Fetal GI Anomalies … Refining Your Diagnosis

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Differential Diagnosis

- Omphalocele
- Gastrochisis
- Duodenal atresia
- Diaphragmatic hernia
- Esophageal atresia/TE fistula
- Small bowel obstruction
- Echogenic bowel
- Abdominal cysts

Omphalocele

Background
Herniation of abdominal contents into base of umbilical cord
Amnio-peritoneal covering
Bowel only versus bowel & liver
Incidence 1:5,000 births
Omphalocele

Development of abdominal wall
- Physiologic gut herniation resolves by 12 wks
- Fusion of ectomesenteric folds by 10 wks
  - Cephalic fold defect: ectopia cordis
  - Caudal fold defect: bladder extrophy
  - Lateral fold defect: omphalocele

Associated anomalies
- Seen in up to 50%
  - Congenital heart disease
  - Genito-urinary
  - Body stalk anomaly
- Malrotation of gut in 100%
- Aneuploidy in 40%
  - Trisomy 18, 13
  - Beckwith-Wiedemann syndrome

Omphalocele & karyotype
- Presence of liver in sac reduces risk
- Abnormal karyotype 1/25 with liver in sac
- Abnormal karyotype 8/15 with liver in abdomen
- Small hernia more likely abnormal karyotype
- Trisomy 18, 13

De Veciana Prenat Diagn 1994;14:487-92
Omphalocele & karyotype

- Presence of liver in sac reduces risk
- Abnormal karyotype 2/18 with liver in sac
- Abnormal karyotype 8/8 with liver in abdomen
- Trisomy 18, 13

Nyberg J Ultrasound Med 1989;8:299-308

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Omphalocele

Associated CHD
37 patients, CHD IN 13 (35%)
- VSD 4
- DORV 3
- Pentalogy of Cantrell 7
Karyotypes from 33/37 (89%)
- Abnormal 5 (15%)
- CHD & omphalocele
- Aneuploidy 4/13 (31%)

Fogel Am J Perinatol 1991;8:411-416

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Omphalocele & karyotype

<table>
<thead>
<tr>
<th></th>
<th>Isolated</th>
<th>MCA</th>
<th>Total</th>
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</thead>
<tbody>
<tr>
<td>Tri 21</td>
<td>1</td>
<td></td>
<td>1</td>
</tr>
<tr>
<td>Tri 18</td>
<td>5</td>
<td>16</td>
<td>21</td>
</tr>
<tr>
<td>Tri 13</td>
<td>1</td>
<td>4</td>
<td>5</td>
</tr>
<tr>
<td>45, X</td>
<td>1</td>
<td>1</td>
<td></td>
</tr>
<tr>
<td>Other</td>
<td>6</td>
<td></td>
<td>6</td>
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<tr>
<td>Total</td>
<td>7</td>
<td>27</td>
<td>34</td>
</tr>
</tbody>
</table>

Barisic Ultrasound Obstet Gynecol 2001;18:309-16
Omphalocele outcomes

<table>
<thead>
<tr>
<th></th>
<th>Live Birth</th>
<th>Stillbirth</th>
<th>TOP</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Isolated</td>
<td>38 (63)</td>
<td>12 (20)</td>
<td>10 (17)</td>
<td>60</td>
</tr>
<tr>
<td>Syndromic</td>
<td>4 (29)</td>
<td>4 (29)</td>
<td>6 (42)</td>
<td>14</td>
</tr>
<tr>
<td>Chrom.</td>
<td>4 (12)</td>
<td>7 (21)</td>
<td>23 (67)</td>
<td>34</td>
</tr>
<tr>
<td>MCA</td>
<td>10 (35)</td>
<td>7 (24)</td>
<td>12 (41)</td>
<td>29</td>
</tr>
<tr>
<td>Total</td>
<td>56 (41)</td>
<td>30 (22)</td>
<td>51 (37)</td>
<td>137</td>
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</table>

Barisic Ultrasound Obstet Gynecol 2001;18:309-16

Beckwith-Wiedemann

- May be up to 20% omphalocele
- Macroglossia
- Polyhydramnios
- Other organs

Fetal GI Anomalies

Profile
Normal cord insertion

Omphalocele

Omphalocele
Omphalocele
Pentalogy of Cantrell

Physiologic bowel herniation

Omphalocele
Omphalocle First Trimester

Omphalocele 13 weeks
Trisomy 18

Omphalocele
### Gastroschisis

**Etiology:**
Premature involution of right umbilical vein
Vascular compromise results in full thickness defect

### Gastroschisis

**Background**
- Paraumbilical full thickness defect
- Usually on right
- No covering membrane
- Liver rarely involved
- Incidence 1:10,000 births
  - Young maternal age
  - Regional differences

### Gastroschisis outcomes

<table>
<thead>
<tr>
<th></th>
<th>Livebirth</th>
<th>Stillbirth</th>
<th>TOP</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Isolated</td>
<td>60 (73)</td>
<td>9 (11)</td>
<td>13 (16)</td>
<td>82</td>
</tr>
<tr>
<td>Syndromic</td>
<td>0</td>
<td>1 (14)</td>
<td>6 (86)</td>
<td>7</td>
</tr>
<tr>
<td>Chrom.</td>
<td>0</td>
<td>0</td>
<td>2 (100)</td>
<td>2</td>
</tr>
<tr>
<td>MCA</td>
<td>2 (13)</td>
<td>3 (20)</td>
<td>10 (67)</td>
<td>15</td>
</tr>
<tr>
<td>Total</td>
<td>62 (59)</td>
<td>13 (912)</td>
<td>31 (29)</td>
<td>106</td>
</tr>
</tbody>
</table>

Barisic Ultrasound Obstet Gynecol 2001;18:309-16
Gastroschisis

Gastroschisis

Gastroschisis
Bedside closure with silo

1 hour after birth

24 hours after birth
48 hours after birth

72 hours after birth

5 months later
### Gastroschisis

**Associated CHD**
- 17 patients, CHD in 2 (12%)
  - VSD
  - CAVSD

**Karyotypes from 16/17 (94%)**
- Abnormal 2 (12%)

**CHD & gastroschisis**
- Aneuploidy 1/2

_Fogel Am J Perinatol 1991;8:411-416_

### Gastroschisis - route of delivery

<table>
<thead>
<tr>
<th></th>
<th>Vaginal</th>
<th>Cesarean</th>
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</thead>
<tbody>
<tr>
<td>Number</td>
<td>41</td>
<td>15</td>
</tr>
<tr>
<td>Weight (grams)</td>
<td>2257</td>
<td>2575</td>
</tr>
<tr>
<td>Meconium</td>
<td>29%</td>
<td>30%</td>
</tr>
<tr>
<td>Meconium</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Death</td>
<td>7.3%</td>
<td>6.7%</td>
</tr>
</tbody>
</table>

_Moretti Am J Obstet Gynecol 1990_

### Gastroschisis - route of delivery

<table>
<thead>
<tr>
<th></th>
<th>Vaginal</th>
<th>Cesarean</th>
</tr>
</thead>
<tbody>
<tr>
<td>Number</td>
<td>14</td>
<td>14</td>
</tr>
<tr>
<td>Days to po feeds</td>
<td>14</td>
<td>19</td>
</tr>
<tr>
<td>Days in hospital</td>
<td>27</td>
<td>34</td>
</tr>
<tr>
<td>Complications</td>
<td>4</td>
<td>5</td>
</tr>
<tr>
<td>Death</td>
<td>0</td>
<td>1</td>
</tr>
</tbody>
</table>

_Bethel J Pediatr Surg 1989_
Omphalocele & Gastrochisis

Route of delivery

- Meta-analysis combined diagnoses
- Rate of 1\textsuperscript{st} repair no difference
  - $N = 430$, RR 1.22, 95\% CI 0.99-1.51
- Rate of neonatal sepsis no difference
  - $N = 262$, RR 0.90, 95\% CI 0.50-1.62
- Mortality rate no difference
  - $N = 615$, RR 1.23, 95\% CI 0.79-1.92

Segel Obstet Gynecol 2001;98:867-73

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Polyhydramnios

Multiple etiologies
- Diabetes
- Multiple gestation
- Erythroblastosis
- Neurologic dysfunction
- Fetal anomalies
  - Bowel atresias (esophageal, duodenal)
  - Diaphragmatic hernia
  - Congenital heart disease
  - Anencephaly
Duodenal atresia

Background
- Failure of primitive bowel to canalize
- Annular pancreas as alternative mechanism
- Aneuploidy in 20%
- Incidence 1:10,000 births

Duodenal atresia

Site of obstruction:
- Preampullary 20%
- Postampullary 80%
- Annular pancreas 21%

Duodenal atresia

Associated CHD
- 15 patients, CHD in 4 (27%)
  - ASD, CAVSD 1 each
  - VSD 2
- Karyotypes from 15/15
  - Abnormal 7 (47%)
- CHD & duodenal atresia
  - Aneuploidy 2/4

Fogel AM J Perinatol 1991;8:411-416
Duodenal atresia

21 WEEKS
Duodenal atresia, 39 weeks

Double Bubble

Double Bubble
Other Findings
Small bowel obstruction

- Site:
  - Jejunum 50%
  - Ileum 43%
  - Multiple 7%

- Etiology:
  - Internal diaphragm 19%
  - Lumen (cord-like) 31%
  - Complete separation 45%

Duodenal atresia?

Small bowel obstruction
**SBO progression – 28 wk**

![Image of SBO progression at 28 weeks]

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**SBO progression – 28 wk**

![Image of SBO progression at 28 weeks]

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**? Esophageal atresia**

![Image of esophageal atresia]

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?Esophageal atresia

Esophageal Atresia/TE Fistula

Diaphragmatic hernia

Associated anomalies
- Structural anomalies
- Secondary anatomic distortion
  - Lung hypoplasia
  - Polyhydramnios
Diaphragmatic hernia

Development of the diaphragm
Fusion of:
  • Septum transversum
  • Dorsal esophageal mesentery
  • Pleuroperitoneal membrane
  • Body wall

Diaphragmatic hernia

Types
Posterolateral - Bochdalek
  • Herniation through pleuropertitoneal canal
Sternal - Morgagni
  • Anterior (pentalogy of Cantrell)
Septum transversum
  • Central tendon
Hiatal hernia
  • Enlarged esophageal orifice

Development of diaphragm
Diaphragmatic hernia

Diaphragmatic hernia

Diaphragmatic hernia
### Diaphragmatic hernia

"Hidden mortality"
- Early series included most favorable postnatal cases compared to prenatal cases
- Effects of multiple anomalies in prenatal series but those patients excluded from postnatal series
- Showed worse mortality for prenatal diagnoses
- Taking stillbirths & deaths prior to transport/full diagnosis into account mortality similar (~75%) pre-ECMO

### Diaphragmatic hernia & CHD

<table>
<thead>
<tr>
<th>N</th>
<th>36</th>
</tr>
</thead>
<tbody>
<tr>
<td>CHD in</td>
<td>5 (14%)</td>
</tr>
<tr>
<td>VSD &amp; tri 18 (n = 2)</td>
<td></td>
</tr>
<tr>
<td>VSD &amp; encephalocele</td>
<td></td>
</tr>
<tr>
<td>VSD &amp; del 8</td>
<td></td>
</tr>
<tr>
<td>VSD &amp; omphalocele</td>
<td></td>
</tr>
<tr>
<td>Aneuploidy in 11 (31%)</td>
<td></td>
</tr>
</tbody>
</table>

Thorpe-Beeston Fetal Therapy 1989;4:21-8
25 1/7 week
Suspected lung mass

28 5/7 weeks
Follow-up scan
Echogenic bowel

- Brighter than bone
- Present in 0.5% normal, 3% T21
- Likelihood ratio ~5
- Watch out for effect of high transducer frequency!
Echogenic Bowel

Hyperechoic Bowel

Echogenic Bowel
Post Processing

Echogenic Bowel

Transducer Frequency
Echogenic Bowel

- Fetal swallowing of blood following amniocentesis or abruption
- Cystic fibrosis
- Down syndrome
- Infection, such as CMV or parvo virus

Echogenic Foci in the Fetal Abdomen

- Echogenic focus in the upper abdomen
  - 26 fetuses with 35 EF in upper abdomen
  - 7 disappeared during pregnancy
  - 9 disappeared in the neonate
  - No abnormalities were noted in any of the neonates

Fetal Abdomen

- Echogenic focus in the upper abdomen
  - 26 fetuses with 35 EF in upper abdomen
  - 7 disappeared during pregnancy
  - 9 disappeared in the neonate
  - No abnormalities were noted in any of the neonates

Where is the Calcification?
It’s in the liver...

Measurements from 3D perpendicular planes: 8 X 4 X 5 mm

Abdominal Cysts

• Ovarian
• Mesenteric
• Bowel duplication
• Bowel obstruction
• Cloaca
Abdominal Cysts

Abdominal Cysts

Abdominal Cysts
• 1. Large Stomach
• 2. Duodenal Atresia
• 3. Jejunal Atresia
• 4. Duplication Cyst
• 5. Normal
• 6. No Clue

Ovarian cyst in female baby. Later images are from 4 weeks later, looks hemorrhagic.
3???

- 1. Normal
- 2. Urachal Cyst
- 3. Ureterocele
- 4. Megacystis Microcolon
- 5. Ovarian Cyst
- 6. No clue

Conclusions

- Remember your anatomy
- Put it all together
- Find other anomalies
- Refer to surgical center as appropriate
- Consider Newer treatments
- Special THANK YOU to Joshua Copel for his contribution of this talk