Congenital Heart Diseases

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Congenital Heart Defects



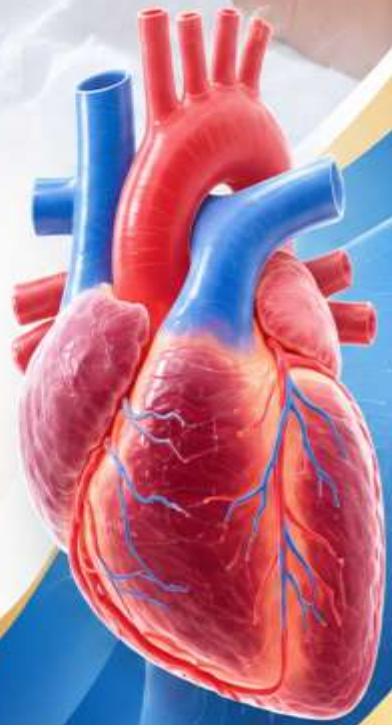
Definition

Abnormalities of cardiac structure that are present **from birth.**

Such developmental abnormalities of the heart typically arise in the **third to eighth week** of gestation.

The first operation for congenital heart disease was patent ductus arteriosus (PDA) ligation by Gross in **1938.**

With the development of neonatal CPB, improved myocardial protection and microsurgical techniques, an increasing nunnber of corrective and palliative operations are possible.



First Operation for Congenital Heart Disease in the World



Dr. Robert Edward Gross
(1905 – 1988)
Pioneer of congential cardiac surgery

First widely recognized successful operation for CHD

Date: August 26, 1938

Surgeon: Dr. Robert E. Gross

Patient: 7-year-old girl, Lorraine Sweeney

Diagnosis: Patent ductus arteriosus (PDA)

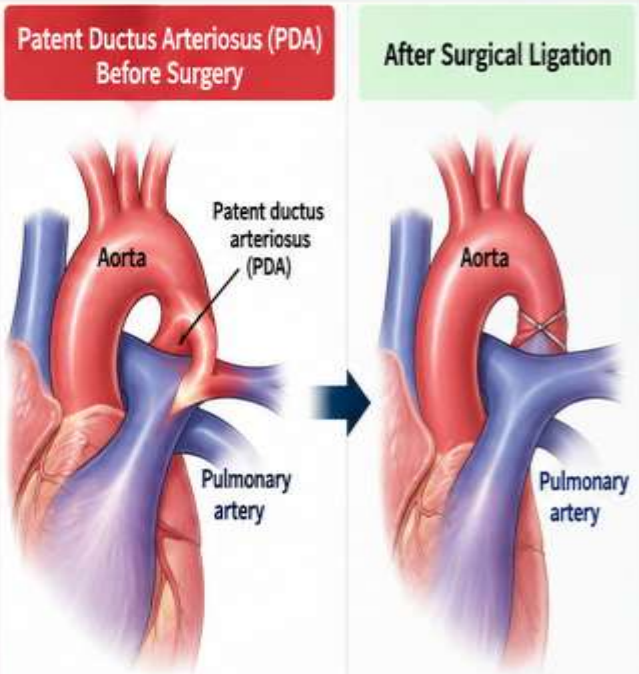
Procedure: Surgical ligation of the PDA

Approach: Left thoracotomy

Outcome: Successful closure and recovery



Dr. Gross performing PDA ligation (1938)



Major Milestones in Congenital Cardiac Surgery

1937 — John Strieder
Attempted PDA closure; patient died

1938 — Robert E. Gross
First widely recognized successful PDA ligation

1944 — Clarence Crafoord
Successful coarctation repair

1944 — Alfred Blalock (with Helen Taussig and Vivien Thomas)
First Blalock–Taussig shunt

1952 — F. John Lewis
First successful direct-vision intracardiac ASD repair using hypothermia and inflow occlusion

1953 — John Gibbon
First successful ASD closure using cardiopulmonary bypass

1954 — C. Walton Lillehei
Landmark intracardiac repairs using controlled cross-circulation

Note: Emil Karl Frey may have successfully ligated a PDA in Germany in or before 1938, but his operation was not published at the time. Dr. Robert Gross's operation remains the conventionally recognized first successful operation for congenital heart disease.

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Discussion Questions

1. How many CHD in IRAQ per year ?
2. How many from CHD need Surgery ?
3. Do you think surgery in Iraq cover all babys ?

Incidence and Prevalence of Congenital Heart Disease (CHD)

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Global Incidence (Birth Prevalence)

8 – 10
per 1,000 live births
(0.8 – 1%)

Global estimate (meta-analysis)
9.41 per 1,000 live births
Based on **260** studies (~130.8 million live births)
Liu et al. JAMA Cardiol. 2019;4:1139–1148.

People living with CHD globally (all ages, 2021)
15.77 million
GBD 2021 estimate

Children living with CHD (0–19 years, 2021)
4.18 million
GBD 2021 estimate

Estimated Burden of CHD in Iraq

Annual live births in Iraq
1,177,772
(≈ 3,227 per day)
(Based on UN World Population Prospects)



Estimated newborns with CHD annually
9,422 – 11,778
(8 – 10 per 1,000 live births)

Estimated newborns with CHD daily
26 – 32
(8 – 10 per 1,000 live births)

Key Points

- CHD is the most common group of major congential structural abnormalities.
- **Affects approximately 1 in every 100 live-born infants.**
- The number of people living with CHD is increasing due to improved diagnosis, medical care and surgical survival.
- **Not all patients require surgical intervention.**

These are estimates based on global data and demographic figures. They do not confirm how many infants require surgery in Iraq.

DISCUSSION |
Can Iraq Meet the Need for CHD Surgery?



1 **Newborns with CHD in Iraq per day**

26 – 32

newborns with CHD per day

(8 – 10 per 1,000 live births)

≈ 1,177,772 live births per year
(≈ 3,227 per day)

2 **Newborns with critical CHD per day**

≈ 6 – 8

newborns with critical CHD per day

(≈ 25% of all CHD)

6 – 8 is NOT the number of daily open-heart operations.
It includes all who will need surgical or catheter intervention (many in the first year).

3 **Require intervention during the first year**



Surgical repair



Catheter intervention

Most infants with critical CHD require surgery or catheter intervention during the first year of life.

Discussion Points

Can Iraq provide timely diagnosis, intervention, PICU care and follow-up for all infants with CHD?

1. National case volume?

- How many infants with CHD are identified each year?
- How many require intervention?

2. Neonatal surgical capacity?

- Is there sufficient surgical and catheter-based capacity?
- Availability of PICU, ECMO and specialist teams?

3. Geographic access?

- Are services available across Iraq or concentrated in major centers?
- Challenges in referral and transport of critically ill neonates?

4. Waiting lists & outcomes?

- What are the current waiting times?
- Are all high-risk infants treated timely?
- How do our outcomes compare with international standards?

Current local data (example)

One Iraqi center reported **> 440 pediatric open-heart operations in 2024.**
This is not a national total.

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Planning estimate based on 8–10 CHD per 1,000 births, ~1.18 million annual births, and CDC estimate ~25% critical CHD. Actual Iraqi capacity and surgical-only demand require registry data.

Sources: CDC CHD Data; Imam Zain Al-Abideen Hospital (2025 report of 2024 activity).

Embryological Development of the Heart and Fetal Circulation

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Embryological development of the heart

Week 3 (18–21 days)



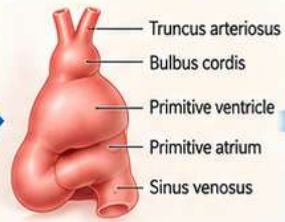
Cardiogenic mesoderm and formation of the primitive heart tube

Week 4 (22–28 days)



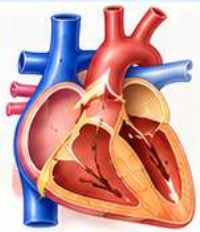
Rightward looping of the heart tube (D-loop)

Week 4–5 (28–35 days)



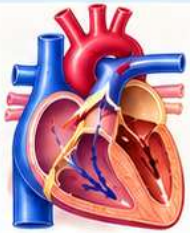
Differentiation of cardiac segments

Week 5–7



Septation of atria and ventricles, formation of valves and outflow tracts

Week 8



Completion of septation and formation of the great arteries

Key points in cardiac development

- The heart is the first functional organ to develop.
- By 12 weeks of fetal life, the primitive vascular tube is fully developed.
- Major cardiac structural abnormalities typically arise during the **third to eighth week of gestation.**
- The heart develops from the cardiogenic mesoderm and undergoes complex processes of looping, septation, and remodeling.
- **Disturbances in these processes lead to congenital heart diseases.**

Fetal Circulation (Right and left ventricles pump in parallel)

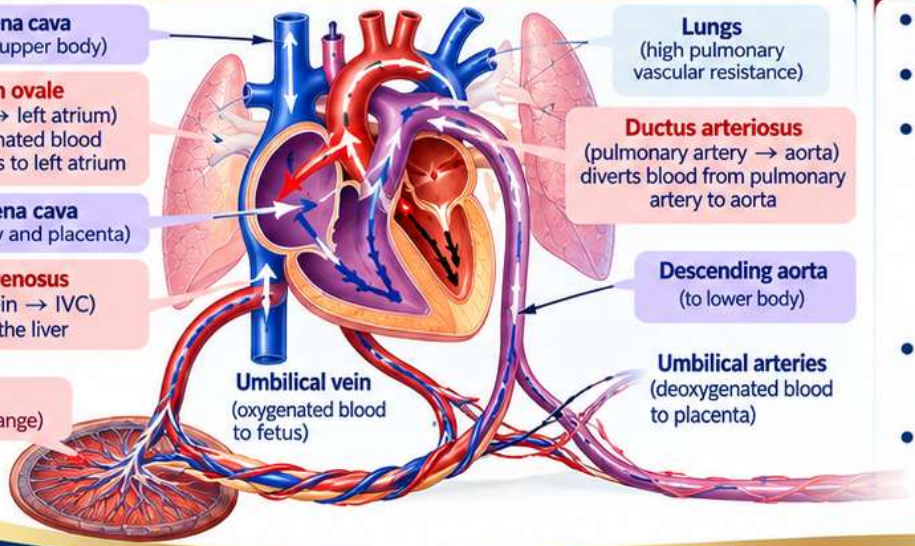
Superior vena cava
(from head and upper body)

Foramen ovale
(right atrium → left atrium) allows oxygenated blood from IVC to pass to left atrium

Inferior vena cava
(from lower body and placenta)

Ductus venosus
(umbilical vein → IVC) bypasses the liver

Placenta
(site of gas exchange)



Lungs
(high pulmonary vascular resistance)

Ductus arteriosus
(pulmonary artery → aorta) diverts blood from pulmonary artery to aorta

Descending aorta
(to lower body)

Umbilical arteries
(deoxygenated blood to placenta)

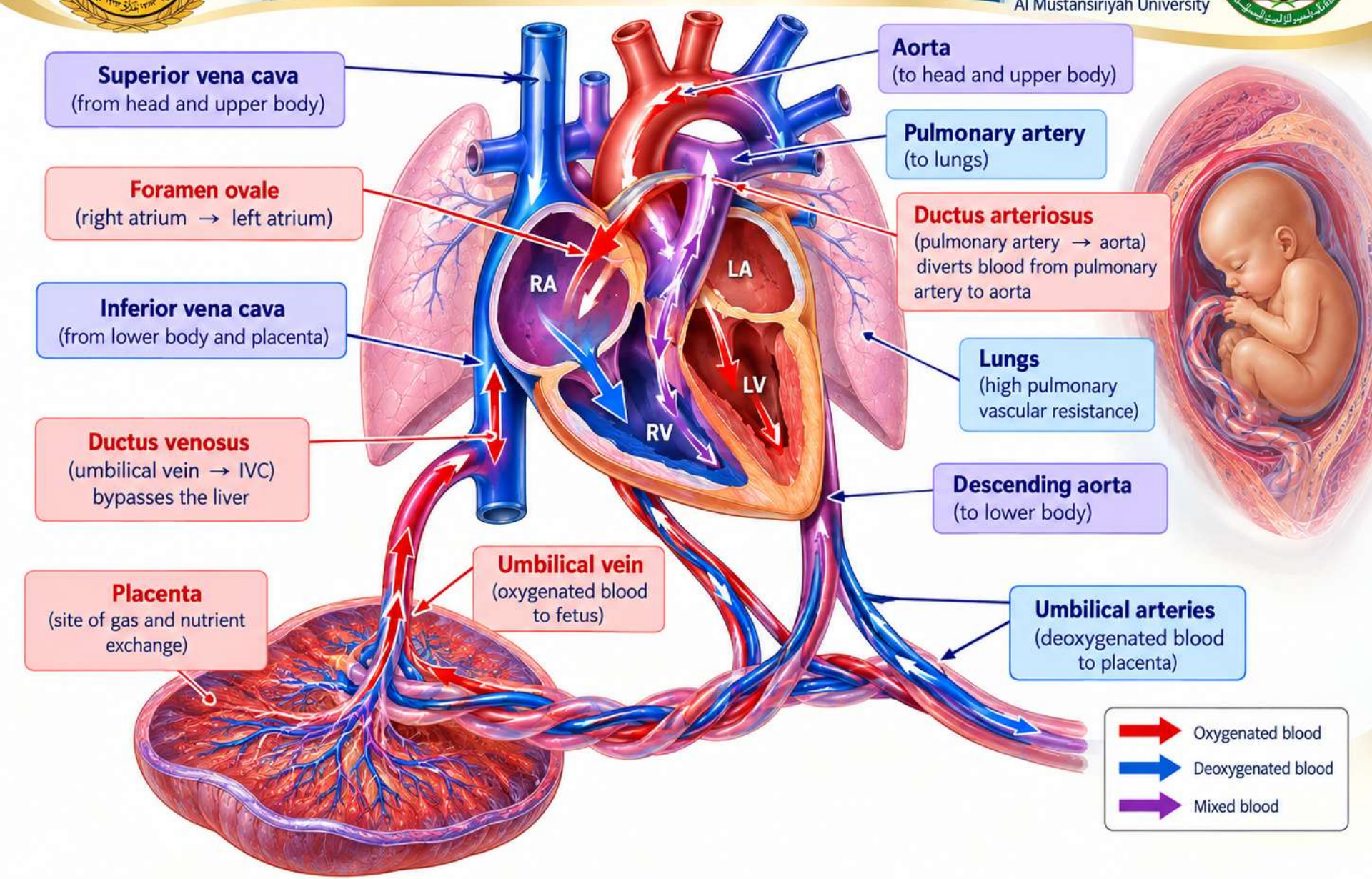
Features of fetal circulation

- **Right and left ventricles pump blood in parallel** rather than in series.
- **The heart** and head receive more **highly oxygenated blood.**
- **Three structural shunts** are present:
  1. **Ductus venosus** (umbilical vein → IVC)
  2. **Foramen ovale** (right atrium → left atrium)
  3. **Ductus arteriosus** (pulmonary artery → aorta)
- Pulmonary vascular resistance is high, so only a small portion of right ventricular output goes to the lungs.
- **Oxygen and nutrients** are provided **by the placenta.**



Fetal Circulation

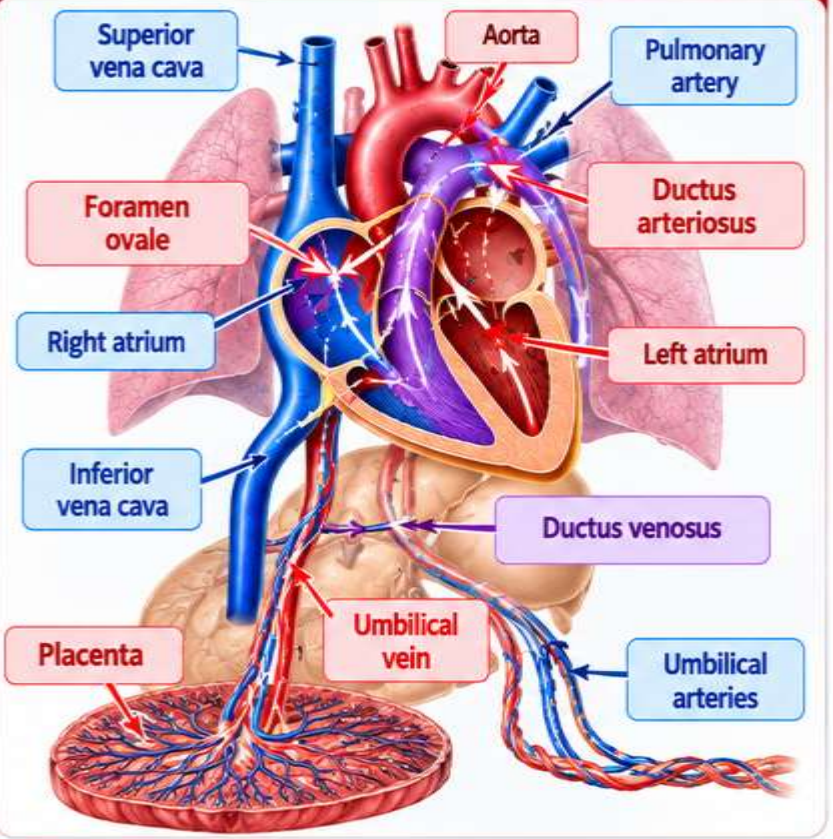
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Fetal Circulation (in utero)

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Fetal circulation (in utero)



Challenge Question

What happens to the fetal circulation at birth?

1. Umbilical venous return stops → .....
2. Inferior vena cava pressure .....
3. Pulmonary vascular resistance .....
4. Pulmonary venous return .....
5. Right atrial pressure .....
6. Left atrial pressure .....
7. Foramen ovale .....
8. Ductus arteriosus .....
9. Ductus venosus .....

Word Bank

decreases | closes (functionally) | decreases | closes (anatomically) | increases

closes | decreases | stops | increases

Timing of Changes in Circulation After Birth

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1	**Cutting and tying of the umbilical cord** stops venous blood flow from the placenta.	**At birth** (within seconds)
2	**Fall in inferior vena cava pressure** due to cessation of placental venous return.	**Within seconds to minutes**
3	**Fall in pulmonary vascular resistance** with the first breaths and lung expansion.	**Within minutes**
4	**Increase in pulmonary venous return** to the left atrium.	**Within minutes**
5	**Fall in right atrial pressure** and rise in left atrial pressure.	**Within minutes to hours**
6	**Functional closure of the foramen ovale** The stopping of venous return from the placenta also causes the ductus venosus to close.	**Within hours to days**

Causes and Recognized Associations with Congenital Heart Disease

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Maternal (environmental) factors

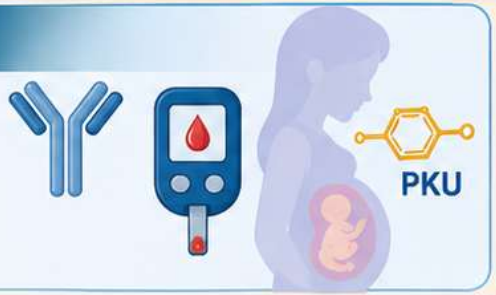
Infections

- Rubella



Maternal diseases

- Systemic lupus erythematosus
- Diabetes mellitus
- Maternal phenylketonuria



Genetic factors

Drugs / medications

- Alcohol abuse
- Warfarin
- Phenytoin
- Lithium
- Thalidomide



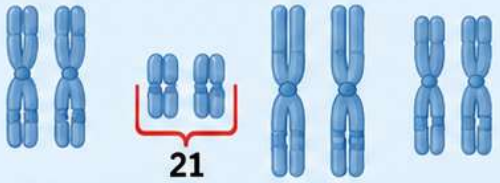
Single gene defects

- Marfan syndrome
- Noonan syndrome
- Holt–Oram syndrome
- Numerous single-gene disorders



Chromosomal defects

- Trisomy 21 (Down syndrome)
- Trisomy 18 (Edwards syndrome)
- Trisomy 13 (Patau syndrome)
- Turner syndrome
- Klinefelter syndrome



Deletions

- DiGeorge syndrome (22q11.2 deletion)
- Williams syndrome (7q11.23 deletion)

Congenital Heart Disease

Causes and Recognized Associations

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1 Abnormalities of cardiac structure may arise from the

- Abnormal development during embryogenesis
- Genetic, environmental or multifactorial causes



2 Persistence of normal fetal channels (PDA, patent foramen ovale)

- Failure of closure after birth
- Examples: PDA, patent foramen ovale



3 Failure of septation

- Atrial septal defect (ASD)
- Ventricular septal defect (VSD)
- Tetralogy of Fallot (conotruncal defect)



4 Stenosis

- Intracardiac (valvular, subvalvular, supravalvular)
- Infravalvular or extracardiac
- Coarctation of the aorta

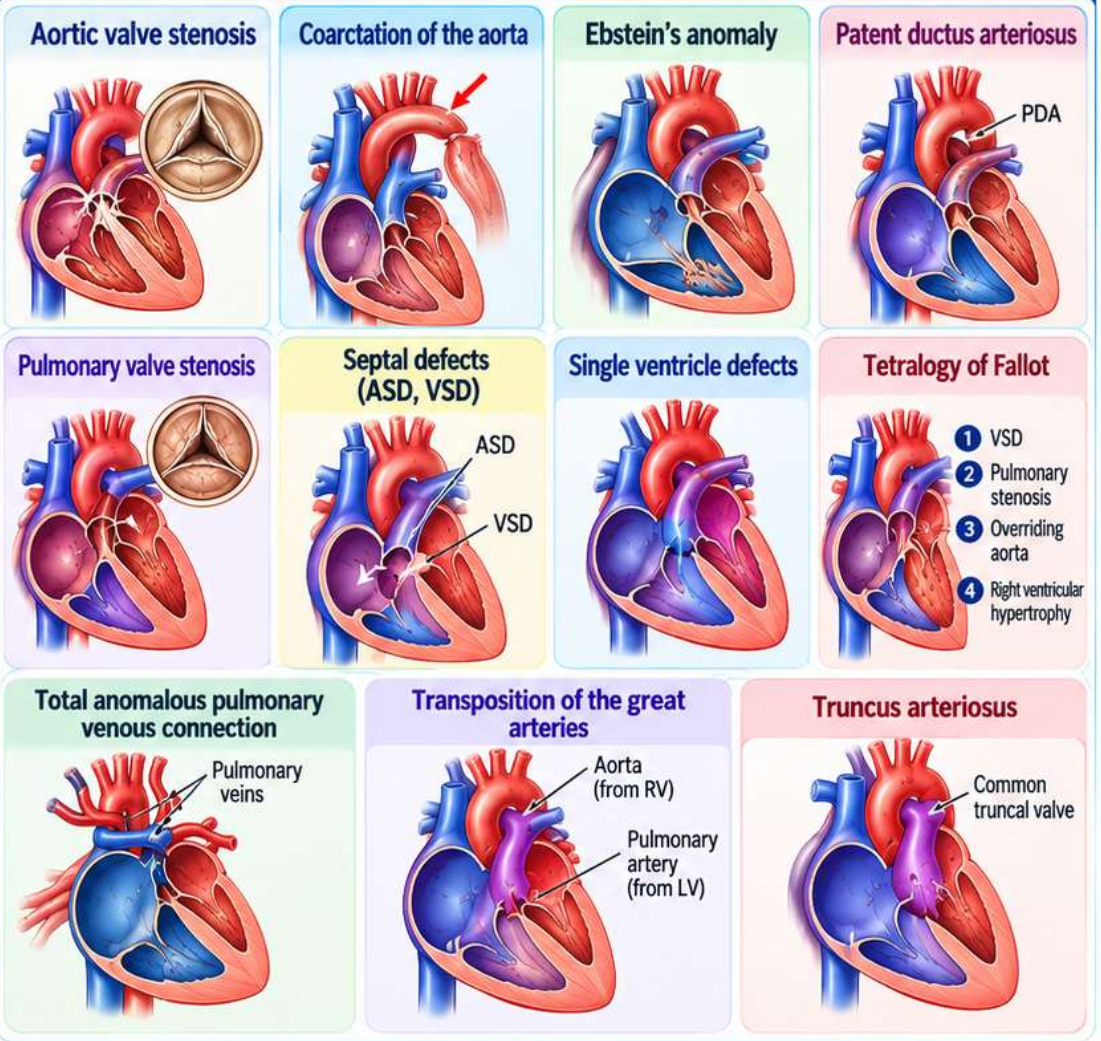


5 Atresia or abnormal connections (transposition of the great vessels)

- Abnormal alignment or connection of the great arteries
- Examples: Transposition of the great arteries, Truncus arteriosus, Total anomalous pulmonary venous connection



Types of Congenital Heart Disease Associated



Classification of Congenital Heart Disease

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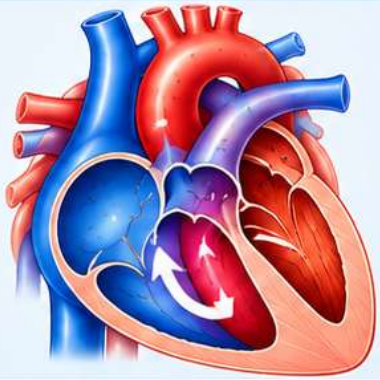
Overview

Cyanotic congenital heart diseases make up **25%** of cases (8–9 per 1000 live births) and are usually more complex, although they do include simple defects.



Cyanotic Congenital Heart Disease

Decreased pulmonary blood flow (right-to-left shunt or mixing)

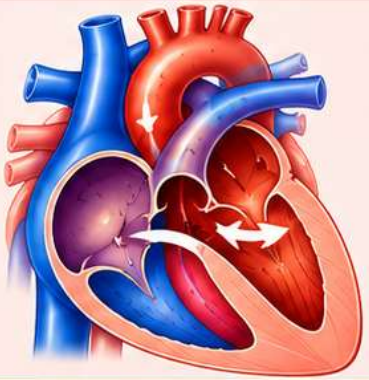


Examples

- Tetralogy of Fallot
- Tricuspid atresia
- Total anomalous pulmonary venous connection (TAPVC)
- Transposition of the great arteries (TGA)
- Other univentricular heart with pulmonary stenosis

Acyanotic Congenital Heart Disease

Left-to-right shunt or obstructive lesions (usually without cyanosis)



Examples

- **Left-right shunt lesions:**
  - Ventricular septal defect (VSD)
  - Atrial septal defect (ASD)
  - Atrioventricular septal defect (AVSD)
  - Patent ductus arteriosus (PDA)
- **Obstructive lesions:**
  - Aortic stenosis
  - Pulmonary valve stenosis
  - Coarctation of the aorta

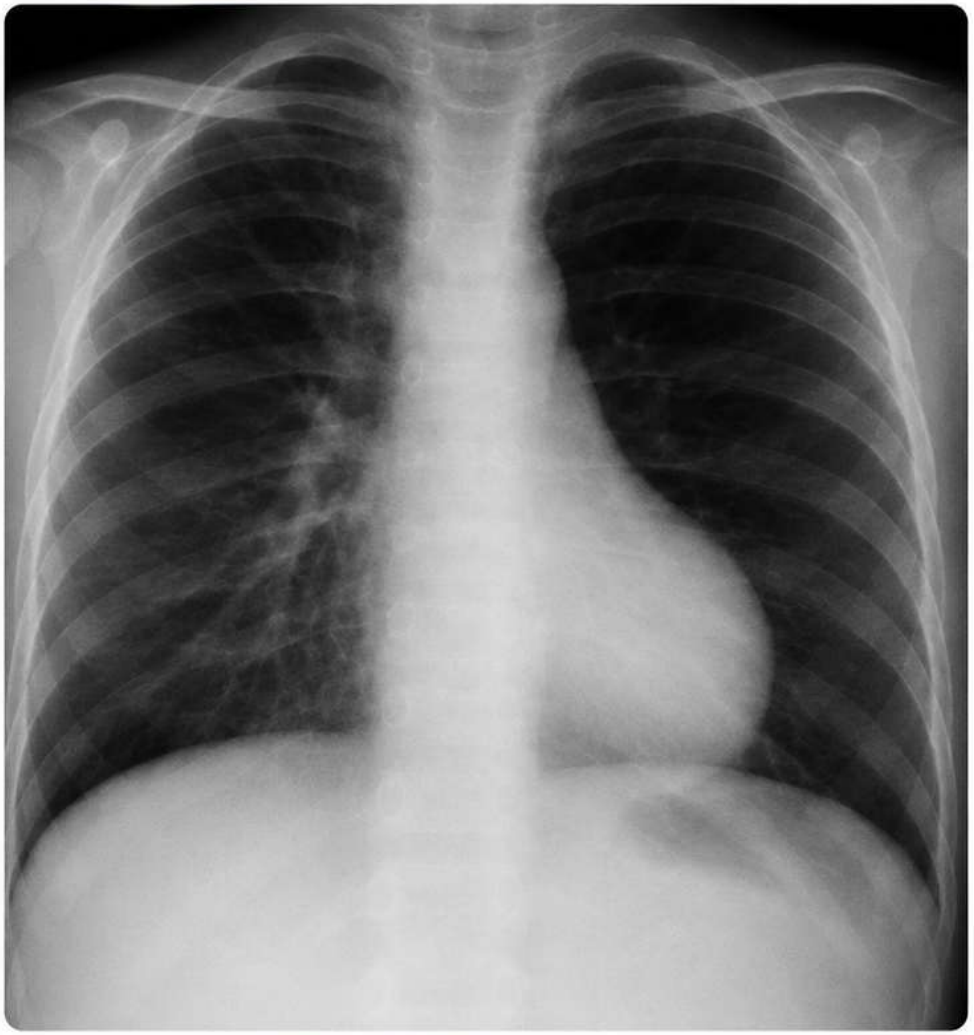
Comparison Between
Cyanotic and Acyanotic Congenital Heart Diseases

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Feature	Cyanotic CHD	Acyanotic CHD
Representative image	 **Example:** Tetralogy of Fallot (cyanotic)	 **Example:** Atrial septal defect (acyanotic)
Definition	CHD with systemic desaturation due to mixing of deoxygenated blood	CHD with normal systemic oxygenation initially
Cyanosis	**Present (central)**	**Absent initially**
Type of shunt	Right → Left or parallel circulation	Left → Right
Arterial O_2 saturation	↓ (<85–90%)	Normal
Pulmonary blood flow	↓ (TOF) or ↑ (TAPVR)	↑
Heart failure	Less common early	Common
Chest infections	Uncommon	Common
Clubbing	Common (chronic)	Rare
Polycythemia	Common	Absent
Squatting	Typical (TOF)	Absent
Main complications	Brain abscess, stroke, endocarditis	Pulmonary HTN, Eisenmenger syndrome
Reversibility	Often fixed anatomy	May reverse (Eisenmenger)
Prostaglandin E1	Often lifesaving	Rarely needed

Exam Challenge

Chest X-ray in Congenital Heart Disease



Question

A 3-year-old boy presents with cyanosis and exertional dyspnoea. A systolic ejection murmur is heard at the left upper sternal border. The following chest X-ray is obtained.

Which of the following congenital heart diseases is most likely?

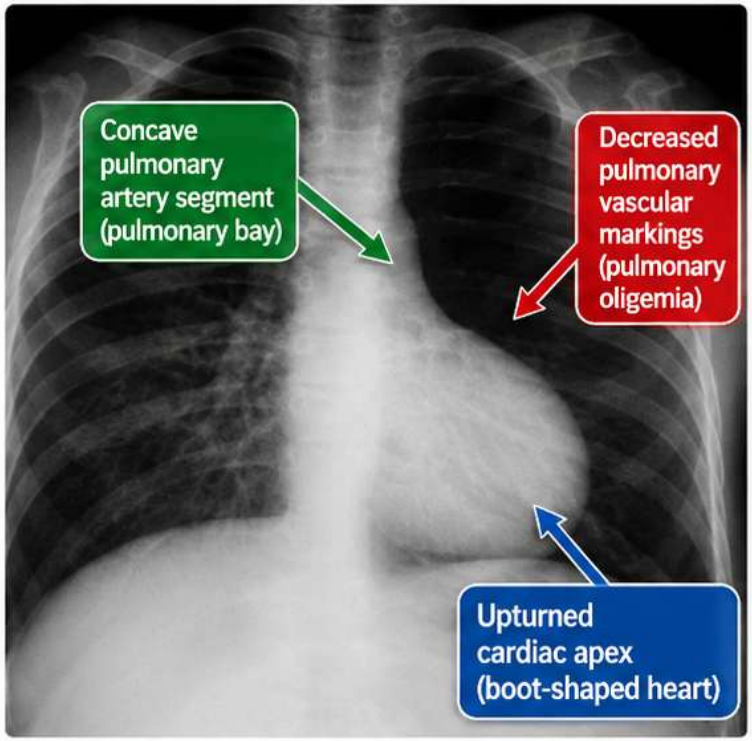
- **A** Atrial septal defect
- **B** Ventricular septal defect
- **C** Tetralogy of Fallot
- **D** Transposition of the great arteries
- **E** Patent ductus arteriosus

Exam Challenge
CXR in Tetralogy of Fallot (TOF)

Question

A 2-year-old boy presents with cyanosis and exertional dyspnoea. A chest X-ray is shown below.
Which of the following is the most likely diagnosis?

- **A** Atrial septal defect
- **B** Ventricular septal defect
- **C** **Tetralogy of Fallot**
- **D** Transposition of the great arteries
- **E** Patent ductus arteriosus



Answer

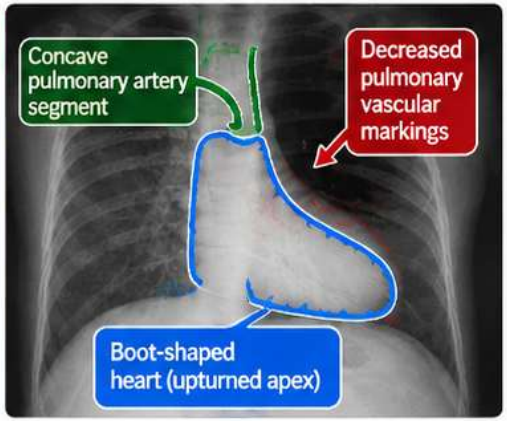
C **Tetralogy of Fallot**

Explanation

- The chest X-ray shows a classic **boot-shaped heart** (coeur en sabot).
- Features include:
  - Upturned cardiac apex due to right ventricular hypertrophy.
  - Concave pulmonary artery segment (pulmonary bay).
  - Decreased pulmonary vascular markings (pulmonary oligemia) due to reduced pulmonary blood flow.
- These findings are characteristic of **Tetralogy of Fallot (TOF).**

Key CXR Features of TOF

1. Boot-shaped heart with upturned apex (RV hypertrophy)
2. **Pulmonary oligemia** (decreased vascular markings)
3. Concave pulmonary artery segment (pulmonary bay)
4. Often normal or mildly enlarged heart size
5. Right-sided aortic arch in 20–25% of cases



Remember

- TOF = VSD + RV outflow tract obstruction (infundibular and/or pulmonary stenosis) + overriding aorta + right ventricular hypertrophy.
- Chest X-ray supports the diagnosis, but echocardiography is the diagnostic test of choice.
- The **boot-shaped** appearance is not present in all cases.

Tetralogy of Fallot (TOF)

Overview

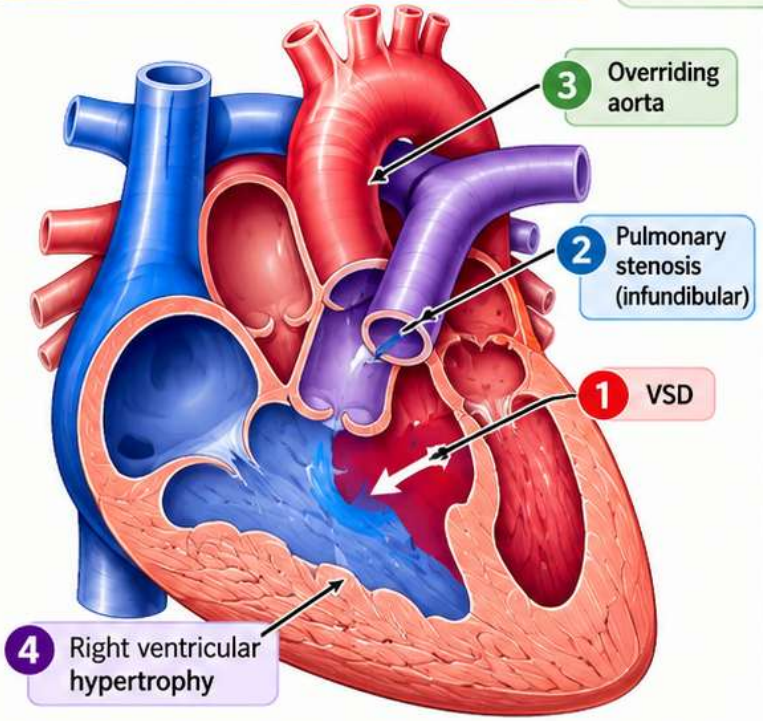
- The most common cyanotic congenital heart disease.
- Accounts for about 4–6% of all congential heart diseases.
- Usually presents in early **infancy and** most children survive to 1 year.

Pathophysiology

- Right ventricular outflow obstruction leads to diversion of deoxygenated venous blood through the VSD into the left ventricle and aorta.
- Systemic circulation is provided with unoxygenated blood causing cyanosis.

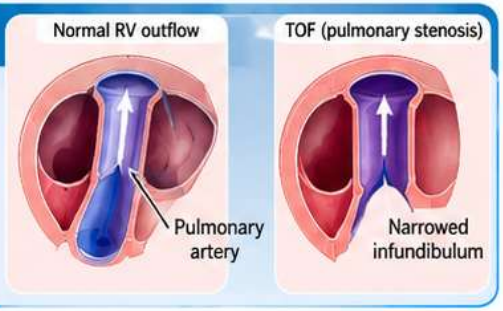
Components (the four lesions)

1. Ventricular septal defect (VSD)
2. Infundibular (subpulmonary) stenosis → right ventricular outflow obstruction
3. Overriding aorta (dextroposition of aorta)
4. Right ventricular hypertrophy (RVH)



Result

- Right → Left shunt (or parallel circulation)
- Decreased arterial oxygen saturation (≈ 85–90%)
- Decreased pulmonary blood flow
- Cyanosis



Clinical features

- Cyanosis with dyspnoea.
- Ejection systolic murmur at left upper sternal border (due to pulmonary stenosis) which diminishes during cyanotic attacks (tet spells).
- Clubbing (chronic cases).
- Polycythemia.
- Less common heart failure in early infancy.
- Chest infections are uncommon.

Investigations

- Echocardiography (diagnostic).
- Cardiac catheterization (if needed).

Complications

- Brain abscess, stroke, endocarditis (due to chronic cyanosis).

Tetralogy of Fallot (TOF)
Management and Treatment

Goals of Treatment

- Increase pulmonary blood flow.
- Relieve right ventricular outflow tract obstruction.
- Correct cyanosis.
- Prevent complications (e.g. brain abscess, hypoxic spells).

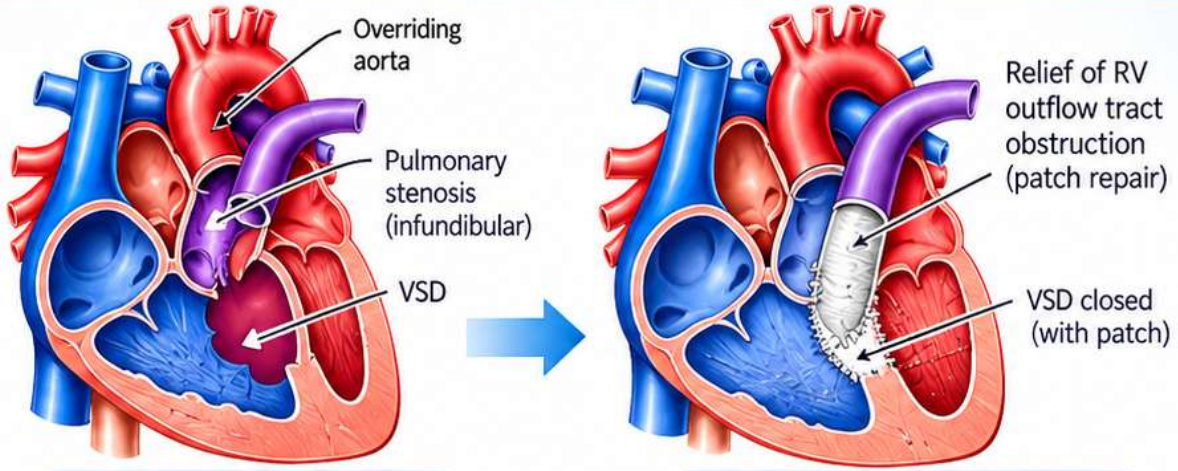
Initial / Medical Management

- Prostaglandin E1 (if ductal-dependent pulmonary blood flow).
- Treat hypercyanotic (tet) spells:
  - Knee–chest position
  - Oxygen
  - Morphine (sedation)
  - IV fluids
  - Beta-blocker (e.g. propranolol)
- Treat anemia (avoid excessive polycythemia).
- Prevent and treat infections.

Timing of Definitive Repair

- Complete surgical repair is usually performed between 3–6 months of age (by 6–12 months).
- Early repair is recommended in most centers to prevent long-term complications.

Surgical Treatment – Complete Repair
(Gold standard)



Pre-operative (TOF) | Post-operative (Repaired TOF)

Surgical Components

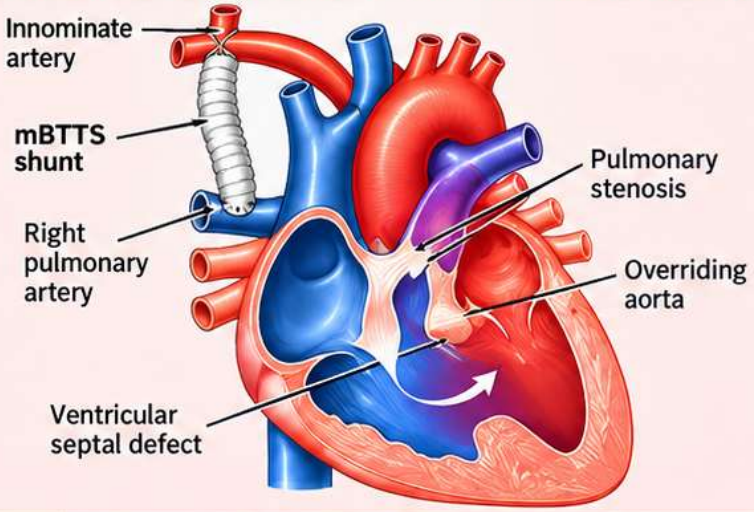
1. Closure of the ventricular septal defect (with a patch).
2. Relief of right ventricular outflow tract obstruction (resection of infundibular muscle, patch enlargement of RVOT and/or pulmonary valve).
3. Preservation of the pulmonary valve if possible.
4. Additional procedures as needed (e.g. transannular patch if severe pulmonary stenosis).

Palliative Procedures
(when complete repair is not possible)

- Systemic-to-pulmonary shunt:
  - Modified Blalock–Taussig shunt (subclavian artery to pulmonary artery).
  - Central shunt (aorta to pulmonary artery).
  - Stented ductus arteriosus.
- Used in neonates or small infants with severe pulmonary stenosis or inadequate pulmonary blood flow, to improve oxygenation until definitive repair.

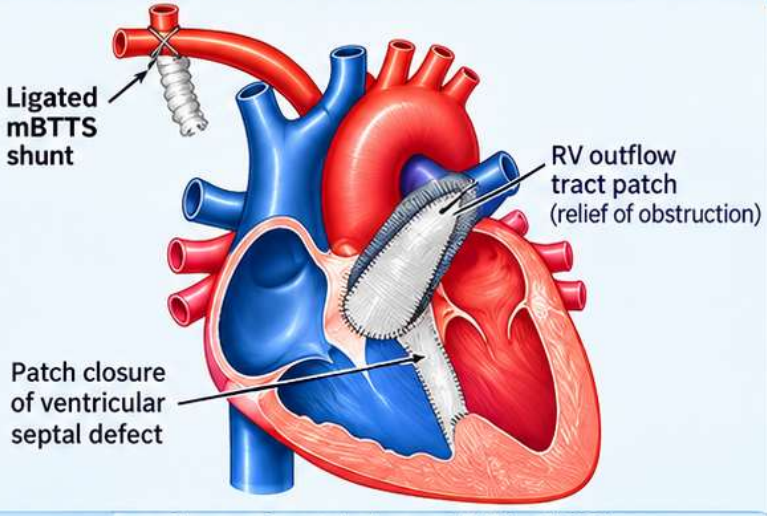
Surgical Management of Tetralogy of Fallot (TOF)

Palliative Surgery
Modified Blalock–Taussig–Thomas Shunt (mBTTS)



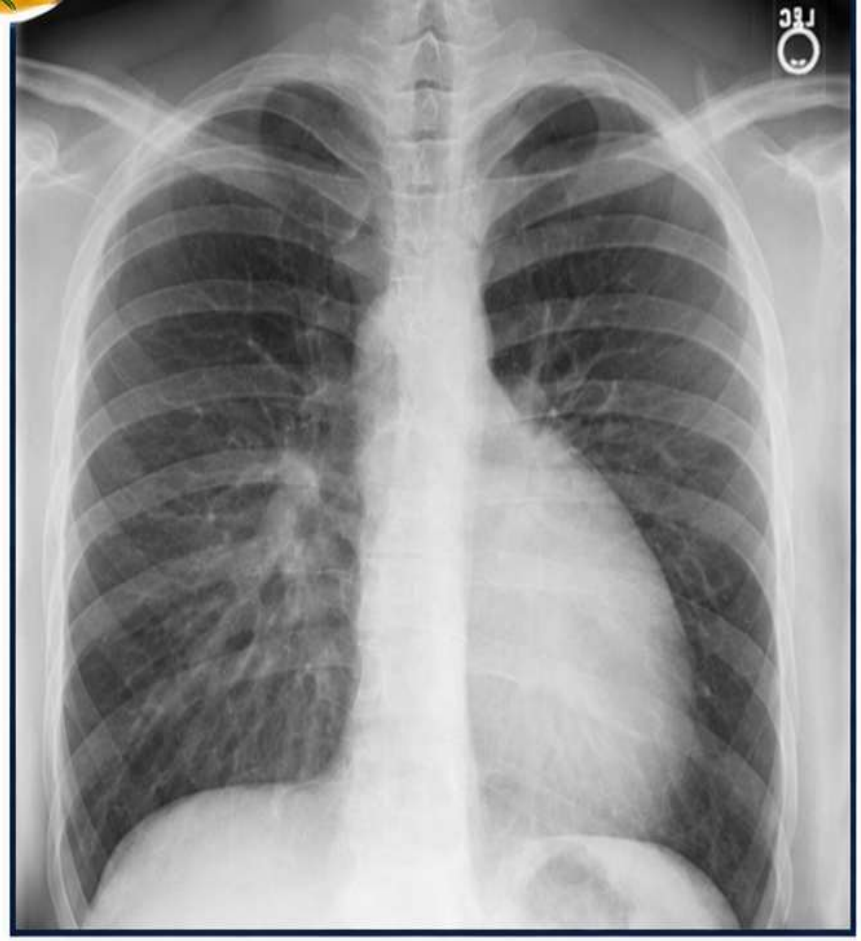
Purpose	• Increase pulmonary blood flow and improve oxygenation (palliative).
Indication	• Symptomatic cyanosis in neonates/infants with severe pulmonary stenosis or small pulmonary arteries, or when primary repair is not feasible.
Procedure	• End-to-side anastomosis of subclavian (innominate) artery to right pulmonary artery using a graft (PTFE) – modified BT shunt.
Effect	• Increases pulmonary blood flow → improves arterial oxygen saturation and relieves cyanosis.
Notes	• Temporary (palliative) procedure. • Shunt is later taken down (ligated) during complete repair.

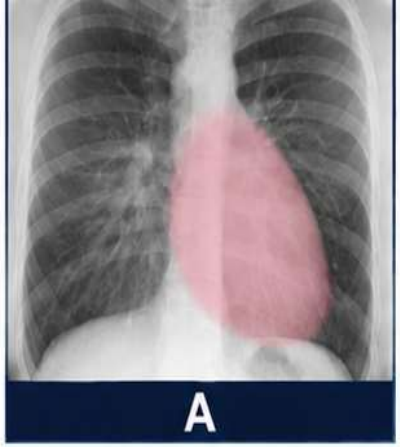
Complete Surgical Repair
(Definitive Treatment)

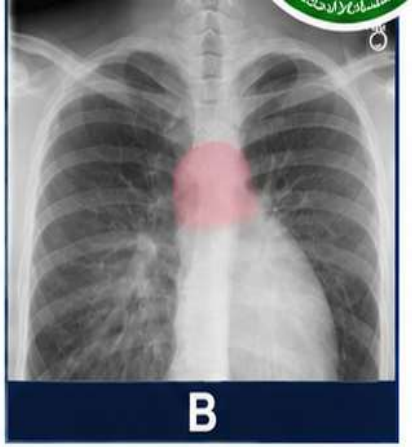


Components	• Closure of ventricular septal defect (VSD). • Relief of right ventricular outflow tract (RVOT) obstruction (resection of infundibular muscle, patch enlargement of RVOT and/or pulmonary valve).
Goals	• Establish normal (or near-normal) intracardiac anatomy and physiology. • Eliminate right-to-left shunt and correct cyanosis.
Procedure	• VSD closure with patch (e.g. Dacron/PTFE). • Relief of RVOT obstruction (transannular patch if needed). • Previous BT shunt is ligated/divided.
Timing	• Usually performed between 3–6 months of age. Early repair favored in many centers.
Outcome	• Good long-term survival with low residual defects in most patients.

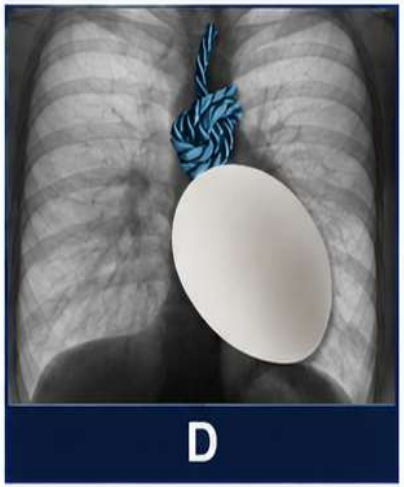
Key Point: **The modified BT shunt is a palliative procedure and not required in every patient.**
Many infants can undergo primary complete repair, while staged palliation is reserved for selected cases.









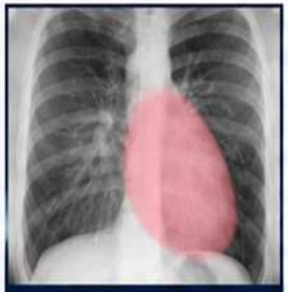


Question

A chest X-ray is shown (main image with additional views A–D).

1. Describe the chest X-ray findings.
2. What is the most likely diagnosis?
3. Mention two other conditions with a similar radiological appearance.





A. Oval cardiac silhouette



B. Narrow vascular pedicle



C. Increased pulmonary vascular markings



D. Egg-on-a-string appearance

1. Description of the chest X-ray findings

A. Cardiac silhouette

- Normal size or mildly enlarged
- Smooth, oval-shaped cardiac silhouette (egg-shaped heart)

B. Mediastinum / vascular pedicle

- Narrow superior mediastinum (narrow vascular pedicle)

C. Pulmonary vascularity

- Increased pulmonary vascular markings (pulmonary plethora)

D. Overall appearance

- Classic 'egg-on-a-string' appearance due to parallel arrangement of the great vessels

2. Most likely diagnosis

Transposition of the Great Vessels (TGV)

(Dextro-transposition, D-TGA)

3. Two other conditions with a similar radiological appearance

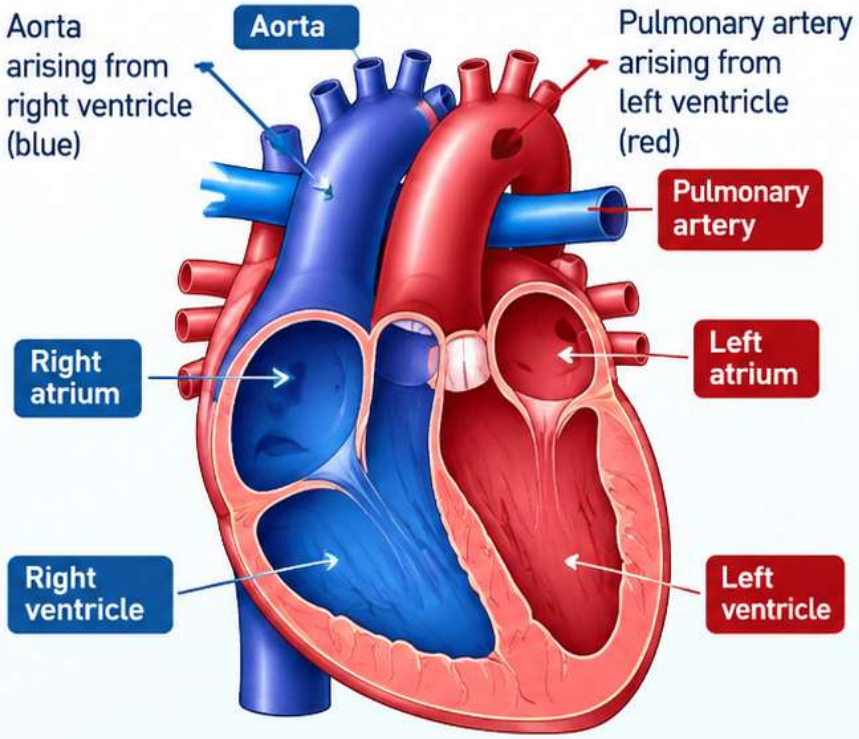
1. **Total anomalous pulmonary venous connection (TAPVC)**
   - Also shows pulmonary plethora and an oval-shaped heart (with narrow vascular pedicle).
2. **Truncus arteriosus**
   - May also present with an oval cardiac silhouette and increased pulmonary vascularity.

Key Teaching Point

- TGV presents with an **egg-shaped heart**, **narrow mediastinum** and **increased pulmonary** vascularity due to parallel circulation (pulmonary and systemic).
- Diagnosis is confirmed by **echocardiography**.

Transposition of the Great Vessels (TGV)

Anatomy of TGV



Parallel circulation: systemic and pulmonary circulations run in series, not in parallel.

Key Points

- TGV is the second most common cyanotic congenital heart disease (after TOF).
- It results from abnormal development of the conotruncal region, with the aorta arising from the right ventricle and the pulmonary artery from the left ventricle.
- Leads to two separate circuits: systemic (deoxygenated) and pulmonary (oxygenated).
- Survival depends on mixing of blood (via ASD, VSD, or PDA) and/or early intervention.

Clinical Features

- Cyanosis (usually appears within hours to days of birth).
- Tachypnea, poor feeding, and respiratory distress.
- Single, loud second heart sound (due to both great vessels arising from the ventricles).
- May present with shock if no mixing is present.

Diagnosis

- Chest X-ray: often normal or shows cardiomegaly with increased pulmonary vascular markings if mixing exists.
- Echocardiography: confirms the diagnosis and defines anatomy.

Important | **TGV is a duct-dependent lesion. Prostaglandin E1 may be required to keep the ductus arteriosus open and maintain pulmonary blood flow.**

Management of Transposition of the Great Arteries (TGA)

1. Initial Stabilization
(Immediately after diagnosis)

- Maintain oxygenation and perfusion to sustain mixing.
- Start prostaglandin E1 (PGE1)
  - 0.05–0.1 µg/kg/min (IV infusion)
  - Keep the ductus arteriosus open.
- Correct acidosis, hypoglycemia, anemia.
- Treat heart failure if present (diuretics, inotropes as needed).
- Monitor oxygen saturation and hemodynamics.

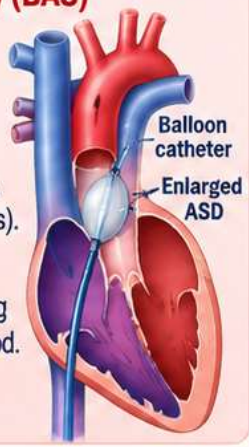
Target

- Ensure adequate mixing of pulmonary and systemic blood.
- Maintain systemic oxygen saturation (usually 75–85%).

2. Increase Mixing
(Emergency or early intervention)

A Balloon Atrial Septostomy (BAS)

- First-line procedure in most neonates with TGA.
- Performed urgently if there is inadequate mixing (severe cyanosis, metabolic acidosis, or low saturations).
- Creates/expands an interatrial communication (ASD) to allow mixing of oxygenated and deoxygenated blood.

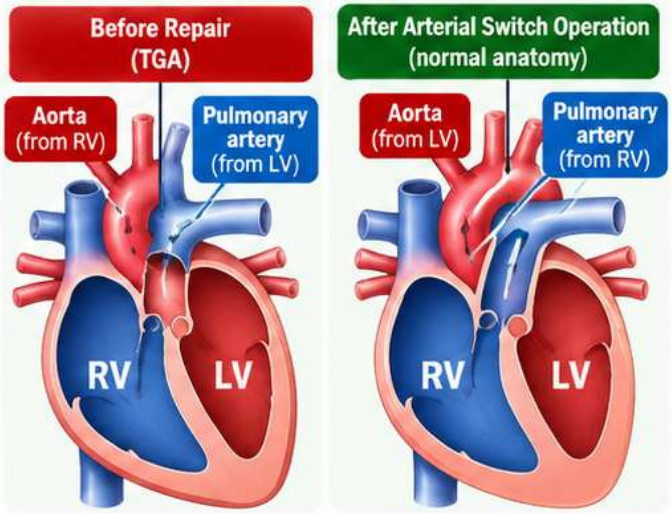


B Alternative / Additional Methods

- Surgical atrial septectomy (if BAS unsuccesful or not available).
- Maintain ductal patency with PGE1.
- In selected cases: stent atrial septostomy or ductal stenting (as a bridge).

3. Definitive Surgical Repair
(Arterial Switch Operation)

- Optimal timing: first 1–2 weeks of life (ideally within the first 2 weeks).
- Procedure: Arterial switch operation (ASO) with transfer of the coronary arteries to the neoaorta.
- Close the atrial septal defect (if present).
- Ligate and divide the ductus arteriosus.
- Most infants achieve excellent long-term outcomes with normal ventricular function and near-normal life expectancy.



Key Points

- Prostaglandin E1 keeps the ductus arteriosus open and is essential immediately after diagnosis.
- Balloon atrial septostomy is performed urgently if there is inadequate mixing.
- Arterial switch operation is the definitive treatment and should be performed early (preferably within 2 weeks of age).
- Long-term outcomes after arterial switch are excellent in most patients.

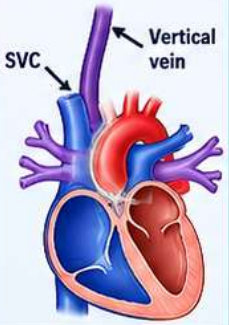
Total Anomalous Pulmonary Venous Drainage (TAPVD)

Definition and Epidemiology

- All pulmonary veins drain anomalously into the systemic venous circulation (not into the left atrium).
- Accounts for 1–2% of congenital heart disease.
- Usually associated with an **atrial septal defect (ASD)** which allows right-to-left shunting and survival.

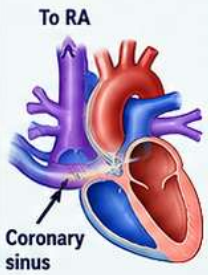
Types of TAPVD (with anatomical diagrams)

Supracardiac (40–50%)



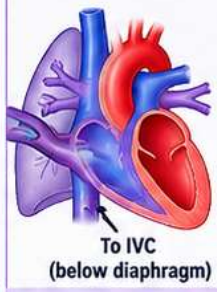
Pulmonary veins join a vertical vein draining to the SVC or brachiocephalic vein.

Cardiac (15–20%)



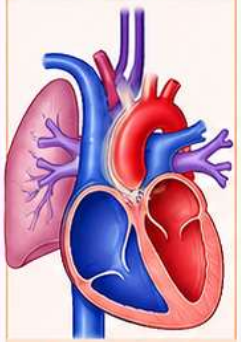
Pulmonary veins drain to the coronary sinus or directly to the right atrium.

Infracardiac (15–25%)



Pulmonary veins drain to the portal vein, hepatic veins or IVC (below diaphragm).

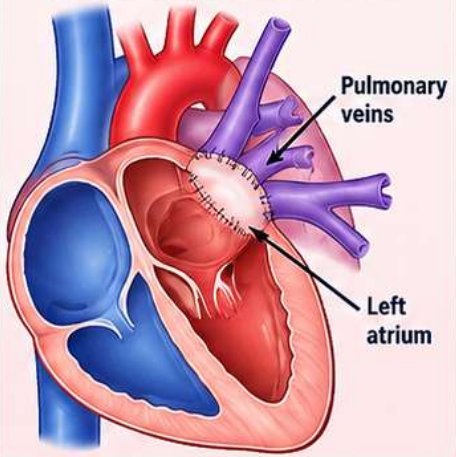
Mixed (5–10%)



Combination of two or more sites of drainage.

Surgical Repair

Anastomosis of the pulmonary venous confluence to the left atrium



Pathophysiology

- Pulmonary venous blood returns to the right atrium or systemic veins instead of the left atrium.
- Mixing of oxygenated and deoxygenated blood occurs at the atrial level → systemic desaturation.
- Pulmonary venous obstruction may be present (↑ pulmonary venous pressure → pulmonary edema).

Clinical Presentation

Unobstructed TAPVD

- **Cyanosis** (mild to moderate) in the first days to weeks of life.
- Tachypnea, failure to thrive, recurrent chest infections (due to high pulmonary flow).

Obstructed TAPVD (medical emergency)

- Severe cyanosis within hours to days of life.
- Respiratory distress and pulmonary edema.
- Metabolic acidosis, shock, rapid deterioration.

Diagnosis

- **Echocardiography (primary investigation):** confirms diagnosis, defines site of drainage, and assesses obstruction.
- **CT angiography:** useful when pulmonary venous anatomy is unclear or for preoperative planning.
- **Cardiac catheterization/ angiography:** rarely required (nowadays).

Management

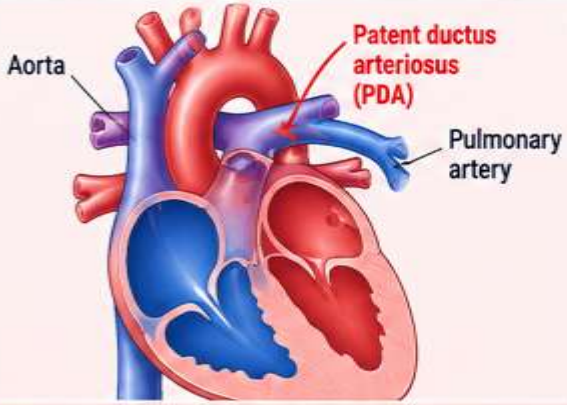
- **Stabilization:** oxygen, ventilation if needed, treat acidosis, diuretics for pulmonary edema, prostaglandin E1 not required.
- **Obstructed TAPVD:** urgent surgical repair.
- **Definitive treatment:** surgical anastomosis of the pulmonary venous confluence to the left atrium (removal of vertical vein or other anomalous channels).
- **Long-term outcome:** generally excellent with low late mortality. Watch for complications: pulmonary venous stenosis, arrhythmias, sinus node dysfunction.

Key Points

- TAPVD = all pulmonary veins drain to systemic venous circulation (not the left atrium).
- Presentation depends on presence of pulmonary venous obstruction (unobstructed: mild–moderate cyanosis; obstructed: severe, early).
- **Echocardiography** is the key diagnostic tool.
- Treatment is surgical: anastomosis of pulmonary venous confluence to left atrium (urgent if obstructed).
- Long-term survival after repair is good.

Patent Ductus Arteriosus (PDA)

Anatomy and Physiology



- The ductus arteriosus is a normal fetal communication between the pulmonary artery and the aorta.
- It allows right-to-left shunting of blood, bypassing the lungs.

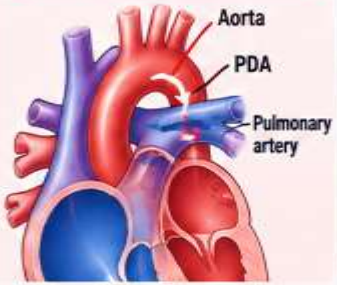
Normal Closure After Birth

1. Increase in oxygen saturation
2. Decrease in pulmonary vascular resistance
3. Reduced prostaglandin levels (triggering smooth muscle contraction) — PGE1

Functional closure within a few hours of birth and anatomical closure within 1–3 weeks.

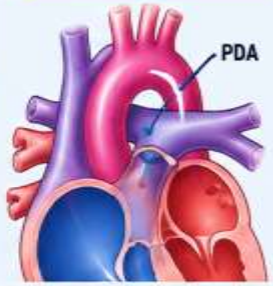
PDA Pathophysiology and Hemodynamics

Left-to-Right Shunt (most common)



- Occurs after birth due to higher aortic pressure.
- Causes pulmonary overcirculation and increased left heart volume load.
- Can lead to heart failure, pulmonary hypertension and ventricular dilatation if large.

Right-to-Left Shunt (uncommon)



- Occurs with severe pulmonary hypertension (Eisenmenger syndrome).
- Results in cyanosis and differential clubbing.

Clinical Presentation

Infants (large PDA)

- Tachypnea, respiratory distress
- Poor feeding and failure to thrive
- Recurrent chest infections
- Signs of heart failure (sweating, poor weight gain)

Older children and adults

- Often asymptomatic (small PDA)
- Exertional dyspnea, easy fatigability
- Palpitations
- Continuous machinery murmur

A continuous "machinery" murmur best heard at the left second intercostal space (under the clavicle).

Diagnosis



Echocardiography (primary investigation)

- Confirms the diagnosis
- Defines the size, shunt direction and hemodynamic effect
- Colour Doppler imaging



CT angiography

- Useful when anatomy is unclear or for pre-procedural planning

Medical Management (Indomethacin / Ibuprofen)



- Cyclo-oxygenase inhibitors reduce prostaglandin synthesis and promote ductal closure.
- Effective in many preterm neonates in the first few days of life.
- Contraindicated in renal failure, NEC, thrombocytopenia or if ductal closure is not desired (e.g. duct-dependent lesions).

Indications for Closure of PDA (in general)

1. Hemodynamically significant PDA (LV volume overload).
2. Symptomatic (heart failure, failure to thrive, respiratory infections).
3. Pulmonary hypertension (reversible).
4. Recurrent endarteritis or risk of endocarditis.
5. Large PDA (even if asymptomatic).
6. In selected cases before pregnancy or strenuous activity.

Methods of Closure

Transcatheter Closure (First choice in most cases)



- Umbrella or coil occlusion device.
- Preferred in suitable anatomy.
- High success rate and low complication rate.

Surgical Closure (when catheter closure is not feasible)



- Left thoracotomy.
- Ligation or division of the PDA.
- Preferred for very large PDA or very small infants, or unsuitable anatomy.

Outcomes

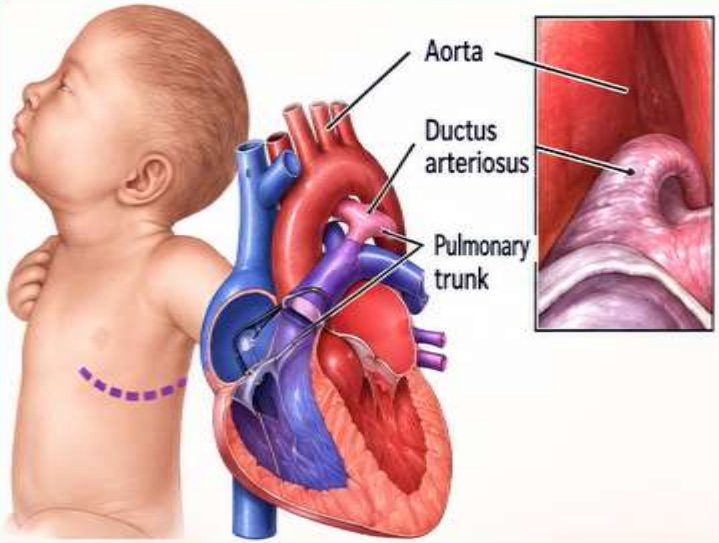
- Most PDAs are closed by preschool age.
- Early closure prevents long-term complications (heart failure, pulmonary hypertension, endarteritis, Eisenmenger syndrome).
- Operative mortality is low and long-term outcome is excellent.

Indications for Surgical Closure of Patent Ductus Arteriosus (PDA)

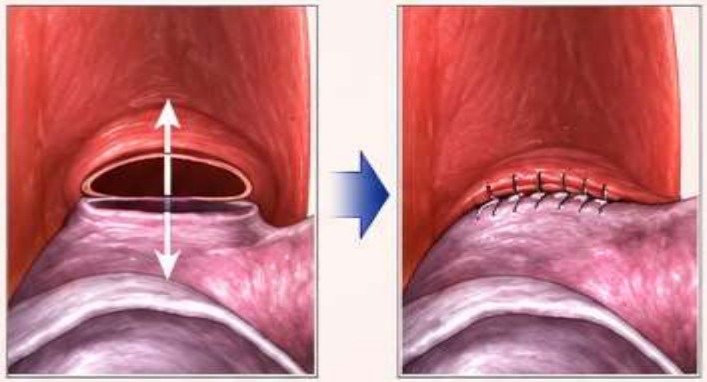
1. **Hemodynamically significant PDA**
   - Left ventricular volume overload with left atrial and left ventricular dilatation on echocardiography.
   - Cardiomegaly on chest X-ray.
   - Even if symptoms are mild, significant shunt physiology warrants closure to prevent long-term complications.
2. **Heart failure**
   - Congestive heart failure not responding adequately to medical therapy.
   - Tachypnea, feeding difficulty, sweating, or poor weight gain in infants.
3. **Pulmonary hypertension (reversible)**
   - PDA should be closed when pulmonary hypertension is **present but still reversible.**
   - Early closure prevents progression to Eisenmenger syndrome.
4. **Failure to thrive**
   - Poor growth and inadequate weight gain due to chronic volume overload.
   - Especially important in infants and young children.
5. **Recurrent respiratory tract infections**
   - Frequent chest infections due to pulmonary overcirculation.
   - Common indication in pediatric practice.
6. **Endarteritis or risk of endocarditis**
   - Previous or active infective endarteritis is an **absolute indication.**
   - Even small but audible PDAs are closed to prevent this complication.
7. **Large PDA**
   - Large PDAs should be closed even if the patient is asymptomatic.
   - Risk of heart failure and pulmonary hypertension is high.
8. **Symptomatic PDA in preterm neonates**
   - Medical treatment (indomethacin or ibuprofen) fails.
   - NSAIDs are contraindicated (e.g. renal failure, NEC, thrombocytopenia).
   - The infant remains ventilator-dependent.
9. **When catheter closure is not feasible**
   - PDA anatomy is unsuitable for device closure.
   - Device closure has failed or caused complications.
   - Surgical closure via left thoracotomy (ligation or division) is preferred.

Patent Ductus Arteriosus (PDA)

Anatomy and Fetal Circulation



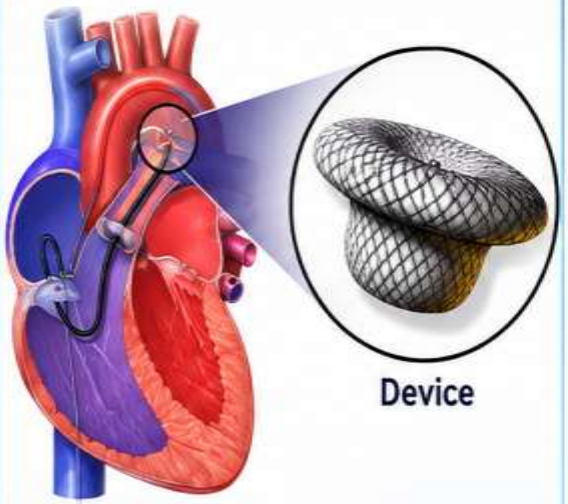
Postnatal Closure of the Ductus Arteriosus



The aorta and pulmonary trunk are separated (during fetal life)

The open ends are closed (after birth)

Transcatheter Device Closure of PDA (Amplatzer Device)

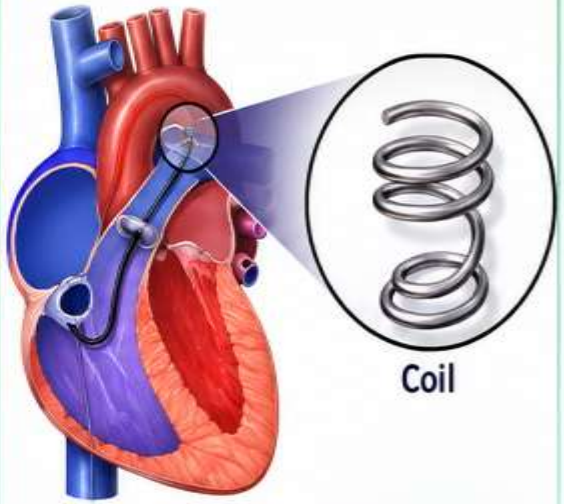


Device Closure of PDA

Amplatzer duct occluder device



Transcatheter Coil Closure of PDA

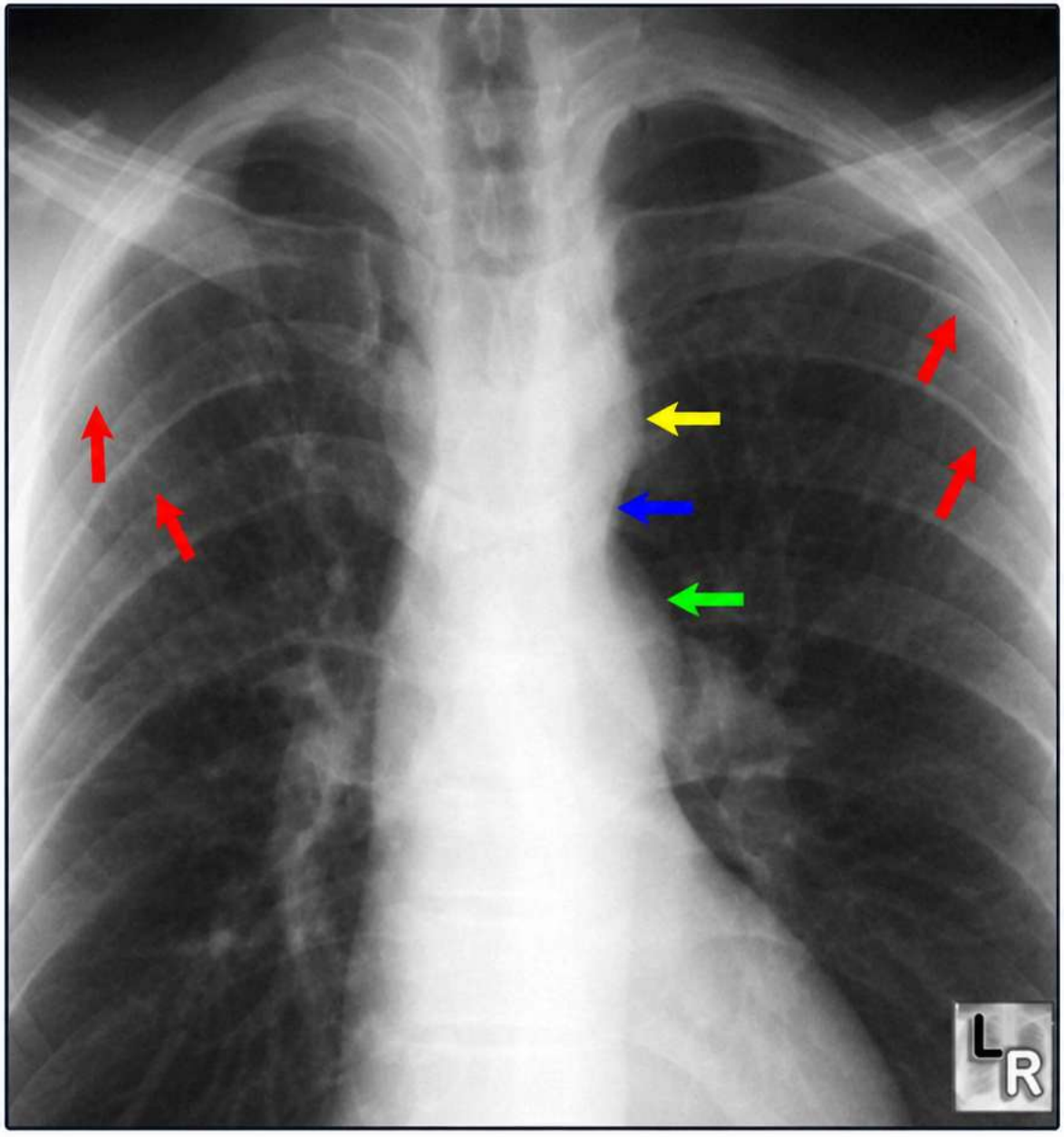


Coil Closure of PDA

Coil device



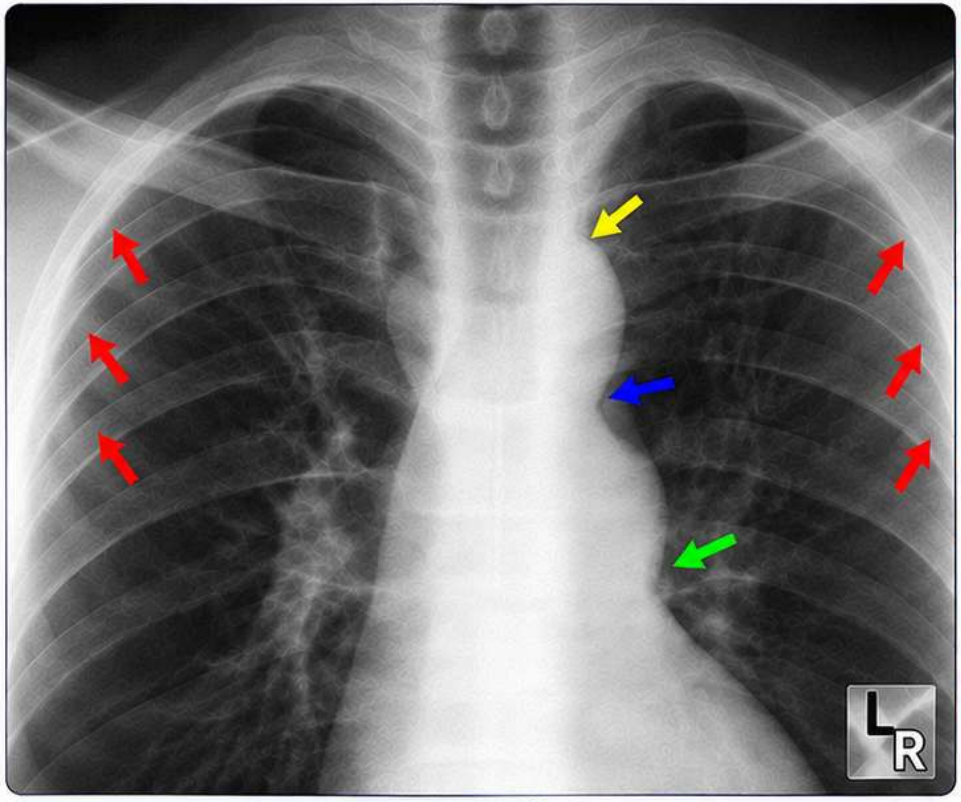
CHALLENGE QUESTION – CHEST X-RAY

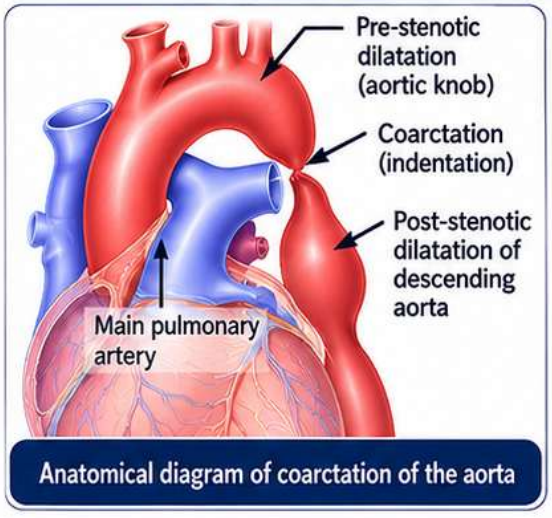


1. Describe the abnormal findings in this chest X-ray.
2. Identify the structures indicated by the yellow, blue and green arrows.
3. What do the red arrows indicate?
4. What congenital heart disease is suggested by these findings?
5. Explain the pathophysiological basis for these radiographic findings.
6. What other investigations would help confirm the diagnosis?

CHALLENGE QUESTION – CHEST X-RAY

MODEL ANSWER





Anatomical diagram of coarctation of the aorta

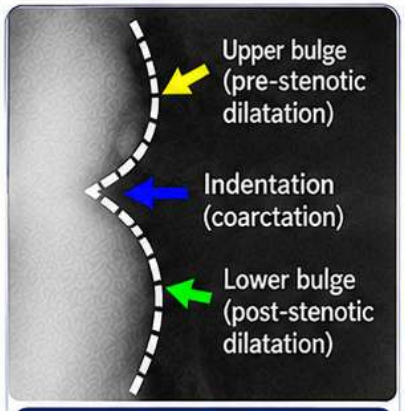


Figure 3 sign
(enlarged view of left mediastinal border)

1 Abnormal findings in this chest X-ray

- **Bilateral rib notching**, more prominent in the inferior ribs (red arrows).
- **Indentation of the aortic knob/left upper mediastinal border** (yellow arrow).
- Prominent main pulmonary artery segment (blue arrow).
- Prominent descending aortic contour below the indentation (green arrow).
- Heart size is usually normal or mildly increased.

2 Identify the structures indicated by the arrows

- **Yellow arrow** – Aortic knob / pre-stenotic dilatation of the aorta.
- **Blue arrow** – Main pulmonary artery (prominent segment).
- **Green arrow** – Post-stenotic dilatation of the descending aorta.
- **Red arrows** – Rib notching due to enlarged intercostal collateral arteries.

3 What do the red arrows indicate?

- Rib notching due to dilated intercostal collateral arteries (classically involving the inferior margins of ribs 3–8) secondary to long-standing coarctation of the aorta.

4 What congenital heart disease is suggested by these findings?

- **Coarctation of the aorta (CoA),** post-ductal (usually just distal to the origin of the left subclavian artery).

5 Explain the pathophysiological basis for these radiographic findings

- Narrowing of the aorta distal to the left subclavian artery → increased resistance to blood flow to the lower body → proximal hypertension (upper limbs, head and neck) and reduced perfusion to the lower body.
- Development of collateral circulation (intercostal, internal thoracic, subscapular arteries) to supply the distal aorta → dilated, tortuous intercostal arteries causing rib notching.
- Pre-stenotic dilatation of the aortic arch (aortic knob) and post-stenotic dilatation of the descending aorta, with an intervening indentation at the site of coarctation, produce the classic **'Figure 3 sign'**.
- Increased afterload → left ventricular hypertrophy (which may not always increase heart size on CXR).

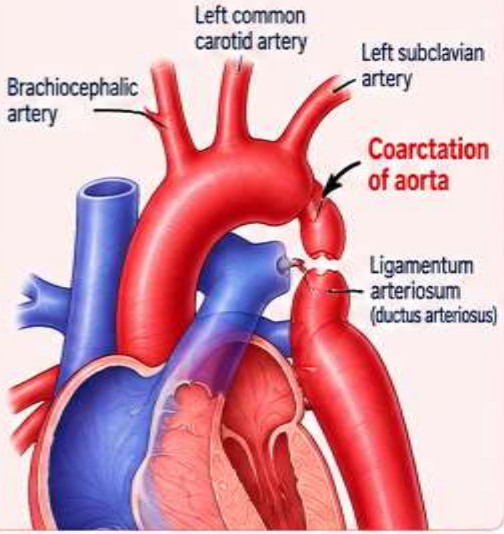
6 Other investigations to confirm the diagnosis

- **Echocardiography with Doppler** – demonstrates the site of narrowing and pressure gradient.
- **CT angiography or MR angiography** – defines anatomy and collateral vessels.
- **Blood pressure measurement** – higher in upper limbs, lower in lower limbs.

Coarctation of the Aorta (CoA)

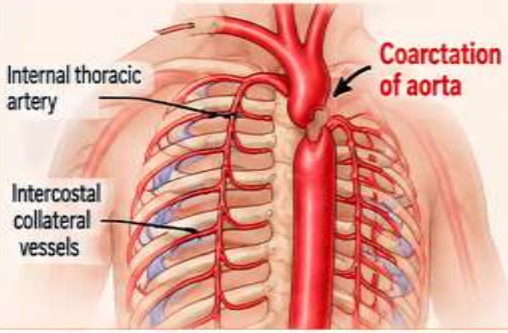
Definition and Anatomy

- Narrowing of the aorta just distal to the origin of the left subclavian artery, usually at the site of ductus arteriosus insertion (juxtaductal).
- Often associated with PDA, VSD, and bicuspid aortic valve.



Pathophysiology

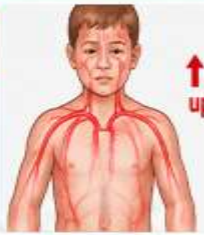
- Narrowing leads to increased resistance to blood flow to the lower body.
- Proximal to the coarctation: increased pressure and flow to upper body, brain and coronary circulation.
- Distal to the coarctation: reduced perfusion to lower body and organs (kidneys, abdomen, lower limbs).
- Development of collateral circulation (intercostal, internal thoracic, subscapular) to supply distal aorta.
- Left ventricular pressure overload → left ventricular hypertrophy.



Clinical Features

Upper body findings (proximal hypertension)

- Raised blood pressure in upper limbs (> lower limbs).
- Headache, dizziness, epistaxis (in older children/adults).
- Increased perfusion to brain and face.



Lower body findings (distal hypoperfusion)

- Lower blood pressure in lower limbs.
- Weak or delayed femoral pulses (radio-femoral delay).
- Cool lower limbs, claudication (in older children).
- Reduced perfusion to kidneys → renin-mediated hypertension.



Other features

- Visible dilated intercostal vessels due to collateral circulation (more obvious when the patient leans forward).
- Left ventricular hypertrophy on examination and ECG/Echo.
- Differential cyanosis is uncommen, but may occur in neonates with a PDA and right-to-left ductal shunting.



Associated Lesions

- Patent ductus arteriosus (PDA)
- Ventricular septal defect (VSD)
- Bicuspid aortic valve
- Other arch anomalies (e.g. interrupted aortic arch, hypoplastic arch)
- Turner syndrome



Investigations

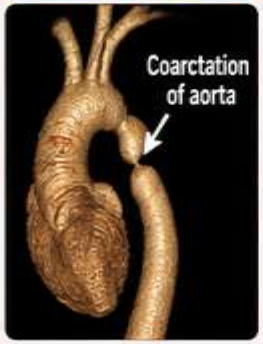
- Upper and lower limb blood pressure (> 20 mmHg difference).
- Weak/delayed femoral pulses.
- Echocardiography (initial test).
- CT angiography or MRI (delineates anatomy, especially in older children and adults).
- Chest X-ray: rib notching due to collateral vessels (in older patients).
- ECG: left ventricular hypertrophy.

Imaging

Echocardiography



CT angiography

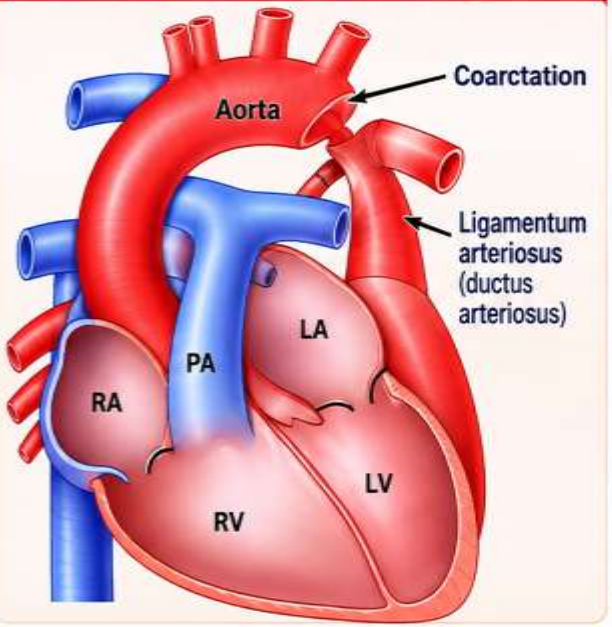


Management

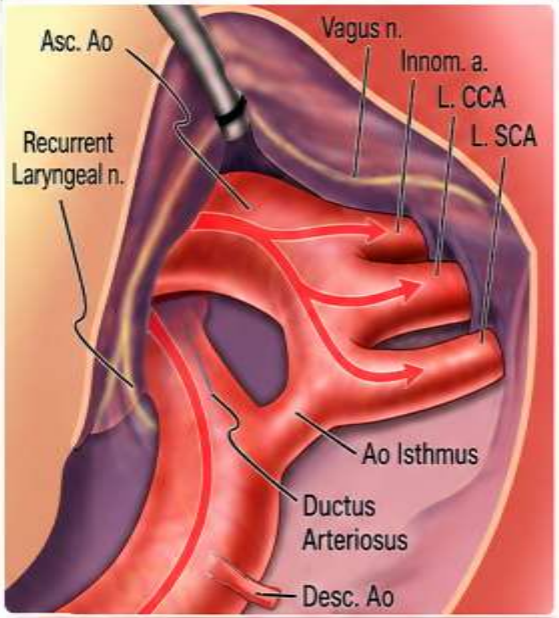
- **Indications:** significant gradient (≥ 20 mmHg), hypertension, symptoms, or significant anatomical narrowing.
- **Methods:**
  - Surgical end-to-end anastomosis (resection with primary anastomosis) – preferred in neonates and children.
  - Transcatheter balloon angioplasty with or without stent – often used in older children and adults.
- Long-term follow up for recoarctation, hypertension, and associated lesions.

Coarctation of the Aorta (CoA)

Anatomy and Site of Narrowing



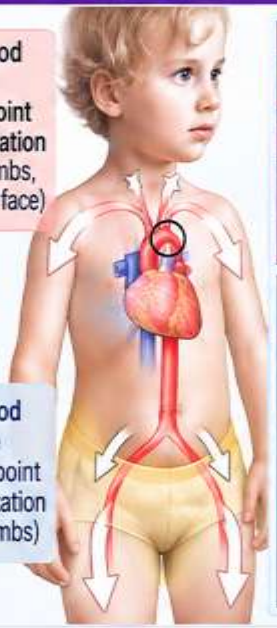
Aortic Arch and Collateral Circulation

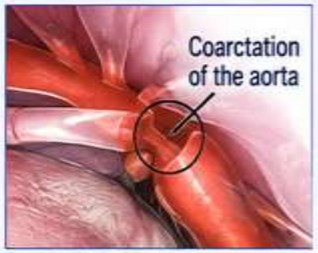


Clinical Features

High blood pressure before point of coarctation (upper limbs, head and face)

Low blood pressure beyond point of coarctation (lower limbs)



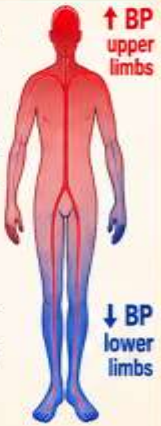




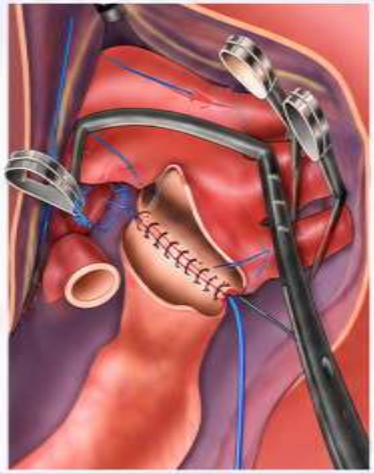
Dilated intercostal vessels (collaterals) (more obvious when leaning forward)

Pathophysiology and Effects

- Narrowing of the aorta → increased resistance to blood flow to the lower body.
- Proximal to the coarctation: increased pressure and flow to upper body, brain and coronary circulation.
- Distal to the coarctation: reduced perfusion to lower body and organs (kidneys, abdomen, lower limbs).
- Development of collateral circulation (intercostal, internal thoracic, subscapular) to supply distal aorta.
- Left ventricular pressure overload → left ventricular hypertrophy.



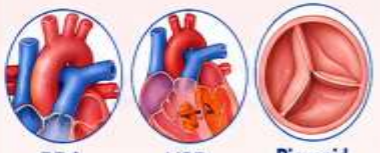
Surgical Treatment (Resection and End-to-End Anastomosis)





Associated Lesions

- Patent ductus arteriosus (PDA)
- Ventricular septal defect (VSD)
- Bicuspid aortic valve
- Other arch anomalies (e.g. interrupted aortic arch, hypoplastic arch)
- Turner syndrome

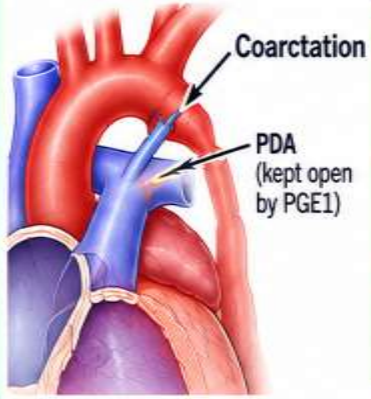


Treatment of Coarctation of the Aorta (CoA)

1 When is treatment indicated?

- Systolic arm–leg blood pressure gradient ≥ 20 mmHg
- Significant anatomical narrowing on imaging (even with lower gradient)
- Symptoms (heart failure, poor perfusion)
- Left ventricular hypertrophy

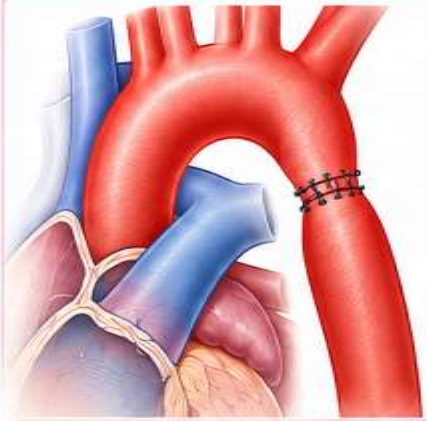
2 Neonates with critical CoA (duct-dependent)



- Start **Prostaglandin E1** (alprostadil) to keep ductus arteriosus patent
- Stabilize circulation (treat shock, heart failure, acidosis)
- **Urgent surgical repair** after stabilization

3 Definitive treatment options

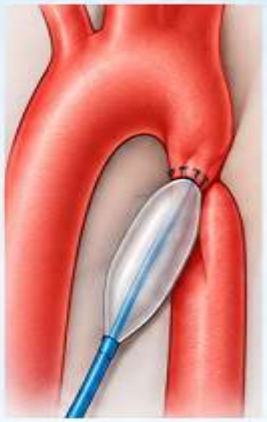
A. Surgical repair



- **Resection with end-to-end anastomosis** (most common)
- **Extended end-to-end anastomosis** in infants/children
- **Subclavian flap aortoplasty** (selected cases)
- **Patch aortoplasty or interposition graft** (for long-segment/complex lesions)

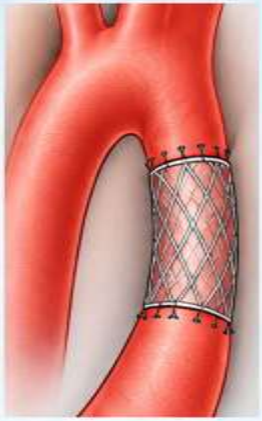
B. Endovascular therapy

Balloon angioplasty



- Useful for recoarctation (after surgical repair)
- Also used in selected native coarctation (particularly in children)

Stent implantation



- Preferred in older children, adolescents and adults
- Used for native or recurrent coarctation

4 Follow-up

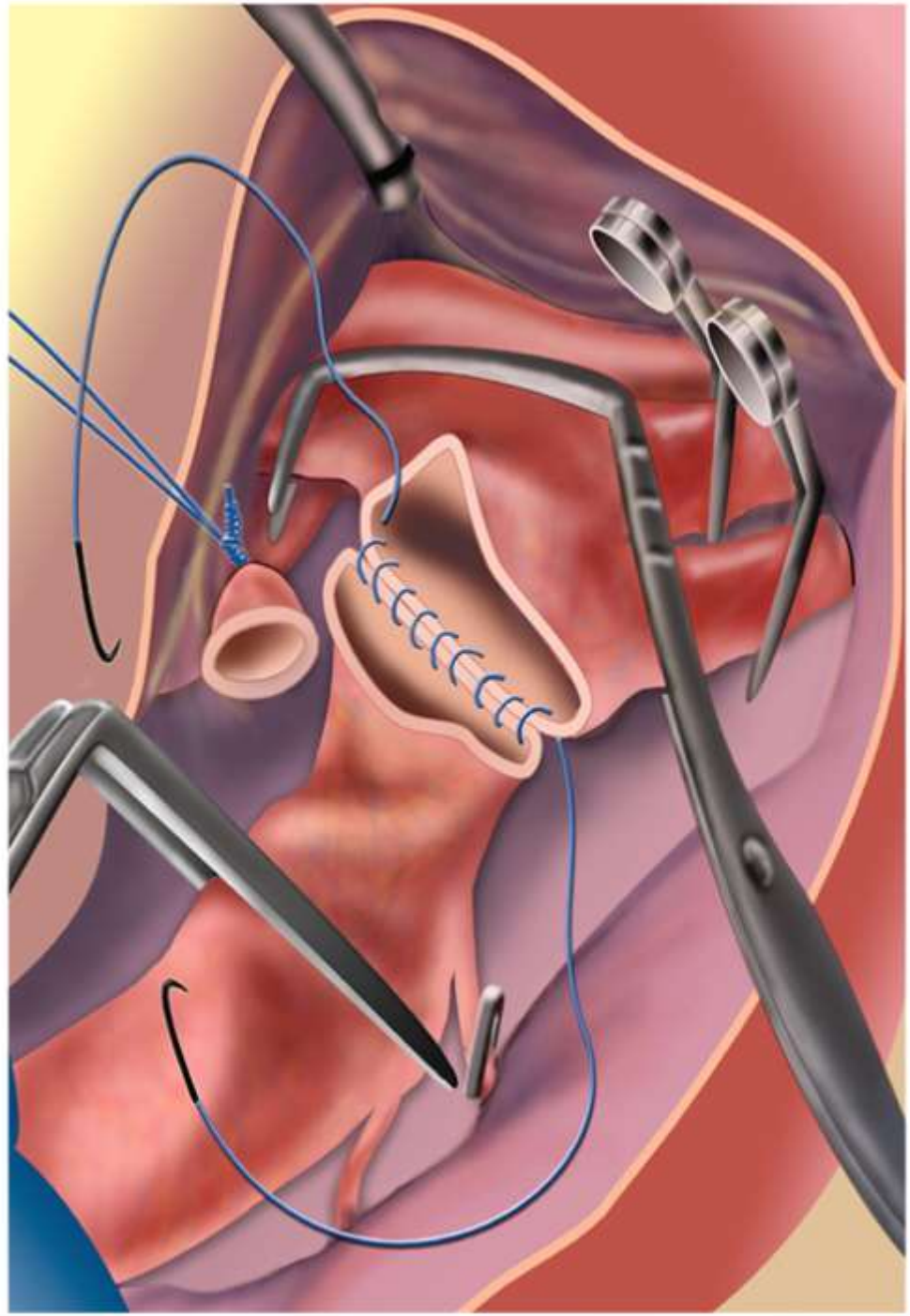
- Lifelong follow-up
- Regular blood pressure monitoring (both arms and legs)
- Periodic imaging (Echocardiography, CT or MR angiography)
- Assess for recoarctation, aneurysm and associated lesions (e.g. bicuspid aortic valve)

5 Complications

- Recoarctation or residual obstruction
- Persistent or recurrent hypertension
- Aortic aneurysm or dissection
- Associated bicuspid aortic valve disease
- Post-surgical complications (e.g. recurrent laryngeal nerve injury, chylothorax – uncommon)





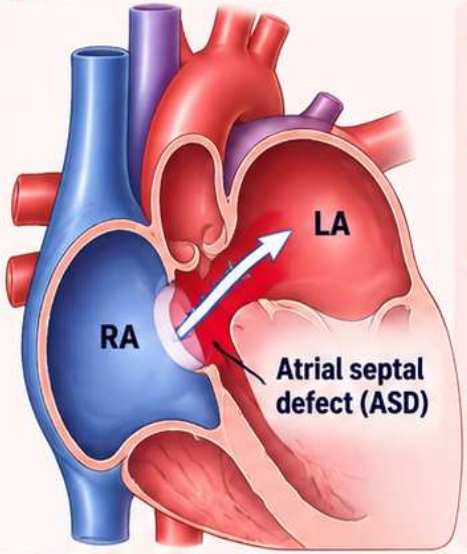




Atrial Septal Defect (ASD)

6–8% of congenital heart defects

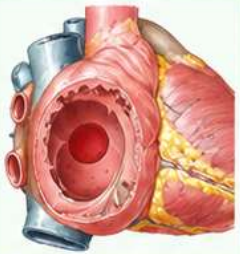
1 Definition and haemodynamics



- A hole in the atrial septum allowing communication between left and right atrium.
- Blood flows from left atrium to right atrium (left → right shunt).
- Causes increased blood flow to right heart and lungs → right heart enlargement and eventual pulmonary hypertension (reversible if early).

2 Types of ASD

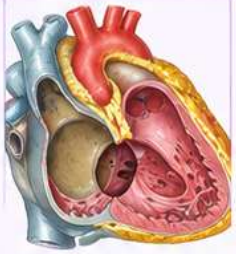
Secundum type (most common)



Ostium secundum defect

- 70% of ASDs
- Oval, in the middle of the atrial septum (fossa ovalis)

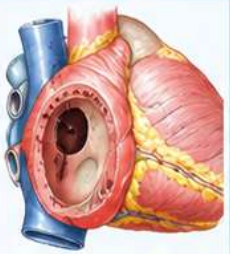
Primum type (less common)



Ostium primum defect

- 15–20% of ASDs
- Lower part of the atrial septum (associated with AV valve abnormalities)

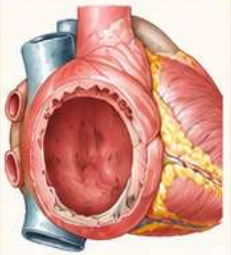
Sinus venosus type (uncommon)



Sinus venosus defect

- 5–10% of ASDs
- Near the opening of superior vena cava (SVC), often with anomalous pulmonary veins

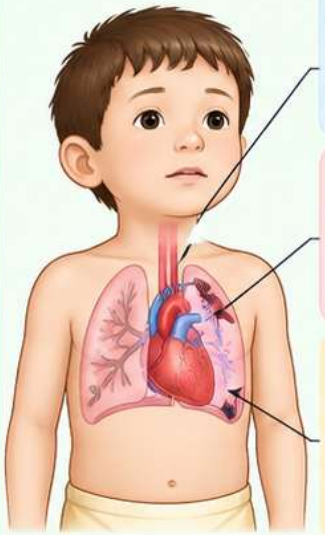
Common atrium (rare)



Common atrium

- Very rare
- Large defect with single atrial cavity

3 Clinical features



Symptoms

- Often asymptomatic in childhood
- Dyspnoea on exertion (older children)
- Exercise intolerance

Signs

- Fixed split second heart sound (wide)
- Systolic ejection murmur at upper left sternal border (due to ↑ flow across pulmonary valve)

Complications (late)

- Pulmonary hypertension
- Right heart failure
- Atrial arrhythmias (e.g. atrial fibrillation)
- Paradoxical embolism (rare)

4 Diagnosis



Echocardiography (with Doppler)

- Shows the defect
- Detects left → right shunt and right heart enlargement



Chest X-ray

- Cardiomegaly (right heart enlargement)
- Increased pulmonary vascular markings



Cardiac catheterisation (when needed)

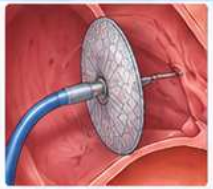
- Measures shunt (Qp/Qs)
- Defines anatomy before closure

5 Treatment

Indications for closure

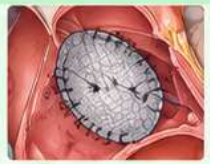
- Significant left → right shunt
- (Qp/Qs ≥ 1.5:1)
- Right heart enlargement
- Symptoms
- To prevent long-term complications

A. Transcatheter closure (most secundum ASD)



- Preferred for suitable secundum ASD
- Less invasive

B. Surgical closure



- Used for primum ASD, sinus venosus ASD, common atrium or unsuitable defects
- Direct suture or patch (pericardium or Dacron)

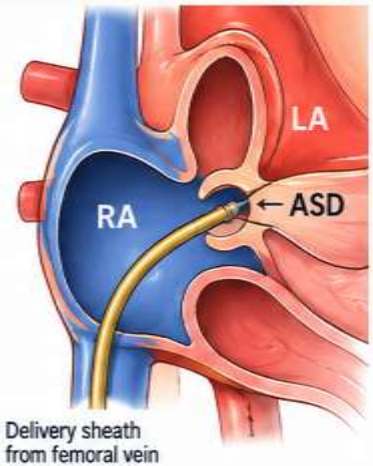
Treatment of Atrial Septal Defect (ASD)

Indications for closure

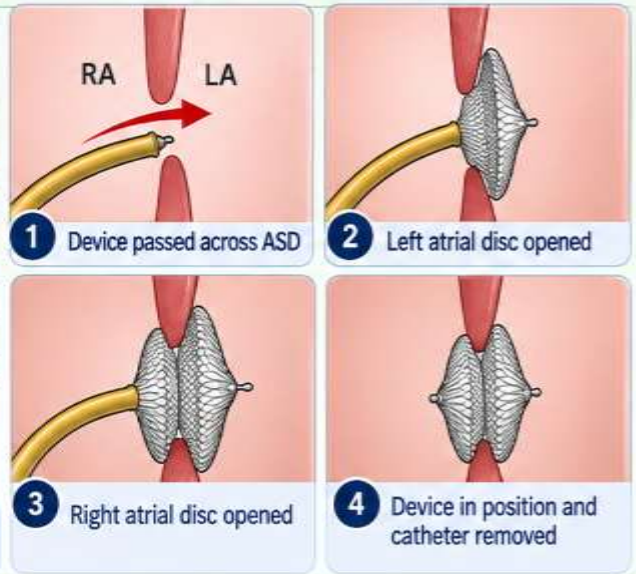
- Significant left → right shunt (Qp/Qs ≥ 1.5:1)
- Right heart enlargement
- Symptoms (dyspnoea, exercise intolerance)
- To prevent long-term complications (pulmonary hypertension, arrhythmias, paradoxical embolism)

1 Transcatheter closure

- Preferred for suitable secundum ASD
- Performed via femoral vein under echocardiographic and fluoroscopic guidance
- Device (e.g. Amplatzer septal occluder) is deployed across the defect



Catheter across atrial septal defect

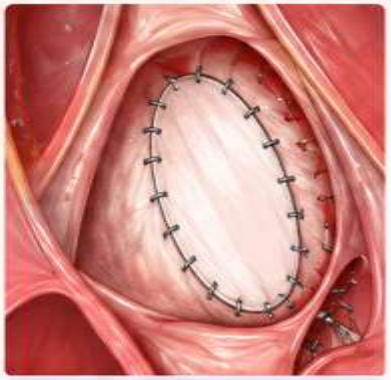


2 Surgical closure

- Indicated when ASD is large, non-secundum (e.g. primum, sinus venosus), multiple defects, or unsuitable for device closure
- Performed via open heart surgery (usually with cardiopulmonary bypass)
- Closed by direct suturing or using a patch (pericardium or Dacron)



Intraoperative view of ASD (secundum type)



Surgical closure using pericardial/Dacron patch

Key points

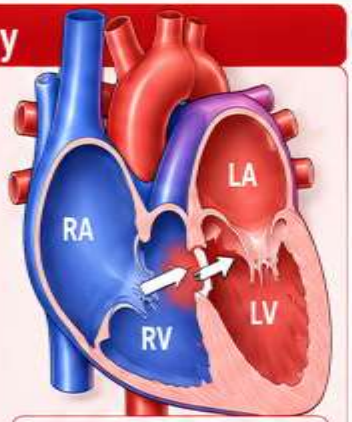
- **Transcatheter device closure** is the treatment of choice for most secundum ASDs.
- Surgical closure is used for large, non-secundum, multiple ASDs or when device closure is not feasible.
- Closure prevents right heart failure, pulmonary hypertension, arrhythmias and paradoxical embolism.

Ventricular Septal Defect (VSD)

20–30% of congenital heart defects

1 Definition and pathophysiology

- A defect (hole) in the ventricular septum allowing blood to flow from left ventricle (LV) to right ventricle (RV) (left → right shunt).
- Causes increased pulmonary blood flow → pulmonary hypertension and ventricular hypertrophy.



Ventricular septal defect (VSD)

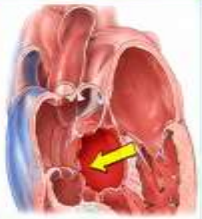
2 Types of VSD

Perimembranous (conoventricular)



- Most common (70–80%)
- Located in the membranous septum, may extend to tricuspid or aortic valve

Muscular (trabecular)



- About 10%
- Located in the muscular part of the septum
- Can be multiple

Atrioventricular (inlet)



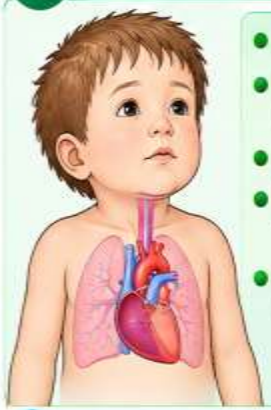
- About 5%
- Located in the atrioventricular canal beneath the tricuspid valve

Subarterial (outlet)



- About 5–10%
- Located in the conal septum immediately beneath the pulmonary valve annulus

3 Clinical features



- Often asymptomatic in small VSD.
- Pansystolic murmur (loudest at left lower sternal border).
- Palpable thrill.
- Wide split second heart sound with pulmonary accentuation.
- Symptoms: dyspnoea, exercise intolerance, recurrent respiratory infections (large VSD).

4 Investigations

Echocardiography (with Doppler)



- Shows the site and size of the defect.
- Demonstrates left → right shunt.
- Assesses right heart enlargement and pulmonary pressure.

Chest X-ray



- Cardiomegaly (right heart enlargement)
- Increased pulmonary vascular markings

Cardiac catheterisation (when needed)



- Measures shunt (Qp/Qs).
- Defines pulmonary pressure and anatomy.

5 Treatment

Spontaneous closure

- 40–50% of small VSDs close spontaneously (especially in the first years of life).
- Chance of closure is inversely related to age (e.g. ~80% at 1 month, ~25% at 12 months).

Indications for closure

- Large defect with significant left → right shunt (Qp/Qs ≥ 1.5:1).
- Right heart enlargement.
- Symptoms (dyspnoea, failure to thrive).
- To prevent long-term complications (pulmonary hypertension, arrhythmias, endocarditis).

6 Methods of closure

Surgical closure



- Used for large VSDs, multiple defects or when device closure is not suitable.
- Closed by direct suturing or using a patch (pericardium or Dacron).

Surgical closure using pericardial patch

Transcatheter device closure



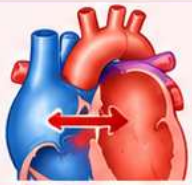
Transcatheter closure using an occluder device (e.g. Amplatzer)

7 Complications (long-term)

- Pulmonary hypertension and Eisenmenger's syndrome (in large, untreated VSD).
- Right heart failure.
- Recurrent respiratory infections.
- Aortic regurgitation (especially with subarterial VSD).
- Arrhythmias.
- Infective endocarditis.
- Residual shunt (after closure).

Indications for Surgical Closure of VSD

1



Large VSD with significant left → right shunt

- Qp/Qs ≥ 2:1
- Evidence of left heart volume overload (left atrial and left ventricular enlargement)

2



Symptoms despite medical therapy

- Heart failure (tachypnoea, poor feeding, poor weight gain)
- Recurrent respiratory infections

3



Evidence of left ventricular enlargement

- Progressive LV dilatation on echocardiography

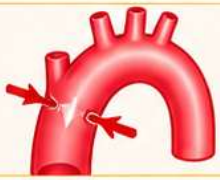
4



Raised pulmonary artery pressure but still reversible

- Pulmonary vascular resistance < 6 Wood units
- or < two-thirds of systemic vascular resistance

5



Development of aortic regurgitation

- Due to a subarterial (outlet) VSD (to prevent progression)

6



Failure to thrive or persistent significant symptoms

- Despite optimal medical management

7



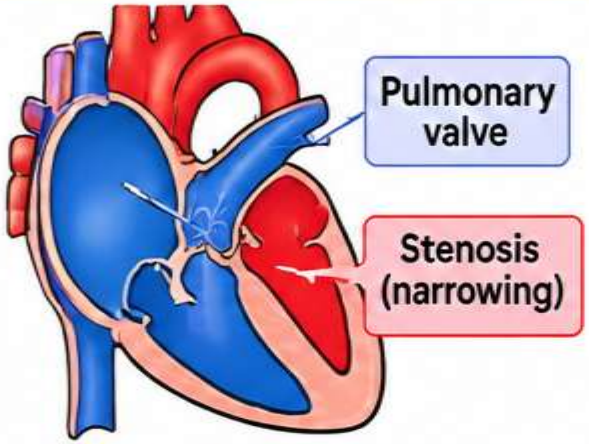
To prevent long-term complications

- Pulmonary hypertension, arrhythmias, infective endocarditis

Pulmonary Stenosis (PS)

What is it?

Narrowing of the pulmonary valve or RV outflow tract → ↑ pressure in RV and less blood to lungs.



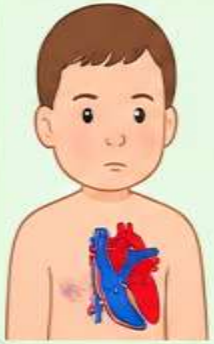
Types

- **Valvular PS** (most common)
- **Supravalvular PS** (above the valve)
- **Branch PS** (R or L pulmonary artery)
- **Peripheral PS** (distal branches)

Stenotic pulmonary valve



Clinical Features



- Often asymptomatic (mild cases)
- Murmur (ejection type, LUSB)
- RV hypertrophy (later)
- Symptoms if severe: dyspnea, cyanosis, fatigue

Investigations

- **Echocardiography** (with Doppler): shows gradient across valve
- **Cardiac catheterisation** (if needed)
- **CXR:** may show post-stenotic dilatation of PA

Indications for Surgery / Intervention

- Large gradient (usually > 50–60 mmHg)
- Symptoms (dyspnea, cyanosis, fatigue)
- RV hypertrophy / dysfunction
- Progressive increase in gradient
- Valvular morphology not suitable for balloon valvuloplasty

Treatment Options

Balloon valvuloplasty (first choice)



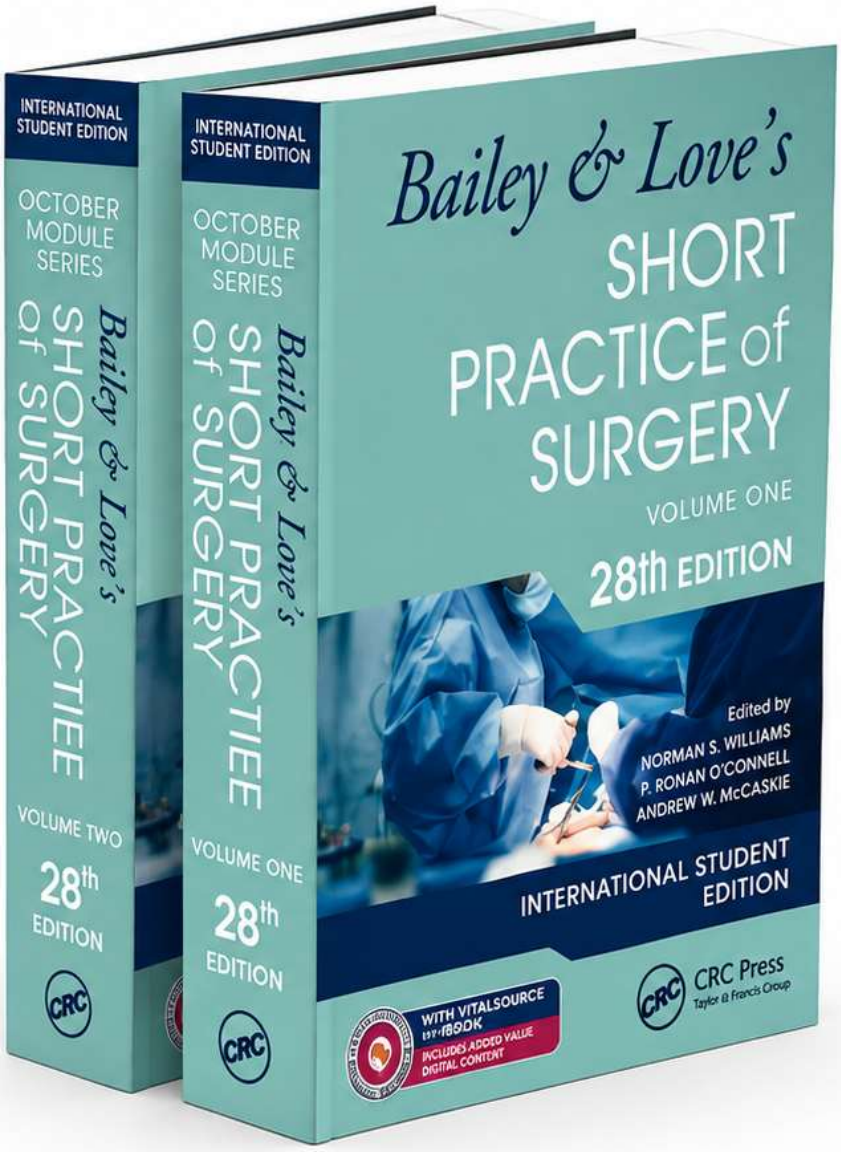
Surgery (if not suitable for balloon)



Key Points

- ✓ PS is one of the common congenital heart lesions.
- ✓ Most patients are asymptomatic.
- ✓ Treatment: balloon valvuloplasty for suitable cases.
- ✓ Surgery if severe or not amenable to balloon.

Reference



Textbook

Bailey & Love's Short Practice of Surgery

28th Edition

Editors

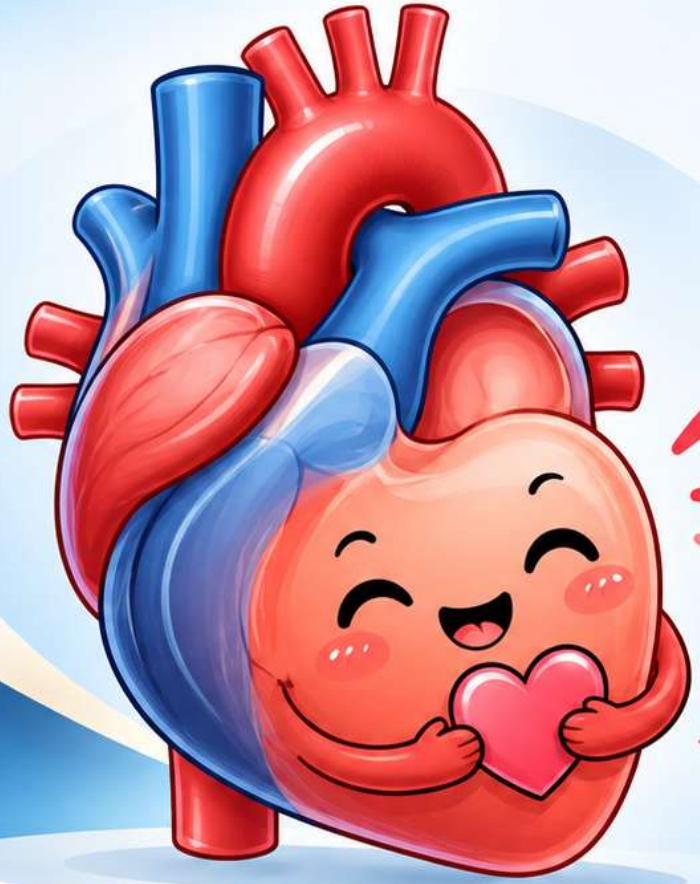
Norman S. Williams,
P. Ronan O'Connell,
Andrew W. McCaskie

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Edition

International Student Edition, 28th Edition



Thank You

for your attention

from the apex of my heart ♡



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