CONGENITAL HEART DISEASE

Overview and scope of disease burden in Uganda

Management, treatment and surgery

Emmanuel Tenywa, Paediatric cardiologist JRRH

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Introduction

- Congenital heart disease, or a congenital heart defect, is a heart abnormality present at birth.
- The problem can affect:
  - the heart walls
  - the heart valves
  - the blood vessels
- Can range from simple conditions that don't cause symptoms to complex problems that cause severe, life-threatening symptoms.

Ugandan situation

- 1.6million children are born every year (UDHS 2016).
- 1% (16,000 children)suffer from CHD.
- 50% (8000 children) have a defect serious enough to require an intervention.
- Burden of CHD in Uganda.

Pattern of congenital heart disease among children presenting to the Uganda Heart Institute, Mulago Hospital: a 7-year review.

(Judith Namuyonga *et al*)

African Health Sciences Vol 20 Issue 2, June, 2020

Table 1: Acyanotic heart diseases

	Number	Overall Percentage in CHD (%) N=3526	Mean age (months)/years	Female n (%)
Isolated VSD	921	26	25(2)	484 (52)
PDA	760	22	19(1.6)	478 (62)
ASD	332	9.4	51(4)	188 (56)
ECD	265	8	17(1)	164(62)
Pulmonary valve stenosis	226	6	38(3)	99(44)
Mitral valve prolapse	63	2	90(7.5)	44(69)
Aortic valve stenosis	35	0.9	100(8)	14(40)
PAPVC	17	0.5	46 (4)	5(29)
COA	14	0.4	80(6.5)	7(50)

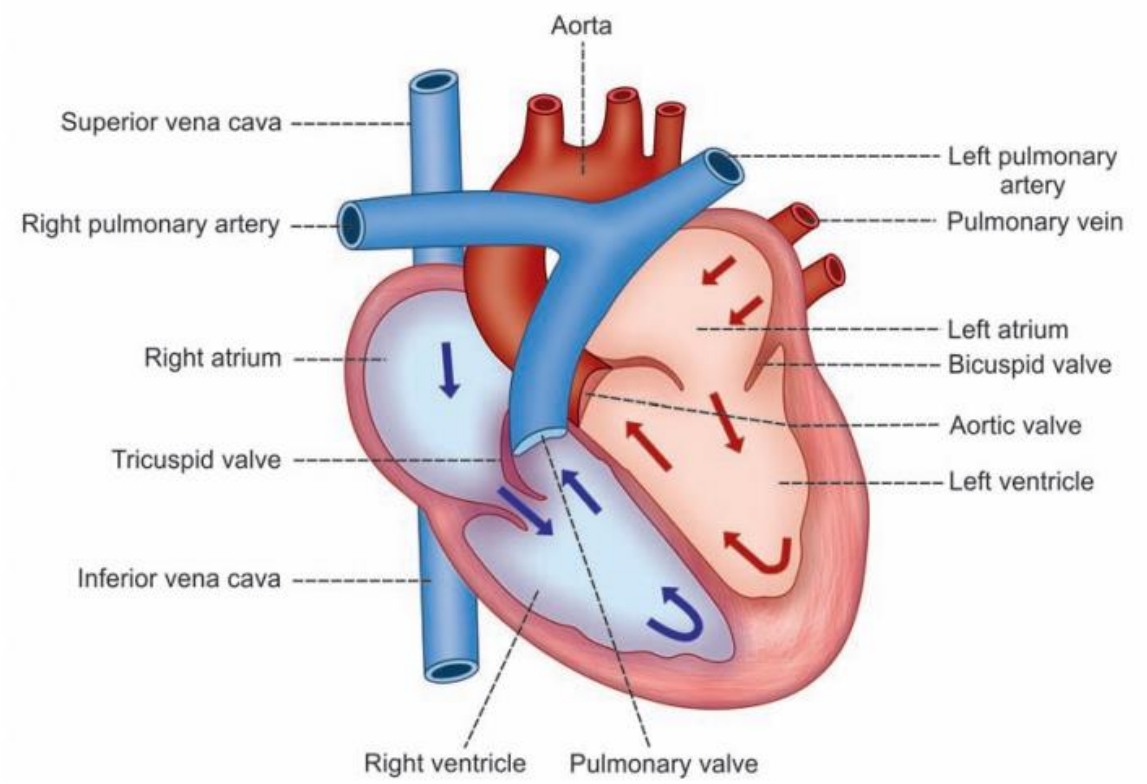
ECD- endocardial cushion defects, VSD- Ventricular septal defect, ASD-Atrial septal defect, PDA-Patent ductus arteriosus, PAPVC- partial anomalous pulmonary connections, COA- coarctation of the aorta

Table 2: Cyanotic heart diseases

Lesion	Numbers	Mean age Years (months)	Female Number (%)	Overall percentage in CHD (N=3526)
Tetralogy of Fallot	247	4(50)	110(44)	**7**
Truncus arteriosus	165	0.4 (5)	92(56)	**5**
DORV	104	1.4(16.5)	56(53)	**3**
Pulmonary atresia	71	2.6(32)	38(53)	**2**
Tricuspid atresia	62	1.6(20)	31(50)	**1.8**
D TGA	53	0.7(9.5)	21 (40)	**1.5**
A PVR	8	0.5(7)	4(50)	**0.2**

D TGA-Transposition of the Great Arteries, APVR- anomalous pulmonary venous return, DORV- Double outlet right ventricle

Normal anatomy of the heart



Classification of CHD

- Acyanotic CHD
- Shunt lesions(volume load lesions)
- Obstructive lesions(pressure load lesions)
- Cyanotic CHD
- Cyanotic Congenital Heart Disease: Lesions Associated With Decreased Pulmonary Blood Flow
- Cyanotic Congenital Heart Disease: Lesions Associated With Increased Pulmonary Blood Flow

Cyanotic Congenital Heart Diseases are classified into two:

A. Cyanotic Congenital Heart Disease: Lesions Associated With Decreased Pulmonary Blood Flow

1. Tetralogy of fallot
2. Pulmonary Atresia With Intact Ventricular Septum

3 Tricuspid Atresia

4 Double outlet right ventricle

5 Transposition of the Great Arteries With Ventricular Septal Defect and Pulmonary Stenosis

6 Ebstein Anomaly of the Tricuspid Valve

B. Cyanotic Congenital Heart Disease: Lesions Associated With Increased Pulmonary Blood Flow

1. Transposition of the Great Arteries.
2. Double-Outlet Right Ventricle Without Pulmonary Stenosis.
3. Double-Outlet Right Ventricle With Malposition of the Great Arteries (Taussig-Bing Anomaly).
4. Total Anomalous Pulmonary Venous Return.
5. Truncus Arteriosus.
6. Single Ventricle (Double-Inlet Ventricle, Univentricular Heart).
7. Hypoplastic Left Heart Syndrome.

ACYANOTIC CONGENITAL HEART LESIONS

- Lesions Resulting in Increased Volume Load

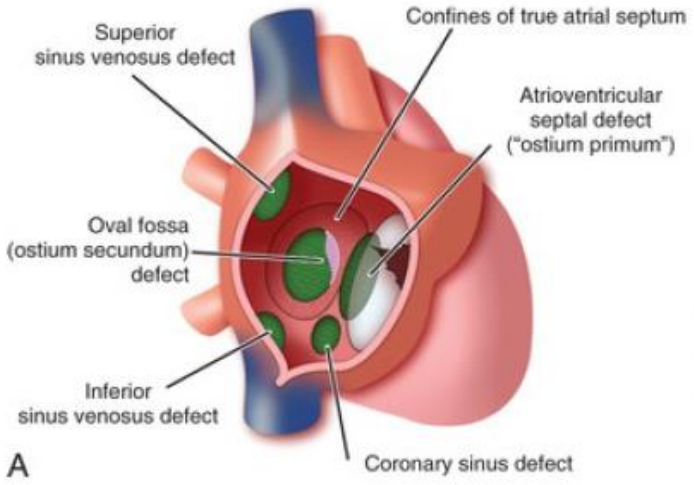
- Those that cause left-to-right shunting:
  - Atrial septal defect (ASD)
  - Ventricular septal defect (VSD),
  - Atrioventricular septal defects
  - Patent ductus arteriosus.

Lesions Resulting in Increased Pressure Load

- Increased pressure load is an obstruction to normal blood flow.
- Valvular pulmonic stenosis
- Valvular aortic stenosis
- Coarctation of the aorta.

Atrial Septal Defect

- Most common form of ASD.
- Can occur in any portion of the atrial septum—secundum, primum, or sinus venosus.
- Commonly occurs in the central part of the atrial septum in the region of fossa ovalis (secundum ASD).
- Majority of cases of ASD are sporadic.
- Autosomal dominant inheritance does occur as part of the Holt-Oram syndrome.


Superior
sinus venosus defect
Confines of true atrial septum
Atrioventricular
septal defect
("ostium primum")
Oval fossa
(ostium secundum)
defect
Inferior
sinus venosus defect
Coronary sinus defect
A

CLINICAL MANIFESTATIONS

- Most patients with ASD are asymptomatic and may remain undiagnosed until later in life.
- Even an extremely large secundum ASD rarely produces clinically evident heart failure in childhood.
- On closer evaluation, however, younger children may show **subtle failure to thrive**, and older children may have varying degrees of **exercise intolerance.**

DIAGNOSIS

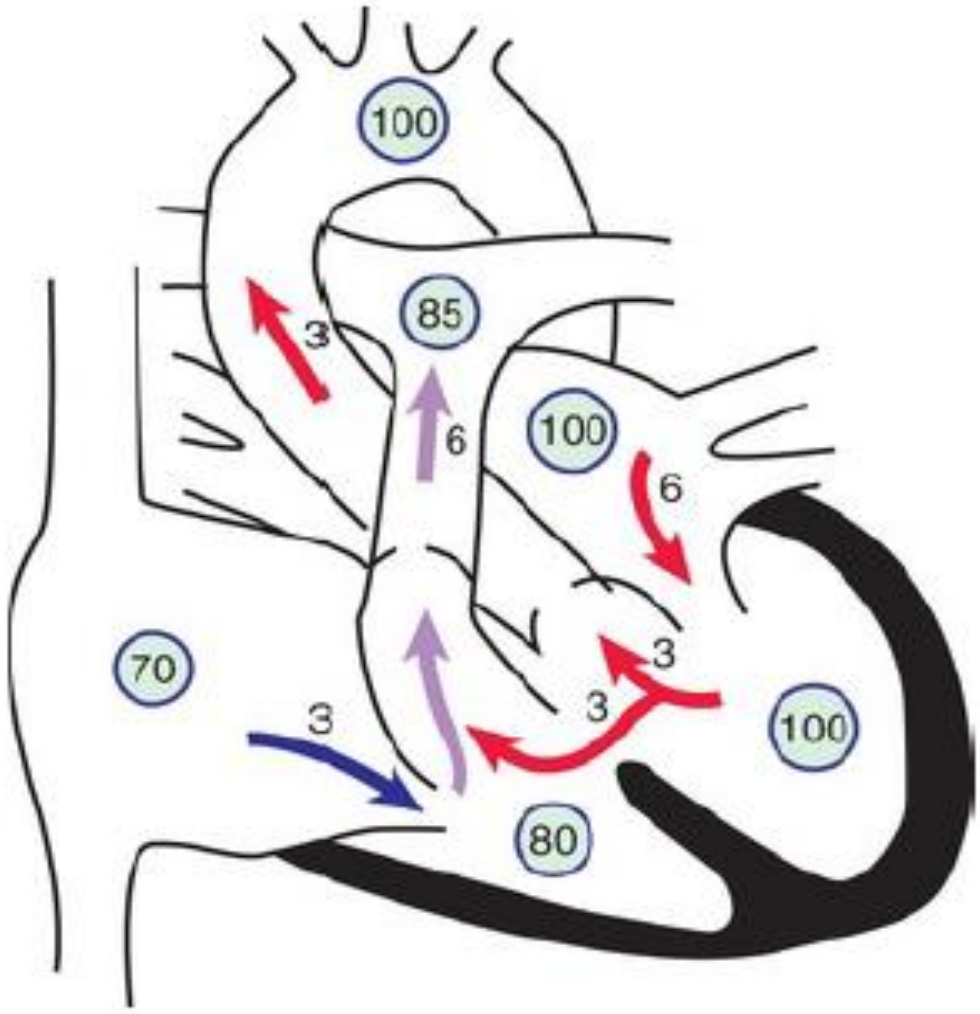
- The chest radiograph shows varying degrees of enlargement of the right ventricle and atrium, depending on the size of the shunt. The pulmonary artery is enlarged, and pulmonary vascularity is increased.
- The echocardiogram is instrumental in defining
- the type of ASD
- its size and shape
- the degree and direction of shunting
- its effect on the right -sided chambers of the heart ,associated lesions, and estimations of right ventricular pressure.

MANAGEMENT

- Most children with an ASD are asymptomatic .
- In rare cases when they are symptomatic, anti congestive therapy with diuretics may be indicated until closure is accomplished.
- Closure of an ASD is indicated if there is a large shunt, that is, Qp:Qs≥1.5

- ✓ Medical management is rarely indicated
- ✓ If significant shunt is still present at around **3 years of age**, closure is usually recommended:
  - Secundum can be closed with catheterization
  - Primum and sinus venous defects: surgical closure

Ventricular septal Defect



- Ventricular septal defect is the most common cardiac malformation.
- Accounts for 25% of congenital heart disease.
- Defects may occur in any portion of the ventricular septum
- Most common are of the membranous type
- VSDs in the mid portion or apical region of the ventricular septum are muscular in type and may be single or multiple (“Swiss cheese” septum)

Classification of VSD

- Membranous
- Inlet Atrioventricular septal defect type
- Muscular
- Supracristal VSD

❖ CLINICAL MANIFESTATIONS

Vary according

- to the size of the defect
- pulmonary blood flow and pressure.

- Small VSDs with trivial left-to-right shunts and normal pulmonary artery pressure (PAP) are the most common.
- These patients are asymptomatic, and the cardiac lesion is usually found during routine physical examination.
- Characteristically, a loud, harsh, or blowing holosystolic murmur is present and heard best over the lower left sternal border.

- Large VSDs with excessive pulmonary blood flow and pulmonary hypertension are responsible for signs of congestive heart failure: dyspnea, feeding difficulties, poor growth, profuse perspiration, and recurrent pulmonary infections in early infancy.

Physical exam

- Infants with large VSDs initially appear quite well .
- As the left -to-right shunt becomes significant , the infant may develop a wasted appearance with diminished subcutaneous tissue.
- Tachypnea and increased work of breathing with subcostal retractions will ensue.
- On cardiac examination , there is an active precordium ,which extends over both the right (parasternal) and the left ventricular (apical) areas.

Diagnosis

- **Small**
- **ECG : LV** hypertrophy
- **Chest X Ray:** Normal or minimal cardiomegaly and a borderline increase in pulmonary vasculature

- **Large**
- **ECG: Biventricular hypertrophy**,)
- **Chest X Ray:**
- Gross cardiomegaly with prominence of both ventricles, left atrium, and pulmonary artery
- Pulmonary vascular marking are increased, frank pulmonary edema, pleural effusions
  - **Echocardiogram**
    1. Confirms the diagnosis
    2. Estimate shunt size

Treatment

Small VSDs

- ✓ Muscular VSDs: are most likely to close up to 80% than membranous VSDs up to 35% during the first 2 years of life
- ✓ Encourage to live a normal life, with no restrictions on physical activity
- ✓ Surgical repair is not recommended
- ✓ Endocarditis prophylaxis is no longer recommended for dental visit and surgical procedures.

- Moderate to Large VSDs symptomatic patients

✓ Normalize growth

- Nutritional support (120kcal/Kg to 150Kcal/Kg /day or more)

✓ Control of congestive heart failure (depending on severity)

- Oxygen
- Diuretics: Mainstay of the therapy
  - Furosemide 1 – 2mg/Kg/dose every 2 to 3 times in a day
  - ACEI/ARBs
  - Spirinolactone 3mg/Kg
- IV Inotropics in severe cases
- Antibiotics if pneumonia

Surgical treatment

1. Large defects in whom clinical symptoms and failure to thrive cannot be controlled medically
2. Moderate to large defects associated with pulmonary hypertension (between 6 and 12 mo of age)
3. Older than 24 mo with a Qp:Qs ratio greater than 2:1
4. All children with Supracristal VSD

Patent Ductus Arteriosus

- Connection between the descending aorta and pulmonary artery.
- A small PDA is usually asymptomatic and is usually diagnosed by the presence of a heart murmur.
- A large PDA will result in heart failure similar to that encountered in infants with a large VSD.
- Retardation of physical growth may be a major manifestation in infants with large shunts.
- The classic continuous murmur is described as “machinery-like” in quality.

Diagnosis

Small PDA: Normal ECG and Chest X Ray

Moderate to large shunt:

Chest X Ray: Cardiomegaly, increased pulmonary vascularity

ECG: normal or evidence of LVH.

if pulmonary hypertension: right ventricular hypertrophy

Treatment of PDA

✓Full-term or preterm newborn?

- ✓ **Full-term newborns:** spontaneous closure **after a few weeks** is uncommon
- ✓ **Small PDAs:** elective closure of (audible or silent) and hemodynamically insignificant is controversial
- ✓ **Moderate and large PDAs:** initially manage with diuretics, but closure with catheterization

- ✓ **Preterm newborns**
- **Indomethacin or ibuprofen: likely to be effective before the age of 2 weeks**

- < 48-hr-old: orally every 12 – 24 hours for three doses
  - 1st dose: 0.2 mg/kg/dose
  - 2nd dose and 3rd dose 0.1mg/kg/dose
- > 7-day-old: 0.25mg/kg/dose

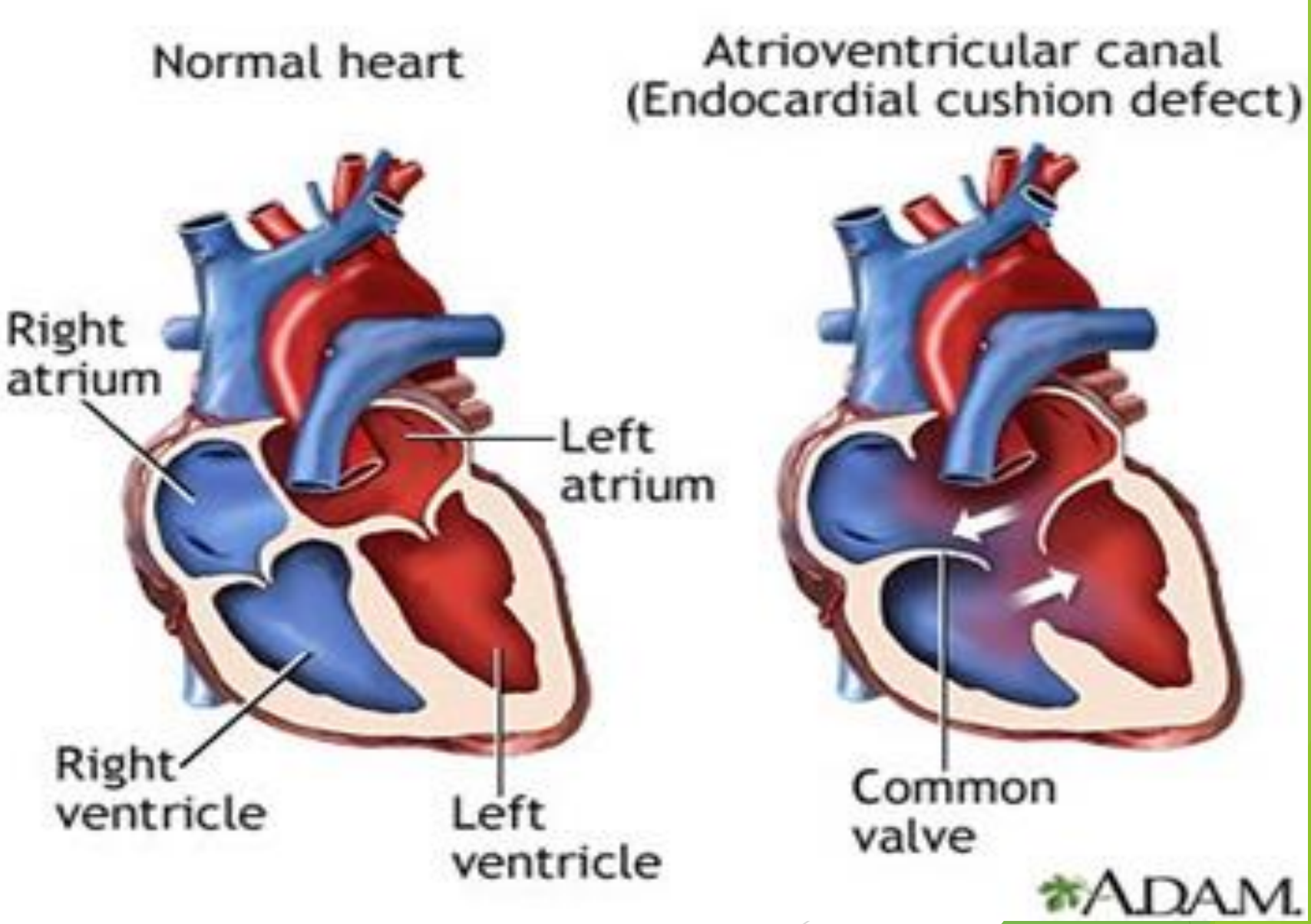
- Closure with catheterization if newborns don't respond to the treatment

Surgical treatment

- ✓ Moderate and large PDA
- ✓ Preterm newborns doesn’t respond to treatment
- ✓ Prior episodes of endocarditis

Atrioventricular (AV) septal defects

- Represent a spectrum of a basic embryologic abnormality, a deficiency of the AV septum.
- The tricuspid valve sits slightly lower (more toward the cardiac apex) than does the mitral valve, and thus a small portion of septum separates the left ventricle from the right atrium.


Normal heart
Atrioventricular canal
(Endocardial cushion defect)
Right
atrium
Left
atrium
Right
ventricle
Left
ventricle
Common
valve
A.D.A.M.

- An AV septal defect, formerly known as an AV canal defect or endocardial cushion defect,consists of a defect of the AV septum and contiguous atrial and ventricular septal defects with a common AV valve.
- Complete AV septal defect is common in children with Down syndrome

- The echocardiogram is diagnostic and shows signs of RV enlargement .
- There is encroachment of the mitral valve into the left ventricular outflow tract; the abnormally low position of the AV valves results in a “gooseneck” deformity of the LVOT.

Treatment

- Surgical treatment of complete AV septal defects is more complex, although highly successful.
- Full correction of these defects can be readily accomplished in infancy.

CYANOTIC CONGENITAL HEART DISEASE

- Congenital heart disease (CHD) produces cyanosis when obstruction to right ventricular inflow or outflow causes intracardiac right-to-left shunting or when complex anatomic defects cause an admixture of pulmonary (deoxygenated) and systemic (oxygenated) venous return in the heart.

- Cyanosis may be caused by persistence of fetal pathways, such as right-to-left shunting across the foramen ovale and ductus arteriosus in the presence of pulmonary outflow tract obstruction or **persistent pulmonary hypertension of the newborn (PPHN).**

Cyanotic Congenital Heart Diseases are classified into two:

A. Cyanotic Congenital Heart Disease: Lesions Associated With Decreased Pulmonary Blood Flow

1. Tetralogy of fallot
2. Pulmonary Atresia With Intact
3. Ventricular Septum
4. Tricuspid Atresia
5. Double outlet right ventricle
6. Transposition of the Great Arteries With Ventricular Septal Defect and Pulmonary Stenosis
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TRANSPOSITION OF THE GREAT ARTERIES

- It is the most common cyanotic lesion to present in the newborn period.
- it is due to ventriculoarterial discordance secondary to abnormalities of septation of the truncus arteriosus.
- **The aorta arises from the right ventricle, and pulmonary artery arises from the left ventricle. This results in:**
  - ✓ Desaturated blood returning to the right side of the heart and being pumped back out to the body,
  - ✓ While well-oxygenated blood returning from the lungs is pumped back to the lungs.
  - ✓ Without mixing of the two circulations, death occurs quickly
-
- Mixing can occur at the ASD, VSD & PDA

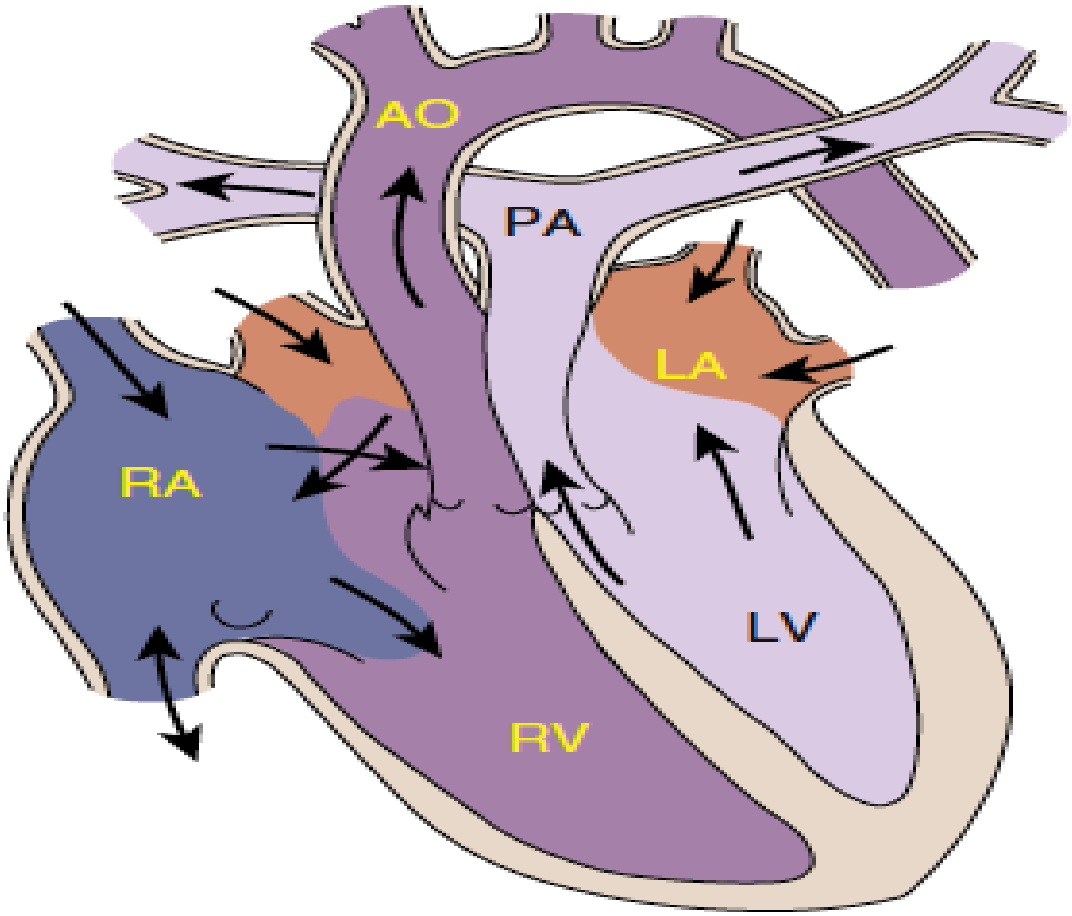


Figure 144-2 Transposition of the great vessels. *AO*, Aorta; *LA*, left atrium; *LV*, left ventricle; *PA*, pulmonary artery; *RA*, right atrium; *RV*, right ventricle.

- Transposition of the great arteries, or vessels, a common cyanotic congenital anomaly, accounts for approximately 5% of all congenital heart disease.

- In this anomaly, the systemic veins return normally to the right atrium and the pulmonary veins return to the left atrium.

- The connections between the atria and ventricles are also normal (**atrioventricular concordance**).

- The aorta arises from the right ventricle and the pulmonary artery from the left ventricle. In normally related great vessels, the aorta is posterior and to the right of the pulmonary artery.

- In d-transposition of the great arteries (**d-TGA**), the aorta is anterior and to the right of the pulmonary artery (the *d* indicates a dextropositioned aorta, *transposition* indicates that it arises from the anterior right ventricle).

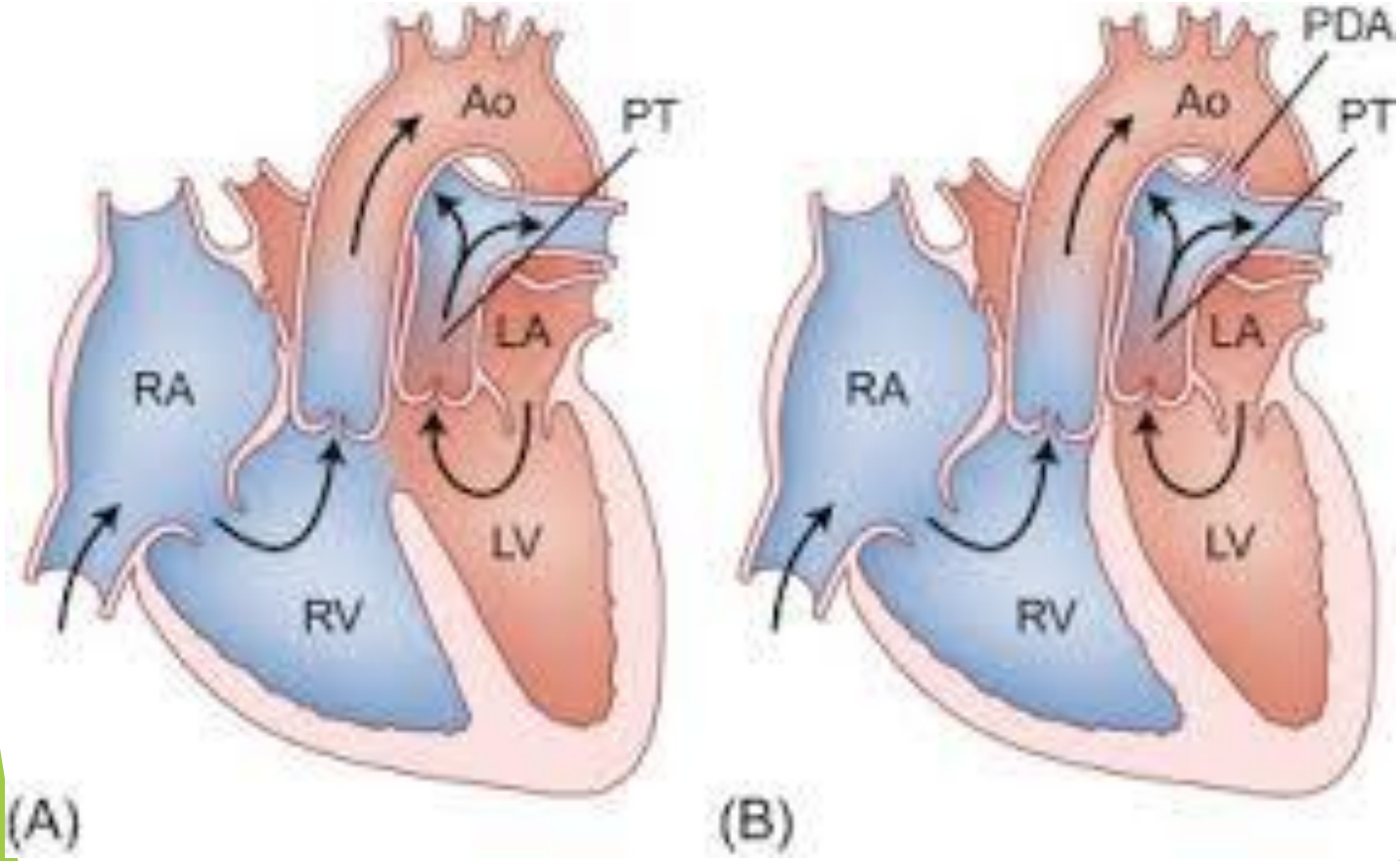
- Desaturated blood returning from the body to the right side of the heart goes inappropriately out the aorta and back to the body again, whereas oxygenated pulmonary venous blood returning to the left side of the heart is returned directly to the lungs. Thus the systemic and pulmonary circulations exist as 2 parallel circuits.

- Survival in the immediate newborn period is provided by the foramen ovale and the ductus arteriosus, which permit some mixture of oxygenated and deoxygenated blood.

D-Transposition of the Great Arteries With Intact Ventricular Septum

- d-TGA with an intact ventricular septum is also referred to as **simple TGA** or **isolated TGA**.

- Before birth, oxygenation of the fetus is only slightly abnormal, but after birth, once the ductus arteriosus begins to close, the minimal mixing of systemic and pulmonary blood by the patent foramen ovale is usually insufficient, and severe hypoxemia ensues, generally within the 1st few days of life.


Ao
PT
LA
RA
LV
RV
(A)
PDA
Ao
PT
LA
RA
LV
RV
(B)

CLINICAL MANIFESTATIONS

- Cyanosis and tachypnea are most often recognized within the first hours or days of life. Untreated, most of these infants would not survive the neonatal period.

- Hypoxemia is usually moderate to severe, depending on the degree of atrial level shunting and whether the ductus is partially open or totally closed.

- This condition is a medical emergency, and only early diagnosis and appropriate intervention can avert the development of prolonged severe hypoxemia and acidosis, which lead to death.

- Physical findings, other than cyanosis, may be remarkably nonspecific.
- The precordial impulse may be normal, or a parasternal heave may be present.
- The second heart sound (S2) is usually single and loud, although it may be split.
- Murmurs may be absent, or a soft systolic ejection murmur may be noted at the mid-left sternal border.

DIAGNOSIS

- The electrocardiogram (ECG) is usually normal, showing the expected neonatal right-sided dominant pattern.
- Chest radiographs may show mild cardiomegaly, a narrow mediastinum (the classic “egg-shaped heart”), and normal to increased pulmonary blood flow.
- In the early newborn period, the chest radiograph is generally normal. As pulmonary vascular resistance (PVR) drops during the first several weeks of postnatal life, evidence of increased pulmonary blood flow becomes apparent.
- Arterial blood partial pressure of oxygen (PaO2) is low and does not rise appreciably after the patient breathes 100% oxygen (hyperoxia test), although this test may not be totally reliable.

- Echocardiography is diagnostic and confirms the transposed ventricular-arterial connections.
- The size of the interatrial communication and the ductus arteriosus can be visualized and the degree of mixing assessed by pulsed and color Doppler examination.
- The presence of any associated lesion, such as left ventricular outflow tract obstruction or a VSD, can also be assessed.
- The origins of the coronary arteries can be imaged, although echocardiography is generally not as accurate as catheterization for this purpose.

- Cardiac catheterization may be performed in patients for whom noninvasive imaging is diagnostically inconclusive, where an unusual coronary artery anomaly is suspected, or in patients who require emergency balloon atrial septostomy (**Rashkind procedure**).

TREATMENT

- When transposition is suspected, an infusion of prostaglandin E1 (PGE1; 0.01-0.20 µg/kg/min) should be initiated immediately to maintain patency of the ductus arteriosus and improve oxygenation.

- Because of the risk of apnea associated with prostaglandin infusion, an individual skilled in neonatal endotracheal intubation should be available.

- Hypothermia intensifies the metabolic acidosis resulting from hypoxemia, and thus the patient should be kept warm.

- Prompt correction of acidosis and hypoglycemia is essential.

- Infants who remain severely hypoxic or acidotic despite prostaglandin infusion should undergo **Rashkind balloon atrial septostomy**.

- A Rashkind atrial septostomy is also usually performed in all patients in whom any significant delay in surgery is necessary.

- If surgery is planned during the 1st 2 wk of life, and the patient is stable, catheterization and atrial septostomy may be avoided.

- Some patients with TGA and VSD may require balloon atrial septostomy because of poor mixing, even though the VSD is large.

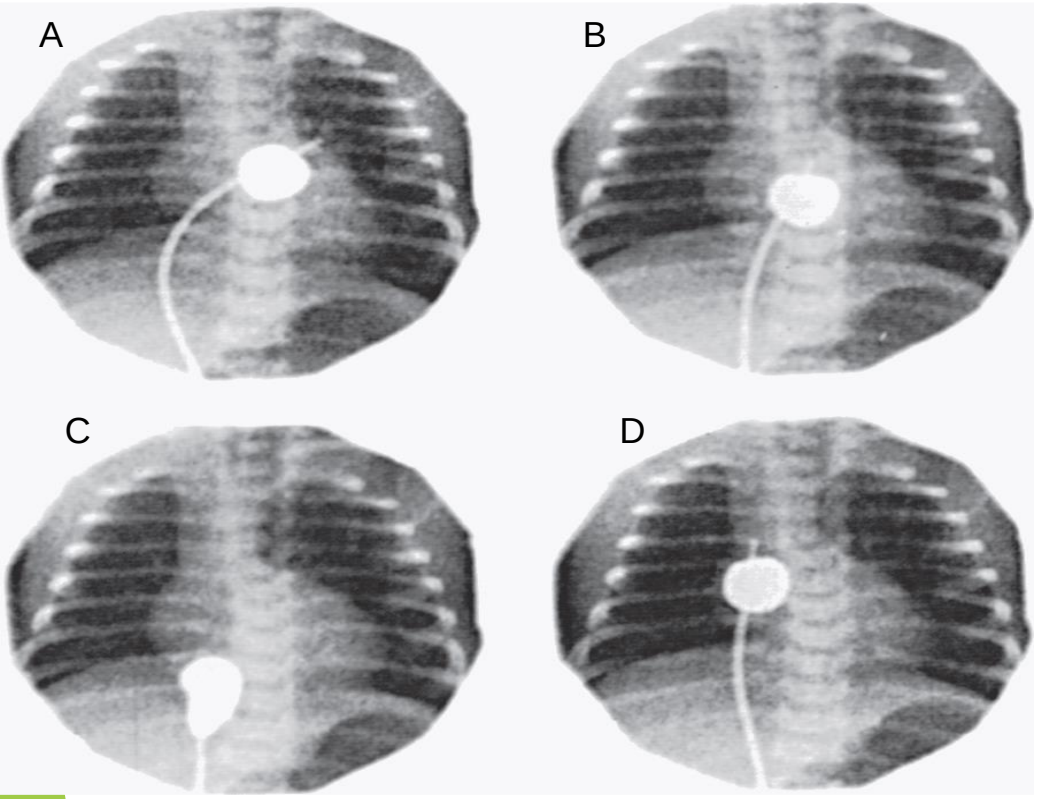
- The **arterial switch (Jatene) procedure** is the surgical treatment of choice for neonates with d-TGA and an intact ventricular septum and is usually performed within the first 2 weeks of life.

- The reason for this time frame is that as PVR declines after birth, pressure in the left ventricle (connected to the pulmonary vascular bed) also declines.

- This pressure drop results in a decrease in left ventricular (LV) mass over the first few weeks of life.

- If the arterial switch operation is attempted after LV pressure (and mass) has declined too far, the left ventricle will be unable to generate adequate pressure to pump blood to the high-pressure systemic circulation.

Rashkind balloon atrial septostomy



- Four frames from a continuous cineangiogram show the creation of an atrial septal defect in a hypoxemic newborn infant with transposition of the great arteries and an intact ventricular septum.

A. Balloon inflated in the left atrium.

B. The catheter is jerked suddenly so that the balloon ruptures the foramen ovale.

C. Balloon in the inferior vena cava.

D. Catheter advanced to the right atrium to deflate the balloon. The time from A to C is <1 sec.

Transposition of the Great Arteries With Ventricular Septal Defect

- If the VSD associated with d-TGA is small, the clinical manifestations, laboratory findings, and treatment are similar to those described previously for transposition with an intact ventricular septum.
- A harsh systolic murmur is audible at the lower left sternal border, resulting from flow through the defect.
- Many of these small defects eventually close spontaneously and may not be addressed at the time of surgery.
- When the VSD is large and not restrictive to ventricular ejection, significant mixing of oxygenated and deoxygenated blood usually occurs and clinical manifestations of cardiac failure are seen.

- cyanosis may be subtle and sometimes may not be recognized until an oxygen saturation measurement is performed.

- The murmur is holosystolic and generally indistinguishable from that produced by a large VSD in patients with normally related great arteries. The heart is usually significantly enlarged.

- Cardiomegaly, a narrow mediastinal waist, and increased pulmonary vascularity are demonstrated on the chest radiograph. The ECG shows prominent P waves and isolated right ventricular hypertrophy or biventricular hypertrophy.

- Occasionally, dominance of the left ventricle is present. Usually, the QRS axis is to the right, but it can be normal or even to the left.

- The diagnosis is confirmed by echocardiography, and the extent of pulmonary blood flow can also be assessed by the degree of enlargement of the left atrium and ventricle.

- In equivocal cases, the diagnosis can be confirmed by cardiac catheterization. Right and left ventriculography indicate the presence of arterial transposition and demonstrate the site and size of the VSD.

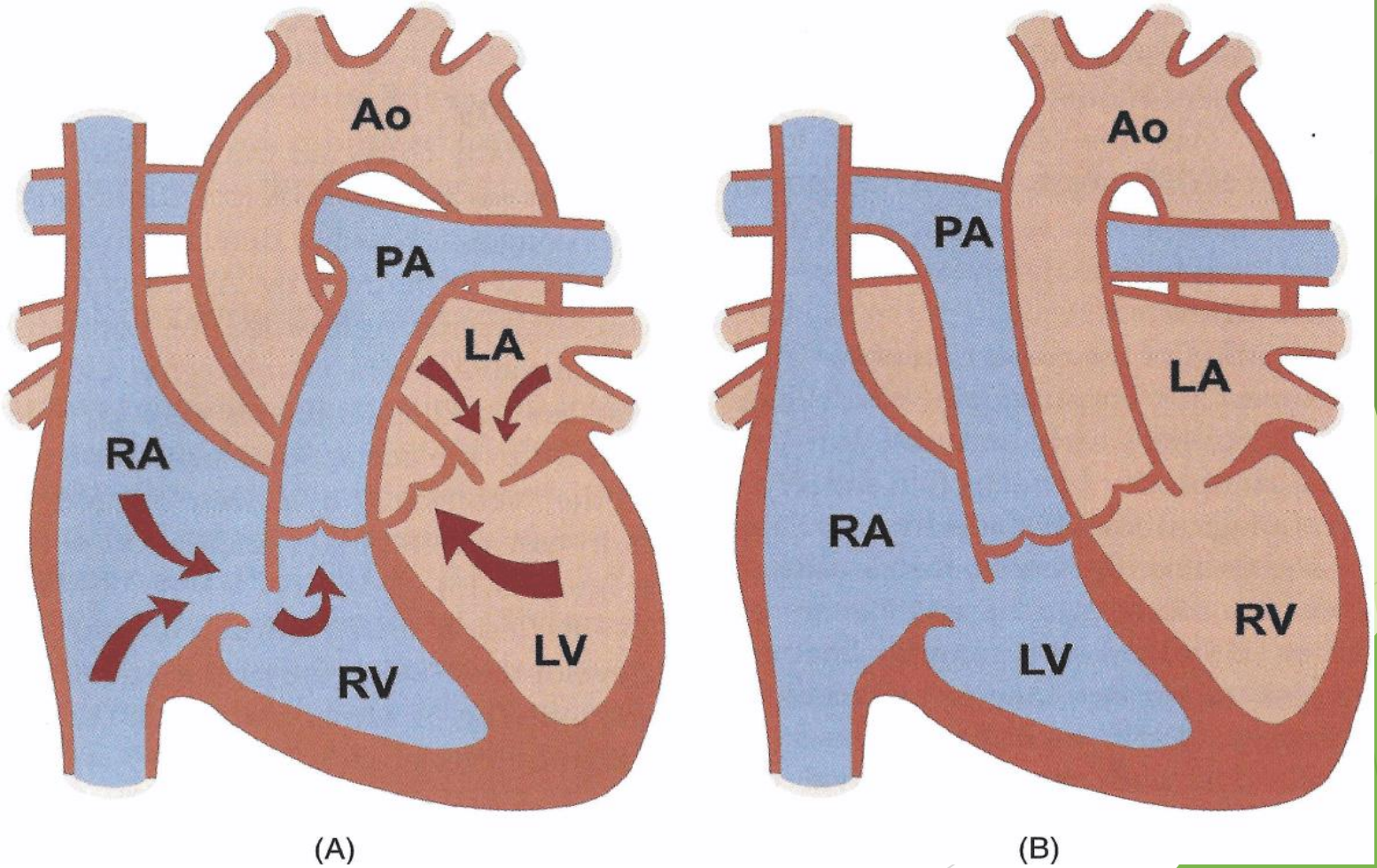
- Systolic pressure is equal in the 2 ventricles, the aorta, and pulmonary artery. Left atrial pressure may be much higher than right atrial pressure, a finding indicative of a restrictive communication at the atrial level.

- At the time of cardiac catheterization, Rashkind balloon atrial septostomy may be performed to decompress the left atrium, even when adequate mixing is occurring at the ventricular level.

- Surgical treatment is advised soon after diagnosis, because heart failure and failure to thrive are difficult to manage and pulmonary vascular disease can develop unusually rapidly in these patients.

- Preoperative management with diuretics lessens the symptoms of heart failure and stabilizes the patient before surgery.

L-Transposition of the Great Arteries



- In l-transposition (l-TGA), the atrioventricular relationships are discordant: the right atrium is connected to a left ventricle and the left atrium to a right ventricle (also known as **ventricular inversion**).

- The great arteries are also transposed, with the aorta arising from the right ventricle and the pulmonary artery from the left.

- In contrast to d-TGA, the aorta arises to the left of the pulmonary artery (thus the designation*l* for levo-transposition).

- The aorta may be anterior to the pulmonary artery, although often they are nearly side by side.

- The physiology of l-TGA is quite different from that of d-TGA.

- Desaturated systemic venous blood returns via the vena cavae to a normal right atrium, from which it passes through a bicuspid atrioventricular (mitral) valve into a right-sided ventricle that has the architecture and smooth wall morphologic features of the normal left ventricle.

- Because transposition is also present, however, the desaturated blood ejected from this left ventricle enters the transposed pulmonary artery and flows into the lungs, as it would in the normal circulation.

Treatment

- Surgical treatment of the associated anomalies, most often the VSD, is complicated by the position of the bundle of His, which can be injured at surgery and result in heart block.

- Identification of the usual course of the bundle in corrected transposition (running superior to the defect) has been accomplished by mapping of the conduction system so that the surgeon can avoid the bundle of His during repair.

- Even without surgical injury, patients with l-TGA are at risk for heart block as they grow older.

- Because simple surgical correction leaves the right ventricle as the systemic pumping chamber, and thus vulnerable to late ventricular failure, surgeons have become more aggressive about trying operations that utilize the left ventricle as the systemic pumping chamber.

- This is accomplished by performing an atrial switch operation, to reroute the systemic and pulmonary venous returns, in combination with an arterial switch operation to reroute the ventricular outflows (**double switch procedure**).

- The long-term benefit of this approach in preserving systemic ventricular function is still under investigation.

Total Anomalous Pulmonary Venous Return

- All of the pulmonary veins fail to connect to the left atrium and return abnormally via the right side of the heart.
  - ✓ They may have supracardiac , infracardiac , cardiac , or mixed drainage .
- Infants without obstruction have minimal cyanosis and may be asymptomatic.
- Infants with obstruction present with cyanosis, marked tachypnea and dyspnea, and signs of right-sided heart failure including hepatomegaly.

Total Anomalous Pulmonary Venous Return

Classification according to connection site	Total Anomalous Pulmonary Venous Return
SITE OF CONNECTION (% OF CASES)	% WITH SIGNIFICANT OBSTRUCTION
Supracardiac (50)	
Left superior vena cava (40)	40
Right superior vena cava (10)	75
Cardiac (25)	
Coronary sinus (20)	10
Right atrium (5)	5
Infracardiac (20)	95-100
Mixed (5)	

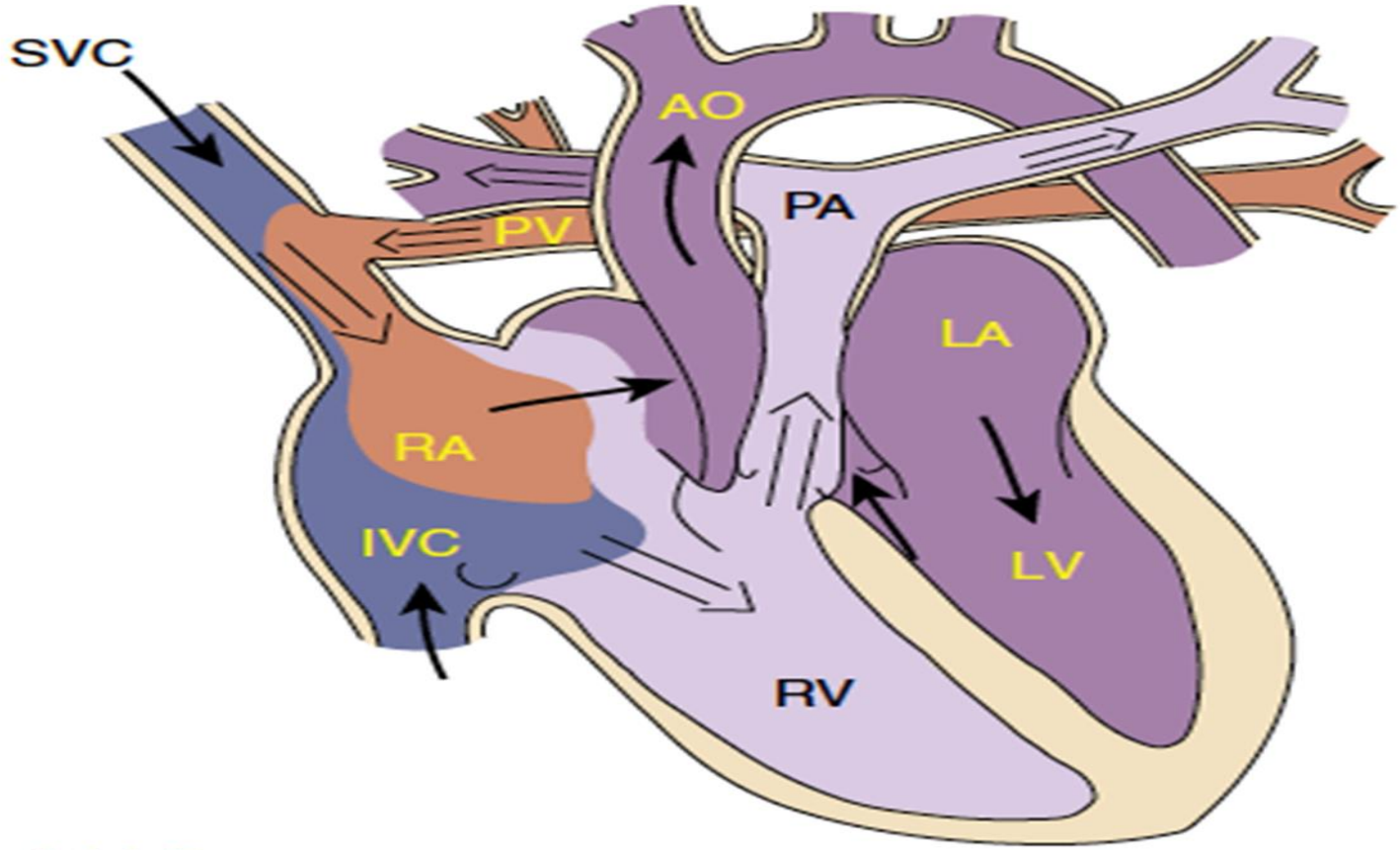


Figure 144-5 Total anomalous pulmonary venous return. *AO,* Aorta; *IVC,* inferior vena cava; *LA,* left atrium; *LV,* left ventricle; *PA,* pulmonary artery; *PV,* pulmonary vein; *RA,* right atrium; *RV,* right ventricle; *SVC,* superior vena cava.

PATHOPHYSIOLOGY

- Partial anomalous pulmonary venous return is usually an acyanotic lesion.

- Total anomalous pulmonary venous return (TAPVR) is associated with total mixing of systemic venous and pulmonary venous blood flow within the heart and thus produces cyanosis.

Clinical manifestations

- The clinical manifestations of TAPVR depend on the presence or absence of *obstruction* of the venous channels.
- If pulmonary venous return is obstructed, severe pulmonary congestion and pulmonary hypertension develop; rapid deterioration occurs without surgical intervention.
- *Obstructed TAPVR is a pediatric cardiac surgical emergency because prostaglandin therapy is usually not effective.*

TREATMENT

- Surgical correction of TAPVR is indicated during infancy, with emergent repair performed for those patients with venous obstruction.

- Surgically, the pulmonary venous confluence is anastomosed directly to the left atrium, the ASD is closed, and any connection to the systemic venous circuit is interrupted.

Truncus Arteriosus

PATHOPHYSIOLOGY

- In truncus arteriosus, a single arterial trunk (truncus arteriosus) arises from the heart and supplies the systemic, pulmonary, and coronary circulations.

- A VSD is always present, with the truncus overriding the defect and receiving blood from both the right and left ventricles. The number of truncal valve cusps varies from 2 to as many as 6, and the valve may be stenotic, regurgitant, or both.

- The pulmonary arteries can arise together from the posterior left side of the persistent truncus arteriosus and then divide into left and right pulmonary arteries (**type I**).

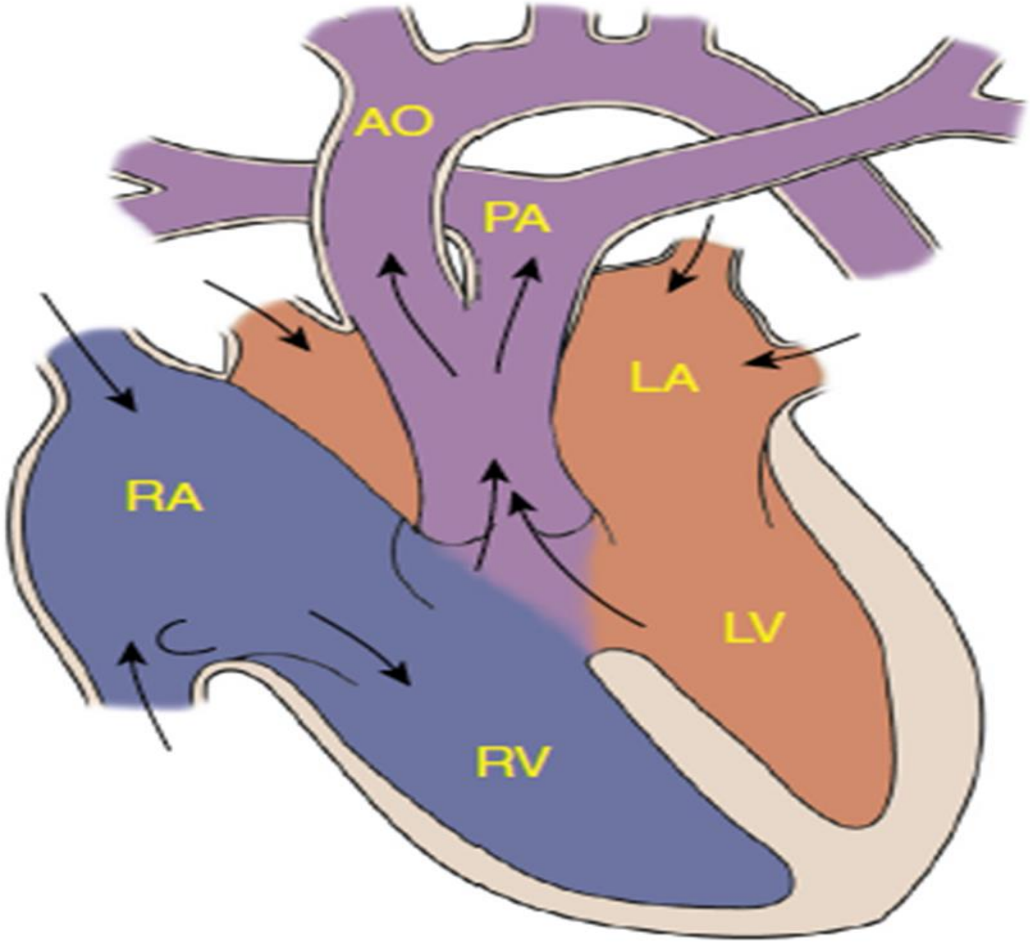


Figure 144-4 Truncus arteriosus. *AO*, Aorta; *LA*, left atrium; *LV*, left ventricle; *PA*, pulmonary artery; *RA*, right atrium; *RV*, right ventricle.

- In types II and III truncus arteriosus, no main pulmonary artery is present, and the right and left pulmonary arteries arise from separate orifices on the posterior (**type II**) or lateral (**type III**) aspects of the truncus arteriosus.

- *Type IV truncus* is a term no longer used because, in this case, there is no identifiable connection between the heart and pulmonary arteries, and pulmonary blood flow is derived from major aortopulmonary collateral arteries arising from the transverse or descending aorta; this is essentially a form of pulmonary atresia.

CLINICAL MANIFESTATIONS

- In the immediate newborn period, signs of heart failure are usually absent; a murmur and minimal cyanosis may be the only initial findings.
- Over the next 1-2 mo of life, pulmonary blood flow begins to become torrential, and the clinical picture is dominated by heart failure, with still mild cyanosis.
- Runoff of blood from the truncus to the pulmonary circulation may result in a wide pulse pressure and bounding pulses.
- These findings will be further exaggerated if truncal valve insufficiency is present.
- The heart is usually enlarged, and the precordium is hyper-dynamic. S2 is loud and single. A systolic ejection murmur, sometimes accompanied by a thrill, is generally audible along the left sternal border.
- Truncus arteriosus is a conotruncal malformation and may be associated with **DiGeorge syndrome**, linked to a deletion of a large region of **chromosome 22q11.**

DIAGNOSIS

- The ECG shows right, left, or combined ventricular hypertrophy. The chest radiograph also shows considerable variation.
- Cardiac enlargement will develop over the 1st several weeks of life and is a result of the prominence of both ventricles.
- The truncus may produce a prominent shadow that follows the normal course of the ascending aorta and aortic knob; the aortic arch is right sided in 50% of patients.
- Sometimes a high bulge left of the aortic knob is produced by the main or left pulmonary artery. Pulmonary vascularity is increased after the 1st few wk of life.

- Echocardiography is diagnostic and demonstrates the large truncal artery overriding the VSD and the pattern of origin of the branch pulmonary arteries.

- Associated anomalies such as an interrupted aortic arch may be noted. Pulsed and color Doppler studies are used to evaluate truncal valve regurgitation.

- If required, cardiac catheterization shows a left-to-right shunt at the ventricular level, with right-to-left shunting into the truncus.

- Systolic pressure in both ventricles and the truncus is similar. Angiography reveals the large truncus arteriosus and more defines the origin of the pulmonary arteries.

PROGNOSIS AND COMPLICATIONS

- Surgical results have been excellent, and many patients with repaired truncus are entering mid-adulthood with several centers reporting 30 and 40 yr old survivors.
- The need to replace the right ventricular–to–pulmonary artery conduit as the child grows means that these patients will need to undergo multiple operations by the time they reach adulthood.
- The development of trans-catheter stent-valves may reduce this in the future.
- When truncus arteriosus is associated with DiGeorge syndrome, the associated endocrine, immunologic, craniofacial, and airway abnormalities may complicate recovery.

TREATMENT

- In the first few weeks of life, many of these infants can be managed with anti-congestive medications; as PVR falls, heart failure symptoms worsen and surgery is indicated, usually within the 1st few mo.

- Delay of surgery much beyond this time period may increase the likelihood of pulmonary vascular disease; many centers now perform routine neonatal repair at the time of diagnosis.

- At surgery, the VSD is closed, the pulmonary arteries are separated from the truncus, and continuity is established between the right ventricle and the pulmonary arteries with a homograft conduit.

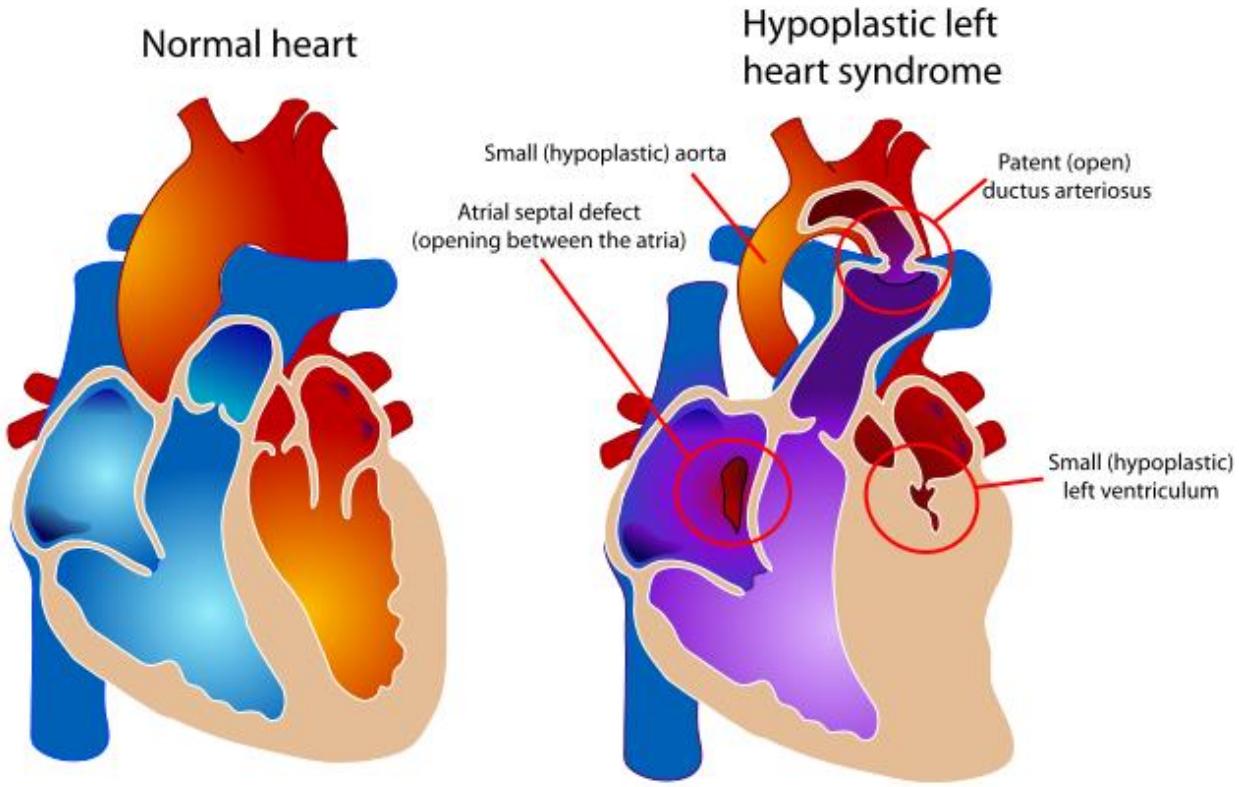
- Immediate surgical results are excellent, but these conduits will develop either regurgitation or stenosis over time and must be replaced, often several times, as the child grows.

- If regurgitation is the primary problem, patients can now be treated with transcatheter stent-valve.

Hypoplastic Left Heart Syndrome

- Hypoplastic left heart syndrome occurs when there is failure of development of the mitral or aortic valve or the aortic arch, resulting:
  - ✓ A small left ventricle that is unable to support normal systemic circulation
- The newborn 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 heart failure from excessive pulmonary blood flow and obstruction of systemic blood flow.

HLHS


Normal heart
Hypoplastic left heart syndrome
Small (hypoplastic) aorta
Patent (open) ductus arteriosus
Atrial septal defect (opening between the atria)
Small (hypoplastic) left ventriculum

TREATMENT

- Surgical therapy for HLHS is associated with improving survival rates, reported as high as 90–95% for the 1st-stage palliation in experienced centers.

- The 1st-stage repair is designed to construct a reliable source of systemic blood flow arising from the single right ventricle using a combination of aortic and pulmonary arterial tissue, and to limit pulmonary blood flow to avoid heart failure and prevent the development of pulmonary vascular disease.

- The surgical procedure typically used is the **Norwood procedure** or the **Sano procedure.**

Cyanotic Congenital Heart Disease
Lesions Associated With Decreased Pulmonary Blood Flow

Tetralogy of Fallot

Tetralogy of Fallot is defined as the simultaneous occurrence of the following four defects:

- Right ventricular outflow obstruction (RVOT)
- Right ventricular hypertrophy
- Ventricular septal defect (VSD)
- Overriding aorta (above the VSD)

- Most common cause of cyanotic CHD

Pathophysiology

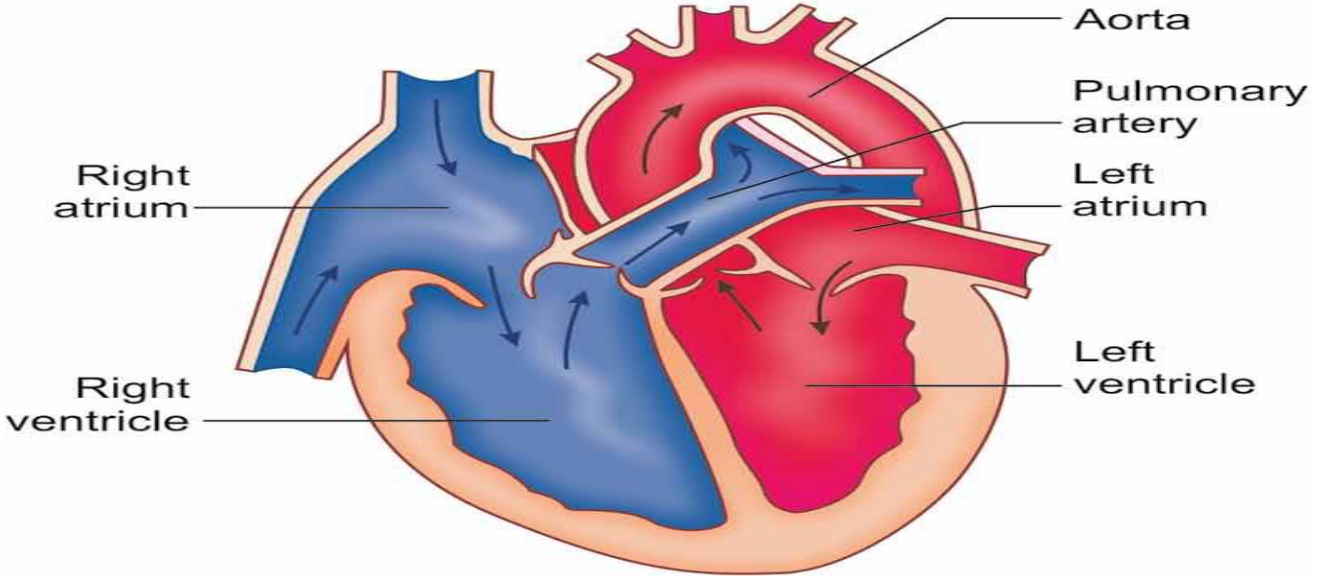
- The extent of right ventricular outflow tract obstruction and central pulmonary hypoplasia determines the severity of hemodynamic dysfunction.
  - Mild obstruction → left-to-right shunt via VSD more pronounced → no cyanosis
  - Severe obstruction → right-to-left shunt via VSD more pronounced → severe cyanosis

- The **VSD** is usually nonrestrictive and large, is located just below the aortic valve, and is related to the posterior and right aortic cusps
- The **aortic arch** is right sided in 20% of cases, and the aortic root is usually large and **overrides the VSD** to varying degrees.

- When the aorta overrides the VSD by >50% (in which case they may also be a subaortic conus) this defect may be classified as a form of DORV;

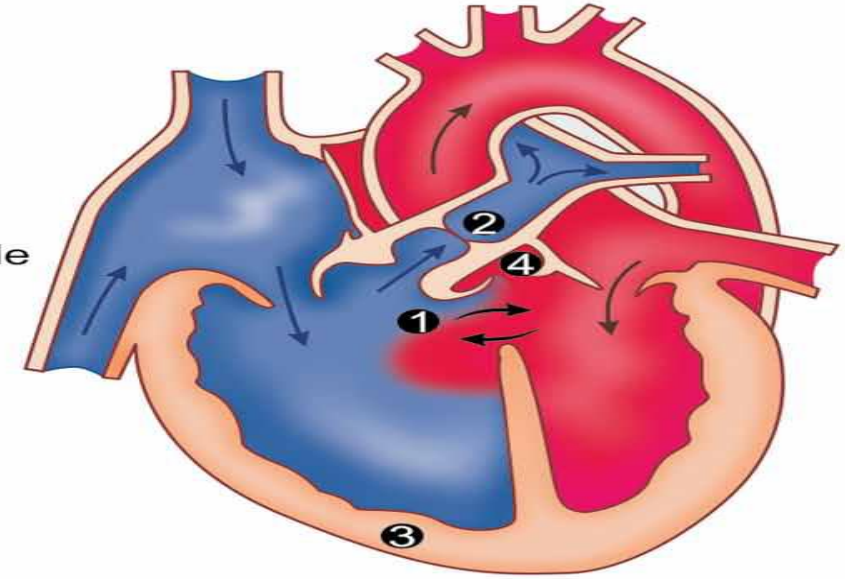
- The degree of RV outflow obstruction determines the timing of the onset of symptoms, the severity of cyanosis, and the degree of **right ventricular hypertrophy** (RVH).

- When obstruction to RV outflow is mild to moderate and a balanced shunt is present across the VSD, the patient may not be visibly cyanotic (**acyanotic** or “pink” tetralogy of Fallot). When obstruction is severe, cyanosis will be present from birth and worsen when the ductus arteriosus begins to close.



Normal heart

❶ Ventricular septal defect
❷ Pulmonary stenosis
❸ Hypertrophy of right ventricle
❹ Overriding aorta



TOF heart

Clinical manifestation

- Infants with **mild degrees** of RV outflow obstruction may initially even have symptoms of heart failure caused by a ventricular-level left-to-right shunt. In these patients, cyanosis is not present at birth; but with increasing hypertrophy of the RV infundibulum as the patient grows, cyanosis occurs later in the 1st few mo of life.

- In contrast, in infants with **severe degrees** of RV outflow obstruction, neonatal cyanosis is noted immediately. In these infants, pulmonary blood flow may be partially or almost totally dependent on flow through the ductus arteriosus.
- When the ductus begins to close in the 1st few hr or days of life, severe cyanosis and circulatory collapse may occur.
- Older children with long-standing cyanosis who have not undergone surgery may have dusky blue skin, gray sclerae with engorged blood vessels, and marked clubbing of the fingers and toes.

- In older children with unrepaired tetralogy, dyspnea occurs on exertion. They may play actively for a short time and then sit or lie down.
- Older children may be able to walk a block or so before stopping to rest. Characteristically, children assume a **squatting** position for the relief of dyspnea caused by physical effort;
- the child is usually able to resume physical activity after a few minutes of squatting.

- These Paroxysmal hypercyanotic attacks are identified as **hypoxic, "blue," or "tet" spells** and they are a problem during the 1st year of life.
-
- Tet spells
  - Intermittent hypercyanotic, hypoxic episodes with a peak incidence 2–4 months after birth
  - Associated with psychological and physical stress (e.g., crying, feeding, defecation)
- Untreated young children tend to squat.
- Auscultatory finding: harsh systolic murmur that is best heard over Erb's point and left upper sternal border ; single second heart sound
- **Diagnostics:**
- Pulse oximetry: ↓ SpO2
- Hyperoxia test: to distinguish cardiac from pulmonary causes of cyanosis especialy in infant

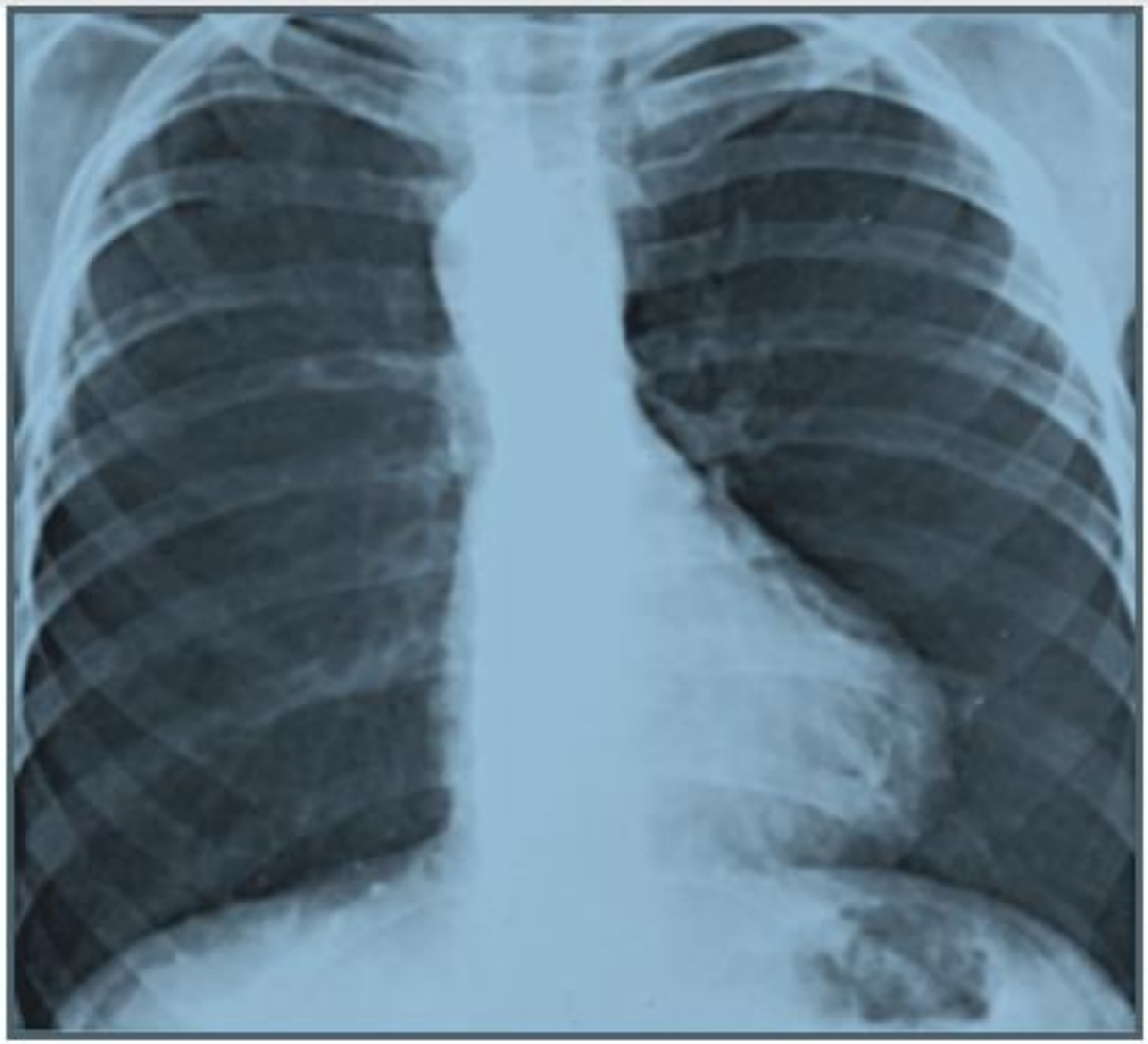


FIG. 457.2 Chest radiograph of 8 yr old boy with tetralogy of Fallot. Note the normal heart size, some elevation of the cardiac apex, concavity in the region of the main pulmonary artery, right-sided aortic arch, and diminished pulmonary vascularity.

The electrocardiogram **(ECG)** demonstrates

- right axis deviation and
- evidence of RVH.
- A dominant R wave appears in the right precordial chest leads (V1 , V2) or an RSR′ pattern.
- In some cases the only sign of RVH may initially be a positive T wave in leads V3 R and V1 . The P wave may be tall and peaked, suggesting right atrial enlargement
- **Two-dimensional (2D) echocardiography** with Doppler establishes the diagnosis

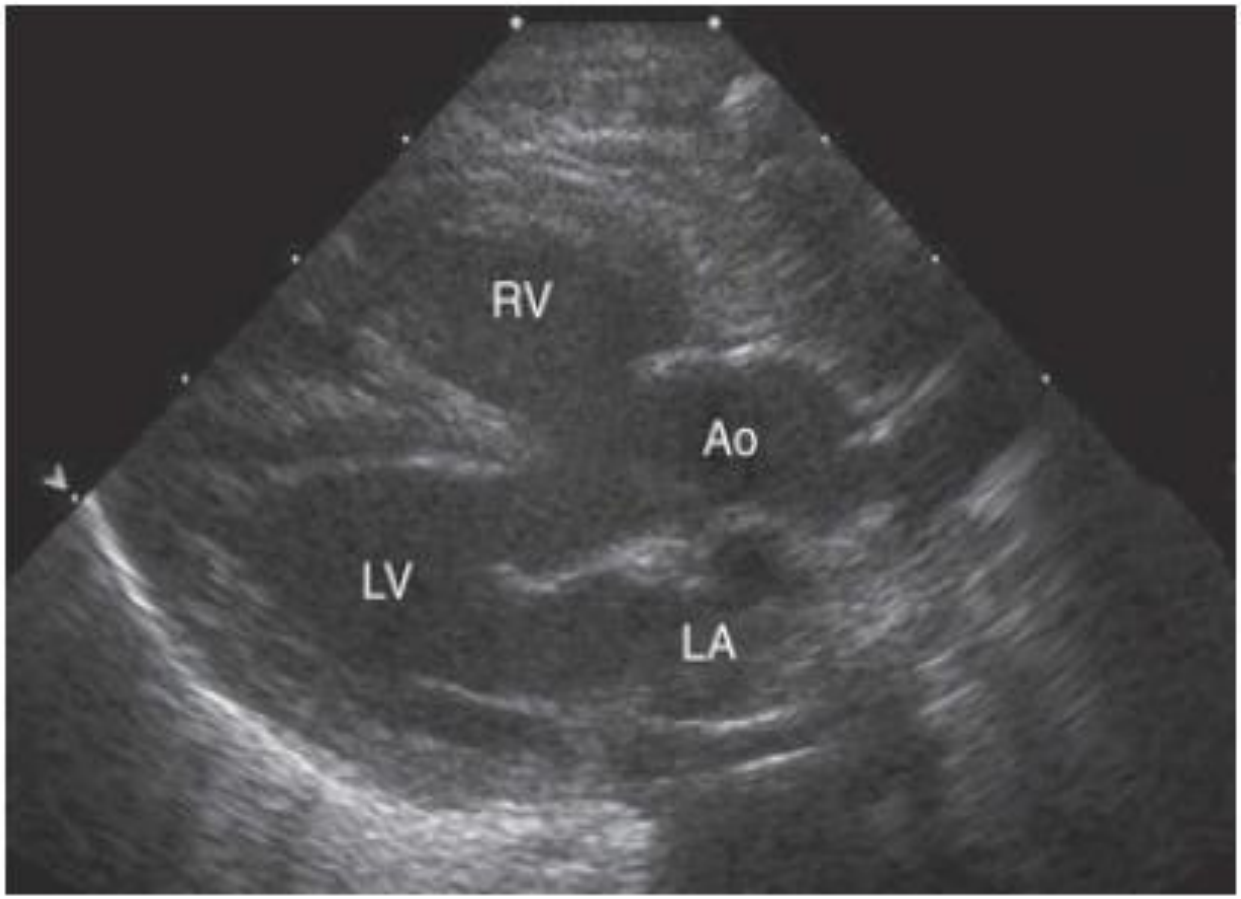


FIG. 457.3 Echocardiogram in patient with tetralogy of Fallot. This parasternal long axis 2D view demonstrates anterior displacement of the outflow ventricular septum that resulted in stenosis of the subpulmonic right ventricular outflow tract, overriding of the aorta, and an associated ventricular septal defect. Ao, Overriding aorta; LA, left atrium; LV, left ventricle; RV, right ventricle.

COMPLICATIONS

- **Cerebral thromboses,** usually occurring in the cerebral veins or dural sinuses and occasionally in the cerebral arteries, are sequelae of extreme polycythemia and dehydration
- **Brain abscess** is less common than cerebrovascular events and extremely rare today. Patients with a brain abscess are usually older than 2 yr.
- **Bacterial endocarditis** may occur in the right ventricular infundibulum or on the pulmonic, aortic. it may complicatepalliative shunts or, in patients with corrective surgery

Management

- Neonates with marked RVOT obstruction may deteriorate rapidly because, as the ductus arteriosus begins to close, pulmonary blood flow is further compromised.
- The IV administration of **prostaglandin E1** (PGE1 ; 0.01-0.20μg/kg/min),is imperative to maintain the PDA

- Acute hypoxia: (tet spells)
  - Administer oxygen
  - Knee to chest position, squatting
  - IV morphine for sedation and fluids
  - If above measures fail: IV beta blockers or oral(Oral propranolol, 2 to 4 mg/kg/day)

- Surgical interventions:
  1. Palliative surgeries: Use of systemic pulmonary shunts:
  - modified Blalock-Taussig shunt) is the procedure of choice in small infants

- Long-term management
  - Surgery: performed within the first year of life, if possible
- Corrective surgical therapy consists of relief of the RVOT obstruction by resecting obstructive muscle bundles and by patch closure of the VSD.
- If the pulmonary valve is stenotic,, a valvotomy is performed.
- If the pulmonary valve annulus is too small or the valve is extremely thickened, a valvectomy may be performed, the pulmonary valve annulus split open, and a transannular patch placed across the pulmonary valve ring.

- Follow-up care is necessary to prevent long-term complications such as heart failure, arrhythmias (e.g., ventricular tachycardia), and neuro-developmental impairment)

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