

# Giant Omphalocele Prenatal Imaging & Diagnosis

The Hendren Project  
Abdominal Defects Center Webinar

Jill Stein, MD

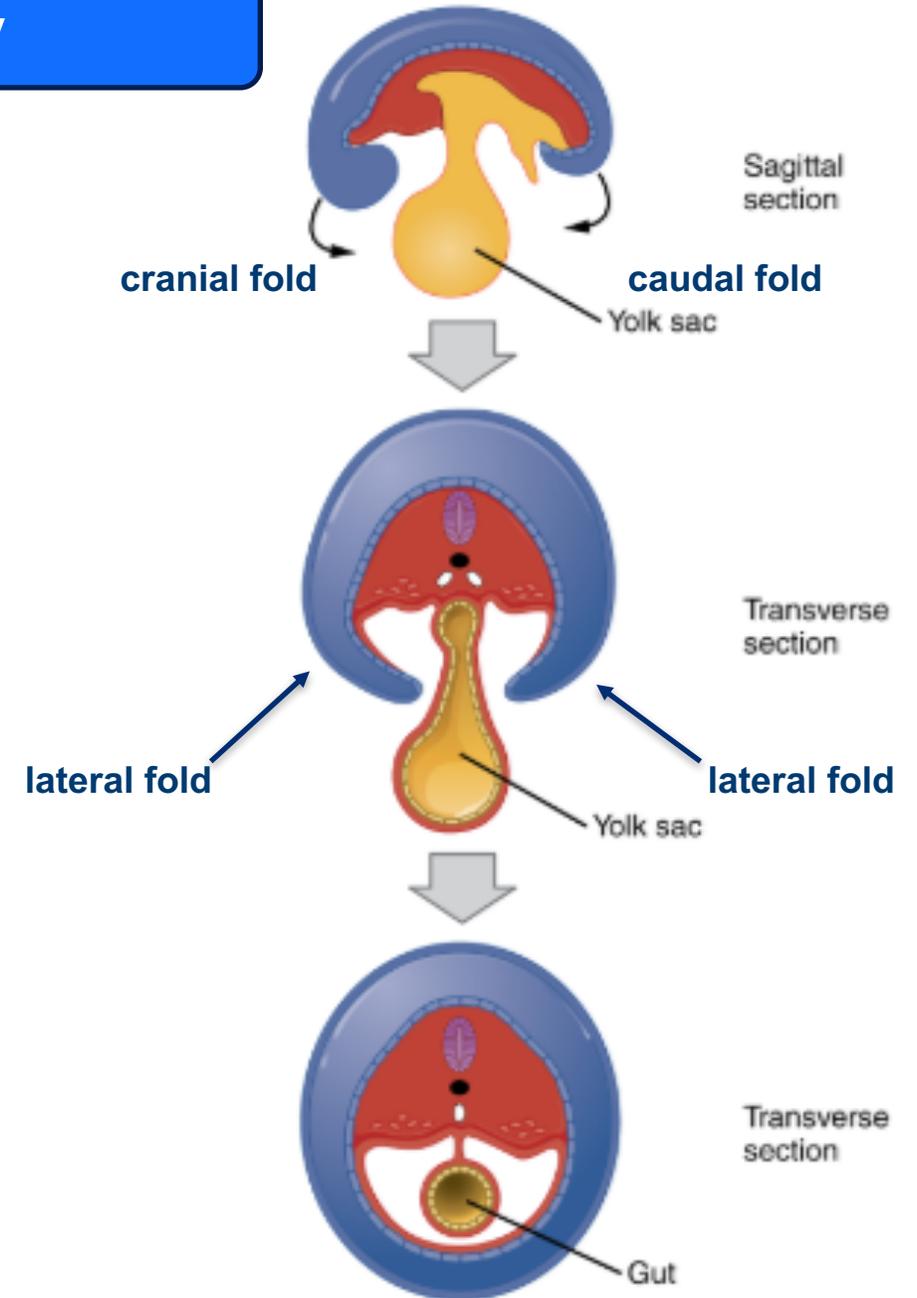
Rony Marwan, MD

Colorado Fetal Care Center  
Children's Hospital Colorado



# Embryology

- Ventral defect at umbilical ring
- Rectus muscles insert more laterally on costal margins
- Classically thought to result from migratory failure of lateral folds to form umbilical ring very early in embryogenesis

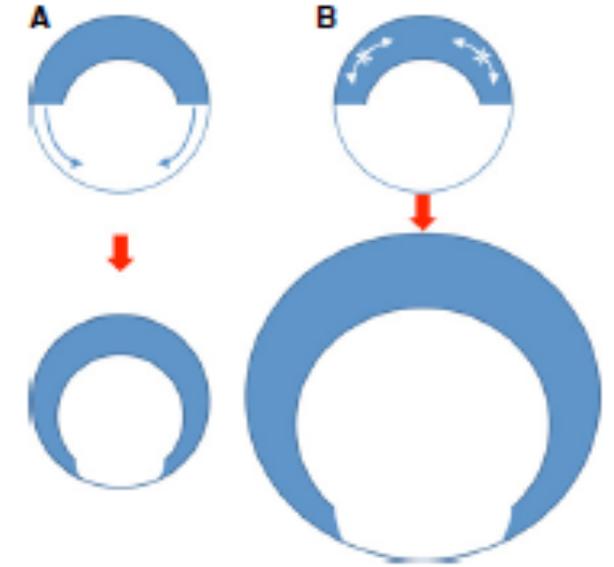


# Embryology

Recent embryologic data suggests differential dorsoventral growth determines ventral body wall closure

*J. Anat.* (2015) **227**, pp673-685

doi: 10.1111/joa.12380



## Development of the ventral body wall in the human embryo

Hayelom K. Mekonen,<sup>1</sup> Jill P. J. M. Hikspoors,<sup>1</sup> Greet Mommen,<sup>1</sup> S. Eleonore Köhler<sup>1</sup> and Wouter H. Lamers<sup>1,2</sup>

<sup>1</sup>Department of Anatomy & Embryology, Maastricht University, Maastricht, The Netherlands

<sup>2</sup>Tygat Institute for Liver and Intestinal Research, Academic Medical Center, Amsterdam, The Netherlands



# Embryology

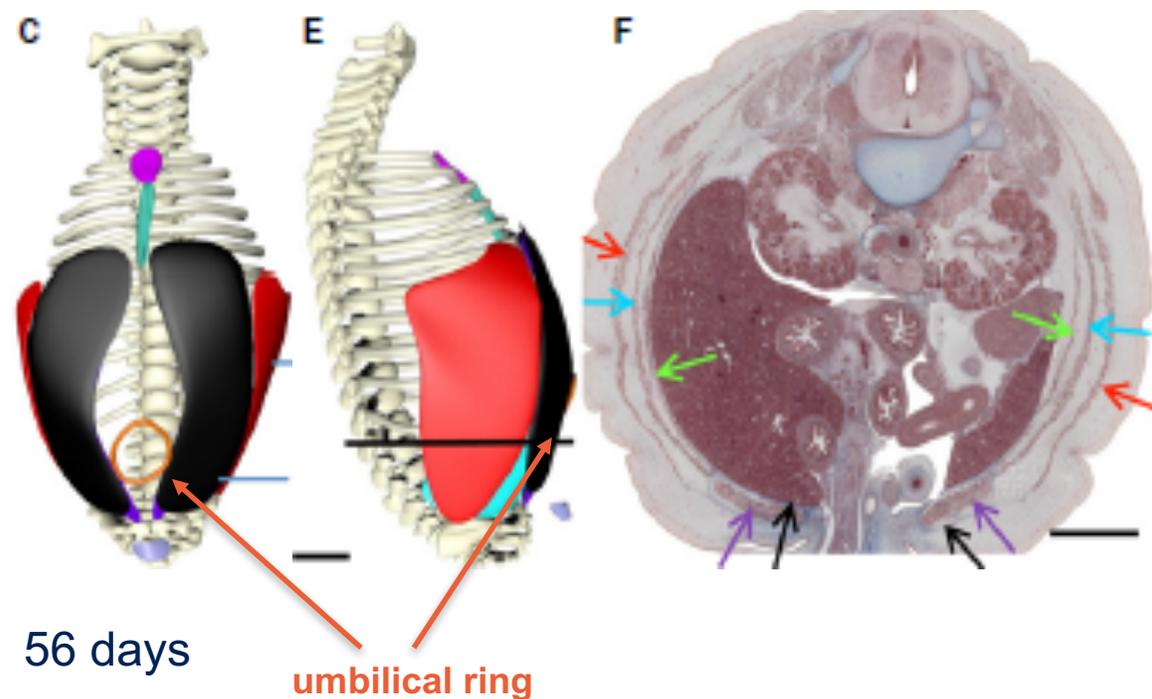
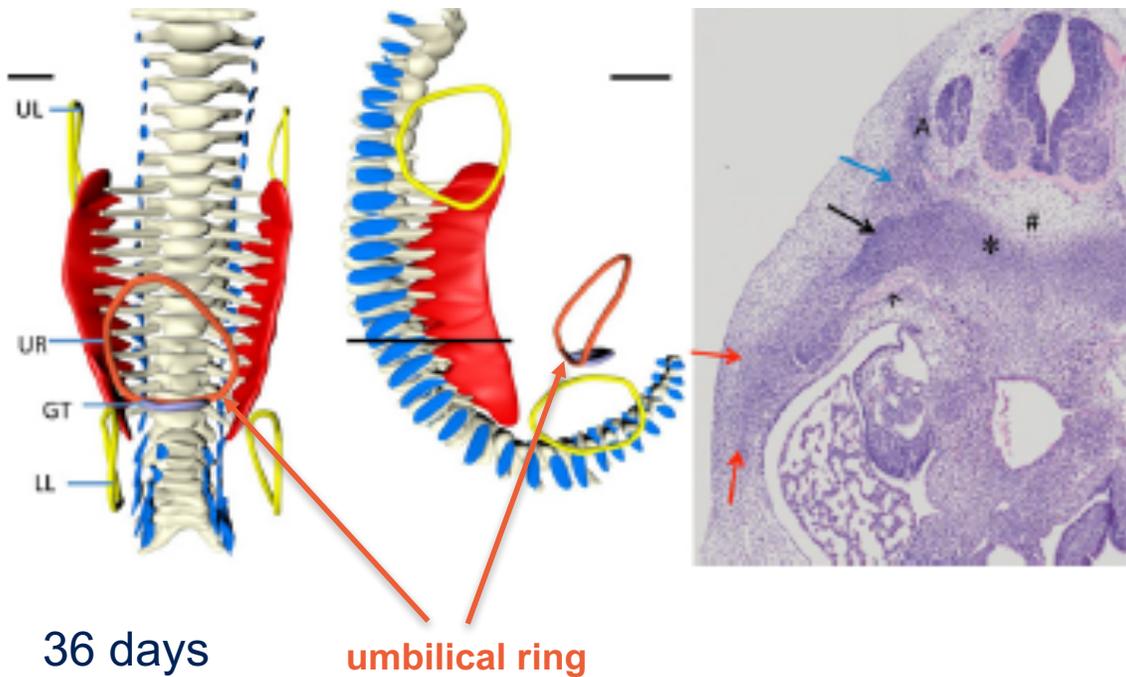
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### Rectus muscles reached the umbilicus at 8 weeks



## Anatomy

- Ventral abdominal wall defect covered by sac
  - Sac layers: peritoneum | Wharton's jelly | amnion
  - Sac may rupture (10-20% prenatally)
- Umbilical cord inserts eccentrically on defect
- Size: small (hernia of the cord) to giant
- Abdominal location:
  - Epigastric omphalocele → Pentalogy of Cantrell | cephalic fold
  - Central omphalocele → classic omphalocele | lateral folds
  - Hypogastric omphalocele → cloacal exstrophy | caudal folds



# Associated Anomalies

Omphaloceles commonly (50-70%) associated with anomalies

- Chromosomal abnormalities 20-30% | Trisomy 18,13, 21

- Beckwith-Wiedemann Syndrome

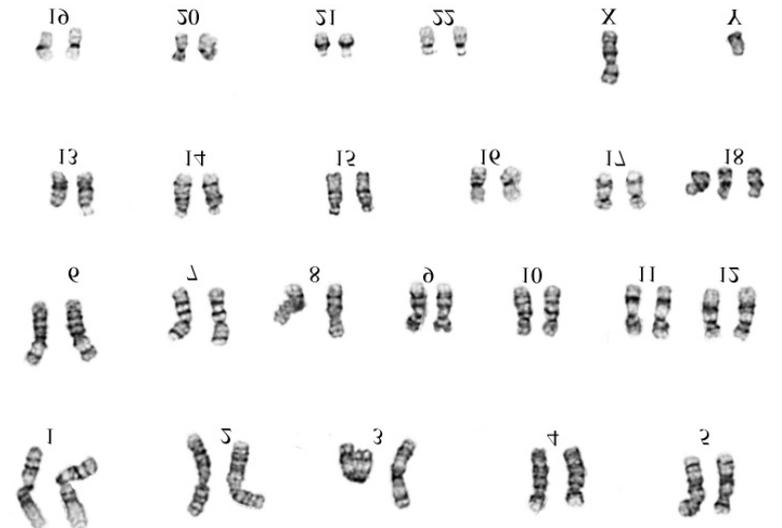
Beckwith-Wiedemann syndrome



Macroglossia    Umbilical hernia    Omphalocele

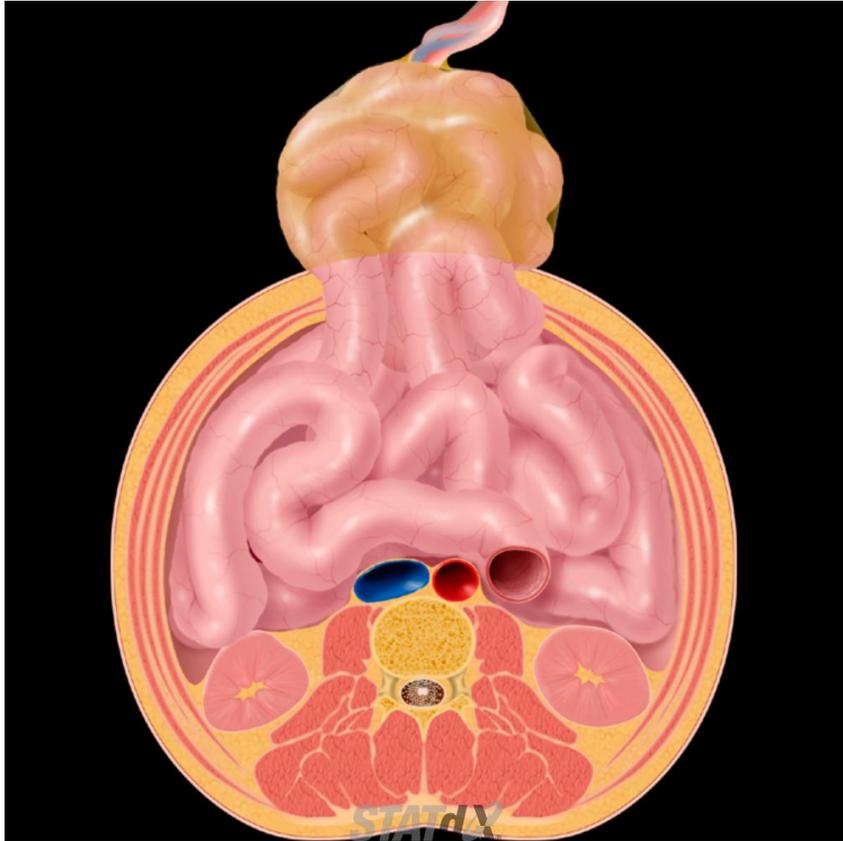
- Others

- Musculoskeletal
- Urinary
- Cardiac
- Bowel

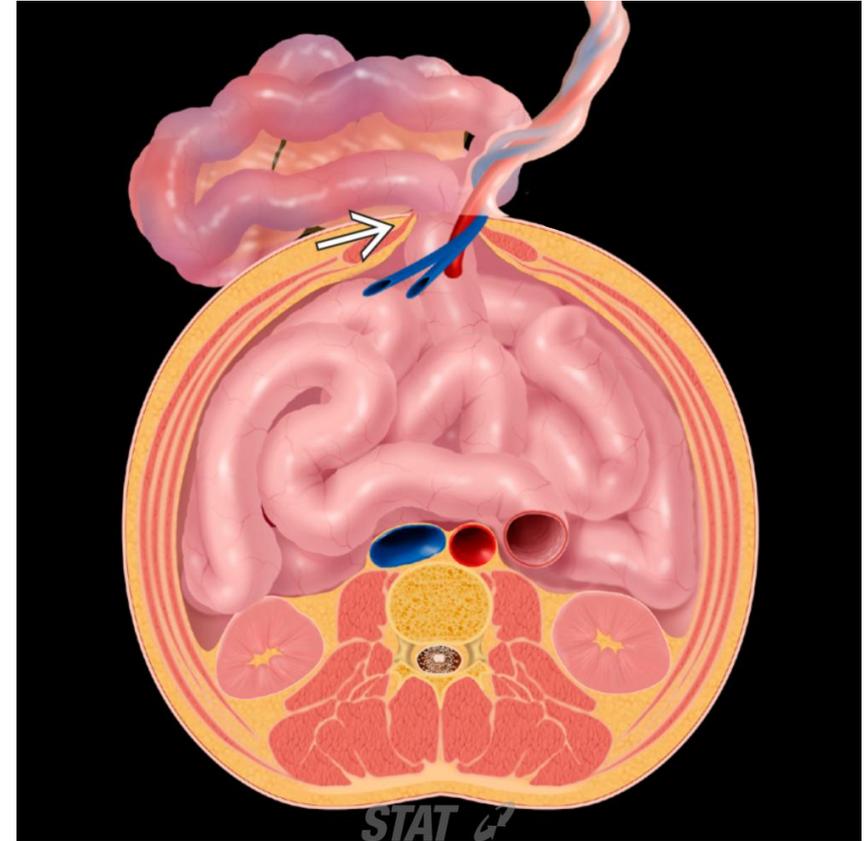


# Ventral Abdominal Wall Defects

## Omphalocele



## Gastroschisis



## Ventral Abdominal Wall Defects

- ✓ Omphalocele
- ✓ Gastroschisis



## Ventral Abdominal Wall Defects

- ✓ Omphalocele
- ✓ Gastroschisis
- ✓ Complex malformations
  - Pentalogy of Cantrell
  - OEIS Complex | Cloacal Exstrophy
  - Limb Body Wall Complex | Body Stalk Anomaly



## Ventral Abdominal Wall Defects

- ✓ Omphalocele
- ✓ Gastroschisis

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- ✓ Other ventral defects
  - Cephalic: Ectopia cordis
  - Caudal: Bladder exstrophy | urachal anomalies



## Ventral Abdominal Wall Defects

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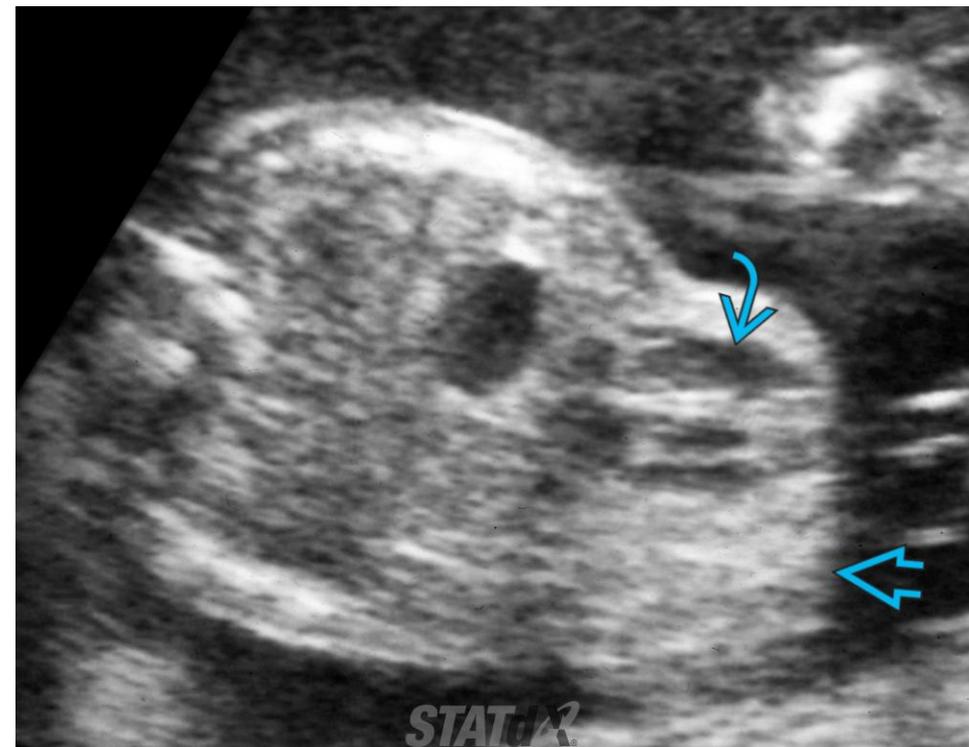
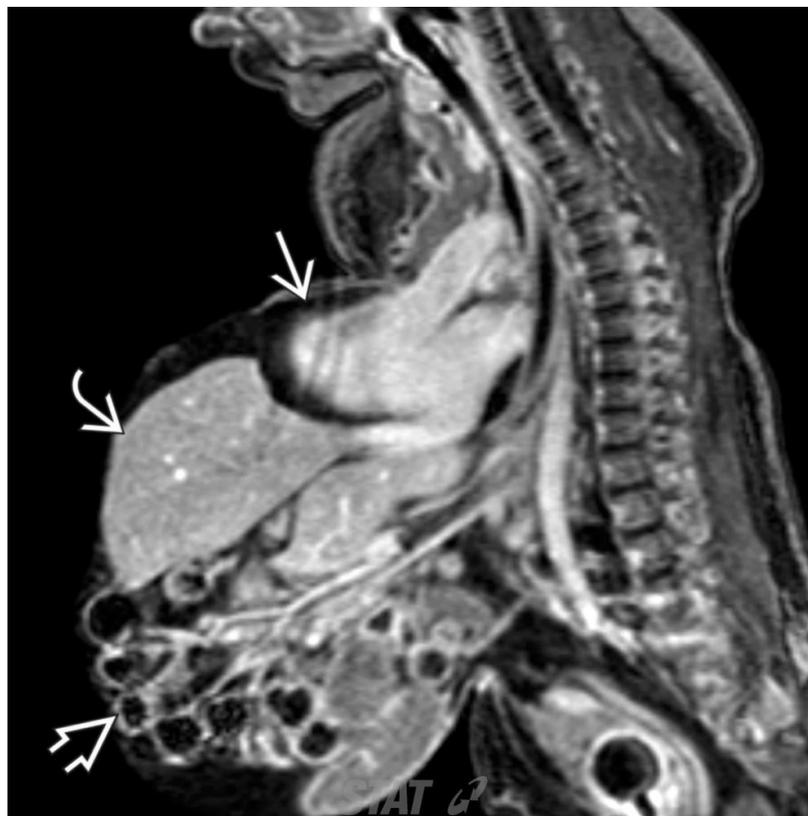
- ✓ Complex malformations
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- ✓ Imaging distinctions
  - Oligohydramnios
  - Fetal positioning



## Complex Malformations

### ✓ Pentalogy of Cantrell

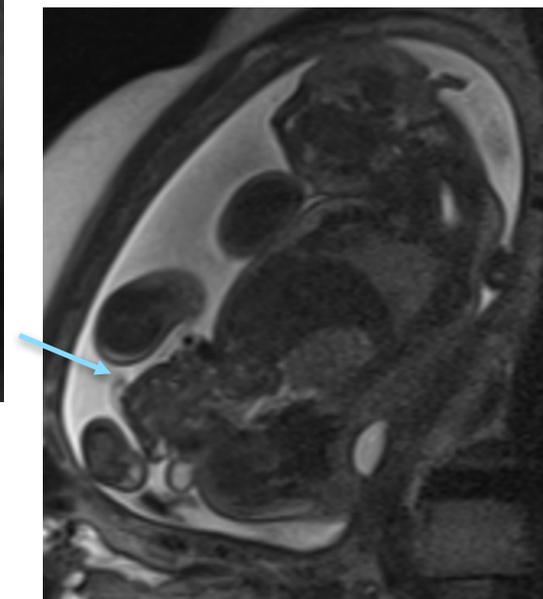
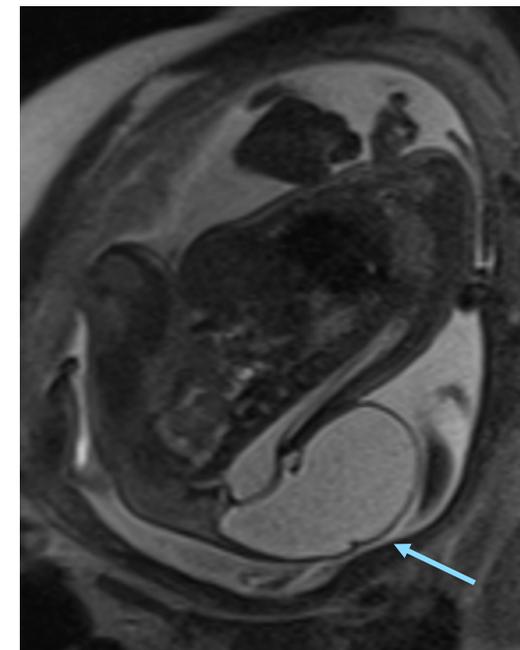
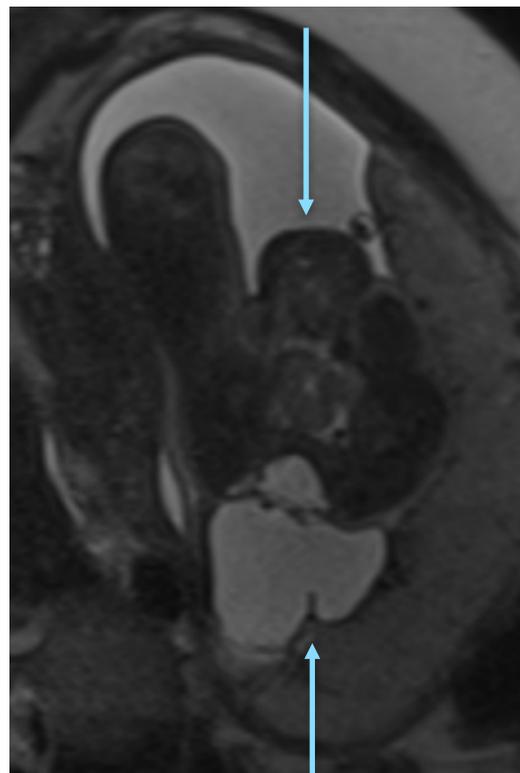
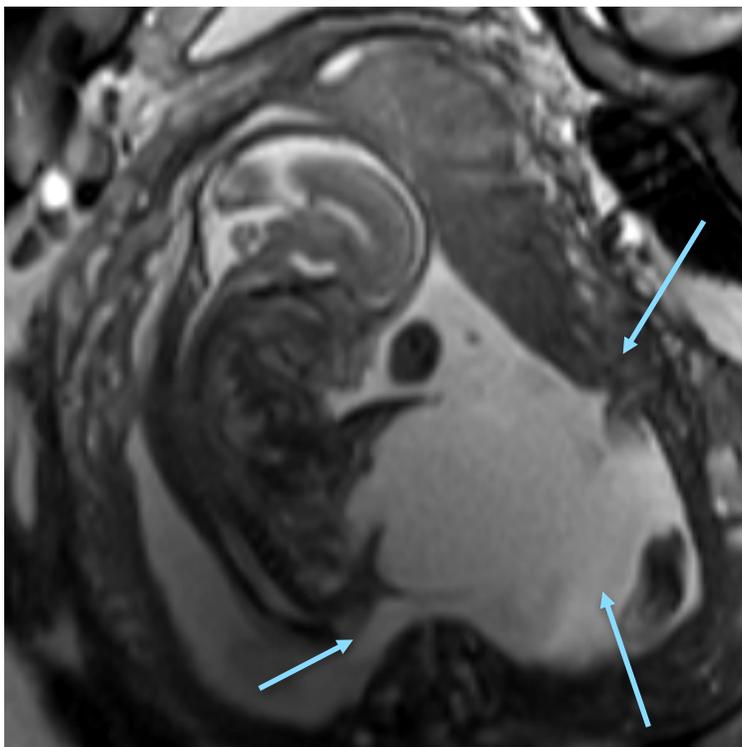
- Omphalocele
- Anterior diaphragmatic hernia
- Distal partial sternal defect
- Pericardial defect
- Cardiac defect



## Complex Malformations

### ✓ OEIS Complex

- Omphalocele
- Cloacal exstrophy
- Imperforate anus
- Spinal defects



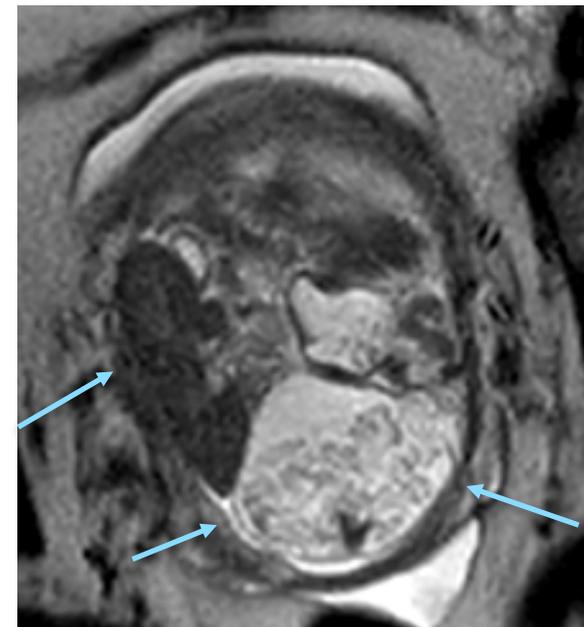
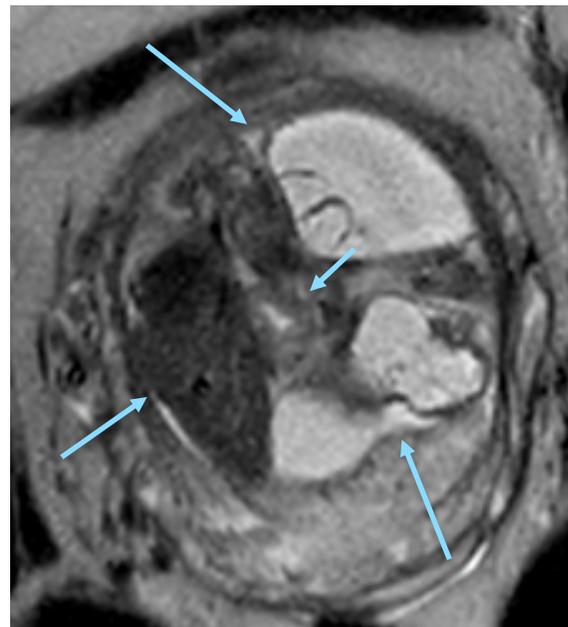
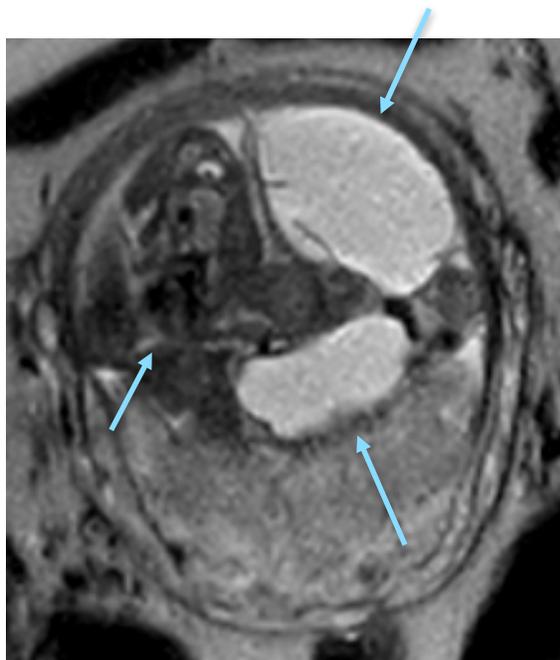
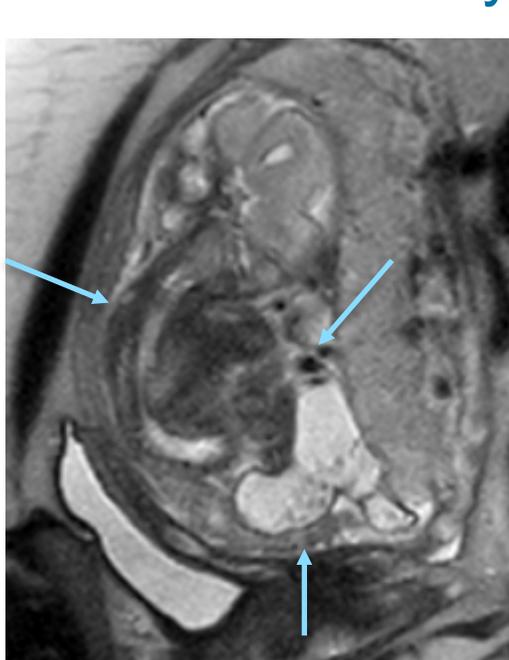
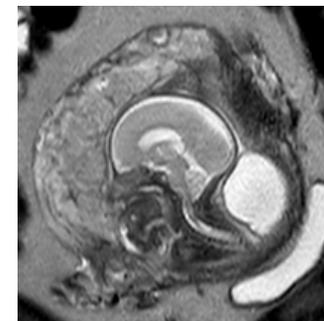
## Complex Malformations

### ✓ Limb Body Wall Complex

- Neural tube defect
- Ventral wall defect
- Limb anomaly

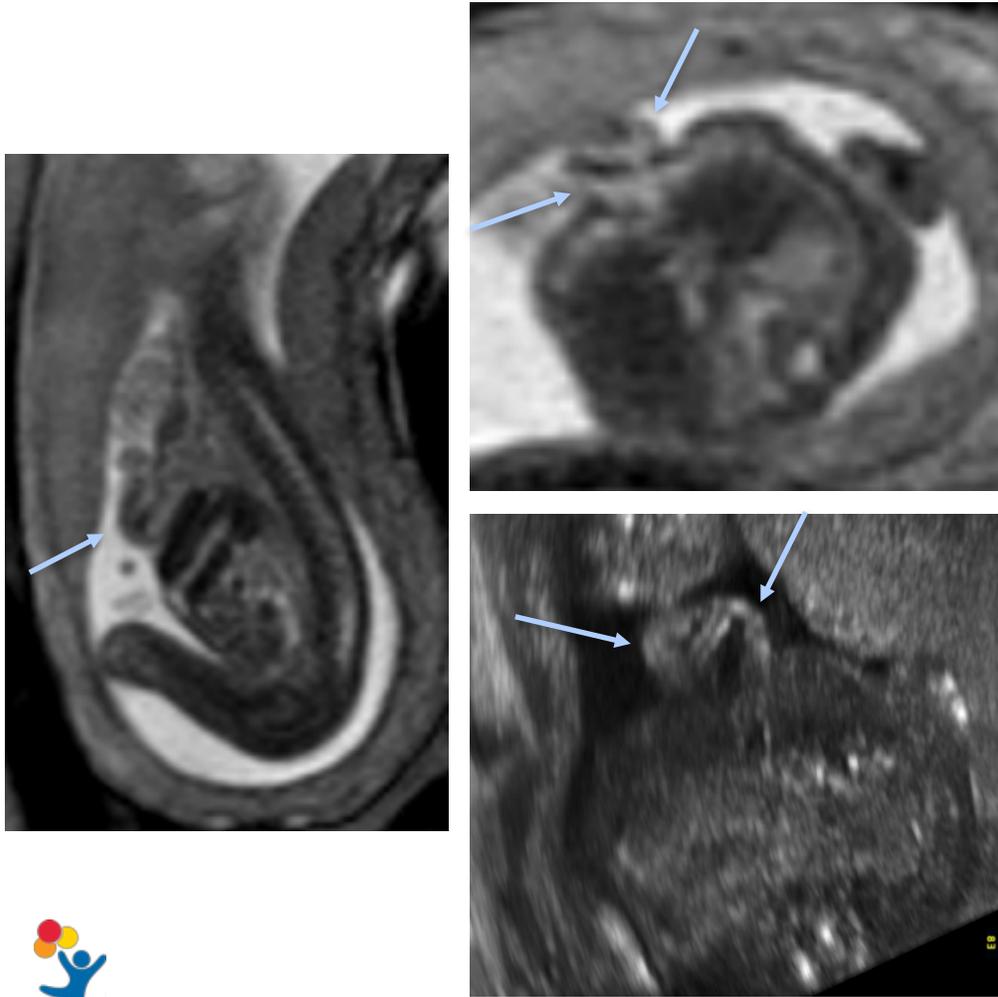
### ✓ Body Stalk Anomaly

- Large abdominal defect
- Abnormal short umbilical cord
- Severe kyphoscoliosis

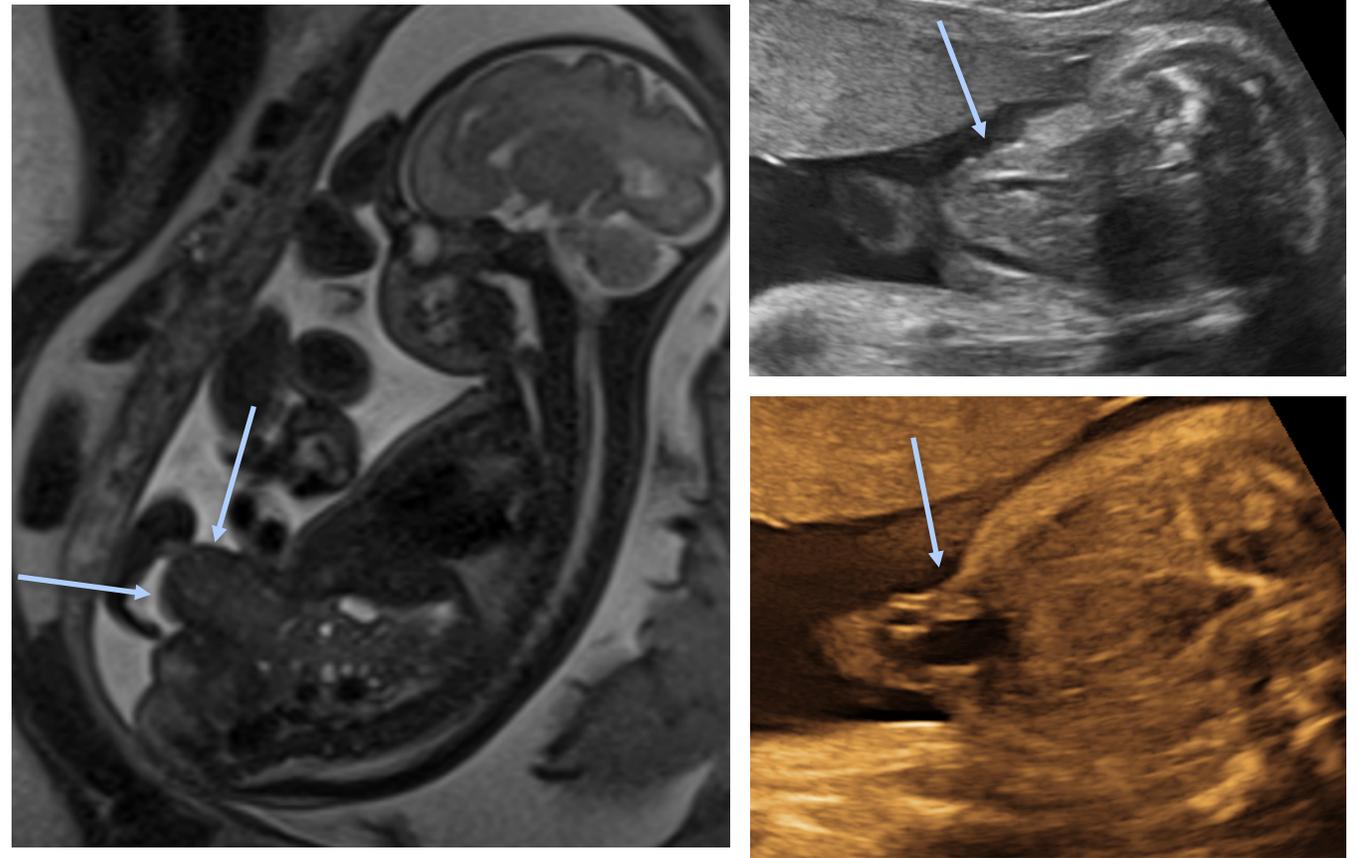


## Other Ventral Defects

### ✓ Ectopia cordis

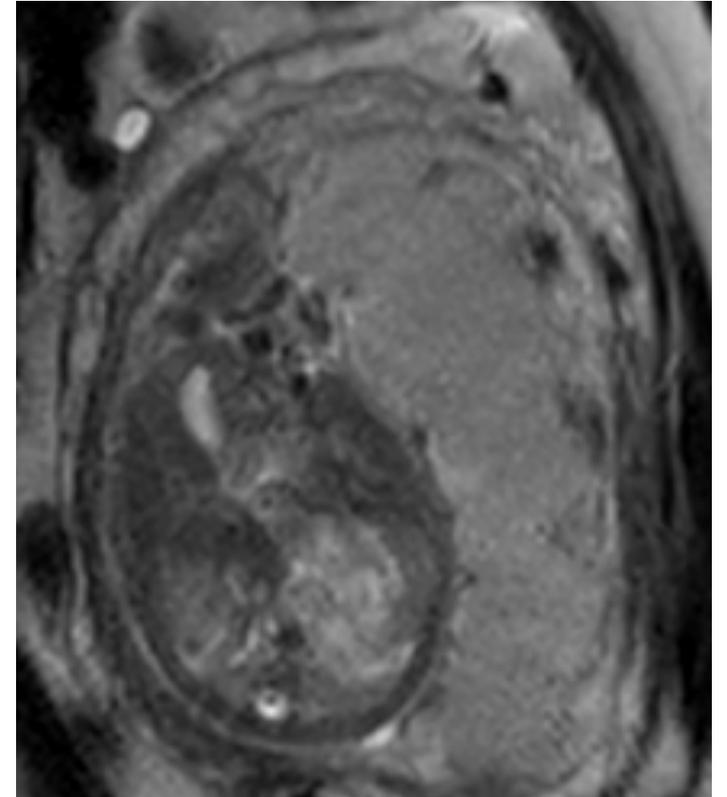
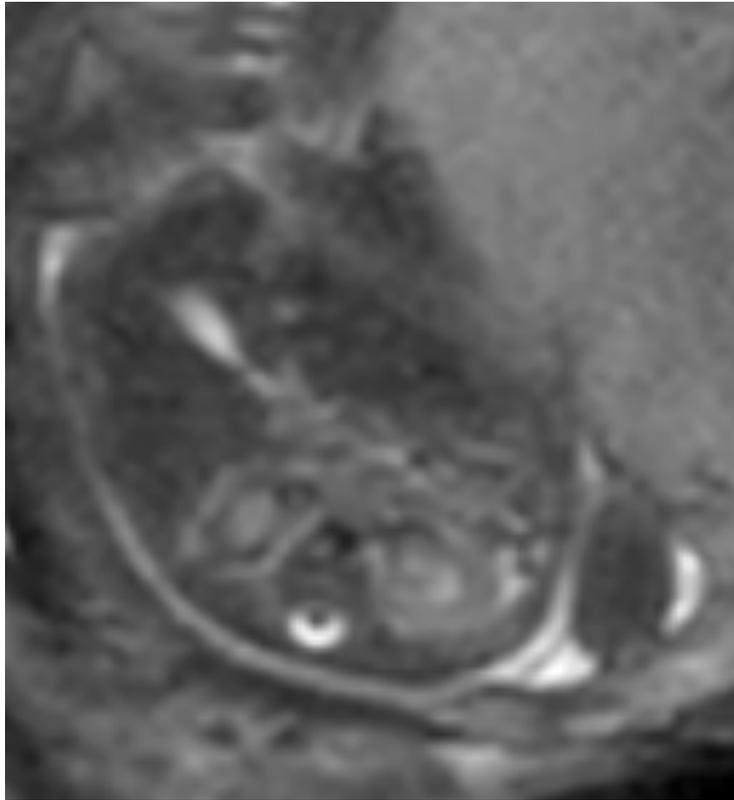


### ✓ Bladder exstrophy



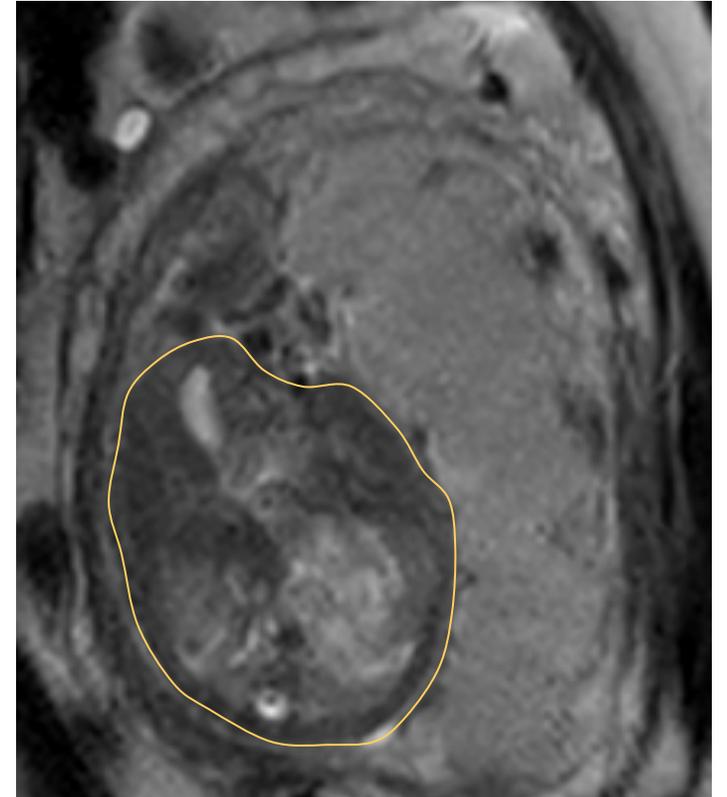
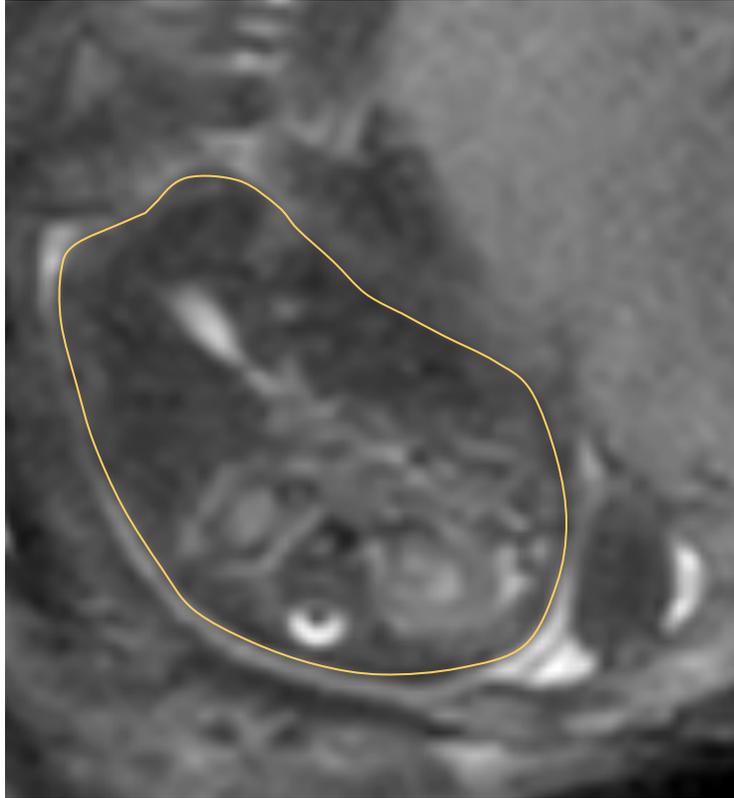
## Imaging Distinctions

### ✓ Oligohydramnios | fetal positioning



## Imaging Distinctions

✓ Oligohydramnios | fetal positioning



# Giant Omphalocele

## Classification

- Small
- Giant (GO) | lack of consensus regarding definition
  - Defect >5 cm
  - >50% liver
- Ruptured



Historically, GO have been defined by various criteria:

- diameter of sac
- diameter of abdominal wall defect
- inability to primarily close the defect
- tissue defect >5 cm
- sac containing liver
- volume disproportion b/w abdominal viscera and cavity



## Giant Omphalocele

### Physiologic & Anatomic Sequelae

- Pulmonary Hypoplasia
- Pulmonary Hypertension
- Systemic Hypertension
- Inguinal hernias and undescended testes
- GERD and feeding difficulties



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## Fetal MRI-calculated lung volumes

- Observed/expected total lung volume (O/E TLV) calculated using normative data by gestational age
  - *Rypens et al.* 2001
  - *Meyers et al.* 2018 | 19-22 weeks GA
- Conclusion: MRI-based O/E TLV <50% predictive of increased postnatal morbidity

Fetal Diagnosis  
and Therapy

Original Paper

Fetal Diagn Ther 2012;31:248–253  
DOI: [10.1159/000334284](https://doi.org/10.1159/000334284)

Received: July 6, 2011  
Accepted after revision: September 26, 2011  
Published online: April 27, 2012

### Fetal MRI-Calculated Total Lung Volumes in the Prediction of Short-Term Outcome in Giant Omphalocele: Preliminary Findings

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Natalie E. Rintoul Mark P. Johnson Alan W. Flake N. Scott Adzick  
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The Center for Fetal Diagnosis and Treatment, The Children's Hospital of Philadelphia and The University of Pennsylvania School of Medicine, Philadelphia, Pa., USA



# Pulmonary Hypoplasia

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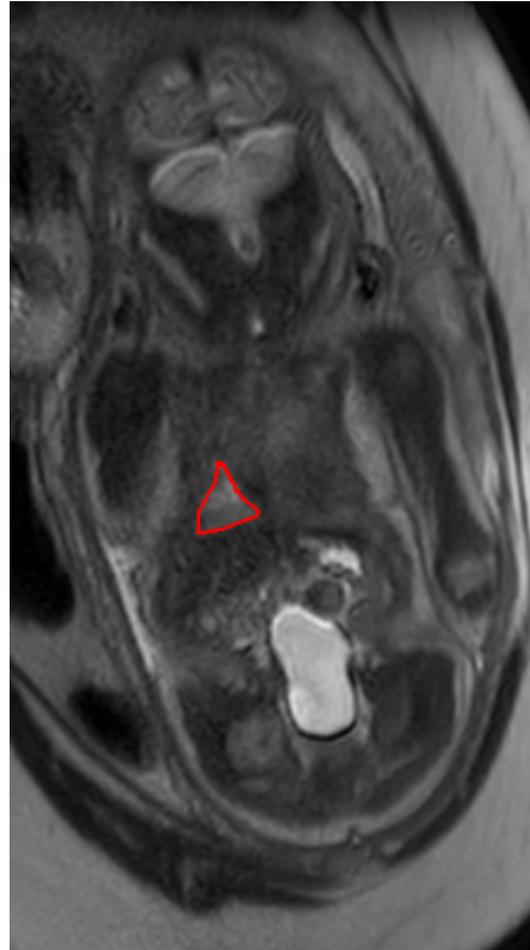
FLV: Descriptive Statistical Data for Gestational Age Classes

Age Class (wk)	FLV Range (mL)	Mean FLV (mL)	Median FLV (mL)	SD	Skewness	95% CI
21.0–25.0 ( <i>n</i> = 13)	16–48	26.15	24	9.15	1.18	13.00, 47.55
26.0–27.5 ( <i>n</i> = 29)	23–66	38.83	37	10.12	0.57	22.33, 63.27
28.0–30.0 ( <i>n</i> = 34)	29–89	52.97	53	14.20	0.59	29.73, 88.00
31.0–31.5 ( <i>n</i> = 26)	35–101	65.04	64	15.91	0.18	37.71, 105.53
32.0 ( <i>n</i> = 32)	38–109	70.22	67.5	18.16	0.41	40.30, 114.62
32.5–33.0 ( <i>n</i> = 34)	47–110	72.29	69	17.18	0.69	44.38, 111.73
33.5–35.0 ( <i>n</i> = 30)	52–129	80.73	77.50	24.32	0.73	43.34, 138.38
35.5–38.0 ( <i>n</i> = 16)	38–150	88.63	77.50	31.77	0.42	39.45, 175.58



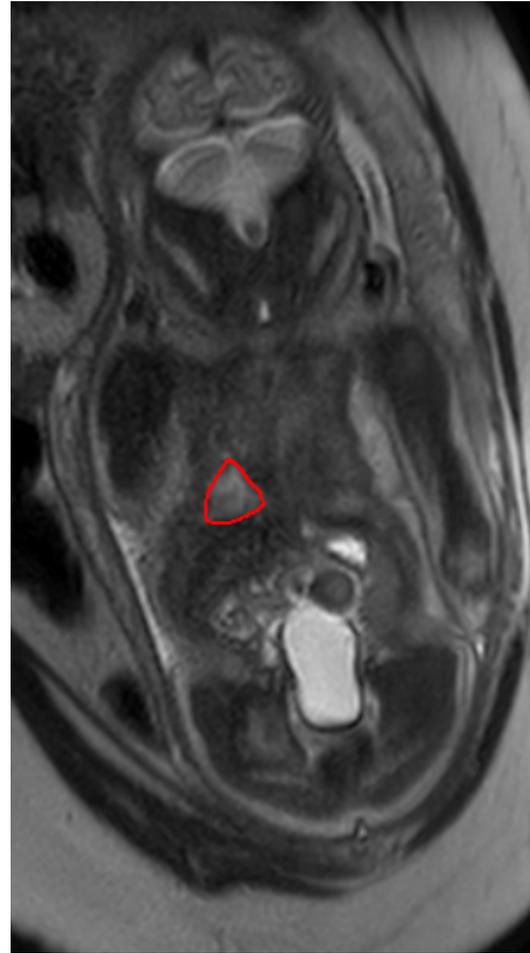
# Pulmonary Hypoplasia

## Fetal MRI-calculated lung volumes | *Technique*



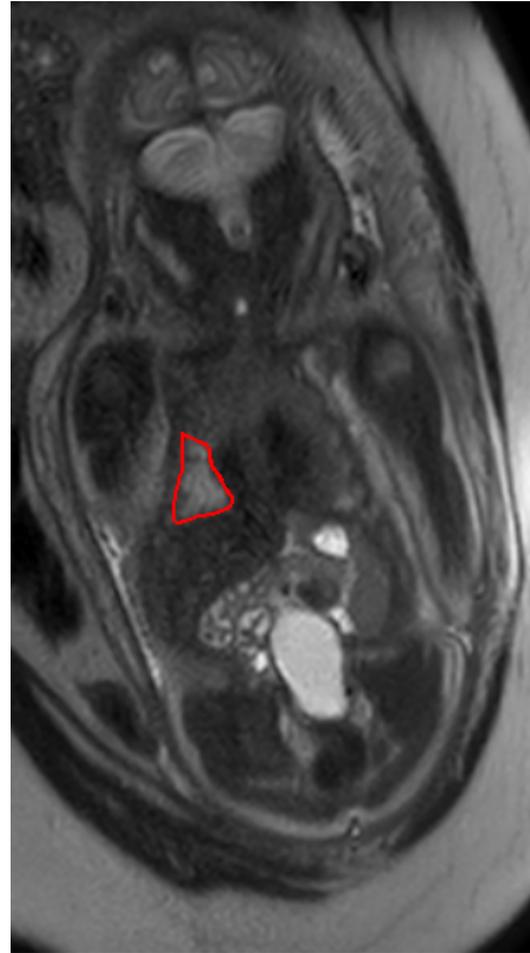
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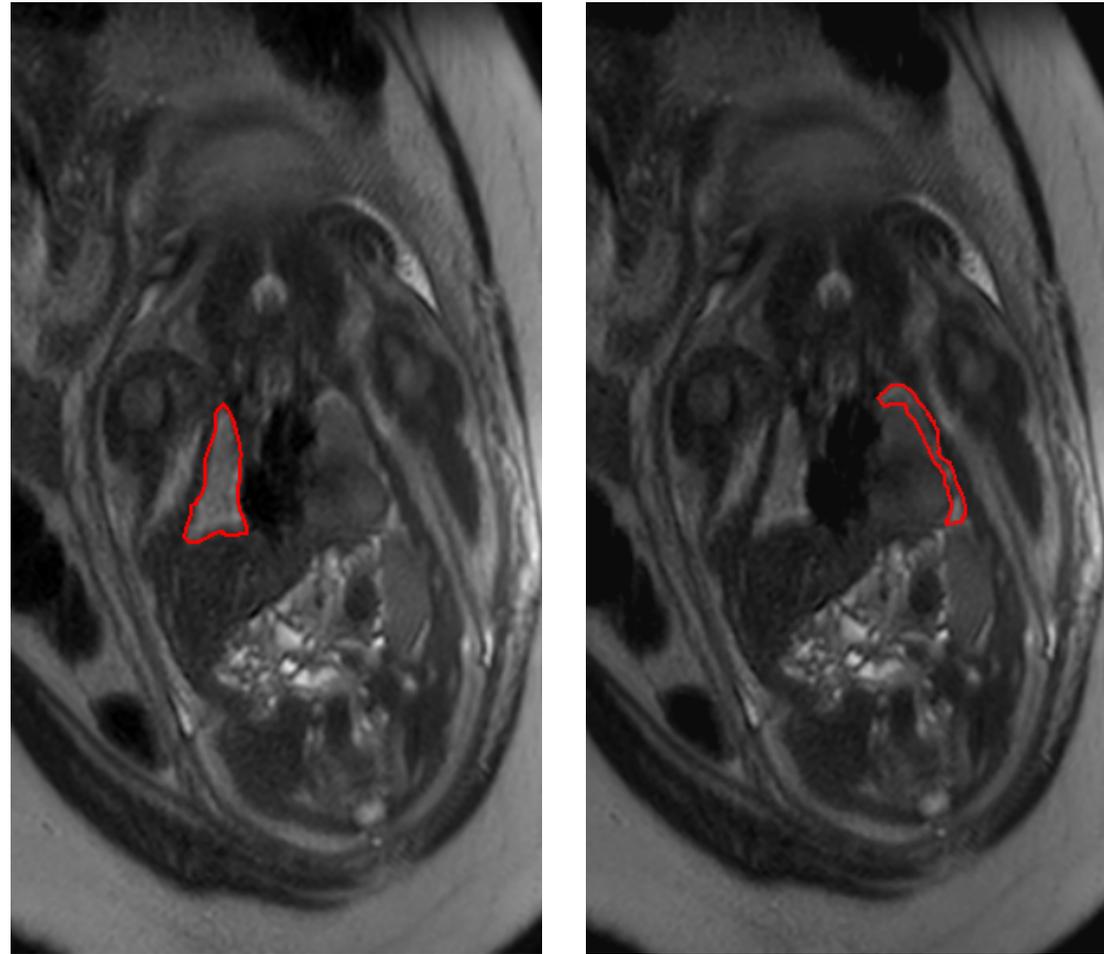
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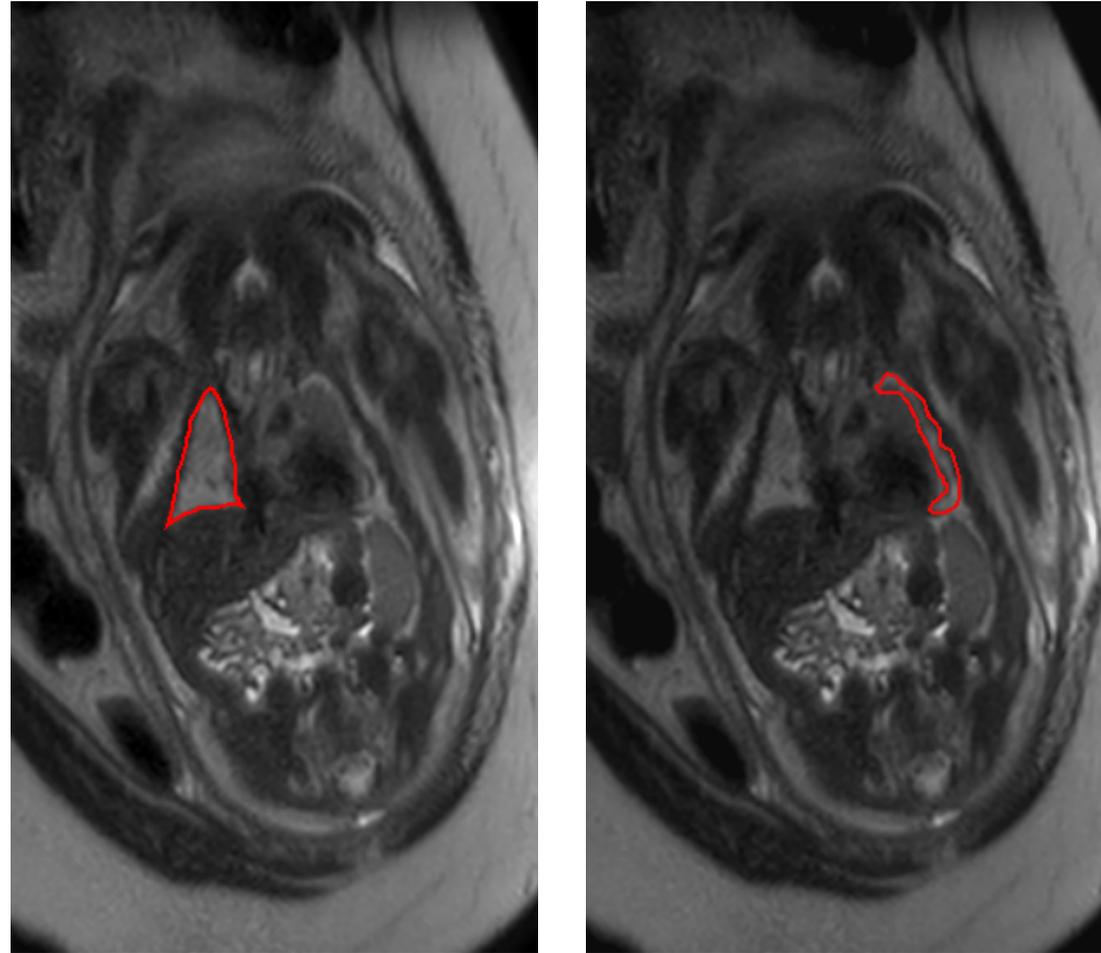
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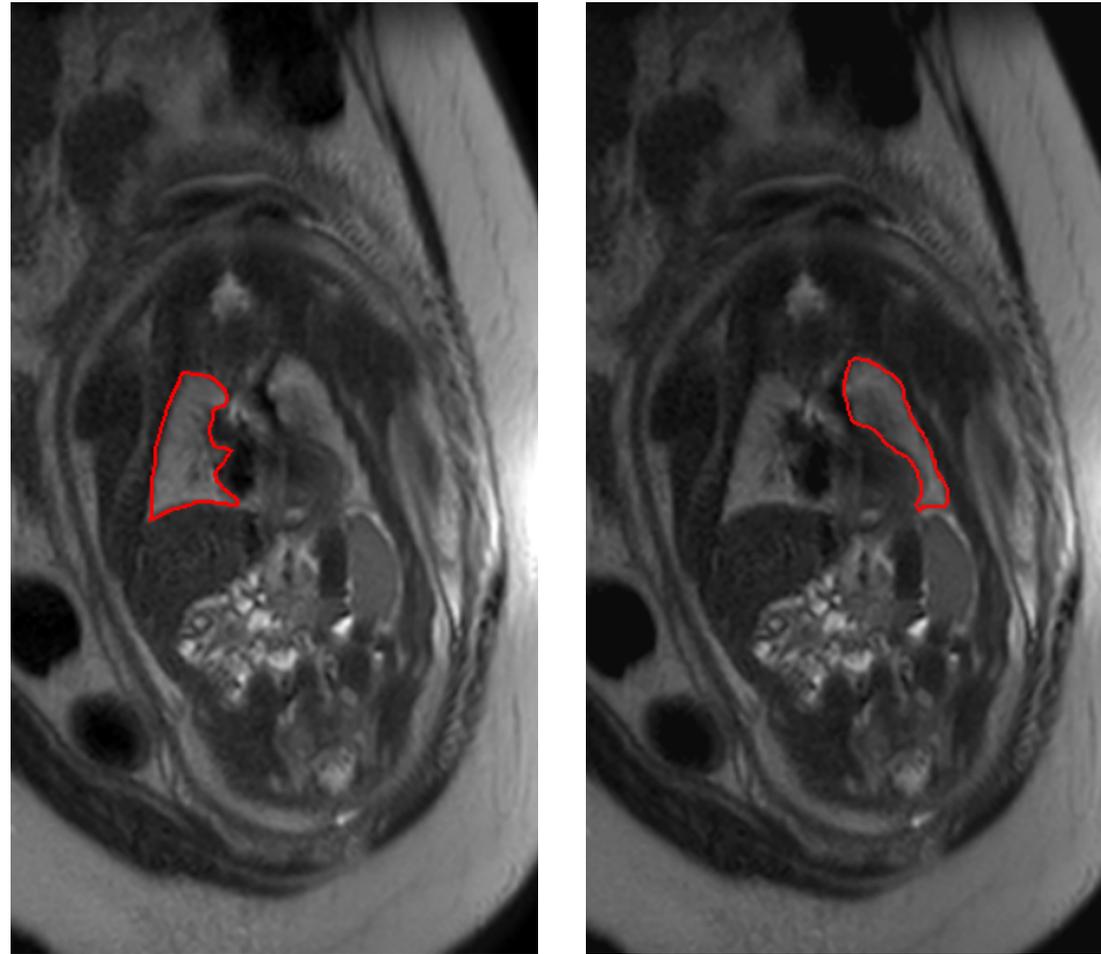
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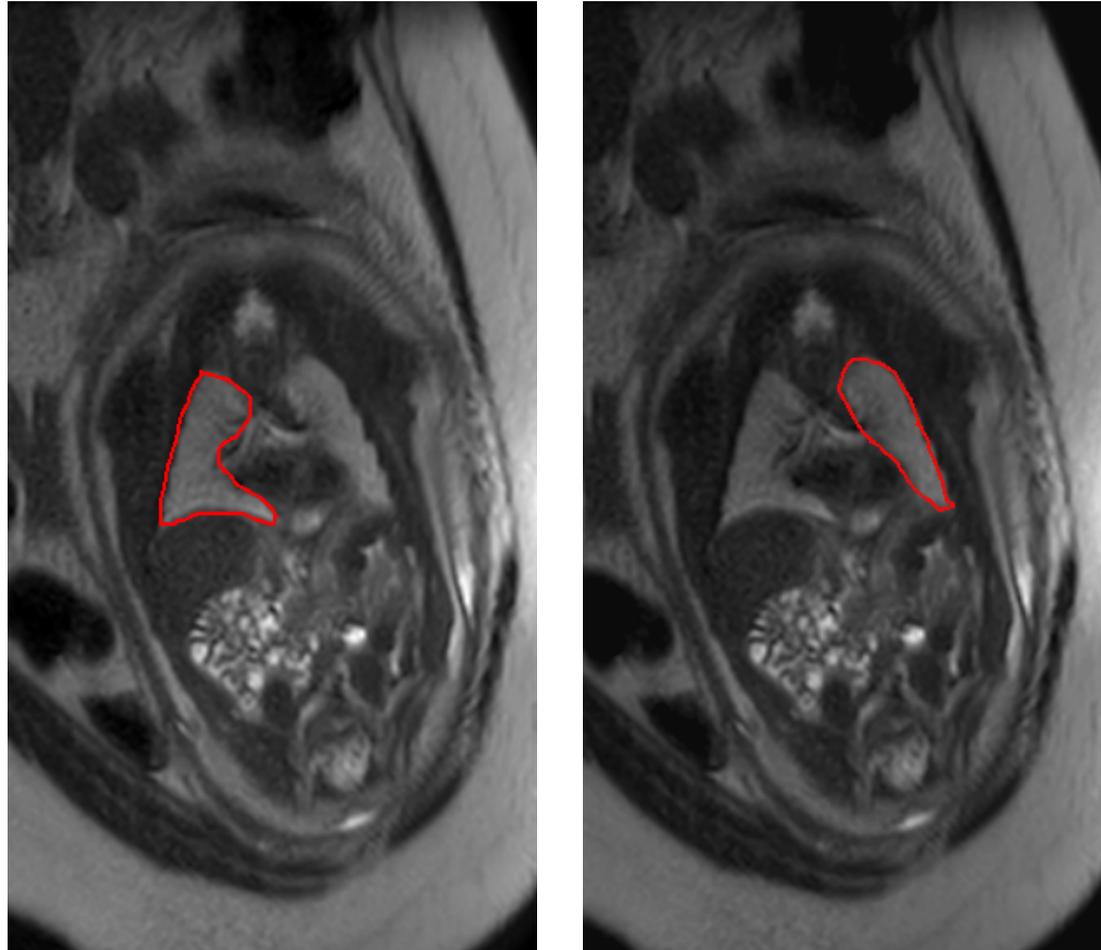
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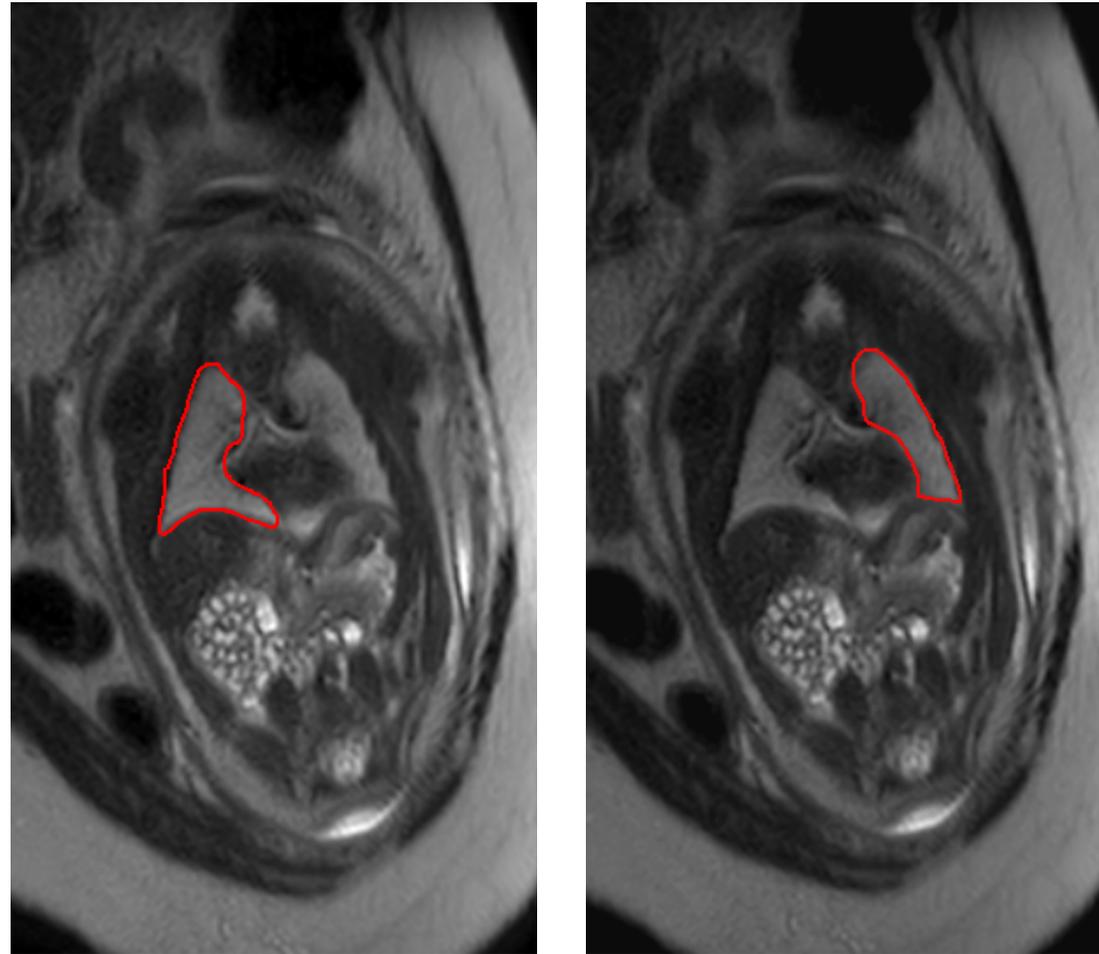
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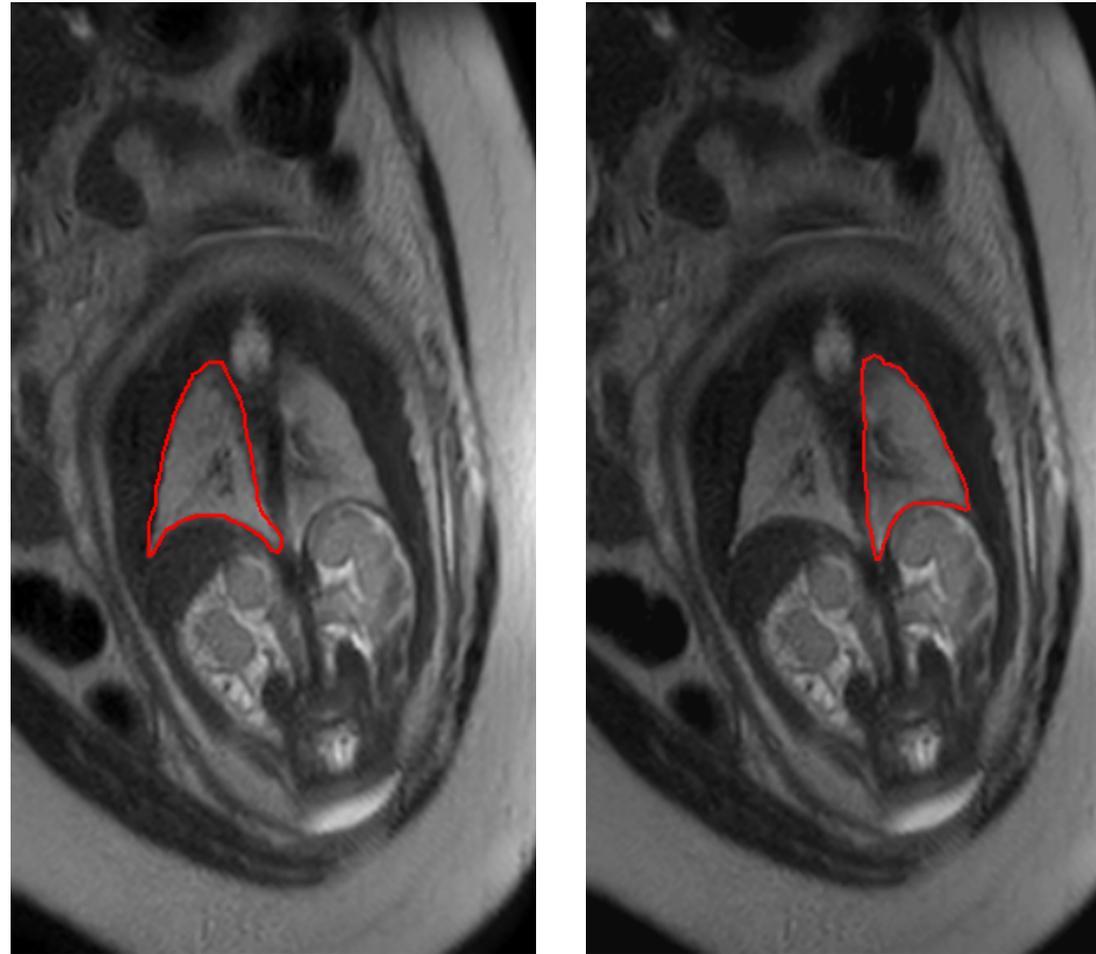
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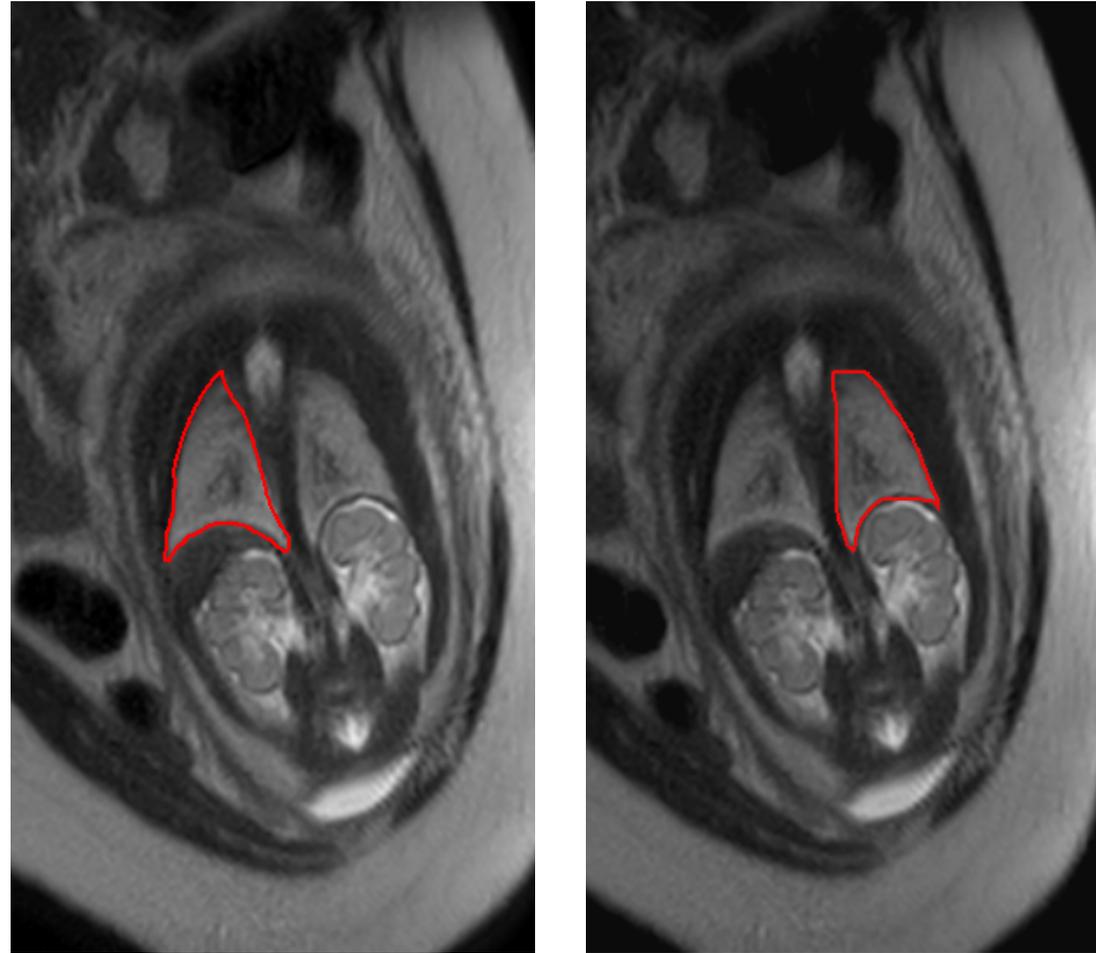
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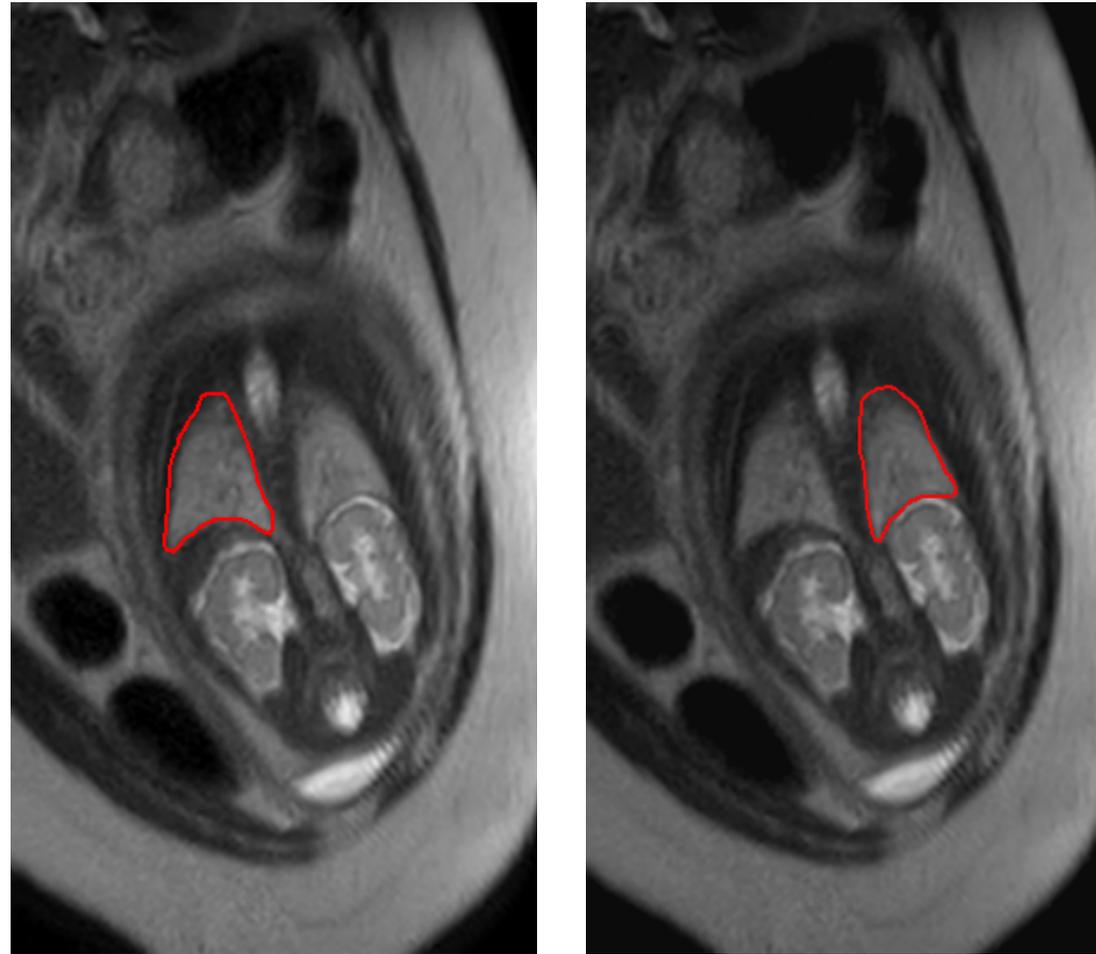
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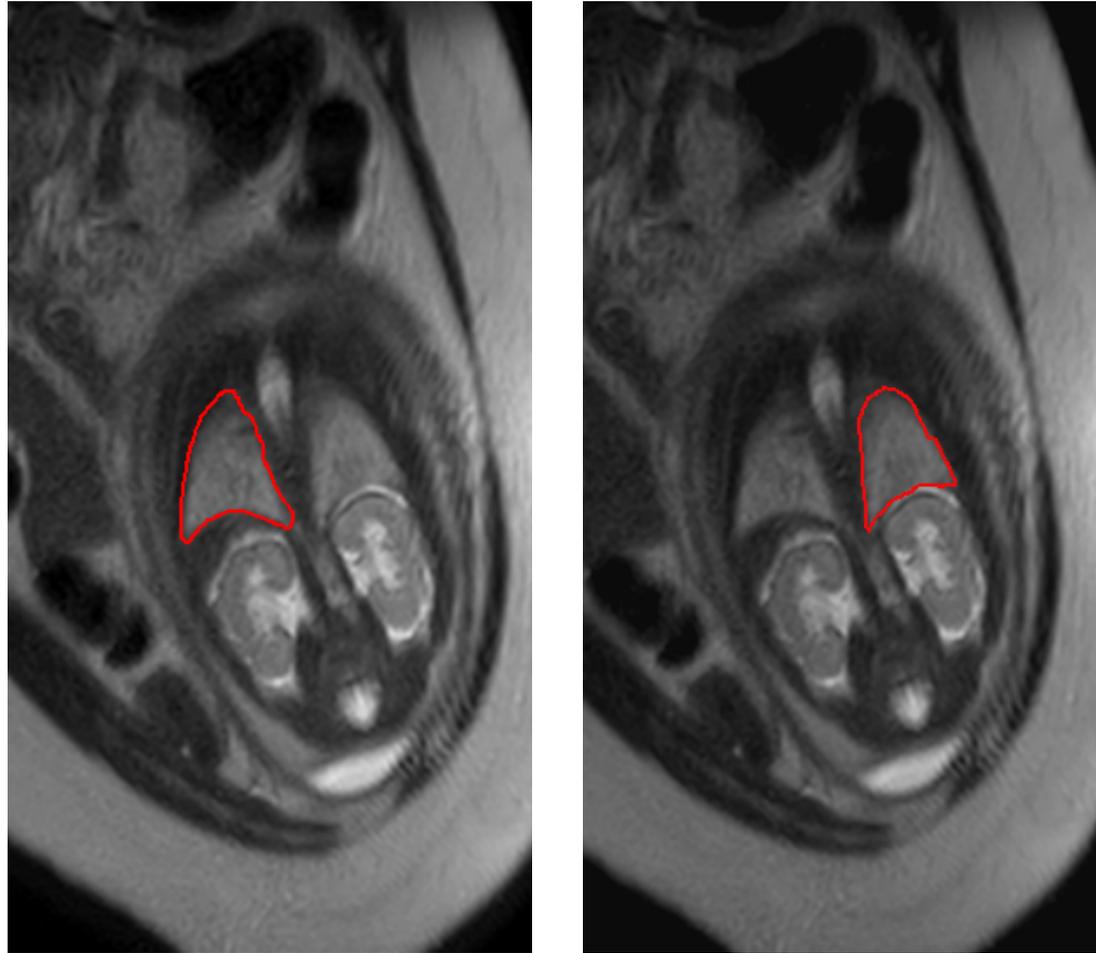
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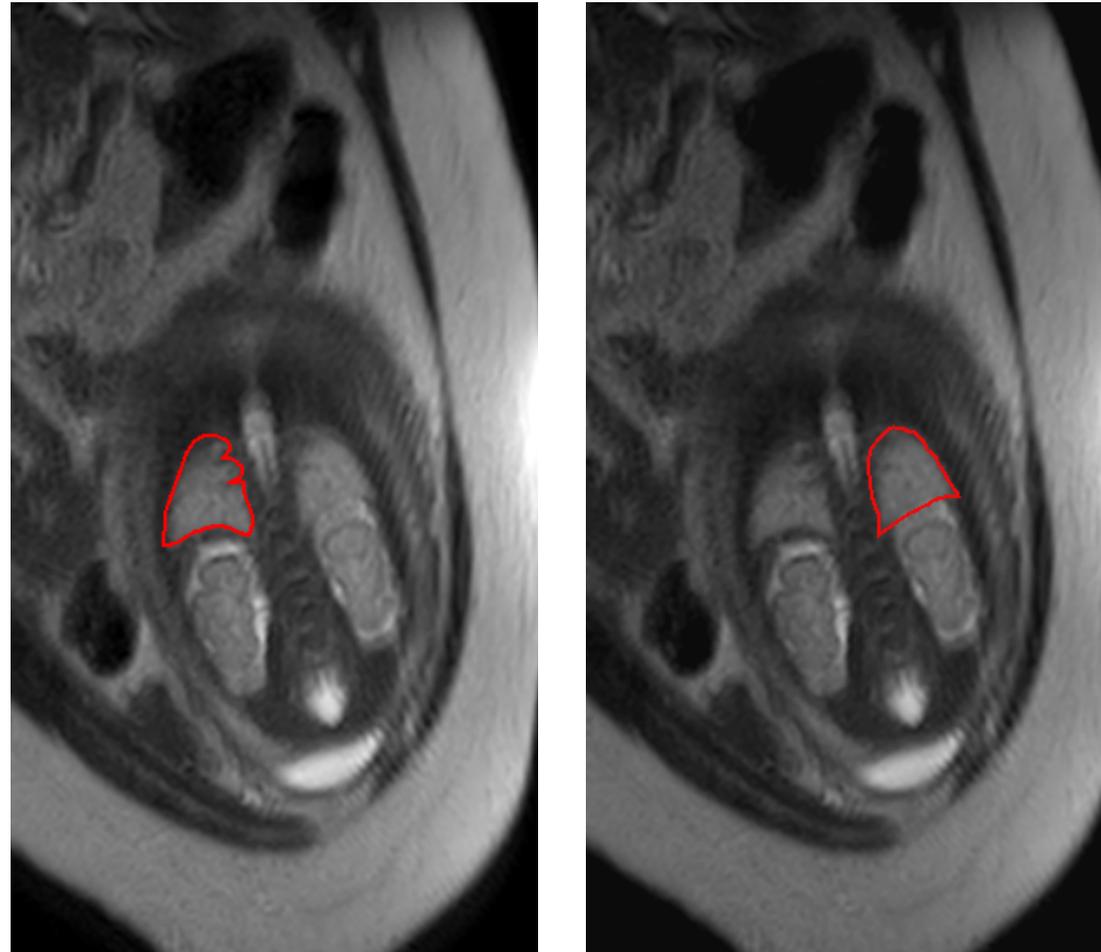
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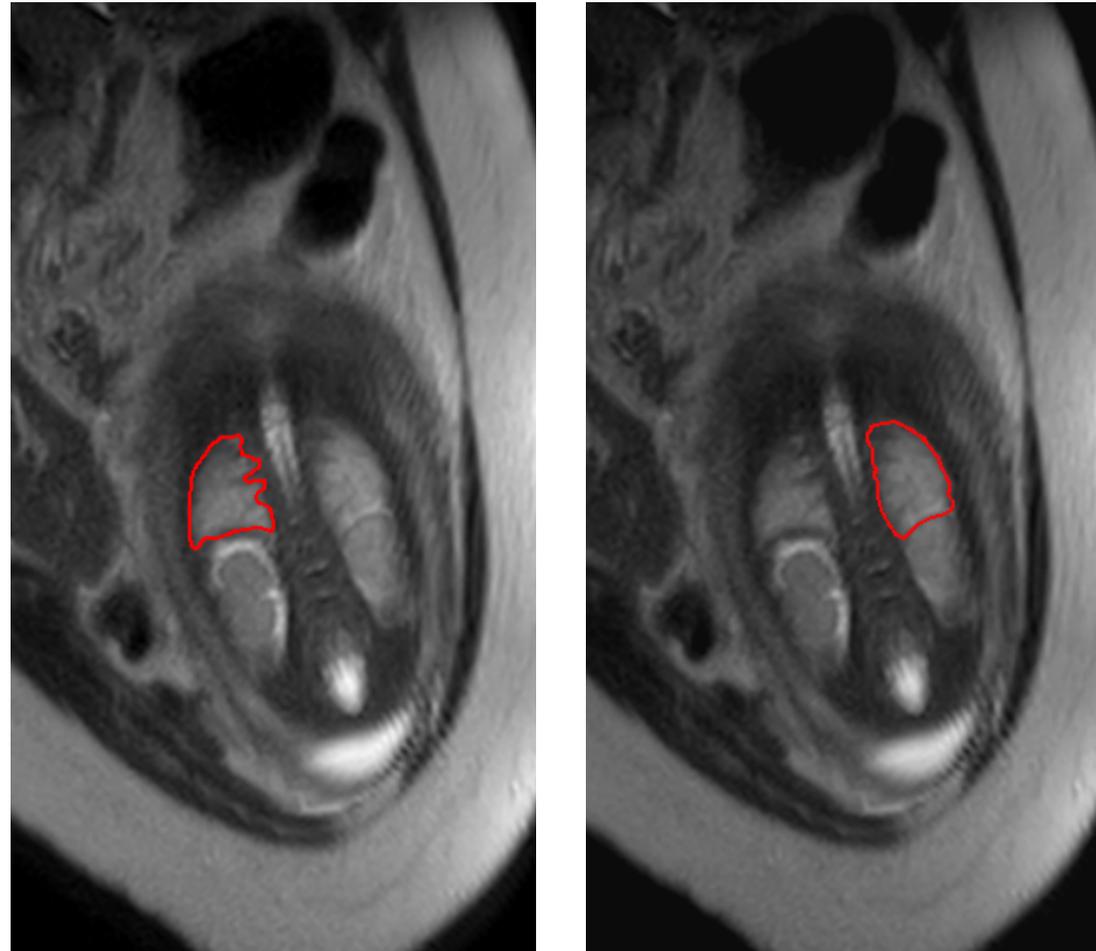
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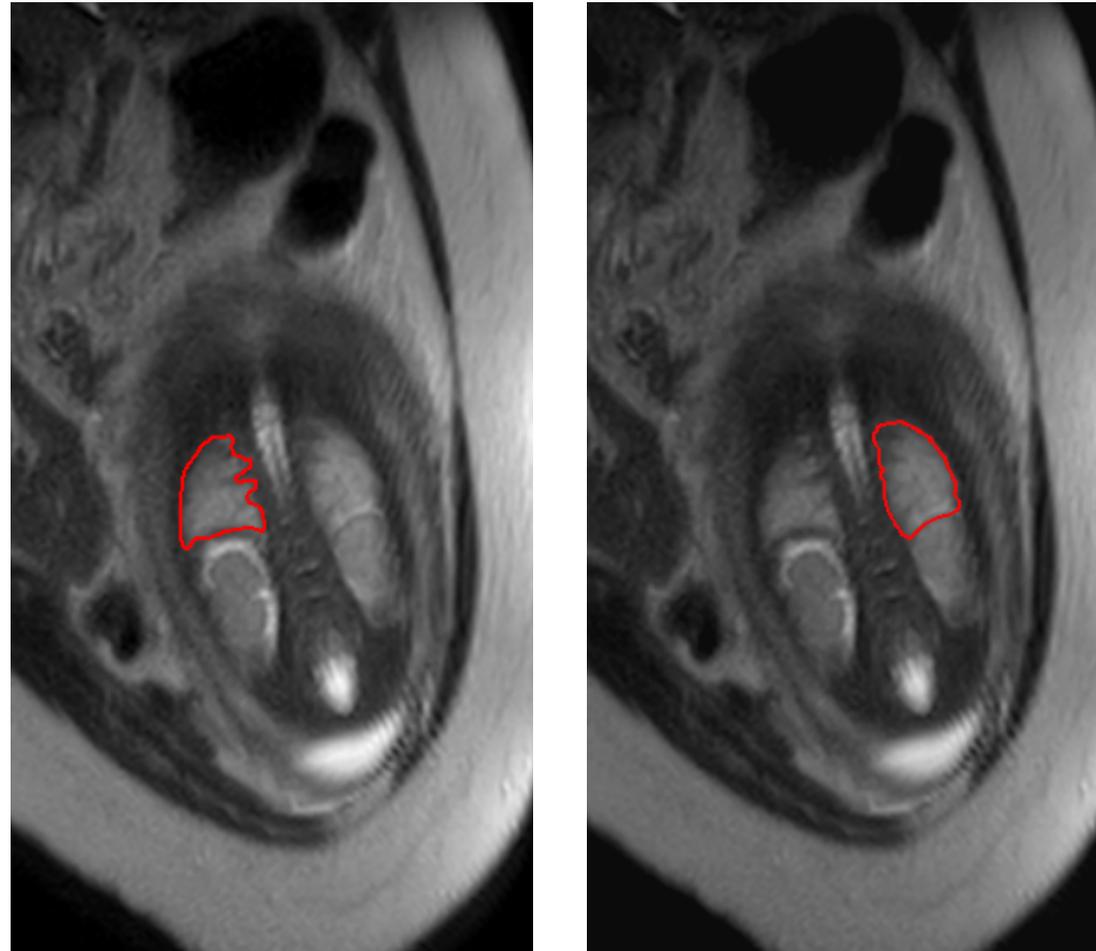
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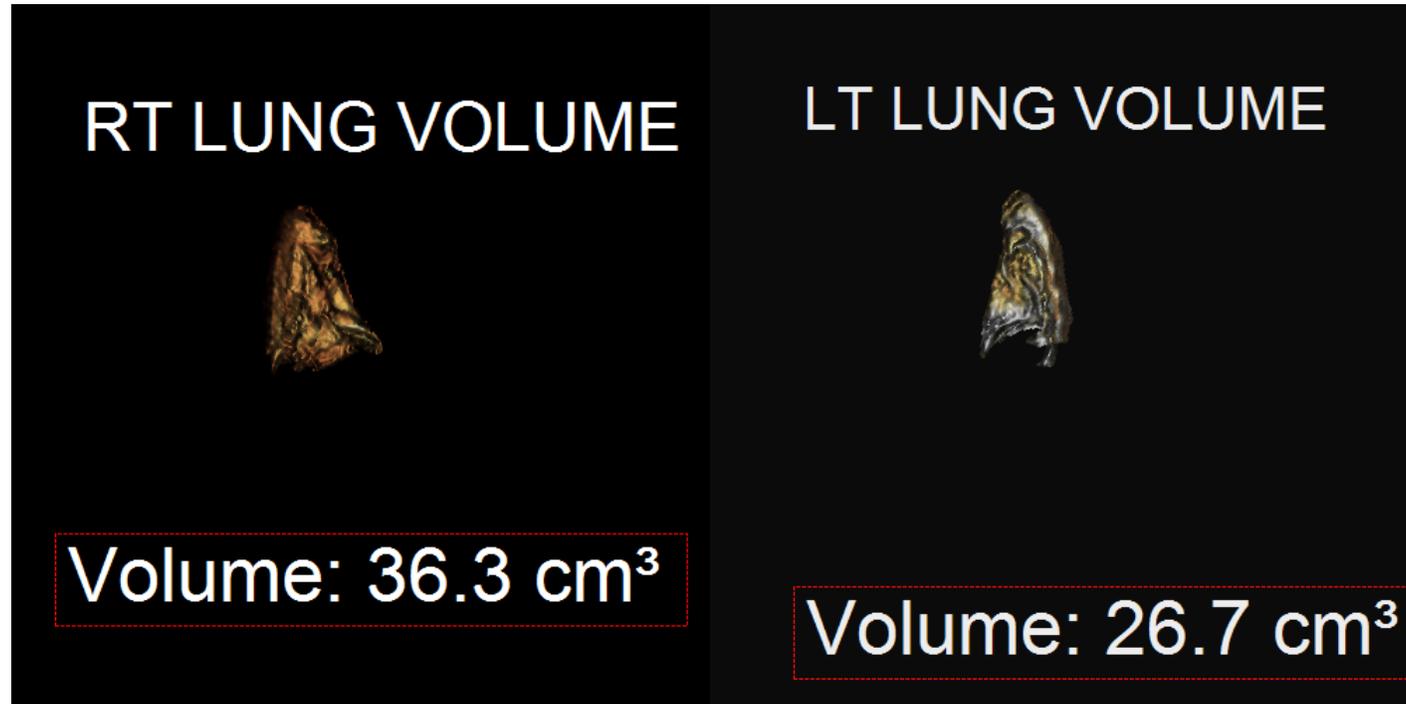
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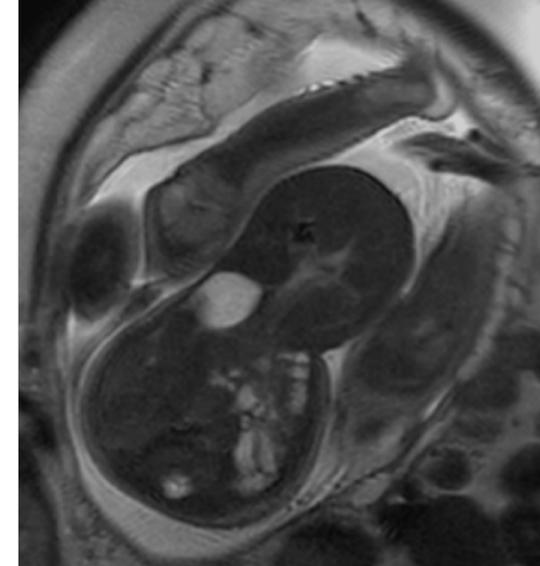
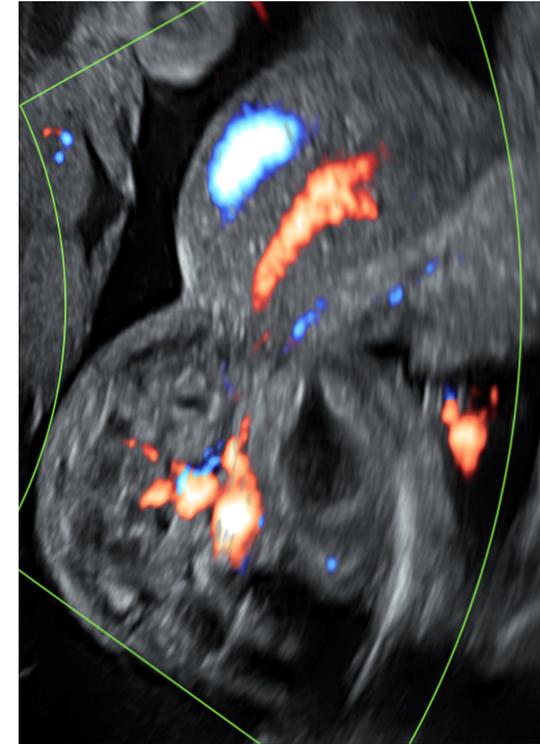
TLV = 63 mL | Mean for GA = 80.7 mL (*Rypens et al.*)

$$O/E = 63/80.7 = 78\%$$



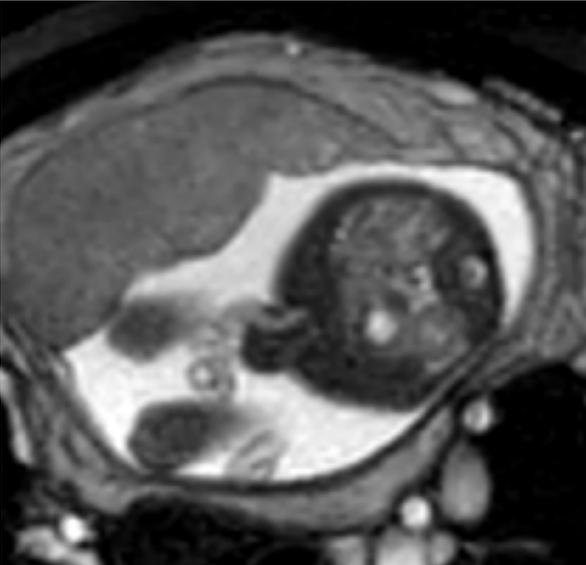
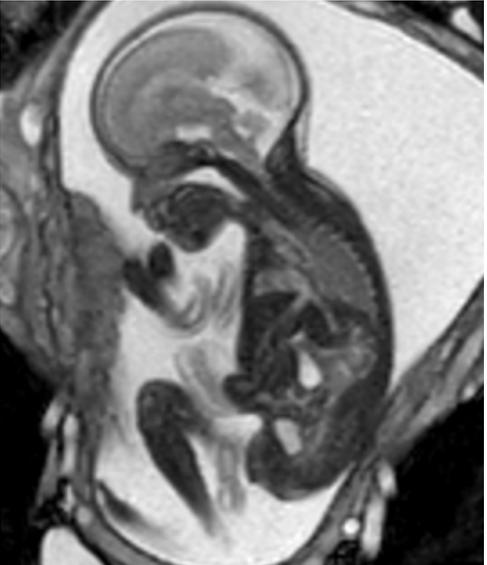
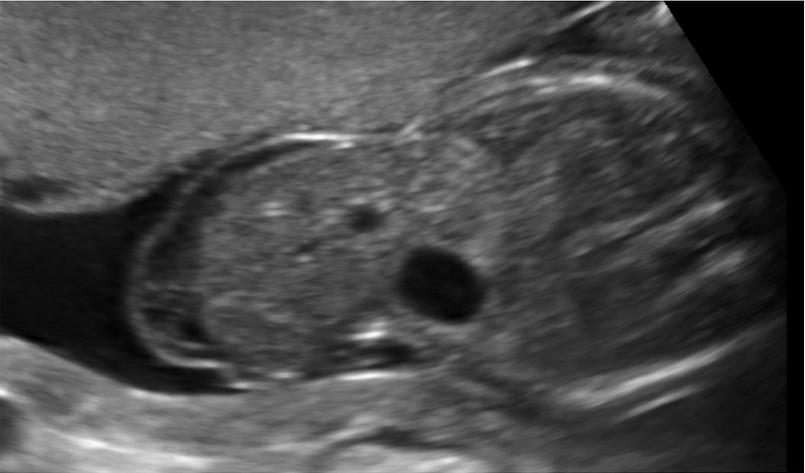
## Imaging Features

- Midline abdominal wall defect located at base of umbilical cord
- Cord inserts onto the defect
- Size variable: small hernia of the cord to giant omphalocele
- Eviscerated organs vary: liver, bowel, stomach
  - Prenatal dx omphalocele contains liver 80%
- Overlying membrane | difficult to visualize or may rupture
  - Contained eviscerated organs as opposed to freely floating
  - Ascites
- Polyhydramnios 10%



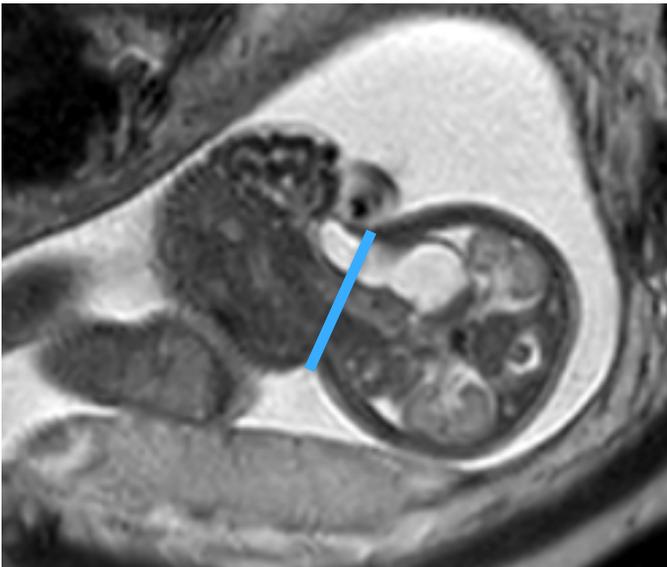
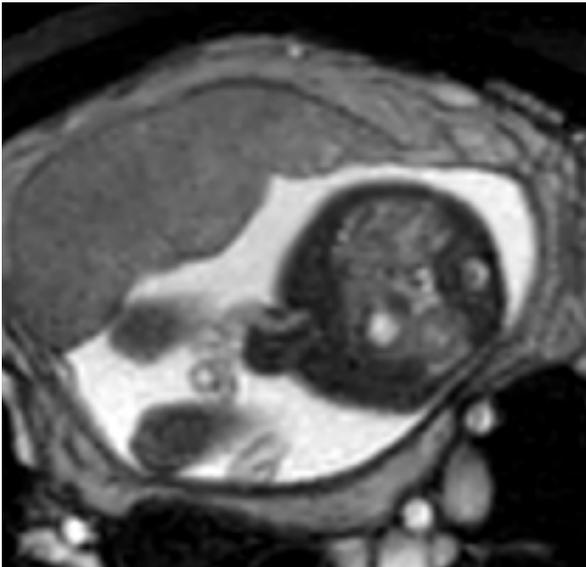
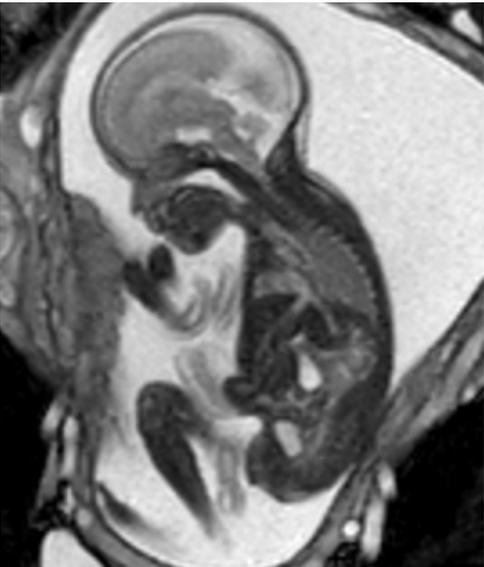
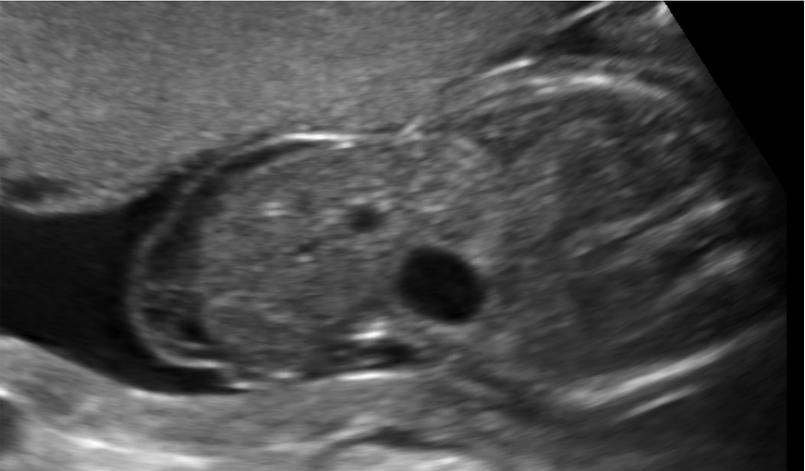
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Size variability



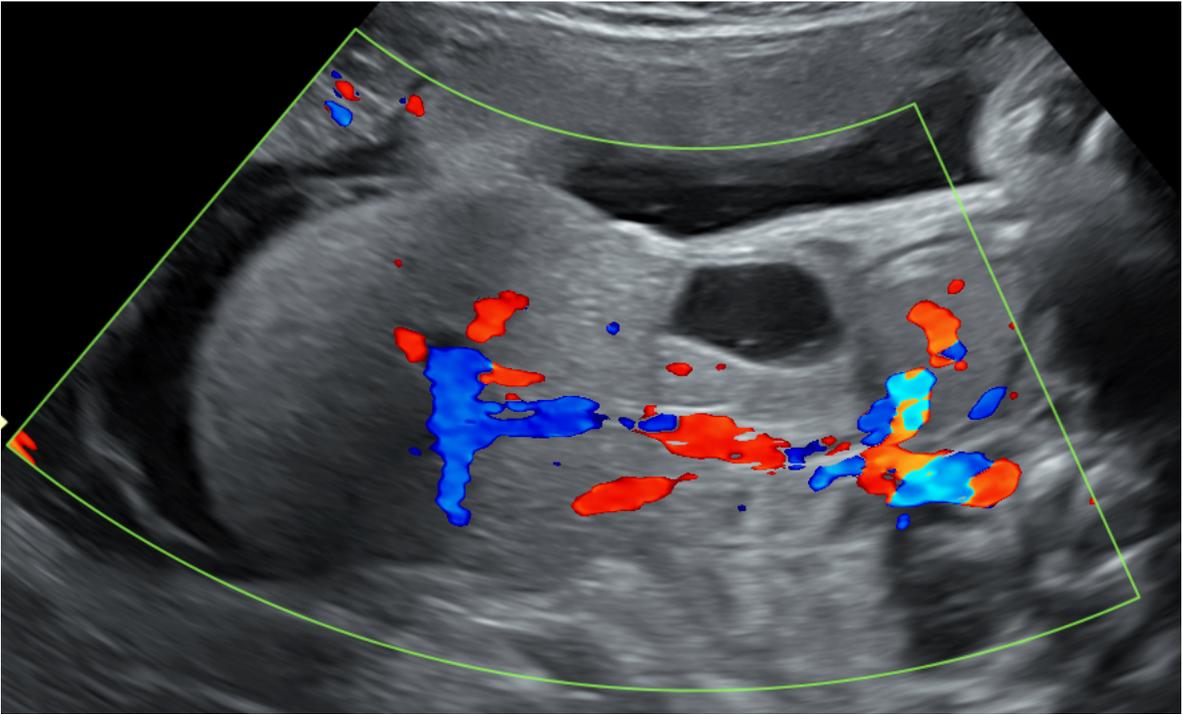
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Size variability



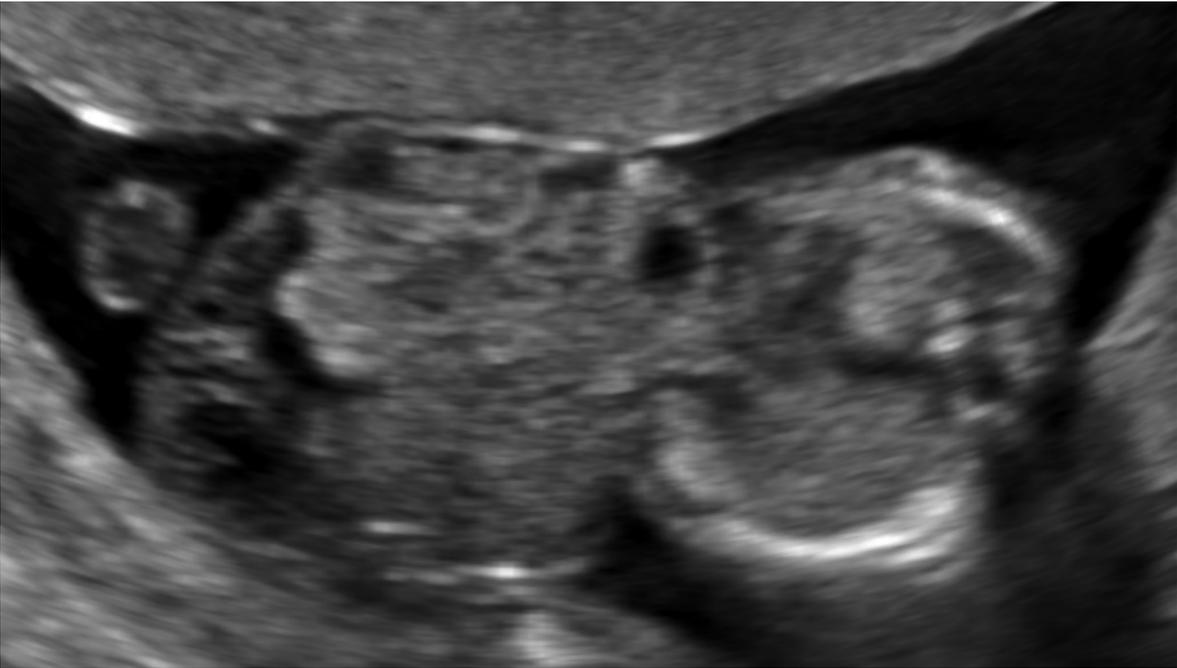
# Imaging Features

## Ultrasound



# Imaging Features

## *Ultrasound*



23 weeks gestation



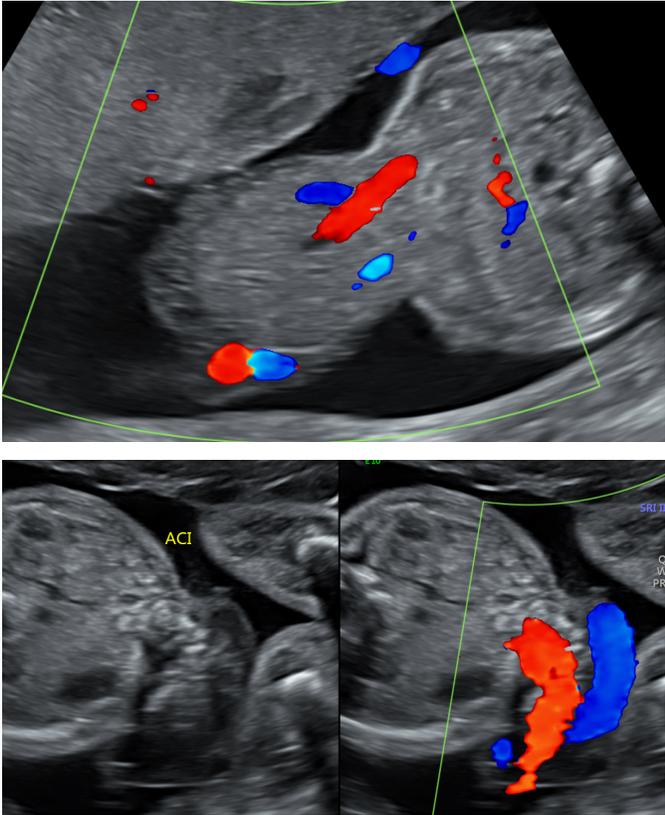
34 weeks gestation



# Imaging Features

## Ultrasound

### Omphalocele



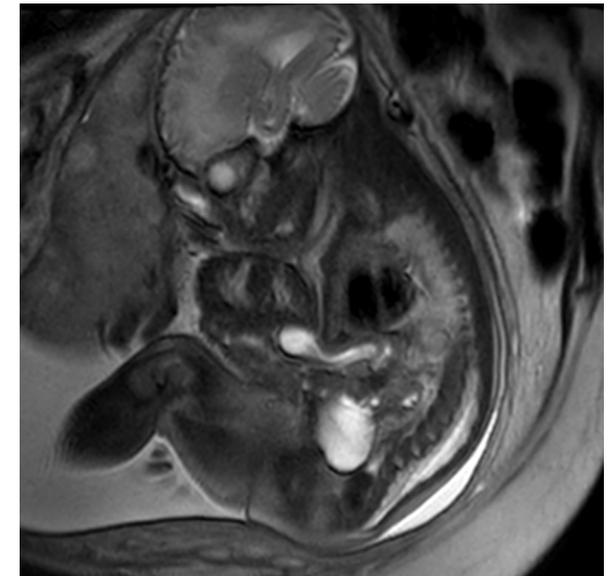
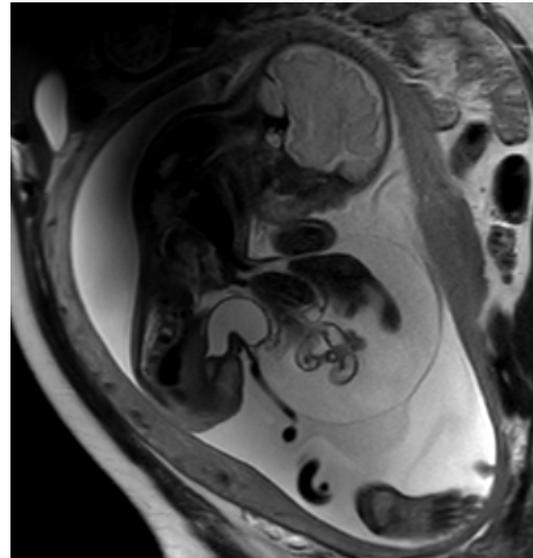
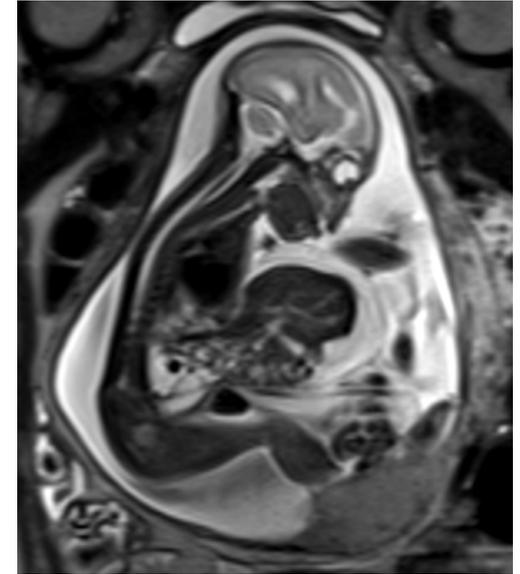
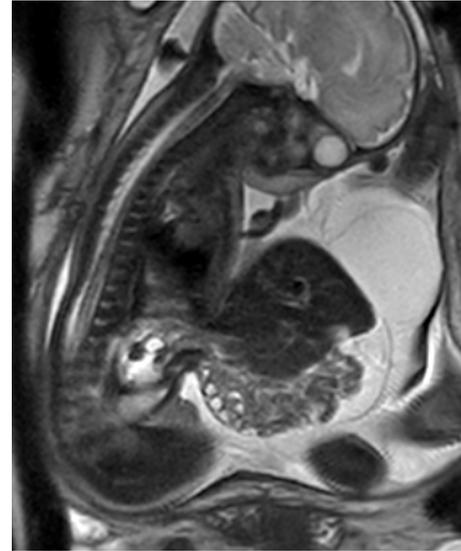
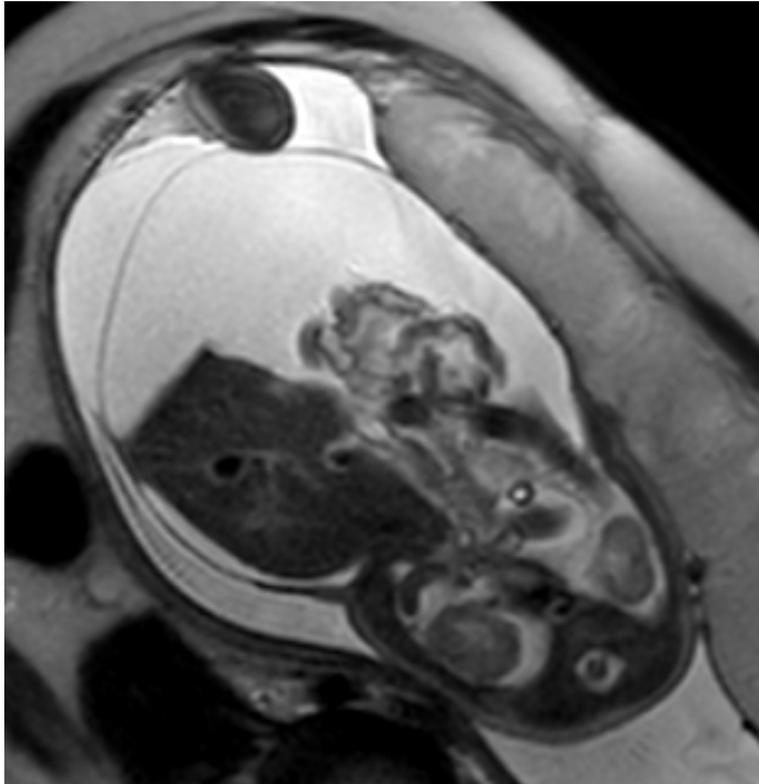
### Gastroschisis



# Imaging Features

## *MRI*

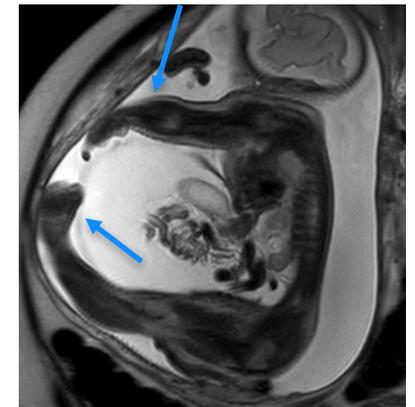
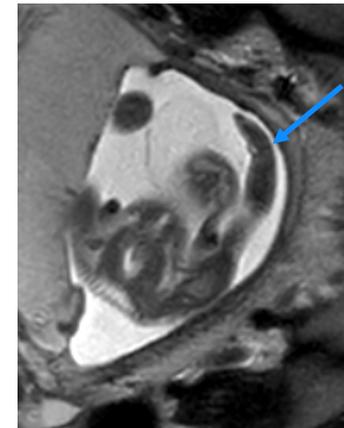
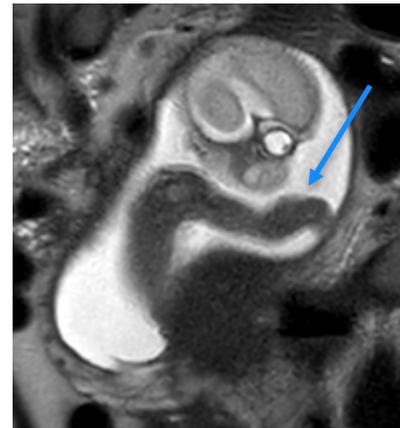
- Liver easily distinguished from bowel
- Quantify defect size
- Useful for associated abnormalities
- Pulmonary hypoplasia



# Imaging Features

## MRI

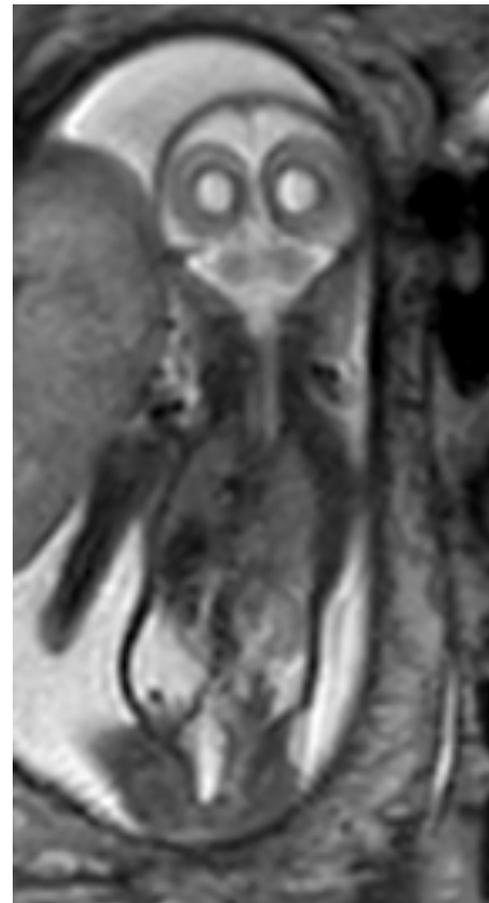
- Liver easily distinguished from bowel
- Quantify defect size
- Useful for associated abnormalities
- Pulmonary hypoplasia



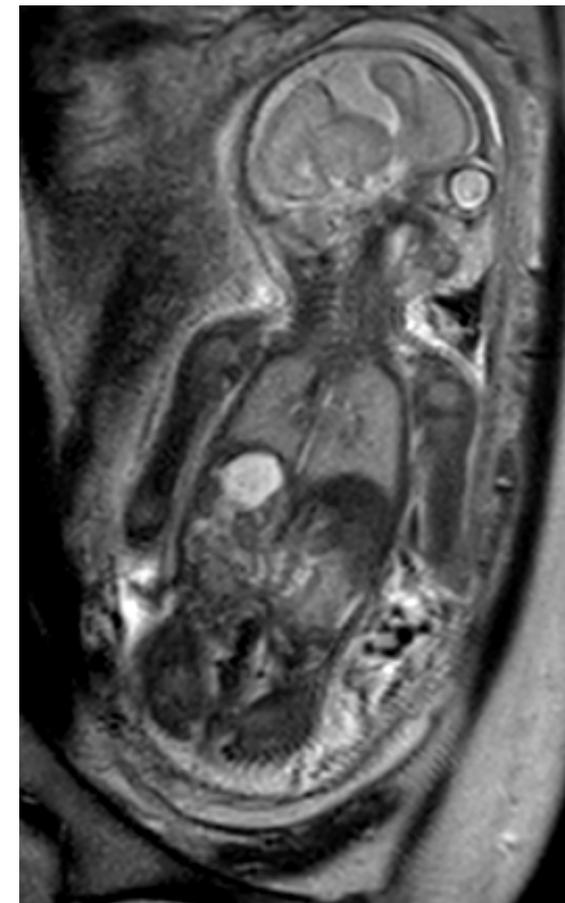
## Imaging Features

### *MRI*

- Liver easily distinguished from bowel
- Quantify defect size
- Useful for associated abnormalities
- Pulmonary hypoplasia
  - Subjective: thoracic shape and lung signal



Hypoplasia



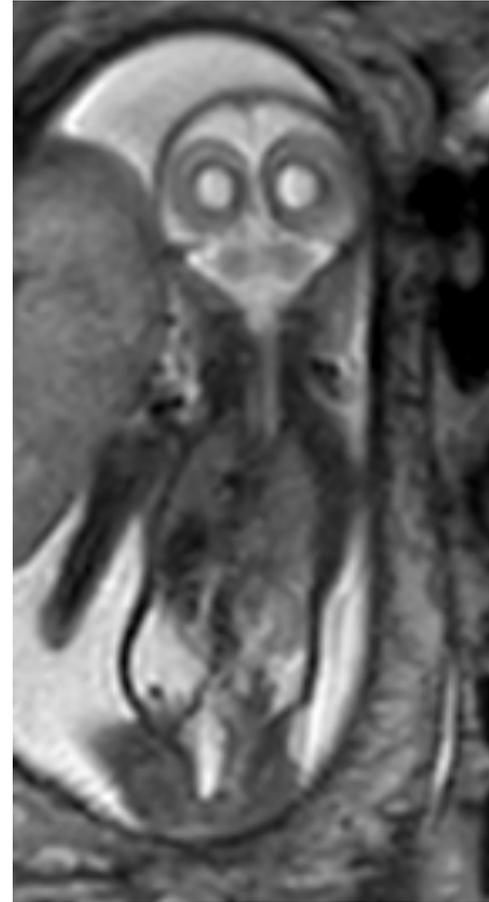
Normal



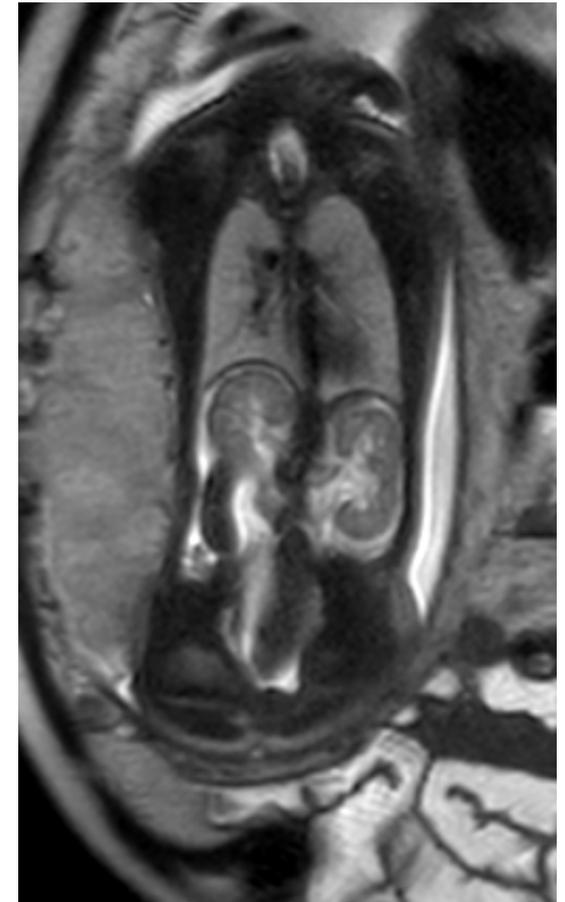
# Imaging Features

## *MRI*

- Liver easily distinguished from bowel
- Quantify defect size
- Useful for associated abnormalities
- Pulmonary hypoplasia
  - Subjective: thoracic shape and lung signal



2<sup>nd</sup> trimester



3<sup>rd</sup> trimester



# Imaging Features

## MRI

- Liver easily distinguished from bowel
- Quantify defect size
- Useful for associated abnormalities
- Pulmonary hypoplasia
  - Subjective: thoracic shape and lung signal
  - Objective quantification of TLV
    - 3<sup>rd</sup> trimester more predictive than 2<sup>nd</sup>



2<sup>nd</sup> trimester



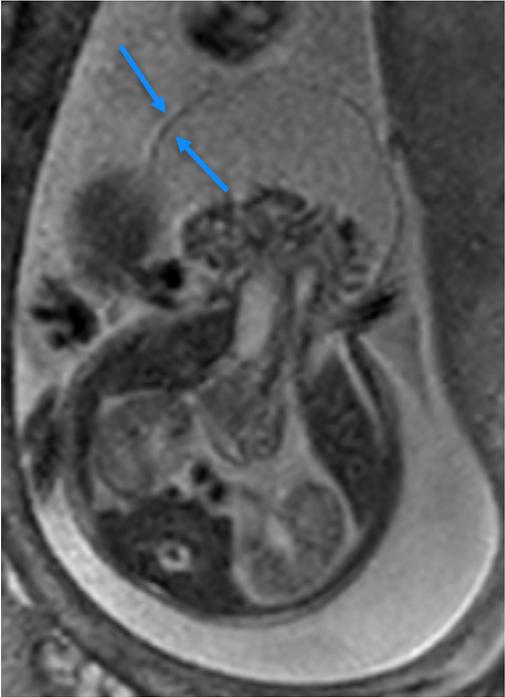
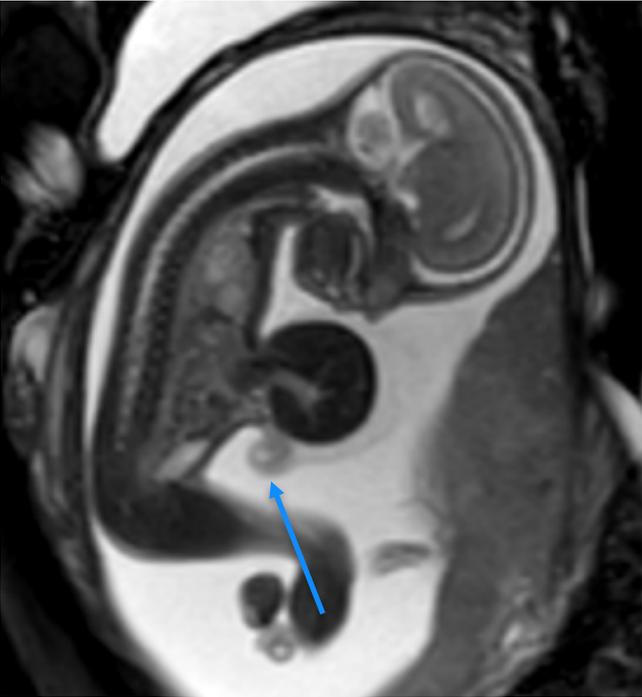
3<sup>rd</sup> trimester



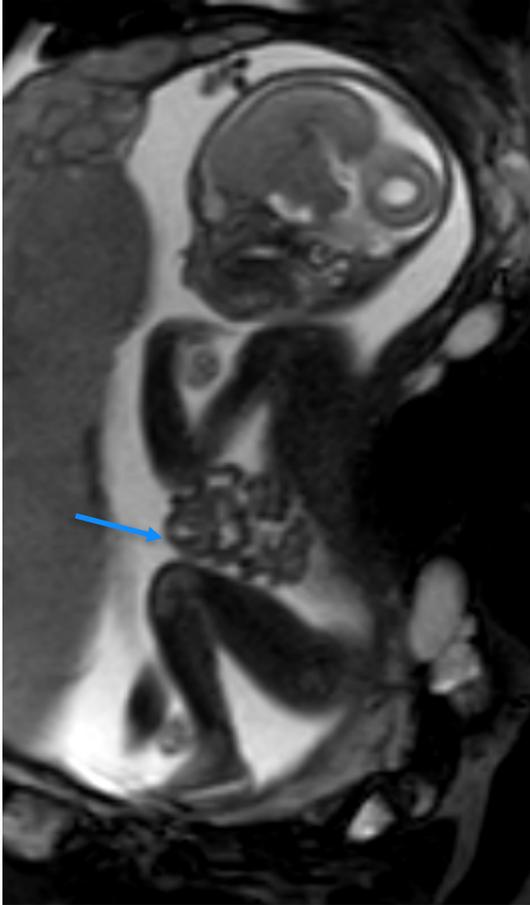
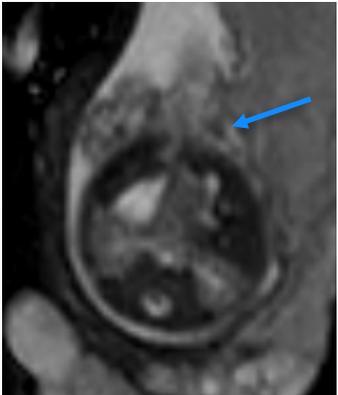
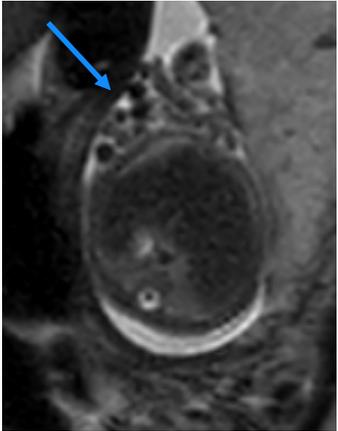
# Imaging Features

*MRI*

## Omphalocele



## Gastroschisis



# Prenatal Diagnosis of Giant Omphaloceles



- 1 Identify ultrasound findings that characterize omphalocele vs other ventral wall defects
- 2 Identify MRI findings that characterize omphalocele vs other ventral wall defects
- 3 Recognize the importance and approach to evaluating pulmonary hypoplasia
- 4 Determine the presence of other associated anomalies that may affect management

