

Identifying Congenital Heart Disease in Infancy

Jessica Mouledoux, MD, MSCI

Background: Congenital Heart Disease

- Most common type of congenital anomaly
- Overall prevalence about 1%
- **Critical congenital heart disease (CCHD)**
 - ¼ of all congenital heart disease
 - severe and life-threatening disease requiring surgical or catheter intervention within the first month of life
 - **risk of morbidity and mortality increases when there is a delay in diagnosis**

Objectives

- Identification of CHD in the nursery
 - Prenatal evaluation
 - Clinical exam
 - Role of pulse ox screening
- Signs and symptoms of CHD in infancy
 - Left heart obstructive lesions
 - Hypoxic lesions
 - Lesions with pulmonary overcirculation

CHD in the Nursery

Diagnosis of CHD: Prenatal

- 18-20 wga OB ultrasound
- Four chamber view
- Identifies 50-60% of CCHD
- Commonly identifies:
 - Single ventricle lesions (hypoplastic left heart)
 - Significant chamber size discrepancy (Ebstein's anomaly)
- Maternal risk factors
 - Diabetes mellitus
 - Hypertension
 - Obesity
 - Phenylketonuria
 - Thyroid disorders
 - Epilepsy
 - Assisted reproductive technology
 - In utero infections

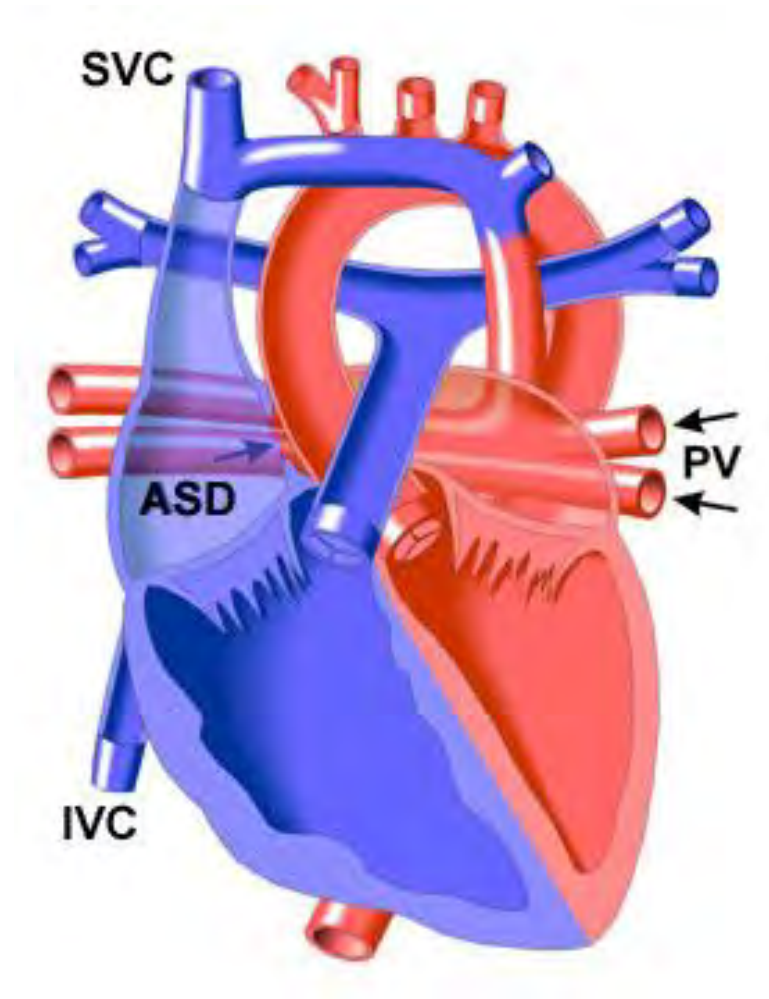


Diagnosis of CHD: Prenatal



Lesions “Missed” by Prenatal Ultrasound

- Outflow tract anomalies
 - Transposition of the great arteries
 - Truncus arteriosus
 - Pulmonary stenosis/atresia
 - Aortic stenosis
- Aortic arch anomalies
 - Coarctation of the aorta
 - Interrupted aortic arch
- Pulmonary vein anomalies
 - Total anomalous pulmonary venous return
- Septal defects
 - Atrial septal defects
 - Ventricular septal defects



Diagnosis of CHD: Postnatal

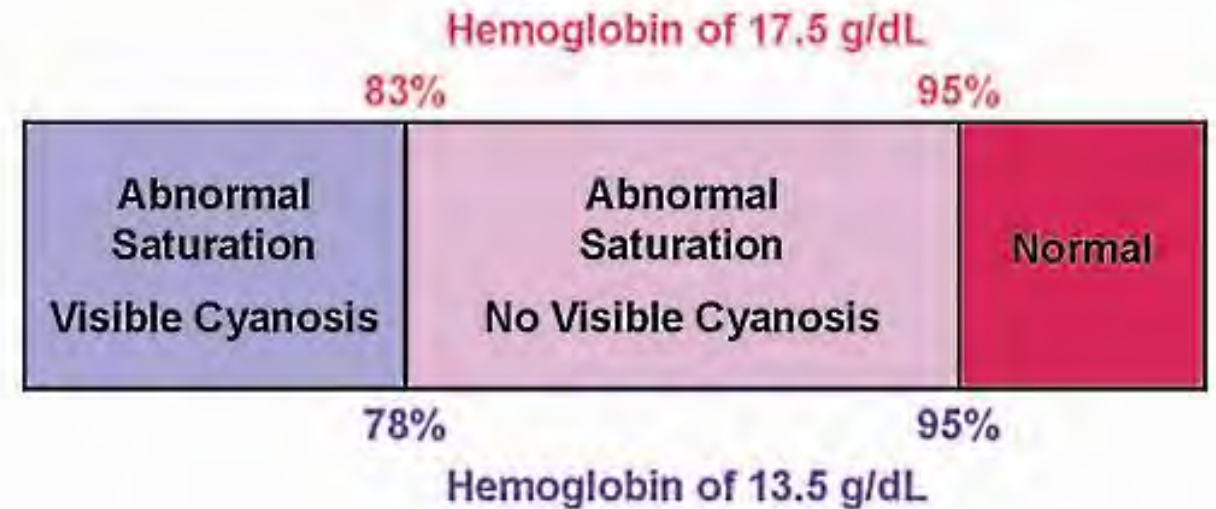
- Suspected due to signs or symptoms of CHD
 - Murmur
 - Cyanosis
 - Respiratory distress
 - Poor perfusion
- Identifies an additional 20-30% of CCHD
- ~20% CCHD undiagnosed by prenatal testing and neonatal exam

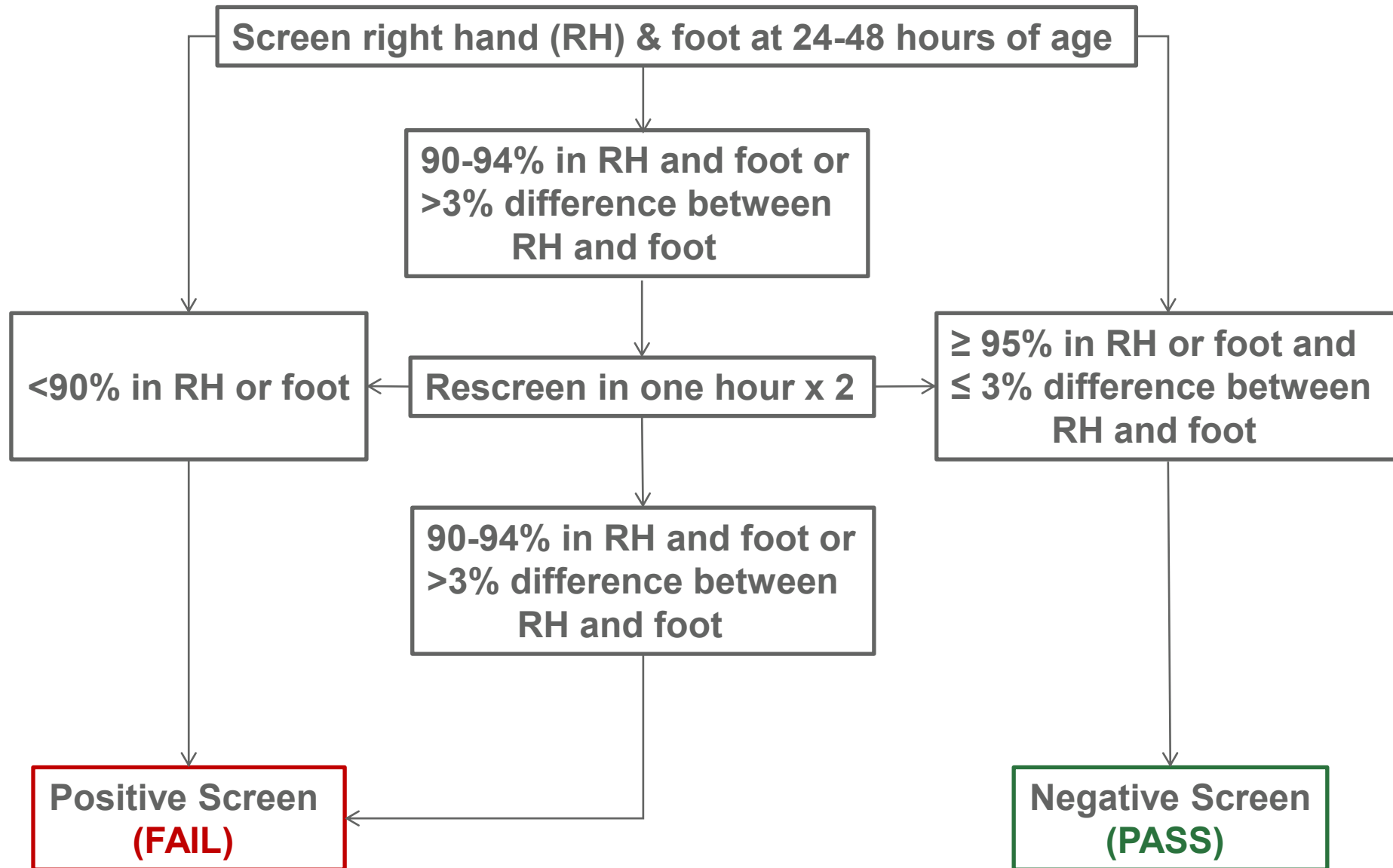


Hypoxemia & Pulse Oximetry in the Detection of Congenital Heart Disease

- Hypoxemia is a feature of many congenital heart defects
- Significant hypoxemia may result in obvious cyanosis (sat <80%)
- Subclinical hypoxemia is common
- Difficult in neonates with darker skin
- Pulse oximetry
 - Non-invasive assessment of oxygen saturation
 - Amount of oxygen dissolved in hemoglobin
 - Normal range >95%

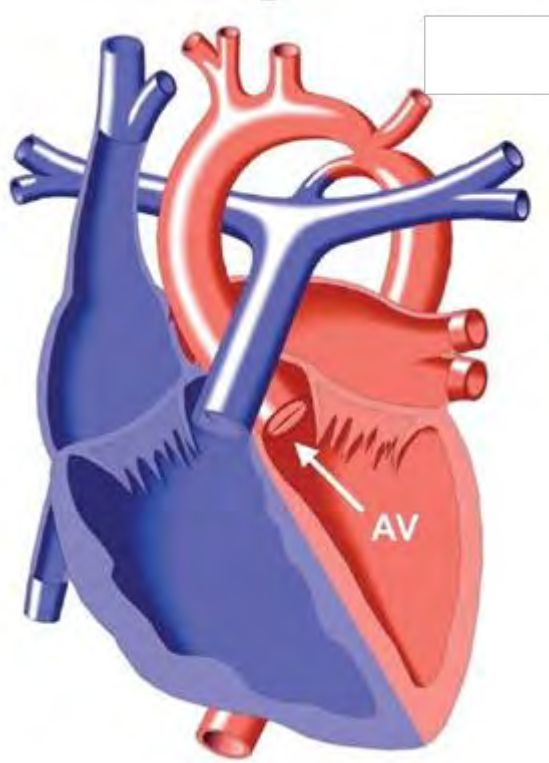
The Cyanotic Blind Spot



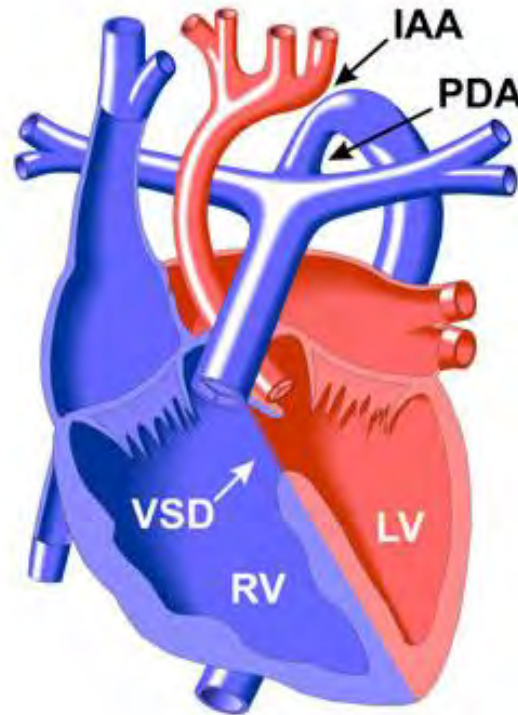


Presentation of Congenital Heart Disease in Infancy: Left Heart Obstruction

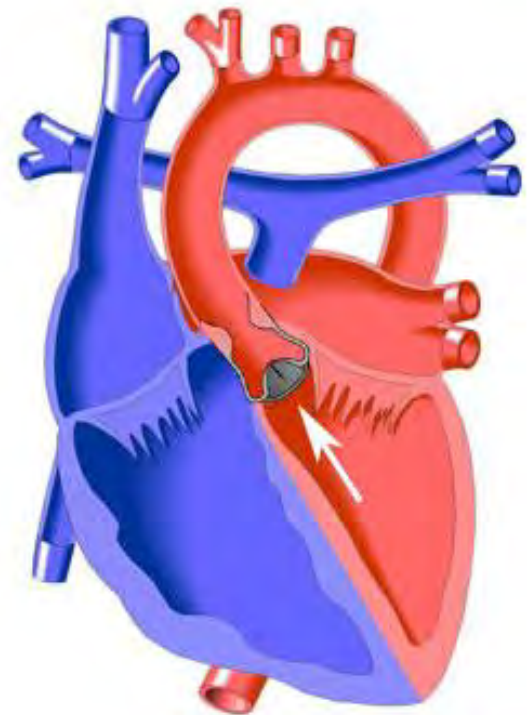
Left Heart Obstructive Lesions May be Missed by Pulse Oximetry



Coarctation of the Aorta



Interrupted Aortic Arch

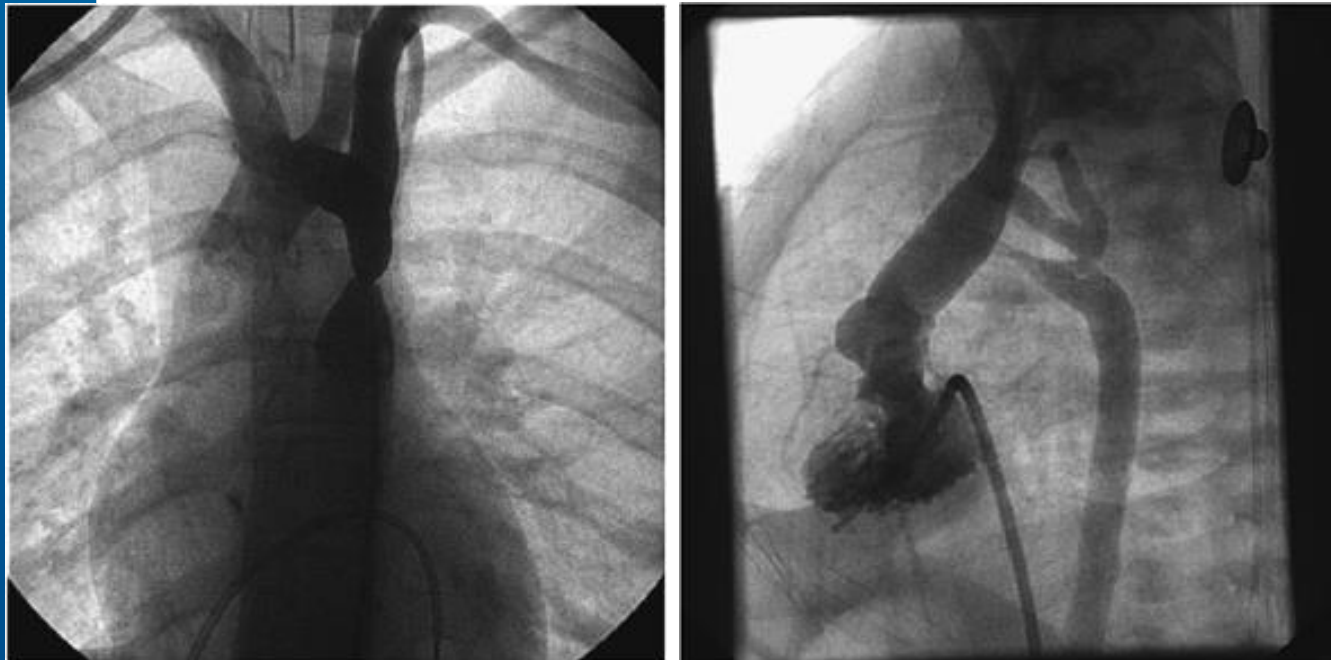
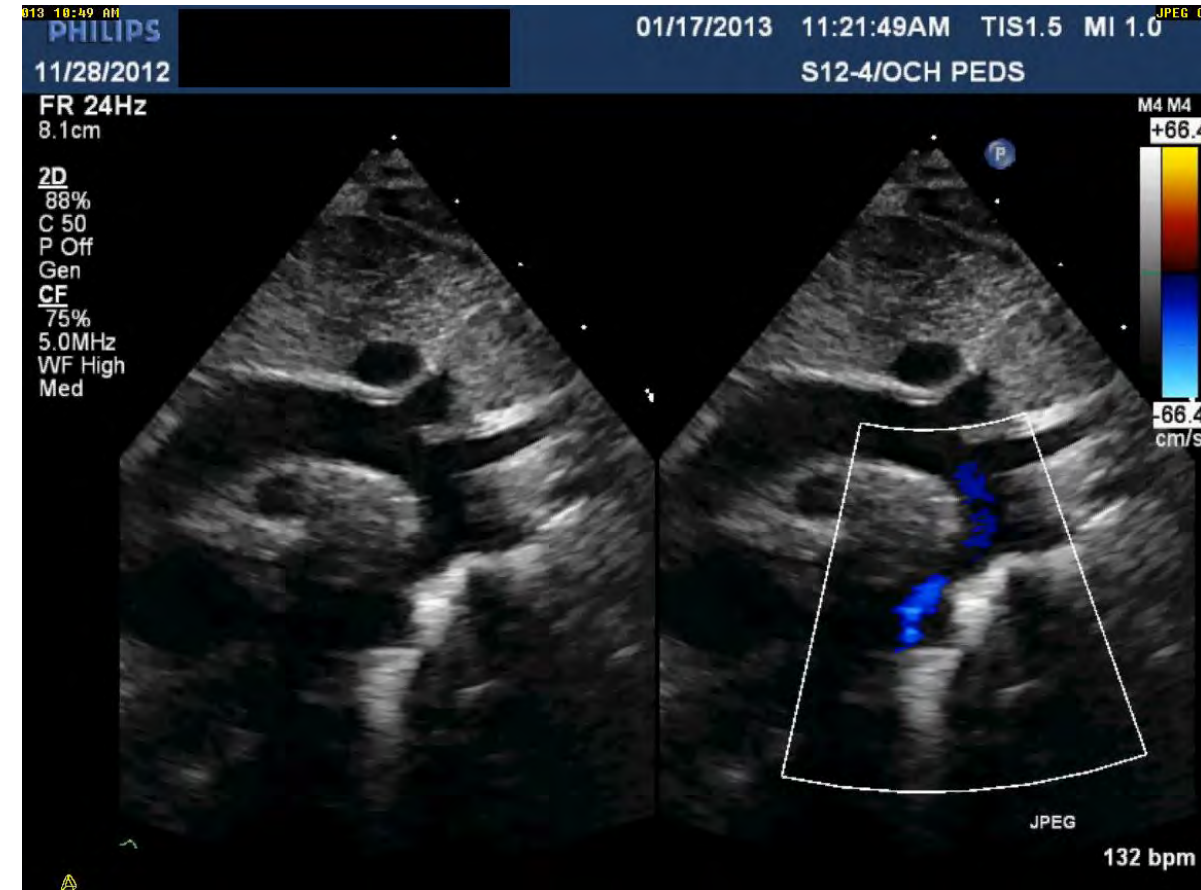


Aortic Stenosis

Coarctation of the Aorta

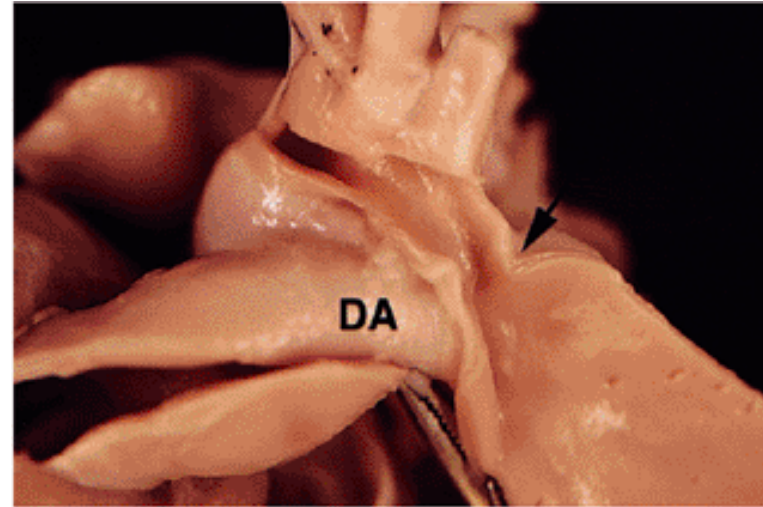
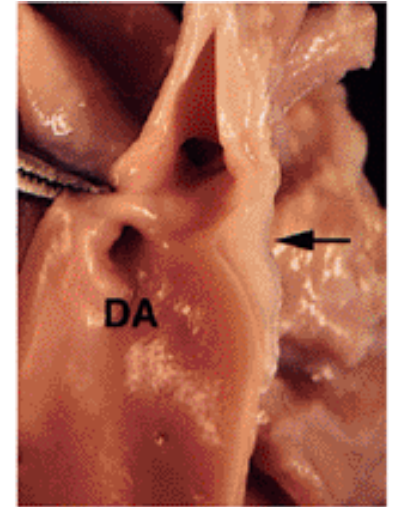
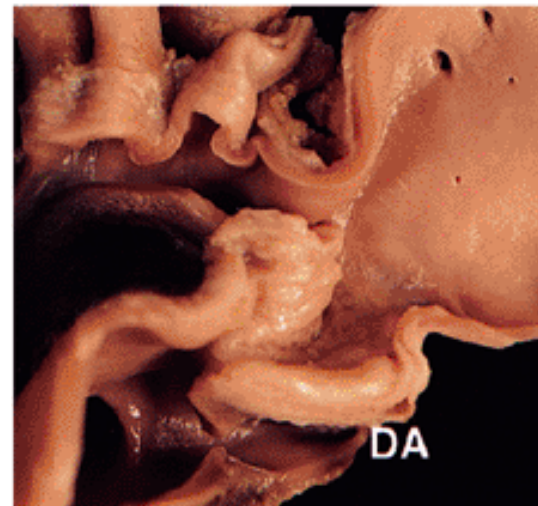
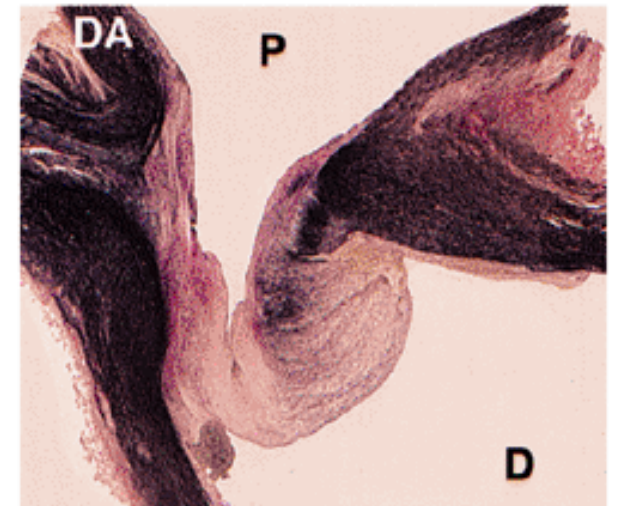
Definition: CoA

- Discrete stenosis of the upper thoracic aorta
- Juxtaductal – at the point of insertion of the ductus
- Long segment
- Diffuse hypoplasia of transverse arch



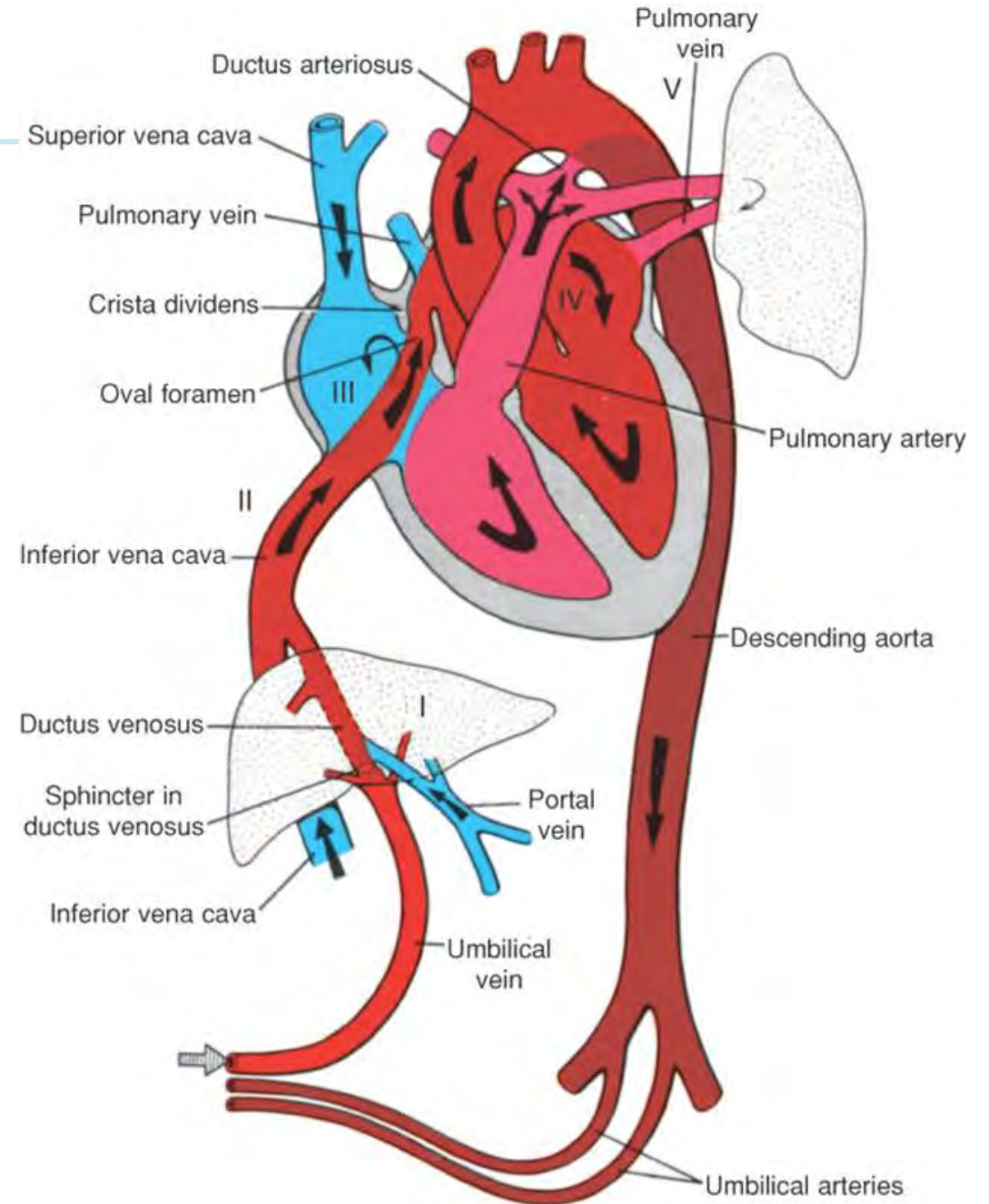
Pathophysiology: CoA

- Malformation of the aortic media
- Prominent posterior in-folding → posterior shelf
- Diffuse tubular hypoplasia
- Incidence:
 - 4/10,000 live births
 - 4-6% of CHD



Pathophysiology: CoA

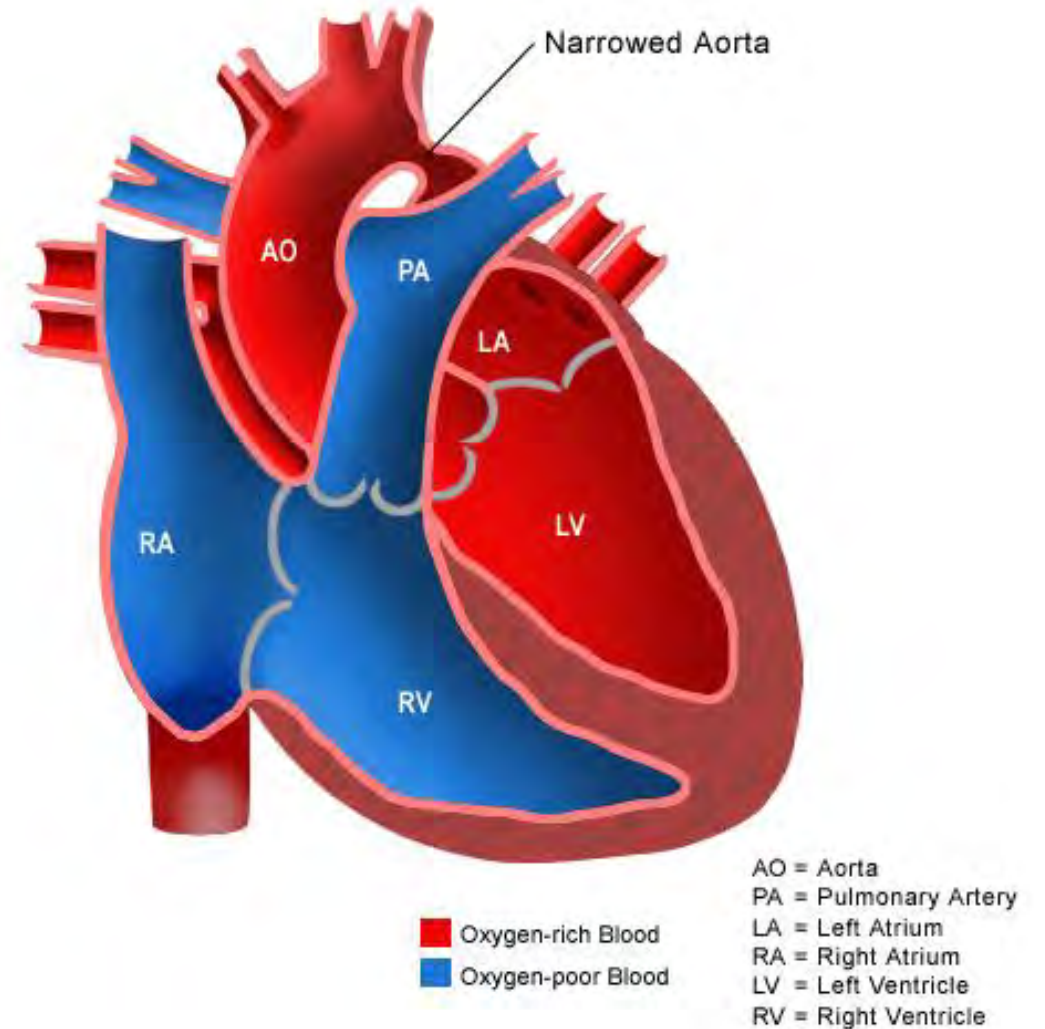
- Fetal circulation: Only 10% of fetal blood flow across the isthmus
- Postnatal: PVR decreases, PFO and PDA close → all cardiac output crosses isthmus
- Sudden increase in ascending aorta and LV pressure
- No time for LVH to develop
 - Depressed function → decreased stroke volume
 - Increased LVEDP → elevated left atrial pressure
 - Pulmonary venous congestion
- Low cardiac output → poor myocardial perfusion and acidosis



Presentation: Critical CoA

- Ductal dependent systemic circulation
- CHF with circulatory collapse
 - Cardiogenic shock and acidosis
 - Typically, 8-12 days of life
 - But can be later
 - Poor perfusion
 - Differential cyanosis due to right to left PDA shunt
 - Lethargy
 - Greyish discoloration
 - Multi-organ system dysfunction
 - Renal failure
 - NEC
- **Can mimic sepsis**

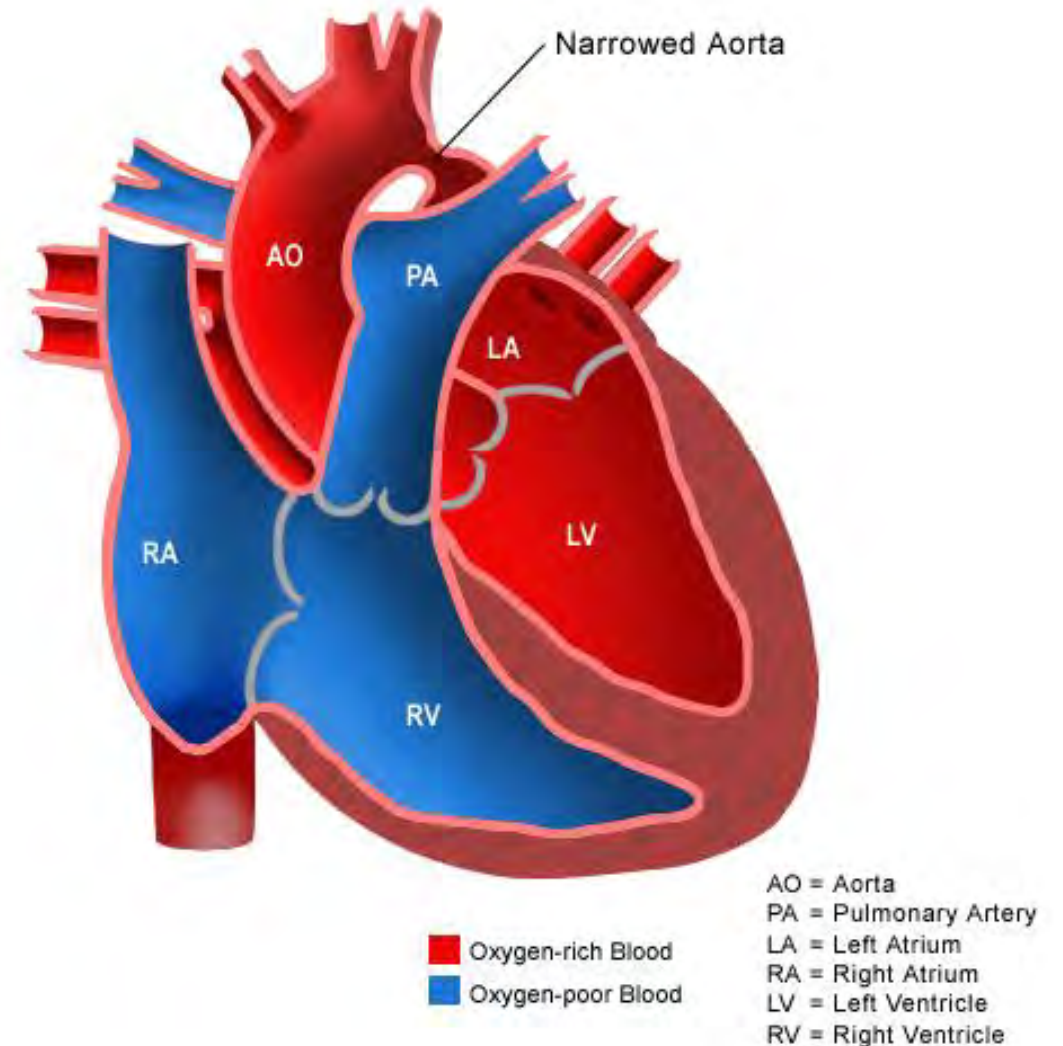
Coarctation of the Aorta



Presentation: CoA

- Diminished pulses
- Upper Ext BP > Lower Ext BP
- +/- Murmur (due to associated defects)
 - 30% with bicuspid aortic valve
- CHF due to poor cardiac output
 - Increased LVEDP and LA pressure = Pulmonary edema
 - Tachypnea
 - Wheezing, cough less common
 - **Can mimic bronchiolitis symptoms**
 - Poor feeding
 - FTT
- **Dysmorphic female = Turner's syndrome**

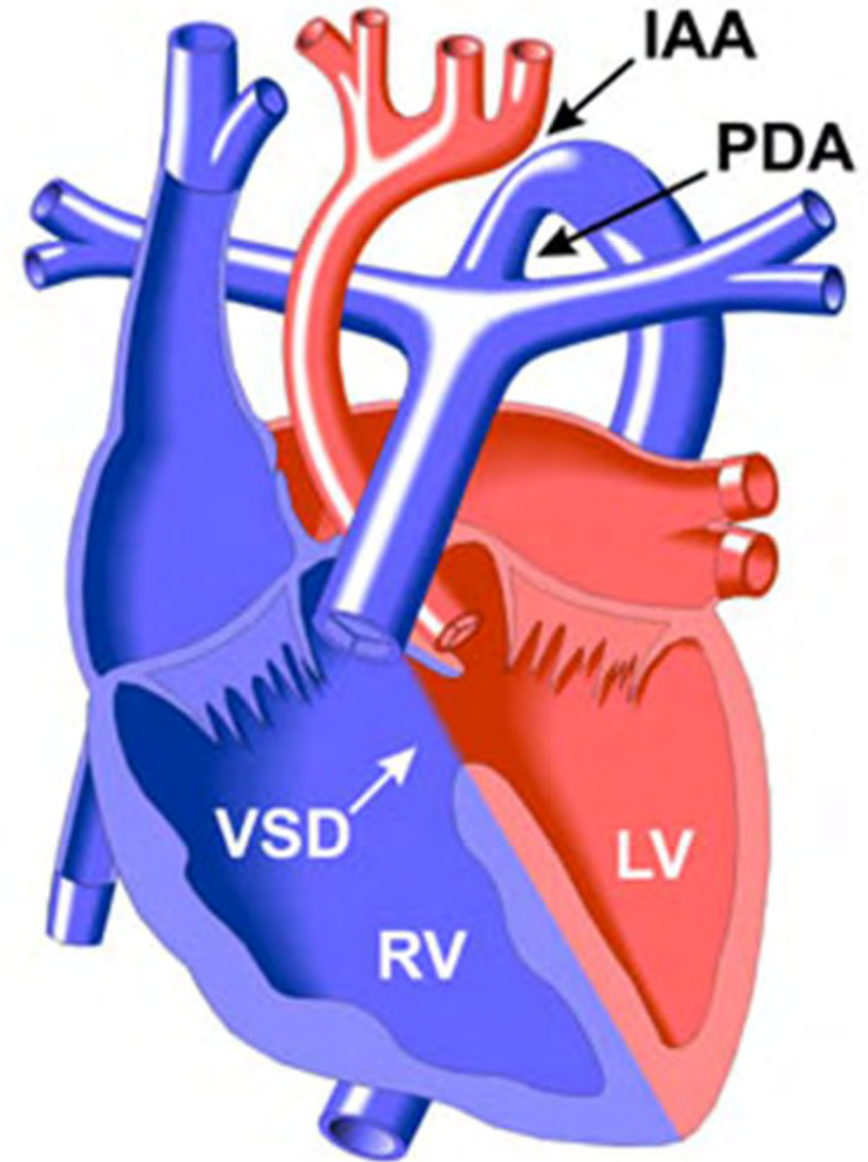
Coarctation of the Aorta



Interrupted Aortic Arch

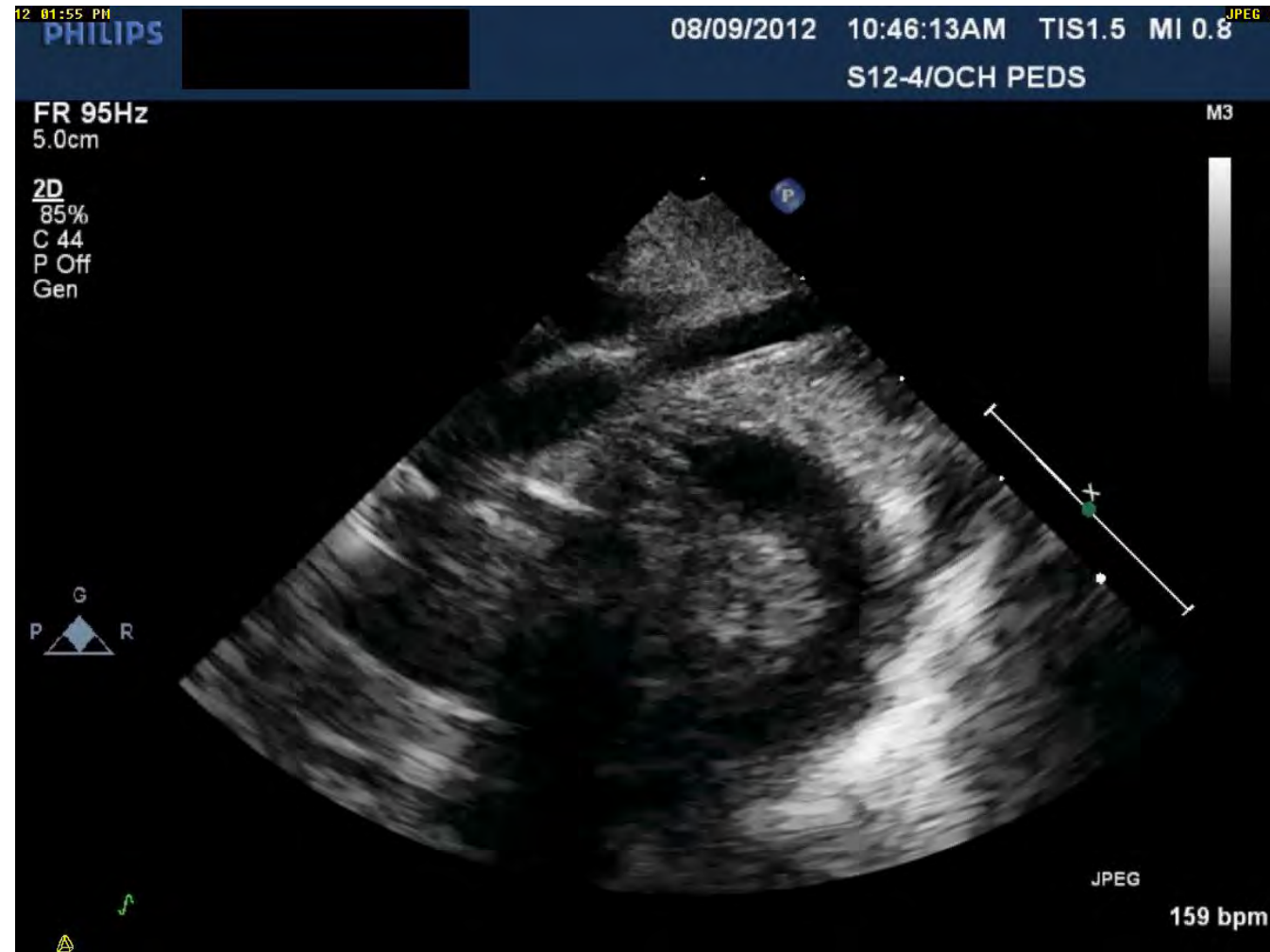
Definition: IAA

- Complete separation of the ascending and descending aorta
- Ductal dependent systemic blood flow
- Incidence:
 - 2/100,000 births
 - 1.5% of CHD



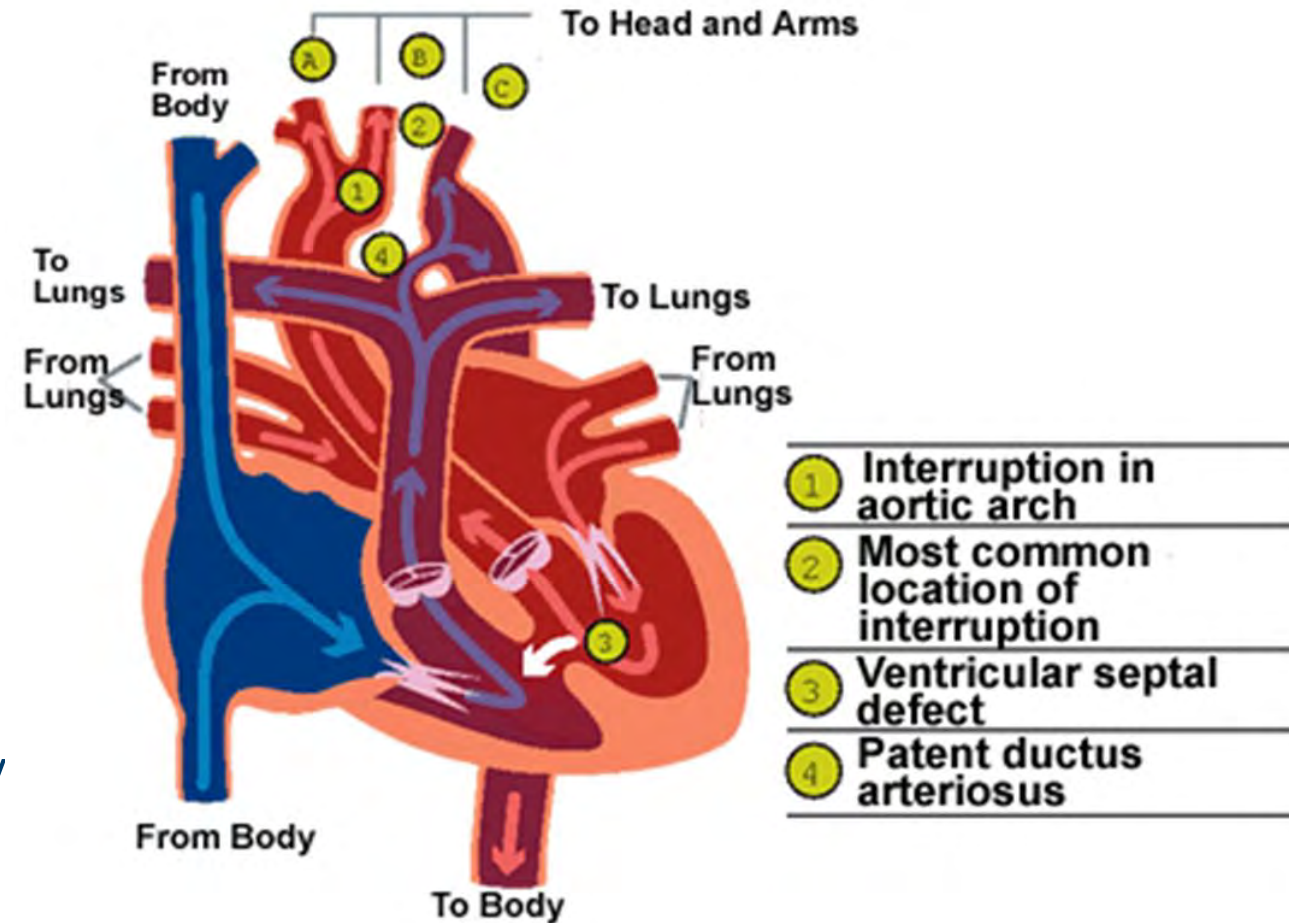
Pathophysiology: IAA

- 50% with deletion of 22q11 (DiGeorge)
- Associated VSD or AP window
- Posterior malalignment of the conal septum = LVOT obstruction
- Over ½ with associated LVOT obstruction

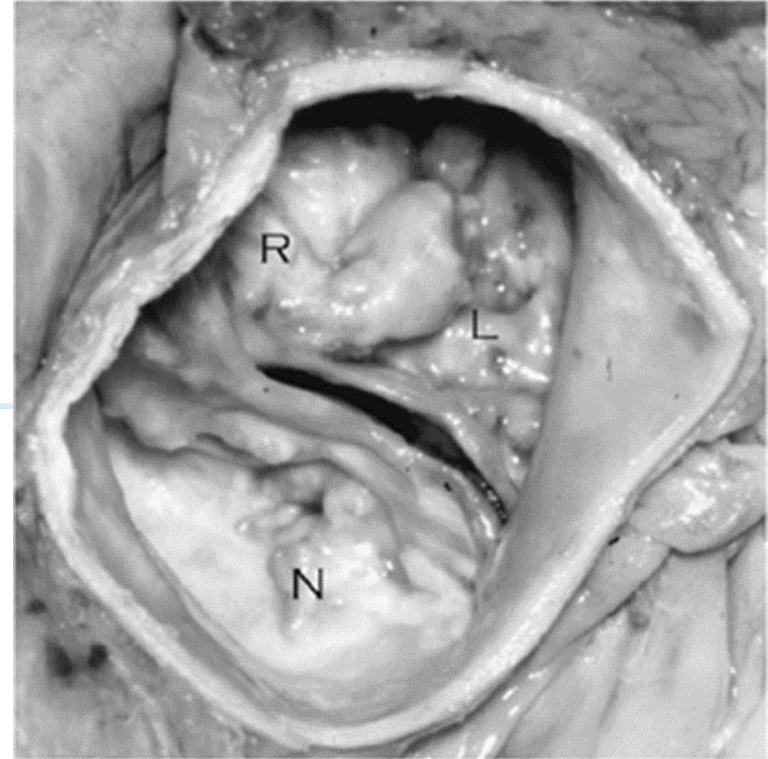


Presentation: IAA

- Ductal closure = limited blood flow to the lower body
- Diminished pulses
- BP differential
- Differential cyanosis (LE cyanosis)
 - Not frequently seen
 - Pulmonary arterial blood (ductal blood) relatively highly saturated due to large VSD
- Systolic ejection murmur usually due to LVOT obstruction
- CHF due to poor cardiac output and pulmonary edema
- Acute cardiovascular collapse
 - Shock and acidosis
 - Multi-organ system dysfunction

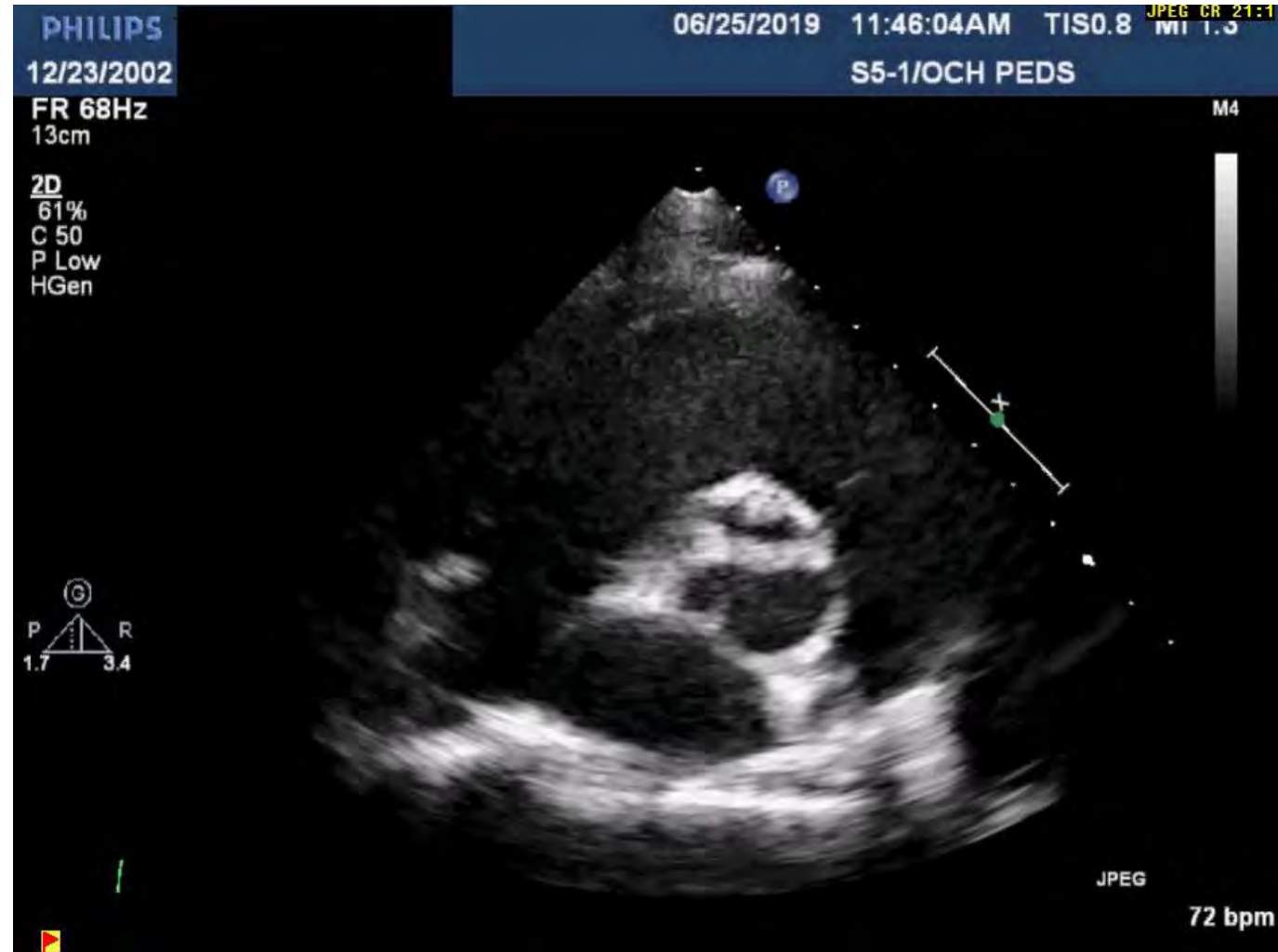


Aortic Stenosis



Definition: AS

- Obstruction of the left ventricular outflow tract
- Secondary to anatomic valve or outflow tract anomaly
- Bicuspid aortic valve is the most common form of CHD
- LVOT obstruction incidence:
 - 3-4/10,000 live births
- Critical AS = ductal dependent systemic blood flow

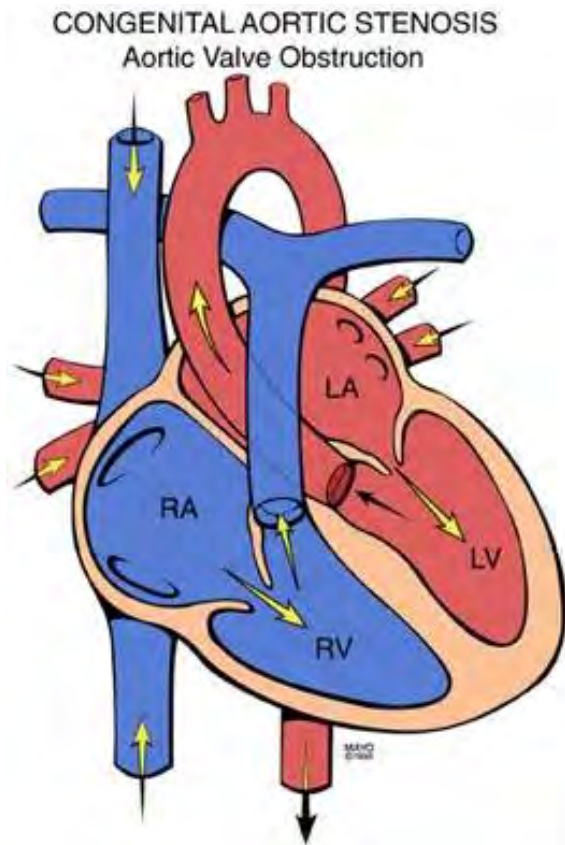
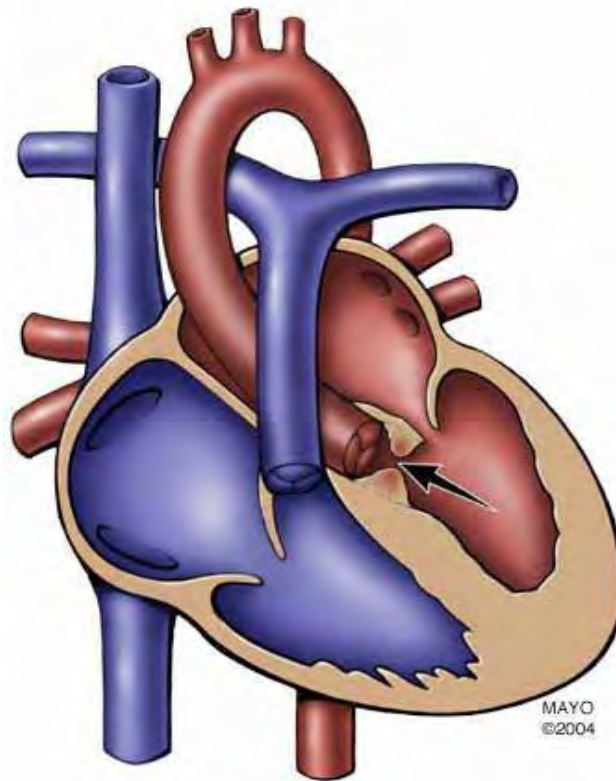
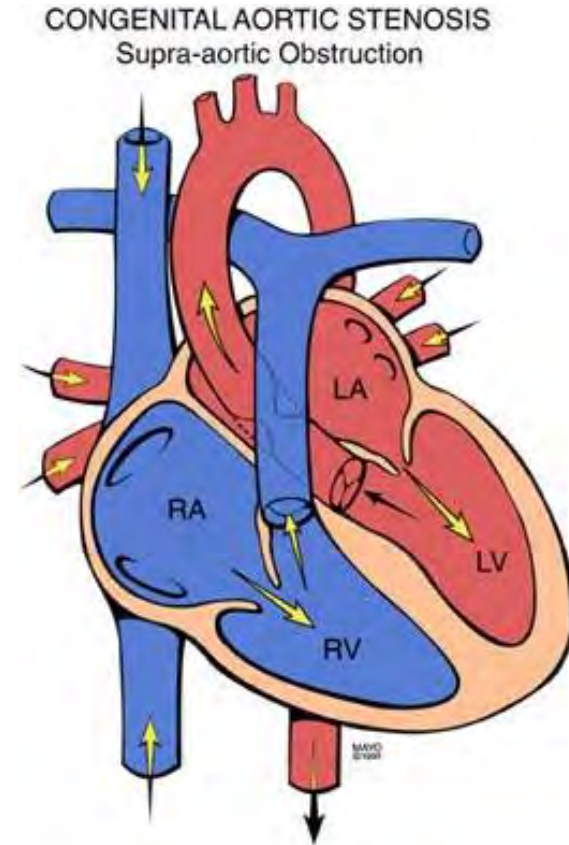


Aortic Stenosis

Valvular

Subvalvar

Supravalvar

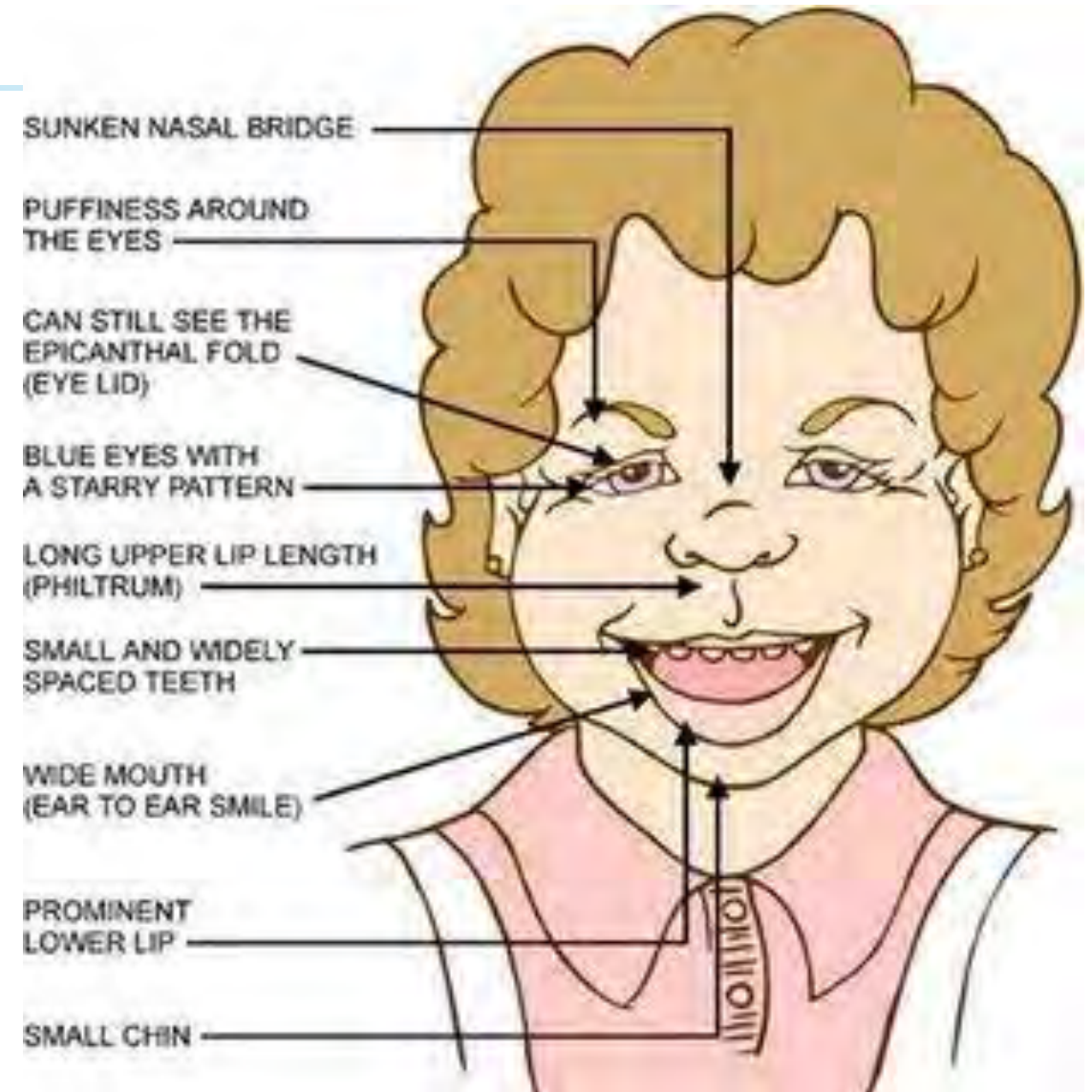


Pathophysiology: Valvar AS in Infancy

- In utero
 - Mild: $\uparrow$ LV pressure $\pm$ mild LVH
 - Moderate: LVH but normal flow, LV size normal
 - Severe: LVH, decreased flow, decreased LV compliance can result in small AoV and MV
- Tolerated in fetus w/ rapid development of symptoms after birth
- Systolic ejection murmur +/- gallop, poor pulses/perfusion
- CHF due to poor cardiac output and pulmonary edema
 - Tachypnea/wheezing
 - Poor feeding
 - FTT
- Critical AS – circulatory collapse w/ PDA closure
- **Murmur quality and echo gradient may not be reliable due to poor LV function**

Supravalvar AS

- Least common type of aortic stenosis
- Most common with **Williams syndrome**
 - Deletion of 7q11
- Can also be seen with familial elastin disorders

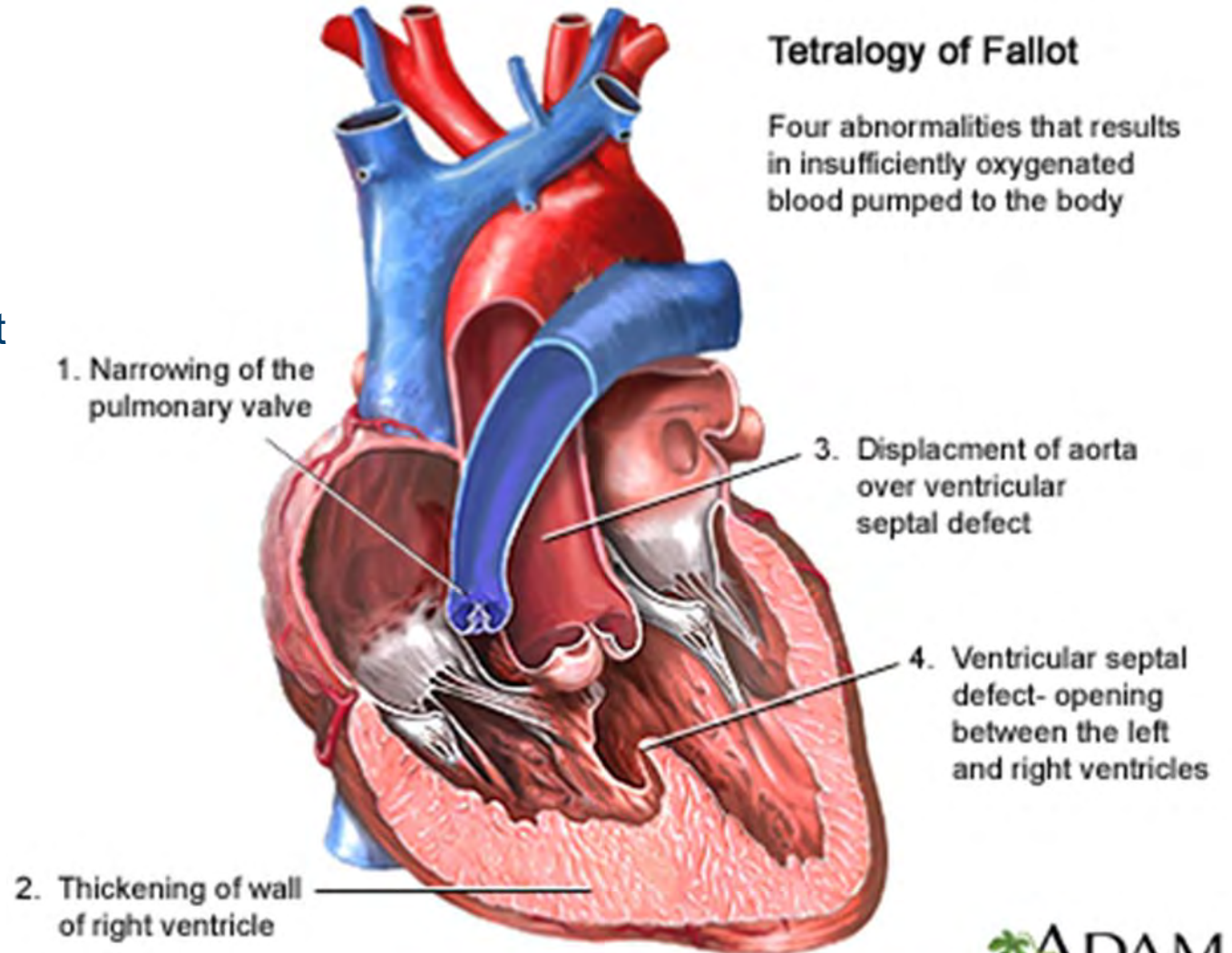


Presentation of Congenital Heart Disease in Infancy: Hypoxic lesions

Tetralogy of Fallot

Definition: Tetralogy of Fallot

- Fallot's tetralogy:
 - 1) Right ventricular outflow obstruction
 - 2) Right ventricular hypertrophy
 - 3) Overriding aorta
 - 4) Large malaligned ventricular septal defect
- Leading form of cyanotic CHD
- Incidence:
 - 4-5/10,000 live births
 - 7-10% of all CHD
- Male > female predominance



PHILIPS

09/21/2016

01:50:46PM TIS1.5 MI 0.8

S12-4/OCH PEDS

FR 95Hz
5.0cm

M4

2D

77%
C 48
P Off
Gen



JPEG

131 bpm

PHILIPS

10:45:30AM TIS1.1 MI 1.2

S8-3/OCH PEDS

FR 20Hz
7.0cm

2D

70%

C 50

P Off

HGen

CF

77%

3.0MHz

WF High

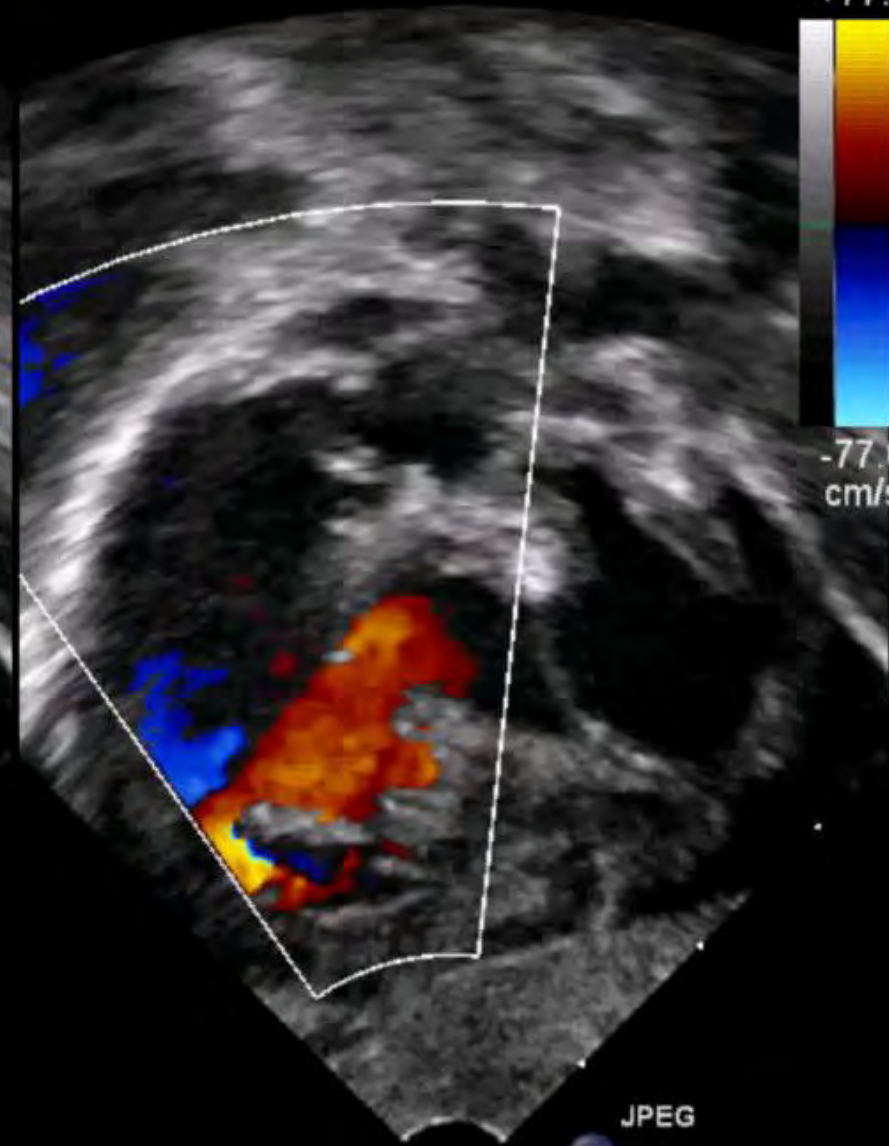
Med

M4 M4

+77.0

-77.0

cm/s



JPEG

P

114 bpm

Pathophysiology: “Pink” vs “Blue”

- Physiology dependent on RVOT obstruction
 - Minimal obstruction - high sats
 - “pink tet”
 - Pulmonary overcirculation and even CHF possible (same physiology as VSD)
 - Severe obstruction – hypoxia
 - “blue tet”
- Almost always large, unrestrictive VSD

Hypercyanotic (Tet) Spell

- Often provoked by crying
- Cry becomes frantic
 - Pain of skeletal muscle/myocardial ischemia
- Worsening cyanosis and breathlessness
- LOC and death
- Acute imbalance between systemic & pulmonary blood flow
- Increased R-L shunt
- Pain and anxiety →
 - Endogenous catecholamine release
 - Tachycardia → reduced RV preload
 - Increased O₂ consumption & reduced mixed venous O₂ content
 - Decreased SVR
- **Markedly diminished RVOT murmur!**



"Tet spell"

Treatment of a Hypercyanotic Spell:

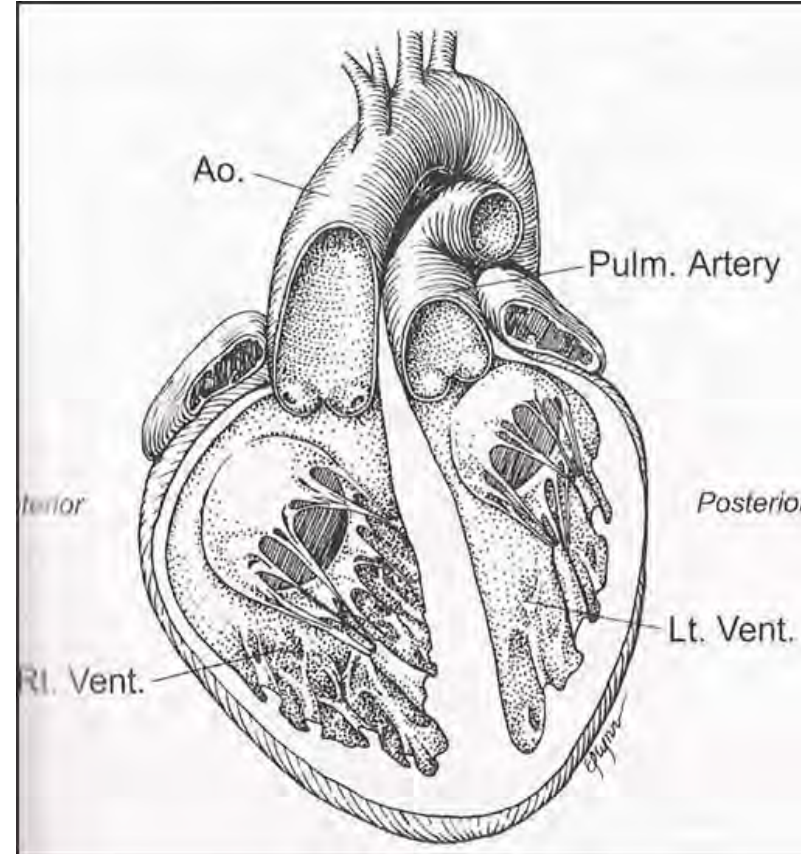
Restore PBF/SBF balance

1. Provoked/worsened with crying:
 - Pick up & comfort
 - Knees/hips flexed
 - Increase SVR
2. Oxygen
 - Decrease PVR
3. IV bolus
 - Increase preload
4. IV/IM Morphine:
 - Alleviate pain/anxiety
 - Decrease catecholamine release
5. IV Propranolol/Esmolol:
 - Decrease HR to increase preload
6. IV sodium Bicarb:
 - Correct acidosis to decrease PVR
7. IV systemic vasoconstrictors
 - Phenylephrine or Norepi
 - Increase SVR
8. Anesthesia, intubate/ventilate
 - Decrease O₂ consumption
9. ECMO

D-Transposition of the Great Arteries (d-TGA)

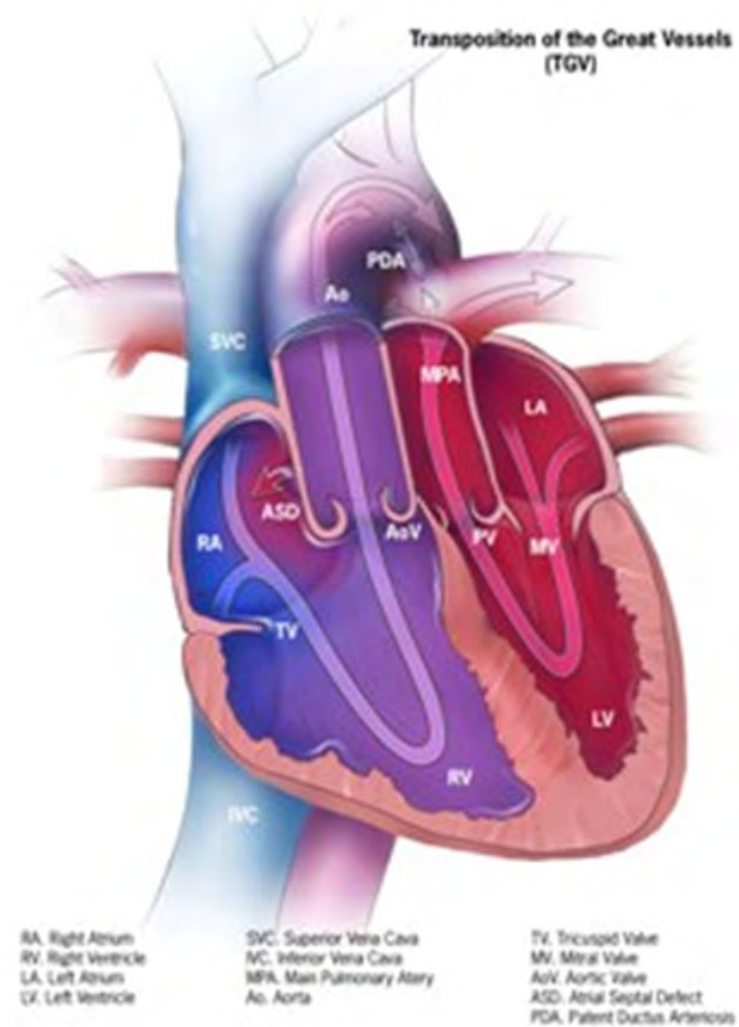
Definition: d-TGA

- Abnormal position of the aorta relative to the pulmonary artery
- Ventriculoarterial discordance
 - Morphologic **RV** gives rise to **aorta**
 - Morphologic **LV** gives rise to **pulmonary trunk**
- Incidence:
 - 2-5/10,000 live births
 - 3% of CHD



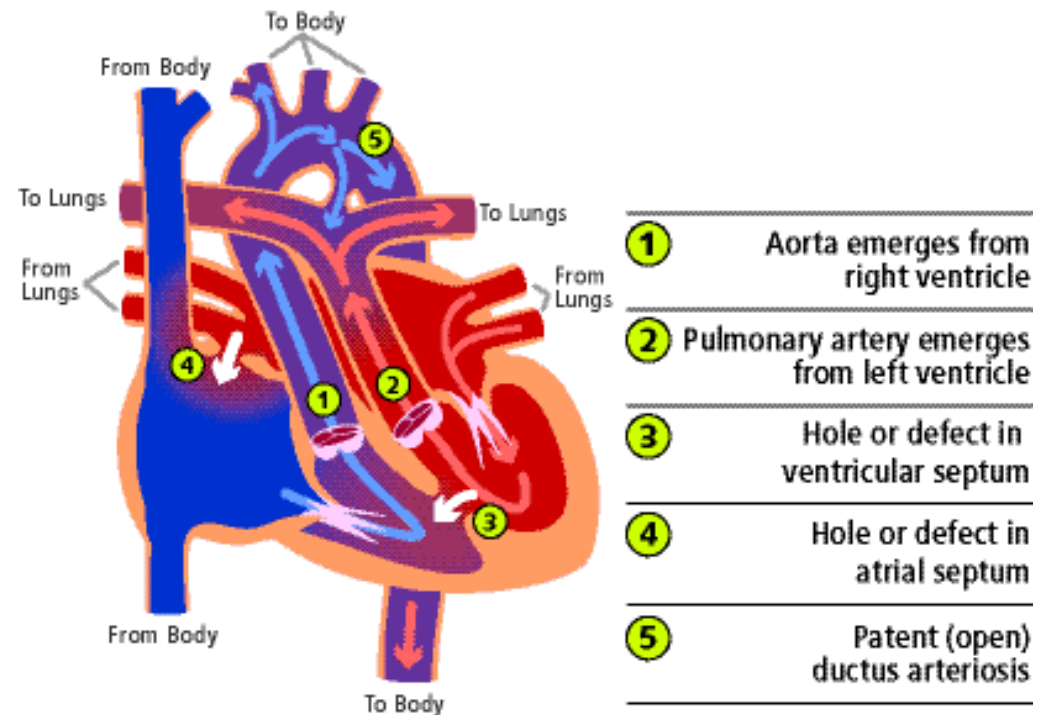
Pathophysiology: d-TGA

- Aorta is rightward and anterior
- Pulmonary artery is leftward and posterior
- 2 parallel circulations
 - Systemic venous blood → RV → aorta
 - Pulmonary venous blood → LV → PA
- Survival depends on extent of mixing
 - ASD
 - VSD
 - PDA



Pathophysiology: d-TGA

- Aorta to PA at the PDA
- LA to RA at the PFO (O₂→body)
- PFO too small?
 - pulmonary circuit overload
 - decreased SaO₂
 - acidosis

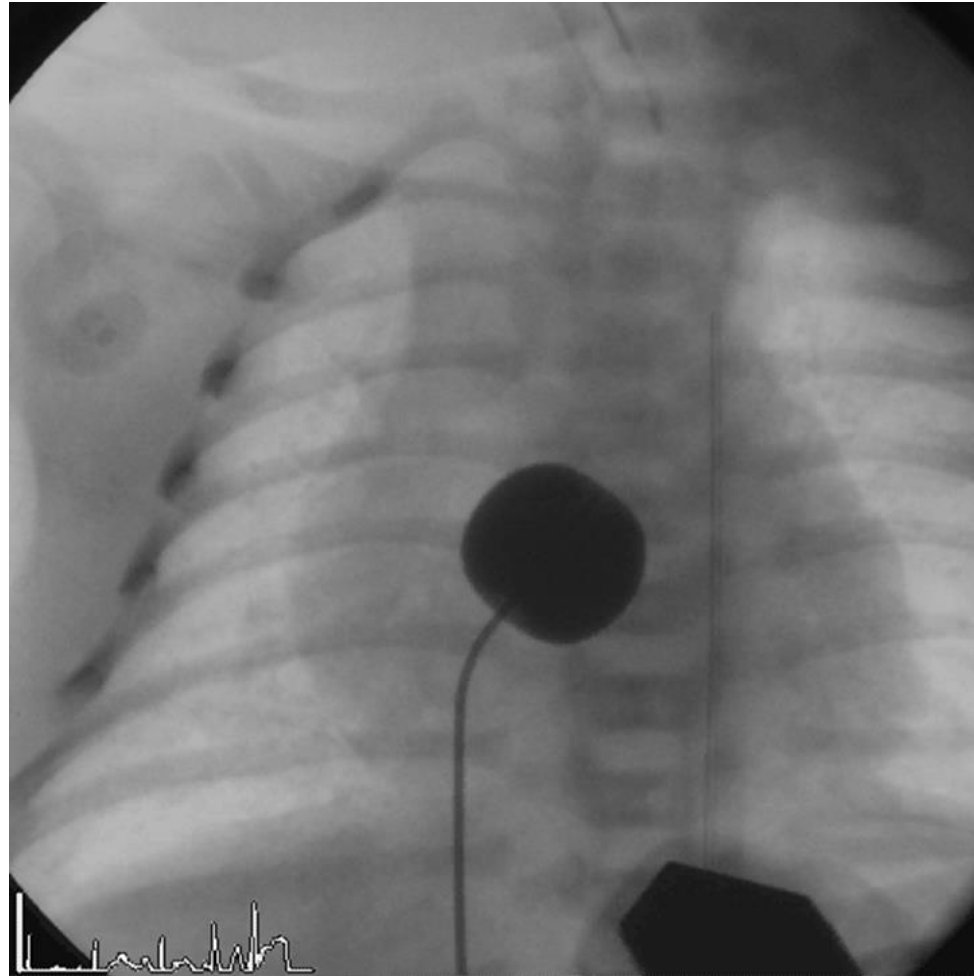


Presentation: d-TGA

- Usually good-sized baby, not dysmorphic
- Cyanosis in the first few hours
 - Unresponsive to supplemental O₂
- Reverse differential cyanosis
 - Coarctation
 - IAA
 - Pulmonary hypertension
- Tachypnea
- Metabolic acidosis



Balloon atrial septostomy



Balloon atrial septostomy

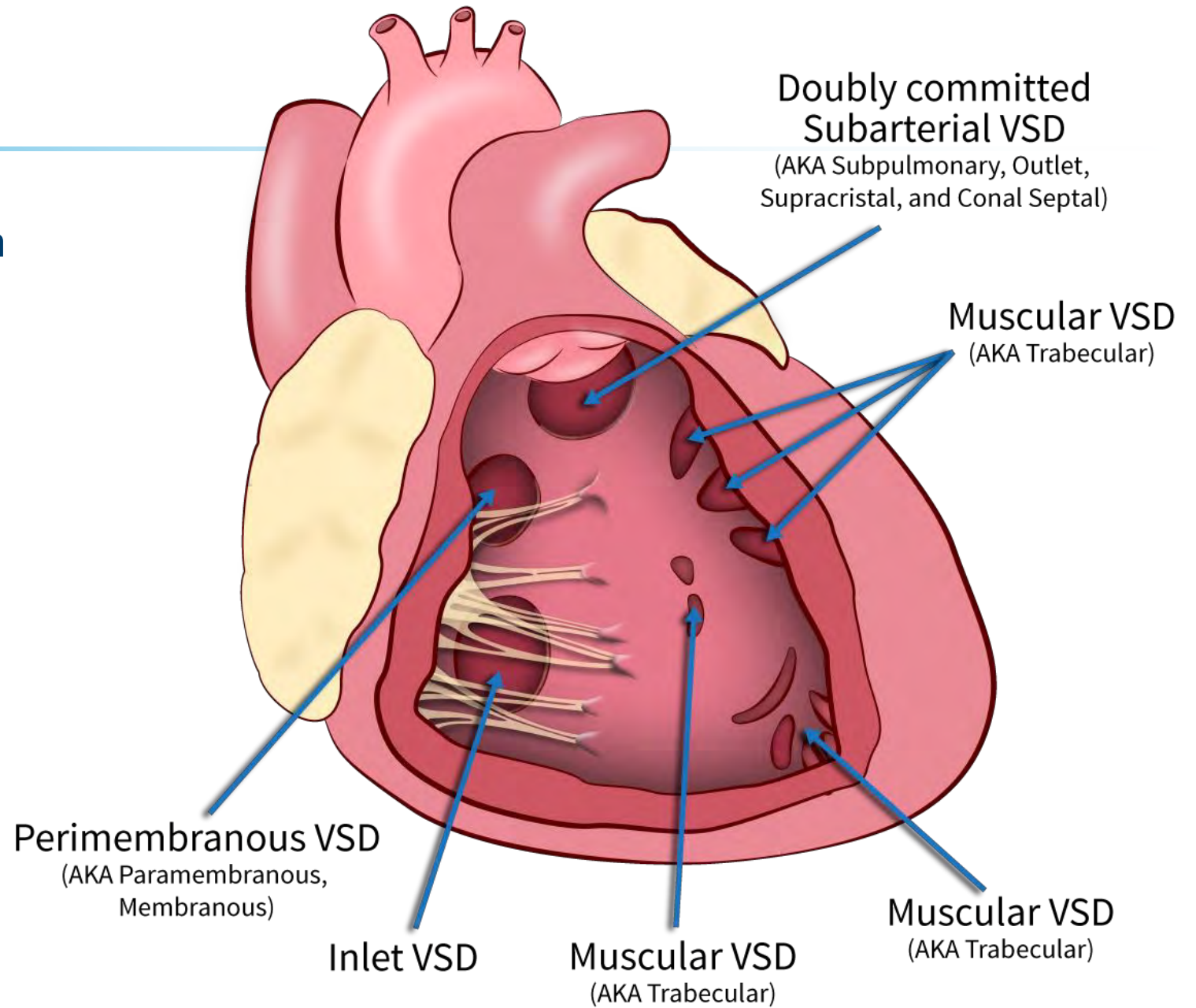


Presentation of Congenital Heart Disease in Infancy: Pulmonary Overcirculation

Ventricular Septal Defect

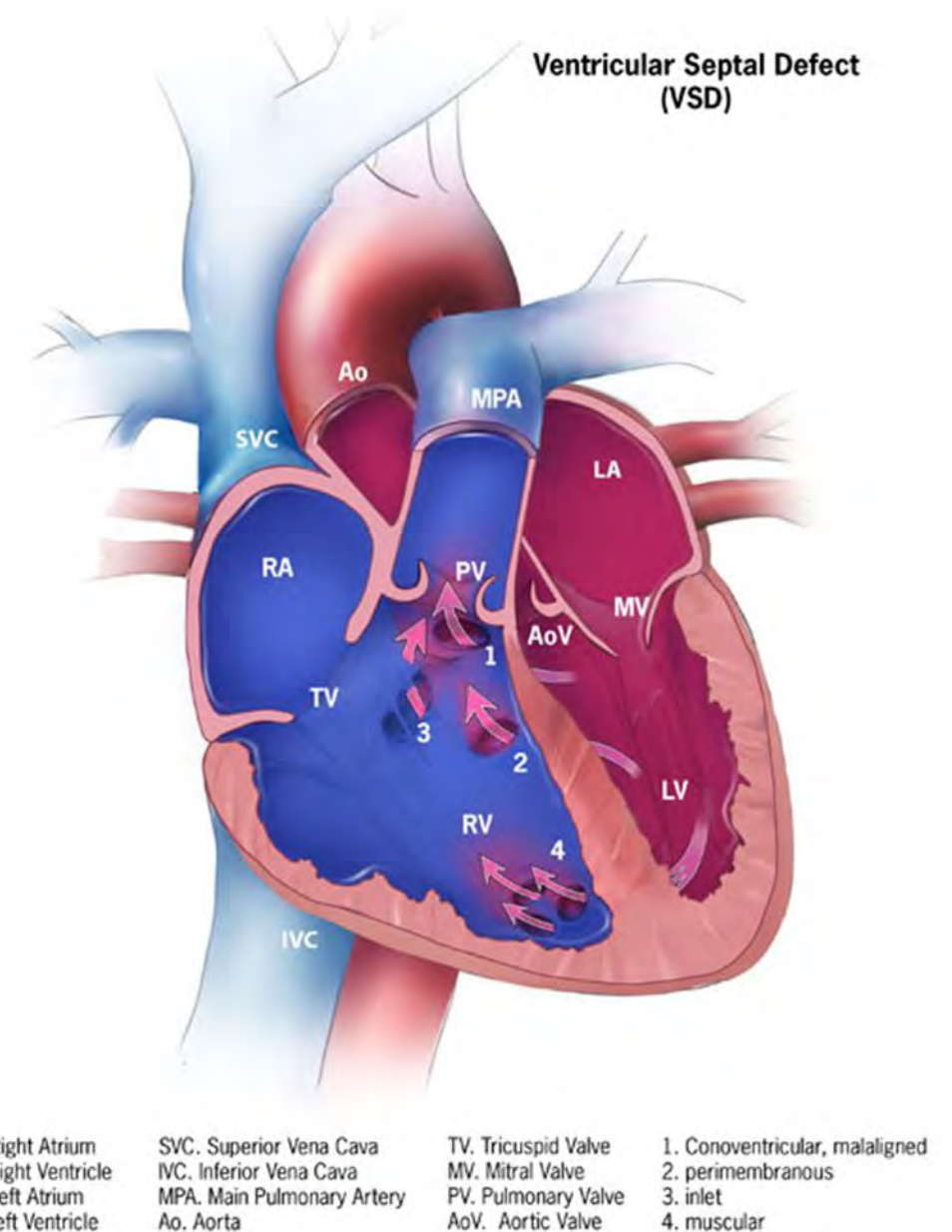
Definition: VSD

- Defect in the ventricular septum
- Multiple types
 - Outlet
 - Perimembranous
 - Inlet
 - Muscular
- 40% of all CHD
- Most common lesion
 - Trisomy 21
 - Trisomy 13
 - Trisomy 18



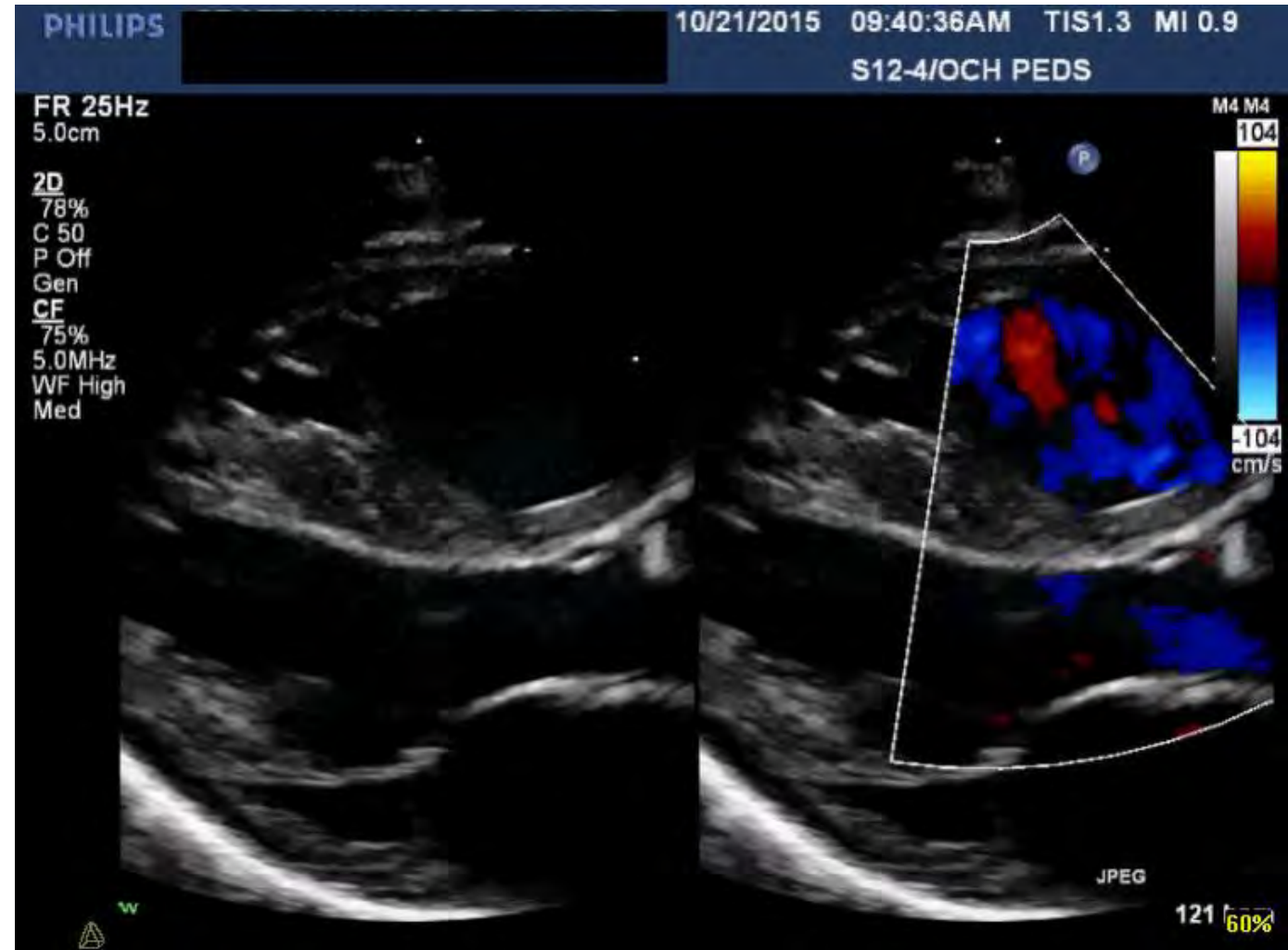
Pathophysiology: VSD

- Left to right ventricular shunt
- Symptoms occur as PVR falls
 - PVR high during fetal life
 - Decreases by 50% in the first 24 hours of life
 - Adult levels by 6-8 weeks
- Increased pulmonary blood flow
- Left heart enlargement



Presentation: VSD

- Determined by the size of the defect
- Murmur may not be appreciable in the nursery
 - Smaller defects – early systolic murmur
 - Larger defects
 - Holosystolic murmur
 - Very large – pulmonary flow murmur
- Symptoms of pulmonary overcirculation @ 6-8 weeks
 - Tachypnea/wheezing
 - Poor feeding/FTT
 - Sweating
 - Hepatomegaly

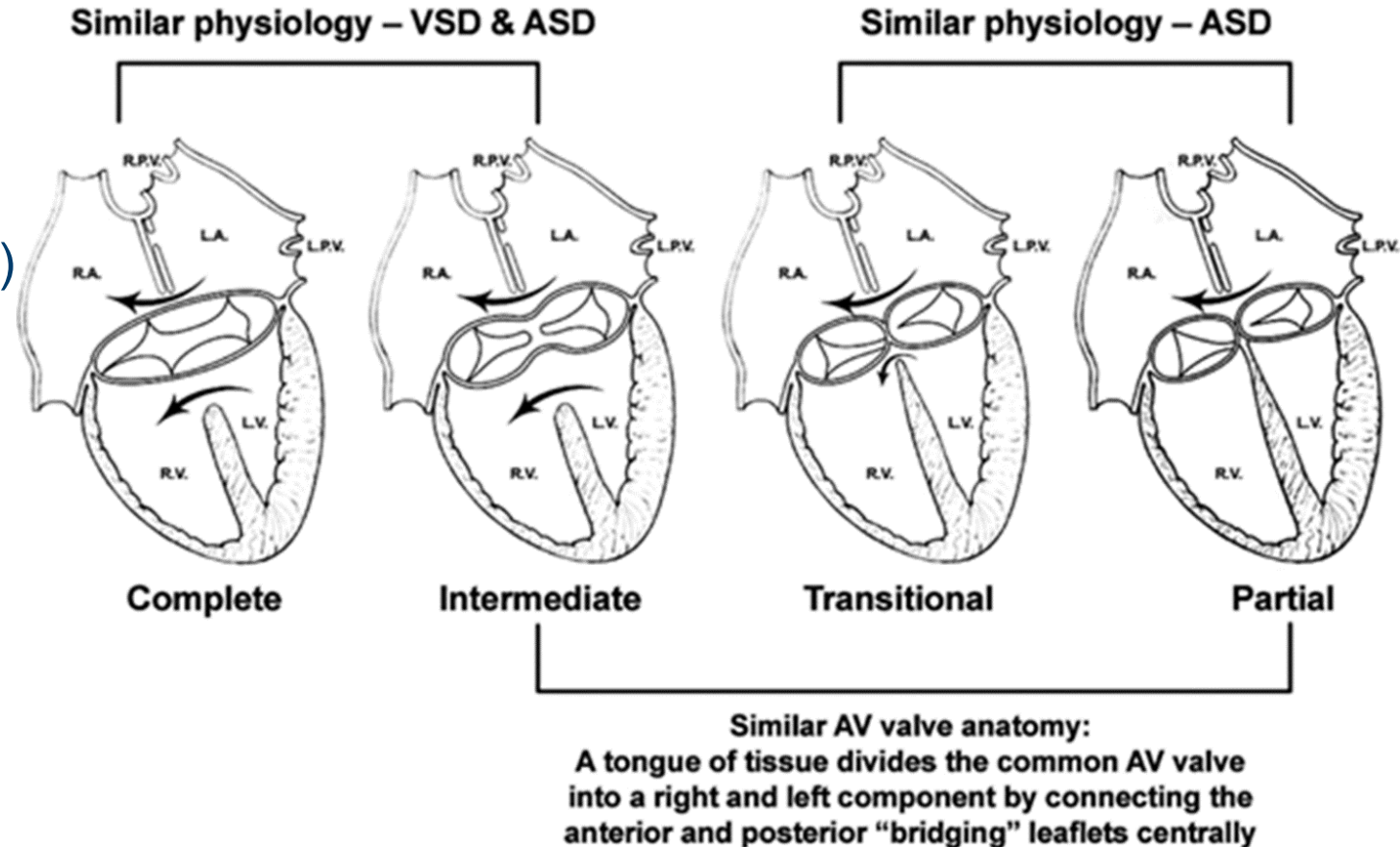


Atrioventricular Septal Defect

Definition: AVSD

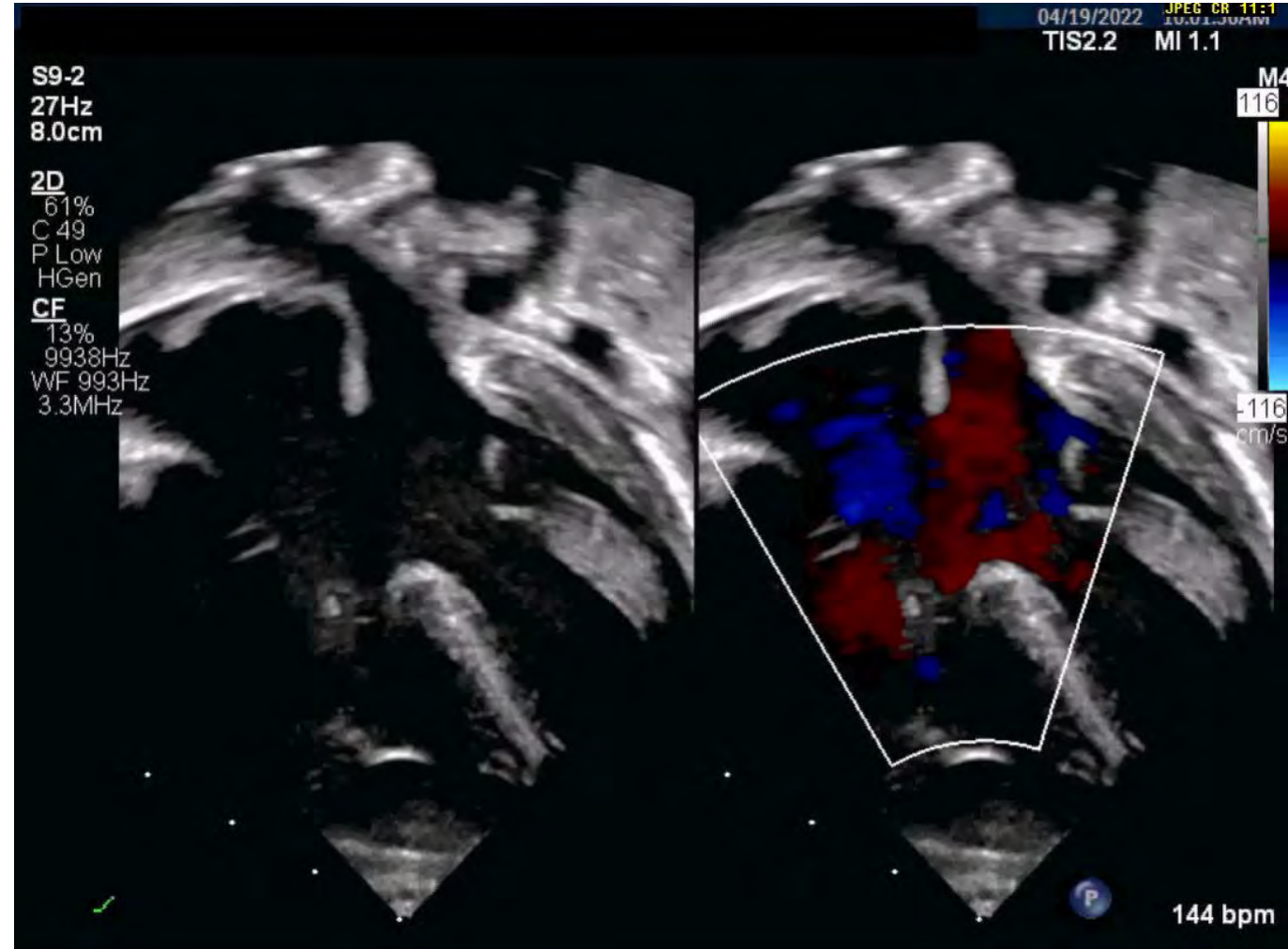
- Defect of the atrioventricular septum
- Abnormalities of the atrioventricular valves
- 4-5% of congenital heart disease (CHD)
- 40-45% of children with Down syndrome (trisomy 21) have CHD
 - 40% of those with CHD have AVSD (complete)

AVSD Summary



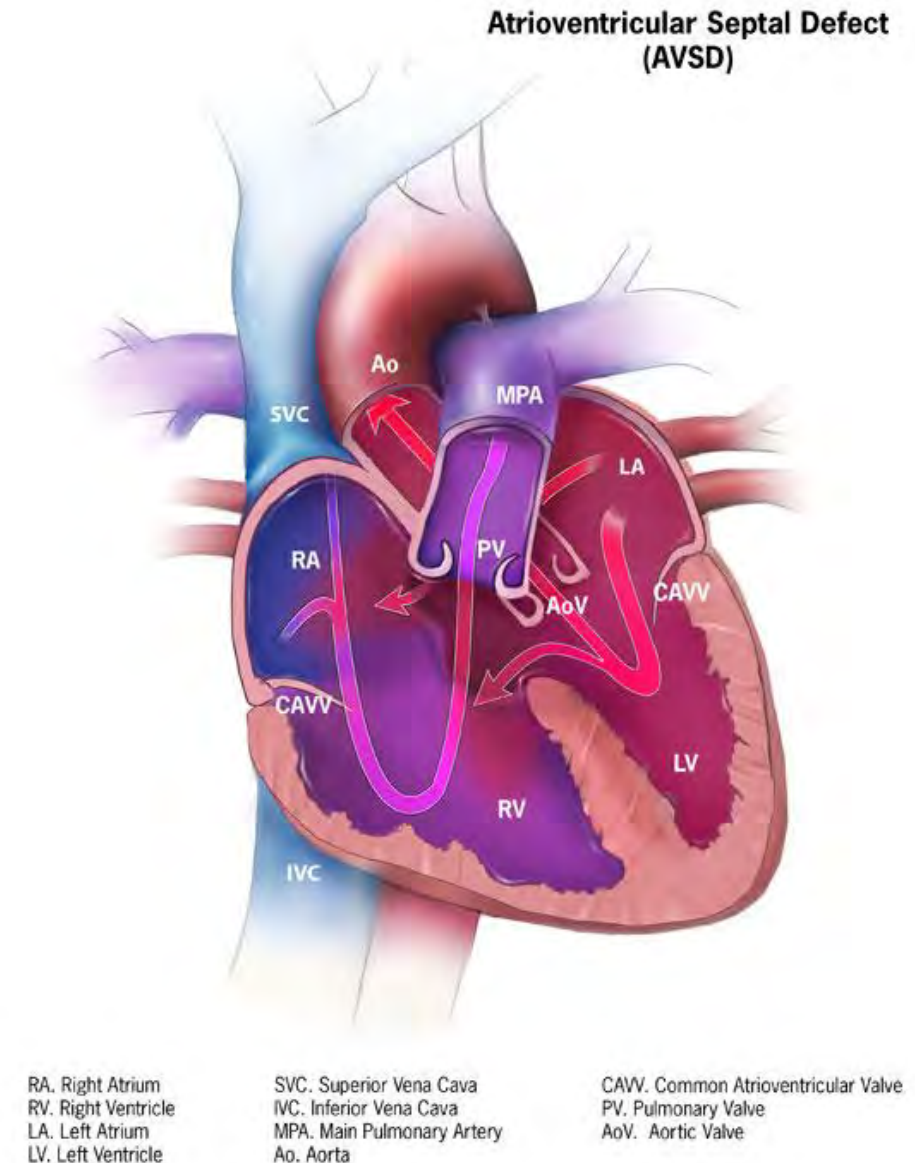
Pathophysiology: Complete AVSD

- Significant intracardiac shunt = ASD + VSD
- Atrial left to right shunt
 - Right heart enlargement
- Ventricular left to right shunt
 - Pulmonary overcirculation
 - Left heart enlargement (LA > LV, large atrial shunt)

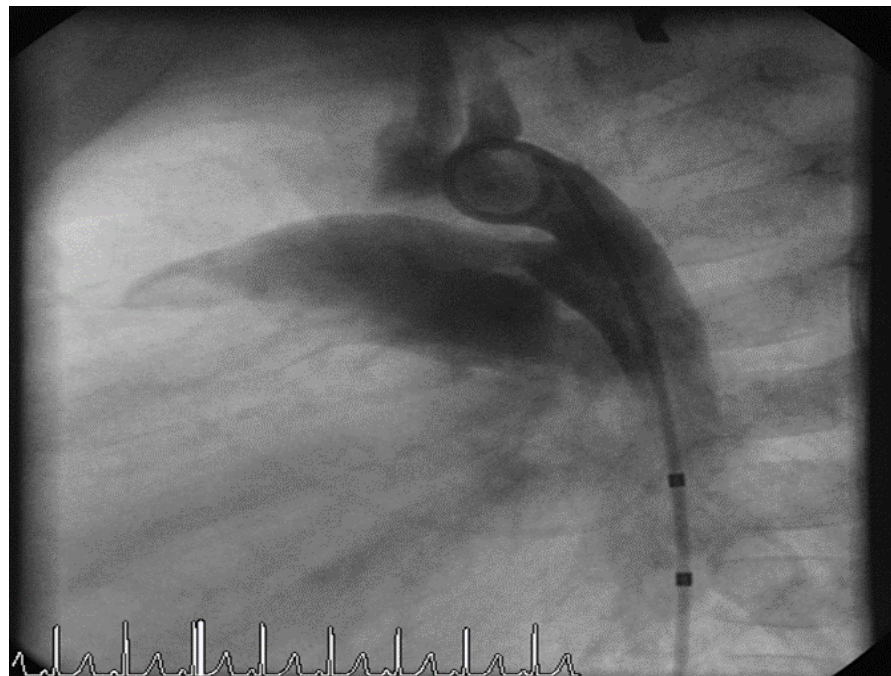


Presentation: AVSD

- Signs/symptoms when PVR drops
- Congestive heart failure due to pulmonary overcirculation
 - Tachypnea/wheezing
 - Sweating
 - Poor feeding/FTT
 - Hepatomegaly

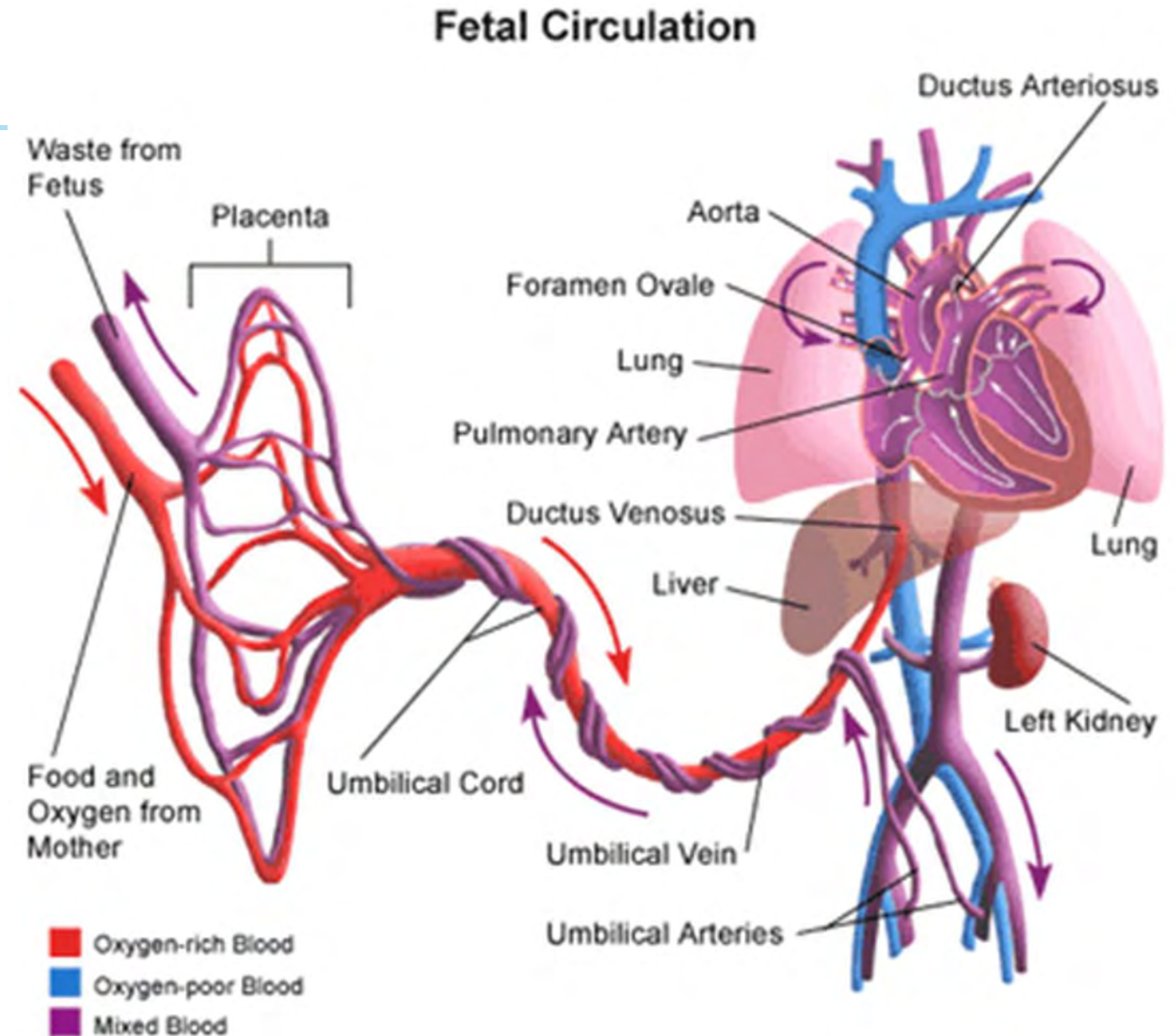


Patent ductus arteriosus



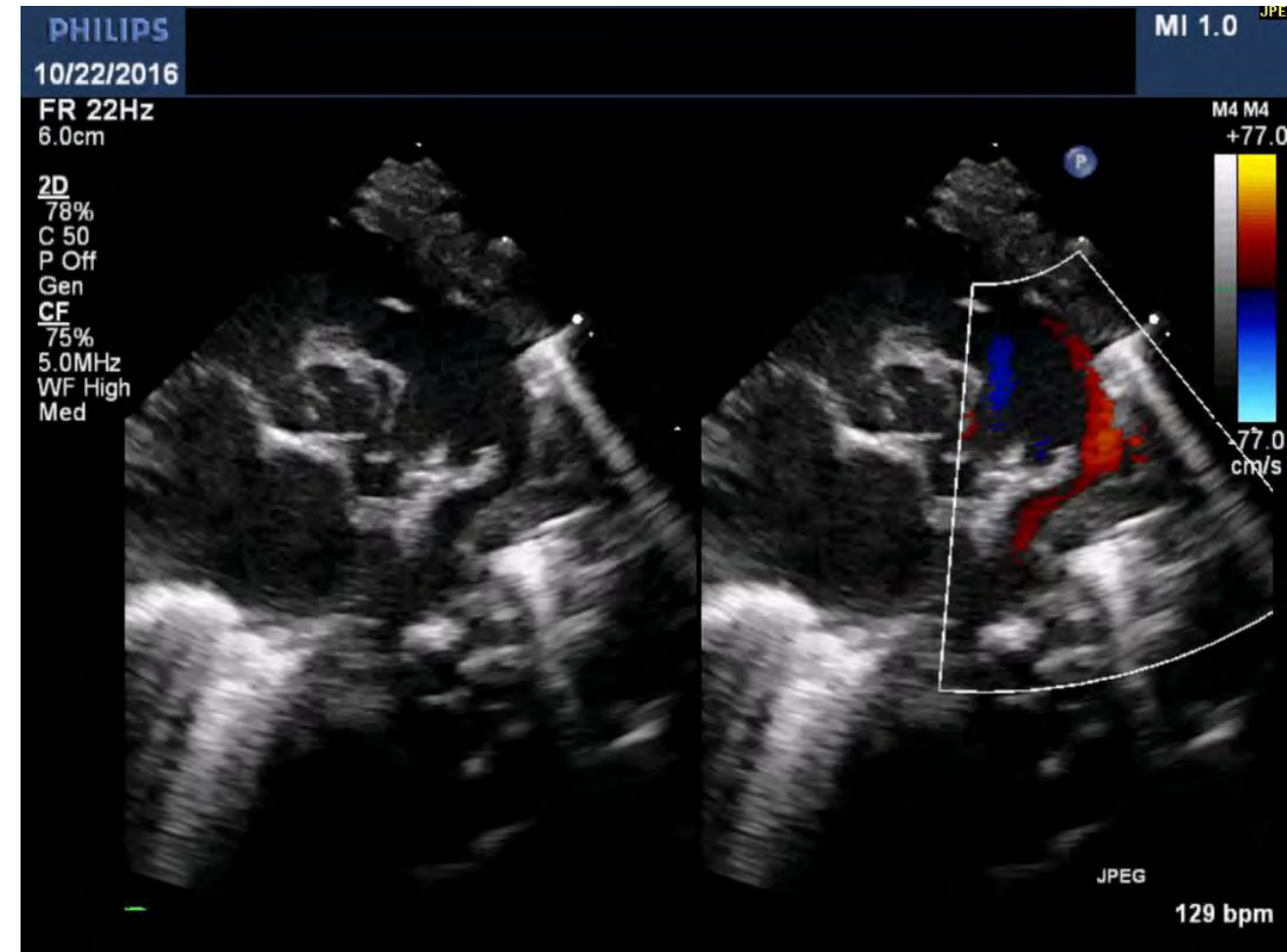
Definition: PDA

- Ductus arteriosus - normal fetal structure connecting the pulmonary artery and the aorta
- In utero most blood bypasses the lungs and is delivered to the body via the DA
- Postnatal constriction promoted by oxygen
 - Hemodynamic closure @ 10-15 hours of life
- PDA = persistently patent ductus arteriosus



Pathophysiology: PDA

- Full term infants
 - Isolated 0.5/1000 live births
 - 5-10% of all CHD
- Preterm infant
 - Overall 8/1000 live births
 - Very high incidence (80%) in low-birth-weight infants (<1200g)
- Dependent upon defect size
- Pulmonary vs. systemic vascular resistance
 - Significant effects of lung disease in premature infants
- Left to right shunt (aorta to pulmonary artery)



Presentation: PDA

- Neonate
 - High PA/RV pressure = primarily systolic flow through the PDA
 - Systolic murmur
- As PVR falls
 - Aortic pressure > pulmonary pressure = continuous flow from the aorta into the PA
 - Continuous machinery like murmur left infraclavicular space
- Small defect
 - PVR and PA pressures decrease normally
 - Minimal increase in pulmonary blood flow
 - Normal left heart size
- Large defect
 - Early congestive heart failure due to pulmonary overcirculation
 - Significant pulmonary edema
 - Left heart enlargement

Review

A 1-week-old full term infant presents to your clinic with complaints of poor feeding and fast breathing.

Vitals: HR 180bpm, RR 70, RA sat 92%, BP 50/25

On exam, the patient is in respiratory distress; there is no murmur.

What is your diagnosis?

- A. VSD
- B. Coarctation of the aorta
- C. Tetralogy of Fallot
- D. D-TGA



What other lesions could present in a similar fashion?

- Hypoplastic left heart
- Interrupted aortic arch
- Critical aortic stenosis

What is the most appropriate intervention for treating the patient's cardiogenic shock?

- A. NS bolus 20ml/kg
- B. Administration of 100% oxygen
- C. Administration of PGE₁
- D. Administration of antibiotics

Review

A 2-week-old full term infant presents to your clinic with concerns of blue lips

Vitals: HR 130bpm, RR 30, sat 98%, BP 75/40

On exam, the patient is breathing comfortably; there is harsh 2-3/6 systolic ejection murmur. When upset, the infant is cyanotic with sat of 75%, the murmur is decreased in intensity.

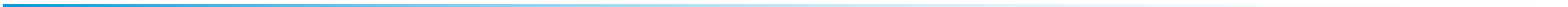
What is your diagnosis?

- A. VSD
- B. Coarctation of the aorta
- C. Tetralogy of Fallot
- D. D-TGA



What is the most appropriate intervention for treating the patient's cardiogenic shock?

- A. NS bolus 20ml/kg
- B. Calm the infant and administration of 100% oxygen
- C. Administration of PGE₁
- D. Administration of antibiotics



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