Identifying and Managing Congenital Heart disease

What primary care providers should know

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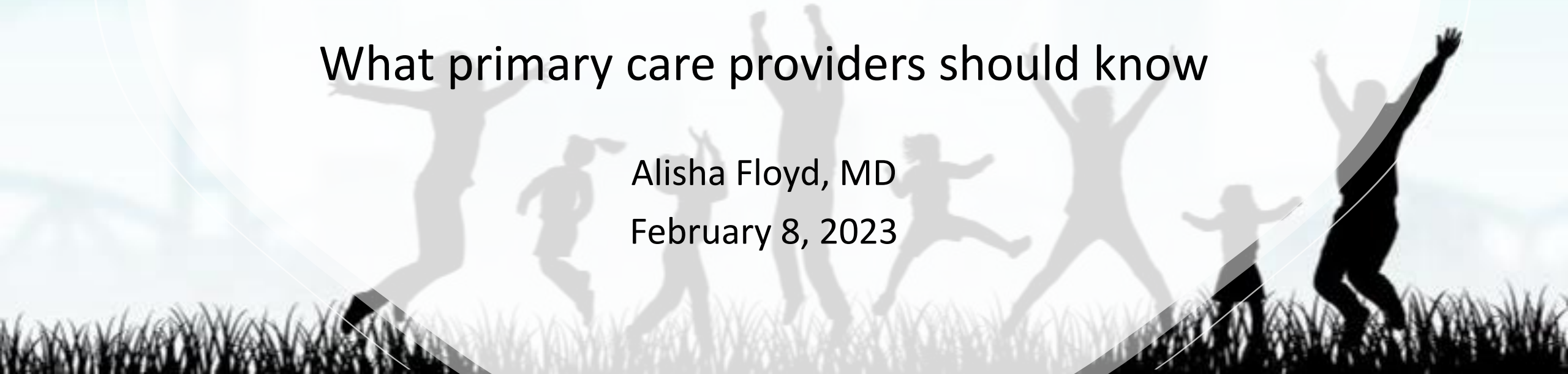


Planning Committee & Faculty Disclosure

The Planning Committee and Faculty have no relevant financial relationships with commercial interests to disclose.

WHAT WE KNOW ABOUT CONGENITAL HEART DISEASE

WHAT WE HAVE TIME TO TALK ABOUT TODAY

“What do you want to know about congenital heart disease?”

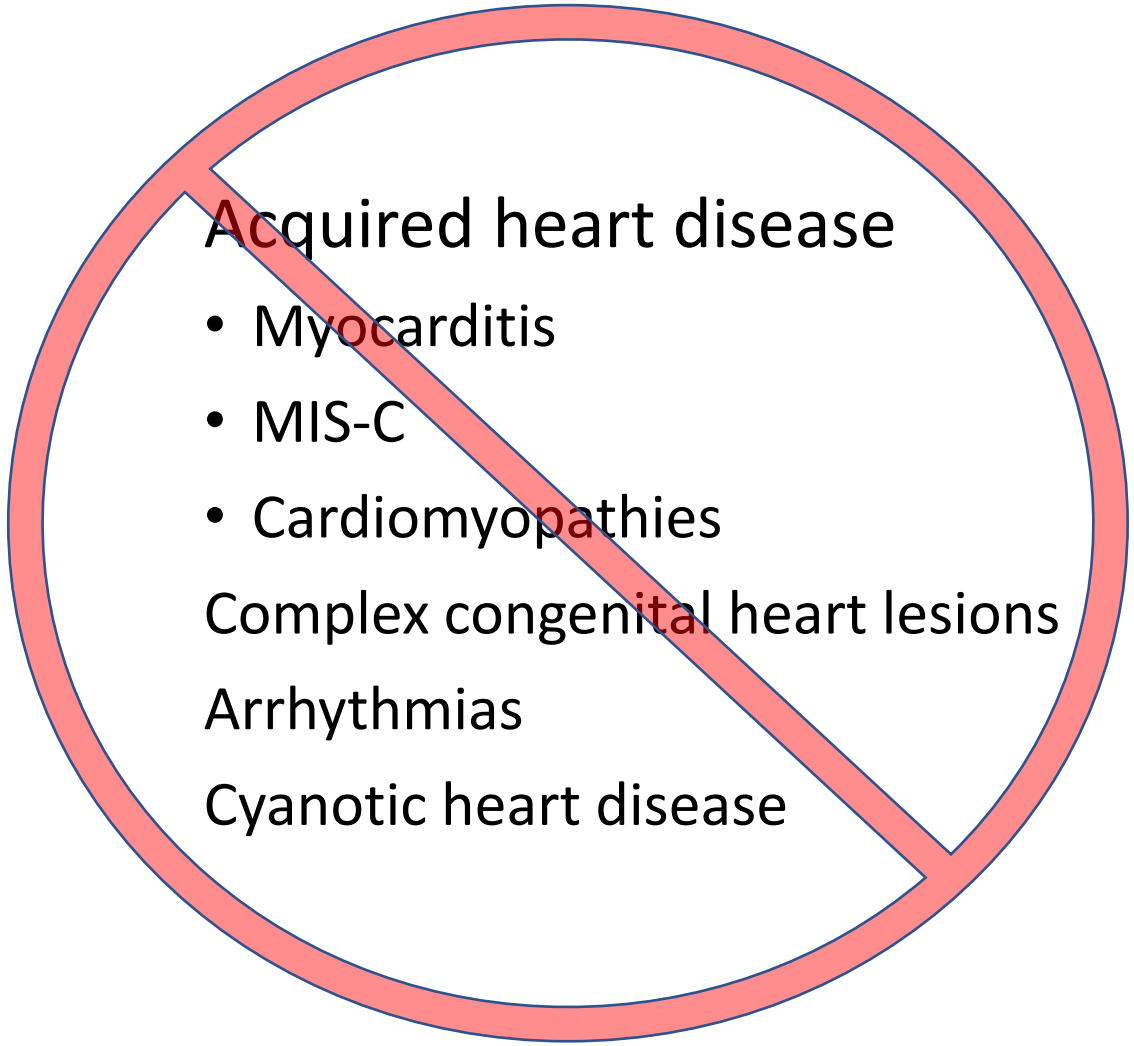
“Everything”

Outline

- Overview of CHD
  - Timing
  - Indication for intervention
  - Pre-op management
  - What not to miss

Congenital heart disease

- Left to right shunts
- Pulmonic Stenosis
- Left sided lesions



Left to right shunts

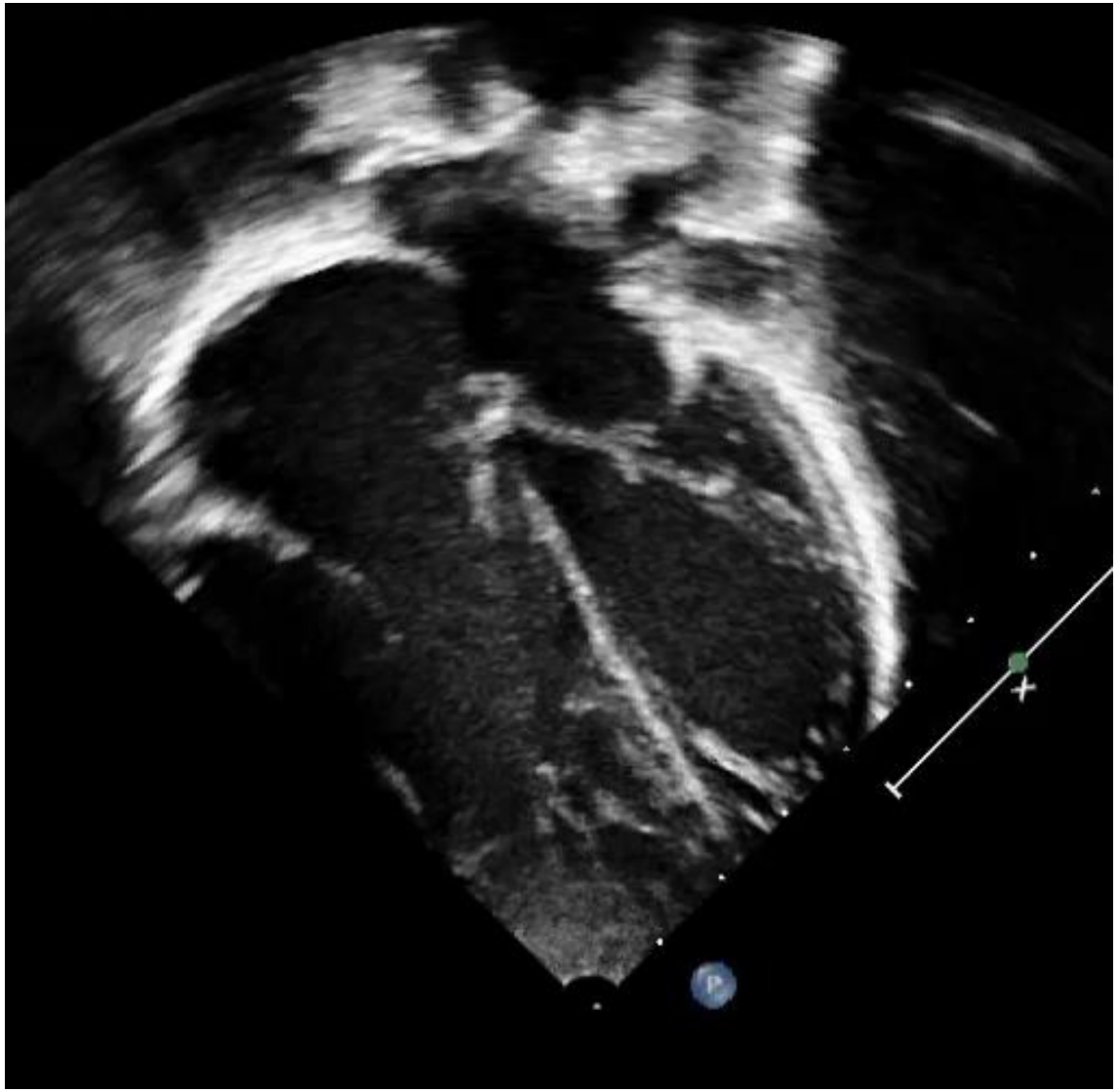
Innocent murmur?

You see a 4-year-old girl in your clinic for the first time and notice a 2/6 non-vibratory systolic ejection murmur loudest in the left upper sternal border. The S2 split is wide and fixed. There is no diastolic component. She is completely asymptomatic. Which of the following cardiac lesions is most likely?

A. VSD
B. PDA
C. ASD
D. Bicuspid aortic valve

Atrial septal defect (ASD)

- 5-10% of congenital heart defects
- More common in females
- 30-50% of children with congenital heart disease
- Associated with mitral valve prolapse and partial anomalous pulmonary veins



ASD: Presentation and Natural History

- Usually asymptomatic
- Widely split and fixed S2
- Grade 2 to 3/ 6 SEM
- Mid diastolic rumble
- Right axis deviation or RBBB pattern on ECG
- Cardiomegaly on chest Xray

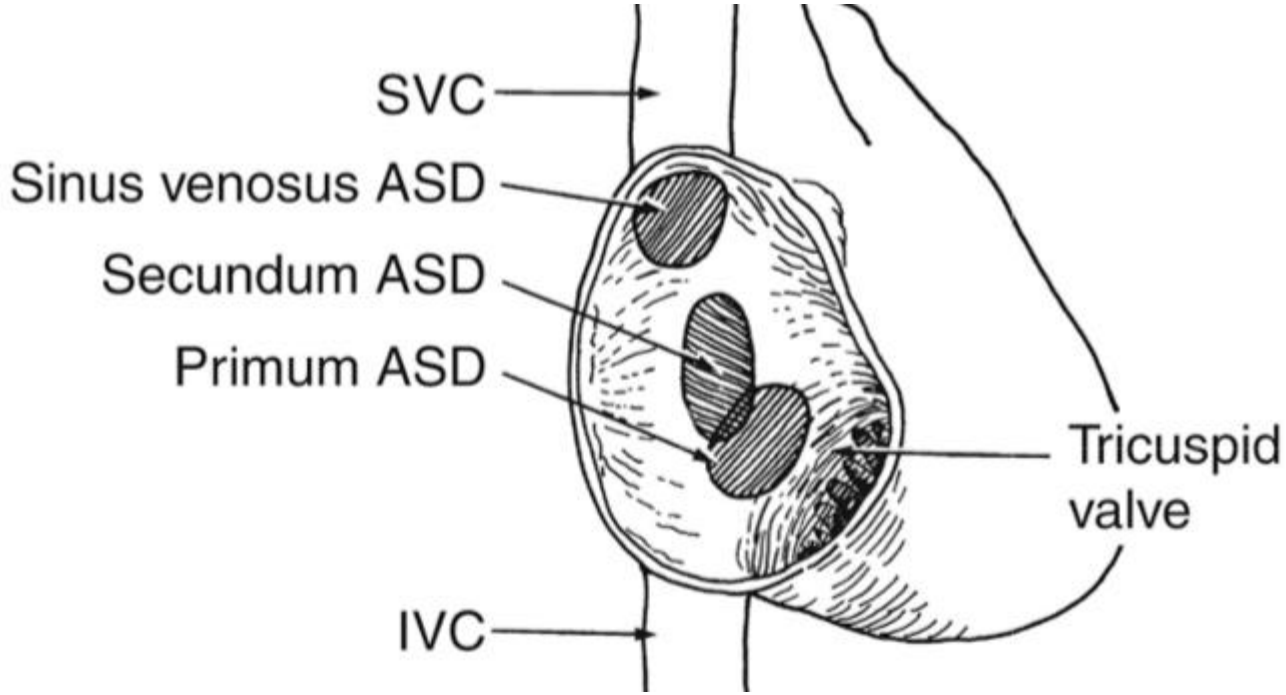
- Most small and medium defects close before age 1 ½
- Large ASDs (> 8 mm) rarely close spontaneously
- Untreated large lesions can lead to heart failure and pulmonary HTN in 20s and 30s
- Can develop atrial arrhythmias
- Stroke from paradoxical emboli is rare

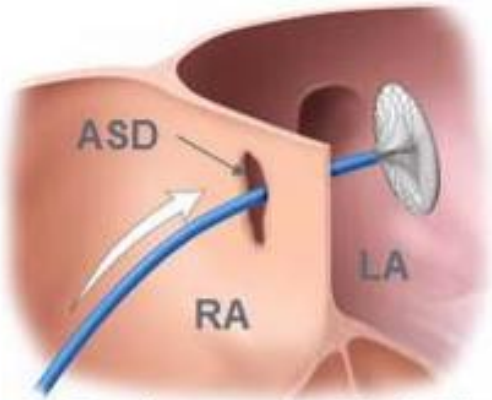
ASD: Management

Depends on size and type

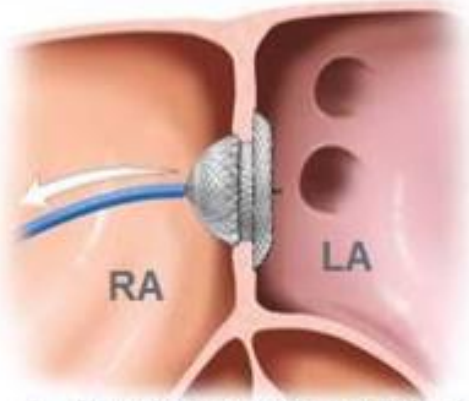
- Secundum (50-70%)
- Primum:
  - 30% (including AVSD)
  - 15% (isolated)
- Sinus vensosus (10%)
- Coronary sinus (rare)

- Surgery vs. device closure
  - Usually around age 4

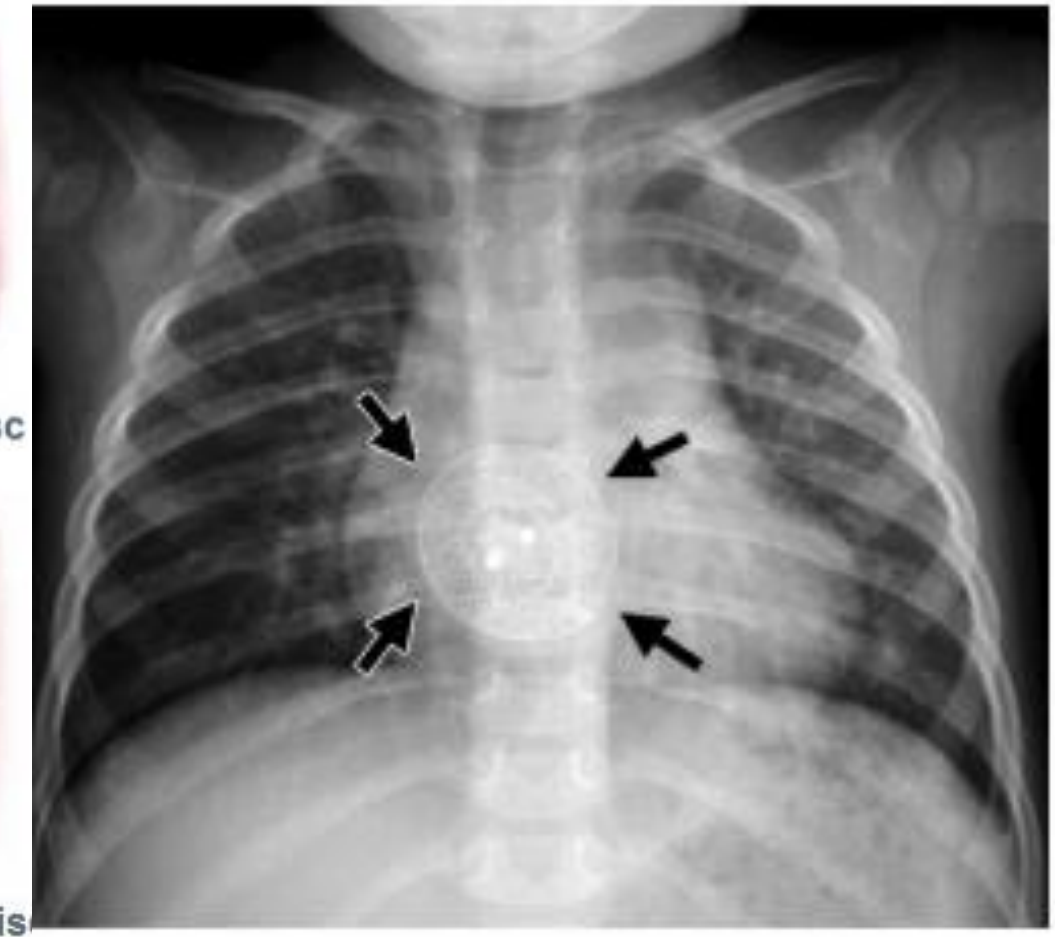


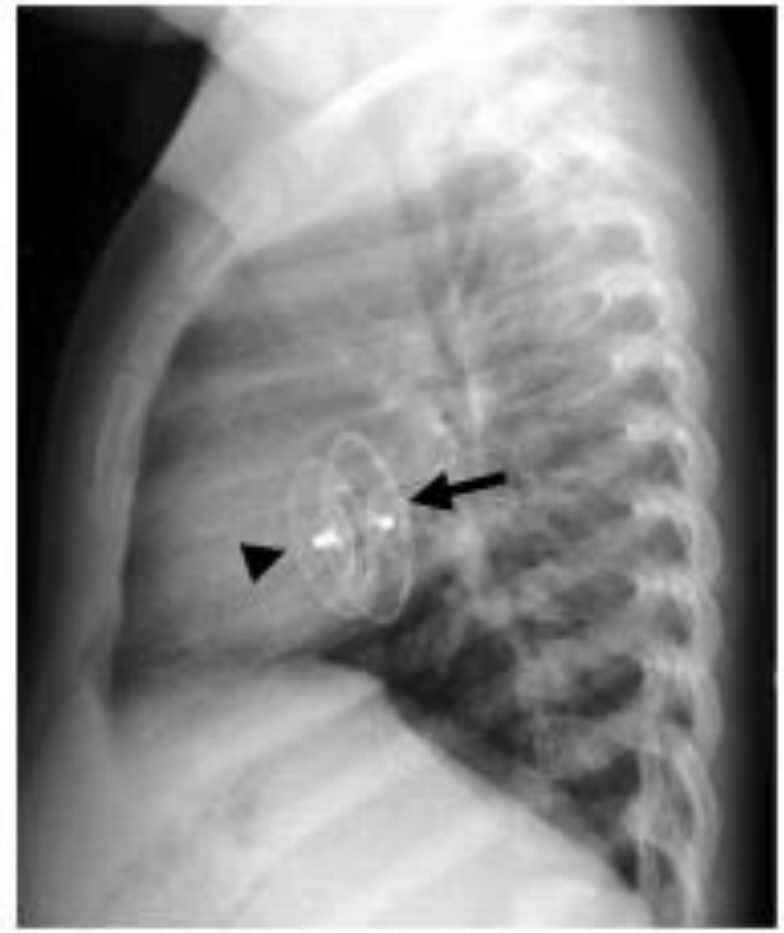


1--Deployment of LA disc



3--Deployment of RA dis





Atrial septal defects

TIMING

- Echo diagnosis to first cardiac evaluation
  - Infants/neonates: 3-6 months
  - Children/teens: non urgent
- Intervention:
  - Around age 4 years
  - Infancy if a symptomatic premature neonate
- Pulmonary hypertension can develop in 20s and 30s if large ASD is not repaired*

INDICATON FOR INTERVENTION

- Right heart enlargement
- Large size

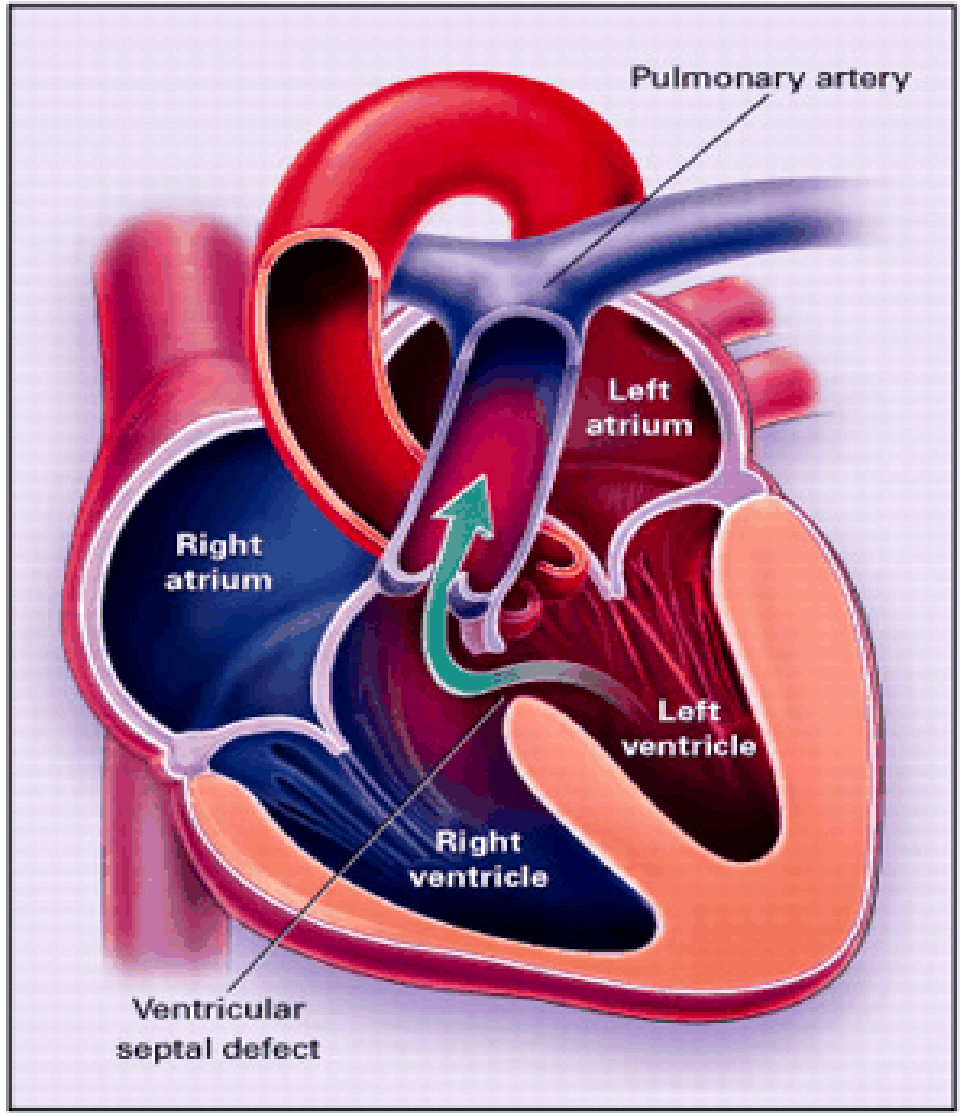
Failure to thrive

You are admitting a 4-month-old for evaluation of failure to thrive. He sweats with feeds and takes 40 min to take 2 oz. On exam he appears comfortably tachypneic. You hear a low pitched holosystolic murmur in the left lower sternal border radiating to the apex. His liver is down 3 cm. Chest xray shows cardiomegaly. Diagnosis?

VSD

Ventricular septal defect (VSD)

- Most common form of congenital heart defects
- Several types
- Presentation determined by VSD size and PVR
  - Asymptomatic if small



Clinical Presentation: Moderate to Large VSD

Symptoms occur as PVR falls

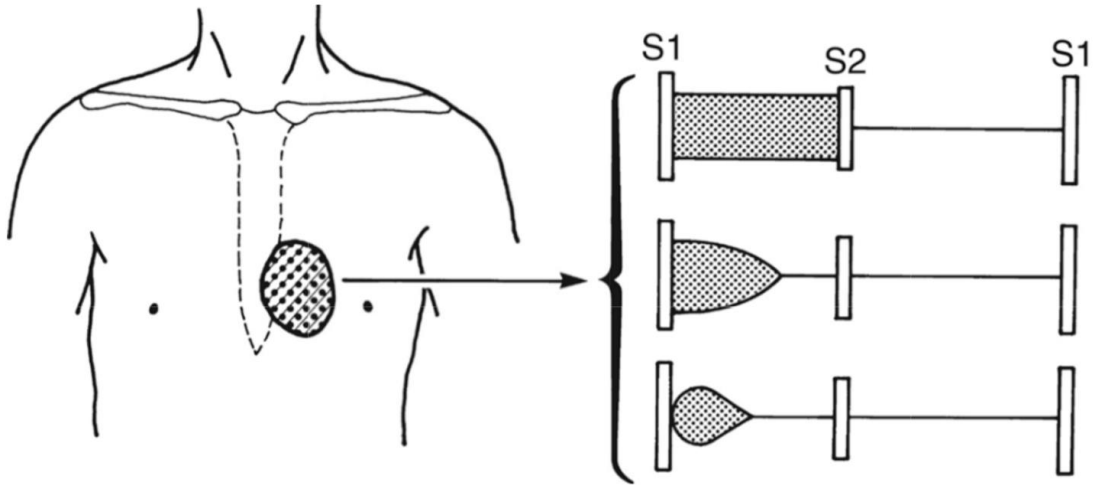
- PVR high during fetal life
- Decreases by 50% in the first 24 hours
- Falls to adult level at 6-8 weeks of life

- Symptoms develop around 6 – 8 weeks of age
- Delayed growth and development
- Decreased feeding/activity tolerance
- Recurrent pulmonary infections

VSD: Physical Examination

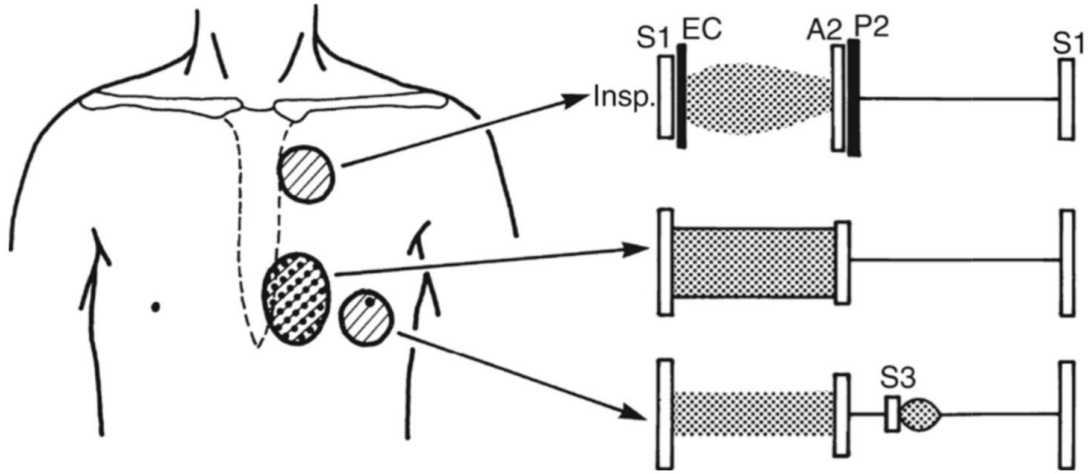
Small VSD

- Asymptomatic
- Normal ECG
- No cardiomegaly



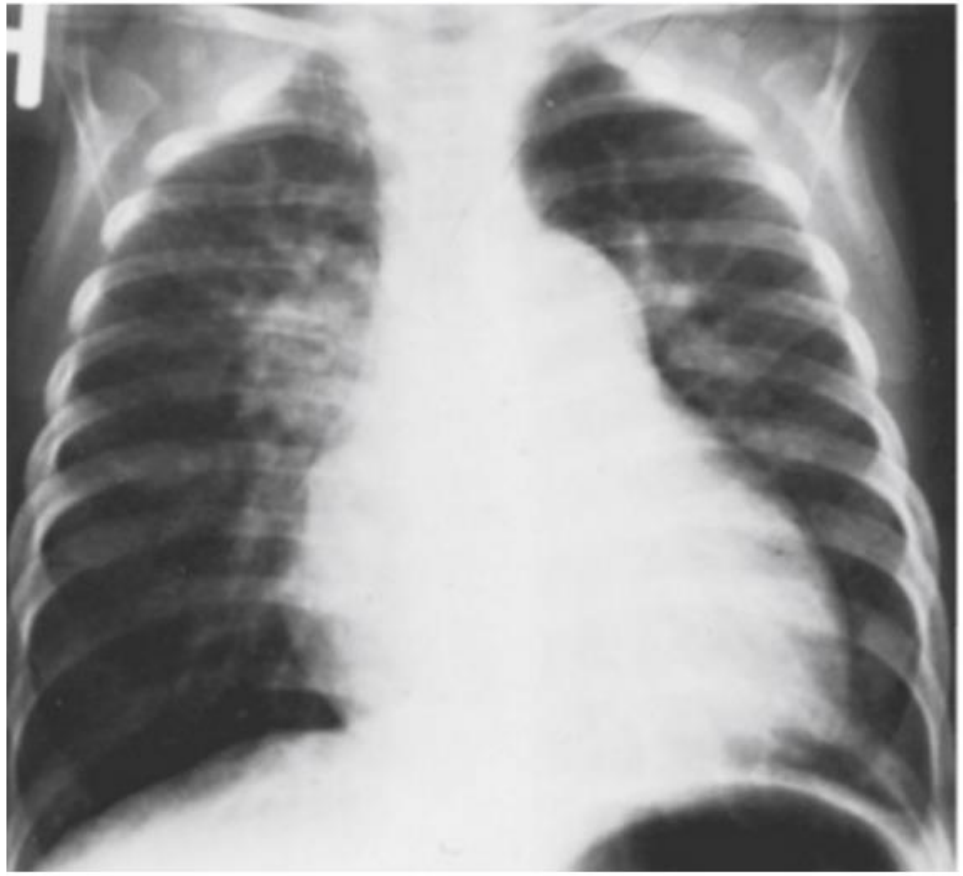
Large VSD

- Tachypneic
- Hepatomegaly
- Underweight

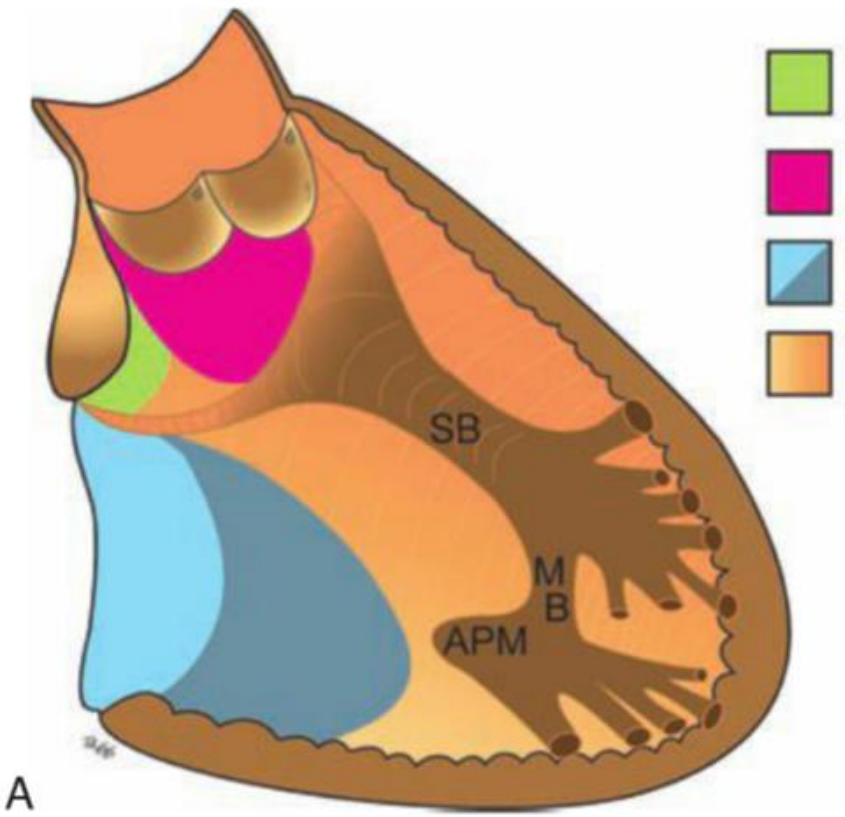


ECG and CXR : Large VSD

- ECG:
  - LVH / LAH
  - Might have biventricular hypertrophy
- Chest Xray:
  - Cardiomegaly
  - Increased pulmonary vascular markings

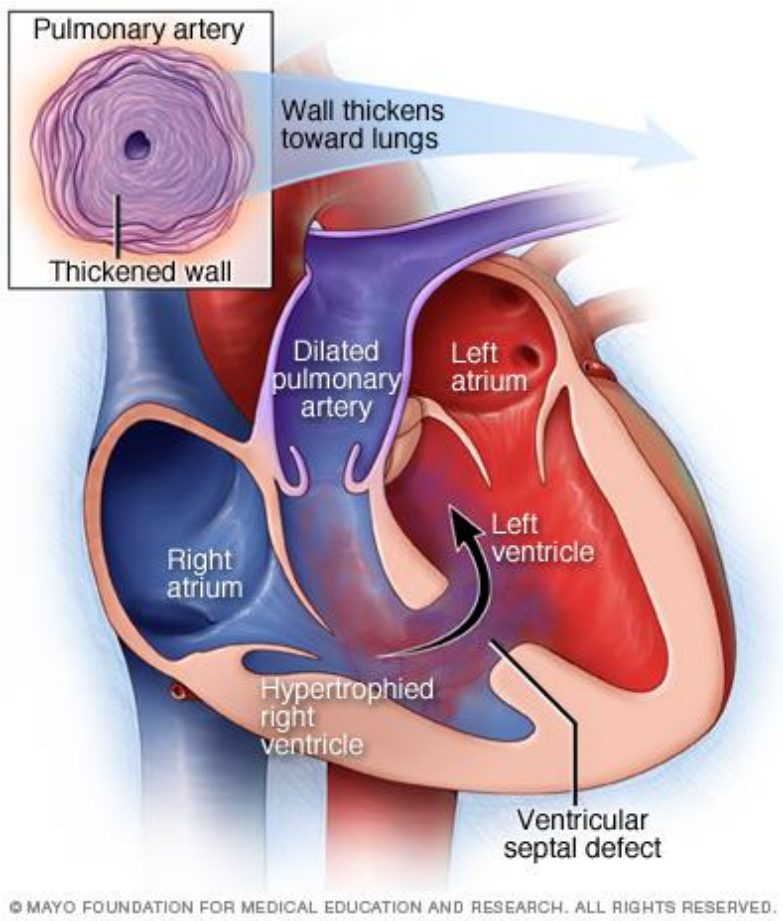


Natural History



- 60% of small to moderate muscular VSDs close spontaneously
- 35% of small perimembranous VSDs close spontaneously
- Inlet / outlet VSDs do not close spontaneously
- Large VSDs
  - Pulmonary overcirculation develops ~ 6 -8 weeks in large VSDs
  - Pulmonary hypertension can develop as early as 6 – 12 months

Pulmonary Hypertension and Eisenmenger Syndrome



- Cyanosis and clubbing
- Single loud S2 on exam
- ECG: RVH
- CXR :
  - PA enlargement
  - Decreased pulmonary blood flow
  - Normal heart size

Management: Large VSD

- Medical*
  - Diuretics
  - Nutritional support
    - Fortification
    - NG tube
    - Weight checks
- Surgical
  - Age 4 – 6 months
  - Indicated if symptoms or significant pulmonary overcirculation
  - Contraindicated if PVR significantly elevated
- Interventional
  - Option if anatomy favorable and patient ~10 kg

Ventricular Septal Defects

TIMING

- Echo diagnosis to first cardiac evaluation
  - Neonate/infant (large VSD): 4 weeks (sooner if in heart failure)
  - Neonate/infant (small VSD): 3-6 months
  - Child/teen (small VSD): non urgent
- Intervention
  - 4-6 months (no later than 12 mo): large shunt, heart failure, not device candidate
  - Unlikely to close if patent at 2 years

INDICATION FOR INTERVENTION

- Persistent moderate/large defects
- Pulmonary hypertension*
- Persistent high velocity small lesion that is a good device candidate

PRE-OP MANAGMENT

- Large defects: regular weight checks
- Synagis if on diuretics

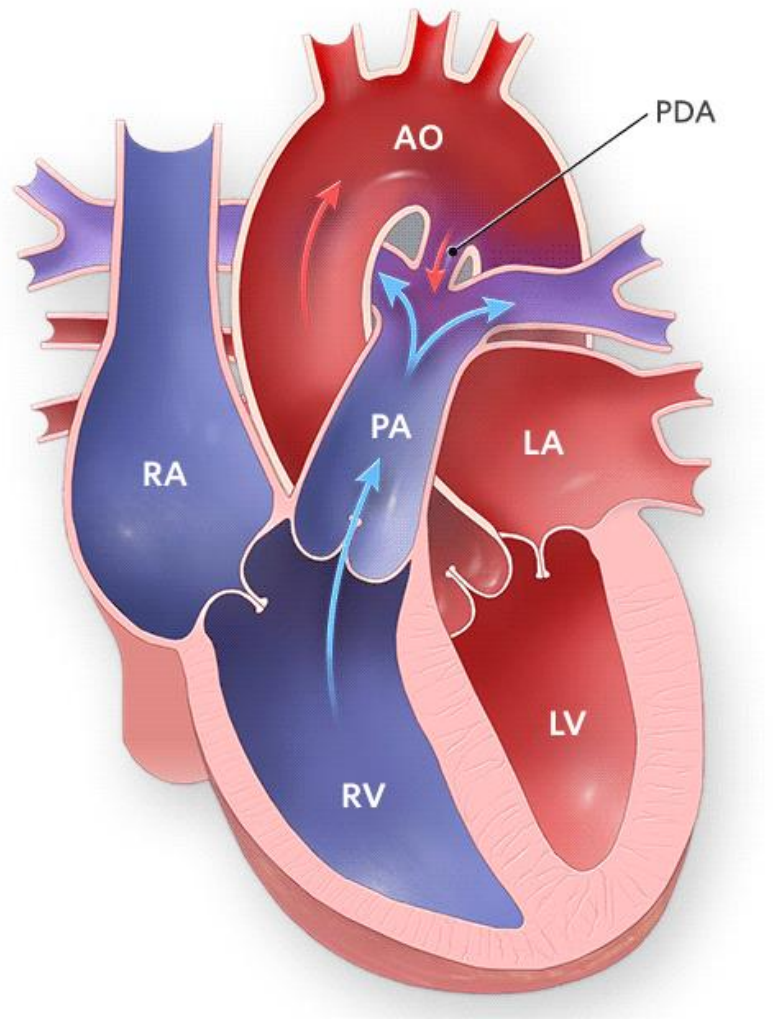
Diagnosis : Murmur

You see a 3-year-old female for a well child check. She has been growing and developing normally and parents have no complaints. You auscultate a “to and fro” murmur in the left upper sternal border. The remainder of her exam is normal. Diagnosis?

PDA

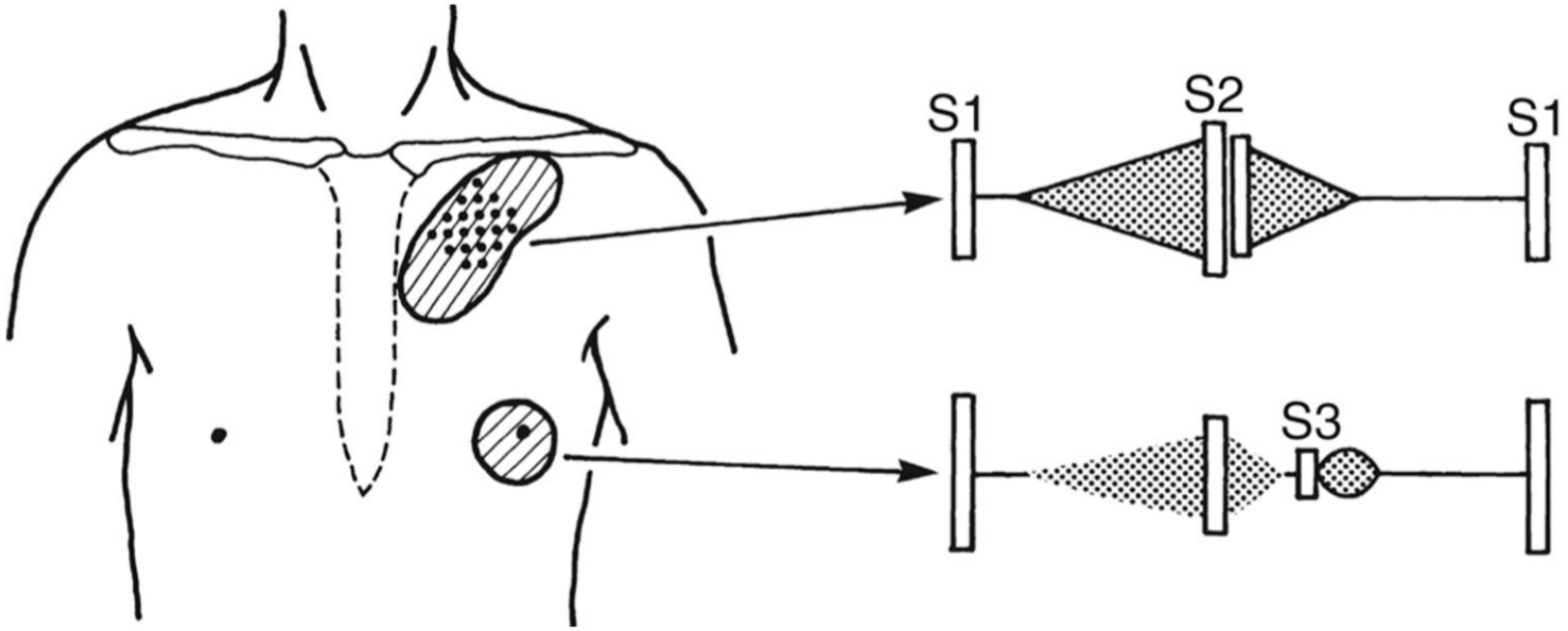
Patent Ductus Arteriosus (PDA)

- 5 – 10% of all CHD (excluding premies)
- Females > males
- Common in premies
- Asymptomatic if small
- Signs of pulmonary over-circulation if large



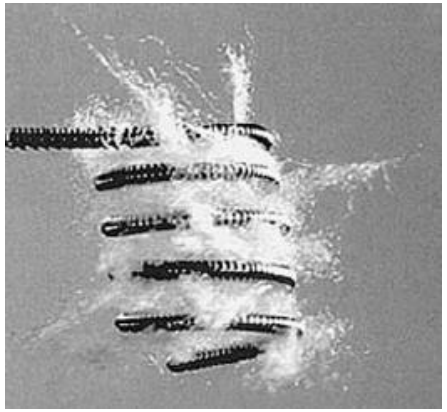
Physical Exam : Mod to Large PDA

- Bounding peripheral pulses
- “Machine-like” murmur
- If pulmonary hypertension -> differential cyanosis
- ECG and CXR findings similar to VSD



PDA: Natural History / Management

- Spontaneous closure rare in full term infants and children
- Pulmonary over circulation develops if shunt is large
- May develop pulmonary hypertension if untreated
- Surgical / cath lab closure





Premature infants with PDA

- Clinical evidence of PDA in:
  - 45 % of infants BW < 1750 g
  - 80% of infants BW < 1200 g
- Ductal tissue is immature
- Management
  - If small PDA and no symptoms: observe; may close spontaneously
  - If symptomatic, pharmacologic, surgical or device closure indicated
    - Indomethacin or ligation through lateral thoracotomy

More murmurs

You see a 4-year-old girl in your clinic for the first time and notice a 2/6 non-vibratory systolic ejection murmur loudest in the left upper sternal border. The murmur radiates to her back. There is no diastolic component. She is completely asymptomatic.

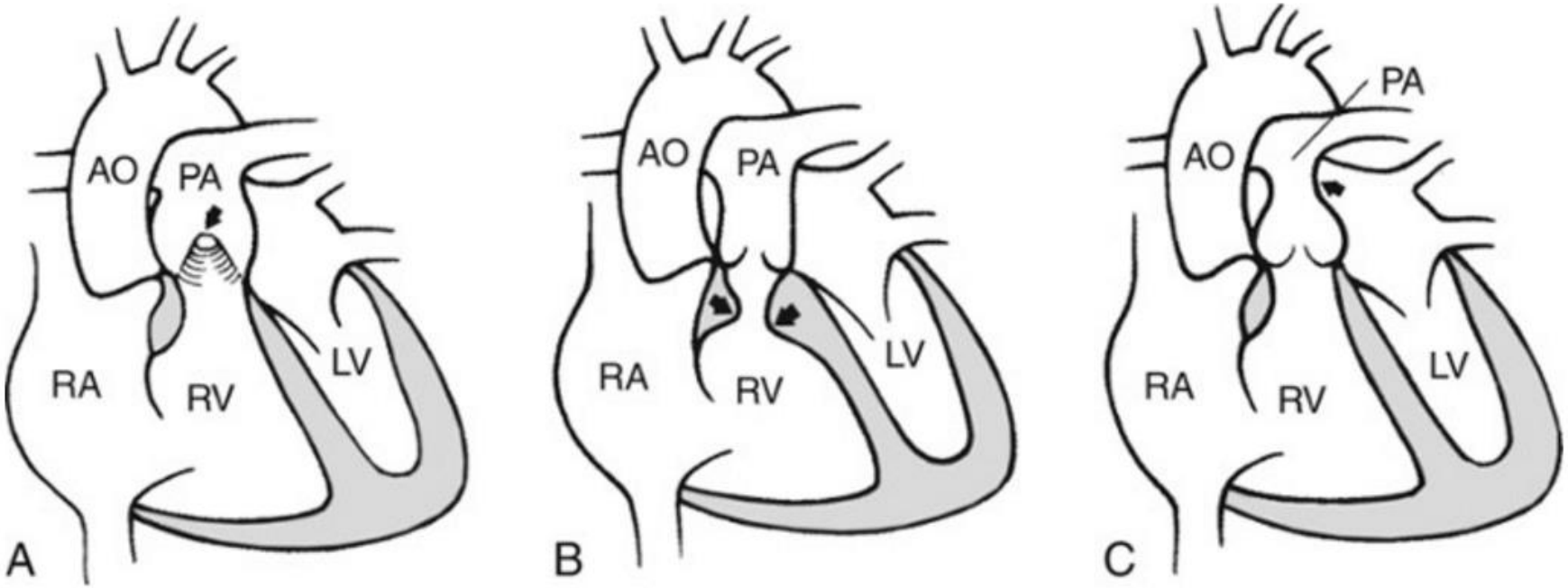
Diagnosis?

Pulmonary valve stenosis

Pulmonary stenosis

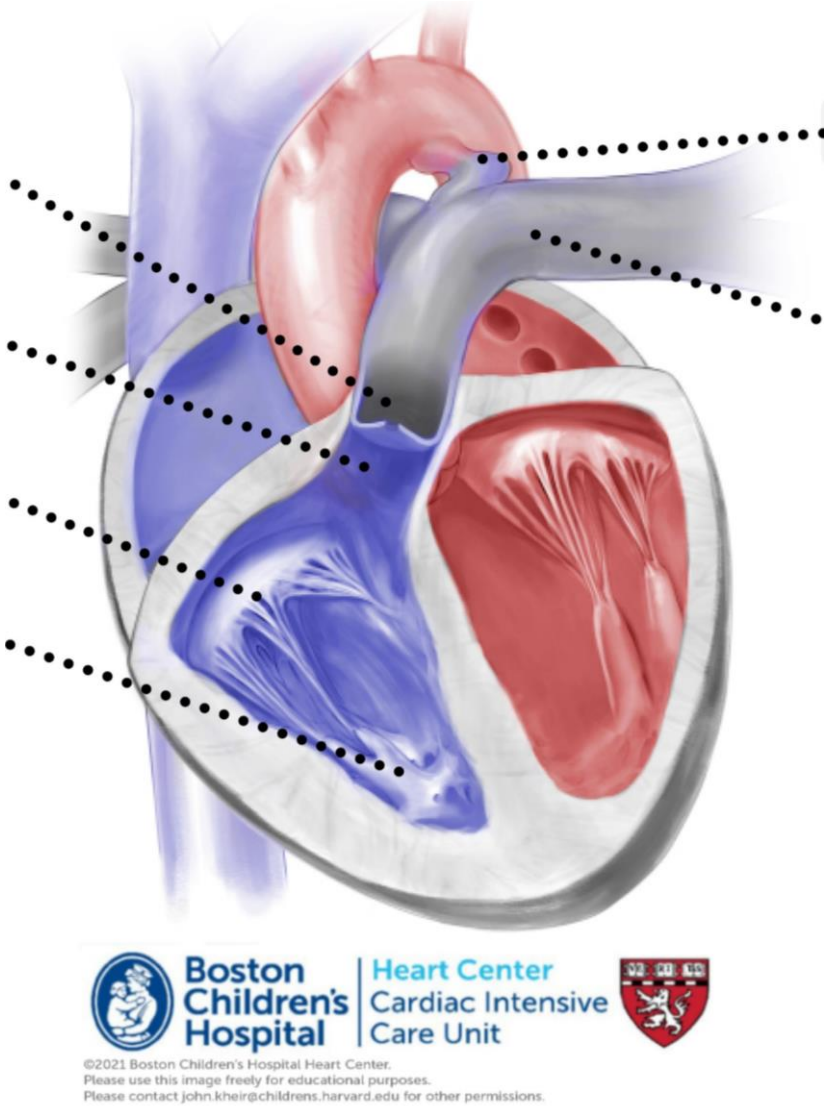
Pulmonary Stenosis

- Isolated PS in 8-12% of CHD
- Valvular, subvalvular, supravalvular, or within the RV cavity

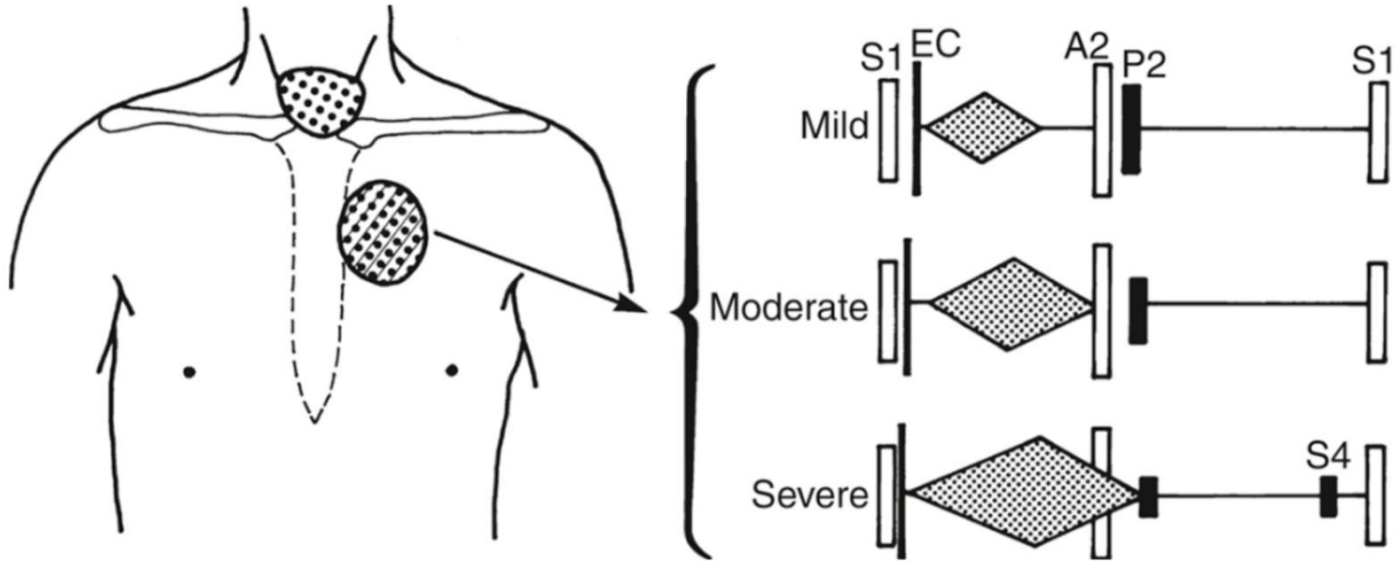


PS: Clinical Presentation

Critical	• Poor feeding • Tachypnea • Cyanosis
Moderate/ Severe AS:	• Exertional chest pain, exercise intolerance, heart failure symptoms
Mild	• Asymptomatic

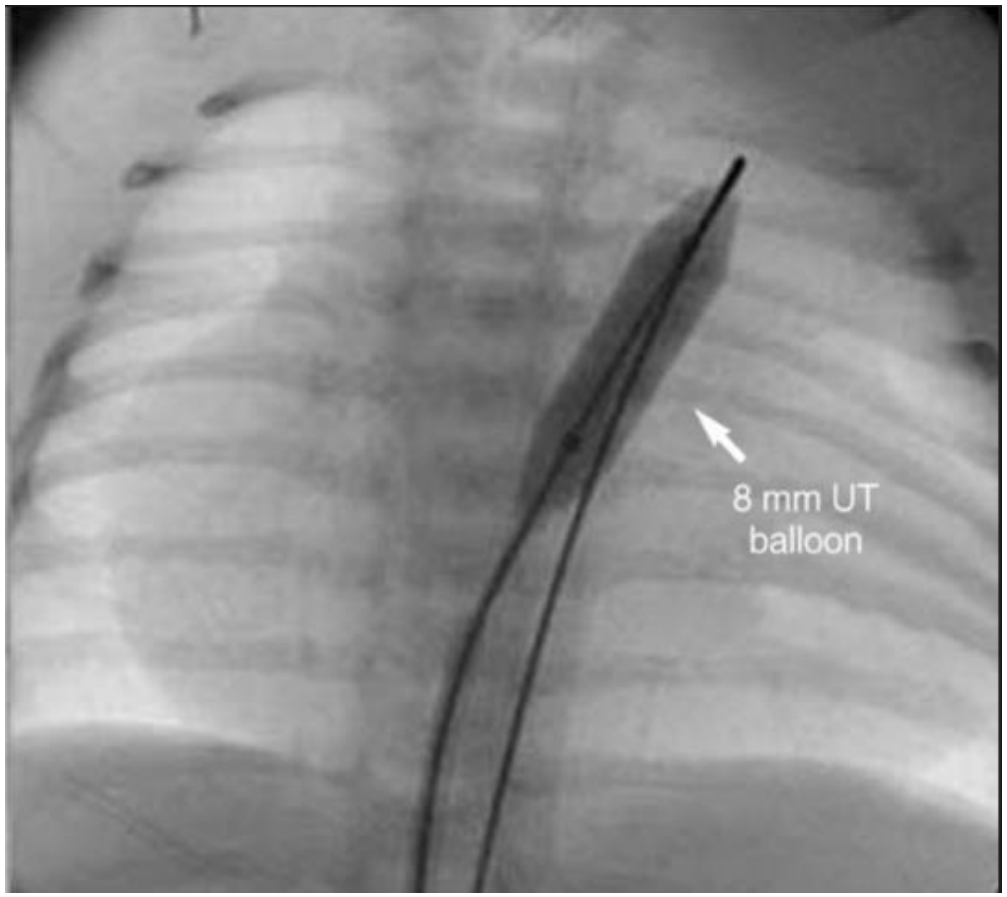


PS: Exam, ECG and CXR



- Mild : Normal ECG and CXR
- Moderate: RAD and RVH, MPA segment may be prominent Severe: RAH and RVH with strain, decreased pulmonary vascular markings
- Critical: LVH , oligemic lung fields, +/- cardiomegaly

PS: Natural progression/ Management



- Critical PS :
  - PGE, balloon valvuloplasty, ductal stent or surgical shunt
  - Patient will die without intervention
- Moderate and severe PS:
  - Severity progresses with age
  - Balloon valvuloplasty
- Mild PS
  - Usually not progressive, and usually no intervention required

PDA

TIMING

- Echo diagnosis to first cardiac evaluation
  - Non urgent outpatient follow up if no heart failure symptoms
- Unlikely to close if full term or premie over the age of 6 months.

INDICATION FOR INTERVENTION

- Left heart enlargement
- Heart failure symptoms
- Pulmonary hypertension

Pulmonary stenosis

TIMING

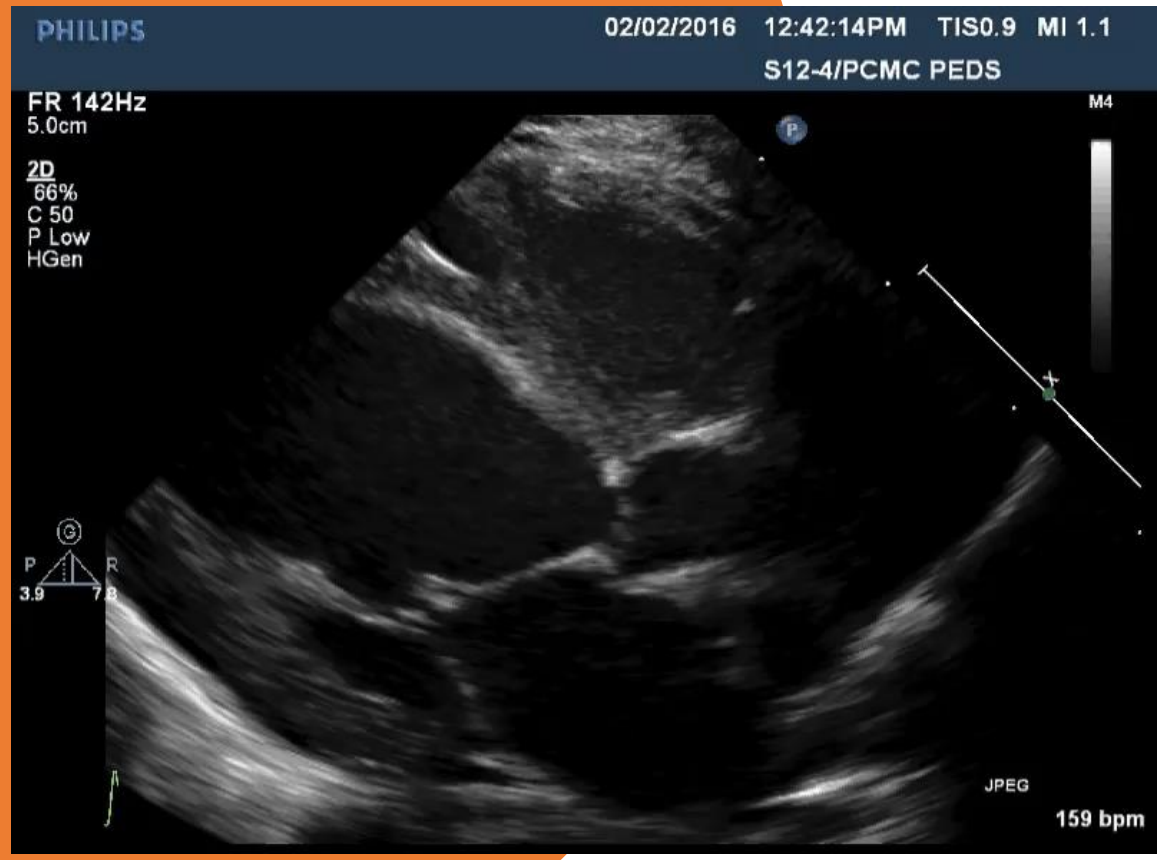
- Echo diagnosis to first cardiac evaluation
  - Infant under 2 months: 4 weeks
  - Older infant/child/teen: routine

INDICATION FOR INTERVENTION

- Left heart enlargement
- Moderate to severe disease with RVH or symptoms

- WHAT NOT TO MISS
- PS can worsen when the PVR falls

Mommy call nightmare



You are a young attending taking mommy call for the first time. You receive a call from a mother of a previously healthy 1 week old who has been pale, irritable, with poor feeding all day. You send them to ED and they are admitted. While stalking the chart the next day, you see the following echo image. Diagnosis?

(Critical) aortic stenosis

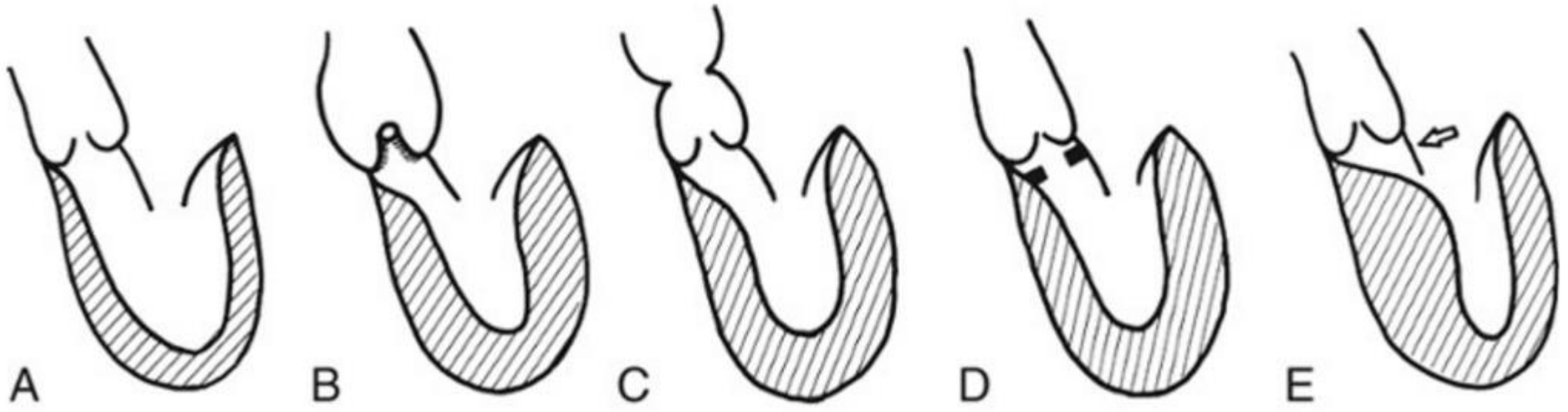
Left heart lesions

Aortic Stenosis

Left ventricular outflow tract obstruction 10% of CHD

71% valvular AS

Males > female



AS: Clinical Presentation

Critical
- Poor perfusion
- Respiratory distress
- Symptoms days to weeks of birth

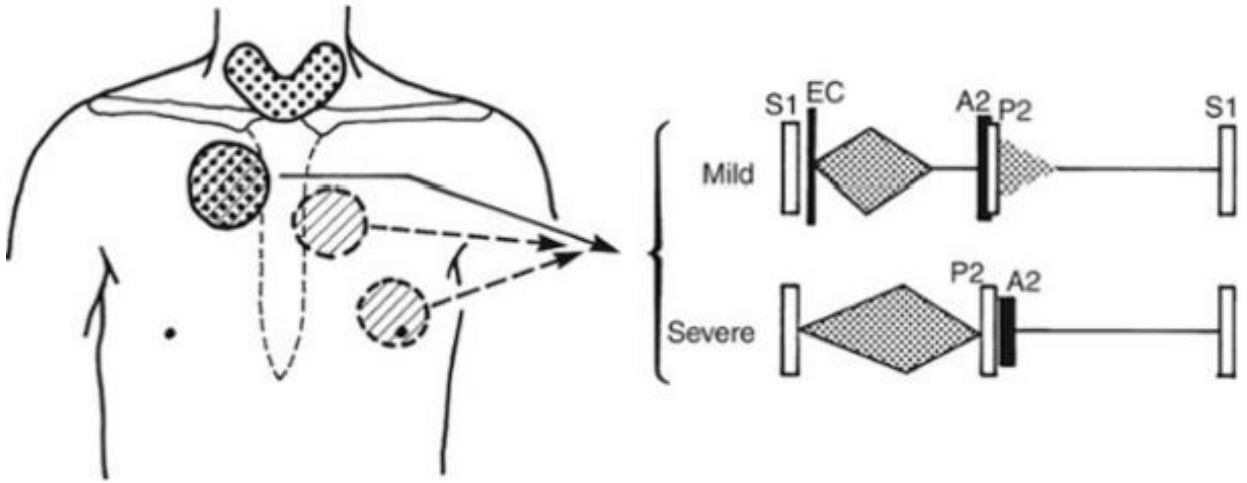
Severe AS:
- Chest pain, easy fatigability or syncope

Mild to moderate AS
- Often asymptomatic
- May have exercise intolerance

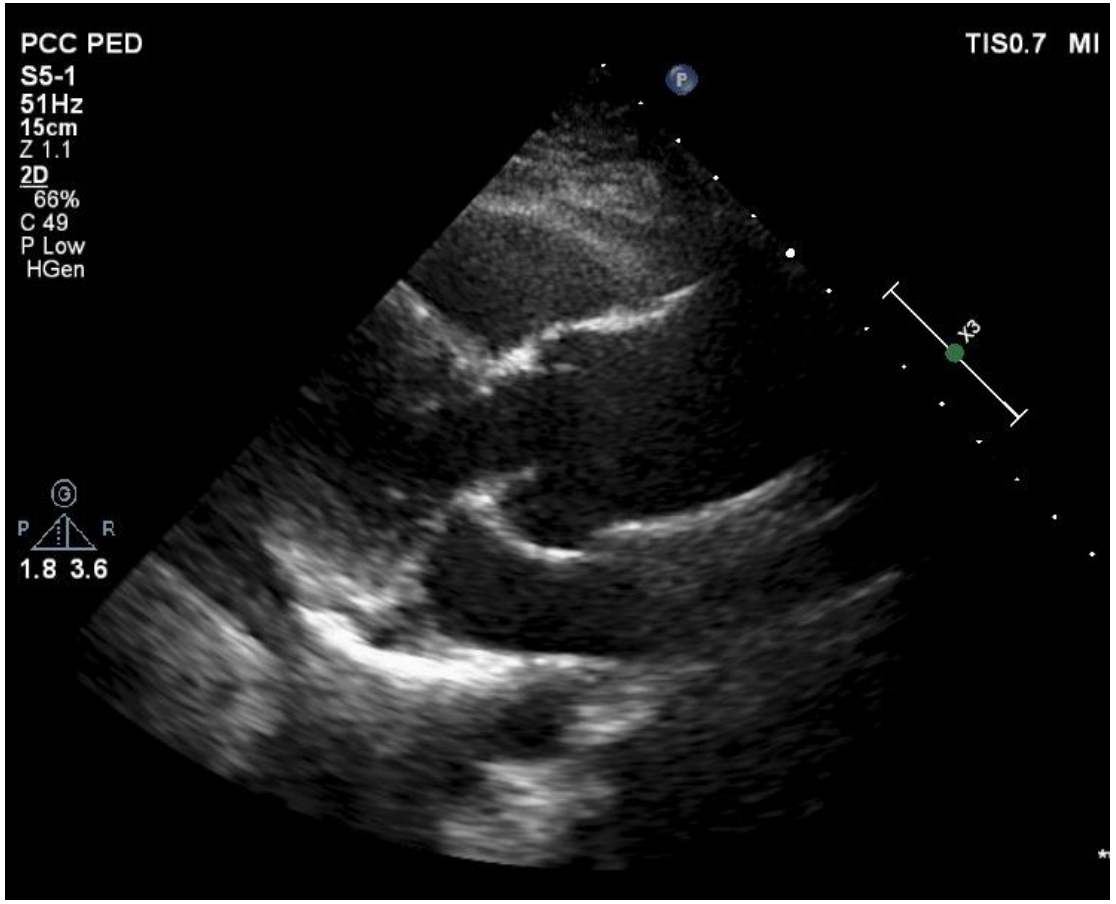
- ECG :
  - Normal in mild cases, LVH if severe
- CXR:
  - Usually no cardiomegaly, can have post stenotic ascending aorta dilation
  - Newborns with critical AS have cardiomegaly with pulmonary venous congestion

AS: Physical Exam

- Acyanotic and normally developed
- Narrow pulse pressure in severe AS (otherwise usually normal)
- Supravalvular stenosis may have R arm BP > L arm BP
- May have elfin facies (Williams syndrome)
- Critical AS can present in shock as ductus closes
  - May not have a murmur

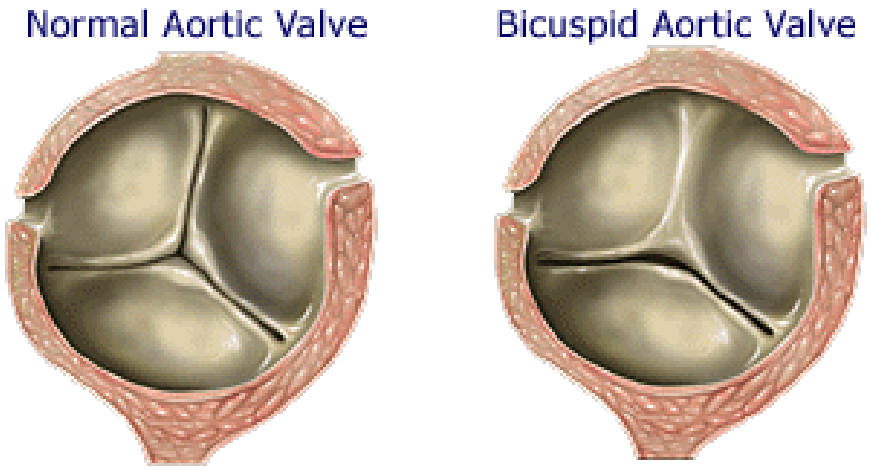


AS: Natural Course and Management



- Severe AS :
  - Chest pain, syncope, and even sudden death (1%–2% of cases)
  - Heart failure
- If patient has a bicuspid aortic valve (BAV)
  - Progressive AS and aortic root dilation
  - Progressive AI
- Management may include balloon valvuloplasty or surgery

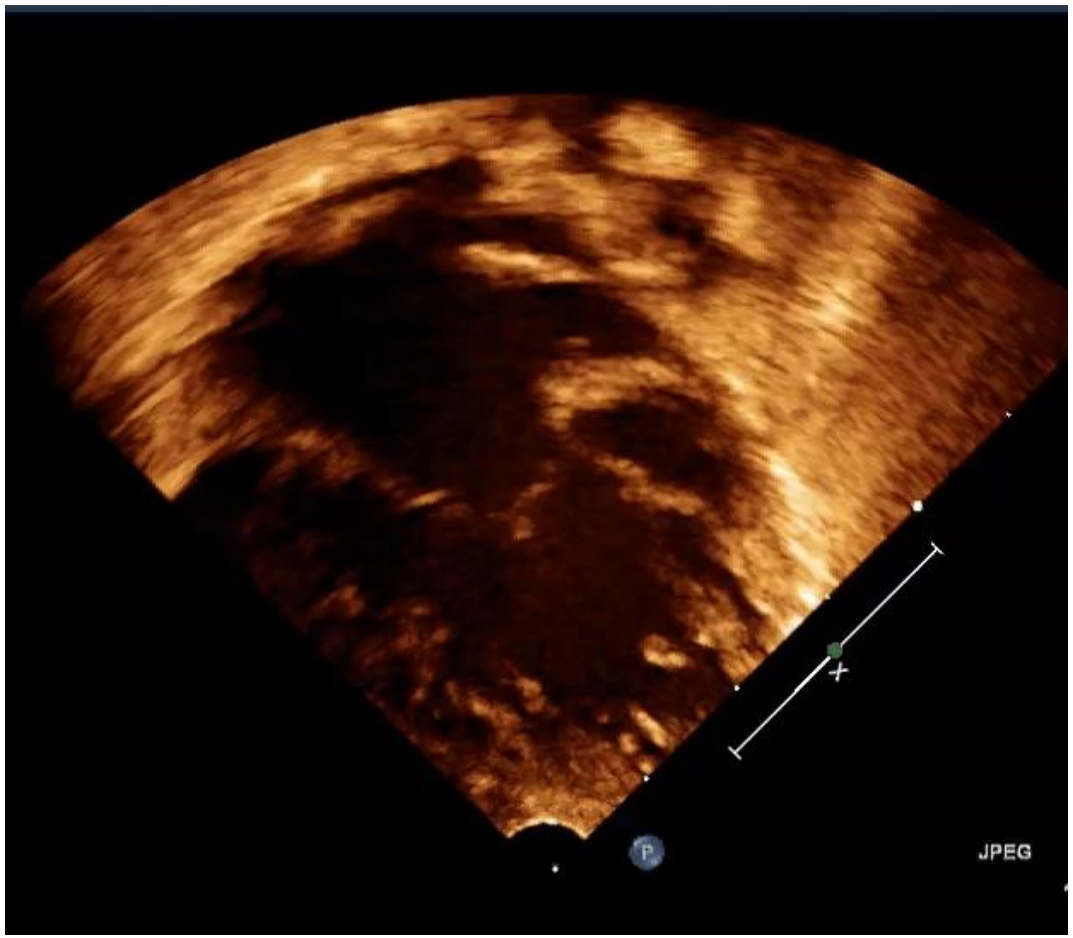
Bicuspid aortic valve (BAV)



- Most common cause of aortic stenosis (75%)
- May hear click and diastolic rumble.
- Isolated AI (younger patients)
- Early calcification in 20s and 30s
  - -> worsening AS and AI
  - May need valve replacement

Hypoplastic Left Heart Syndrome

- 1 : 5000 live births
- Males > females
- 23 % of cardiac deaths in the first week of life, 15% in the first month
- Presentation
  - SHOCK/Poor systemic perfusion
  - Pulmonary overcirculation (tachypnea, dyspnea, crackles, pulmonary edema)
- Management: PGE, surgery



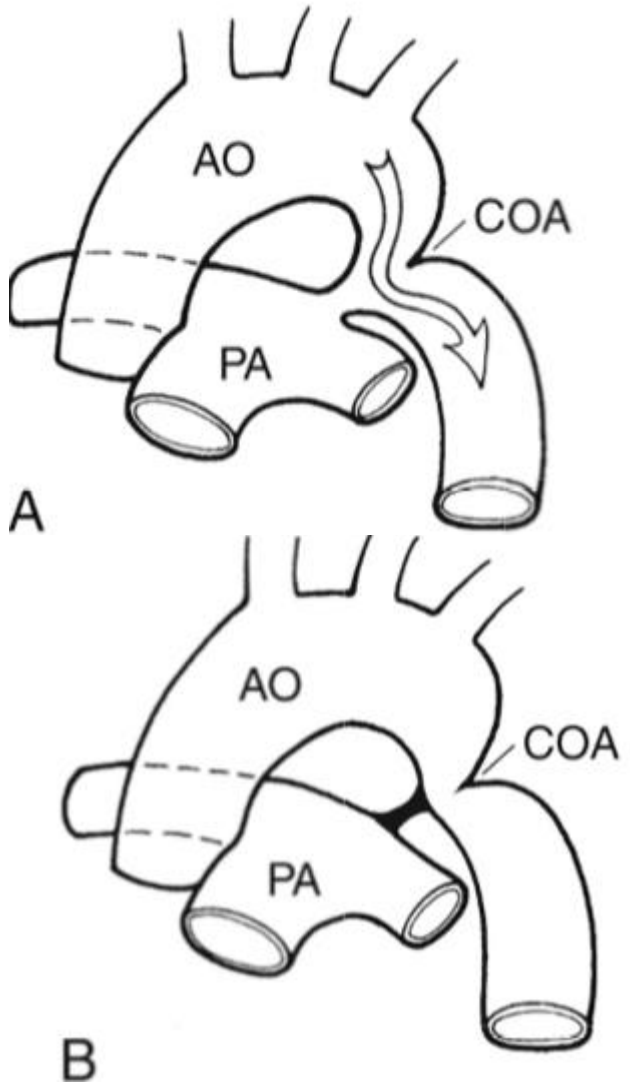
Just another day in clinic....

You see a 2-week-old male in clinic for a well child check. Femoral pulses are 1+ bilaterally. The upper extremity systolic blood pressures are 20 points higher than the lower extremity blood pressures. What should you do?

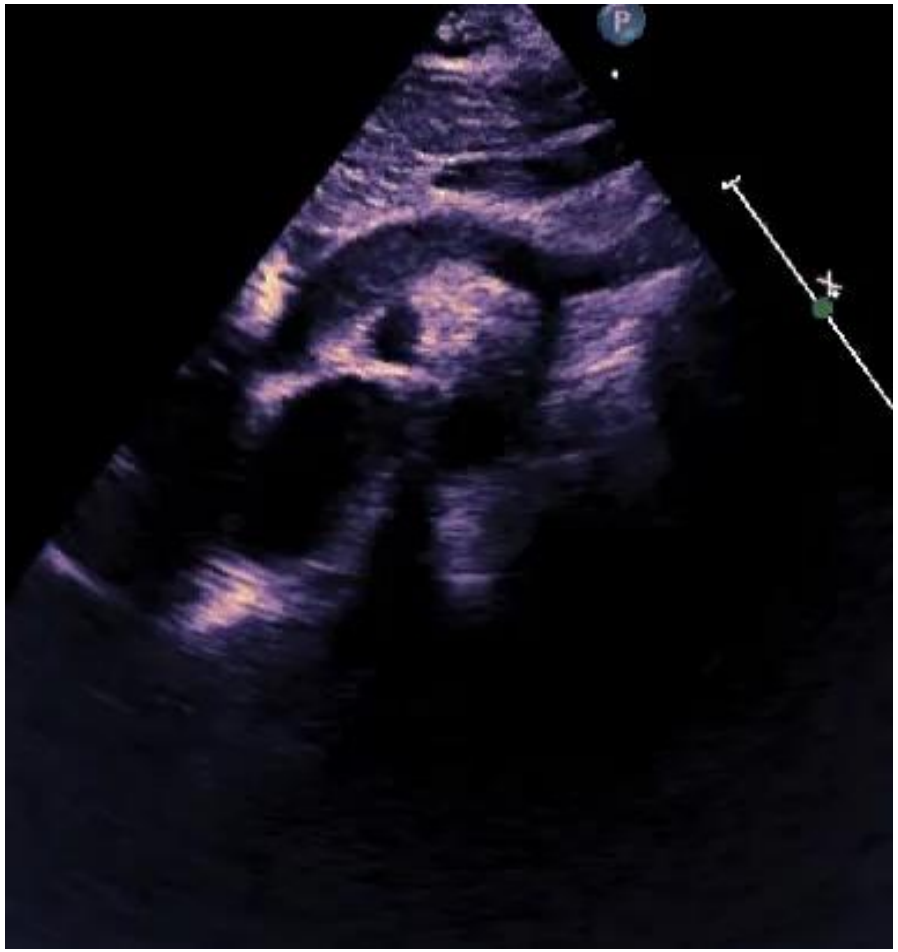
A. Have the patient follow up in your clinic in 1 week; 20 points isn't so bad
B. Schedule an outpatient appointment with cardiology for the next day next week.
C. Call cardiology directly and speak to someone about arranging for an echo today.
D. Have the patient follow up in your clinic at 2 months for their next well child check.

Coarctation of the aorta

- Occurs in 8-10% of congenital heart disease cases
- 30 % of Turner syndrome patients have CoA
- 50 – 85% of patients with CoA have bicuspid aortic valve
- More common in males (2: 1)
- Usually juxtaductal



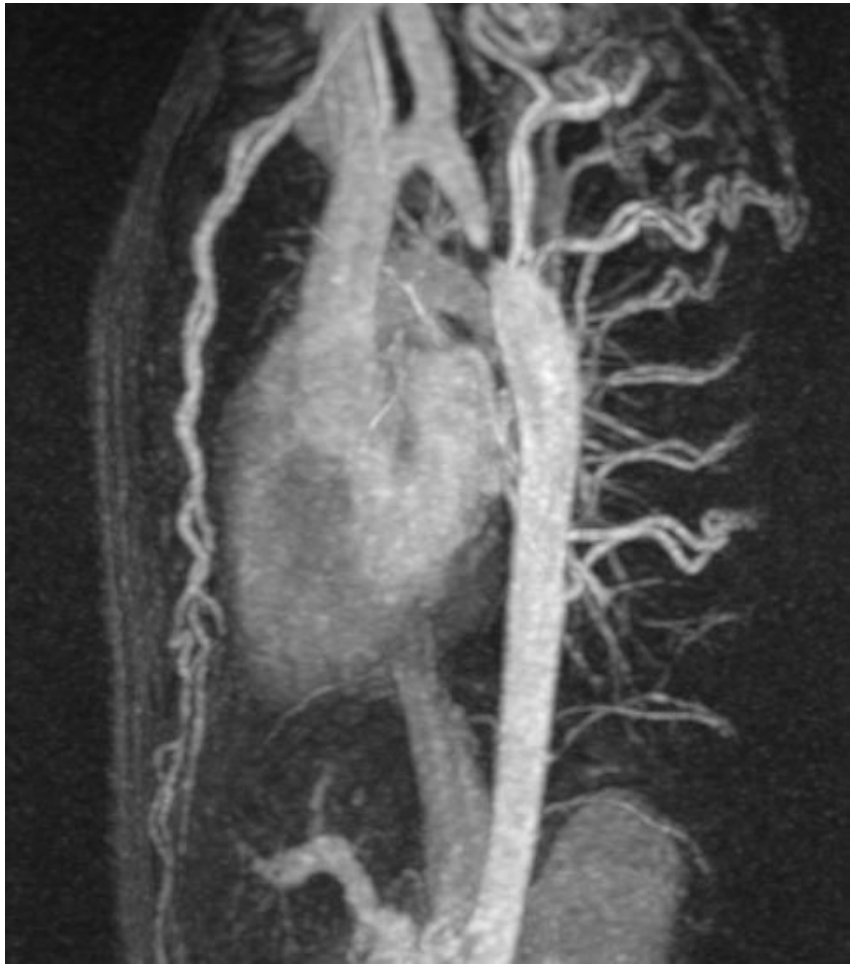
Co A Presentation: Symptomatic Infants



- Duct is the source of systemic blood flow (no good collaterals)
- Decreased peripheral pulses, blood pressure differential
- Poor feeding, dyspnea, poor urine output, acidosis, shock
- Single loud S2 on exam, gallop
- 50% sick infants do not have a murmur
- RVH or RBBB on EGC

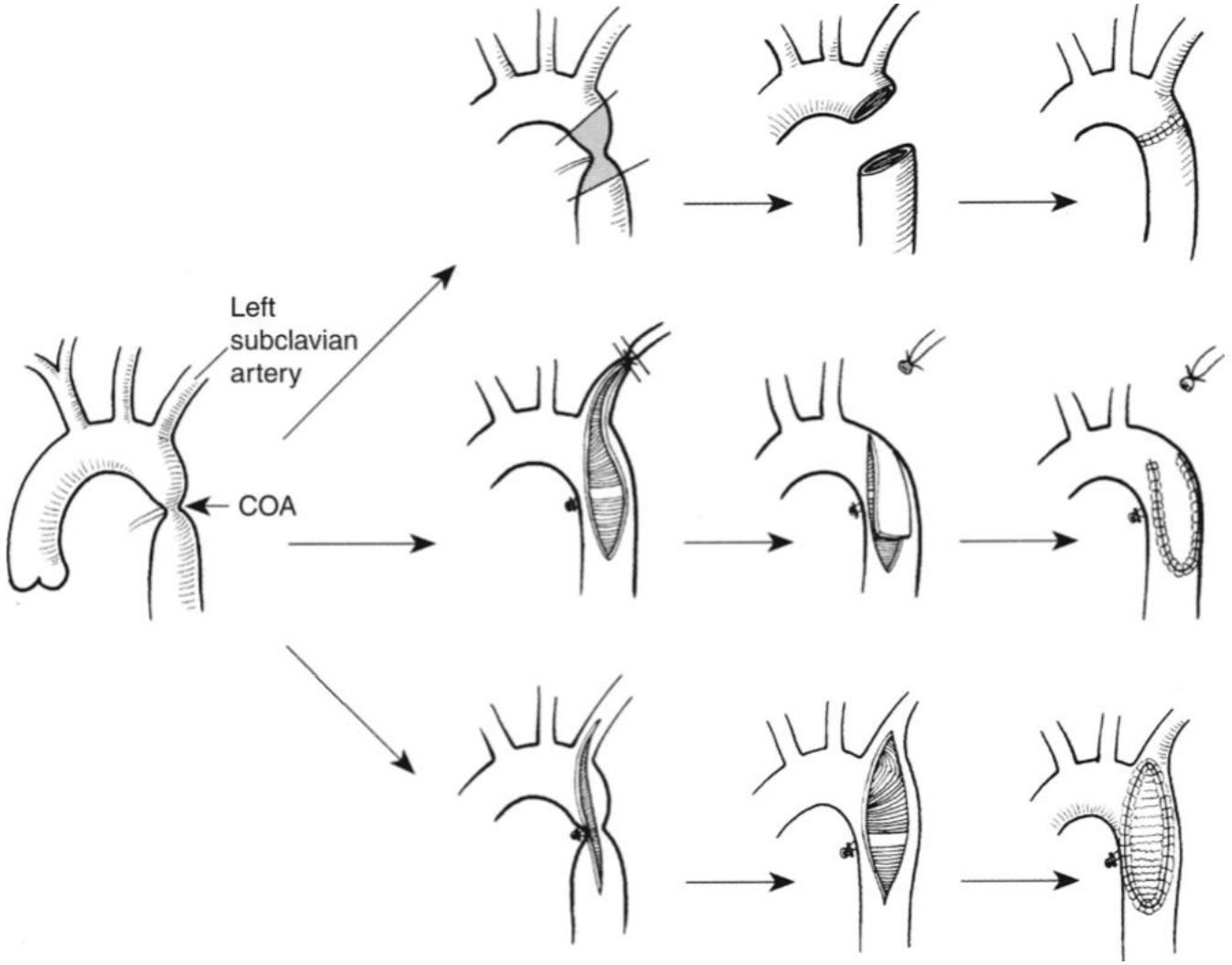
Co A: Asymptomatic infants and children (and teens and adults)

- Can be asymptomatic due to good collateral circulation
- Lower extremity pulses absent or delayed.
- 2-4/6 SEM RUSB or LM-LLSB, → back
- May have systolic thrill SSN, accentuated A2 or ejection clinic (from BAV or HTN)
- Hypertension, pain/weakness in legs after exercise
- Leftward axis LVH on ECG



Co A: Management

- Initial management
  - Start PGE
  - May need inotropic agents
  - Treat hypertension
- Surgical repair
- Balloon angioplasty / stent placement



Co A: Post op / Follow up

- Post op complications
  - Recoarctation (uncommon if surgery performed after 2 years of age)
  - Abdominal pain
- Risk of late hypertension
  - More common if repaired after late childhood
  - May be as high as 10-20%, even if repaired in infancy
- Aortic aneurysms
  - More common in patch repairs and balloon angioplasty
- Patients need regular cardiology follow up

Aortic Stenosis and Coarctation of the Aorta

TIMING

- Echo diagnosis to first cardiac evaluation
  - Infants: Urgent (especially if ~2 weeks old)
  - Children/teen: Within a few weeks of diagnosis*
- Intervention

INDICATION FOR INTERVENTION

- Severe stenosis or symptoms (AS)

WHAT NOT TO MISS

- Poor femoral pulses
- Hypertension
- Low blood pressure in the lower extremities

Take home points

- Left to right shunts
  - Can result in pulmonary overcirculation (clinical diagnosis)
  - Prolonged overcirculation can -> pulmonary htn and eisenmenger physiology
- If a neonate presents in shock, think of ductal dependent lesions
  - Critical PS and AS require balloon valvuloplasty, coarctation of the aorta need surgery.
- Patients with repaired cardiac lesions need regular follow up with cardiology
- If you're not sure what to do, ask

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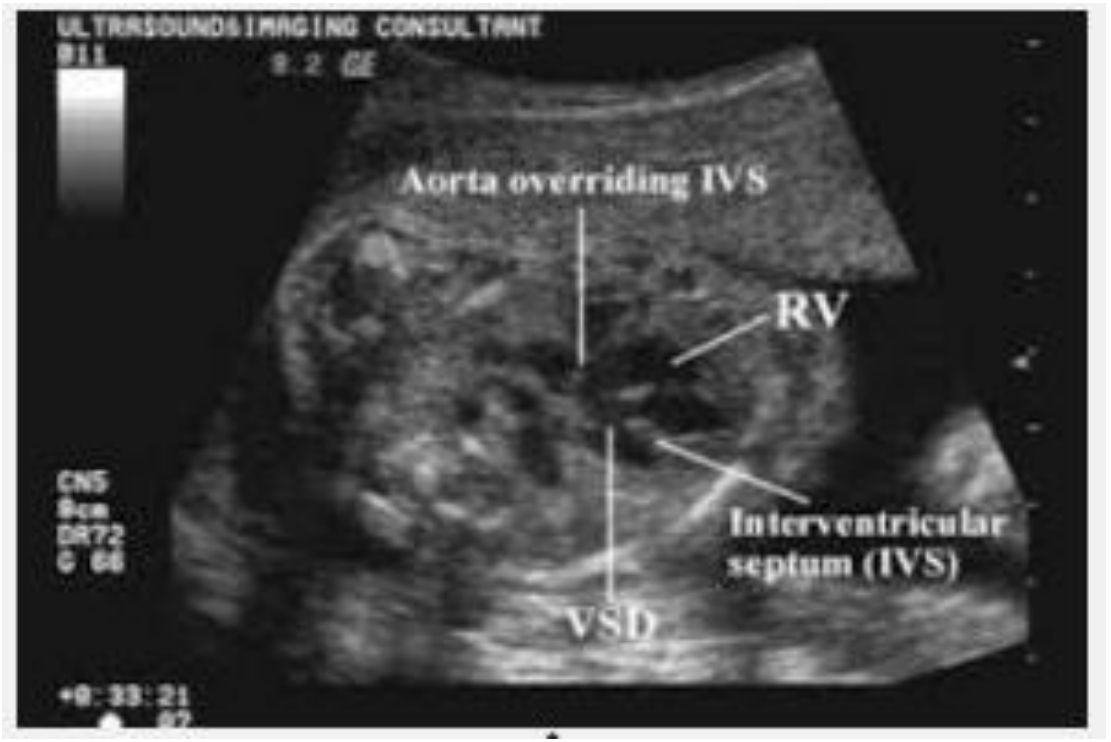
Questions?

What about cyanotic heart disease?



- Most cyanotic heart disease is diagnosed prenatally
  - Tetralogy of Fallot diagnosed prenatally in 70-90% of cases
- Many have surgery as a neonate
- Acyanotic lesions can also be diagnosed on fetal echocardiogram
  - AVSD, large ASD/VSD, coarctation of the aorta, severe valve stenosis, etc
  - Fetal echo can miss subtle BAV and pulmonary vein anomalies

Indications for SBE prophylaxis

- Cyanotic heart disease
- Cardiac surgery within the last 6 months
- ASD or VSD device placed within the last 6 months
- Residual VSD or ASD near surgical patch or cath device after 6 months
- History of cardiac transplant
- Single ventricle physiology
- Significant aortic valve disease*

Who needs Synagis?

- Babies on medication for pulmonary overcirculation
- Babies with cyanotic heart disease

PFO

- Normal variant present in up to 25% of adults
- Usually closes by 1 year of age
- Not audible on exam