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Updates in the Screening and Diagnosis of Congenital Heart Disease

Prashant Minocha, MD

Pediatric Cardiologist, Non-invasive imager

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No Disclosures

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Outline

- Fetal screening and diagnosis of Congenital Heart Disease
- A new frontier- Fetal Cardiac MRI
- Updated pulse oximetry screening algorithm for neonates
- Sport Pre-Participation Cardiac Clearance

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Background & Importance

- **Prevalence & Mortality**
  - Congenital heart defects impact ~8 per 1,000 live births; critical forms (CCHD) affect 2–4 per 1,000 and are a leading cause of infant death.

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Fetal screening and Diagnosis of Congenital Heart Disease

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Fetal Screening for CHD

- The current standard of care for screening for congenital heart disease (CHD) by obstetricians in the United States is second-trimester obstetric ultrasound with dedicated cardiac views as part of the routine fetal anatomic survey.

- This screening includes, at minimum, the **four-chamber view of the heart, as well as views of the left and right ventricular outflow tracts, and, when technically feasible, the three-vessel and three-vessel-and-trachea views.**

- If the screening ultrasound identifies a suspected cardiac abnormality **referral for fetal echocardiography is indicated**.

- The prenatal detection rate of critical congenital heart defects ranges from 13% to 87% across different countries, with a median detection rate of 50%.

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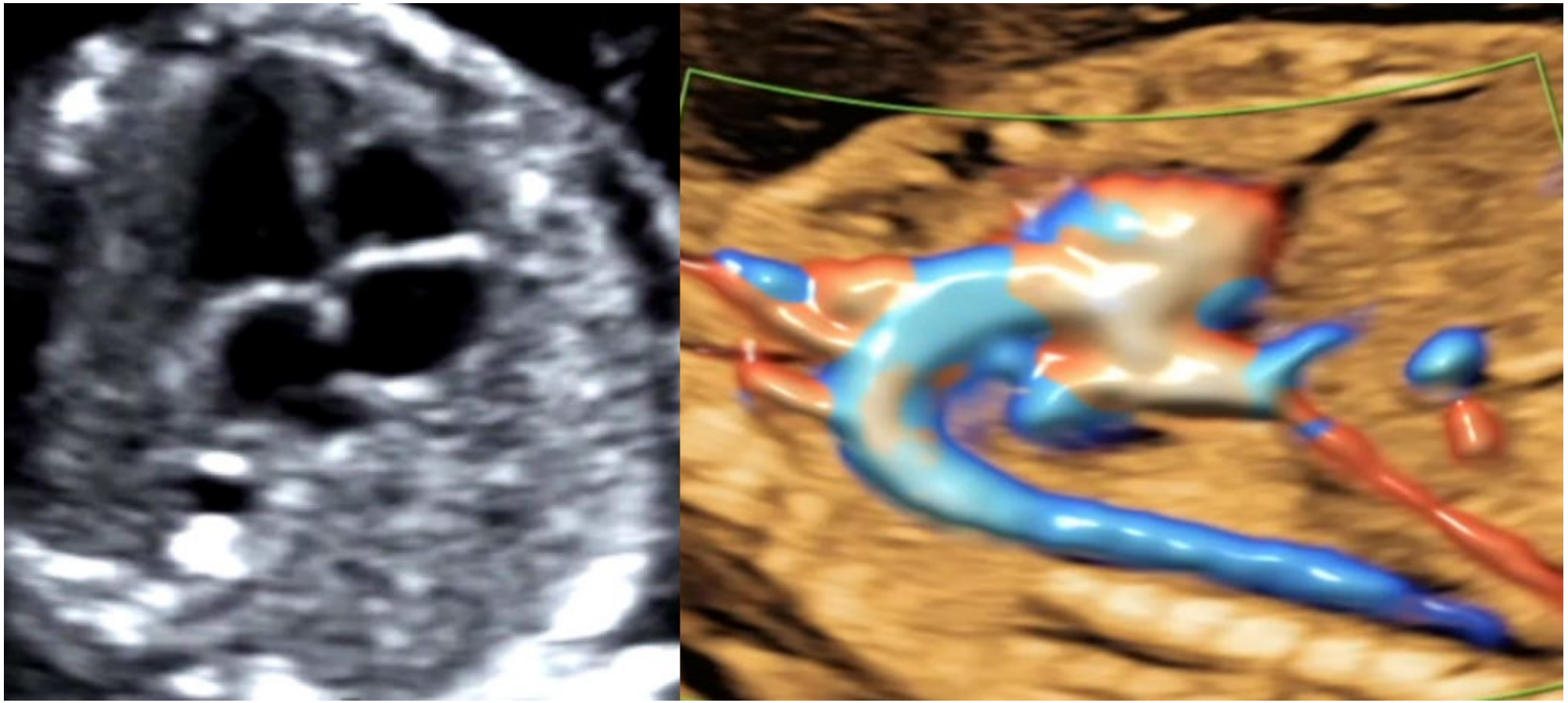
Fetal Echo



What is a fetal echo

- A detailed ultrasound examination of the fetal heart
- Typically performed after 18 weeks gestation
- Aimed at diagnosing **congenital heart defects (CHDs)** and **rhythm abnormalities**

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Maternal Risk Factors

- Pre-existing diabetes mellitus (especially poorly controlled)
- Autoimmune disorders (e.g., SLE, Sjögren's – SSA/SSB antibodies)
- Teratogen exposure (e.g., lithium, retinoic acid, alcohol)
- In vitro fertilization (IVF)
- Maternal infections (e.g., rubella, CMV)

Fetal Risk Factors

- Increased nuchal translucency (>3.5 mm)
- Suspected or diagnosed chromosomal abnormality (e.g., Trisomy 21)
- Extracardiac anomalies (renal, CNS, etc.)
- Non-immune hydrops fetalis
- Fetal arrhythmias
- Suspicious findings on obstetric ultrasound (e.g., cardiomegaly, pericardial effusion)

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Family History

- Previous child with CHD
- Parental CHD or genetic syndromes
- Family history of inherited arrhythmias (e.g., long QT syndrome)

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Goals of Fetal Echocardiography

- Assess cardiac anatomy and function
- Detect congenital heart defects
- Evaluate heart rhythm and conduction
- Guide perinatal management and delivery planning

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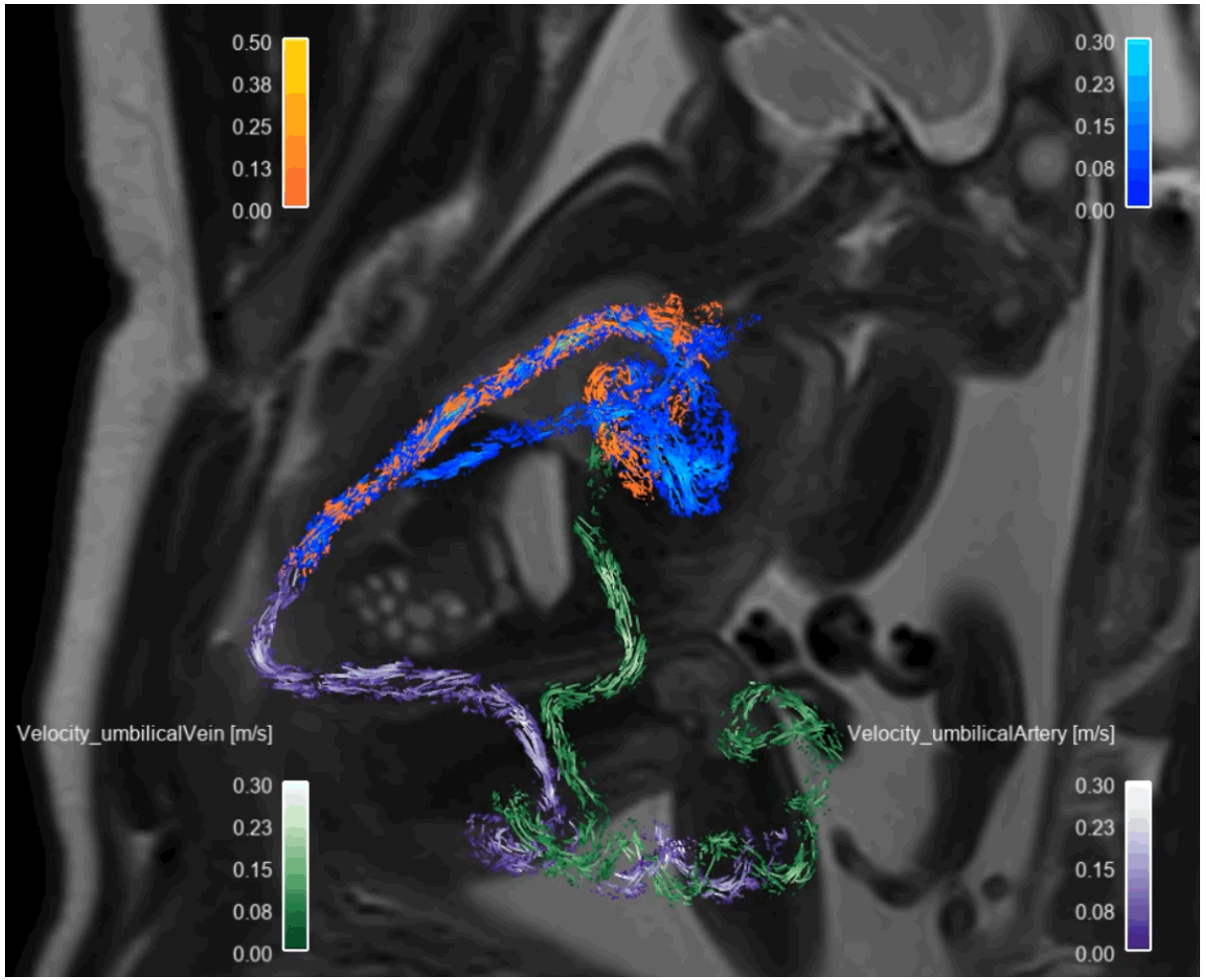
Fetal Cardiac MRI



Cardiac MRI

- Cardiovascular anatomical MRI is an essential diagnostic tool in the evaluation of the heart postnatally.
- It is non-invasive, provides excellent soft tissue contrast and has the ability to perform multi-planar imaging in any orientation.
- Dynamic MRI of the heart necessitate synchronization of image acquisition with the cardiac phase, which is most often facilitated by an electrocardiogram (ECG) signal.

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Fetal CMR

- New FDA-approved MRI-compatible Doppler ultrasound probe that produces a gating signal from the fetal heart introduced into mainstream clinical practice in 2021.
- The Doppler ultrasound probe detects blood flow through the fetal heart, which can be used to trigger MRI data acquisition and produce dynamic images of the fetal heart.
- In situations where the fetus is diagnosed with congenital heart disease (CHD) through routine care (feta echo) the patients may be referred to our MRI program for additional fetal body and or cardiovascular assessment.

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Normative Volumes for Ventricular Size

Reference Values for Fetal Cardiac Dimensions, Volumes, Ventricular Function and Left Ventricular Longitudinal Strain Using Doppler Ultrasound Gated Cardiac Magnetic Resonance Imaging in Healthy Third Trimester Fetuses

Prashant K. Minocha, MD,[1] Erin K. Englund, PhD,[2] Richard M. Friesen, MD,[1] Takashi Fujiwara, PhD,[2] Sarah A. Smith, MHA,[2] Mariana L. Meyers, MD,[2]

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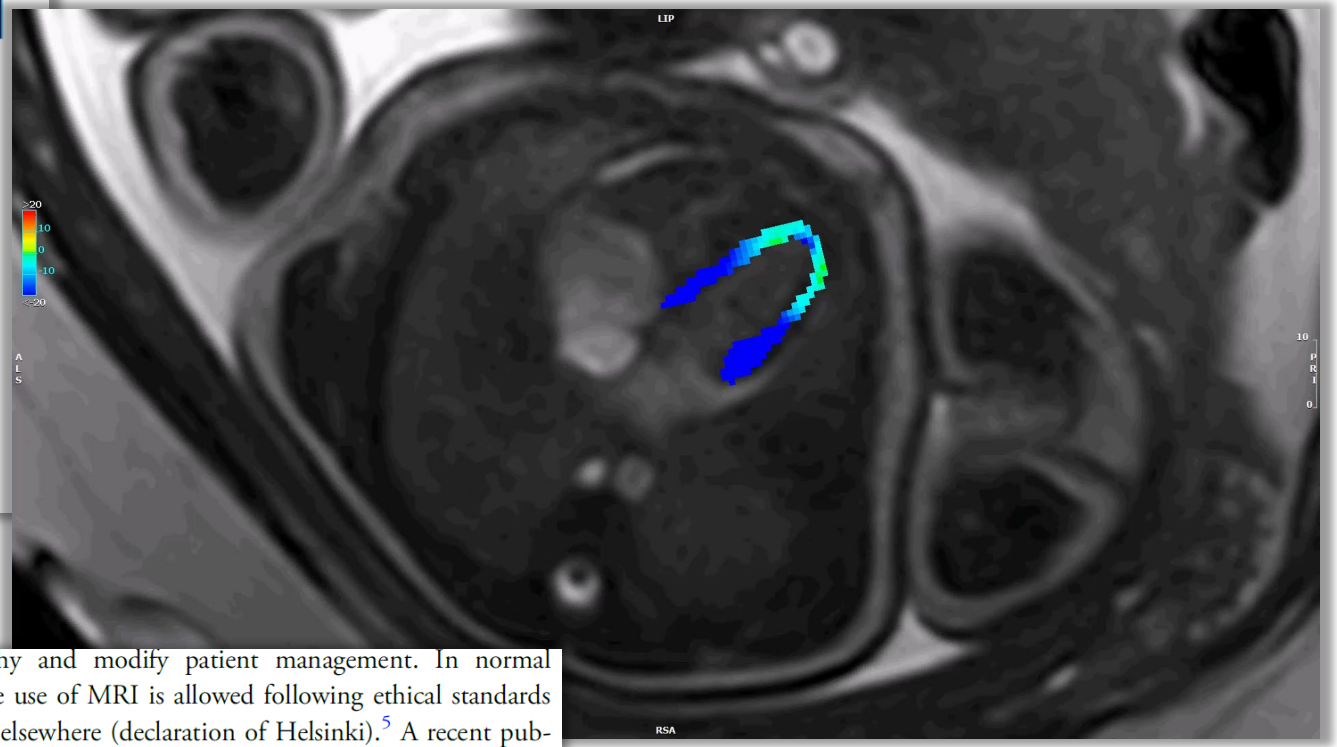
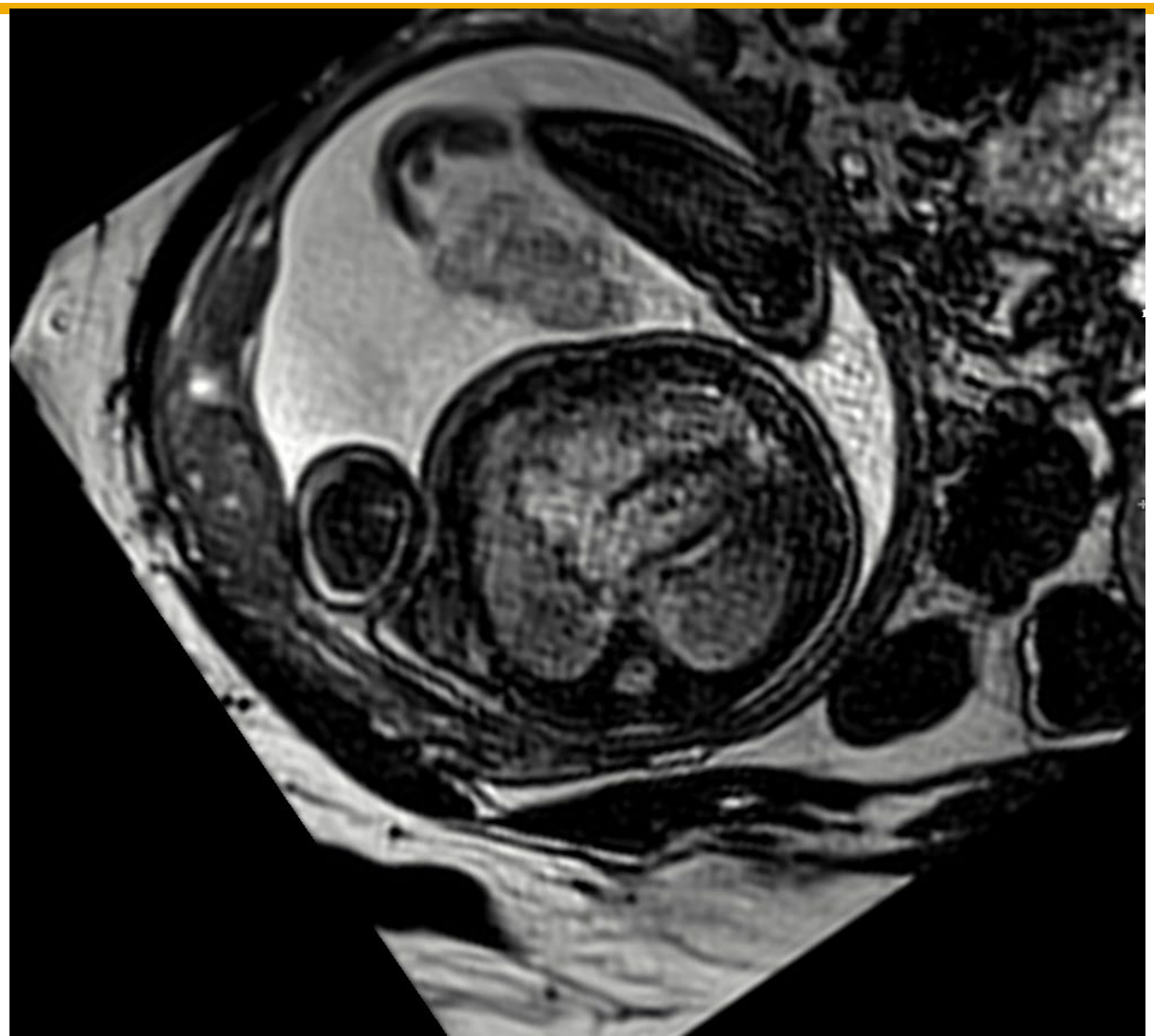


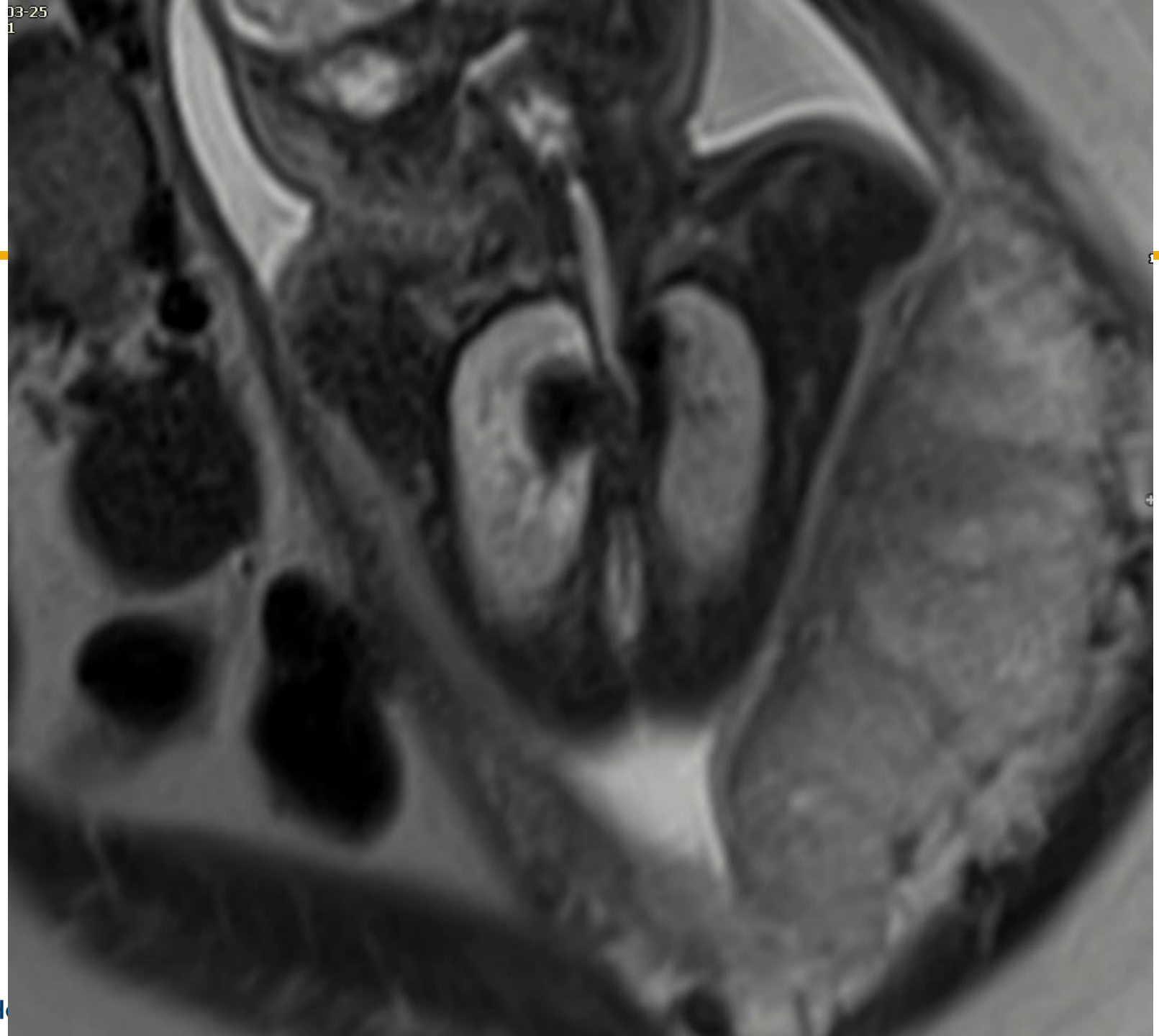
EDITORIAL

Editorial for "Reference Values for Fetal Cardiac Dimensions, Volumes, Ventricular Function and Left Ventricular Longitudinal Strain Using Doppler Ultrasound Gated Cardiac Magnetic Resonance Imaging in Healthy Third Trimester Fetuses"

cardiography and modify patient management. In normal fetuses, the use of MRI is allowed following ethical standards as defined elsewhere (declaration of Helsinki).[5] A recent publication by Barker et al in the Journal of Magnetic Resonance Imaging (JMRI) is one of the first publications to present normative cardiac MRI data for a healthy fetal population, eg, cardiac dimensions, volumes, and function.[6] This is an important advance, as establishing a range of normal structural and functional values allows more for effective diagnosis.

Tetralogy Absent Pulmonary Valve





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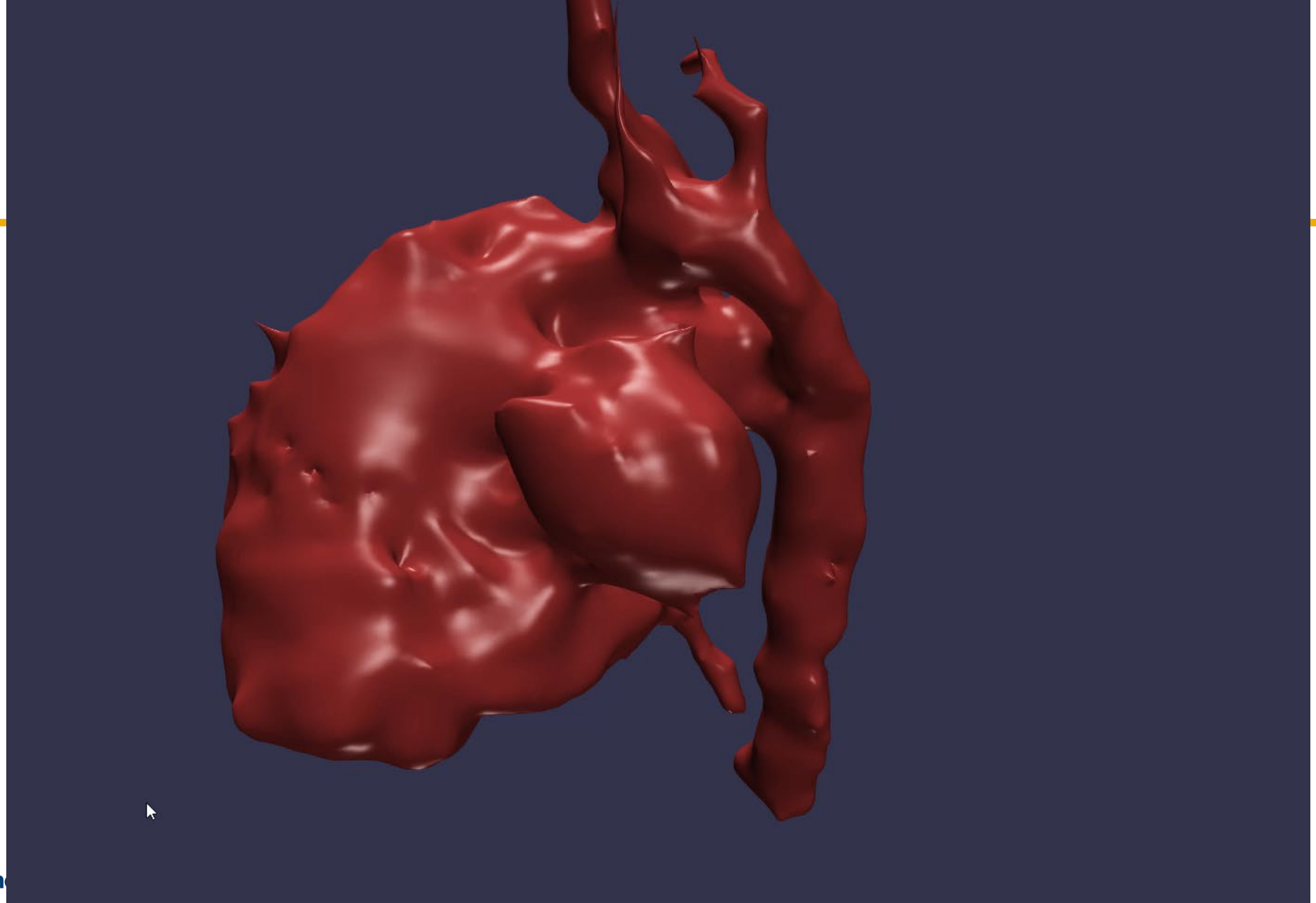
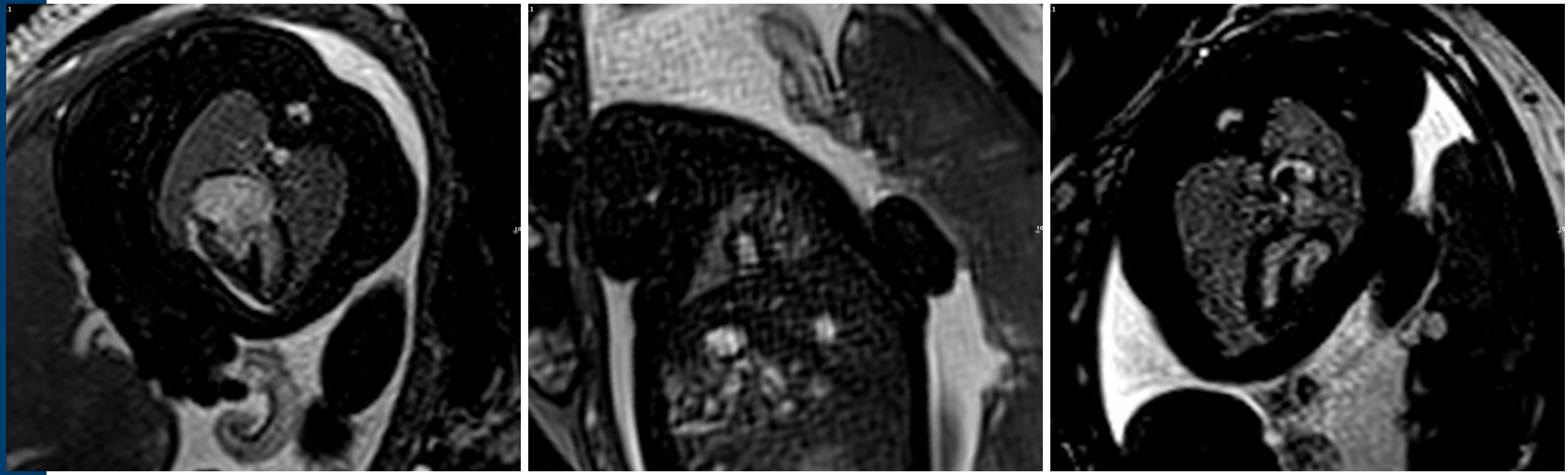
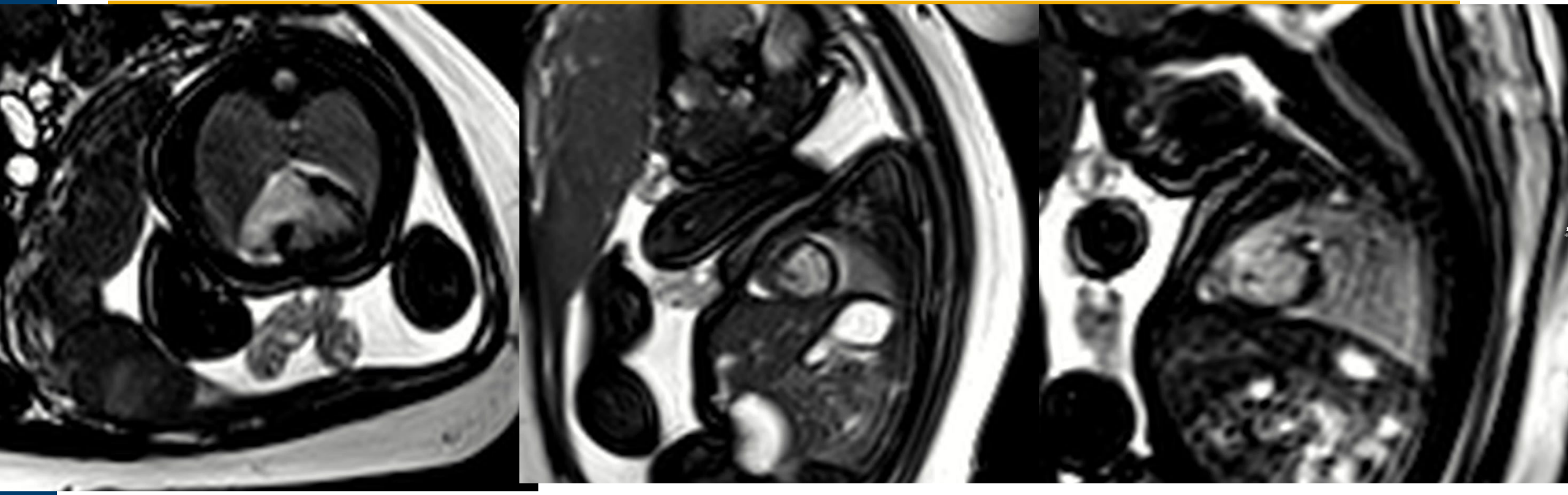


Unbalanced, RV Dominant AV canal



HLHS (Mitral and Aortic Atresia)



Benefits of Gated Fetal CMR

- Ochsner is the first and only institution in Louisiana to offer this service.
- Can help clarify diagnoses in fetuses with CHD who have had a fetal echo
- Assist with counseling and delivery planning.
- This will offer opportunities for research within cardiology, radiology and MFM

Updated Newborn Pulse Oximetry Screening Algorithm

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Universal CCHD screening in the USA

- Universal pulse oximetry screening was added to the Recommended Uniform Screening Panel in 2011 and implemented across all U.S. states by 2018
- Screening has led to a ~33% reduction in early infant mortality and decreased emergency hospitalizations for CCHD

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Pulse Oximetry Screening Protocol

Timing
Recommended ≥24 hours after birth or closest to discharge; can align with newborn hearing screen.

Measurement Sites
Pre-ductal: Right hand
Post-ductal: Either foot

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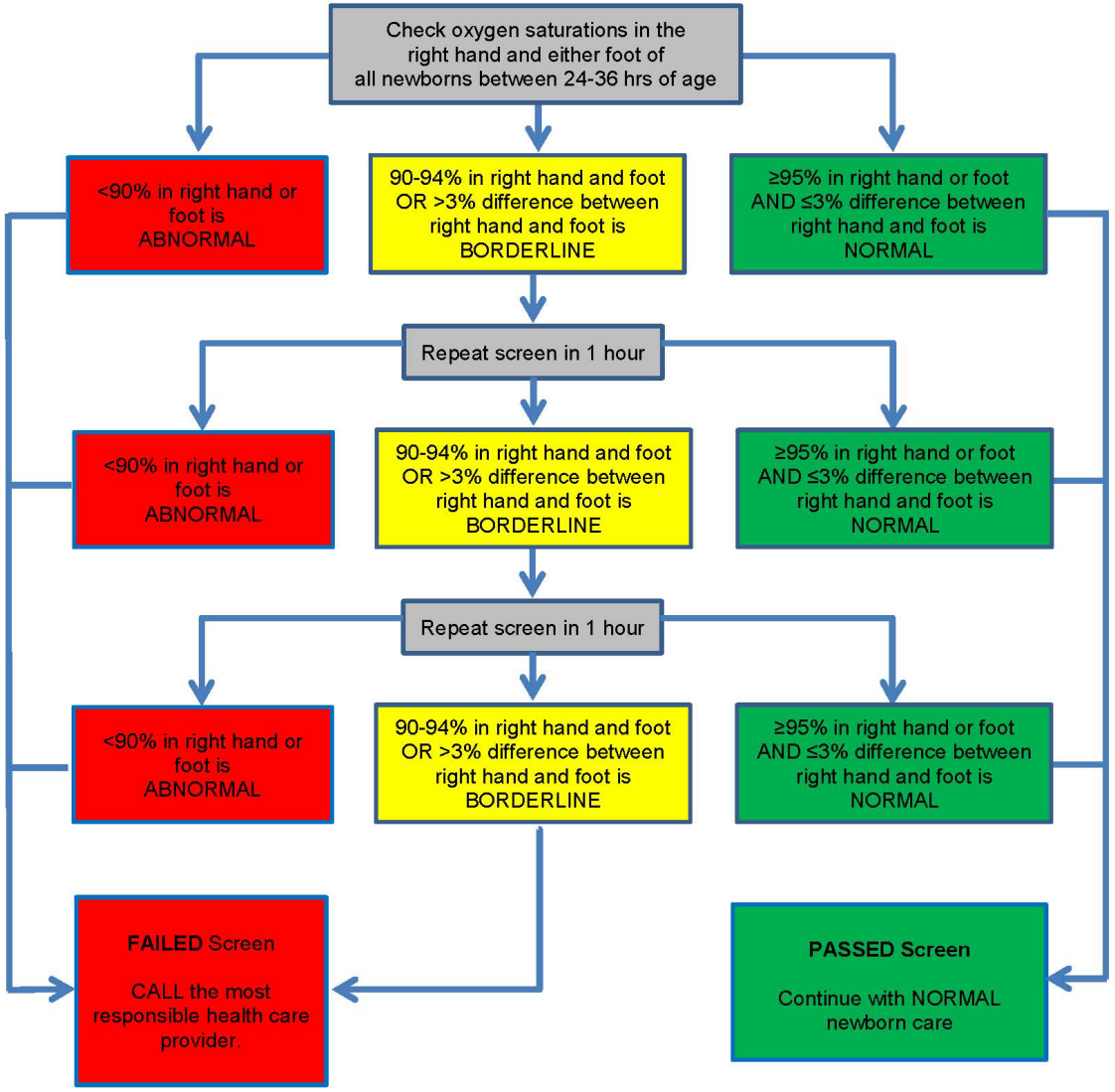
Old Algorithm: Implemented in 2011



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New AAP Algorithm Updated in 2025

Pass Criteria
≥ 95% SpO_2 in both pre- **AND** post-ductal readings (previously required either)

Retest Policy
Only **one** repeat measurement if initial result is below threshold or has >3% hand-foot difference (instead of two).

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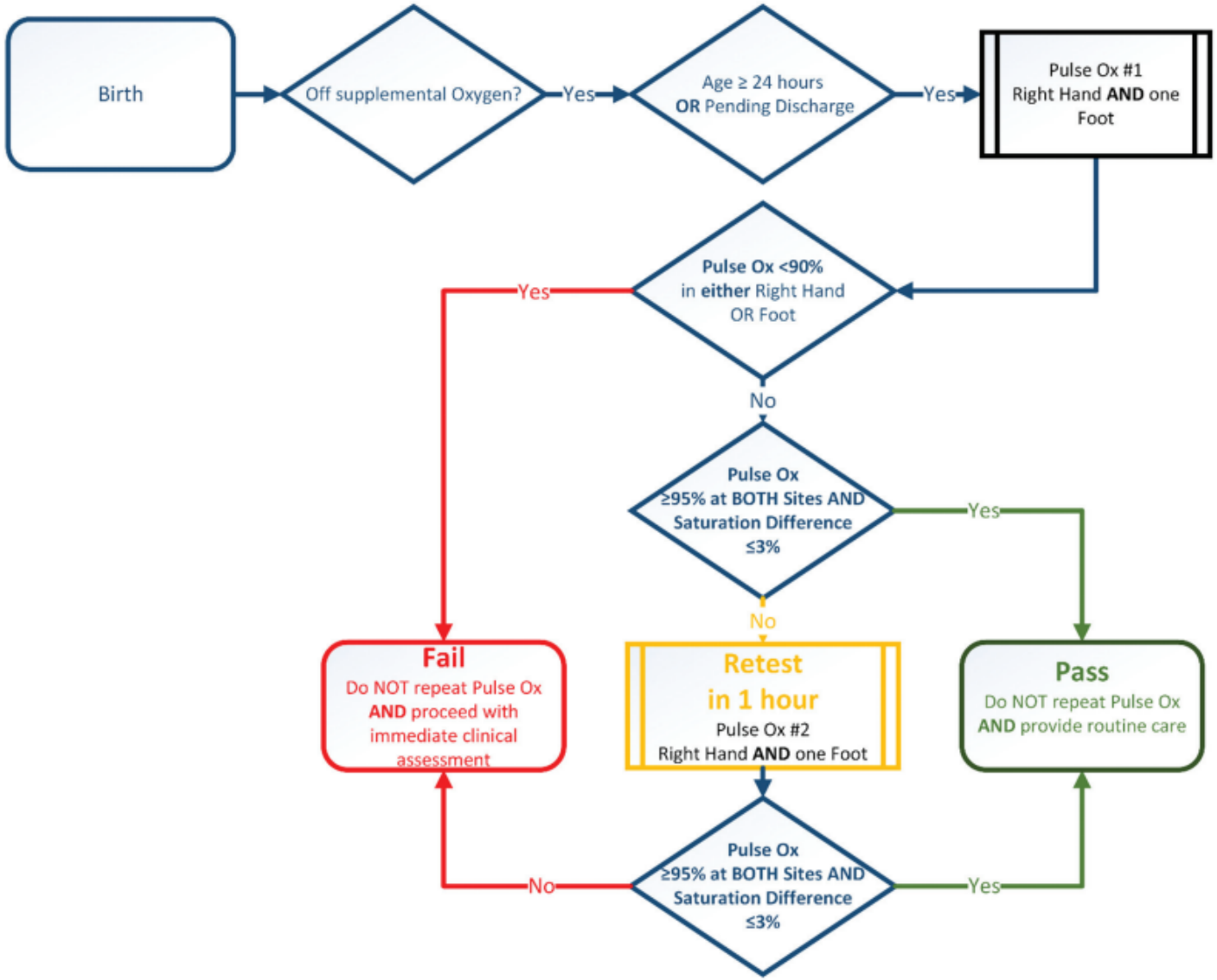

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Screening Outcomes

Pass
≥95% on both sites and ≤3% difference → Screening ends (with note that rarer CCHD may still be missed).

Indeterminate
Either site 90–94% or >3% difference → Retest once.
If still indeterminate → Fail.

Fail
Any reading <90% or persistent SpO_2 <95% or >3% difference post-retest → Further evaluation (echocardiogram, infection/respiratory check)

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Implications & Benefits

Simplified Decisions

Clear pass/fail thresholds reduce confusion.

Faster detection

One retest shortens time to diagnosis without meaningful increase in false positives.

Broader detection

Can sometimes identify non-cardiac hypoxic conditions like sepsis or pneumonia

ECG in Newborns

- Normal ECG parameters in neonates are highly dynamic and age-dependent, reflecting rapid physiological changes during the transition from fetal to neonatal circulation. This results in significant interindividual variability in ECG findings, making interpretation challenging and reducing sensitivity for detecting congenital heart disease or arrhythmias in the immediate newborn period.

- Adding ECG to the screening pathway for CHD does not increase diagnostic yield.

- Frequently generates unnecessary additional evaluation.

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Screening for heart disease in Children and Adolescents

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Epidemiology of Sudden Cardiac death in Athletes

- Incidence: ~1 in 50,000 to 1 in 80,000 athletes/year
- Higher in males, Black athletes, and certain sports (basketball, football)
- Common causes:
  - Hypertrophic cardiomyopathy
  - Arrhythmogenic right ventricular cardiomyopathy
  - Congenital coronary anomalies
  - Long QT syndrome

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Pre-Participation Evaluation (PPE)

Standard approach includes:

- Personal history
- Family history
- Physical examination
- Additional testing (e.g., ECG) based on risk

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AHA 14-Point Questionnaire for Children

AHA's 14-Point PPE
Personal history
1. Chest pain, discomfort, tightness, or pressure related to exertion
2. Unexplained syncope or near-syncope not felt to be vasovagal or neurocardiogenic in origin
3. Excessive and unexplained dyspnea or fatigue or palpitations associated with exercise
4. Previous recognition of a heart murmur
5. Elevated systemic blood pressure
6. Previous restriction from participation in sports
7. Previous testing for the heart, ordered by a physician
8. Family history of premature death (sudden and unexpected or otherwise) before 50 y of age attributable to heart disease in ≥1 relative
9. Disability from heart disease in close relative <50 y of age
10. Hypertrophic or dilated cardiomyopathy, LQTS, or other ion channelopathies, Marfan syndrome, or clinically significant arrhythmias; specific knowledge of genetic cardiac conditions in family members
Physical Examination
11. Heart murmur, not felt to be innocent
12. Femoral pulses to exclude aortic coarctation
13. Physical stigmata of Marfan syndrome
14. Brachial artery blood pressure (sitting position), preferably taken in both arms

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History – Red Flags

- Chest pain with exertion
- Syncope or near-syncope
- Palpitations
- Excess fatigue
- Family history of SCD, cardiomyopathy, or inherited arrhythmia

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Physical Exam

- Blood pressure check
- Cardiac auscultation (supine, standing, Valsalva)
- Marfan features (tall, arm span > height, lens dislocation)
- Femoral pulses (coarctation screening)

Role of ECG in Screening

The American Heart Association notes that universal ECG screening is not recommended for the general pediatric population due to limited sensitivity and specificity, as well as the potential for false positives and negatives

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American vs. European Guidelines

Feature	AHA/ACC (USA)	ESC (Europe)
ECG required?	No	Yes
Emphasis	Cost-effectiveness, targeted screening	Broad population screening
ECG criteria	Conservative	Sensitive

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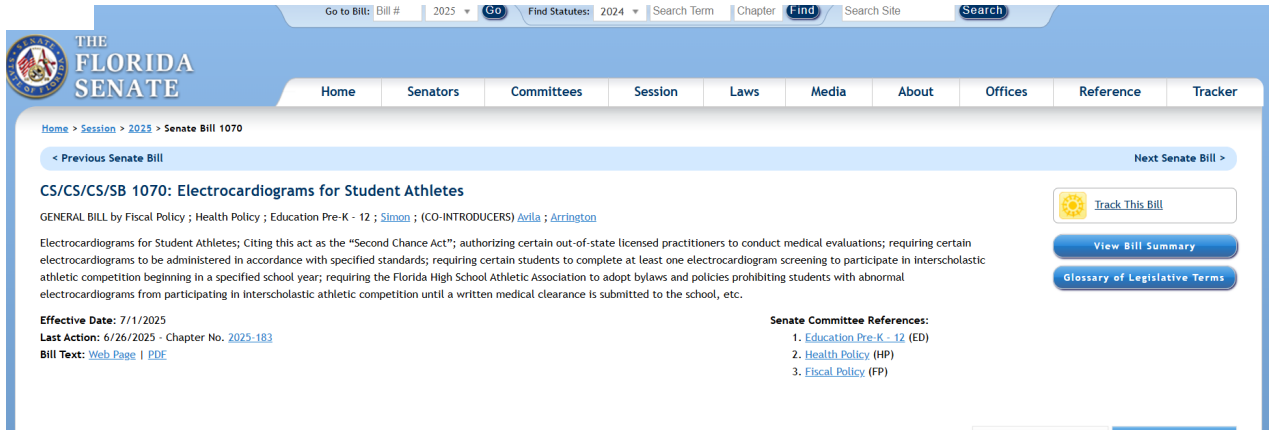
Education

Florida is the first state to require student-athletes get life-saving EKGs

Central Florida Public Media | By Danielle Prieur
Published June 29, 2025 at 7:03 AM EDT



Automated external defibrillator (AED) in Schools
Secondary prevention

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Project Adam

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When to Refer to Cardiology

- Abnormal history or exam
- Positive ECG findings (e.g., T-wave inversions, delta waves)
- Murmurs suspicious for obstructive lesions
- Syncope or chest pain on exertion

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Summary

- Screening and diagnosis of CHD starts in fetal life
- The new neonatal CCHD screen will allow for simplified decisions and faster detection of CHD
- Cardiac screening saves lives
- Careful history and exam are essential
- Collaboration between primary providers and cardiologists is crucial

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Thank You

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Questions?

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