

11. VISCERAL AND PARIETAL ANOMALIES

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ABDOMINAL WALL DEFECTS

Definitions

- Omphalocele (also called exomphalos)
 - Defect is central, involving the umbilicus
 - Often covered by a membrane, “protecting” viscera; membrane may be ruptured
 - >30 % have associated anomalies
 - Chromosomal anomalies: trisomy 18
 - “Upper midline” defects: Pentalogy of Cantrell (including heart, pericardium, pleural)
 - “Lower midline” defects: cloacal exstrophy
 - Etiology: depends on type; unclear
- Gastroschisis (laparoschisis)
 - Defect is always to the side of an intact umbilicus; usually right of midline
 - Never covered by a membrane
 - Associated anomalies are rare, except (presumably secondary) intestinal ones (atresia)
 - Correlation with young maternal age
 - 4-5 times more common than omphalocele

Diagnosis

- Ultrasound very sensitive, and highly specific:
 - Position of defect and umbilical cord insertion (omphalocele vs. gastroschisis)
 - Difficult < 16 weeks; not possible < 12 weeks (‘physiologic’ omphalocele at 8-11 wk)
 - Importance of finding associated anomalies:
 - Chromosomal (amniocentesis) (omphalocele)
 - Heart, bladder, pelvis (exstrophy), limbs: omphalocele
 - Major limb/body wall deformities: rare form of severe amniotic band syndrome sometimes associated with gastroschisis – highly lethal

- Gastrointestinal anomalies: intestinal loop distension not specific/sensitive for atresia
- Grading: “giant” omphalocele contains liver; small omphalocele = “hernia of the cord”
- Alpha-fetoprotein (AFP) elevated (amniotic fluid and maternal serum): reflects ‘leakage’ of body proteins through any breach in fetal skin: gastroschisis, spina bifida

Prenatal management

- Alterations in time, place and mode of delivery
 - Decision to deliver in tertiary center (with neonatal ICU and surgical services)
 - C/Section: usually not necessary, except for giant omphalocele (liver trauma)
 - Early delivery:
 - Controversial. Rationale: prolonged exposure of intestinal loops to “caustic” amniotic fluid increases bowel wall damage and impairs intestinal function
 - Definitely not applicable to covered defects: omphalocele with intact membrane
 - No evidence that it improves outcome or bowel appearance in gastroschisis (or ruptured omphalocele)
 - Recent studies: worse outcome (closure of defect, return of bowel function) if infant born near- (35-37 wk), rather than at term (>37 wk)
- Most important prenatal intervention: parental counseling and preparation for perinatal care
- Other interventions (i.e., infusion of saline to dilute or replace “caustic” amniotic fluid): no

Postnatal management and long-term outcome

- If possible: primary repair of defect
- In 30-40%: not possible without damage to intestines, liver, compartment syndrome
 - Temporary silo: sterile coverage of intestines to avoid infection, dessication
 - Delayed closure of defect (5-10 days later)
 - Difficult cases: giant omphaloceles (kinking of vena cava, non-compliant liver)
 - Risk of chronic ventral hernia
 - Gastroschisis: concomitant small bowel atresia in 10-15%
- Long-term outcome: normal life expectancy, intestinal function after 1st year; survival >80%
 - Omphalocele: prognosis depends mostly on associated conditions (chromosomal, ...)

INTESTINAL OBSTRUCTION

Differential diagnosis

- High obstruction (esophageal atresia, duodenal atresia)
 - Fetus cannot swallow amniotic fluid → polyhydramnios

- Low obstruction (mid-, distal small bowel, colon)
 - Normal amniotic fluid volume
 - Distended intestinal loops on ultrasound
- Heterogeneous etiologies
 - Isolated esophageal, duodenal atresia
 - Duodenal atresia and trisomy 21 (Down syndrome)
 - Malrotation/midgut volvulus
 - Meconium ileus: associated to cystic fibrosis
 - Hirschsprung disease
 - Combinations: meconium ileus → obstruction → volvulus → local bowel ischemia → segmental necrosis → atresia

Prenatal management

- If underlying condition suspected: prenatal diagnosis
 - Meconium ileus and testing for cystic fibrosis (see chapter 5c)
 - Duodenal atresia and karyotyping (Down; see chapter 5c)
- No indications for prenatal intervention
 - Delivery in tertiary center recommended

ECHOGENIC BOWEL

Definition

- Bright appearance of bowel or bowel wall on ultrasound
- Etiology unclear – is a secondary effect of several possible causes:
 - Meconium ileus (cystic fibrosis)
 - Intestinal obstruction
 - Swallowed blood (e.g. days after amniocentesis)
 - Most often idiopathic

Significance

- Unclear
- Further testing recommended:
 - Infectious screen
 - Amniocentesis, consider cystic fibrosis; chromosomal anomaly (trisomy)
- If associated with maternal bleeding: poor pregnancy outcome

- If underlying condition: prognosis depends on that condition
- If intestinal obstruction: good prognosis, but postnatal surgical intervention likely

ABDOMINAL MASSES AND CYSTS

Differential diagnosis

- Intestinal obstruction (see above)
- Meconium peritonitis: intestinal rupture/perforation → sterile peritonitis → inflammation causes pseudocyst/walled off meconium-stained pocket. Impressive abdominal distension, but rarely an emergency; prognosis depends on underlying condition (often cystic fibrosis)
- True cyst: etiology difficult to establish; process of elimination
 - Ovarian cyst (check gender)
 - Choledochal/liver cyst
 - Mesenteric cyst/lymphangioma
 - Cystic kidney disease (see chapter on urologic anomalies)
 - Adrenal cyst: adrenal hemorrhage, cystic neuroblastoma
 - Intraabdominal pulmonary sequestration
 - Cystic teratomas (ovary, retroperitoneum)
 - Anterior neural tube defect/neurenteric cyst

Management

- Can almost always wait until after delivery
- If very large: risk of dystocia/C-section may be indicated
- Ovarian cyst: if large (>4-5 cm): risk of torsion and loss of entire ovary
 - Antenatal intervention (needle aspiration) justified? Balance against risk to the fetus
 - Usually best managed (immediately) postnatally
 - Etiology: maternal hormonal stimulation → regression postnatally
 - Heterogeneous appearance, debris, mixed cystic/solid: suspect teratomas or torsion

FETAL TUMORS

Fetal tumors, benign or malignant, are rare

Even malignancies usually have a more benign course than their counterparts in children

- Neuroblastoma (foci present in 1/100 adrenal glands at 20 weeks gestation; spontaneous regression/apoptosis)

- May require excision at birth, or observation (often regresses)
- Rare cases are (or become) metastatic
- No indication for prenatal intervention
- Teratoma
 - Sacrococcygeal and ovarian most common
 - Giant sacrococcygeal teratoma may cause high output cardiac failure and hydrops
 - Indication for fetal surgery if large and symptomatic <20 weeks gestation
 - Risk of preeclampsia-like syndrome in mother ("mirror syndrome")
 - Retroperitoneal teratoma
 - Cervical teratoma
 - Very rare
 - May be voluminous – risk of airway compromise at birth → consider EXIT procedure (see chapter 12: Fetal treatment)
- Wilms tumor (kidney)
 - Exceedingly rare in fetus

NECK MASSES

Significance:

- Severe, acute respiratory compromise at birth
 - Obstruction of upper airway
 - Distortion of face/mouth/nose
- May require prenatal planning, EXIT procedure (see chapter 12)

Types of masses:

- Teratoma (see above)
- Hemangioma
- Cystic hygroma (giant lymphangioma)