

Hope. Heal. Learn.

CARDIOLOGY
2025 

When A Fetal Cardiologist Could Use A Fetal Medicine Consult

February 18, 2025

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Richard D. Wood Jr. Center
for Fetal Diagnosis and
Treatment

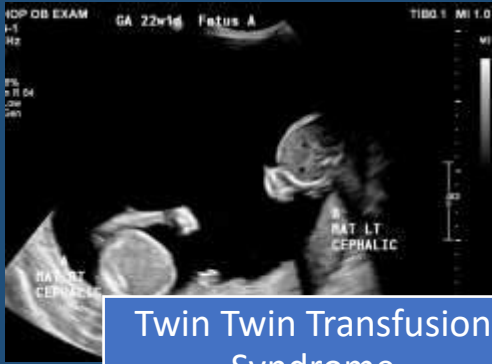


Children's Hospital
of Philadelphia

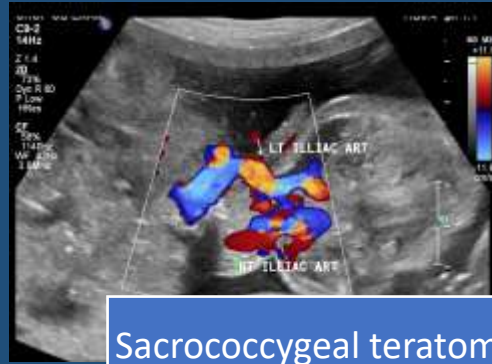
Richard D. Wood Jr.
Center for Fetal Diagnosis & Treatment



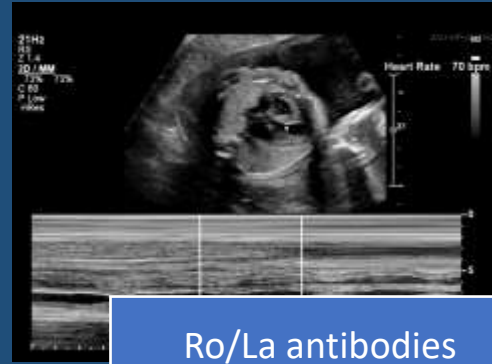
We all know there are
times when a fetal
medicine specialist needs
a fetal cardiologist.



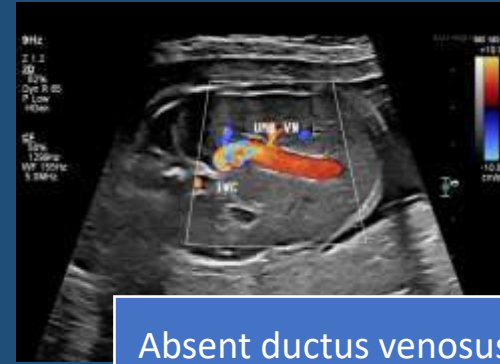
Twin Twin Transfusion Syndrome



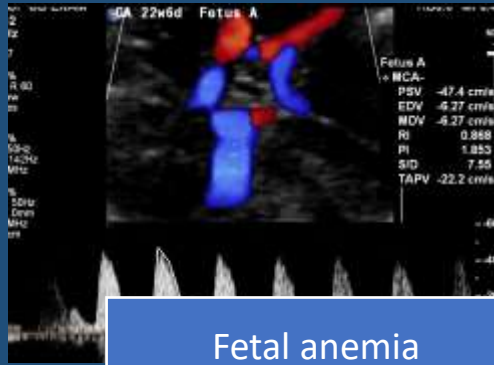
Sacroccocygeal teratoma



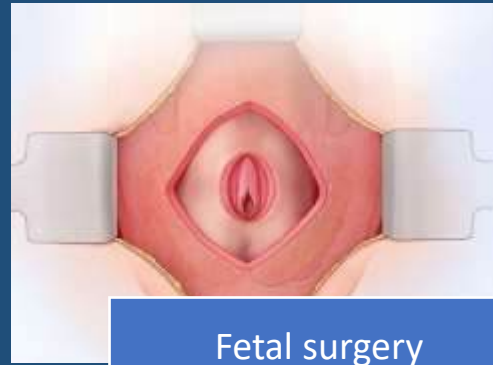
Ro/La antibodies



Absent ductus venosus



Fetal anemia



Fetal surgery

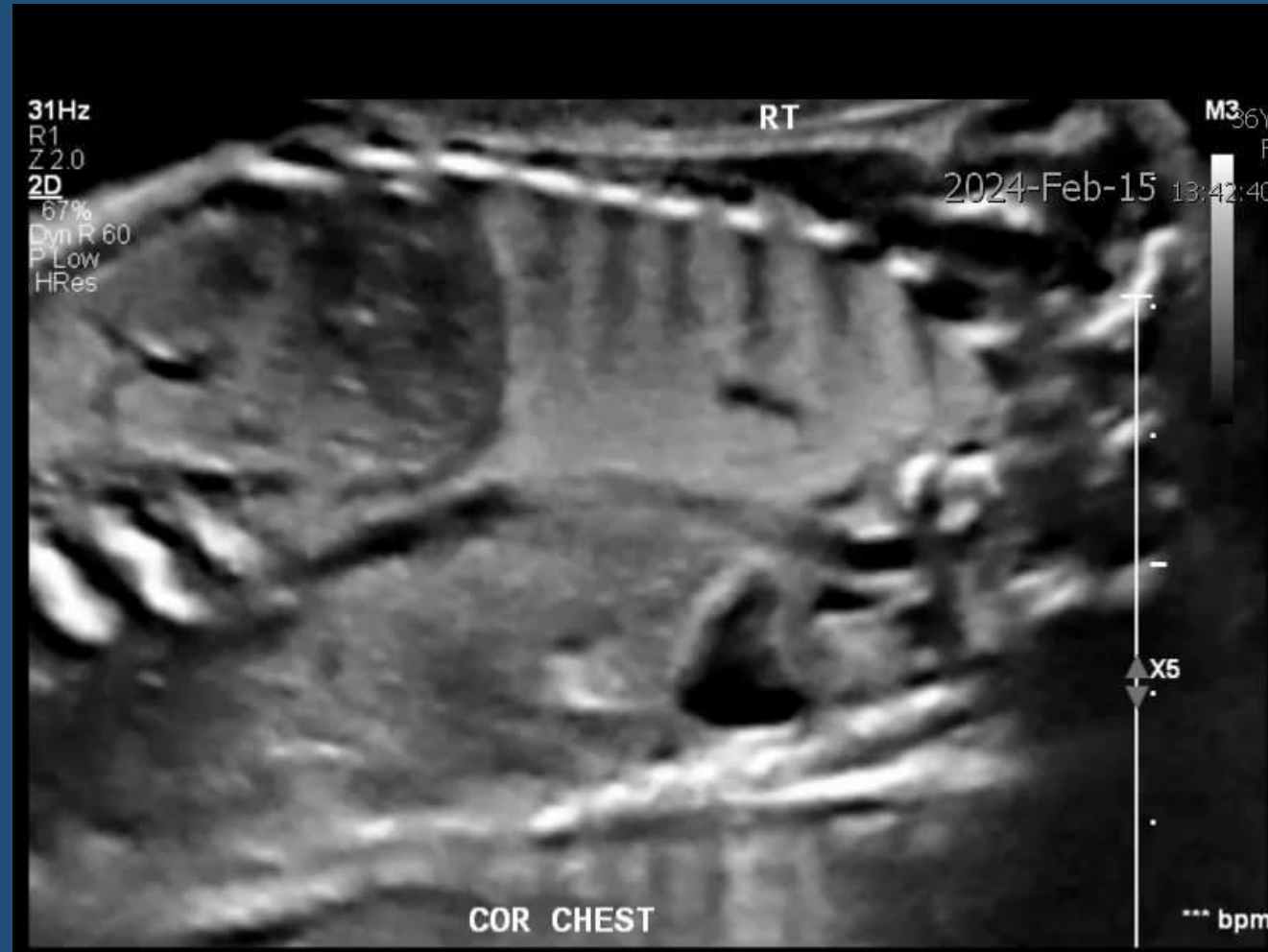


Hydrops

Are there times when a fetal cardiologist could benefit from a fetal medicine specialist?

Times when we can help
optimize postnatal care...







Ok, it's pulmonary agenesis, can't they just follow-up after birth?

Results: pulmonary agenesis location



13 (72%) Right



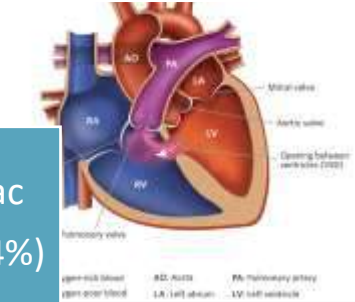
3 (17%) Left



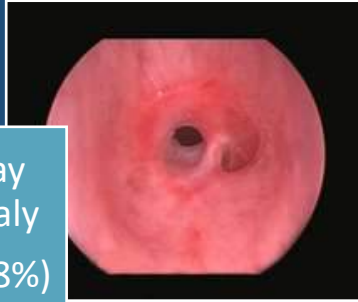
2 (11%) Bilateral

Associated anomalies (n=16)

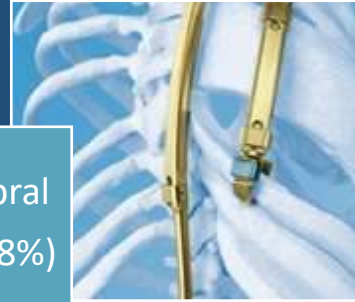
Cardiac
n=7 (44%)



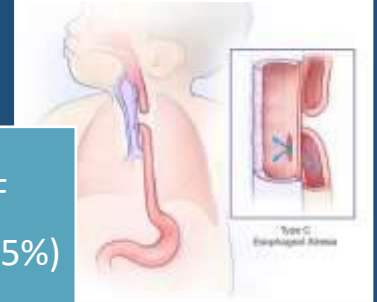
Airway
anomaly
n=6 (38%)



Vertebral
n=6 (38%)



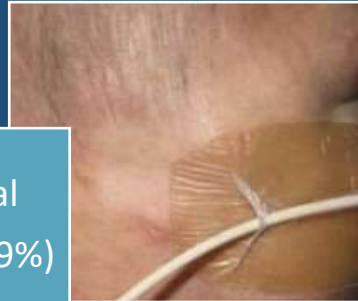
TEF
n=4 (25%)



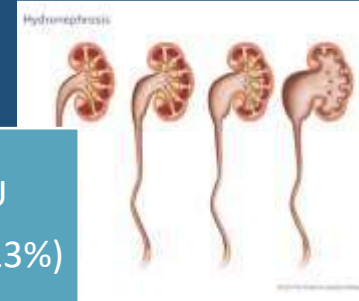
Other GI
n=4 (25%)



Facial
n=3 (19%)



GU
n=2 (13%)



Associated anomalies

Prenatally diagnosed	Postnatally diagnosed
<p>Vertebral: segmentation anomalies</p> <p>Cardiac: VSD, truncus arteriosus, DORV/VSD</p> <p>GI: suspected imperforate anus, TEF/EA</p> <p>Facial: anophthalmia, hypoplastic ear</p> <p>Renal: pelvic kidney, urinary tract dilation</p> <p>Limb: oligodactyly, short right radius/ulna</p> <p>GU: hypospadias</p>	<p>Vertebral: segmentation anomalies, scoliosis</p> <p>Cardiac: ASD</p> <p>GI: Imperforate anus, microgastria, TEF</p> <p>Facial: absent right ear</p> <p>Neuro: tethered spinal cord</p> <p>Airway: tracheomalacia, bronchial stenosis, complete tracheal rings</p>

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Airway issues

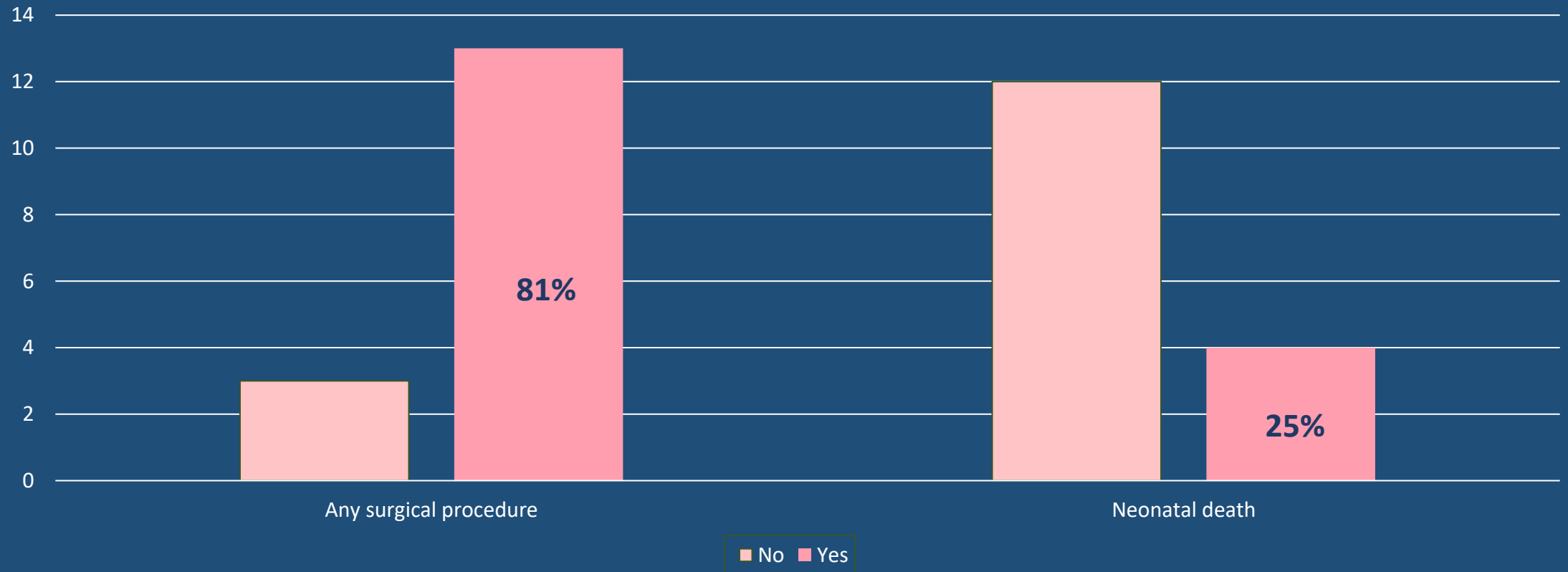


Tracheal or mainstem
bronchus stenosis

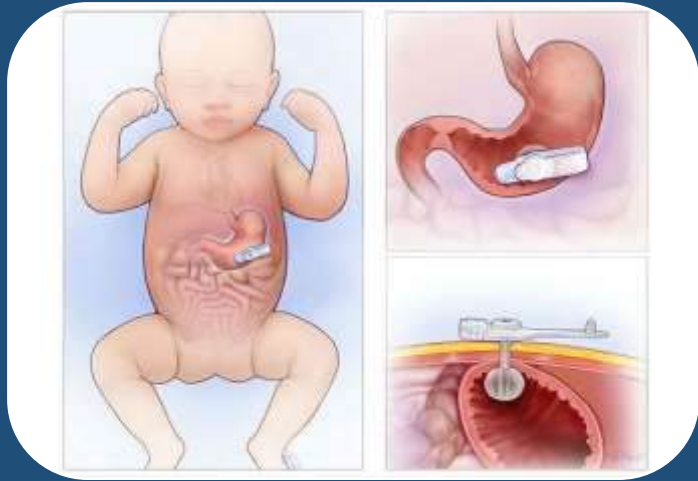


Complete tracheal
rings

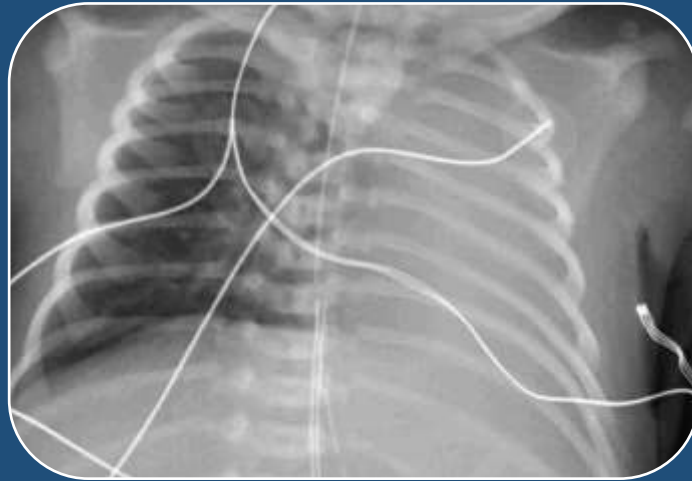
Neonatal/childhood outcomes (n=16)



Long-term survivors (n=12)



Gastrostomy
n=7 (58%)



Pulmonary hypertension
n=4 (33%)

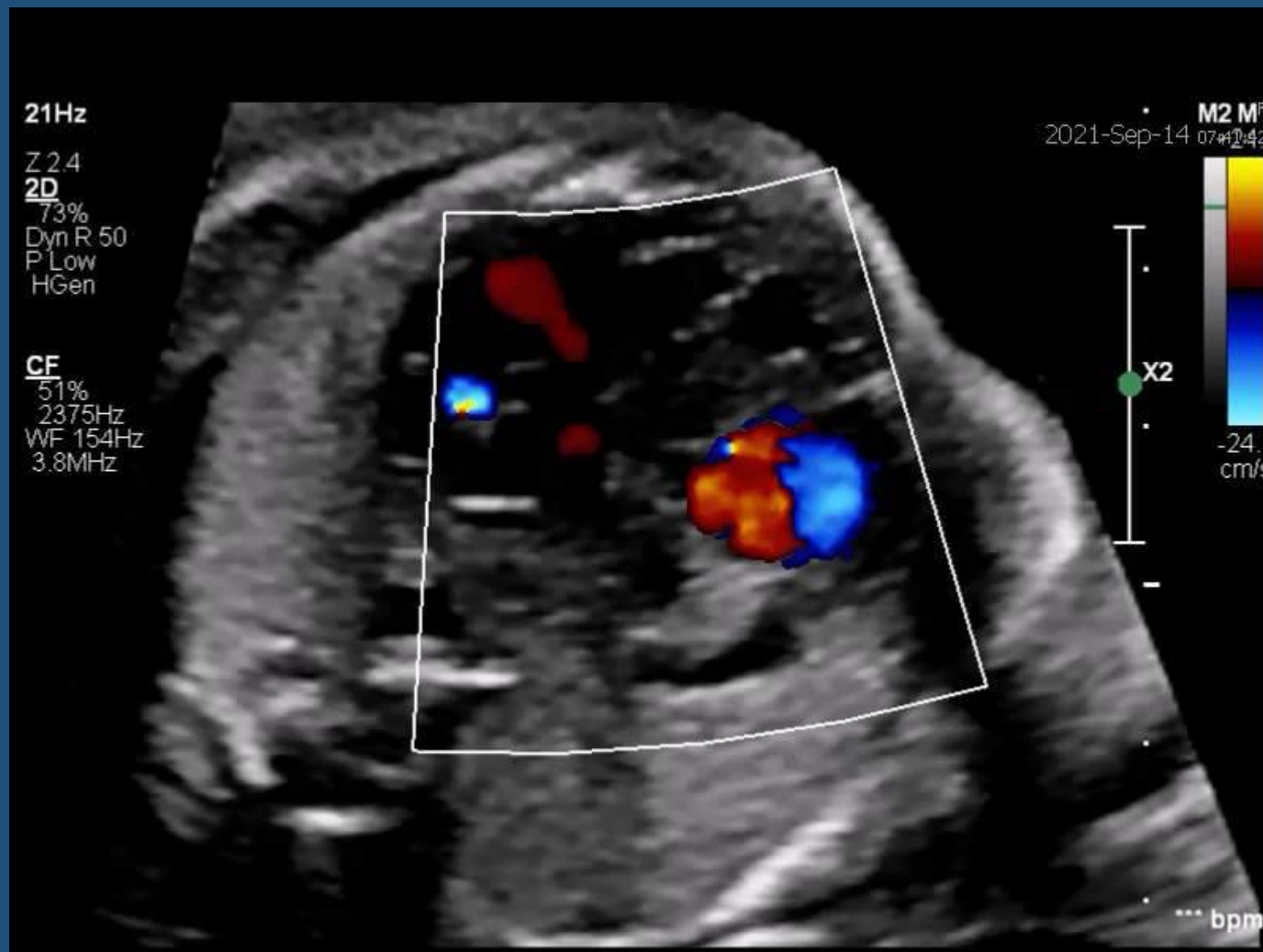


Long-term oxygen
n=1 (8%)

Times we can help nail
the diagnosis...



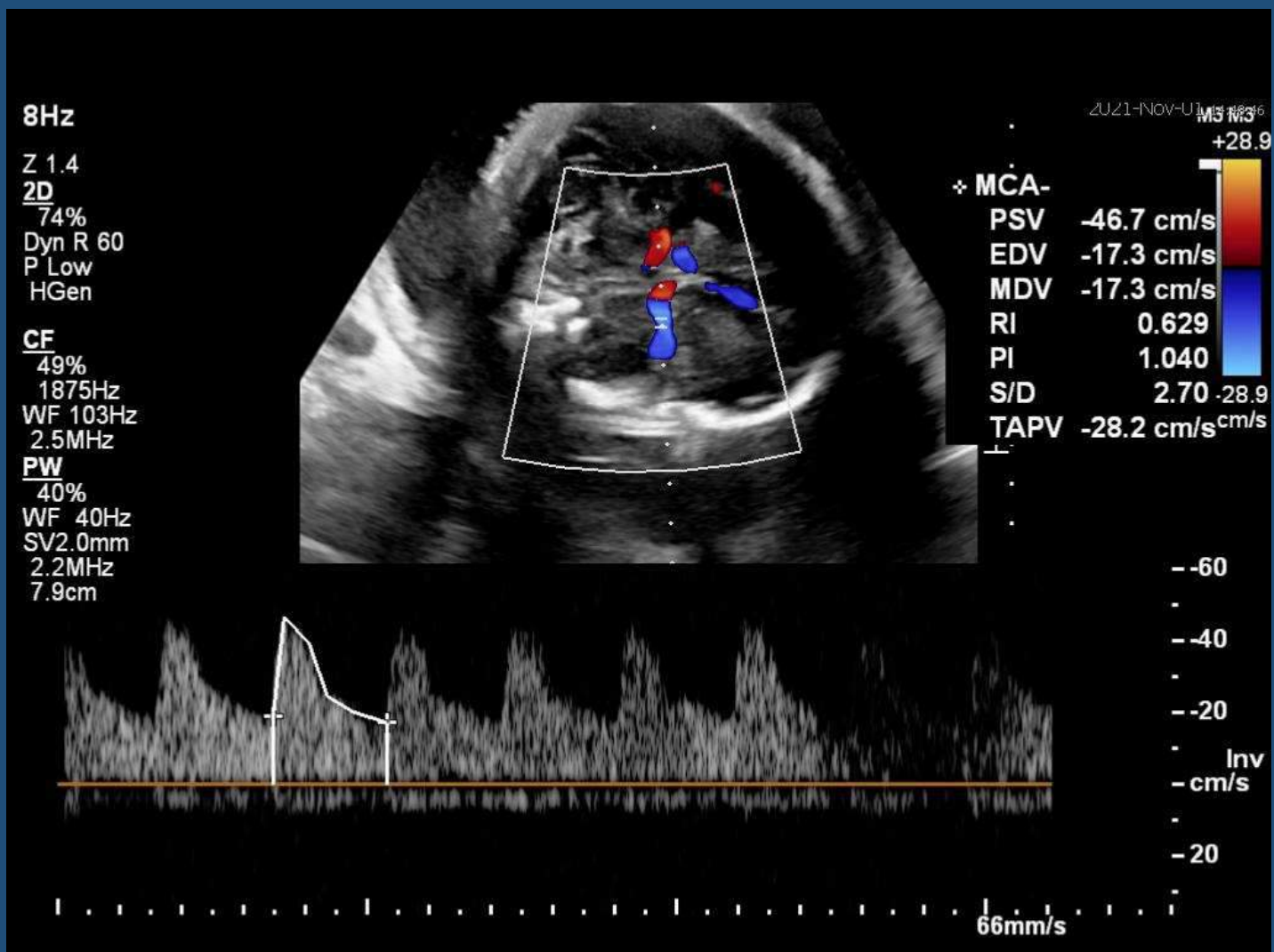














33 weeks 3 days

Decreased fetal
movement

BPP 6/10

IMPACT
delivery

Blue rubber bleb nevus syndrome



Test Requested: Somatic Overgrowth & Vascular Malformations Panel (v3)

Sample(s) Analyzed:

- 1: Genomic DNA isolated from blood
- 2: Genomic DNA isolated from Left foot frozen tissue sample
- 3: Genomic DNA isolated from Left foot FFPE sample

Result Summary: POSITIVE Two Somatic Pathogenic Variants in *TEK*

Identification of Variant(s):

Sample	Gene	Nucleotide Position	Amino Acid Position	Variant Frequency	Classification
2	TEK	c.2690A>T	p.Tyr897Phe	10.3 - 12.5%	Pathogenic
2	TEK	c.2753G>A	p.Arg918His	9.9 - 13.7%	Pathogenic

22q11
del



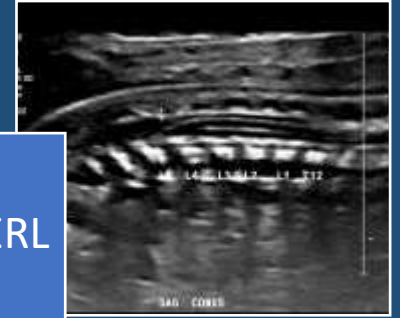
T21



Noonan



VACTERL



T18/13



Kabuki



CHARGE



Cri du
chat



Times we can help provide
the most compassionate
care...





What is CFDT CARE?

Complex Antenatal Resuscitation Evaluation

Built as a multidisciplinary
support system

For families with a potentially
life-threatening fetal
diagnosis



CFDT COMPLEX ANTENATAL RESUSCITATION EVALUATION (CARE) CONSULT

Who:



- Concern for life-threatening diagnosis at any point in the pregnancy (+/- a confirmed genetic diagnosis)
- Concern for immediate neonatal demise
- Parents express interest in “NON-INTERVENTION” OR perinatal palliative

What:



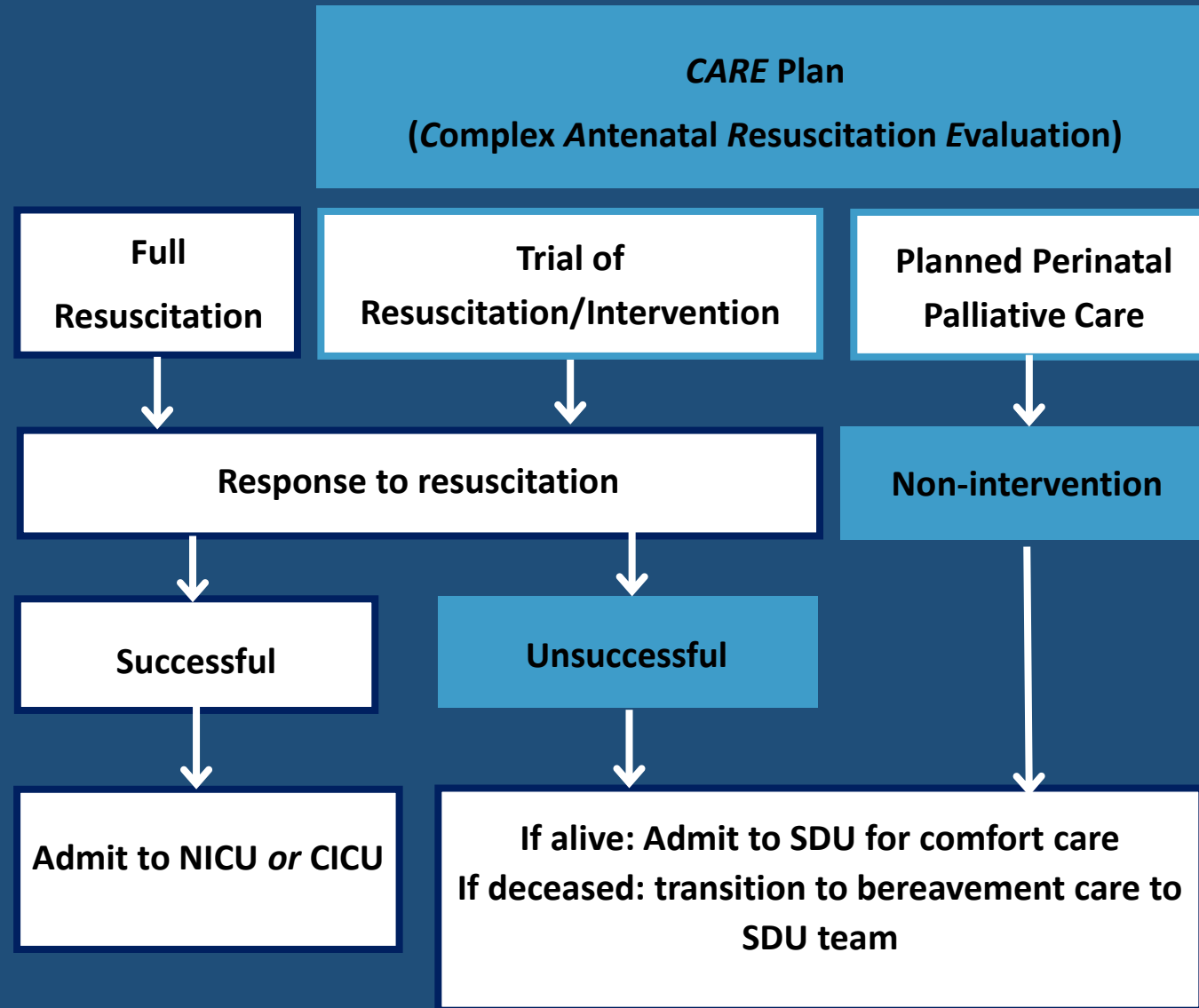
- Multidisciplinary collaborative consult
- Ongoing planning and support in pregnancy, delivery room and postpartum

Why:

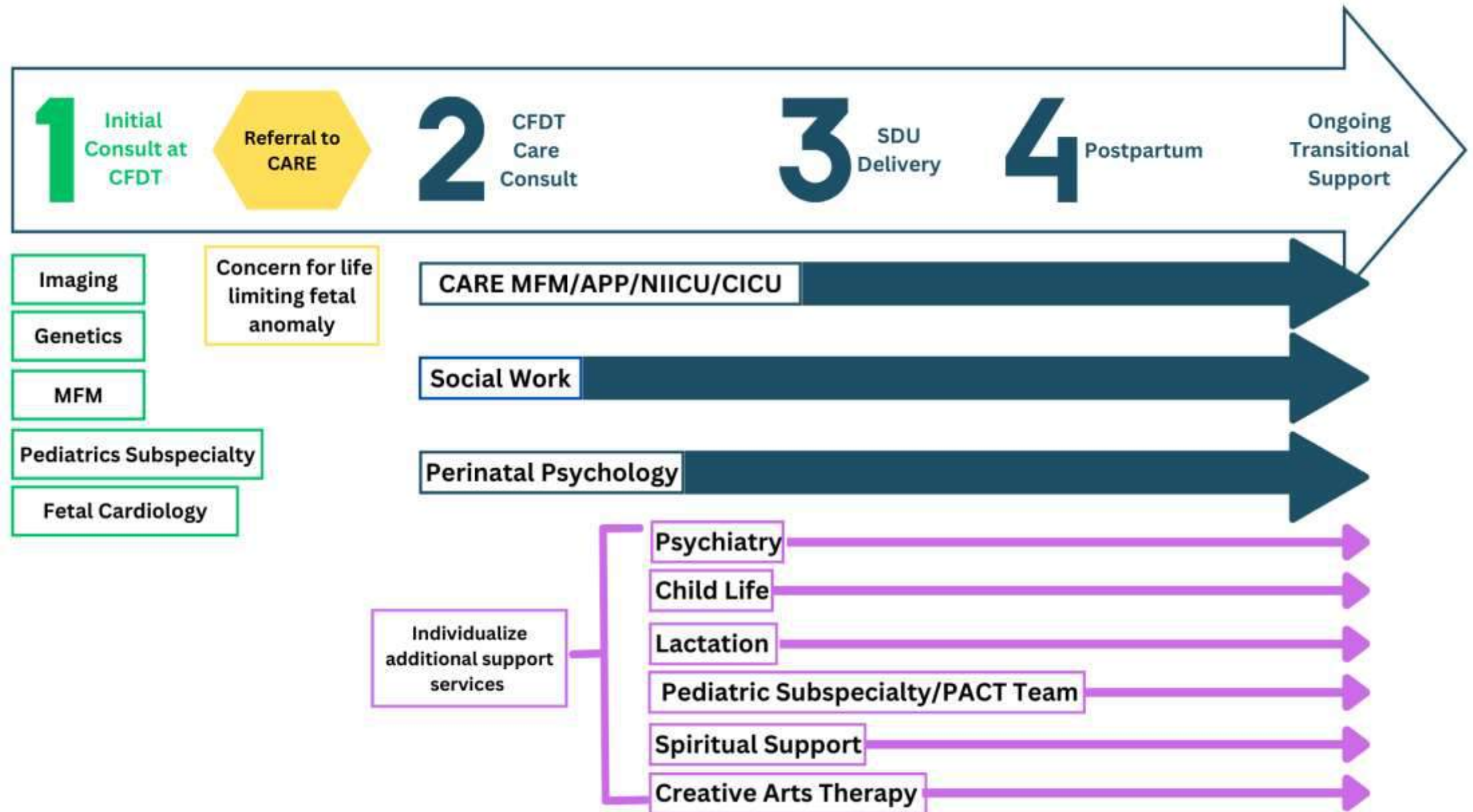


- Individualized goals and expectations
- Pregnancy planning: imaging, monitoring, mode and timing of delivery
- Neonatal routes of care
- Recommendations
- Support

Neonatal Care ALGORITHM



CFDT Care Consult Workflow





Thank you!

