



Society for
Maternal • Fetal
Medicine

DETAILED FIRST TRIMESTER ULTRASOUND

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TEXAS CHILDREN'S HOSPITAL PAVILION FOR WOMEN

HOUSTON, TX



Baylor
College of
Medicine



Pavilion
for Women



Disclosures

Topic author &
reviewer for UpToDate



Here's the problem

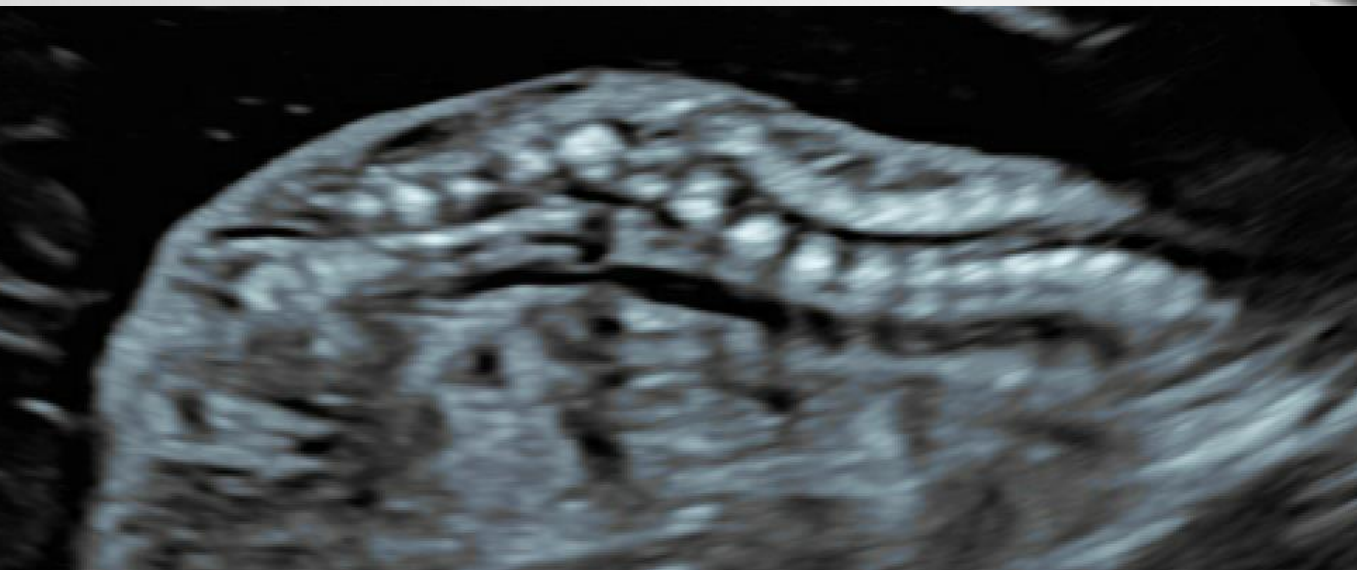
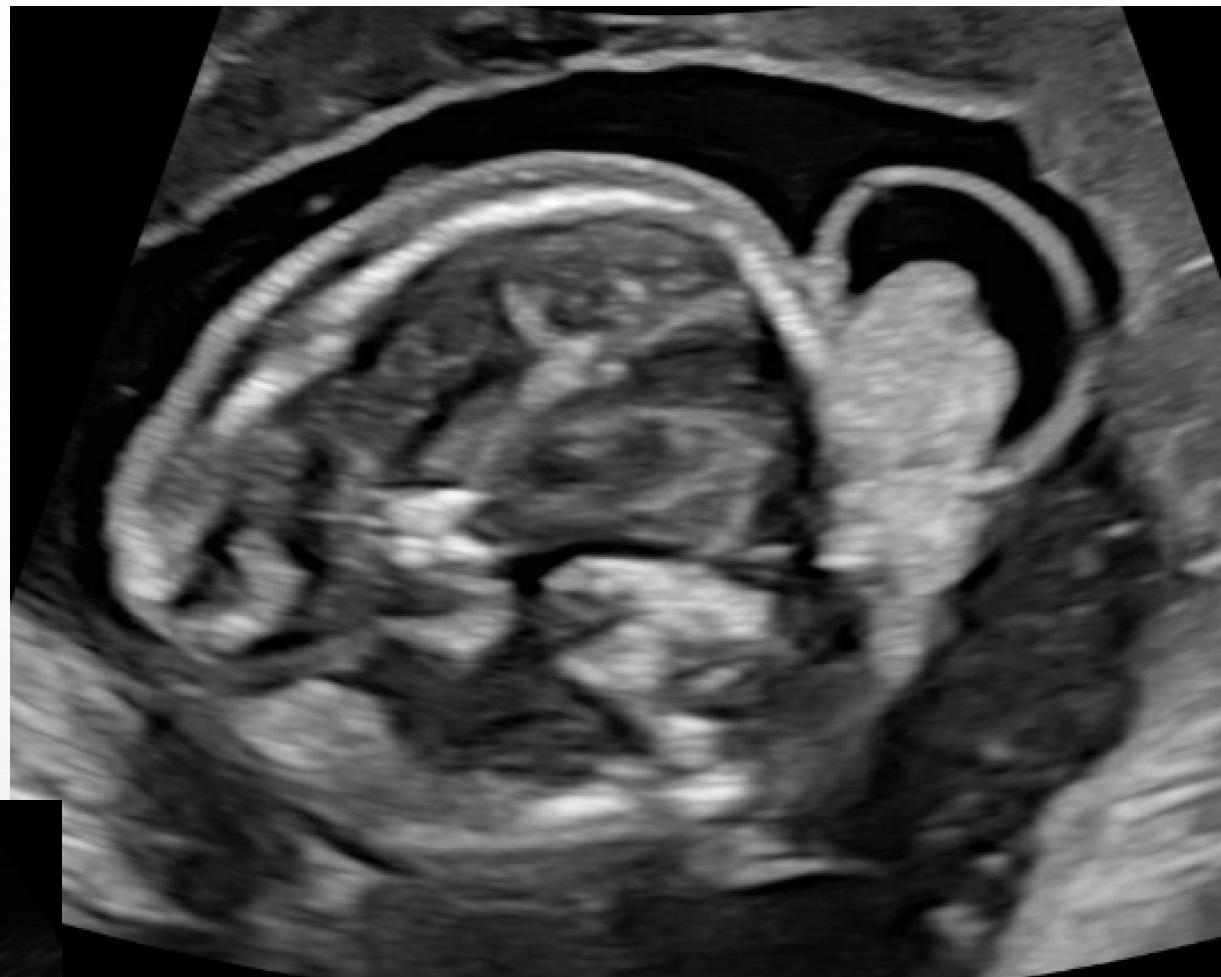
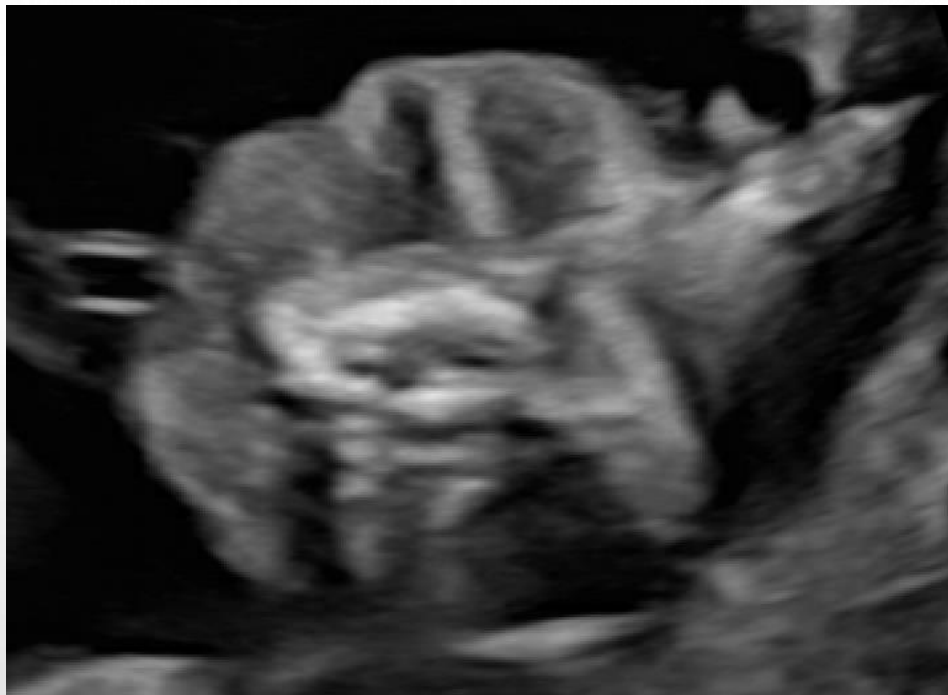
--27 yo G1P0 at 19+0 weeks

--BMI=20.67

--**MSAFP=7.68 MoM**

--Is our baby okay?

--Both parents are in healthcare



HOW DID THIS HAPPEN?

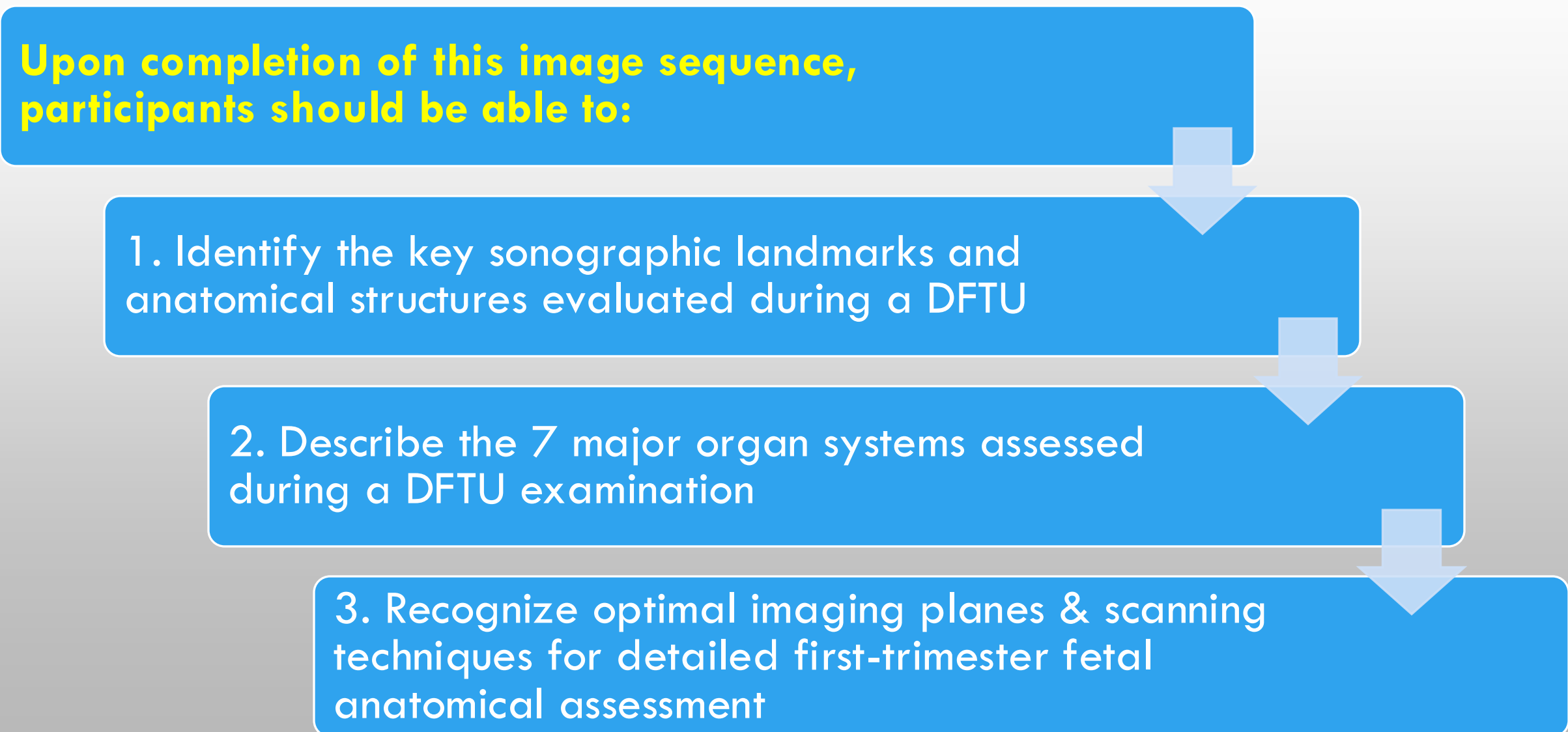
We had a
normal 7-week
ultrasound in our
doctor's office

Our NIPT was
normal

Could we have
seen this earlier?

OBJECTIVES

Upon completion of this image sequence, participants should be able to:



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graph TD; A[Upon completion of this image sequence, participants should be able to:] --> B[1. Identify the key sonographic landmarks and anatomical structures evaluated during a DFTU]; B --> C[2. Describe the 7 major organ systems assessed during a DFTU examination]; C --> D[3. Recognize optimal imaging planes & scanning techniques for detailed first-trimester fetal anatomical assessment];
```

1. Identify the key sonographic landmarks and anatomical structures evaluated during a DFTU

2. Describe the 7 major organ systems assessed during a DFTU examination

3. Recognize optimal imaging planes & scanning techniques for detailed first-trimester fetal anatomical assessment

AIUM 2021: 1ST TRIMESTER PRACTICE PARAMETER

AIUM Practice Parameter
for the Performance of Detailed
Diagnostic Obstetric Ultrasound
Examinations Between 12 Weeks
0 Days and 13 Weeks 6 Days

First trimester obstetric ultrasound



In most cases, transabdominal ultrasound is sufficient



TV approach can optimize visualization by providing a higher resolution & recommended if TA sonography is suboptimal or if an anomaly is suspected



Assure adequate magnification, appropriate depth, sector size & correct focal zone placement at the area of interest



Power & color Doppler imaging may used to complement grayscale

First trimester obstetric ultrasound

Aneuploidy assessment

Accurate dating

Optimal assessment of
amnionicity &
chorionicity in multiple
gestations

Opportunity to
evaluate structural
assessment of the fetus

Many anomalies
historically diagnosed in
the 2nd trimester can be
identified in the latter
part of the 1st trimester

1ST TRIMESTER IMAGING - HISTORY



- Nuchal translucency (NT) screening - Nicolaides, 1992
- Combined first-trimester screen (serum analytes & NT) - mid-2000's
- Cell-free DNA screening for common autosomal trisomies - Oct 2011
 - ✓ Rapid adoption of cf-DNA
 - ✓ Challenges associated with NT measurement & need for yearly certification



- ✓ Use of **cf-DNA alone without an early anatomic survey** will result in **many anomalies** that are typically detected in the 1st trimester **NOT** being identified



1ST TRIMESTER SCREENING



- ACR-ACOG-AIUM-SMFM-SRU Practice Parameter for the Performance of Standard Diagnostic Obstetric Ultrasound Examination
 - Minimal key elements required for fetal anatomic assessment
 - Can be performed by a variety of clinicians in a range of practice settings
- AIUM Practice Parameter for the Performance of Detailed Diagnostic Obstetric Ultrasound Examinations Between 12 Weeks 0 Days and 13 Weeks 6 Days
 - Includes extended evaluation of the fetal heart (4-ch view, outflow tracts, color Doppler)
 - Performed by sonographers/sonologists with extensive obstetric imaging experience

ACR-ACOG-AIUM-SMFM-SRU Practice Parameter for the performance of stand diagnostic obstetrical ultrasound, 2023.

AIUM Practice Parameter for the Performance of Detailed Diagnostic Obstetric Ultrasound Examinations Between 12Weeks 0 Days and 13 Weeks 6 Days. J Ultrasound Med, 2021;40: E1-E16.

Bromley B, Platt L. First trimester ultrasound screening in Routine Obstetric Practice. Obstet Gynecol 2024;00:1-15

ISUOG Practice Guidelines: Performance of 11-14-week ultrasound scan. Ultrasound Obstet Gynecol 2023;61:127-145

DETAILED FIRST-TRIMESTER ULTRASOUND (DFTU) EXAM



DFTU

- **“Indication-driven”**
- For patients at increased risk for fetal or placental abnormalities that are potentially detectable between 12 weeks 0 days and 13 weeks 6 days’ gestation.
- Requires advanced training, knowledge & skills, and ability to effectively communicate findings to the patient and referring physician
- Performance is rare outside of referral practices

AIUM Practice Parameter for the Performance of Detailed Diagnostic Obstetric Ultrasound Examinations Between 12 Weeks 0 Days and 13 Weeks 6 Days. J Ultrasound Med, 2021,40: E1-E16.

Bromley B, Platt L. First trimester ultrasound screening in Routine Obstetric Practice. Obstet Gynecol 2024;00:1-1545

DFTU: INDICATIONS



- Previous fetus or child with a congenital, genetic, or chromosomal anomaly
- Known or suspected fetal abnormality detected by ultrasound in current pregnancy
- Fetus at increased risk for a congenital anomaly
 - ✓ 35 years or older at delivery; Pregestational diabetes; Pregnancy conceived by IVF; Multiple gestation
 - ✓ Teratogen exposure; Enlarged nuchal translucency.
 - ✓ Positive screening test results for aneuploidy (cf DNA, serum-only or combined 1st-trimester screening)
- ✓ Other conditions possibly affecting the pregnancy/fetus
 - ✓ Maternal BMI of 30 kg/m² or higher
 - ✓ Placental implantation covering the internal cervical os, under a cesarean scar site, or cesarean scar pregnancy diagnosed in index gestation



Detailed First Trimester

[Home](#) / Practice Topics

Practice Topics

From specialty areas to research arenas to cutting-edge medical procedures, ultrasound impacts the practice and delivery of medical care.

Explore the topics below as each contains medical ultrasound information, peer-reviewed research results, helpful resources, and educational components.

Artificial Intelligence

Basic Science and Instrumentation - AIUM

Cardiovascular Ultrasound

Contrast-Enhanced Ultrasound

Dermatology

Elastography

Fetal Echocardiography

General and Abdominal Ultrasound

Gynecologic Ultrasound

High Frequency Clinical and Preclinical Imaging

Interventional-Intraoperative Ultrasound

Musculoskeletal Ultrasound

Neurosonology

Obstetric Ultrasound

Pediatric Ultrasound

Point-of-Care Ultrasound

Sonography

Therapeutic Ultrasound

Ultrasound in Global Health

Ultrasound in Medical Education

Urology

Training **Guidelines** [View More »](#)

Training Guidelines for Physicians Who Perform and/or Interpret Diagnostic Ultrasound Examinations

Training Guideline for Licensed Medical Providers Including Advanced Clinical Providers (PAs, NPs, CNMs/CMs), DPTs, and DCs Performing and/or Interpreting Diagnostic Ultrasound Examinations

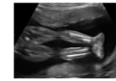
Training Guidelines for Licensed Medical Providers Who Perform and Interpret Ultrasound-Guided Procedures

Practice **Parameters** [View More »](#)



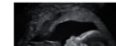
Obstetric (Detailed 1st Trimester) * (2020)

[Article](#) | [Image Library](#)



Obstetric (Detailed 2nd/3rd Trimester Anatomy) * (2019)

[Article](#) | [Image Library](#)



Obstetric (Standard) * (2024)

Practice **Parameters** [View More »](#)



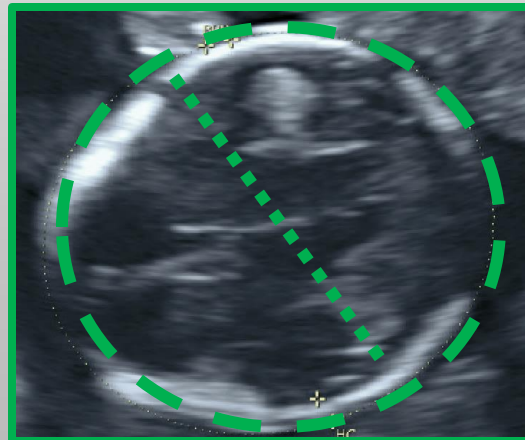
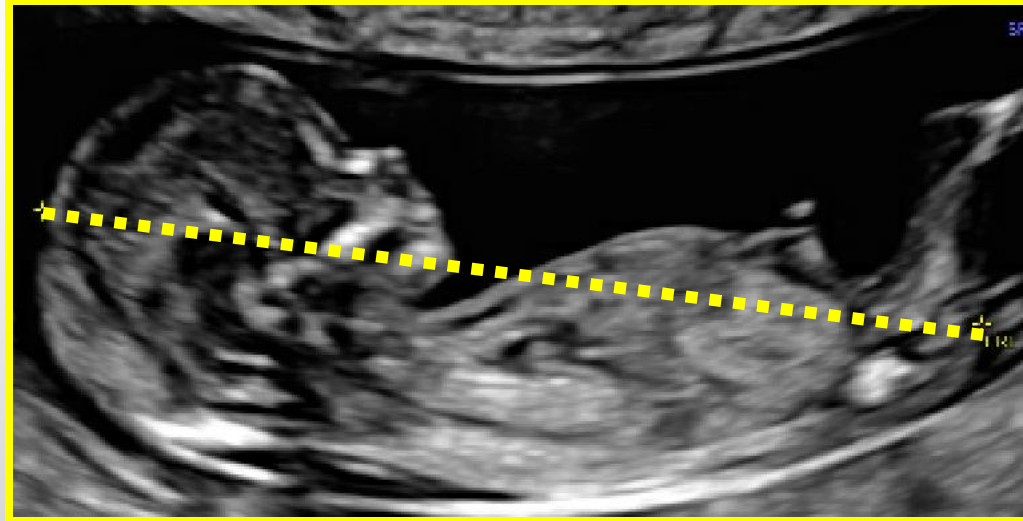
Obstetric (Detailed 1st Trimester) * (2020)

[Article](#) | [Image Library](#)



FETAL BIOMETRY

- CRL
- HC and BPD
- AC
- FL



DFTU: IMAGING CHECKLIST

- Evaluation of 7 major organ systems
 - ✓ Head & brain
 - ✓ Face & neck
 - ✓ Thorax
 - ✓ Heart
 - ✓ Abdomen
 - ✓ Spine
 - ✓ Limbs

Table 5.1 • Major Fetal Abnormalities That can Potentially be Easily Identified on First Trimester Ultrasound

- Early hydrops
- Anencephaly
- Alobar holoprosencephaly
- Body stalk anomaly
- Ectopia cordis
- Large omphalocele
- Large gastroschisis
- Megacystis
- Molar placenta

Diagnosis of fetal non-chromosomal abnormalities on routine ultrasound examination at 11–13 weeks' gestation

A. SYNGELAKI¹ , A. HAMMAMI¹, S. BOWER¹, V. ZIDERE¹ , R. AKOLEKAR^{2,3}  and K. H. NICOLAIDES¹

¹Fetal Medicine Research Institute, King's College Hospital, London, UK; ²Fetal Medicine Unit, Medway Maritime Hospital, Gillingham, UK; ³Institute of Medical Sciences, Canterbury Christ Church University, Chatham, UK

Retrospective study of prospectively collected data (~101,000 pregnancies)

Standardized protocol; continuing pregnancies had additional scan at 18–24 weeks

Final dx of fetal abnormality based on result of postnatal exam or findings of last US in cases of pregnancy interruption, SAB or stillbirth

Diagnosis of fetal non-chromosomal abnormalities on routine ultrasound examination at 11–13 weeks' gestation

A. SYNGELAKI¹ , A. HAMMAMI¹, S. BOWER¹, V. ZIDERE¹ , R. AKOLEKAR^{2,3}  and K. H. NICOLAIDES¹

¹Fetal Medicine Research Institute, King's College Hospital, London, UK; ²Fetal Medicine Unit, Medway Maritime Hospital, Gillingham, UK; ³Institute of Medical Sciences, Canterbury Christ Church University, Chatham, UK

Detected ALL cases of

- Acrania
- Encephalocele
- Holoprosencephaly
- Tricuspid/Pulmonary Atresia
- Ectopia cordis
- Gastroschisis/Omphalocele
- Body stalk anomaly

>50% cases of

- Open spina bifida
- HLHS
- AVSD
- Complex heart defect
- LUTO
- Absence of extremities
- Lethal skeletal dysplasia

First-trimester ultrasound detection of fetal heart anomalies: systematic review and meta-analysis

J. N. KARIM¹, E. BRADBURN¹, N. ROBERTS² and A. T. PAPAGEORGHIU^{1,3} , for the ACCEPTS study[#]

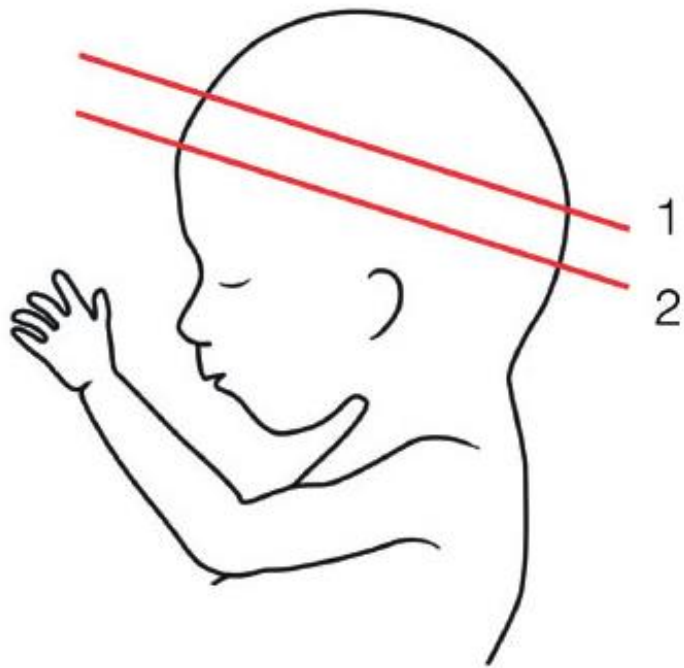
¹Nuffield Department of Women's & Reproductive Health, University of Oxford, Oxford, UK; ²Bodleian Health Care Libraries, University of Oxford, Oxford, UK; ³Oxford Maternal & Perinatal Health Institute, Green Templeton College, University of Oxford, Oxford, UK

- Review of 63 studies & 328,262 fetuses
- 1st trimester US of the fetal heart identified over half of fetuses affected by major cardiac pathology
- Importance of (1) a structured anatomical assessment protocol and (2) visualization of the outflow tracts and use of color-flow Doppler to optimize detection of cardiac anomalies

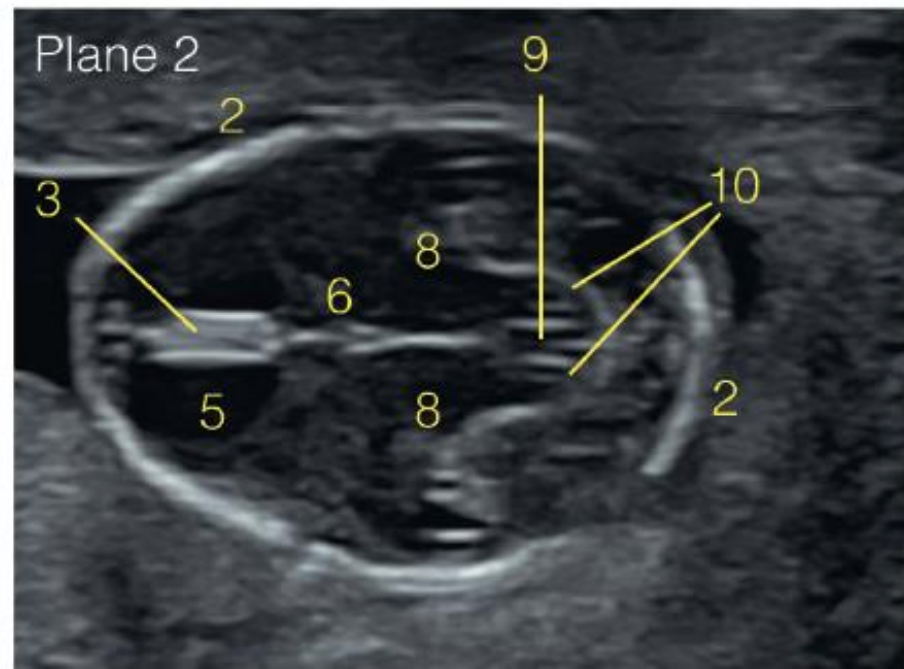
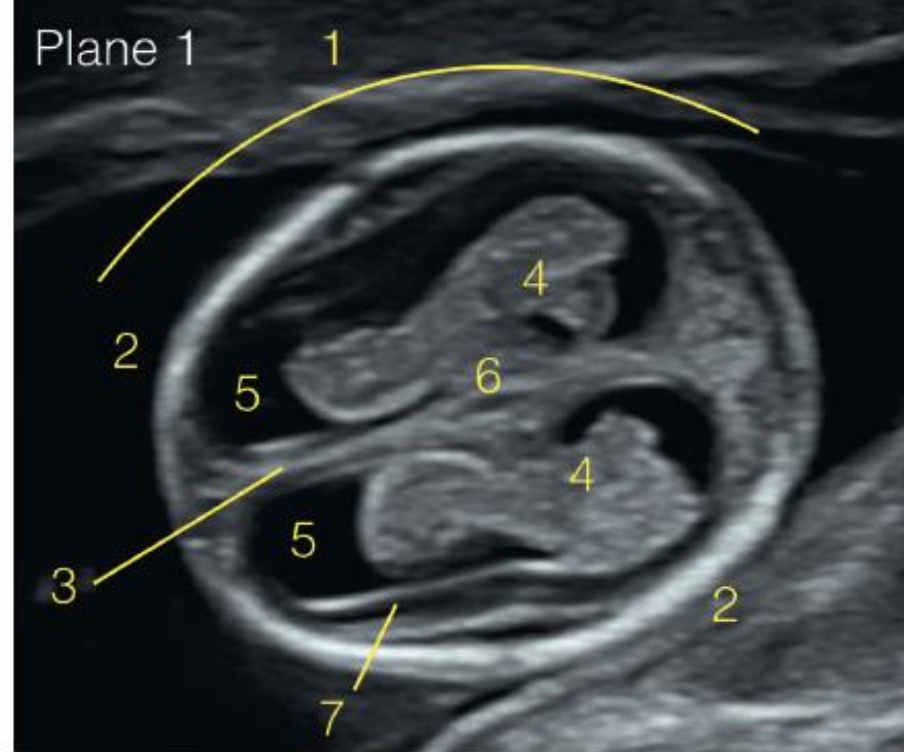


I. HEAD & BRAIN IMAGING





1. Oval head shape
2. Skull bones
3. Falx cerebri
4. Choroid plexus
5. Lateral ventricles
6. Third ventricle
7. Cortex
8. Thalami
9. Aqueduct of Sylvius
10. Cerebral peduncles



Voluson™
E10

1ST TRIM

TIs 0.2
TIB 0.2
MI 1.0

02/11/2021
2:57:09 PM
C2-9-D

46Hz/ 6.3cm
40°/1.2

Routine Res./OB
HD Res 15.90 - 3.80

Gn -2

C7/M8

FF2/E2

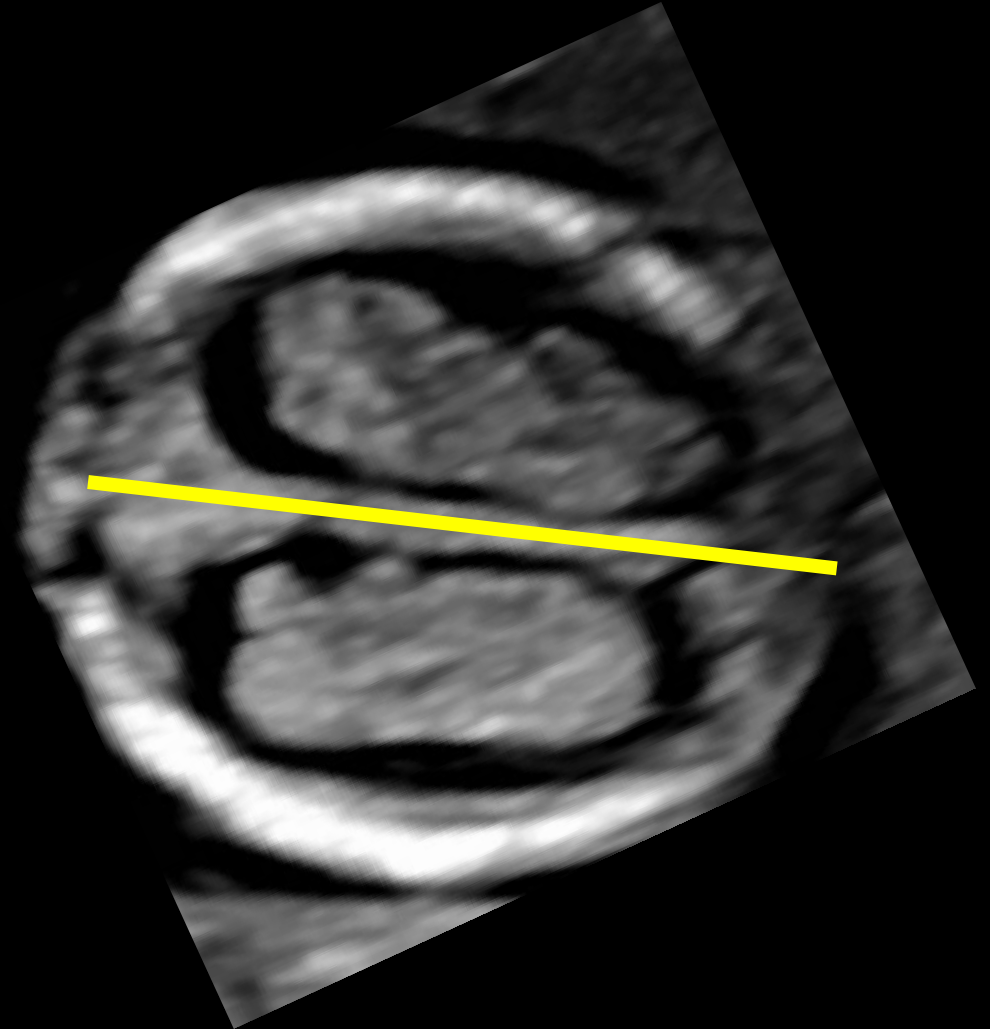
SRI II 3/CRI 3

Voluson
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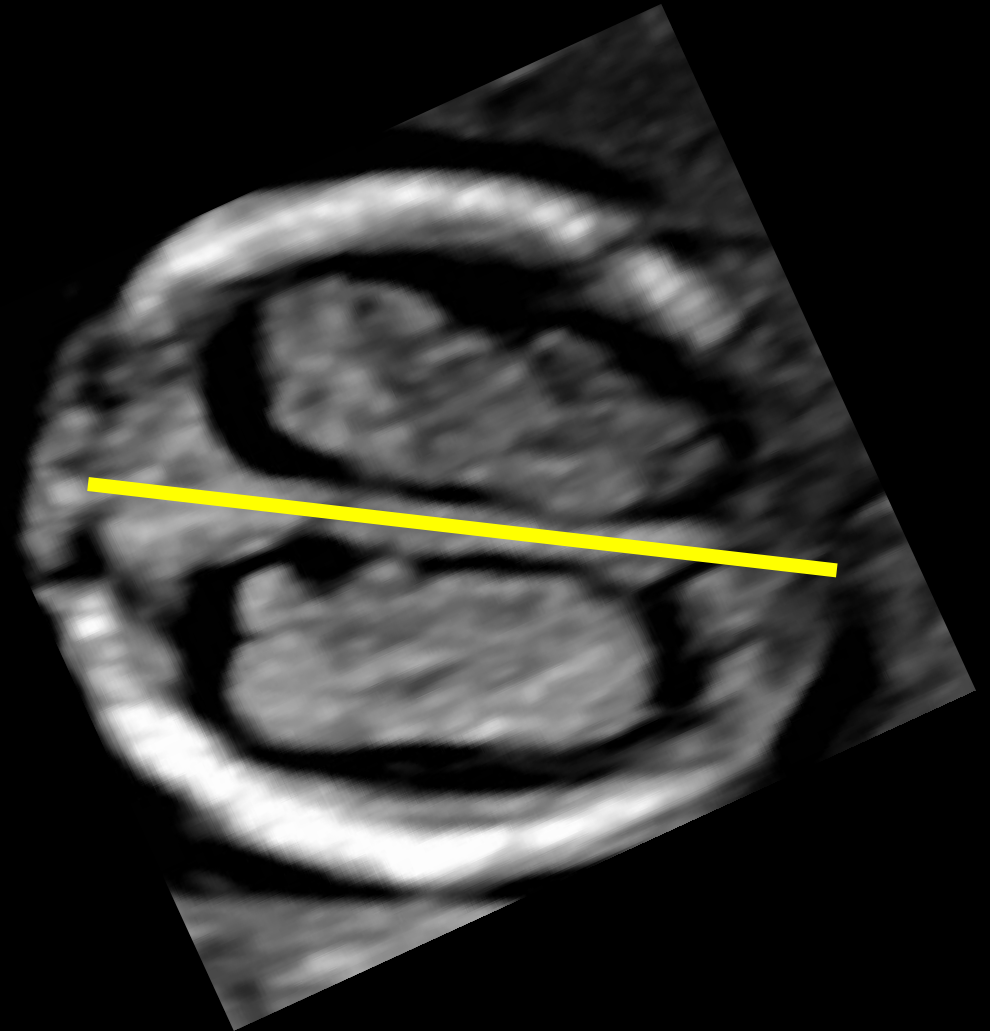
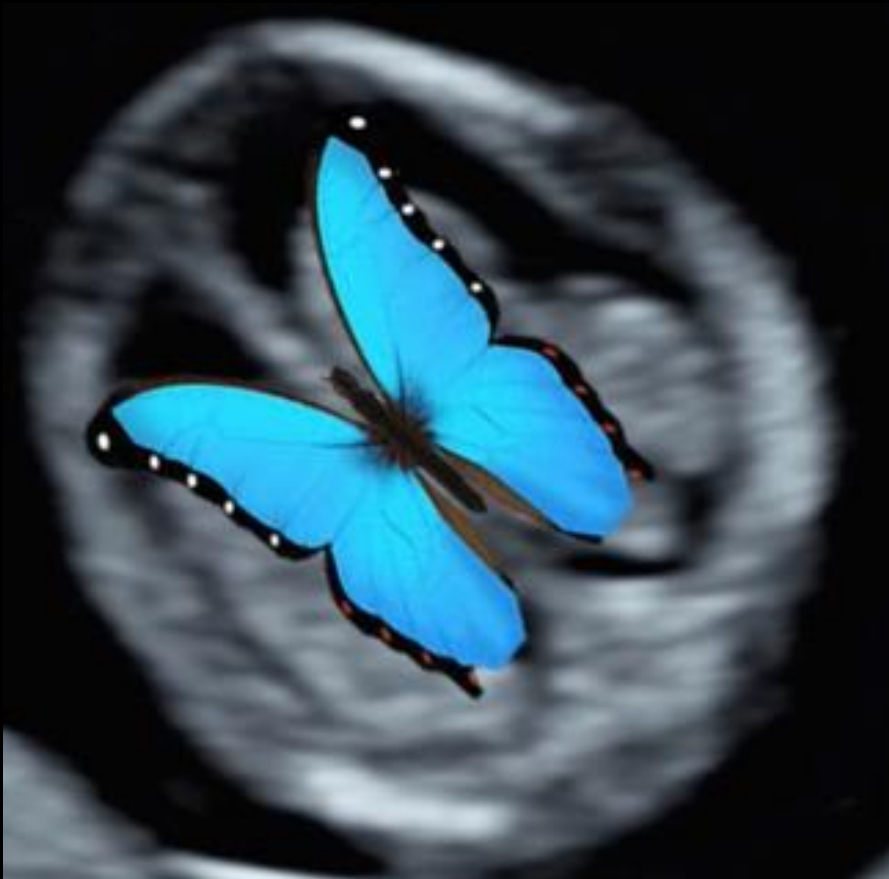
Head: Transventricular

[cranium, falx, ventricles, brain]



Head: Transventricular

[cranium, falx, ventricles, brain]

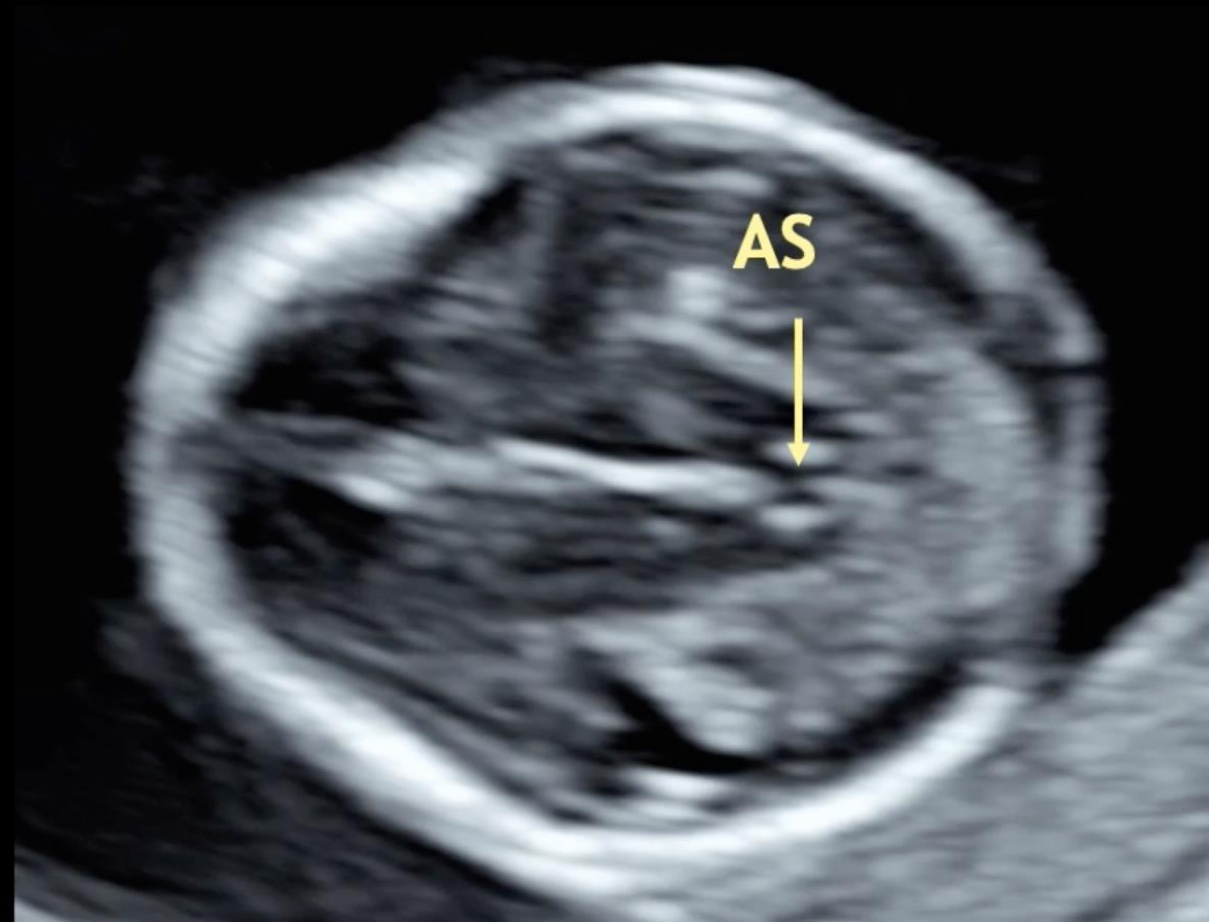
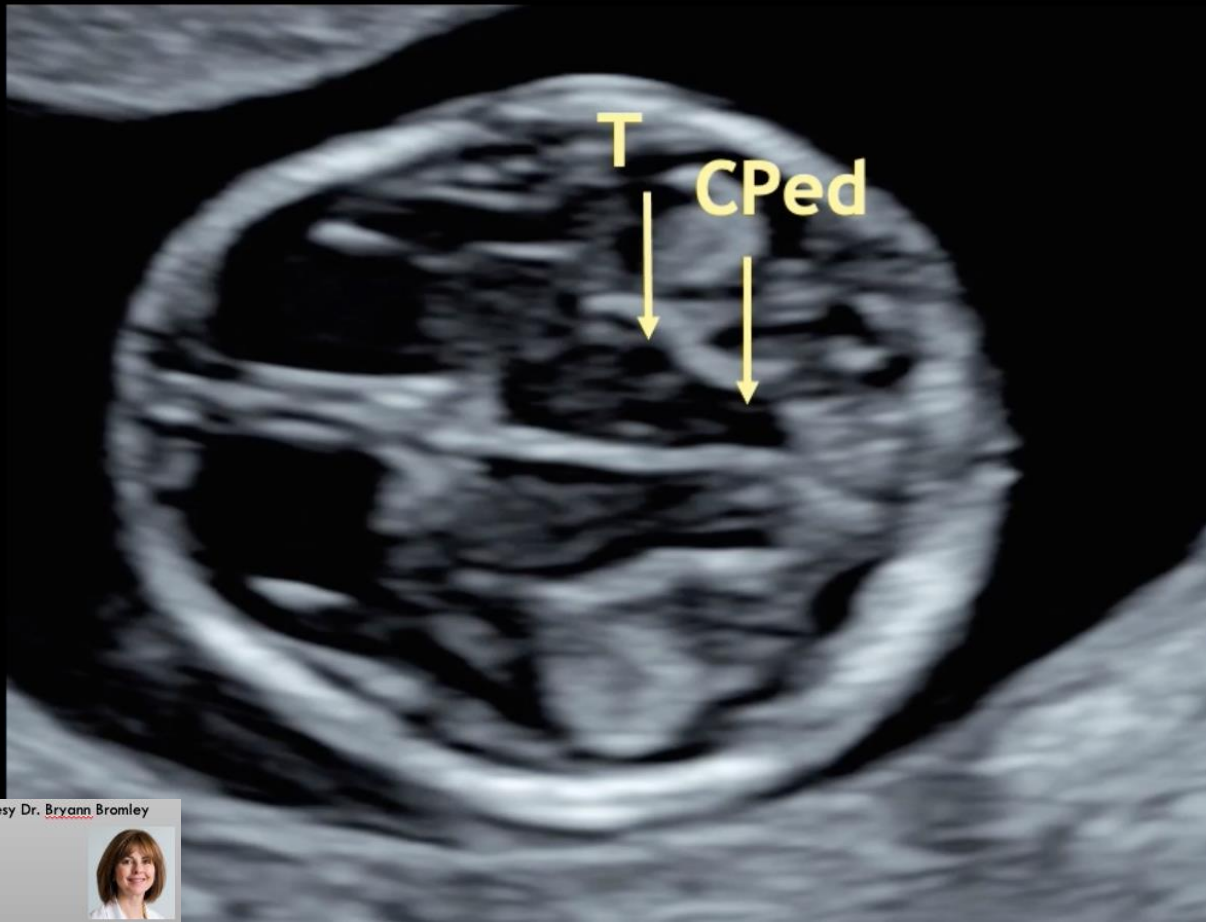


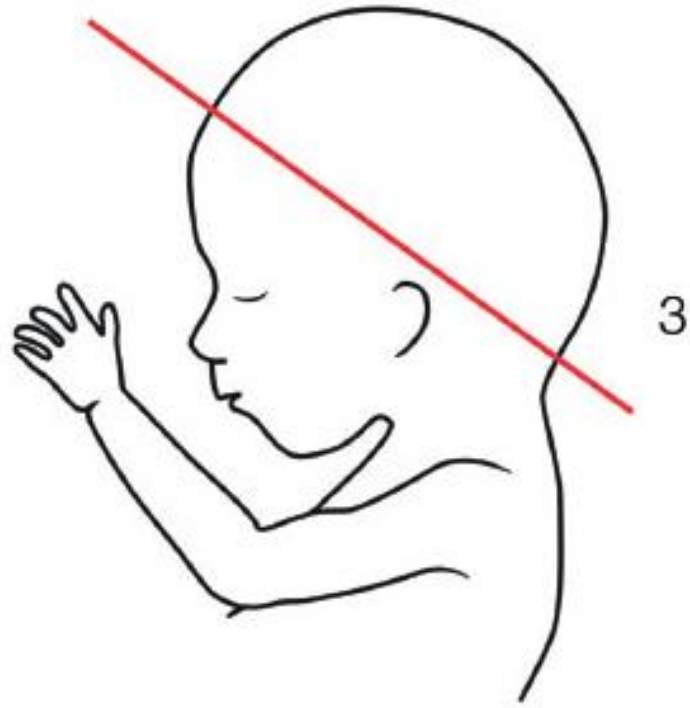
Head: **Transthalamic** [cranium, falx, thalami, CPed, AS]



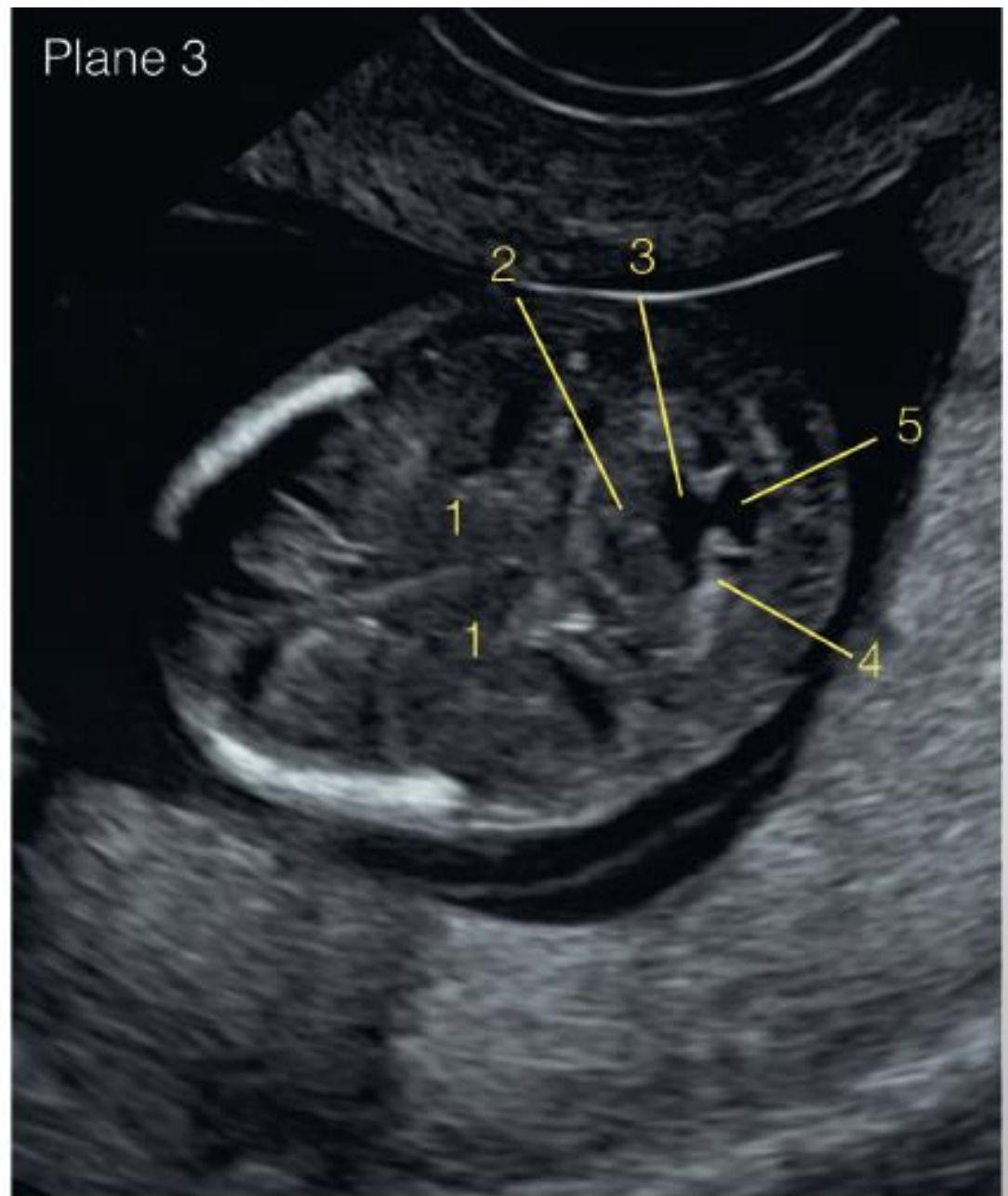
Head: 2-Transthalamic

[Cranium, Falx, Thalami, CPed, AS]

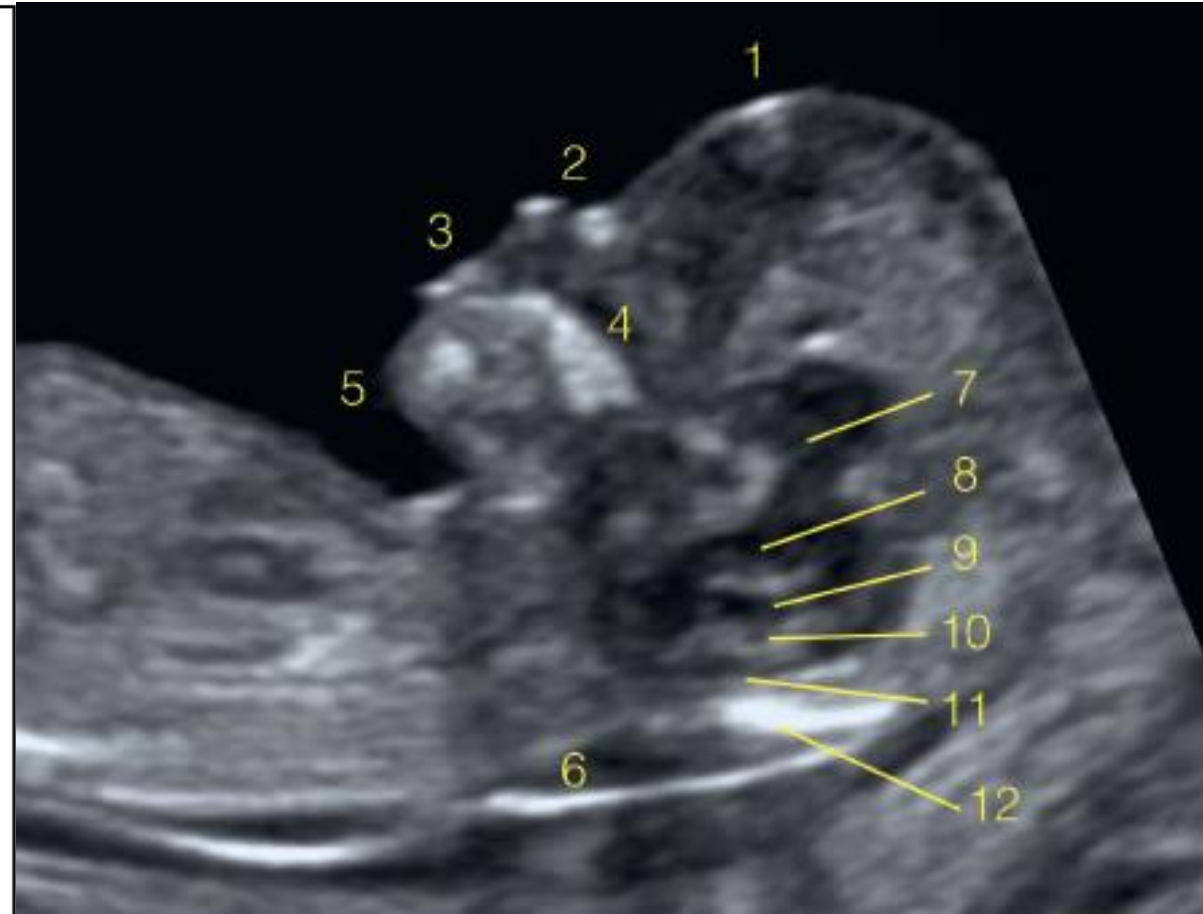
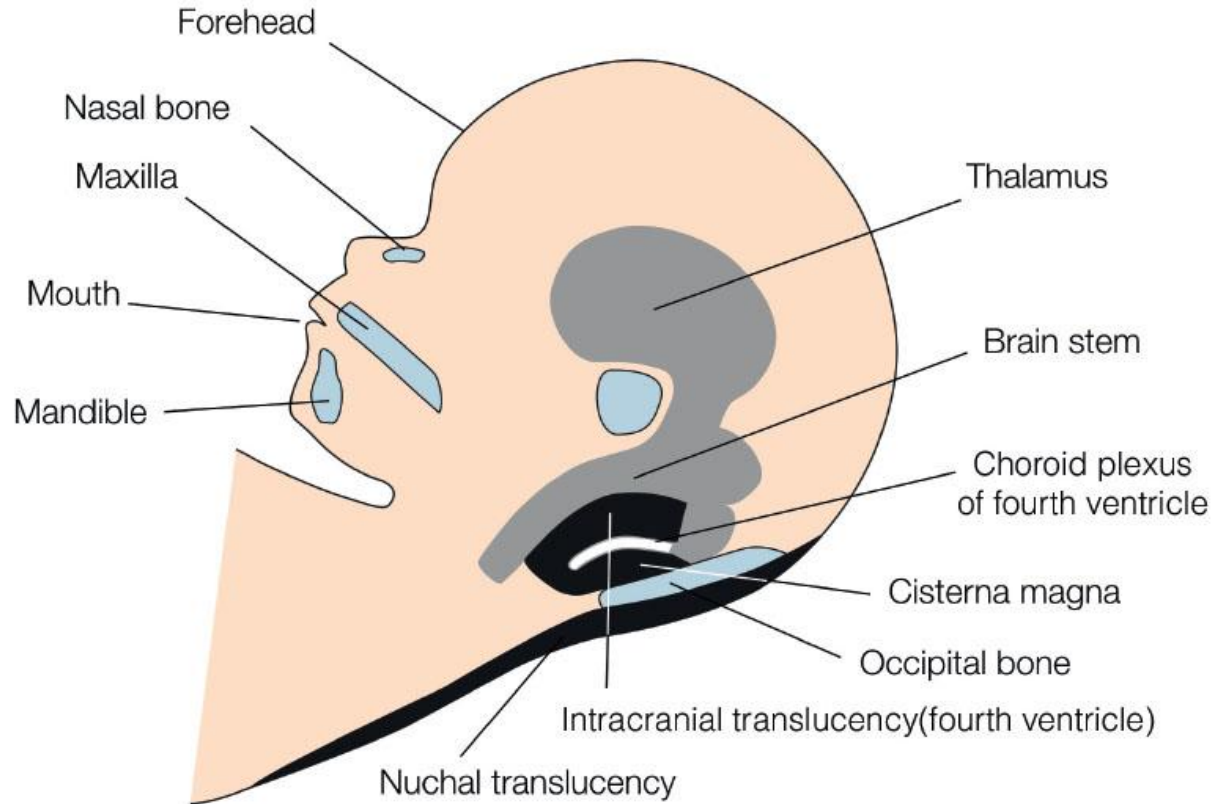




1. Thalami
2. Developing cerebellum
3. Fourth ventricle (IT)
4. Choroid plexus of Fourth ventricle
5. Future cisterna magna



Normal face

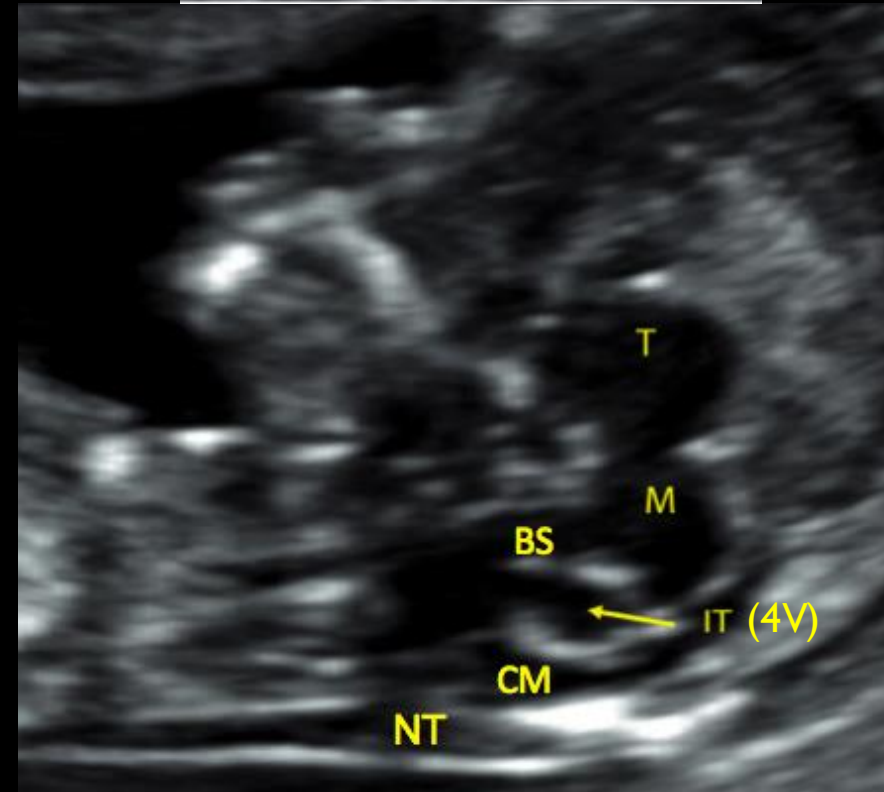
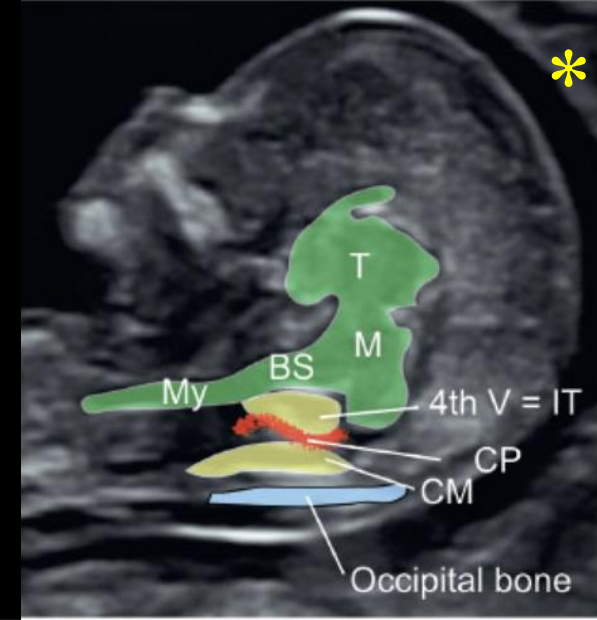




Head: Midsagittal

[Thalamus, midbrain, brainstem, IT (4th V), CM]

- 4th ventricle is a translucency parallel to the NT
- With open spina bifida be an absence of the IT owing to caudal displacement of brain



Anencephaly



WHAT DO YOU SEE?

EXENCEPHALY/ANENCEPHALY SEQUENCE

Absence of cranial bone cephalad to the orbits

Varying amount of supratentorial brain

Neural tissue wears away during gestation

Result of fetal movement & exposure to amniotic fluid

EXENCEPHALY/ANENCEPHALY SEQUENCE

CRL less than expected

Protuberant eyes

Normal head contour is absent

Echogenic fluid





WHAT DO YOU SEE?



ENCEPHALOCELE

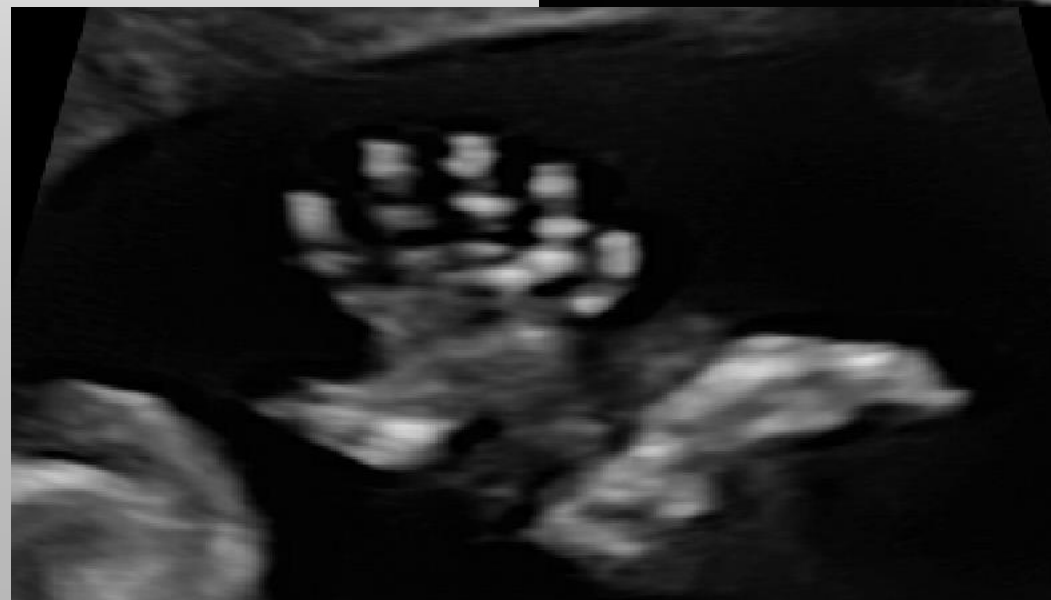
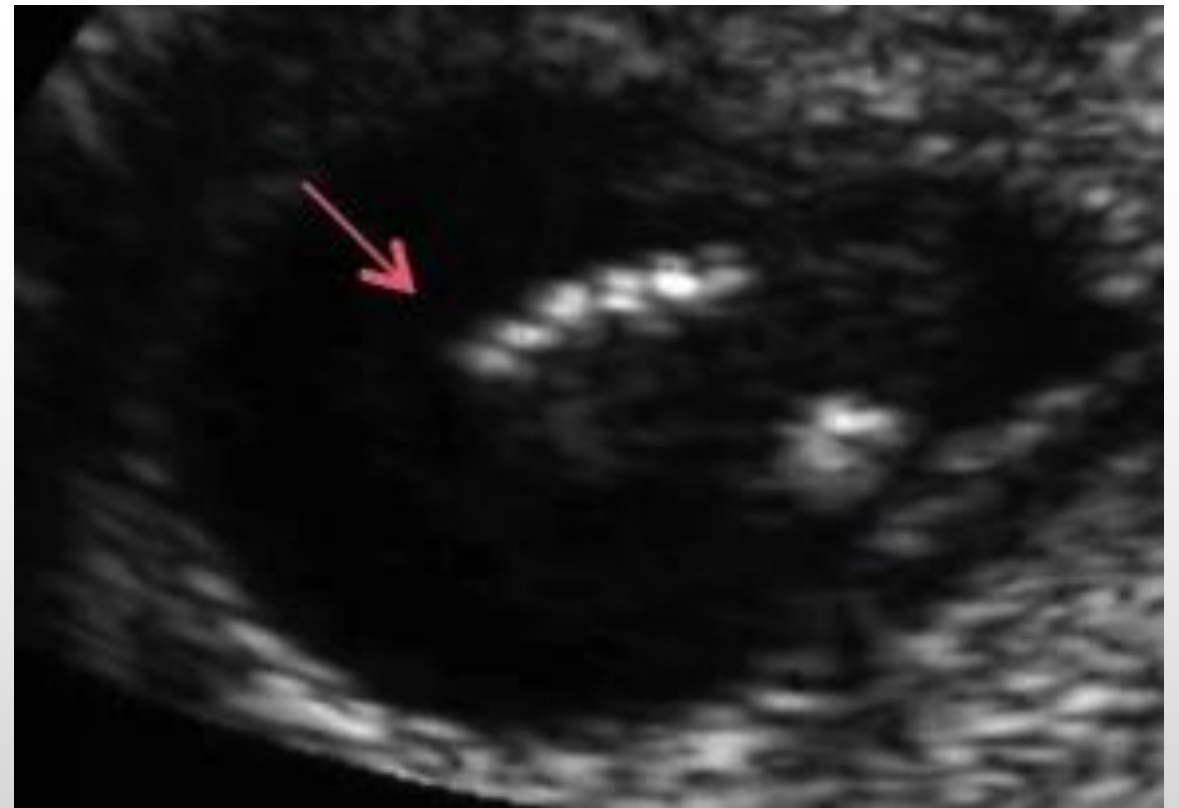
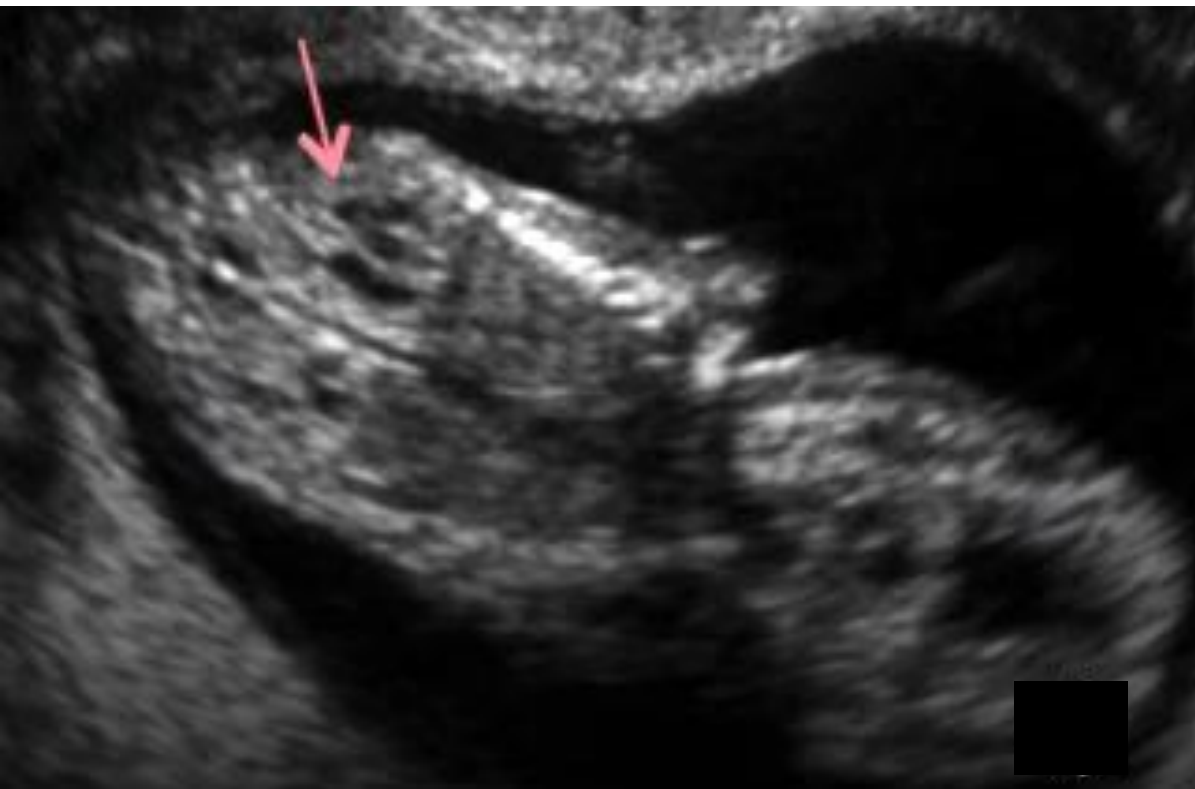
Cranium is present

Neural tissue protrudes through defect

- Most commonly occipital
- Frontal
- D/Dx scalp masses, amniotic band sequence, anencephaly/acrania

Microcephaly

Ventriculomegaly



Courtesy Dr. Bryann Bromley



MECKEL-GRUBER SYNDROME

Triad of findings

- Renal cystic dysplasia (95-100%)
- Encephalocele/other CNS abnl (90%)
- Postaxial polydactyly (55-75%)

At least 2 of 3 classic features in fetus with normal karyotype

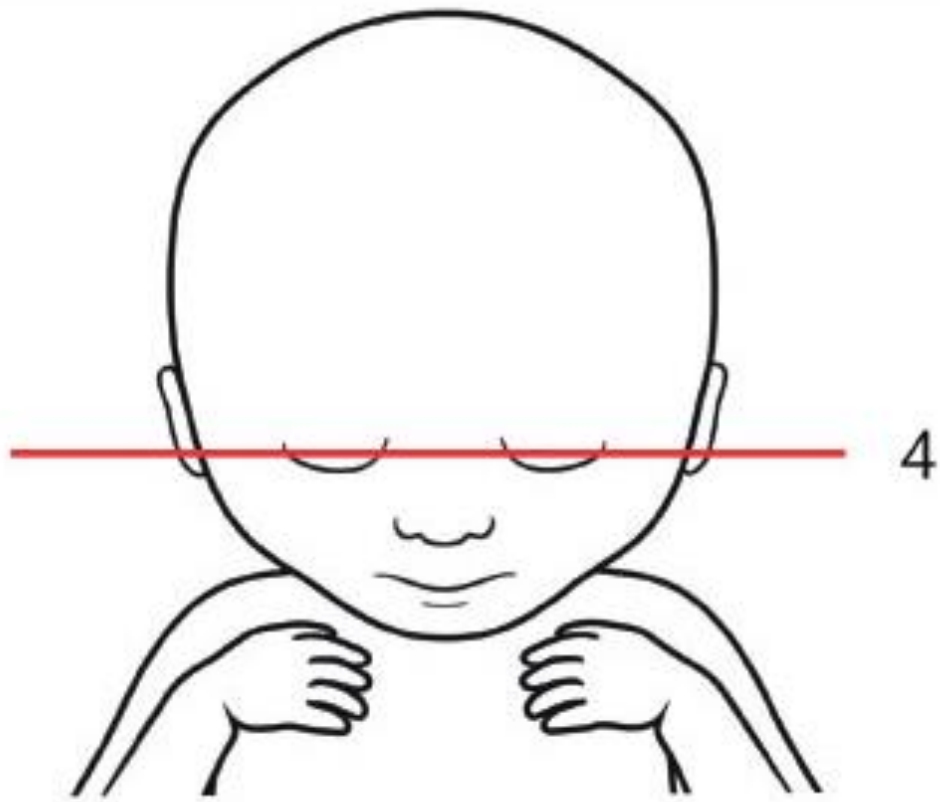
- Significant overlap (d/dx=trisomy 13)
- AR inheritance



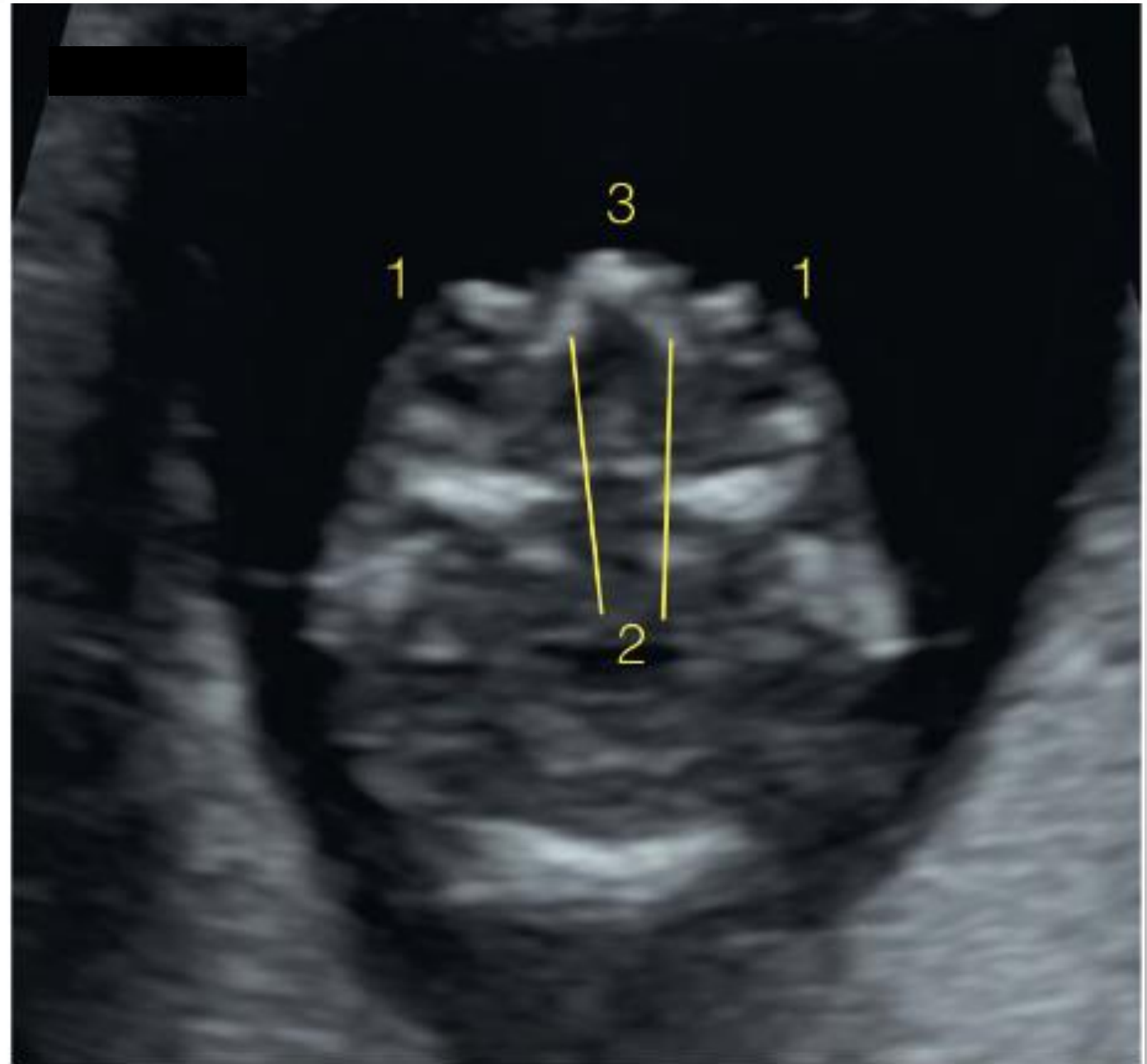


II. FACE & NECK IMAGING





1. Eyes/orbits
2. Maxillary processes
3. Nose

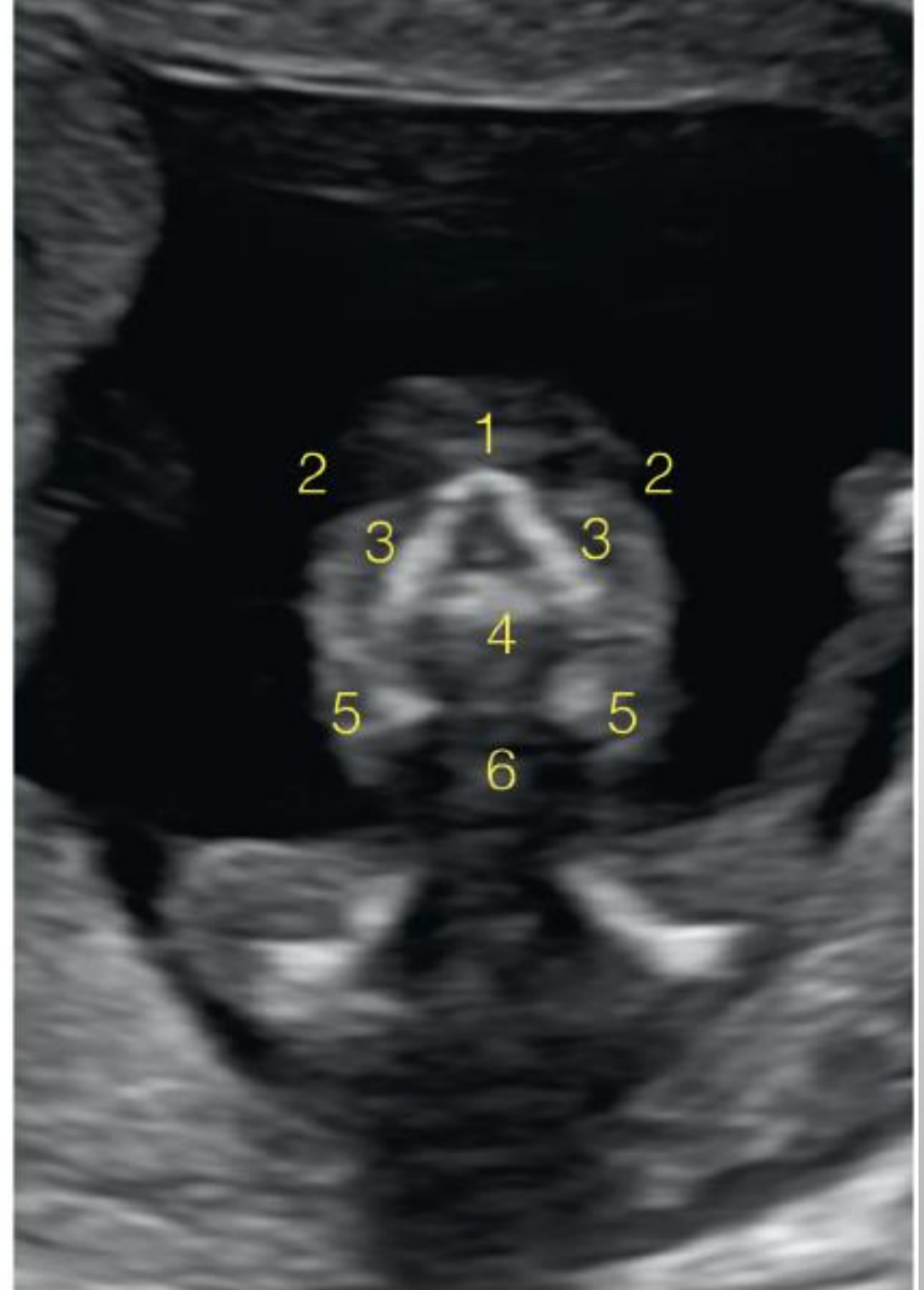


Face: Orbits, Lenses





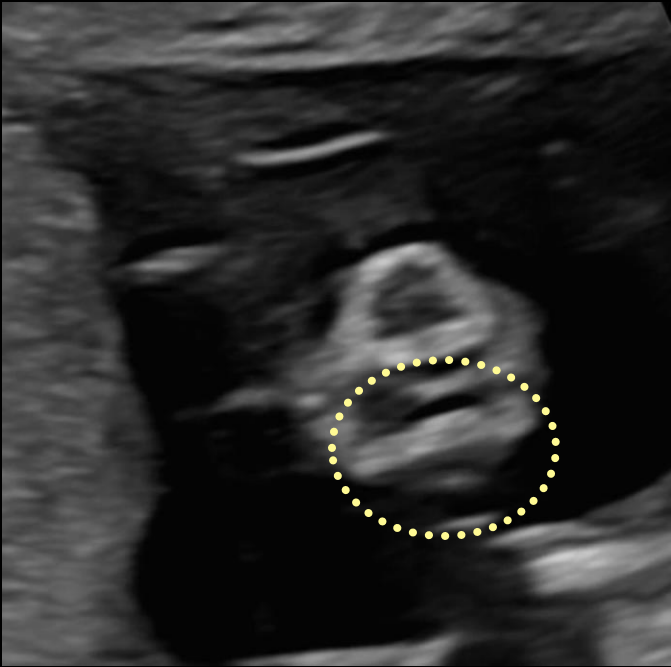
1. Nasal bones
2. Orbits/eyes
3. Maxillary processes
4. Maxilla-alveolar ridge
5. Mandible
6. Mandibular gap



Face: Retronasal triangle, nasal bone, mandible



Absent mandibular gap: **Micrognathia/Retrognathia**



Profile (mid sagittal) including mandible



Sagittal maxilla



Maxillary gap at 11–13 weeks' gestation: marker of cleft lip and palate

R. CHAOUI*, G. OROSZ†, K. S. HELING*, A. SARUT-LOPEZ* and K. H. NICOLAIDES†

*Center for Prenatal Diagnosis and Human Genetics, Berlin, Germany; †Harris Birthright Research Centre for Fetal Medicine, King's College Hospital, London, UK

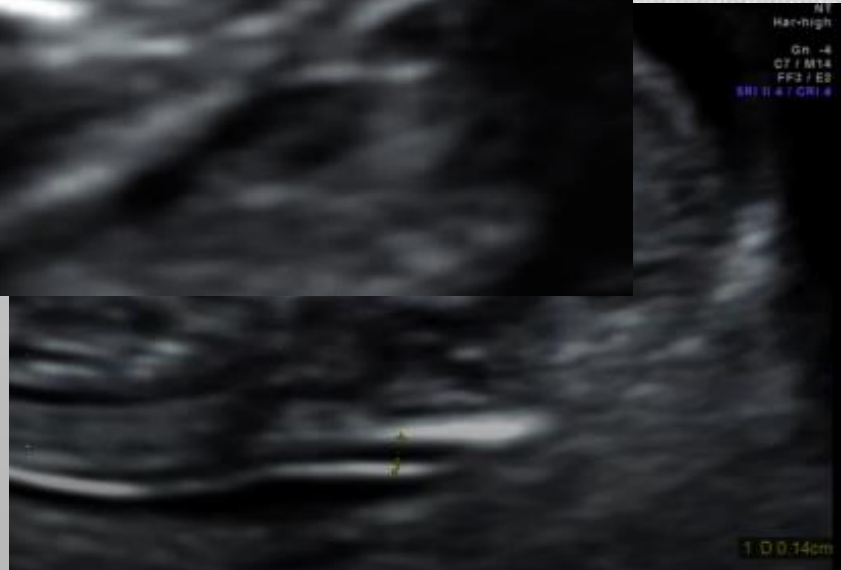
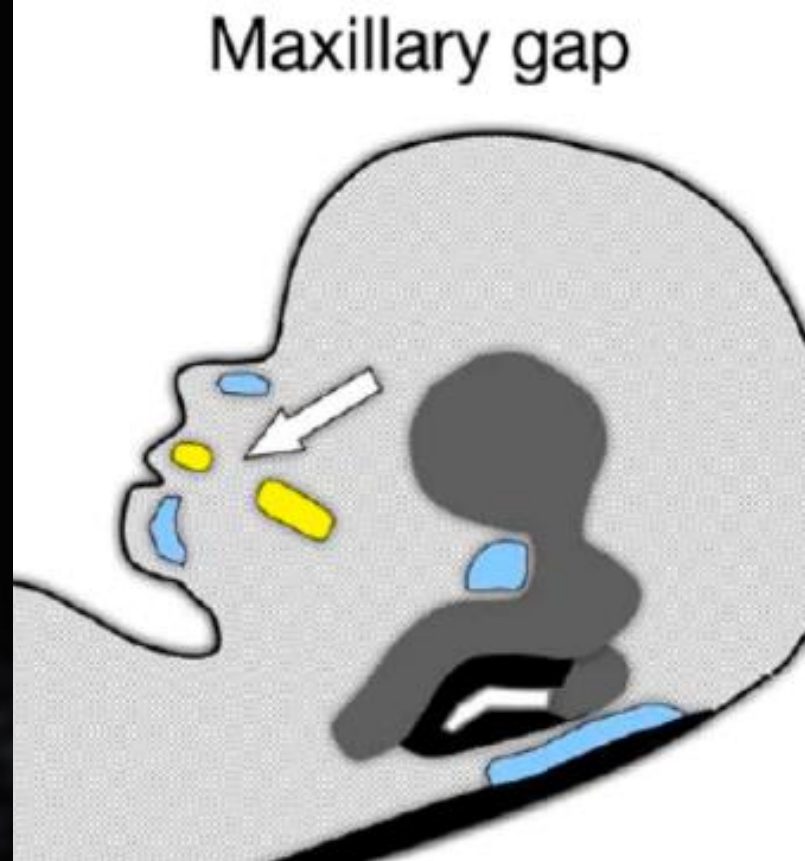
Appearance of maxilla - useful marker to detect facial clefts

Retrospective study of stored images from 86 fetuses with clefts & 86 controls

Maxillary gap identified in 96% of fetuses with CLP with additional findings & in 65% with isolated cleft & in 7% of normal fetuses

Threshold (for abnormality) => gap size of >1.5 mm

- 69% showed a large gap >1.5 mm in fetuses with facial cleft & additional findings
- 35% showed a large gap of >1.5 in fetuses isolated clefts identified
- No large gaps were identified in normal controls





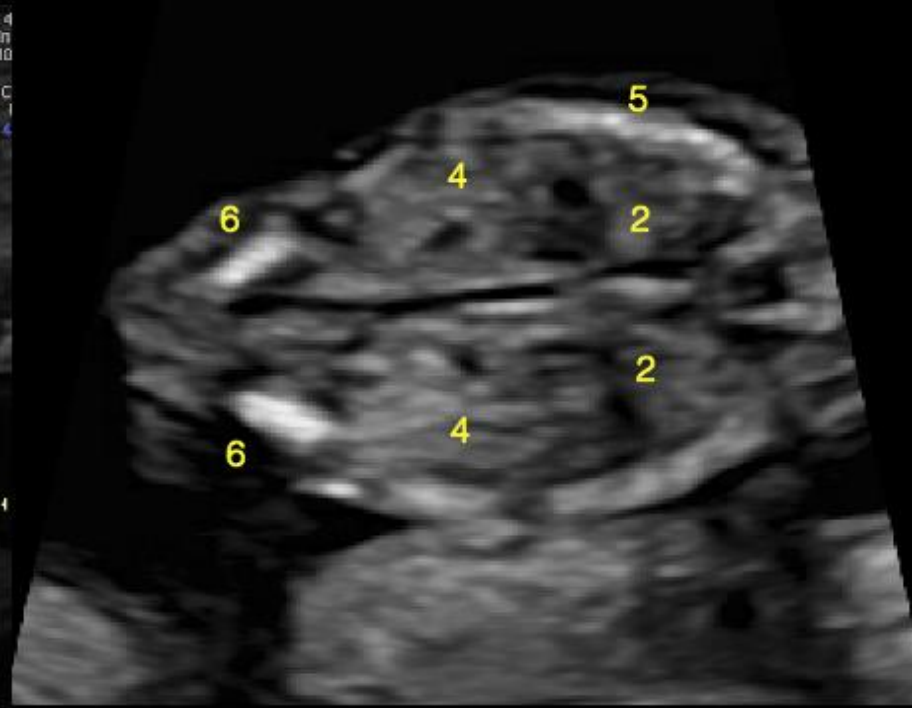
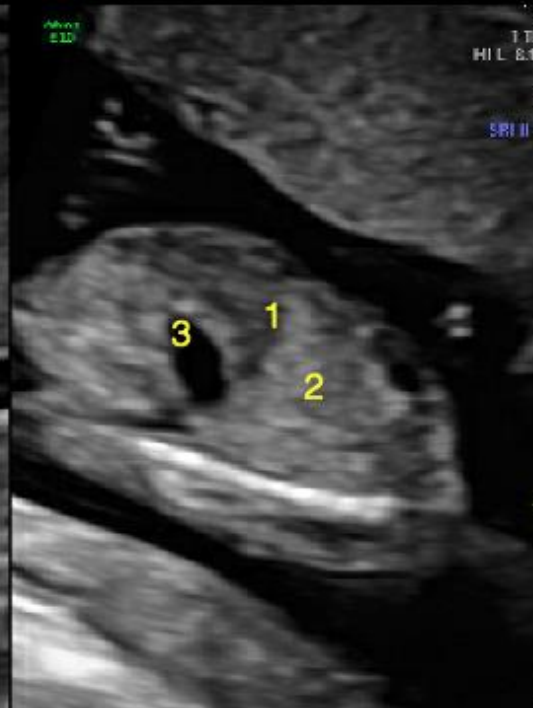


III. THORAX IMAGING

Thorax:

Shape of thoracic wall, lung fields, diaphragmatic continuity

1. Diaphragm
2. Lungs
3. Stomach in abdomen
4. Kidneys
5. Ribs
6. Pelvic bones



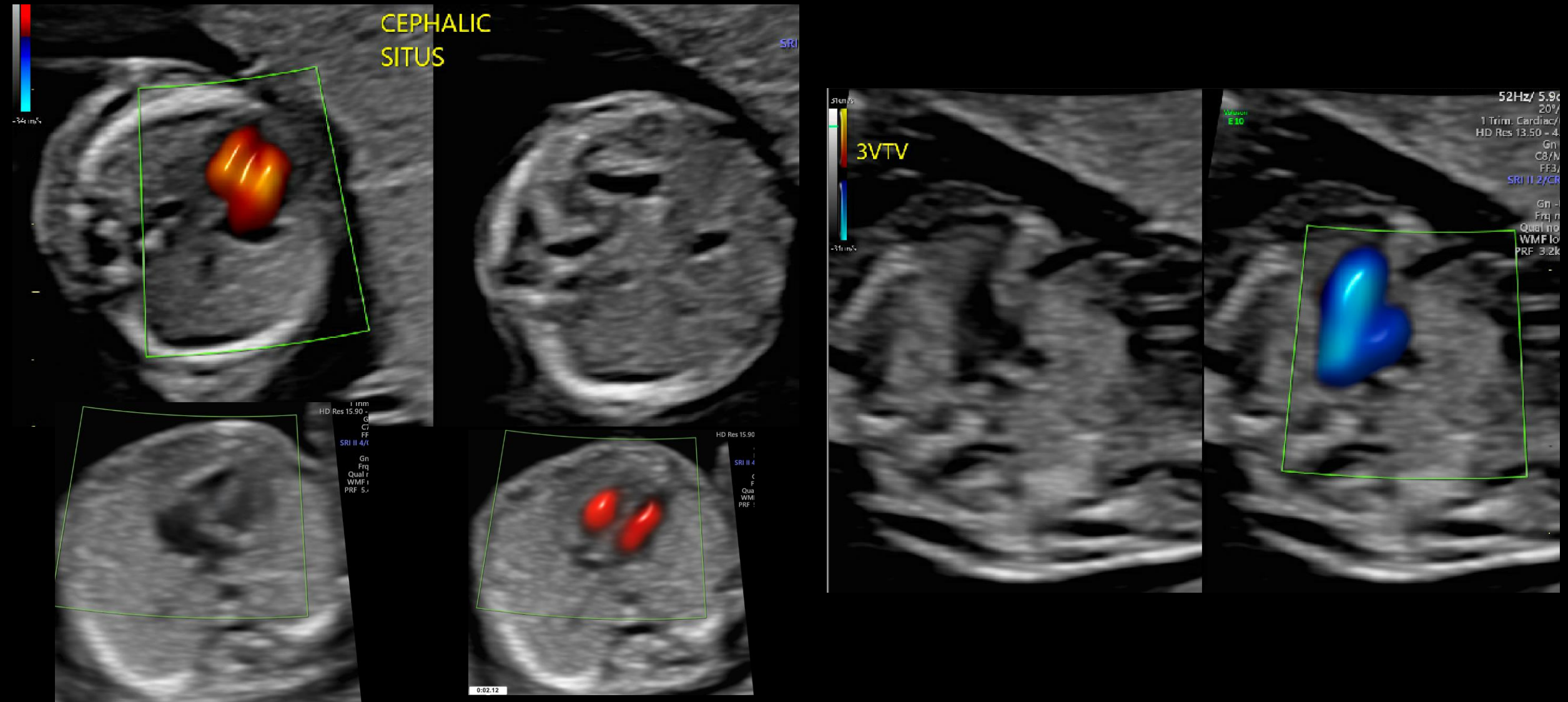




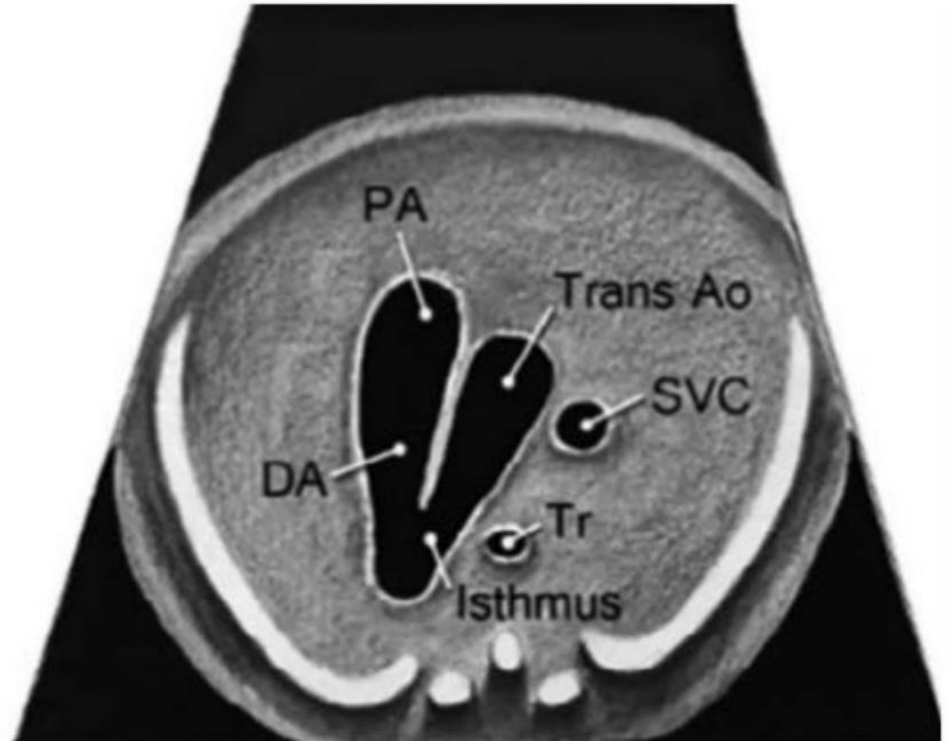
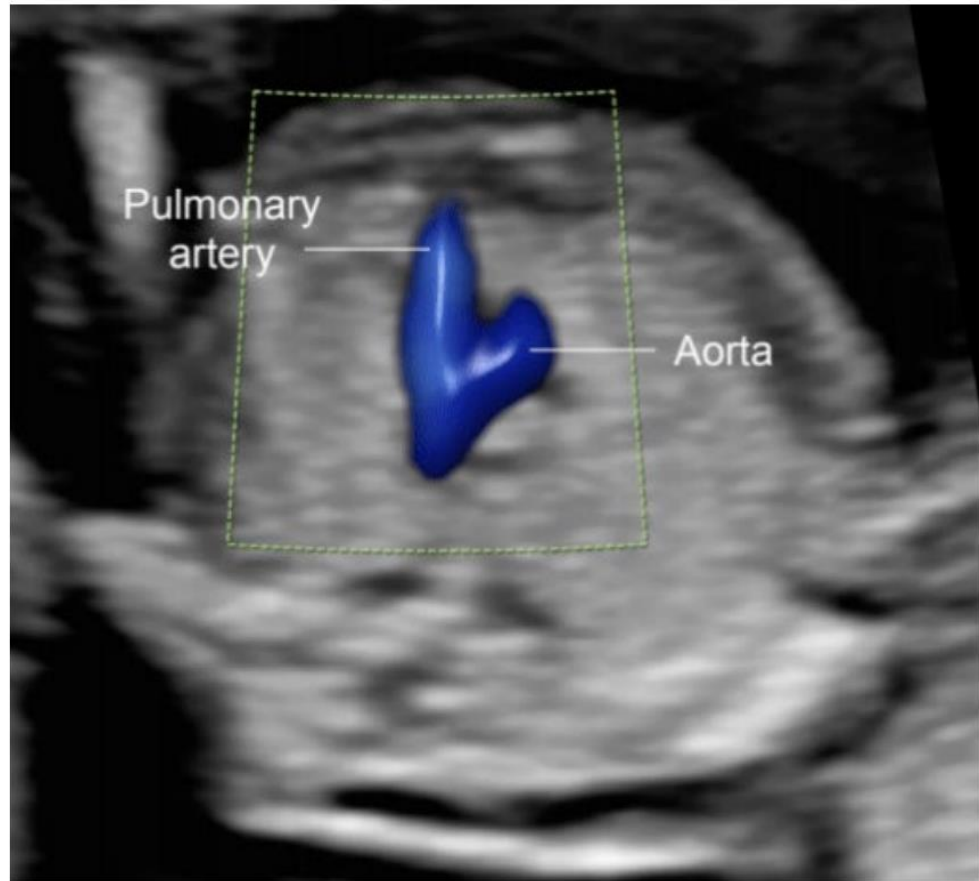
IV. CARDIAC IMAGING

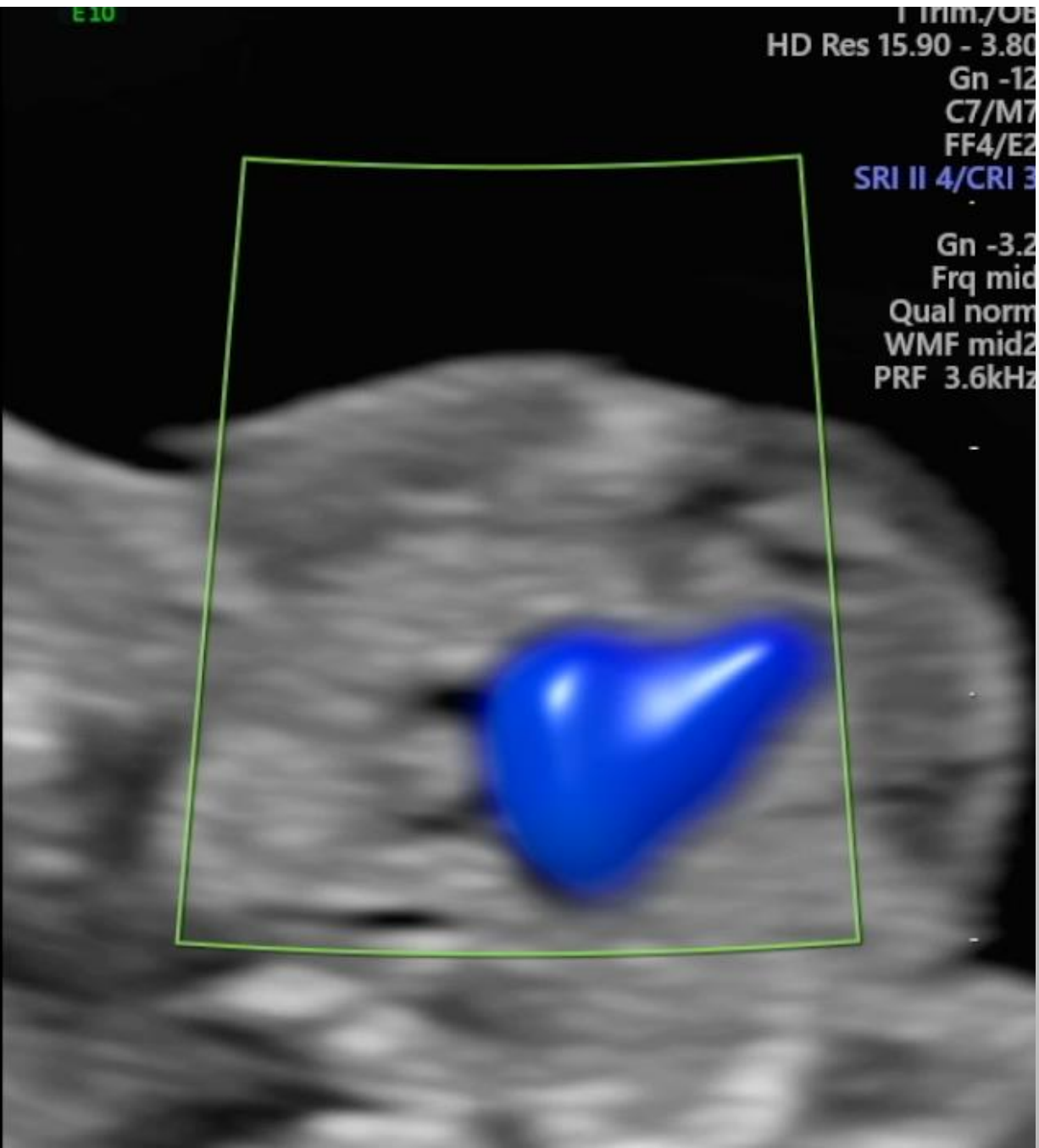
Cardiac

Most major anomalies identified in these views

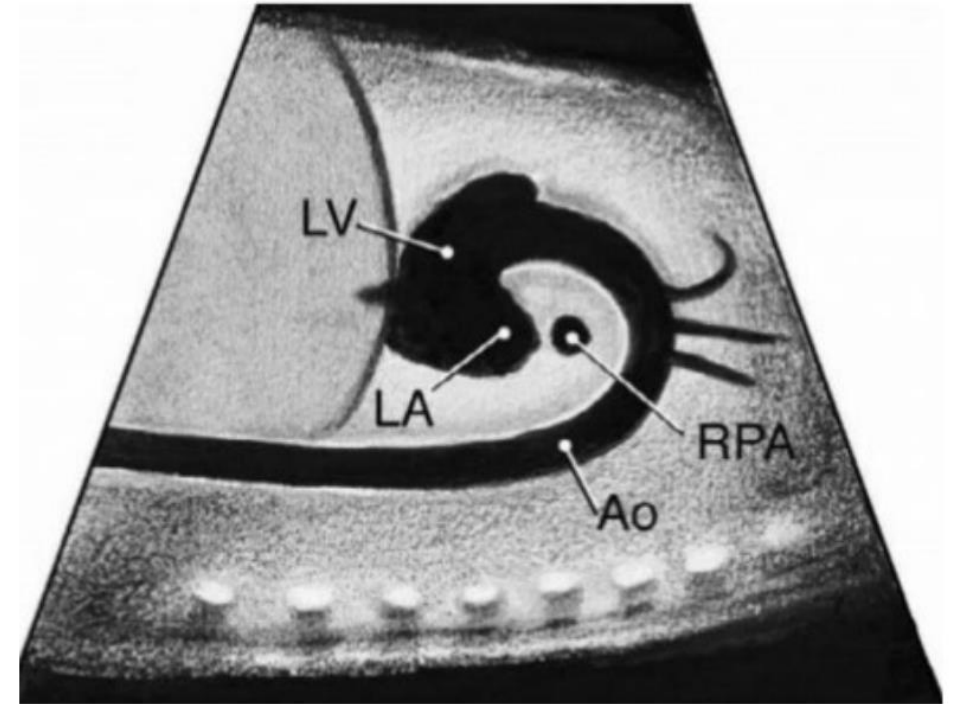
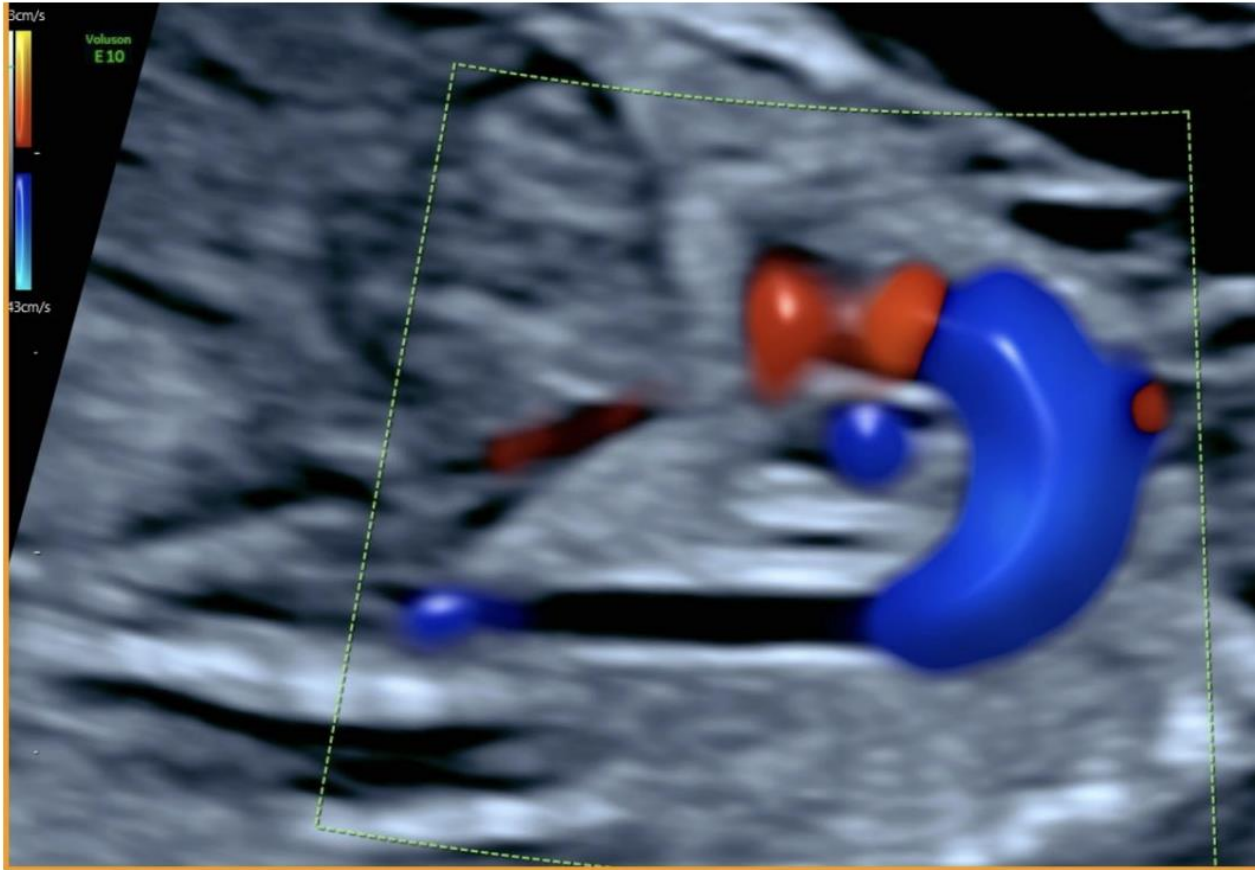


3 vessel trachea view with color

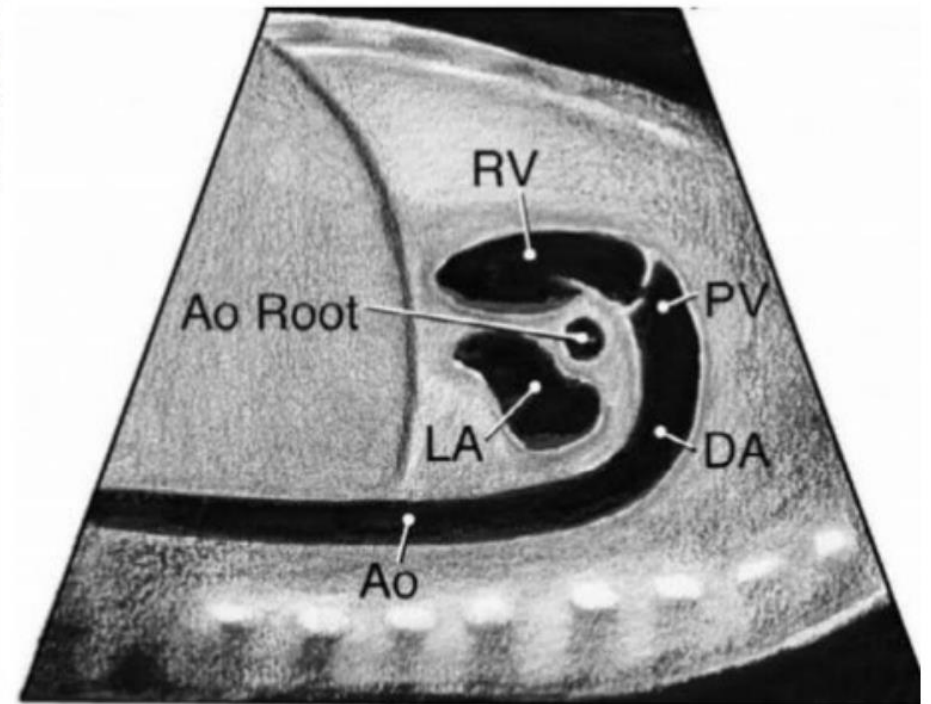
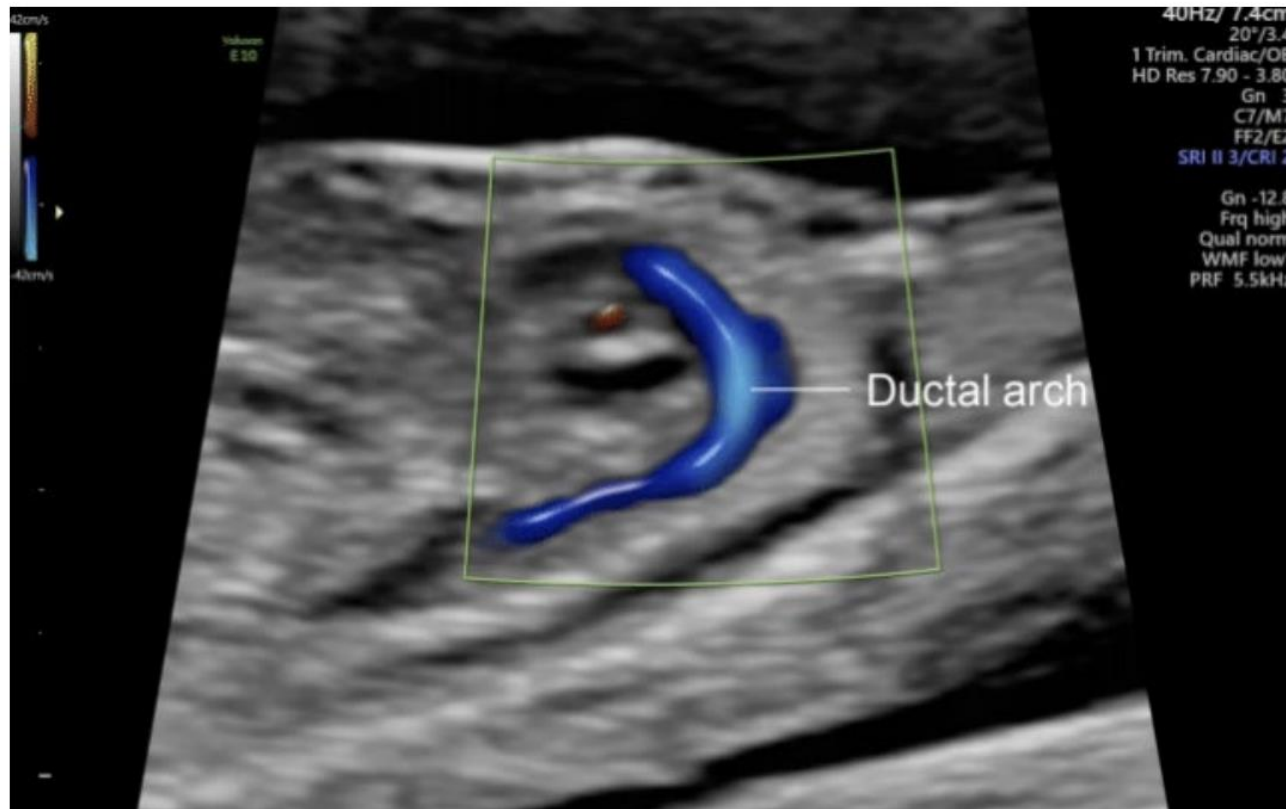




Sagittal aortic arch with color



Sagittal ductal arch with color

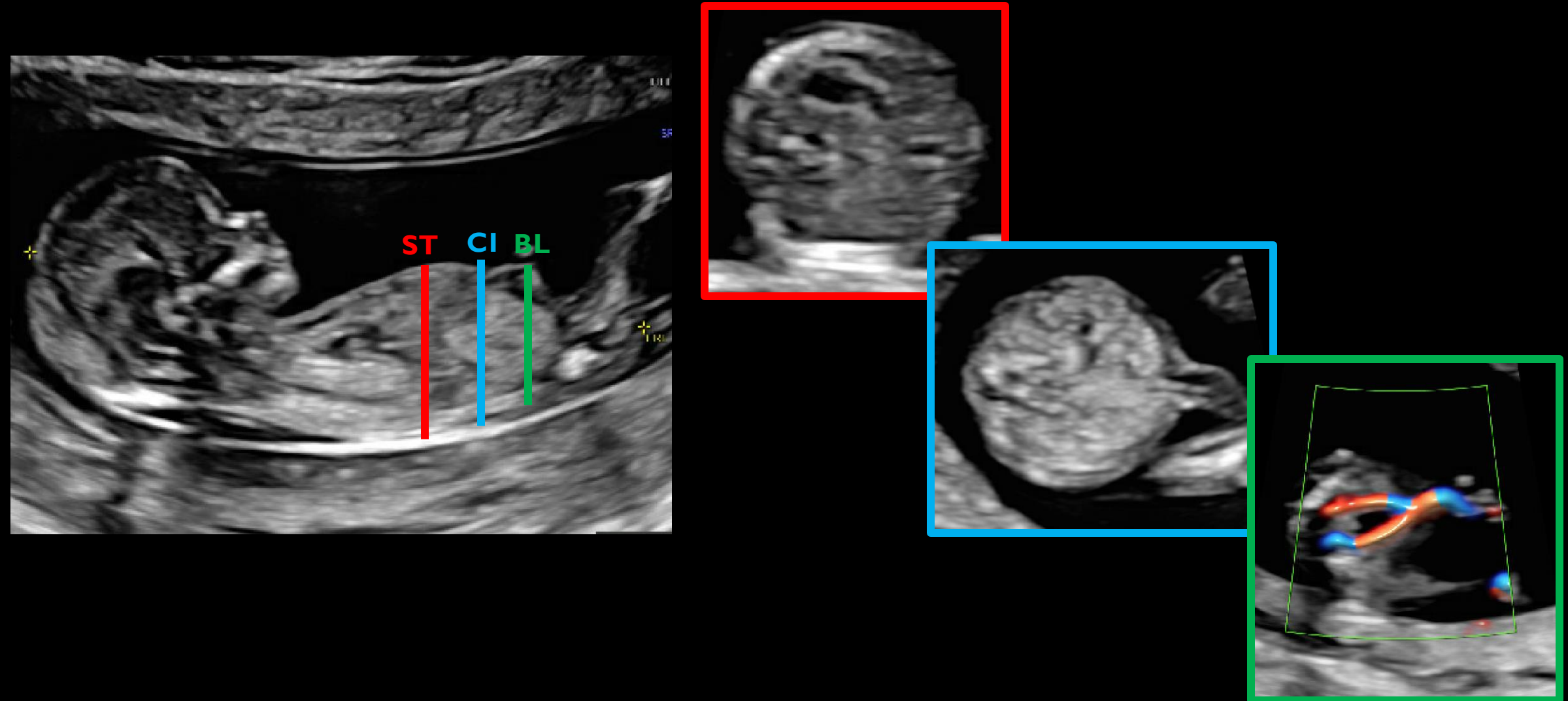


5



ABDOMINAL IMAGING

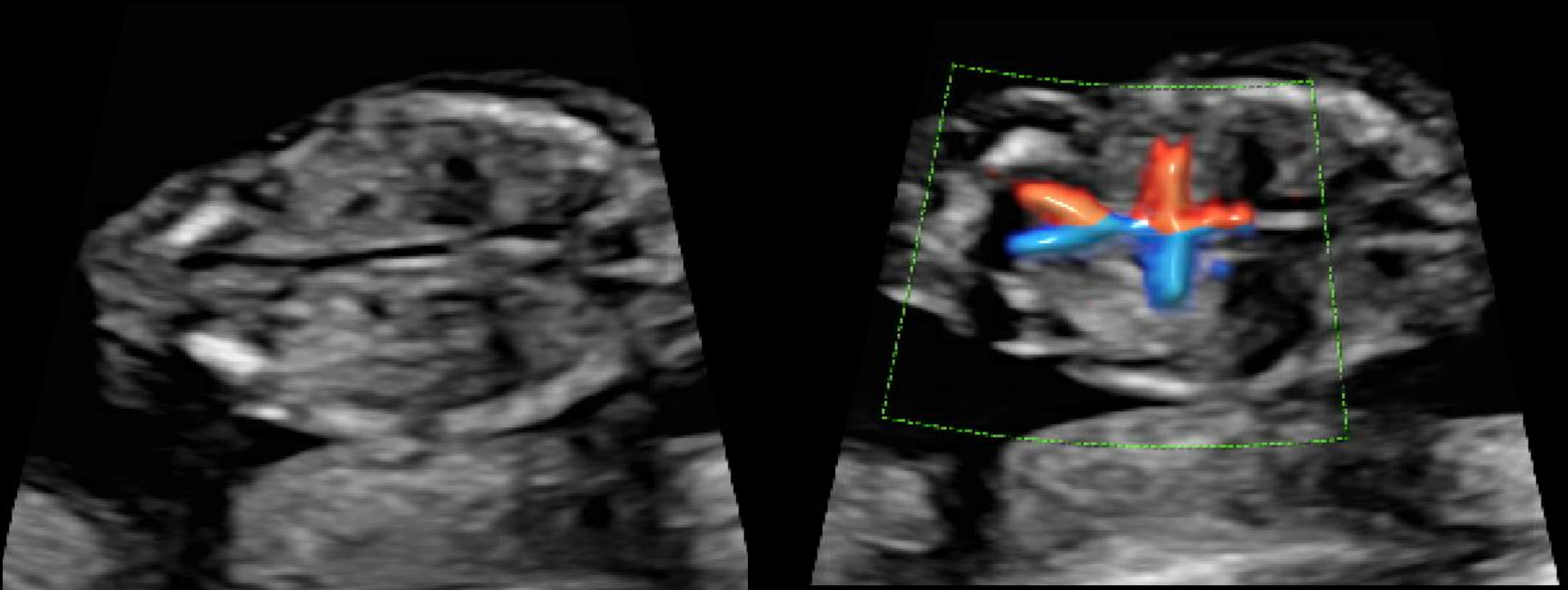
Abdominal: axial imaging - 3 levels



Coronal plane at the level of the kidneys



Abdominal: coronal imaging - kidneys



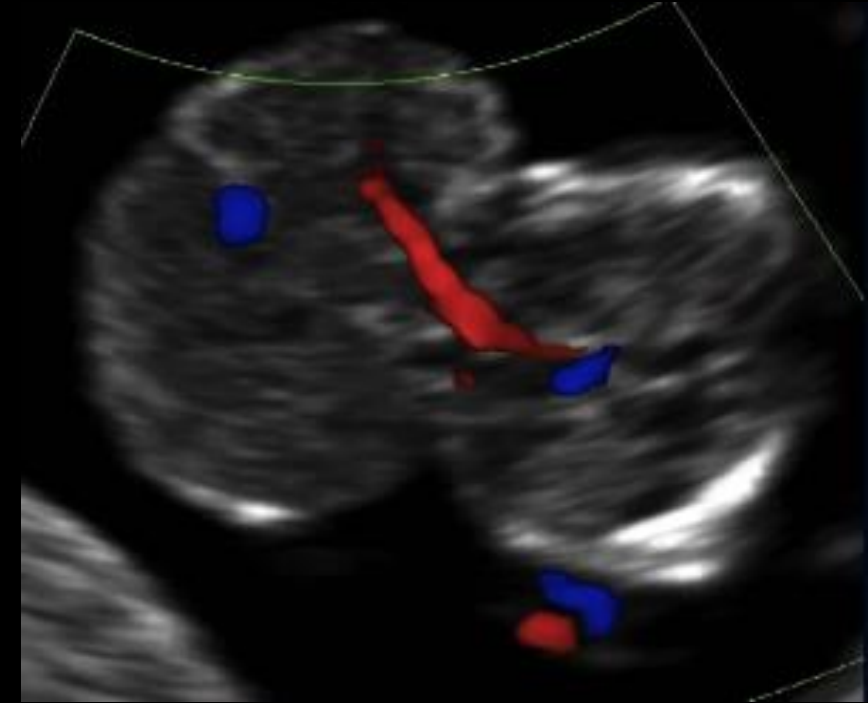
Abdominal Wall: Axial imaging



Normal

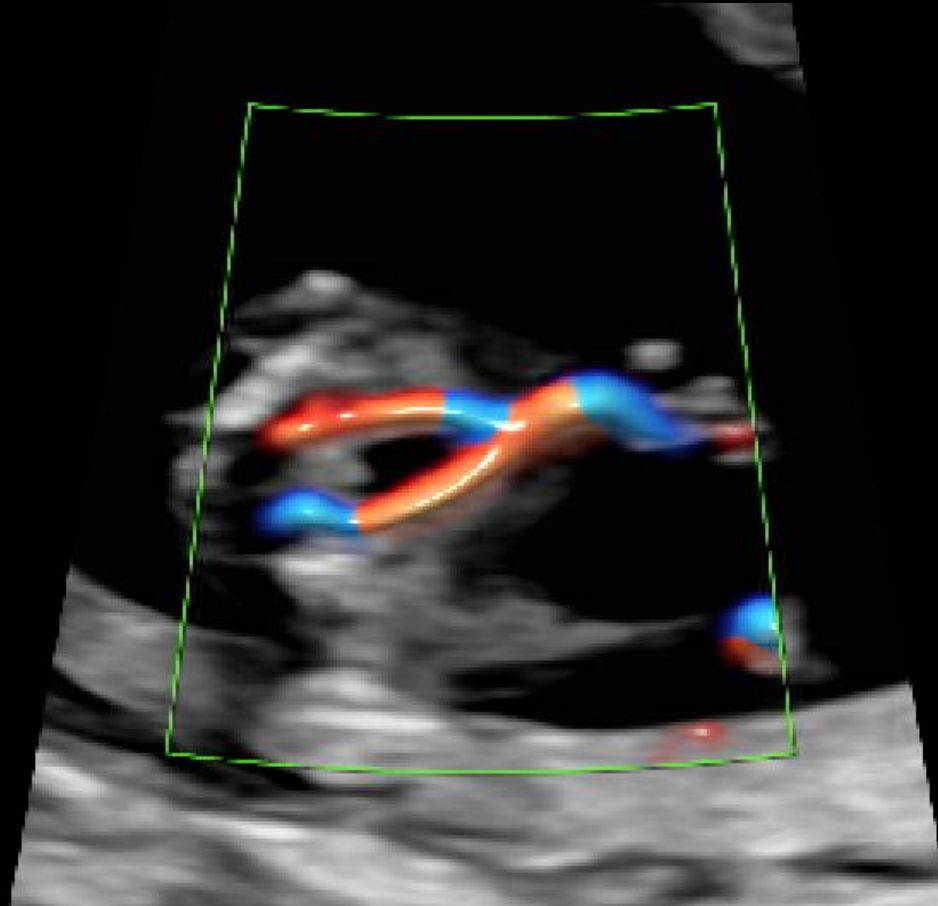


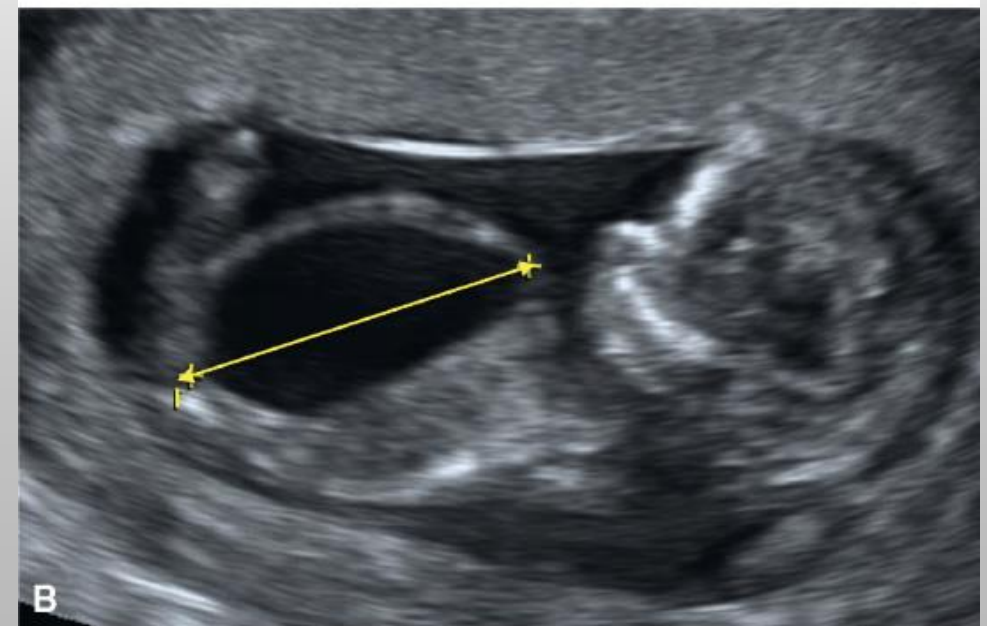
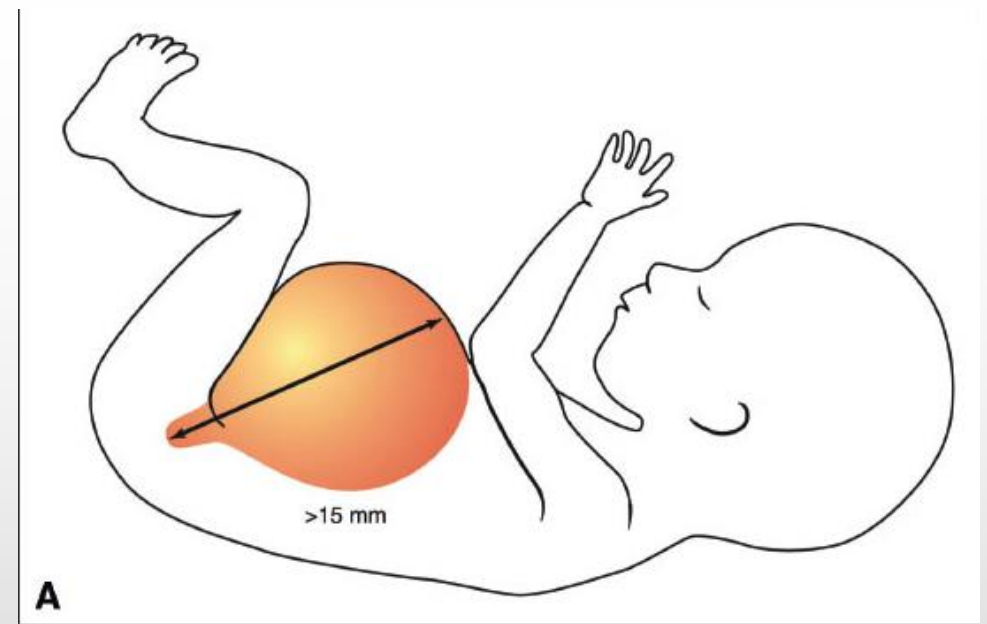
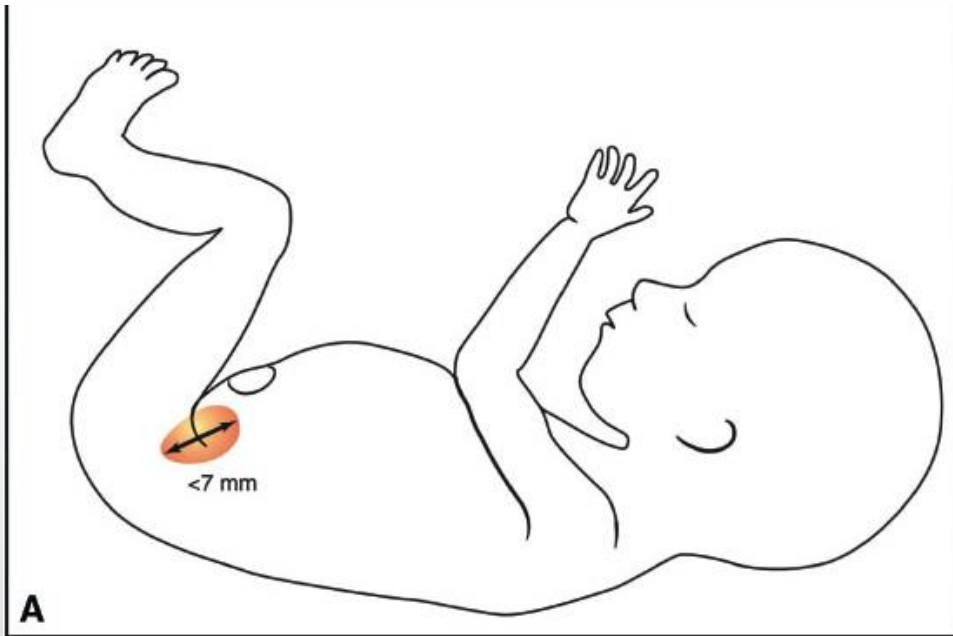
Gastroschisis



Omphalocele

Abdominal: Axial imaging urinary bladder & umbilical arteries



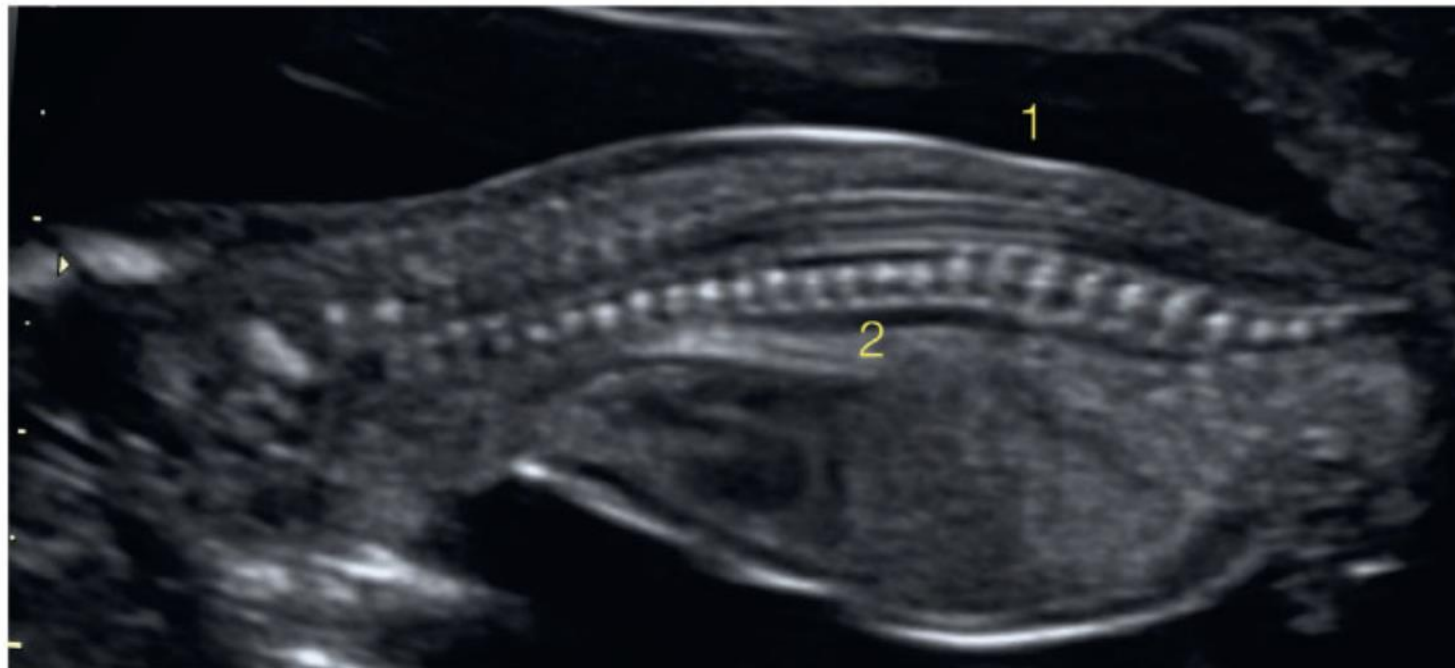
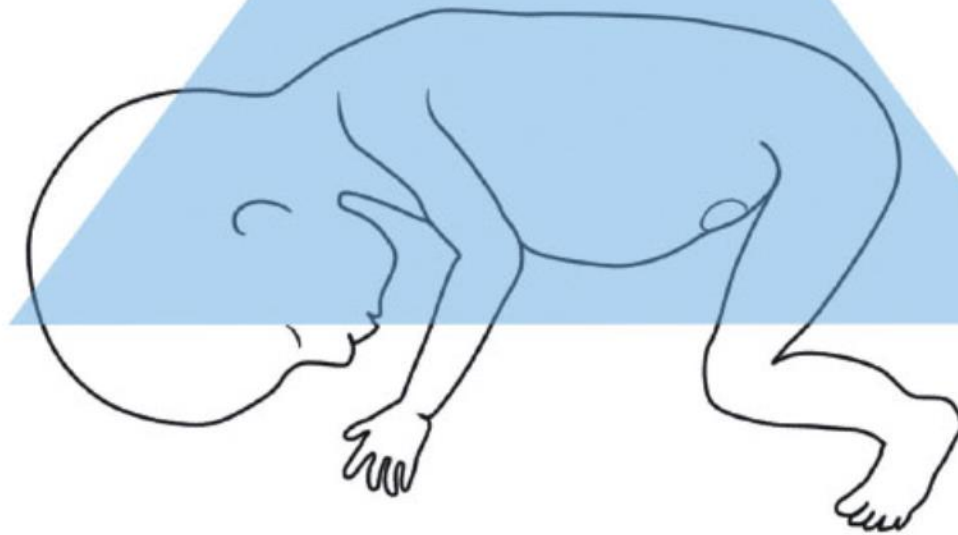






VI. SPINE IMAGING

1. Skin
2. Spine

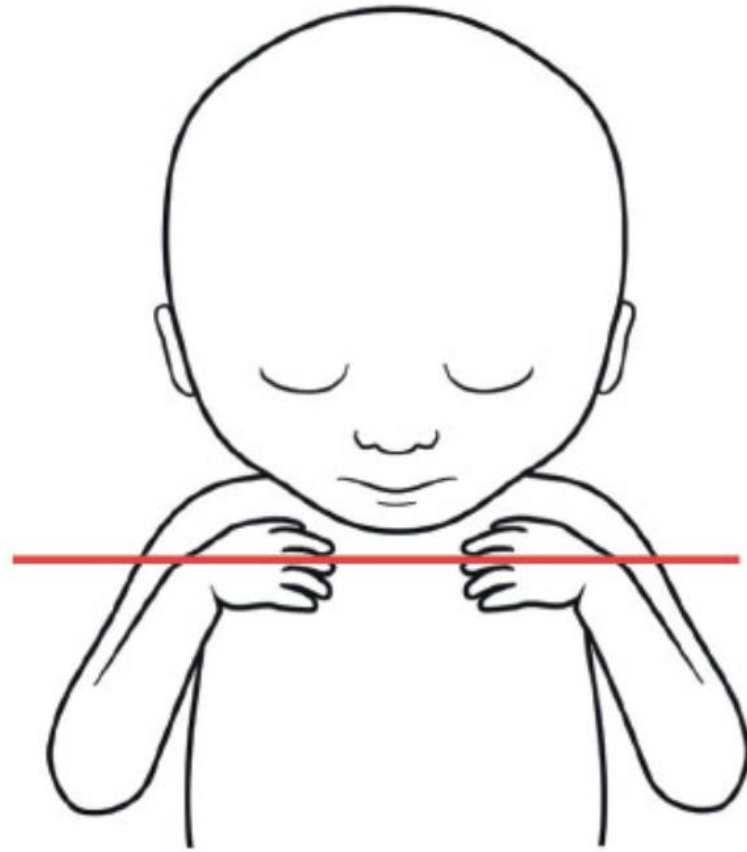




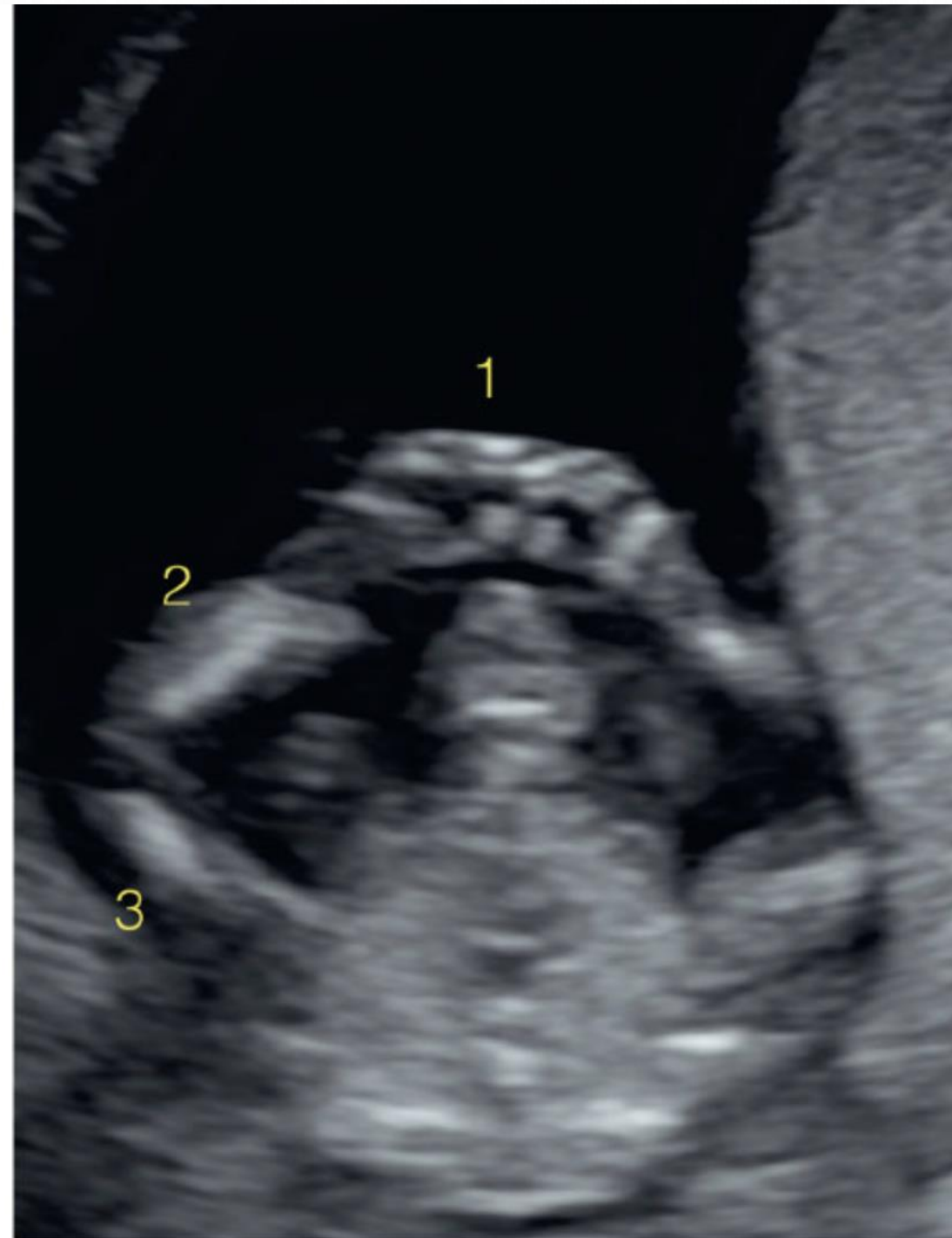


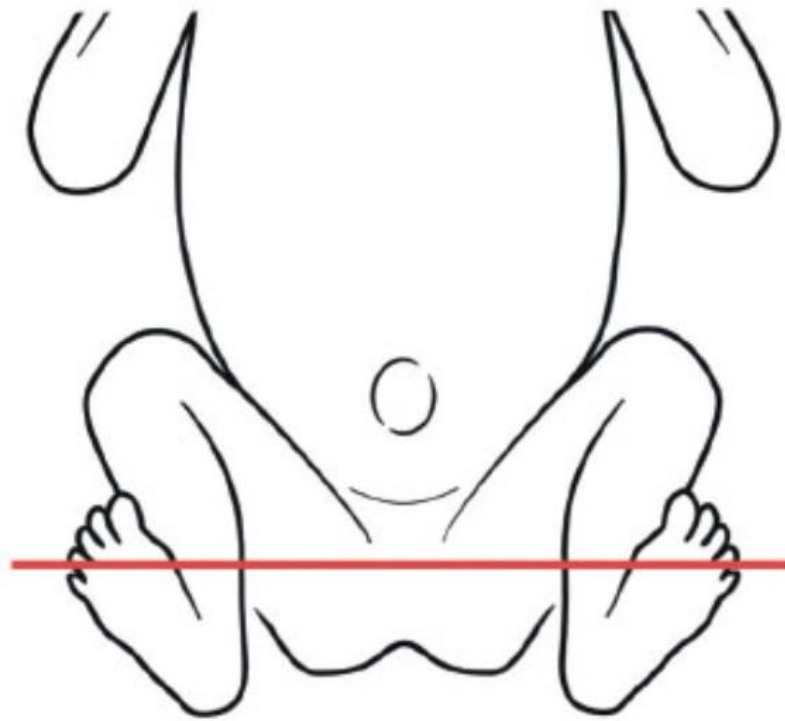


VII. LIMBS

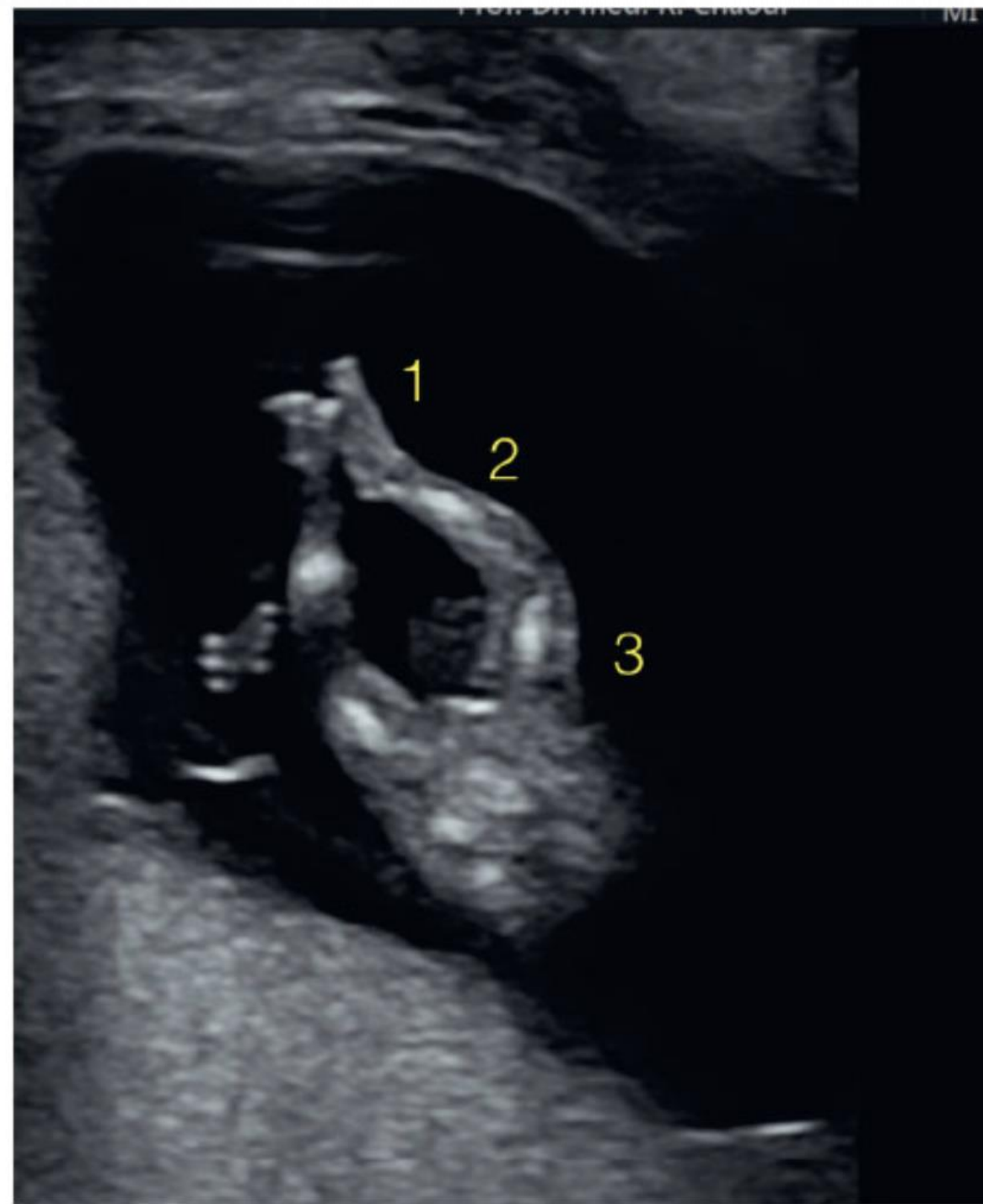


1. Hand and fingers
2. Lower arm
3. Upper arm



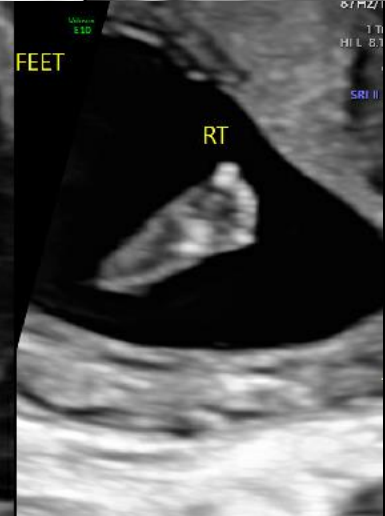


1. Foot
2. Lower leg
3. Upper leg



Extremities/limbs:

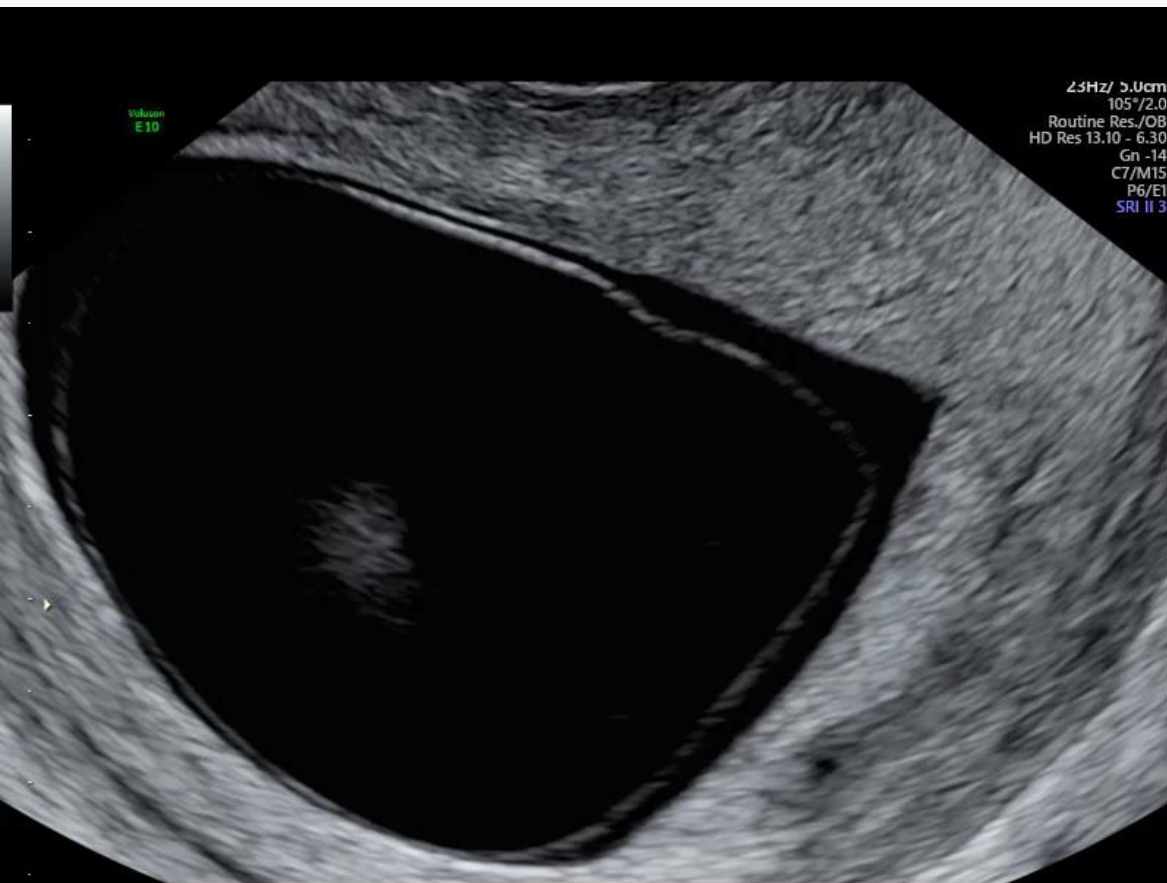
Upper with 3 segments/Lower with 3 segments





**CLINICAL
EXAMPLES:
WHAT DO
YOU THINK?**





- Omphalocele
containing heart

**Suspected ectopia
cordis**



CASE

- 34-year-old G2P1
- 13w0d gestation
- Abnormal cell-free DNA



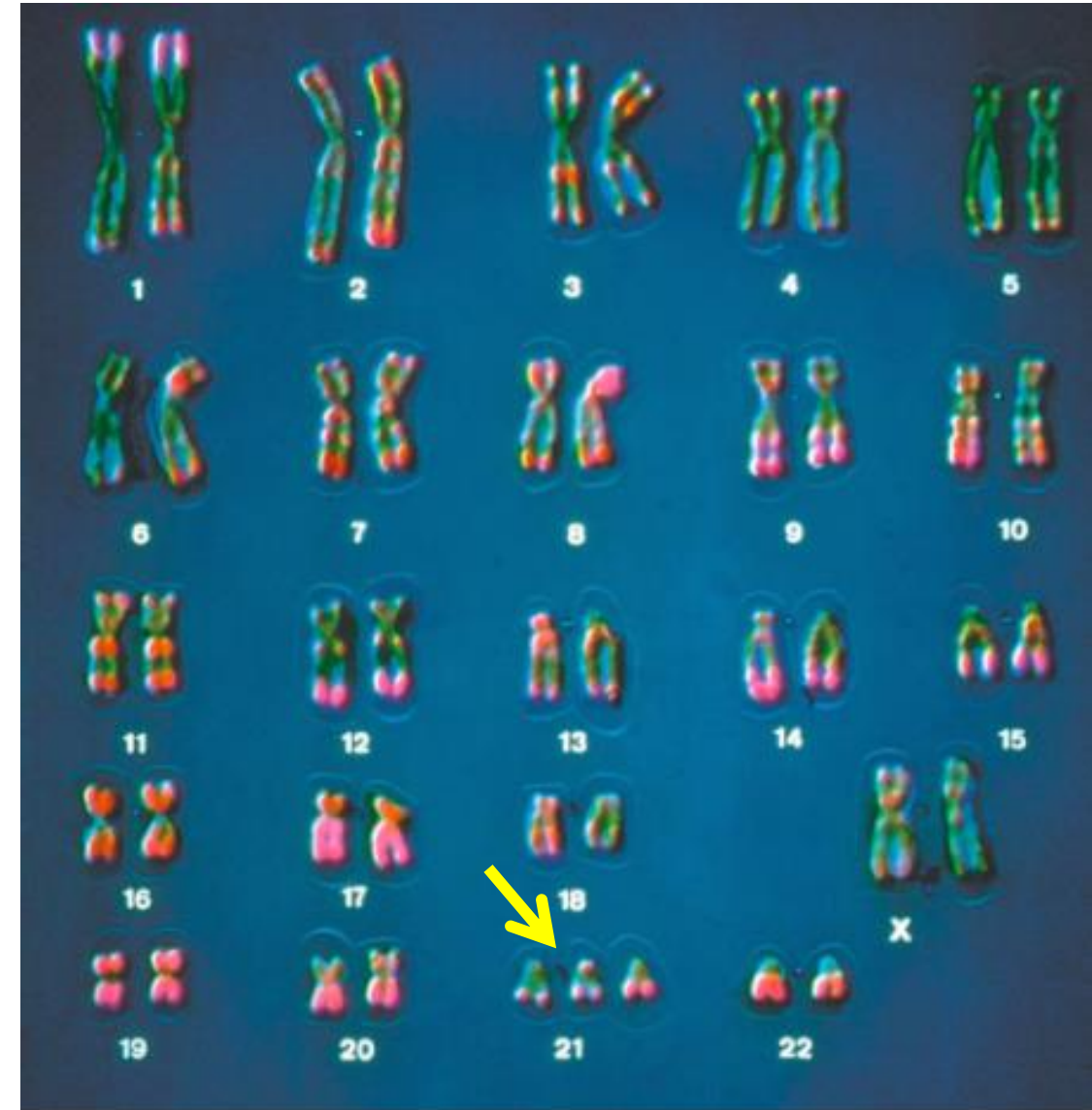
Courtesy Dr. Bryann Bromley





CASE 2 DIAGNOSIS

- TRISOMY 21



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CASE

29-year old G1

12w3d

cfDNA collected

No significant PMHx

No significant FHx

HEAD AND BODY IMAGES AT 12W3D

ASYMMETRY in head & body size

NORMAL head to body size



HEAD AND BODY IMAGES

12W3D, measures 11W4D



14W0D, measures 12W4D



14w0d: **Discordant** Head & Abdomen measurements



CASE DIAGNOSIS

- TRIPLOIDY

Asymmetric FGR with a
large head & small body
=> **Think triploidy**



CONCLUSION

- Autosomal trisomies represent a small portion of the total genetic & structural risk to the fetus, especially in younger patient
- An early anatomic scan, even at the standard level (minimal key elements) is a vital component of pregnancy care
- If pregnancy screening is limited to cf-DNA alone, many anomalies will not be identified until the second-trimester anatomic survey
- The detailed first trimester anatomic survey should be performed & interpreted by appropriately trained clinicians and sonographers

