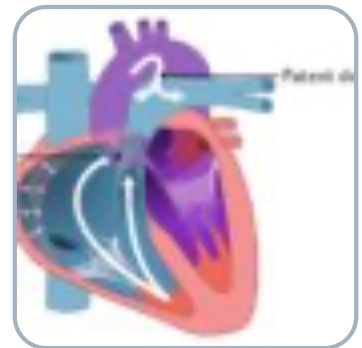


Clinical findings ^[59]

- The clinical presentation of Ebstein anomaly varies according to the severity of the abnormality. ^[65]
 - Mild apical displacement → asymptomatic presentation throughout adulthood
 - Moderate tricuspid leaflet displacement → cyanosis or heart failure in infancy or childhood
 - Severe displacement → in utero heart failure → nonimmune hydrops fetalis → death
- Neonates and children
 - Cyanosis
 - Severe cardiomegaly and heart failure
 - Respiratory distress
- Adolescents and adults
 - **Arrhythmias** e.g., Wolff-Parkinson-White syndrome ^[66]
 - Exertional dyspnea and fatigue
 - Palpitations and sudden cardiac death
- Cardiac examination findings
 - Loud S1 ^[66]
 - Widely split S1 and S2 ^[66]
 - Holosystolic murmur at the left sternal border ^[66]

Diagnostics

- **Prenatal diagnosis:** fetal echocardiography
- **Imaging**
 - **Echocardiography** (confirmatory test)
 - Enlarged right heart
 - Tricuspid regurgitation ^[66]
 - Chest x-ray ^[66]
 - Mild to severe cardiomegaly
 - RA enlargement
 - MRI ^[66]: apical displacement of the septal and posterior tricuspid leaflets
- **ECG**
 - Tall and broad P waves ^[66]
 - First-degree atrioventricular block ^[66]
 - Right axis deviation ^[66]
 - Right bundle branch block
 - Findings specific for WPW syndrome (e.g., delta wave)
- **Pulse oximetry:** ↓ SpO₂



Treatment

- **Initial postnatal management:** stabilization of cardiorespiratory status until PVR decreases and surgery can be performed
 - PGE1 infusion ^[66]
 - Inhaled nitric oxide or infusion of milrinone ^[66]
- **Surgical repair**
 - Objective: creation of a systemic-pulmonary shunt, repair of the tricuspid valve, and reconstruction of the right ventricle
 - Age-related considerations
 - Neonates and infants: should be avoided in infancy and delayed as long as possible ^[66]
 - Children and adults: recommended if concomitant heart failure or progressive RV dysfunction is present ^[67]
- **Arrhythmias:** catheter ablation of the aberrant pathway ^[68]

Prognosis

- Varies according to the severity of disease
- Around 30% of patients die within the first year of life. ^[65]

FEEDBACK

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