Abnormalities of the Fetal GI Tract

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Objectives

- Recognize the sonographic features of the normal fetal abdominal structures.
- Describe sonographic findings associated with fetal abdominal abnormalities.
- Correlate other findings associated with fetal abdominal abnormalities.
- Determine sonographic images needed to assist interpreting physician with diagnosis.

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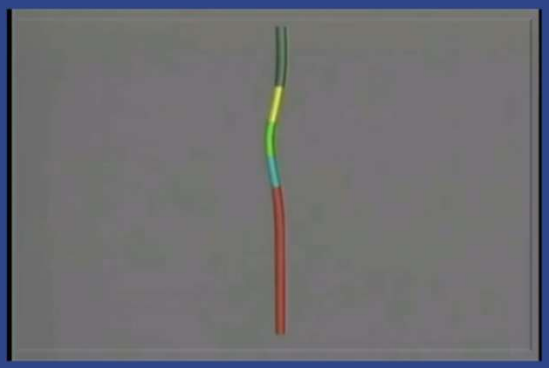
Embryology of the gi tract

- 4th Week Formation
  - Yolk sac is part of the foregut, hindgut, & midgut
- 6th Week
  - Herniation beginning
- 7-10 Weeks
  - Rotation & return to abdomen
  - Returned by 13 weeks gestation
- 20-25 Weeks
  - Haustral folds & accumulation of meconium



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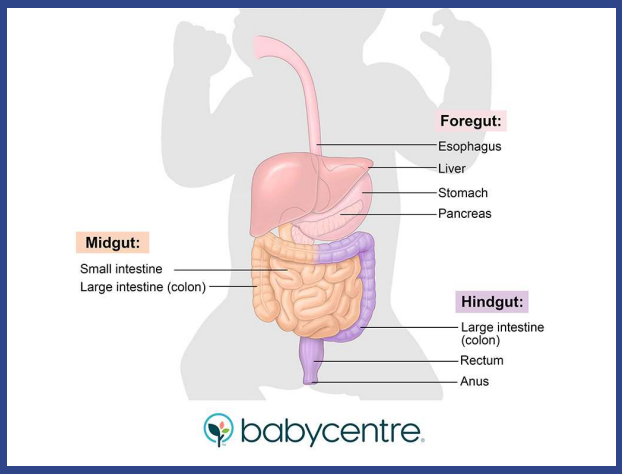
Fetal GI Structures

- Foregut
  - Espophagus
  - Liver
  - Stomach
  - Pancreas
- Midgut
  - Small intestine
  - Large Intestine
- Hindgut
  - Large intestine
  - Rectum
  - Anus



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Sonographic Features - Esophagus

- Can be visualized as early as 15 weeks
- Two echogenic parallel lines located in the neck and posteri chest
- After 26 weeks may have an alternative shape
  - Two parallel echogenic lines
  - Several parallel echogenic lines
- May see swallowing intermittently

https://www.fetalultrasound.com/online/text/3-017.HTM

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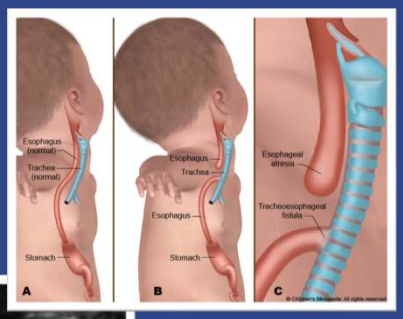
Fetal Esophagus Abnormalities

Esophageal Atresia

- 90% of esophageal atresia cases are associated with tracheoesophageal fistula
- Associated Anomalies
  - Aneuploidy
  - VACTERL
- Sonographic Findings Small to absent stomach bubble
  - Polyhydramnios
  - Esophageal pouch



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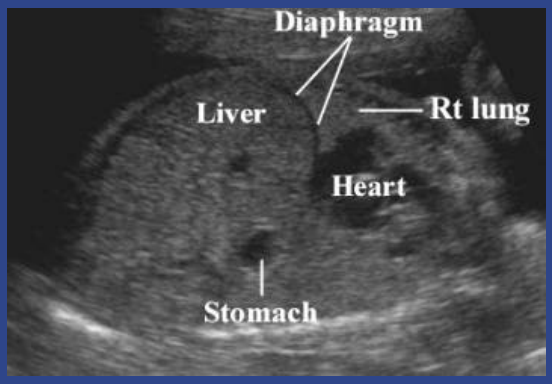
Sonographic Features - Liver

- Homongeneous texture similar to the adult liver
- The confluence of the umbilical vein and portal vein is usually slightly right of the midline
- The fetal liver occupies a large portion of the abdomen, making it important for estimating the fetus's weight.



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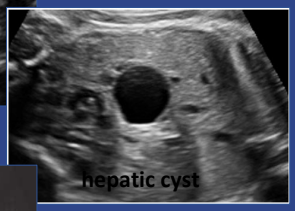
Fetal Liver Abnormalities

- Choledochal cysts
  - Most often fusiform dilatation of the CBD
  - Caroli's Disease
- Hepatic calcifications
  - Idiopathic
  - Hepatic Tumors
  - Vascular insult
  - Infection or aneuploidy – CMV or Trisomy 21
- Hepatic tumors (*rare*)
  - Evaluate with color Doppler to distinguish vascular lesions
  - Highly vascular lesions can lead to high cardiac output and hydrops



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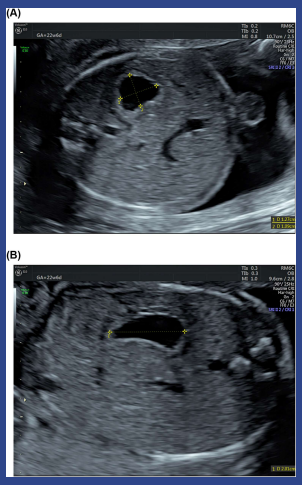
Sonographic Features - Stomach

- Seen by week 7 gestation
- Noted routinely by weeks 13-14
- Normal Position: Upper left abdomen
- The stomach appears as an oval or round, echo-poor structure filled with amniotic fluid



https://obgyn.onlinelibrary.wiley.com/doi/abs/10.1002/pd.5990

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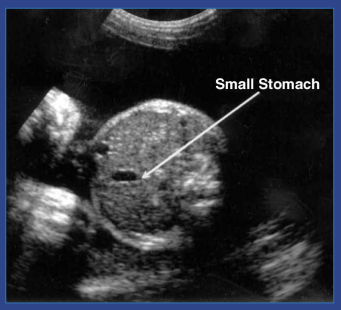
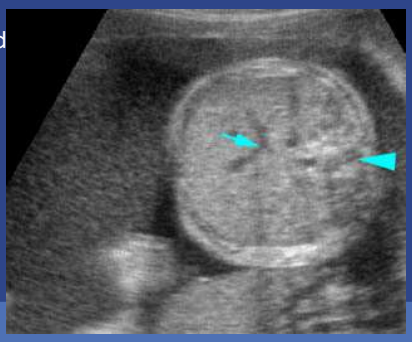
Fetal Stomach Abnormalities

- Absent Stomach
  - Give ample time to fill (30 min)
  - Can be seen with other anomalies
    - Oligohydramnios
    - Aneuploidy (Tracheoesophageal fistula)
  - Other causes:
    - Microgastria – interruption during embryological d
    - Neuromuscular disorder – impairs swallowing
    - Micrognathia
    - Face/neck mechanical obstruction
      - Orofacial cleft
      - Epignathus,
      - Large neck mass/ Teratoma
      - Esophageal Atresia
  - Follow up ultrasound indicated for surveillance





https://w1.med.cmu.ac.th/obgyn/lessons/absent-stomach/

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Sonographic Features - Gallbladder

- Seen between 20-32 weeks, can be seen sometimes throughout 2nd and 3rd trimester
- Often confused for hepatic cyst
- Normal- oblong, teardrop echolucent structure, RUQ of fetal abdomen, right of ML, inferior to UV

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Fetal Gallbladder Abnormalities

- Non-visualized GB is associated with
  - Cystic fibrosis
  - Gallbladder/Biliary atresia
- Gallstones/Sludge
  - 2nd/3rd trimester
  - Can resolve postnatally
- Choledochal Cyst
  - More common in females
  - Most commonly represents dilatation of the CBD
  - Sonographically: GB + Cyst

https://thefetus.net/content/choledochal-cyst-7

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Sonographic Features – Abdominal Wall

- Forms during the 4th week
  - Layers may not close completely resulting in abdominal wall defect
- Anterior abdominal wall does not fuse, becomes a ventral abdominal wall defect
- The differential diagnosis for a ventral abdominal wall defect includes several anomalies but the most common are omphalocele and gastroschisis.

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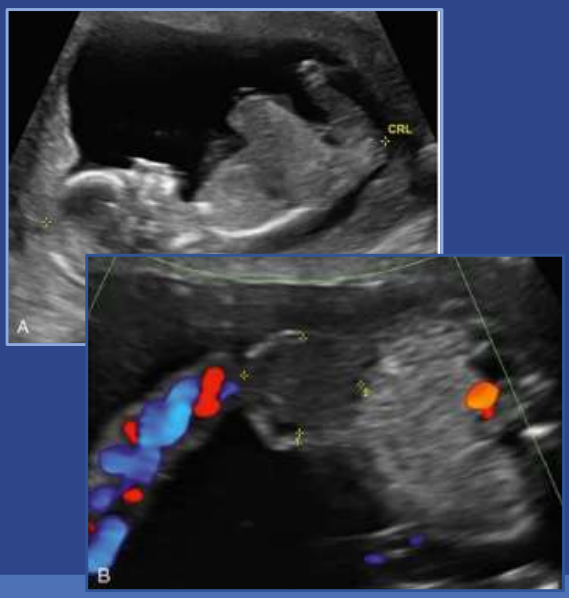
Omphalocele

- Midline defect in anterior abdominal wall with abdominal content herniated into the base of the umbilical cord
- Covered by peritoneum and amnion
- Associations
  - Advanced maternal age (AMA)
  - Aneuploidy
    - Trisomy 18 – most common
    - Trisomy 13
    - Beckwidth-Wiedemann syndrome
- Sonographic findings
  - Umbilical cord inserts into the base of the omphalocele sac
  - Membranous sac covering abdominal contents
  - Visualization of the liver in the omphalocele has decreased likelihood for chromosomal abnormality if seen in the 2nd trimester



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Gastroschisis

- Congenital malformation with abdominal contents herniating through a right-sided abdominal wall defect
- Seen adjacent to the normal umbilical cord insert
- **Not covered by a membrane**
- Almost all cases are right sided and do not involve liver
  - Free floating loops of bowel
- Maternal age younger than 20
- Elevated MSAFP
- Associated GI abnormalities
  - Atresia, stenosis, perforation

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3D imaging

- Provides Multiplanar imaging and surface rendering provide additional information
- May result in Better estimation of fetal weight
- Better estimation of volume of amniotic fluid
- Provide additional information in 51% of cases with fetal anomalies
- Helpful in counseling and improving of understanding of the patients about fetal anomalies

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Sonographic Features – fetal bowel

- Fetal Small Bowel
  - 12 – 16 weeks appears homogeneous
  - Located centrally within fetal abdomen
  - Increasing in echogenicity as gestation progresses
  - Maximum diameter of normal fetal small intestine: 7mm
- Fetal Colon
  - Visualized as early as 22 weeks gestation
  - Located peripherally in the fetal abdomen
  - Hypoechoic bowel segments
  - Variable in size
  - Haustral folds

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Fetal Bowel Abnormalities

- Echogenic Bowel
  - **Aneuploidy (T21**)**
  - **Cystic Fibrosis**
  - **Infection**
  - Edema of the bowel wall
  - Decreased amniotic fluid volume
  - Presence of meconium
  - Intestinal ischemia causing hypomobility
  - Swallowed blood after intra-amniotic bleeding

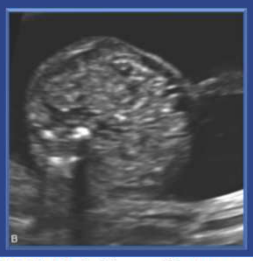

TABLE 14-4 Adverse Outcomes Associated With Echogenic Bowel

Aneuploidy, in particular Down syndrome
Cystic fibrosis
Growth restriction and fetal demise
Congenital infection, in particular cytomegalovirus (CMV)
Gastrointestinal obstruction

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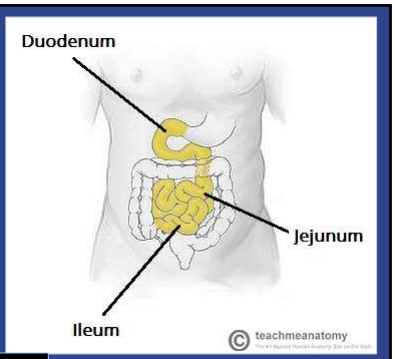
Jejunoileal Obstruction

- Jejunoileal Atresia
  - Most frequent cause of fetal bowel obstruction
  - Commonly associated with cystic fibrosis
- Sonographic appearance
  - Hallmark sign of distal small bowel obstruction is dilated bowel loops
  - Mid-abdominal loops of bowel
  - Enlarged stomach
  - Abdominal calcifications
  - Ascites
- Other Findings
  - Polyhydramnios
  - Dilated stomach
  - Echogenic bowel – due to meconium
- High incidence of prematurity and growth restriction



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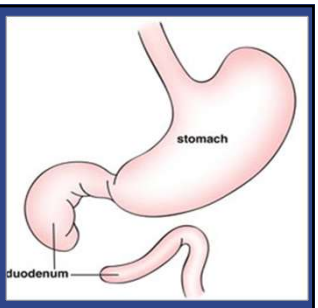
Duodenal obstruction

- Most common perinatal intestinal obstruction
- Cause
  - Intrinsic – Atresia, Annular pancreas
  - Extrinsic – Constriction
- Sonographic Appearance
  - Double bubble sign
    - Continuation of dilated duodenum with fluid in the stomach, crossing the fetal midline
  - Polyhydramnios
- Associated anomalies and structural abnormalities
  - Trisomy 21 (50 %)
  - VACTERL
  - Congenital Heart Disease



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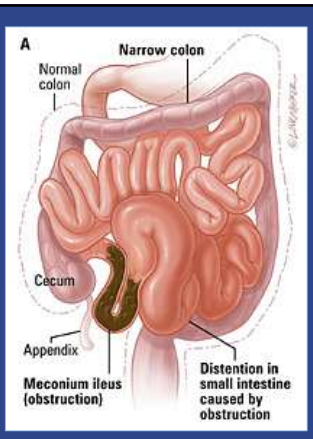
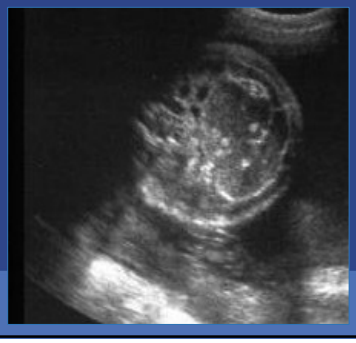
Meconium ileus

- Associated with cystic fibrosis
- Sonographic features
  - Dilated fluid filled loops of bowel above the ileum
  - Echogenic dilated bowel (meconium)
  - Small distal colon





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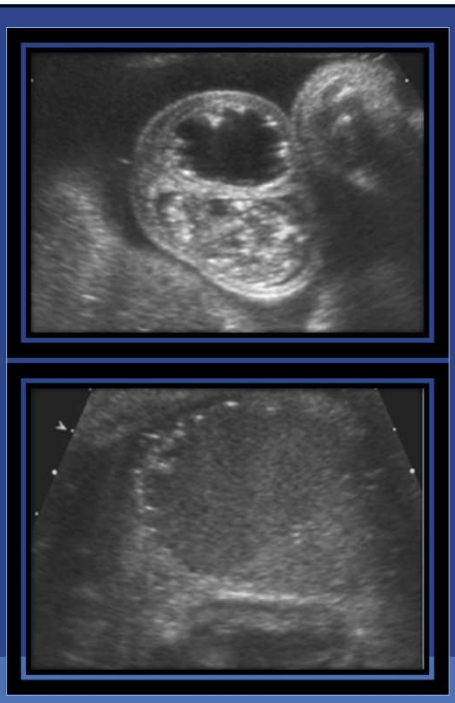
Meconium peritonitis

- Results from inflammation that occurs in response to intestinal rupture
- Associated with cystic fibrosis
- Perforation due to
  - Bowel atresia, volvulus, intussusception, and vascular compromise
  - Unknown etiology
- Sonographic features
  - Intraperitoneal calcifications
    - Surface of the liver or under the diaphragm, outlining bowel
  - Ascites
  - Polyhydramnios



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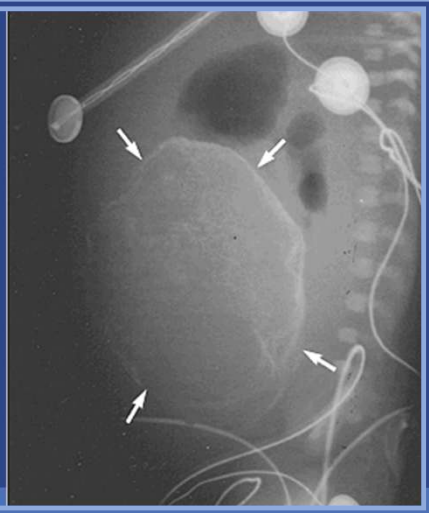
Meconium psuedocyst



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Persistent cloaca

- Most severe form of anorectal malformation
  - Rare
  - Only in females
- Distal portion of urinary tract, vagina, and rectum all lead to a single opening into the perineum

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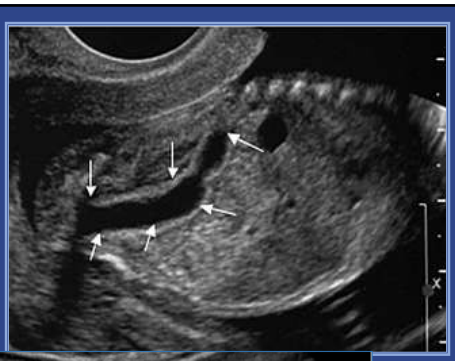
Anal atresia

- Most common large colon atresia
- 98% have associated anomalies
- Associations
  - VACTERL sequence
  - Chromosomal and GU anomalies
- Sonographic findings
  - V or U shaped dilated bowel in pelvis
  - Intraluminal calcification



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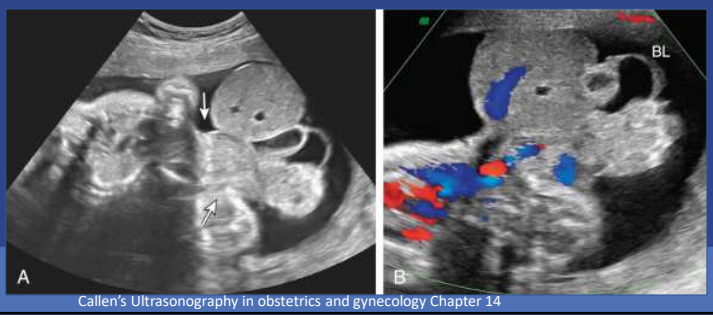
Limb-body-wall complex

- AKA Body Stalk Anomaly
- Rare complex abnormalities resulting from failure of closure of the ventral body wall
- Two or more of the following abnormalities:
  - Wall defects
  - Excencephaly
  - Encephalocele
  - Scoliosis of the spine (77%)
  - Limb defects (95%)
  - Facial & cranial anomalies



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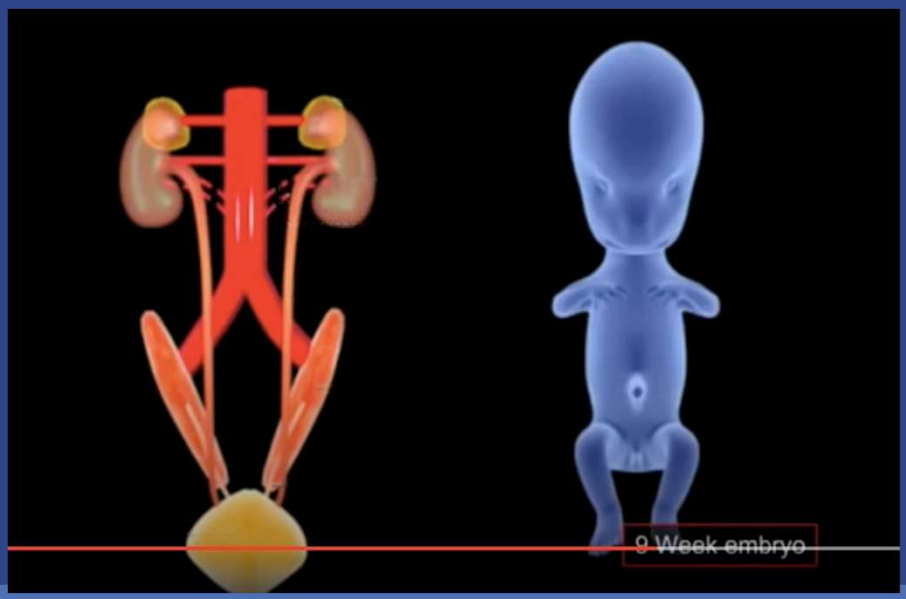
Cloaca



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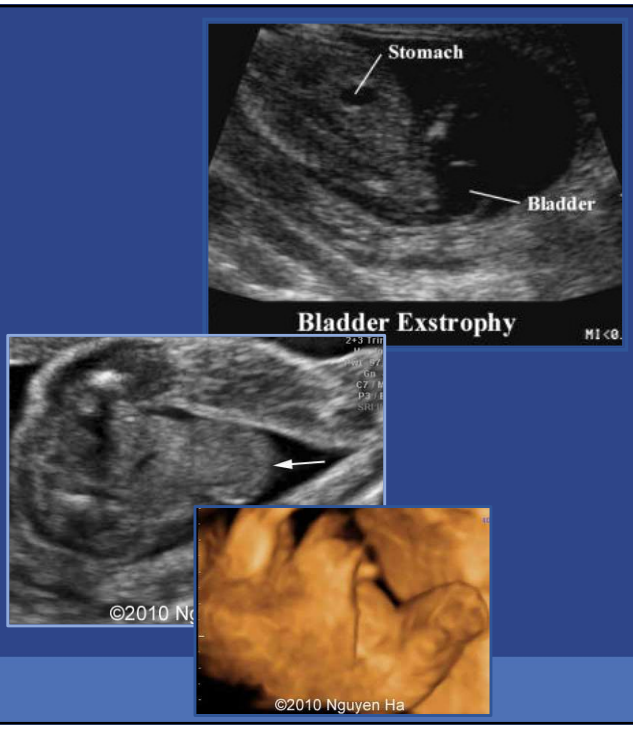
Bladder exstrophy

- Bladder located externally on abdominal wall
- Associated findings
  - Absent bladder
  - Epispadias and undescended testicles in males - Bladder exstrophy- Epispadias complex (BEEC)
  - Lower abdomen bulge representing bladder
  - Wide separation of pubic bones
  - Low cord insertion



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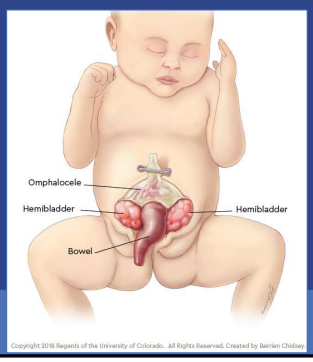
Cloacal exstrophy

- Persistent Cloacal Membrane
  - Interferes with abdominal wall closure
  - Results in failure of the separation of the urogenital septum from the rectum
  - Combination of omphalocele, exstrophy, imperforate anus & spinal defects (OEIS)
- Sonographic Features
  - Solid mass herniating from an infraumbilical defect
  - Bladder is not present internally
  - Large anechoic mass (cloaca)
  - Abnormal genitalia
  - Hemi-bladder



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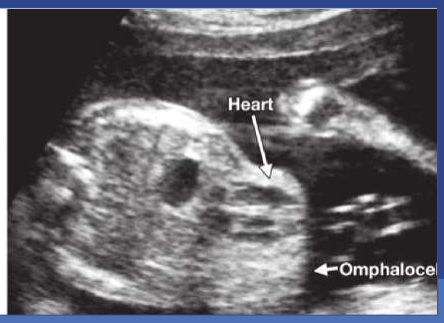
Ectopia cordis

- Midline defect with all or part of the heart extruded from the thorax
  - With or without a membrane covering the heart
- Pentalogy of Cantrell
  - Anterior diaphragmatic hernia
  - Omphalocele
  - Cardiac defects
  - Defect of the pericardium
  - Lower sternal defect



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