

Congenital heart diseases

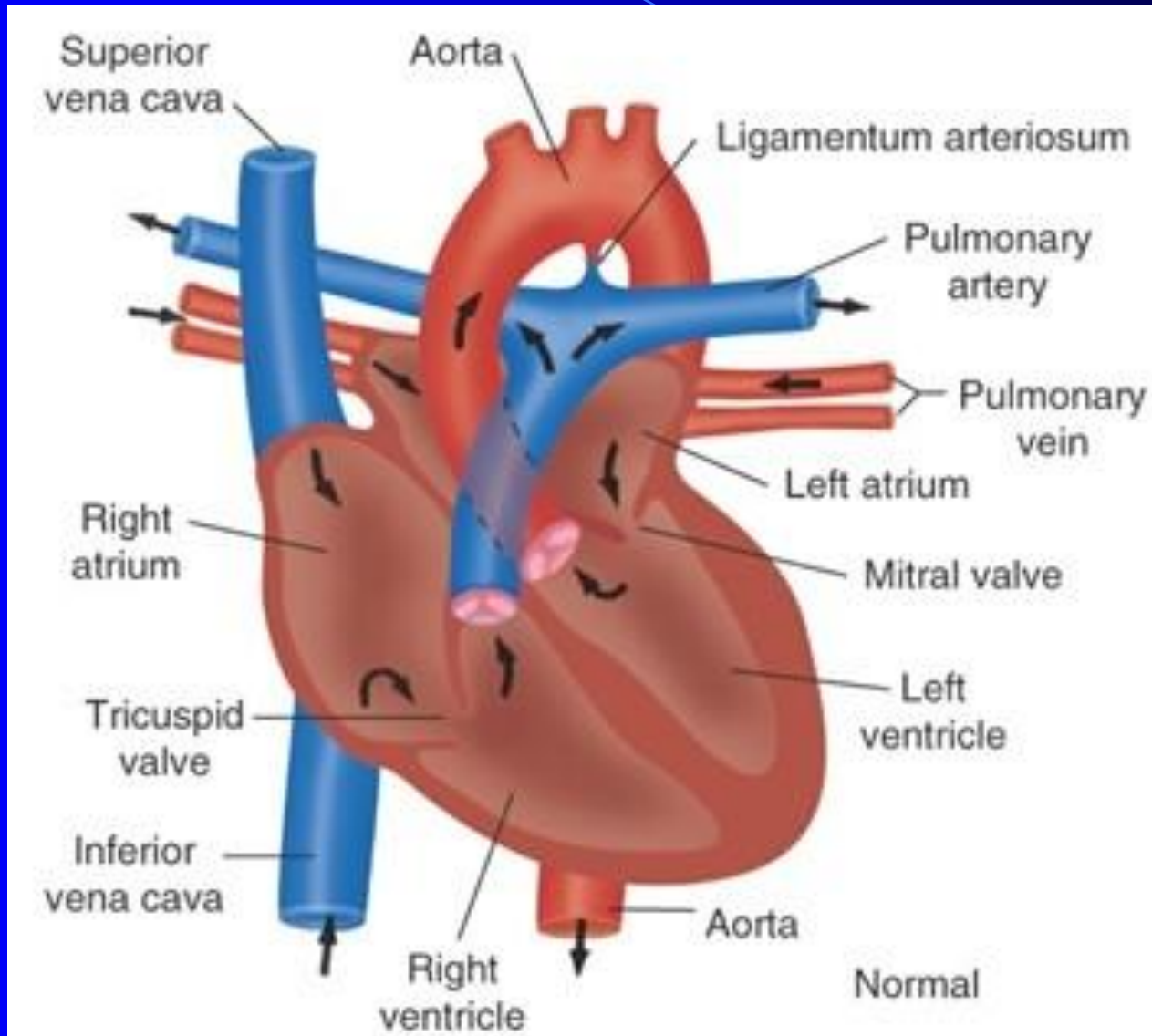
Dr Zuriel – UON- Human Pathology.

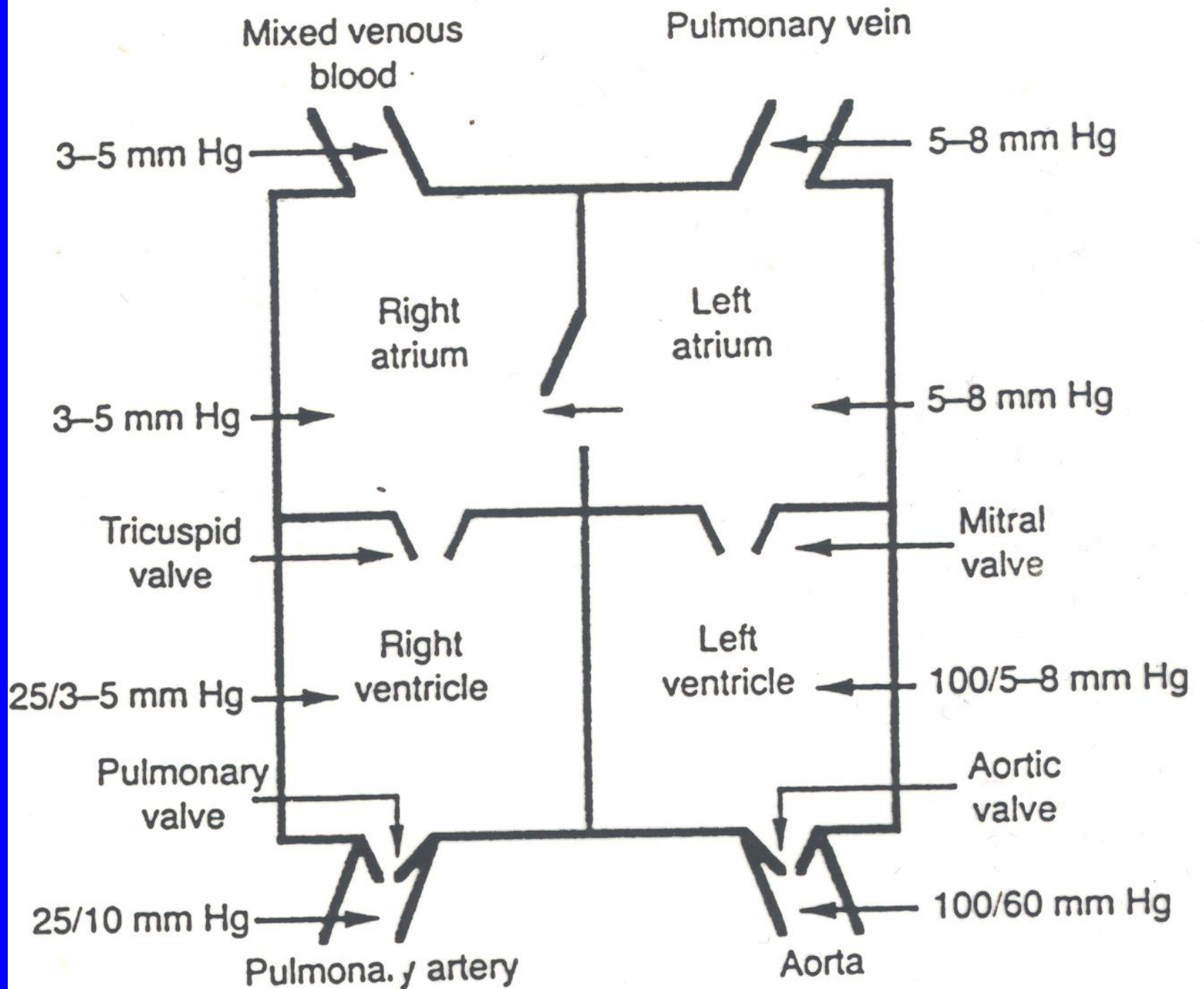
2016

Expected outcomes

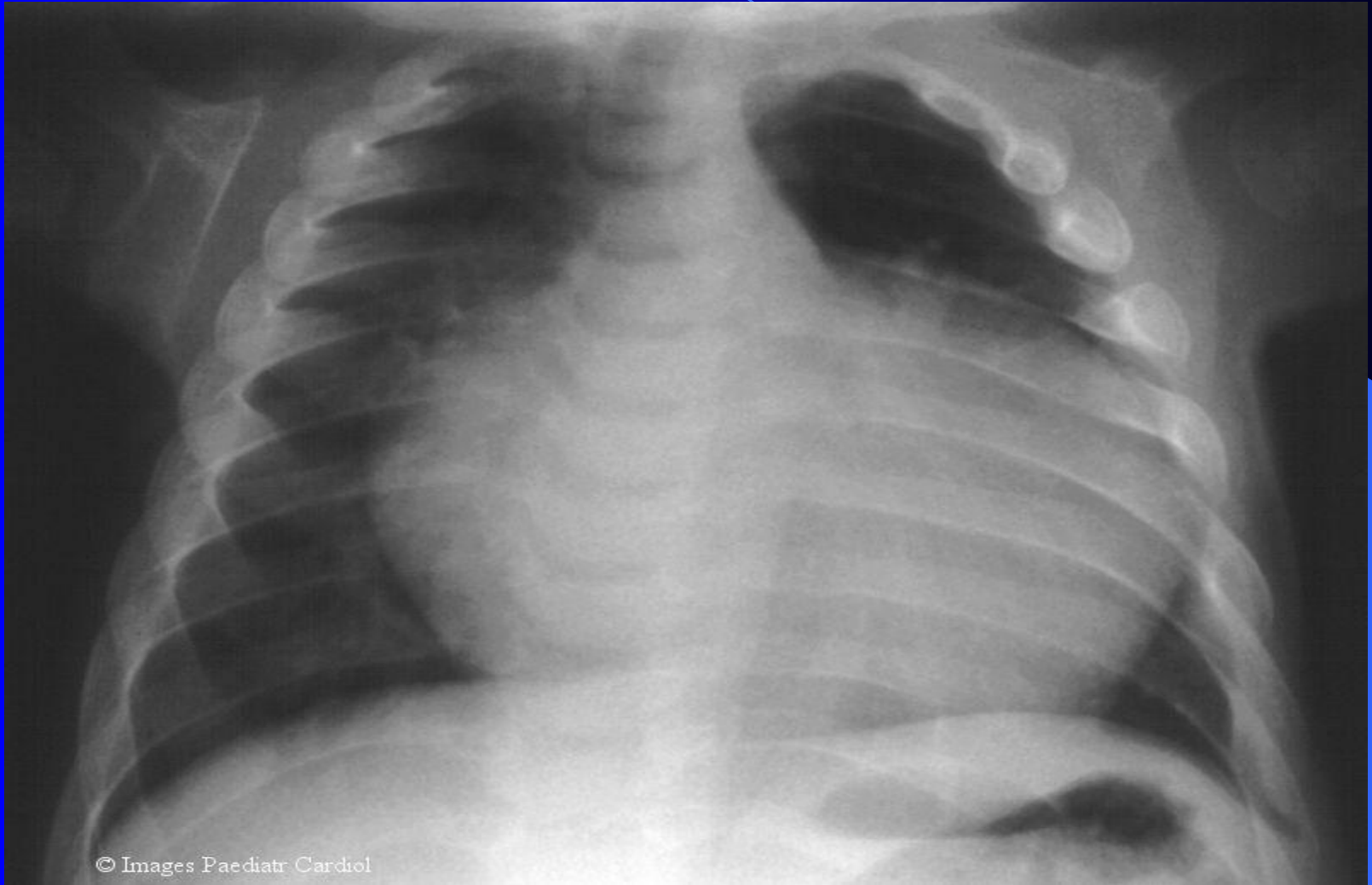
- Understand the different types
- Understand the structural defects
- Known risk factors and etiologies
- Role of inheritance
- Clinical presentations
- Outcomes

Normal Cardiac Blood Flow





Cardiomegaly



Congenital heart disease

- ◆ congenital cardiac malformations resulting from defective embryonic development.
- ◆ Between 3-8 weeks “gestation” all the fetal heart structures are formed “organogenesis”

Incidence

- ✓ 5-10/1000 live births
- ✓ Incidence is more in :-
 - a-Premature
 - b-abortions
 - c-still births
- ✓ Incidence increased for siblings.

SYNDROMES

- Down's: Incidence 50% . AV canal defects.
- Turner's : 10%. Coarctation , bicuspid aortic valve
- Williams's : Supravalvar aortic stenosis, PPS
- Alagille : Peripheral pulmonic stenosis (PPS)
- Noonan : PPS and HCM
- Marfan's : Aortic root dilatation, MVP

Syndromes

- DiGeorge: Truncus Arteriosus, Interrupted aortic arch.
- Catch 22 : conotruncal abn. such as VSD, TOF, collaterals, right aortic arch
- Kartagener : Dextrocardia, situs inversus, immotile cilia
- Holt-Oram: Limb abnormalities with ASD
- Ellis-van Creveld: ASD
- Pompe's D: Hypertrophic cardiomyopathy

Congenital Heart Disease-Etiology- Environmental Factors/Toxins

- Lithium: Ebstein's anomaly
- Ethanol: ASD, VSD (Fetal Alcohol Syndrome)
- Anticonvulsants: PS, AS, TOF
- Retinoic Acid: Transposition
- Rubella: PDA, PPS
- Coxsackie B : Neonatal myocarditis
- Maternal Diabetes: HCM, TGA
- Maternal Lupus: Complete heart block
- PKU: VSD, ASD, complex CHD

✓ Syndrome complexes

-VACTREL syndrome -

Vertebral, Anorectal, Cardiac (VSD, TOF and others), tracheal, Renal, Oesophageal and Limb abnormalities.

-CHARGE syndrome

“Coloboma, Heart (VSD, TOF, A-V canal), Atresia choanal, Retardation, Genital, Ear abnormalities.

-Kartagener syndrome - Dextrocardia

Majority of cases of the
congenital heart diseases are of
unknown cause

Congenital Heart Disease- Structural

- *Acyanotic*

Shunts (L to R) :

- ASD
- VSD
- PDA

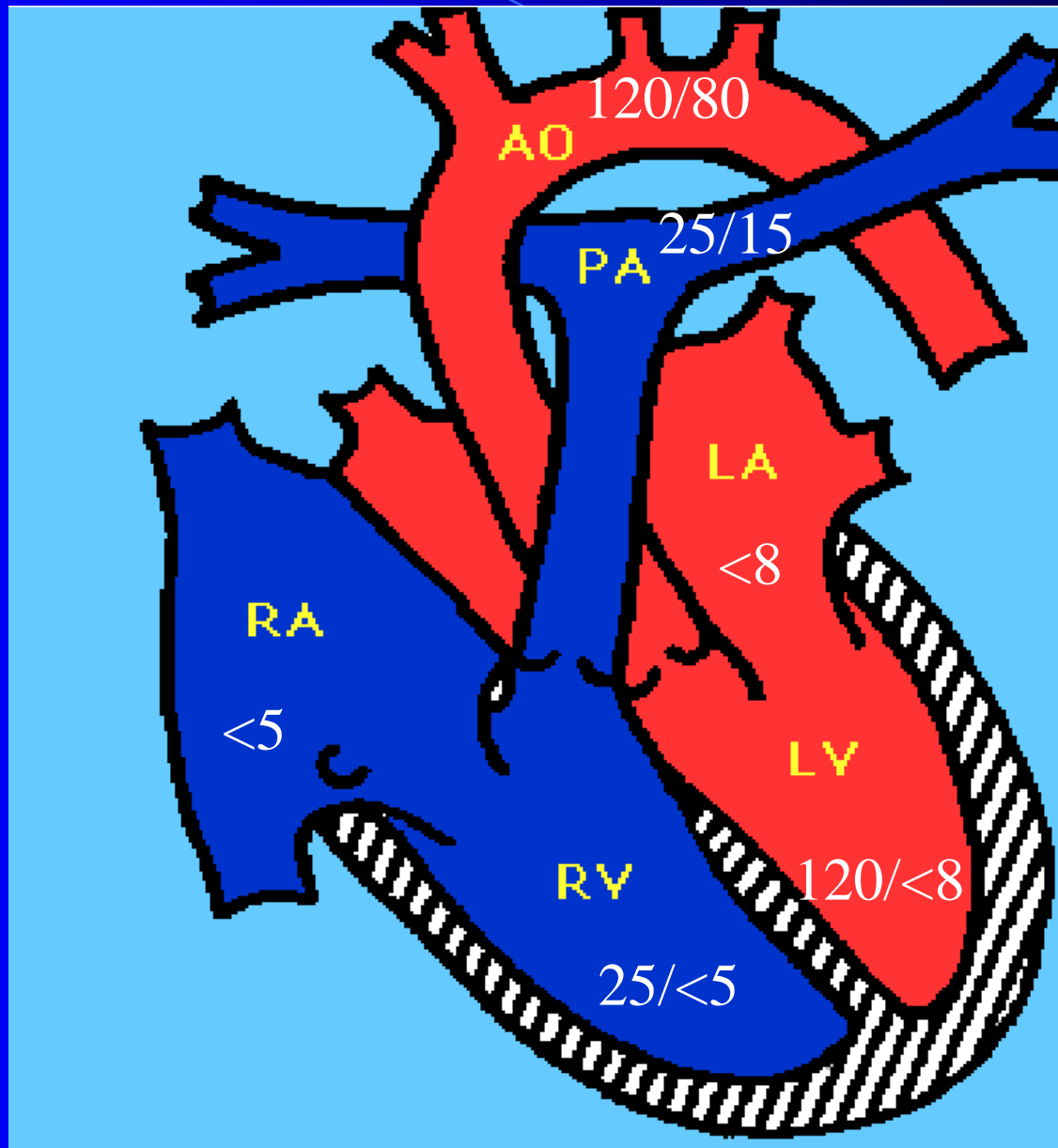
Stenosis:

- AS
- PS
- Coarctation

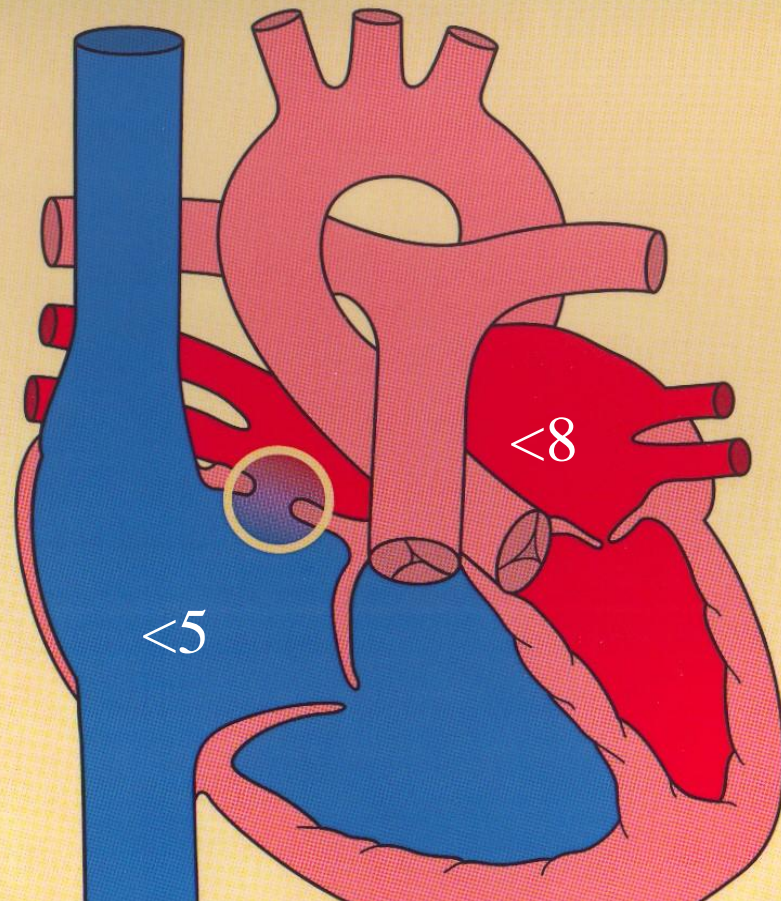
- *Cyanotic*

- TOF
- TGA
- Tricuspid atresia
- Truncus
- TAPVR
- Ebstein's
- Single ventricle

Normal Cardiac Pressures



ATRIAL SEPTAL DEFECT



ATRIAL SEPTAL DEFECTS (ASD)

- Three types exist : primum, secundum and sinus venosus
- The most common is the *secundum* type
- Symptoms: *None* in childhood, arrhythmias in the 3rd decade

ASD- cont...

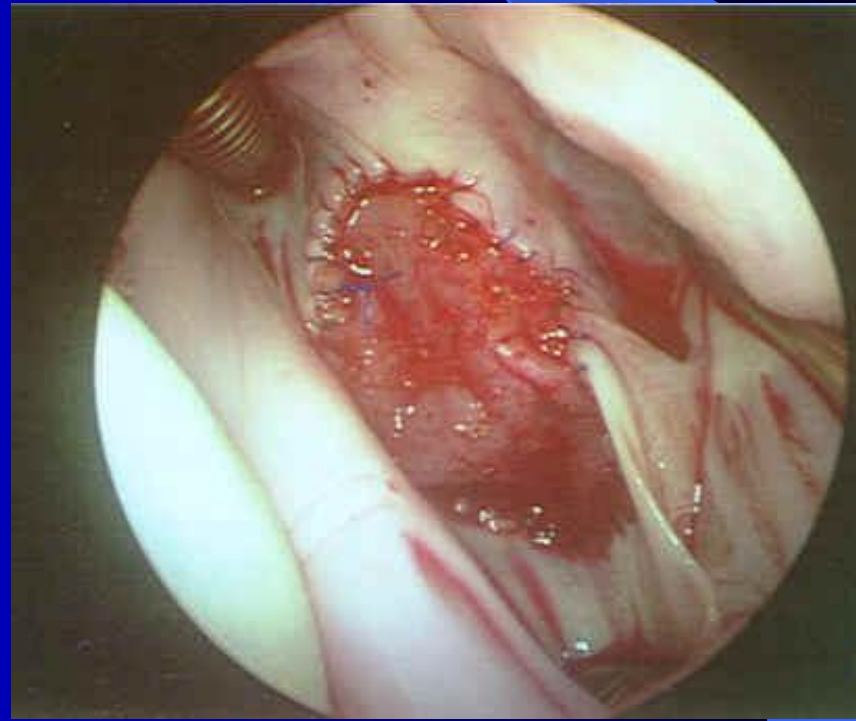
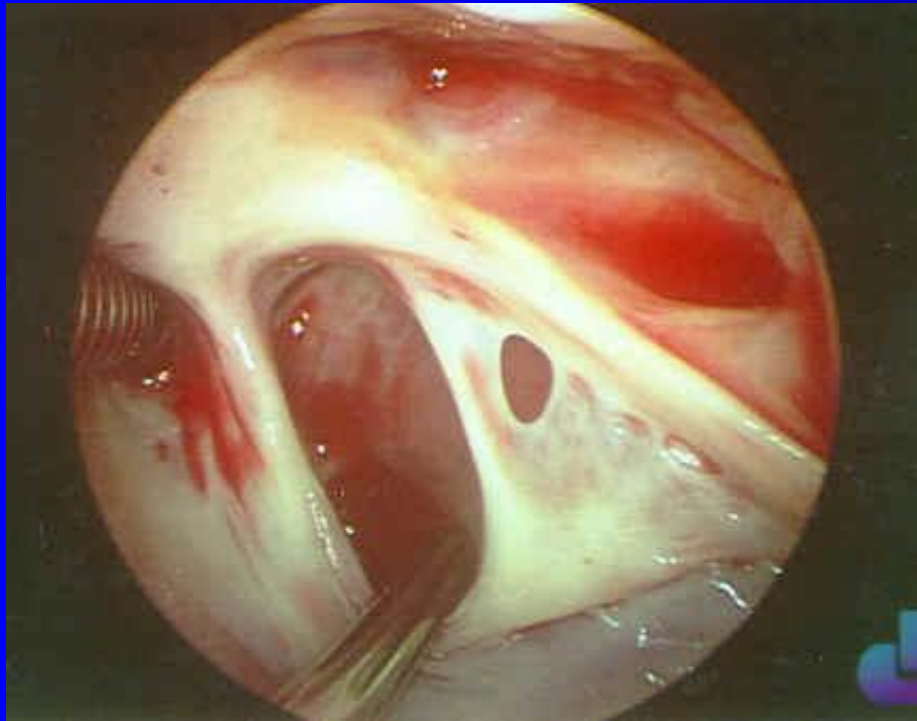
- Clinical signs include a *fixed wide split S2*
- A large ASD causes right ventricular enlargement
- Ecg

ASD - cont...

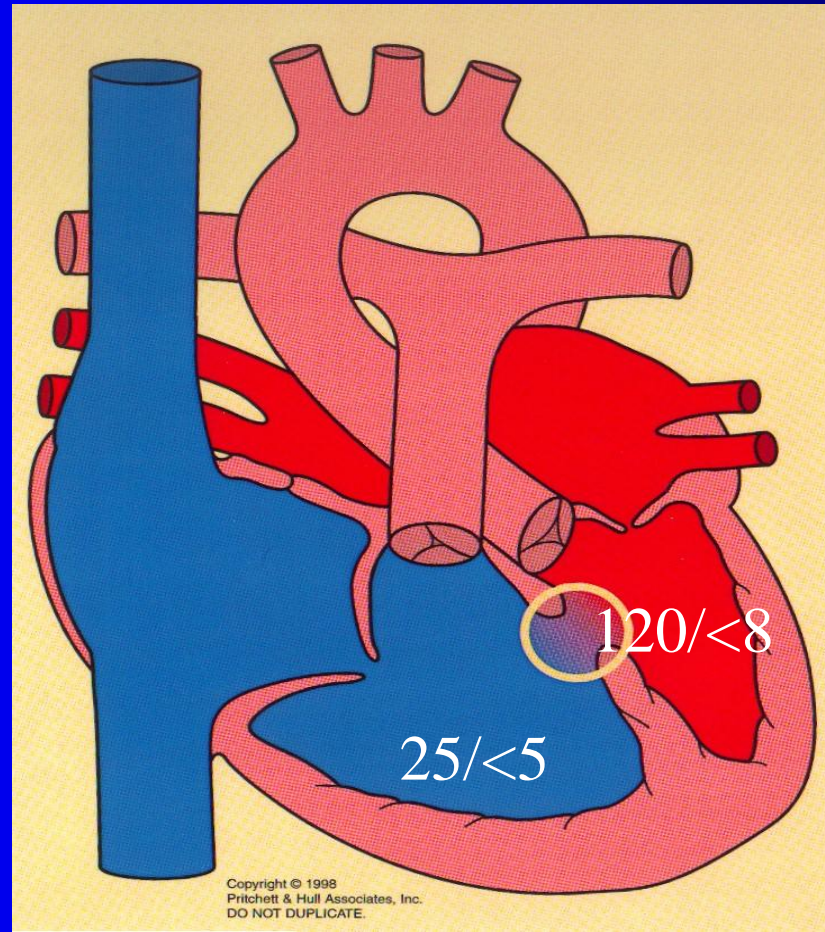
- **ECHO: Diagnostic**
- **Natural History: Arrhythmias and pulmonary obstructive vascular disease in the 3rd and 4th decade.**
- **Treatment : Surgical vs. transcatheter closure**

- Surgical closure.
- Non-Surgical closure via cardiac catheterization.

Treatment - ASD



VENTRICULAR SEPTAL DEFECT



VENTRICULAR SEPTAL DEFECTS (VSD)

- This is the most common form of CHD
- The VSDs are subdivided according to the part of the septum they occur in :
Muscular, perimembranous, inlet, outlet
- A large VSD causes left ventricular enlargement
- With a small VSD there is normal growth and development

VSD - cont.....

- **With a large defect there may be CHF(usually at 6-8 weeks), pulmonary infections and delayed growth**
- **Clinical signs : Loud 4-5/6 , harsh holosystolic murmur, middiastolic rumble and a loud P2**

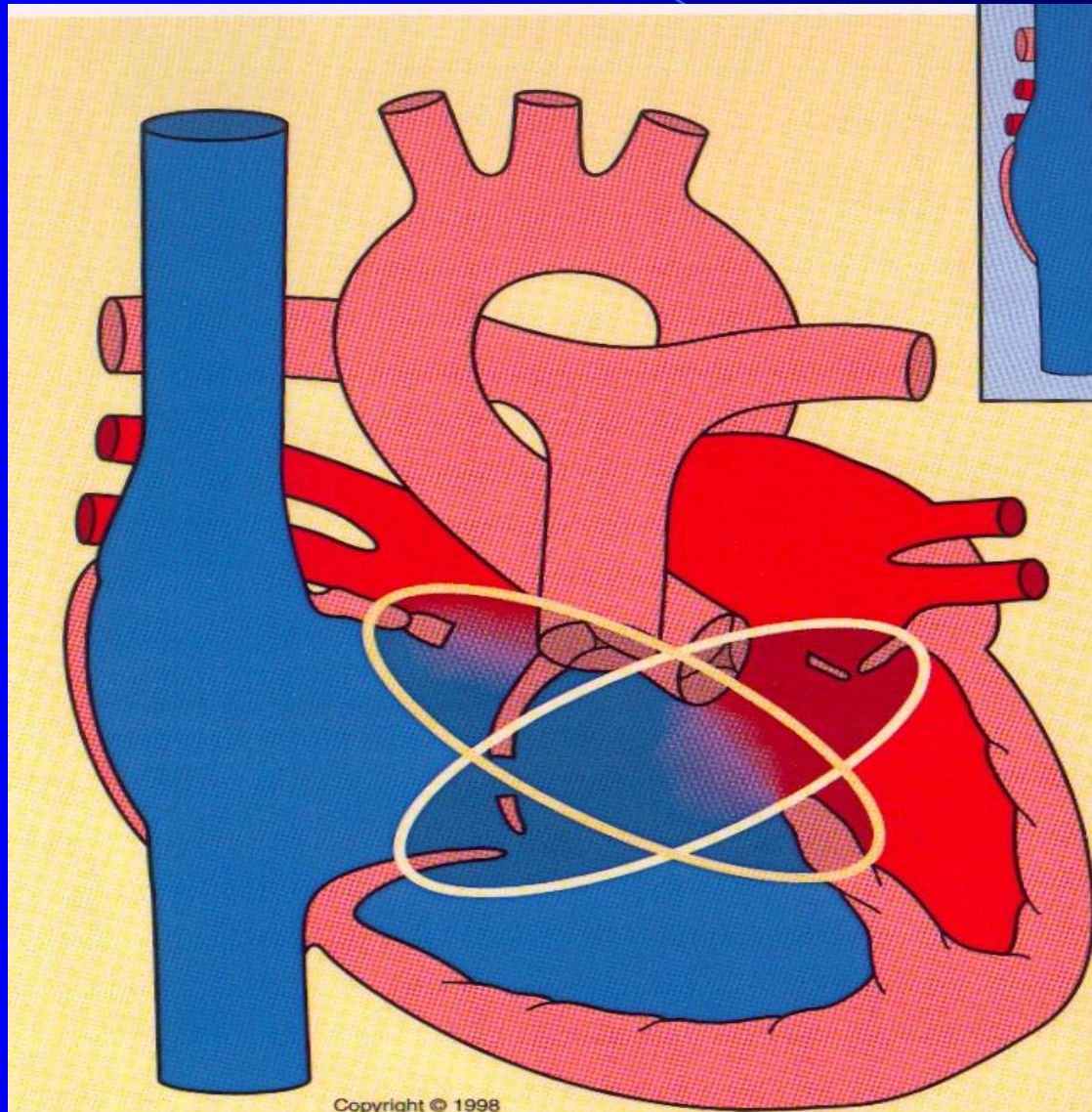
VSD -cont...

- **Natural history : Small VSDs close spontaneously depending on the site.**
- **Unrepaired the large defects may lead to Eisenmenger's syndrome.**

VSD - cont.....

- Large VSDs are closed surgically by 6 months of age.

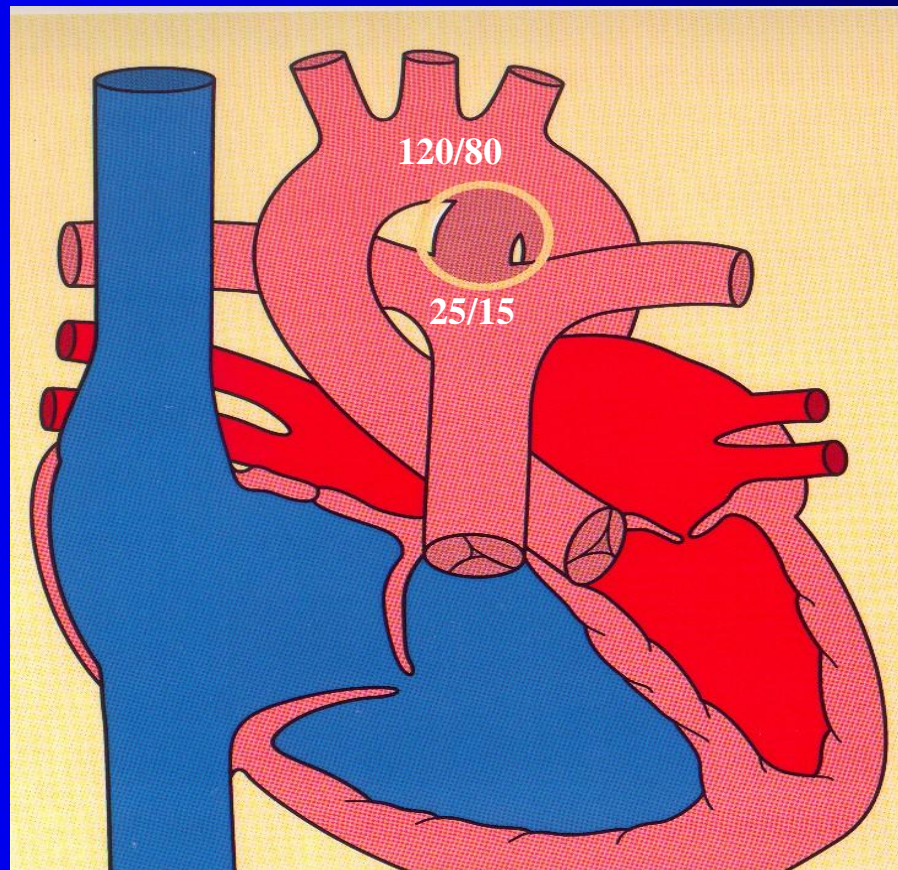
ENDOCARDIAL CUSHION DEFECTS



AVSD - cont...

- **1/3rd of babies with this have Down syndrome**
- **EKG : Characteristic with a superior left axis.**
- **Echo : Confirmatory**
- **Management : Anticongestive medications and surgery at 4-8 months of age.**

PATENT DUCTUS ARTERIOSUS



PATENT DUCTUS ARTERIOSUS (PDA)

- It is a connection between the aorta and the pulmonary artery.
- Very common in preterm babies.
- Usually closes in the first 2 weeks of life.

PDA - cont.....

- **Symptoms :**
- **a) None if small**
- **b) If large can cause CHF at 6-8 weeks in a term infant**

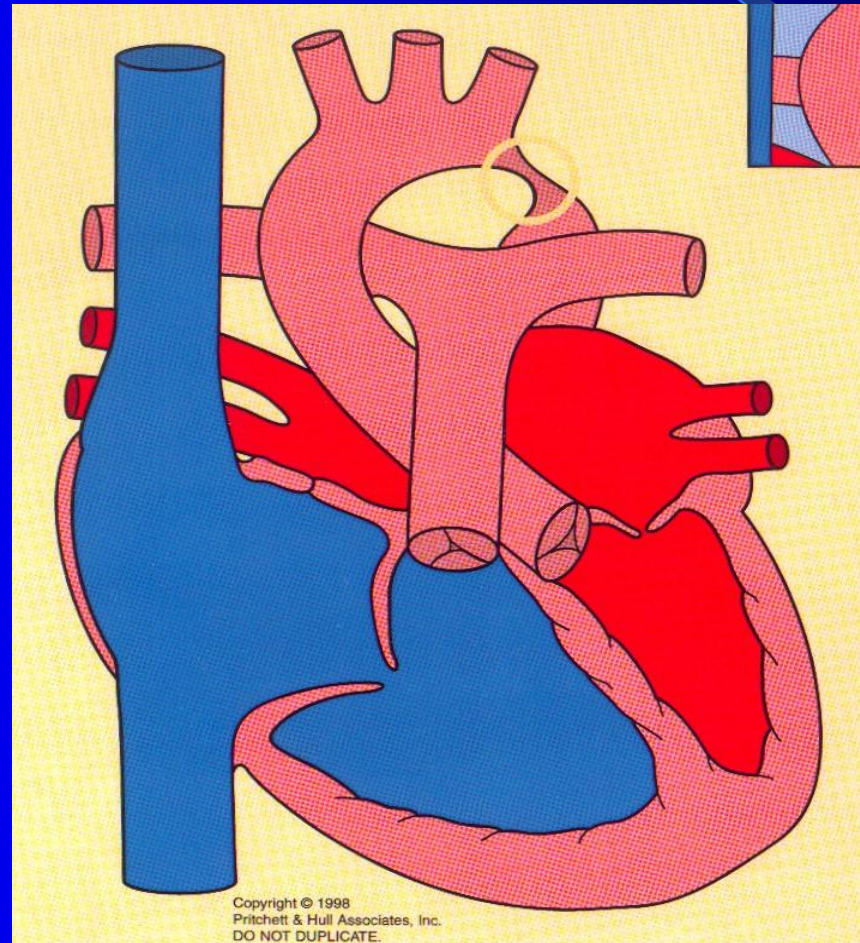
PDA

- **Signs: Systolic murmur in a newborn and a continuous “train in a tunnel” murmur in an older child. Best heard below the left clavicle.**
- **A large PDA causes LA and LV enlargement.**
- **Treatment : Preterm vs. term baby.**

PDA - cont...

- **In a preterm it can be closed medically using indomethacin.**
- **In a term baby if still open at 3 months of age then coil closure by cardiac catheterization is the method of choice.**

COARCTATION OF THE AORTA



Coarctation of the Aorta (CoA)

- **More common in males**
- **Almost always juxtaductal**
- **85% of children with CoA have a bicuspid aortic valve.**

CoA - cont....

- **Symptoms and Signs:**
 - SEVERE : Shock**
 - MODERATE : CHF,**
 - MILD : Headaches, leg claudication**
- **Decreased femoral pulses are an important sign esp. in neonates.**
- **BP lower in the lower limbs**

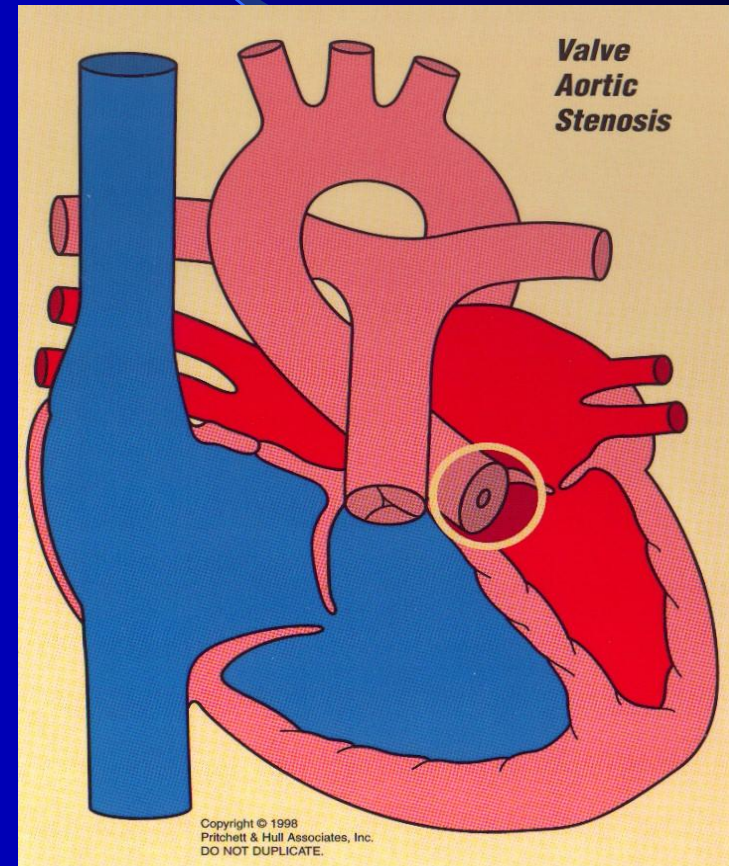
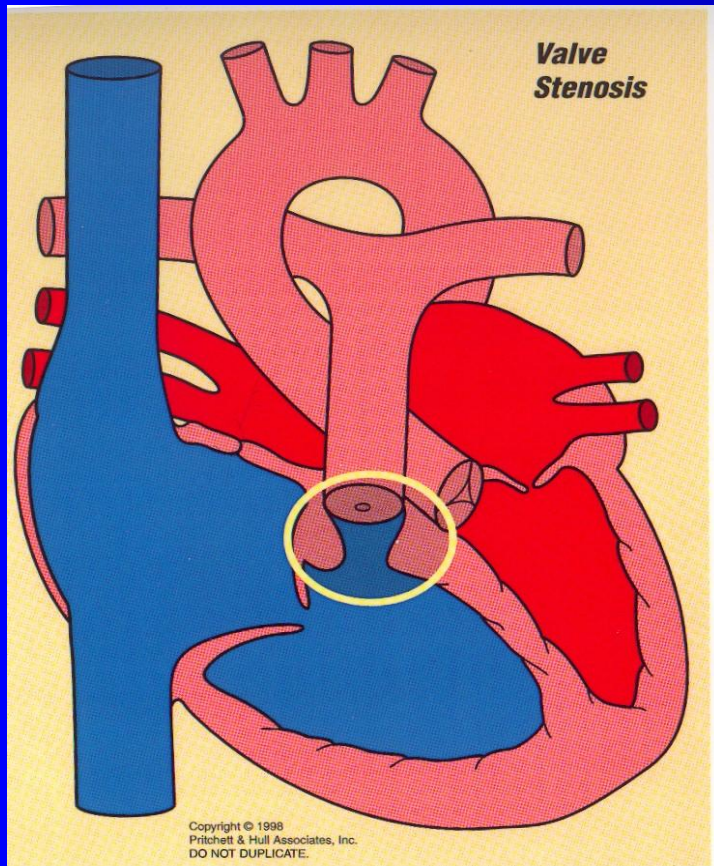
CoA - cont....

- **ECHO : Diagnostic**
- **Treatment: For an infant in shock - PGE1 immediately.**
- **Surgical vs. transcatheter repair.**

Hypoplastic Left Heart Syndrome

- Varying degrees of left heart hypoplasia at multiple levels
- Babies present in cardiogenic SHOCK once the ductus closes.

Pulmonic/ Aortic Stenosis



Stenosis

Pulmonic

- This may be at the valve, subvalvular or supra-avalvular.
- Symptoms: None in mild or moderate stenosis. Cyanosis is seen only with critical PS.
- Signs: ejection click and a harsh SEM.
- ECHO : Diagnostic
- Treatment: Ballooning

Aortic

- Stenosis possible at the valve, subvalvular or supra-avalvular.
- This is a more significant and a dangerous lesion compared to PS.
- More common in males.
- Valvular AS is usually associated with a bicuspid aortic valve.
- Treatment: Ballooning

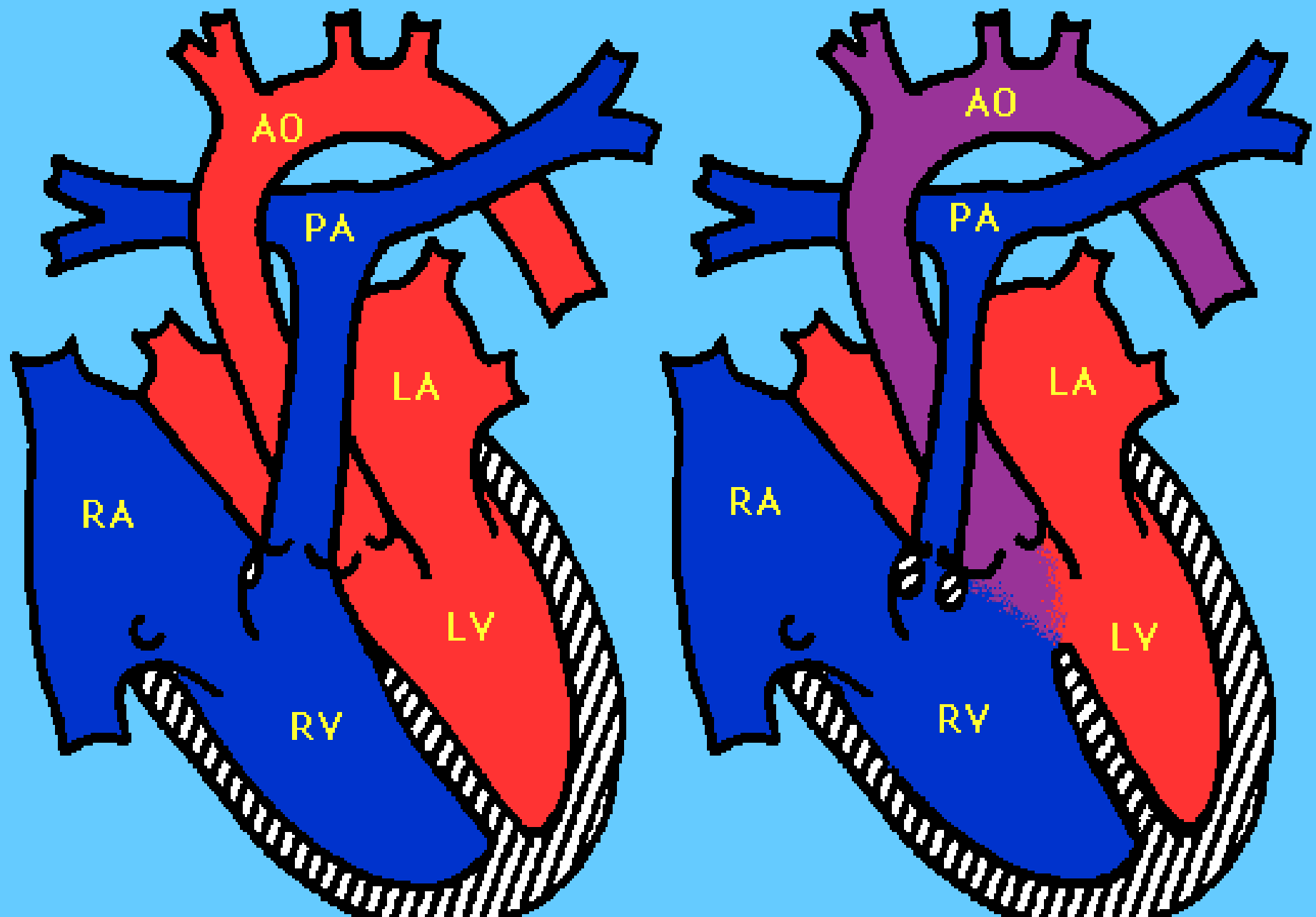
Cyanotic lesions.

There has to be a RIGHT to
LEFT shunt to cause
cyanosis

Tetralogy of Fallot

- **Most common cyanotic heart disease.**
- **The four abnormalities include:**
 - **Pulmonary stenosis**
 - **RVH**
 - **VSD**
 - **Overriding Aorta**
- **Signs include cyanosis, murmur, squatting and spells.**

Tetralogy of Fallot



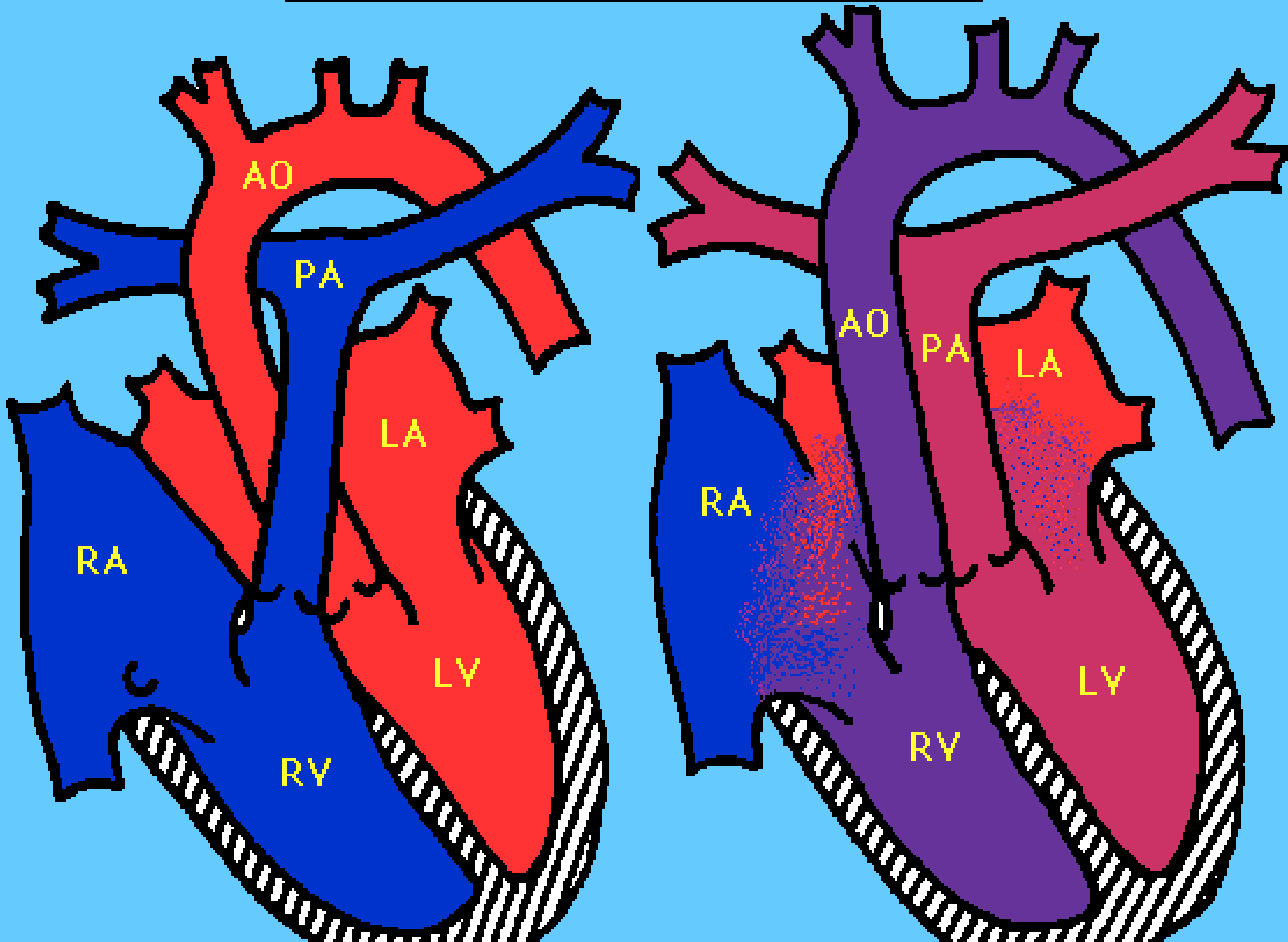
TOF cont..

- A “tet” spell consists of rapid breathing and increased cyanosis. Any event like crying or increased physical activity can initiate the spell.



TRANSPOSITION OF THE GREAT ARTERIES

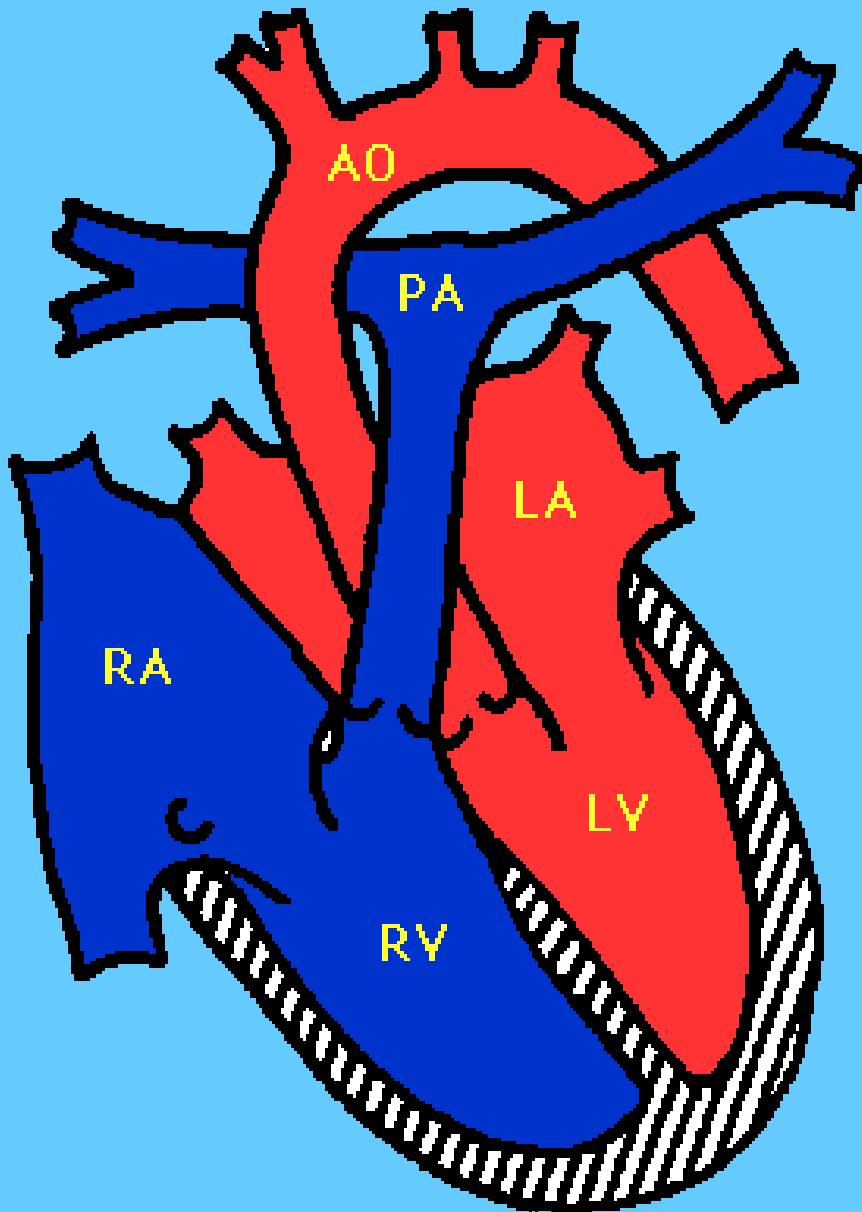
Transposition of the Great Vessels



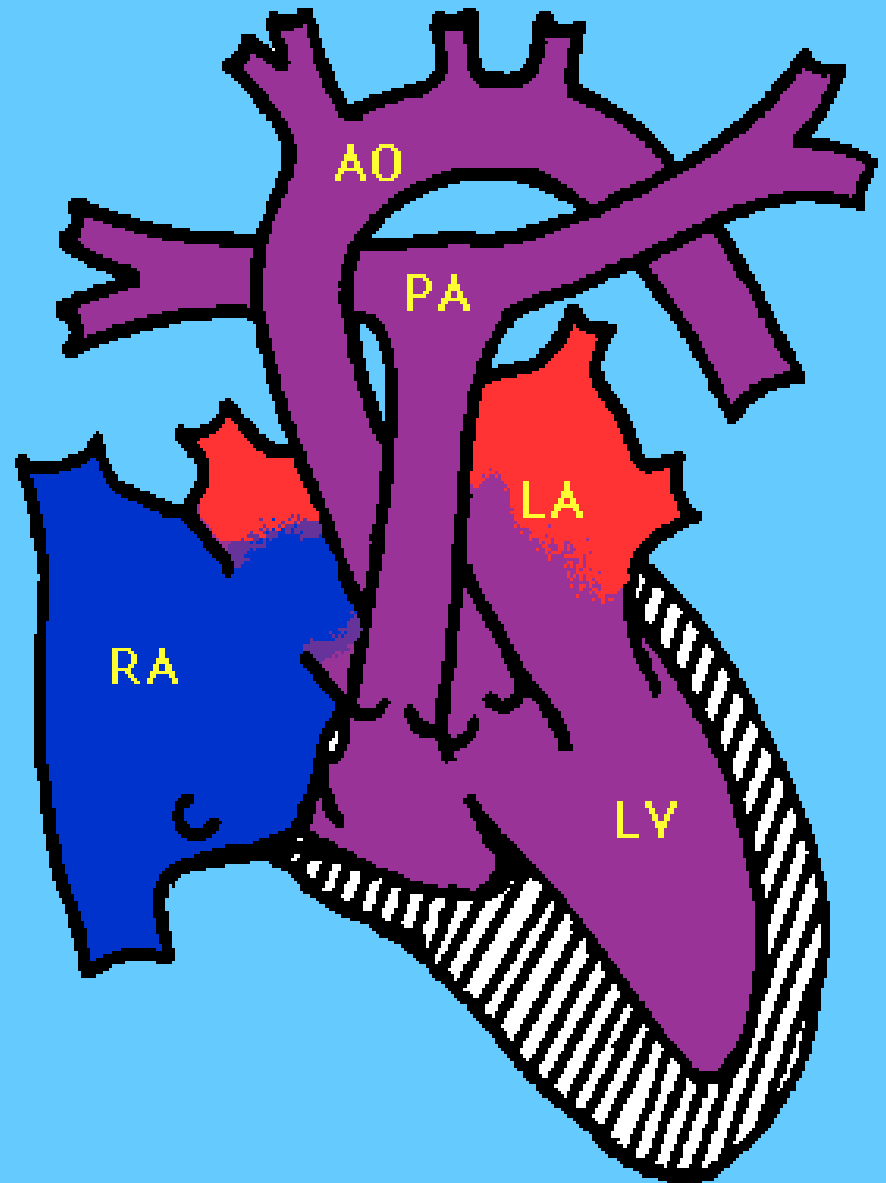
Transposition of the great Arteries

- The aorta arises from the right ventricle and the pulmonary artery from the left.
- The mixing of the blood occurs at the PFO and the PDA.
- The signs include cyanosis and cardiomegaly.
- There may be no murmur.
- An echocardiogram is diagnostic.

Tricuspid Atresia

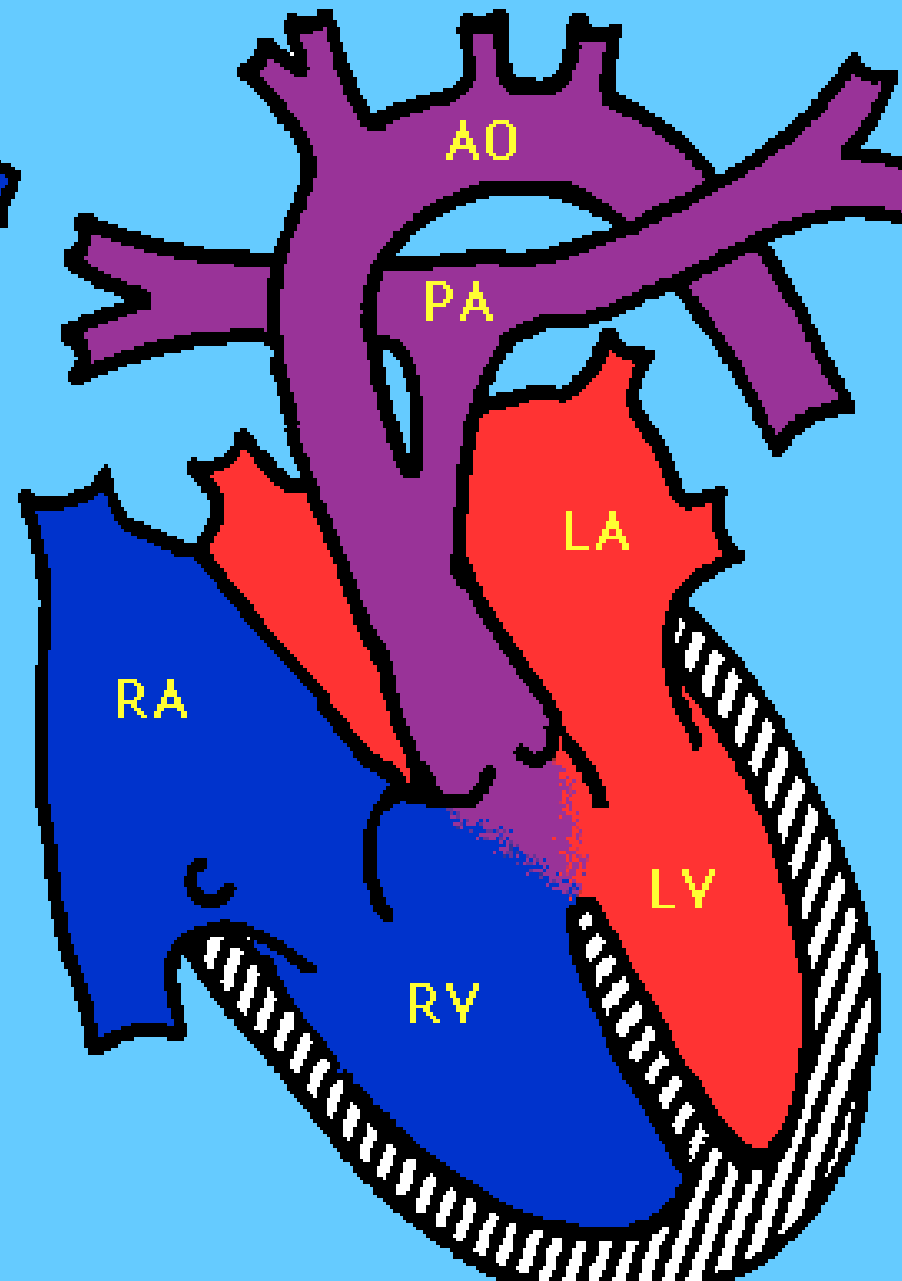
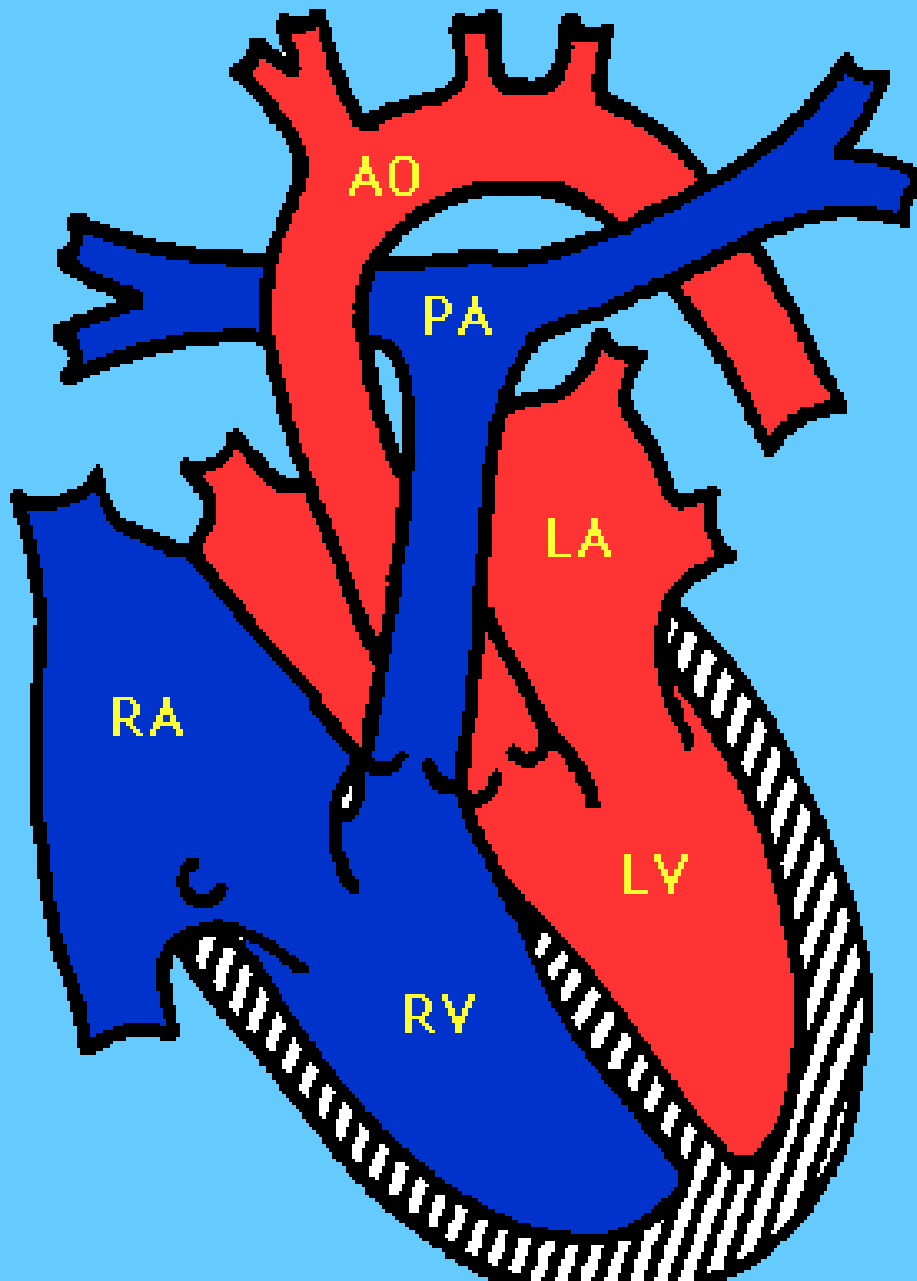


Normal



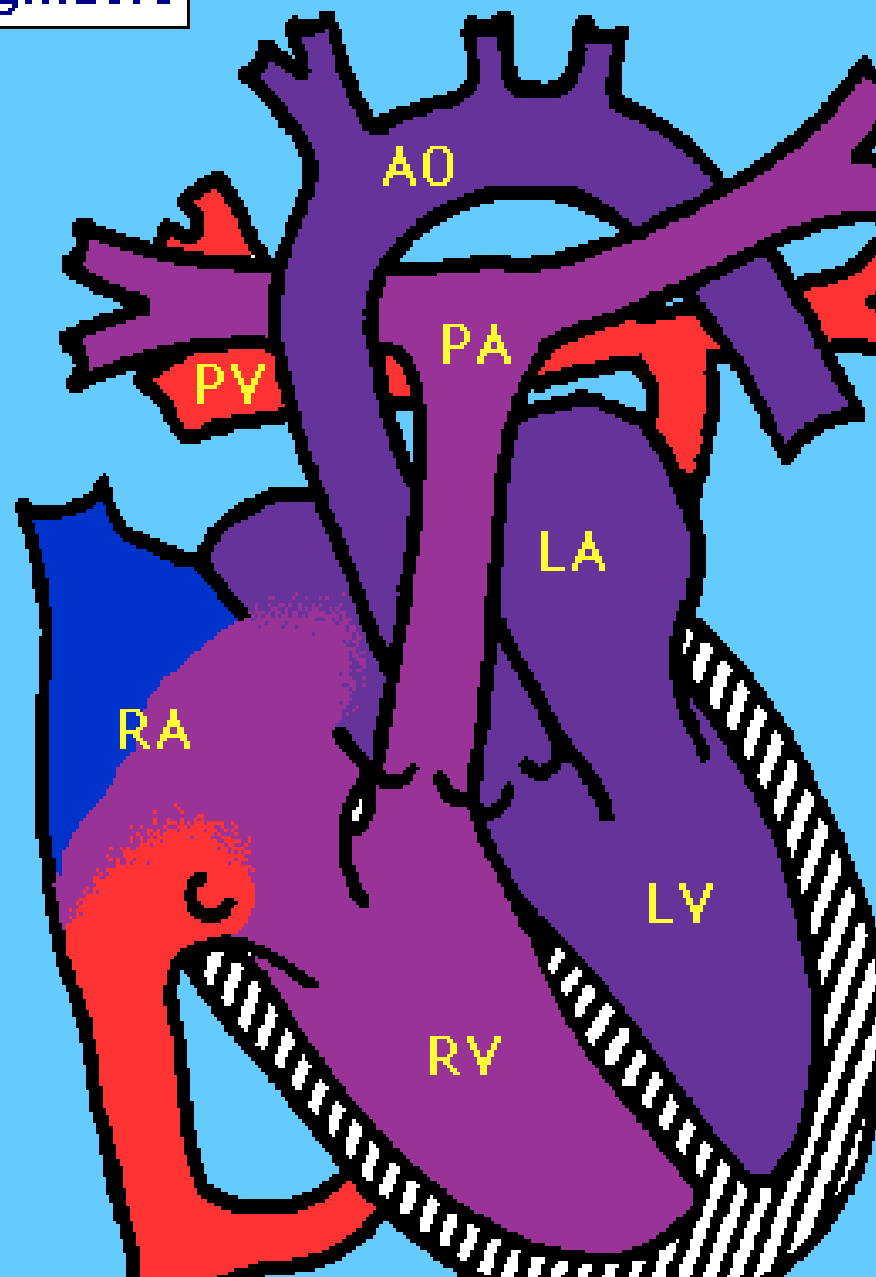
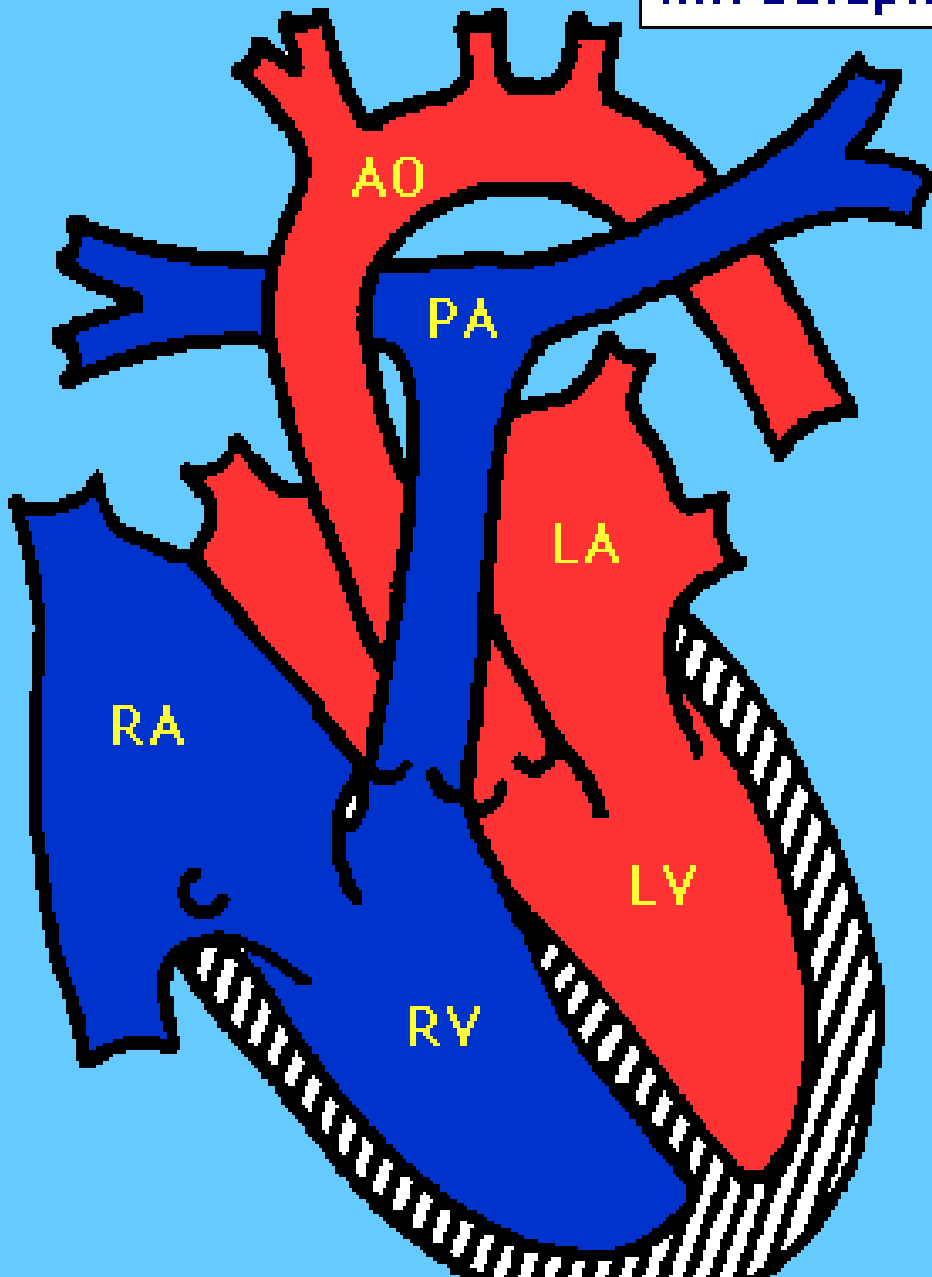
Tricuspid Atresia

Truncus Arteriosus



Total Anomalous Pulmonary Venous Return

Infradiaphragmatic



Ebstein Anomaly

