Congenital Heart Disease: Cyanotic Lesions

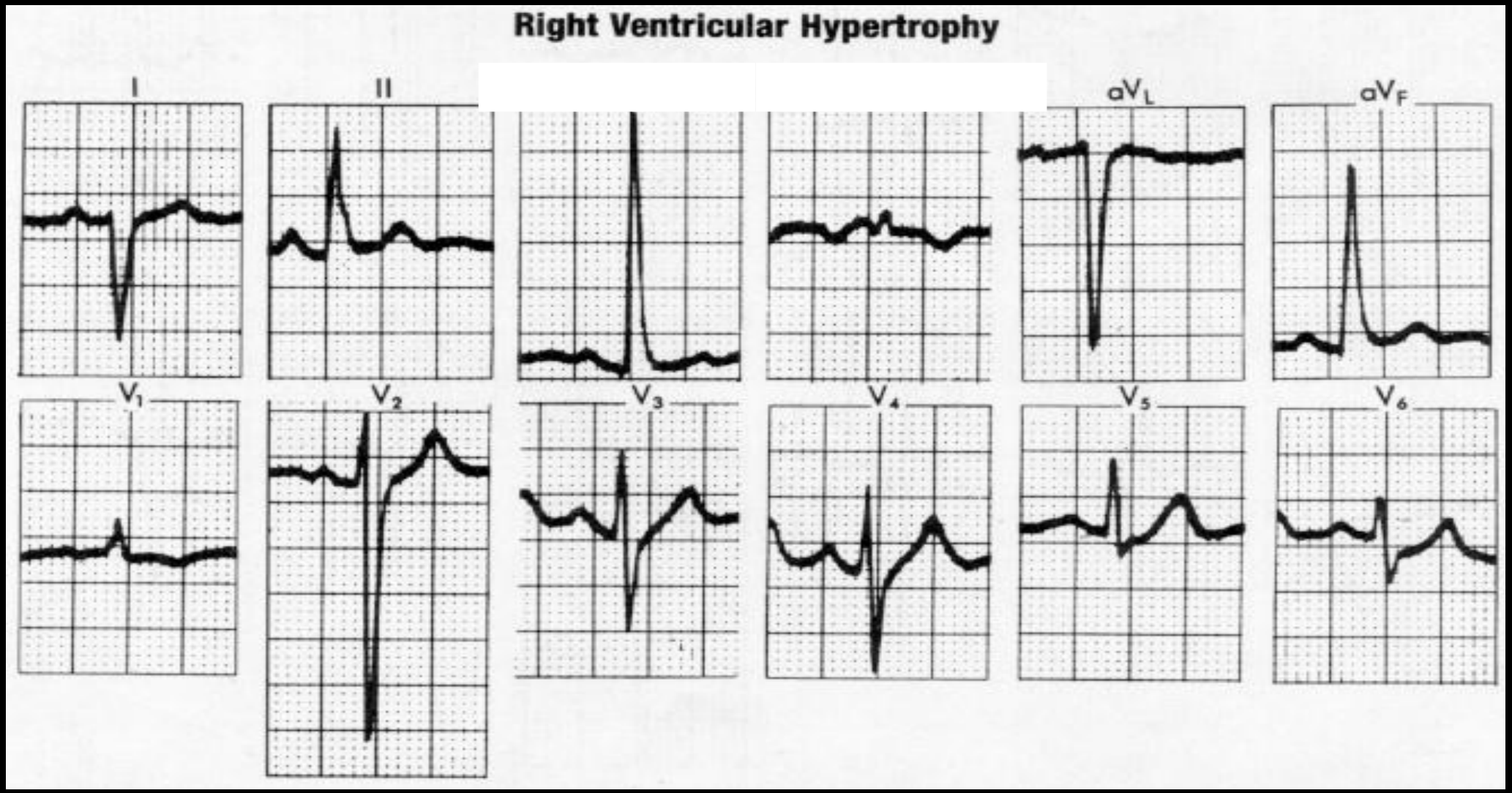
Amitesh Aggarwal

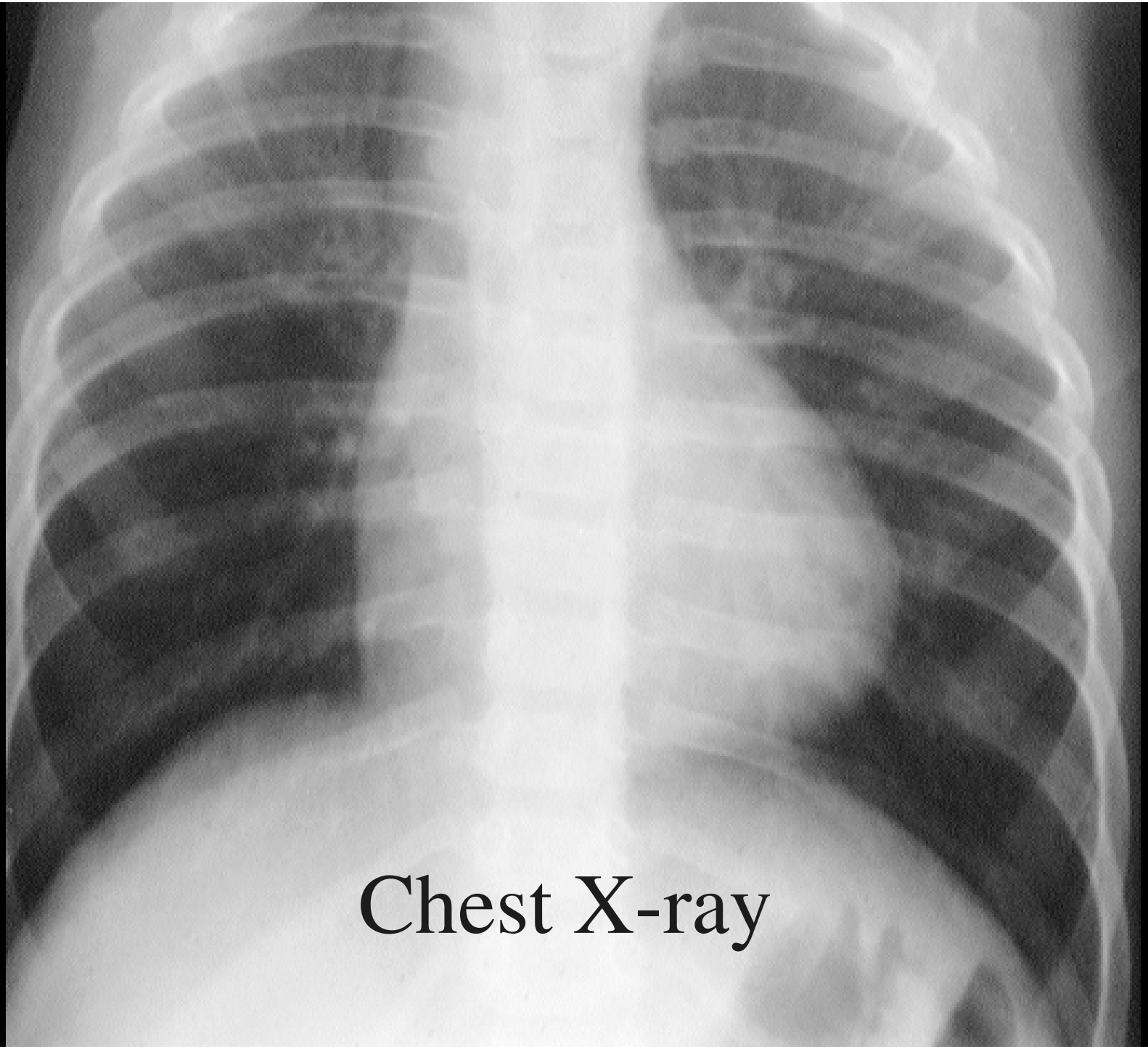
- 12 y/o male admitted because of dyspnea and cyanosis
- Patient has been cyanotic since few months after birth
- Has episodes of tachypnea and worsening cyanosis which improved with squatting
- Known history of heart disease but mother has refused surgery

Physical Exam

- Bluish discoloration
- Clubbing of fingers and toes
- Parasternal lift
- S1:normal;S2:single
- Grade 3/6 systolic ejection murmur at the 2nd and 3rd ICS, LSB

ECG





Chest X-ray

Evaluation of cyanosis: 100% O_2 test

measure pO_2 in room air and 100% O_2

Lung disease:

1. Room air pO_2 30 mmHg
   O_2 sat=60%
2. 100% O_2 pO_2 110 mmHg
   O_2 sat=100%

Cardiac disease:

1. Room air pO_2 30 mmHg
   O_2 sat=60%
2. 100% O_2 pO_2 40 mmHg
   O_2 sat=75%

pO_2 >100 mmHg suggests lung disease
Little or no change in pO_2 suggests cyanotic heart disease

Evaluation possible congenital heart

- Exam: rate, rhythm, impulse, murmur, pulses (brachial and femoral)
- Oxygen saturation
- ABG
- Chest x ray
- Hyperoxia test
- Echocardiogram

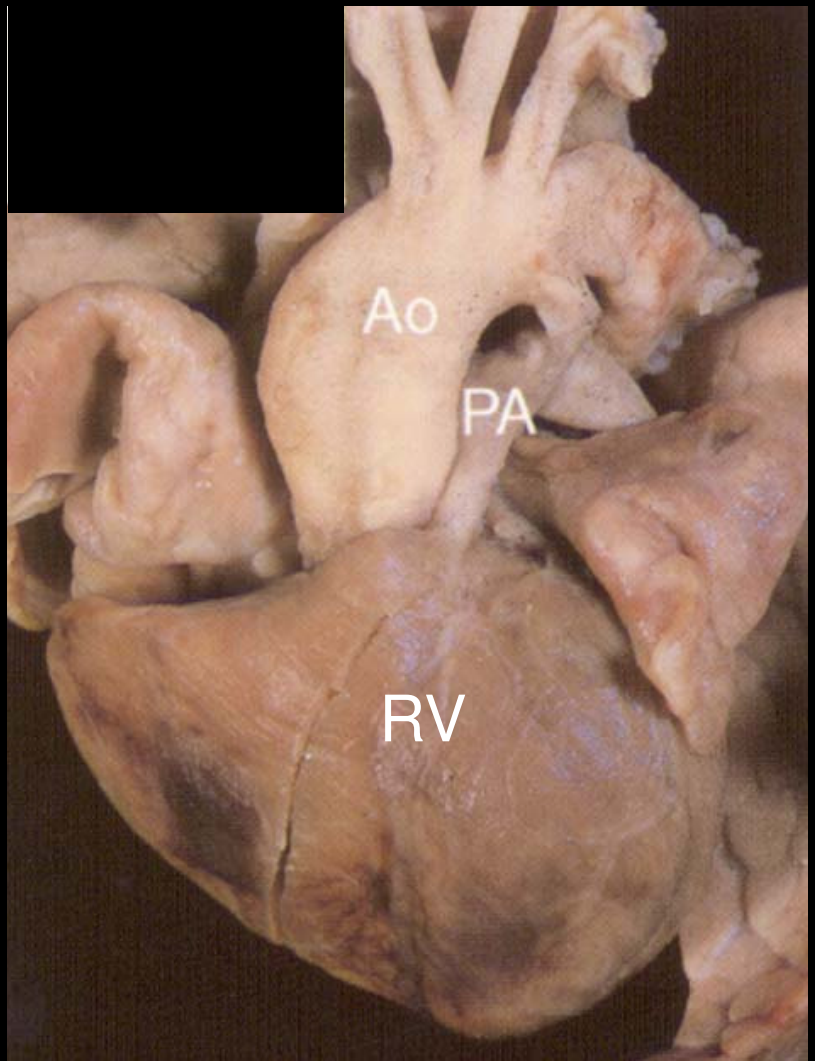
Cyanotic Lesions

- **CXR helps in diagnosis**
  - oligaemic lungfields
    - ↗ PS, pulmonary atresia, TOF, Tricuspid atresia
  - plethoric lung fields
    - ↗ TGA, TAPVC, TA
  - massive cardiomegaly
    - ↗ Ebstein's

The 5 T's of cyanotic heart disease

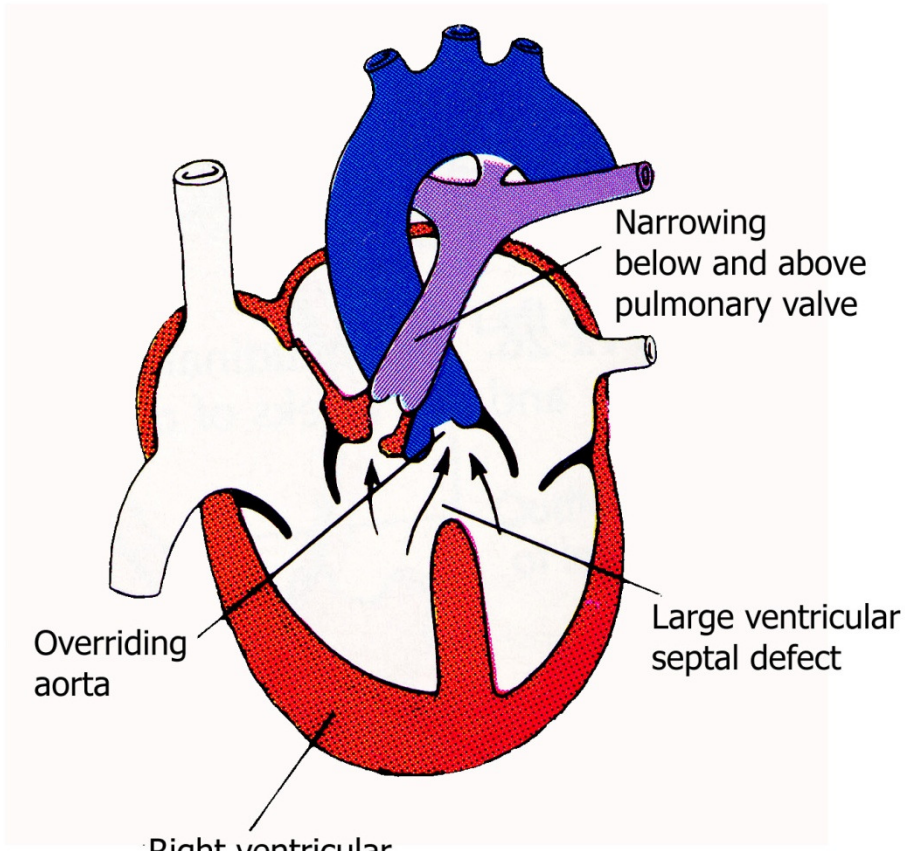
- Tetralogy of Fallot
- TGA (d-transposition of the great arteries)
- Truncus arteriosus
- Total anomalous pulmonary venous communication
- Tricuspid atresia / Pulmonary atresia
- Ebstein's malformation of Tricuspid valve

Tetralogy of Fallot

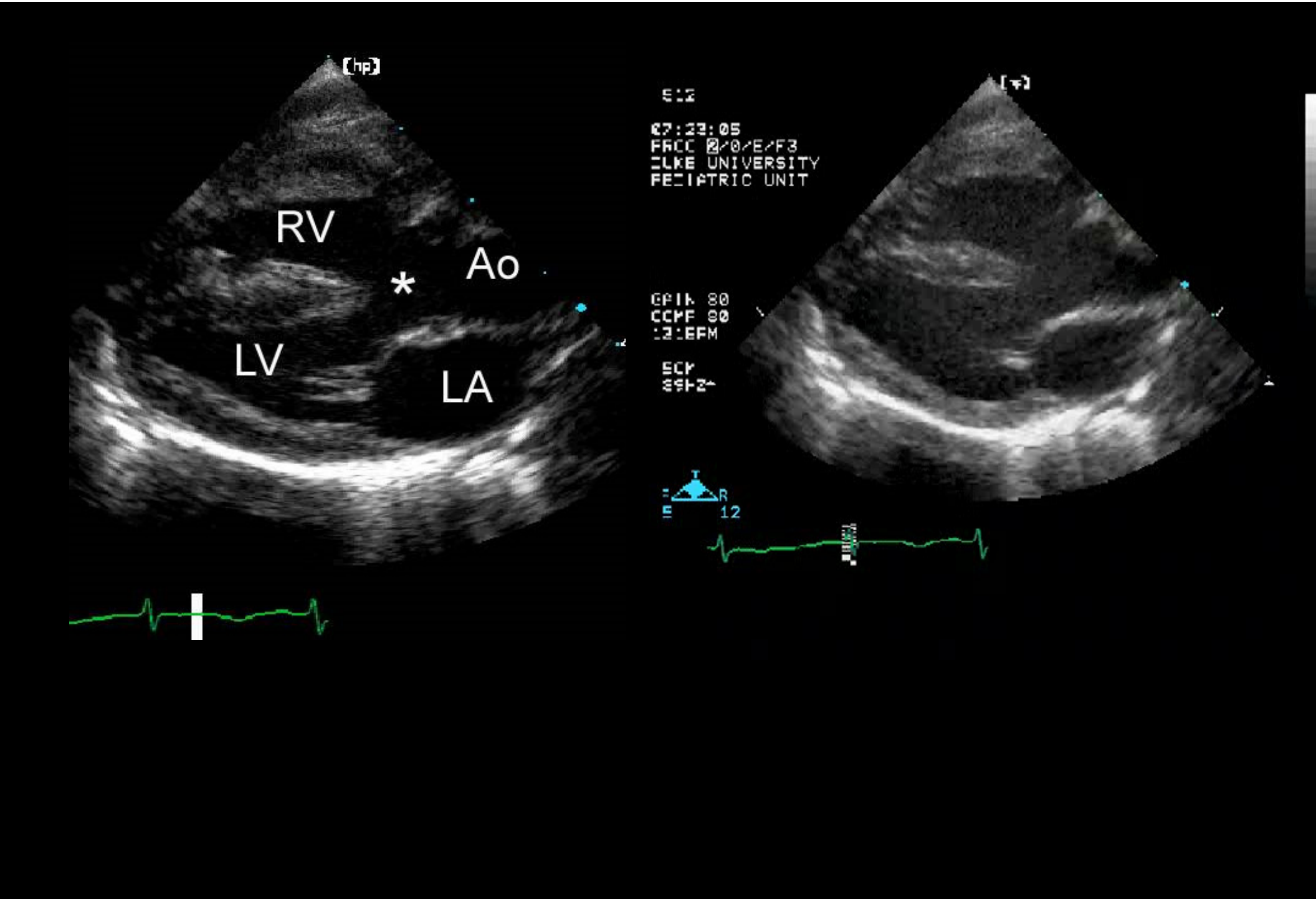


- 6 % of all congenital heart disease
- 1:3600 live births
- Most common cause of cyanosis in infancy/childhood
- Severity of cyanosis proportional to severity of RVOT obstruction

Tetralogy of Fallot



1. Pulmonary stenosis
2. Large VSD
3. Overriding aorta
4. Right ventricular hypertrophy


RV
Ao
*
LV
LA
12

TOF - problems

- Reduced blood flow to the lungs
- Low 02 blood pumped up Ao (shunting)
- Reduced Sa02 in circulation
- Cyanosis – baby appears blue (lips/skin)
- Increased RV pressure (RVH)

History

- Cyanosis develops within the first few years of life.
- Symptoms progress secondary to hypertrophy of infundibular septum.
- Cyanosis occurs and demands surgical repair.
- Dyspnea on exertion common.
- Squatting is uniquely characteristic of a right-to-left shunt.
- Hypoxic "tet" spells are potentially lethal, unpredictable episodes that occur even in noncyanotic patients with TOF.
- Birth weight is low.
- Growth is retarded.
- Development and puberty may be delayed.

Physical

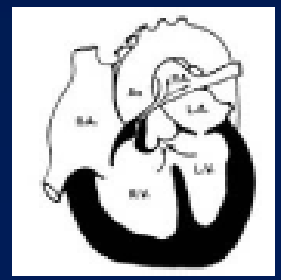
- Right ventricular predominance (later)
- Systolic thrill at the lower left sternal border
- Aortic ejection click
- Single S2 - Pulmonic valve closure not heard
- Systolic ejection murmur - Varies in intensity inversely with the degree of RVOT obstruction
  - More cyanotic patients have greater obstruction and a softer murmur.
  - An acyanotic patient with TOF (pink tet) has a long, loud, systolic murmur with a thrill along the RVOT.
- Cyanosis and clubbing - Variable
- Scoliosis - Common

TOF frequently is associated with the following:

- Fetal hydantoin syndrome
- Fetal carbamazepine syndrome
- Fetal alcohol syndrome
- Maternal phenylketonuria (PKU) birth defects
- DiGeorge syndrome
- Branchial arch abnormalities

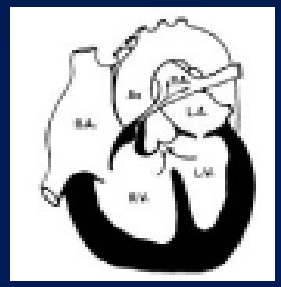
Tetralogy of Fallot
Critical Component

- Degree of pulmonic stenosis
  - Regulates degree of R → L shunt
  - Regulates overriding of aorta
    - Greater the stenosis, the greater the aortic overriding



Tetralogy of Fallot
Other anomalies

- Right aortic arch in 25%
  - Mirror image type
- Left superior vena cava
- ASD
- Tricuspid valve abnormalities
- Anomalies of coronary arteries
  - Aberrant left anterior descending coronary artery arising from right coronary artery



Trilogy of Fallot

- Pulmonic valvular stenosis
- ASD
- Right ventricular hypertrophy

PENTOLOGY OF FALLOT

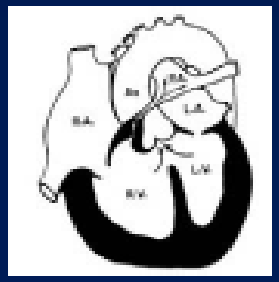
TOF+ASD

ACYANOTIC FALLOT

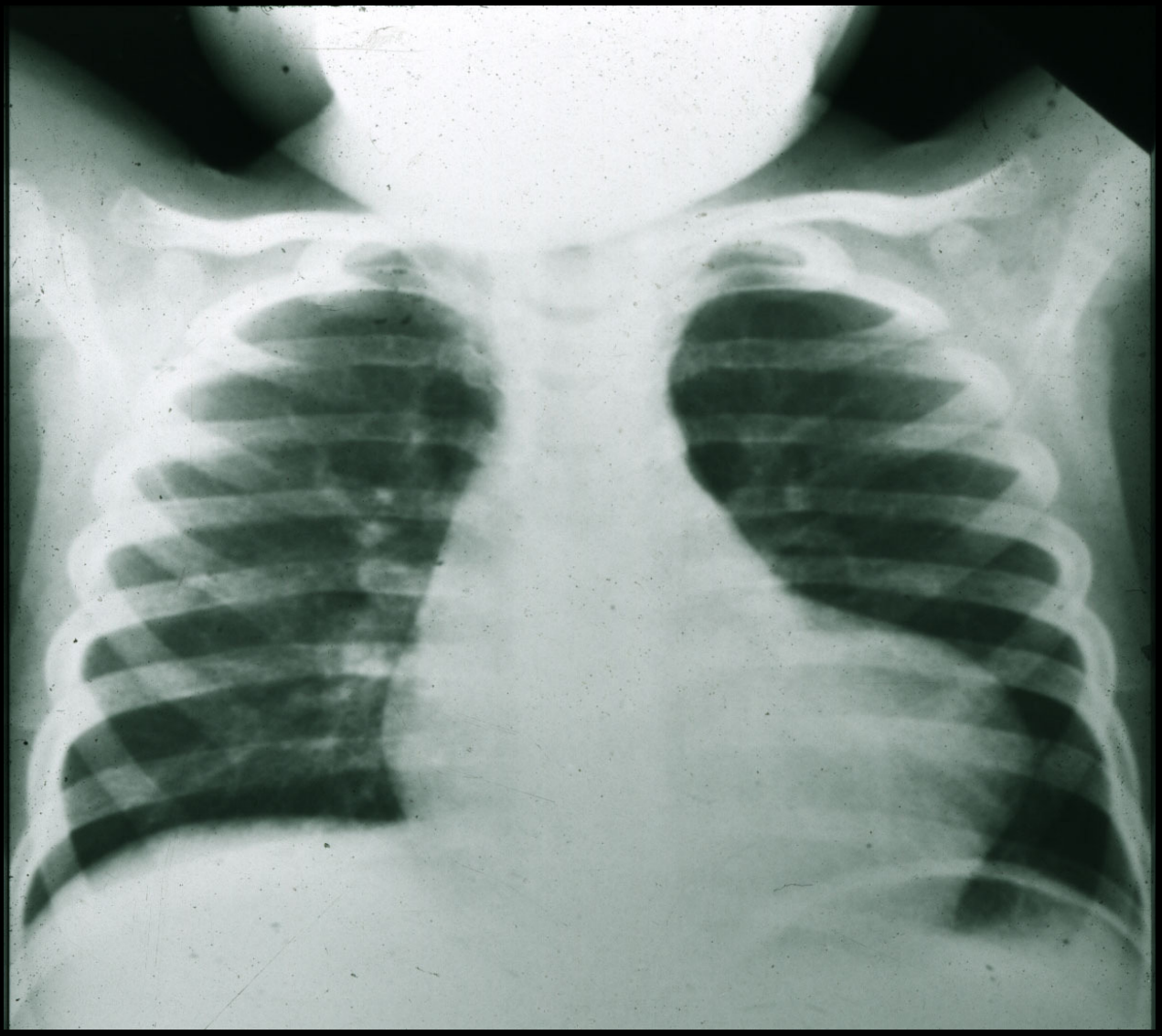
SLIGHT OBS+ BIDIRECTIONAL SHUNT

Tetralogy of Fallot
Imaging Findings

- Heart size normal
  - Rarely enlarged
- Cardiac apex displaced upward "coer en sabot"
- PA segment concave
- Decreased vasculature
- R aortic arch in 25%



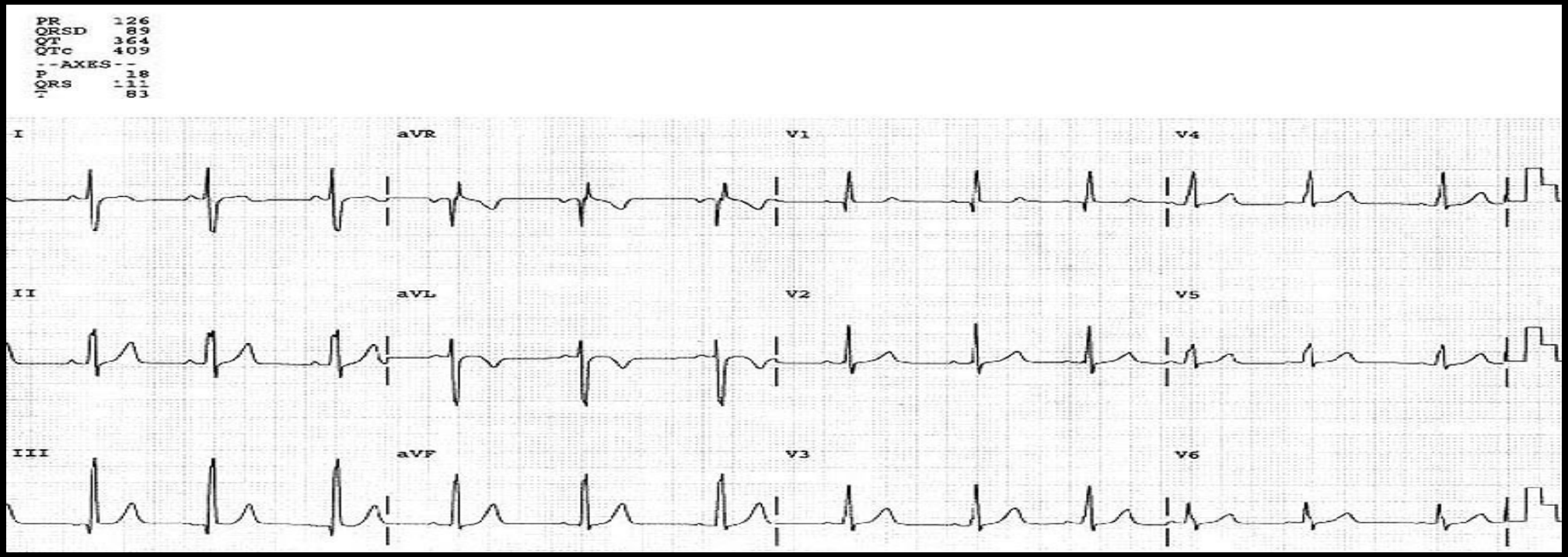
Tetralogy of Fallot - CXR



- Typical "boot-shaped" heart secondary to RVH and small main pulmonary artery segment
- Pulmonary vascular markings are decreased

Electrocardiogram

- Right axis deviation
- Right or combined ventricular hypertrophy
- Right atrial hypertrophy



Natural History

- Prognosis is poor without surgical repair or palliation
- The survival rate of repaired TOF is worse compared to age-matched control
  - Sudden cardiac death: VT
  - Atrial arrhythmias
  - Infective endocarditis
  - Pulmonic Regurgitation

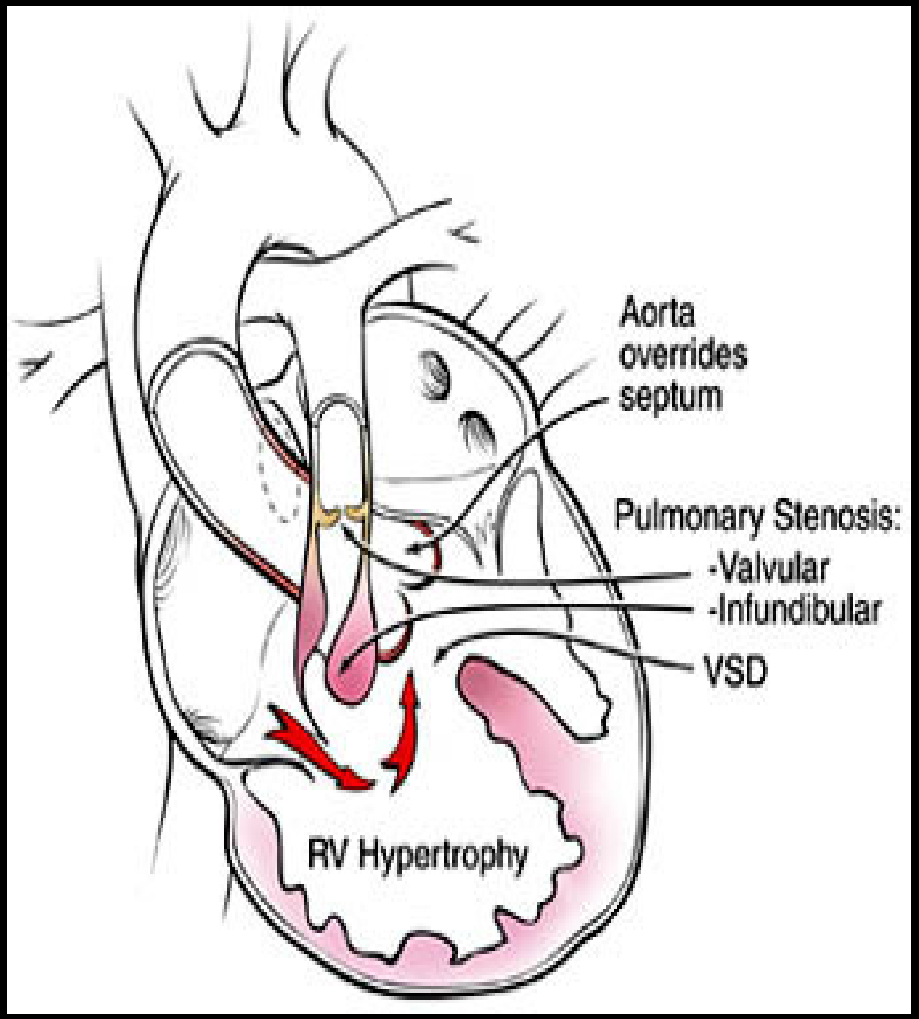
Treatment

- Drugs – increase pulm blood flow/Sa02
- Complete surgical repair: VSD closure and relief of RVOT obstruction
- Blalock-Taussig operation: SCA-PA
- Potts operation: DA-LPA
- Waterston-Cooley: AA-RPA
- Total surgical correction with patch closure of the VSD and relief of the ventricular outflow obstruction is preferred

Hypercyanotic "tet" spell

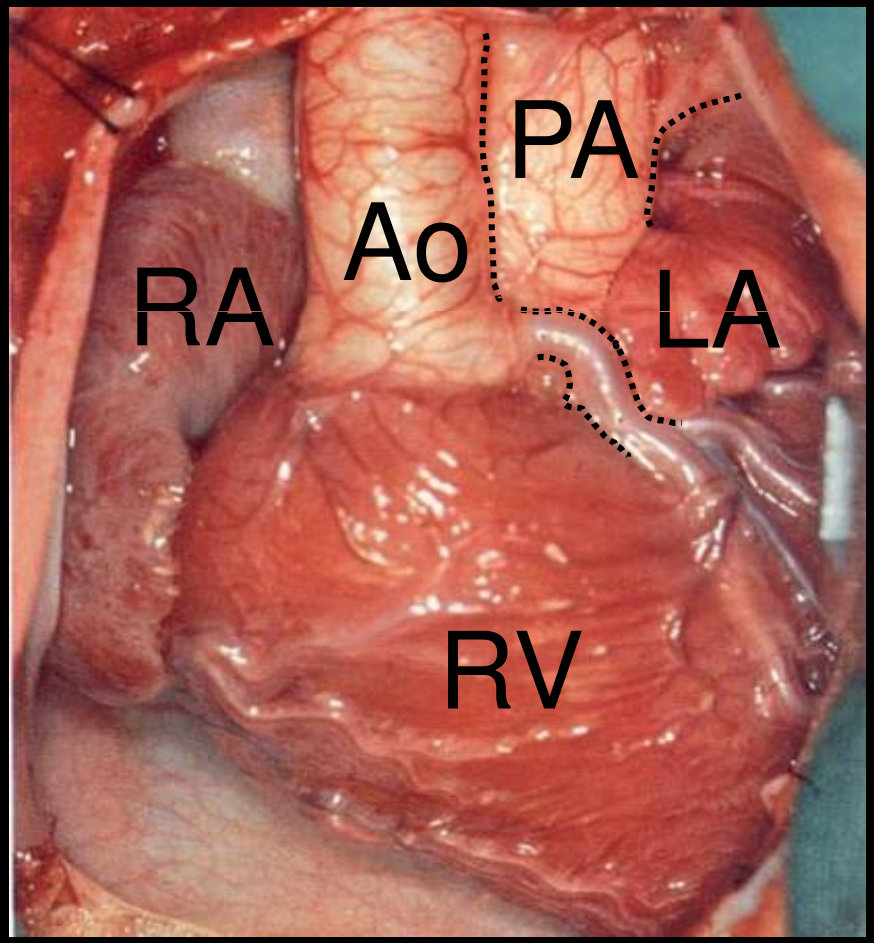
- Paroxysmal hypoxemia due to acute change in balance between PVR and SVR
- ↓ SVR causes an increase in R→ L shunt, increasing cyanosis
- ↓ SVR (hot bath, fever, exercise)
- Agitation → dynamic subpulmonic obstruction
- Life-threatening if untreated
- Hypercyanotic episodes are characterized by paroxysms of hyperpnea, prolonged crying, intense cyanosis, and decreased / absent intensity of the murmur of pulmonic stenosis.
- If left untreated, may result in syncope, seizure, stroke, or death

Management of "tet" spell:
goal is to ↑ SVR and ↓ PVR

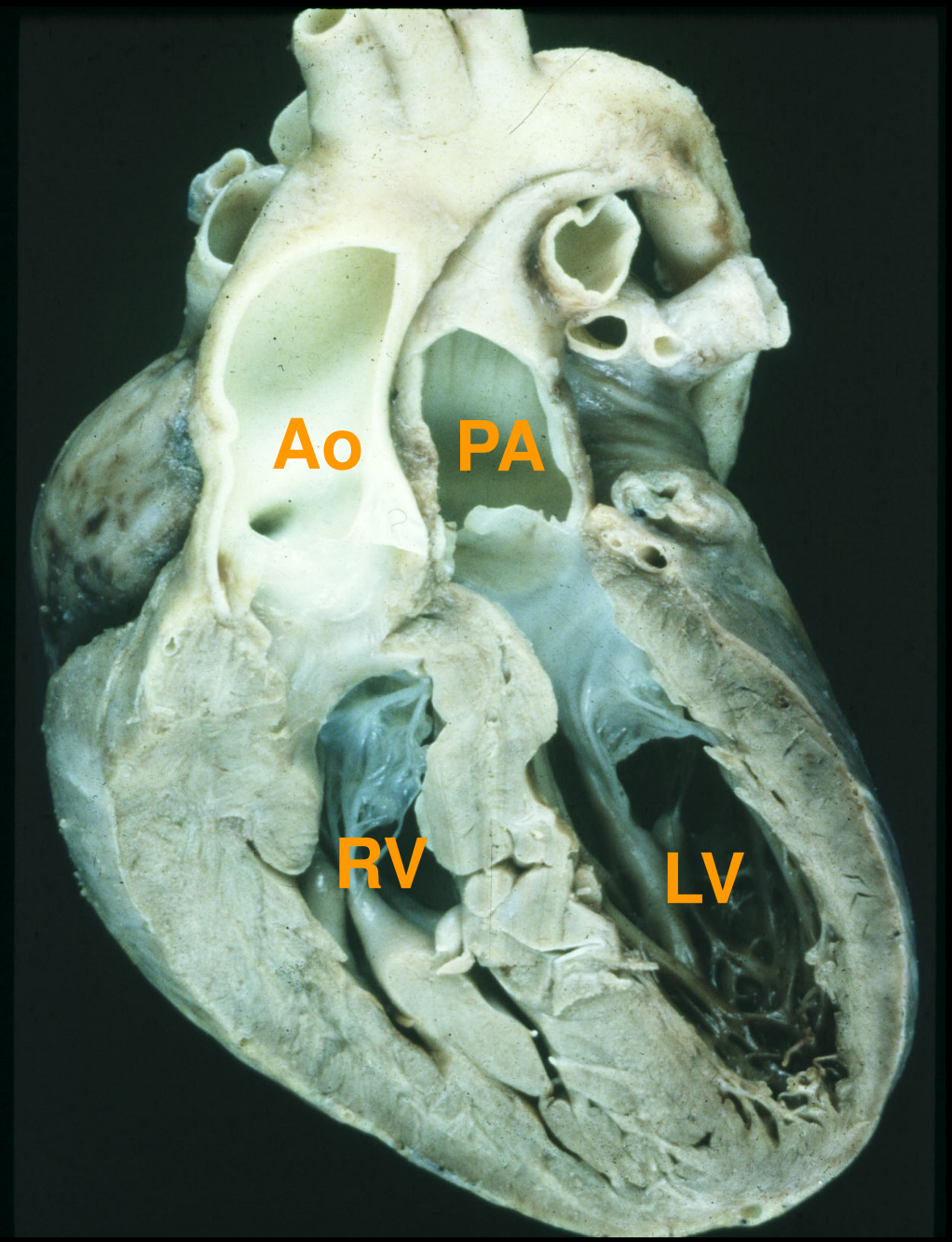


1. Knee-chest position (↑ SVR)
2. Supplemental O_2
3. Fluid bolus i.v. (↑ SVR)
4. Morphine i.v. (↓ agitation, ↓ dynamic RVOT obstruction)
5. $NaHCO_3$ to correct metabolic acidosis (↓ PVR)
6. Phenylephrine to ↑ SVR
7. β-blocker to ↓ dynamic RVOT obstruction

D-transposition of great arteries



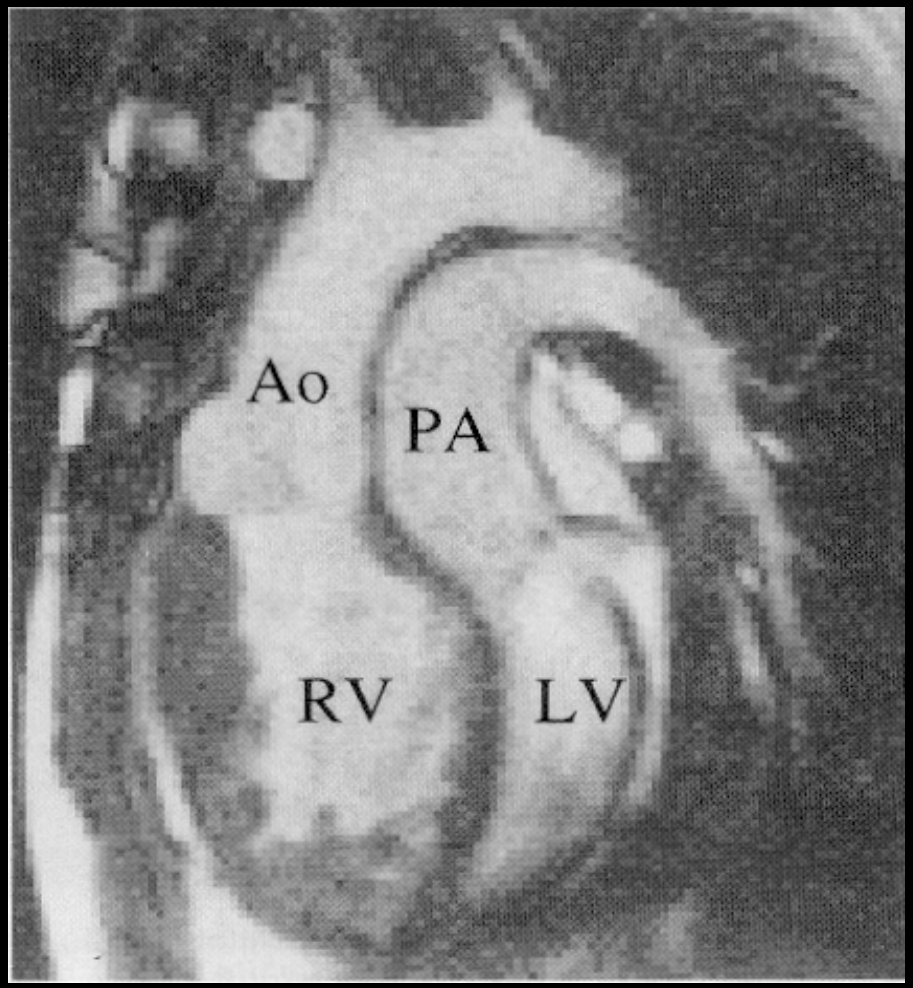
- 5% of all congenital heart disease
- Most common cause of cyanosis in neonate
- Second most common in infants
- Male:female 2:1



D-Transposition of the Great Arteries

- Ao is anterior, arises from right ventricle
- PA posterior, arises from left ventricle
- Systemic venous (blue) blood returns to RV and is ejected into aorta
- Pulm venous (red) blood returns to LV and is ejected into PA

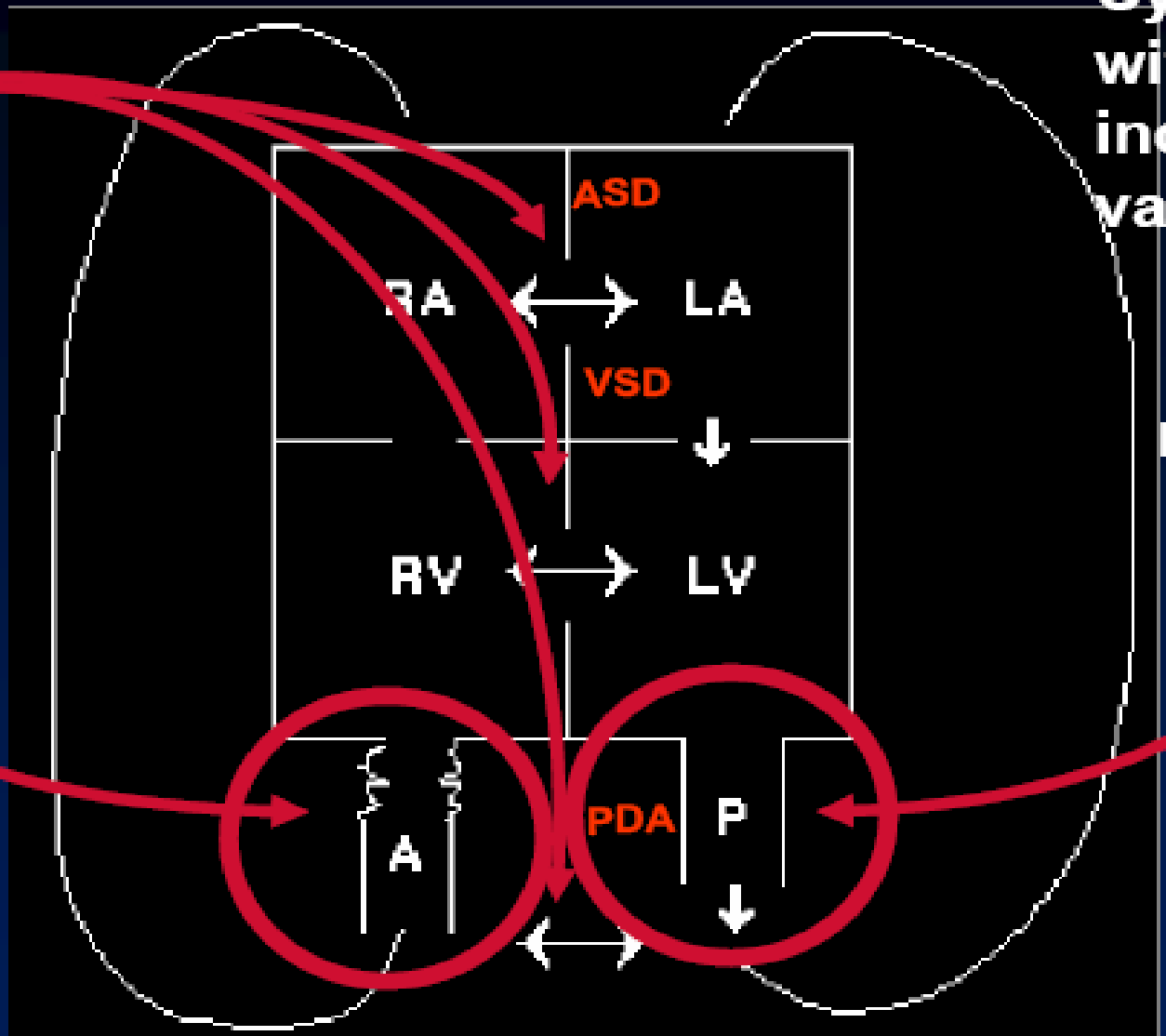
D-transposition of great arteries



- Systemic and pulmonary circulations are in parallel, rather than in series
- Mixing occurs at atrial and ductal levels
- Severe, life-threatening hypoxemia

Obligatory shunt since there are 2 separate circulations

Cyanotic with increased vasculature



Aorta arises from pulmonic infundibulum

PA arises from anatomic left ventricle

Transposition of the Great Vessels (D-Trans)

- Presence or absence of associated cardiac anomalies defines the clinical presentation and surgical management of a patient with TGA.

  The primary anatomic subtypes are
- (1) TGA with intact VS
- (2) TGA with VSD
- (3) TGA with VSD and LVOTO
- (4) TGA with VSD and pulmonary vascular obstructive disease
- In one third patients with TGA, the coronary artery anatomy is abnormal

clinical course and manifestations depend on extent of inter circulatory mixing and presence of associated anatomic lesions

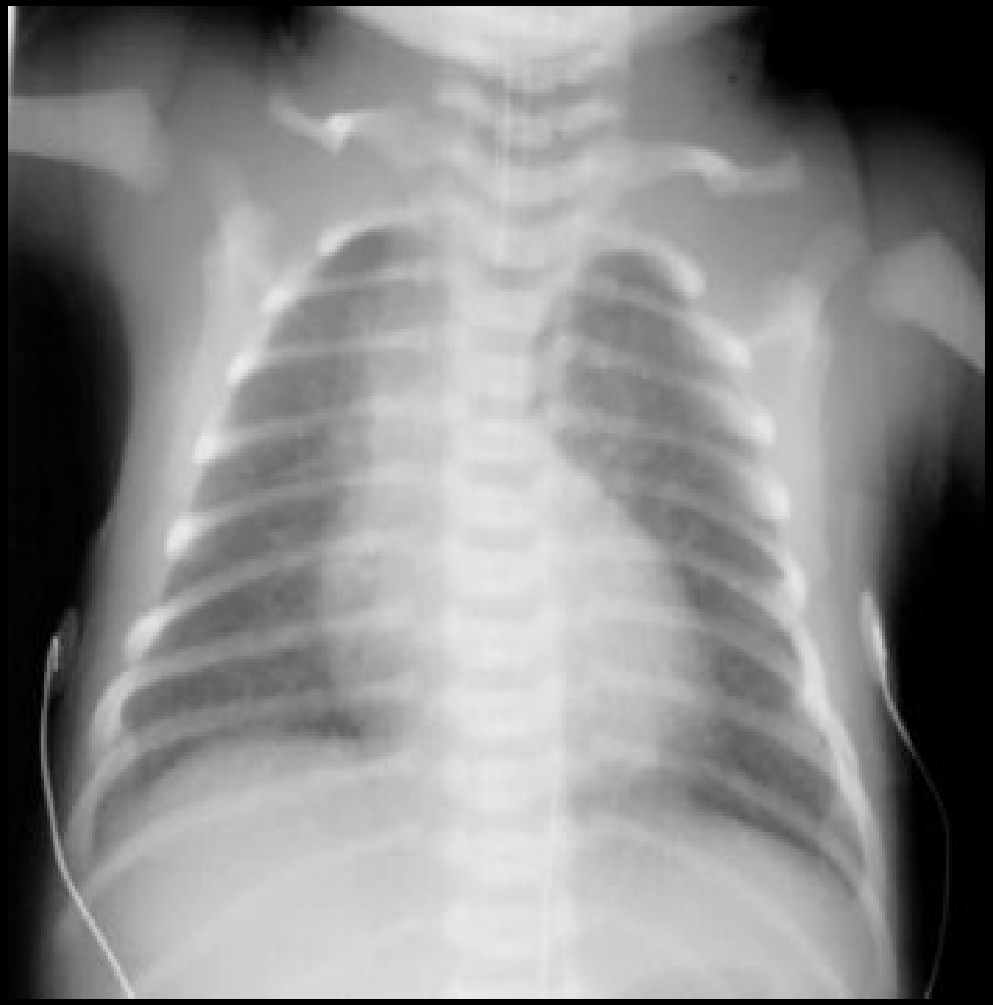
TGA with intact VS: Prominent and progressive cyanosis within first 24 hours of life

- TGA with large VSD
  - Infants may not initially manifest symptoms of heart, although mild cyanosis is often noted.
  - Signs of CHF may become evident over the first 3-6 weeks
- TGA with VSD and LVOT obstruction
  - Infants often present with extreme cyanosis at birth, proportional to the degree of left ventricular (pulmonary) outflow tract obstruction.
- TGA with VSD and pulmonary vascular obstructive disease
  - Progressively advancing pulmonary vascular obstructive disease can prevent this rare subgroup of patients from developing symptoms of congestive heart failure, despite a large ventricular septal defect.

- Newborns with TGA are usually well developed, without dysmorphic features.
- Physical findings at presentation depend on the presence of associated lesions.

- TGA with intact VS
  - progressive central cyanosis
  - Other unremarkable
- TGA with large VSD
  - Cyanosis mild initially, more apparent with stress or crying
  - increased RV impulse, prominent grade 3-4/6 holosystolic murmur, S3, mid-diastolic rumble, and gallop rhythm.
- TGA with VSD and LVOT obstruction
  - Cyanosis prominent at birth, findings similar to tetralogy of Fallot
  - A single, or narrowly split, diminished second heart sound and a grade 2-3/6 systolic ejection murmur may be present
- TGA with ventricular septal defect and pulmonary vascular obstructive disease
  - Cyanosis is usually present and can progress despite palliative therapy
  - No murmur (despite the ventricular septal defect)
  - second heart sound is often single, with increased intensity
  - In later childhood or adolescence, a high-pitched, blowing, early decrescendo diastolic murmur of PI and blowing apical murmur of MI

d-TGA CXR: “egg on a string”



- Narrow mediastinum due to anterior-posterior orientation of great arteries and small thymus
- Cardiomegaly is present with increased pulmonary vascular markings

TGA Treatment

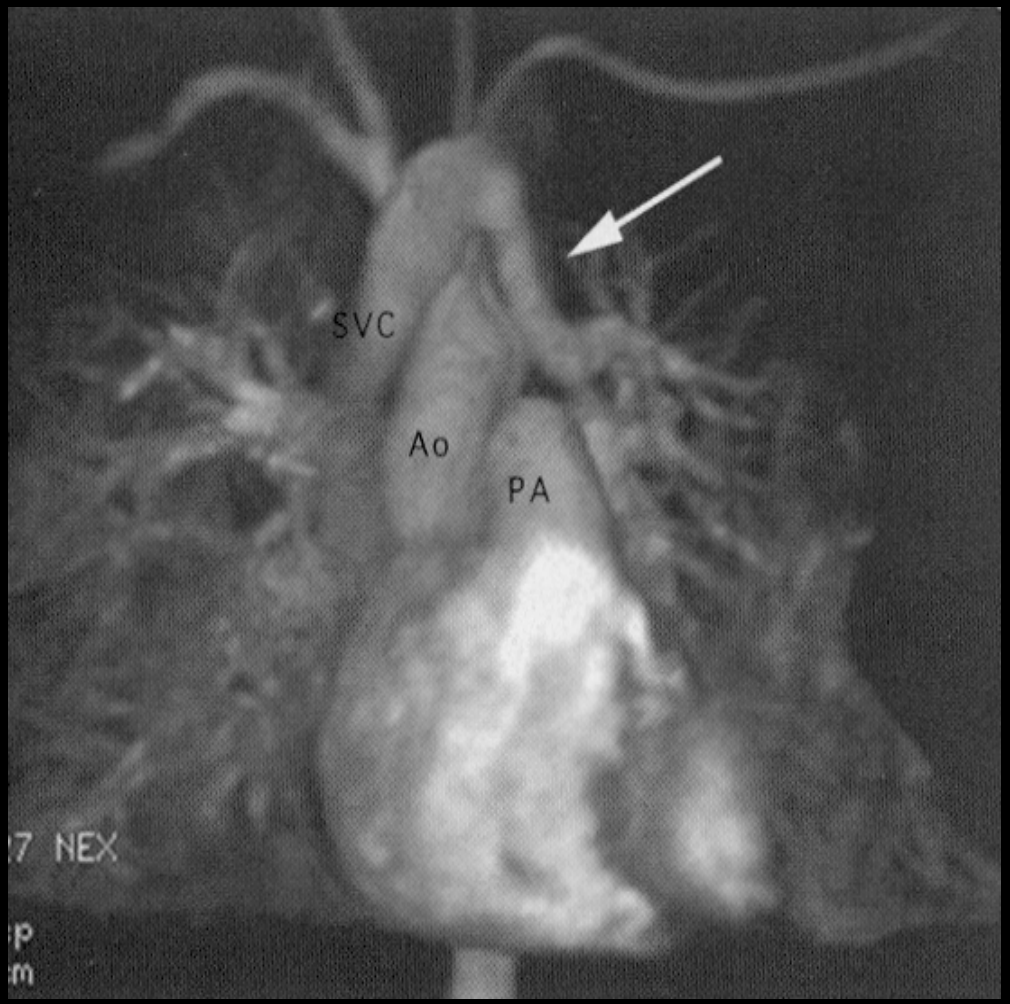
- Maintaining ductal patency with continuous IV PG E1 infusion to promote pulmonary blood flow, increase left atrial pressure, and promote left-to-right intercirculatory mixing at the PDA level.
- Balloon atrial septostomy in severely hypoxemic patients with an inadequate atrial level communication and insufficient mixing. (Rashkind procedure)
- Metabolic acidosis should be corrected
- Ultimately, the patient requires surgical repair or palliation early in life

Surgical management of d-TGA: Arterial switch procedure

The arterial trunks are transected and "switched" to restore "normal" anatomy

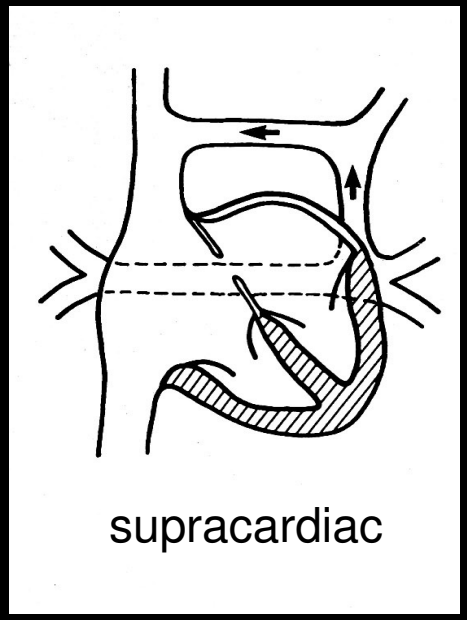
The coronary arteries are resected and re-implanted.

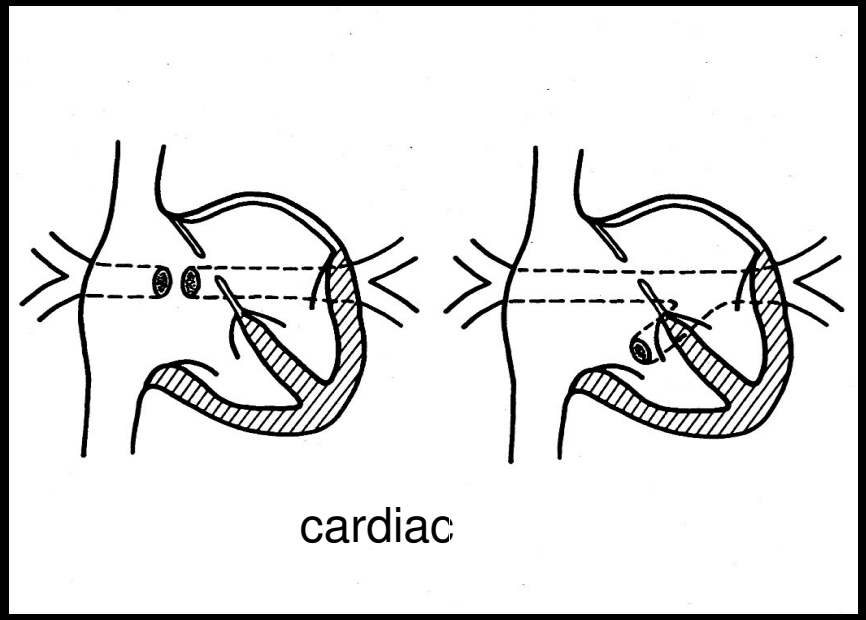
Total anomalous pulmonary venous return (TAPVR)

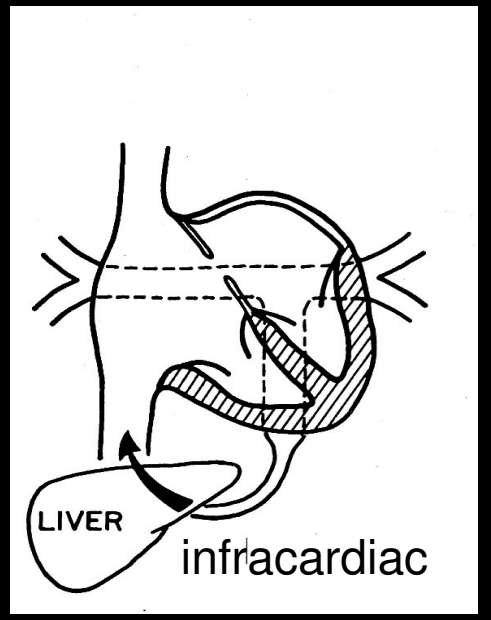


- Failure of pulm veins (PV) to fuse with developing left atrium
- PV drainage occurs through embryological remnants of systemic veins
- Incidence: rare

Venous connections in TAPVR







Supracardiac:
Ascending vertical vein
most common

Cardiac:
RA or coronary sinus

Infracardiac:
Descending vein
to portal system

Clinical manifestation of TAPVR

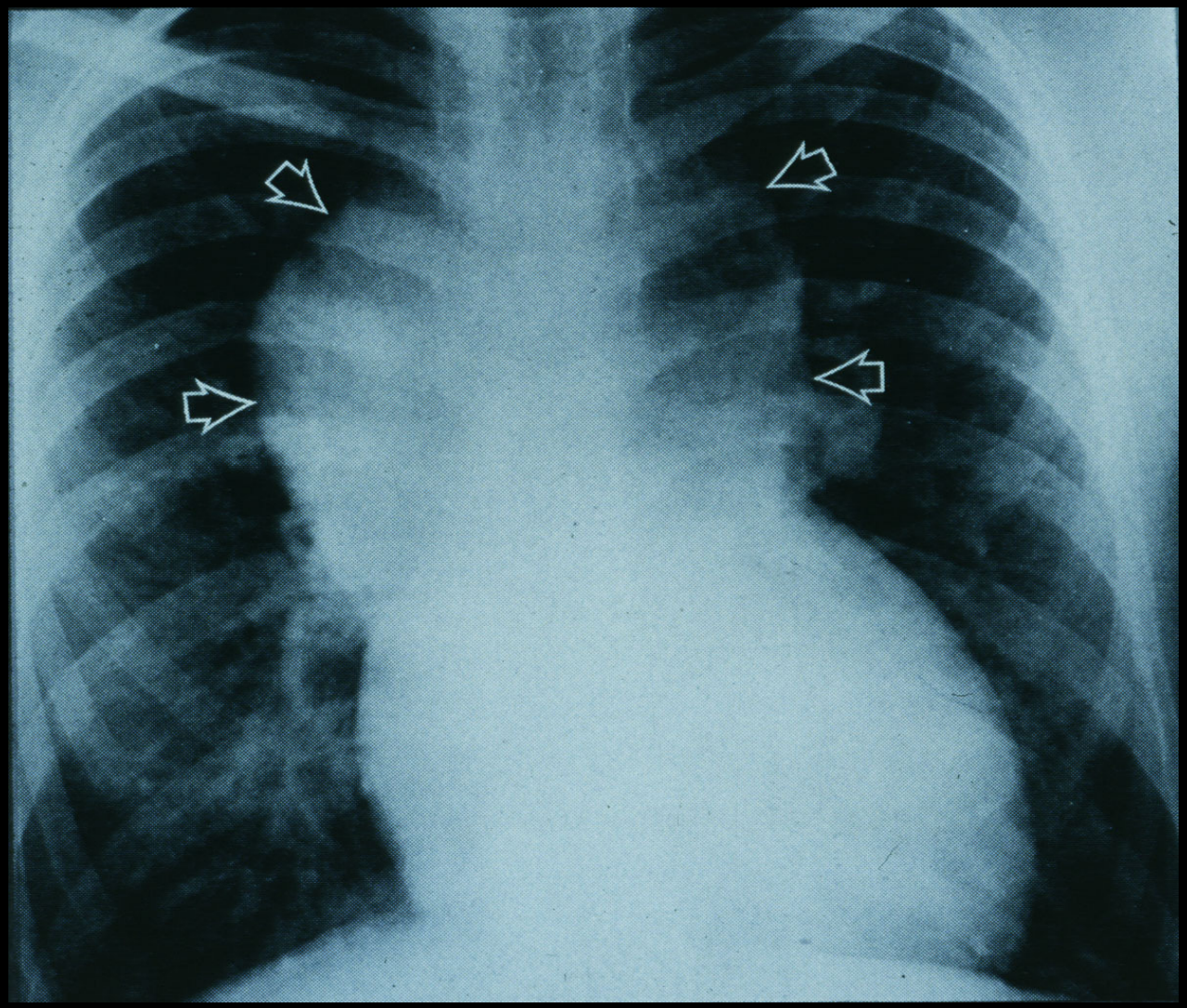
Obstructed (infra cardiac)

- Severe pulmonary edema, cyanosis, shock
- Surgical emergency

Unobstructed

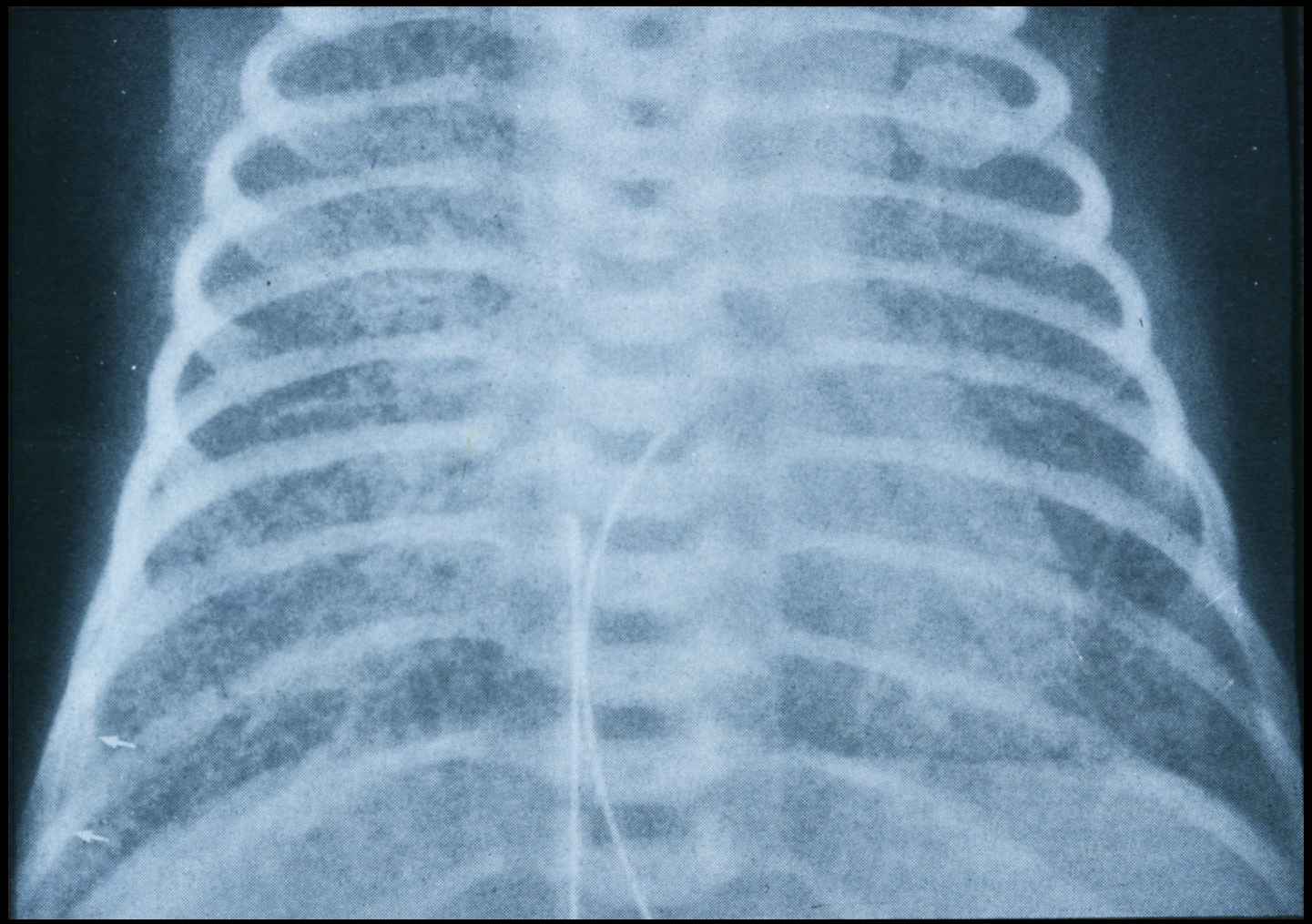
- Mild to moderate congestive heart failure and cyanosis
- Surgery in first 6 months

Supracardiac TAPVC - CXR

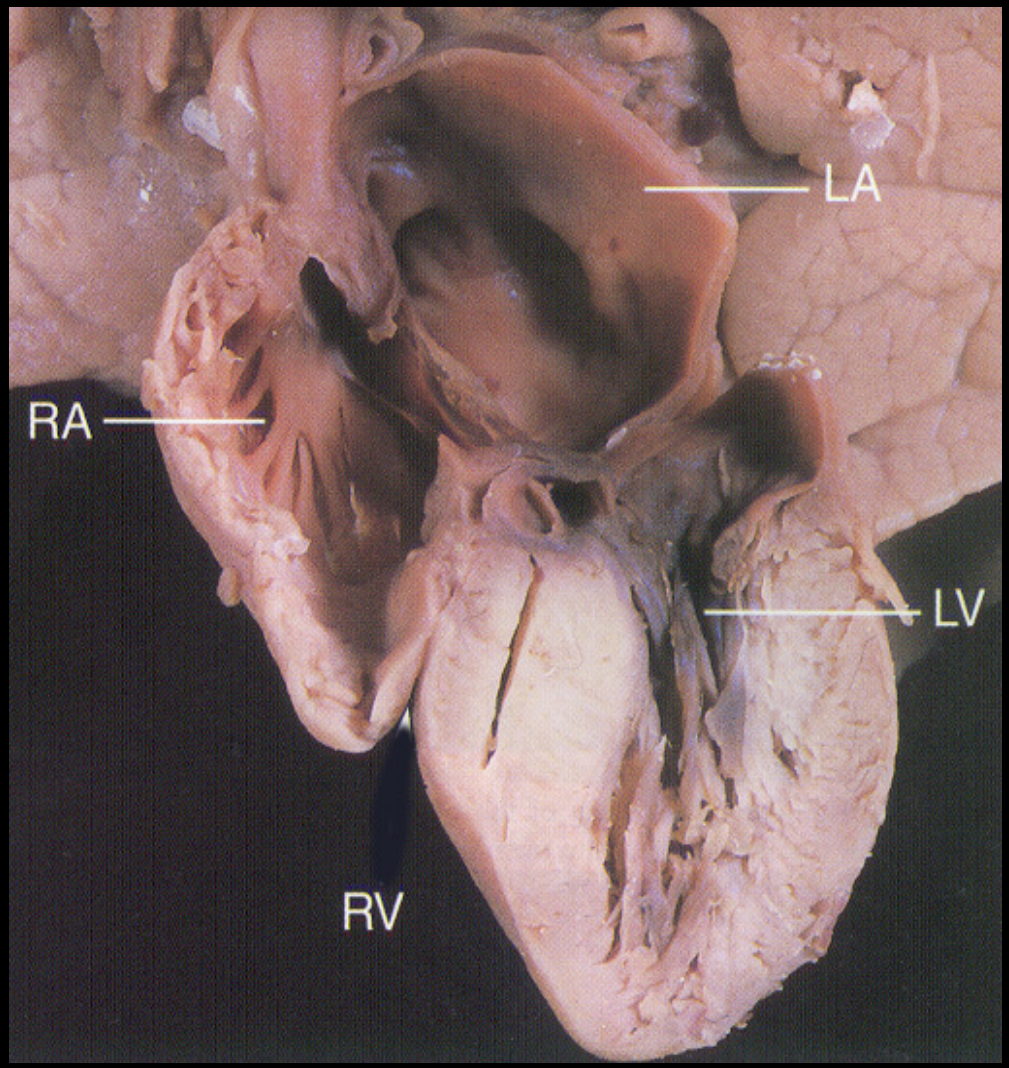


"Snowman" appearance secondary to dilated vertical vein, innominate vein and right superior vena cava draining all the pulmonary venous blood

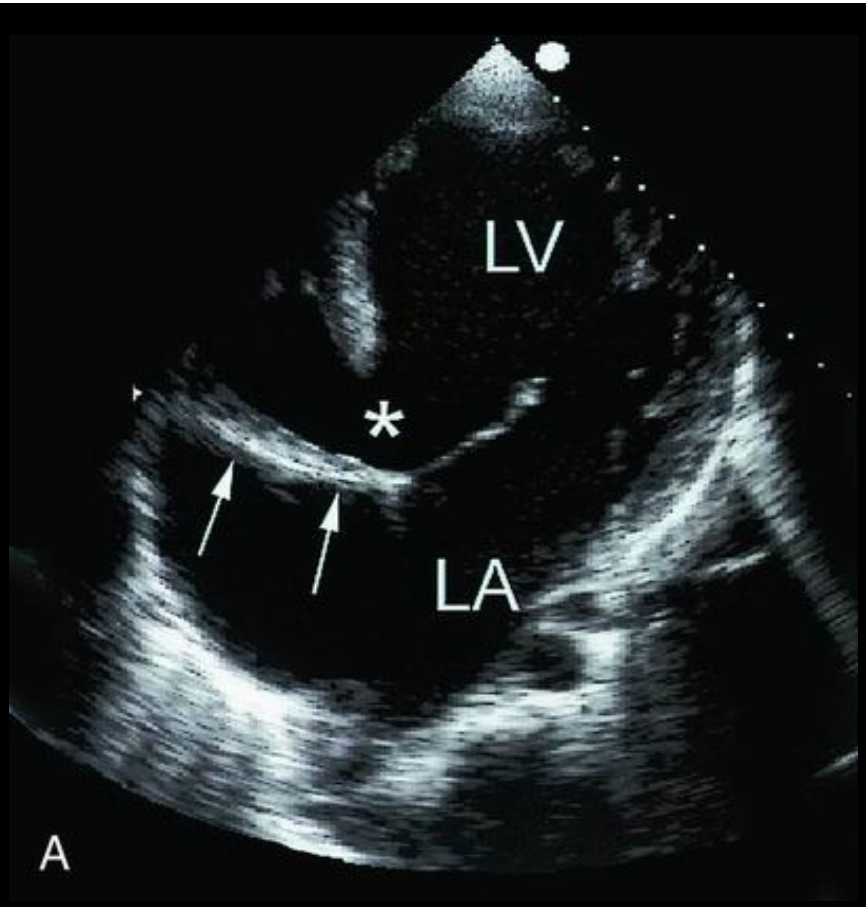
TAPVR - CXR
Infracardiac = Surgical Emergency

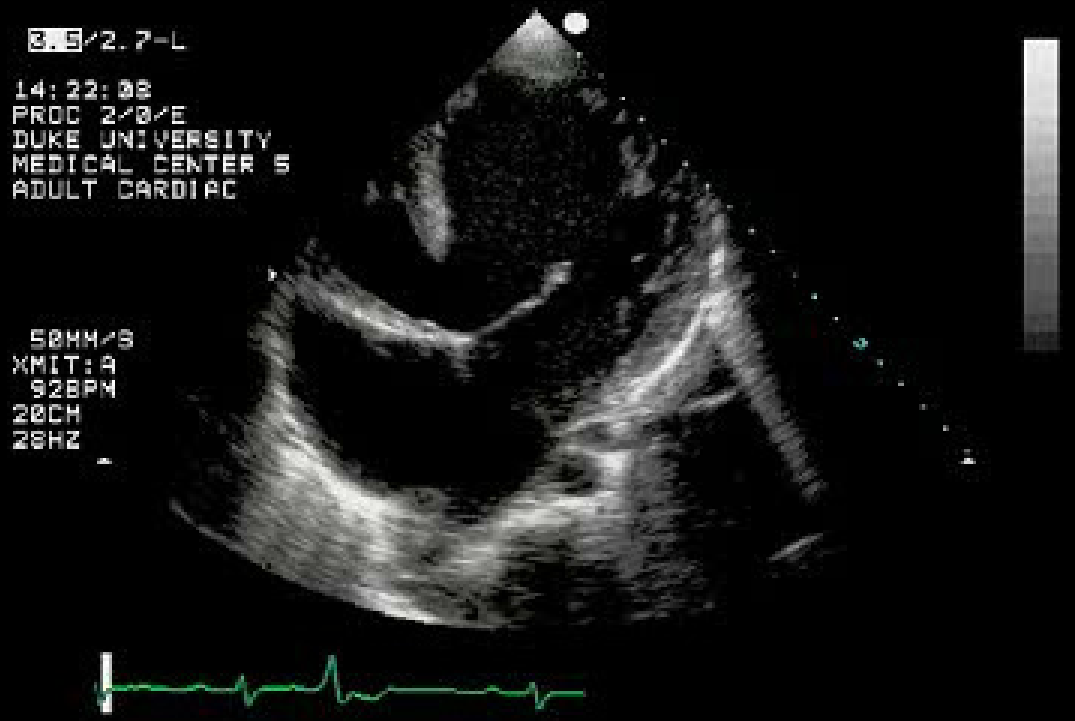


Tricuspid / Pulmonary atresia



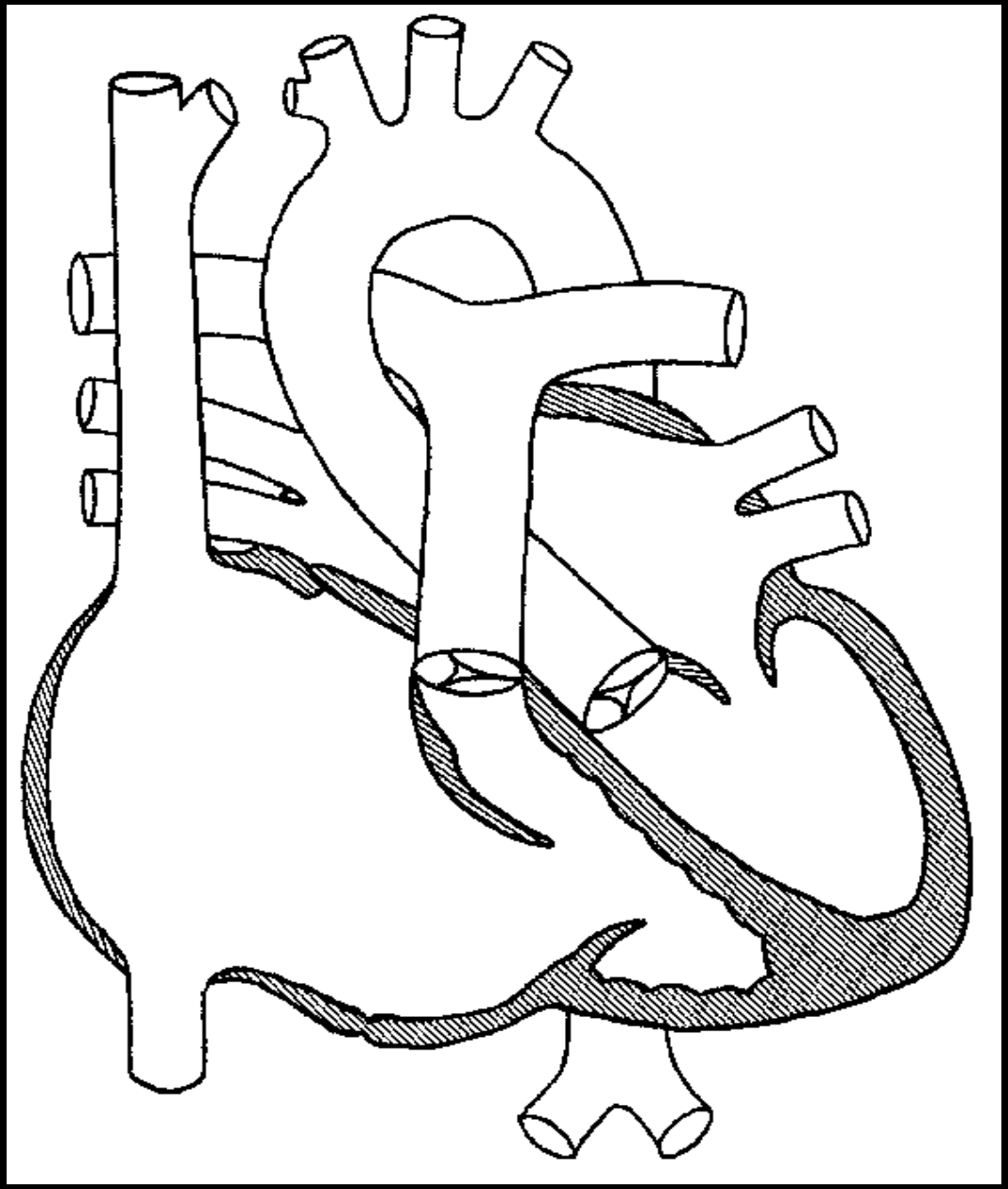
- Tricuspid/ Pulmonary valve may be normal/ stenotic/ atretic
- Degree of cyanosis proportional to degree of pulmonary stenosis
- Pulmonary arteries often normal
- Hypoplastic RV (TA), RVH (PA)


LV
*
LA
A


3.5/2.7-L
14:22:08
PROC 2/0/E
DUKE UNIVERSITY
MEDICAL CENTER S
ADULT CARDIAC
50MM/S
XMIT:A
92BPM
20CH
2SHZ

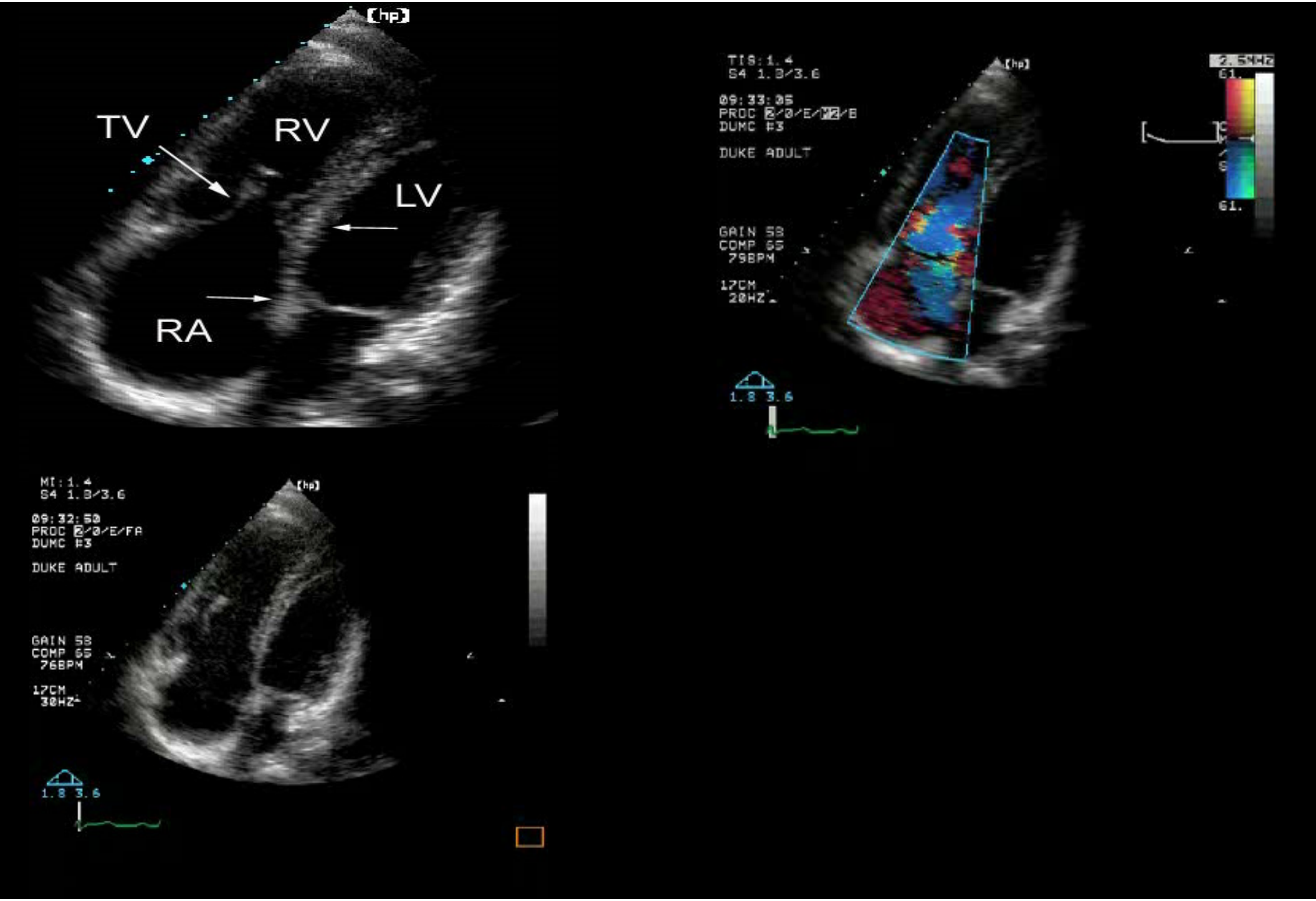
Tricuspid / Pulmonary atresia

- Ductal-dependent lesion
- Requires PGE_1 to maintain oxygenation
- Therapy directed at opening atretic valve in cath lab or surgery
- Prognosis depends upon size and compliance of hypoplastic RV

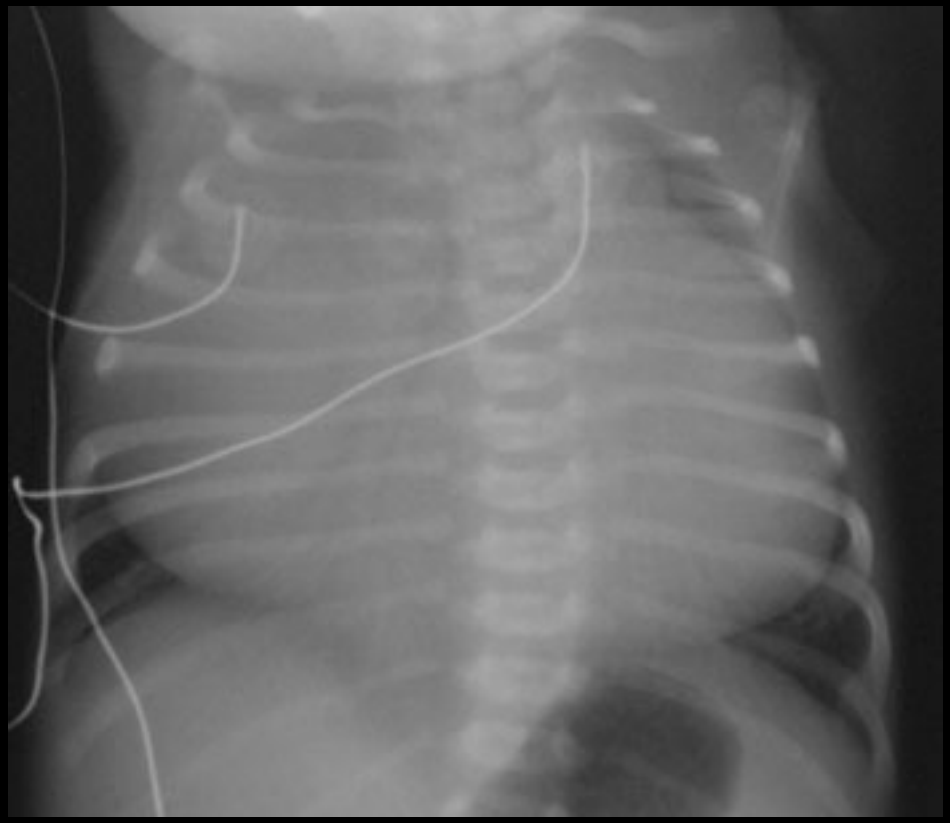


Ebstein's anomaly

The tricuspid valve is abnormal and inserts well down into the RV. There is often severe trisuspid regurgitation, which can lead to death in the fetus or infant. Usually also with ASD so right-to-left flow results in cyanosis.

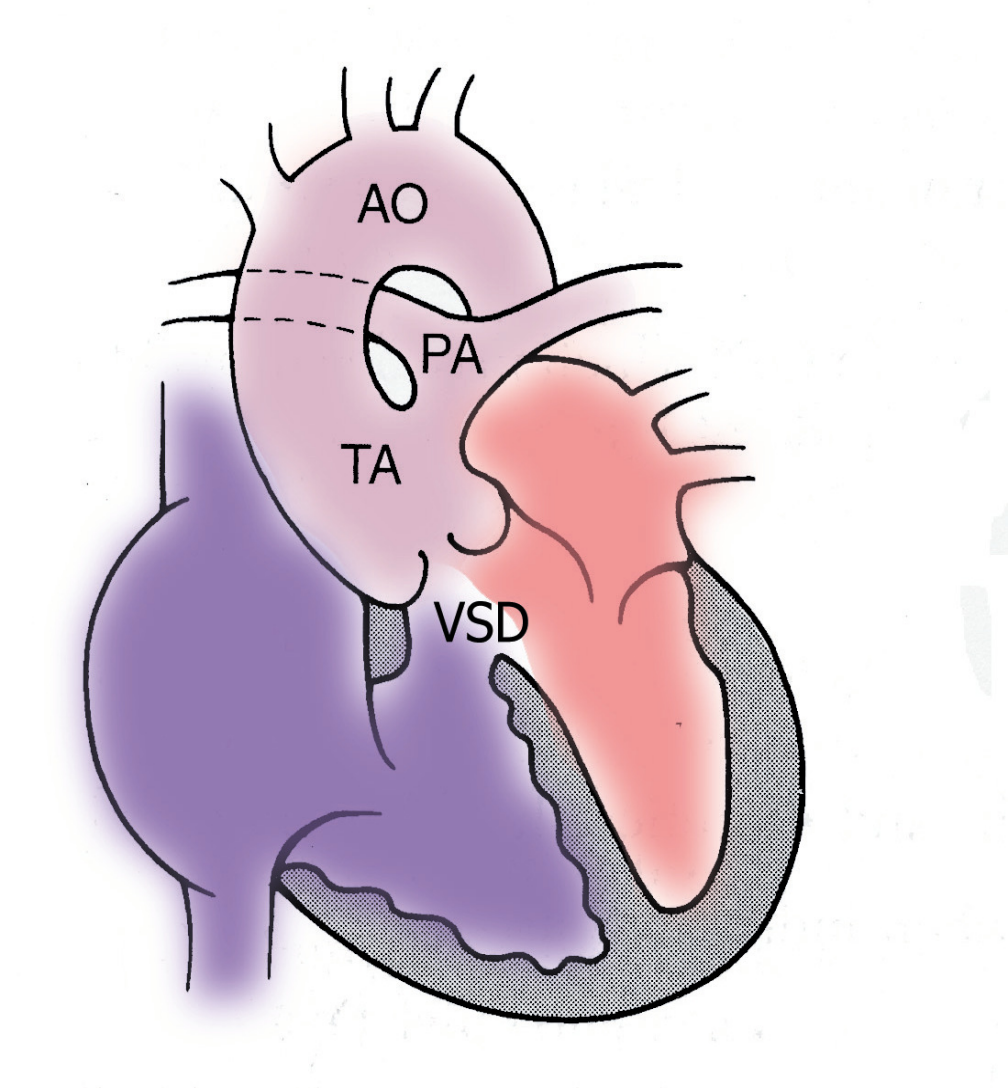

TV
RV
LV
RA
TIS:1.4
S4 1.8/3.6
09:33:05
PROC 2/0/E/V2/B
DUMC #3
DUKE ADULT
GAIN 58
COMP 65
79BPM
17CM
20HZ
1.8 3.6
MI:1.4
S4 1.8/3.6
09:32:50
PROC 2/0/E/FA
DUMC #3
DUKE ADULT
GAIN 58
COMP 65
76BPM
17CM
30HZ
1.8 3.6

Ebstein's malformation of TV
(Wall to wall heart)



- Massive RA dilation due to severe TR
- R to L shunt at atrial level causes cyanosis
- Degree of cyanosis related to size and compliance of functional RV
- Cyanosis usually decreases as PVR falls shortly after birth

Truncus arteriosus



- Aorta, pulmonary arteries, and coronary arteries arise from single vessel
- Truncus sits over large ventricular septal defect
- Failure of septation of embryonic truncus
- Uncommon (1.4% CHD)

Thank you