Prenatal Diagnosis of Gut Herniations by Ultrasound

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Uses of Ultrasound (US) in Obstetrics

- establish the presence of a living embryo/fetus.
- estimate the age of the pregnancy.
- diagnose congenital abnormalities of the fetus.
- evaluate the position of the fetus.
- evaluate the position of the placenta.
- determine if there are multiple pregnancies.
- determine the amount of amniotic fluid around the baby.
- check for opening or shortening of the cervix or mouth of the womb.
- assess fetal growth.
- assess fetal well-being.

http://www.radiologyinfo.org
Fetal US: Transverse Planes

Transverse plane through brain

Transverse plane through abdomen

Meet Our Patient

39 yo F, G4P0, wanted First Trimester Screening

- Previous spontaneous abortion and 2 D&C elective abortions
- No significant PMH or other surgeries
About First Trimester Screening

- Performed between 11 and 13 weeks LMP
- Evaluates risk of Trisomy 21, 18 and 13
- 85% sensitive, 5% false positive rate

Parameters:
- Maternal age
- Serum levels of βhCG and pregnancy-associated plasma protein A (PAPP-A)
- US measurement of nuchal translucency
Our Patient’s Fetal US: Nuchal Translucency

First Trimester Screening Results:

• Risk of Trisomy 21 = 1:9

• Risk of Trisomy 13 or 18 = 1:73

Nuchal Translucency = 3.0 mm (normal <3.0 mm)
Our Patient’s Fetal US:
Gut Herniation

12 weeks LMP

Transverse plane through mid-abdomen shows a prominent gut herniation at the umbilical cord insertion site.
Overview of GI Embryology

FIGURE 41-22 Normal development of the anterior abdominal wall. A, Herniation into the base of the proximal umbilical cord (9 weeks’ gestation). B, This bowel undergoes a 90-degree rotation along the axis of the superior mesenteric artery. C, At approximately 12 weeks’ gestation, the bowel returns to its normal position in the abdominal cavity, undergoing an additional 180-degree rotation along the axis of the superior mesenteric artery.

From: Rumack, Diagnostic Ultrasound, 3rd Ed. 2005
Companion Patient 1

- 32 yo F, G3P1, presented for First Trimester Screening
- Unsure of dates, IUD removed 2 mo ago
  - ~ 11.5 weeks
- Previous Cesarean delivery at 39 weeks for breech
- Previous elective D&C abortion
- No significant PMH
Companion Patient 1 Fetal US: Gut Herniation

Gestational age by CRL: 11 weeks 1 day
Companion Patient 1 F/U US: Normal Cord insertion

Transverse view shows normal cord insertion without gut herniation

Doppler US confirms that the umbilical cord contains only patent vessels
Physiologic Gut Herniation

- Gut herniation is physiologic up to 12 weeks LMP
- >12 weeks LMP, gut herniation suggests an anterior abdominal wall defect
Returning to Our Patient

- Chose to have Sequential Screening Test at 16 weeks
  - Parameters:
    - alpha-fetoprotein (AFP)
    - unconjugated estriol (uE3)
    - β hCG
    - Dimeric inhibin A (DIA)
  - Results:
    - Risk of Trisomy 21: 1:5
    - Risk of Trisomy 13 or 18: 1:61

- Had a F/U ultrasound at 16 weeks to reassess gut herniation
Our Patient’s F/U Fetal US: Anterior Abdominal Wall Defect

Transverse plane through mid-abdomen still shows a prominent gut herniation at the umbilical cord insertion site indicating an anterior abdominal wall defect.
About Anterior Abdominal Wall Defects

- Detection rate by US is 65-98% (largely reflects operator variability)

- Four major types:
  - Omphalocele
    - Most common (1:4000 live births)
    - Herniation of abdominal contents into base of umbilical cord
  - Gastroschisis
    - Occurs in 1:10,000 live births
    - Loops of bowel protrude through all layers of abdominal wall
  - Pentology of Cantrell
    - Omphalocele, a diaphragmatic defect, a pericardial defect, ectopic heart and disruption of the sternum, craniofacial anomalies
  - Limb-body wall complex
    - A neural tube defect, an anterior abdominal wall defect, and limb anomalies, often severe scoliosis

- Cause elevated $\alpha$-fetoprotein (AFP) in amniotic fluid and maternal serum
Our Patient’s F/U Fetal US: Omphalocele

Transverse plane with doppler US shows the gut herniation is centrally located within the umbilical cord insertion site indicating it is an omphalocele.
Two chamber view of the heart shows a \textit{ventricular septal defect}.

Doppler US confirms that there is communication between the R and L ventricles.
Our Patient’s Outcome

Because of high risk results of First Trimester and Sequential Screening exams, our patient chose to have amniocentesis performed at 18 weeks LMP.

The amniocentesis proved the fetus had Trisomy 18, Edwards syndrome.

- Trisomy 18 is associated with abdominal wall, kidney, and cardiac defects and multiple structural abnormalities
- 67% of fetuses detected by amniocentesis die before term
- 90% live-born die by 1 year of age (median survival= 8 wks)

Our patient underwent elective abortion of the fetus at 18 weeks LMP.
About Omphalocele

- Located centrally within umbilical cord
- 30-40% are associated with chromosomal abnormalities (rare if liver is involved)
- 75% associated with other structural defects, especially cardiac, other GI and GU
- 5-10% are part of Beckwith-Weidemann syndrome: gigantism, renal tumors, hemihypertrophy, and macroglossia

From: Rumack, Diagnostic Ultrasound, 3rd Ed. 2005
Example of Giant Omphalocele on Fetal US

Transverse plane shows a large gut herniation containing liver (L) with the portal vein (PV), small bowel (SB) and the stomach (S)
Companion Patient 2

- 19 yo F, G2P1 presented for First Trimester Screening
- No significant PMH
- Prior delivery at 39 weeks, no complications
- Smokes ½ ppd; boyfriend smokes 1 ppd
Companion Patient 2 Fetal US: Gut Herniation

Sagittal plane shows a gut herniation
Transverse plane with doppler US shows the herniation is lateral to the umbilical cord insertion.

From: Rumack, Diagnostic Ultrasound, 3rd Ed. 2005
Omphalocele vs. Gastroschisis

**Omphalocele**
- Midline cord insertion site
- Variable size (2-10 cm)
- Liver can be involved
- Membrane present
- Often ascites and bowel wall thickening
- Often cardiac, GI and GU defects
- Often chromosomal abnormalities

**Gastroschisis**
- Paraumbilical location
- Small (2-4 cm)
- Bowel only
- No membrane
- Bowel wall thickened but usually no ascites
- Rarely associated with other structural or chromosomal abnormalities
- Associated with IUGR and smoking
Omphalocele on 3D US

Ultrasound Obstet Gynecol 2002; 20: 635–637
Omphalocele on MR Imaging

Sagittal T2-weighted MRI of a fetus with omphalocele containing liver, some bowel and part of the stomach
Summary

In this presentation you learned:
- The uses of US in obstetrics and common US planes for assessing the fetus.
- The appearance of gut herniations on US and how to differentiate physiologic gut herniation from abdominal wall defects.
- How to distinguish omphalocele from gastroschisis on fetal US.

You were offered fetal US examples of:
- Omphalocele
  - Small-bowel only
  - Giant with liver involvement
  - 3D US and MRI views for comparison
- Gastroschisis
- Physiologic gut herniation
- Ventricular septal defect
- Abnormal nuchal translucency
Take-home Pearls

- Fetal gut herniation can be seen on US in transverse and sagittal planes during first trimester fetal US.

- Gut herniation is physiologic up to 12 weeks LMP; if present after 12 weeks, it suggests an anterior abdominal wall defect.

- Omphalocele is seen on fetal US as a central umbilical cord herniation.

- Gastrochisis is seen on fetal US as a lateral umbilical cord herniation.
References


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