Visceral and Parietal Anomalies

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Role of fetal diagnosis

Prenatal findings

Do we need to intervene prenatally?
Do we need to deliver this baby early?
What do we expect for this baby postnatally?

Postnatal diagnoses
Abdominal compartment

• **Prenatally we can detect:**
• Defects in the abdominal wall
• Intra-abdominal variations
  – In the caliber/thickness of the bowel
  – In the appearance of bowel
• Intra-abdominal masses/cysts
Parietal defects

- Defects of the abdominal wall
  - Gastrochisis
    - full-thickness/off center
  - Omphalocele
    - covered muscular defect/central
  - Prunebelly syndrome
    - Laxity of intact wall
4th week of development

Progressive cranio-caudal folding by excessive elongation of neural plate

http://www.embryo.chronolab.com
4th week of development

Lateral edges of germ disk
Fold and fuse on midline

http://www.embryo.chronolab.com
Clinical impact

• Two very different entities

Omphalocele

Gastroschisis
Omphalocele

• Newborn Characteristics
  – Incidence 1/4000 – 1/7000 Live births
  – Midline defect covered with membrane
  – Liver typically within sac
  – Associated anomalies: - 50% none
    • with chromosomal (trisomy 13 and 18) 10-20%
    • cardiac anomalies 30-35%
    • overgrowth syndromes – Beckwith Wiedeman

• Maternal Characteristics
  – Advanced age,
  – MS-AFP elevated
Fetal diagnosis

• Fetal U/S Characteristics
  – Contour change of abdominal wall
    • Liver extends beyond confines of abdominal cavity
  – Membrane covers AWD and cord attaches to it.
    • Cord insertion site appears distant from abdominal wall.
  – May be difficult to determine size of defect
    • Hernia of cord, omphalocele, giant omphalocele
Omphalocele

• Postnatal management
  – Protection of viscera
  – Evaluation for associated anomalies
    • Genetic eval, Echo, hypoglycemia
  – Closure abdominal wall –
    • Primary or staged operations

• Outcomes
  – Related to morbidity of associated anomalies
  – When isolated defect – outcome related to size of defect
Omphalocele - spectrum

Hernia of the cord

Large Omphalocele

Pentalogy of Cantrell

Open diaphragm & pericardium

Giant omphalocele
Prenatal discussion

• Geneticist, Neonatologist, Cardiologist and Pediatric surgeon
  – Diagnosis can be virtually certain
    • MSAFP elevated, fetal U/S findings
  – Associated anomalies need to be determined
    • Role of amniocentesis, fetal echo
  – Elevated risk for IUFD, IUGR, preterm labor
    • Incidence of omphalocele in live and stillborns 1/300- 1/4000 i.e. high IUFD rate
  – Delivery at tertiary care center
    • Assure specialists available, possible C/S for giant omph.
Gastroschisis - the other AWD

- Full thickness defect in the abdominal wall
  - to the right of the umbilicus
- Embryology
  - Weakness of abdominal wall?
  - Consequence of involution of right umbilical vein?
Gastroschisis

• Newborn characteristics
  – Abd wall defect to right of umbilical cord
  – Variable size (<5 mm to 3 cm)
  – Variable amount intestine +/- stomach,

• Maternal characteristics
  – Young – under age 20 4x increased risk
  – Elevated MS AFP – better predictor than for omphalocele
  – Predisposing factors
    • Smokers – 1.6 fold risk
    • ? Use of vasoactive medicines – pseudoephedrine increased risk 3 fold
Prenatal issues

• Fetal diagnosis
  – Diagnosis can be certain
  – Fetal intestine adjacent to umbilical cord and external to abdominal wall

only pitfall = ruptured omphalocele - rare
Prenatal issues

• Prenatal discussion
  – Neonatologist / Pediatric surgeon
  – Focus on uni-system issues
    • Only associated anomaly is within same system = atresia ( <30% cases)
    • Significant impact only if SBS
  – Importance of delivery at tertiary care center
    • Optimal premature infant care

• Prenatal management
  Monitor growth
  Monitor condition of intestine
Fetal U/S: gastroschisis

Marked small bowel dilatation

Dilation of intra-abdominal bowel suggests obstruction at fascial level
Gastroschisis

- Postnatal management –
  - Protection/coverage of eviscerated intestine
  - Gradual reduction/expansion of abdominal cavity (silo) vs. Primary closure
  - Prolonged parenteral nutritional support
  - Evaluation for atresia
    - (<30% of infants)
    - Remains a leading cause of Short bowel syndrome
Gastroschisis - outcomes

Survival approaches 100%
Hospitalization 4-8 weeks unless complicated by atresia/SBS
Prenatal issues

• Fetal intervention?
  – Defect life-threatening? – NO
  – Intervene to improve condition/function of bowel at birth?
    • Is exposure to amniotic fluid toxic?
    • Early delivery?
    • C/S delivery?
Fetal intervention for gastroschisis

– Alter place of delivery
  • Yes: deliver in center with neonatal/surgical access

– Alter mode of delivery
  • C/Section: promoted to “protect” the intestine and shorted hospital stay due to more rapid advance to enteral feeding
  • Evidence does not support these assertions
Fetal intervention for gastroschisis

– Alter timing of delivery
  • If amniotic fluid is caustic, early delivery makes sense
  • Prospective series at Brown:
    – No recommendations of early delivery
    – Parameters: age at closure, age at first and full feeds
    – Results: *No rationale for early delivery*
Gastroschisis

Gestational age (weeks)

Age at definitive closure

33 34 35 36 37 38 39 40

1 week

Gestational age (weeks)
Abdominal wall defects

• Prenatal counseling
  – Excellent overall prognosis in absence of associated defects
  – Often prolonged neonatal ICU stay
  – No long-term sequelae
Visceral abnormalities

• Intestinal obstructions
  – mechanical and functional
• Duplications
• Disorders of rotation
• Internal hernias
Visceral anomalies

• Prenatal diagnosis depends on:
  – Alterations in amniotic fluid volume
    • polyhydramnios
  – Alterations in appearance of intestine:
    • dilated, edematous, or smaller than expected
  – Alterations in content of intestine
    • Echogenic material
  – Alterations within abdominal cavity
    • Calcifications, ascites
Intestinal Obstruction

• Postnatal classification –
• Proximal
  – Esophageal atresia, duodenal atresia, proximal jejunal atresia
• Distal
  – Jejuno-ileal atresia, meconium ileus, colon atresia, imperforate anus, Hirschsprung’s
Intestinal Obstruction

• Is it possible/important to provide prenatal diagnosis?
  – Important anomalies
  – All amenable to correction
  – But….are there associated anomalies that would impact on survival?
Possible Prenatal U/S findings

- Esophageal atresia
- Duodenal atresia
- Jejunoileal atresia

Polyhydramnios with

- Small stomach
- Double Bubble
- Dilated loops of intestine
Possible Prenatal U/S findings

- Esophageal atresia
- Duodenal atresia
- Jejunoileal atresia

Look for other anomalies

- VACTERL association
  - Vertebral, anorectal, cardiac, tracheoesophageal, renal, limb

- Chromosomal
  - Cardiac
  - Other atresia

- Generally isolated to intestine
Distal intestinal obstruction

• Not as easily evident
  – May not have abnormal amniotic fluid volume
  – Dilation may be diffuse
• Content of intestine may be best clue
  – Higher density content = echogenic
  – Must consider anatomic and functional problems
• Examples:
  – Distal small bowel obstruction, colon atresia, Hirschsprung’s Disease, imperforate anus
• Prenatal Dx at this time is unreliable
Rotation and Fixation
Disorders of rotation

- Can these be identified prenatally?
Disorders of rotation

• Generally – no!

• Postnatal U/S Dx depends on:
  – Abnormal relation of SMA/SMV
  – Spiraling of bowel with volvulus

• Only if there are prenatal complications
  – Paucity of fluid filled intestine, echogenic content
Malrotation predisposes to volvulus – prenatal or postnatal
Abnormality in intestinal content

– “Echogenic bowel”

• Echogenicity = Brightness of fetal bowel with transducer frequency of 5MHz or less
• Term that applies in 2\textsuperscript{nd} trimester only
• 3 gradations
  – 1 close to normal
  – 2 about as bright as liver
  – 3 as bright as bone (iliac crest)
• Incidence – estimated 0.2-2\% in 2\textsuperscript{nd} trimester
Echogenic bowel

• In most cases this finding is TRANSIENT and has no adverse sequelae.
• It may be associated with:
  – Swallowed blood/maternal bleeding
  – Cystic fibrosis (meconium ileus variants)
  – Aneuploidy (13, 18, 21)
  – Infection (CMV, parvo)
  – GI obstruction, volvulus
  – IUGR, fetal alcohol syndrome, twin gestation
Echogenic bowel

– Association with aneuploidy

• In studies of pts at risk for aneuploidy incidence of this finding varies between 5.5% - 14%

• 75% of these pts had other suspicious findings on U/S, only 3% had this as an isolated finding

• Limited conclusions to be reached in normal risk population with this finding

• In 9067 pregnancies identified 56 pt with EB – 47 agreed to genetic counseling- 22 agreed to amnio 3 cases tri21 one case tri 18 one case cmv. 12 with adverse outcomes only 3 had EB as only finding
Echogenic bowel

- Association with cystic fibrosis
- About 3% will be diagnosed with CF
  - 10 studies of 309 fetuses w/ echogenic bowel
eleven (3.6%) had CF
  - Largest series 244 cases of EB – at least 2 cases
of CF = 20x expected incidence
  - Incidence of CF depends on ethnic background of
sample population
  - Other findings which enhance dx:
    - polyhydramnios, ascites, calcifications, other
structural anomalies
    - clinical postnatal correlate – thick, inspissated
meconium
Echogenic bowel

– If echogenic bowel resolves – outcome is usually normal
– In 182 pregnancies with EB – 13% resulted in IUFD, 5% IUGR and 5% required GI surgery.
– In the majority of cases – the isolated finding of echogenic bowel will have no clinical consequence.
Newborn diagnoses related to the abdomen

- Intra-abdominal masses
- Cystic and solid
- Fetal tumors
- Hepatobiliary anomalies
Fetal cystic abdominal masses

• Differential diagnosis often difficult
  – Ovarian cyst (female fetus)
  – Distended bladder? Urachal cyst
  – Choledochal cyst, liver cyst
  – Mesenteric cyst (rare)
  – Cystic kidney disease (see urology)
  – Adrenal cyst (neuroblastoma, hemorrhage, sequestration)
  – Teratoma
  – Neurenteric cyst (rare)
  – Rarely important to establish definitive dx!
    • **Standard: treatment after birth**
Ovarian cyst

- Ovary under maternal hormonal stimulation
- Cyst size may be large: >4-5 cm
- Risk of torsion related to size?
- Value of antenatal intervention?
  - Needle aspiration
  - Risk to fetus/mother outweighs benefit
  - Potential for spontaneous resolution after birth
Ovarian cyst

Potential for spontaneous resolution

Consider aspiration postnatally if concerned for torsion based on size
Fetal abdominal masses

- Adrenal masses
  - Location is subdiaphragmatic
  - Adrenal hemorrhage
  - Neuroblastoma
  - Intra-abdominal pulmonary sequestration

- Prenatal treatment not indicated
Sacrococcygeal teratoma

- These tumors can present primarily within the abdominal cavity as cystic or solid lesions
- ...Or they can be exophytic lesions
- ...Or anywhere in between!
Sacrococcygeal teratoma

- Most often benign
  - May contain immature elements: evolve to malignant
  - Good prognosis if operated before 2 months of age
- If early prenatal diagnosis & large tumor:
  - Risk of hydrops and fetal demise
  - Risk to mother ("mirror" syndrome)
- Intervention to save mother (and fetus):
  - Attempt at open fetal surgery – few successful cases
  - Radiofrequency ablation technique- catastrophic
  - Only fetal operation for mother’s sake? or termination
Prenatal diagnosis of abdominal abnormalities

- Gastroschisis and Omphalocele
  - Diagnosis and implications
- Proximal intestinal obstruction/ Malrotation
  - Diagnosis and outcomes
- Echogenic bowel-
  - Considerations in prenatal counseling
Prenatal diagnosis of abdominal abnormalities

• screen for associated anomalies
  – chromosomal disorders or potentially life-threatening structural disorders

• prepare parents for postnatal event
  – Counsel regarding length of gestation, mode and site of delivery

• rarely require fetal intervention
Questions?