THE FETAL THORAX
The Top 5 Do Not Miss Diagnoses
OBJECTIVES

- To discuss the 5 *“Do Not Miss”* fetal thoracic abnormalities that can be definitively diagnosed with very detailed, high resolution ultrasound
- To emphasize *“Tips”* to definitive US diagnosis of these conditions which I chose because they are the most common referral diagnoses at CHOP that are questioned or even misdiagnosed
- To illustrate how it is possible to confidently change or add to the suspected diagnoses
Examiner the Fetal Chest

- **“Best View”** – axial/transverse with the fetus supine
- Lung Parenchyma – very homogeneous in texture & intermediate in echogenicity
- The lungs ↑ with age, symmetric volumes, R slightly > L
- Cardiothoracic ratio – constant 0.25-0.3 in 2\textsuperscript{nd}-3\textsuperscript{rd} trimesters
- Thoracic circumference & lung volumes – nomograms for gestational age

THE FETAL THORAX
The Top 5 Do Not Miss Diagnoses
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- Congenital High Airway Obstruction Sequence
- Bronchopulmonary Sequestration
- Congenital Cystic Adenomatoid Malformation
- Hydrothorax/Chylothorax
- Congenital Diaphragmatic Hernia
Congenital High Airway Obstruction Sequence

Narrow Larynx with Dilated, Fluid Filled Trachea
**Congenital High Airway Obstruction Sequence**

- Laryngeal > tracheal obstruction
  
  Causes = atresia, agenesis, stenosis, webs, cysts

- "Symmetric" involvement with enlarged, echogenic lungs → flattened or inverted diaphragm

- "Tip" = Small, midline heart, compressed by lungs

- Ascites is very common and dominates if NIH develops

- Polyhydramnios > oligohydramnios
Congenital High Airway Obstruction Sequence

Distended Trachea from Larynx to Bifurcation
Congenital High Airway Obstruction Sequence

- Sporadic with unknown incidence; associated with syndromes, aneuploidy, familial, autosomal dominant with variable expression.

- Anomalies in $\geq 50\%$; VACTERL association (vertebral, anal atresia, cardiac, tracheoesophageal fistula, renal and limb anomalies).
Congenital High Airway Obstruction Sequence

Tracheal Atresia with NIH & Abnormal Left Hand

Limited

Motion
Congenital High Airway Obstruction Sequence

NIH
Oligohydramnios

NND
Congenital High Airway Obstruction Sequence

NIH

ascites, scalp/body edema
Congenital High Airway Obstruction Sequence

CHAOS Severe NIH
Resolved on F/U Scan
**Bronchopulmonary Sequestration**

- Nonfunctioning pulmonary tissue that receives all or most of its blood supply from anomalous systemic vessels with no tracheobronchial communication.

**Classification – 2 varieties**

- **Intralobar:** shares the same pleural investment with normal lung.
- **Extralobar:** separate pleura from the normal lung; may be intrathoracic or subdiaphragmatic.

- 23% of all fetal lung masses; extralobar BPS is much more common in the fetus and neonate.
Extralobar Bronchopulmonary Sequestration
Intralobar Bronchopulmonary Sequestration

*Ref at 21 wks for CCAM*

Aorta Flow

Pulm Vein
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**Bronchopulmonary Sequestration**

- "Tip" = solid, echogenic mass with feeding vessel usually off the descending thoracic or abdominal aortic. Venous drainage usually pulmonary for ILS and vena cava, azygous or hemiazygous systems for ELS. Associated findings = mediastinal shift, pleural effusion, polyhydramnios and hydrops
- 90% occur on the left in the posterior basal segment; ELS is associated with foregut anomalies and CDH in 58%
- 85-90% supradiaphragmatic 10-15% subdiaphragmatic
- Natural history is variable depending on whether BPS is isolated. No known predisposition or recurrence risk; 50-75% regress; Rx = surgical resection
Extralobar Bronchopulmonary Sequestration

Ref as echogenic mass at 21 wks
Extralobar Bronchopulmonary Sequestration

Supradiaphragmatic BPS
Extralobar Bronchopulmonary Sequestration

*Retroperitoneal BPS with Cysts and Multiple SFV’s*
Extralobar Bronchopulmonary Sequestration

Supradiaphragmatic BPS with Solitary Cyst

Ref as CCAM at 23 wks
Extralobar Bronchopulmonary Sequestration

Ref as CCAM at 20 weeks

Art Flow

Pulm Flow
RUL BPS and RLL CCAM

Ref as CCAM at 21 weeks

Pulm Flow

Art Flow
**Congenital Cystic Adenomatoid Malformation**

- Benign hamartoma caused by cessation of bronchiolar maturation with overgrowth of mesenchymal elements

**Classification – 2 varieties**

- Macrocystic contains numerous cysts >5mm in diameter
- Microcystic contains smaller cysts difficult to detect with US, producing a homogenously echogenic mass

- “Tip” = lung mass with only pulmonary vascular supply

- 95% unilateral and usually one lobe; rarely multifocal

- Associated structural anomalies are uncommon; may regress

- Prognosis is excellent unless NIH and polyhydramnios develop
Congenital Cystic Adenomatoid Malformation

Small Microcystic RLL CAM

Volume 2.8 mL

CVR 0.14
Is This a Large Microcystic CCAM?

Ref as CCAM at 23 weeks
Congenital Lobar Emphysema

Transient Dilated, Fluid Filled Trachea
Congenital Cystic Adenomatoid Malformation

Large Macrocystic LLL
CVR = CCAM Volume Ratio

Volume of a prolate ellipse = $W \times D \times L \times 0.523$

Head Circumference

Congenital Cystic Adenomatoid Malformation

CVR Estimation of LUL Macrocystic CCAM

CCAM

Mild shift

NL Lung

Largest Cyst
CCAM Volume Ratio (CVR)

- **Retrospective study**  
  November 1998-August 1999
  32 patients with CCAM: (p<.001)  
  Hydrops: CVR mean 8.3±1.1  
  No Hydrops: 0.74±0.48

- **Prospective Study**  
  September 1999-March 2001
  58 patients with CCAM: (p<.005)
  Low Risk with CVR≤1.6
    - Patients: 42  
    - Hydrops: 7 (6/7, dominant cyst and if these excluded only 1/36 had hydrops (2.8%))
  High Risk with CVR >1.6
    - Patients: 16  
    - Hydrops: 12 (75%)

**IMAGING OF THE FETAL THORAX**

**Therapeutic Options for Lung Masses Complicated by NIH**

- If lungs mature, EXIT (ex utero intrapartum therapy) for large masses
- If lungs immature;
  - Macrocystic
  - Microcystic
  - Cyst Aspiration
  - Open Fetal Surgery
  - Thoracoamniotic Shunt Placement
- Survivors of fetal intervention often require ventilatory support, sometimes only for brief periods.
- ECMO not usually necessary
- Elective postnatal resection advised due to↑ risk of infection and malignant transformation

**THE FETAL THORAX**

The Top 5 Do Not Miss Diagnoses
Congenital Cystic Adenomatoid Malformation

Macrocystic RLL CCAM, Vol 50 CVR 2.8 with NIH
Congenital Cystic Adenomatoid Malformation

Thoracoamniotic Shunt
“Hybrid” Lung Lesions

**Ultrasound Features:**
1. Systemic and pulmonary vessels in mass of mixed echotexture with solid and cystic components
2. Systemic and pulmonary arterial supply in echogenic solid mass

**Pathologic Features: BPS and CCAM Histology**
1. Extralobar
2. Intralobar

Hybrid CCAM has a more favorable prognosis!!

Macrocystic Intralobar Hybrid

RLL Lesion Vol 13 & CVR 0.76

Aortic Feeder

Pulm Flow
Macrocystic Intralobar Hybrid

Ref at 23 weeks as CCAM

Macrocysts on US & MR
Macrocystic Intralobalbar Hybrid

RLL Vol 25 \(\rightarrow\) 20 CVR 1.19 \(\rightarrow\) 0.87

Arterial Flow

Pulmonary Flow
The Top 5 Do Not Miss Diagnoses

**Hydrothorax/Chylothorax**

**PRIMARY**
- Chylothorax, unilateral, isolated and usually right sided

**SECONDARY**
- Bilateral effusions, typical of hydrops fetalis
- Fluid may occur with masses – CCAM, CDH, BPS
- Idiopathic hydrothorax – abnormal chromosomes, esp T21, 45 XO; congenital heart disease (5%)
- Pulmonary lymphangiectasia

THE FETAL THORAX
Small Idiopathic Right Pleural Effusion
Asymmetric Bilateral Pleural Effusions

$R \gg L$
### The Fetal Thorax

#### The Top 5 Do Not Miss Diagnoses

**Fetal Pleural Effusions: Prognostic Indicators**

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>%Survival</th>
<th>Characteristic</th>
<th>%Survival</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Age at diagnosis</strong></td>
<td></td>
<td><strong>Age at delivery</strong></td>
<td></td>
</tr>
<tr>
<td>&lt; 33 wk</td>
<td>43</td>
<td>&lt; 35 wk</td>
<td>30</td>
</tr>
<tr>
<td>≥ 33 wk</td>
<td>80</td>
<td>≥ 35 wk</td>
<td>79</td>
</tr>
<tr>
<td><strong>Spontaneous resolution</strong></td>
<td></td>
<td><strong>Hydrops</strong></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>100</td>
<td>Yes</td>
<td>52</td>
</tr>
<tr>
<td>No</td>
<td>52</td>
<td>No</td>
<td>100</td>
</tr>
<tr>
<td><strong>Bilateral</strong></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>52</td>
<td></td>
<td></td>
</tr>
<tr>
<td>No</td>
<td>100*</td>
<td></td>
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</table>

*without mediastinal shift

Pleuroamniotic Shunt Placement
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The Top 5 Do Not Miss Diagnoses

Features suggestive of chylothorax

- Pleural effusion is an isolated or initially isolated finding
- Size of effusion is disproportionately large compared to other effusions, if present
- Aspirated fluid is clear, straw-colored with lymphocytes
- Polyhydramnios is common, 60-70%; possibly due to swallowing or altered amniotic fluid production by compressed lungs
- Mortality rate for chylothorax ~ 5% have abnormal chromosomes
Unilateral Right Chylothorax
Is This a Large Chylothorax?

Ref as a left pleural effusion
Is This a Unilateral Left Effusion?

Ref as a right pleural effusion with NIH
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**Congenital Diaphragmatic Hernia**

- "**Tip**" – lung mass with mediastinal shift and absent stomach in the abdomen
- Stomach adjacent to heart and peristalsing bowel loops is pathognomonic
- Pulmonary hypoplasia develops from compression of the developing lungs by the herniated viscera
- Spectrum of embryologic defects but most common is foramen of Bochdalek hernia; 85-90% are left sided, < 10% right and < 5% bilateral defects
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_*Left CDH Containing Stomach and Bowel*_
**THE FETAL THORAX**

The Top 5 Do Not Miss Diagnoses

**Congenital Diaphragmatic Hernia**

- Usually sporadic but occurs in various syndromes such as Fryn’s; associated with maternal ingestion of drugs such as *(quinine, bendectine, antiepileptic agents)*

- Chromosomal anomalies occur in 10-20% of prenatal cases, esp Trisomies 21, 18 and 13.

- Structural anomalies occur in 25-57% of all cases

- Poor prognostic indicators –
  - Large defect
  - Dilated intrathoracic stomach
  - Early diagnosis before 25 weeks
  - NIH and polyhydramnios
  - Intrathoracic liver
  - Other significant anomalies
Left CDH with Stomach and Bowel

**Peristalsing Bowel Loops with SMA Flow**
Bowel Only Left CDH

Ref as mass adjacent to stomach at 26 wks
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The Top 5 Do Not Miss Diagnoses

Left CDH Containing Bowel, Left Kidney, Adrenal & Spleen

Defect

Spleen
# Frequency of Associated Anomalies Among 108 Cases of Congenital Diaphragmatic Hernia

<table>
<thead>
<tr>
<th>%</th>
<th>Organ System</th>
<th>Selected examples</th>
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<tbody>
<tr>
<td>23</td>
<td>Cardiovascular</td>
<td>coarctation, tetralogy of Fallot, transposition, pulmonic stenosis</td>
</tr>
<tr>
<td>15</td>
<td>Urinary</td>
<td>renal agenesis, cystic renal dysplasia, ureteropelvic junction</td>
</tr>
<tr>
<td></td>
<td></td>
<td>obstruction with hydronephrosis</td>
</tr>
<tr>
<td>10</td>
<td>Central Nervous</td>
<td>arrhinecephaly, holyprosencephaly, hydrocephalus, myelodysplasia</td>
</tr>
<tr>
<td>9</td>
<td>Musculoskeletal</td>
<td>talipes equinovarus, hemivertebrae, absent ribs</td>
</tr>
<tr>
<td>8</td>
<td>Genital</td>
<td>ambiguous genitalia, bicornuate uterus, microphallus, cryporchidism</td>
</tr>
<tr>
<td></td>
<td></td>
<td>ureterovaginal atresia</td>
</tr>
<tr>
<td>7</td>
<td>Gastrointestinal</td>
<td>annular pancreas, imperforate anus, absent gallbladder</td>
</tr>
</tbody>
</table>

THE FETAL THORAX
The Top 5 Do Not Miss Diagnoses

Left CDH – Liver, Stomach, Bowel & Anomalies
THE FETAL THORAX
The Top 5 Do Not Miss Diagnoses

*Left CDH – Liver, Stomach, Bowel & Anomalies*
THE FETAL THORAX
The Top 5 Do Not Miss Diagnoses

*Right CDH with Thick 2VC*
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The Top 5 Do Not Miss Diagnoses

**Bilateral CDH**
### Fetal Thorax

**LHR vs. Survival in \*CDH**

<table>
<thead>
<tr>
<th>LHR</th>
<th>% Survival</th>
</tr>
</thead>
<tbody>
<tr>
<td>&lt; 0.6</td>
<td>0% (0/5)</td>
</tr>
<tr>
<td>&gt; 0.6 and &lt; 1.35</td>
<td>57% (16/28)</td>
</tr>
<tr>
<td>&gt; 1.35</td>
<td>100% (5/5)</td>
</tr>
</tbody>
</table>

*CDH diagnosed before 25 weeks gestation

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### Fetal Thorax

The relationship between liver herniation and right lung size in 38 fetuses < 25 weeks gestation

<table>
<thead>
<tr>
<th>Liver herniation</th>
<th>No.</th>
<th>LHR*</th>
</tr>
</thead>
<tbody>
<tr>
<td>None</td>
<td>6</td>
<td>1.33  + 0.30</td>
</tr>
<tr>
<td>Moderate</td>
<td>7</td>
<td>1.25  + 0.34</td>
</tr>
<tr>
<td>Severe</td>
<td>25</td>
<td>0.87  + 0.34</td>
</tr>
</tbody>
</table>

*LHR-right lung to head circumference ratio

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*Large Left CDH with LHR 0.79 & BPS*
Therapeutic Options for CDH

Delivery near term when lungs mature ± ECMO support
Postnatal surgical resection once infant stabilized
EXIT ➔ ECMO procedure (CHOP, Boston)

In utero

1) Fetal CDH repair (failed)
2) Fetal tracheal clip procedure (failed)
3) Fetal balloon occlusion of trachea (Multi-center Trial)
THE FETAL THORAX
The Top 5 Do Not Miss Diagnoses

Take Home Message

- High resolution US with color Doppler can make more specific diagnoses earlier than ever before
- The most frequently encountered Tip in most thoracic anomalies is mass effect with/without abnormal lung echotexture
- The “Top 5 Do Not Miss” fetal thorax diagnoses are CHAOS, BPS, CCAM, Hydro/Chylothorax and CDH
- Imaging results virtually always impact pregnancy counseling and management decisions
THE FETAL THORAX
The Top 5 Do Not Miss Diagnoses

*LIVE LONG and PROSPER*