Anomalies of the Fetal Gastrointestinal Tract

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Learning Objectives

After completing this presentation, the participant should be able to:

1. Recognize the sonographic appearance of normal small and large bowel in the fetus in the second and third
2. Determine the level of fetal bowel obstruction based the sonographic findings
3. Distinguish omphalocele from gastroschisis and understand the clinical importance of differentiating the two.
4. List a differential diagnosis for a cyst in the fetal abdomen based on the location and appearance of the cyst on sonogram

Outline

Fetal gastrointestinal tract
I. Normal sonographic appearance
II. Abnormal
A. Absent stomach
B. Echogenic Bowel
C. Gastrointestinal obstruction
   1. Esophagus
   2. Stomach
   3. Small bowel
   4. Volvulus
C. Ventral wall defects
   1. Omphalocele
   2. Gastroschisis
   3. Pentalogy of Cantrell
D. Intra-abdominal cysts
E. Liver anomalies
F. Intra-abdominal calcifications

Fetal Gastrointestinal Tract

- Esophagus
- Stomach
- Small bowel
- Large bowel
- Liver and biliary tree
- Ventral wall

Disclosures

No Relevant Financial Relationships
### Abdominal Measurement

Assess Stomach presence
To Exclude Esophageal atresia
Diaphragmatic hernia

Stomach location
Situs inversus
Situs ambiguous

Stomach size
Bowel obstruction

Fluid collections
Duodenal obstruction
Dilated bowel loops
Intraabdominal cysts

### Absent Stomach in Left Upper Quadrant

**Normal finding**

**Anomalies**
- Esophageal atresia
- Left diaphragmatic hernia
- Cleft lip
- Central nervous system anomalies
- Situs abnormality
- Severe oligohydramnios

### Bilateral Cleft Lip Diagnosed at 21 weeks

### No stomach seen at 18 weeks gestation

### Situs inversus – 20 weeks
**Normal Bowel**

**Small bowel**
- Located centrally
- Usually more echogenic than colon
- Visible peristalsis in 3rd trimester

**Colon**
- Located around small bowel
- +/- Echogenic in 3rd trimester
- +/- Hypoechoic in 3rd trimester

**Heterotaxy**

- Midline Stomach

- Heterotaxy Stomach Right
  - Right & Left Umbilical Veins

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31 weeks

33 weeks

40 weeks
**Echogenic Bowel**

- Second trimester
  - Bowel as echogenic as bone
- Associated with
  - Cystic Fibrosis
  - Intrauterine growth restriction
  - Trisomies 21 and 18
  - Swallowed blood

**Echogenic bowel at 18w**
- Trisomy 21
- Atrioventricular Canal

**Echogenic bowel at 16w**
- Intrauterine growth restriction at 28w

**Echogenic bowel at 18w**
- Subchorionic hematoma

**Amniotic Fluid Volume**

- Homeostasis depends on
  - Renal function
  - Swallowing and gastrointestinal resorption

**Amniotic Fluid Volume**

- Diminished or absent swallowing
- Inadequate fluid resorption in gastrointestinal tract
  - Polyhydramnios
Severe polyhydramnios due to Myopathy preventing swallowing

Gastrointestinal Obstruction
- Esophageal
- Gastric Outlet
- Duodenal
- Jejunal
- Ileal
- Colonic
Degree of polyhydramnios decreases the more distal the obstruction

Esophageal Atresia
Absence of a segment of esophagus
+/- Tracheo-esophageal fistula
58% have associated anomalies
- Cardiac 15 - 29%
- Gastrointestinal
- Genitourinary

Ultrasound findings
- Severe polyhydramnios
- Absent or small stomach
Esophageal Atresia & Tracheo-Esophageal Fistula

Gastric Outlet Obstruction
- Rare form of gastrointestinal obstruction
- Due to pyloric atresia
  - Primary
  - Secondary to vascular insult
- Ultrasound findings
  - Polyhydramnios
  - Dilated stomach

Pyloric Atresia
- 35 weeks

Duodenal Obstruction
- 50% have associated anomalies
  - Vertebral & Skeletal: 37%
  - Gastrointestinal: 26%
  - Cardiac: 20%
  - Genitourinary: 8%
  - Trisomy 21: 30%

Duodenal atresia – Trisomy 21
Jejunal atresia
Ileal atresia/obstruction
Colonic obstruction

Etiology
- Vascular accident
- Volvulus
- Meconium plug
- Intussusception

Ultrasound findings
- Dilated loops of bowel
- Polyhydramnios

Peritoneal inflammation after bowel perforation

Ultrasound findings
- Ascites
- Cysts with wall calcifications
- Peritoneal calcification with resorption of cystic fluid
Meconium Peritonitis

25 weeks

Small Bowel Volvulus

Ultrasound findings
- Polyhydramnios – acute onset
- Dilated loops of small bowel
- Single, larger, more dilated loop
- +/- “Whirlpool” sign

Volvulus

Volvulus

Volvulus – 34 weeks
Born with dead bowel

Anterior Abdominal Wall

Assess
- Umbilical cord insertion

To Exclude
- Omphalocele
- Gastroschisis
- Amniotic band syndrome
Physiologic Bowel Herniation

- 8 – 11 weeks gestation
- CRL 19 – 41 mm
- Herniated midgut 4 – 7 mm

Omphalocele

- Ventral wall defect at umbilicus
- Herniation of abdominal contents through defect
- Covered by peritoneal membrane
- Commonly associated with other anomalies & aneuploidy

Omphalocele

Associated with:
- Trisomies 13 or 18 (10-20%)
- Other anomalies (50-75%)
- Cardiac anomalies (47%)
- Genitourinary anomalies (40%)
- Neural tube defects (39%)
- Amniotic band syndrome
Beckwith-Wiedemann Syndrome

Macroglossia
Gigantism
Visceromegaly
+/- Omphalocele

60% have anomalies

Ultrasound findings

Polyhydramnios
Large for gestational age
Enlarged kidneys
+/- Macroglossia
+/- Omphalocele

Beckwith-Wiedemann Syndrome

Omphalocele
Anterior diaphragmatic defect
Sternal cleft
Ectopia cordis
Cardiac anomalies
Ventricular septal defect
Left ventricular diverticulum

Pentalogy of Cantrell

Omphalocele
Anterior diaphragmatic defect
Sternal cleft
Ectopia cordis
Cardiac anomalies
Ventricular septal defect
Left ventricular diverticulum
Gastroschisis

- Paraumbilical abdominal wall defect
- Herniation of abdominal contents into amniotic cavity
- Not covered by peritoneal membrane
- Not usually associated with other anomalies

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Gastroschisis

- Young mothers (early 20s)
- Chromosomes usually normal
- ~40% have prenatal bowel dilatation
- ~80% have postnatal complications
  - Infection
  - Gastrointestinal complications:
    - vomiting, reflux, constipation
    - fistulas

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Ultrasound findings
- Bowel loops in amniotic cavity
- No covering membrane
- Normal cord insertion
- Polyhydramnios

Gastroschisis

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Gastroschisis

17 weeks

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Gastroschisis

16 weeks

34 weeks

Intra-abdominal Cysts

- Gastrointestinal duplication cyst
- Mesenteric or omental cyst
- Meconium cyst
- Ovarian cyst
- Choledochal cyst
- Hepatic cyst
- Splenic cyst
- Renal cyst
- Umbilical venous varix

Ultrasound assessment
- Cyst location
- Surrounding structures
- Sex
- Color Doppler

Left upper quadrant
Gastrointestinal duplication cyst

Hepatic cyst
**Liver Anomalies**

- Cysts
- Masses
- Hemangioendothelioma
- Calcifications
- Persistent right umbilical vein

**Hemangioendothelioma**

- Most common fetal hepatic mass
- Ultrasound findings
  - Vascular, solid liver mass
  - ± Displacement of umbilical vein

**Hepatomegaly**

- Usually due to systemic problem
  - Heart failure
  - Malignancy (rare, e.g., leukemia)
Hepatosplenomegaly

Intra-abdominal Calcifications
- Peritoneum
  - Meconium peritonitis
  - TORCH infection
- Liver
  - Hemangioendothelioma
  - TORCH infection
  - Vascular insult
- Gallbladder
- Gallstones

Peritoneal calcifications & Deformed extremities
Due to cytomegalovirus (CMV)

Gallstones – two different patients
- 25 weeks
- 32 weeks

Conclusions
- Ultrasound plays a valuable role in prenatal diagnosis of anomalies of the gastrointestinal tract
- Prenatal diagnosis allows time for patient counseling and planning postnatal care

The End
Key References


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