

American Pediatric Surgical Association

Prenatal Counseling Series

Omphalocele



APSA
American Pediatric
Surgical Association
Saving Lifetimes™

from the
Fetal Diagnosis and Treatment Committee

of the
American Pediatric Surgical Association

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Prenatal Counseling Series Omphalocele

Definition and Etiology

- Omphalocele is one of the two most common abdominal wall defects encountered by pediatric surgeons.
- Etiology: failure of the migration of lateral folds to form the umbilical ring and failure of the herniated midgut to return to the abdominal cavity early in gestation.
- Incidence: 2.5/10,000 to 4/10,000 (1)



Axial ultrasound and sagittal MR images of a fetus with a ventral abdominal wall defect that contains a small portion of the liver. The umbilical cord inserts onto the defect.

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Anatomy

- Ventral abdominal wall defect covered by a sac
 - Sac layers: Peritoneum, Wharton's jelly, Amnion
 - 10-20% of sacs may rupture prenatally
- Anomalous umbilical cord insertion into the omphalocele membrane as opposed to the abdominal wall
- Size: ranges from small (hernia of the cord) to giant (variable definition in literature based on liver involvement and size of the defect) (2, 3)
- Abdominal location:
 - Epigastric omphalocele → Pentalogy of Cantrell | Cephalic fold
 - Central omphalocele → Classic Omphalocele | Lateral folds
 - Hypogastric omphalocele → Cloacal Exstrophy | Caudal Folds



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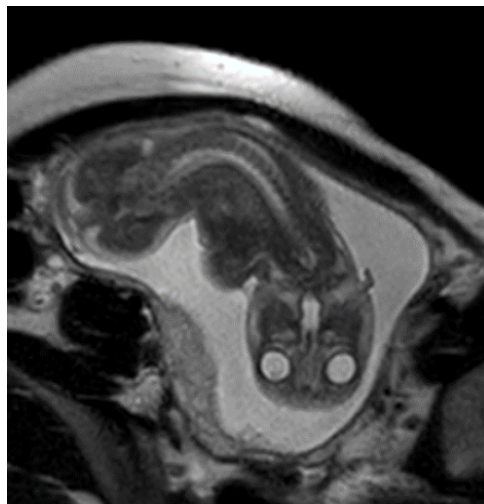
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Differential Diagnosis

Differential diagnosis of a prenatally diagnosed congenital abdominal wall defect includes:

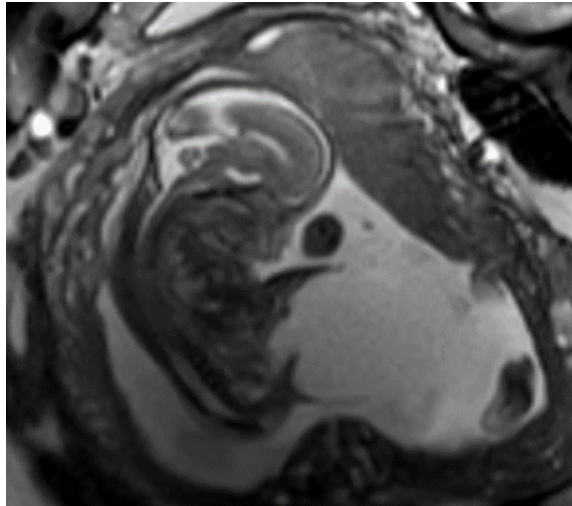
- Ecopia cordis
- Bladder exstrophy
- Cloacal exstrophy
- Urachal anomalies
- Complex abnormalities
 - Pentalogy of Cantrell (abdominal wall defect, anterior diaphragmatic hernia, cardiac anomaly, pericardial defect and sternal cleft)
 - Limb-body wall complex/body stalk anomaly
 - OEIS: Omphalocele, Exstrophy, Imperforate anus and spinal dysraphism

Body Stalk Anomaly



Coronal MR images of the maternal uterus and fetus with body stalk anomaly demonstrate a large omphalocele containing liver, stomach, and bowel in addition to marked kyphoscoliosis. The umbilical cord is abnormally short, and the ventral abdominal defect is approximated to the placenta throughout the examination, raising concern for tethering. The fetus also has bilateral clubfoot.

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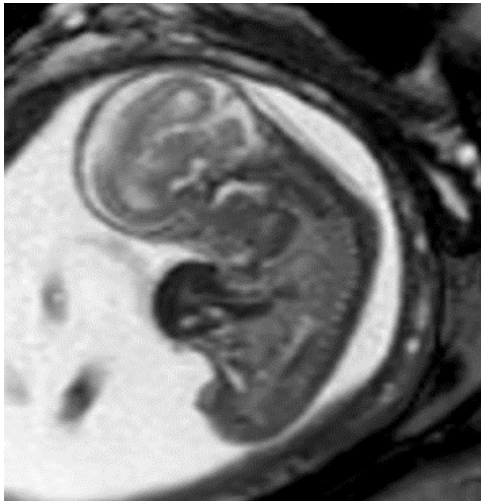


OEIS

Sagittal MR image of a fetus with OEIS (omphalocele, cloacal exstrophy, imperforate anus, spinal defects) complex shows a large ventral wall defect involving the abdomen and pelvis with dominant cystic component consistent with omphalocele and cloacal exstrophy. Also, there is a lumbosacral spinal defect with a dorsal cyst.

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Pentalogy of Cantrell

Sagittal and axial MR images of a fetus with a ventral wall defect involving the upper abdomen with extracorporeal liver. The cardiac apex is located at the superior margin of the defect, raising concern for Pentalogy of Cantrell.

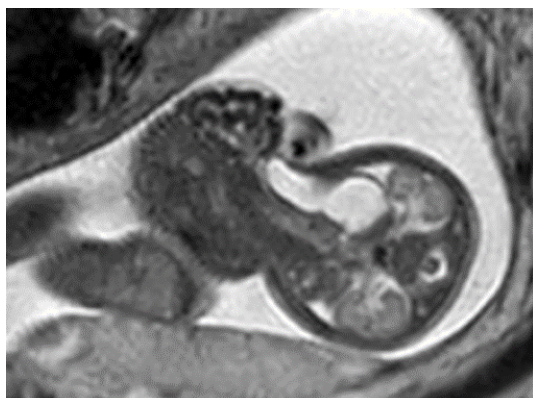
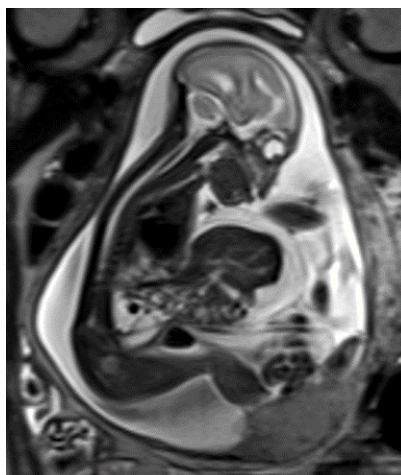
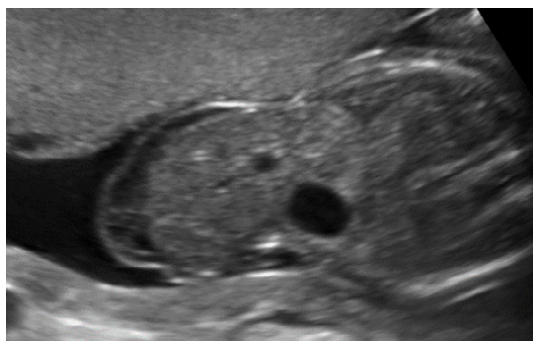
Associated Anomalies

- Omphaloceles are commonly associated with other anomalies (50-70%)
- Chromosomal abnormalities (20-30%), especially Trisomy 18, 13, and 21
- Beckwith-Wiedemann Syndrome
- Bladder exstrophy
- Congenital heart defects (3)

Giant Omphalocele

Giant omphalocele lacks a consensus definition (2,3):

- Historically defined by various criteria including: the diameter of the sac or the abdominal wall defect, inability to primarily close the defect, liver evisceration, and volume disproportion between the abdominal viscera and abdominal cavity.
- Based on recent larger series, omphaloceles are generally considered giant when:
- Defect size: >5 cm
- Liver herniation: >50% of the liver within the sac
- Giant omphaloceles can be associated with varying degrees of (4):
 - Pulmonary hypoplasia
 - Pulmonary hypertension
 - Systemic hypertension
 - Inguinal hernias
 - Undescended testes
 - GERD
 - Feeding difficulties



Axial ultrasound, axial and sagittal MR images of a fetus with a giant omphalocele show a large ventral abdominal wall defect covered by a thin membrane containing the majority of the liver, bowel and the gastric antrum.

Initial Evaluation

Obstetrical ultrasound
Fetal echocardiography
Fetal magnetic resonance imaging (MRI)
Genetic testing

Obstetrical ultrasound (5,6,7)

- Assess for protrusion of viscera through the abdominal wall
- Presence of an overlying membrane (sac)
- Insertion of umbilical cord into the defect
- Presence of associated anomalies
- Measure the size of the defect
- Presence of extracorporeal liver

Echo

- Cardiac structure and function
- Assess for any cardiac anomalies

Fetal MRI

MRI is complementary to ultrasound and is particularly useful for distinguishing liver from the bowel that at times may appear similar by ultrasound

- Measure the proportion of extracorporeal liver
- Quantify defect size
- Useful for associated abnormalities
- Total lung volume (degree of pulmonary hypoplasia)
- Observed/expected total lung volume (O/E TLV) calculated using normative data by gestational age
- O/E TLV <50% predictive of increased postnatal morbidity

Genetic Testing

- Should be performed regardless of whether it is a small omphalocele or hernia of the cord
- ↑ in maternal serum AFP and B-HCG
- Offer genetic testing for Beckwith-Wiedemann syndrome
- Aneuploidies

Prenatal Counseling

- Can be diagnosed as early as 10-12 weeks on prenatal screening ultrasound
- Serial ultrasound examinations to assess fetal growth and amniotic fluid volumes
- Monitor pregnancy for preterm labor and intrauterine growth restriction
- Close antenatal surveillance is recommended due to the possibility of late gestational fetal demise
- Isolated omphalocele defects have a good prognosis

Giant Omphalocele

- Referral to a fetal center for multidisciplinary counseling including pediatric surgery, MFM, neonatology and genetics
- Obstetrical ultrasounds every four weeks
- Consider fetal MRI at 34 weeks in giant omphaloceles to assess for pulmonary hypoplasia and to obtain an objective evaluation of total lung volume
- Twice weekly non-stress tests or biophysical profiles starting at 32 weeks
- Major consideration for the mode of delivery is related to the risk of rupture
 - Pregnancy should be allowed to proceed as close to term as possible
 - For giant omphaloceles, Cesarean section may be justified
- **Rupture** is an emergent situation!

Postnatal Considerations

- The clinical management of these defects varies from straight forward to complex. The morbidity and mortality are often linked to the associated congenital anomalies and size of the defect.
- Evaluation for syndromic features (enlarged tongue, heart defects, and hemihypertrophy)
- Full genetic evaluation as the presence of chromosomal anomalies, cardiac defects, and syndromic conditions impacts outcomes and timing of surgical repair
- Assess for pulmonary hypertension and pulmonary hypoplasia
- Surgical repair via either primary closure or delayed primary/secondary closure

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