

Congenital Heart Disease

An Approach for Simple and Complex Anomalies



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Disclosures

- Consultant to Fuji Medical Imaging

ASCeXAM

- Contains questions on general congenital heart disease, not “adult” CHD
- Study guide contain all of the information in this talk plus addition topics that will be helpful for the exam
- There have been a few questions on fetal echo which have appeared on the ASCeXAM which are covered in the handout
- Insider information provided in study guide – topics that have appeared on prior exams (last page of study guide)

Which heart defect is more common?

19% 1. Tetralogy of Fallot

20% 2. Transposition of the Great Arteries

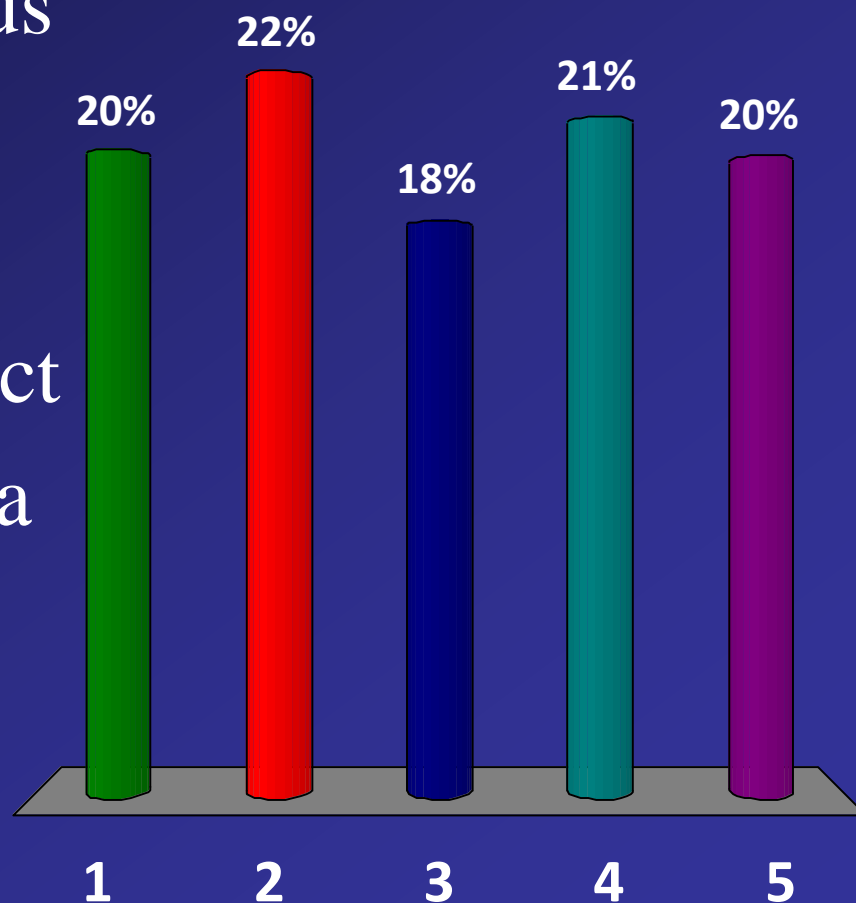
22% 3. Aortic Stenosis

19% 4. Coarctation of the Aorta

19% 5. Atrioventricular Canal

What is the most common defect seen with Down syndrome (trisomy 21) ?

1. Patent Ductus Arteriosus
2. Tetralogy of Fallot
3. Aortic Stenosis
- ✓ 4. Ventricular septal defect
5. Coarctation of the Aorta



Congenital Heart Disease

Spectrum of Congenital Heart Disease - Incidence

- 0.5–0.8% of live births *
- >30,000 individuals/year in U.S.
- 50% simple shunts (ASD, VSD, PDA)
20% simple obstruction
30% complex

* Excludes MVP (4-6%) & Bicuspid AV (1-2%)

Congenital Heart Disease

Spectrum of Congenital Heart Disease - Frequency

<u>Cardiac Malformation</u>	<u>% of CHD</u>	<u>M:F Ratio</u>
Ventr. Septal Defect	18-28	1:1
Patent Ductus Arter.	10-18	1:2-3
Tetralogy of Fallot	10-13	1:1
Atrial Septal Defect	7-8	1:2-4
Pulmonary Stenosis	7-8	1:1
Transp. of Grt. Art.	4-8	2-4:1
Coarctation of Aorta	5-7	2-5:1
AV Septal Defect	2-7	1:1
Aortic Stenosis	2-5	4:1
Truncus Arteriosus	1-2	1:1
Tricuspid Atresia	1-2	1:1
Tot. Anom Pulm Veins	1-2	1:1

Congenital Heart Disease

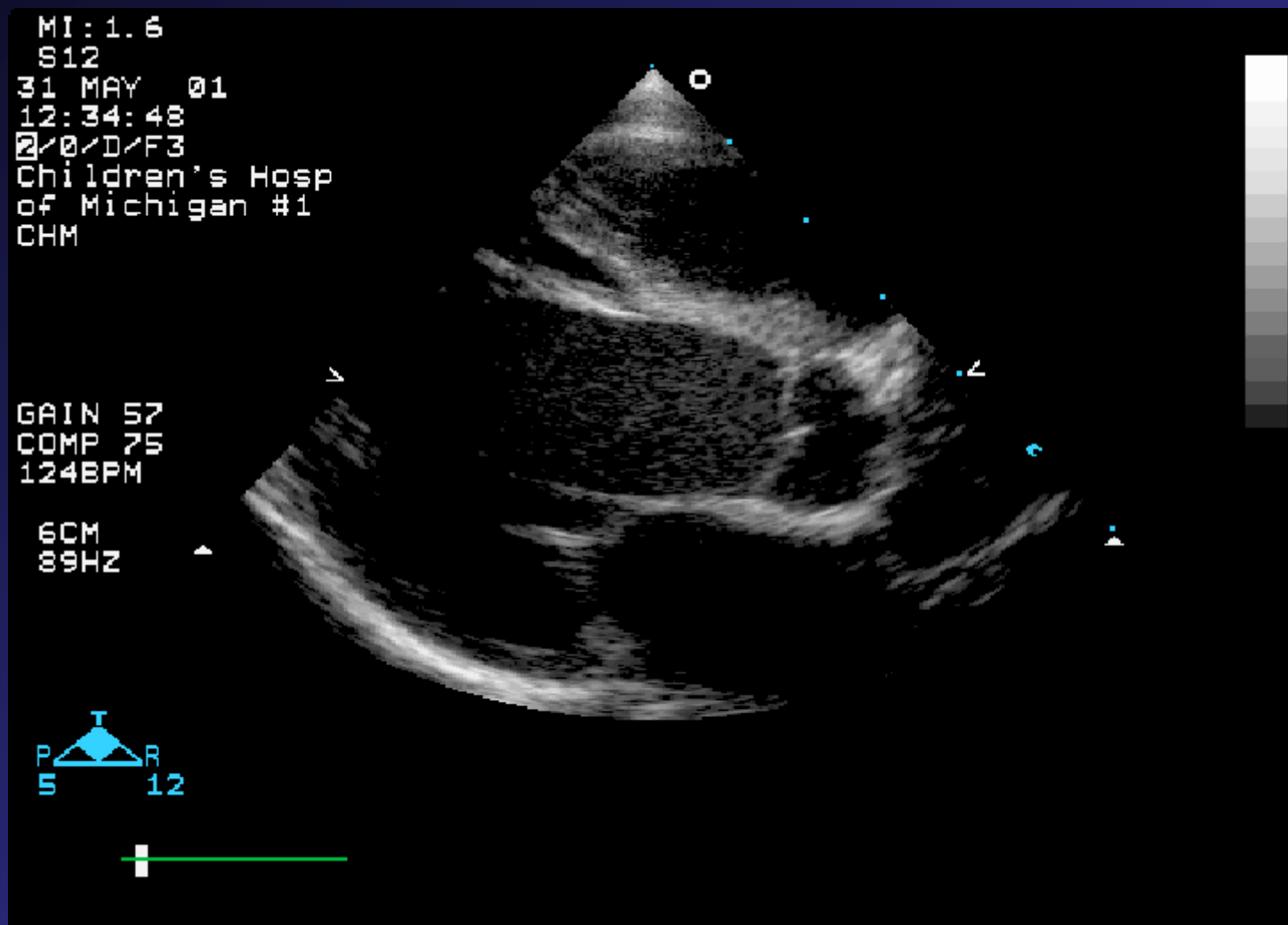
Common Syndromes/Chromosomal Anomalies

<u>Anomaly</u>	<u>Associations</u>
Trisomy 21	VSD, AV Canal
Trisomy 18	VSD, PDA
Trisomy 13	VSD, PDA, Dextrocardia
Turner	Coarctation, AS
Noonan	PS, HCM
Williams	Supravalvar AS, Peripheral PS
Holt-Oram	ASD
Marfan	Aortic root dilation, MVP
DiGeorge	VSD, arch anomalies, TOF

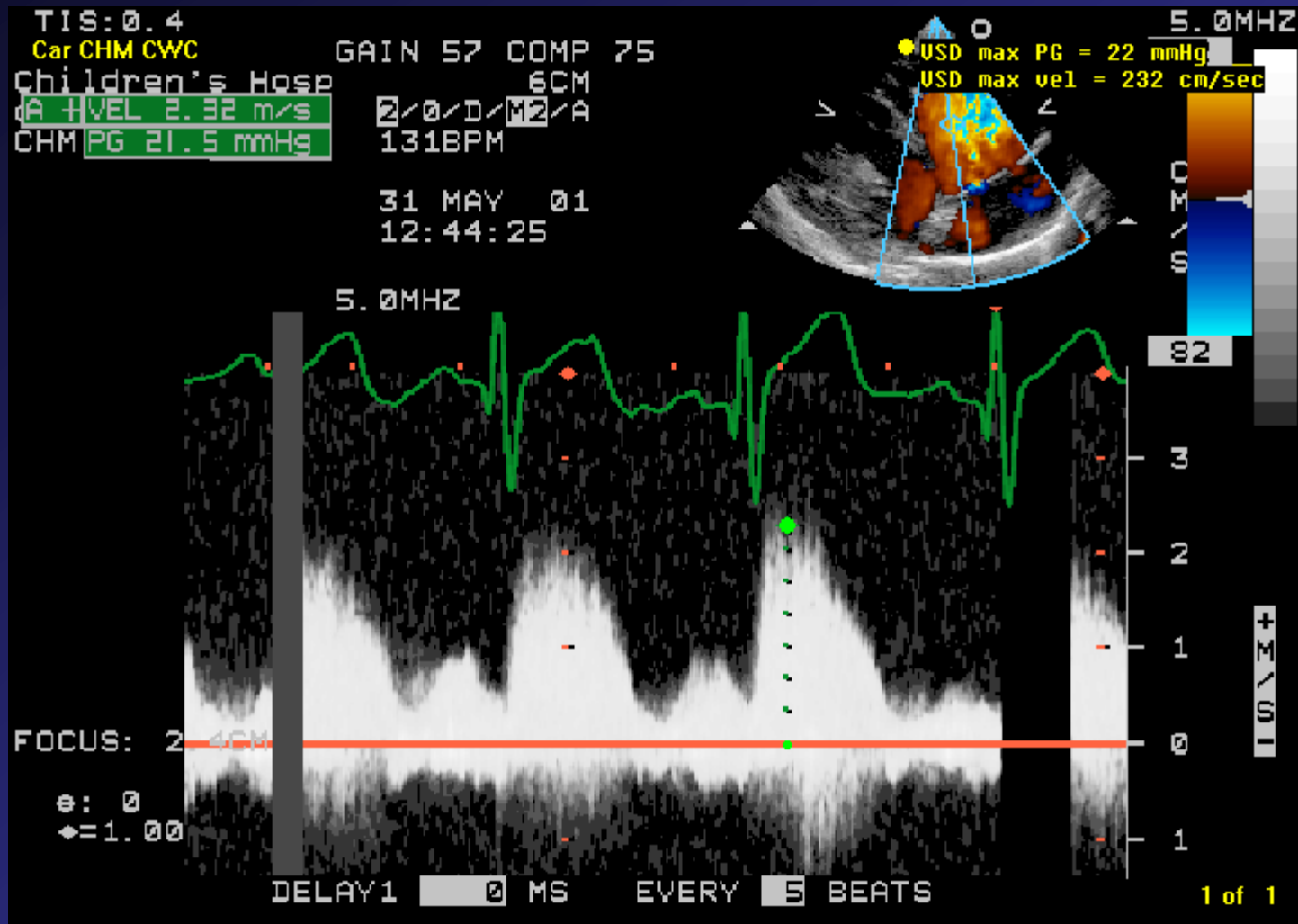
Congenital Heart Disease

- Currently over 1 million patients over 18 with CHD are alive in this country
- This increases at 4-5% per year
- 20,000 cardiac surgical procedures for CHD/year
- >90% of children with CHD survive to adulthood
- The majority of adult CHD patients will be post-op
 - The ASCeXAM does not cover much post-op disease, but has asked questions about operations

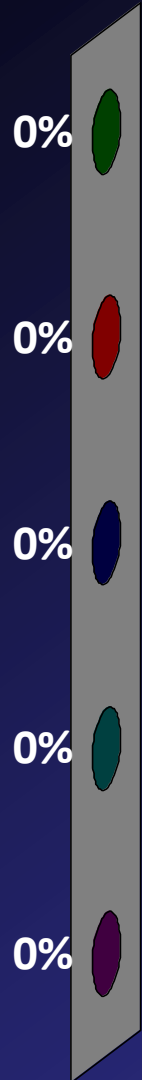
Case 1 - 3 month old with a murmur, tachypnea and failure to thrive



Case 1 - 3 month old with a murmur, tachypnea and failure to thrive



The defect shown in this example is:



1. Secundum VSD

2. Sinus Venosus VSD

★ 3. Perimembranous VSD

4. Inlet VSD

5. Supracristal VSD

An isolated VSD will generally produce enlargement of which chamber(s):

- 0% 1. Left atrium, Left ventricle
- 0% 2. Right ventricle
- 0% 3. Right ventricle, pulmonary artery
- 0% 4. Aorta
- 0% 5. Right ventricle, right atrium

What is the right ventricular pressure?

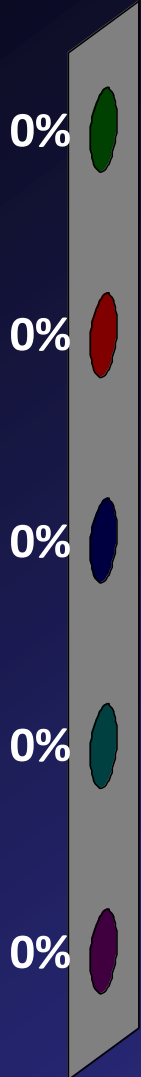
1. Normal

2. Supra systemic

→ 3. Systemic

4. Can't tell from information given

5. Want to go home now



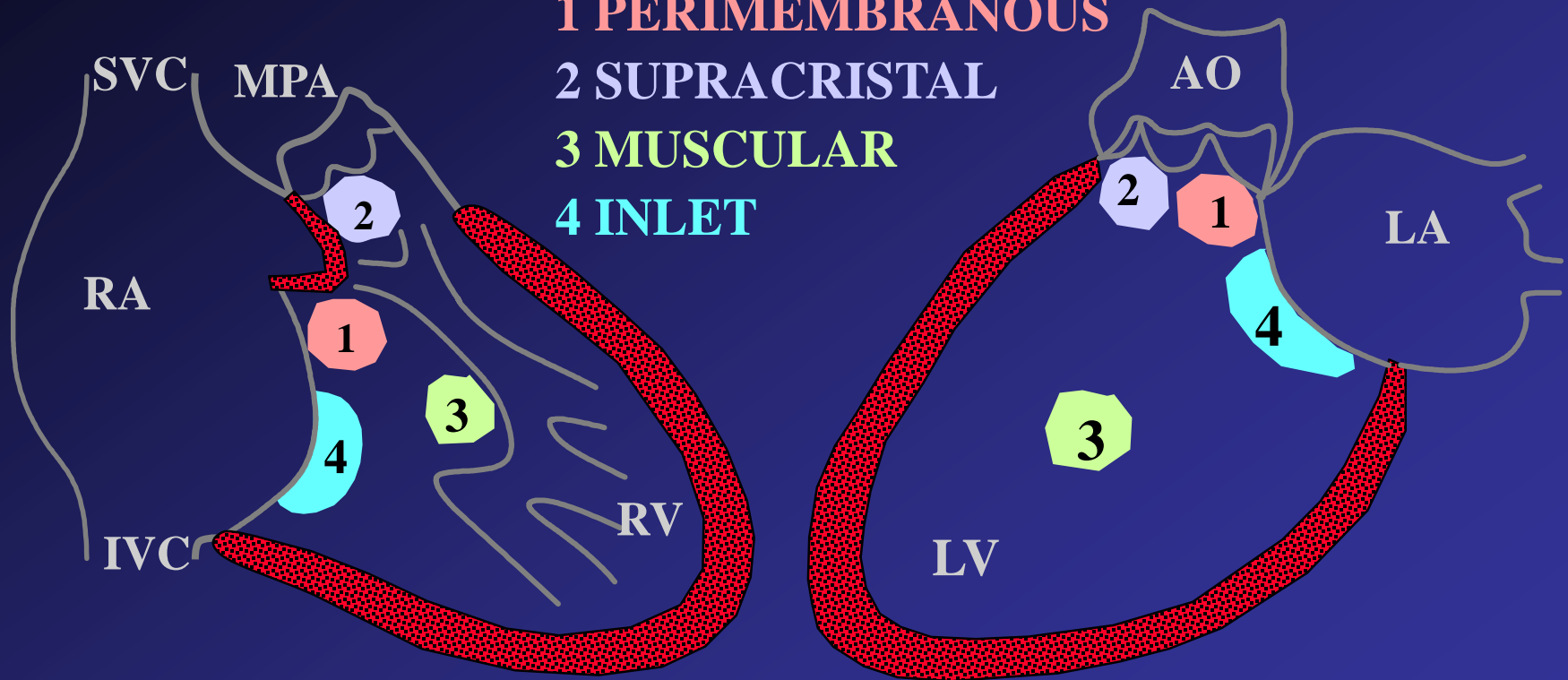
Ventricular Septal Defect

Clinical

- Most common defect, 25% of CHD
- Shunt flow should be left to right
- Symptoms depend on the size of the hole
 - Large - $>50\%$ of aortic annulus size
 - Medium - 25-50% of annulus size
 - Small - $<25\%$ of annulus size
- Large VSDs result in pulmonary edema
 - tachypnea, poor feeding, failure to thrive in infants
- In un-operated patients with large defects pulmonary vascular disease develops → shunt reversal and cyanosis (Eisenmenger's complex)

Ventricular Septal Defect Anatomy

- 1 PERIMEMBRANOUS
- 2 SUPRACRISTAL
- 3 MUSCULAR
- 4 INLET



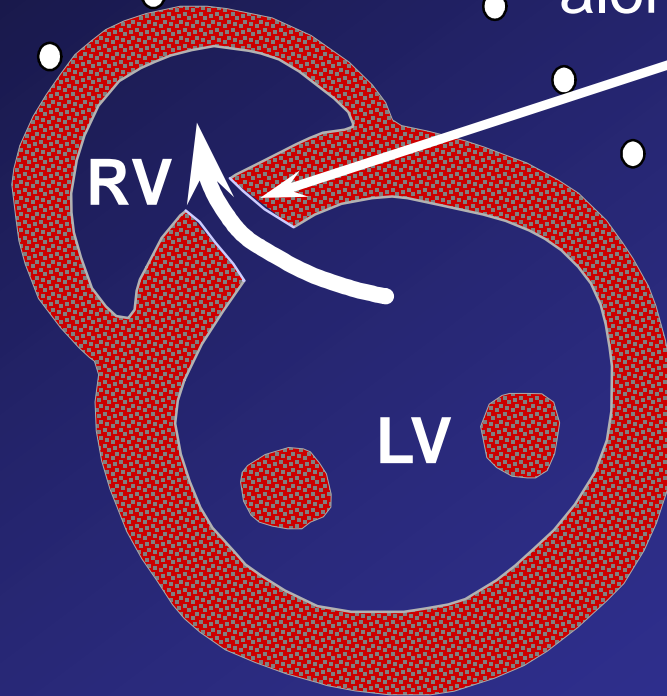
RIGHT VENTRICULAR VIEW

LEFT VENTRICULAR VIEW

Ventricular Septal Defect

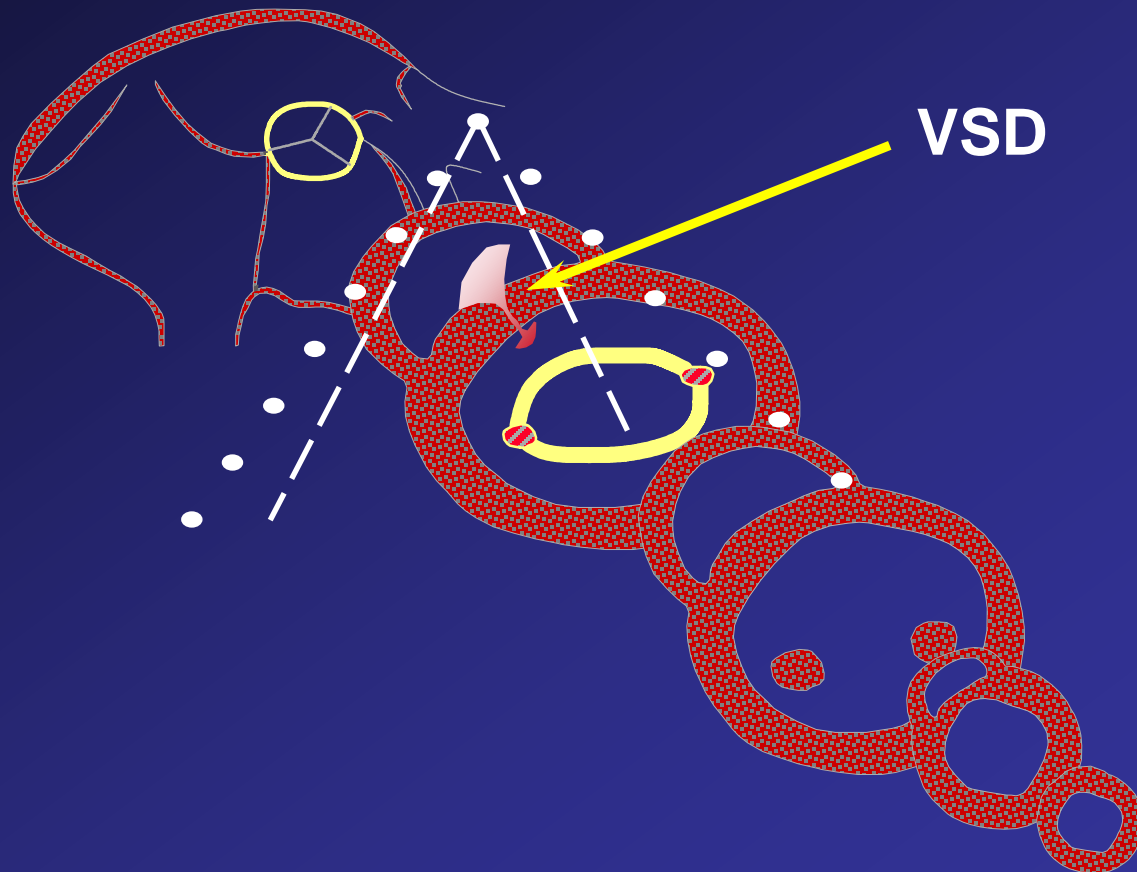
Parasternal Short-Axis - Mid-Ventricle

Muscular (trabecular)
VSD's can be seen anywhere
along the septum from this view



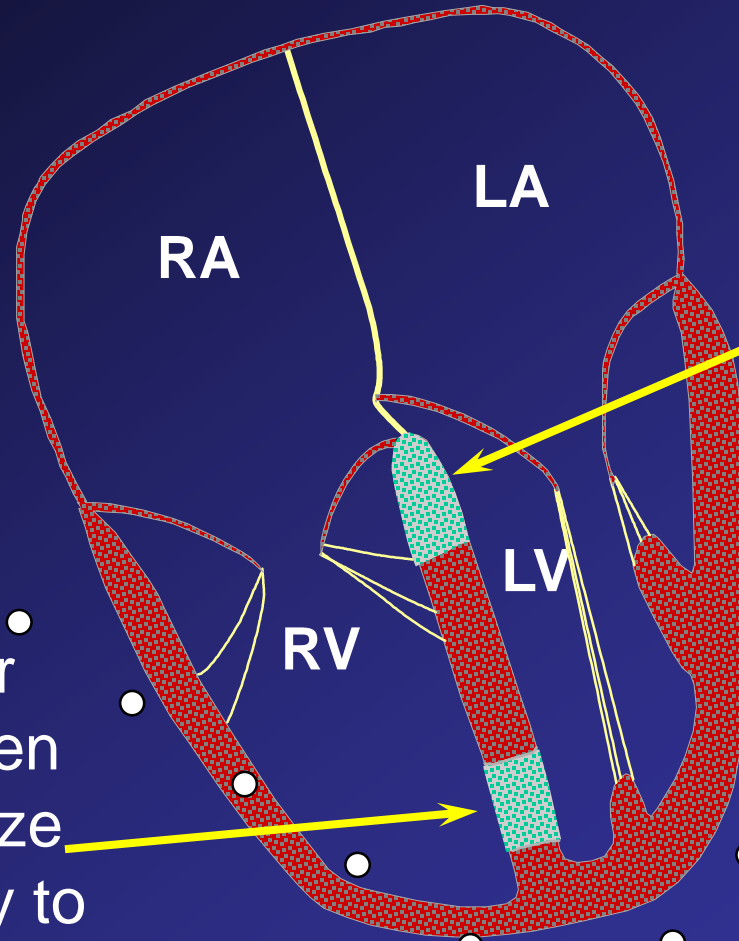
Ventricular Septal Defect

Parasternal Short-Axis "Sweep"



Ventricular Septal Defect

Apical Four-Chamber View

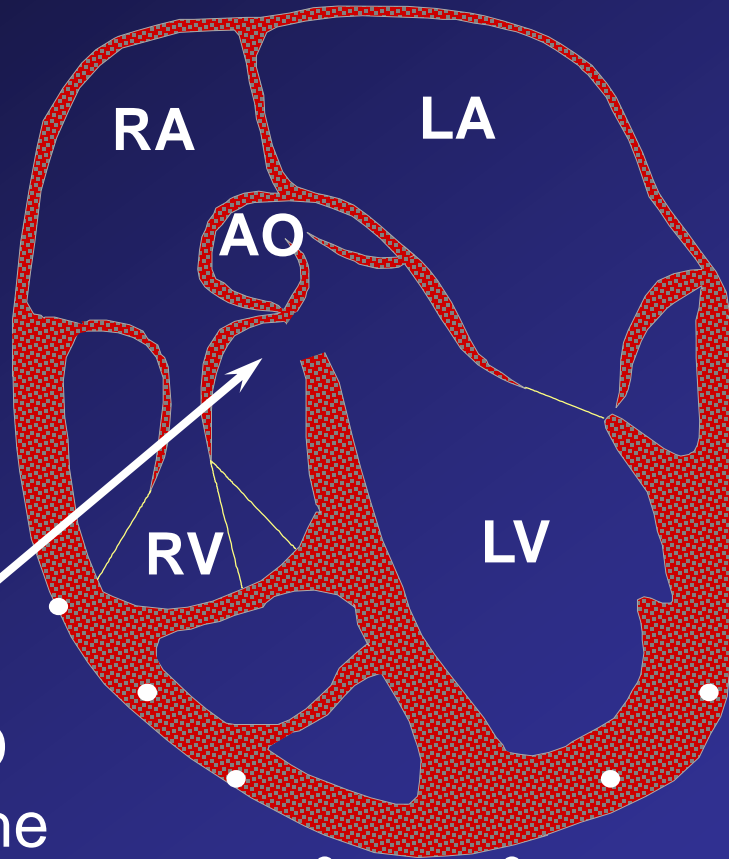


Inlet VSD's are best seen from this view.

Apical muscular VSD's can be seen from this view. Size may vary from tiny to large.

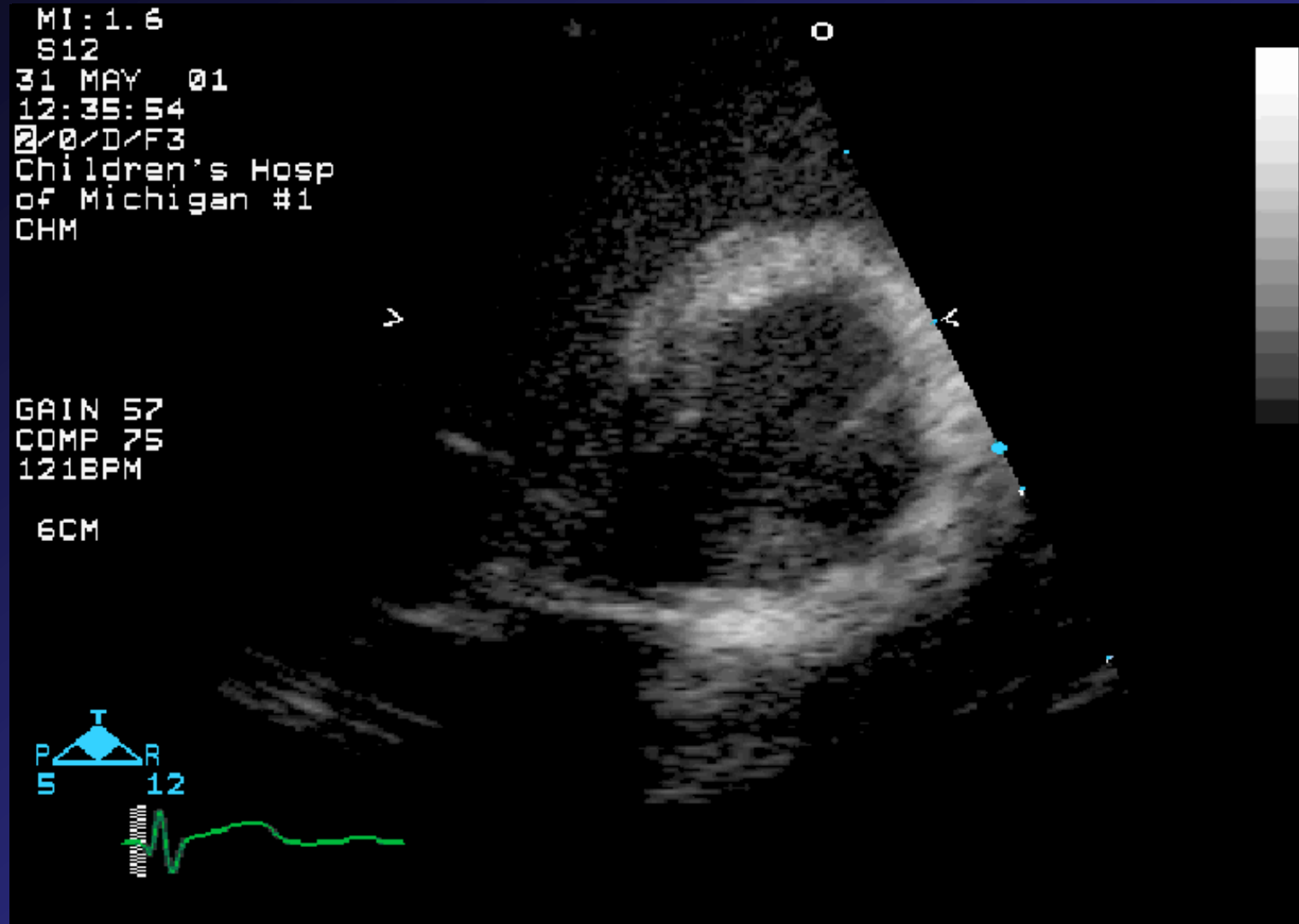
Ventricular Septal Defect

Apical Five-Chamber View



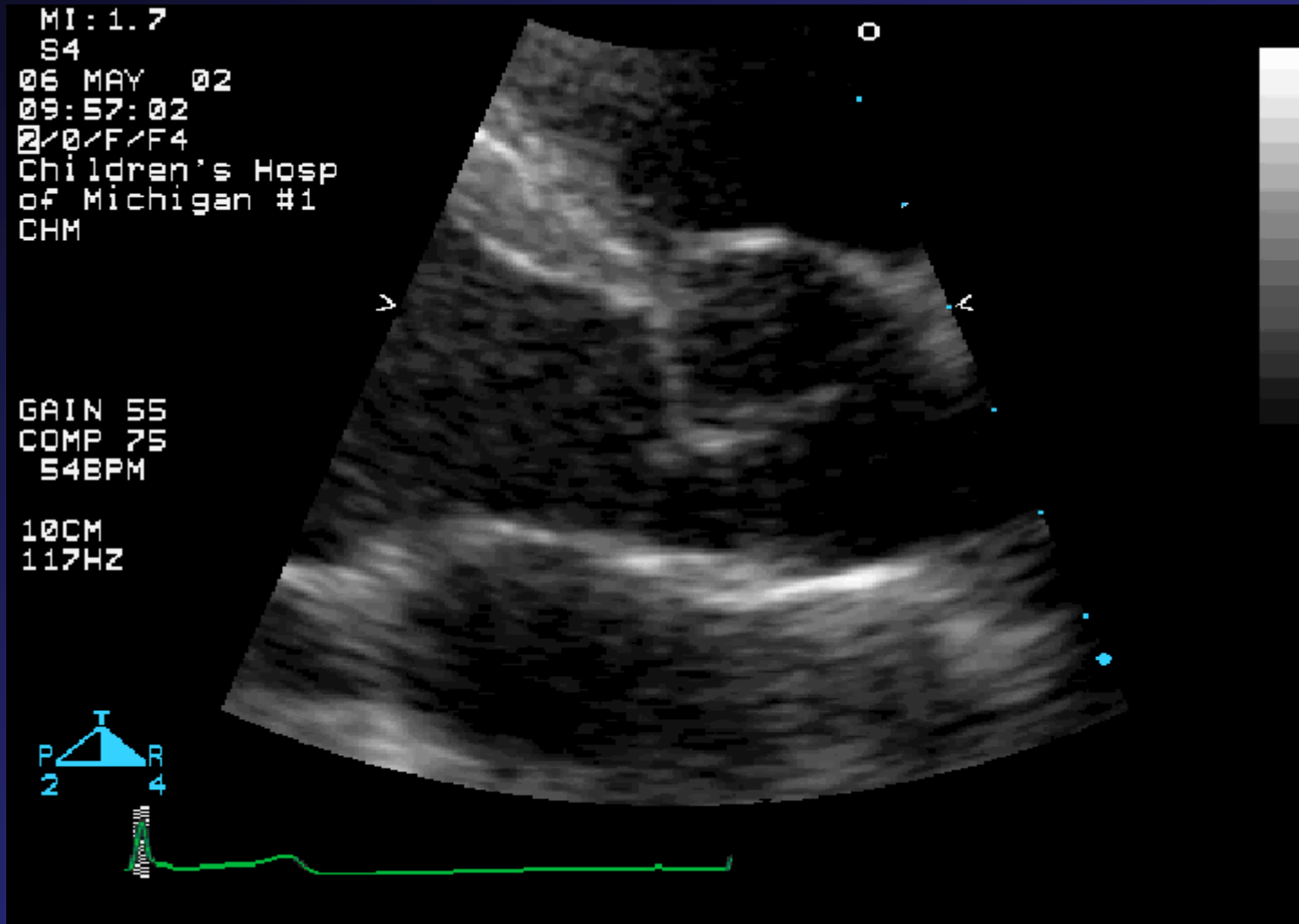
This view nicely demonstrates a perimembranous VSD and its relationship to the aortic and tricuspid valves

Case 1- Review



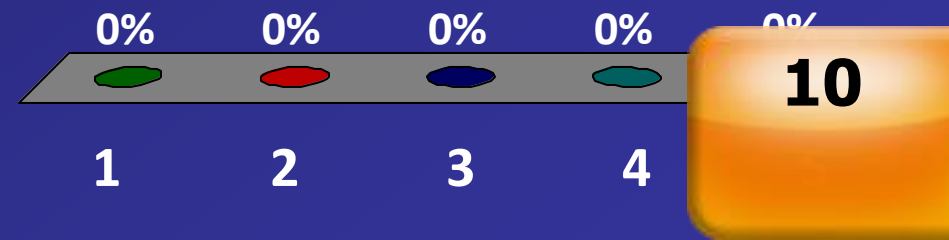
Ventricular Septal Defect

Case 2 – 8 y.o. with asymptomatic murmur



The defect shown in this example is:

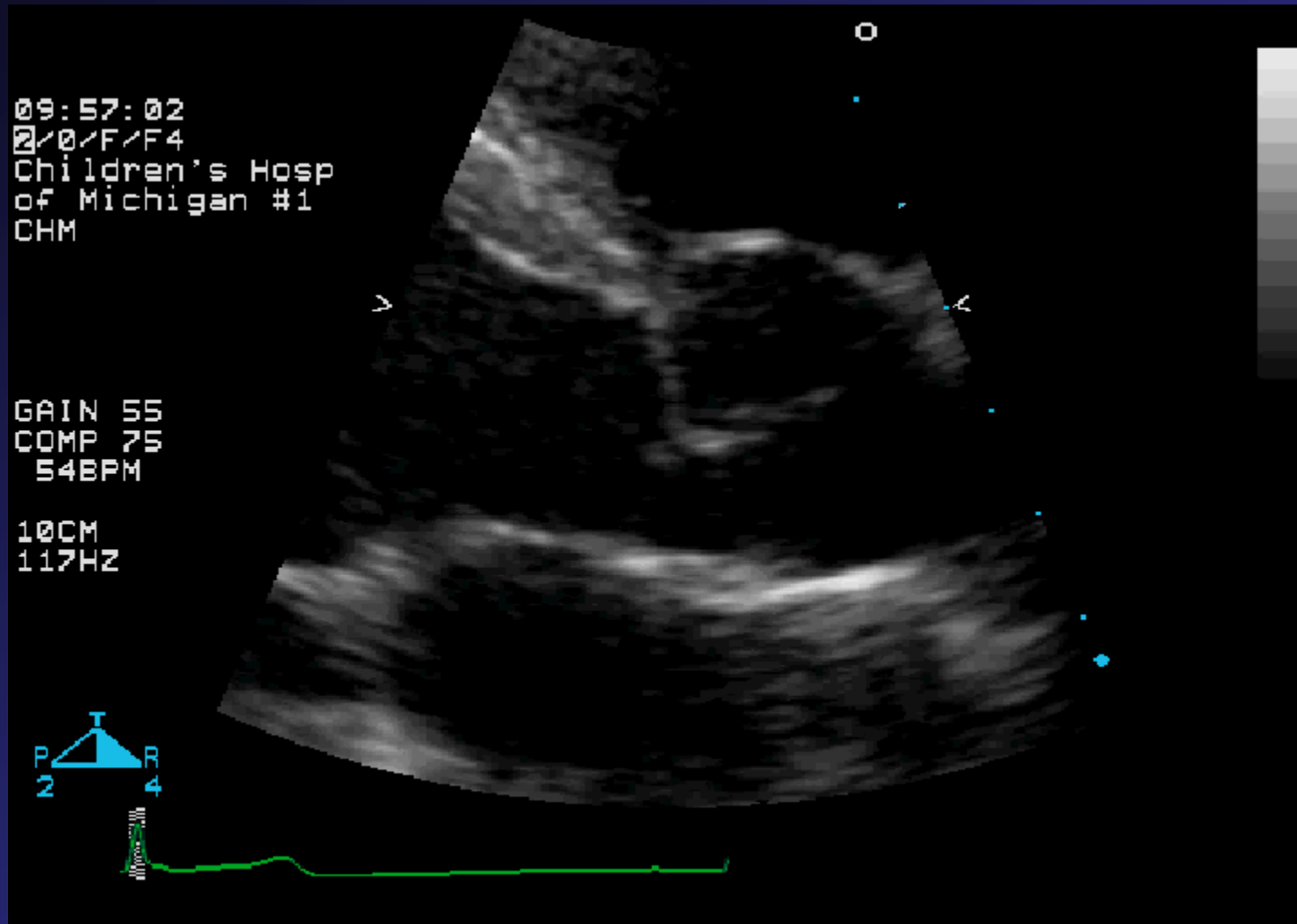
1. Secundum VSD
2. Sinus Venosus VSD
3. Perimembranous VSD
4. Inlet VSD
- 😊 5. Supracristal VSD



Question 10 - A common complication of this defect is:

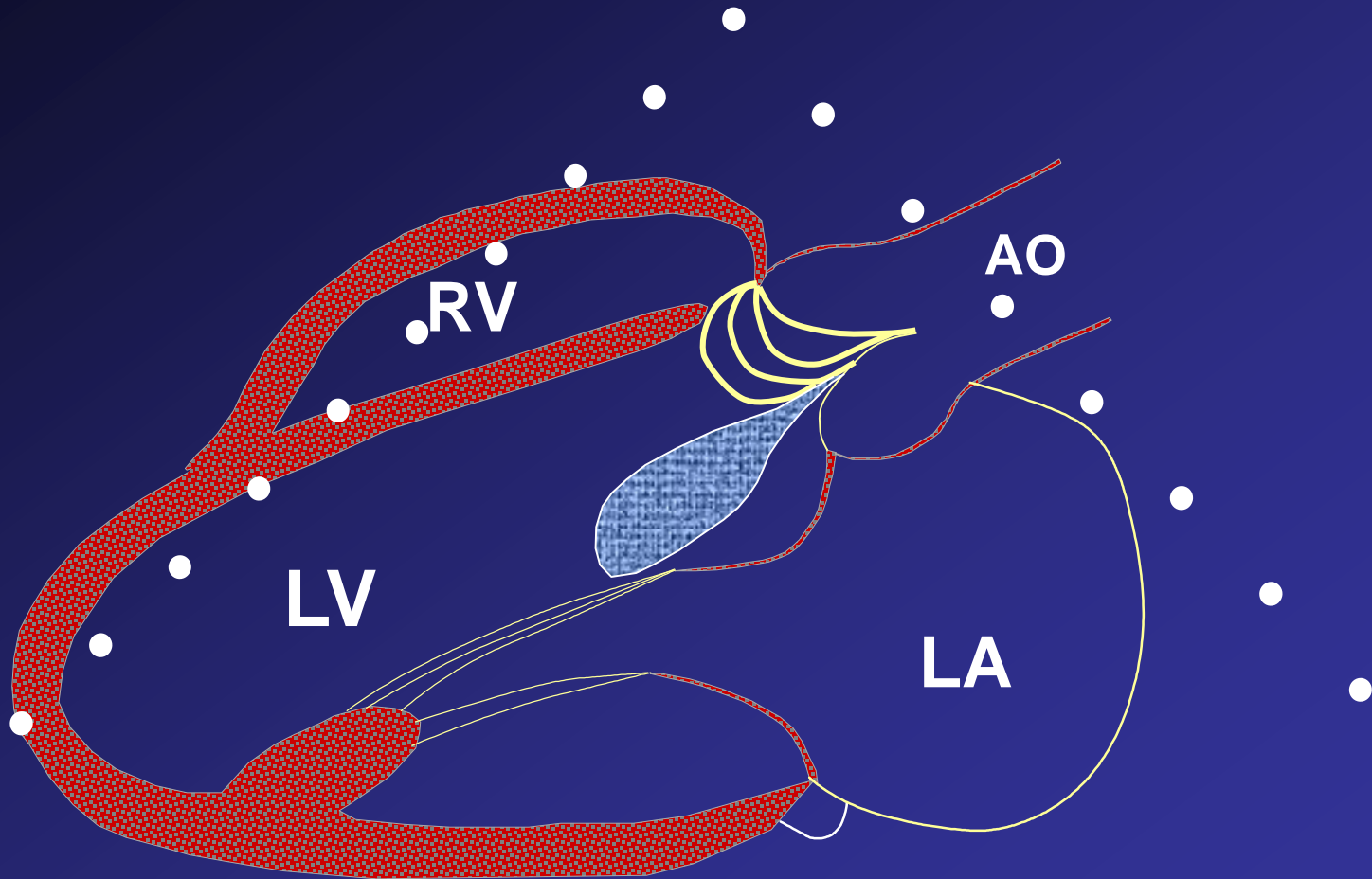
- 0% 1. Pulmonary valve endocarditis
- 0% 2. Aortic regurgitation
- 0% 3. Aortic dissection
- 0% 4. Tricuspid regurgitation
- 0% 5. Right ventricular enlargement

Case 2- Review



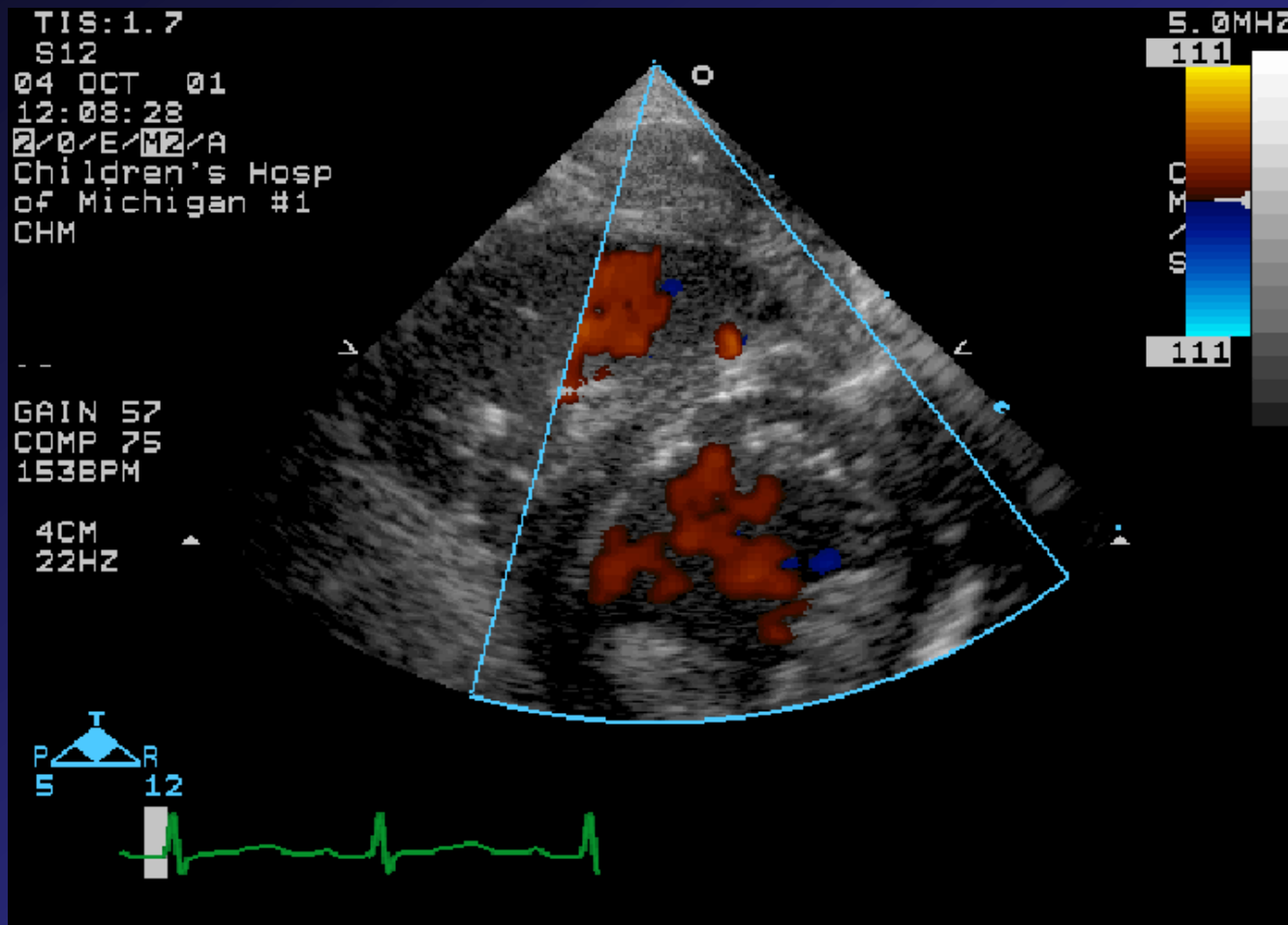
Supracristal VSD

Aortic Cusp Prolapse



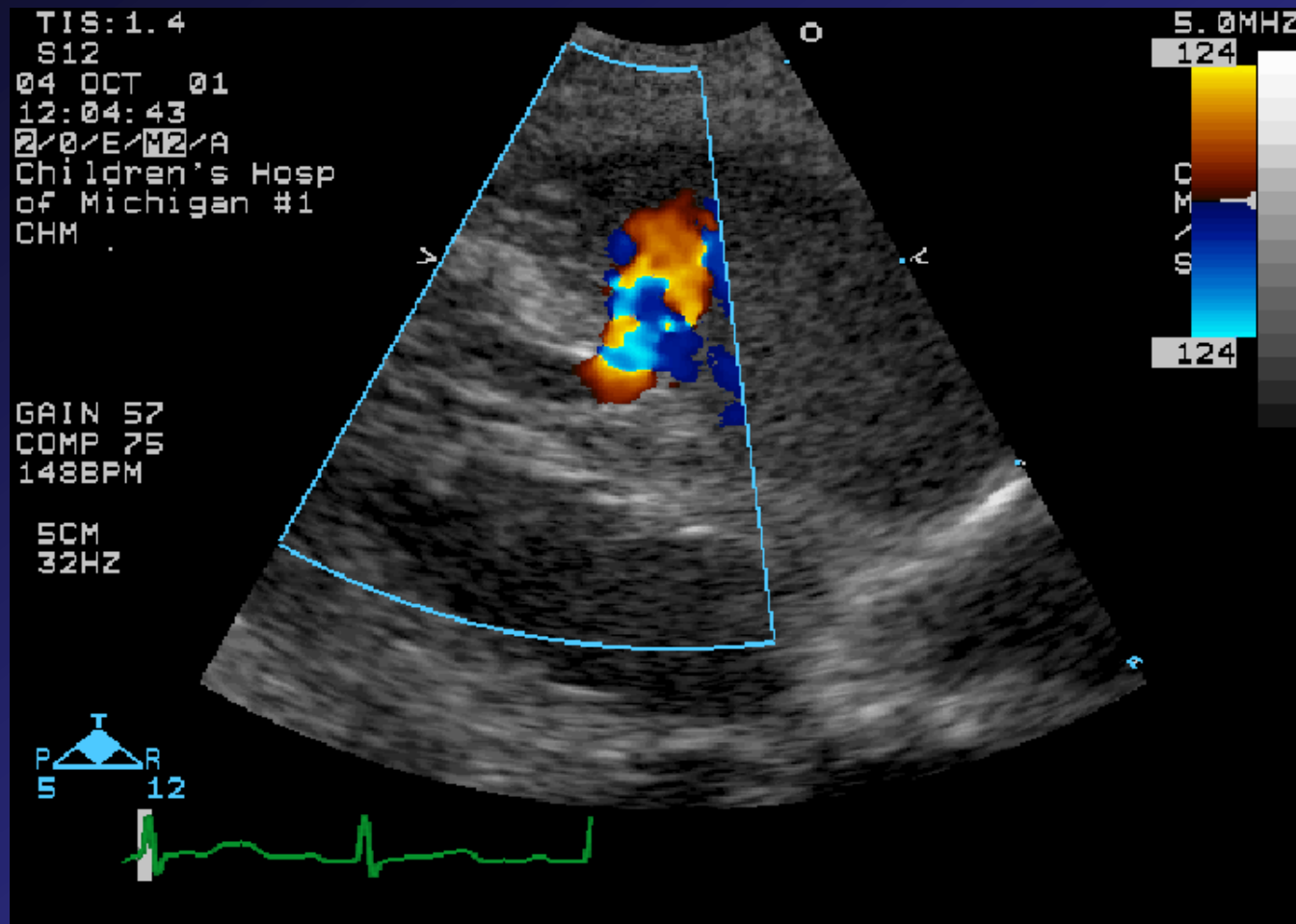
Ventricular Septal Defect

Case 3 – No questions



Ventricular Septal Defect

Case 3 – No questions



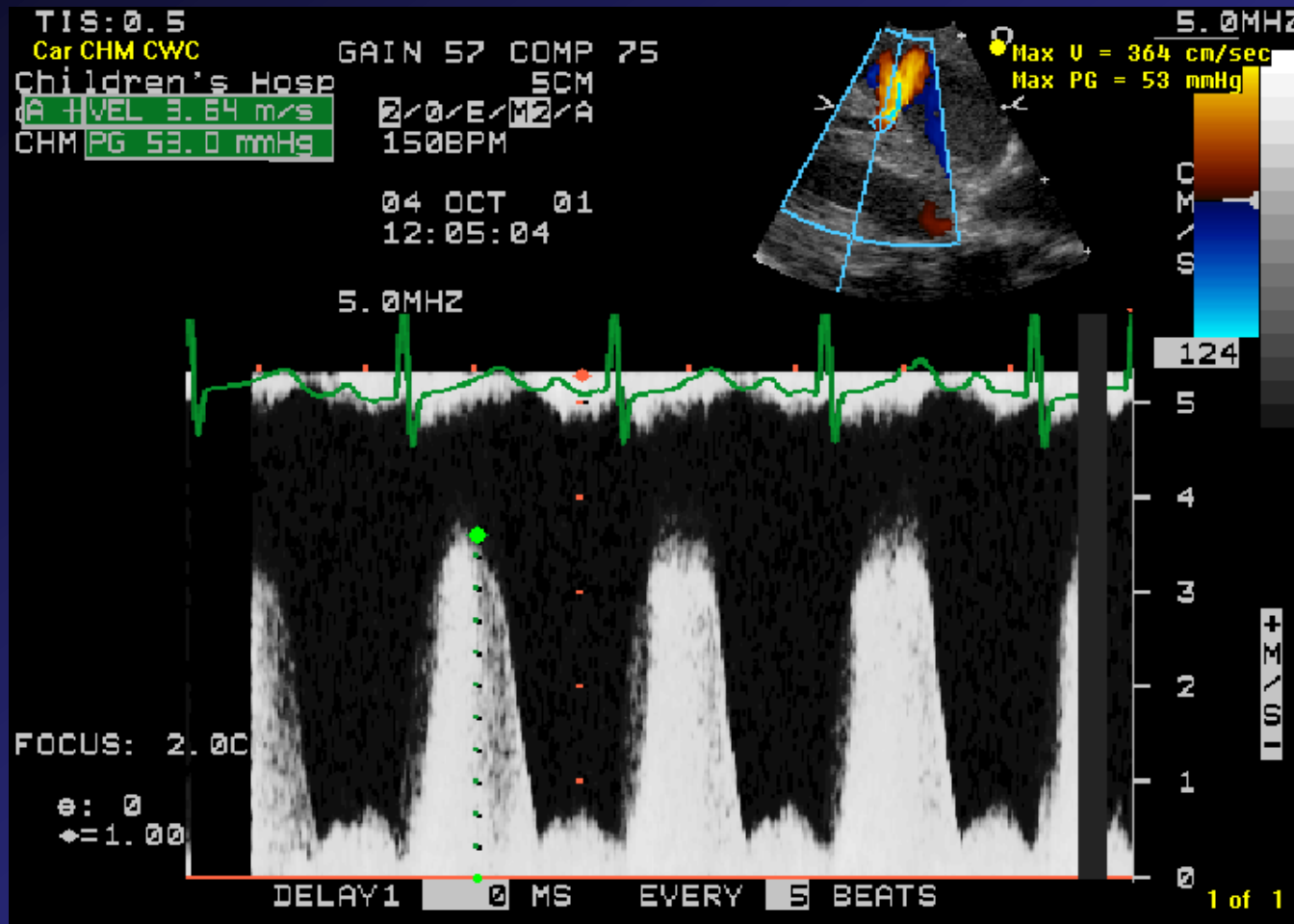
Ventricular Septal Defect

Case 3 – No questions

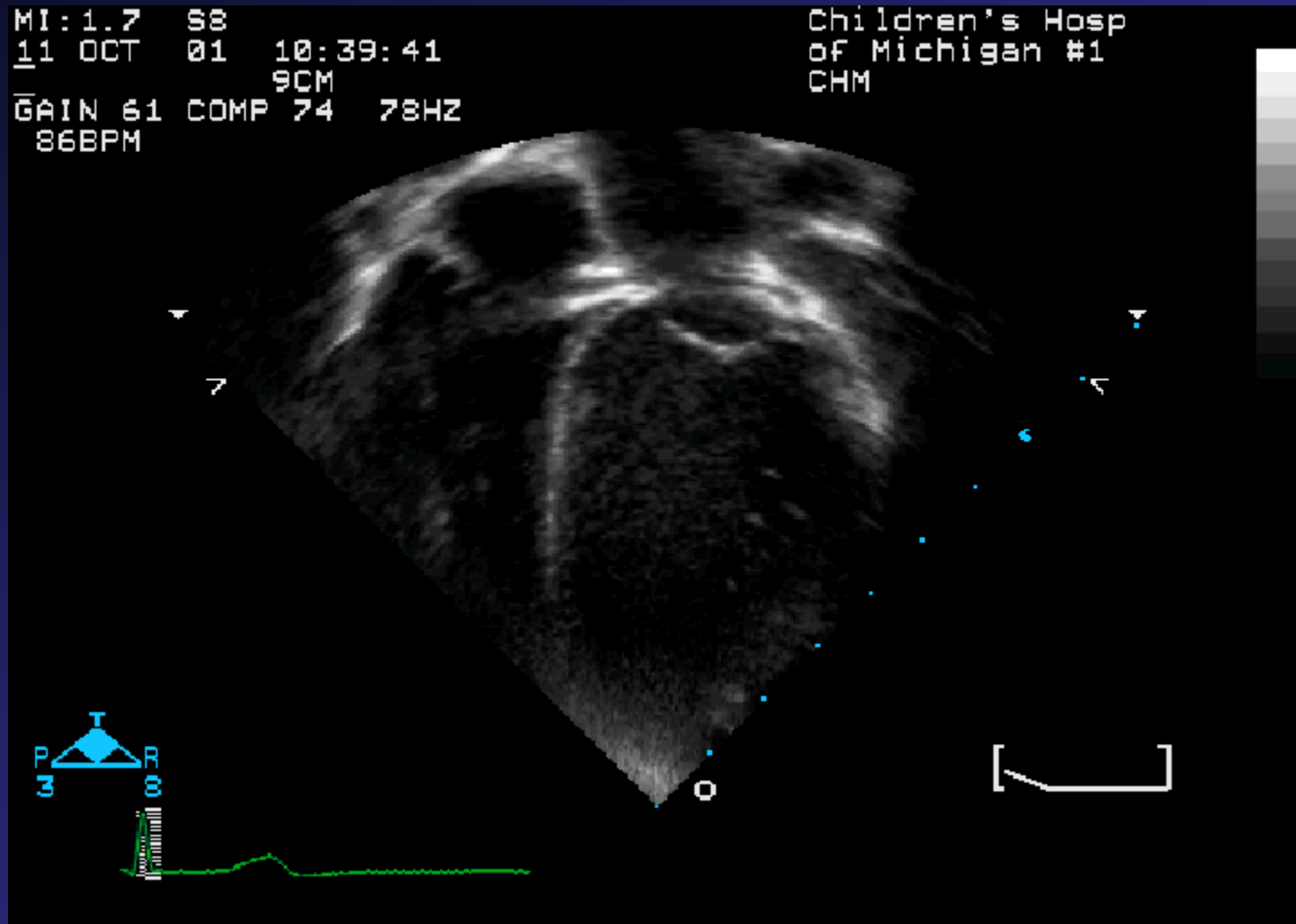


Ventricular Septal Defect

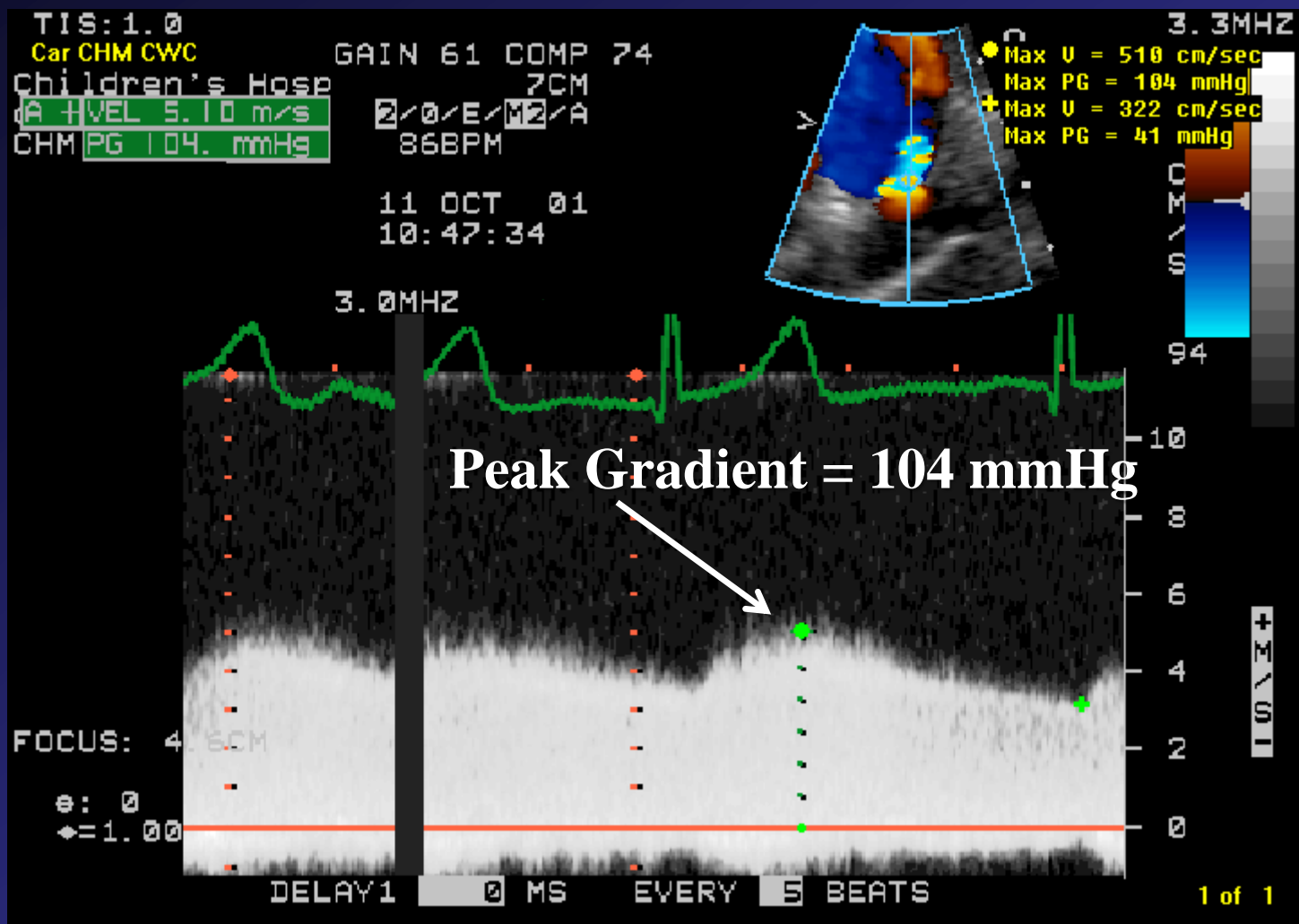
Case 3 – No questions



Case 4 – 6 y.o. with continuous murmur

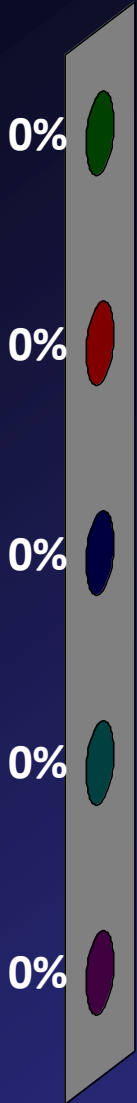


Case 4 – 6 y.o. with continuous murmur



The Doppler tracing in this case implies:

1. Severe pulmonary hypertension
2. Severe systemic hypertension
3. Severe coarctation of the aorta
- ✓ 4. Normal pulmonary artery pressure
5. Severe pulmonary artery stenosis



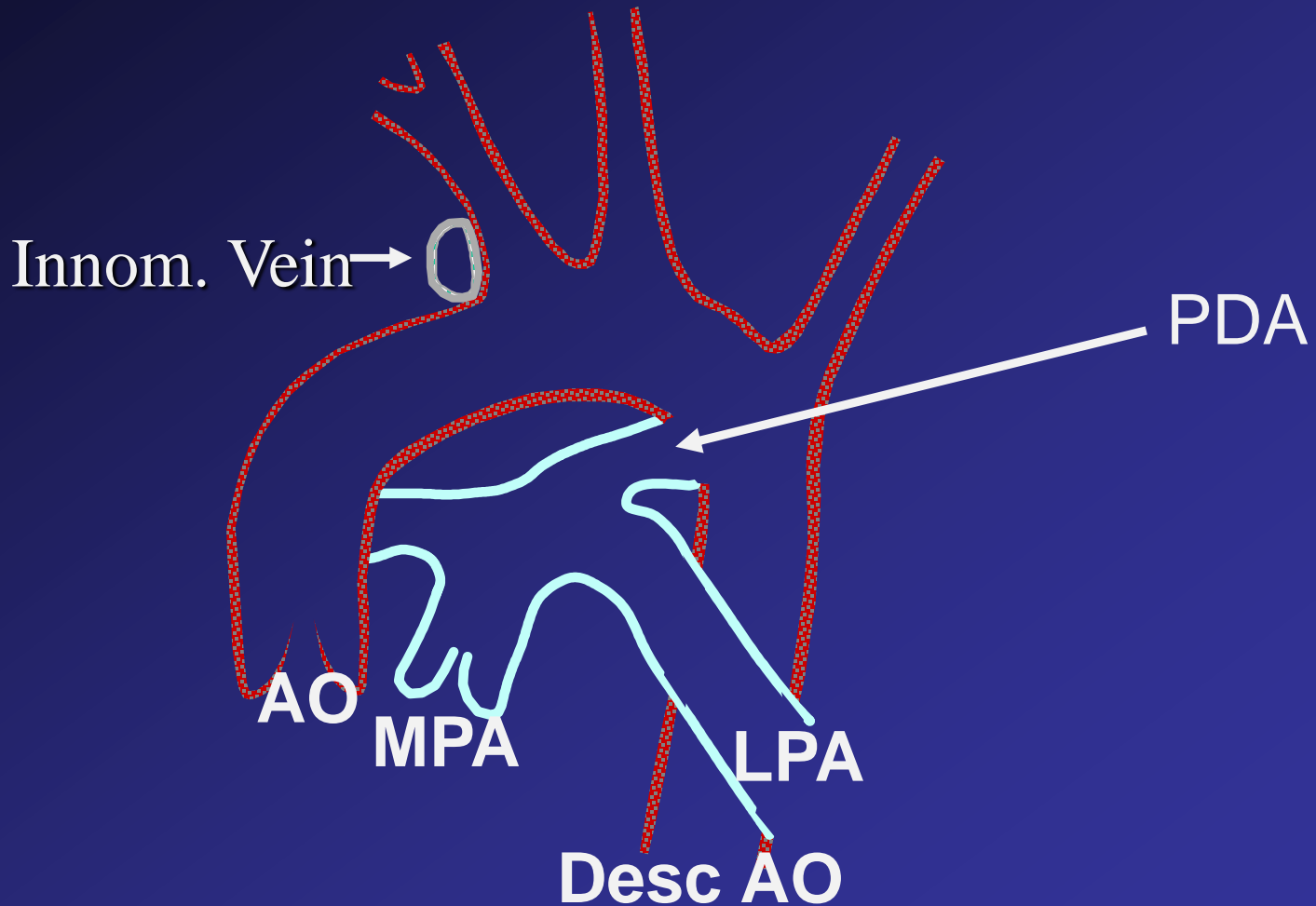
Patent Ductus Arteriosus

Clinical

- Continuous murmur in older patients
- Bounding pulses, wide pulse pressure, respiratory symptoms in neonates with a large PDA
- Large PDA will act much like a large VSD, producing pulmonary over-circulation and signs/symptoms of congestive heart failure
- A small PDA is generally hemodynamically insignificant but is at risk for endarteritis

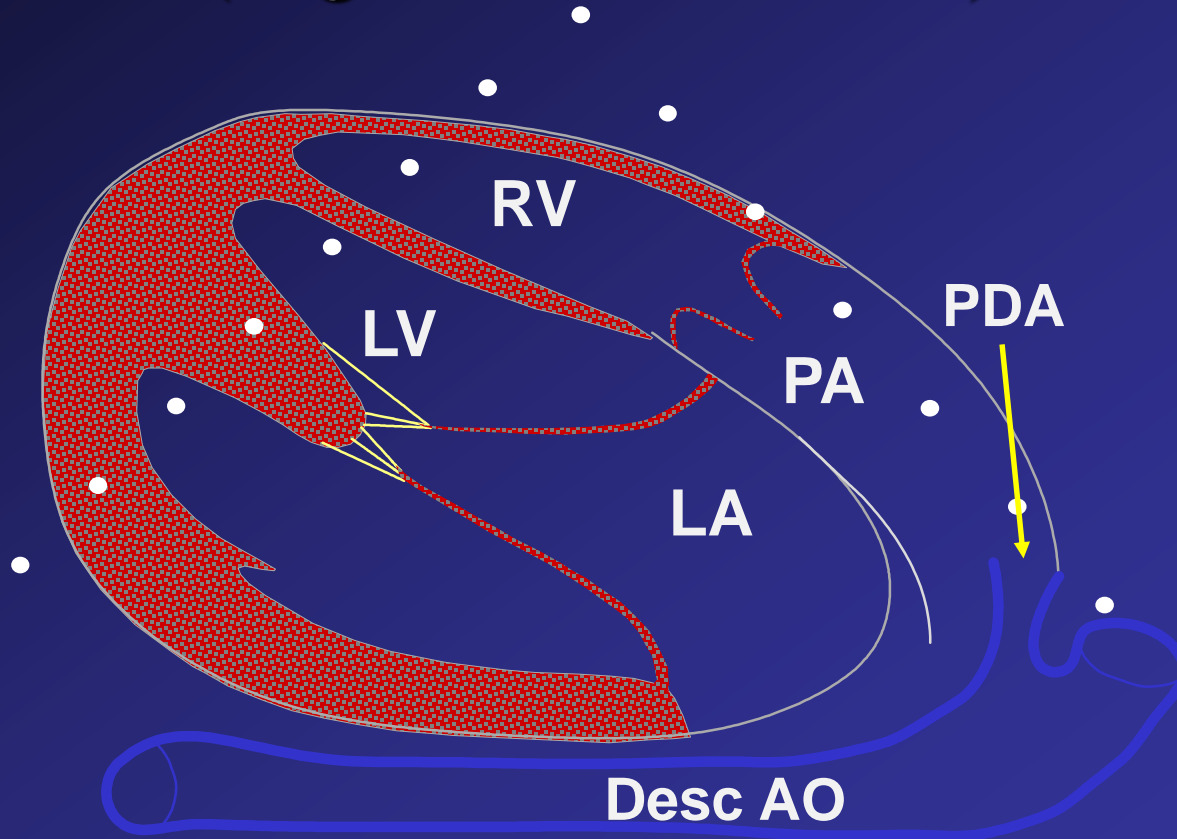
Patent Ductus Arteriosus

Anatomy



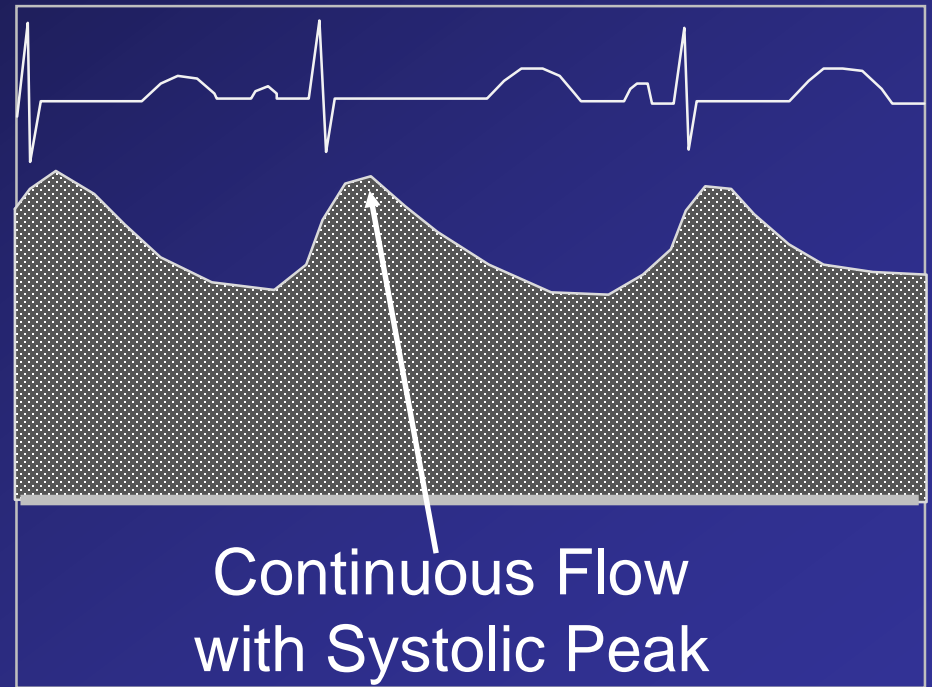
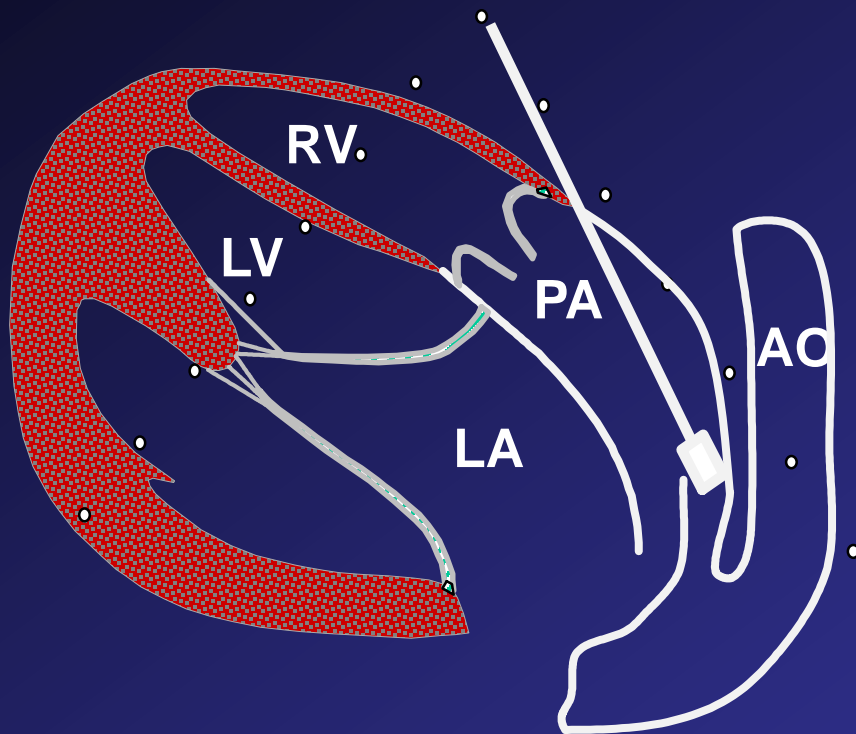
Patent Ductus Arteriosus

Ductal View
Parasternal Ductal View
(High Left Parasternal)



Patent Ductus Arteriosus

Doppler Flow Pattern



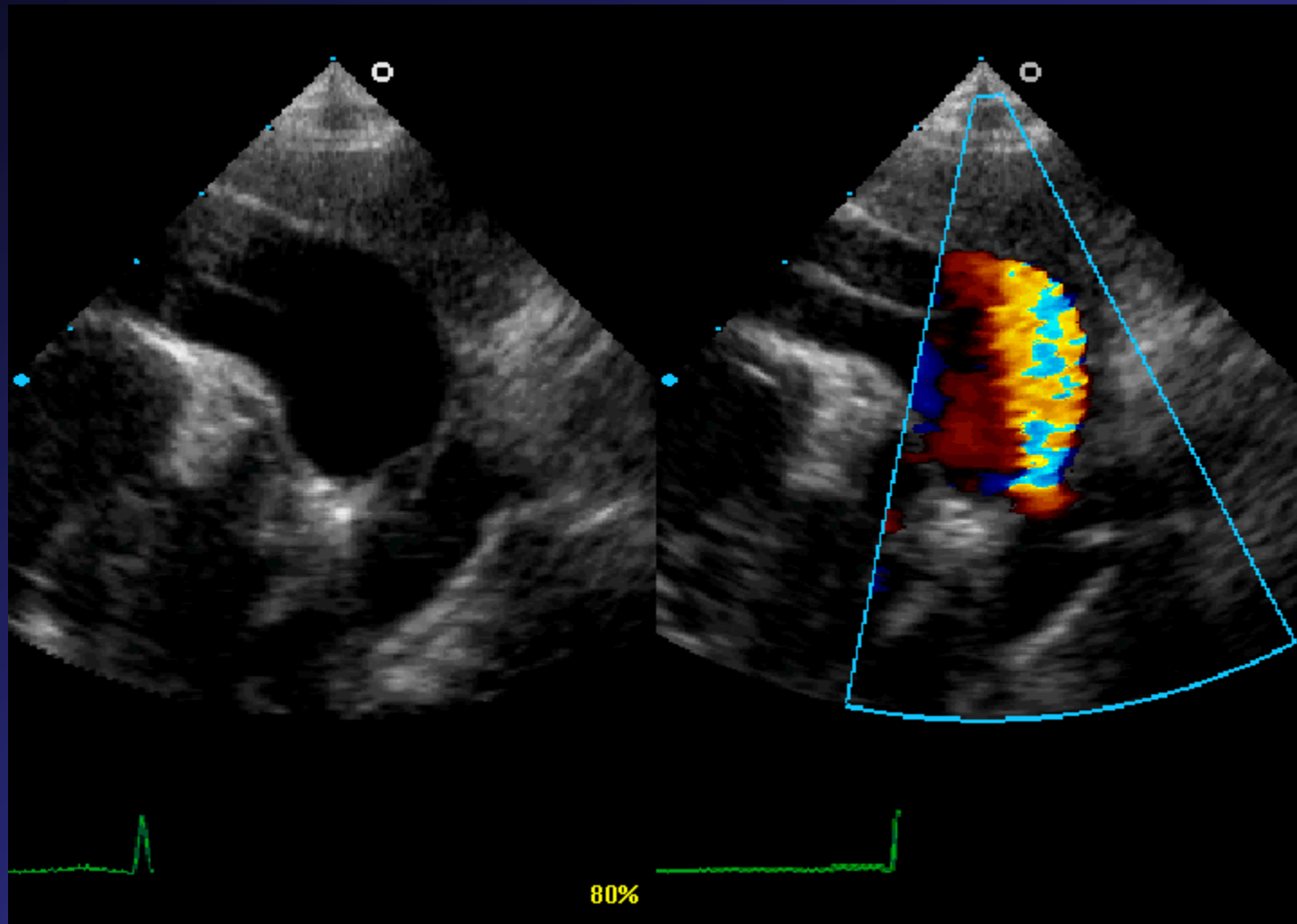
$$\text{Systolic PA pressure} = \text{SBP} - 4V_{\text{PDA}}^2$$

Patent Ductus Arteriosus

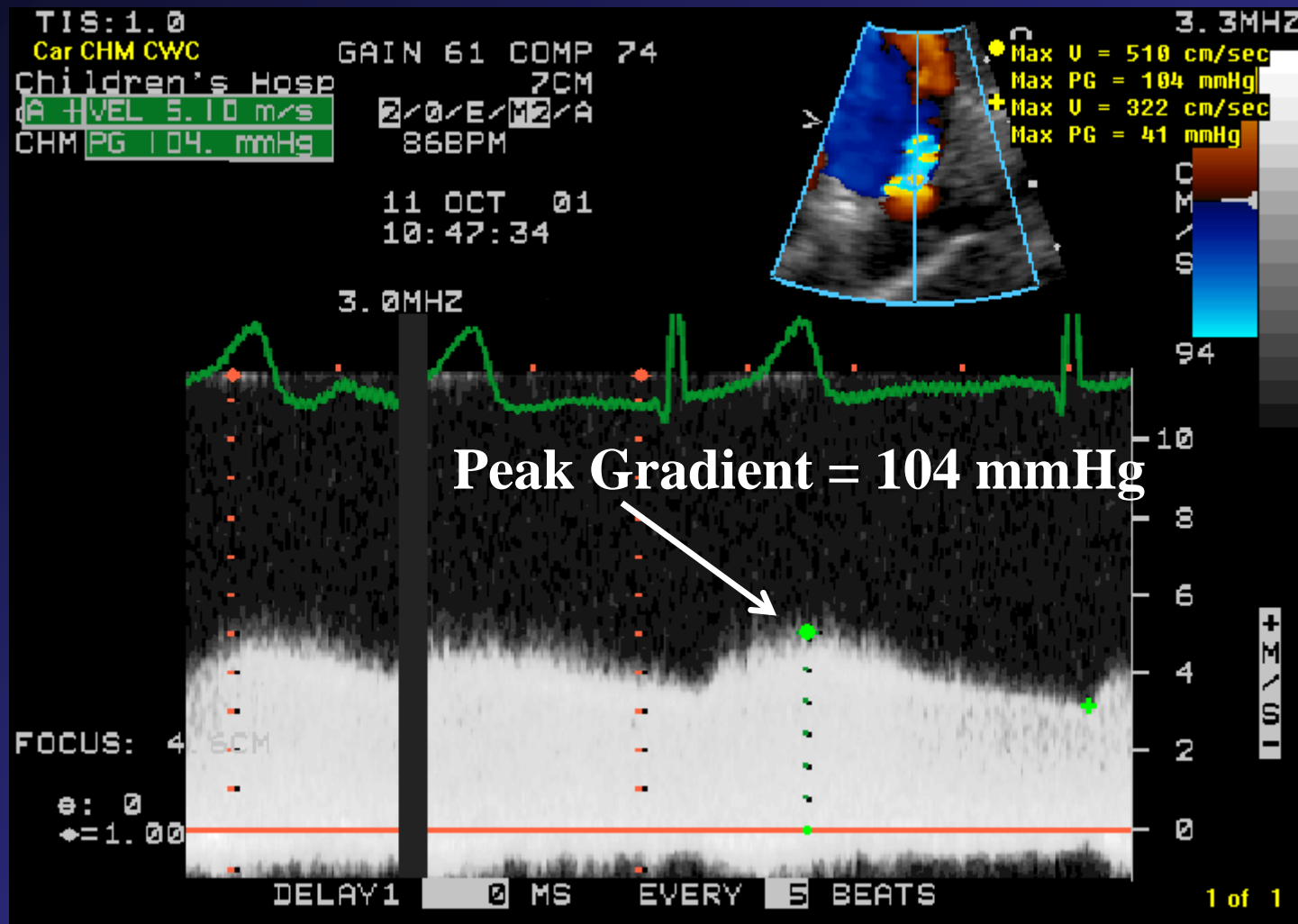
Clinical management

- Large PDAs in preterm infants
 - Pharmacologic closure – indomethacin
 - Surgical closure – left lateral thoracotomy
- Small PDA in older infants and children
 - Catheter closure – device or coil

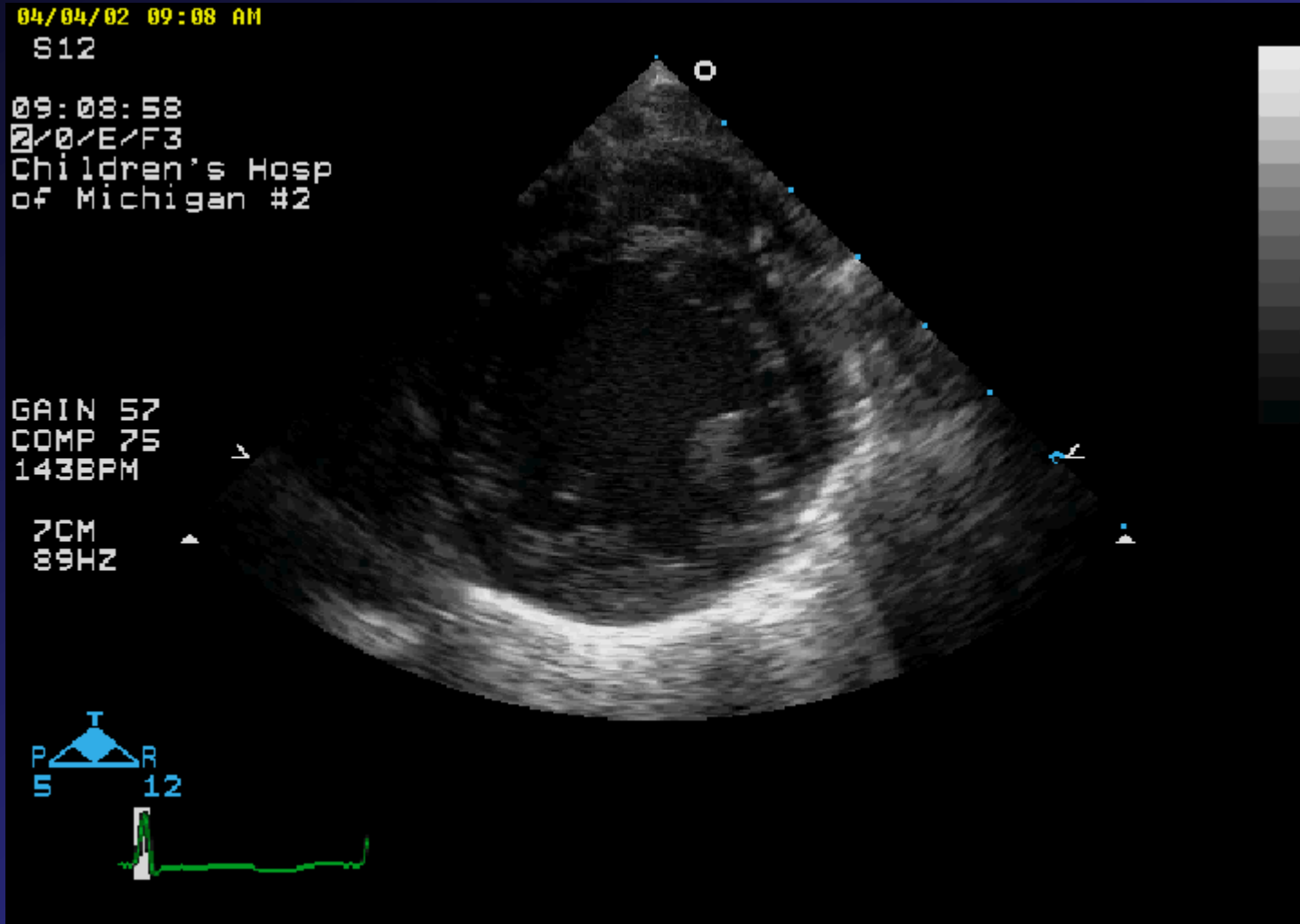
Case 4 - Review



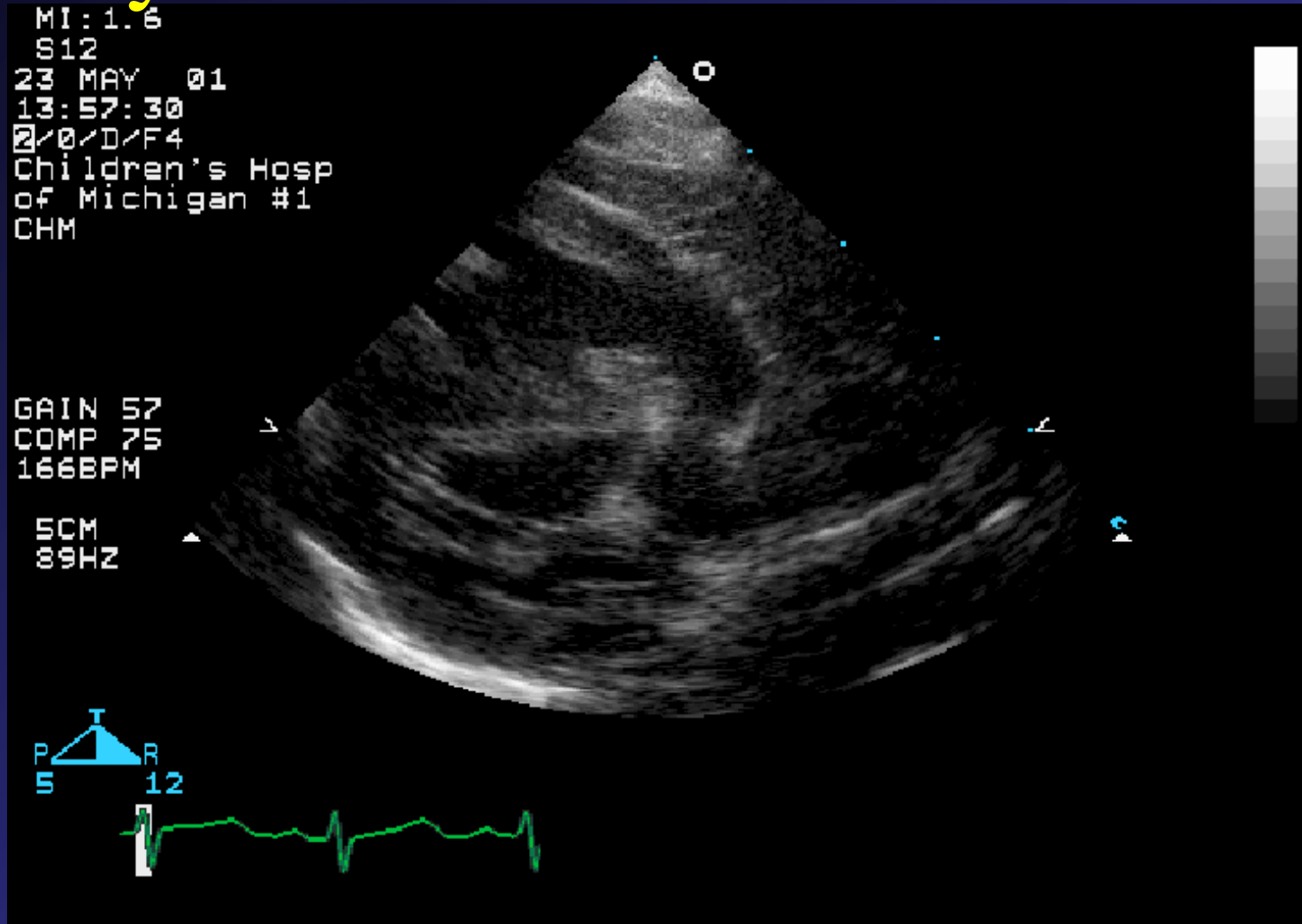
Case 4 - Review



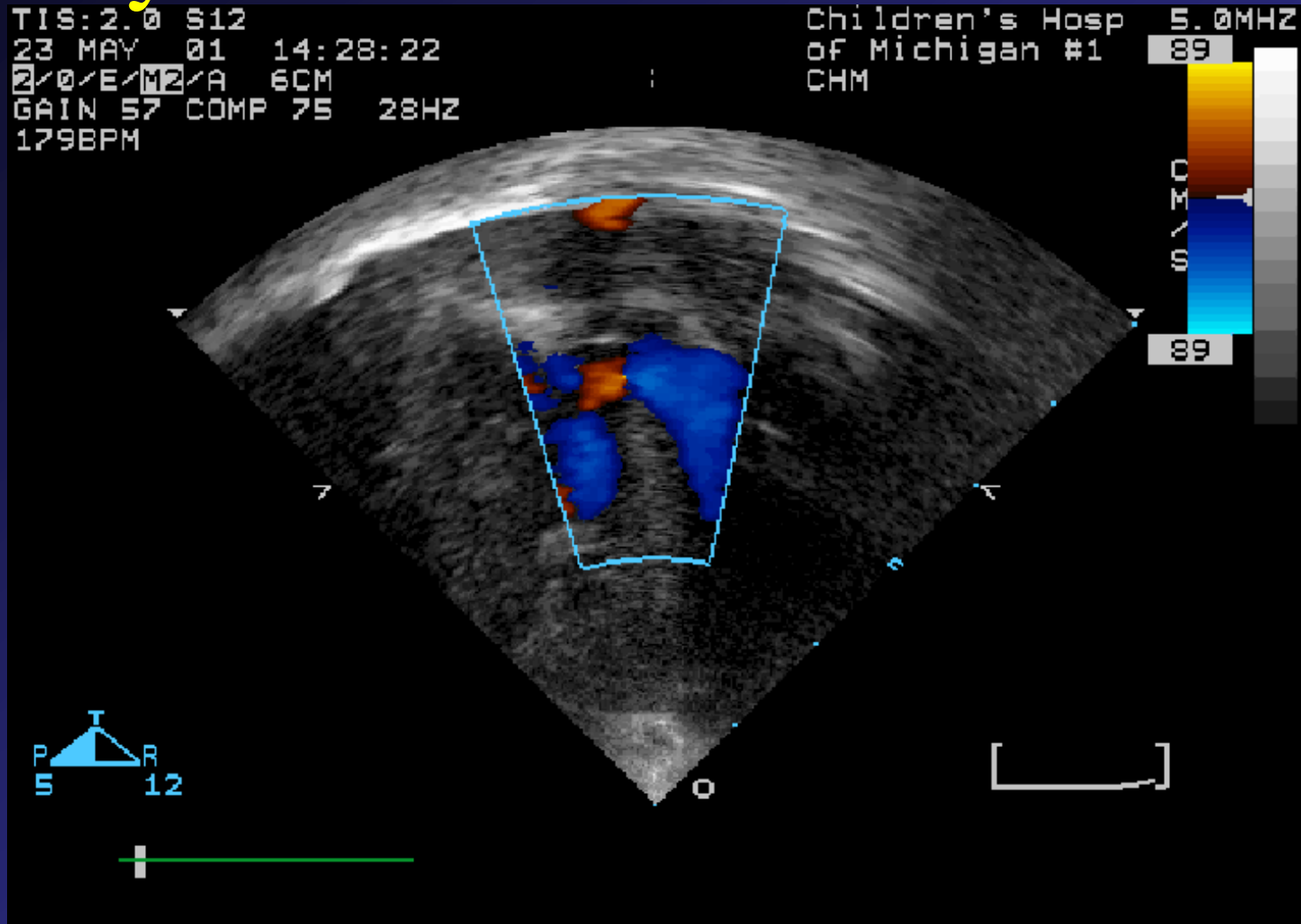
Case 5 – Large PDA



Case 6 – Asymptomatic 3 month old with cyanosis and a cardiac murmur

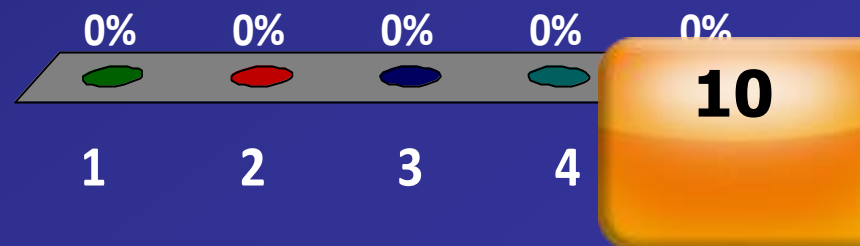


Case 6 – Asymptomatic 3 month old with cyanosis and a cardiac murmur



The defect shown in this example is:

1. Single ventricle
2. Transposition of the great arteries
3. Perimembranous VSD
- 😊 4. Tetralogy of Fallot
5. Complete atrioventricular canal



Tetralogy of Fallot

Background

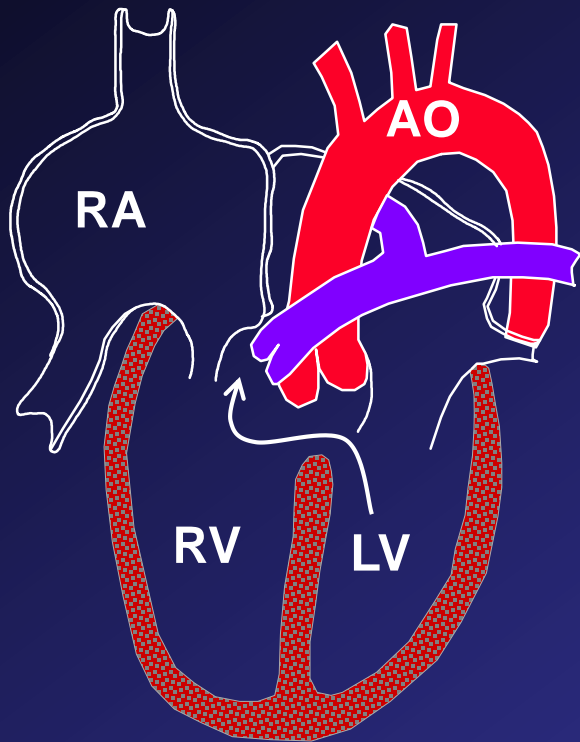
- Accounts for 10-13% of congenital heart disease
- Most common cyanotic CHD
- Usually present as asymptomatic murmur
 - Cyanosis usually develops/progresses with time
- Anatomy
 - Ventricular Septal Defect
 - Overriding Aorta
 - RV outflow obstruction
 - RV hypertrophy



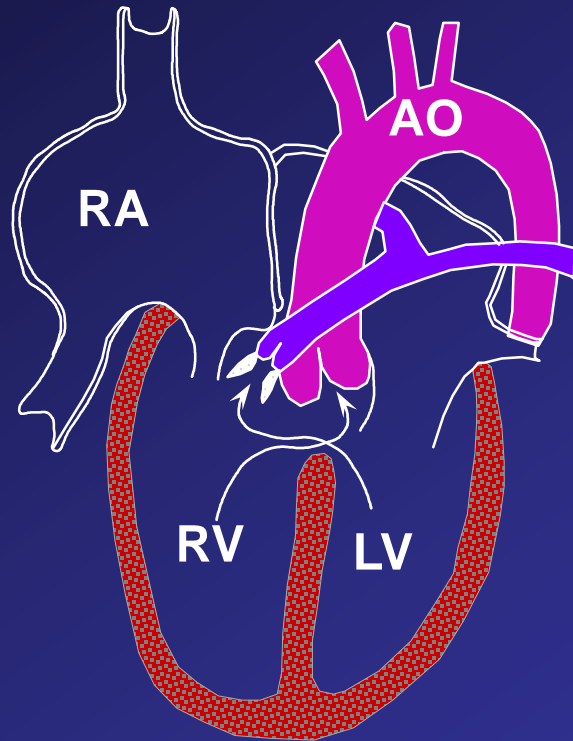
Conal Septum
Malalignment

Tetralogy of Fallot

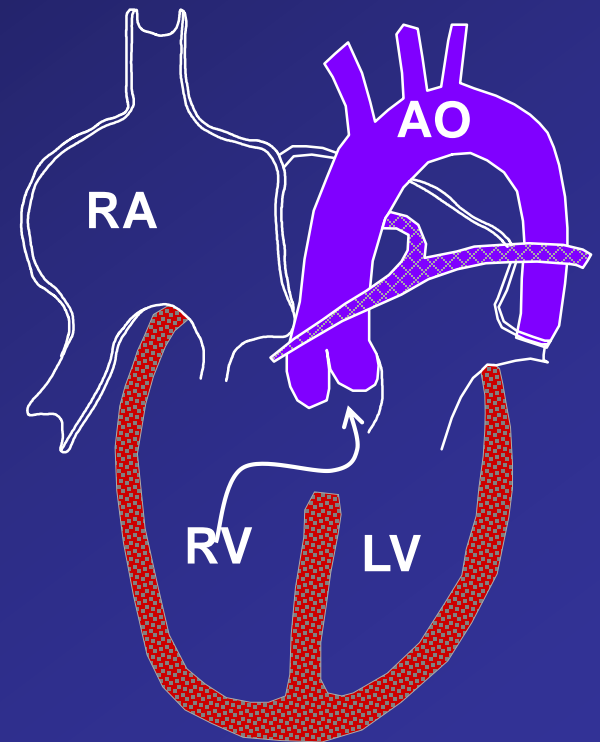
Anatomic Variables



"Pink Tetralogy"
Mild Pulmonary Stenosis



Classic Tetralogy



Severe Tetralogy or
Pulmonary Atresia

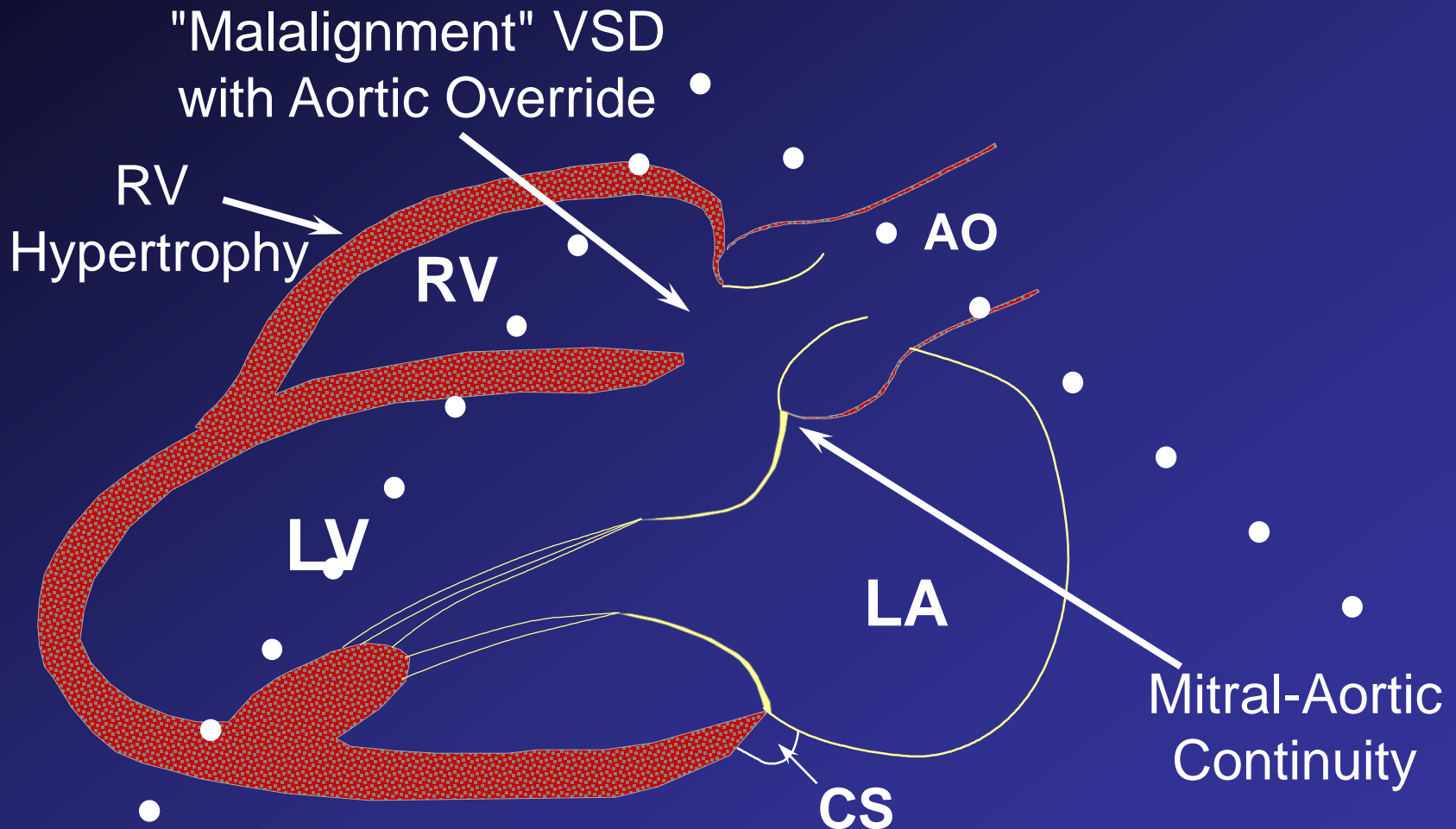
Tetralogy of Fallot

Associated Anomalies

- Valvular pulmonary stenosis - 50-60%
- Right aortic arch - 25%
- Atrial septal defect - 15%
- Coronary anomalies - 5%
- Muscular VSD - 2%

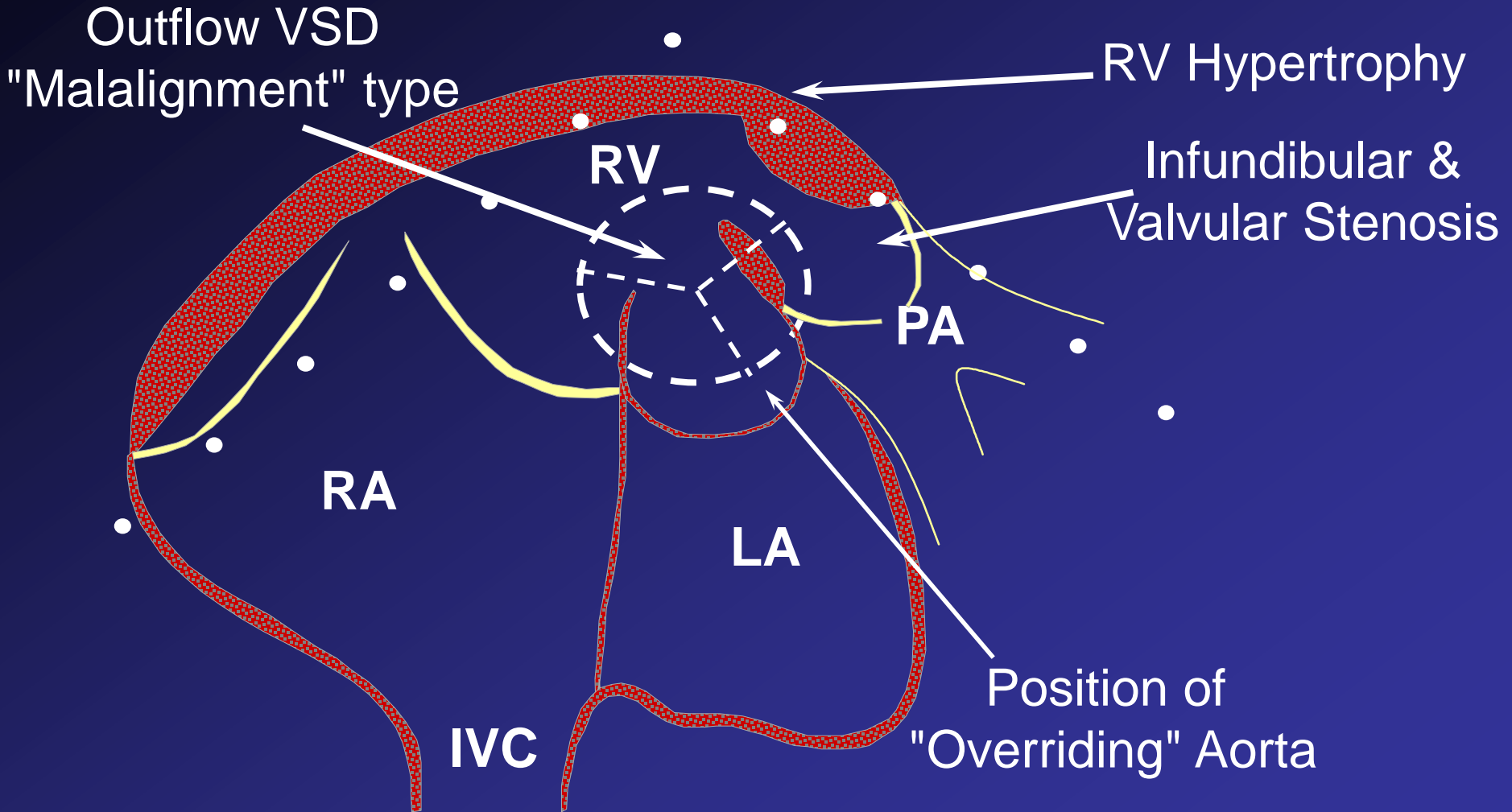
Tetralogy of Fallot

Parasternal Long Axis View



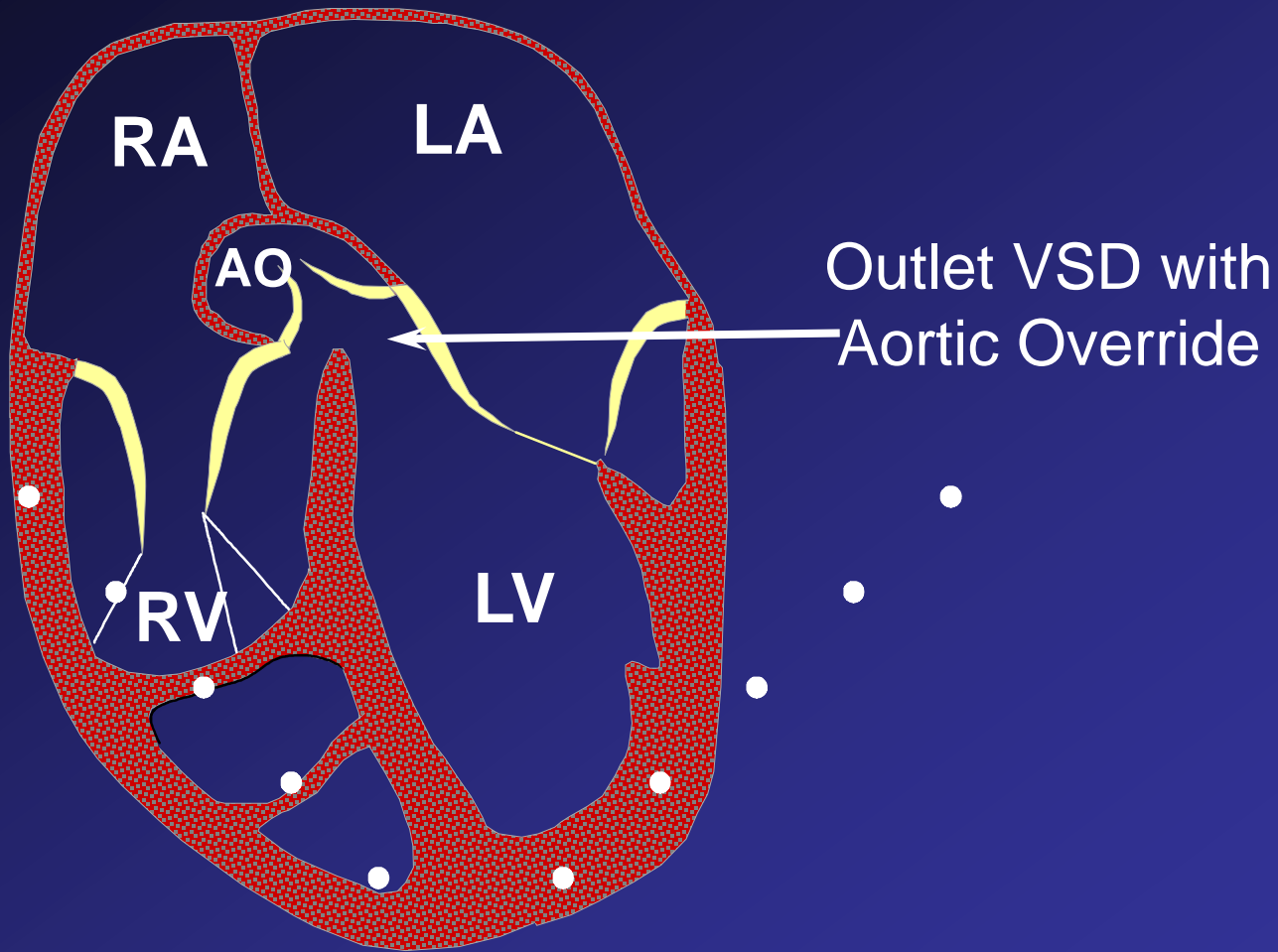
Tetralogy of Fallot

Parasternal Short Axis - Base



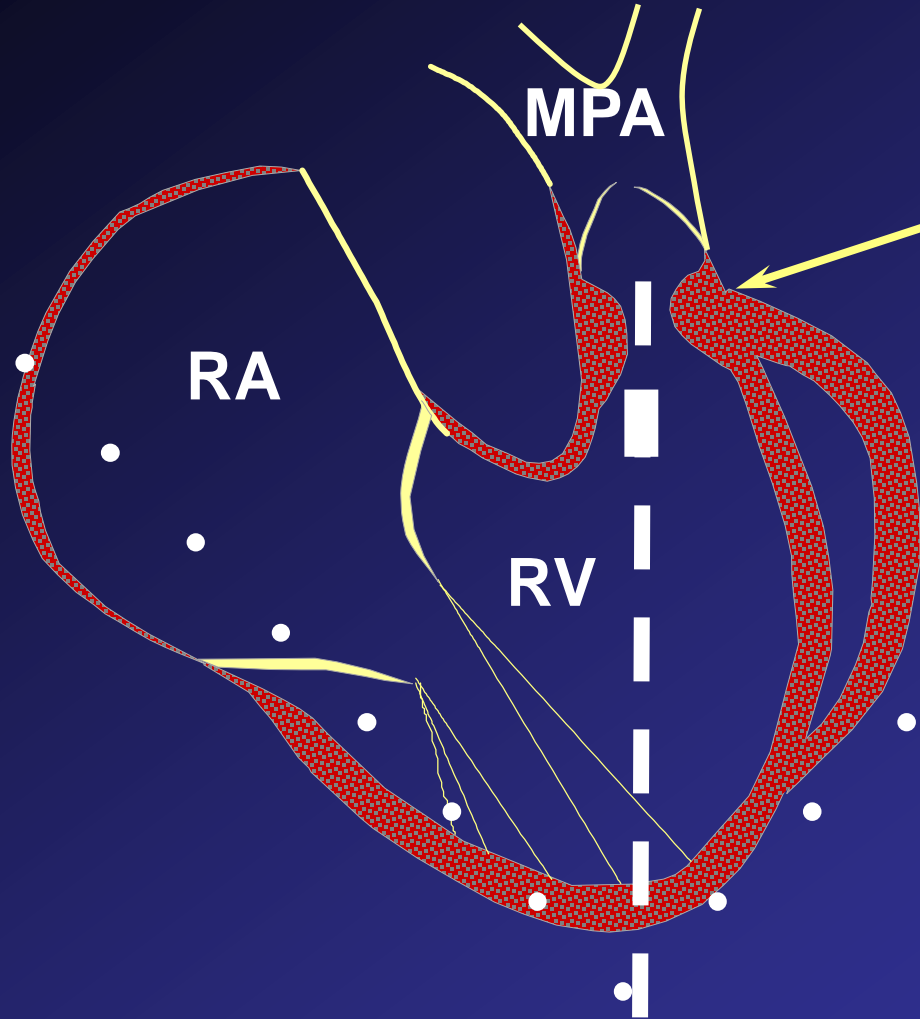
Tetralogy of Fallot

Apical Five-Chamber



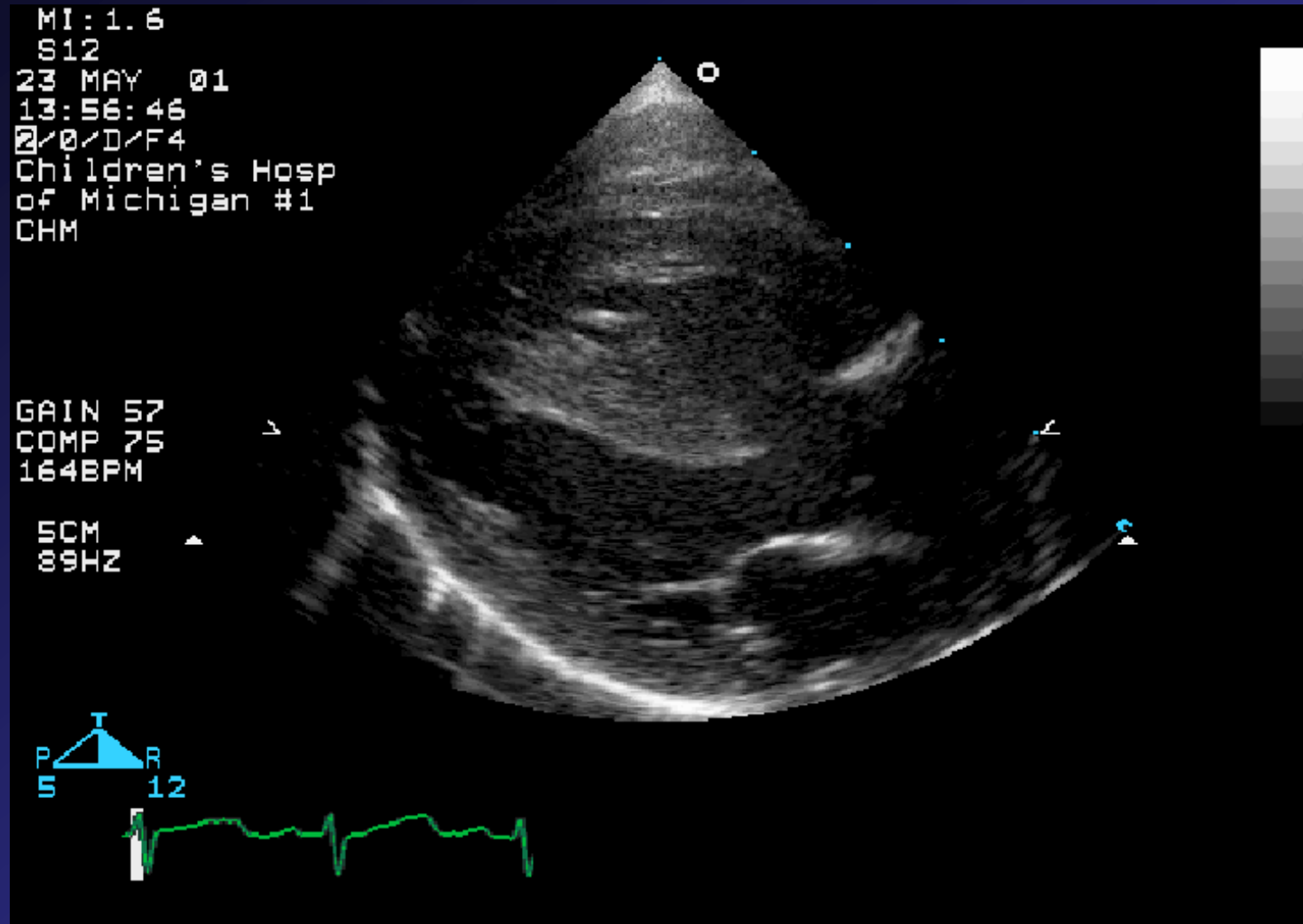
Tetralogy of Fallot

Subcostal RV Inflow/Outflow View



The right ventricular outflow obstruction is seen in this view. This is often the best angle for Doppler interrogation of the RVOT

Case 6 - Review

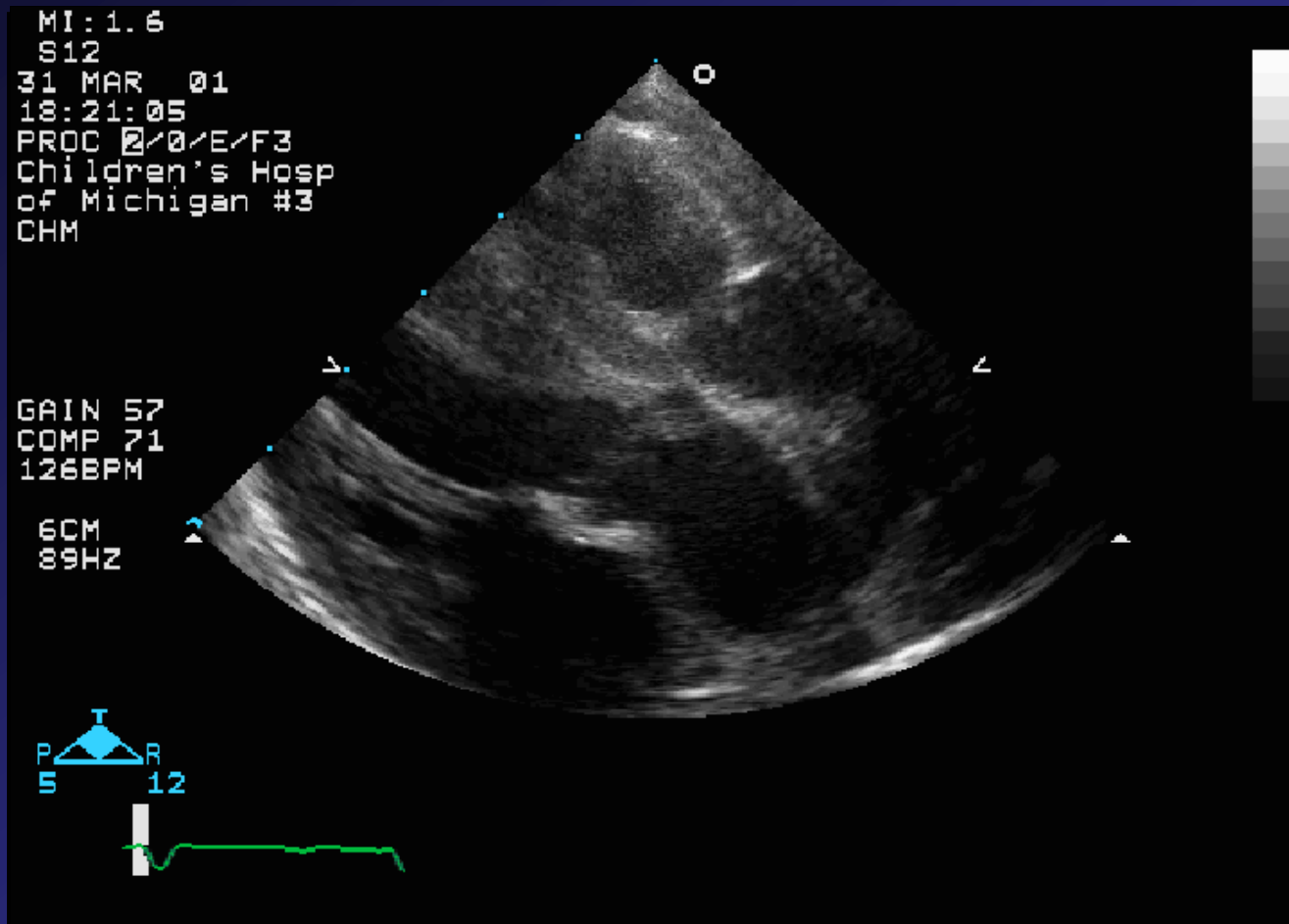


Tetralogy of Fallot


Surgical Intervention

- Timing – usually during first 6 months
- VSD closure, relief of RVOTO obstruction
- Many repairs require a trans annular RV outflow patch with results in chronic severe pulmonary regurgitation
 - Likely need for late pulmonary valve replacement
- Rastelli type repair (VSD closure + RV to pulmonary artery conduit) may be required for complex anatomy – pulmonary atresia, coronary anomalies

Case 7 – 1 day old infant with tachypnea and SaO₂ of 76%

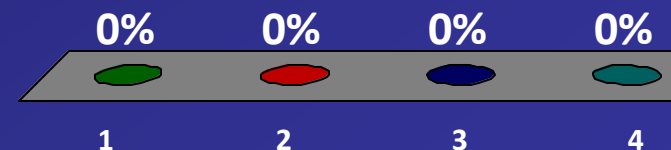


What congenital heart defect is shown:

- 0% 1. Perimembranous VSD
- 0% 2. Truncus arteriosus
- 0% 3. Corrected transposition of the great arteries (L-TGA)
-  0 4. Complete transposition of the great arteries (D-TGA)
- 0% 5. Tetralogy of Fallot

Which of the following is the preferred surgical palliation of this defect?

1. Rastelli operation
2. Mustard operation
3. Jatene operation
4. Konno operation
5. Fontan operation

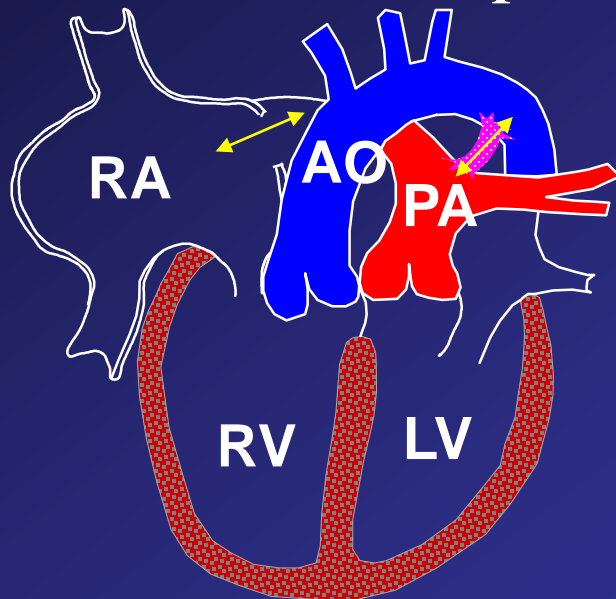


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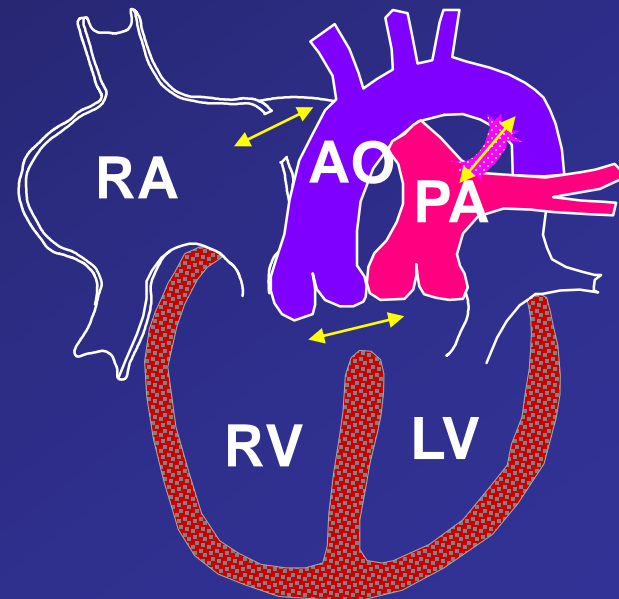
D-Transposition of the Great Arteries

- Most common cyanotic CHD presenting in the newborn nursery
- 4-8% of CHD
- Very high mortality without intervention (90% at 1 year of life)

D-TGA Intact Septum



D-TGA w/ VSD



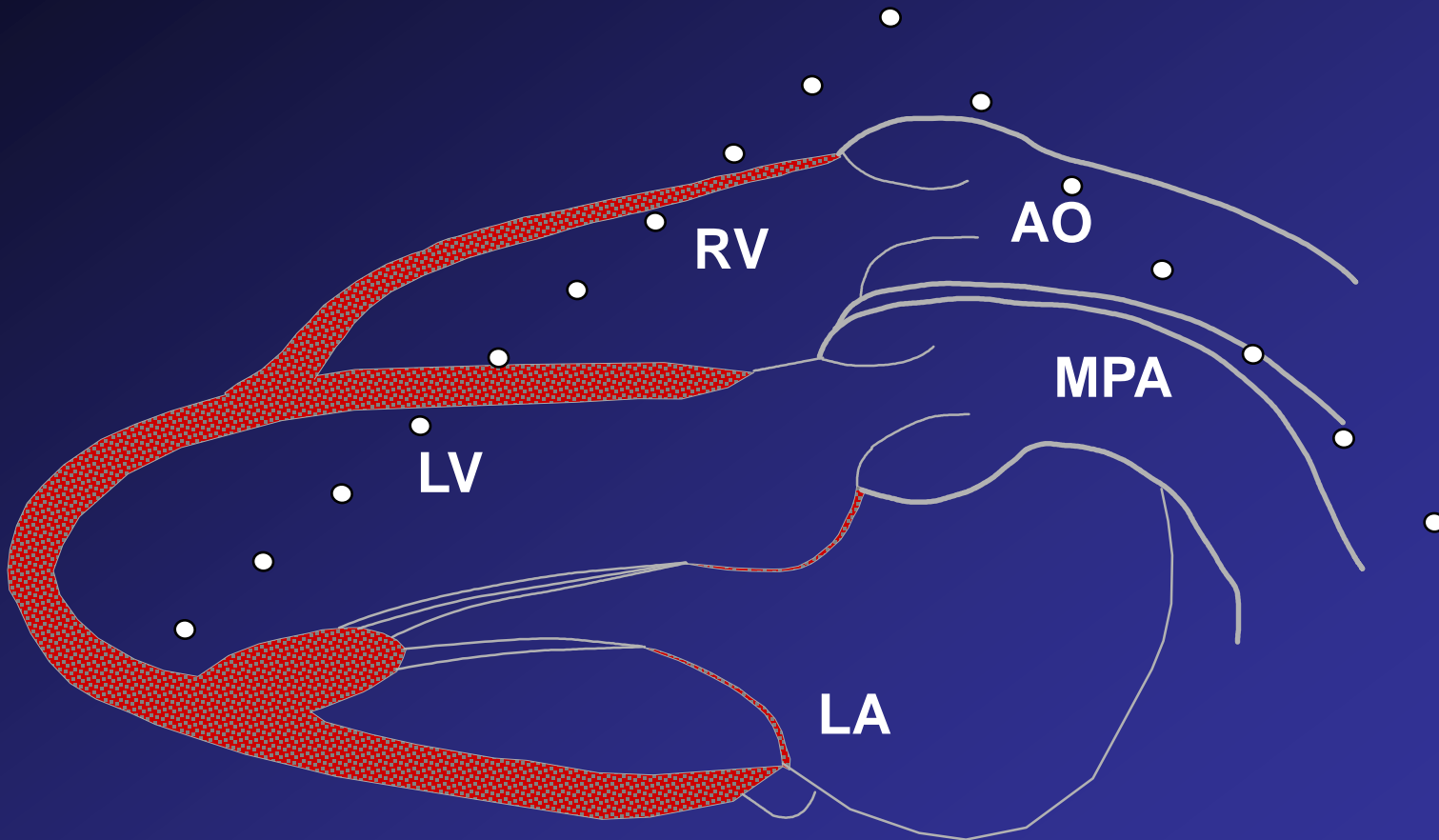
D-Transposition of the Great Arteries

Associated Anomalies

- VSD 40-45%
- Coronary anomalies ~ 40%
- Pulmonary stenosis (valve or sub valve) - 25%
- ASD
- PDA
- Coarctation - 5%

D-Transposition of the Great Arteries

Parasternal Long Axis View



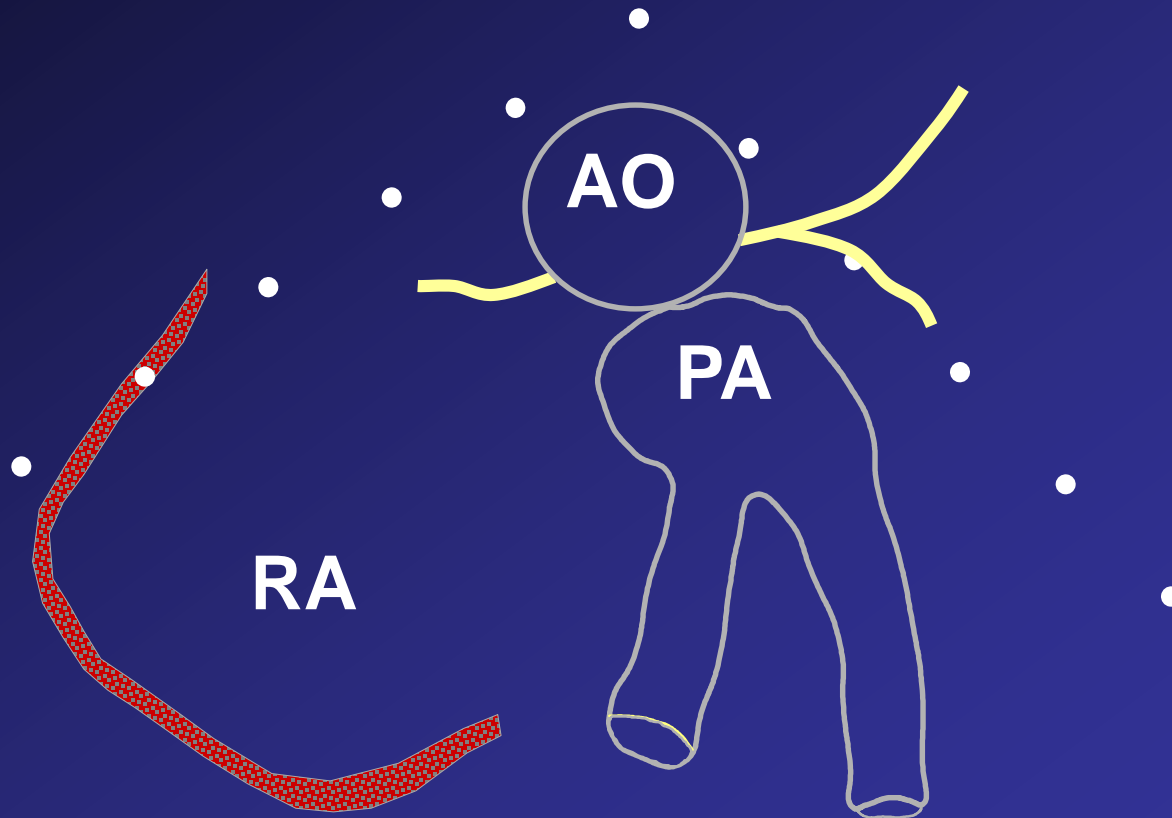
D-Transposition of the Great Arteries

Parasternal Short Axis - Base

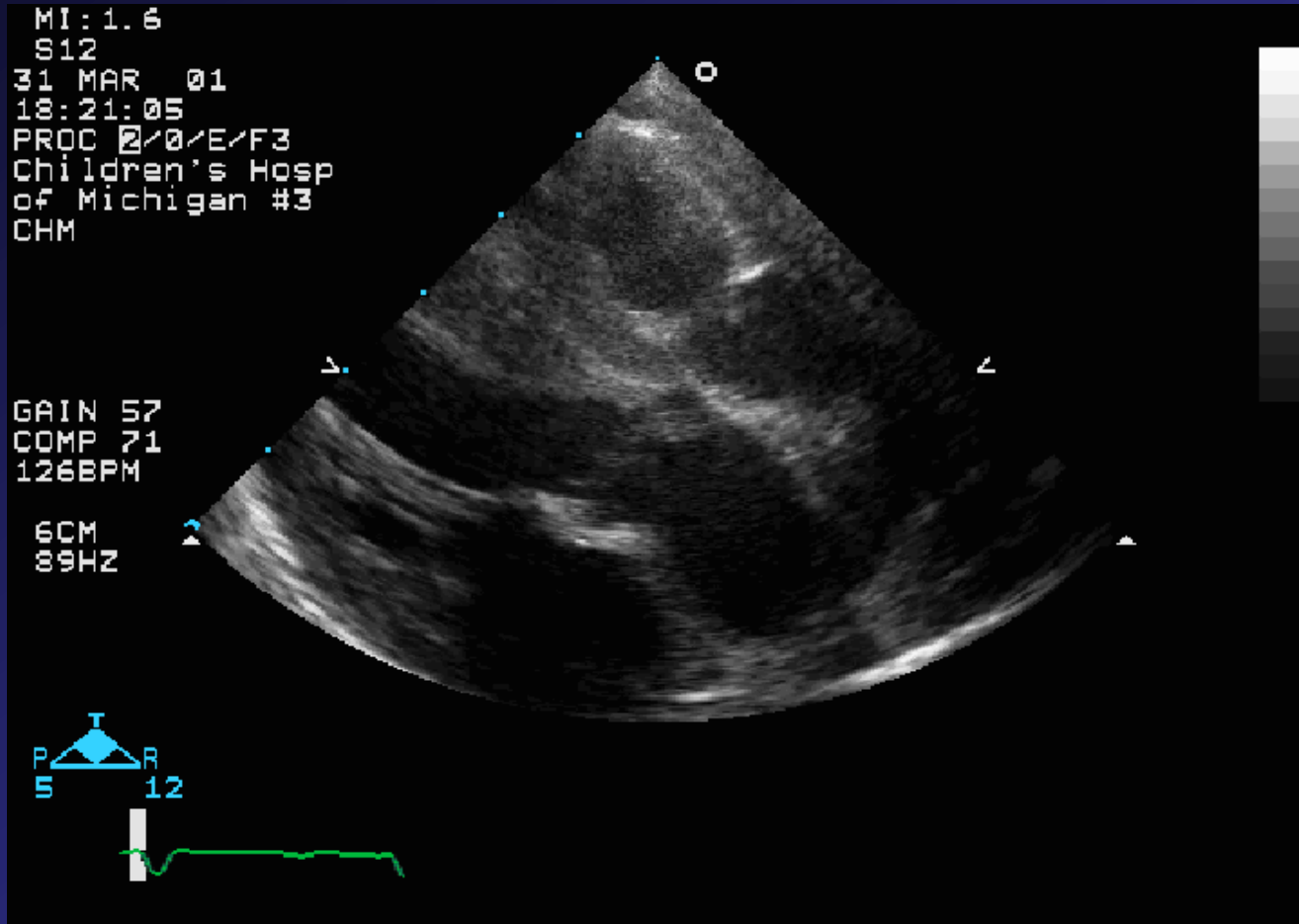


D-Transposition of the Great Arteries

High Parasternal Short Axis - Base



Case 7-Review



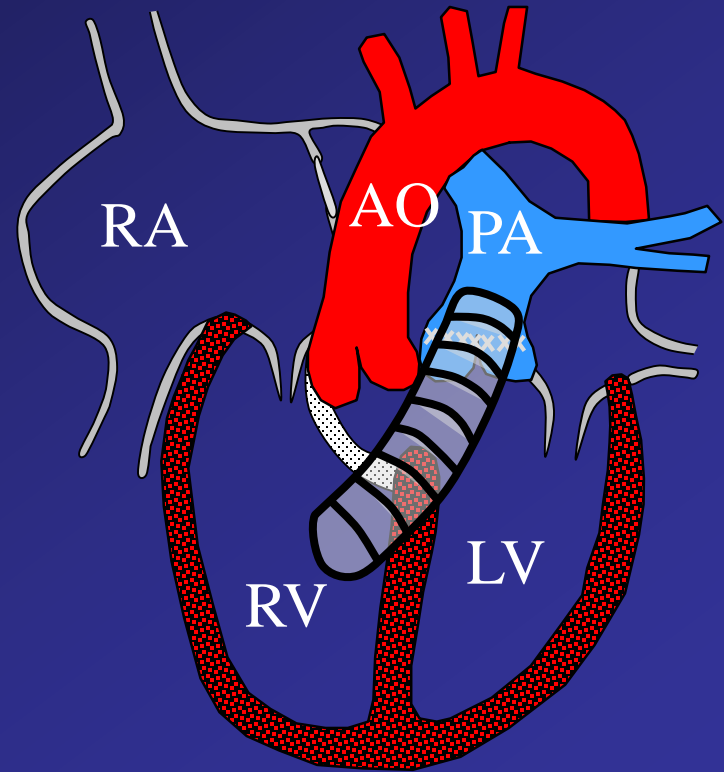
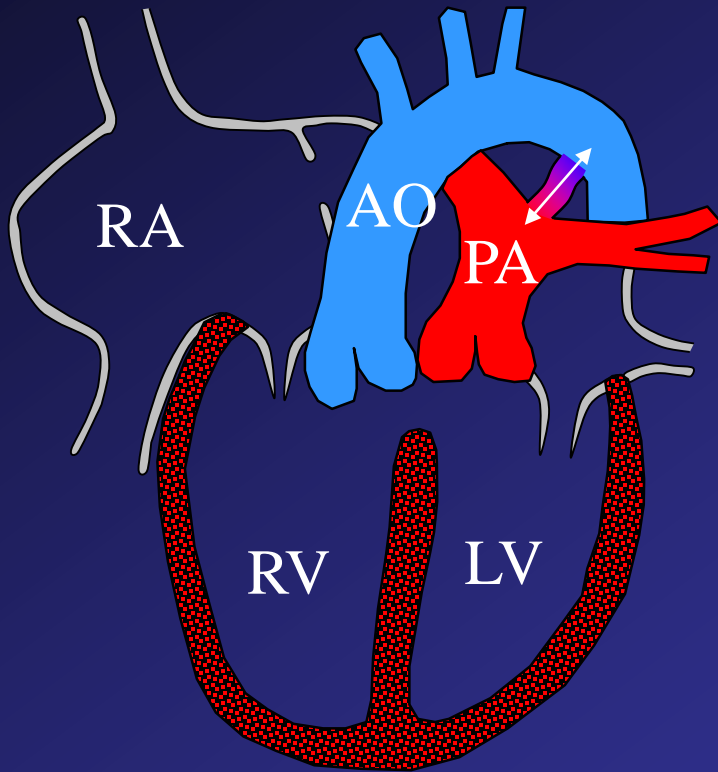
D-Transposition of the Great Arteries

Surgical Options

- Arterial switch (Jatene)
 - ♥ within first 1-2 weeks of life
- Atrial switch (Mustard/Senning)
 - ♥ Has been largely abandoned
- VSD closure/ RV-PA conduit (Rastelli)
 - ♥ within first few months of life
 - ♥ Used in the setting of d-TGA with PS/sub-PS
 - ♥ requires conduit replacement/ future surgery

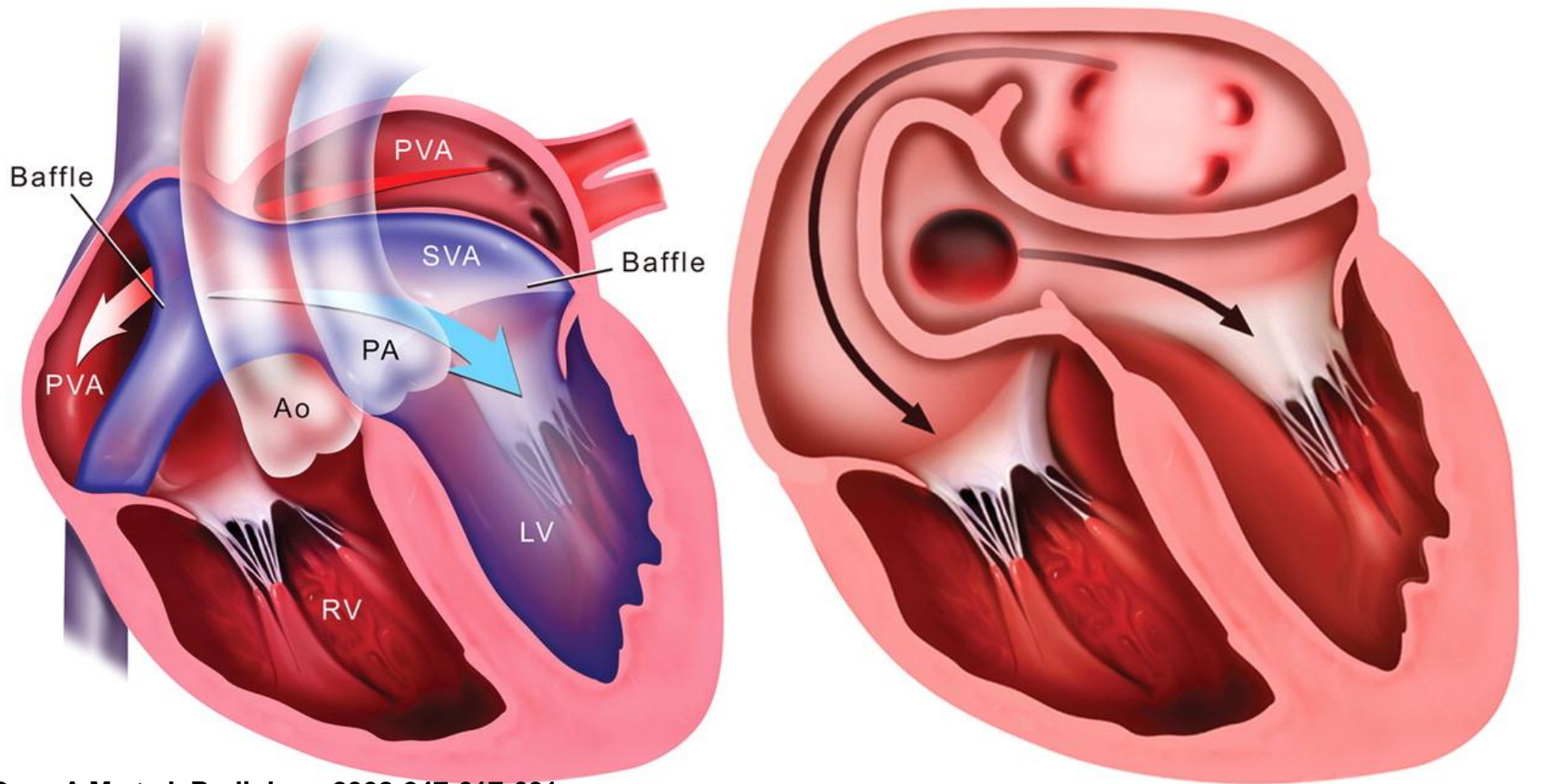
D-TGA

Intervention-Rastelli Procedure



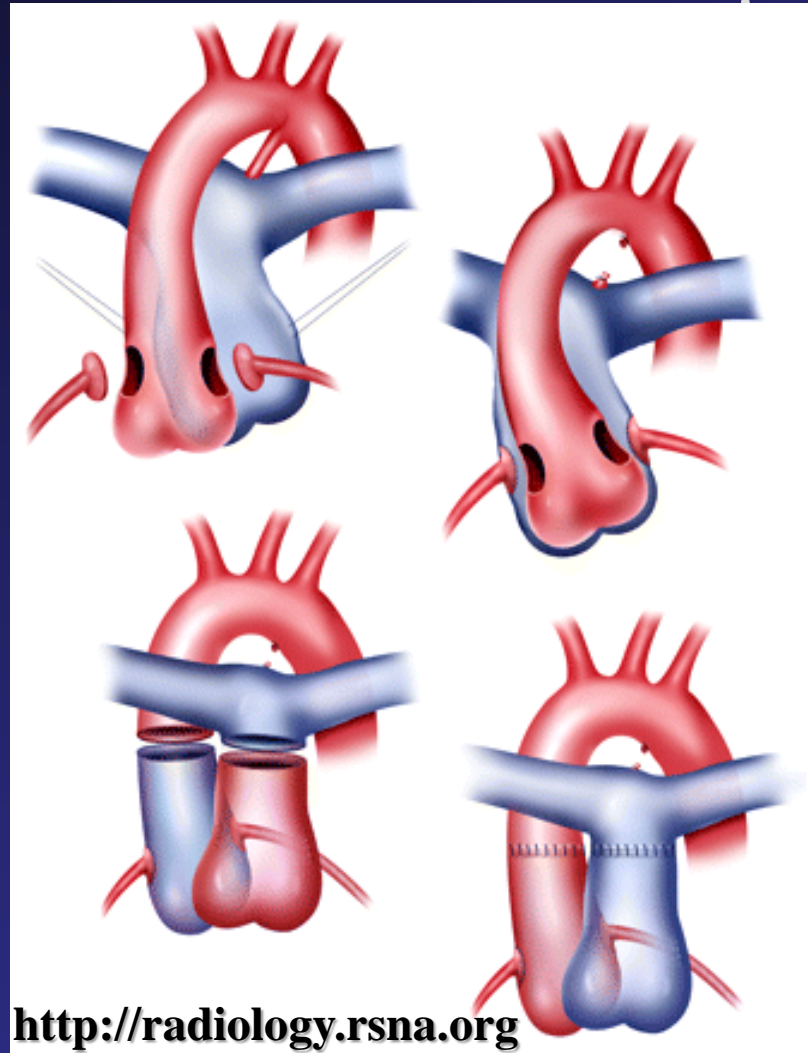
D-TGA

Atrial Switch (Mustard/Senning) Operation

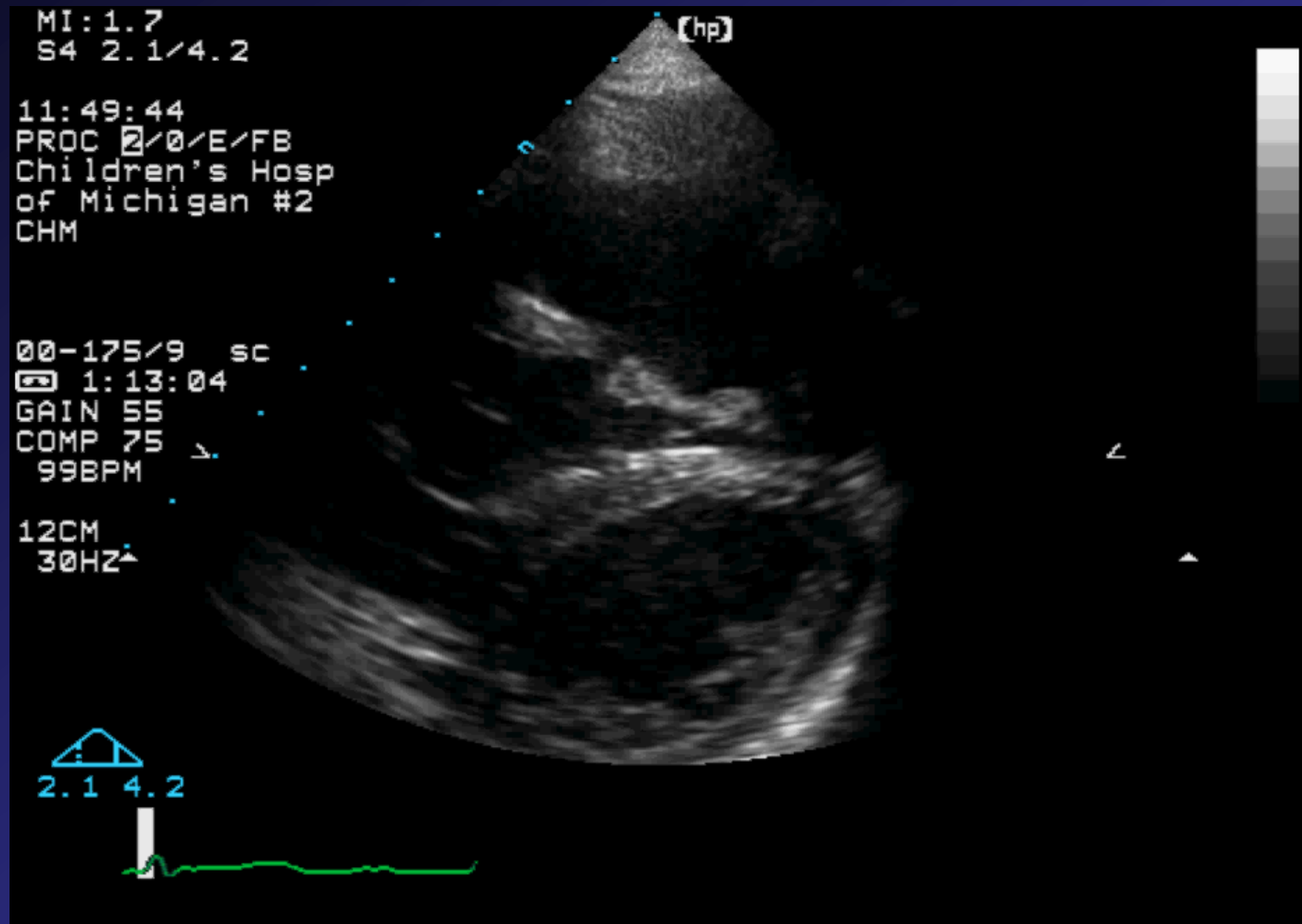


TGA

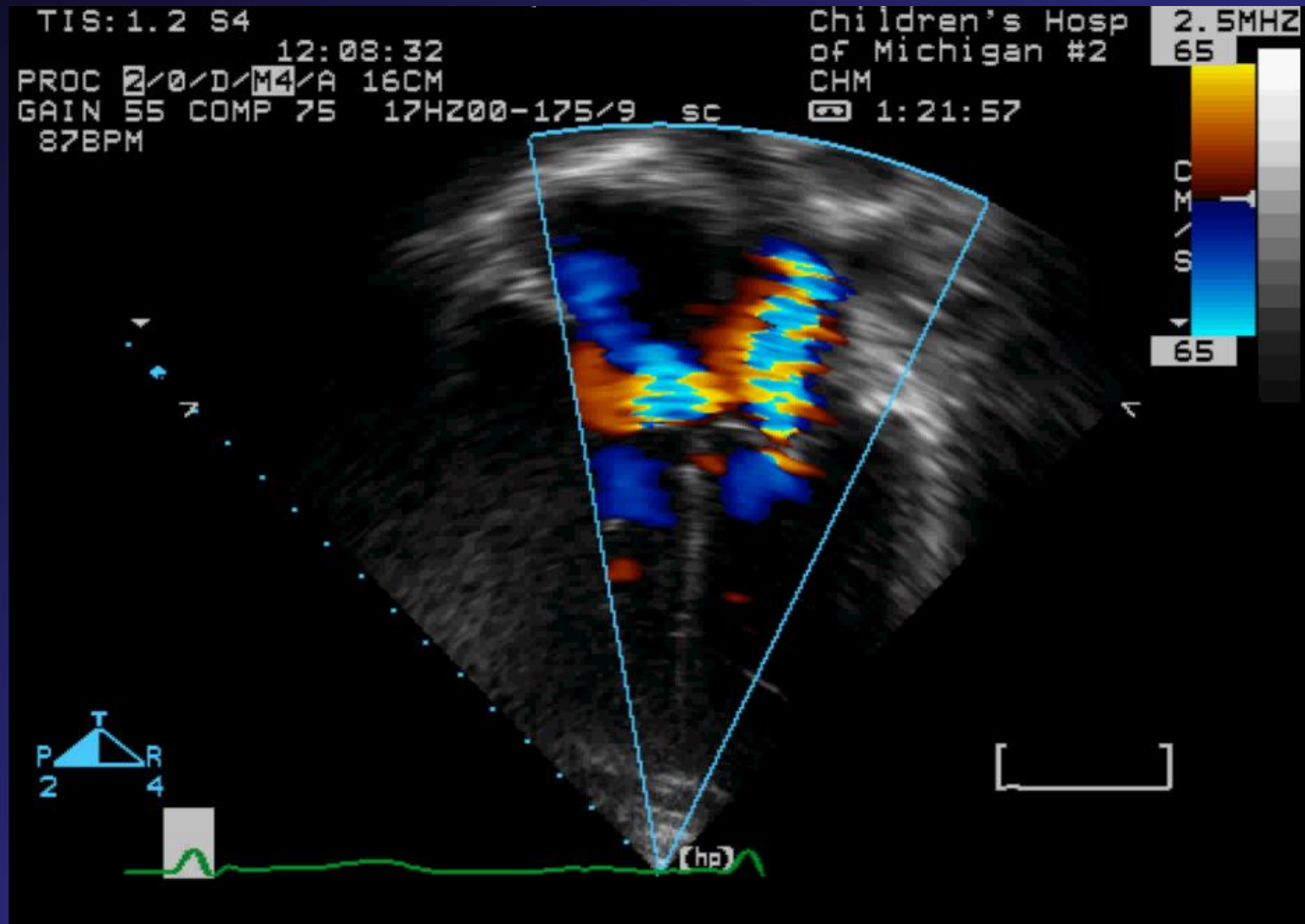
Jatene Arterial Switch Operation



Case 8 – 12 year old with asymptomatic murmur



Case 8 – 12 year old with asymptomatic murmur



The defect shown in this example is:

- 0% 1. Secundum ASD
- 0% 2. Sinus Venosus ASD
- 0% 3. Perimembranous ASD
- 0% 4. Primum ASD
- 0% 5. Coronary sinus ASD

A common associated defect with this anomaly, shown in this case, is:

1. Bicuspid aortic valve
2. Perimembranous VSD
3. Patent ductus arteriosus
4. Coarctation of the aorta
5. Cleft mitral valve

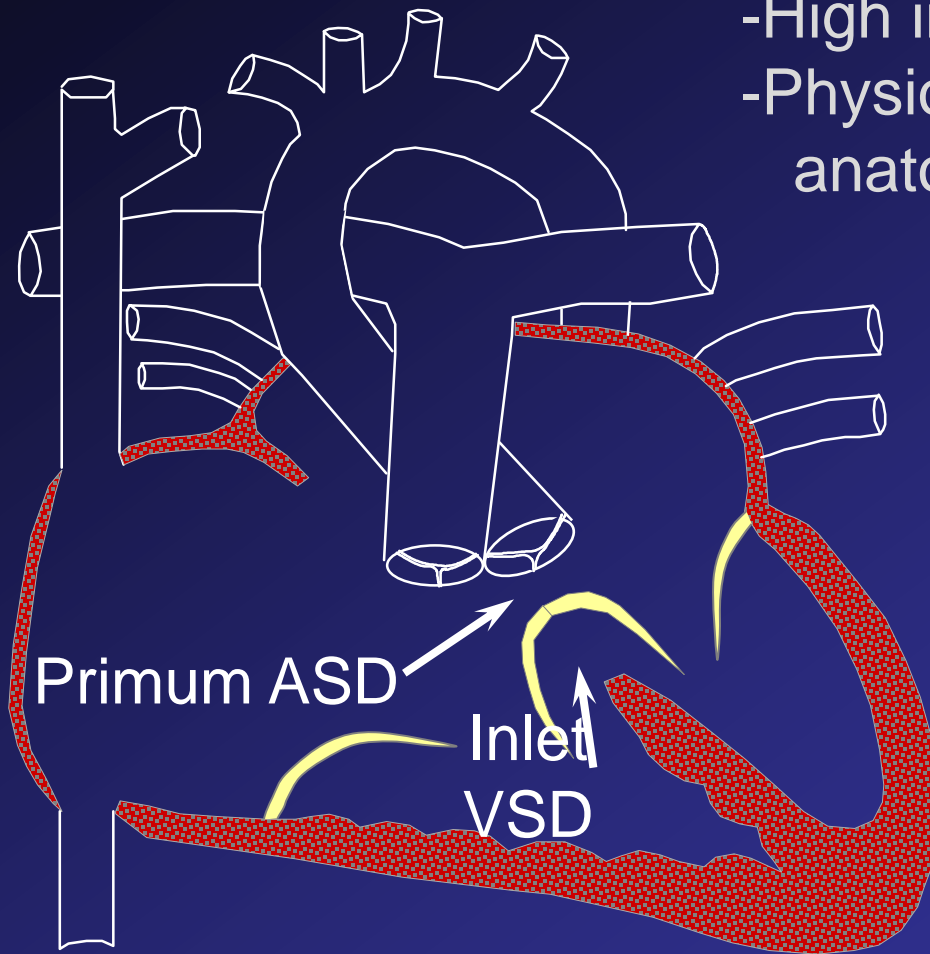


Atrioventricular Septal Defects

-3-5% of CHD

-High incidence in Down Syndrome

-Physiology depends on which anatomic defects are present



Complete AVSD

1. Primum ASD
2. Inlet VSD
3. Common AV Valve

Partial AVSD

1. Primum ASD
2. No VSD
3. Cleft Mitral Valve

Complete Atrioventricular Canal

Associated Anomalies

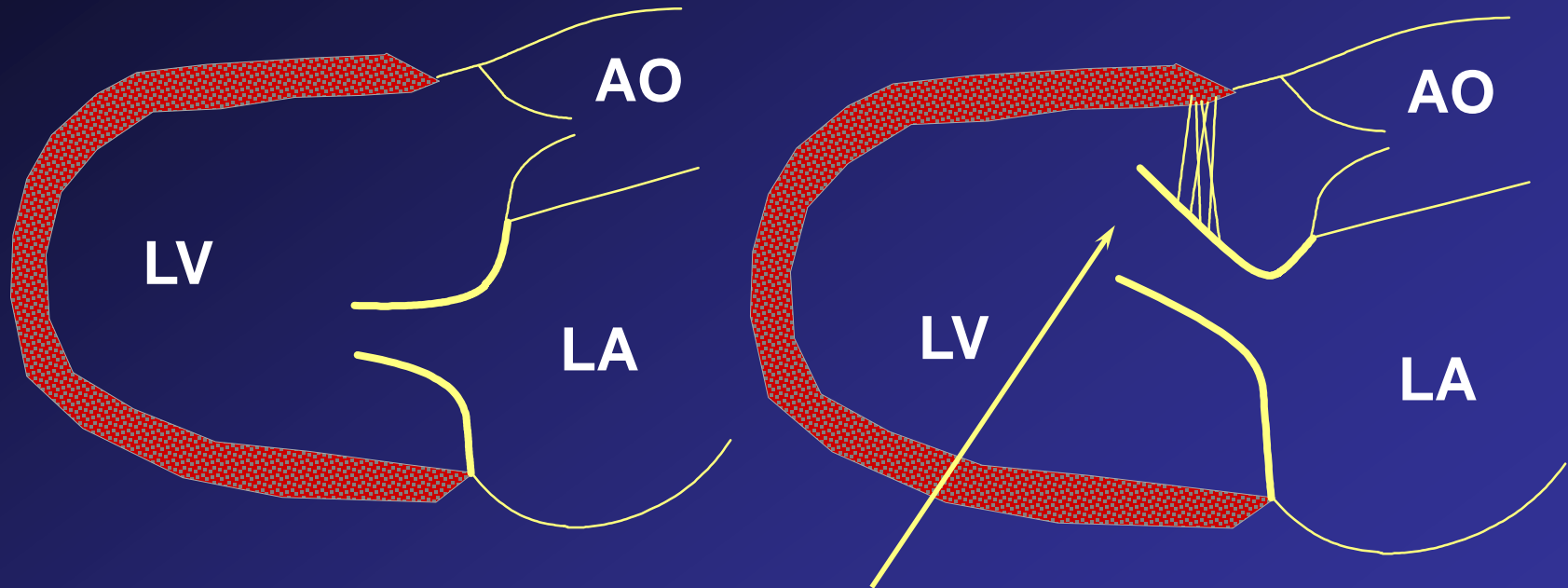
- Patent ductus arteriosus
- Hypoplasia of one ventricle
- AV valve problems - regurgitation
- LVOT obstruction

Atrioventricular Canal

Long Axis View

Normal

AV Canal

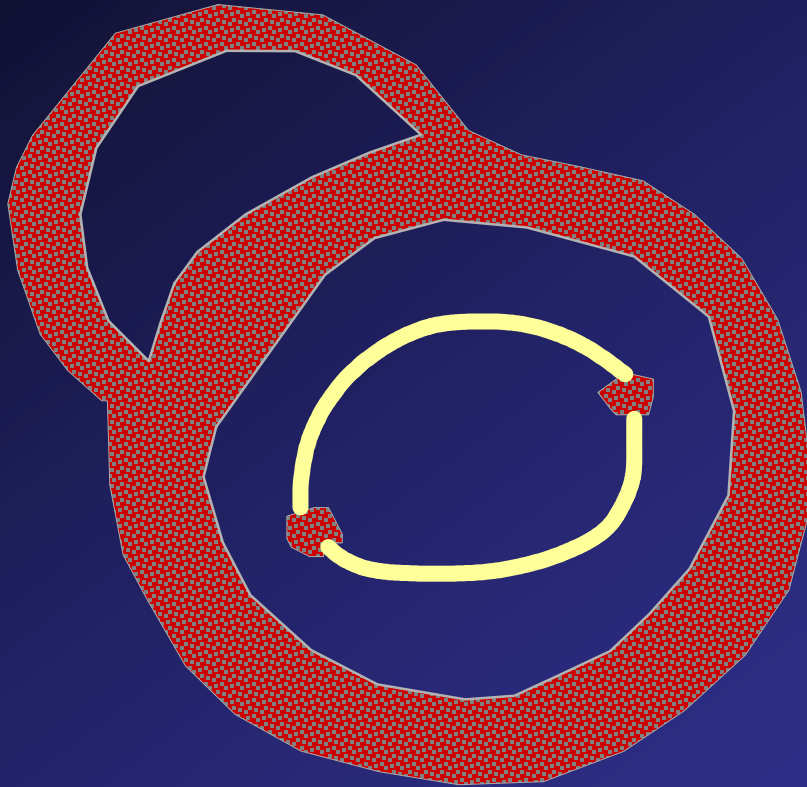


Mitral valve has abnormal orientation
and often has abnormal chordal
attachments across the LV outflow area

Cleft Mitral Valve

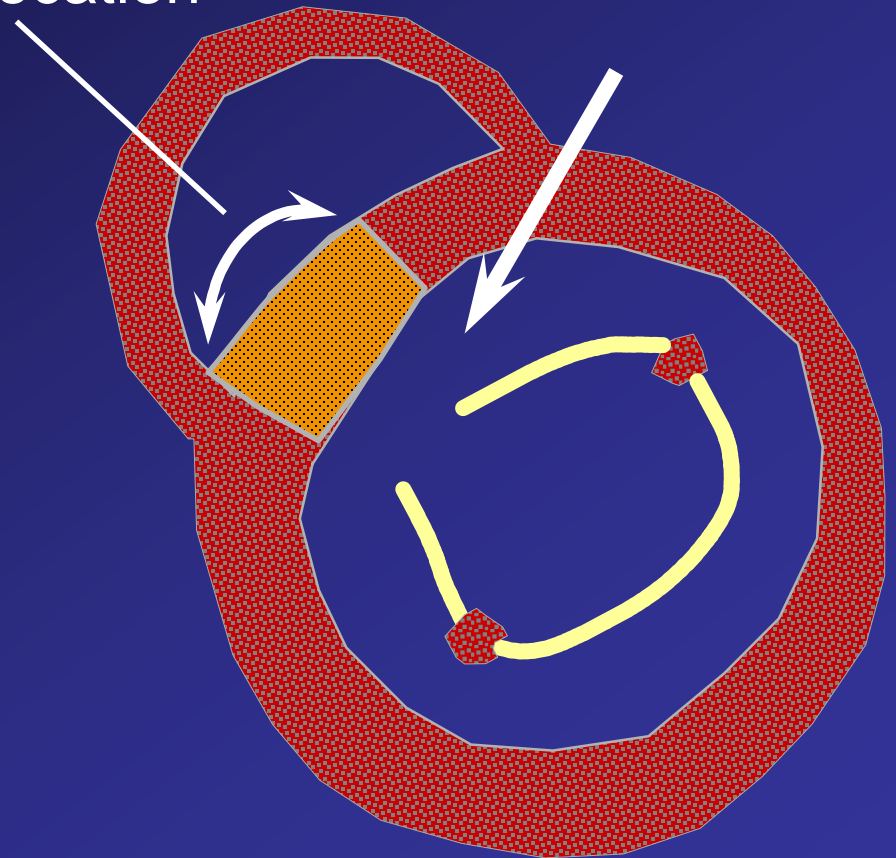
Parasternal Short-Axis View

Normal



Inlet VSD's occur
in this location

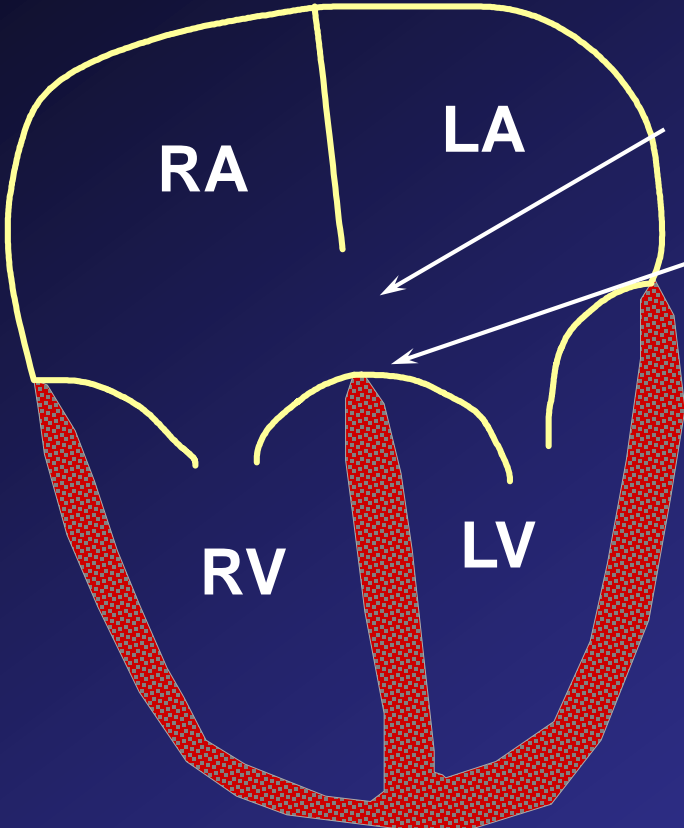
Cleft



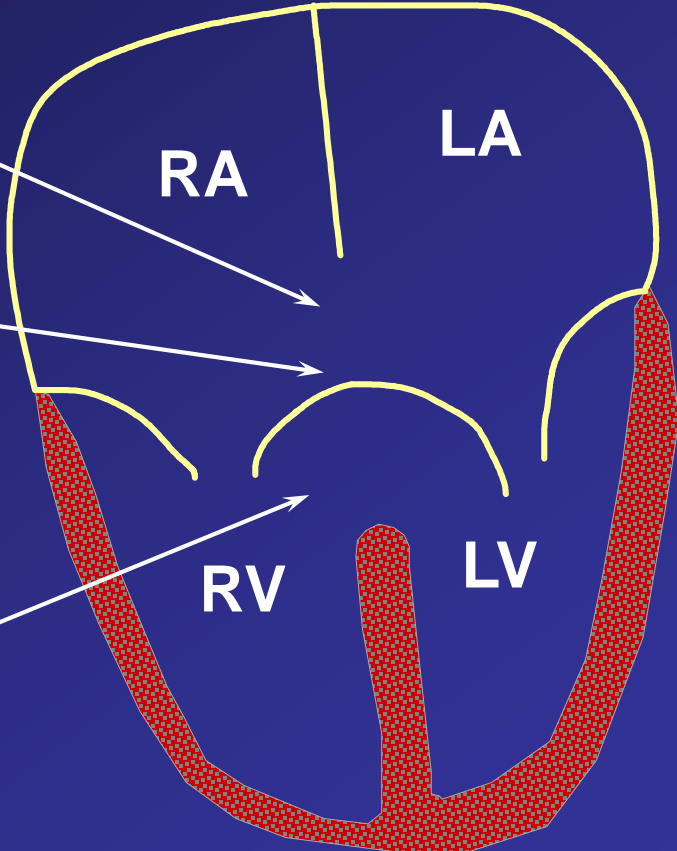
Atrioventricular Canal

Apical 4-Chamber View

Partial



Complete



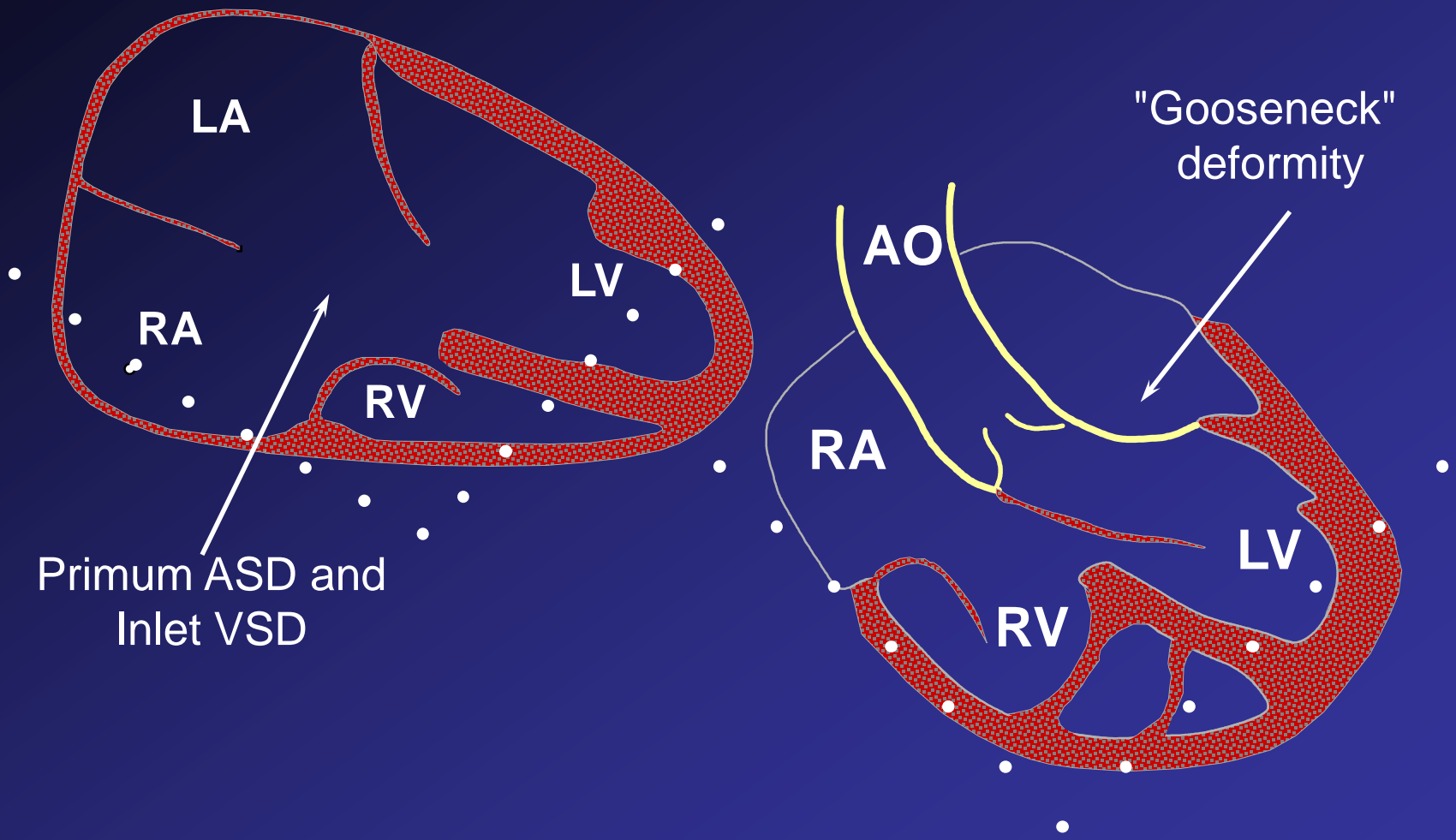
Primum ASD

AV Valves
at same level

Inlet VSD

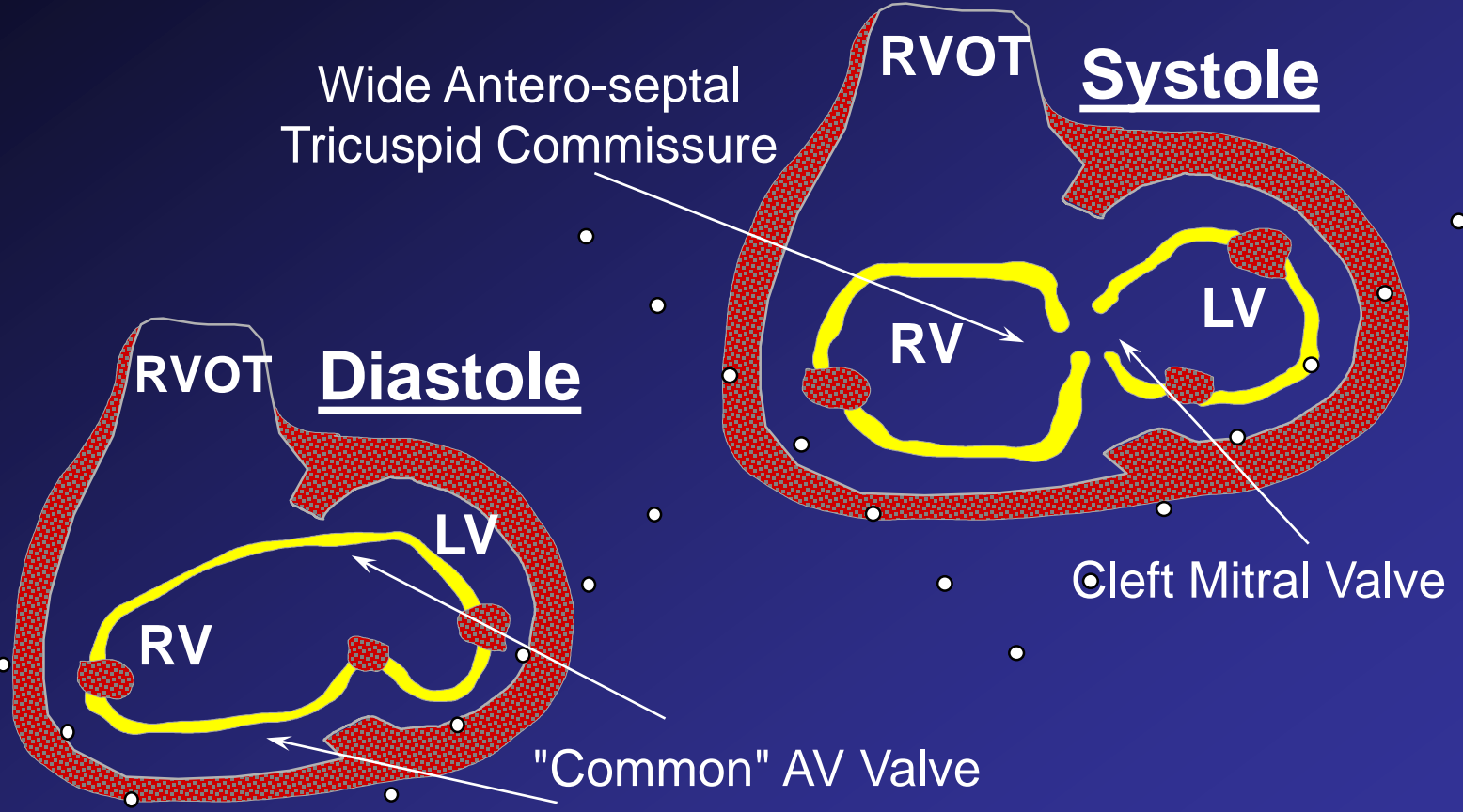
Atrioventricular Canal Defects

Subcostal Views

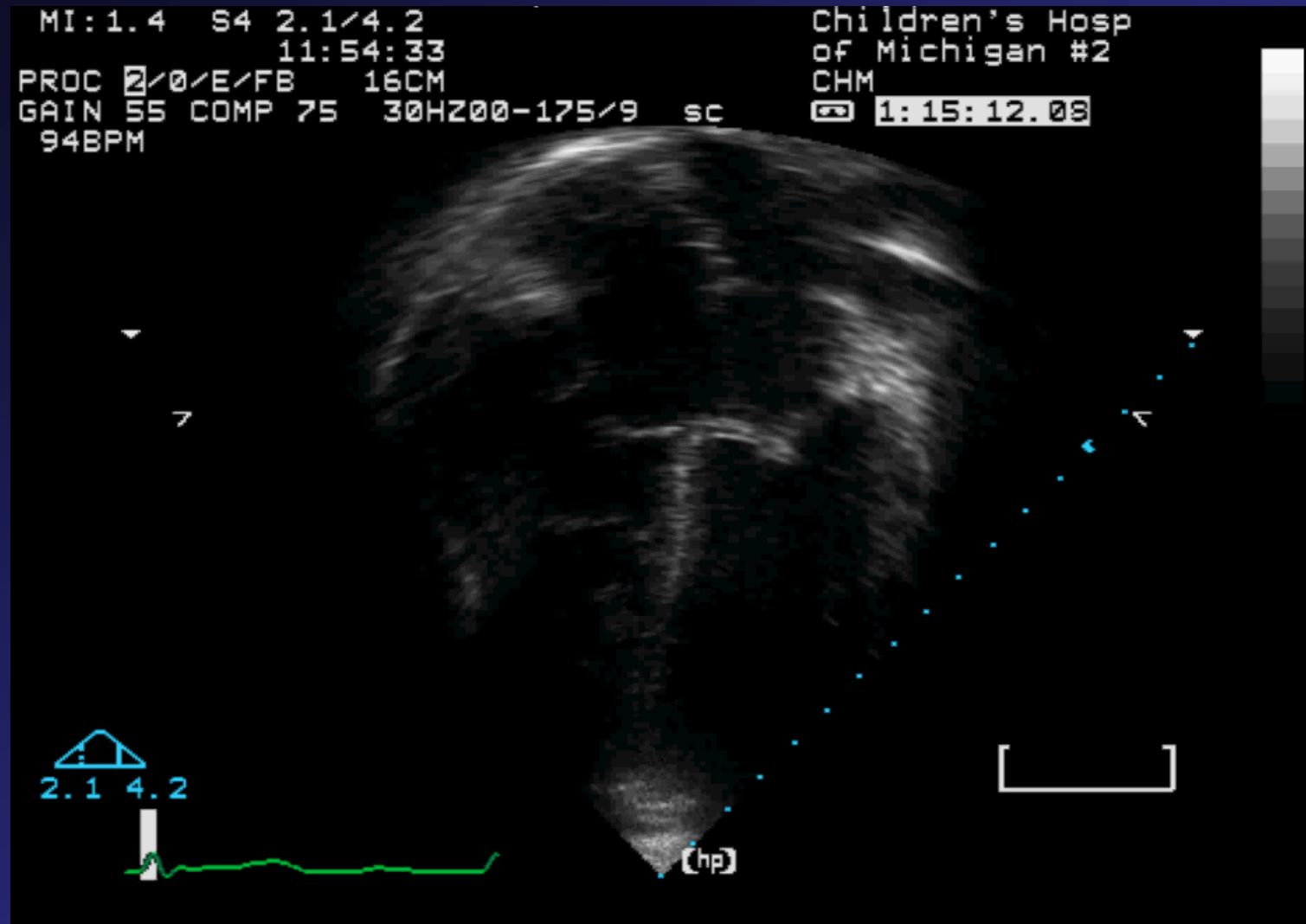


Atrioventricular Canal Defects

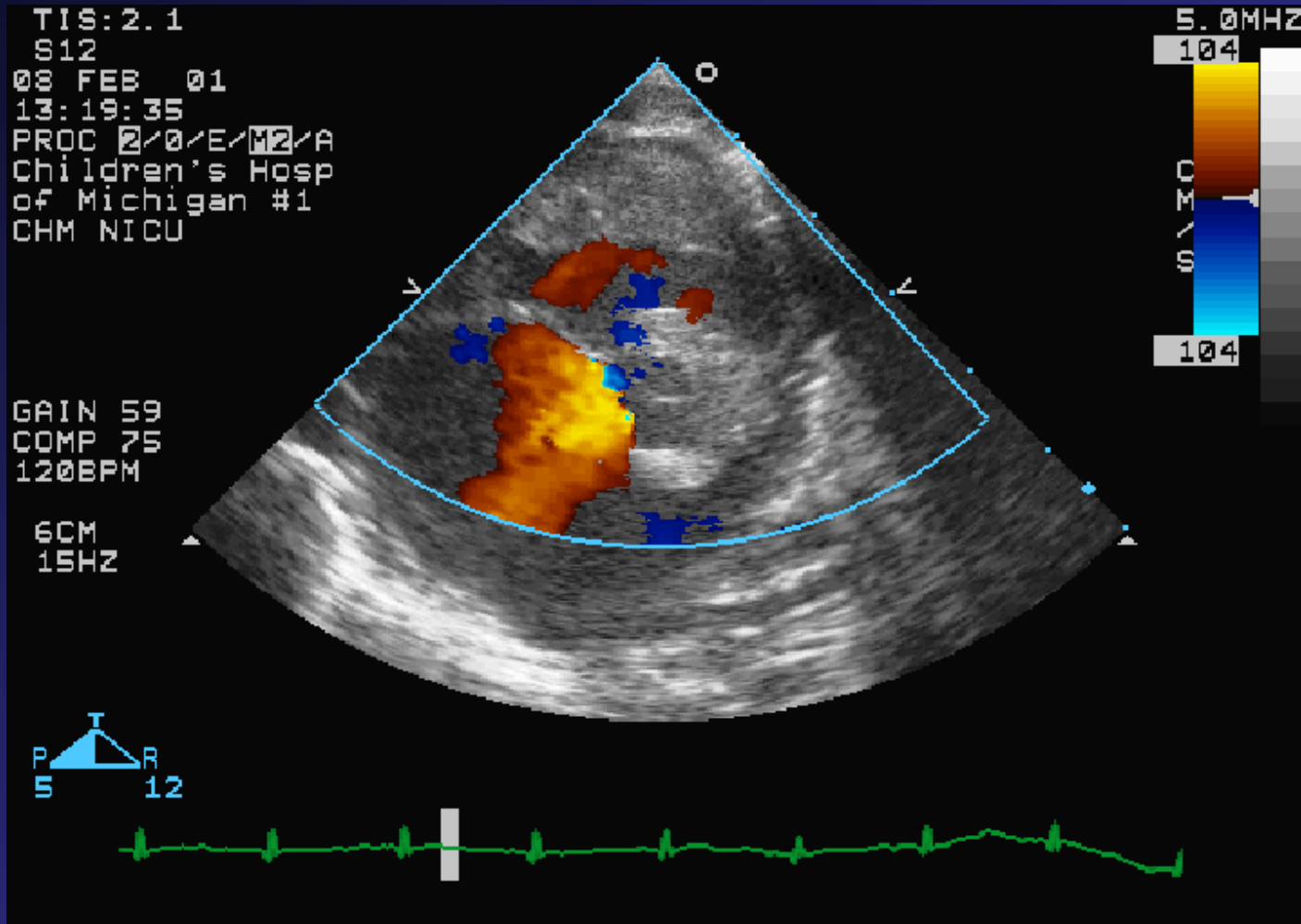
Subcostal Short Axis Views



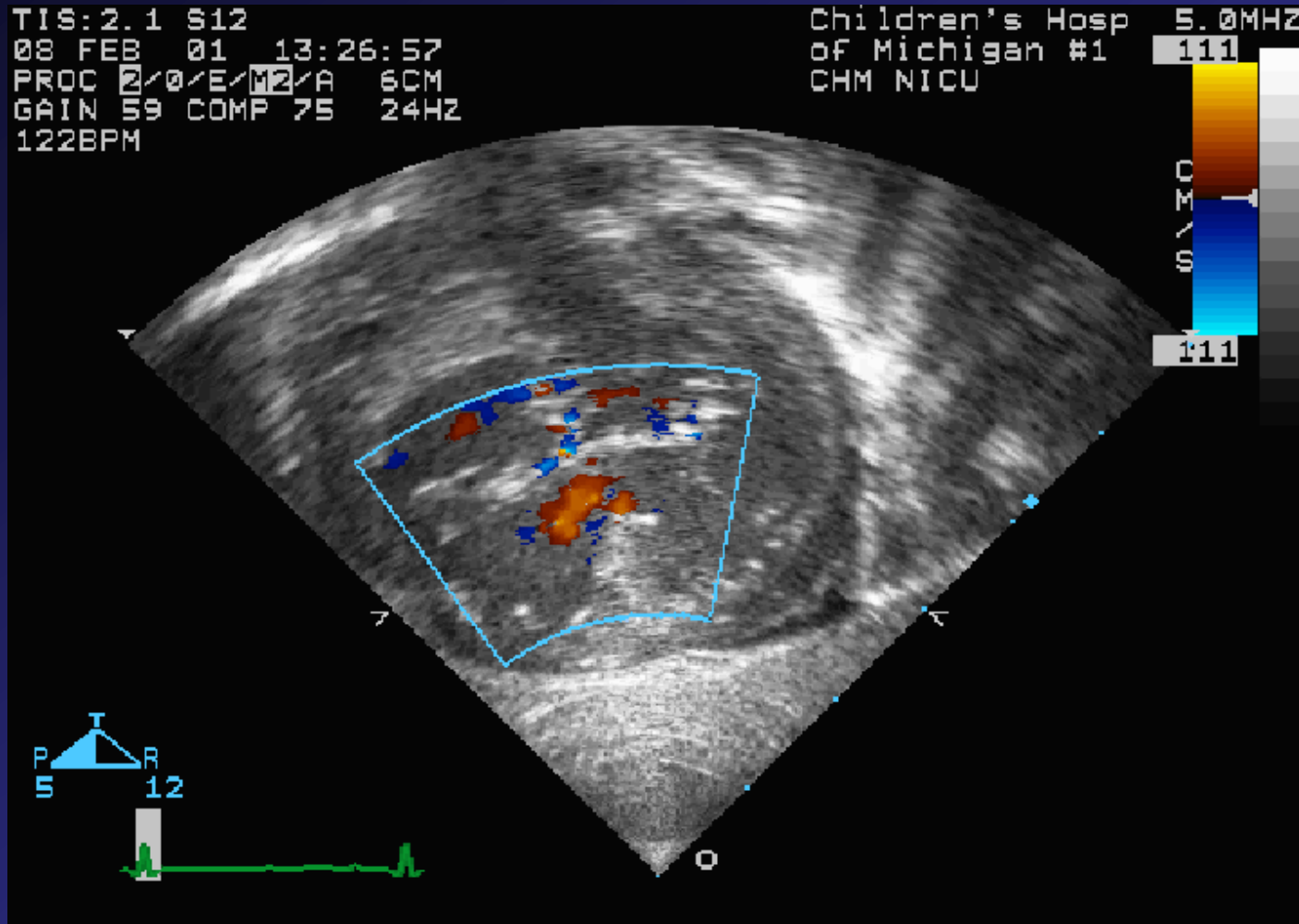
Case 8- Review



Case 9 – Complete AV Canal



Case 9 – Complete AV Canal



AV Septal Defects

Physiology

- Physiology dependent on which components of AV septal defect are present
- If 1° ASD and no VSD - physiology similar to isolated ASD (right sided volume overload)
- Complete AVSD - marked volume and pressure overload (VSD shunt physiology)
- AV valve regurgitation may exacerbate volume overload and symptoms of heart failure

AV Septal Defects

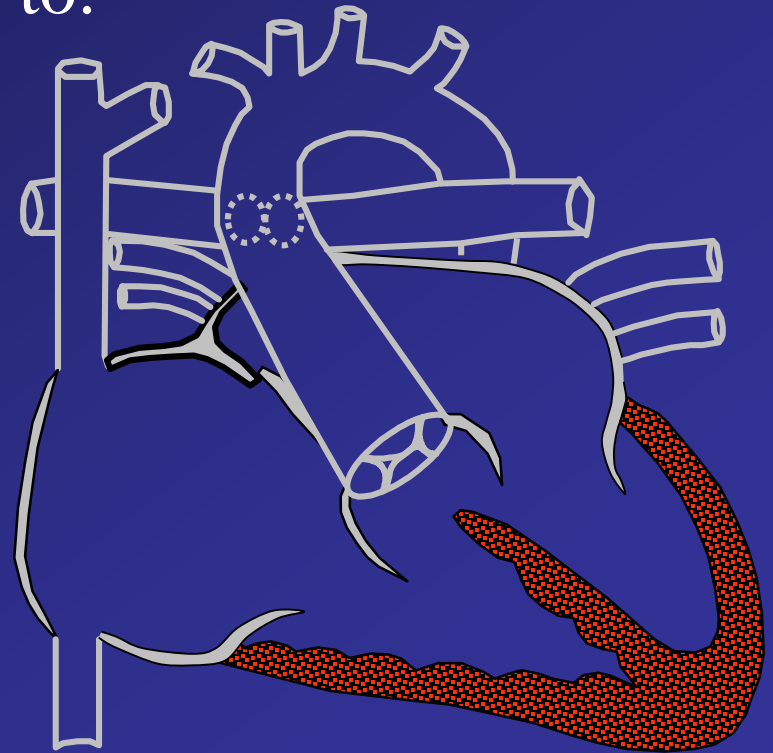
Surgical Intervention

- Partial AVSD
 - Usually electively repaired age 2-4 years
 - Complicating features (AVV regurg., LVOTO) may necessitate earlier intervention
- Complete AVSD
 - Usually repaired by 6 months of age (earlier in trisomy 21) to prevent pulmonary vascular obstructive disease

Truncus Arteriosus

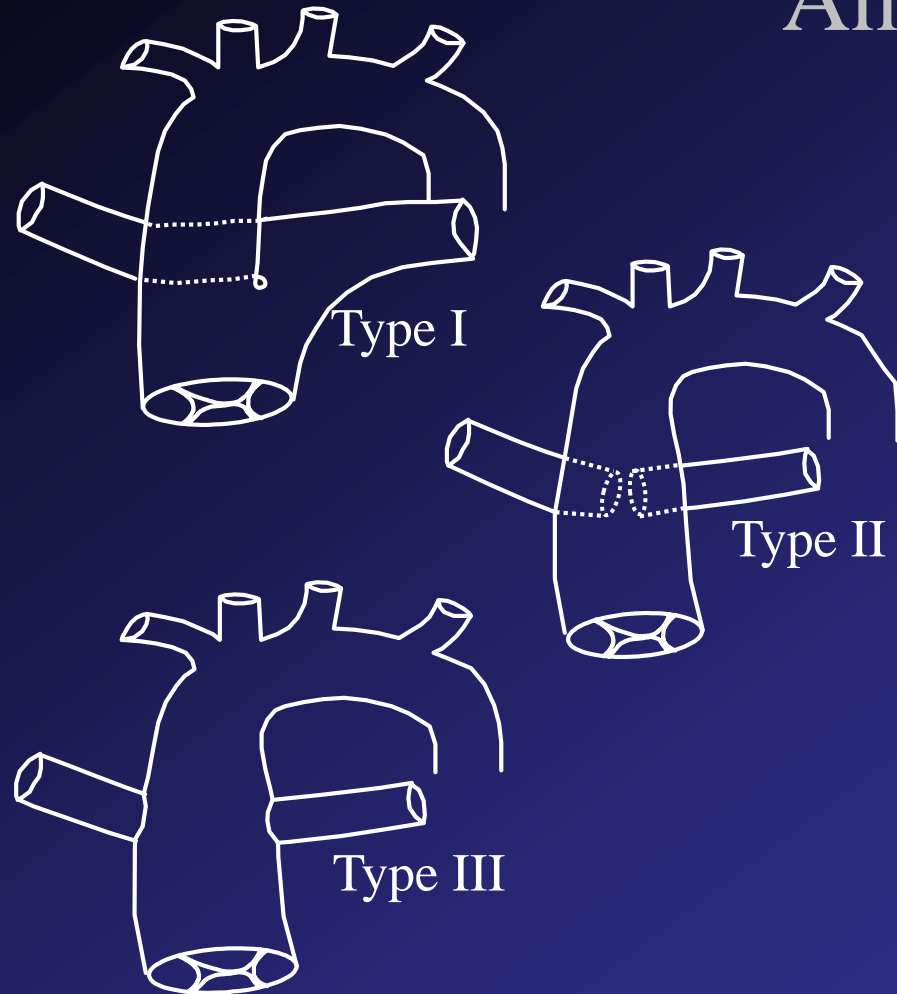
Anatomy

- Characteristic anatomy characterized by:
 - Single arterial vessel that arises from the base of the heart and gives origin to:
 - ♥ Systemic arteries
 - ♥ Pulmonary arteries
 - ♥ Coronary arteries
 - Single semilunar valve



Truncus Arteriosus

Anatomy



Associated Defects

- Abnormal coronaries (37-49%)
- Right aortic arch (30%)
- Abnormal truncal valve
- Absent pulmonary artery (16%)
- Interrupted aortic arch (15%)
- Left SVC (12%)
- Secundum ASD (9-20%)

Truncus Arteriosus

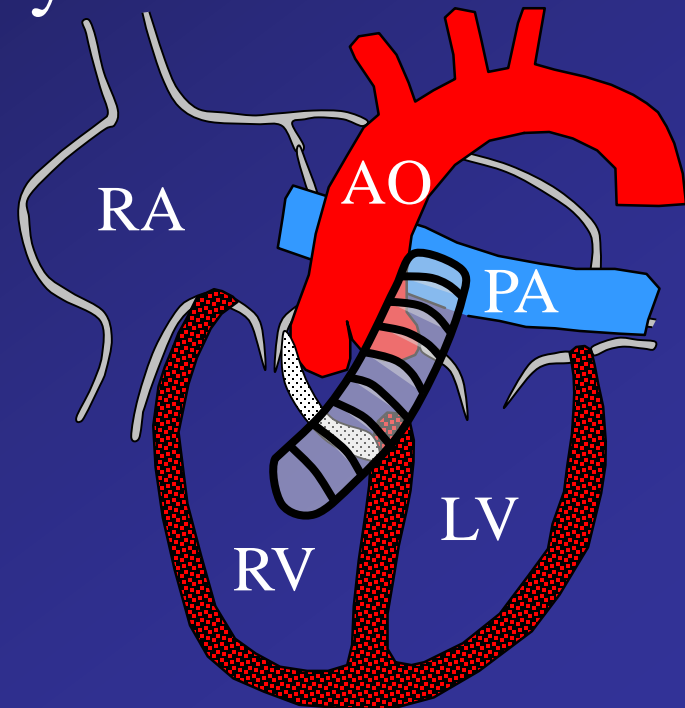
Clinical Aspects

- Patients usually present due to the presence of a cardiac murmur
- Complete mixing of systemic and pulmonary venous blood results in cyanosis
- Excessive pulmonary blood flow causes sign and symptoms of congestive heart failure
- The cyanosis is generally mild

Truncus Arteriosus

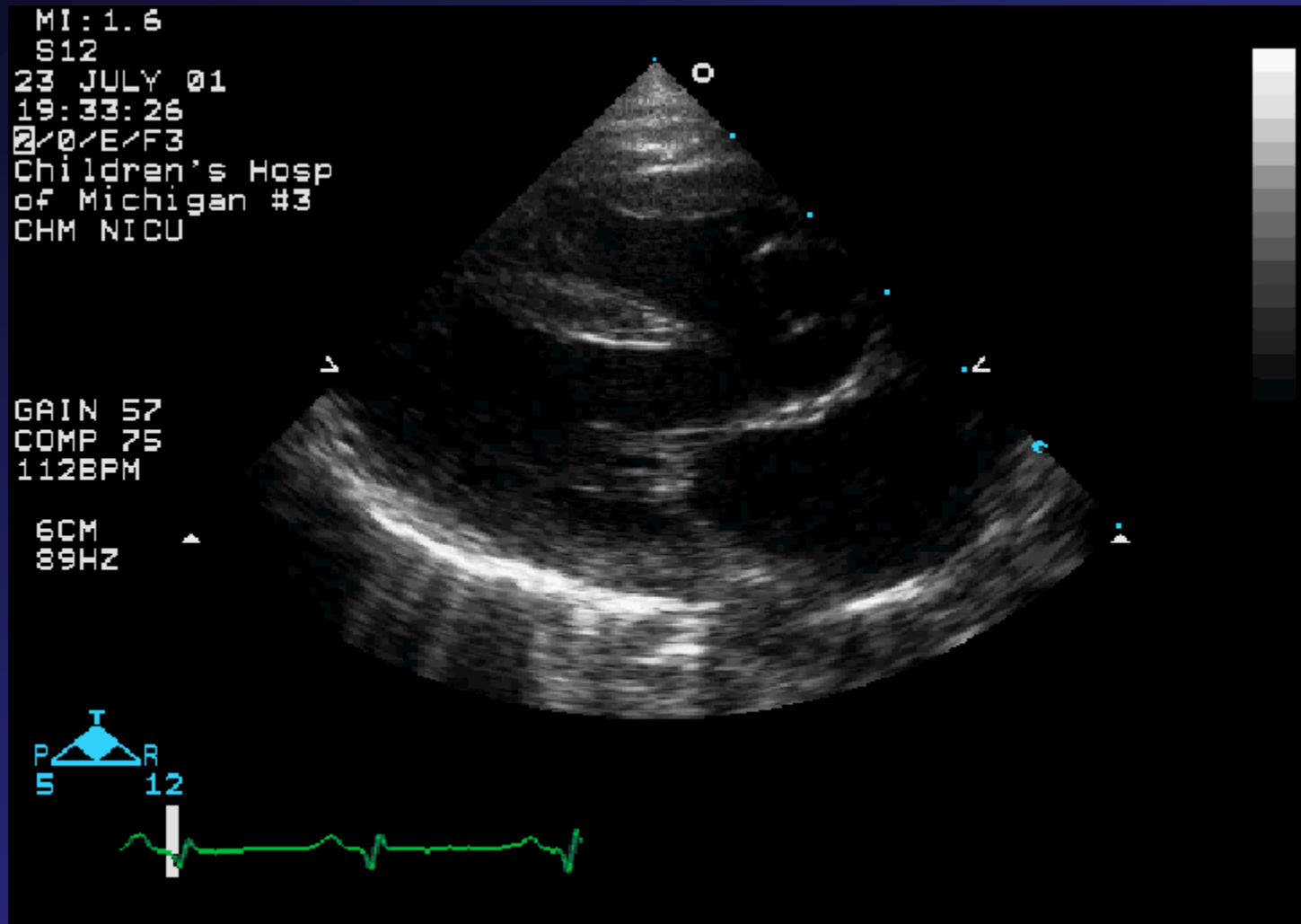
Treatment

- Requires surgical repair in the first weeks of life
- Median sternotomy requiring bypass
- Palliative PA band rarely used in the current era
- Rastelli type repair
 - Close VSD to truncus
 - Disconnect PAs
 - RV to PA conduit



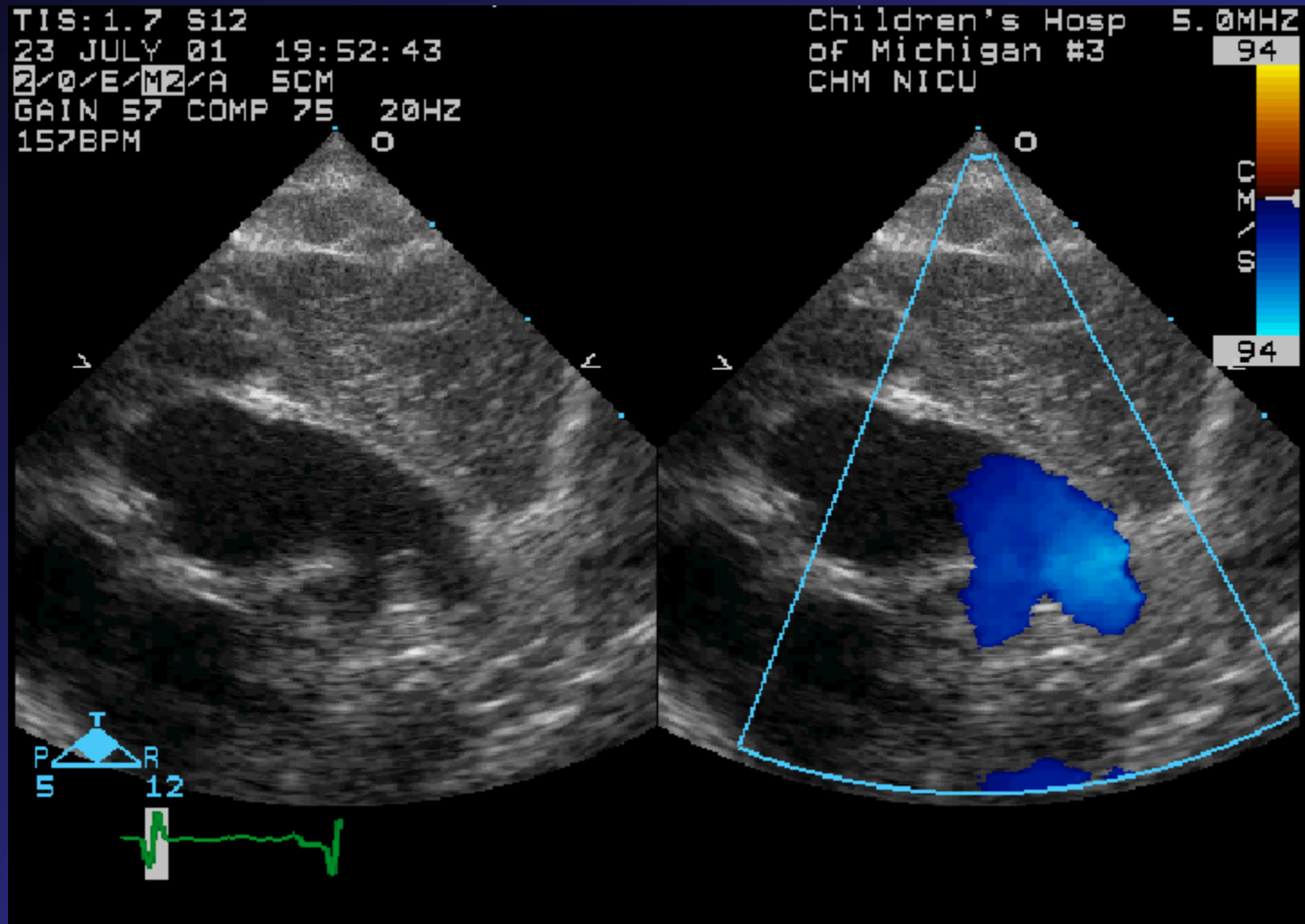
Case 10

Truncus Arteriosus



Case 10

Truncus Arteriosus



Anomalous Left Coronary Artery

From the Pulmonary Artery - ALCAPA



- Rare congenital anomaly
- Usually presents at 2-3 m of age
- Results in severe LV ischemia
- Present as dilated CM, CHF
- ECG often diagnostic
- Patients survive w/ collateral flow
- Surgery done to re-implant vessel

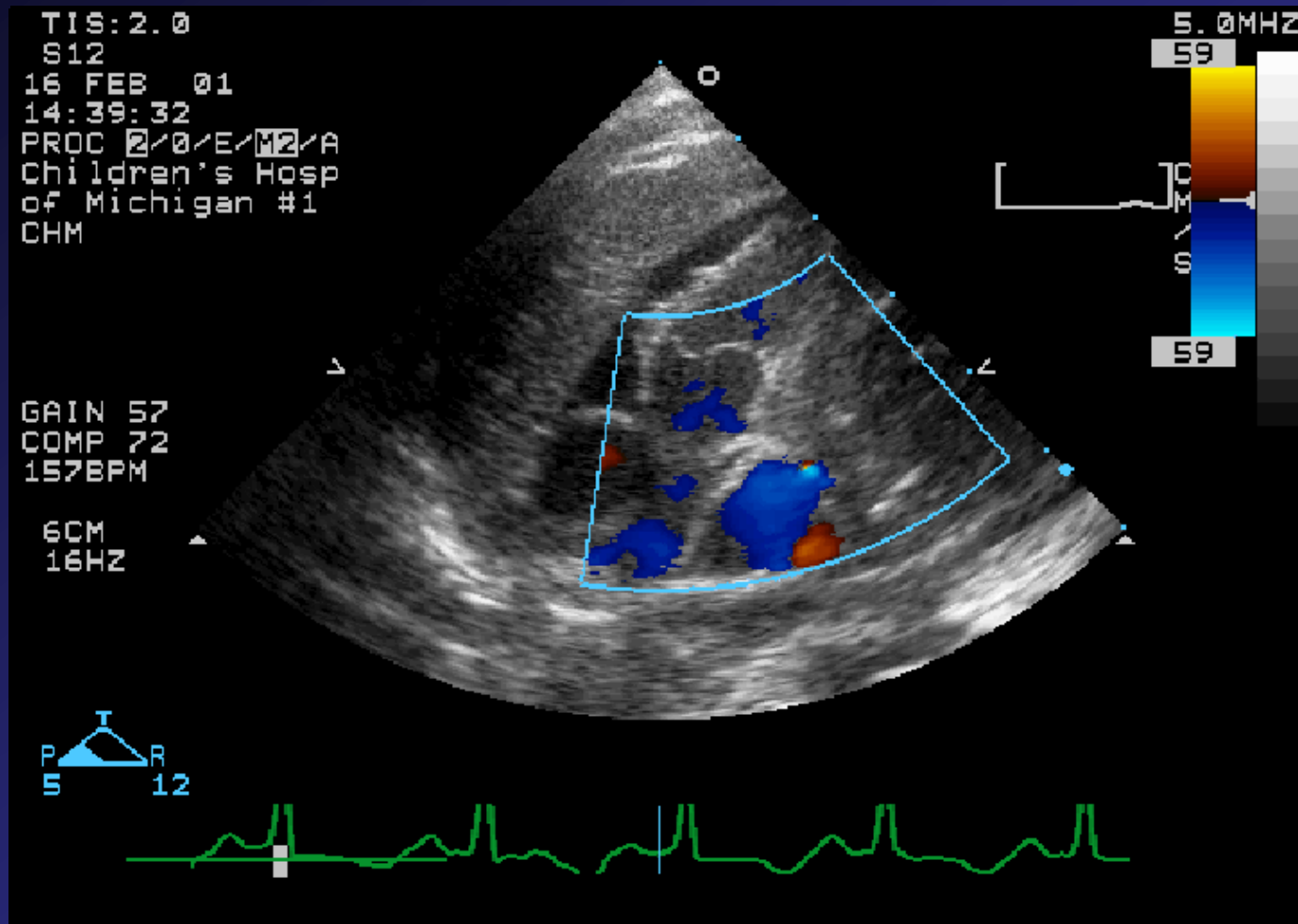
Anomalous Left Coronary Artery

Echocardiographic Clues

- Left ventricular dysfunction (usually severe)
- Mitral insufficiency – due to LV dilation/dysfunction, papillary muscle infarction
- Endocardial fibroelastosis of LV and/or papillary muscles
- Failure to identify proximal LCA from aorta
- Unusual flow into main pulmonary artery

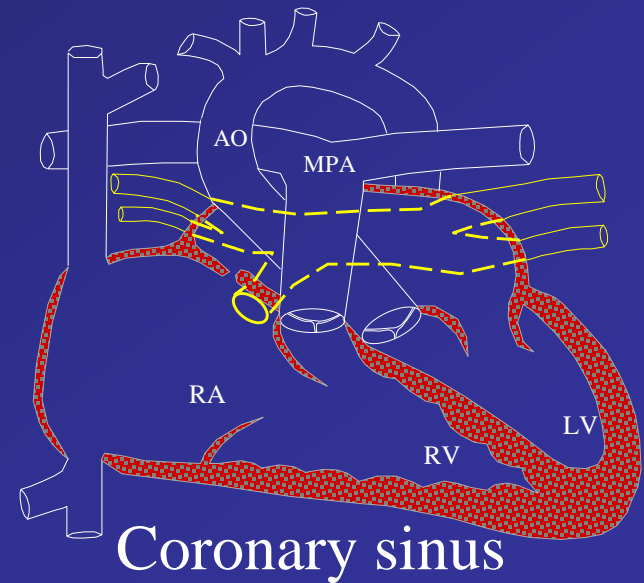
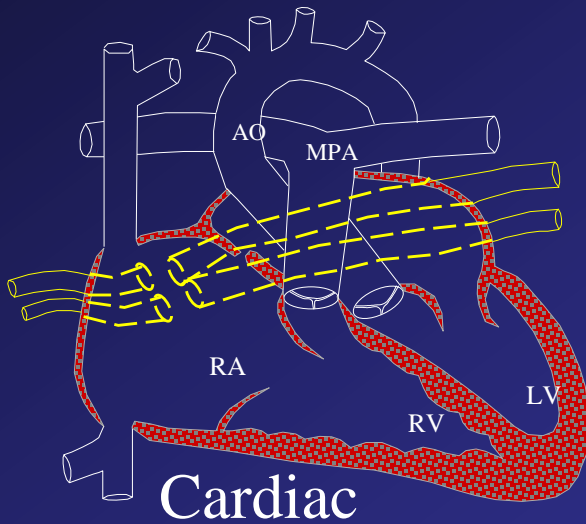
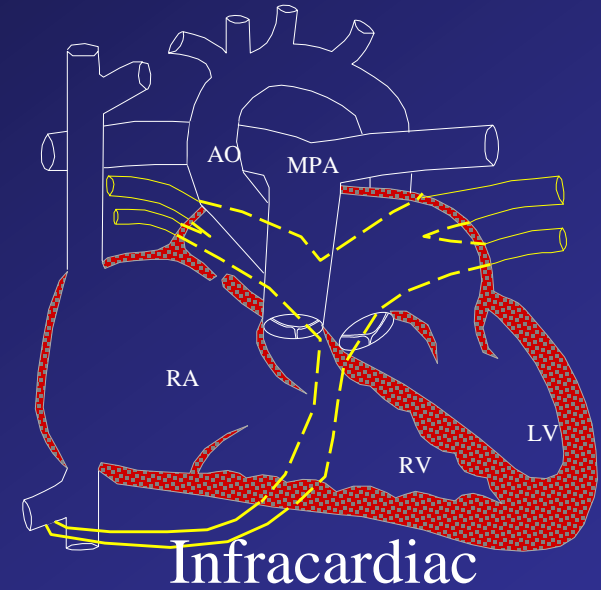
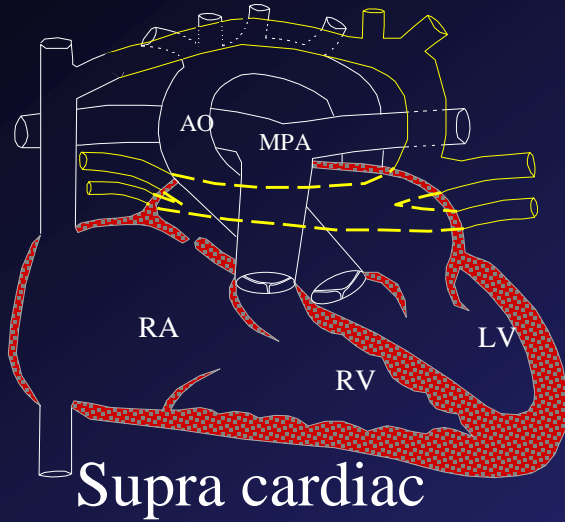
Case 11

ALCAPA



Total Anomalous Pulmonary Venous Return

Anatomic Types



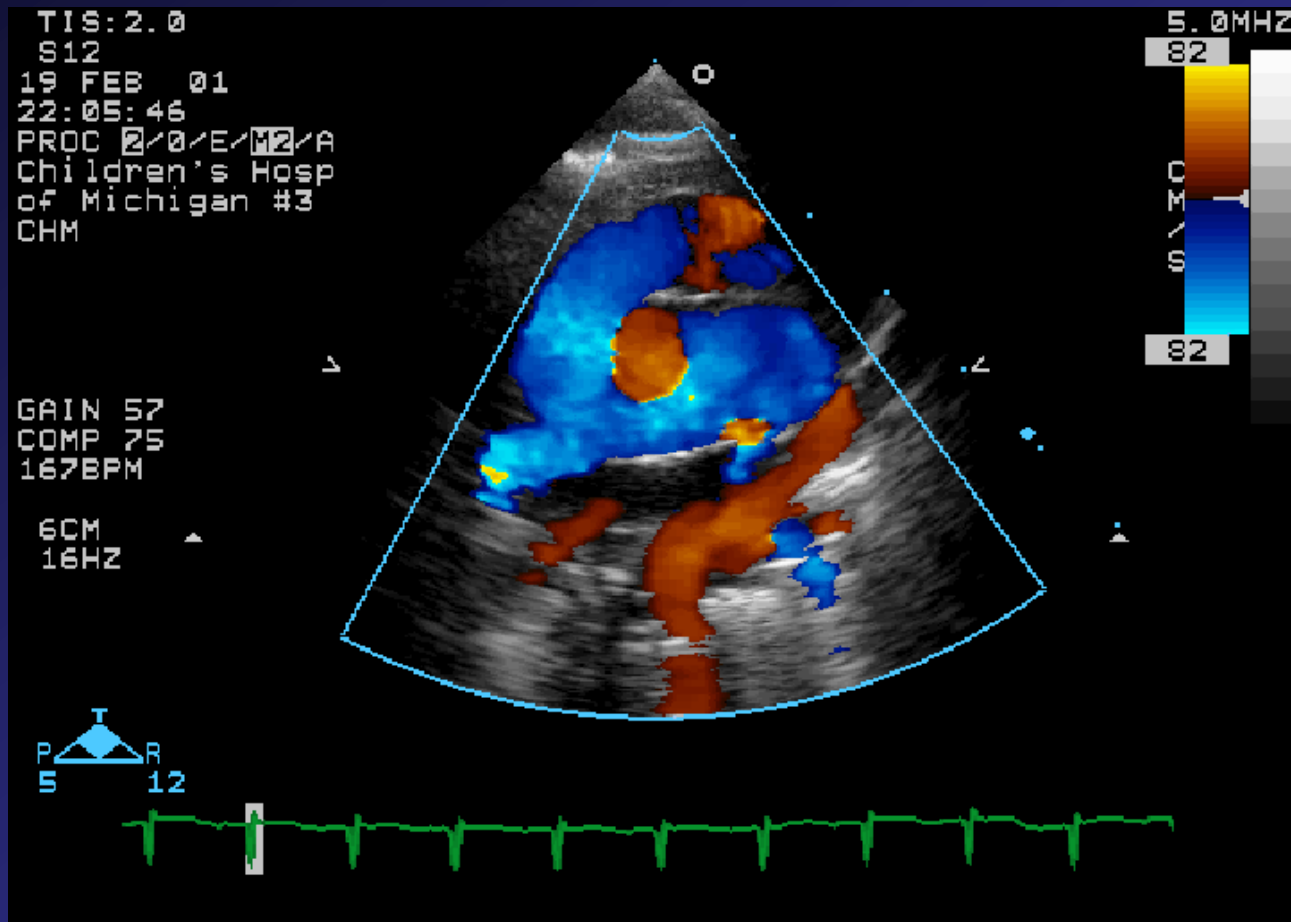
Total Anomalous Pulmonary Venous Return

Echo Clues

- Enlarged right heart
- Right to left atrial shunting
- Unusual “membranes” in left atrium
- Abnormal flow in systemic venous system
- Obstruction may occur at different levels
 - Most common - infracardiac
- May be remarkably asymptomatic (in absence of obstruction)

Case 12

Supra cardiac TAPVR



Good Luck On Your Exam



Questions?

michael_petterson@pediatrix.com