Heterotaxy Syndrome: When Your Organs Decide to Mix It Up!

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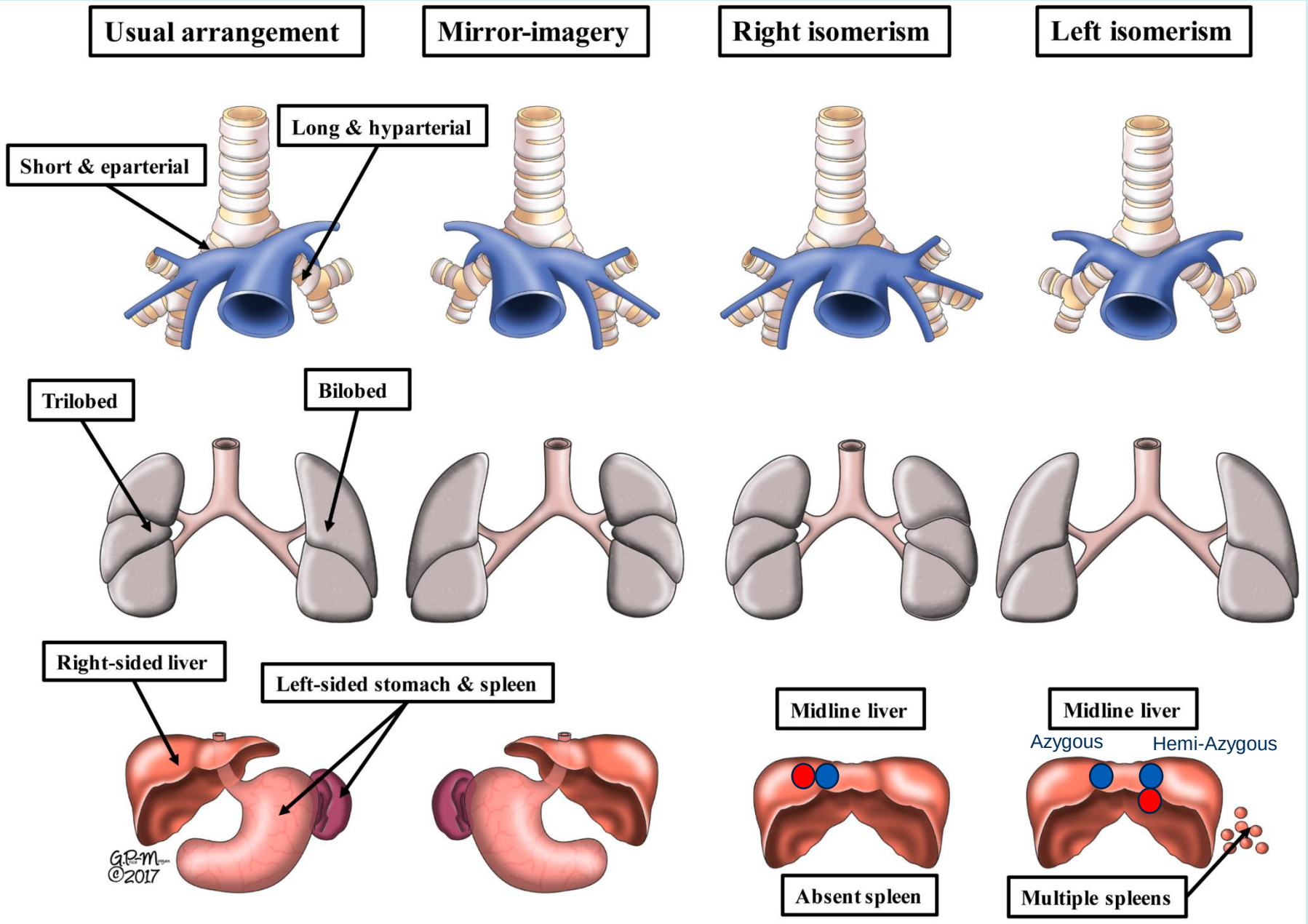
Children's Mercy Wichita

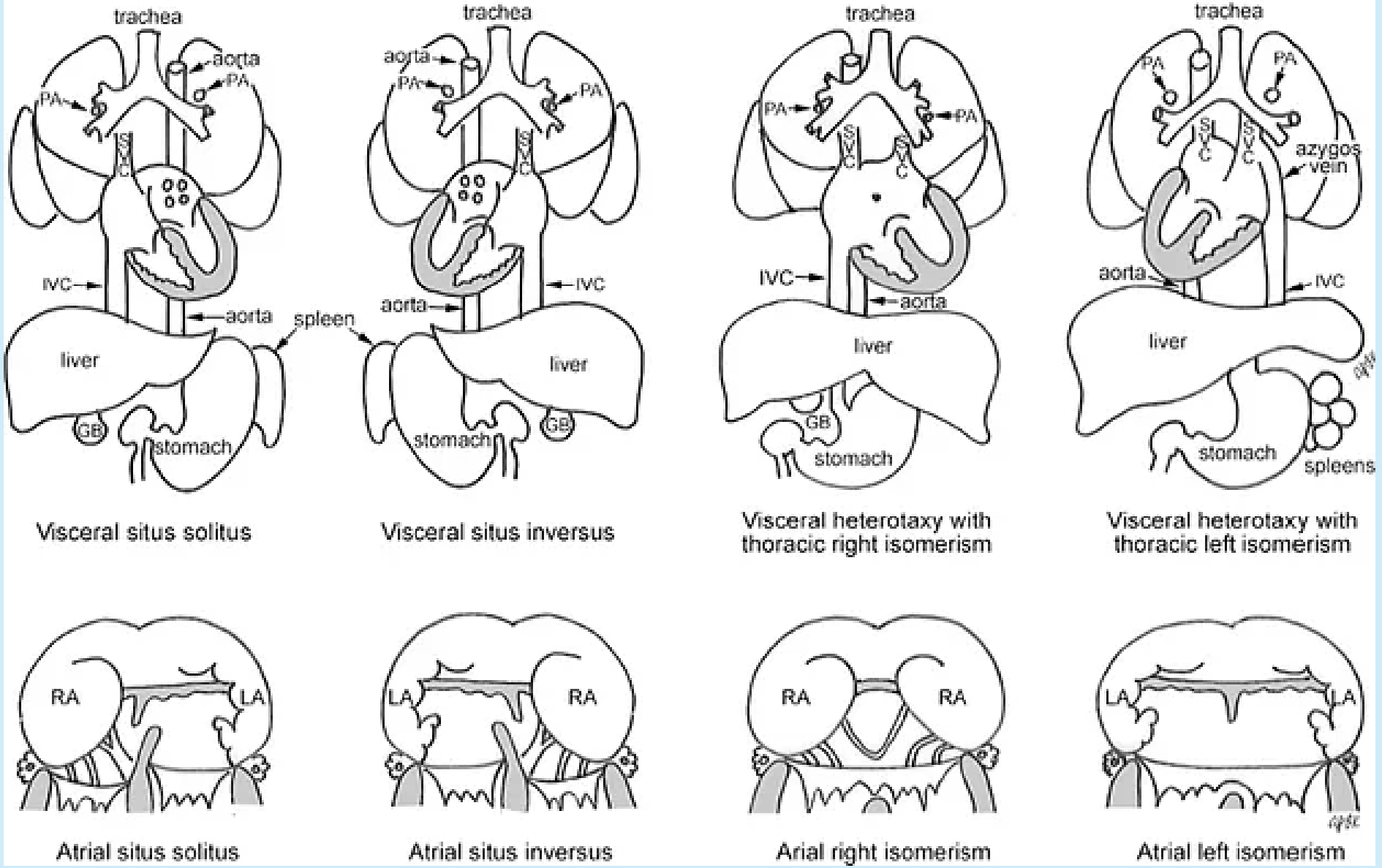
What is Heterotaxy?

- Greek meaning Heteros (other) Taxis (arrangement)

- Abnormal lateralization of abdominal and thoracic viscera
- Birth prevalence 1 in 10,000
- Normal R/L asymmetry is established very early in embryonic development by ciliary motion
  - Most known Primary Ciliary Dyskinesia
  - Many other genes have been implicated in either right or left atrial isomerism



Anderson, R. H., Spicer, D. E., & Loomba, R. (2018). Is an Appreciation of Isomerism the Key to Unlocking the Mysteries of the Cardiac Findings in Heterotaxy? *Journal o Cardiovascular Development and Disease*, *5*(1), 11. https://doi.org/10.3390/jcdd5010011


trachea
aorta
PA
PA
SVC
IVC
aorta
liver
GB
stomach
spleen
Visceral situs solitus
trachea
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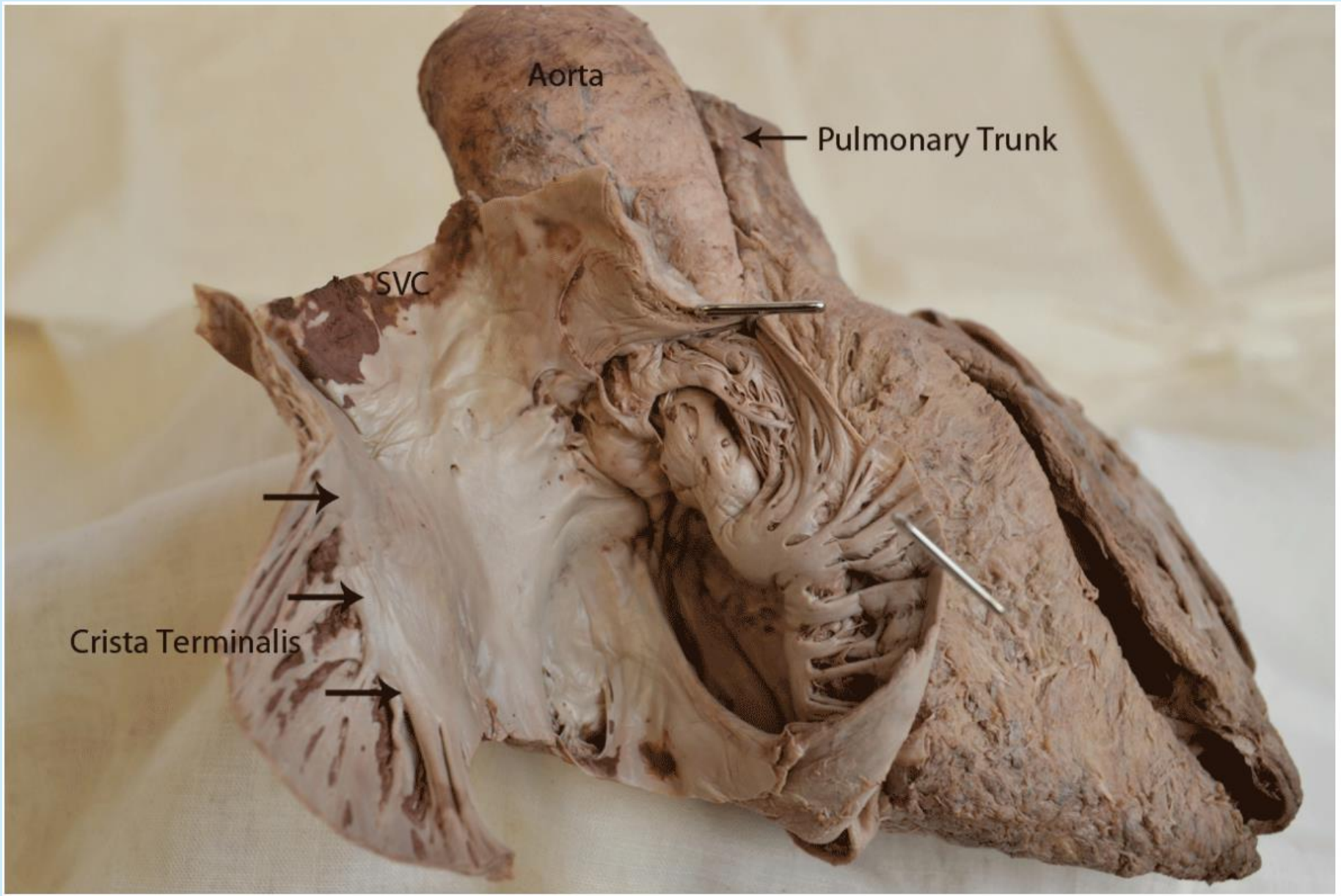
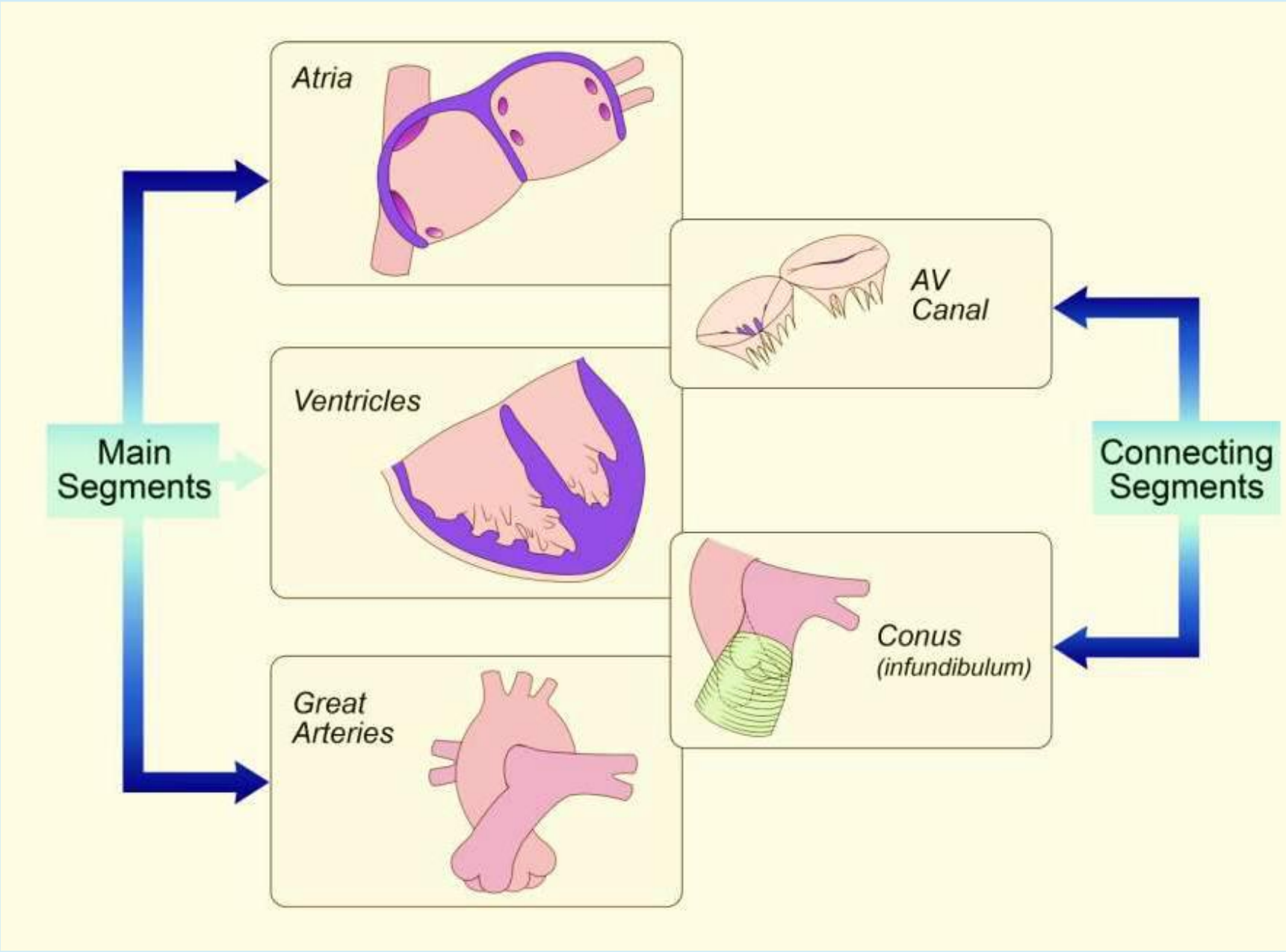
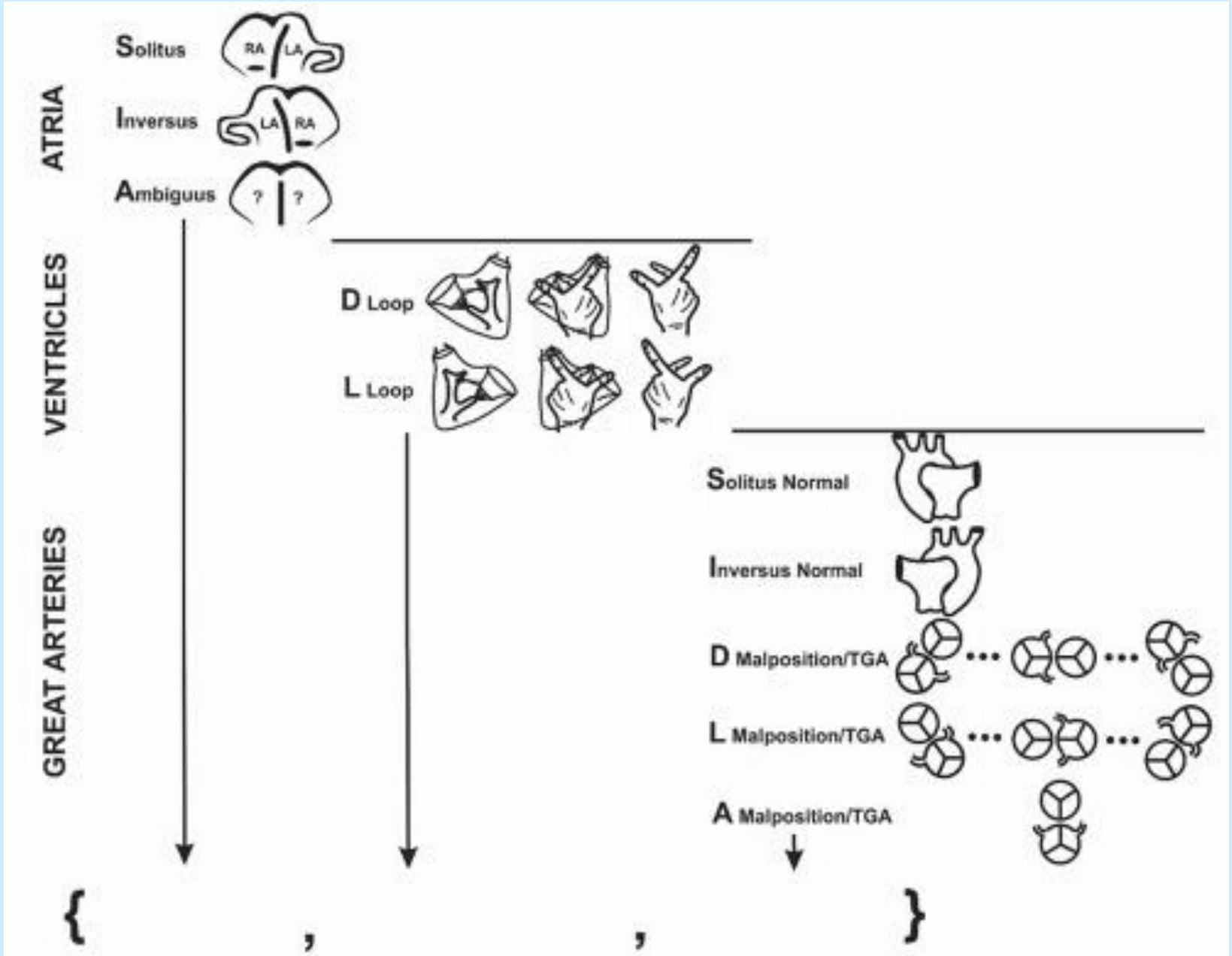
PA
SVC
IVC
aorta
liver
GB
stomach
Visceral situs inversus
trachea
PA
PA
SVC
IVC
IVC
aorta
liver
GB
stomach
Visceral heterotaxy with thoracic right isomerism
trachea
PA
PA
SVC
IVC
azygos vein
aorta
IVC
liver
stomach
spleens
Visceral heterotaxy with thoracic left isomerism
RA
LA
Atrial situs solitus
LA
RA
Atrial situs inversus
RA
RA
Arial right isomerism
LA
LA
Atrial left isomerism



Human heart disssection, courtesy of Rocky Vista University; College of Osteopathic Medicine. Parker, CO




ATRIA
VENTRICLES
GREAT ARTERIES
Solitus
RA
LA
Inversus
LA
RA
Ambiguus
?
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Inversus Normal
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A Malposition/TGA
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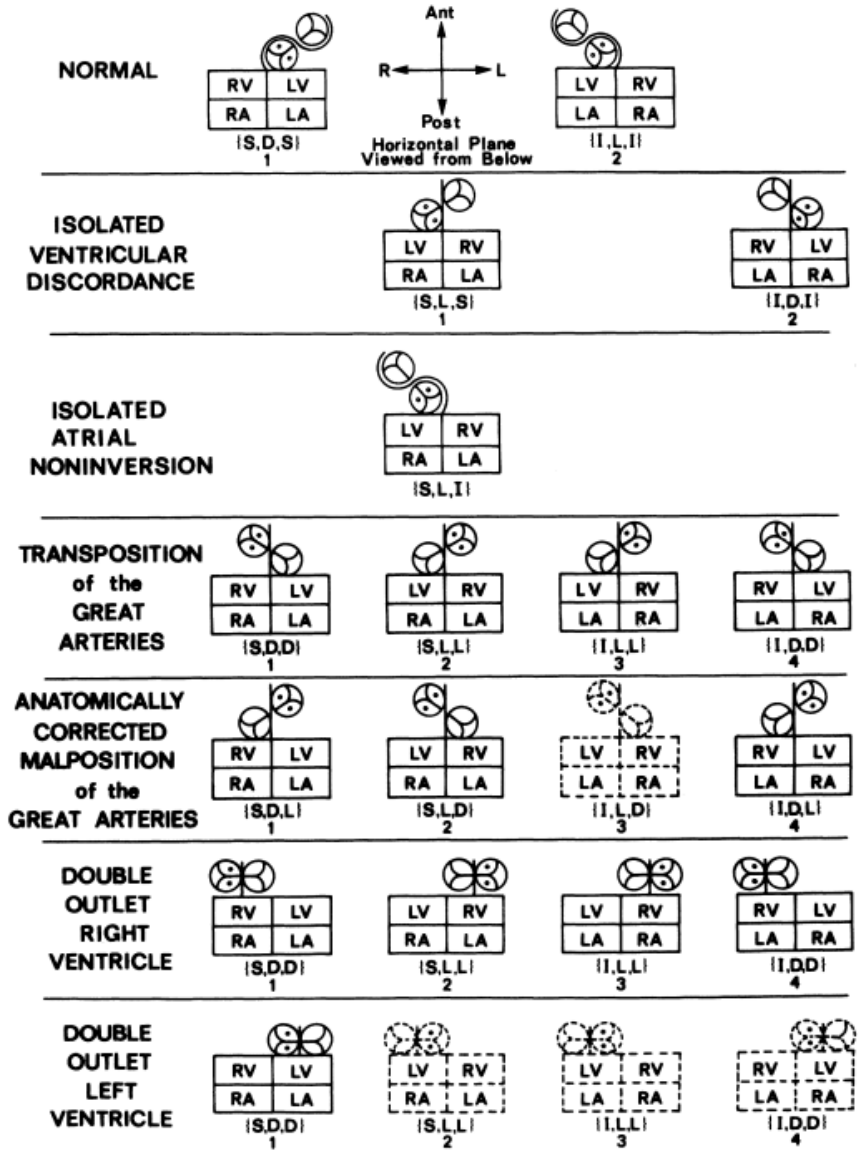

FIGURE 1. *Diagram of various types of heart, viewed from below. (Reproduced with permission from Van Praagh.[4])*

Left isomerism

- Absent/rudimentary sinus node, heart block
- Bilateral SVC
- Absent or interrupted IVC (90% cases)
  - Enlarged azygous veins (to SVC)
  - Hepatic veins directly connected to atrium
- Ipsilateral pulmonary vein connections but can be variable

Left isomerism – cont,

- AV Canal
- LVOTO
- Coarctation
- Arch side, topology of ventricles, position of heart, position of heart is random

- Polysplenia, often abnormal function
- Bilobed lungs

Right isomerism

- Asplenia
- Dual SA nodes, atrial tachycardia, EAT
- Bilateral SVC
- IVC and descending aorta same side spin
- Absent coronary sinus
- TAPVR (supra, infra, intra or mixed)

Right isomerism – cont.

- Atrioventricular Septal Defect
  - Unbalanced
- DORV
  - Pulmonary stenosis or atresia with single outlet with aorta originating from RV
- Arch side, position of heart, position of heart is random

	Right Isomerism	Left Isomerism
AV Connections		
Atrioventricular Canal	Almost 100%	~2/3 of patients
VA Connections	DORV	>50% normal VA connection
	Pulmonary stenosis or atresia	Sub-aortic/aortic stenosis
Pulmonary Veins	TAPVR	Ipsilateral PV drainage
Systemic Veins	Bilateral SVC	Bilateral SVC
	Absent coronary sinus	Interrupted IVC
		Abnormal hepatic vein connection to heart
Conduction System	Bilateral sinus nodes	Complete heart block
		Paired AV nodes

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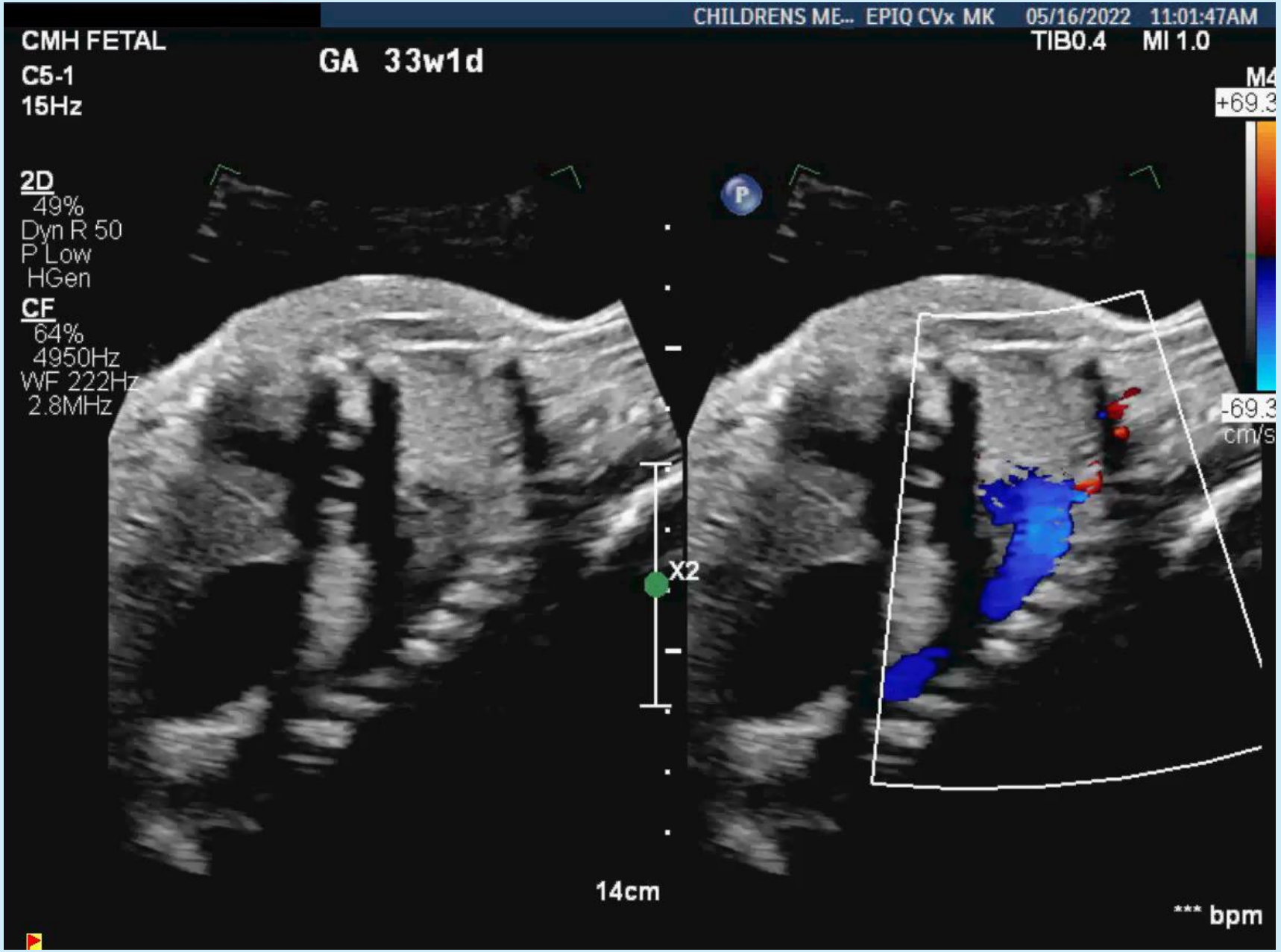

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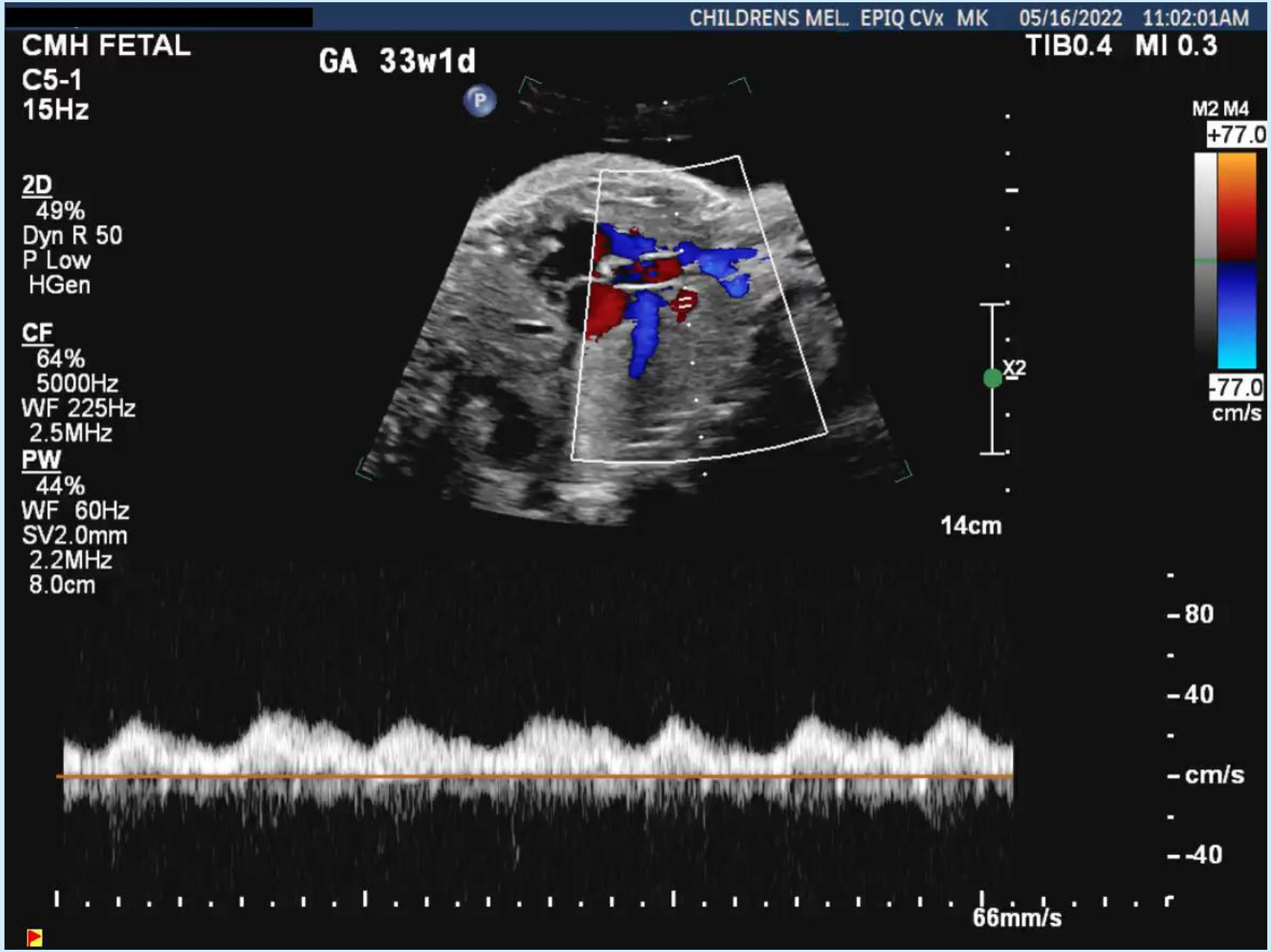

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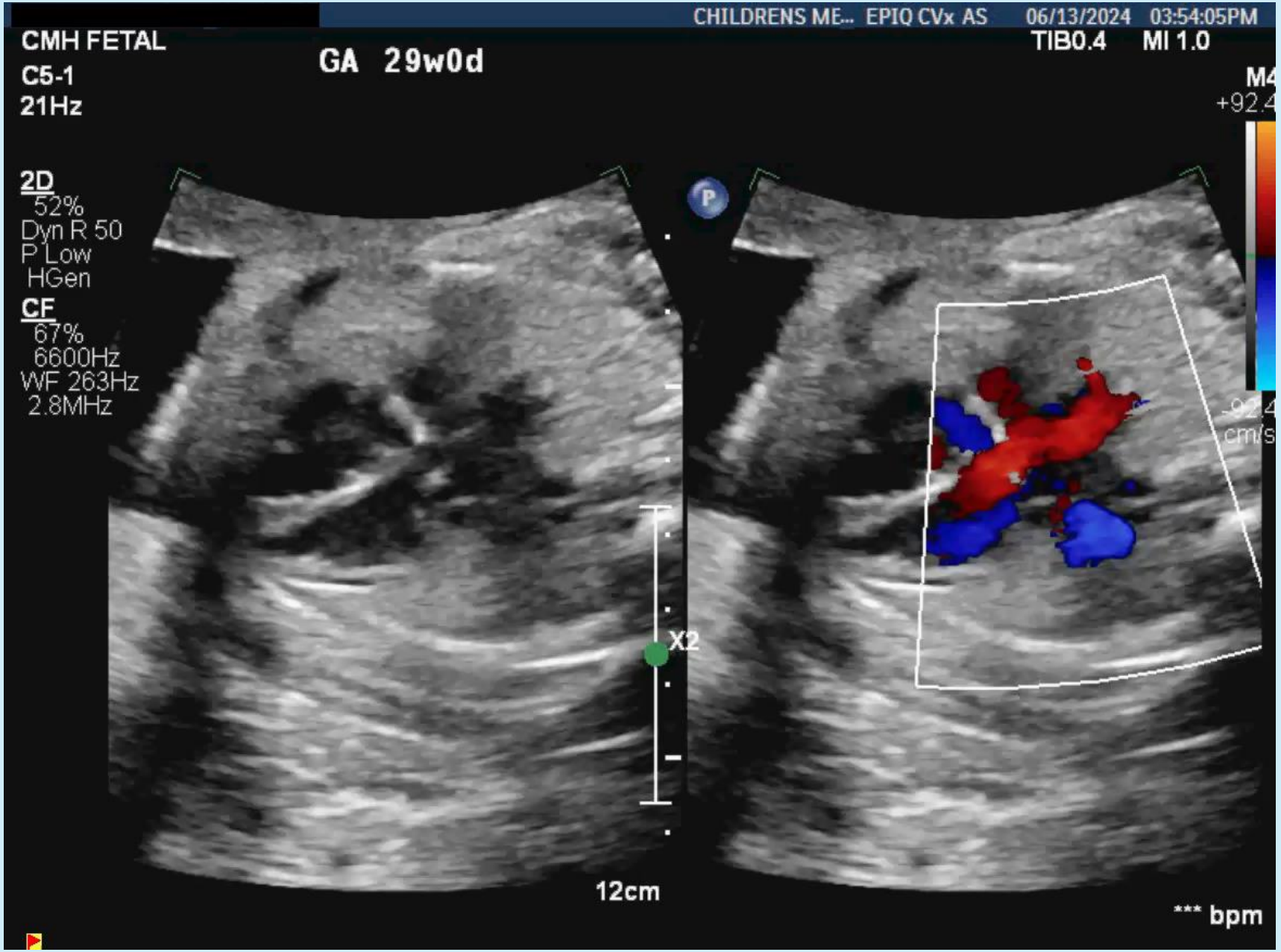
CHILDRENS ME... EPIQ CVx AS
06/13/2024 03:27:51PM
TIB0.3 MI 1.0
CMH FETAL
C5-1
91Hz
RS
2D
48%
Dyn R 50
P Low
HGen
GA 29w0d
M2
X2
11cm
*** bpm
P
R
2.0 4.0



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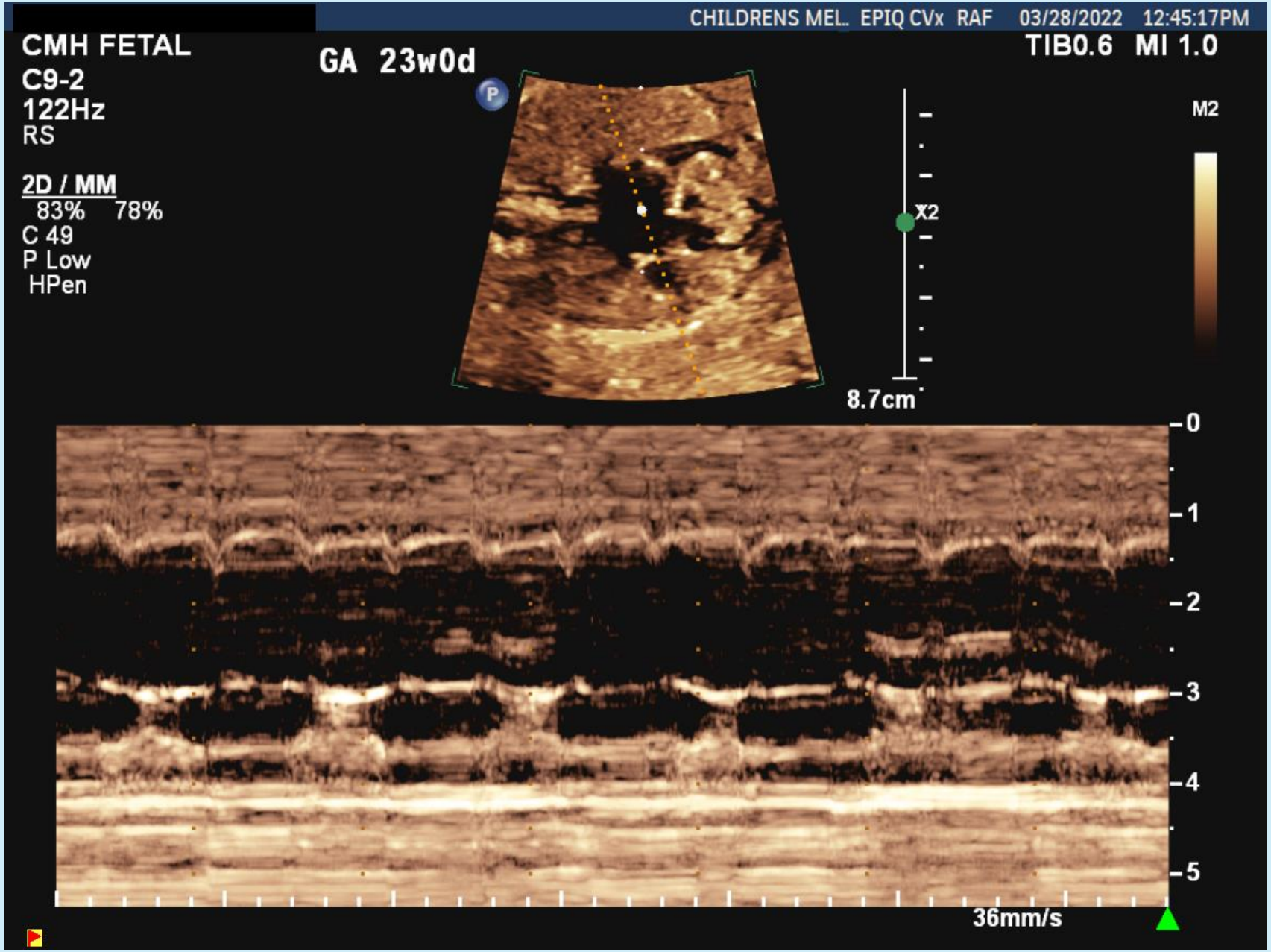

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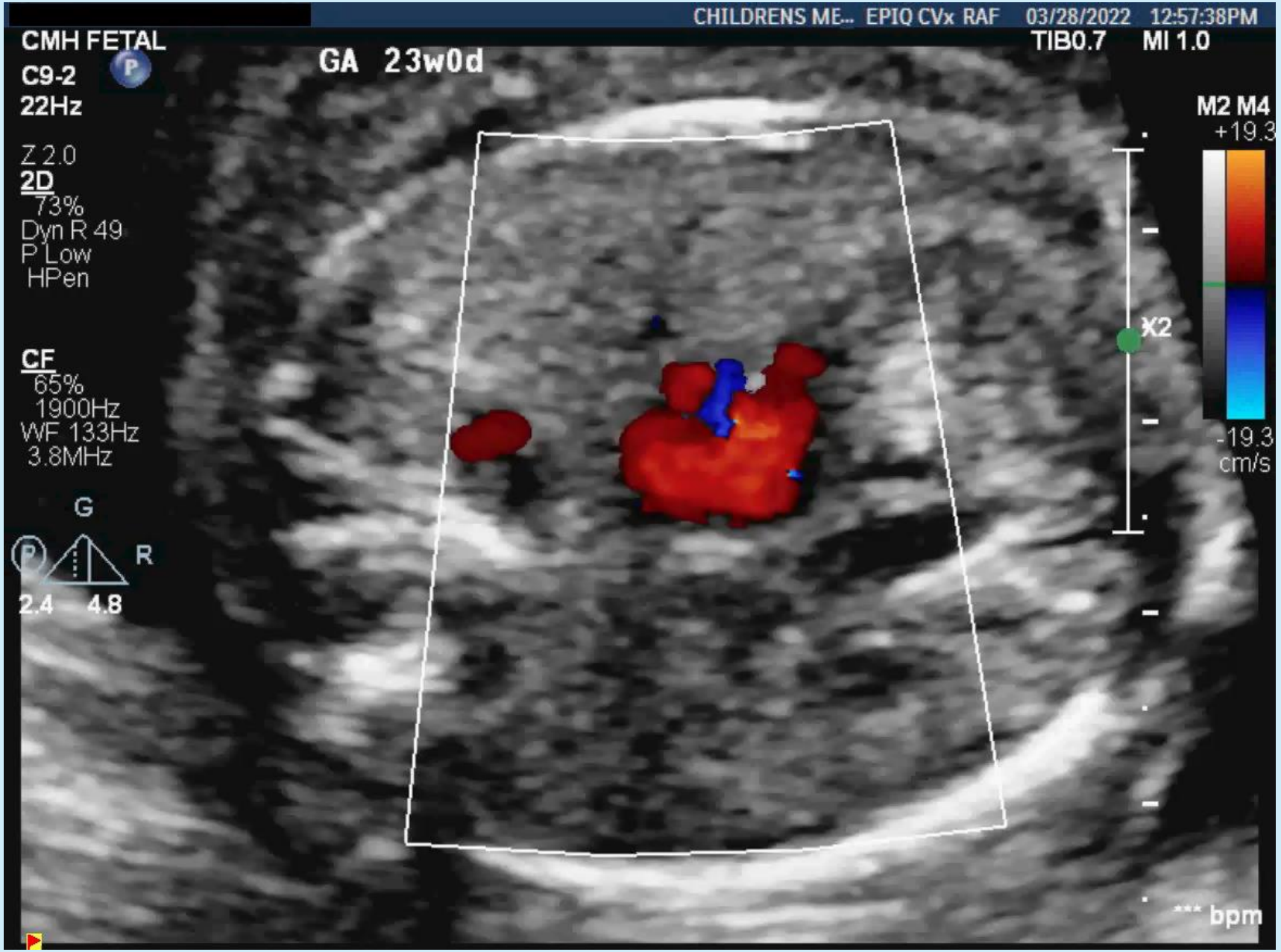

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CHILDRENS ME... EPIQ CVx RAF 03/28/2022 01:46:46PM
CMH FETAL
C5-1
44Hz
RS
Z 1.4
2D
44%
Dyn R 50
P Low
HPen
GA 23w0d
TIB0.1 MI 1.0
M2
G
P
R
1.7 3.4
X2
*** bpm

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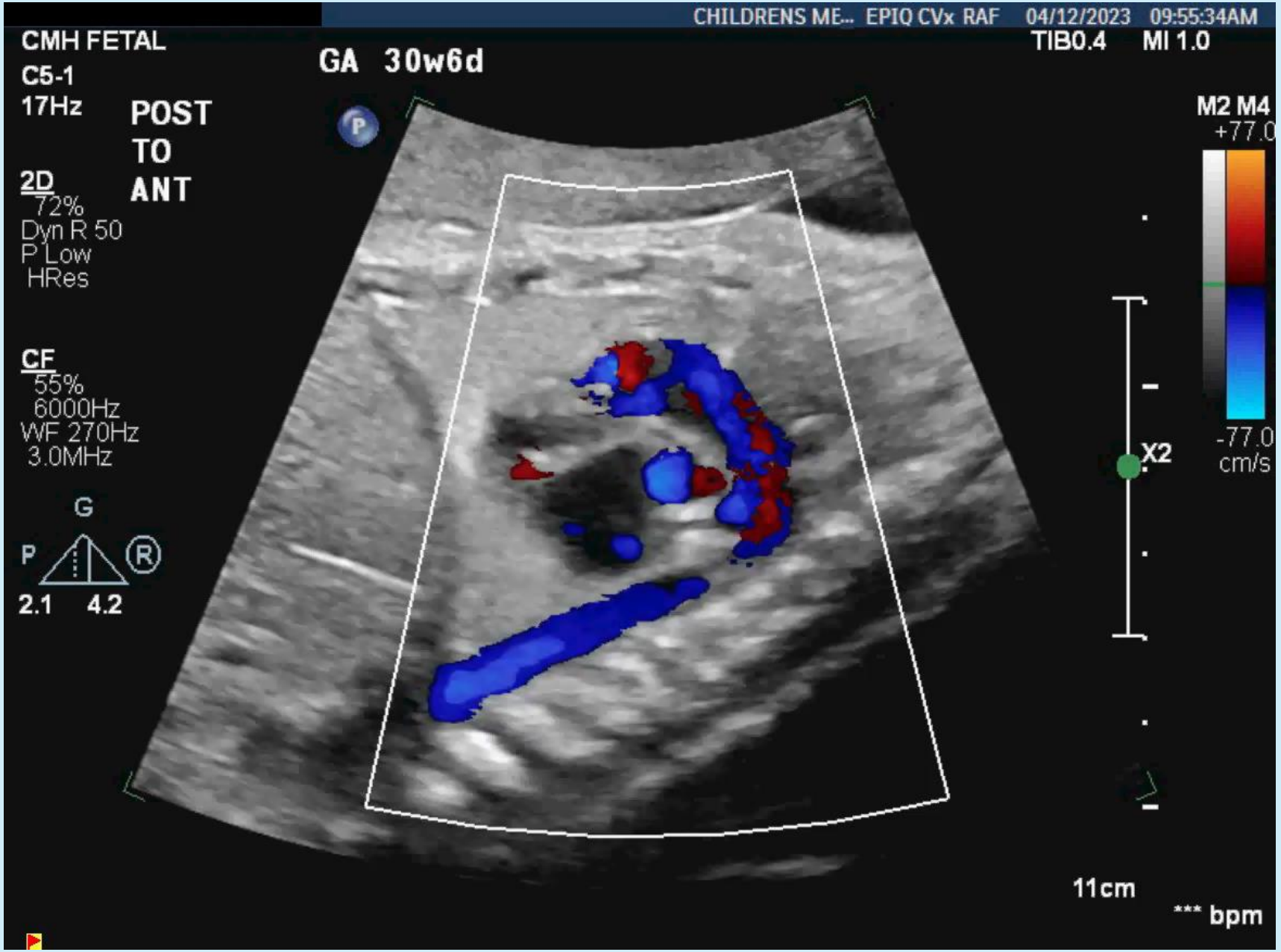
CHILDRENS ME... EPIQ CVx RAF 04/12/2023 09:55:24AM
CMH FETAL
C5-1
70Hz
RS
POST
TO
ANT
2D
60%
Dyn R 46
P Low
HRes
GA 30w6d
TIB0.2 MI 1.0
M2
X2
G
P
R
2.1 4.2
11cm
*** bpm



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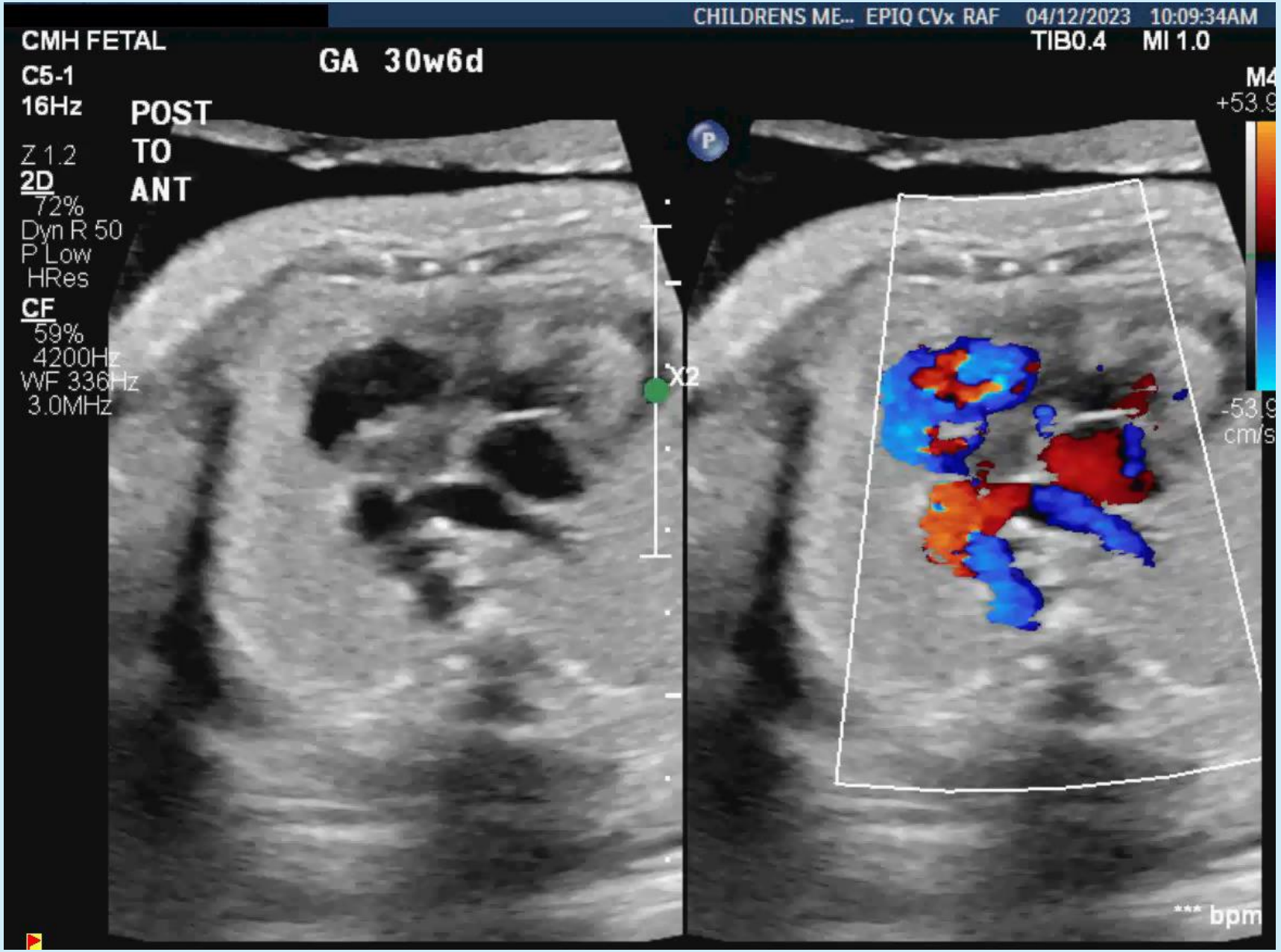

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