

# Congenital Heart Diseases



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# Paediatric Cardiology

- Congenital heart disease

- Acquired heart disease

## Congenital heart diseases

Commonest single group of congenital abnormalities

Accounts for 30% of all congenital abnormalities

Incidence - 8 / 1000 - live births

# Congenital Heart Defects

- Acyanotic
- Cyanotic heart defect

# Acyanotic Heart Diseases

- ✦ Ventricular septal defect
- ✦ Patent ductus arteriosus
- ✦ Atrial septal defect
- ✦ Pulmonary stenosis
- ✦ Coarctation of aorta
- ✦ Aortic stenosis

# Acyanotic Congenital Heart Defects: left to right shunts

✦ Ventricular septal defect

✦ Patent ductus arteriosus

✦ Atrial septal defect

- Associated with Increased pulmonary blood flow

# Acyanotic Congenital Heart Disease: obstructive lesions

- Pulmonary stenosis
- Aortic stenosis
- Coarctation of Aorta

# Cyanotic Congenital Heart Defects

✦ Tetralogy of Fallot

✦ Transposition of great arteries

- Pulmonary atresia
- Tricuspid atresia
- Double outlet right ventricle
- Ebstein anomaly of tricuspid valve
- Total anomalous pulmonary venous drainage
- Hypoplastic left heart syndrome

# Cyanotic heart defects with reduced pulmonary blood flow

- Tetralogy of Fallot
- Pulmonary atresia
- Tricuspid atresia
- Double outlet right ventricle
- Ebstein Anomaly of tricuspid valve



# Cyanotic defects associated with increased pulmonary blood flow

- Transposition of great arteries
- Total anomalous pulmonary venous drainage
- Truncus arteriosus
- Hypoplastic left heart syndrome

# Congenital Heart Diseases

Aetiology- what caused it?

- ✦ Large majority - no cause can be identified
- ✦ Genetic factors -
  - Chromosomal abnormalities
  - Down / turner syndrome
  - Certain defects – ASD - tendency to run in families

# Aetiology

## Environmental factors in utero

### Maternal Drugs

Alcohol - VSD,PDA  
Phenytoin  
Antidepressants

### Maternal Infections

Rubella, herpes simplex,  
Cytomegalovirus

### Maternal illness

Diabetes mellitus,  
Systemic Lupus Erythematosus.

# Congenital Heart Diseases

## Diagnosis

## When?

### Asymptomatic

- ★ Neonatal period - postnatal examination
- ★ In infancy - routine examination – during immunization
- ★ Coincidental finding when examining for another complaint – Upper Respiratory Tract Infections

### Symptomatic

- ★ Failure to thrive, recurrent chest infections
- Cyanosis

# Diagnosis

- History      Examination      Investigations

## Examination

- Inspection –                      Look
- Palpation –                      Feel
- Auscultation -                      Listen

# Diagnosis - History

- Antenatal period – maternal infections etc.
- Perinatal period – premature delivery, cyanosis, shortness of breath
- Infancy – cyanosis, shortness of breath on feeding, recurrent chest infections

# Examination

## Inspection - Look

- Failure to thrive - growth parameters
- Dysmorphic features
- Cyanosis
- Clubbing
- Breathlessness
- Precordial bulge
- Harrison sulci
- Ankle/sacral odema
- Head sweating
- Dental caries

# Examination

## Palpation - Feel

- Pulses      radial & femoral pulses
- Measurement of blood pressure
- Precordium –
  1. Apex beat
  2. Parasternal heave
  3. Thrills( palpable murmur)
  4. Heart sounds - palpable 2<sup>nd</sup> heart sound in the pulmonary area
- Abdomen
  - Tender Hepatomegaly – heart failure
  - Splenomegaly – infective endocarditis



# Examination

## Auscultation - Listen

- Heart sounds    Normal ?  
    Abnormal -            loud/soft  
    fixed split of 2<sup>nd</sup> heart    sound
- Additional sounds murmurs ?  
    systolic -    pan/ejection systolic  
    diastolic -    mid/early diastolic
- Lungs – basal crepitations in heart failure

# Congenital Heart Diseases

## Investigations -

CXR- telechest

ECG

Echocardiography

Cardiac Catheterization

# Acyanotic Defects

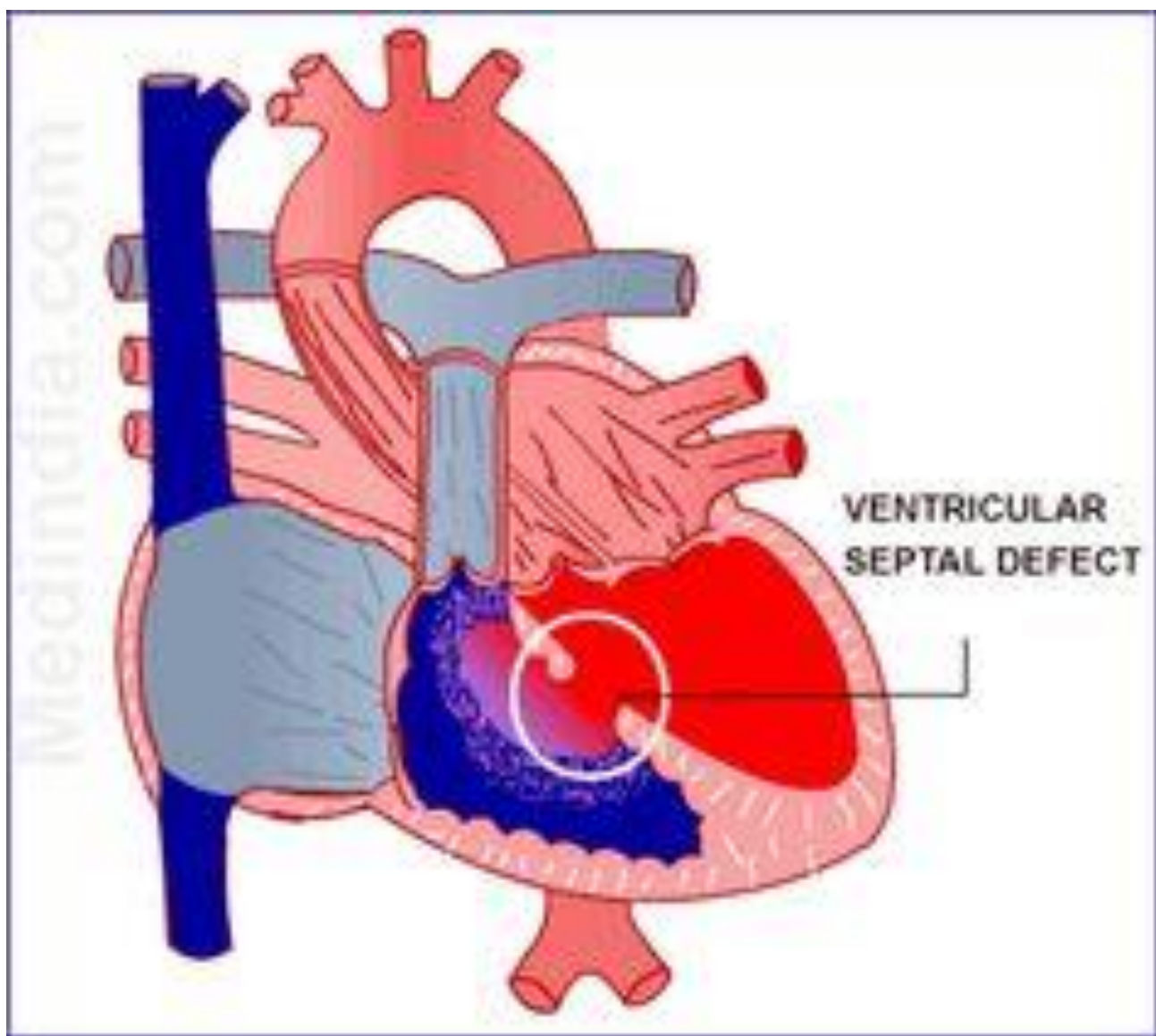
✦ Left to right shunts

ASD, VSD, PDA

# Acyanotic Defects: Left to right shunts

## VENTRICULAR SEPTAL DEFECT

- ⦿ Commonest congenital heart defect
- ⦿ Presentation - symptoms and signs vary with size of defect
- ⦿ Small VSD – asymptomatic  
Murmur found during examination



# Ventricular Septal Defect

## Large Defect

Presentation usually in the first 2 months of life

### Symptoms/signs

- ⦿ Difficulty in feeding
- ⦿ Failure to thrive, recurrent chest infections
- ⦿ Heart failure- dyspnoea, basal crepitations  
oedema, tender hepatomegaly

# Ventricular Septal Defect

## Examination

- ⊙ Inspection - Failure to thrive, dyspnoea
- ⊙ Palpation
  - pulses and blood pressure - normal
  - cardiomegaly +/- hepatomegaly +/-
  - Thrill (Palpable murmur)  
over left 3/4th intercostal space
  - Palpable 2nd heart sound in pulmonary area if  
pulmonary hypertension has developed

# Ventricular Septal Defect

## ⊙Auscultation

- **Heart sounds** - normal but if complicated by pulmonary hypertension loud 2<sup>nd</sup> heart sound in the pulmonary area
- **Murmur** = pansystolic murmur at left lower sternal edge
- Mid diastolic murmur at apex in large VSD.  
Pulmonary : systemic flow > 2:1. Due to a large volume of blood flowing through a normal mitral valve.



# Ventricular Septal Defect

## Investigations

### Chest X'ray

Small VSD – normal

Large VSD - cardiomegaly

- pulmonary plethora

- prominent pulmonary artery.

### ECG

Small VSD – normal

Large VSD- right + left ventricular hypertrophy

### Echocardiography

Demonstrates VSD size and position

# Ventricular Septal Defect

## ☉ Management

A significant number of VSDs close spontaneously large percentage require no treatment

Conservative management – medical management

### ★ Prevention of infective endocarditis

good dental hygiene

antibiotic prophylaxis prior to major/ minor surgery

# Ventricular Septal Defect

## Medical Management

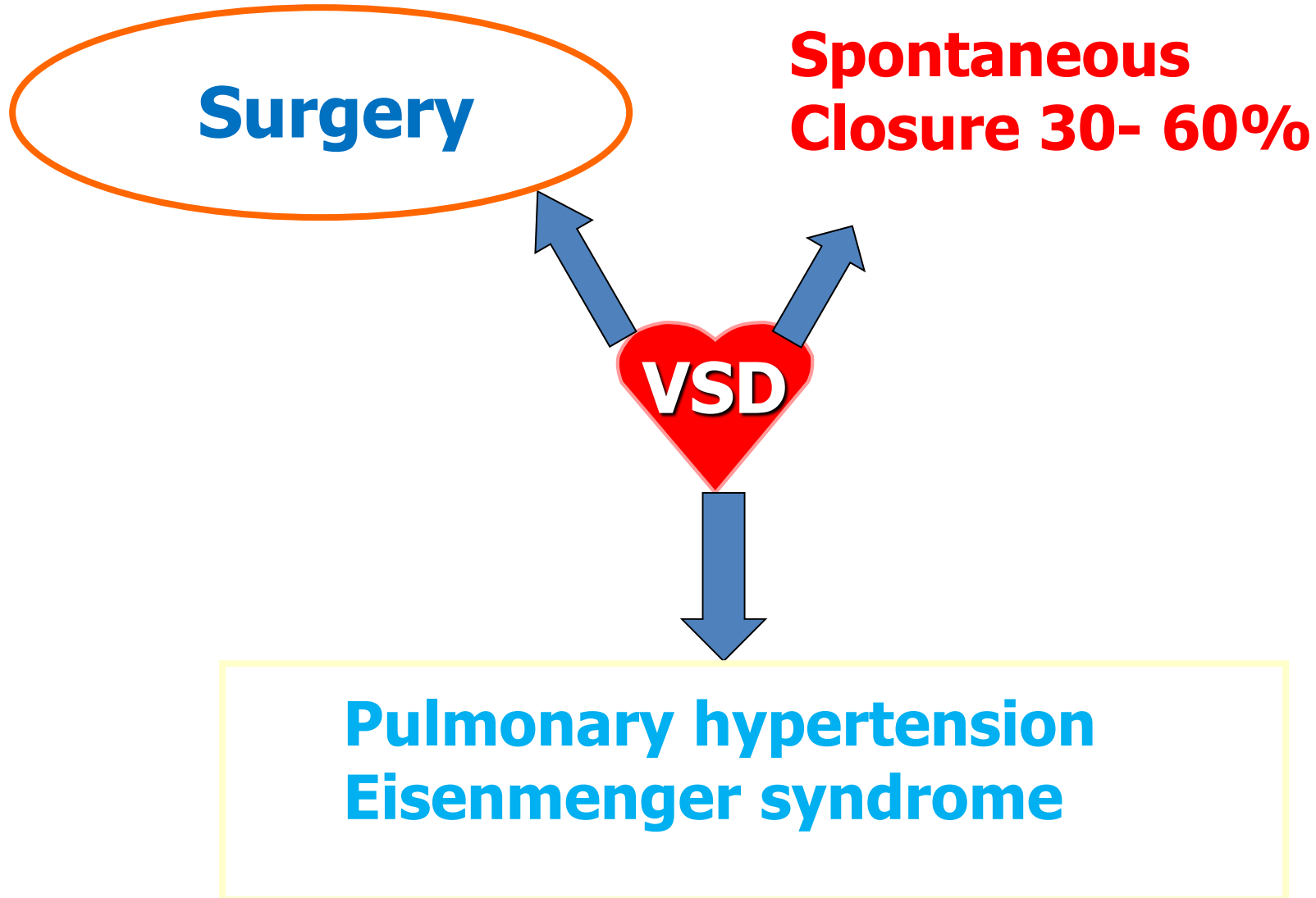
Follow up till VSD is closed

Look for complications

Heart failure, chest infections – treat appropriately

Pulmonary hypertension  Refer for early surgery

# Ventricular Septal Defect



# Ventricular Septal Defect

## Indications for Surgical Closure of VSD

Heart failure not responding to medical  
management

Early pulmonary hypertension

Pulmonary to systemic flow  $> 2:1$

# Atrial Septal Defect

## Simple ASD

### Ostium Secundum Defect

Failure of closure of ostium secundum in the upper part of the inter-atrial septum

# Atrial Septal Defect



Complex ASD

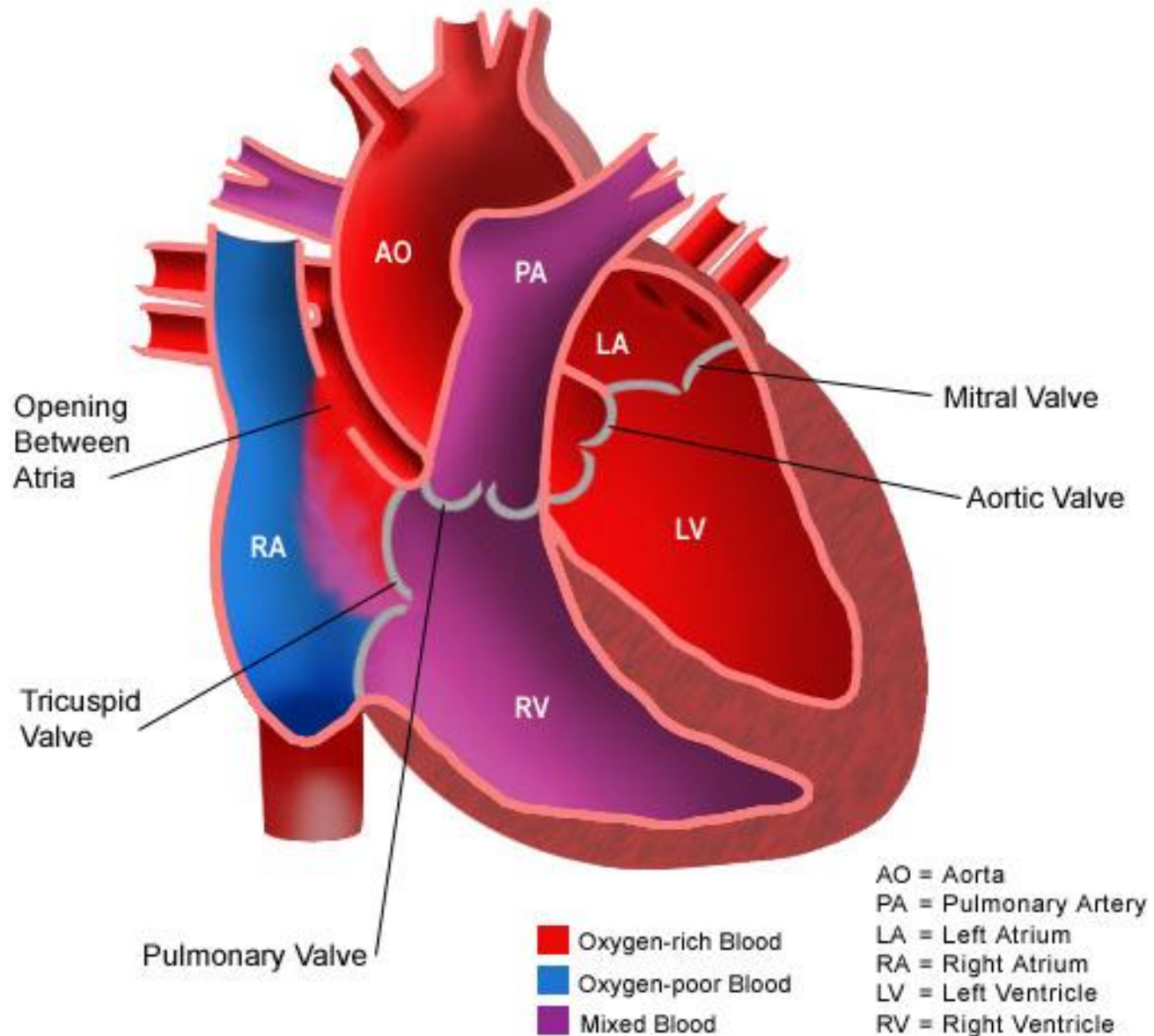
## Ostium Primum Defect

Failure of closure of the ostium primum in  
the lower part of the septum +  
Mitral Regurgitation

## Atrio Ventricular Septal Defect -

Ostium primum + mitral & tricuspid valve  
involvement + high VSD

# Atrial Septal Defect (ASD)





# Atrial Septal Defect

## Ostium Secundum Defect

- Majority - asymptomatic
- Diagnosis usually detected on routine examination
- Few with large defects – failure to thrive, dyspnoea, chest infections, head sweating
- Inspection - FTT+/- , shortness of breath+/-
- Palpation
  - Pulses/ BP – normal. Cardiomegaly +/-
  - RV impulse + left parasternal heave

# Atrial Septal Defect

## Auscultation

1st heart sound - normal

2nd heart sound - wide fixed split no variation with respiration

Ejection systolic murmur in the pulmonary area.

Due to excessive flow of blood through the pulmonary valve

The murmur is not due to flow through defect

Large defect, no major pressure gradient between left and right atria, no turbulence

# Atrial Septal Defect

## Auscultation

**Large Defect** Mid diastolic murmur in tricuspid area due to increased flow through valve

## Investigations

CXR

Cardiomegaly +/- and Pulmonary plethora

ECG

Right axis deviation + right bundle  
branch block

ECHO

Confirms Diagnosis

## Management

Catheter closure with device, or surgical repair 4-5 years

# Atrial Septal Defect

- ⦿ Ostium primum defect  
Management - surgical repair

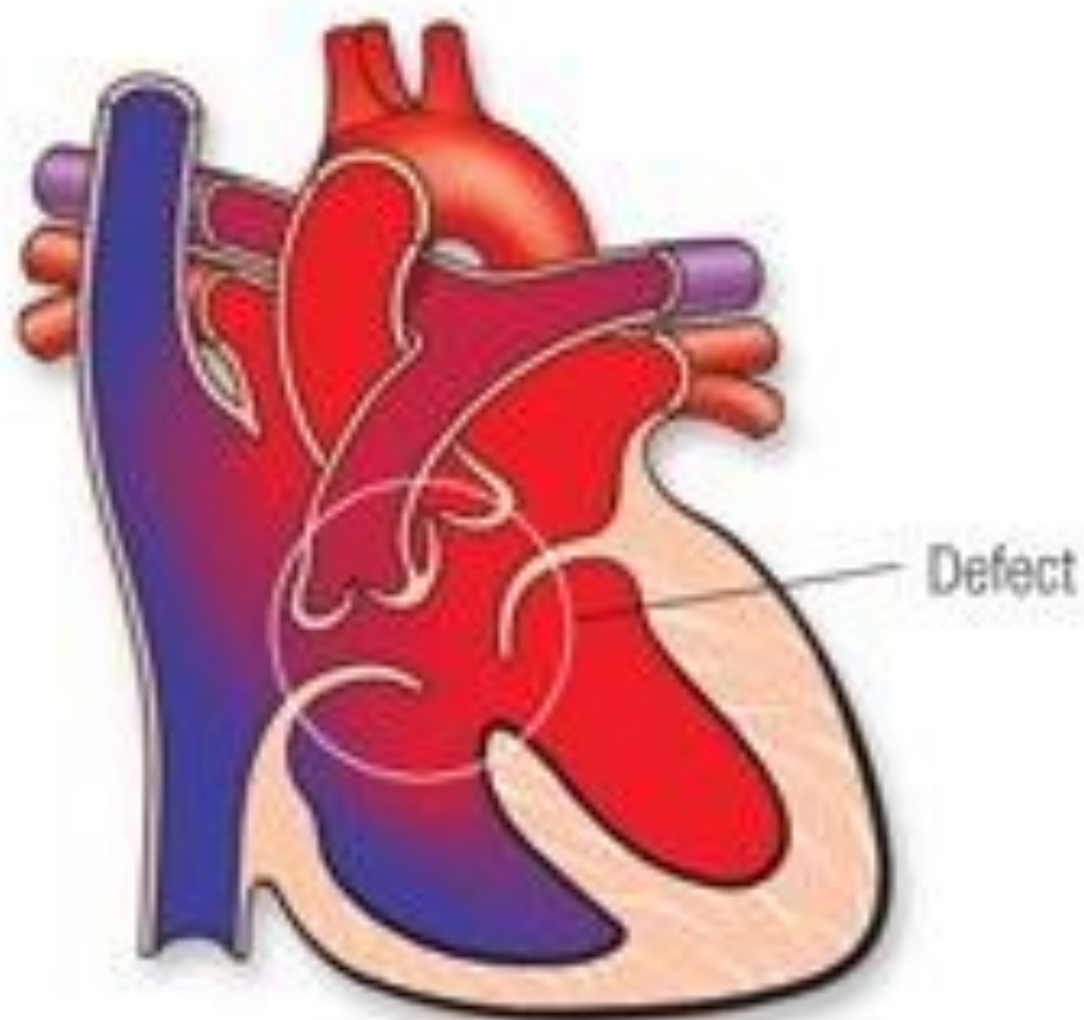
- ⦿ Atrio Ventricular Canal Defect

Poor prognosis

Early surgical repair important  
pulmonary hypertension develops soon

Commonest defect in DOWN SYNDROME

## Atrioventricular Canal Defect



# Patent Ductus Arteriosus

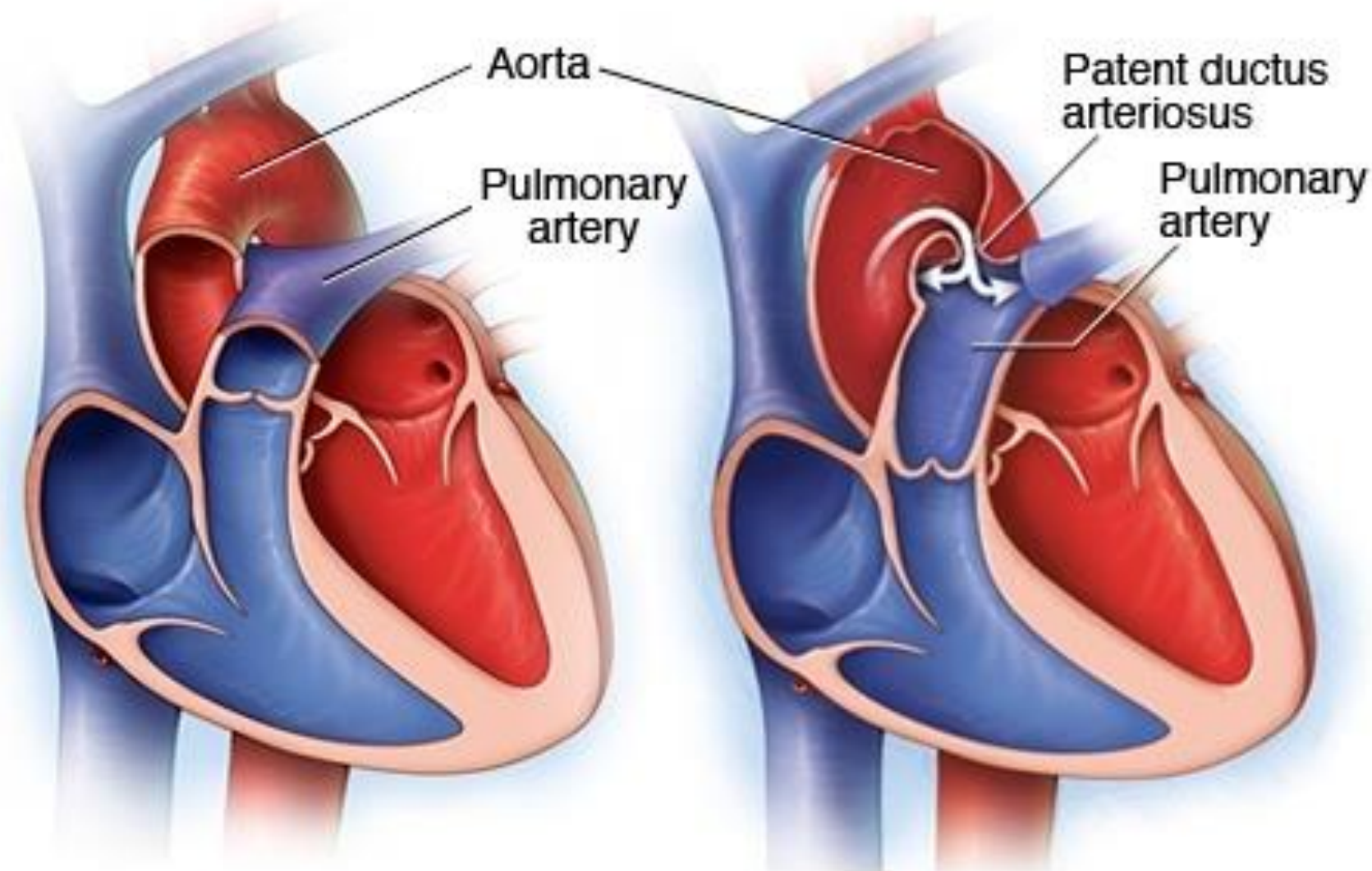
- Failure of the ductus to close after birth

Normally closes 10-15 hrs after birth, due to contraction of specialized tissue present from 25 weeks of IU life.  
Gradually matures over next 10-12 weeks.

- ◎ Premature infant – Maturation defect
- ◎ Mature infant - primary anatomical defect

Normal heart

Patent ductus arteriosus



# Patent Ductus Arteriosus

Clinical features - depends on size

## Inspection

Small - asymptomatic

Large - FTT, heart failure, chest infections

## Palpation

Pulse - large volume, collapsing

BP - increased pulse pressure

Thrill – pulmonary area

Auscultation - Heart sounds normal  
(Pul. Hypertension- loud P2)



# Patent Ductus Arteriosus

## Auscultation

Continuous murmur – systolic + diastolic murmur (machinery) in pulmonary area

Radiation - under clavicle and inter-scapular region posteriorly

## Investigations

Telechest - Cardiomegaly RV+LV,  
Pulmonary plethora

ECG - RV+LV hypertrophy

ECHO - Confirms diagnosis

# Patent Ductus Arteriosus

Treatment - surgical Closure

Pre Term Infant

Medical - Diuretics, fluid restriction and Indomethacin  
Prostaglandin inhibitor

Complications

Heart failure, Failure to thrive, infective Endocarditis,  
Pulmonary hypertension → shunt reversal

# Cyanotic Congenital Heart Disease

## Cyanosis

Bluish discolouration of skin & mucous membranes due to an increase in the amount of reduced haemoglobin in the blood

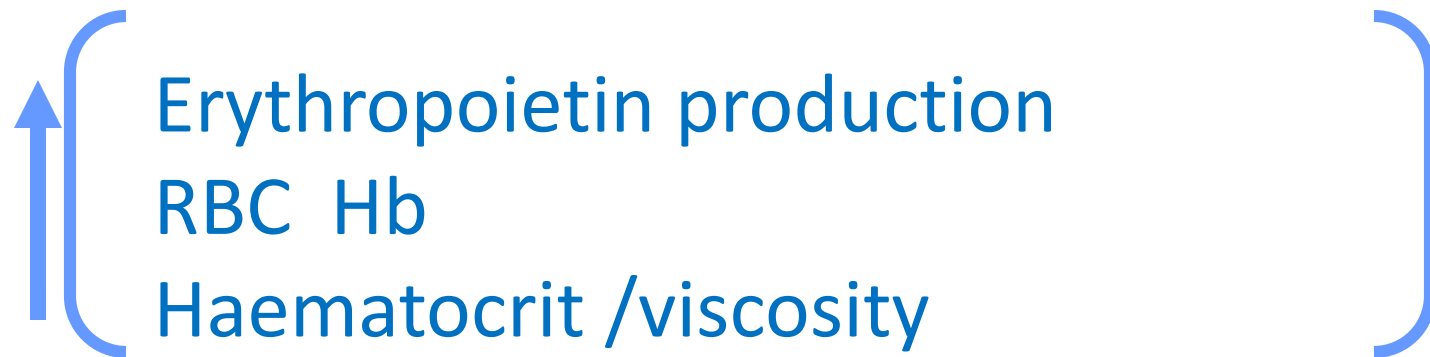
### Central Cyanosis

1. Imperfect oxygenation of blood heart failure, lung disease

# Cyanotic Congenital Heart Disease

## Central Cyanosis

2. Mixing of venous and arterial blood  
Right → left shunt  
generalized cyanosis



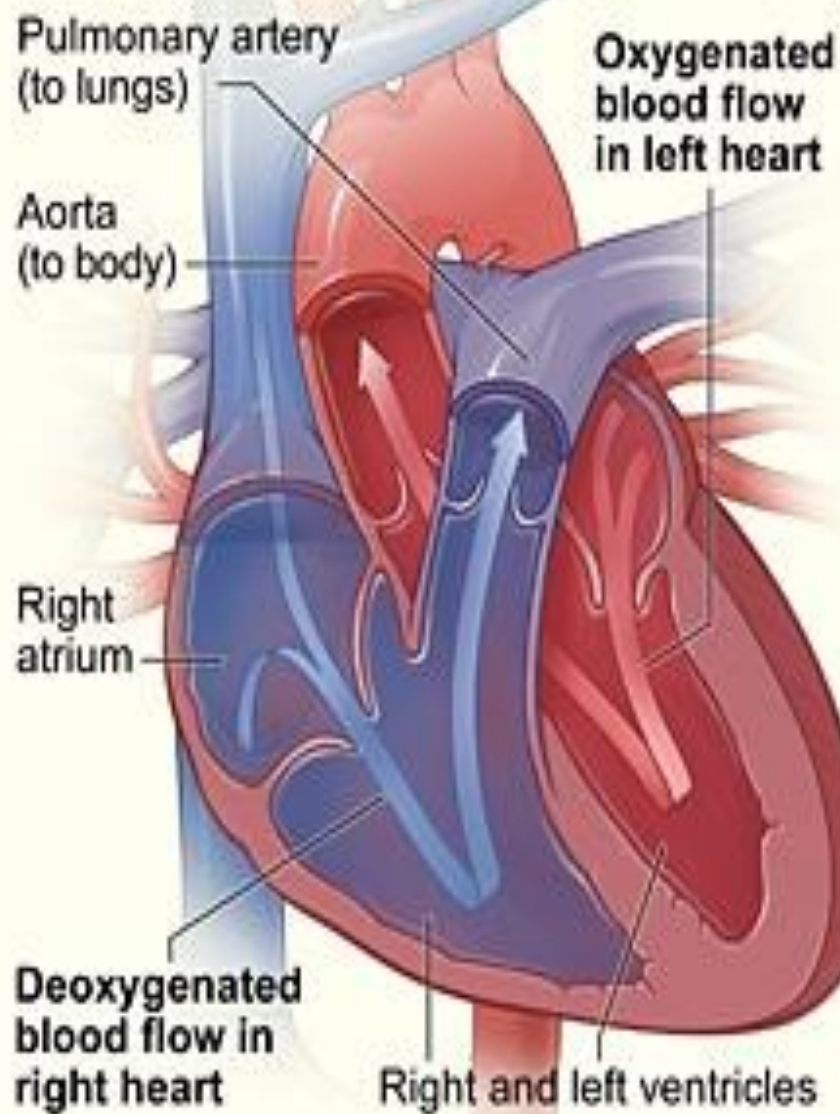
clubbing of fingers and toes

# Cyanotic Congenital Heart Disease

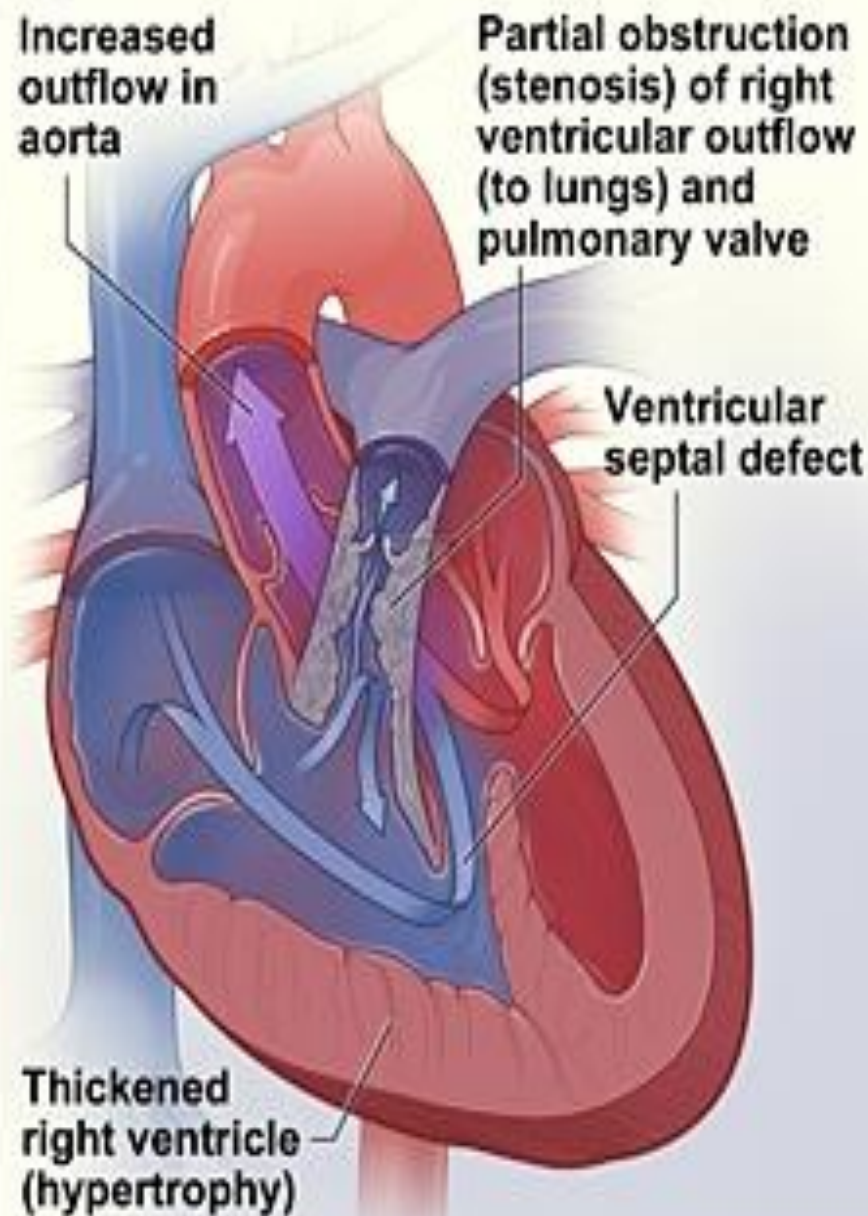
## TETRALOGY OF FALLOT 4 defects

- ⊙ VSD
- ⊙ Pulmonary stenosis
- ⊙ Over riding of Aorta
- ⊙ Right ventricular hypertrophy

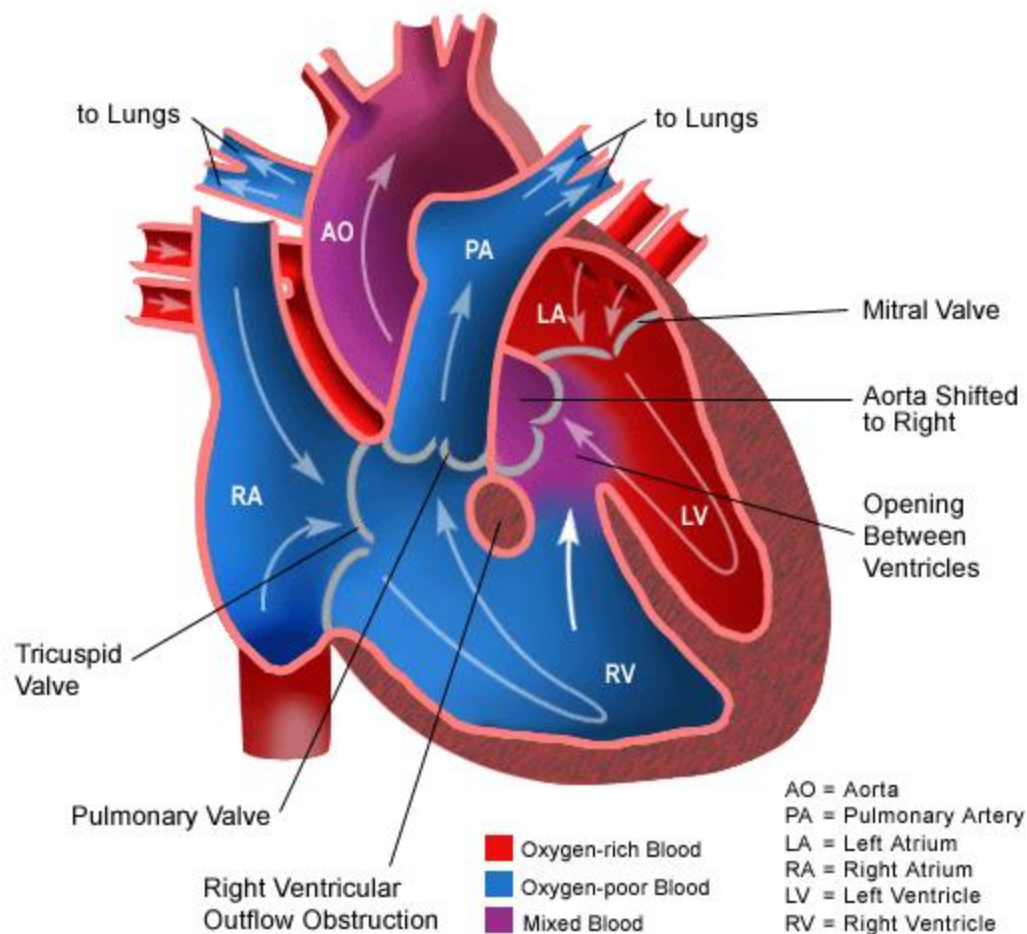
**A** Normal heart



**B** Heart with tetralogy of Fallot



## Tetralogy of Fallot (TOF or "Tet")



# TETRALOGY OF FALLOT

- ♥ Severity depends on the degree of pulmonary stenosis  
If pulmonary stenosis is mild most of the blood enters the pulmonary artery
- ♥ Immediately after birth PS is very mild  
Left to Right shunt occurs through VSD. No cyanosis
- ♥ As the child grows PS worsens  
Right to Left shunt → cyanosis



# TETRALOGY OF FALLOT

## Clinical Presentation

- ⊙ Pink at birth → Cyanosis
- ⊙ Hypercyanotic attacks 3- 6 months of age due to spasm of the infundibulum
- ⊙ Squatting

# Tetralogy of Fallot

## Clinical Presentation

### Examination

- Cyanosis
- Clubbing
- Polycythaemia
- Thrill & ejection systolic murmur in pulmonary area
- 2<sup>nd</sup> heart sound is single or the pulmonary component is soft

# Tetralogy of Fallot

CXR - No cardiomegaly. “Boot shaped heart”

RV enlarged. Pulmonary bay

Oligaemic lung fields

ECG - RV hypertrophy

ECHO

Blood count - increased Haemoglobin level and Haematocrit

Complications

Cerebral Thrombosis / abscess

Infective Endocarditis



# Tetralogy of Fallot

## Management of hypercyanotic attacks

- Knee chest position
- 100% O<sub>2</sub>
- Subcutaneous morphine/ Intravenous propranolol
- Till shunt/definitive surgery is done- oral propranolol

## Surgery

Palliative - Blalock/ Taussig shunt  
subclavian artery + R/L pul. artery

- Definitive surgery

# Transposition of Great Arteries

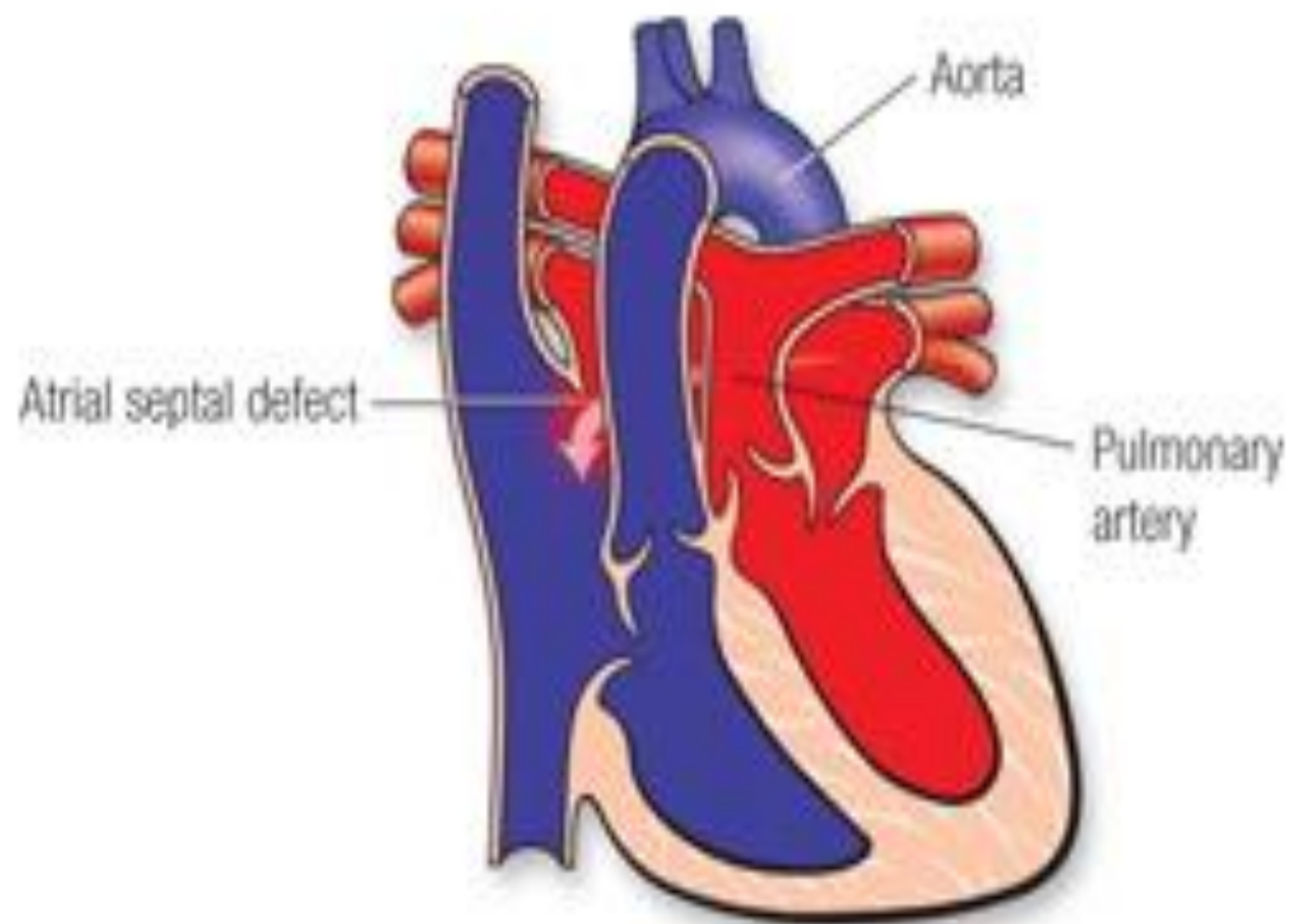
- Aorta arises from the right ventricle
- Pulmonary artery from left ventricle
- Two separate circulations  
needs a PDA/ASD/VSD for survival when ductus arteriosus closes after birth. Cyanosis

## Management

↖ Palliative - Atrial septostomy

↖ Definitive - Arterial switch

## Transposition of the Great Arteries



# Inoperable Heart defects

Management of heart failure – frusemide, captopril and spironolactone

Cyanotic defects

Prevention of complication due to polycythemia

If symptomatic and PCV over 65% venesection

Prevention of dehydration especially with intercurrent infections - gastroenteritis to prevent cerebral thrombosis



# Further reading

- ✦ Pulmonary stenosis
- ✦ Coarctation of aorta
- ✦ Pulmonary atresia
  - Tricuspid atresia
  - Ebstein Anomaly of tricuspid valve
  - Total anomalous pulmonary venous drainage
  - Hypoplastic left heart syndrome
- ✦ Basic knowledge needed