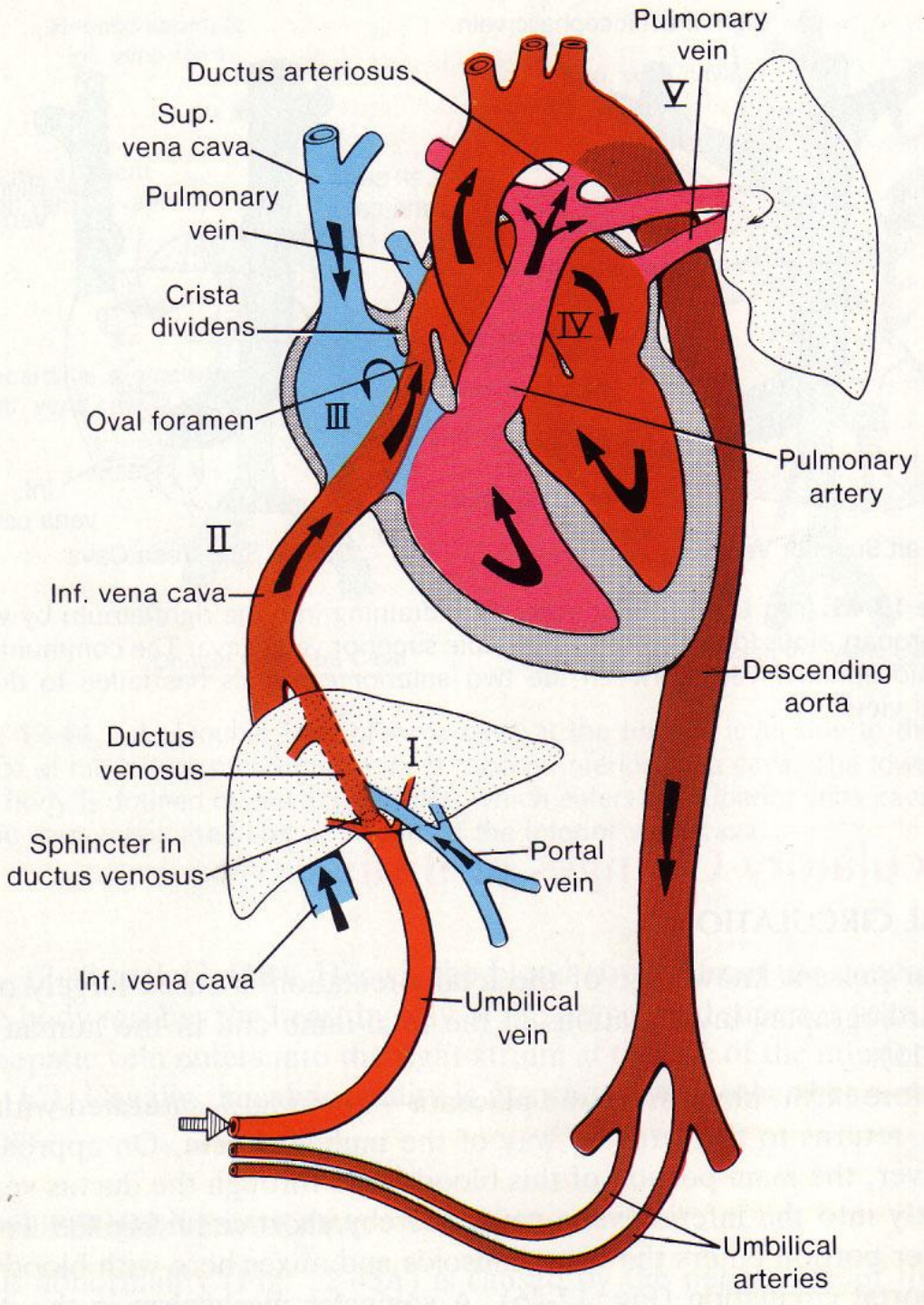
Congenital heart diseases

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

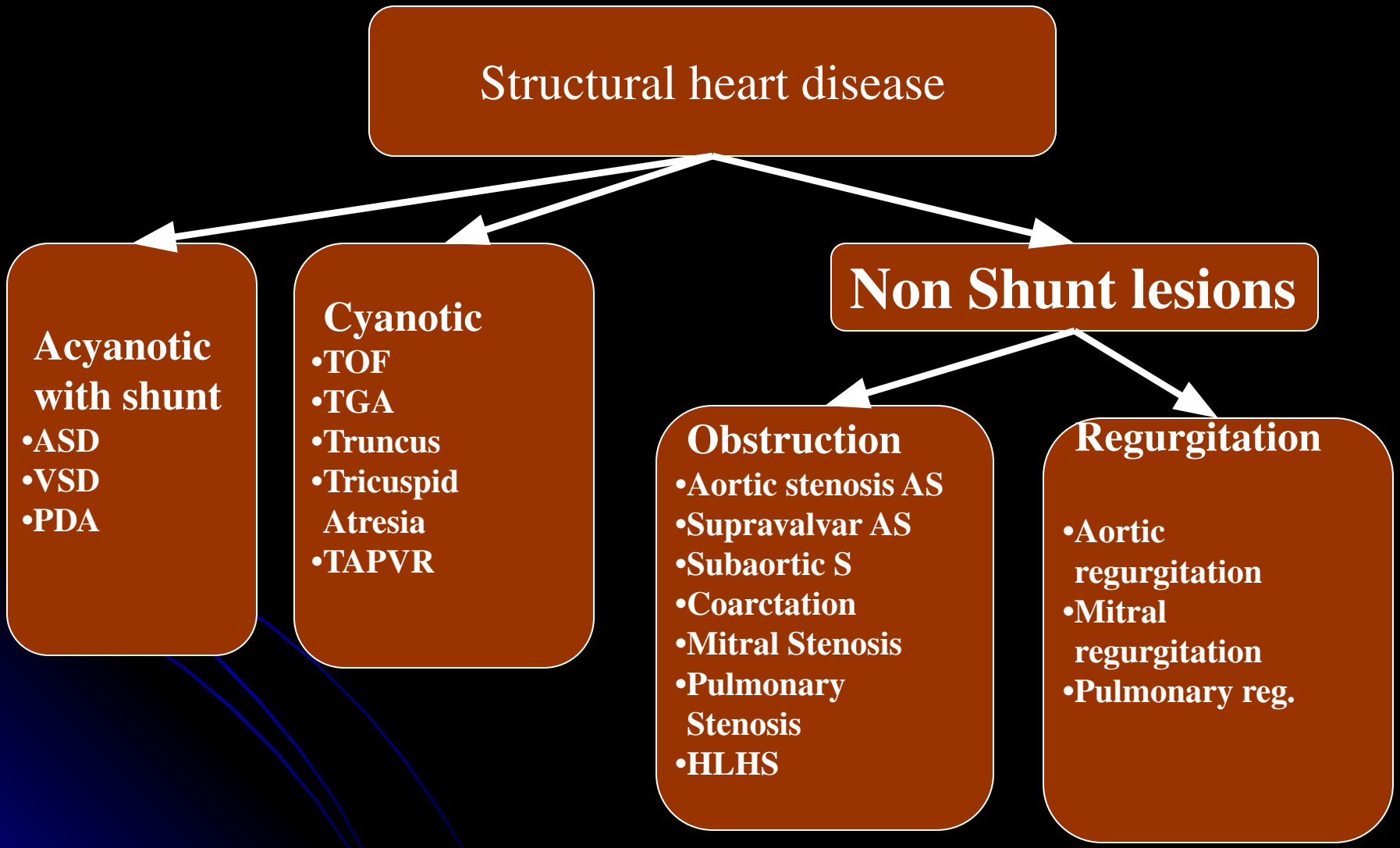
CARDIAC EVALUATION

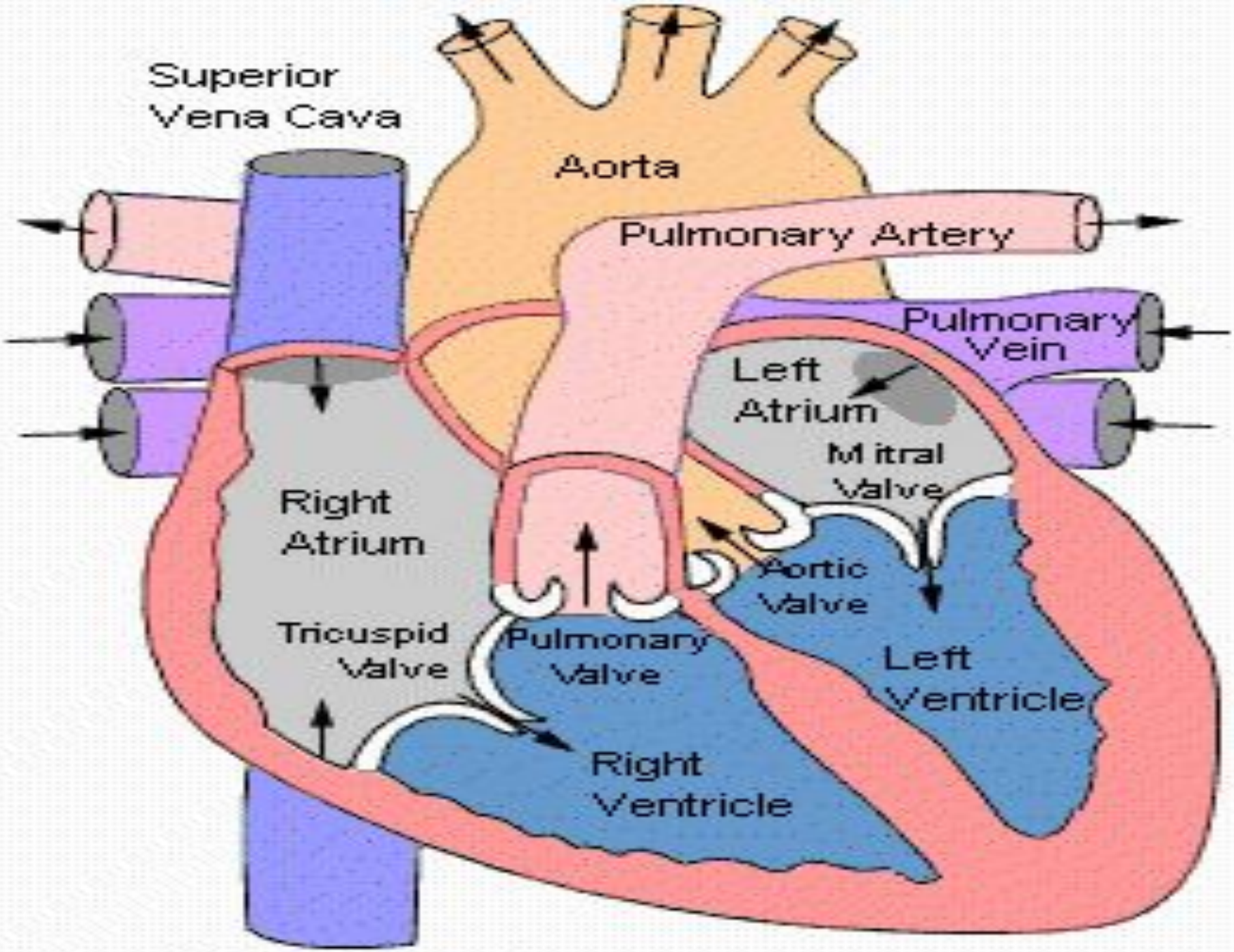
Murmur

Innocent versus pathologic

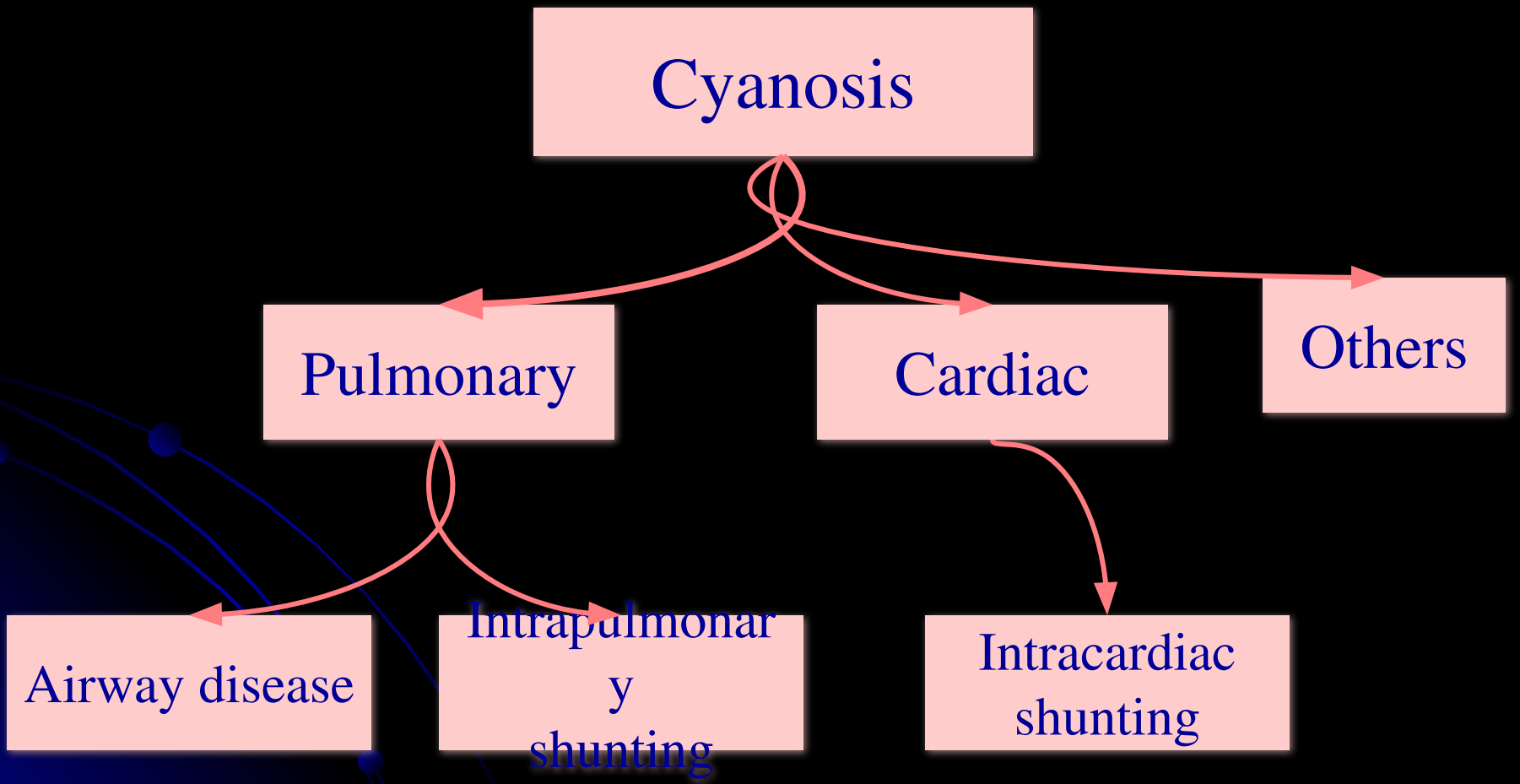
Murmur is pathologic if one or more :

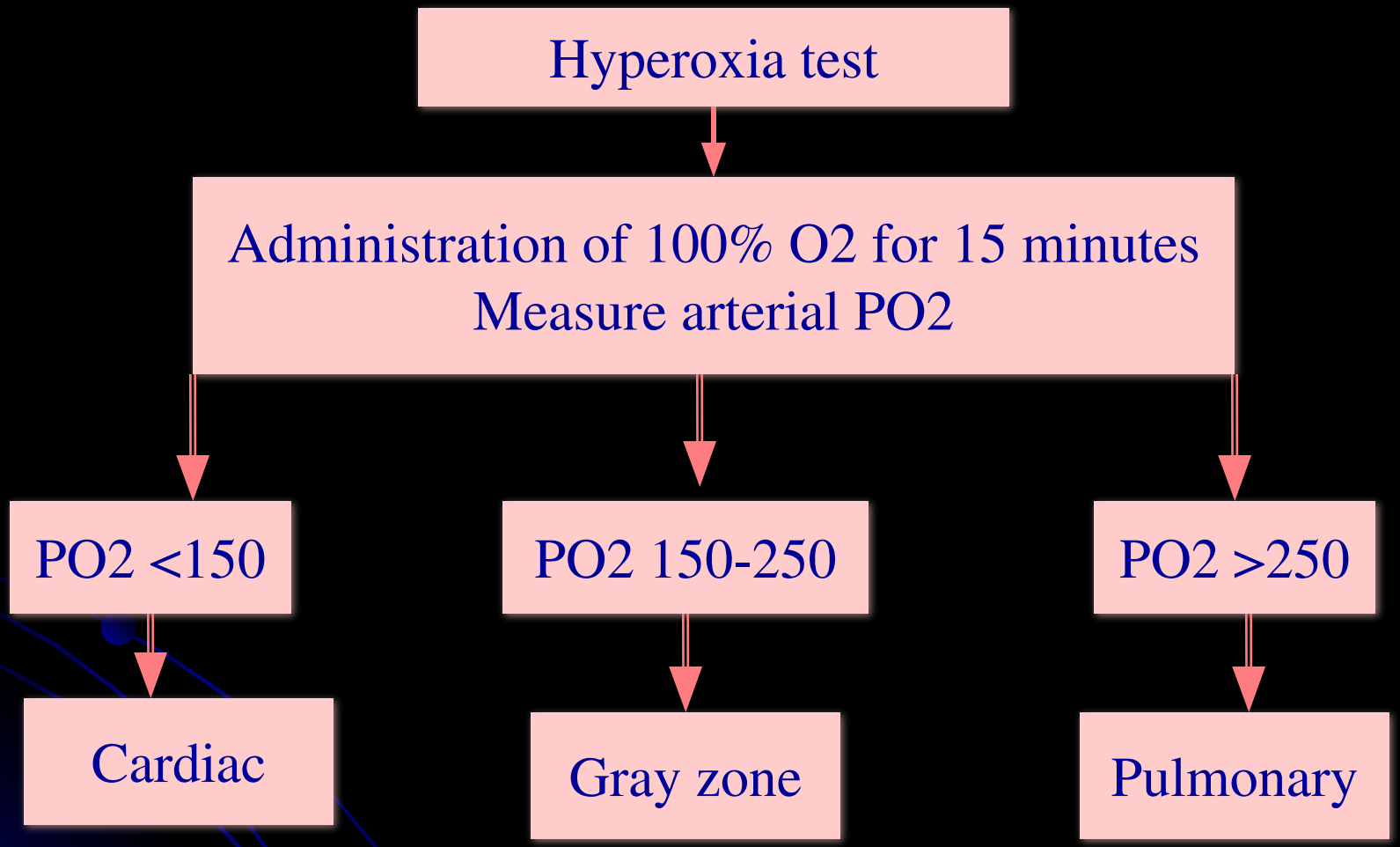
- **Symptoms**
- **Cyanosis**
- **Grade 3/6 or higher**
- **Thrill**
- **Diastolic**
- **Abnormal heart sounds**
- **Abnormally strong or weak pulse**


Structural heart disease
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.


Superior
Vena Cava
Aorta
Pulmonary Artery
Pulmonary
Vein
Left
Atrium
Mitral
Valve
Right
Atrium
Aortic
Valve
Tricuspid
Valve
Pulmonary
Valve
Left
Ventricle
Right
Ventricle

General causes of Cyanosis




Hyperoxia test
Administration of 100% O2 for 15 minutes
Measure arterial PO2
PO2 <150
PO2 150-250
PO2 >250
Cardiac
Gray zone
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 INCREASE Pulmonary Art Blood Flow

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

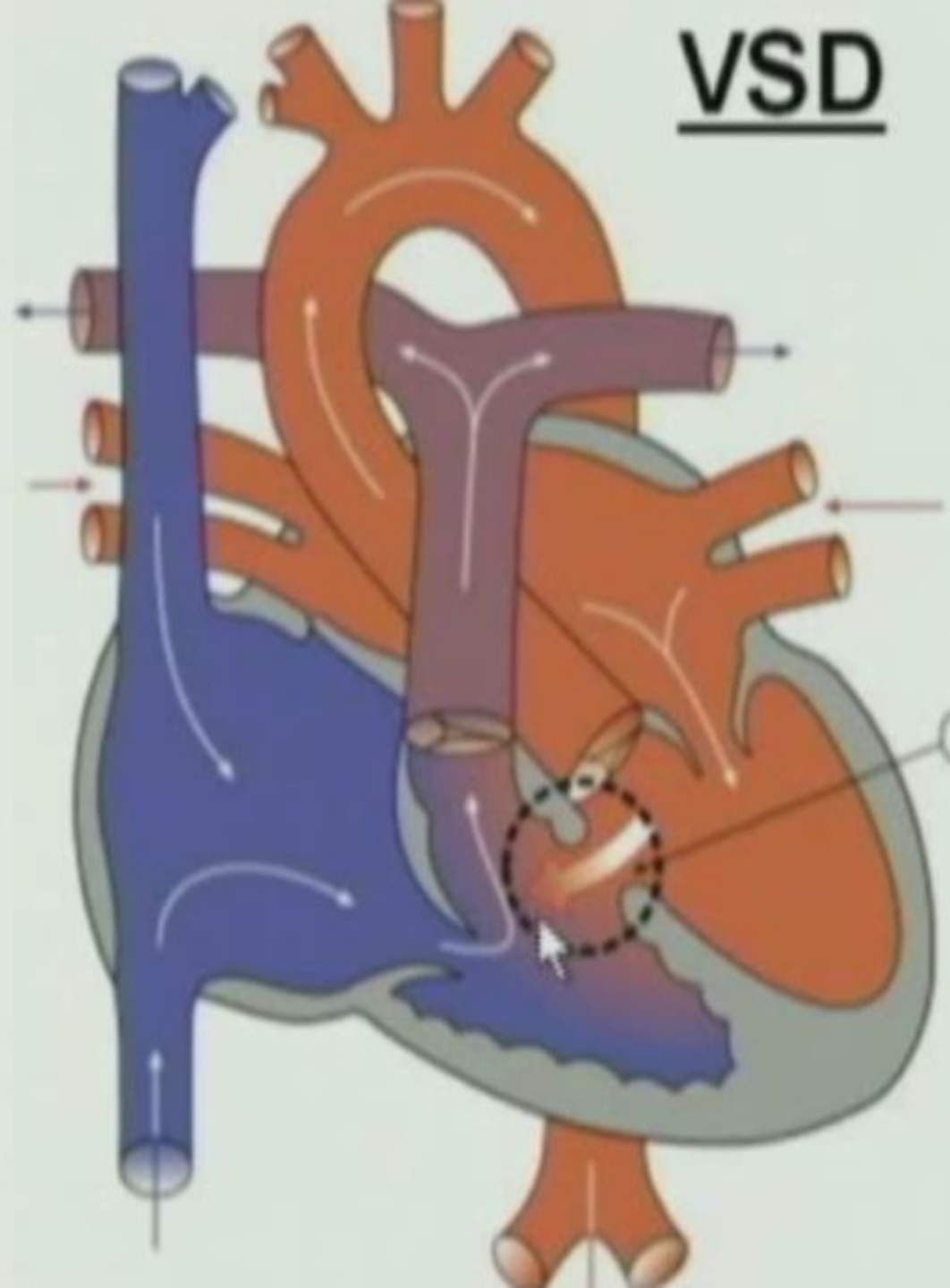
Congenital Heart Disease

- **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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KAPLAN MEDICAL

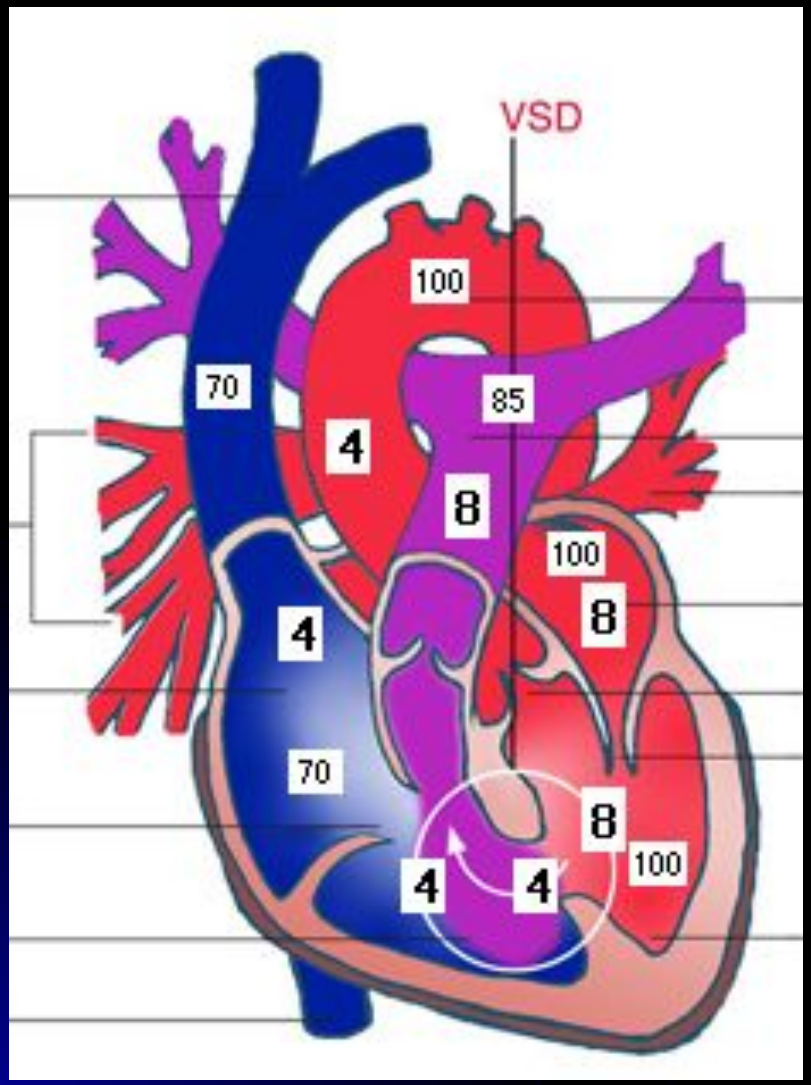
Ventricular septal defect (VSD)

- **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 halosystolic murmur over lower left sternal border +/- thrill
- Male affected as female
- Small defect <2cm and large one >2cm

Type

- Perimembranous (conoventricular 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 Sh
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

Mana

- 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

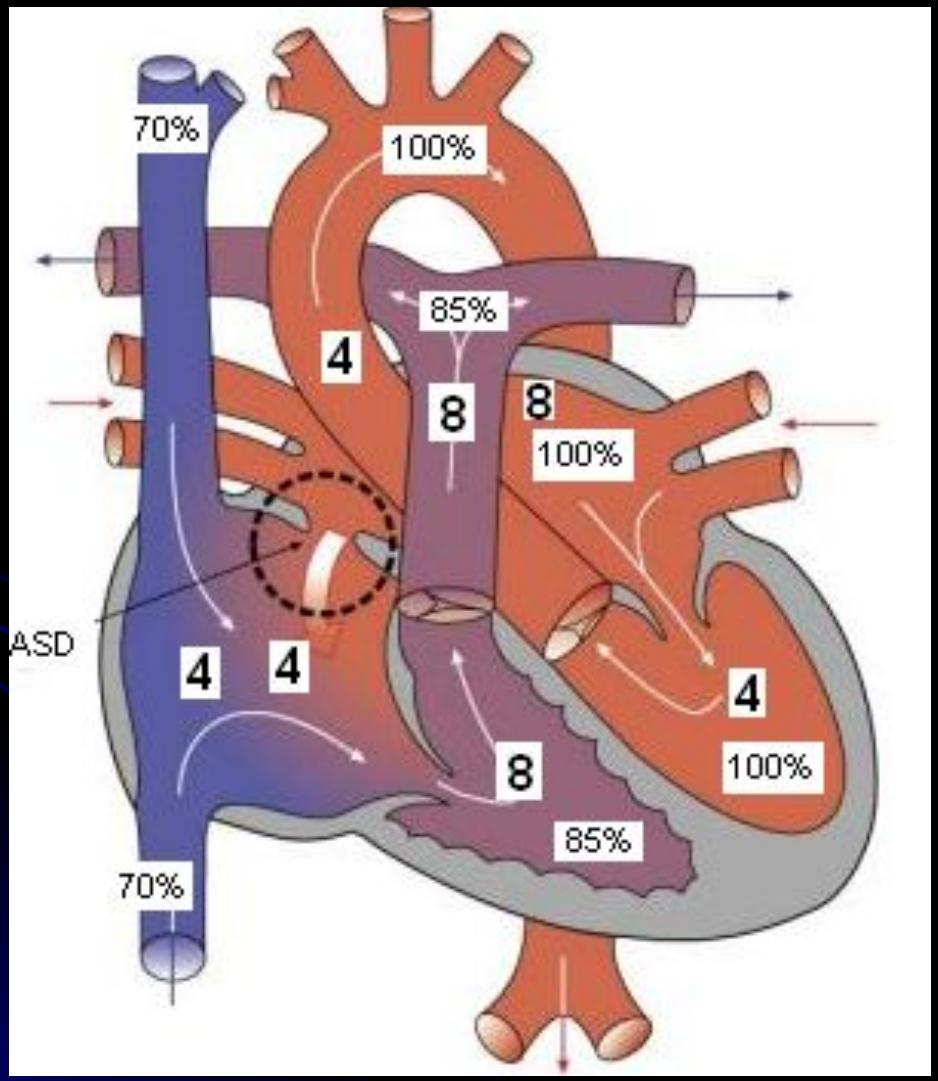
Atrial Septal D

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

Type

- 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 mital 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

Exa

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

Atrial spetal 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.

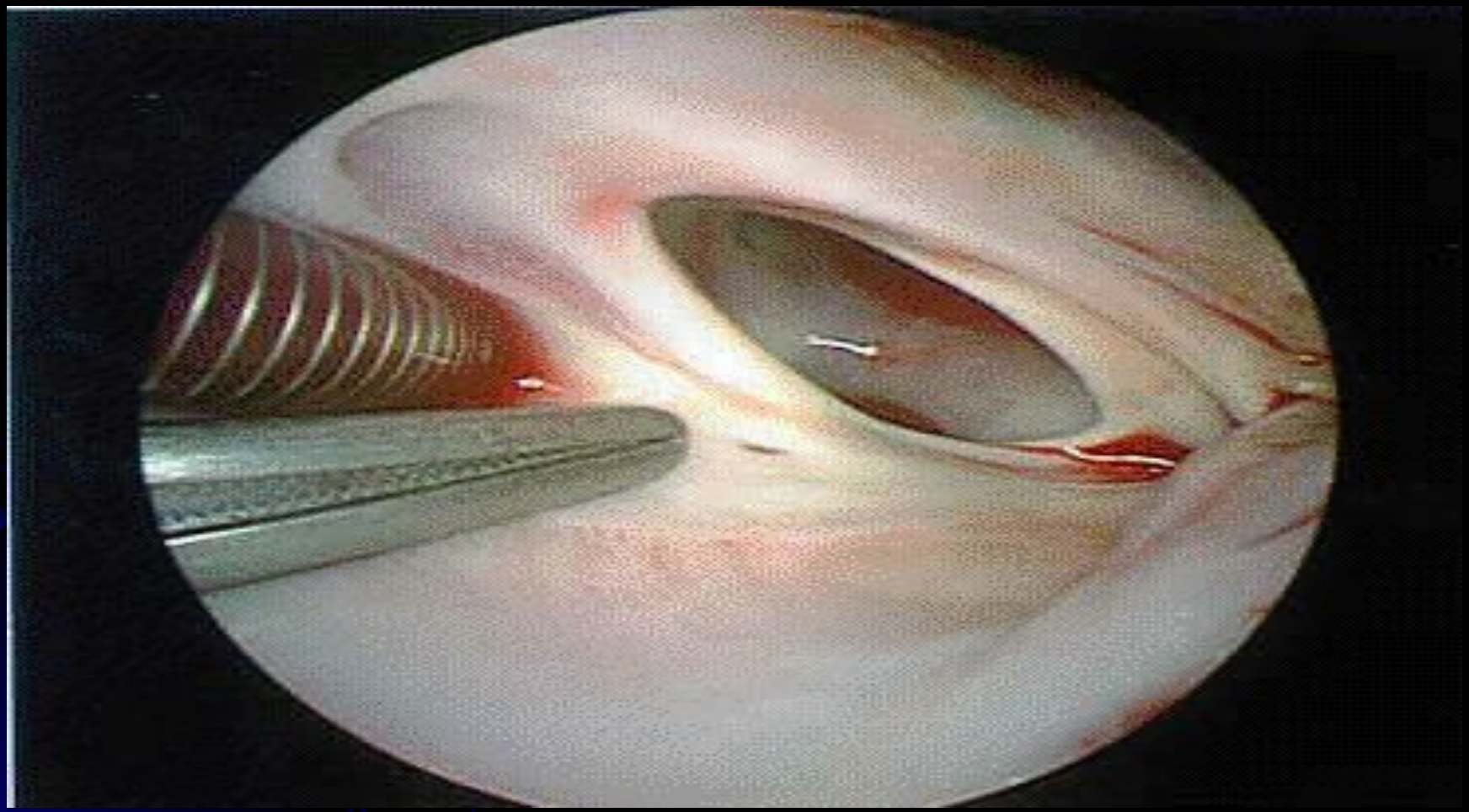
Complications

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

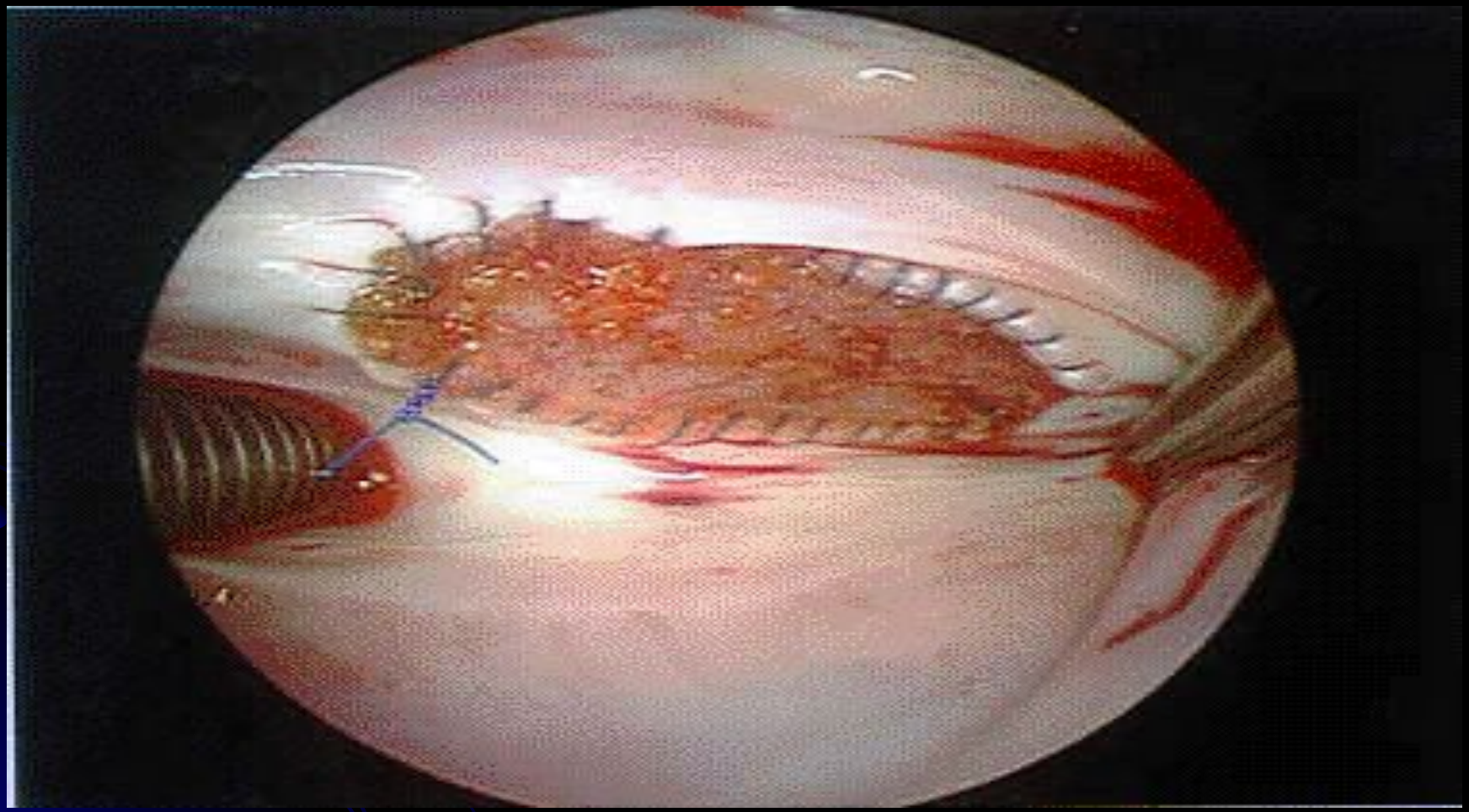
Mana

- 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

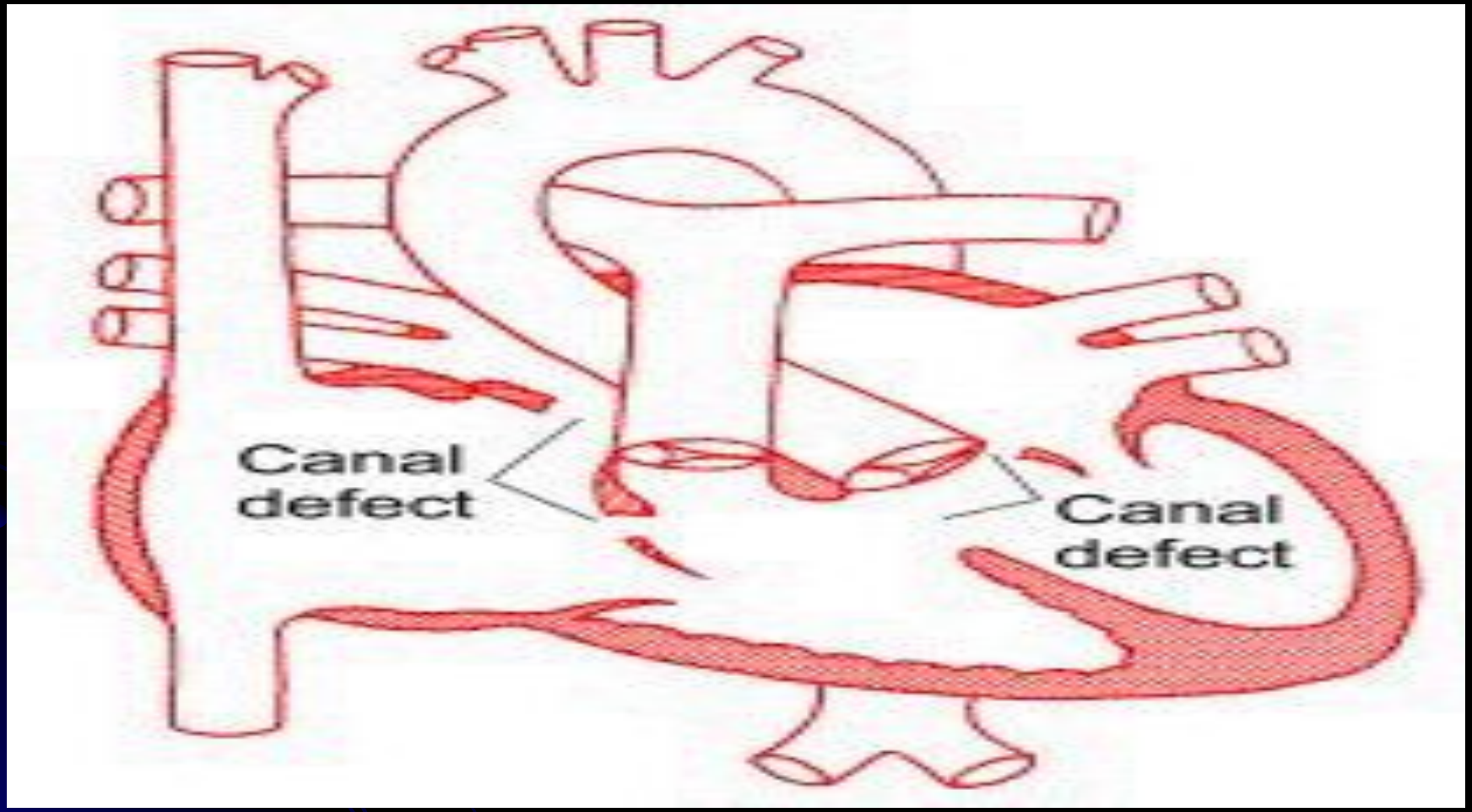
Atrial Septal D



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.

Congenital Heart Disease

- **Patent Ductus Arteriosus (PDA)**

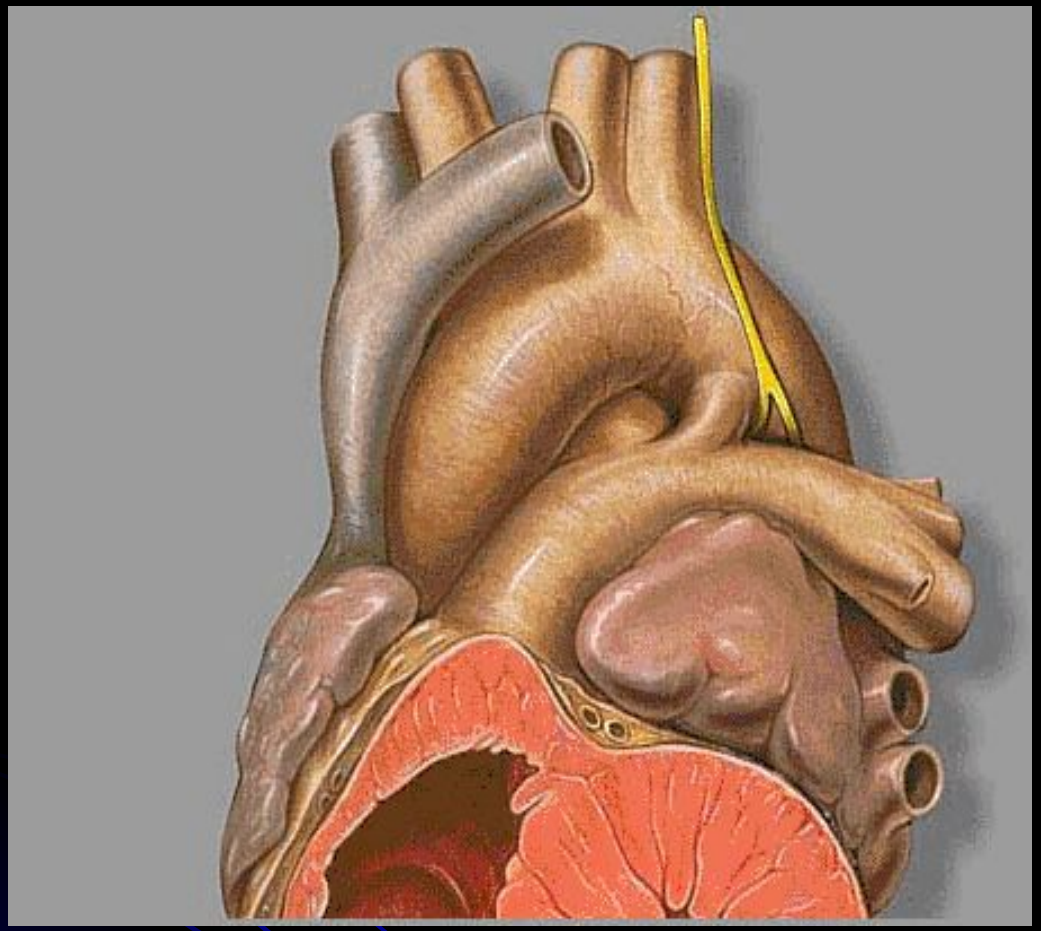
results when the ductus arteriosus fails to close;this leads to blood flow from the aorta to the pulmonary artery

Risk factors

- **More common in girls by2:1**
- **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

- **<u>Associations:</u>** 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 pO2 (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 precodium

Mana

- 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 (righ
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 t
DECREASE Pulmonary Arte
Blood Flow

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

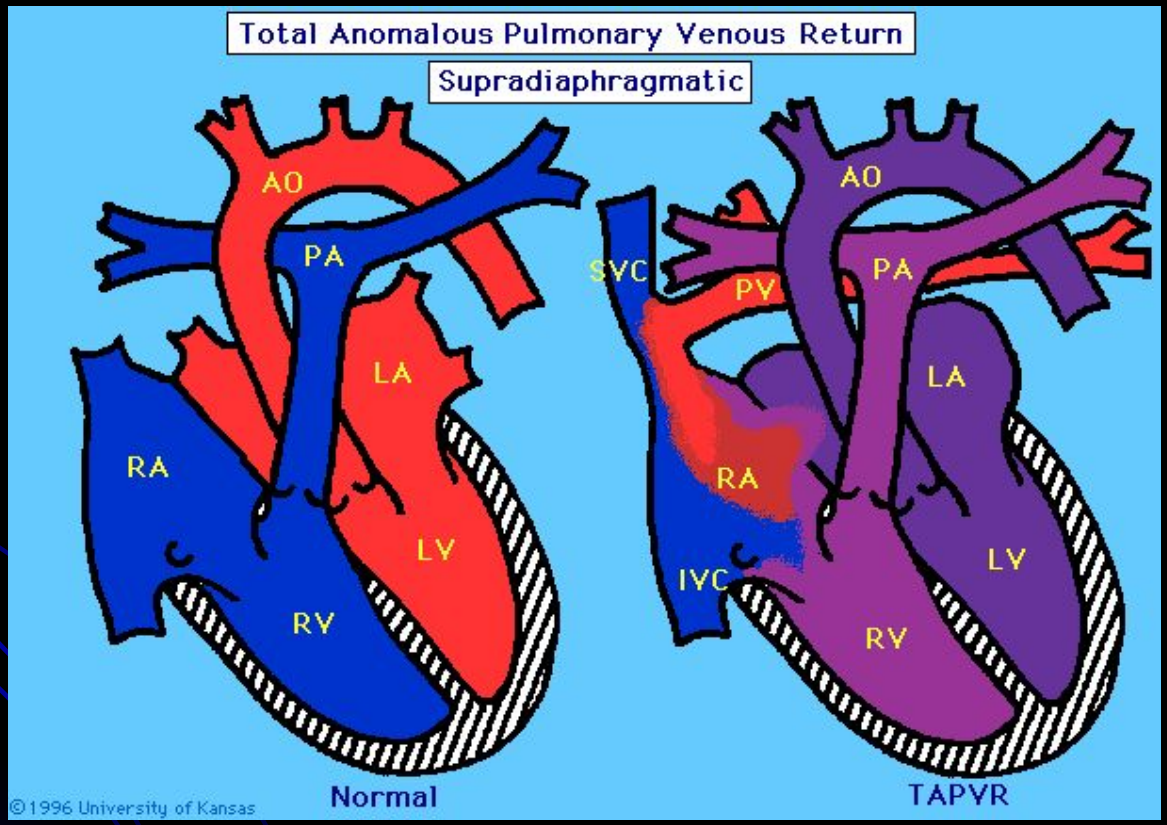
Total Anomalous Pulmona Venous Connection

- 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.

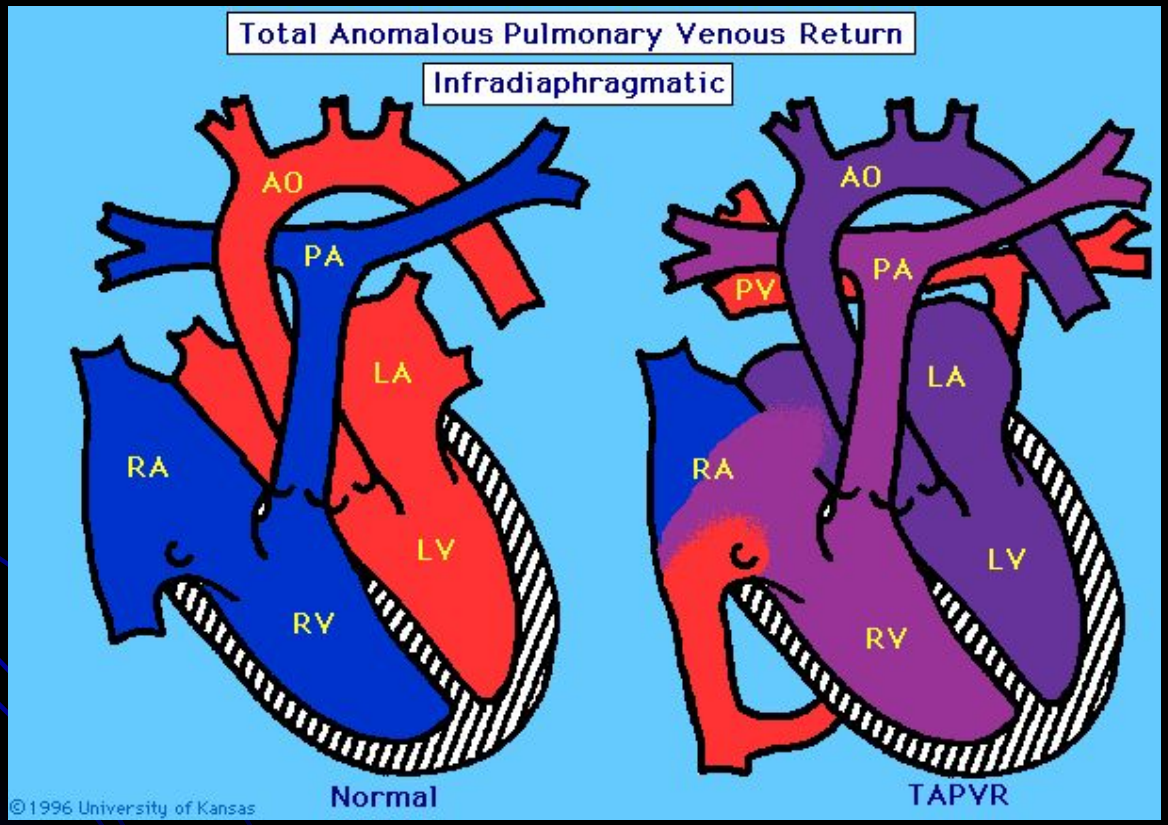
Total Anomalous Pulmona Venous Connection

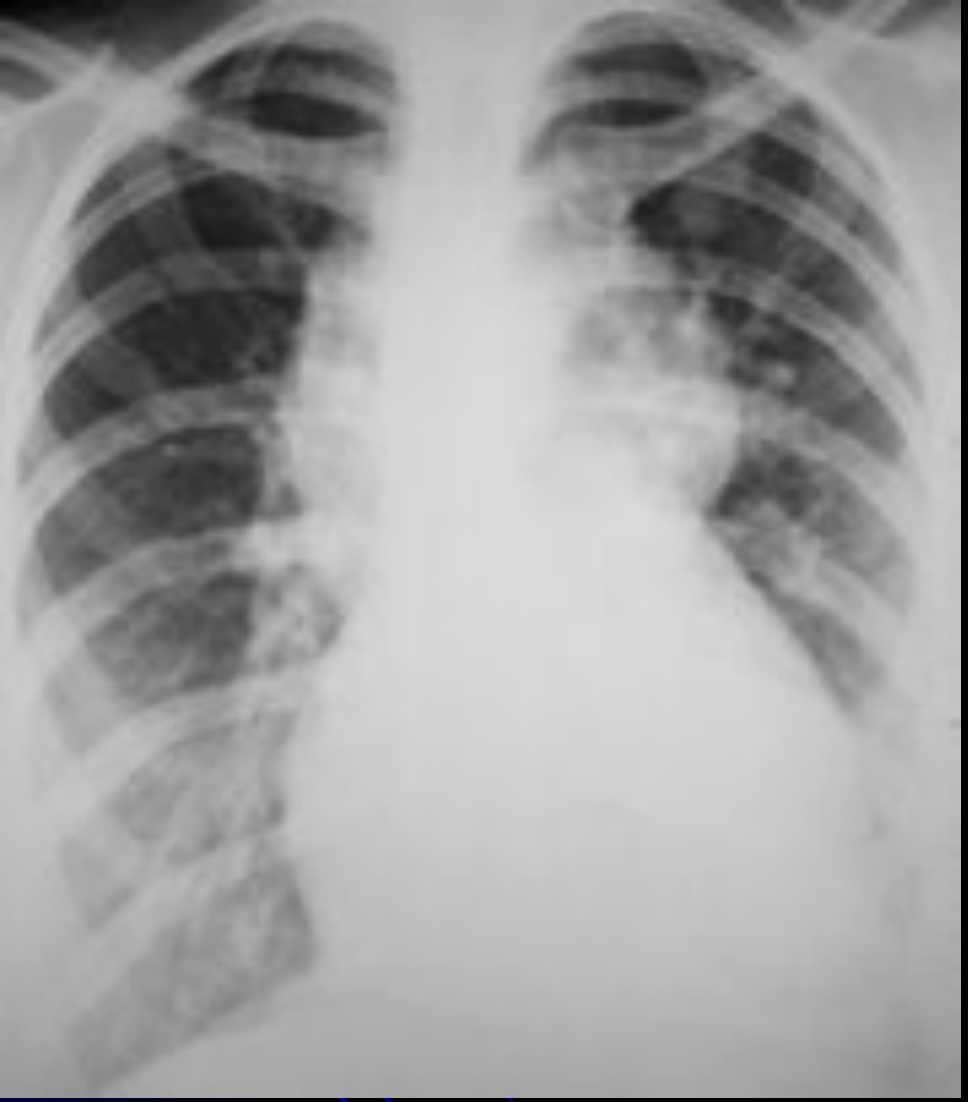
- Diagnosis by cardiac catherization or echocardiography.
- Operative repair in all cases.

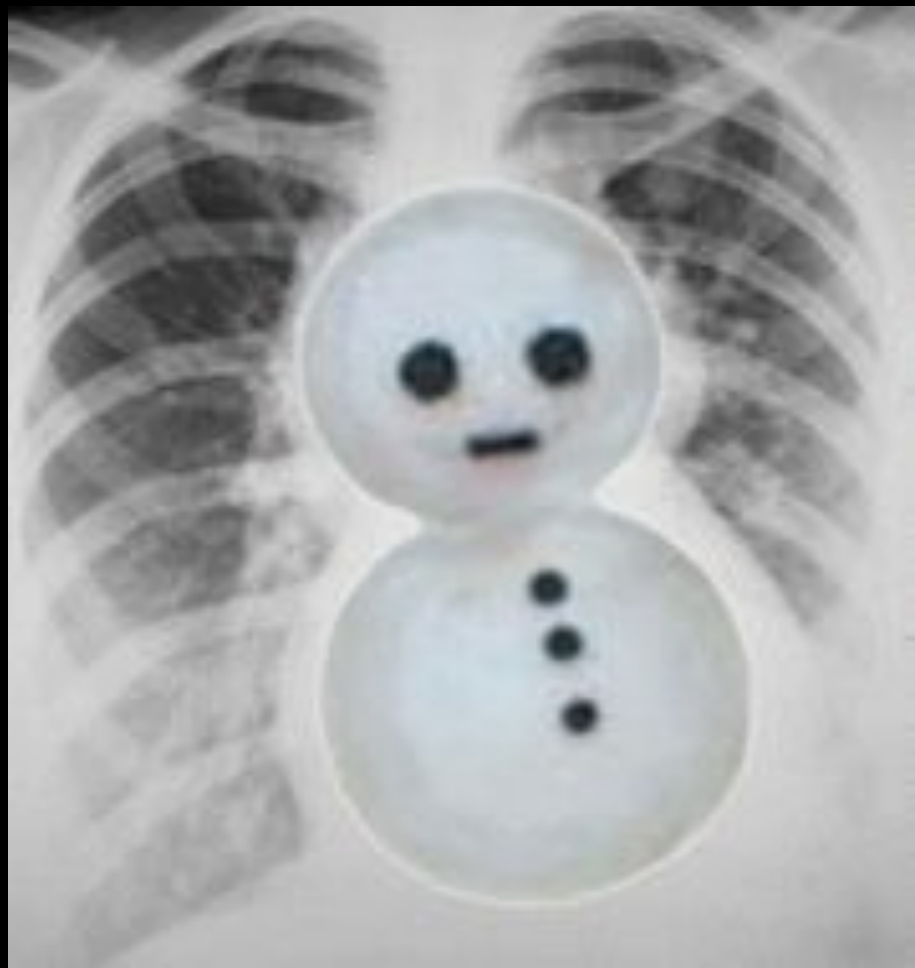
Total Anomalous Pulmonary Venous Return (TAPVR)



TAPVR- Infracar





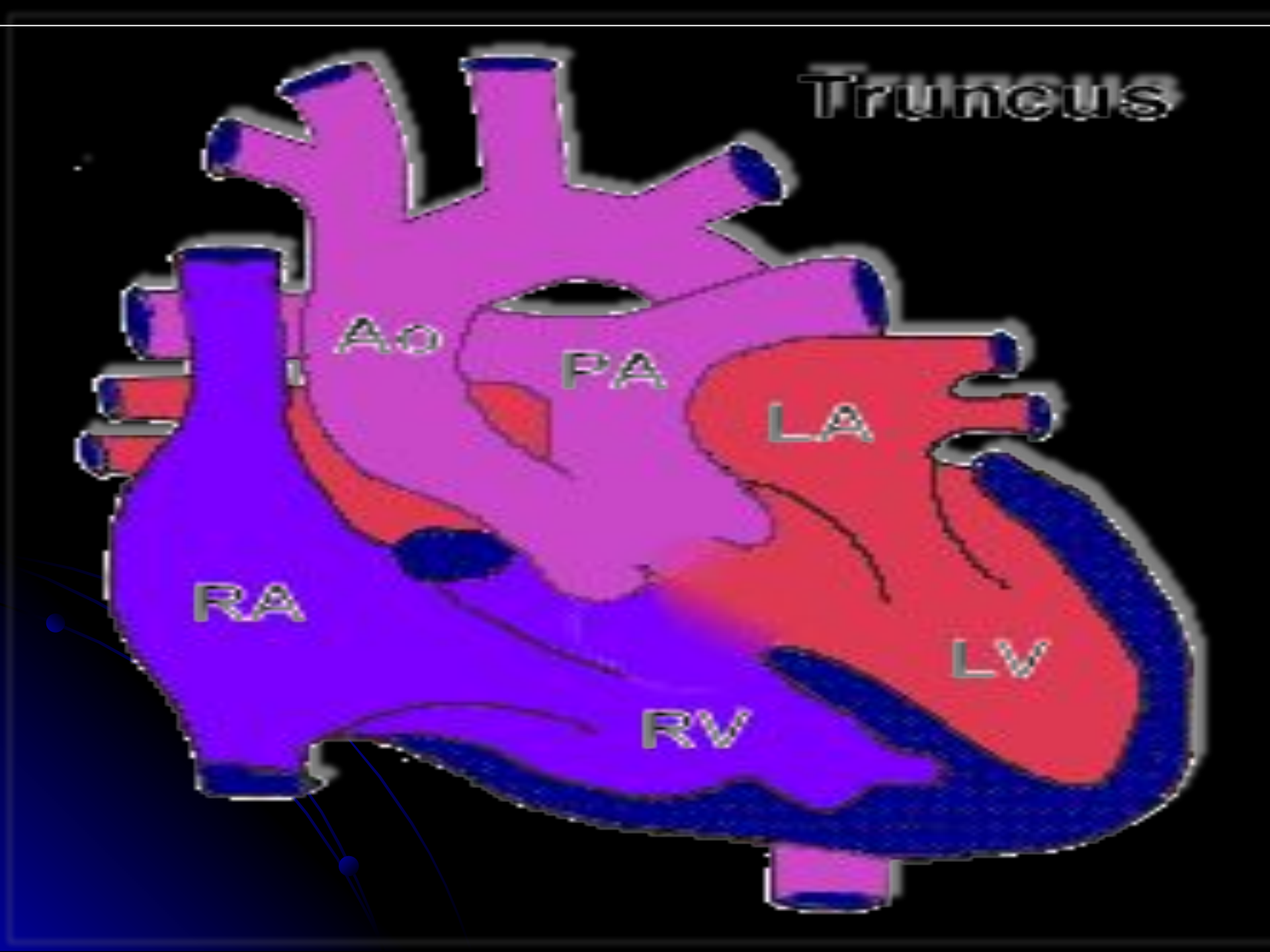


Truncus Arteri

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

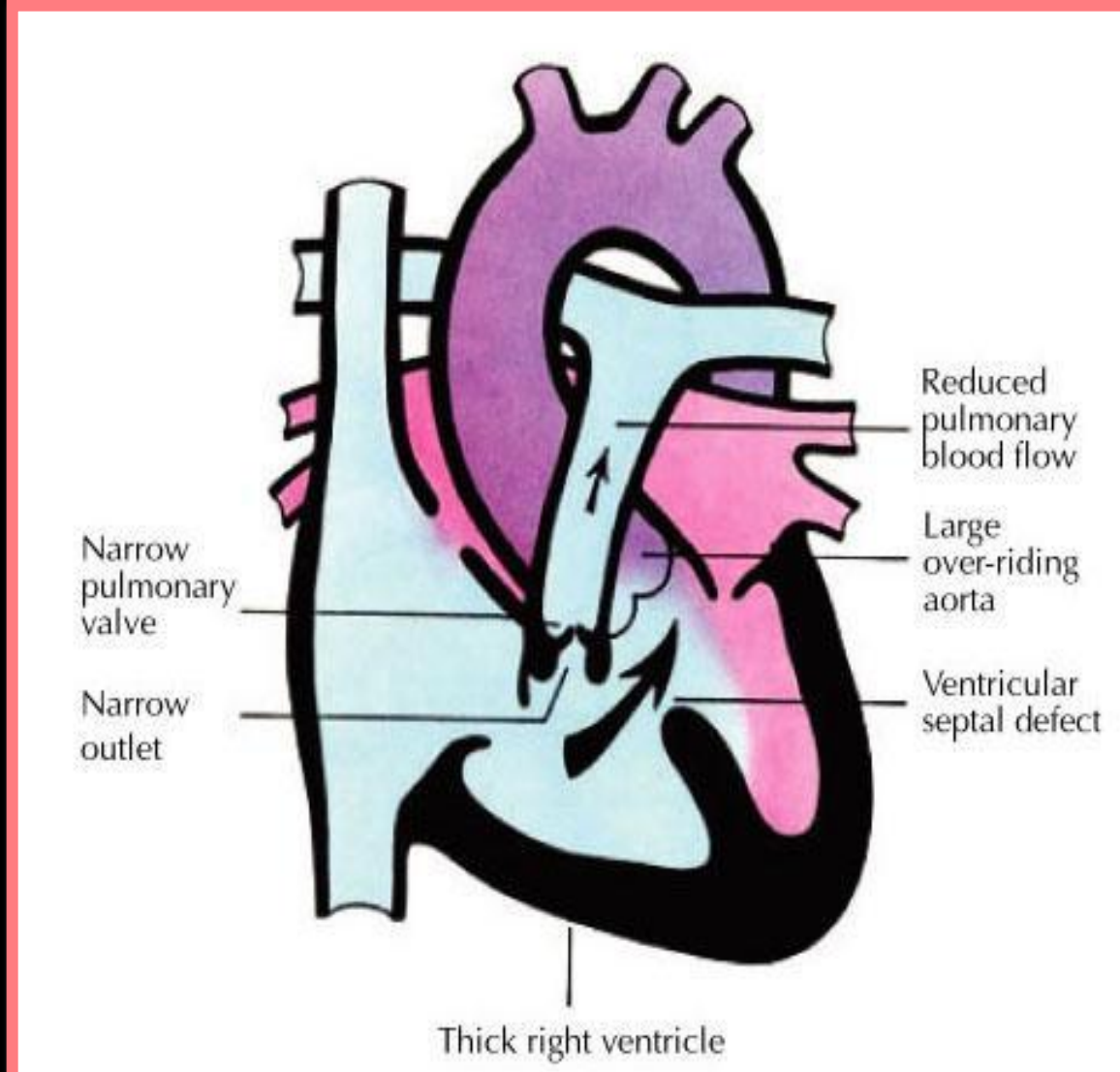
Truncus Arteri

- 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
Ao
PA
LA
RA
LV
RV

Tetralogy of Fallot (TO

- RVOT obstruction
- VSD
- Overriding aorta
- RV hypertrophy



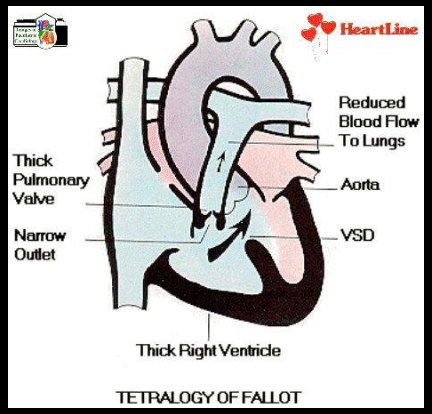
Tetralogy of

- 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)



Morphine

Hypovolemia

Fluid

Syncope

Agitation

Tachycardia

Oxygen

Cyanosis

Impaired RV filling

Knee-chest

Rt Lt shunt

RVOT obstruction

PVR

Phenylepherine

Age

Tetralogy of Fallot

- Diagnosis

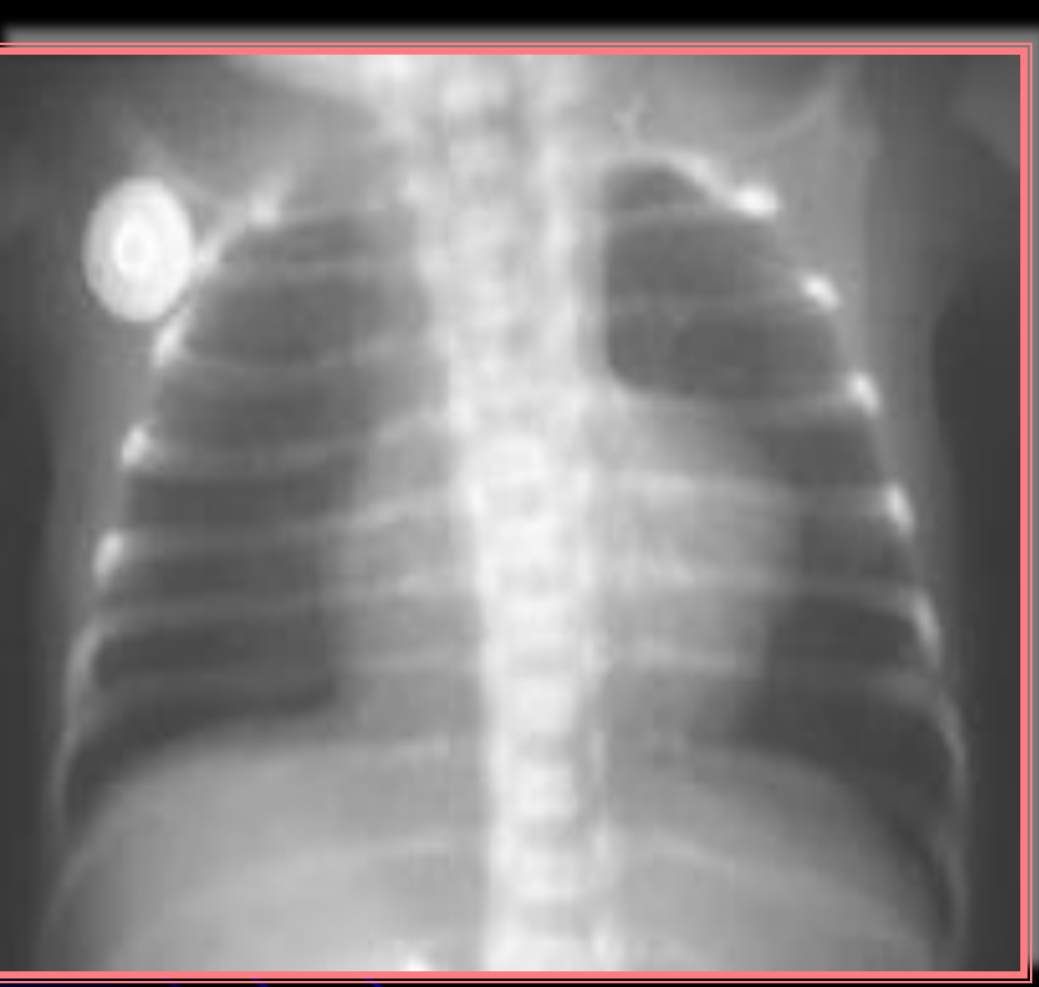
Chest X-ray: Boot-shaped heart, dark lung fields

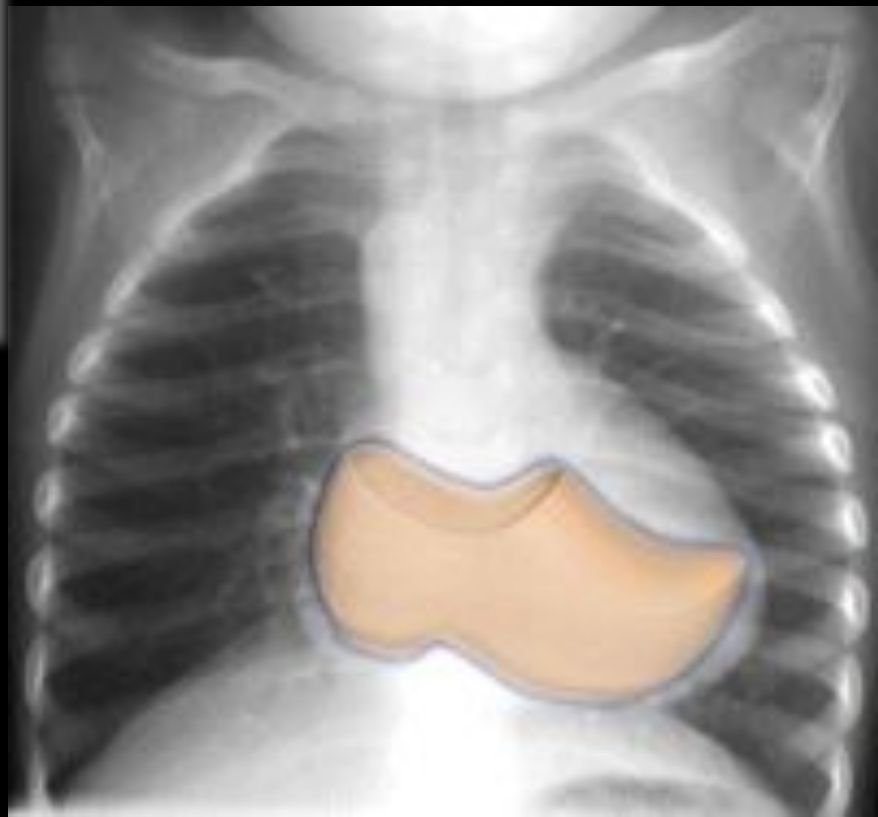
ECG: right axis deviation, RVH

Echocardiogram (gold standard)

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

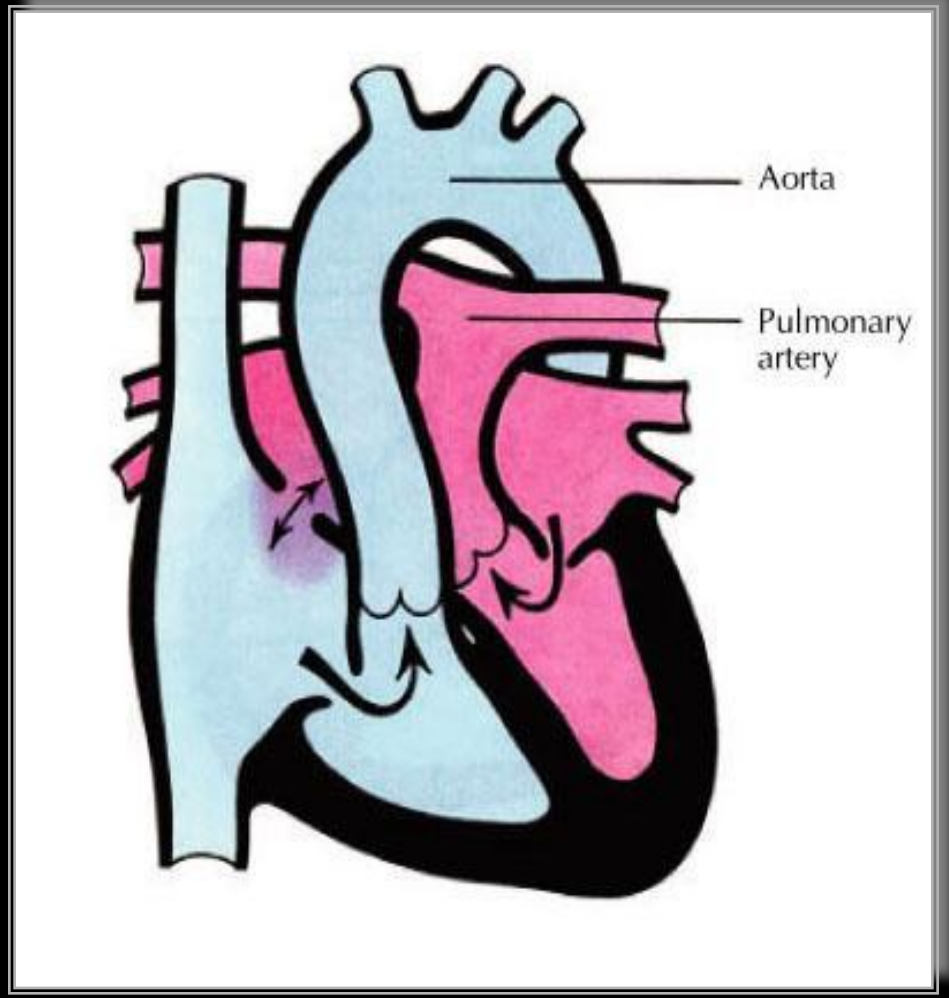
management: surgical repair including closure of VSD and widening of RVOTO





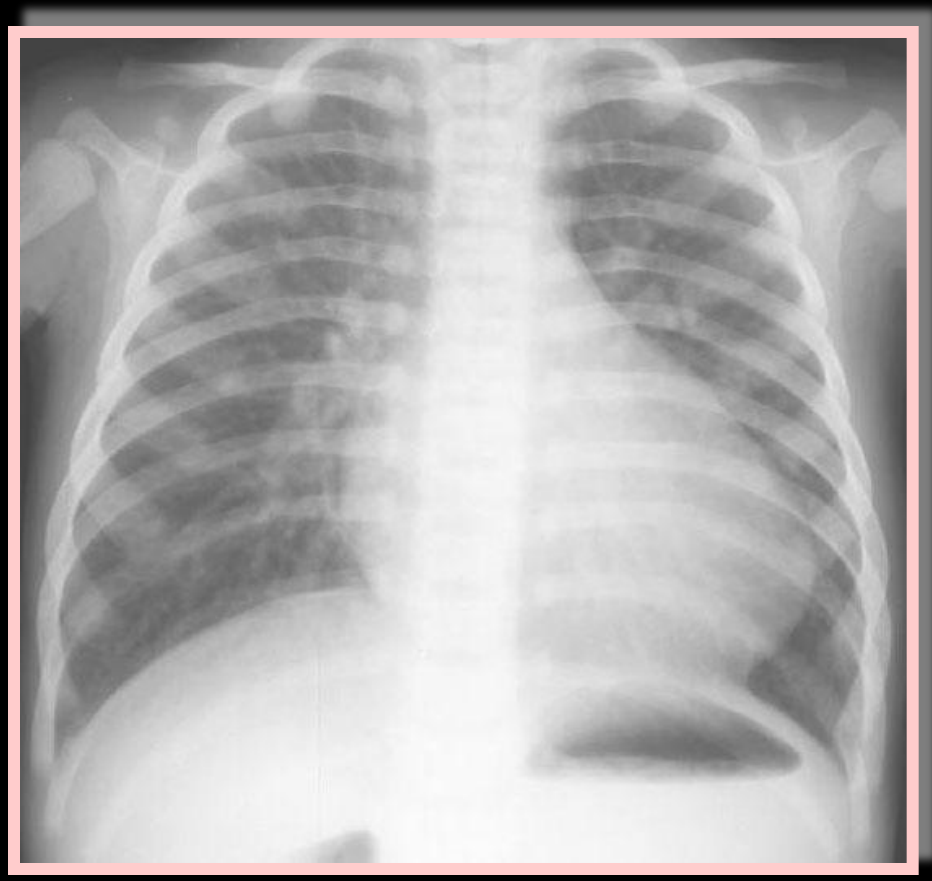
Transposition of Great (TGA)

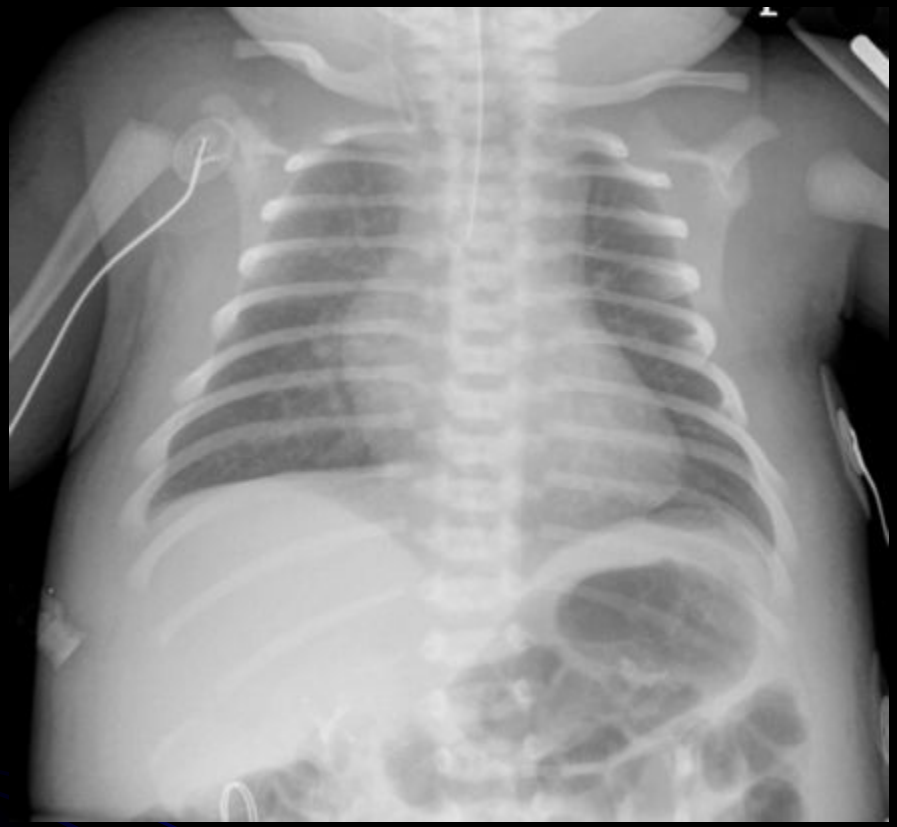
- Aorta originating from the right ventricle, and pulmonary artery originating from the left ventricle
- Accounts for 5-7% of all congenital heart disease

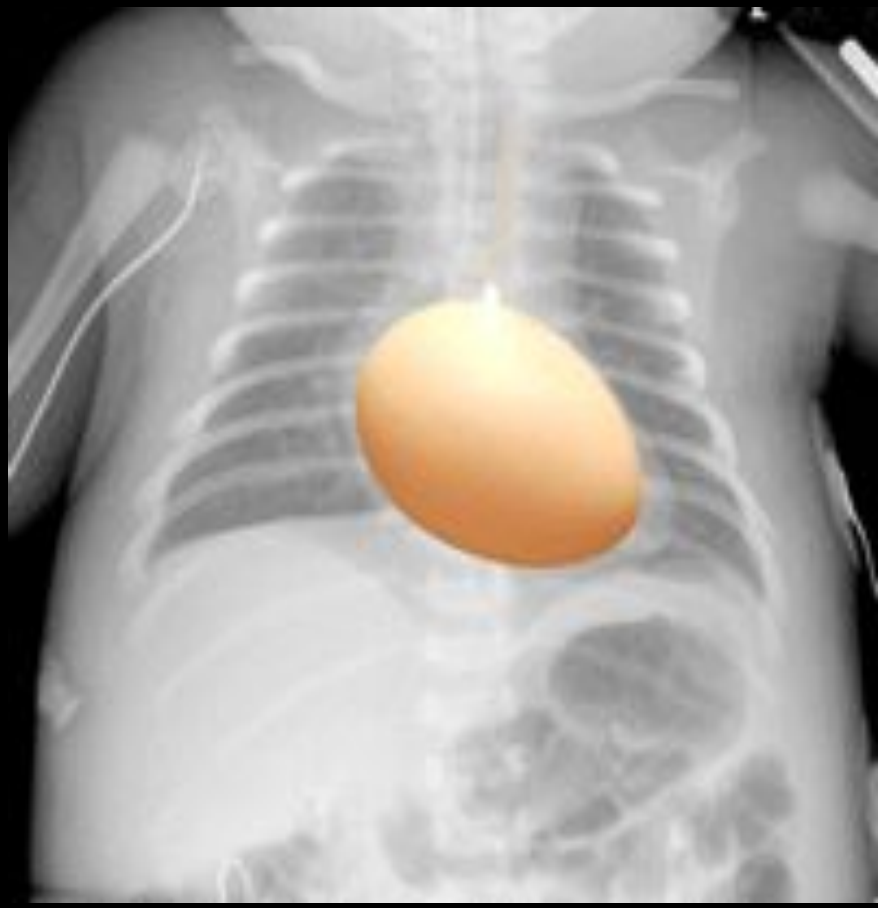


- Survival is dependent on the presence of mixing between the pulmonary and systemic circulation
- Atrial septal defect is essential for survival
- 50% of patients have a VSD
- Usually presents in the first day of life with profound cyanosis
- More common in boys

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







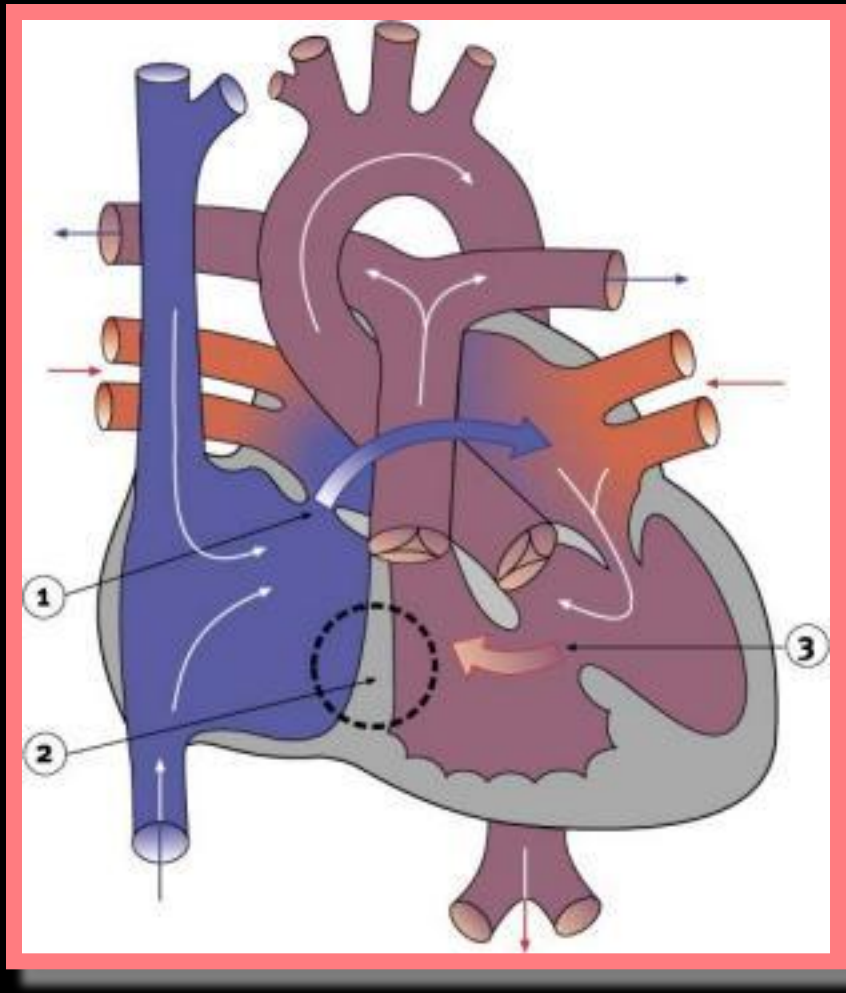
Transposition of the Great Arteries (TGA)

- **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

Trcuspid A

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



Mana

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

No O2
Afterload reduction
Diuretics

Tricuspid A

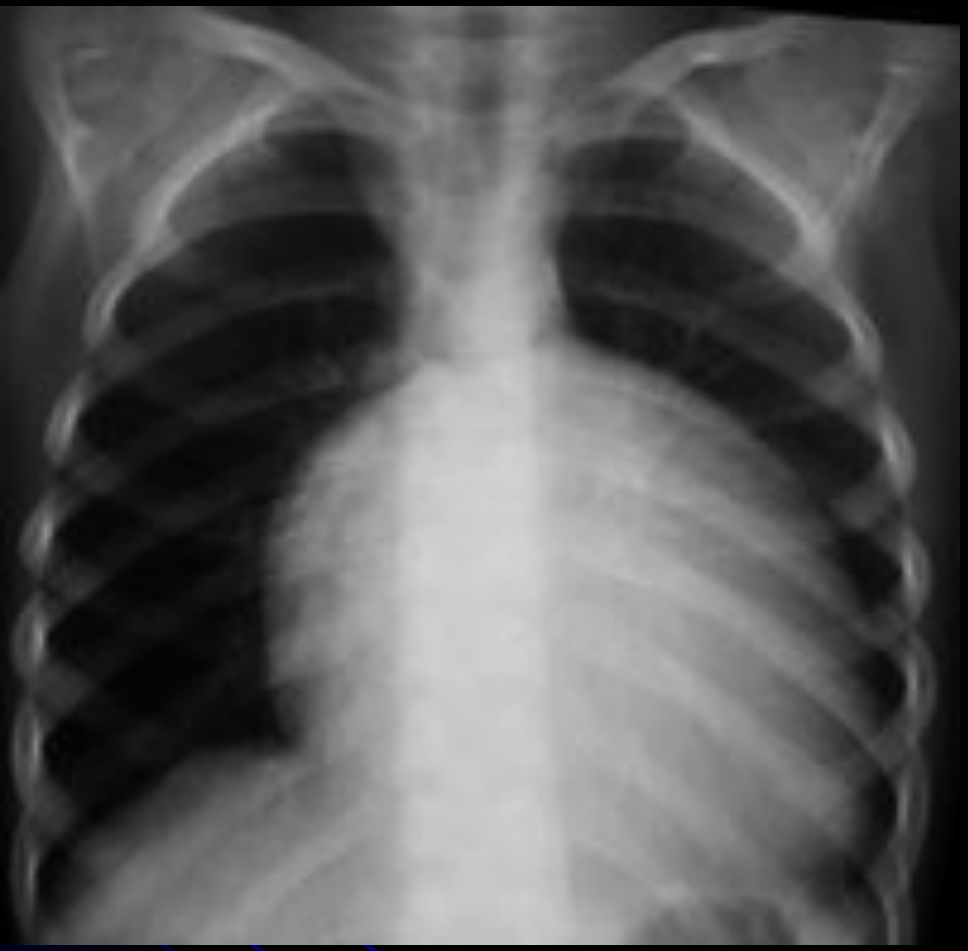
- Repair consists of shunt from right atrium to pulmonary artery or rudimentary right ventricle (Fontan procedure).

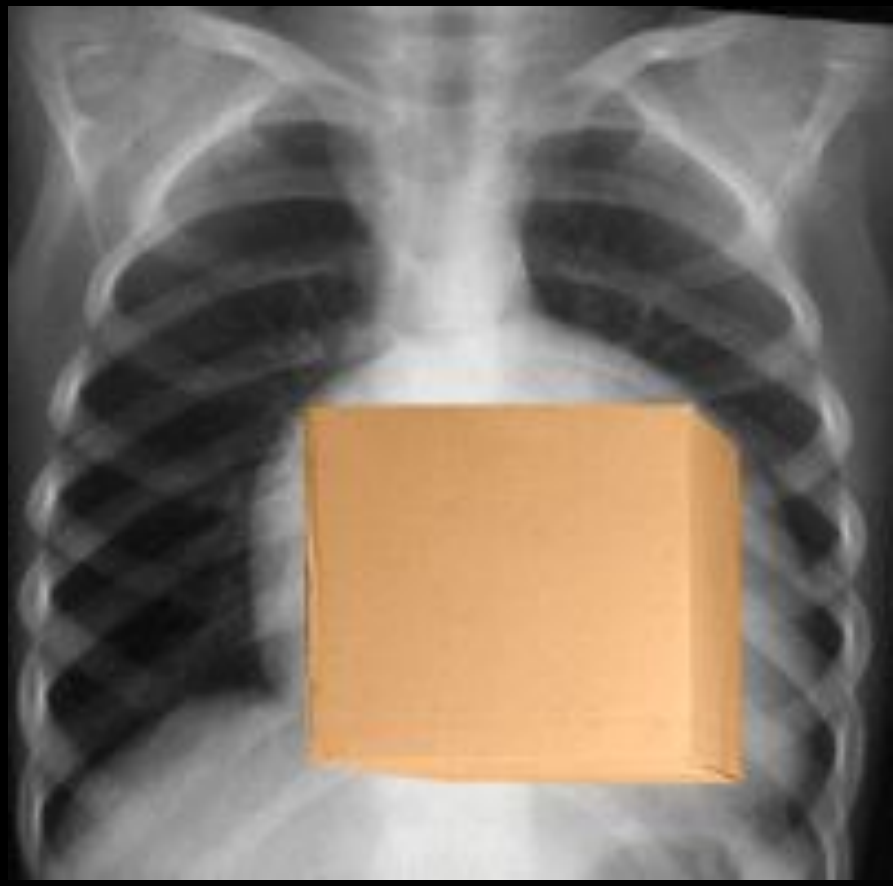
Ebstein's Ano

- 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 Ano

- 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

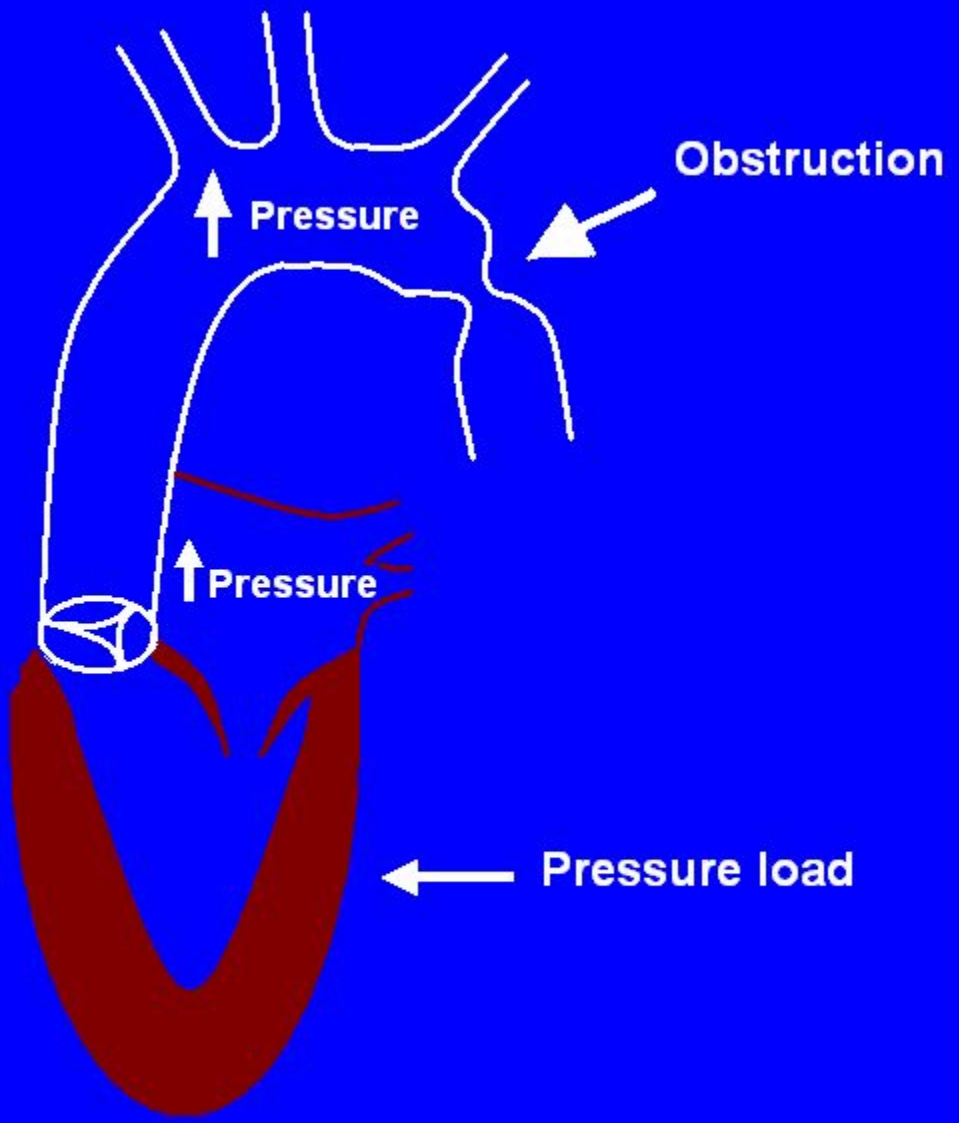
- Absent or weak femoral pulses.
- Systolic pressure higher in upper extremities than in lower extremities; diastolic pressures are similar.
- Harsh systolic murmur heard in the back.

Hypertension
Pressure gradient

Left ventricular
Hypertrophy

Heart Failure
Pulmonary Edema

Pulmonary
Hypertension



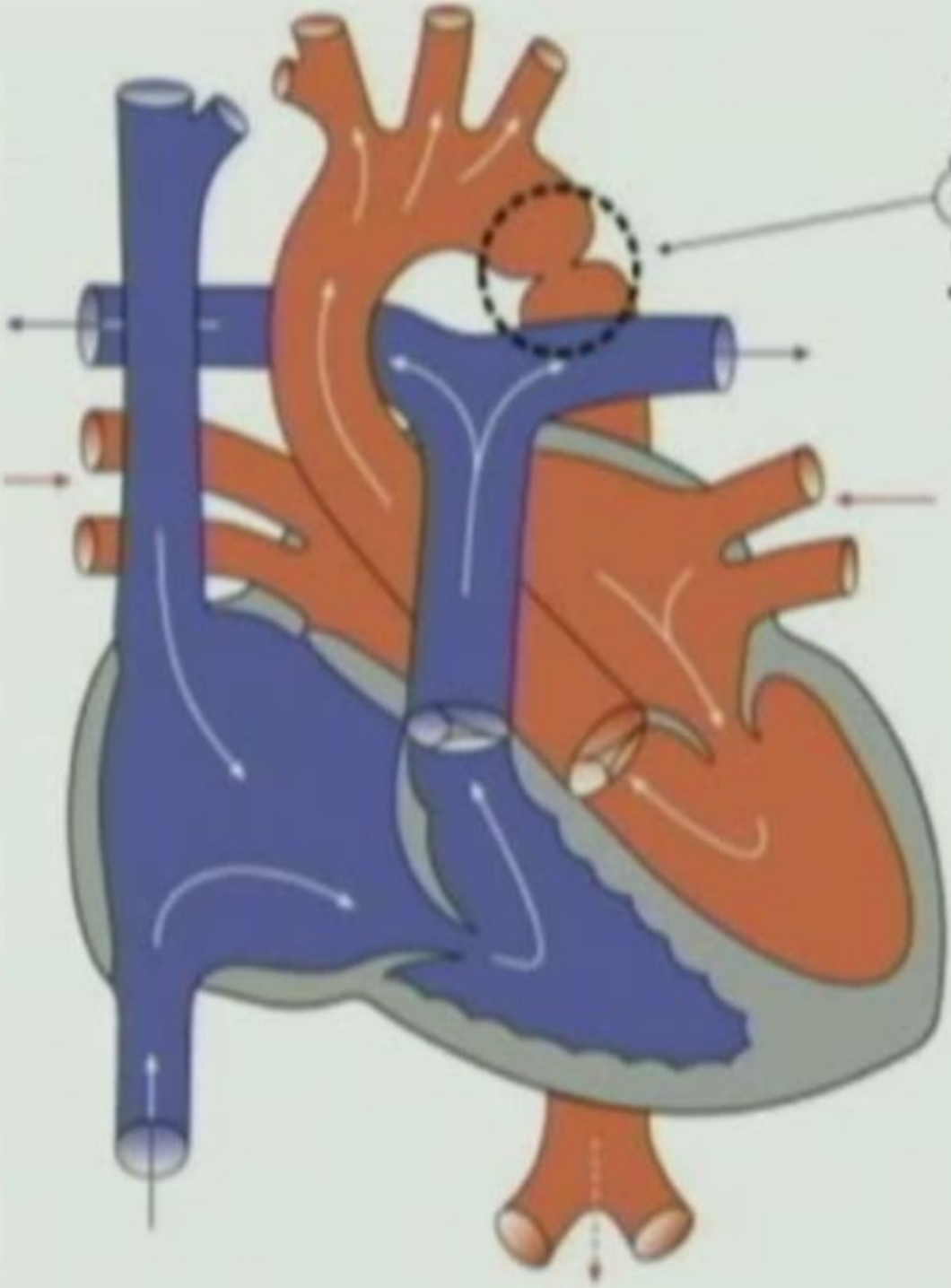
Coarctation of the Aorta

- 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 subclavcian artery



Coarctation of the Aorta

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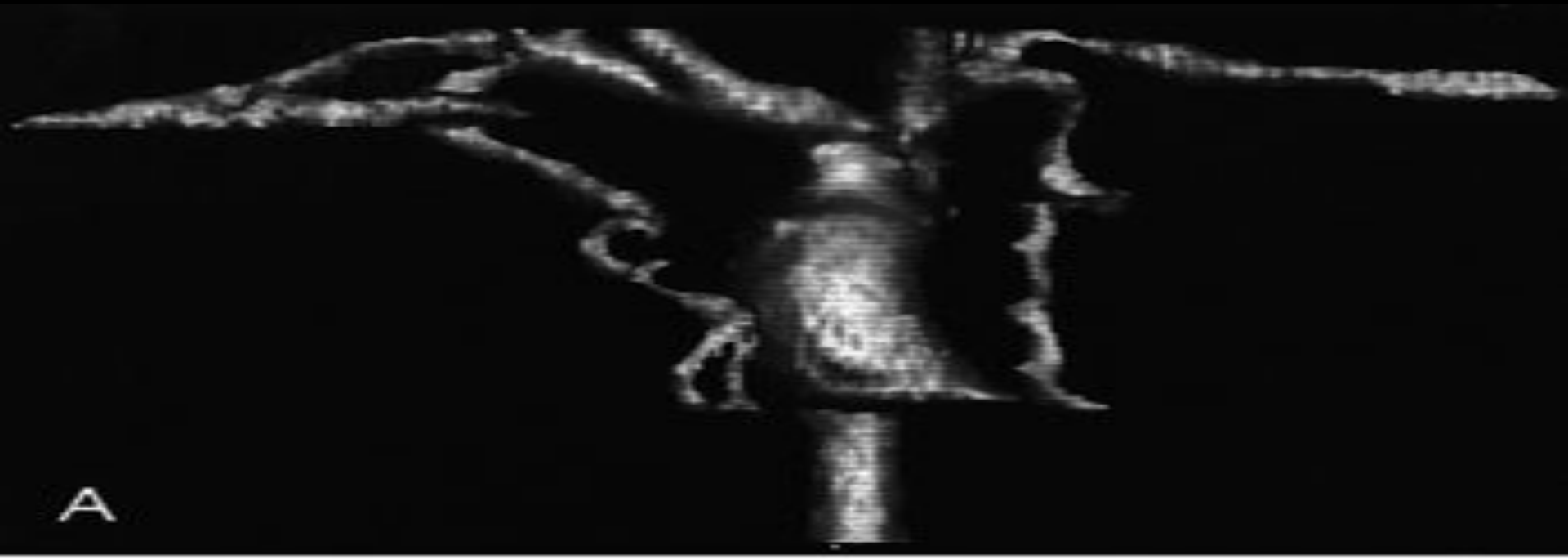
KAPLAN MEDICAL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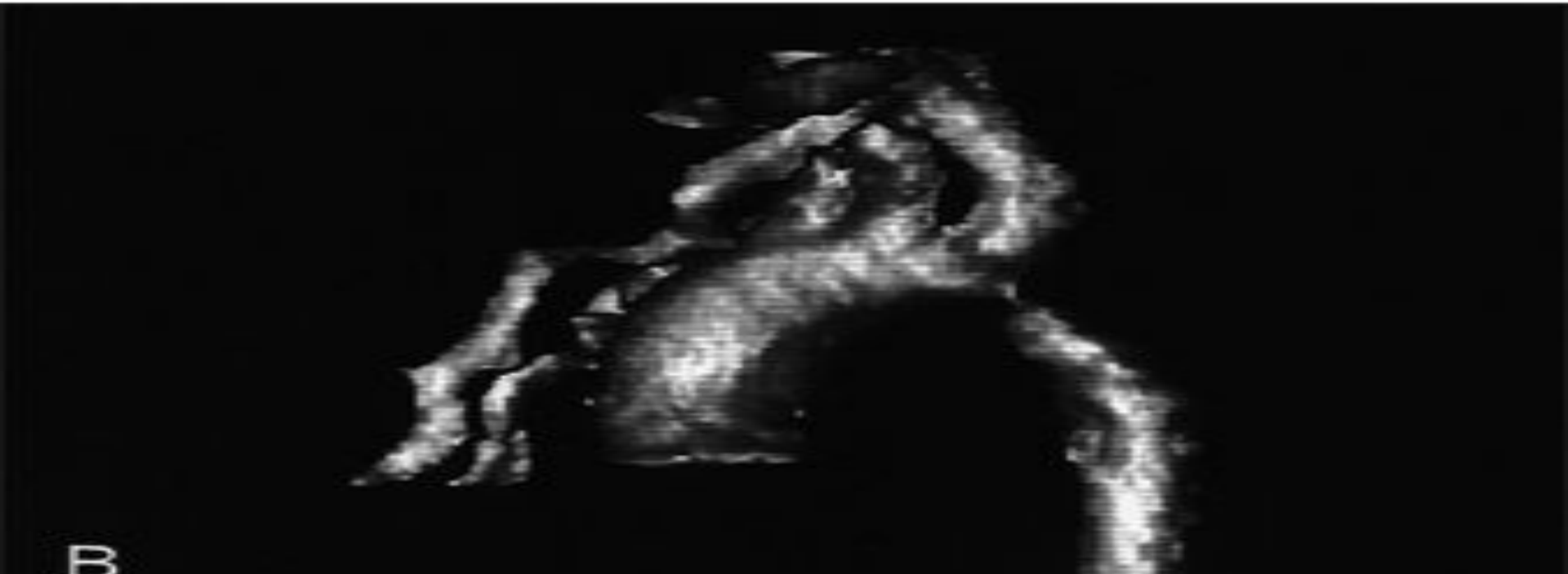

B

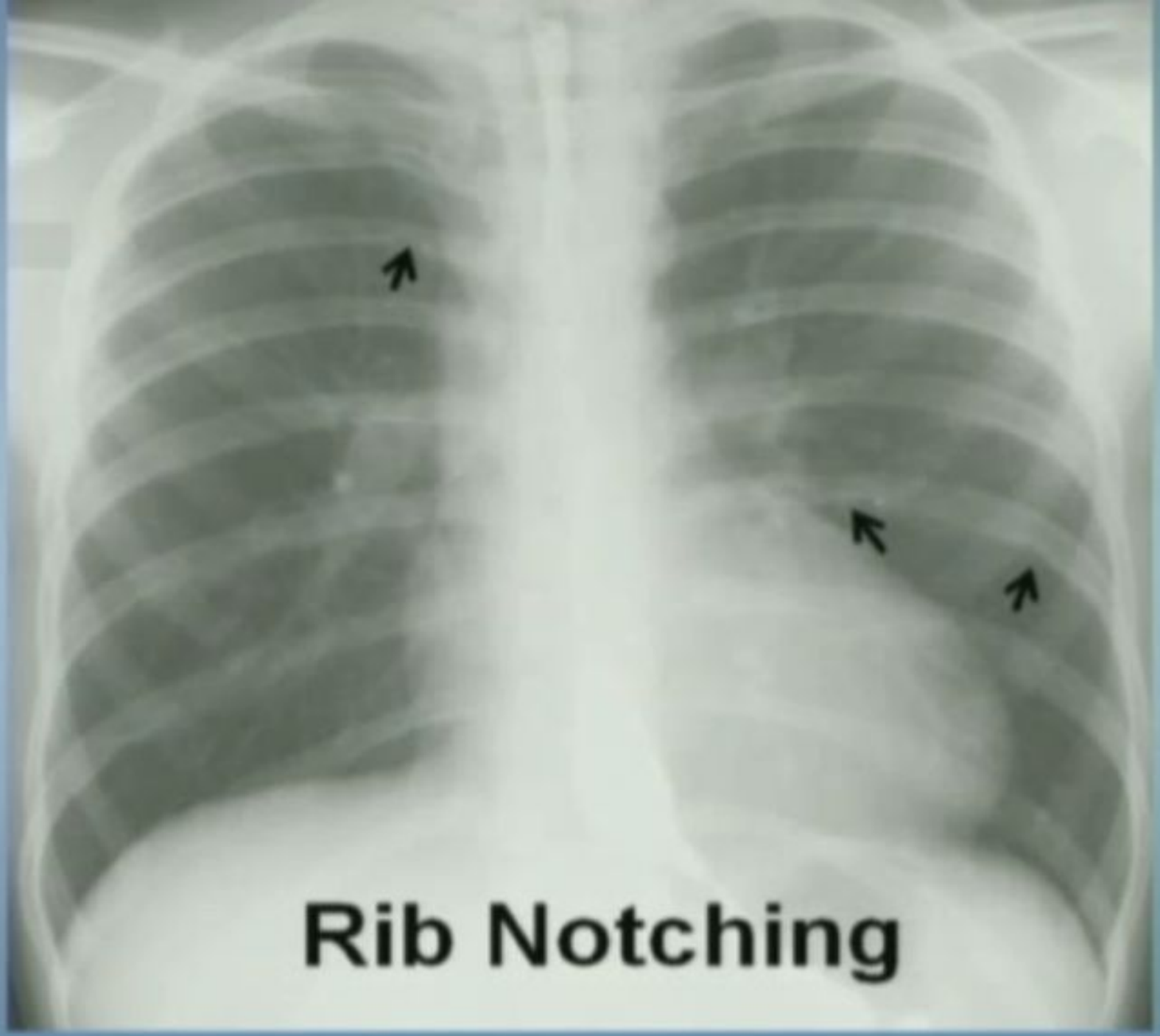
Coarctation of the Aorta

- 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 :

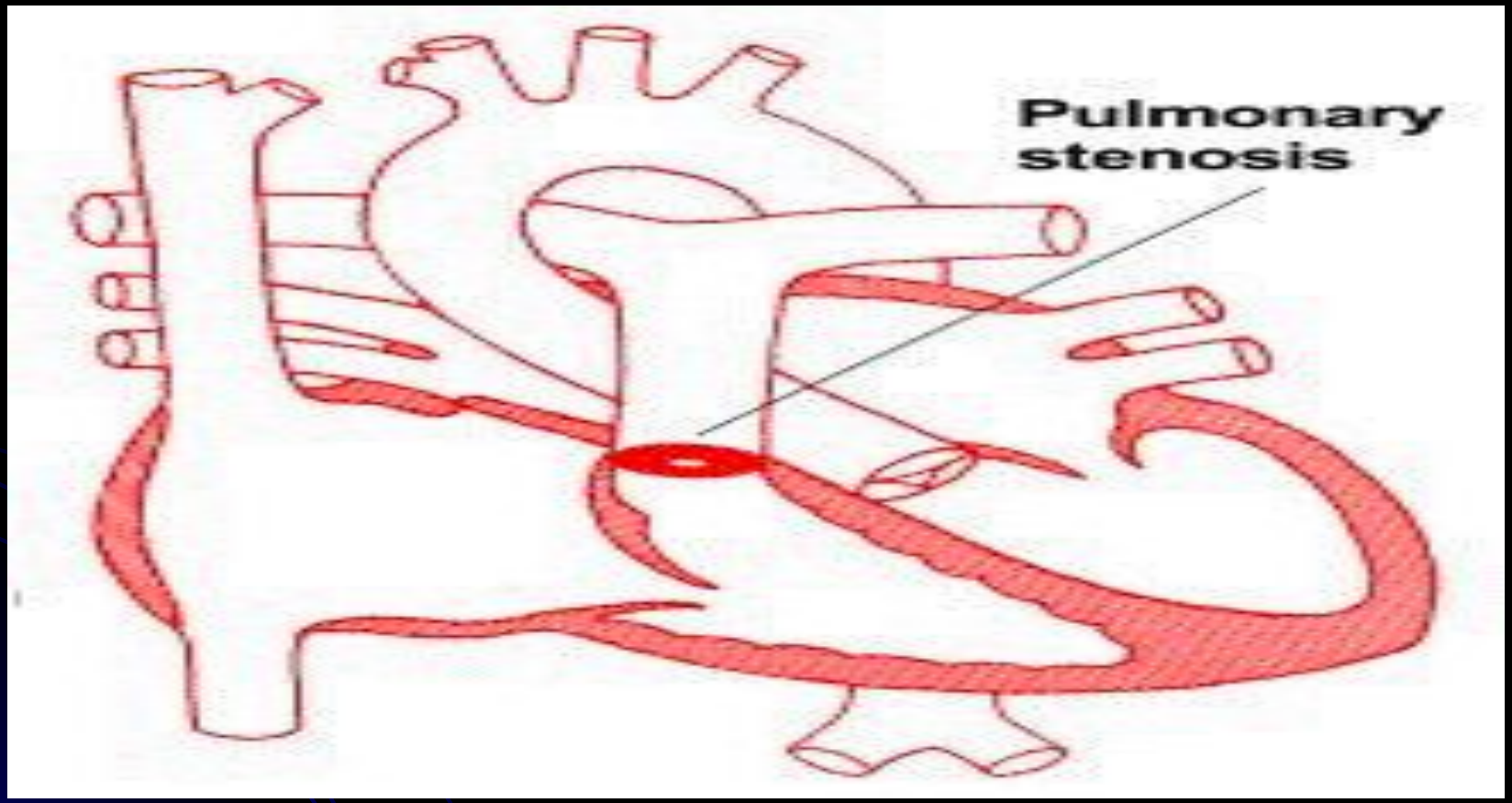
- PGE1
- balloon arterioplasty or surgical correction after stabilization

Older : treat hypertension then surgery

Complications: essential hypertension , aortic aneurysms

endocarditis

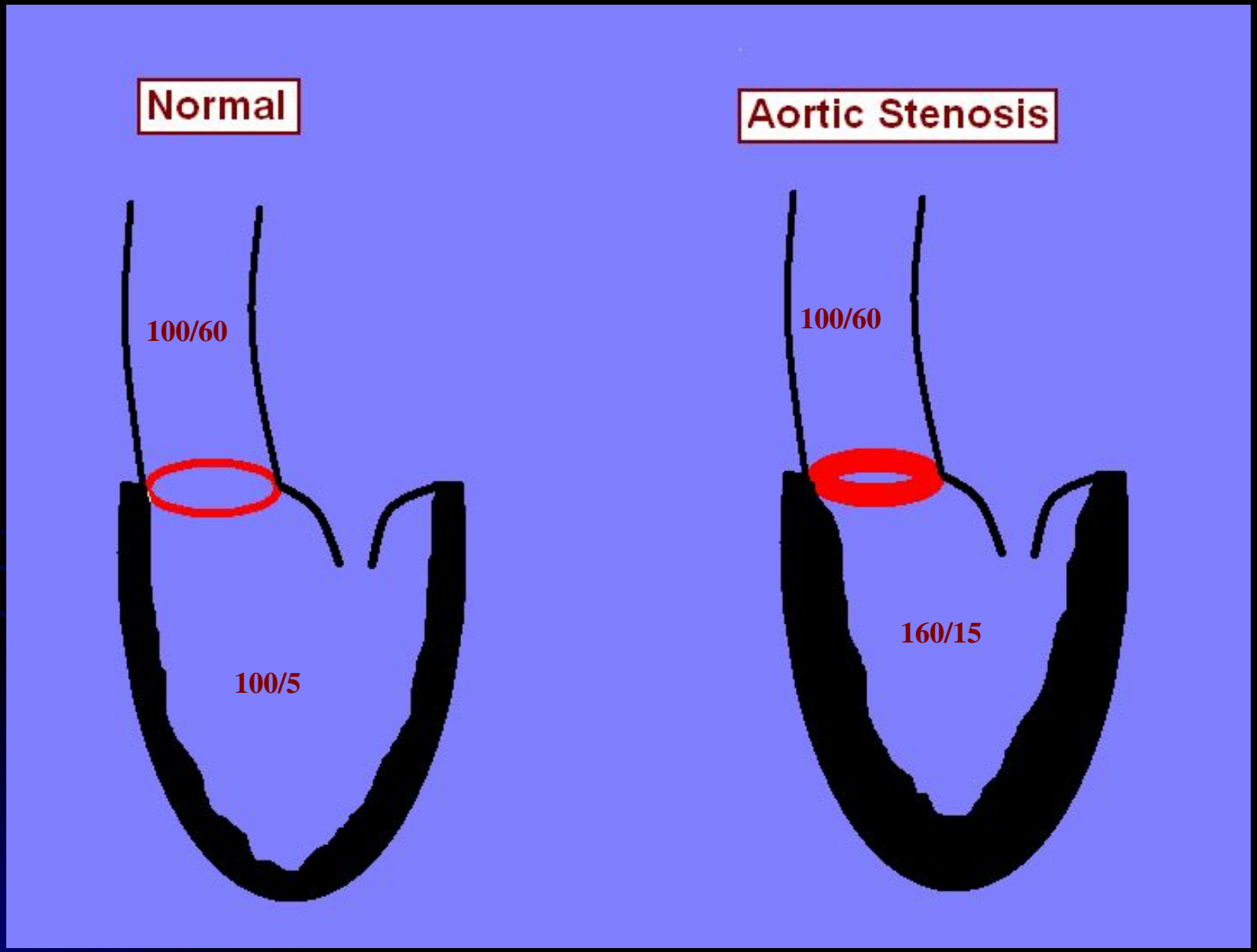
Pulmonary Sten



Pulmonary Sten

- 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

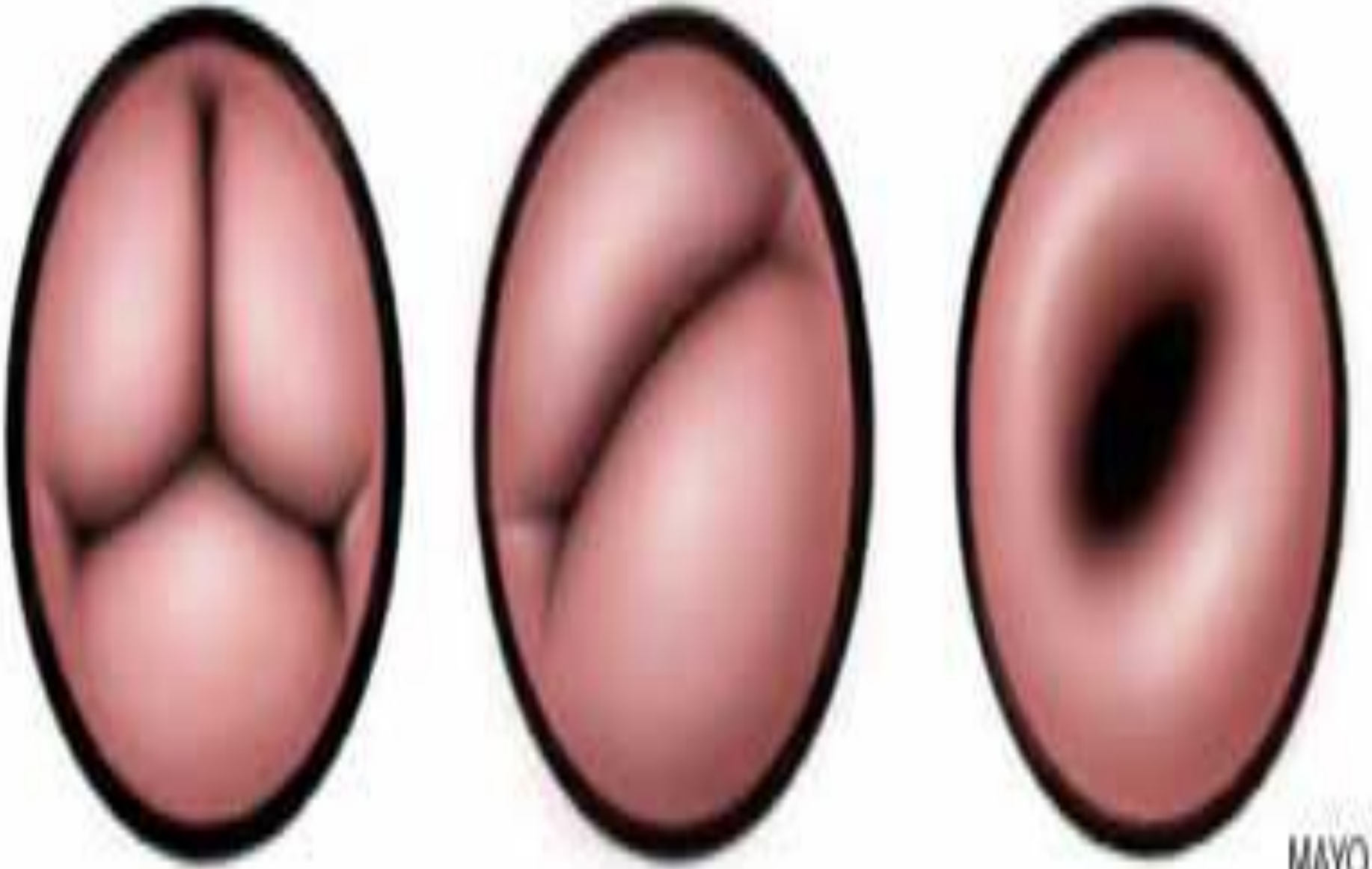


Aortic St

- 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.


MAYO
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Valvular Aortic Steno

- 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 >> periphral 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
- Kartagner 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 failu

- 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 comm

- 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