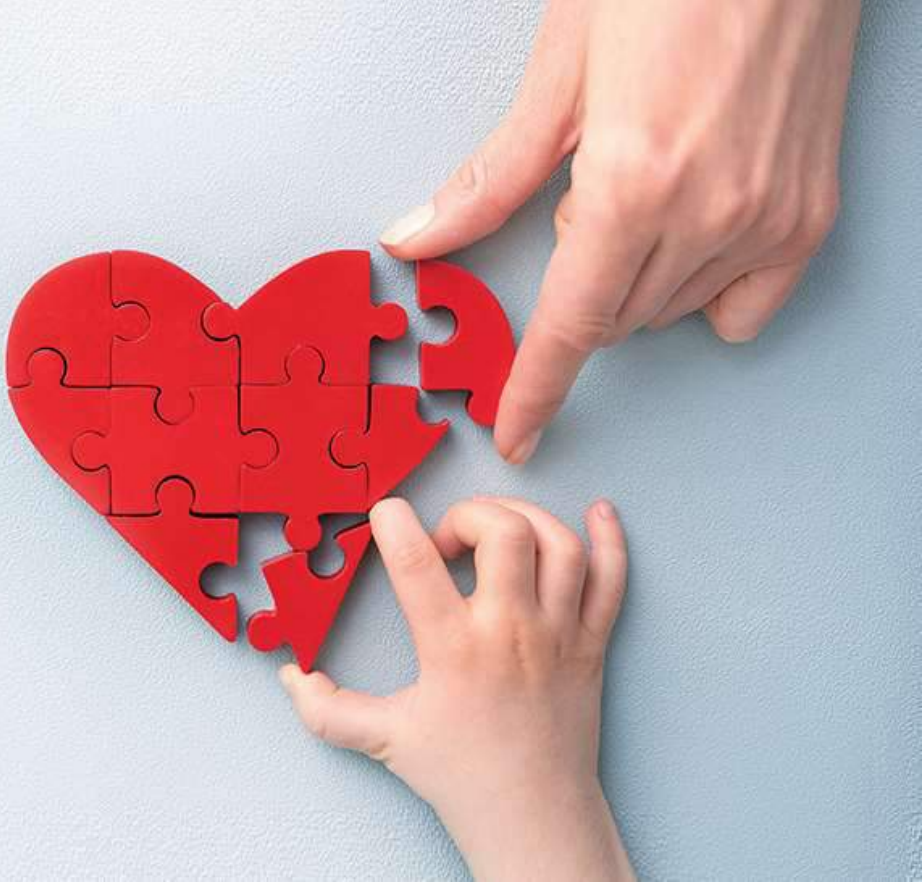
CARDIOLOGY 2024

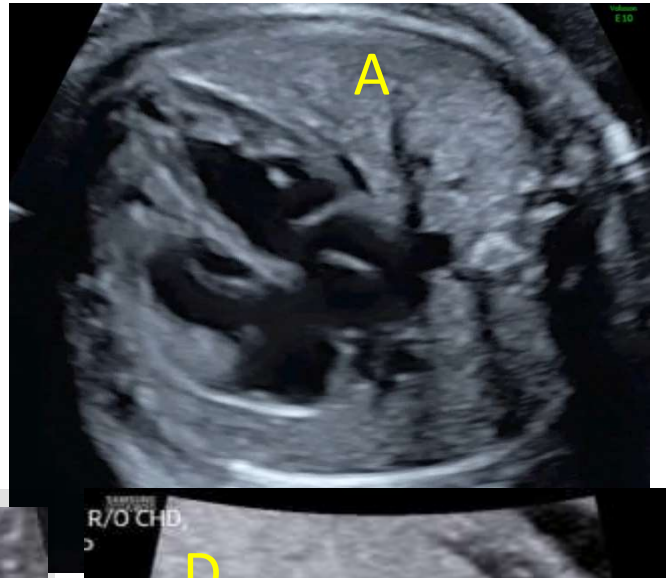
Sunrise Session :
Principles and Clinical Pearls for imaging CHD

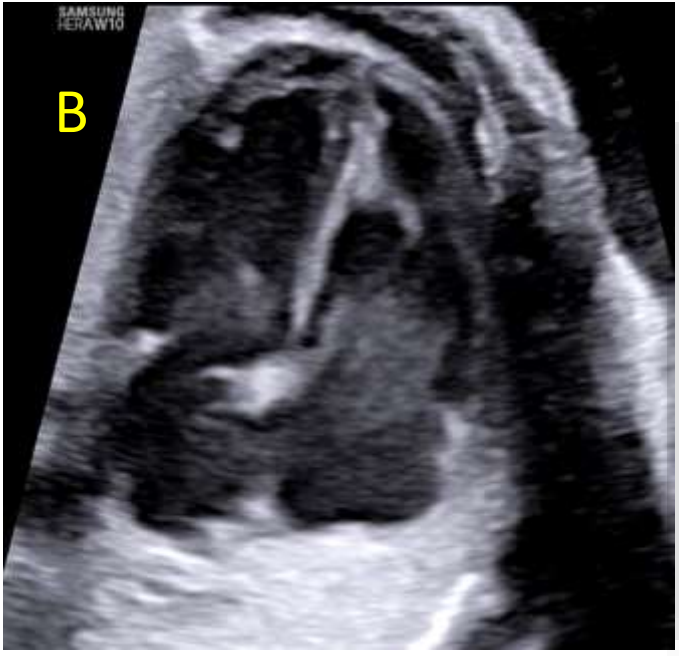


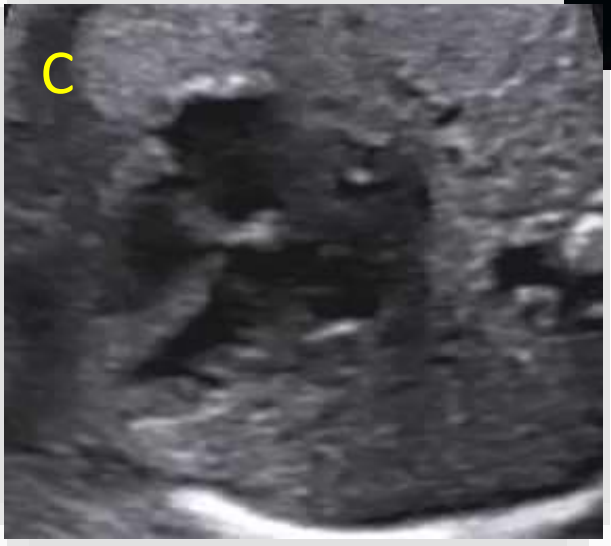
Amanda Shillingford MD
Medical Director, Community Fetal Outreach
Children's Hospital of Philadelphia
Sunday, February 18, 2024

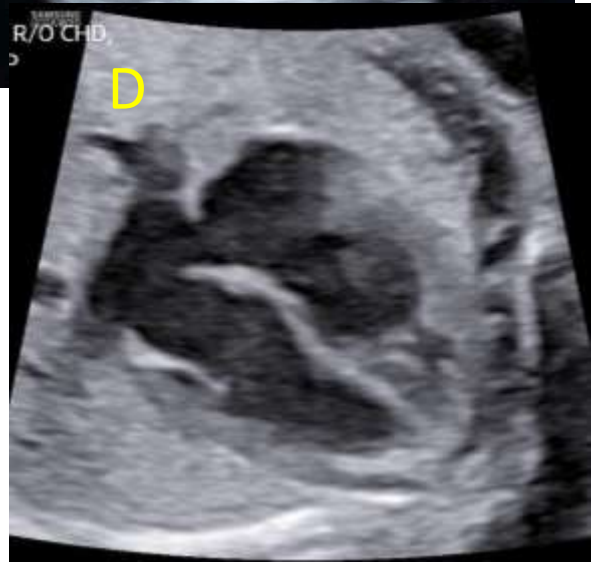
Children's Hospital of Philadelphia
Cardiac Center

Which fetus has CHD?



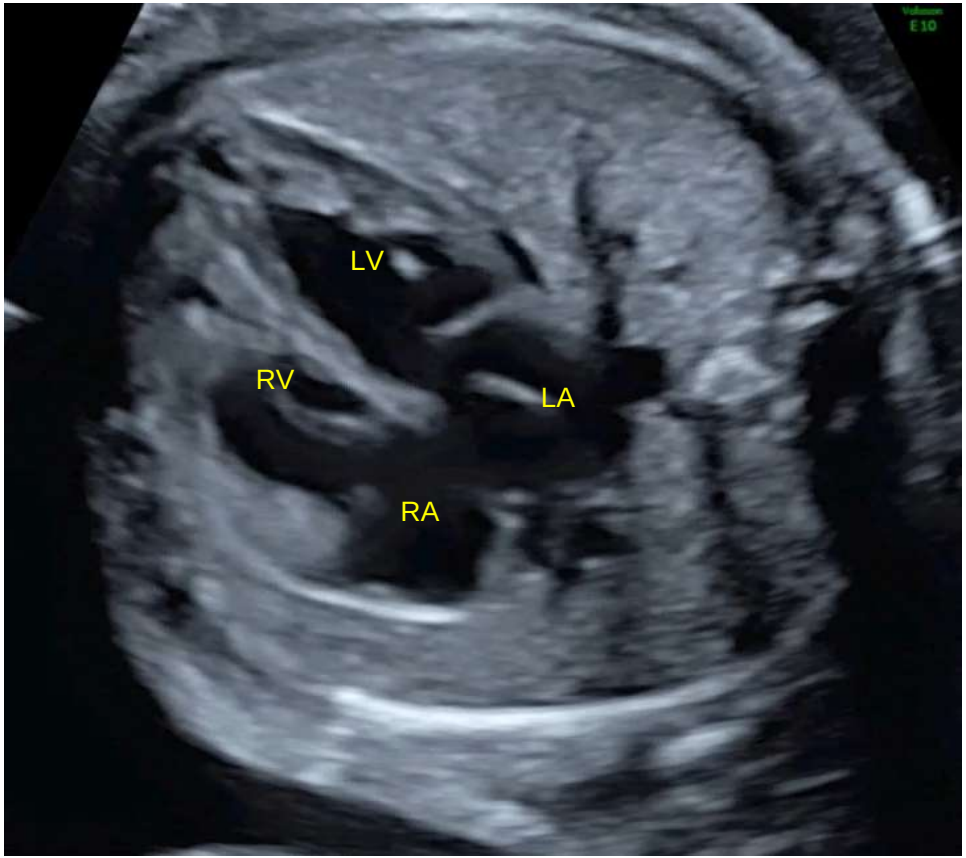


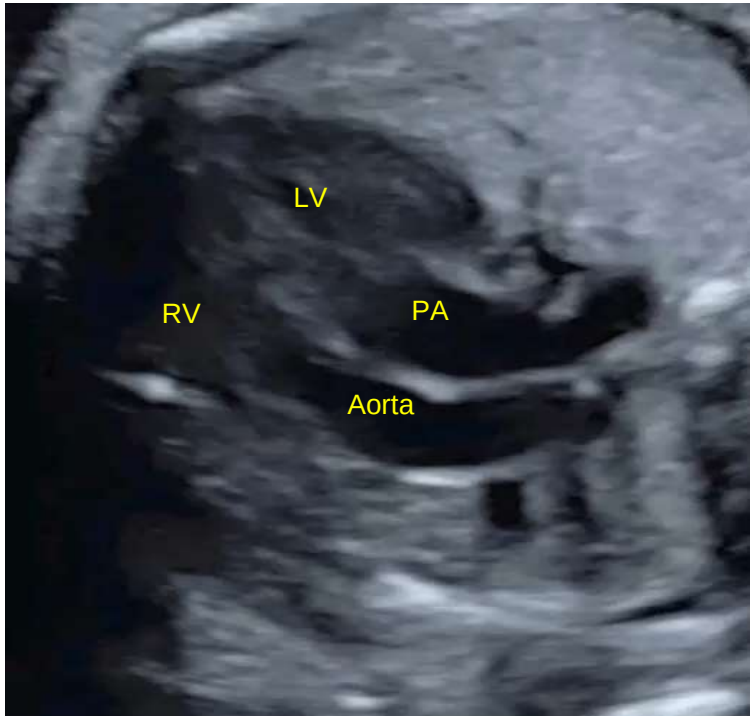




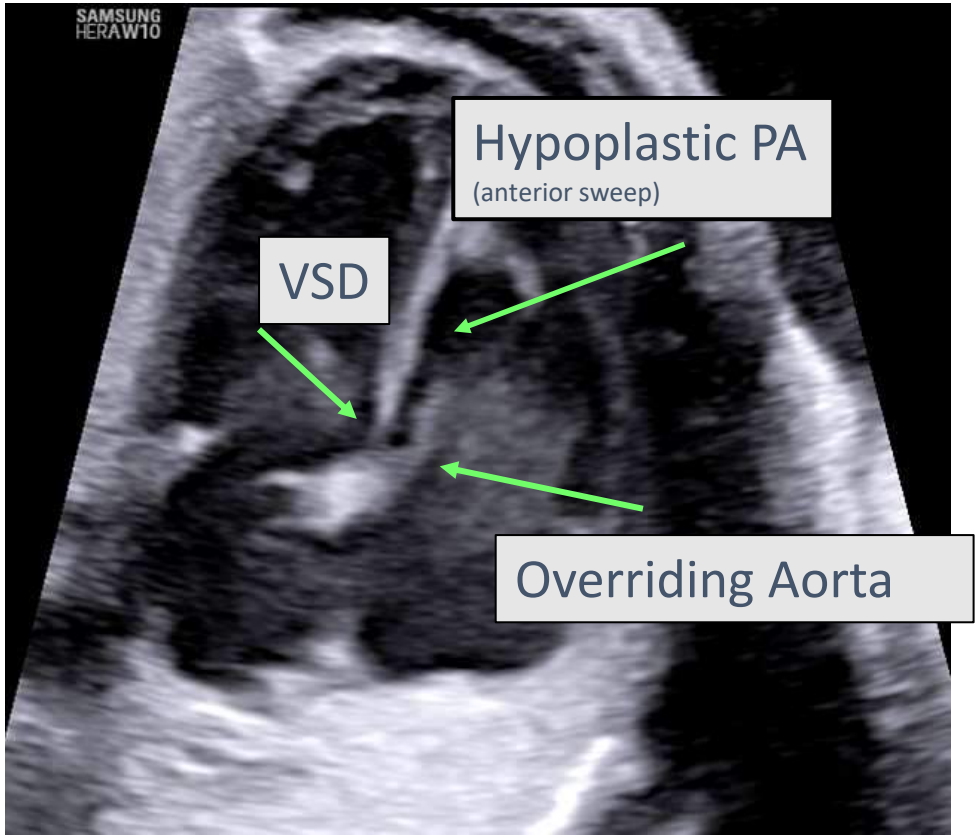
Option A

Transposition of the Great Arteries



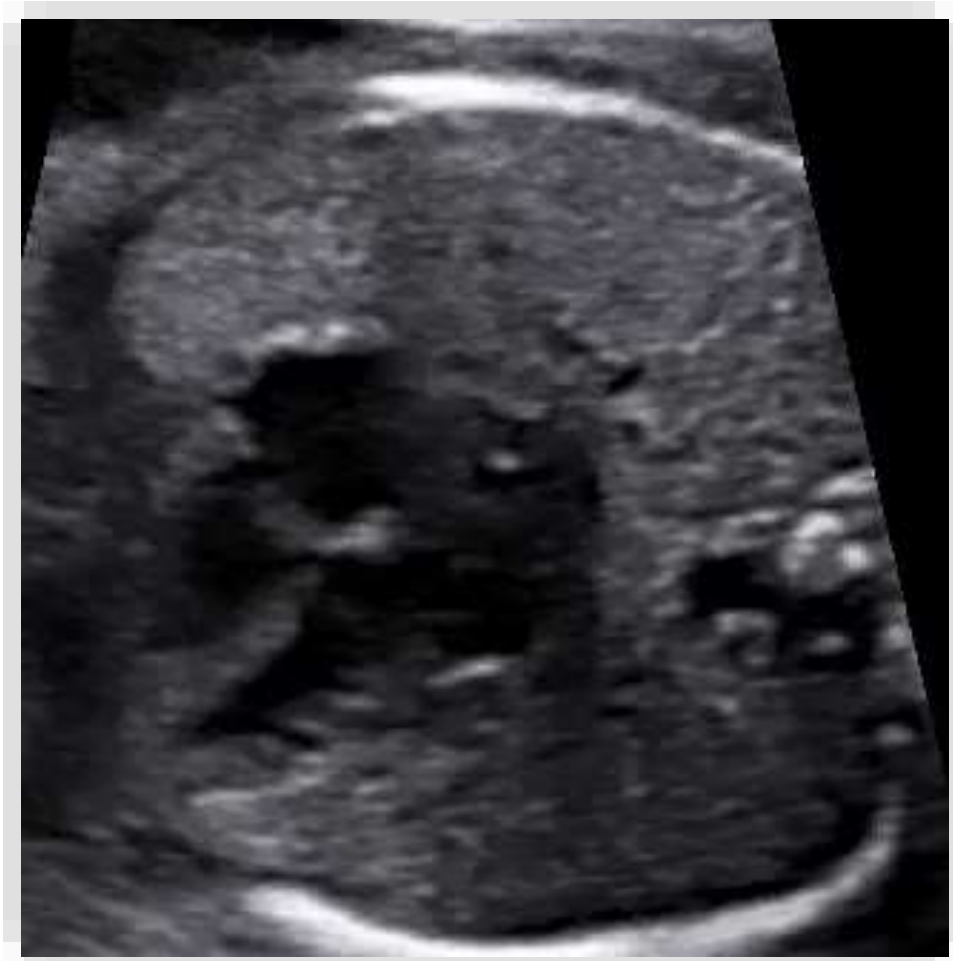


Option B

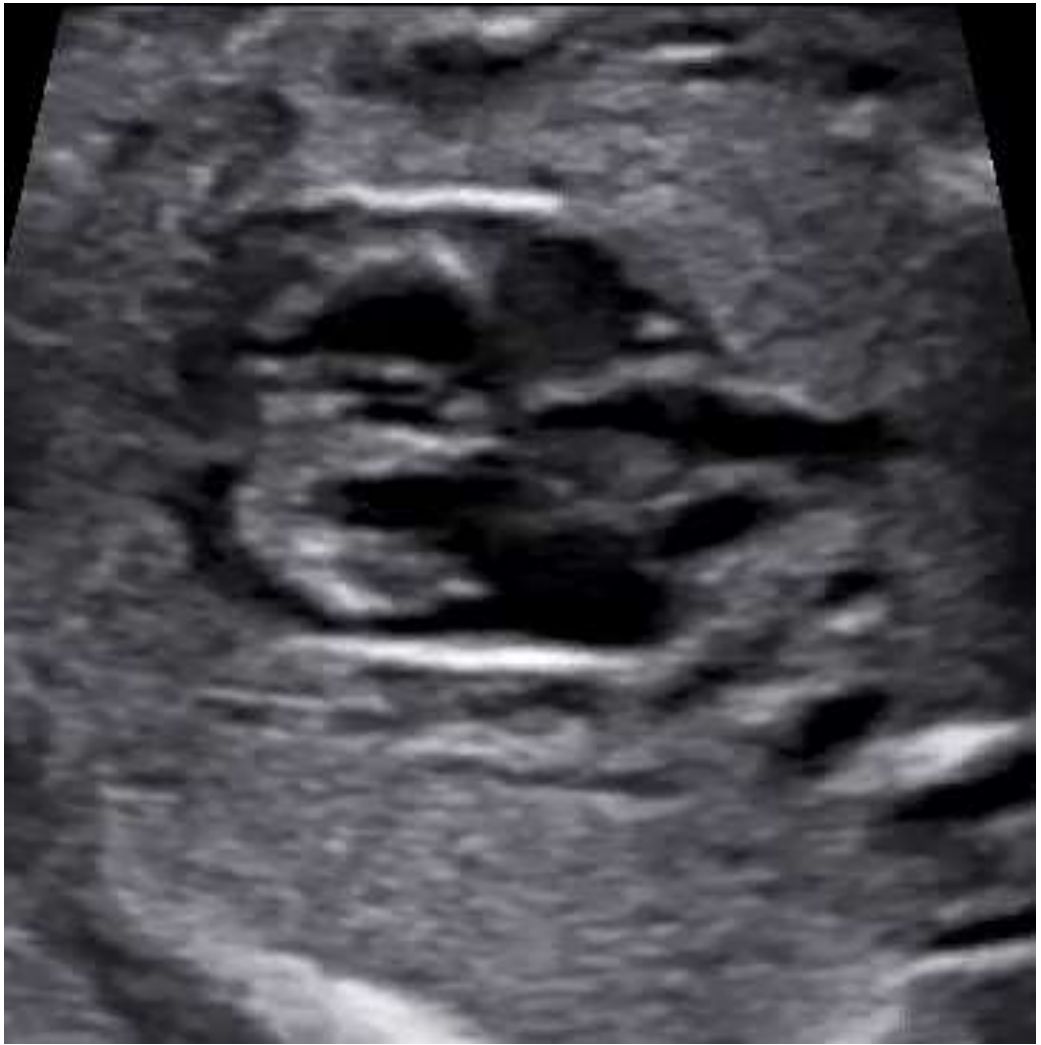


Tetralogy of Fallot

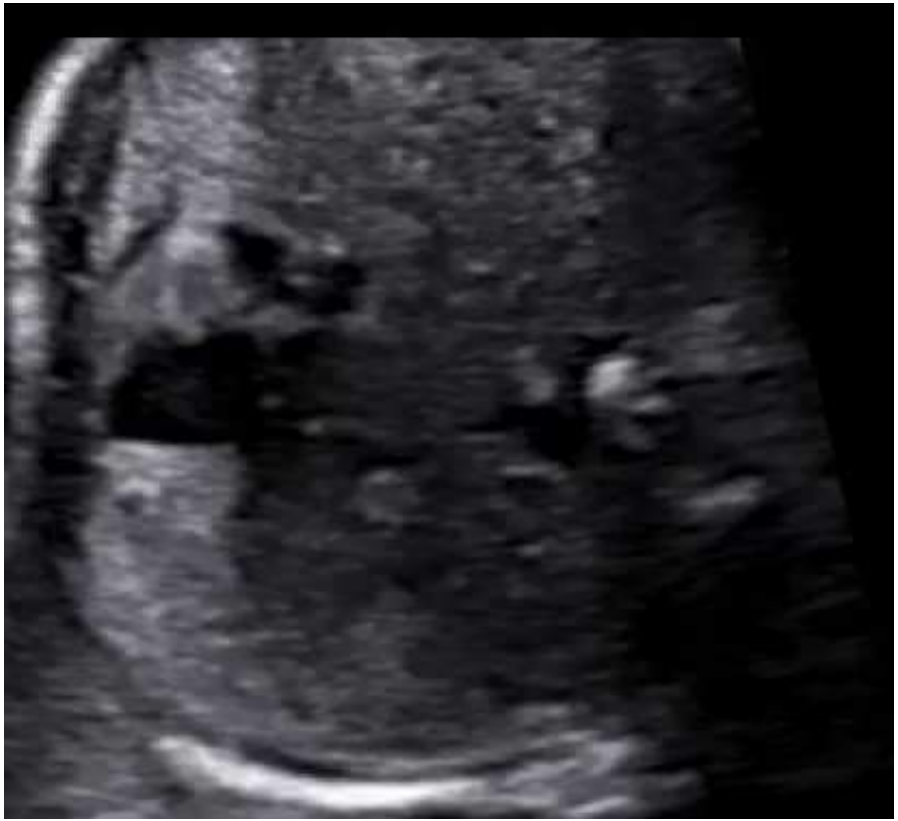
Option C

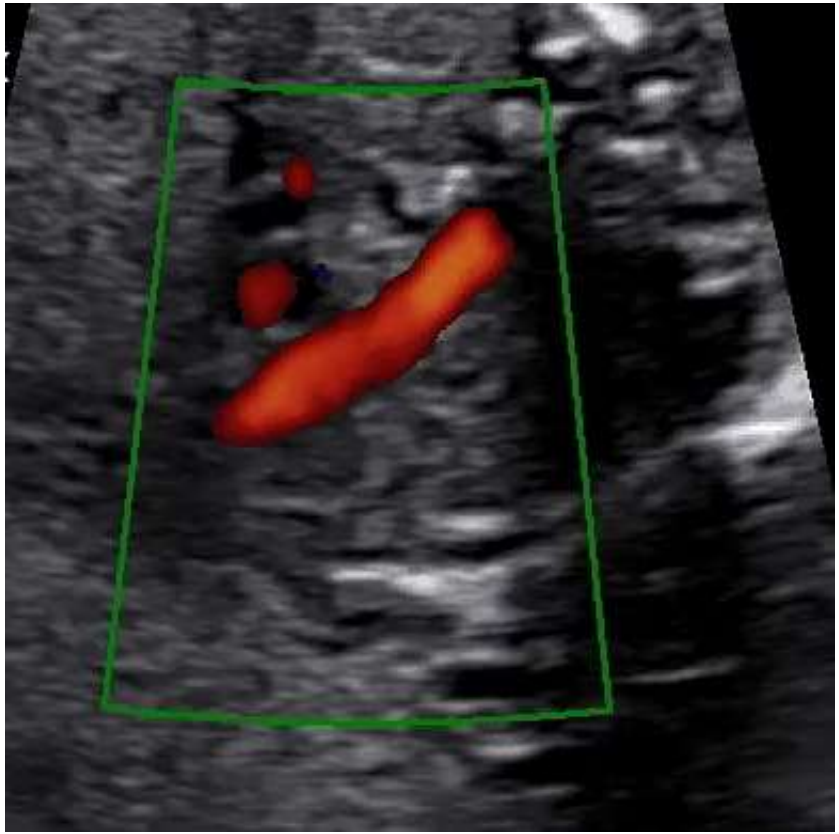


Option C



Total Anomalous Pulmonary Venous Return

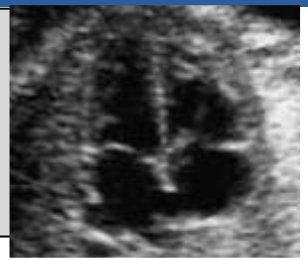




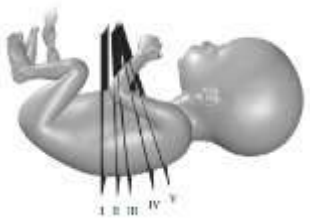
Cardiac Screening with Routine Fetal Ultrasound

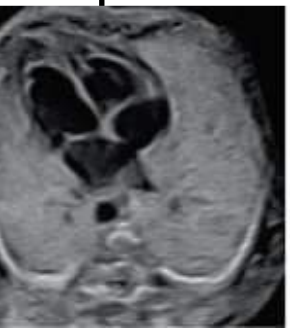
"Basic Screening"
- 4 chamber view

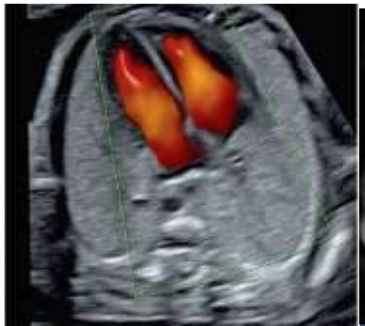
"Extended Screening"
- + outflow tracts *if possible*
- *(no Color Doppler)*

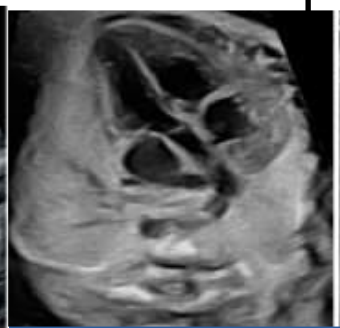


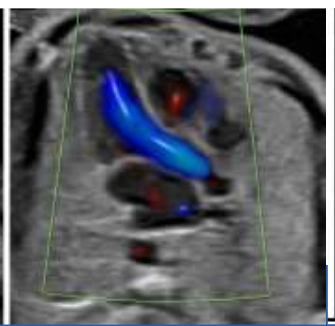
***UPDATED*:**
- Multiple axial place
- 4 chamber + outflow tracts
- *Color Doppler encouraged*

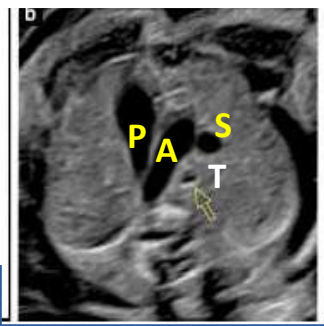


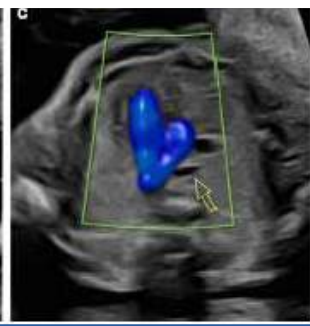








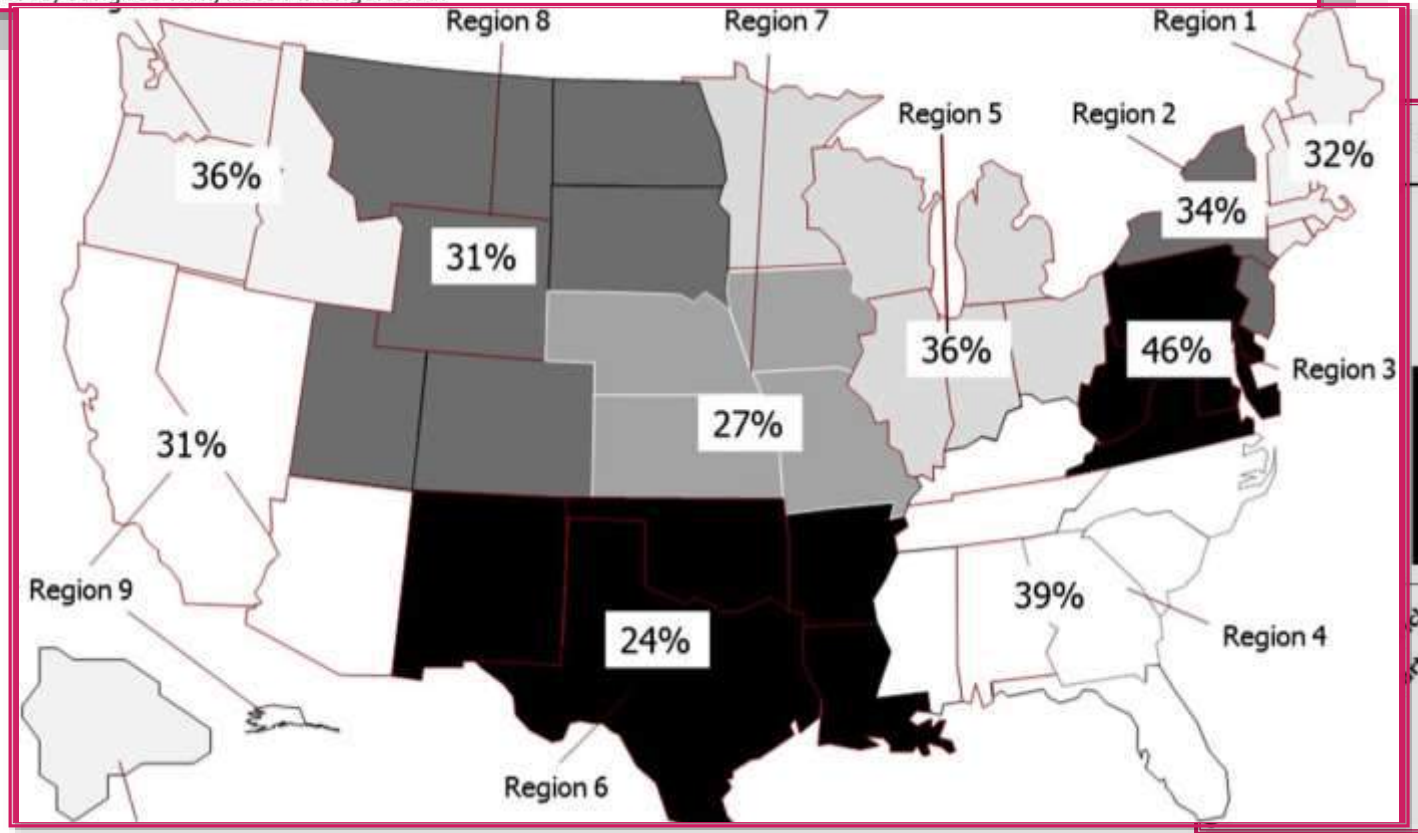


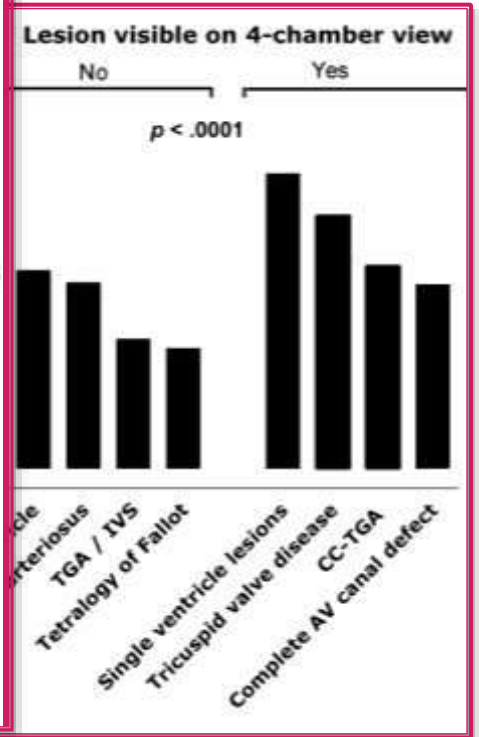


Pediatrics
August 2015, VOLUME 136 / ISSUE 2
Article

Variation in Prenatal Diagnosis of Congenital Heart Disease in Infants

Michael D. Quartermain, Sara K. Pasquali, Kevin D. Hill, David J. Goldberg, James C. Huhta, Jeffrey P. Jacobs, Marshall L. Jacobs, Sunghee Kim, Ross M. Ungerleider





Prenatal diagnosis and prevalence of critical congenital heart defects: an international retrospective cohort study

Bakker MK, *et al*. *BMJ Open* 2019;**9**:

Marian K Bakker,[1] Jorieke E H Bergman,[1] Sergey Krikov,[2] Emmanuelle Amar,[3]

Table 5 Proportion (%) of CCHD prenatally diagnosed, by CCHD type and programme, International Clearinghouse for Birth Defects Surveillance and Research (ICBDSR) Critical Congenital Heart Defects (CCHD) Prenatal Diagnosis study 2000–2014*†

ICBDSR programme by geographic region	Selected CCHD												
	HLHS	SV	PulmA	TriA/HRH	TOF	DTGA	DORV	PTA	IAA	COA	AoS	TAPVR	Overall
France-Rhone Alpes	95.2	100.0	100.0	94.6	84.1	90.2	95.2	70.6	100.0	66.3	61.3	50.0	**86.7**
Italy-Lombardy	100.0	50.0	100.0	100.0	85.7	50.0	75.0	100.0		57.1	66.7		**76.4**
Italy-Emilia Romagna	81.9	68.6	48.6	64.9	50.6	58.6	74.5	75.0	28.6	42.4	28.0	27.8	**57.6**
Italy-Tuscany	84.3	78.9	56.3	71.4	48.9	36.8	82.1	54.5	50.0	25.0	35.3	0.0	**52.3**
USA-Atlanta	77.9	76.9	60.0	70.4	50.7	43.7	63.6	61.1	50.0	34.7	33.3	16.1	**50.5**
Canada	57.8	38.3	52.5	42.0	48.6	34.1	56.0	52.3	50.0	42.4	42.4	46.4	**45.5**
Czech Republic	72.2	59.5	61.4	37.9	29.0	29.9	55.3	53.3	80.7	23.3	30.9	19.5	**40.9**
UK-Wales	88.1	77.1	48.1	66.7	36.8	37.2	57.7	71.1	27.3	17.8	11.8	17.8	**41.5**
Argentina	54.0	55.1	38.1	40.0	36.7	21.6	53.3	29.6	38.9	31.5	25.0	20.0	**38.6**
Germany-Saxony Anhalt	66.1	66.7	50.0	71.4	30.9	33.3	40.0	46.2	40.0	28.0	18.5	25.0	**38.0**
Netherlands-Northern	71.6	63.2	41.4	30.0	24.3	25.6	68.0	11.1	20.0	11.1	4.1	6.7	**31.7**
USA-Arkansas	42.1	32.3	14.8	38.9	14.7	10.4	25.6	36.0	14.3	5.4	2.6	5.1	**17.5**
Malta	25.0	25.0	16.7	25.0	12.5	13.0	33.3	0.0	0.0	0.0	0.0	0.0	**13.5**
Slovak Republic	24.3	10.0	3.2	11.8	6.5	5.8	18.8	21.0	9.1	13.0	5.8	0.0	**13.2**

Case:

- 39yo G2P1 presenting at 20 4/7 weeks CFDT evaluation of suspected enlarged posterior fossa and Dandy Walker
- Fetal echo performed as part of diagnostic work-up
- Mother with sickle cell disease, h/o first trimester bleeding
- No known Family History of CHD
- Male fetus, appropriate growth

No obvious

2023 ASE GUIDELINES: JASE VOL 30

- Pre-gestational DM
- Family Hx of CHD: 1st degree
  - *risk can vary with type of CHD*
- SSA/SSB antibodies
- Medications
- IVF

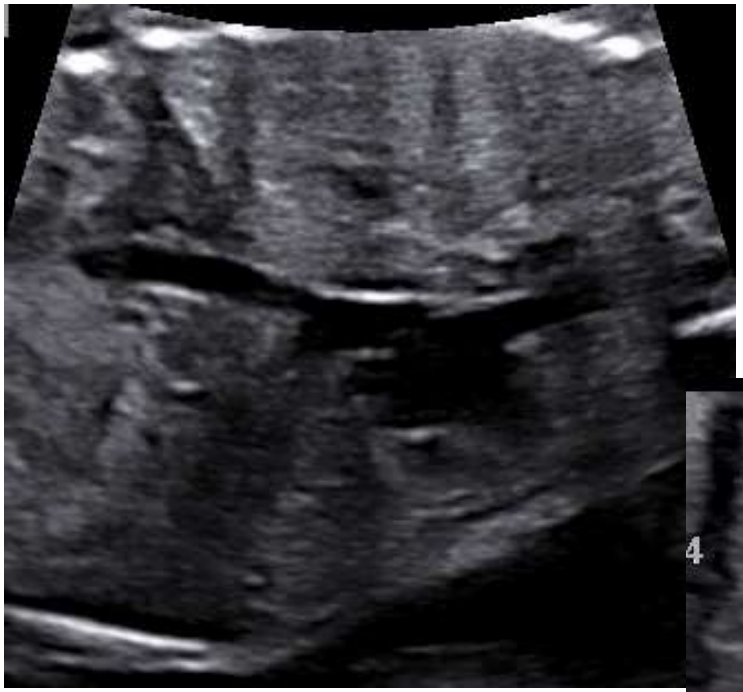
- Fetal Hydrops
- Extracardiac Abnormality
- Chromosome Abnormality
- Monochorionic Twinning
- Increased NT

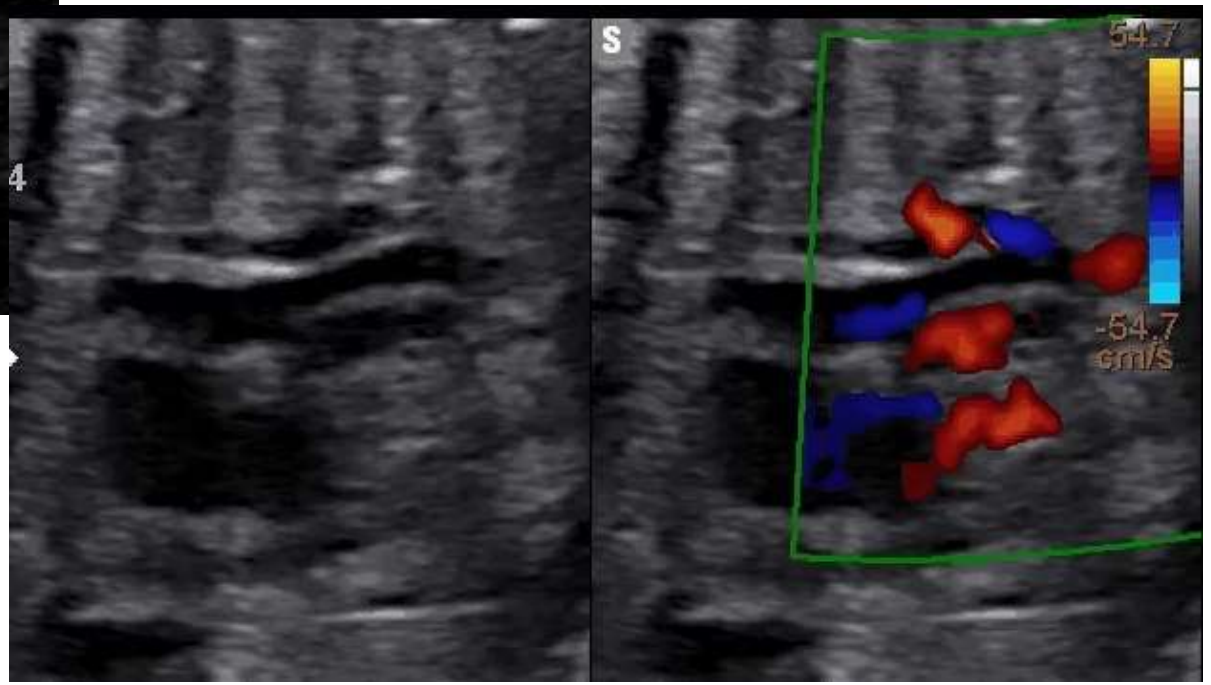
Table 1 Potential indications for fetal echocardiography

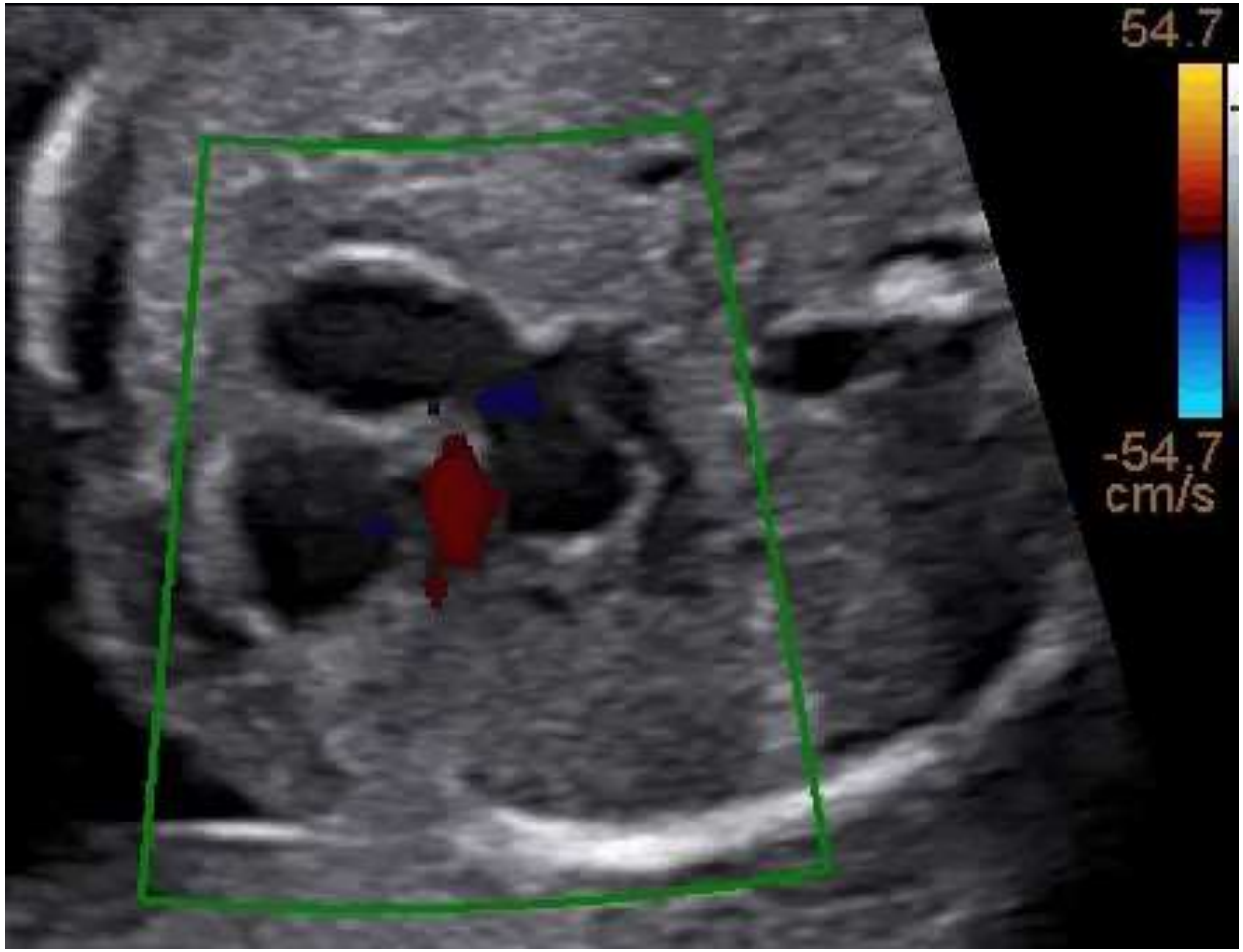
	ASE 2023 recommendation	AIUM 2020[4]	AHA 2014[2•]
Maternal factors (absolute risk)[1]			
Pre–gestational diabetes (3%-5%)	Is indicated	Is indicated	I (indicated)
Gestational diabetes diagnosed after second trimester (<1%)	Not indicated	Not indicated	III (no benefit)
Phenylketonuria (12%-14%)	Is indicated	Is indicated	I (indicated)
Autoimmune disease: SSA/SSB positive (1%-5%)[‡]	Is indicated	Is indicated	IIa (probably indicated)
In vitro fertilization (1.1%-3.3%)	May be considered[§]	Is indicated	IIa (Probably indicated)
Maternal infection: rubella (3%-4%)	Is indicated	Is indicated	I (indicated)
Family history of CHD: first-degree relative (3%-20%)[¶]	Is indicated	Is indicated	I (indicated)
Family history of CHD: second-degree or more distant relative (<2%)[‖]	Not indicated	May be indicated	IIb (may be indicated)
Obesity (BMI > 30 kg/m^2) (1-2%)	Not indicated	Not indicated	—
Retinoids (8%-20%)	Is indicated	Is indicated	I (indicated)
ACE inhibitors (3%)	May be considered[§]	May be indicated	IIa (probably indicated)
Paroxetine (3%)	May be considered[§]	May be indicated	IIb (may be indicated)
Other selective serotonin reuptake inhibitors (1%-2%)[6,7]	Not indicated	Not indicated	III (no benefit)
Anticonvulsants (1%-2%)	Not indicated	May be indicated	IIb (may be indicated)
Lithium (1%-2%)	Not indicated	May be indicated	IIb (may be indicated)
Warfarin (<1%)[8]	Not indicated	Not indicated	III (no benefit)
Fetal factors identified during screening (absolute risk)			
Fetal hydrops (15%-20%)[9]	Is indicated	Is indicated	I (indicated)
Extracardiac anomaly (20%-45%)[10,11]	Is indicated	Is indicated	I (indicated)
Chromosomal abnormalities (10%-90%)	Is indicated	Is indicated	I (indicated)
Monochorionic twinning (2%-10%)	Is indicated	Is indicated	I (indicated)
Nuchal translucency 3.0-3.4 mm (~3%)	May be considered[§]	May be indicated	IIa (probably indicated)
Nuchal translucency ≥3.5 mm (6%-60%)	Is indicated	Is indicated	I (indicated)
Single umbilical artery in isolation (1.2%-1.8%)[12]	Not indicated	Not indicated	IIb (may be indicated)

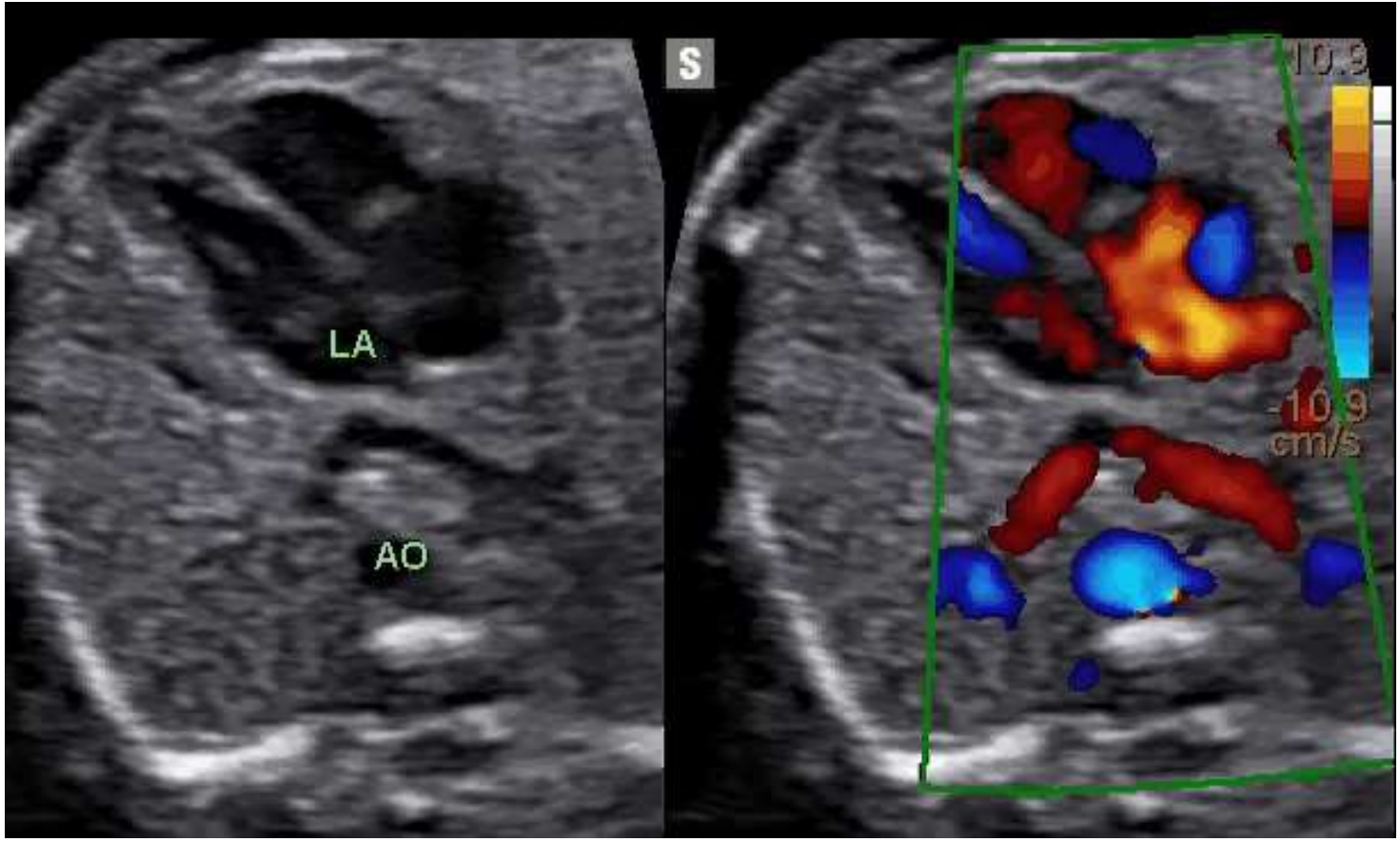
CARDIOLOGY 2024

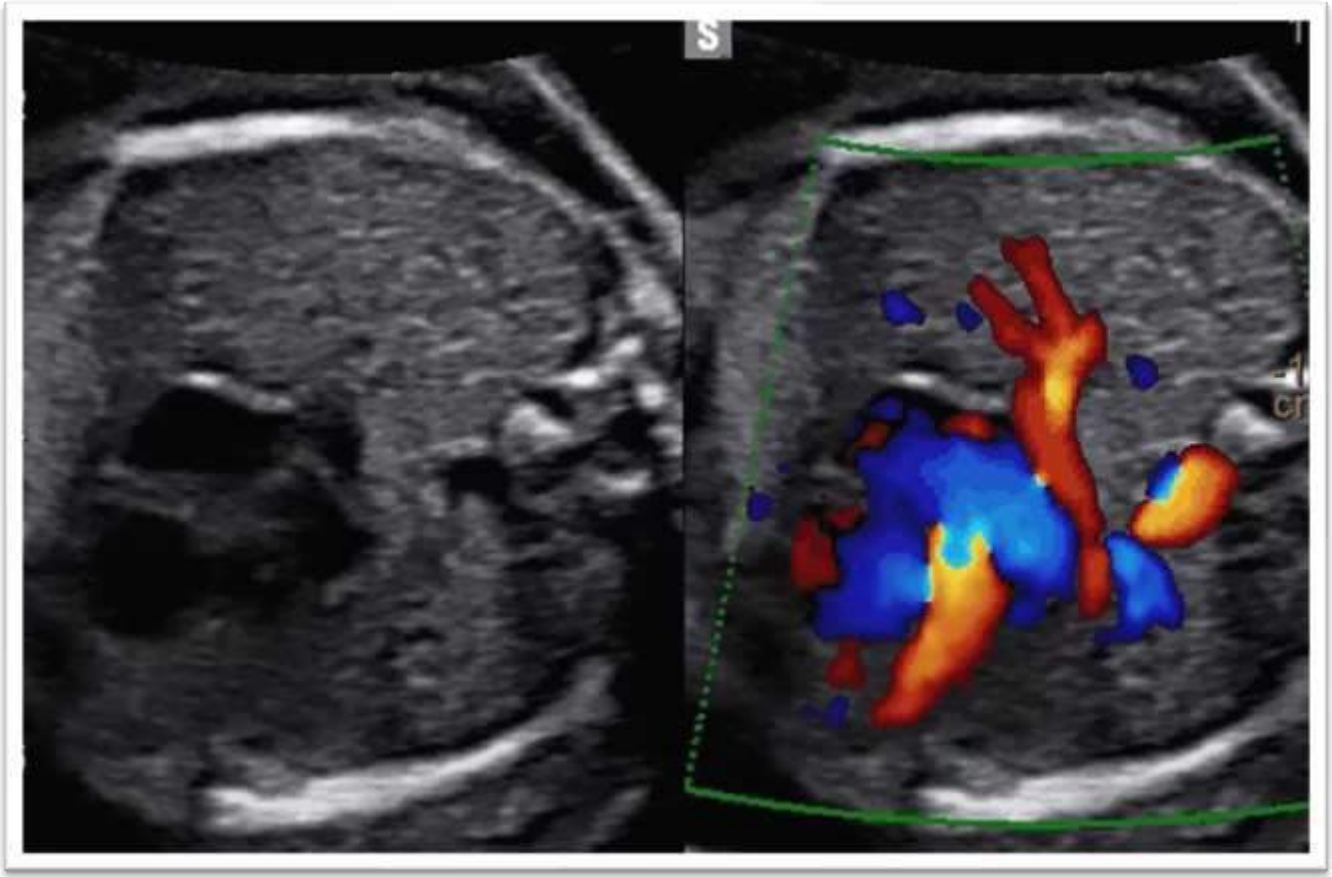
- Central nervous system defects
  - -hydrocephalus --->> 5-15%
  - -agenesis of corpus callosum--->> 15%
- Mediastinal/respiratory defects
  - -TEF/esophageal atresia--->> 15-40%
- Gastrointestinal defects
  - -omphalocele--->> 19-32%
  - -diaphragmatic hernia--->> 10-25%
- Genitourinary defects
  - -renal agenesis--->> 20-40%
  - -horseshoe kidney--->. 40%
- Skeletal defects
  - -arthrogryposis--->> 10-20%
  - -Holt-Oram-->> 75%

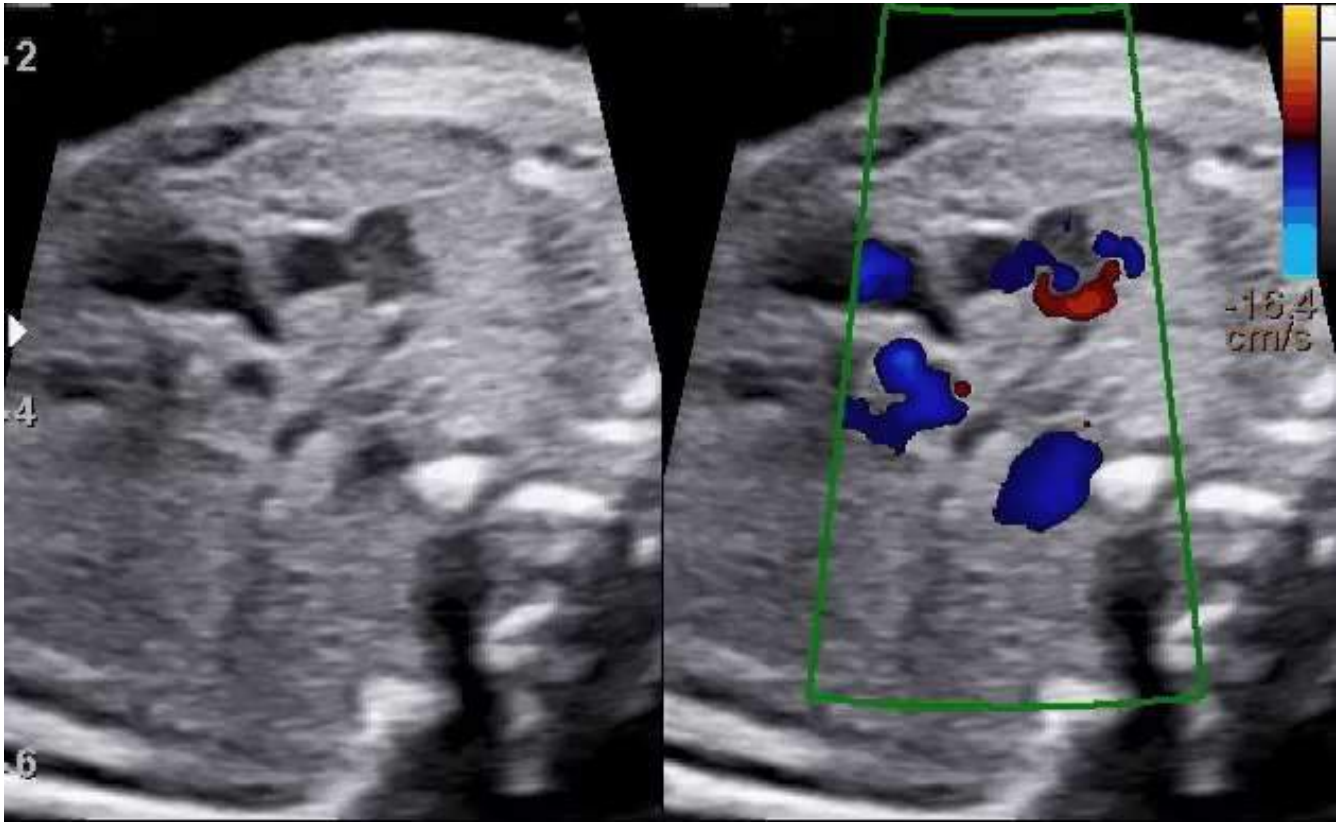



4
S
54.7
-54.7
cm/s

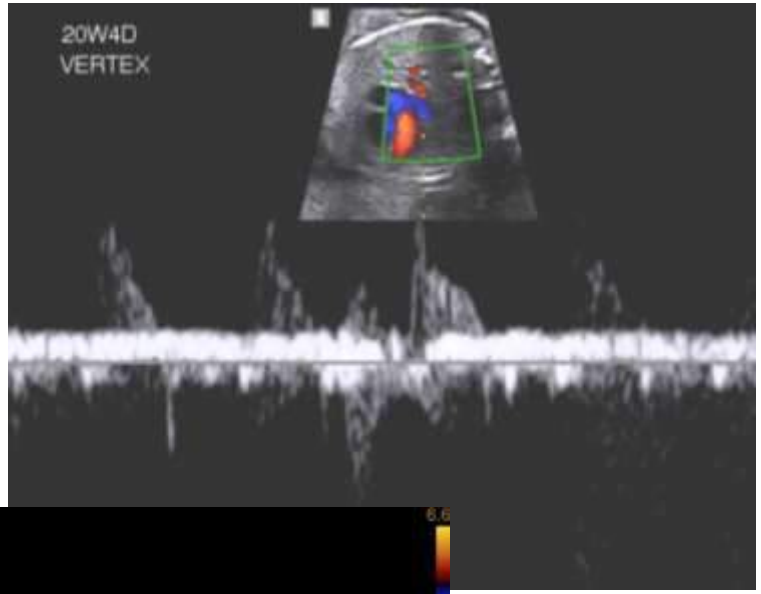

54.7
-54.7
cm/s


S
LA
AO
10.9
-10.9
cm/s

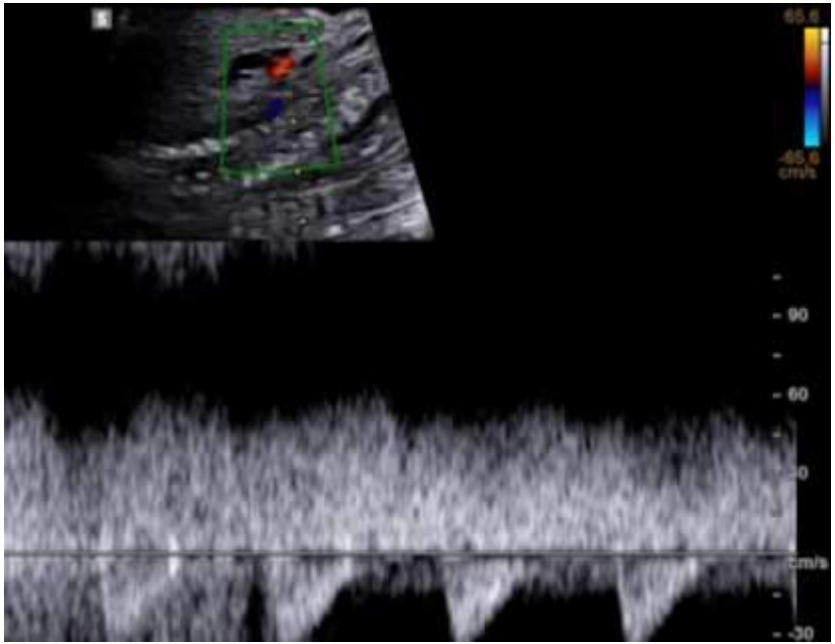


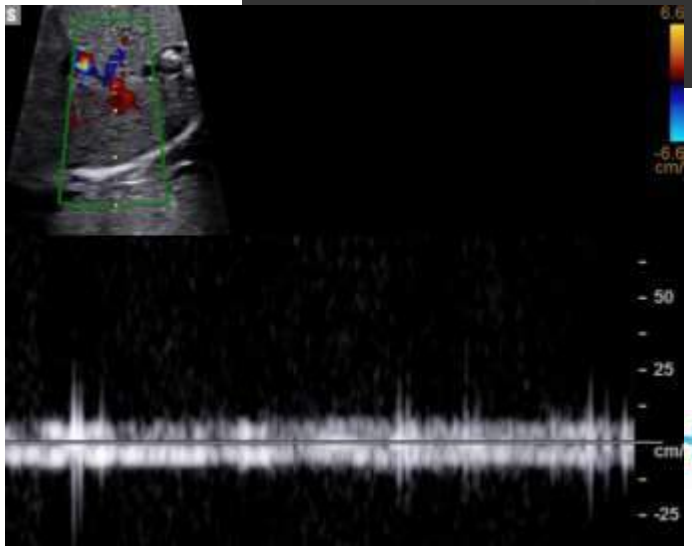

2
4
6
-16.4
cm/s

ABNORMAL DOPPLER



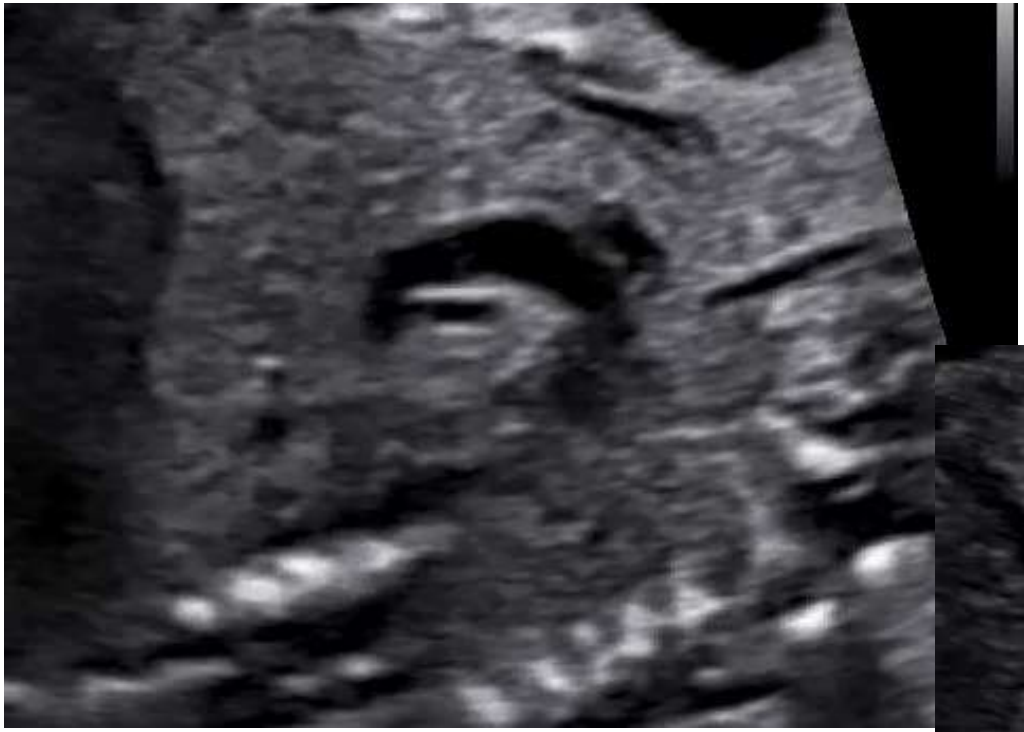
HIGH VELOCITY
MINIMAL PHASIC CHANGES

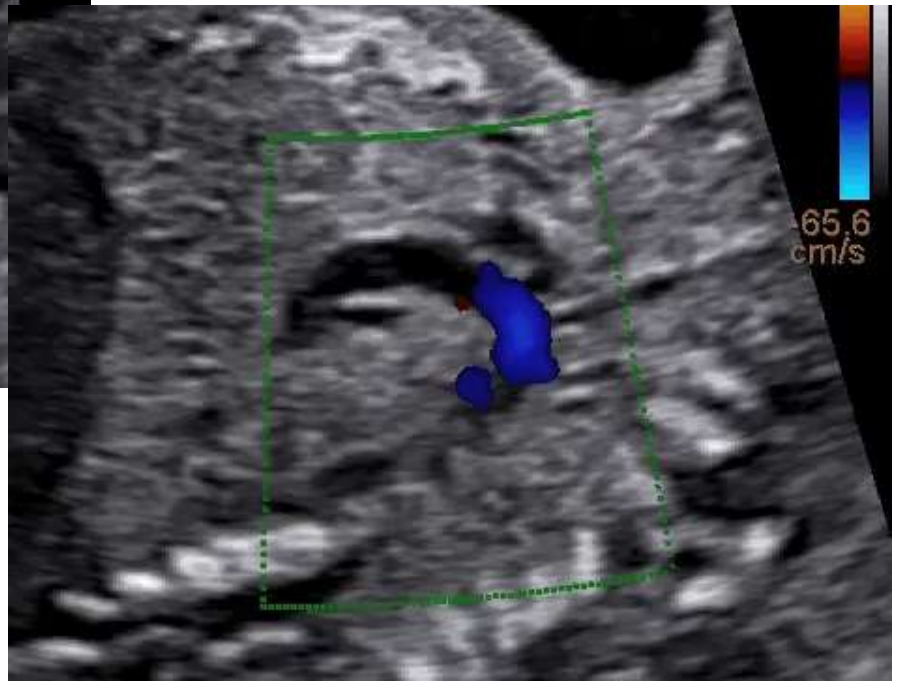


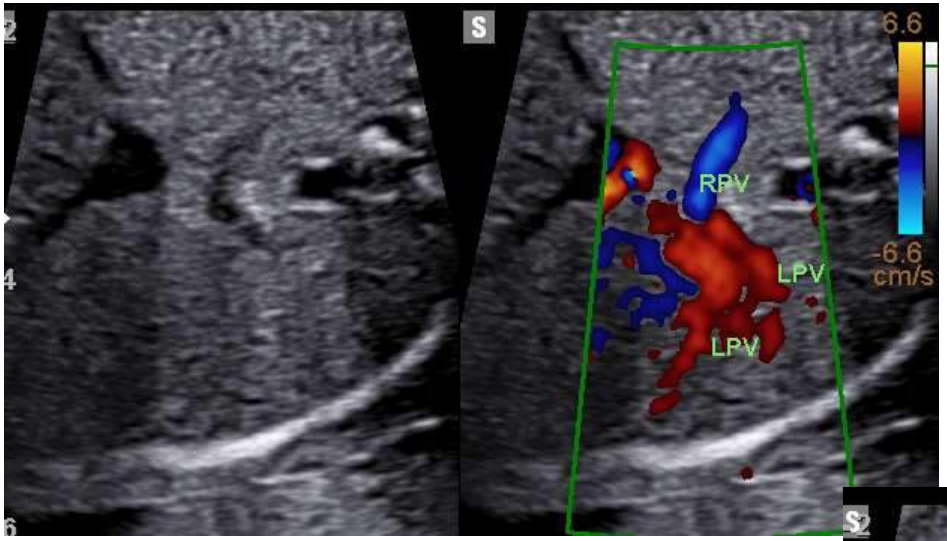


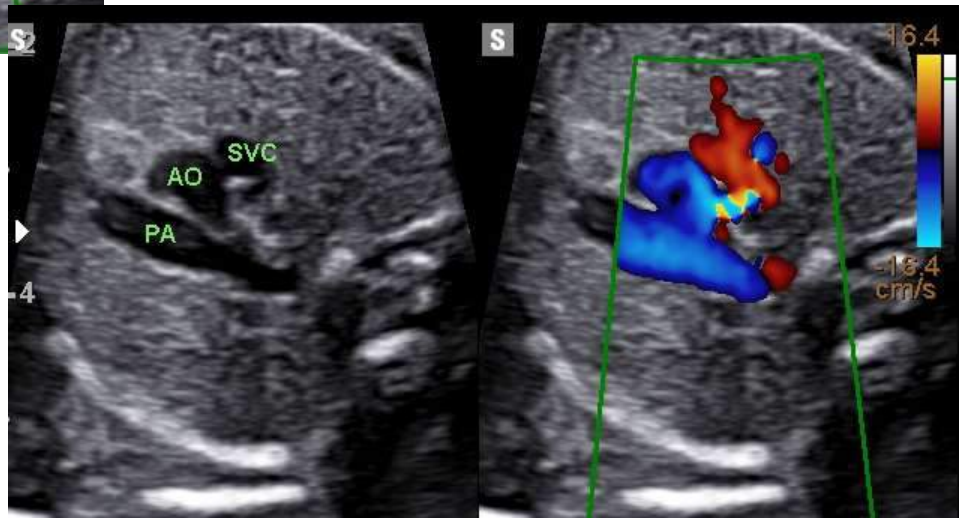
LOW VELOCITY, NONPHASIC

Children's Hospital of Philadelphia
Cardiac Center

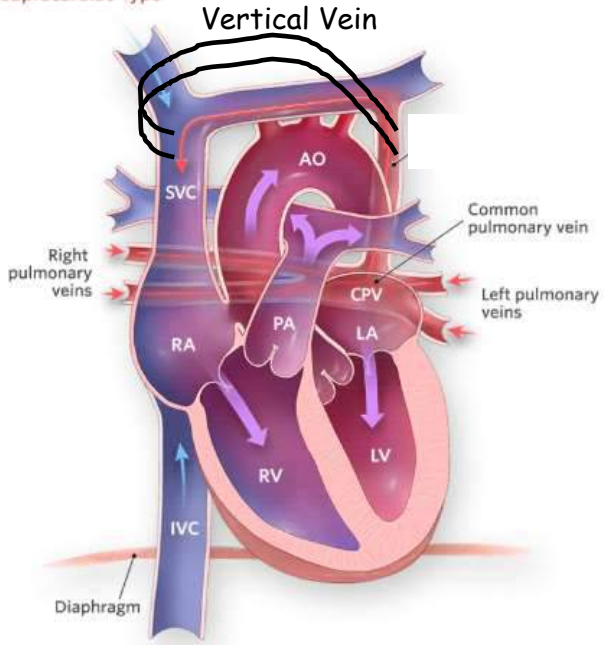



-65.6
cm/s


S
6.6
RPV
LPV
LPV
-6.6
cm/s


S
SVC
AO
PA
S
16.4
-16.4
cm/s

Total Anomalous Pulmonary Venous Return (TAPVR)
Supracardiac Type



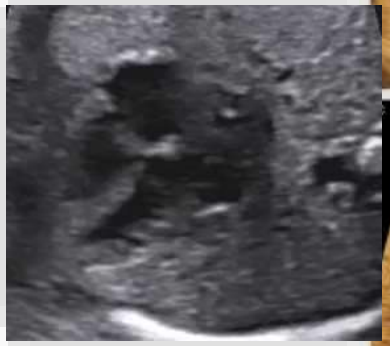
- Oxygen-rich blood
- Oxygen-poor blood
- Mixed blood
- Mixed blood

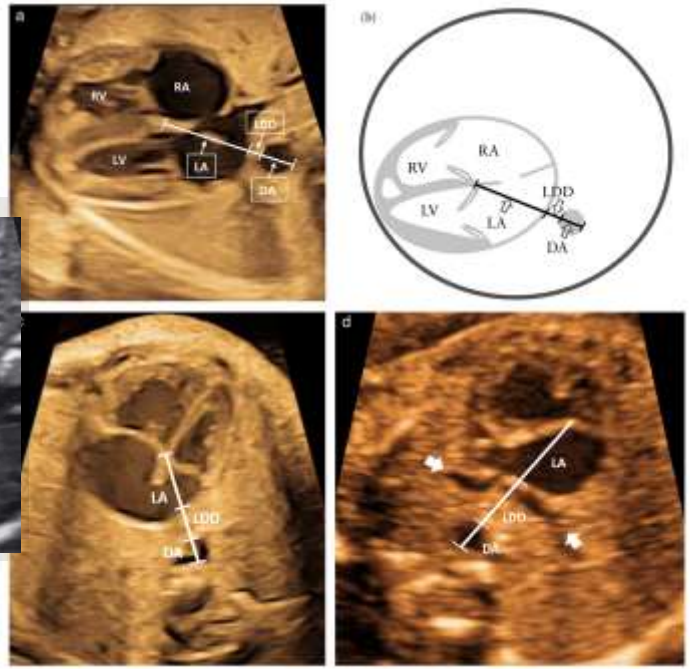
AO: Aorta
PA: Pulmonary artery
LA: Left atrium
LV: Left ventricle
RA: Right atrium
RV: Right ventricle

Ultrasound Obstet Gynecol 2023; **61**: 488–496
Published online 28 February 2023 in Wiley Online Library (wileyonlinelibrary.com). **DOI:** 10.1002/uog.26072

Fetal left-atrial posterior-space-to-diagonal ratio at 17–37 weeks' gestation for prediction of total anomalous pulmonary venous connection

S. ANUWUTNAVIN[1], V. UNALOME[1], T. REKHAWASIN[1], F. TONGPRASERT[2] and P. THONGKLOUNG[1]





- Left-atrial posterior-space-to-diagonal (LAPSD)
  - ≥ 0.35
- post-left atrium space index (PLAS)
  - ≥1

Echo "clues"

- RV/LV size discrepancy
- Dilated SVC or IVC, hep veins
- Lack of PV to LA connection
- Confluence Visualized

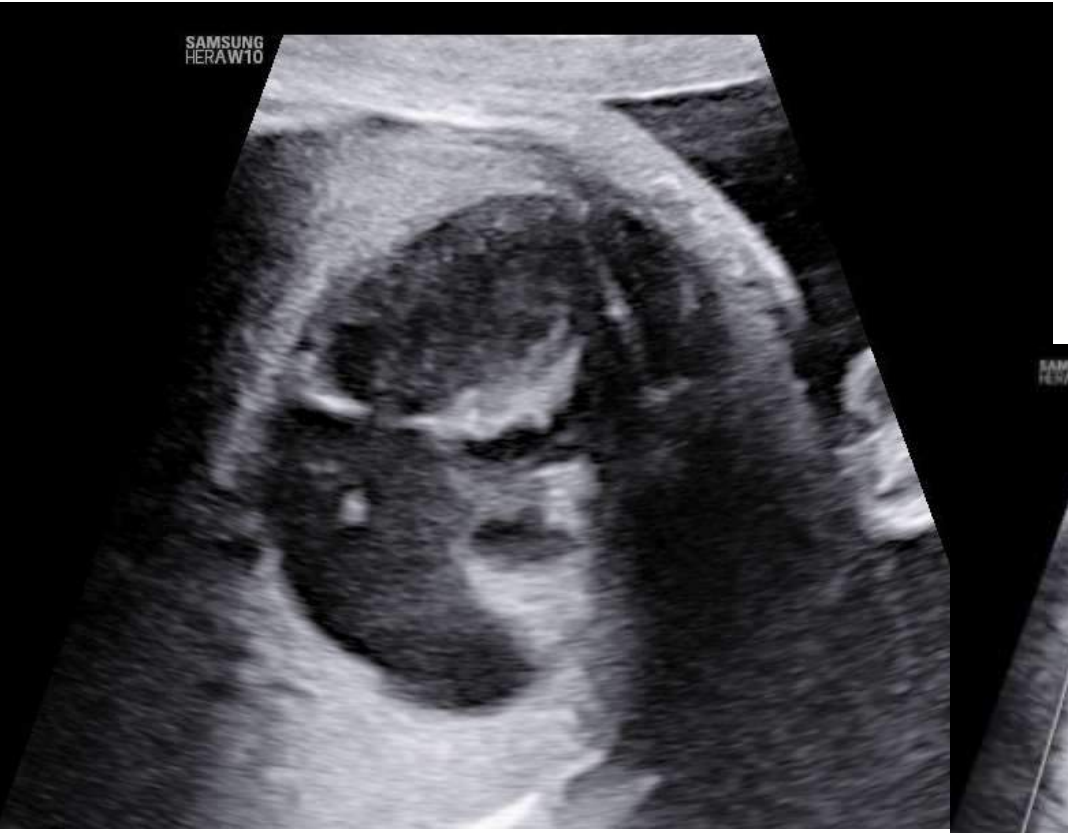
Case 1 Follow-up:

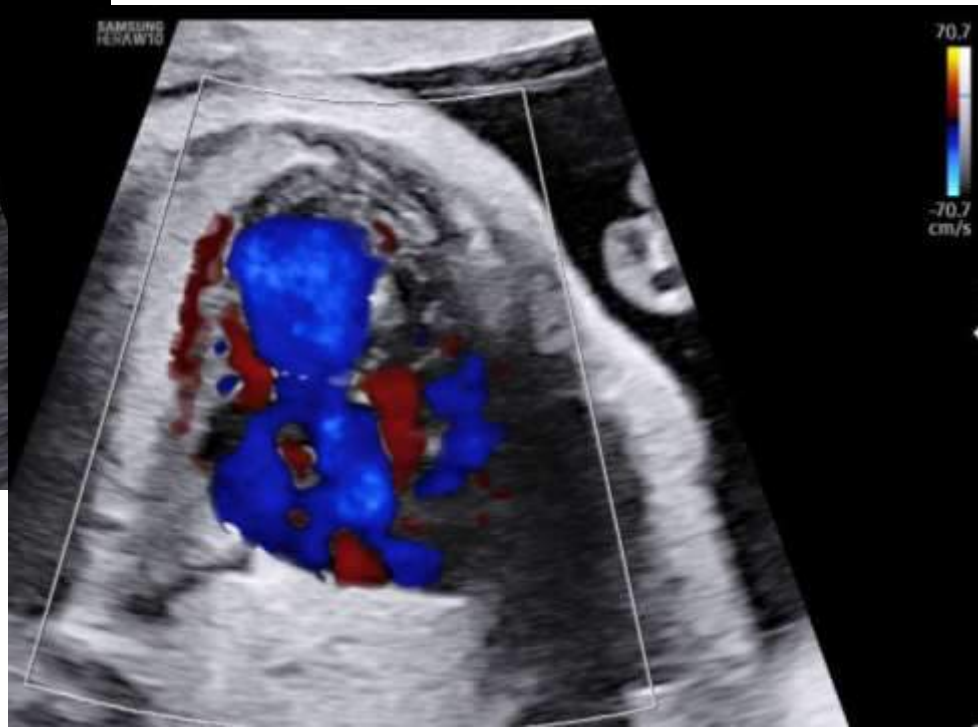
- Presented at 28 weeks with PPROM, PTL
- → decision to pursue comfort care

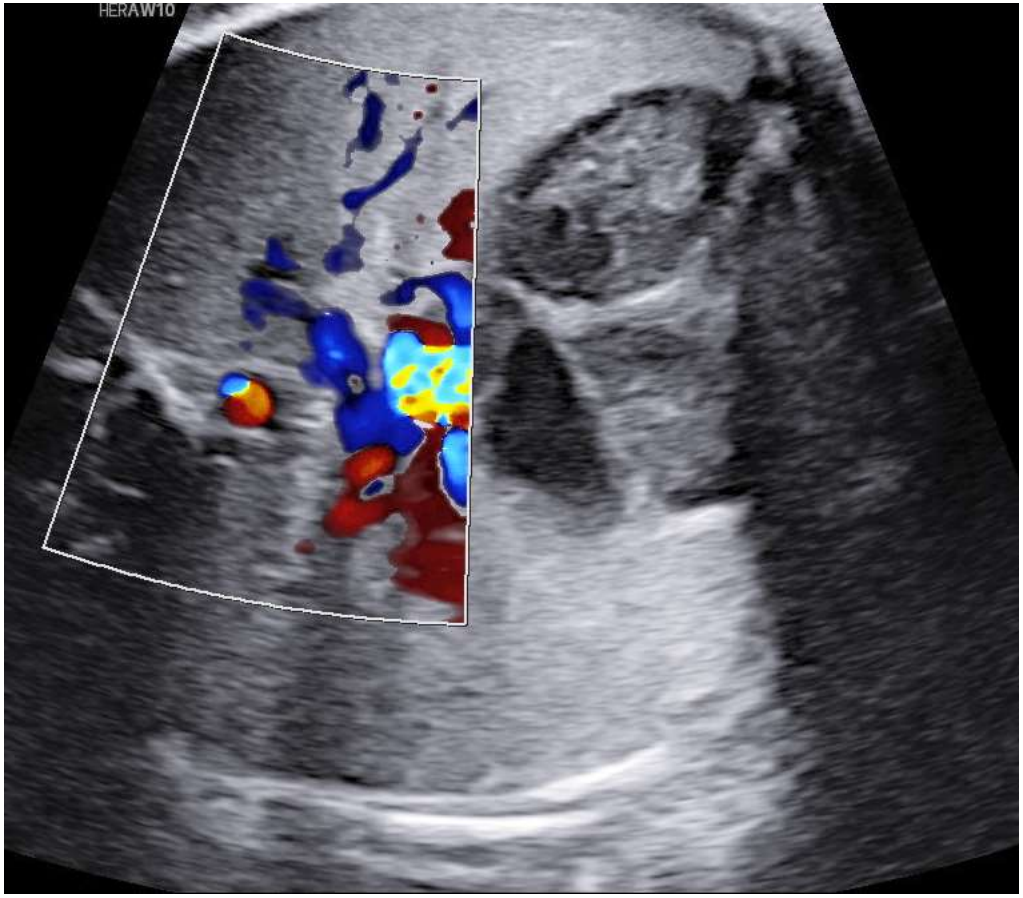
Case 2:

- 36 yo G3P1 with unremarkable medical/family history
- History of “normal” 20 week ultrasound
- 28 week growth US with new MFM group
  - Concern for Common AV Canal Defect
  - → refer to CHOP FHP

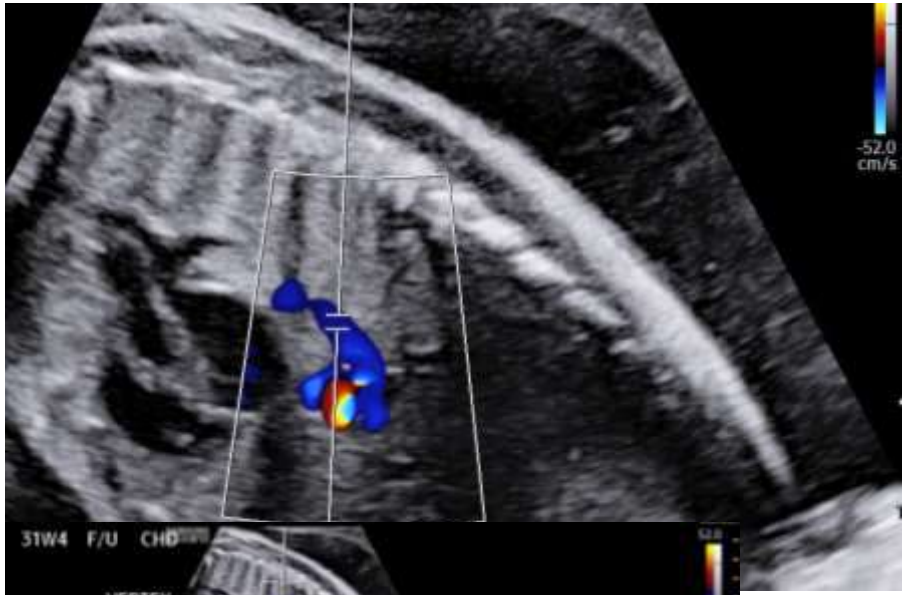
AV CANAL DEFECT - MOD AVVR

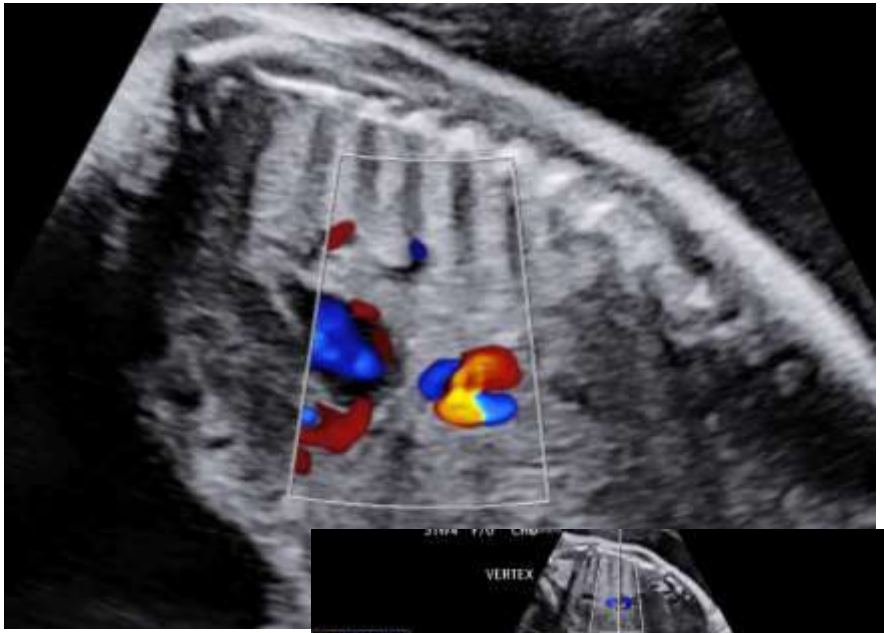


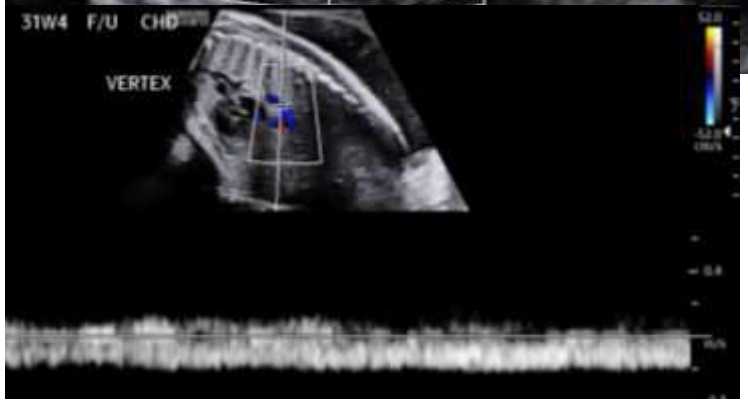


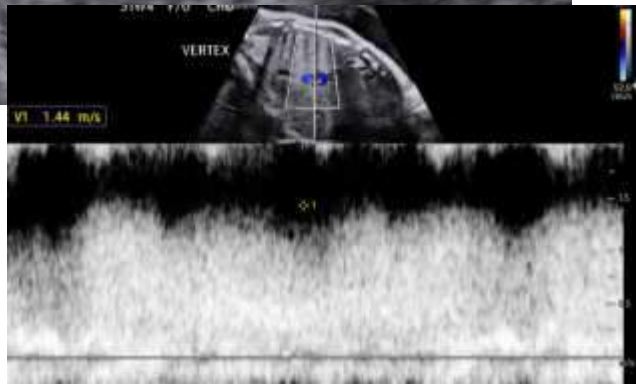

HERAW10

TAPVR - infracardiac









RISK STRATIFICATION

TRANSITION FROM FETAL CIRCULATION

- 100% of blood flow to lungs and need to return to left atrium
- If pulmonary vein return to LA obstructed
  - →Pulmonary Vein Hypertension
  - →Pulmonary edema

- Limited medication options
- NEED Surgery

CHOP DELIVERY CLASSIFICATION SCHEME

	Definition	Action	Examples
CLASS I	Simple cardiovascular anomaly or disease, no hemodynamic instability anticipated.	Neonatology manages	AV Canal, pink TOF
CLASS II	Cardiovascular anomaly or disease of moderate severity, including ductal dependant lesions. Hemodynamic instability is possible, but unlikely and not anticipated.	Neonatology manages	HLHS no risk, pulmonary atresia
CLASS III	Cardiovascular anomaly or disease of important severity with the possibility or likelihood of hemodynamic instability.	Neonatology + Cardiology manages	TGA, TAPVC
IMPACT	Cardiovascular anomaly or disease in which hemodynamic instability is anticipated at separation from placental circulation. Immediate post-partum access to cardiac therapy (IMPACT).	Cardiac services manages	HLHS + IAS, Ebstein's anomaly, CHB, hydropic fetus

Fetal Doppler Echocardiographic Assessment Predicts Severe Postnatal Obstruction in Total Anomalous Pulmonary Venous Connection

Journal of the American Society of Echocardiography
Volume 35 Number 11

Matthew J. Campbell, MD, Brian R. White, MD, PhD, Jack Rychik, MD, Jarrett Linder, MD, MS, Jennifer A. Faerber, PhD, Zhiyun Tian, MD, RDCS, and Meryl S. Cohen, MD, *Philadelphia, Pennsylvania; Wilmington, Delaware; Chicago, Illinois*

Normal

Obstructed pattern

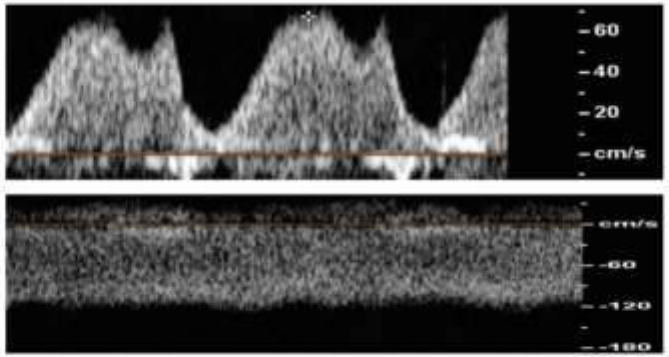


Figure 1 TAPVC vertical vein Doppler patterns. The Doppler tracing at the *top* shows a Doppler signal from a fetus with unobstructed TAPVC, demonstrating phasic variability, which translates to a high PVVI. The Doppler tracing at the *bottom* is obstructed and does not show phasic variability, leading to a low PVVI.

- Velocity correlates to obstruction
  - PVVI
    - Pulm vein max – pulm vein min / mean
  - Minimum velocity may be best predictor
  - No large scale studies

Review

Utility of Fetal Echocardiography with Acute Maternal Hyperoxygenation Testing in Assessment of Complex Congenital Heart Defects

Children 2023, 10, 281

Sheetal R. Patel [1,*], Nitin Madan [2], Pei-Ni Jone [1] and Mary T. Donofrio [3]

Diagnosis	Baseline Fetal Echocardiogram Findings Suggestive of Hemodynamic Instability after Birth	Expected Changes with MH Performed in the Third Trimester Suggestive of Hemodynamic Instability after Birth	Delivery Room Recommendations
TAPVR with significant Obstruction	Pulmonary vein Doppler [6] – Monophasic non-pulsatile pulmonary venous flow – Fetal vertical vein Doppler peak velocity > 0.74 m/s [31]	Mean gradient in the vertical vein after MH correlates with the severity of TAPVR obstruction seen postnatally [12]	– Intubation with mechanical ventilation. – Peripheral IV and/or umbilical line – Initiation of PGE1 infusion (may relax the ductus venosus smooth muscle for infra diaphragmatic TAPVR) – Plan for immediate surgical intervention.

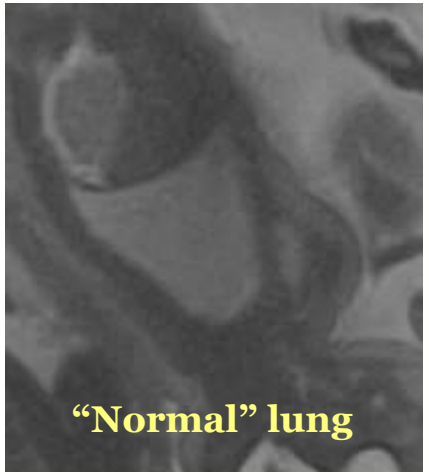
- Assess for velocity change in obstructed vessel
- > 35 weeks GA

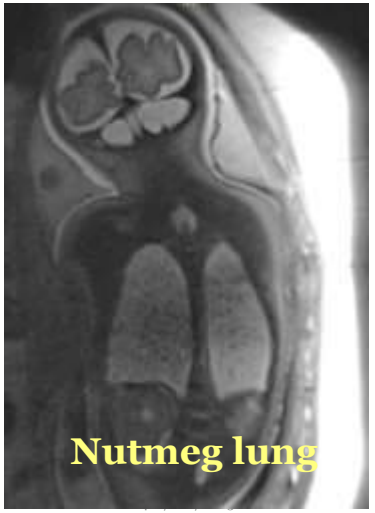
DELIVERY PLANNING FOR FETAL TAPVR

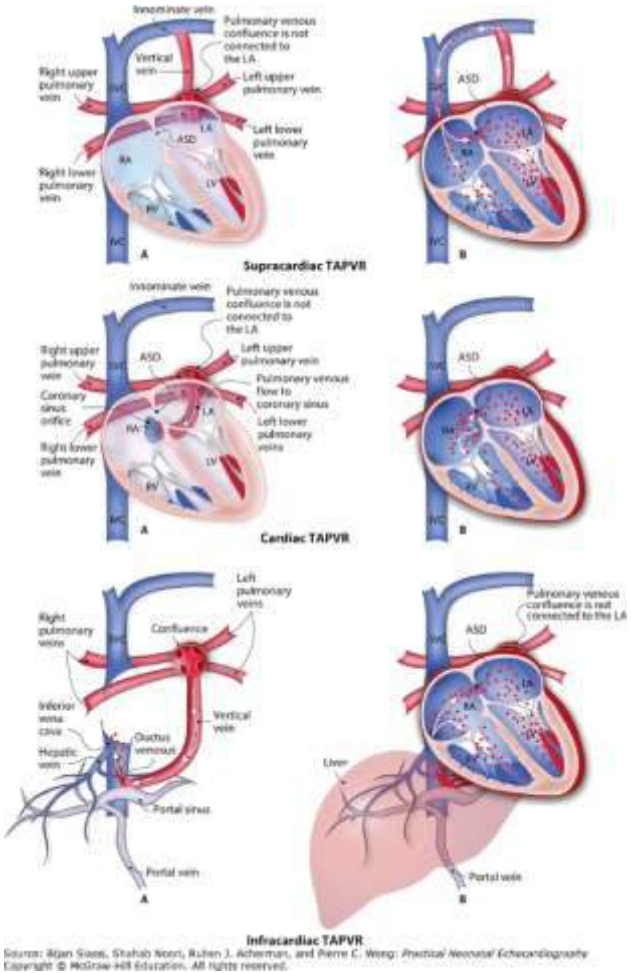
- Obstruction?
  - Abnormal Dopplers with elevated velocities, nonphasic flow

- Maternal Hyperoxygenation
  - →Increase velocity in obstructed venous connection
    - → IMPACT delivery (surgical OR available for immediate intervention)
  - →No change of minimal Doppler abnormalities at baseline
    - → Consider "Class 3 delivery" (CICU available for support at delivery)

- Fetal Lung MRI
  - →"nutmeg lung" (pulmonary lymphangiestasia)
    - →Unlikely to offer IMPACT procedure
    - →Palliative care involvement, consideration of non-intervention







TAPVR

- *Failure of the left atrium to link with the pulmonary venous plexus ; retention of primitive connections*

Supracardiac – (45-50%)

- retained pulmonary vein connections to the cardinal venous systems.
- pulmonary veins from both lungs course to a confluent chamber that is located just posterior to the left atrium. From this chamber, blood ascends through a vertically oriented vein that most often connects to the left innominate vein.
    - right-sided superior vena cava, azygous vein, or a persistent left superior vena cava to coronary sinus.

Cardiac – (15-20%)

- retained pulmonary vein connections to the cardinal venous systems
- pulmonary veins connect to the posterior aspect of the coronary sinus or to the right atrium itself

Infracardiac – (20-25%)

- retained pulmonary vein connections to the umbilicovitelline venous system.
- pulmonary veins drain into a common vertical vein that courses inferiorly from the mediastinum, through the diaphragm via the esophageal hiatus, and inserts most often into the portal vein
- Hepatic vein, DV, IVC

Mixed – (10%)

- any combination of connections that enter 2 or more different levels

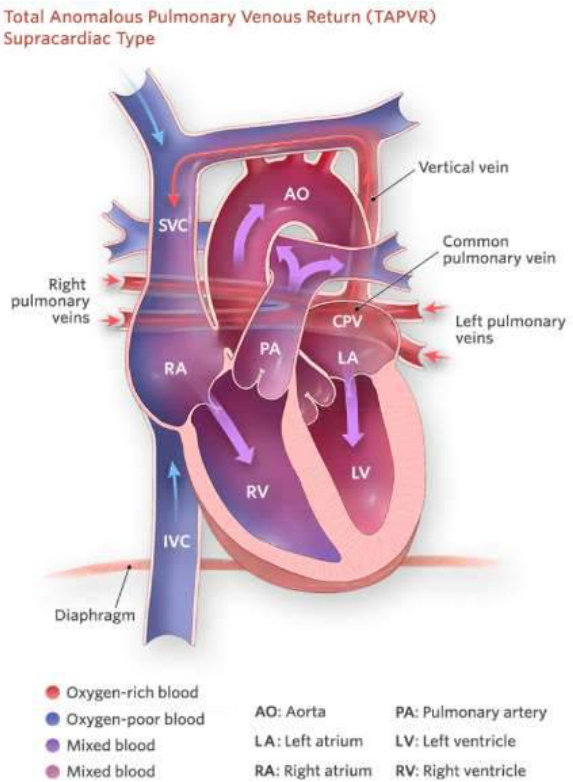
Levels of pulmonary venous obstruction

Supracardiac – Obstruction may occur where the vertical vein courses between the left pulmonary artery and left mainstem bronchus

Infracardiac – Obstruction usually occurs in cases of infracardiac TAPVC. Constriction of the vertical vein commonly occurs at the level of the diaphragm, within the liver in cases where the connection involves the ductus venous, or by the liver parenchyma when the connection is with the portal vein

Cardiac – In some cases of cardiac TAPVC, obstructed flow may be due to stenosis at the mouth of the coronary sinus where the pulmonary veins connect.

Other – Other sources of obstruction include stenotic, tortuous, or atretic pulmonary and common vertical veins, or a restrictive atrial septum.



TAPVR

- Pulmonary veins originate from lung development
  - Lungs develop from primitive foregut
  - Share a common vascular plexus (splanchnic plexus) which initially drain into common cardinal and umbilicovitelline venous system
  - Portion of splanchnic plexus differentiates into primitive pulmonary vascular bed
  - Primitive left atrium forms evagination (Common pulmonary veins) which grows to join the splanchnic plexus.
- Failure of the left atrium to link with the pulmonary venous plexus ; retention of primitve connections