

# Congenital heart diseases

Dr . Owis Khater



# Principle differences in fetal circulation compared to post-natal circulation:

Combined ventricular output

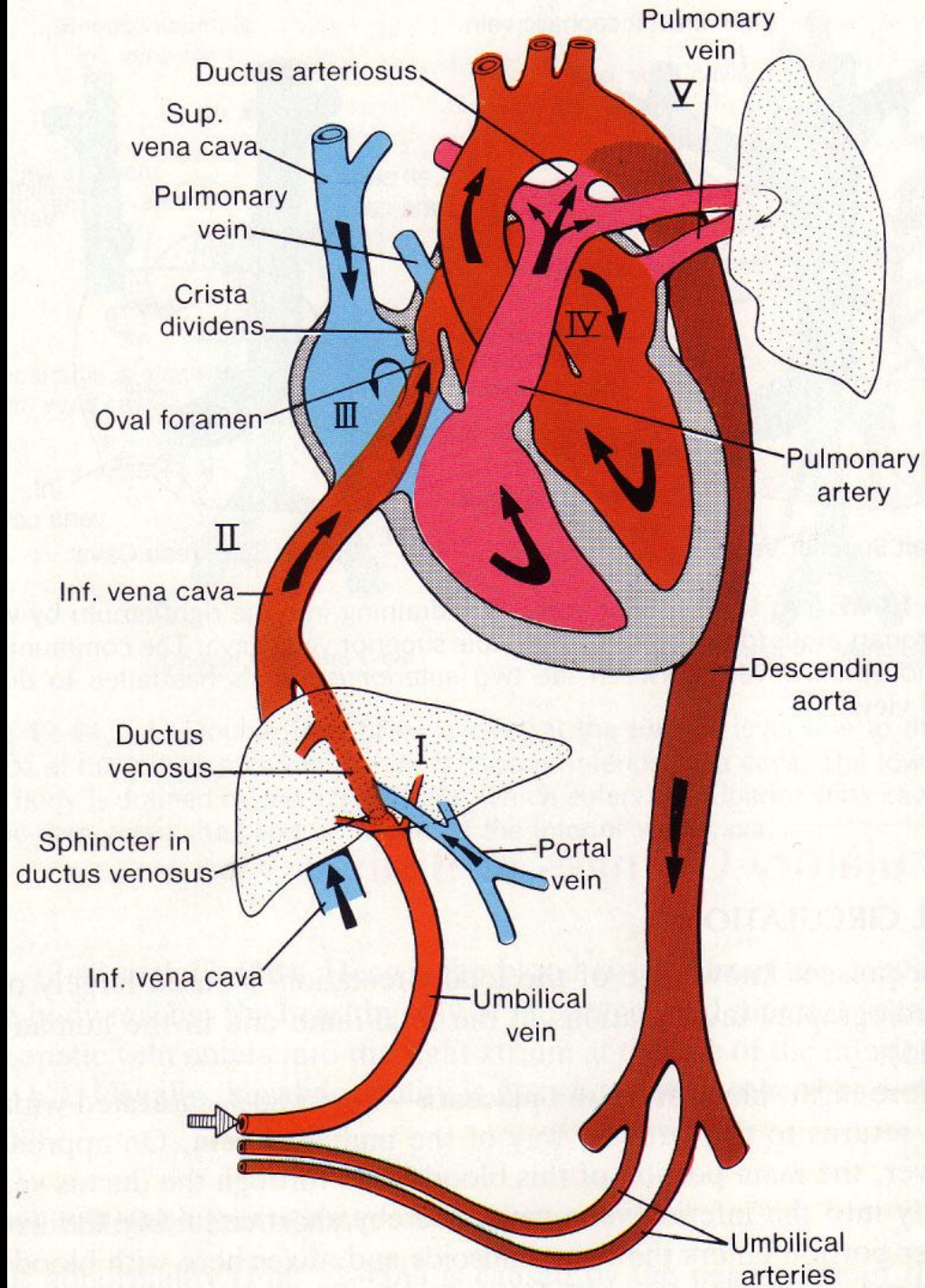
Three critical anatomic communications

PFO (from RA to LA)

PDA (from PA to AO)

Ductus venosus (from UV to IVC)

Organ responsible for oxygenation is Placenta



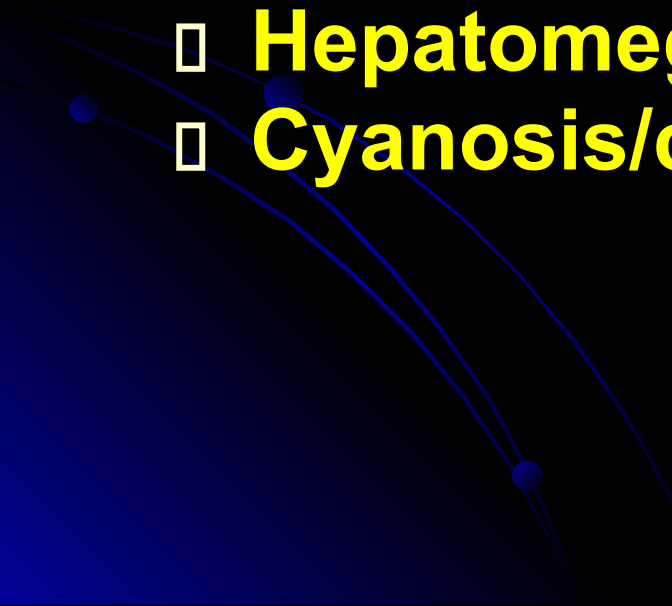
# CARDIAC EVALUATION

## *History*

- **Infants**
  - **feeding difficulties**
  - **Easily fatigued**
  - **Sweating while feeding**
  - **Tachypnea**
  - **Poor weight gain**
- **Older children:**
  - **shortness of breath**
  - **dyspnea**

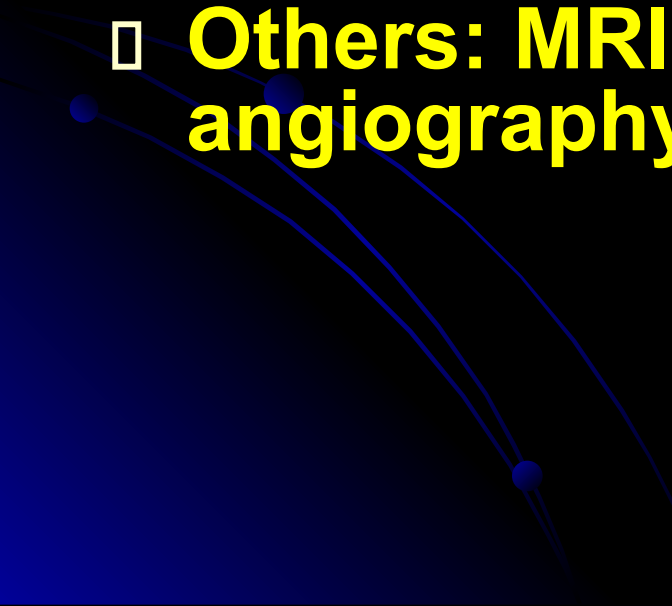
# CARDIAC EVALUATION

## Physical examination

- HR , RR
  - Assess adequate growth
  - Upper/lower BP
  - Rales
  - Hepatomegaly
  - Cyanosis/clubbing
- 

# CARDIAC EVALUATION

## Diagnostic tests

- Chest X-ray
  - ECG
  - Echocardiography
  - Others: MRI ,cardiac catheterization , angiography, exercise testing.
- 

# Cardiac evaluation

## Murmurs

Grade	Quality
1	Soft, difficult to hear
2	Easily heard
3	Louder but no thrill
4	Thrill
5	Thrill + audible with edge of stethoscope
6	Thrill + audible with stethoscope just off chest

# CARDIAC EVALUATION

## Murmur

### Innocent versus pathologic

Murmur is pathologic if one or more :

- **Symptoms**
- **Cyanosis**
- **Grade 3/6 or higher**
- **Thrill**
- **Diastolic**
- **Abnormal heart sounds**
- **Abnormally strong or weak pulse**

# Structural heart disease

```
graph TD; A[Structural heart disease] --> B[Acyanotic with shunt]; A --> C[Cyanotic]; A --> D[Non Shunt lesions]; B --> B1[• ASD]; B --> B2[• VSD]; B --> B3[• PDA]; C --> C1[• TOF]; C --> C2[• TGA]; C --> C3[• Truncus]; C --> C4[• Tricuspid Atresia]; C --> C5[• TAPVR]; D --> E[Obstruction]; D --> F[Regurgitation]; E --> E1[• Aortic stenosis AS]; E --> E2[• Supravalvar AS]; E --> E3[• Subaortic S]; E --> E4[• Coarctation]; E --> E5[• Mitral Stenosis]; E --> E6[• Pulmonary Stenosis]; E --> E7[• HLHS]; F --> F1[• Aortic regurgitation]; F --> F2[• Mitral regurgitation]; F --> F3[• Pulmonary reg.];
```

## Acyanotic with shunt

- ASD
- VSD
- PDA

## Cyanotic

- TOF
- TGA
- Truncus
- Tricuspid Atresia
- TAPVR

## Non Shunt lesions

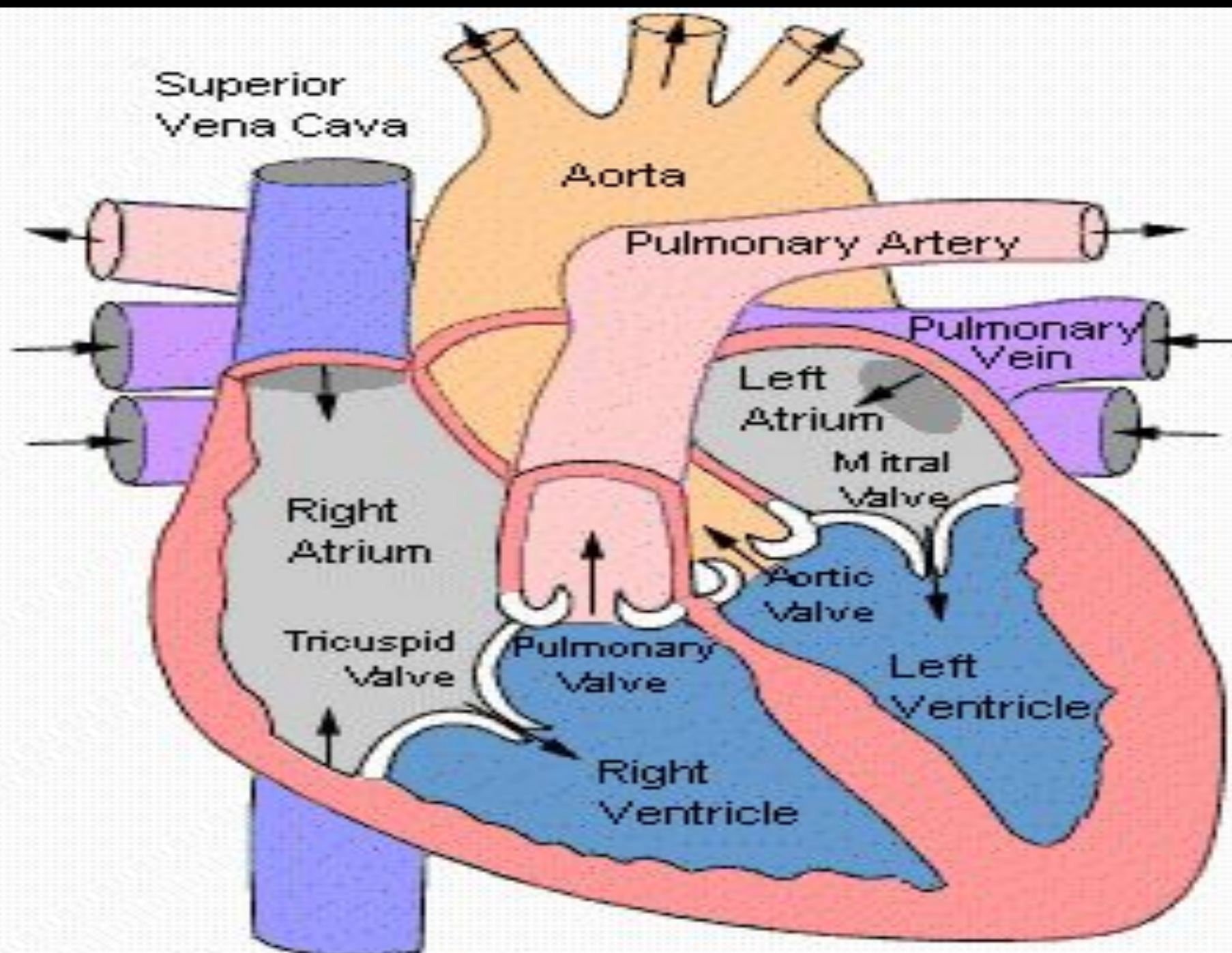
### Obstruction

- Aortic stenosis AS
- Supravalvar AS
- Subaortic S
- Coarctation
- Mitral Stenosis
- Pulmonary Stenosis
- HLHS

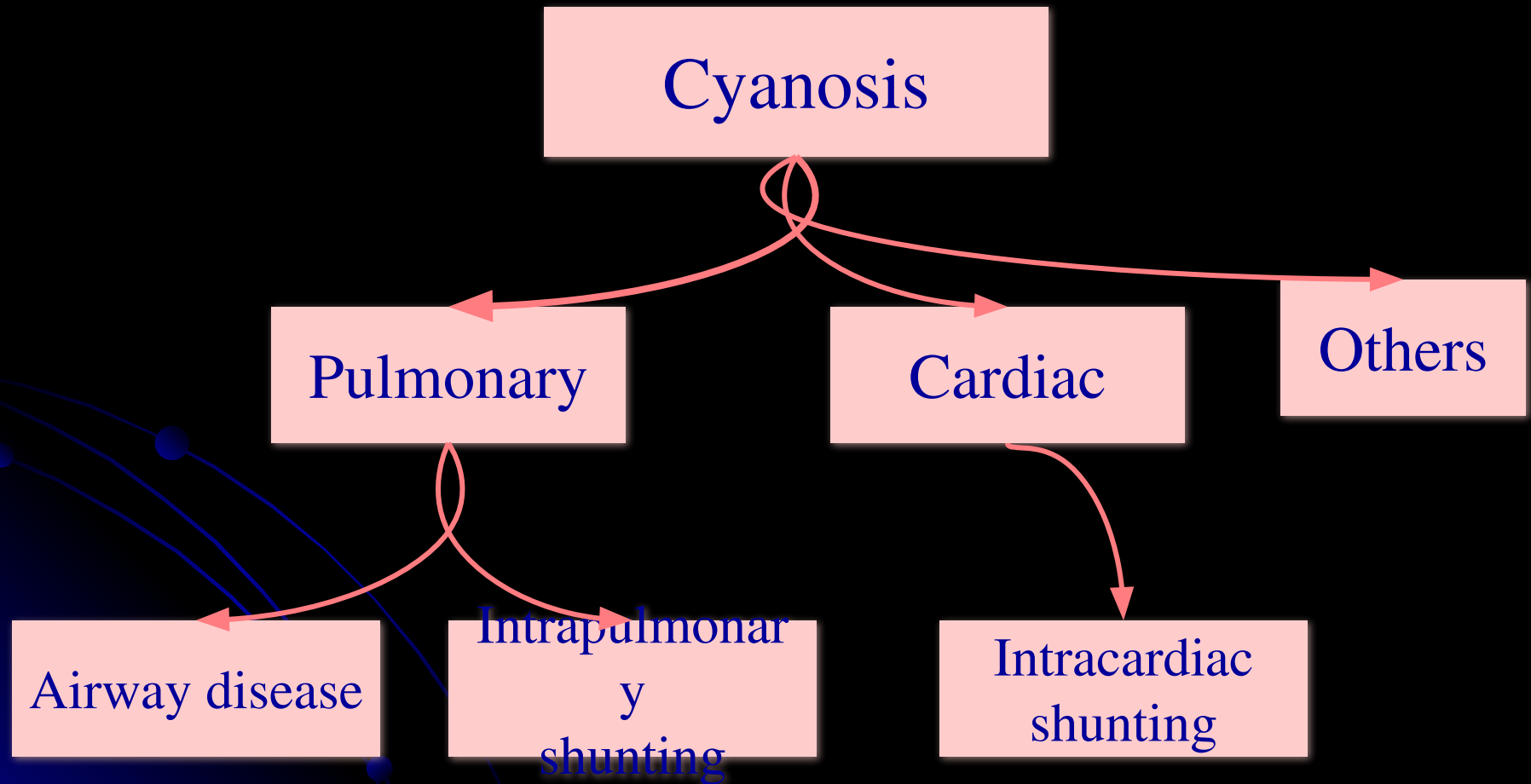
### Regurgitation

- Aortic regurgitation
- Mitral regurgitation
- Pulmonary reg.





# General causes of Cyanosis



Hyperoxia test

Administration of 100% O<sub>2</sub> for 15 minutes  
Measure arterial PO<sub>2</sub>

PO<sub>2</sub> <150

Cardiac

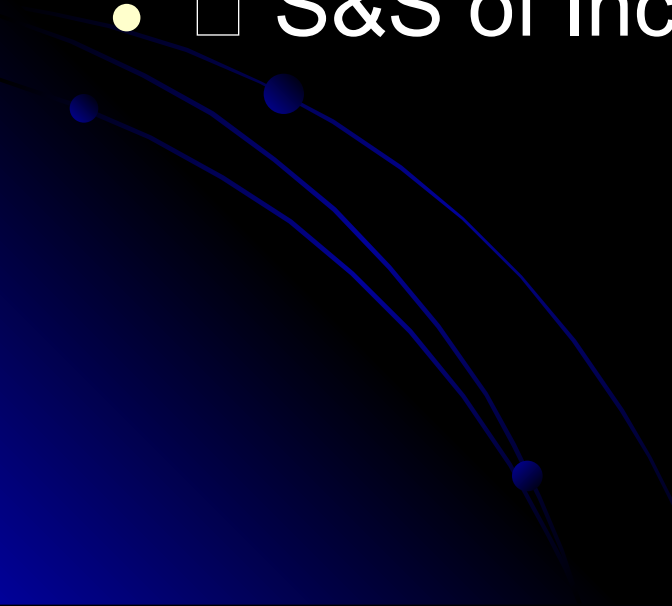
PO<sub>2</sub> 150-250

Gray zone

PO<sub>2</sub> >250

Pulmonary

# Left to Right s

- Portion of fully oxygenated pulmonary venous blood bypassing the systemic flow and going back to the lungs
  - □ In-effective pulmonary blood flow
  - □ S&S of Increased pulmonary blood flow
- 


# Left to right sh

Physiologic effect of the shunt is dependent  
:on three factors

Location of the shunt

Size of the defect

Relative pulmonary and systemic vascular  
resistance (or ventricular compliance in  
case of atrial level shunts)



# Congenital Heart Lesions that INCREASE Pulmonary Arterial Blood Flow

- Atrial Septal Defect
- Complete Atrioventricular Canal
- Ventricular Septal Defect
- Patent Ductus Arteriosus
- Total Anomalous Pulmonary Venous Connection
- Truncus Arteriosus

# Congenital Heart Disease

- **ACYANOTIC CONGENITAL HEART DISEASE**

- *Left to right shunts*

## **Ventricular septal defect (VSD)**

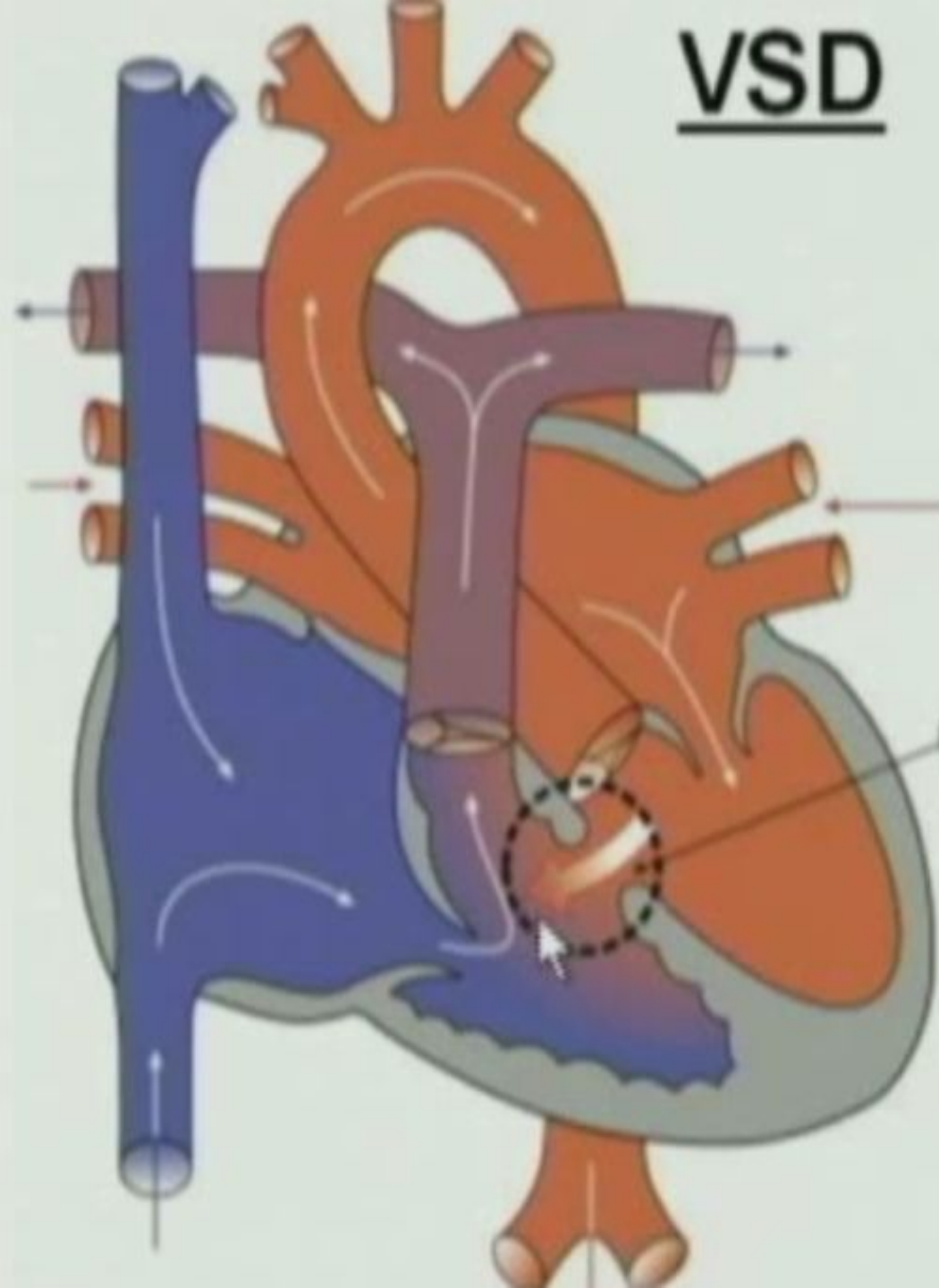
- **Most common congenital heart disease**

- **Shunt determined by the ratio of PVR to SVR**

- **As PVR falls in first few weeks of life , shunt increases**

- **When  $PVR > SVR$  , Eisenmenger syndrome.**

# VSD



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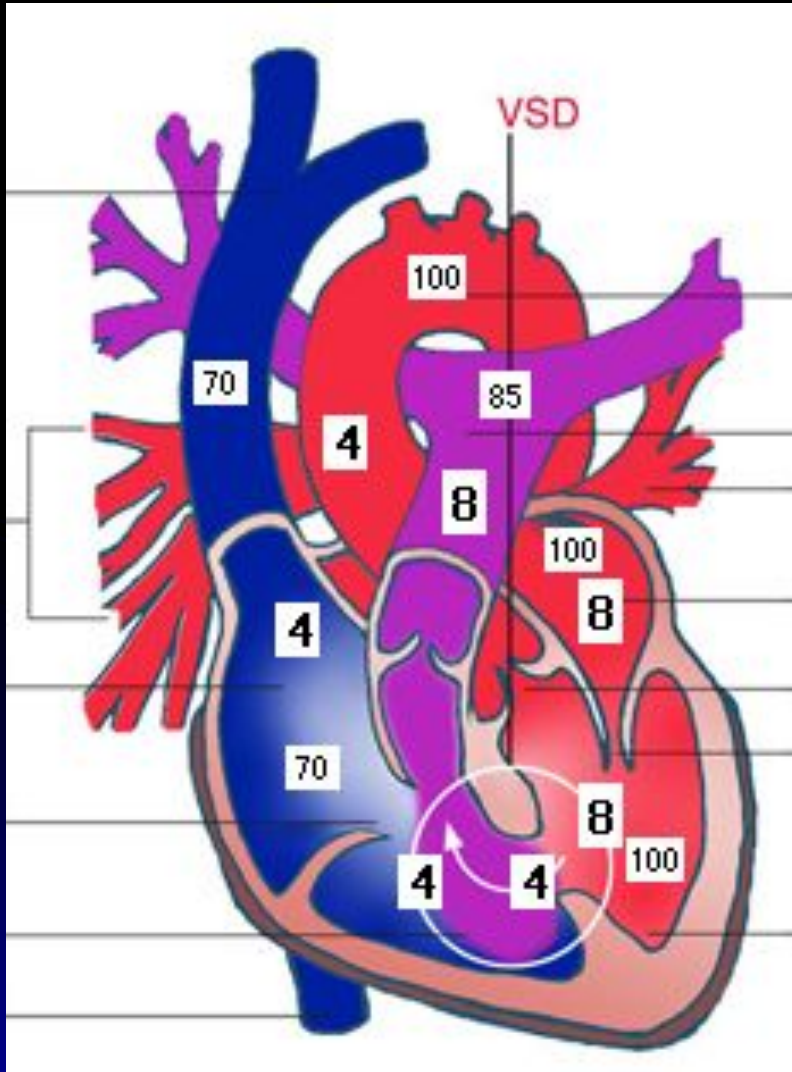
# Ventricular septal defect (VSD)

- **Clinical findings**

- Asymptomatic if small defect with normal pulmonary artery pressure (most)
- Large defect- dyspnea , feeding difficulties , poor growth , sweating , pulmonary infection , heart failure
- Harsh holosystolic murmur over lower left sternal border +/- thrill
- Male affected as female
- Small defect <2cm and large one >2cm

- Perimembranous (conovertricular defect)
  - Commonest type of VSD
  - Defect is under the aortic valve □ There is incidence of aortic valve prolapse and AI
- Muscular VSD
  - Can be single or multiple
- Inlet Type VSD (AV canal type)
  - Complete AV canal is common in Trisomy 21
- Sub-pulmonary (conal septal hypoplasia VSD)
  - Rare
  - Incidence of aortic valve prolapse

# Ventricular Level Shunt Physiology



- VSD causes Pressure load on the right ventricle causing RVH, and Volume load on the left atrium and ventricle leading to dilation
- Shunt occurs during Systole (ventricular emptying)
- Cardiac output is well maintained even in large VSD's

# Diagnostic st

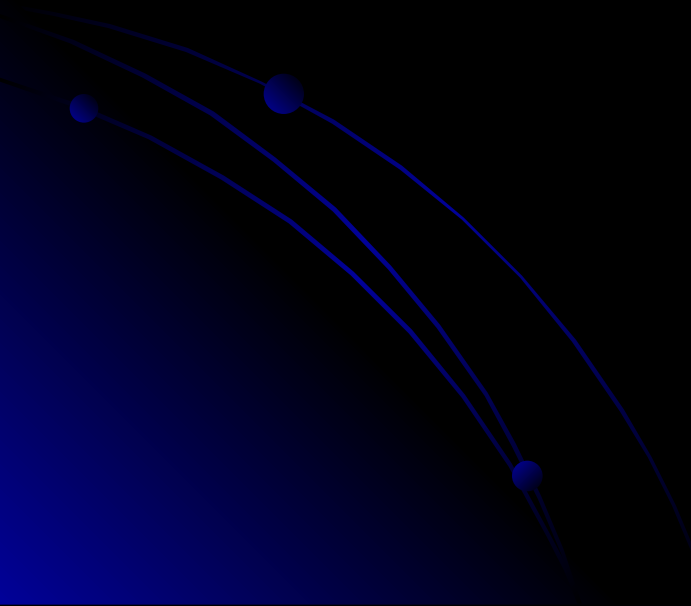
- ECG: (beyond infancy)
  - Left axis deviation
  - LVH
  - Left atrial dilation
  - Northwest (superior) axis in AV canal defects
- CXR shows cardiomegally, and increased pulmonary vascular markings in significant VSD's

- No restriction from activity
- No SBE prophylaxis (the newer guidelines)
- Spontaneous closure is common in small and moderate perimembranous and muscular defects
- AV canal type VSD's don't close spontaneously
- Surgical treatment is the standard treatment for symptomatic VSD's

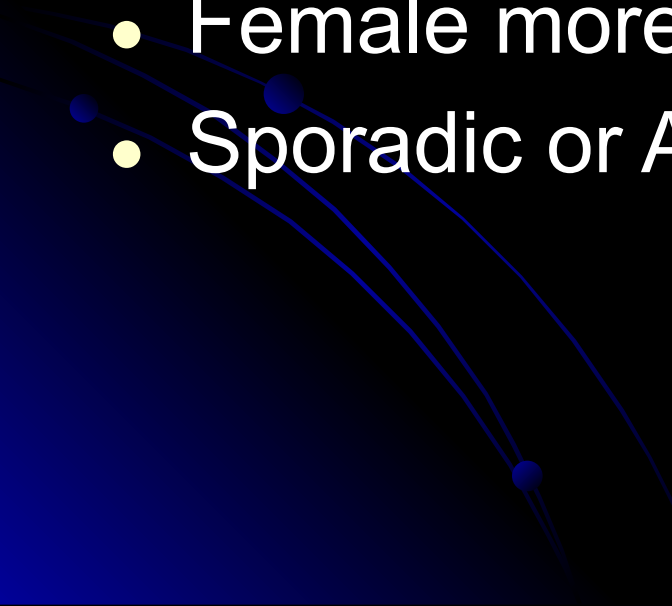
# Ventricular septal defect (VSD)

## Complications

- Large defects lead to HF, failure to thrive
- Endocarditis
- Pulmonary hypertension



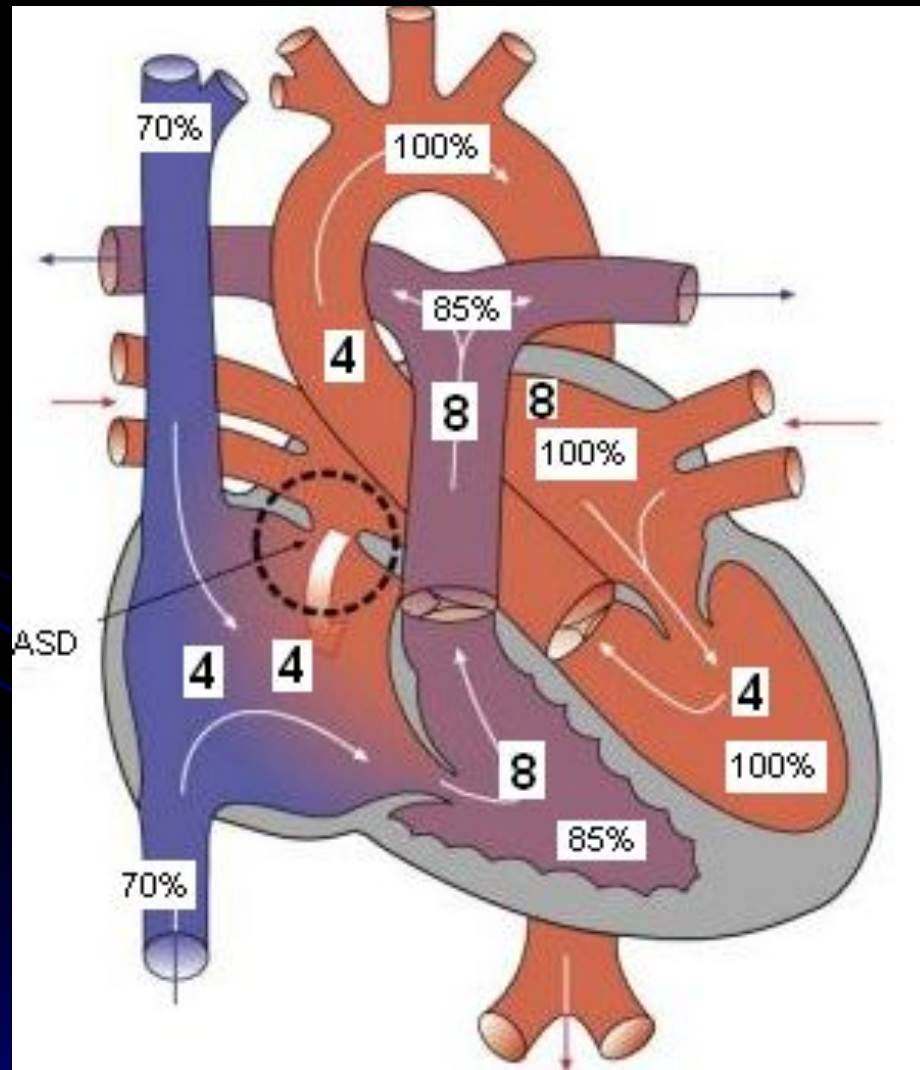
# Atrial Septal D

- Acyanotic; asymptomatic, or dyspnea on exertion.
  - Right ventricular lift.
  - Fixed, widely split second heart sound.
  - Female more affected (3 to 1 )
  - Sporadic or AD
- 

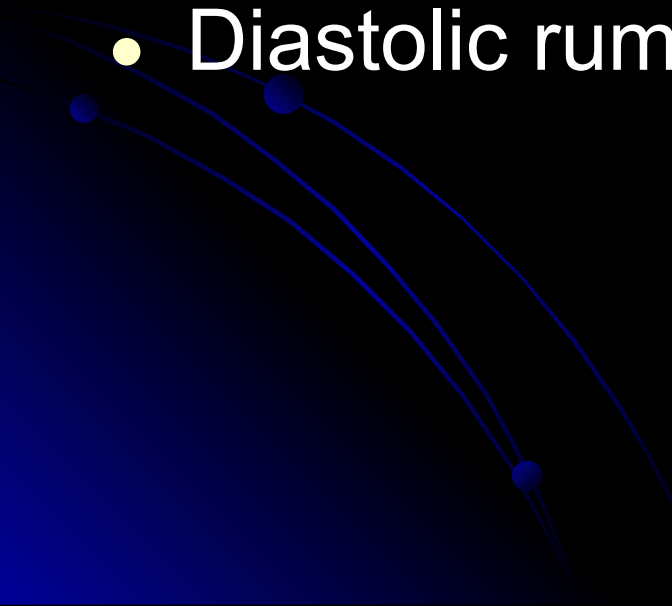
- Ostium secundum ASD:
  - Commonest type
  - Deficiency of septum primum
  - Can be one defect or multiple (fenestrated)
  - Mostly isolated
- Ostium Primum ASD
  - Also called partial AV canal defect (No VSD component)
  - Frequently associated with cleft mitral valve with MR
- Sinus Venosus ASD
  - SVC type much more common than IVC type
  - Majority associated with partial anomalous pulmonary venous return
- Coronary sinus ASD
  - Rare
  - Defect is the os of the coronary sinus with partial or complete un-roofing of the sinus



# Atrial Level Shunt: Physiology



- ASD causes **volume** load on the right atrium, and right ventricle leading to dilation of these structure
- Majority of the shunt occurs during diastole (ventricular filling)
- Cardiac output is well maintained even in large ASD's

- Normal in young infants
  - Prominent RV heave
  - Wide, fixed S2
  - Ejection systolic murmur
  - Diastolic rumble
- 

# Atrial septal defect

## □ **Treatment**

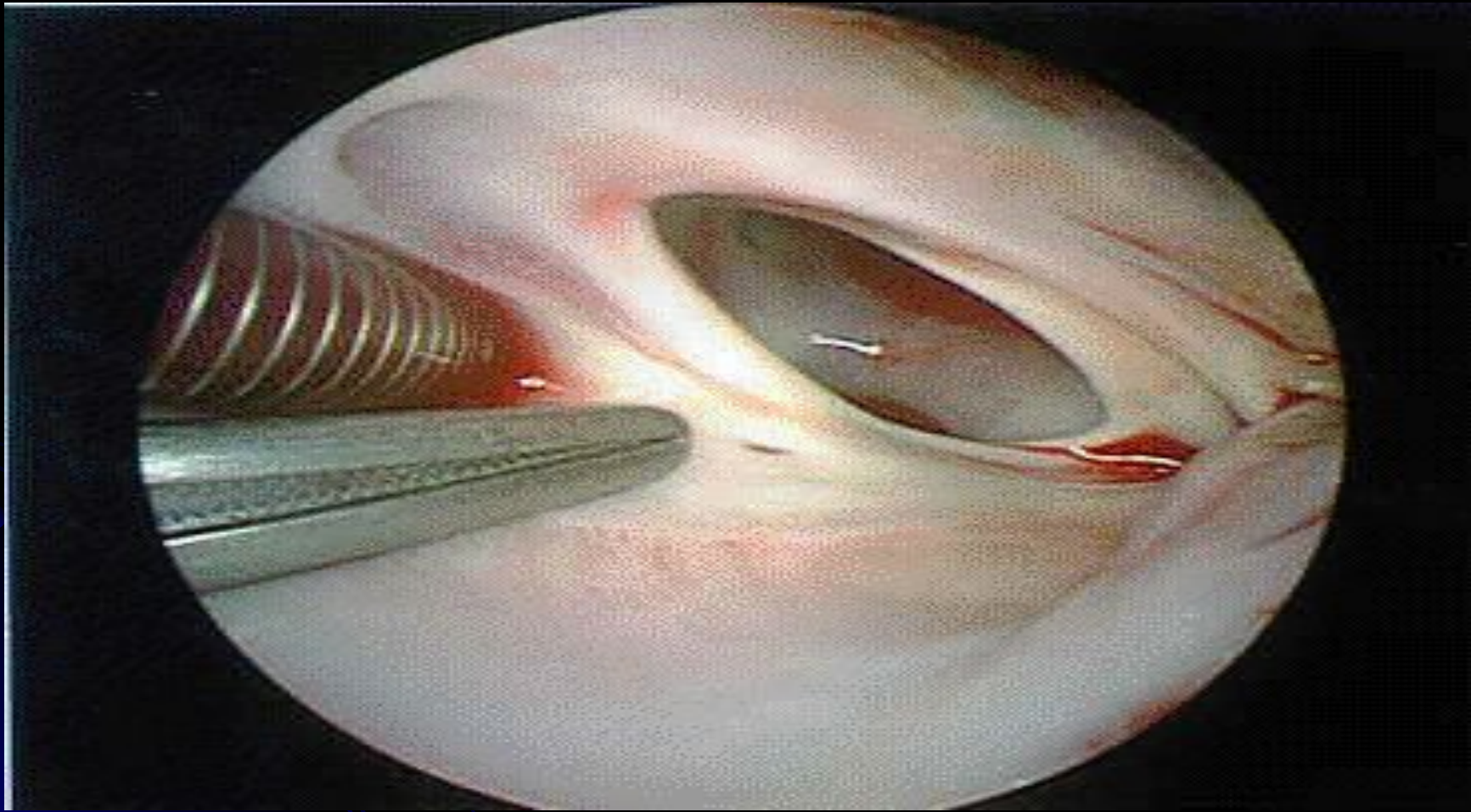
- Most in term infants close spontaneously; symptoms often do not appear until third decade
- Surgery or transcatheter device closure for all symptomatic patients.

## **Complications**

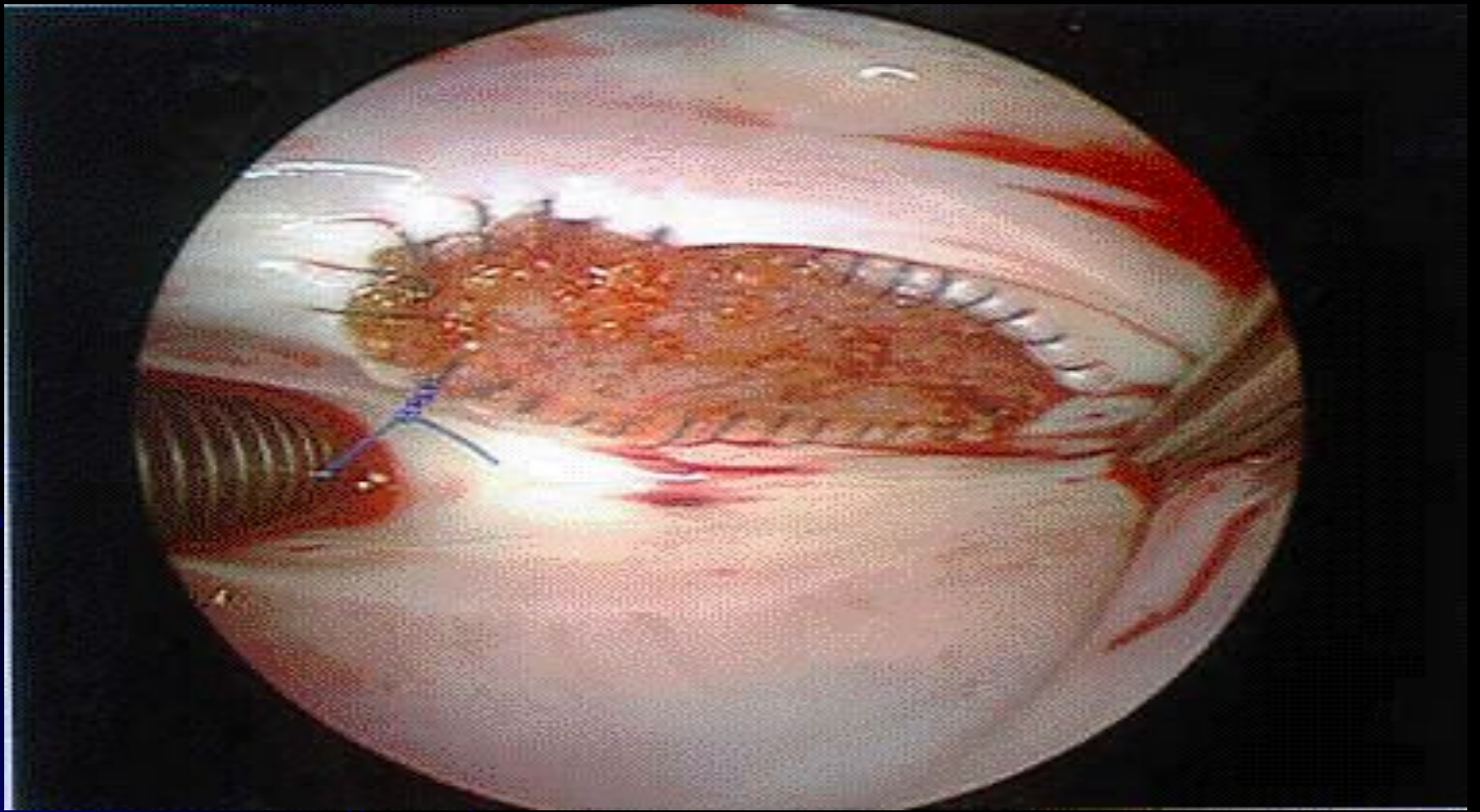
- Dysrhythmia
- Low-flow lesions; does not require endocarditic prophylaxis

- No restriction from activity
- No SBE prophylaxis
- No medications
- Observation for spontaneous closure if secundum type and no significant volume overload on the right ventricle
- Closure is indicated for significant secundum ASD's, and all primum and SV ASD's

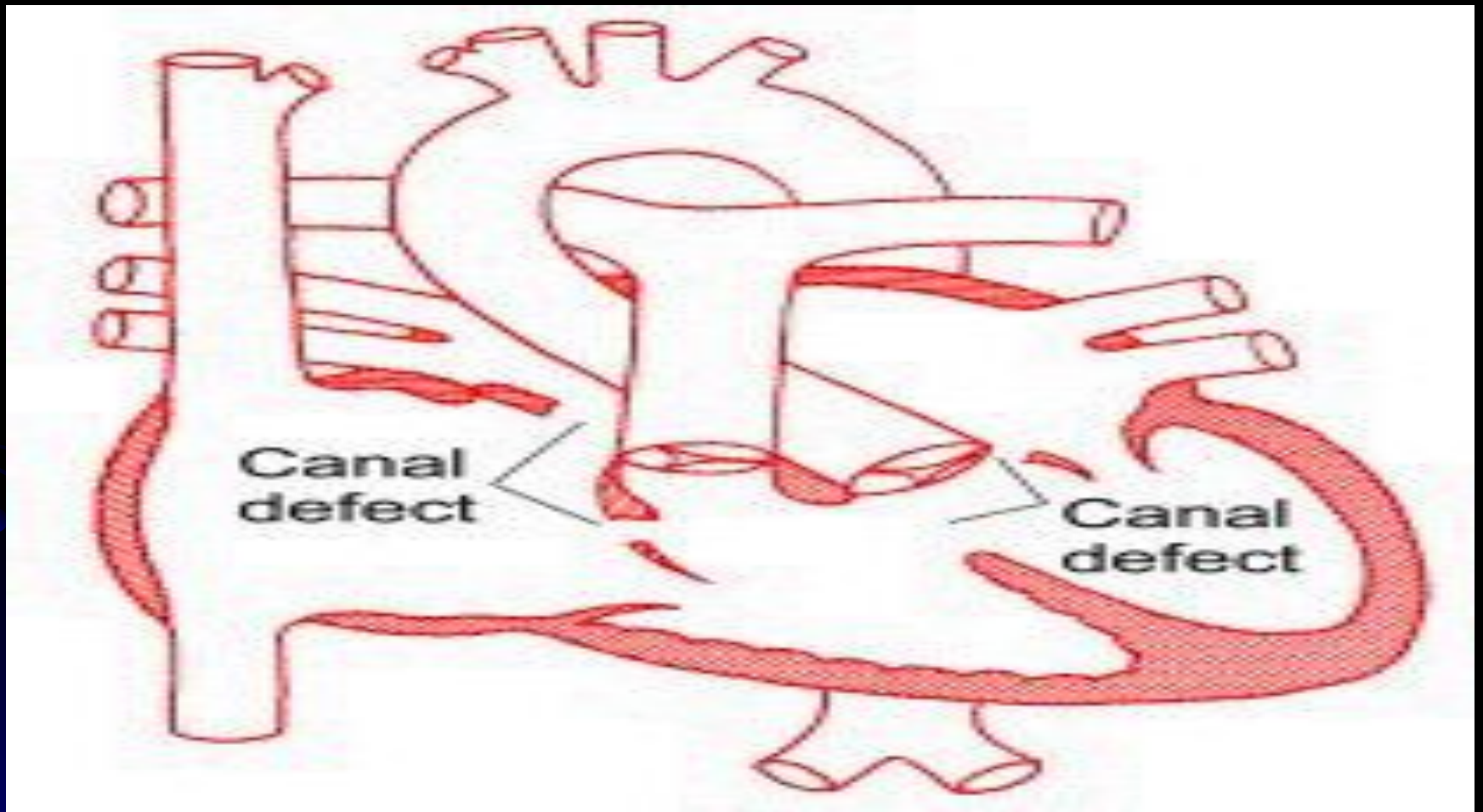
# Atrial Septal D



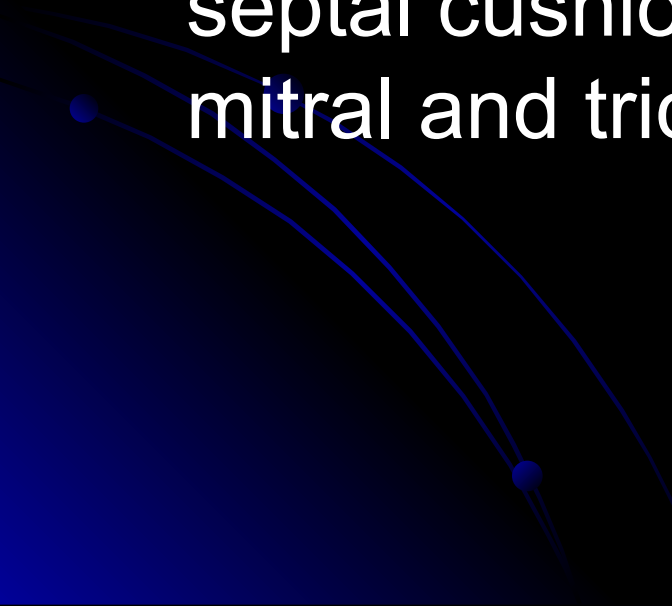
# Atrial Septal D



# Complete Atrioventricular Canal

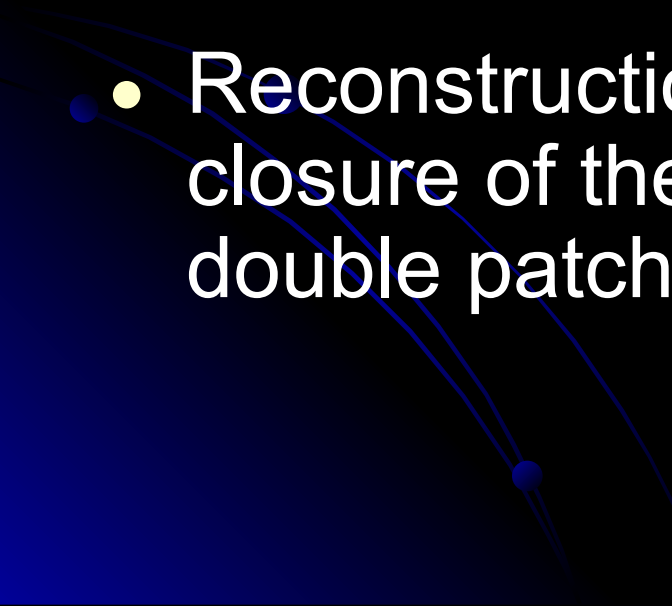


# Complete Atrioventricular Canal

- Heart failure common in infancy.
  - Cardiomegaly, blowing pansystolic murmur, other variable murmurs.
  - Deficiencies of both atrial and ventricular septal cushions and abnormalities of both mitral and tricuspid valves.
- 



# Complete Atrioventricular Canal

- Partial and complete AV canal defects frequently accompany Down's syndrome.
  - Early surgical correction.
  - Reconstruction of the AV valves and closure of the septal defects by a single or double patch technique.
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# Congenital Heart Disease

- **Patent Ductus Arteriosus (PDA)**

**results when the ductus arteriosus fails to close; this leads to blood flow from the aorta to the pulmonary artery**

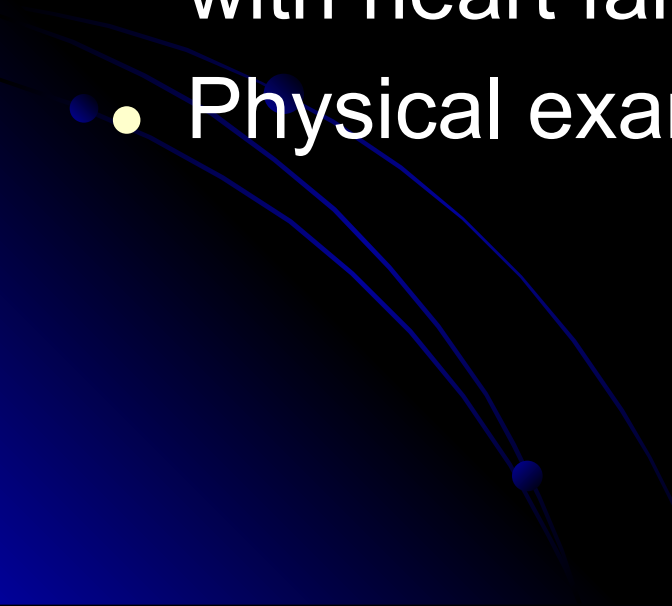
## **Risk factors**

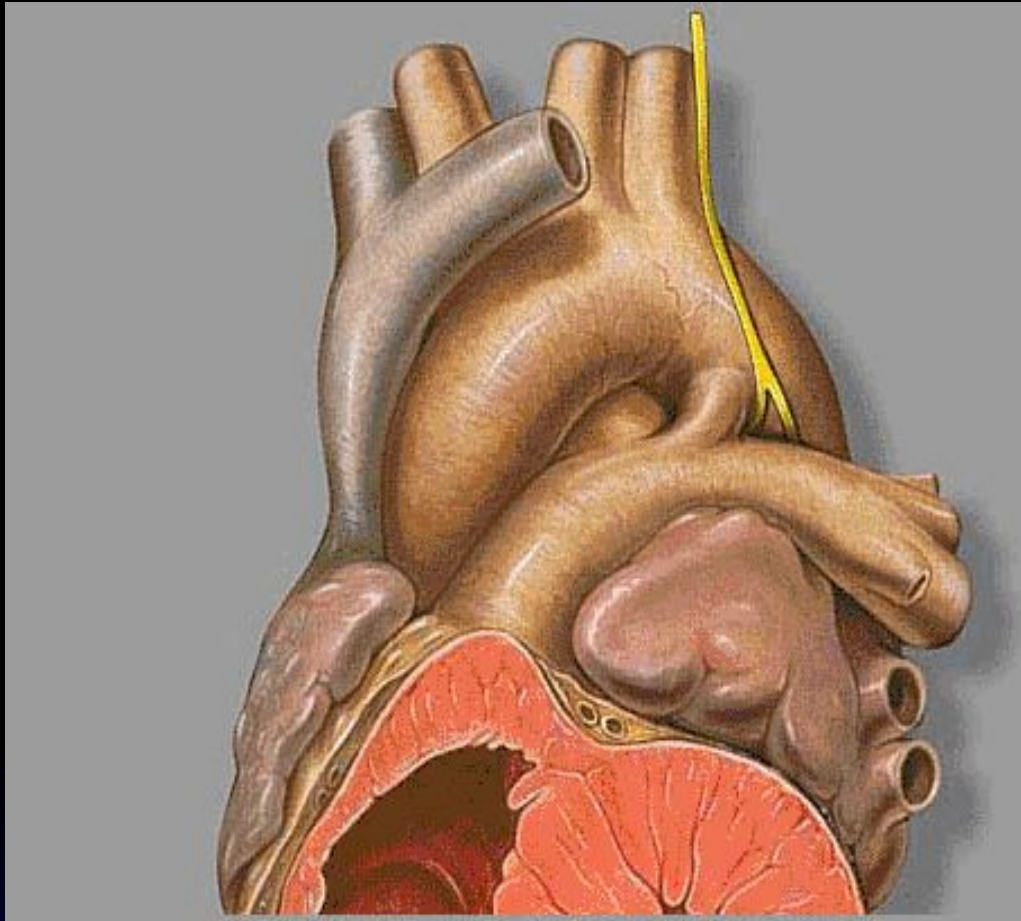
- **More common in girls by 2:1**

- **Association with maternal rubella infection**

- **Common in premature infants**

# Patent Ductus Arterios

- Murmur usually systolic, sometimes continuous, “machinery”
  - Poor feeding, respiratory distress, and frequent respiratory infections in infants with heart failure.
  - Physical exam and echocardiography.
- 



- Located just distal to the origin of the left subclavian artery
- 1 / 2500 to 1 / 5000 live births
  - Increased incidence with prematurity
- 12% of all CHD
- Female to male ratio = 2:1

- **Associations:** Asphyxia, Chromosomal anomalies, Birth at high altitudes, Congenital Rubella, Drugs, Genetic?

# Normal postnatal closure

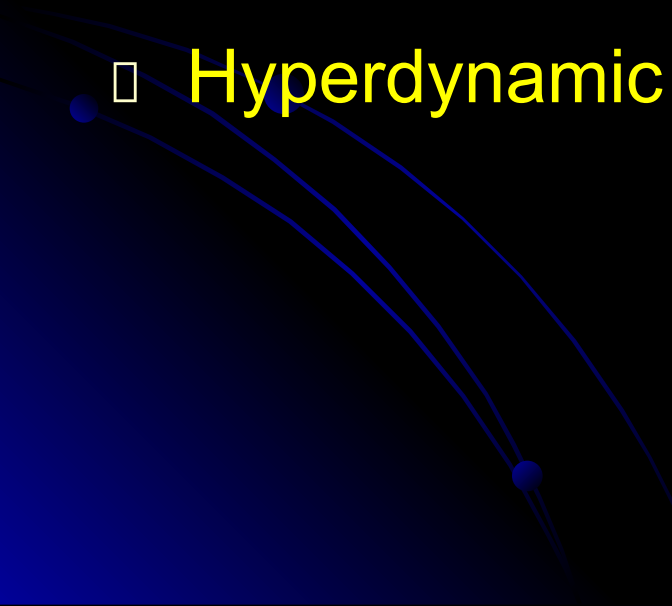
## Functional closure

- Usually occurs within the first 24 hours
- Stimulated by:
  - High pO<sub>2</sub> (more prominent effect with increased GA)
  - Interruption of Prostaglandins
    - Most important factor in ductal patency
    - Produced by the placenta and the ductal tissue
    - Metabolized by the lungs
    - Ductal tissue is much more sensitive to prostaglandins at earlier gestations

## Complete “anatomic” closure (fibrosis)

- Usually occurs in the first 2-3 weeks

# Patent Ductus Arteriosus (PDA)

- **Presentation**
    - If small – possibly no symptoms
    - If large –heart failure, a wide pulse pressure -bounding arterial pulses .
    - Continuous murmur
    - Hyperdynamic precordium
- 

- Asymptomatic PDA's require no treatment before age of 1 year, elective closure can usually be done by catheterization
- Symptomatic PDA's
  - Symptomatic treatment of CHF (diuresis, inotropic support, and vasodilators)
  - Avoid lowering PVR (avoid oxygen, alkalosis, NO)
  - Medical closure (Indomethacin IV, preferably before day 10 of life, Ibuprofen IV has similar effect)
  - Surgical closure in refractory cases

Cyanotic heart disease (right  
left shunt)



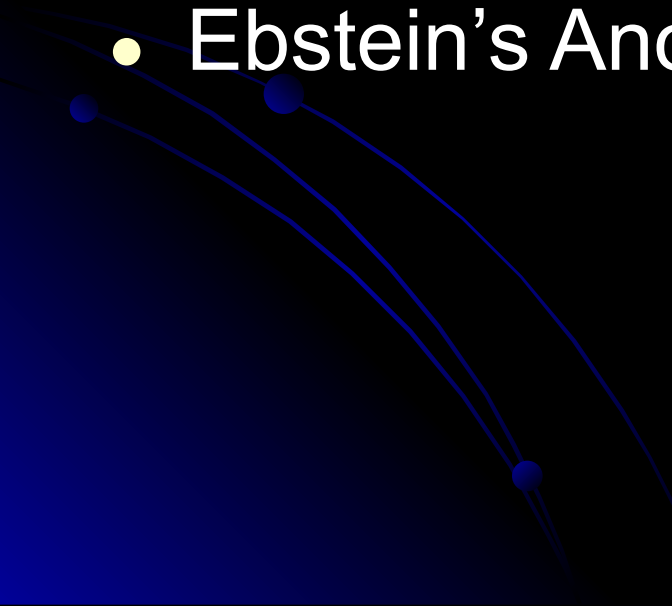


# CYANOTIC CONGENITAL HEART DISEASE

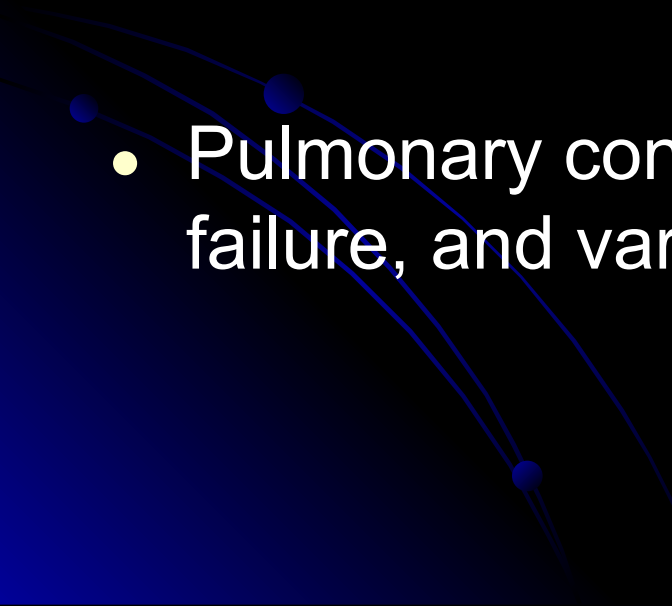
Common cyanotic heart disease ( 5 Ts & a P )

- Tetralogy of fallot
- Transposition of the great vesseles
- Trancus arteriosis
- Total anomalous pulmonary venous return
- Tricuspid atresia
- Pulmonic stenosis

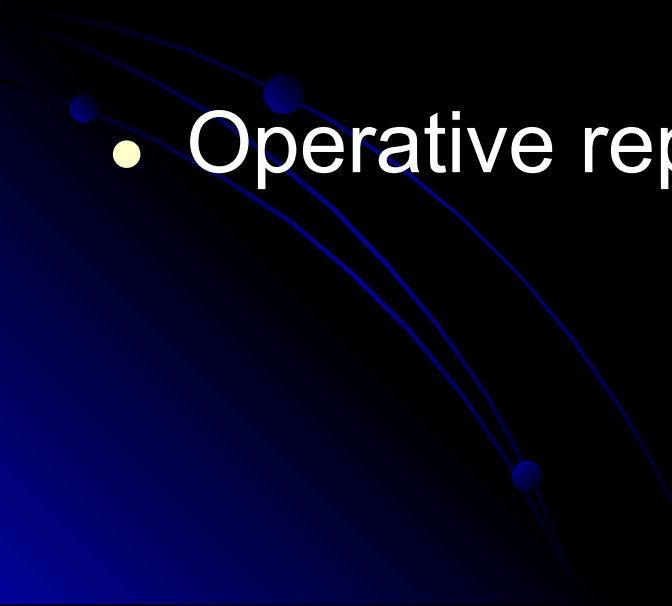
# Congenital Heart Lesions that DECREASE Pulmonary Arterial Blood Flow

- Tetralogy of Fallot
  - Transposition of the Great Arteries
  - Tricuspid Atresia
  - Ebstein's Anomaly
- 

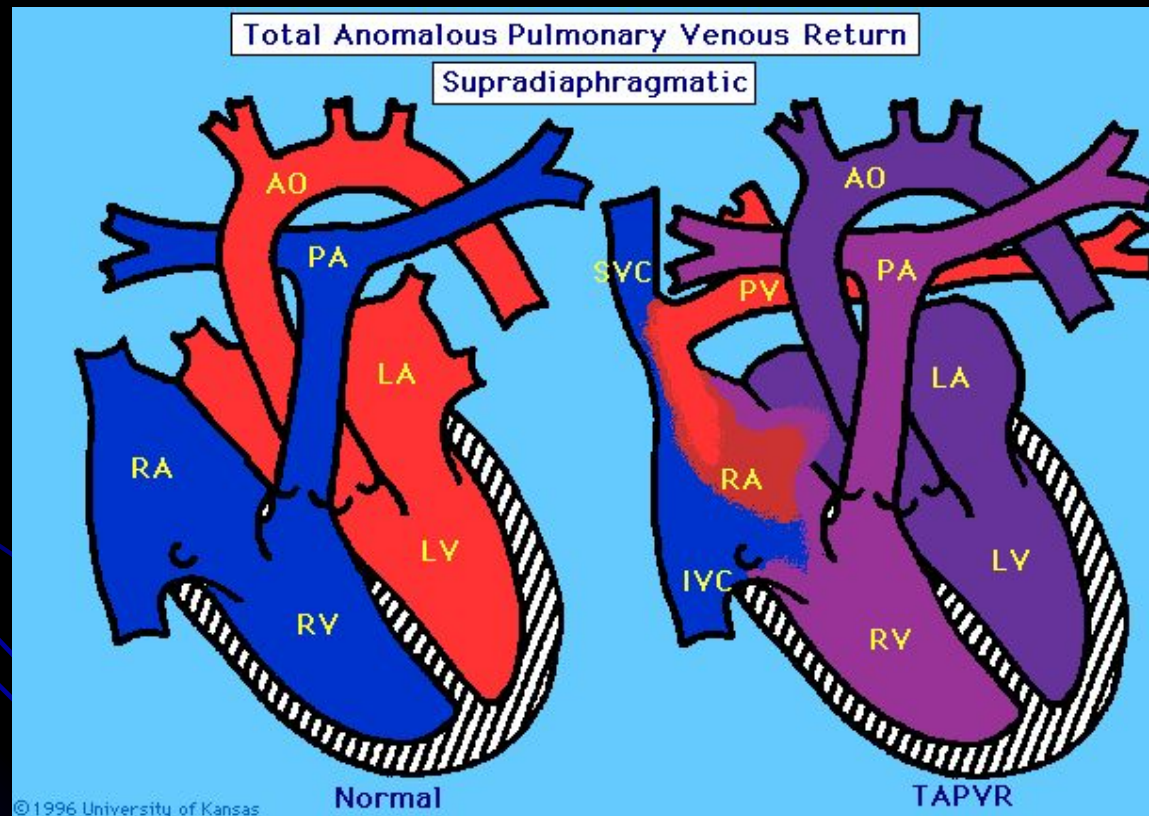
# Total Anomalous Pulmonary Venous Connection

- Pulmonary veins do not make a direct connection with the left atrium.
  - Blood reaches the left atrium only through an atrial septal defect or patent foramen ovale.
  - Pulmonary congestion, tachypnea, cardiac failure, and variable cyanosis.
- 

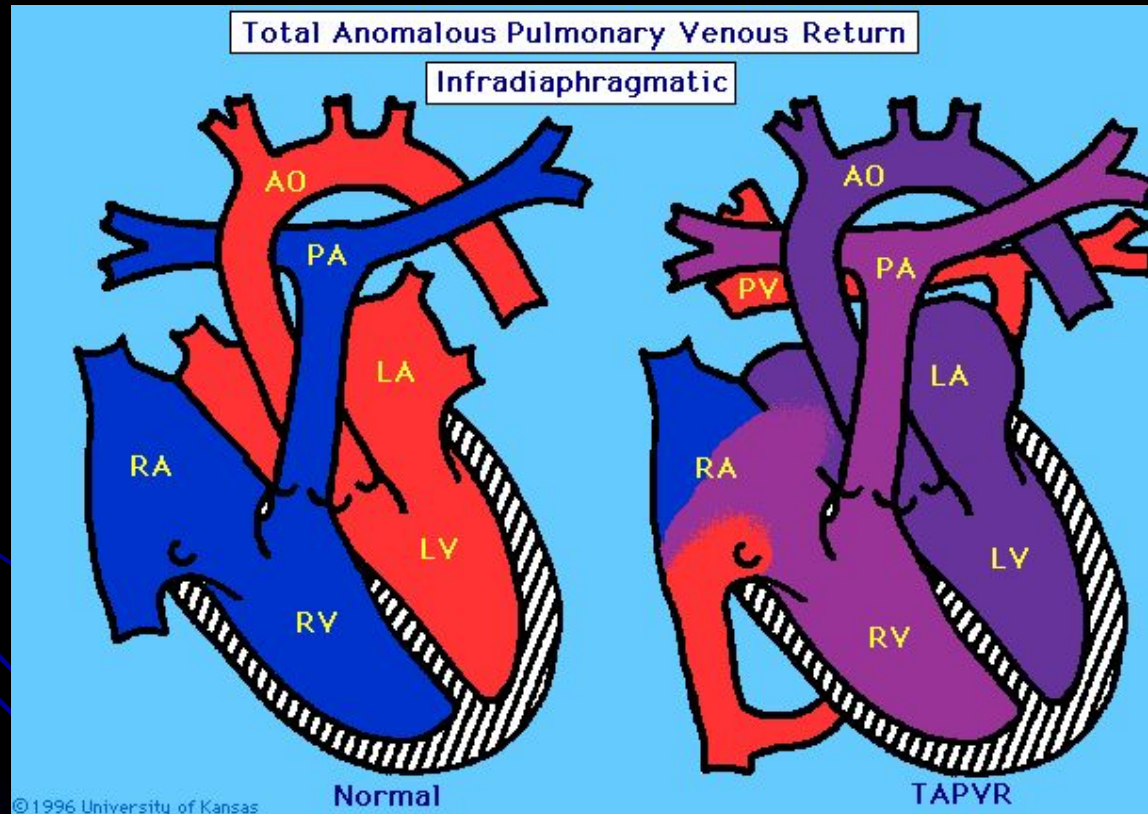
# Total Anomalous Pulmonary Venous Connection

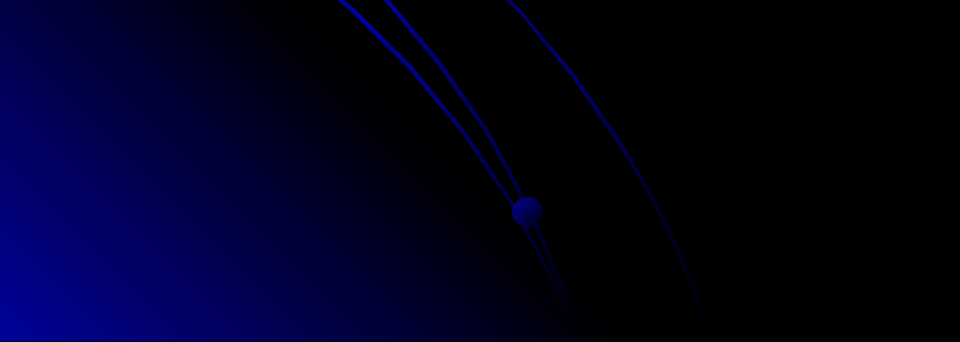
- Diagnosis by cardiac catheterization or echocardiography.
  - Operative repair in all cases.
- 

# Total Anomalous Pulmonary Venous Return (TAPVR)

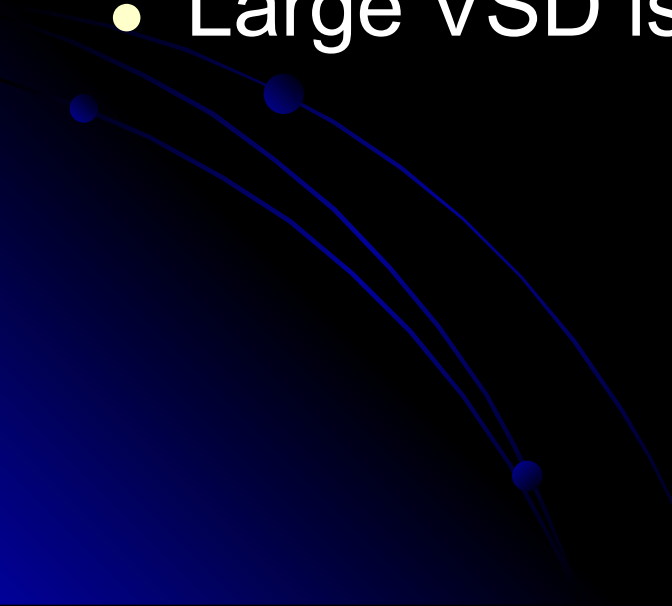


# TAPVR- Infracar



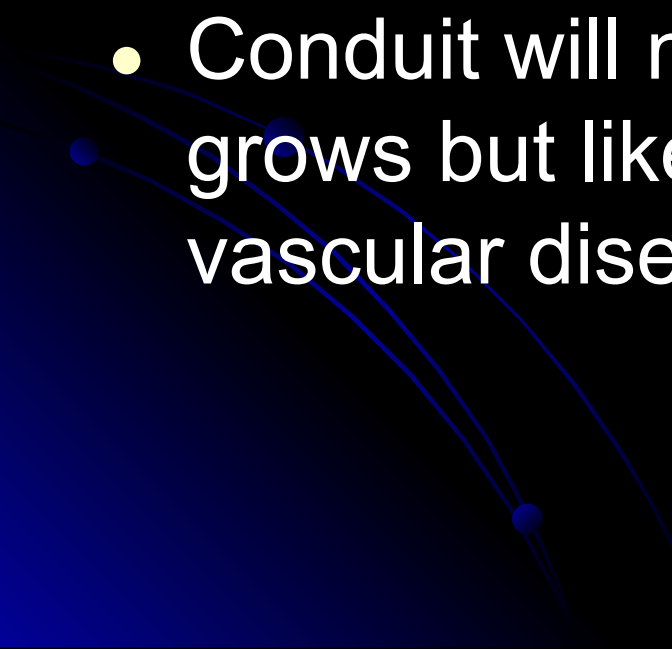


# Truncus Arteri

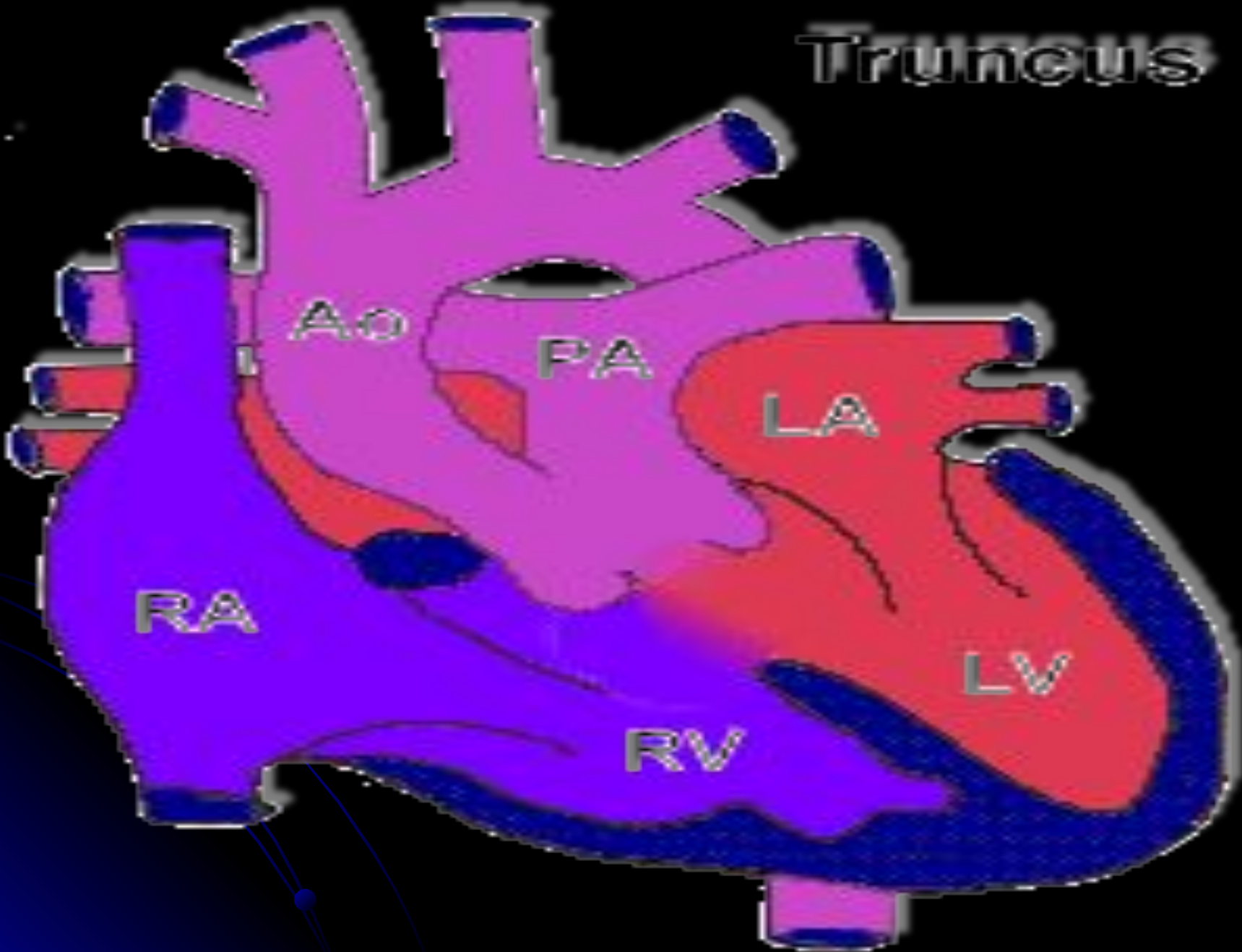
- Single large vessel overrides the ventricular septum and distributes all the blood ejected from the heart.
  - Large VSD is present.
- 



# Truncus Arteri

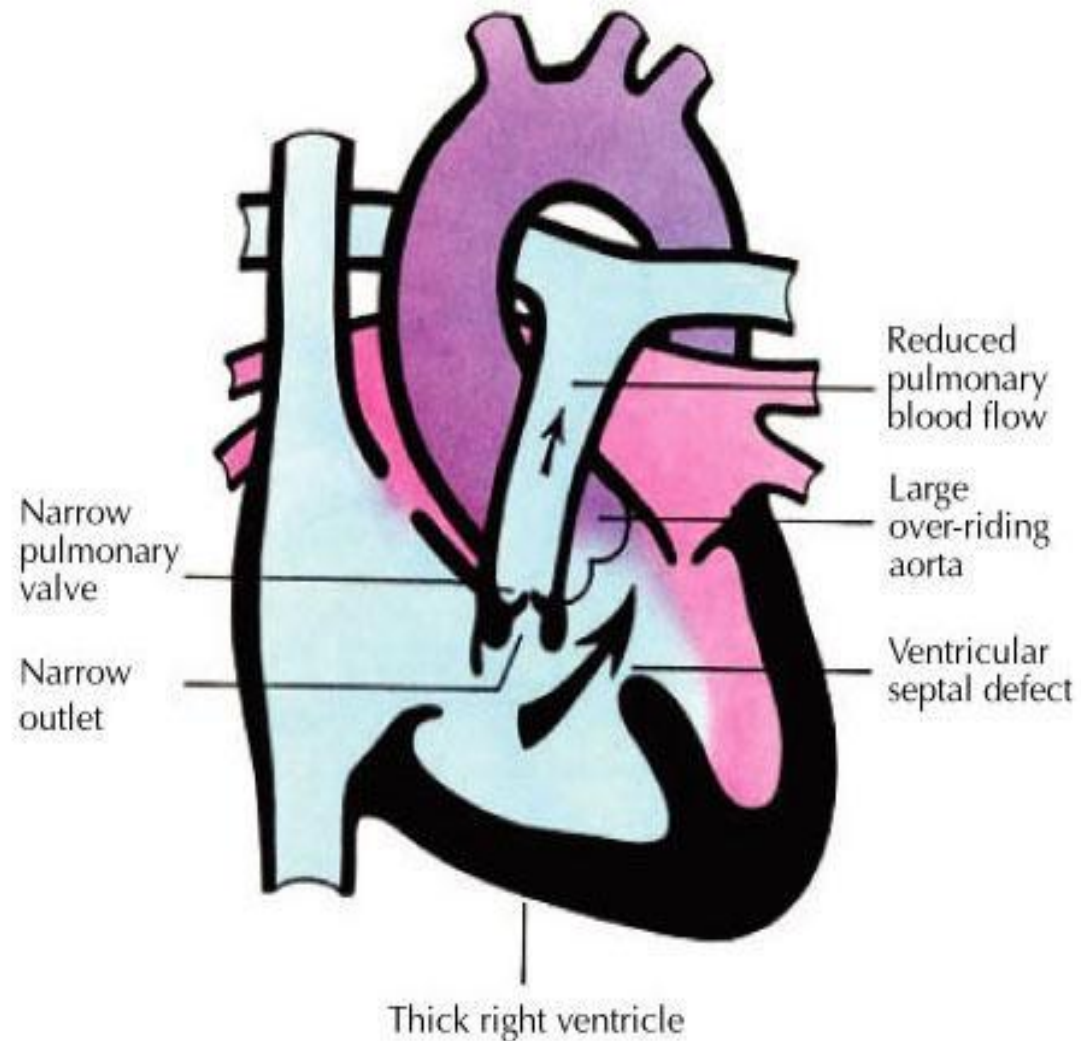
- Corrective operation with a valved conduit between right ventricle and pulmonary vessels.
  - Conduit will need to be changed as child grows but likelihood to develop pulmonary vascular disease is greatly reduced.
- 

# Truncus

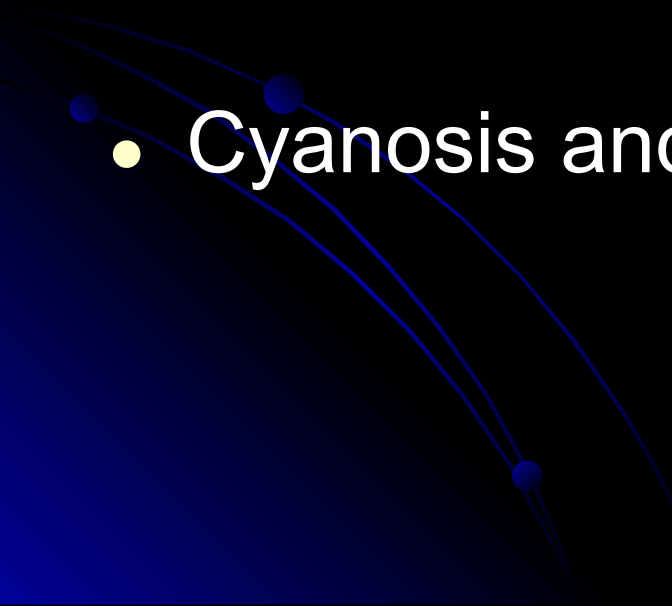


# Tetralogy of Fallot (TO

RVOT  
obstruction  
VSD  
Overriding aorta  
RV hypertrophy



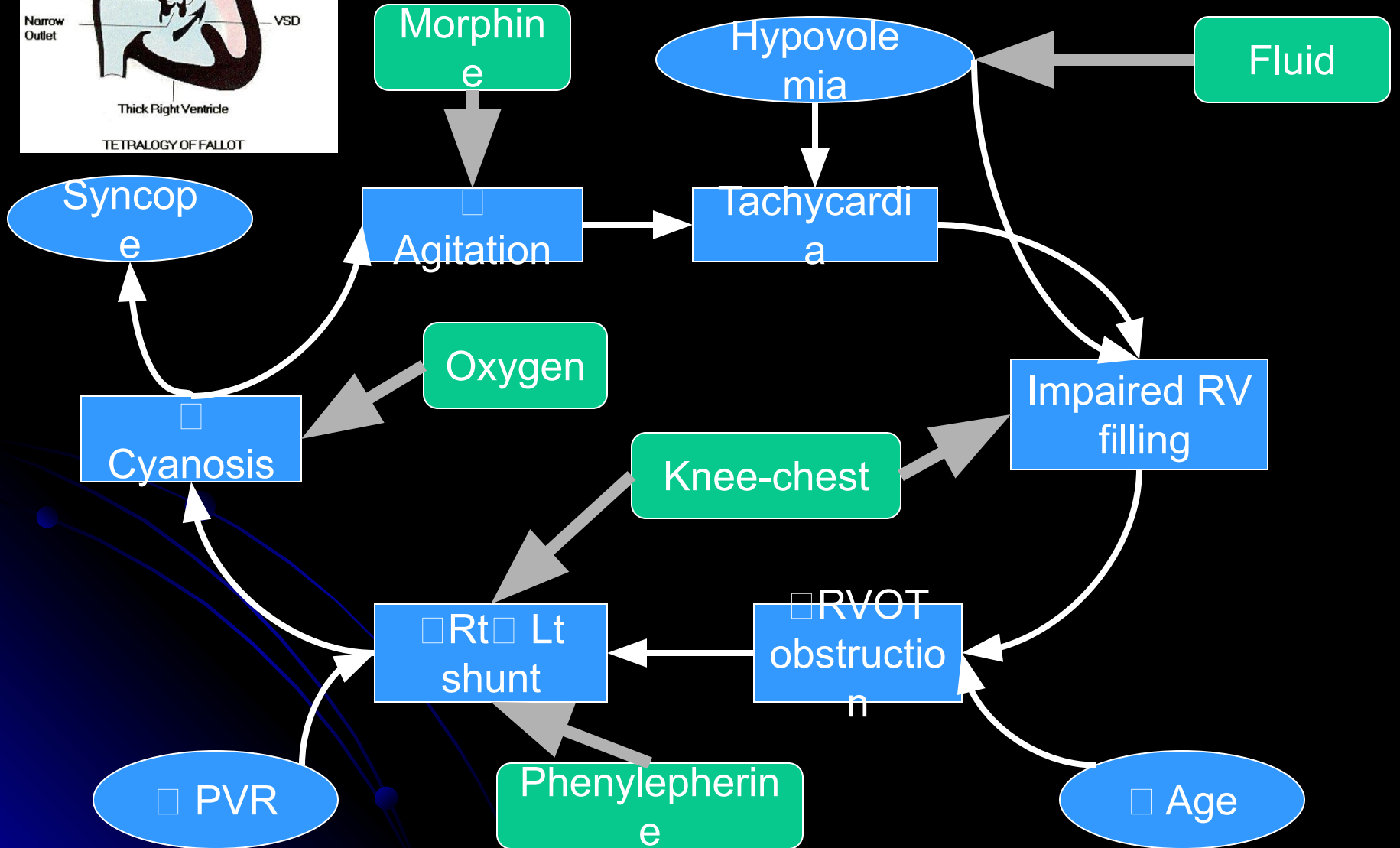
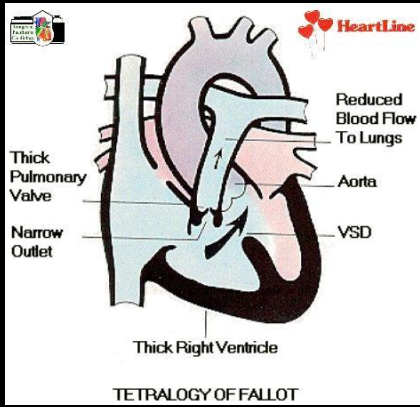
# Tetralogy of

- Addition of an atrial septal defect falls in the category of Pentalogy of Fallot.
  - Hypoxic spells and squatting.
  - Cyanosis and clubbing.
- 

# Clinical Fea

- Asymptomatic infant with murmur is very common in the usual TOF patients
- Murmur of RVOT obstruction can be confused with VSD in infancy
- Cyanosis
  - Typically appears between 6wks and 6 months in the unrepaired infant
  - Nail beds and mucous membranes
  - May be present at rest or only with agitation/exercise
- Persistent cyanosis and clubbing if not repaired

# Hypercyanotic Spells ( TET spells)



# Tetralogy of Fallot

- **Diagnosis**

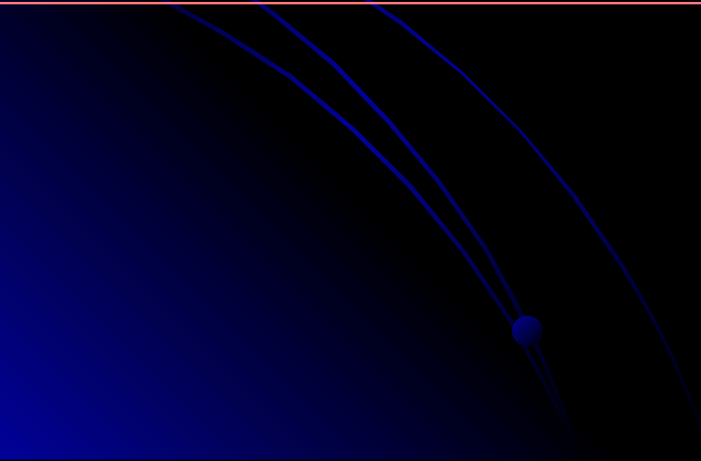
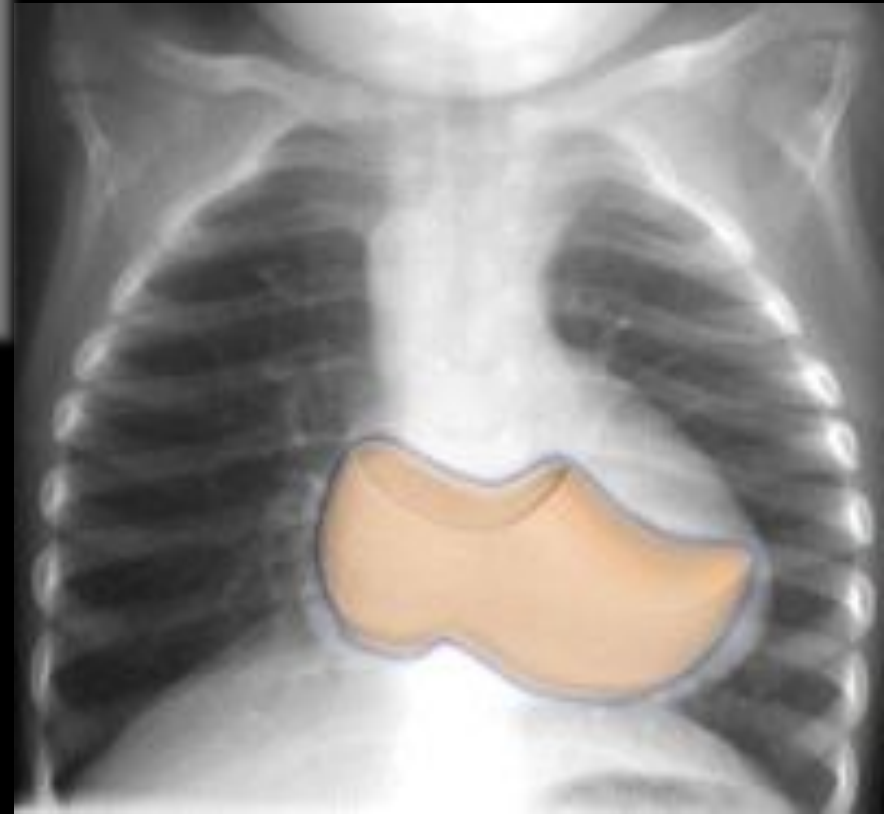
Chest X-ray: Boot-shaped heart, dark lung fields

ECG: right axis deviation, RVH

Echocardiogram (gold standard)

**Pre-correction complications:** cerebral thromboses, brain abscess, bacterial endocarditis, HF.

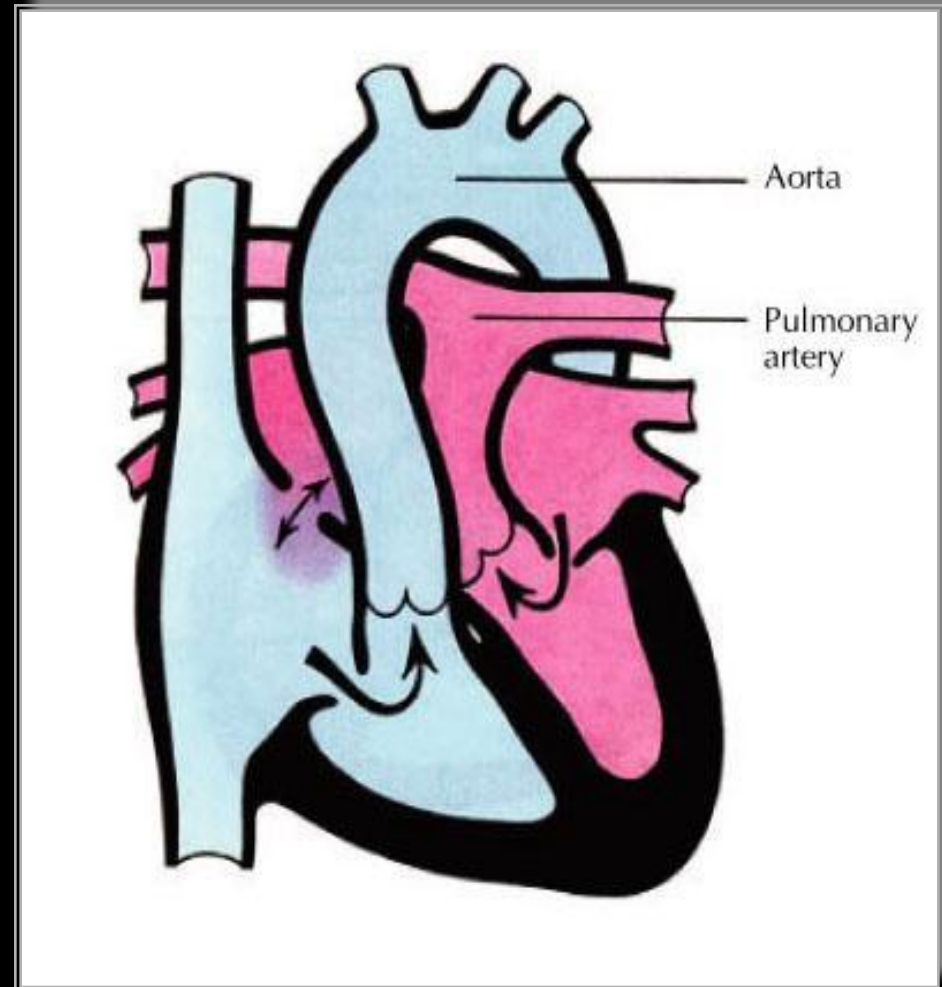
**management:** surgical repair including closure of VSD and widening of RVOTO

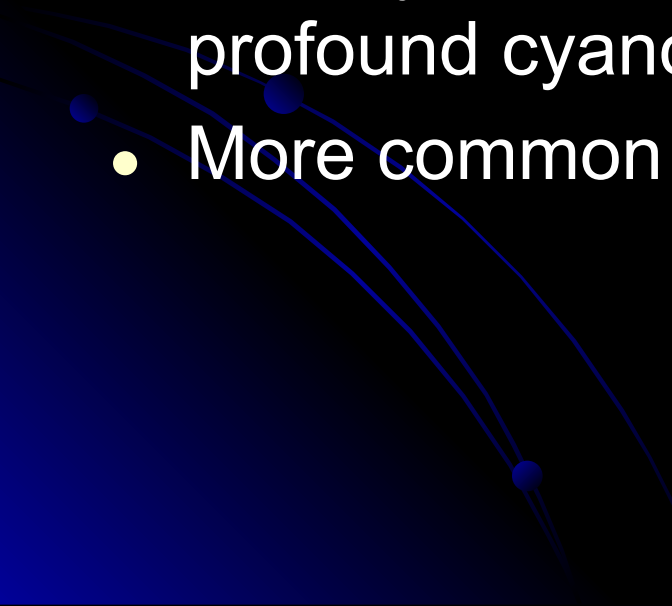




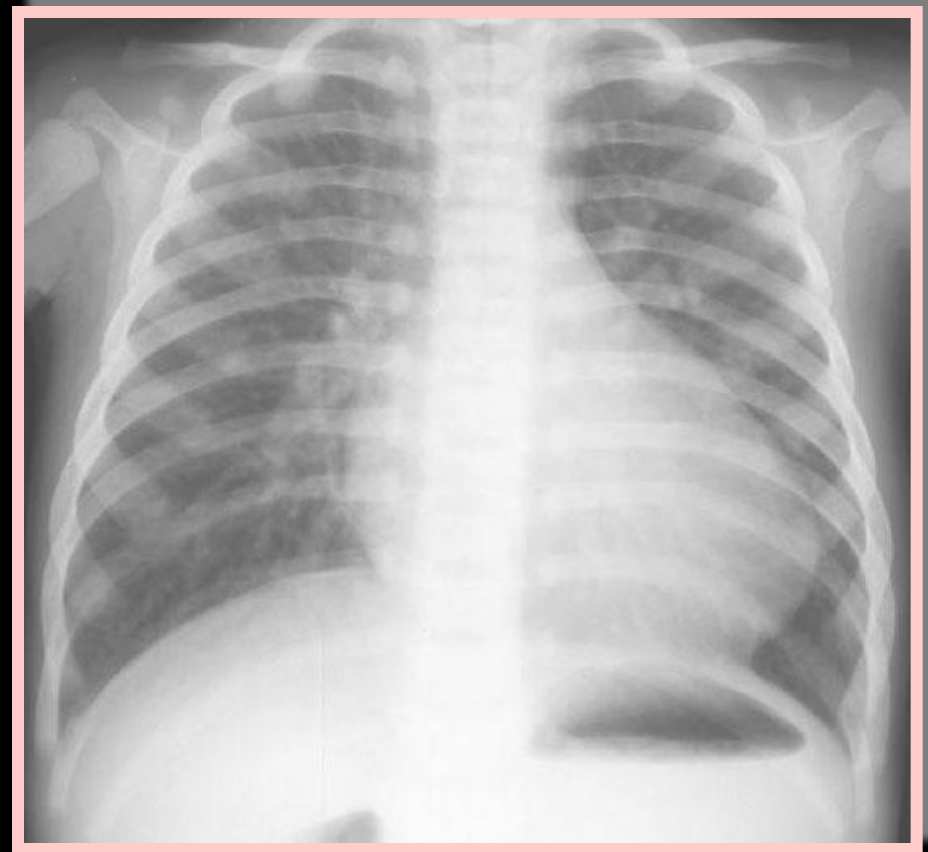
# Transposition of Great (TGA)

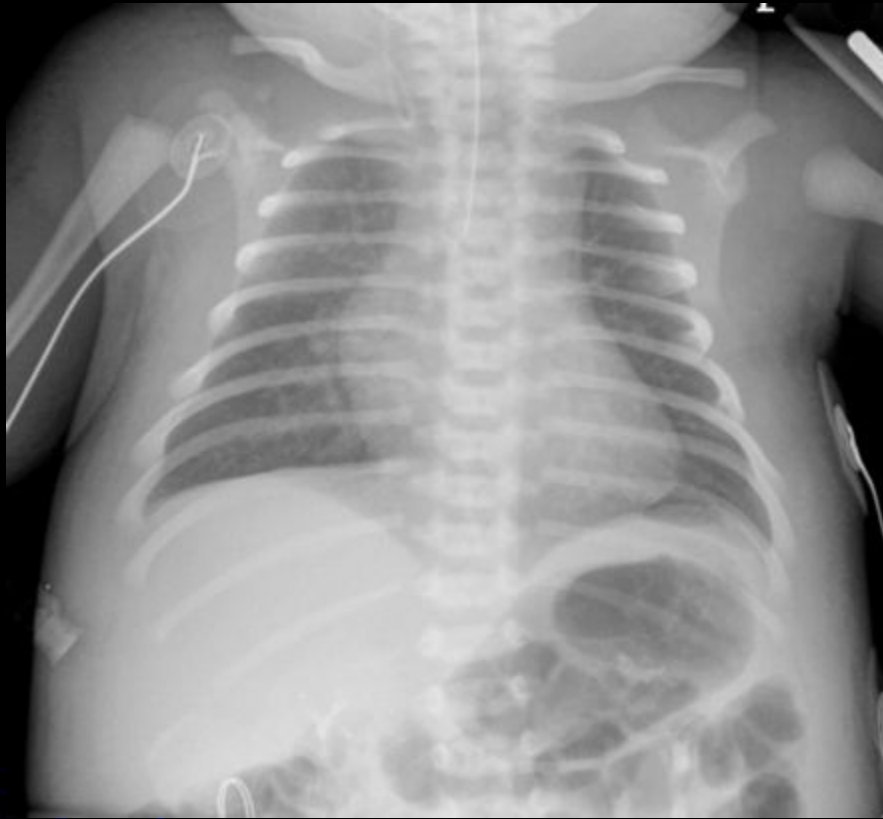
- Aorta originating from the right ventricle, and pulmonary artery originating from the left ventricle
- Accounts for 5-7% of all congenital heart disease



- Survival is dependent on the presence of mixing between the pulmonary and systemic circulation
  - Atrial septal defect is essential for survival
  - 50% of patients have a VSD
  - Usually presents in the first day of life with profound cyanosis
  - More common in boys
- 

- Exam :
  - cyanosis in an otherwise healthy looking baby
  - Loud S2 ( aorta is anterior )
- CXR :
  - Egg on side
  - Narrow mediastinum





# Transposition of the Great Arteries (TGA)

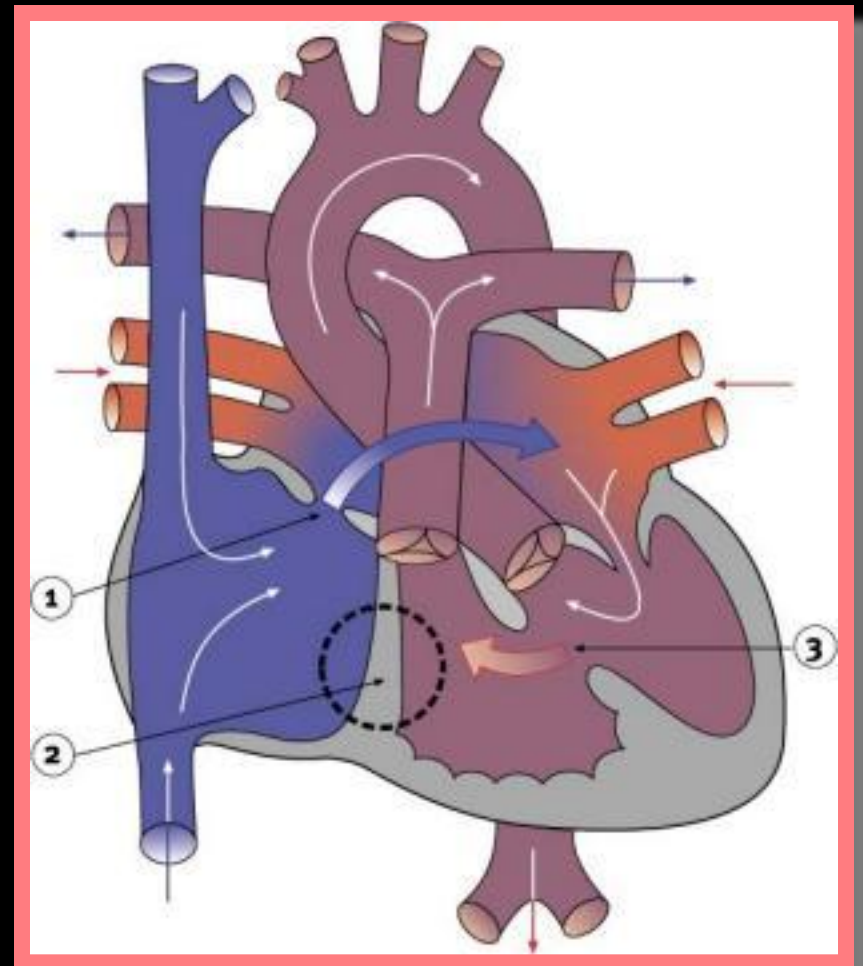
- **Management**

- prostaglandin E1 (PGE1) infusion to keep ductus open until septotomy or surgery
- balloon atrial septostomy with catheter
- surgical correction: arterial switch procedure

infants without VSD must be repaired within 2 weeks to avoid weak LV muscle

# Tricuspid A

- Complete absence of communication between the right atrium and right ventricle
- About 3 % of congenital heart disease





PGE-1, and minimal supplemental O2 to maintain ductal patency

No O2  
Afterload reduction  
Diuretics

# Tricuspid A

- Repair consists of shunt from right atrium to pulmonary artery or rudimentary right ventricle (Fontan procedure).





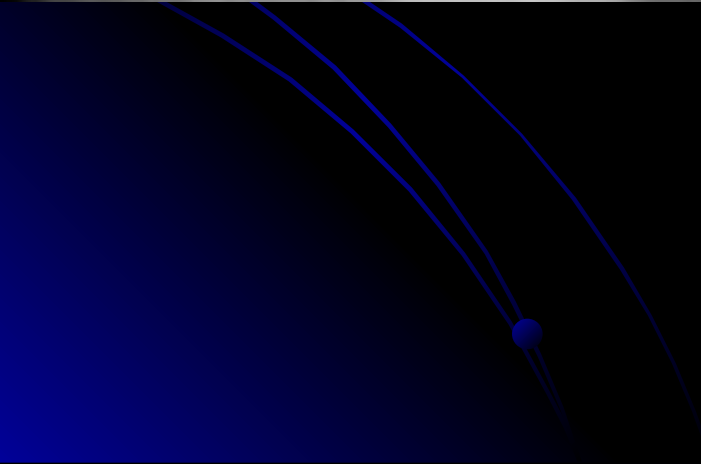
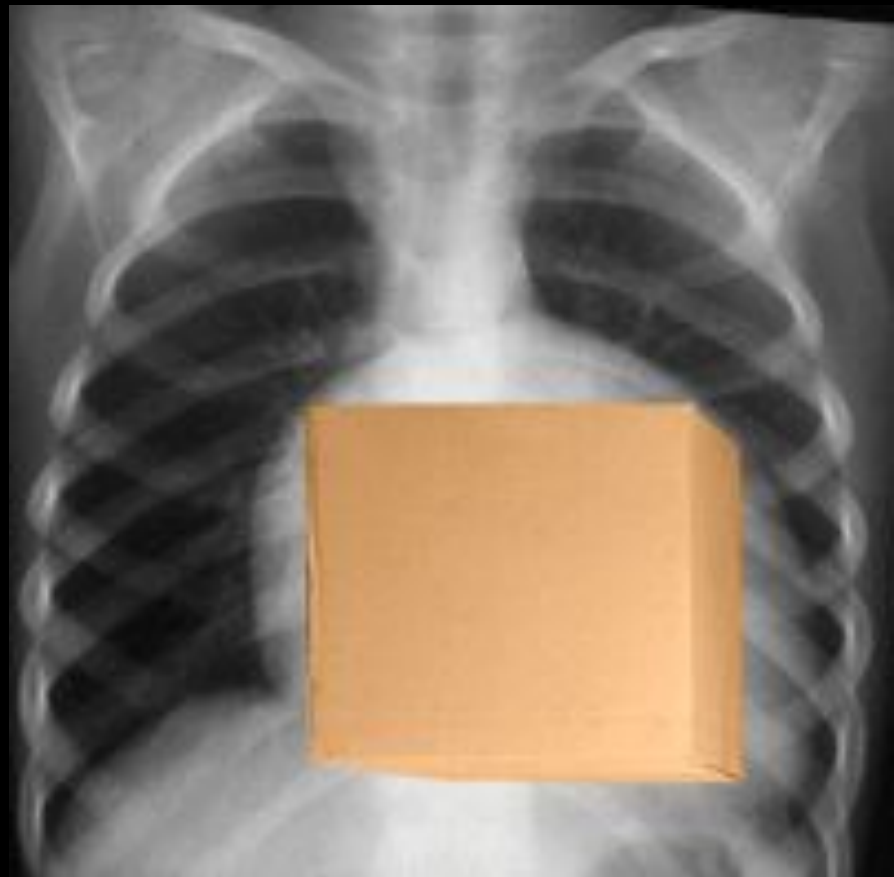
# Ebstein's Anom

- Septal and posterior leaflets of the tricuspid valve are small and deformed, usually displaced toward the right ventricular apex.
- Most patients have an associated ASD or patent foramen.
- Cyanosis and arrhythmias in infancy are common.

# Ebstein's Anom

- Right heart failure in half of patients.
- Operative repair with tricuspid valve replacement.





non-shunt  
Acyanotic heart lesions

**Obstruction**

- Aortic stenosis AS
- Supravalvar AS
- Subaortic S
- Coarctation
- Mitral Stenosis
- Pulmonary Stenosis

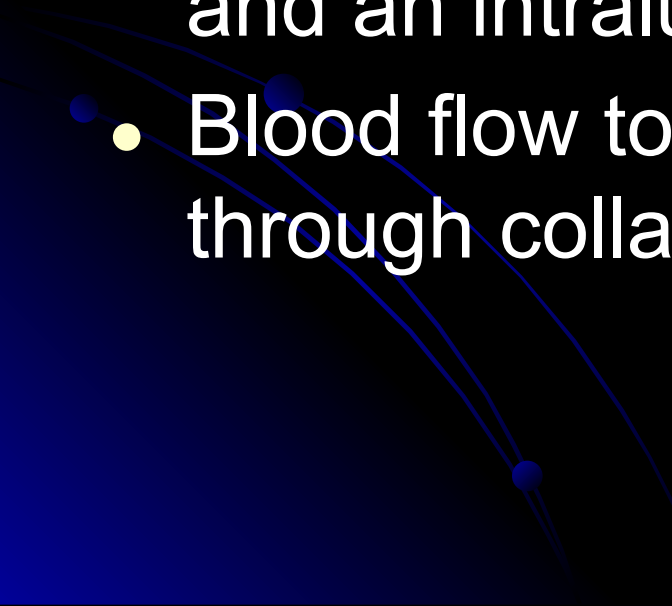
Generally cause  
pressure overload

**Regurgitation**


- Aortic regurgitation
- Mitral regurgitation
- Pulmonary regurgitation

Generally cause  
Volume overload

# Coarctation of the Ao

- Males twice as frequently as females.
  - 98% of all coarctations at segment of aorta adjacent to ductus arteriosus.
  - Produced by both an external narrowing and an intraluminal membrane.
  - Blood flow to the lower body maintained through collateral vessels.
- 

# Coarctation of the Ao

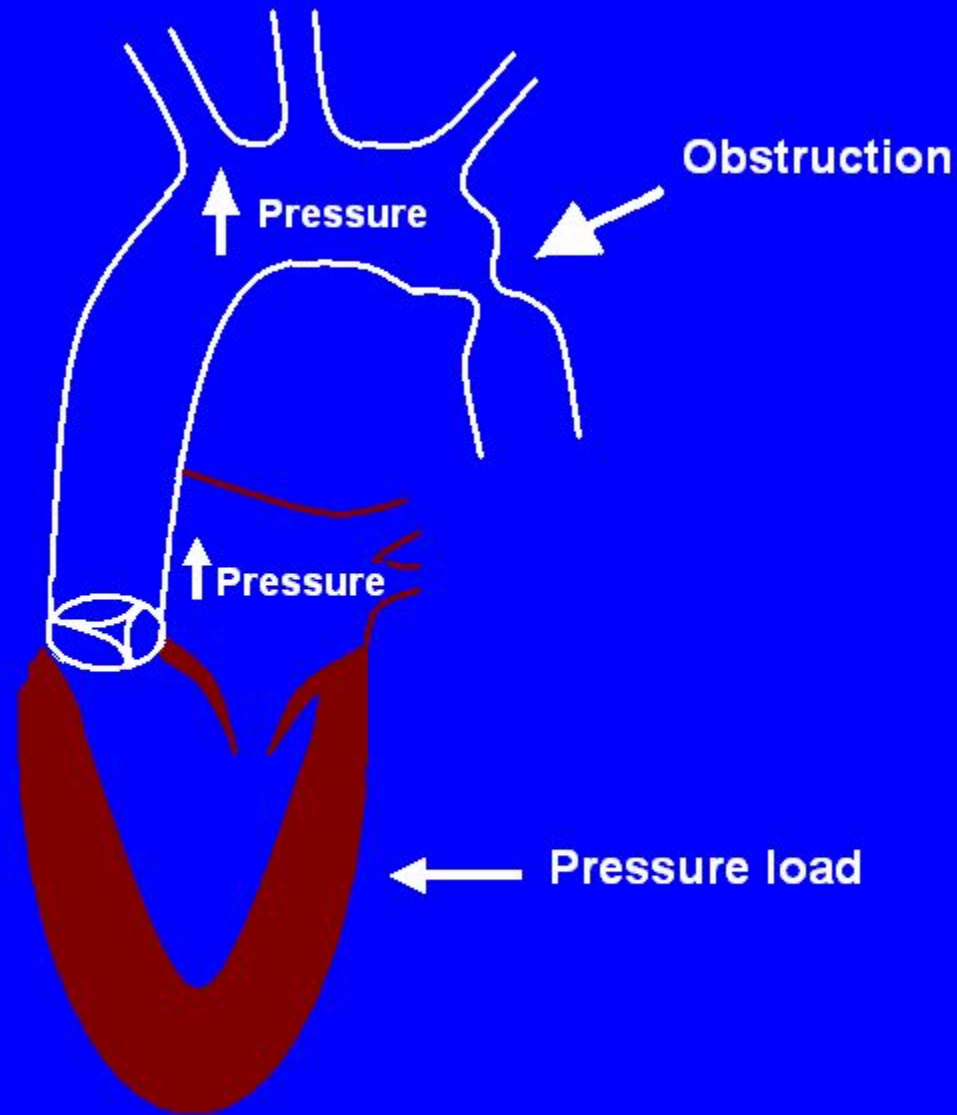
- Absent or weak femoral pulses.
  - Systolic pressure higher in upper extremities than in lower extremities; diastolic pressures are similar.
  - Harsh systolic murmur heard in the back.
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**Hypertension**  
**Pressure gradient**

**Left ventricular**  
**Hypertrophy**

**Heart Failure**  
**Pulmonary Edema**

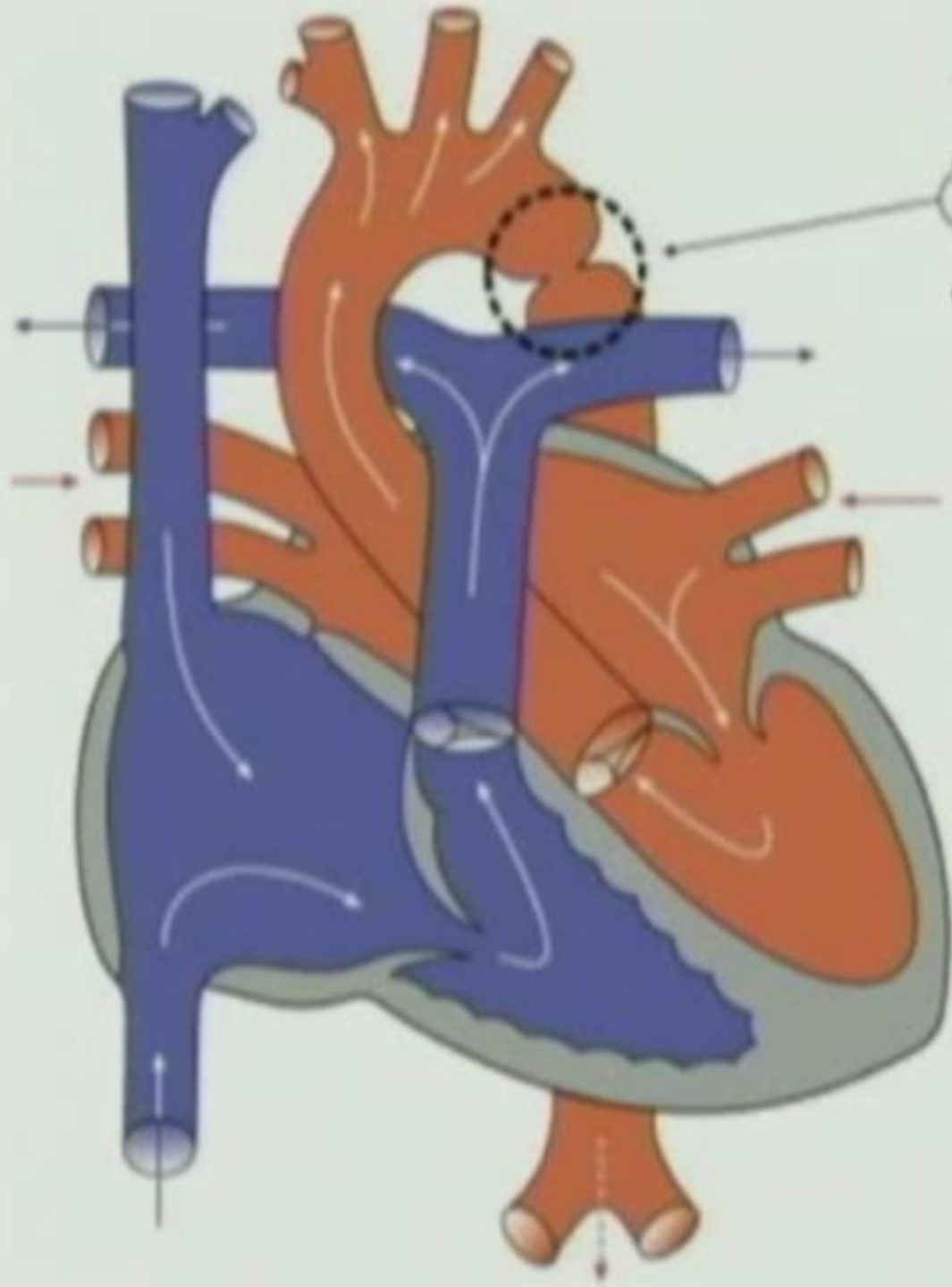
**Pulmonary**  
**Hypertension**



# Coarctation of the Aorta

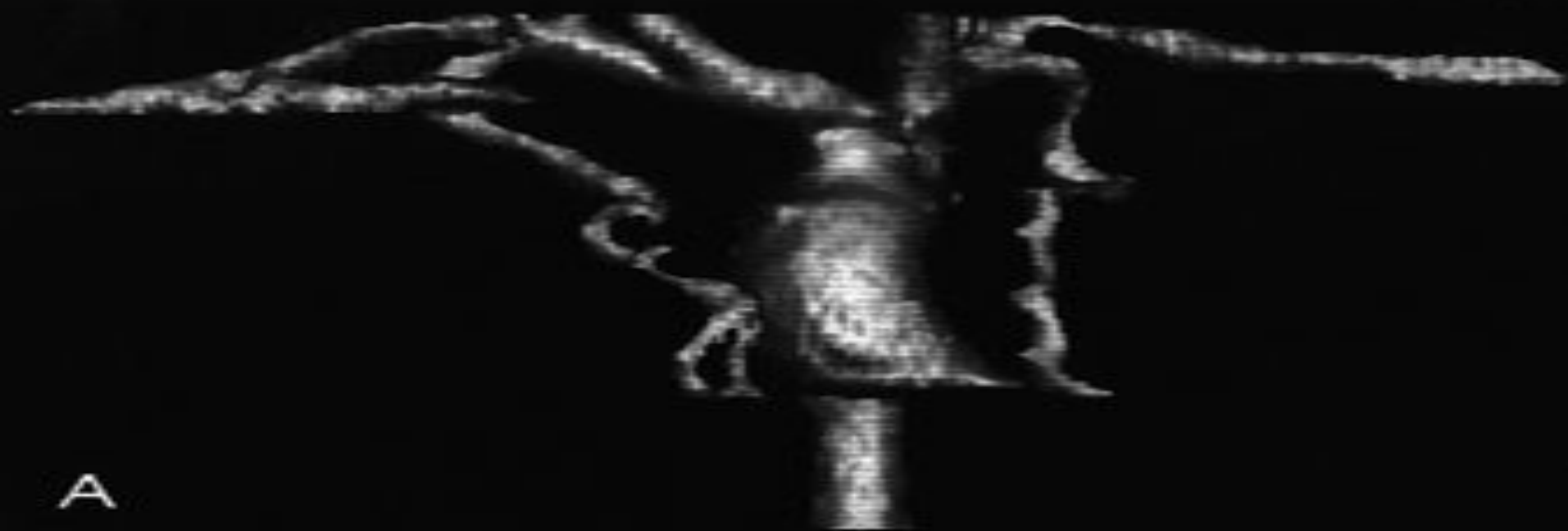
- Older child if milder “juxtaductal” – 90% turner syndrome.
- May hear murmur
- May present with hypertension  
lag of femoral pulse
- Four- extremity BP –decrease ( $>5$ ) in lower extremities
- If pressure : right arm  $>$  left arm = involving left subclavian artery





# Coarctation of the Aorta

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# Coarctation of the Aorta

- **Diagnostic tests**

## Chest X-ray

- Notching of inferior border of ribs
- Poststenotic dilatation of ascending aorta

ECG: RVH early in infancy, LVH later in childhood



**Rib Notching**

# Coarctation of the Aorta

- **Management:**

Neonats :

- **PGE1**

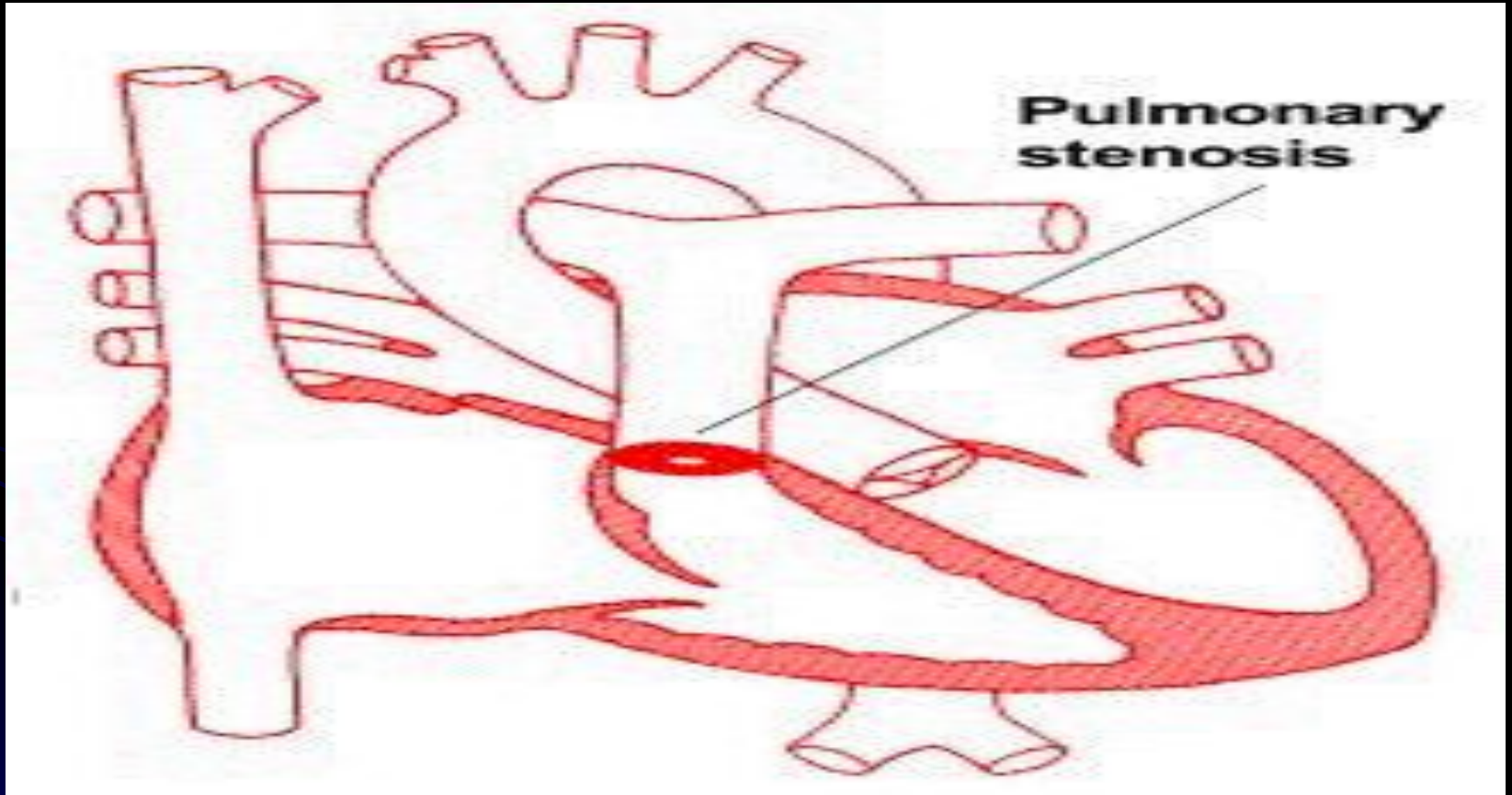
- **balloon arterioplasty or surgical correction after stabilization**

- **Older** : treat hypertension then surgery

**Complications:** essential hypertension , aortic aneurysms

endocarditis

# Pulmonary Stenosis

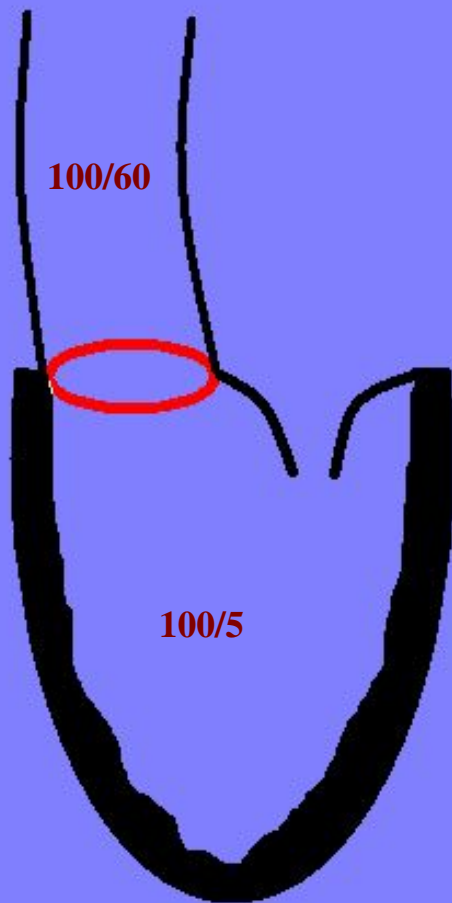


# Pulmonary Ster

- No symptoms in mild or moderately severe lesions.
- Cyanosis and right-sided heart failure in patients with severe lesions.
- High pitched systolic ejection murmur maximal in second left interspace.
- Ejection click often present.

# Aortic st

Normal

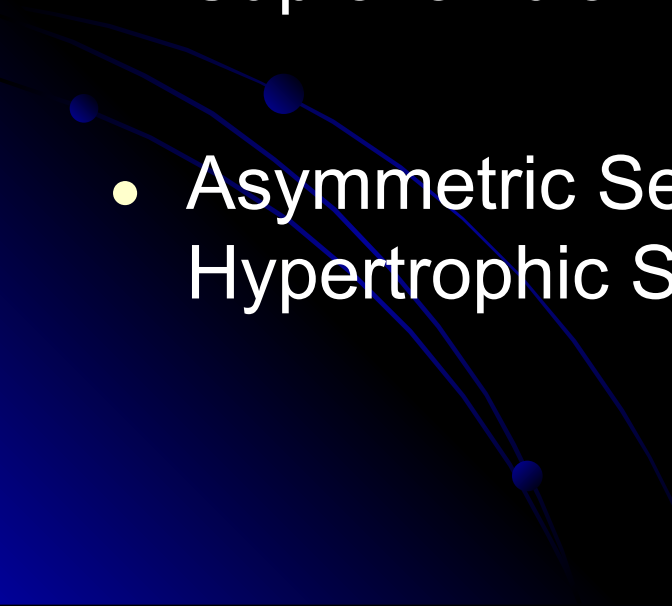


Aortic Stenosis





# Aortic St

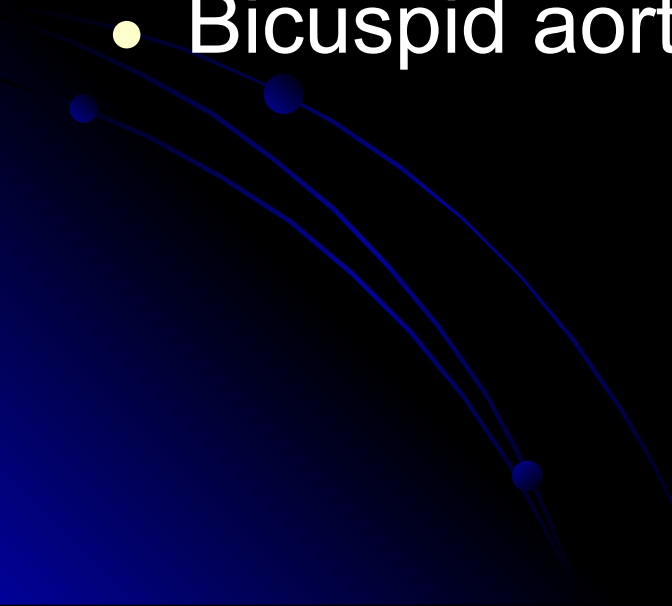
- Valvular Aortic Stenosis
  - Subaortic Stenosis
  - Supravalvular Aortic Stenosis
  - Asymmetric Septal Hypertrophy (Idiopathic Hypertrophic Subaortic Stenosis)
- 

# Valvular Aortic Steno

- Most common type, usually asymptomatic in children.
- May cause severe heart failure in infants.
- Prominent left ventricular impulse, narrow pulse pressure.
- Harsh systolic murmur and thrill along left sternal border, systolic ejection click.



# Valvular Aortic Stenosis

- Predominantly in males
  - Thickened, fibrotic, malformed aortic leaflets.
  - Fused commissures
  - Bicuspid aortic valve.
- 

# Congenital malformat syndrome with CH

- Down syndrome >>>endocardil cushion defect , VSD and ASD
- Trisomy 18 >>> VSD , ASD ,PDA ,Coarctation
- Trisomy 13 >>> VSD , ASD ,PDA ,Coarctation
- Turner syndrome >>> bicuspid aortic valve and coarctation of aorta
- Fragile x syndrome >> mitral valve prolapse and aortic root dilatation
- CHARGE syndrome >> VSD ,ASD ,PDA ,TOF ,Cushion defect

- Di George syndrome >>aortic arch anomalies
- Alagille syndrome >> peripheral pulmonary stenosis , supravalvular aortic stenosis
- VATER>> VSD ,TOF ,ASD ,PDA
- Asplenia syndrome >> complex CHD
- Polysplenia syndrome >> ASD ,VSD ,PDA
- Congenital rubella >>PDA
- Maternal PKU>>ASD .VSD, PDA , coarctation

- Polycystic kidney disease >> mitral prolapse
- Diabetes >> hypertrophic cardiomyopathy ,VSD,TGA
- Kartagener syndrome >> dextrocardia
- Noonan syndrome >> pulmonary stenosis ,ASD, cardiomyopathy
- Marfan syndrome >> aortic dissection , aortic regurgitation , mitral prolapse

# CXR finding with C

- Figure of 8 ( snowman ) >> total anomalous
- Boot shape >>> TOF
- Notching of ribs >>coarction of aorta
- Flask shape >>> pericardial effusion
- Egg on strip >> TGA



# Causes of heart failure

- In neonatal period >>CoA , hypoplastic left heart , sever aortic stenosis and truncus arteriosus
- 3-4 months>>> TOF
- 4 months – 1 year >> VSD , PDA and total anomalous
- Adulthood >>ASD

.... The most common

- Murmur >> innocent murmur
- CHD>>VSD
- Cyanotic CHD >>in less than 1 month..TGV, more than 1 month TOF
- CHD diagnosed at adulthood >>ASD
- CHD in premature >>> PDA
- Fatal CHD in neonate >> left hypoplastic heart
- Causes of plethoric lungs>>> VSD, ASD PDA
- Dysrhythmia >>> supraventricular tachycardia