OMPHALOCELE & OTHER CONGENITAL ABDOMINAL WALL DEFECTS

AN INTERESTING CASE FROM KINGS COUNTY HOSPITAL CENTER
MADHURI RAO, MD, PGY-3
CASE PRESENTATION

- Baby – male, 4000gm, 39 week gestation
- Mother – 26 yo G4P2 with inadequate prenatal care at an outside institute
- Prenatal sonogram suggestive of omphalocele. s/p amniocentesis- normal karyotyping
- Normal vaginal delivery
SOON AFTER BIRTH…

- No acute distress
- Macroglossia, macrosomia, ear creases
- 3.5 cm defect in abdominal wall at the umbilicus with bowel covered by a sac
- Labs – Hypoglycemic FSG 30 mg/dL
IMMEDIATE MANAGEMENT

- Wet gauze over omphalocele by NICU team and pediatric surgery consulted
- Baby placed in warmer
- OGT inserted
- Rectal exam – meconium evacuated
- Omphalocele reduced at bedside and dressing placed over it
- NPO
- IVF and IV Abx started
SURGICAL MANAGEMENT

PRESURGICAL TESTING AND OPTIMIZATION

- Echocardiogram – PDA (1.7mm), PFO
  Rpt. Echo prior to discharge- normal
- Renal U/S – left hydronephrosis

Repair in the operating room planned for DOL 2
SURGICAL MANAGEMENT (Continued)

OPERATIVE DETAILS

 General anesthesia
 Excision of sac
 Ligation of umbilical vessels
 Dissection of fascia
 Closure of fascia with multiple figure of 8s with 3-0 PDS suture
 Closure of skin with circumferential inverted 4-0 PDS suture
POST OPERATIVE COURSE

- POD 1 – Extubated
- POD 2
  - BMs +ve
  - PO feeds
- In house work up of associated abnormalities (r/o Beckwith-Wiedemann syndrome)
  - Syphilis –ve
  - Chromosome karyotype normal
- Discharged home on POD 8 (DOL 10)
## Congenital Abdominal Wall Defects

<table>
<thead>
<tr>
<th>Defect</th>
<th>Site</th>
<th>Contents</th>
<th>Associated Anomalies</th>
</tr>
</thead>
<tbody>
<tr>
<td>Omphalocele – lateral fold</td>
<td>Umbilicus</td>
<td>Liver, intestine, spleen, gonad</td>
<td>Chromosomal, cardiac</td>
</tr>
<tr>
<td>Pentalogy of Cantrell-cephalic fold</td>
<td>Superior umbilicus</td>
<td>Liver, intestine</td>
<td>Cardiac, sternal cleft, pericardial defect, central tendon diaphragm defect</td>
</tr>
<tr>
<td>Cloacal extrophy-caudal fold</td>
<td>Inferior umbilicus</td>
<td>Intestine</td>
<td>Bladder extrophy, imperforate anus, epispadias</td>
</tr>
<tr>
<td>Umbilical cord hernia</td>
<td>Umbilicus</td>
<td>Intestine</td>
<td>Uncommon</td>
</tr>
<tr>
<td>Gastrochisis</td>
<td>Right umbilicus</td>
<td>Intestine</td>
<td>Intestinal atresia</td>
</tr>
<tr>
<td>Ectopia cordis thoracis</td>
<td>Midline sternum</td>
<td>Heart</td>
<td>Cardiac</td>
</tr>
</tbody>
</table>

HISTORY

- Abdominal wall defects – first century AD by Aulus Cornelius Celsus, fifth century by Paulus Aegineta

- Omphalocele
  - 16th century by Ambrose Par
  - First successful repair by Hey in 1802
  - Stage reduction with prosthetic material by Schuster in 1967

- Gastroschisis
  - 16th century by Lycosthenes
  - Confused with ruptured omphalocele
  - Moore and Stokes described it as a separate entity
  - First successful repair by Visick in 1873
INCIDENCE

- 1 in 2000 live births
- Omphalocele – 1-2.5 per 5000 births, M > F
- Gastroschisis – 1 in 4000 births
- Increased detection with increasing antenatal U/S, ms AFP testing
EMBRYOLOGY

- **Week 3-5**
  Folding of embryonic disk in 4 planes converging at the umbilicus. Rapid growth of intestines.

- **Week 6**
  Physiological midgut herniation

- **Week 10-12**
  Reduction of PMH into abdomen for rotation and fixation
# Embryogenesis of Defects

**Omphalocele**
- Failure of extra embryonic gut to return into abdominal cavity – simple omphalocele
- Failure of lateral body folds to migrate medially – larger defect with liver
- Covered by 2 layer amniotic peritoneal membrane
- Defect at the umbilicus
- Occurs early in embryogenesis

**Gastrochisis**
- In-utero vascular accident
  - Right umbilical vein involution → necrosis in abdominal wall → defect
  - Premature right vitelline artery involution → weakening of abdominal wall → rupture a/w intestinal atresia
- Not covered by membranes
- Defect to the right of the umbilicus
- Intestinal damage later in pregnancy
# Clinical Features

## Omphalocele vs. Gastroschisis

<table>
<thead>
<tr>
<th></th>
<th>Omphalocele</th>
<th>Gastroschisis</th>
</tr>
</thead>
<tbody>
<tr>
<td>Full term</td>
<td>Full term</td>
<td>Likely premature</td>
</tr>
<tr>
<td>Central abdominal</td>
<td>Central abdominal wall defect</td>
<td>Defect to right of umbilicus</td>
</tr>
<tr>
<td>wall defect</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Sac</td>
<td>Sac</td>
<td>No sac</td>
</tr>
<tr>
<td>Contents</td>
<td>Contains midgut, liver and other organs</td>
<td>Contains midgut and sometimes gonad</td>
</tr>
<tr>
<td>Associated conditions</td>
<td>• Cardiac (45%) – VSD most common</td>
<td>Associated conditions</td>
</tr>
<tr>
<td></td>
<td>• Chromosomal (40%)</td>
<td>• Intestinal atresia</td>
</tr>
<tr>
<td></td>
<td>• Cryptorchidism (30%)</td>
<td>• Small for gestational age</td>
</tr>
<tr>
<td></td>
<td>• Macrosomia</td>
<td>• GERD (16%)</td>
</tr>
<tr>
<td></td>
<td>• GERD</td>
<td>• Cryptorchidism (15%)</td>
</tr>
<tr>
<td></td>
<td>• Musculoskeletal</td>
<td></td>
</tr>
<tr>
<td></td>
<td>• Neural tube defects</td>
<td></td>
</tr>
</tbody>
</table>

**References:**

[www.downstatesurgery.org](http://www.downstatesurgery.org)
# Omphalocele - Associated Syndromes

<table>
<thead>
<tr>
<th>Syndrome</th>
<th>Features</th>
</tr>
</thead>
<tbody>
<tr>
<td>Beckwith-Wiedemann</td>
<td>Most common Omphalocele, macroglossia, hypoglycemia, nevus flammeus</td>
</tr>
<tr>
<td>OEIS</td>
<td>Omphalocele, Exstrophy, Imperforate anus, Spinal anomalies</td>
</tr>
<tr>
<td>Gershoni-Baruch</td>
<td>Omphalocele, diaphragmatic hernia, cardiovascular anomalies, radial ray defects</td>
</tr>
<tr>
<td>Donnai-Barrow</td>
<td>Diaphragmatic hernia, exomphalos, absent corpus callosum, hypertelorism, myopia, sensorineural deafness (autosomal recessive)</td>
</tr>
<tr>
<td>Fryns</td>
<td>Diaphragmatic hernia, coarse facies, acral hypoplasia, omphalocele</td>
</tr>
<tr>
<td>Trisomy 13, 18, 21</td>
<td></td>
</tr>
</tbody>
</table>
MANAGEMENT – PRENATAL

- Antenatal U/S
  - Omphalocele – Sac +ve, Liver +/-, sensitivity = 75%
  - Gastrochisis – Sac -ve, Liver -ve, sensitivity = 83%
  - Look for other associated anomalies

- Amniotic fluid – elevated AFP, AChE
- Maternal serum – elevated AFP

Helps prenatal counseling and planning
MANAGEMENT – DELIVERY

- Plan for delivery at a tertiary care center
- Involvement of obstetric, NICU and pediatric surgical team
- No benefit of C-section over vaginal delivery * (practiced in some cases of giant omphalocele because of potential liver injury or sac rupture)
- Plan for repair ASAP
  - Worsening bowel edema in gastroschisis
  - Enlarging liver with omphalocele

MANAGEMENT – NEONATAL

Principles of surgical management

- Safely reduce evisceration
- Cosmetic closure of defect
- Identify and treat associated anomalies
- Nutritional support
- Management of abdominal, wound, or bowel complications
Immediate care

- OGT/NGT
- Rectal exam
- Maintain body temperature
- Ventilatory support
- Antibiotics

**Omphalocele**
- Dressing to minimize heat loss
- Maintenance IV fluids
- Look for associated anomalies

**Gastroschisis**
- Heat preservation with bowel bag
- Resuscitative IV fluids
- Plan for immediate OR
Primary vs. staged vs. delayed closure based on size and condition of the neonate

**PRIMARY CLOSURE**
- Reduce abdominal contents with intact sac
- Circumferential dissection to rectus
- Excise, invert and ligate sac with vessels and urachus
- Fascial closure with mattress suture
- Purse string suture for skin to recreate umbilicus
Larger defects – techniques to facilitate primary fascial closure

- Creation of flaps
- Component separation
- Bioabsorbable mesh
- Tissue expanders
STAGED CLOSURE

- Sac excised and silo bag sewn to fascia
- Can use sequential ligation of a thick sac
- Slow reduction over 7-8 days
- Delayed fascial closure in the OR
DELAYED CLOSURE WITH ESCHAROTIC THERAPY

- Severe congenital anomalies preventing closure
- Gradual epithelialization of the omphalocele sac – Silvadene, Povidone-iodine, Mercurochrome (not used)
- Repair of formed ventral hernia after epithelialization
MANAGEMENT – NEONATAL (cont.)

OPERATIVE MANAGEMENT – GASTROSCCHISIS

- Primary reduction with operative closure of fascia
- Silo placement, serial reductions and delayed fascial closures
- Primary or delayed reduction without fascial closure
- ‘Plastic’ sutureless closure

Sutureless closure
SHORT TERM

- Ventilatory support
- Compartment syndrome from tight closure
  - Monitor ventilatory pressures, IAH, urine output and distal perfusion
- Delayed bowel function
  - Need for TPN/PPN
- NEC – risk factors of IAH, prematurity, birth weight, delayed feeding
- Wound failure
- Infection and sepsis
DELAYED

- Ventral hernia (repaired after age 1 year)
- Need for umbilicoplasty
- Adhesive SBO – 25% of gastroschisis and 13% of omphalocele repairs

PROGNOSIS

OUTCOME MEASURES

- Survival
  - Associated anomalies
  - Intestinal length and function
  - Viscero-abdominal disproportion (influences risk of complications)
- Nutritional autonomy
- Cosmesis
PROGNOSIS (cont.)

- **Omphalocele**
  - 94% overall survival
  - Mortality from cardiac and chromosomal anomalies
  - Long term problems including GERD, pulmonary insufficiency, failure to thrive in 60% of infants
  

- **Gastroschisis**
  - 99% overall survival
  - Prognosis depends on intestinal atresia and degree of irreversible bowel damage
LITERATURE REVIEW

Review of the evidence on the closure of abdominal wall defects

Management of omphalocele

- Based on case reports and small case series
- Results of a retrospective review from Kings College Hospital, London (1995-2002)
  - 35 infants: 11 (minor) – primary closure, 13 (major) – primary closure, 11 (major) – staged closure
  - Infants with staged closure needed significantly longer periods of ventilation, longer hospital stay and more delayed bowel function
LITERATURE REVIEW

Reports on techniques for repair of large omphalocele

<table>
<thead>
<tr>
<th>Method</th>
<th>Reference, year</th>
<th>No. of patients</th>
<th>Survival</th>
<th>Complications</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Alaish 2006, JPS 41:e37 [57]</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>VAC</td>
<td>Kilbride 2006, JPS 41:214 [58]</td>
<td>3</td>
<td>3</td>
<td>None</td>
</tr>
<tr>
<td>Tissue expanders</td>
<td>Martin 2009, JPS 44:178 [59]</td>
<td>6</td>
<td>6</td>
<td>Infection, extrusion in one</td>
</tr>
<tr>
<td></td>
<td>Foglia 2006, JPS 41:704 [74]</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>De Ugarte 2004, JPS 39:613 [75]</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Bax 1993, JPS 28:1181 [76]</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Method of epithelialization</th>
<th>Reference, year</th>
<th>Number of patients</th>
<th>Number of patients who survived</th>
<th>Complications</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mercurochrome</td>
<td>Used in 1960–1980s primarily [2]</td>
<td>n/a</td>
<td>n/a</td>
<td>Mercury poisoning, not used currently</td>
</tr>
<tr>
<td>Povidone-iodine</td>
<td>Whitehouse 2010, JPS [67]</td>
<td>6</td>
<td>5 (CHD)</td>
<td>Transient increase TSH, not clinically significant</td>
</tr>
<tr>
<td>Silver sulfadiazine/</td>
<td>Lee 2006, JPS 41:1846 [63]</td>
<td>22</td>
<td>19</td>
<td>Two ruptured sacs, one bleeding sac</td>
</tr>
</tbody>
</table>
LITERATURE REVIEW

Management of gastroschisis

- Grade C evidence – no survival difference between immediate and delayed closure
- Routine use of a SILASTIC spring-loaded silo for infants with gastroschisis: a multicenter randomized controlled trial.


- 27 infants – primary closure, 27 infants – spring loaded silo and elective closure
- No significant difference in LOS, time on TPN, sepsis, NEC
Management of gastroschisis

- Some evidence for using IAP to guide closure*
  - Limit of 20mmHg for primary closure
  - Also used to guide sedation, paralysis and silo reduction


SUMMARY

- Spectrum of congenital abdominal wall defects (Omphalocele and gastroschisis most common)
- Embryology and embryogenesis
  - Omphalocele – failure of migration of lateral fold
  - Gastroschisis – various theories (intrauterine ischemic event)
- Difference in clinical features between omphalocele and gastroschisis
  - Omphalocele – looks better but associated with more lethal anomalies
  - Gastroschisis – looks worse but has a better prognosis
SUMMARY (cont.)

- **Management**
  - Prenatal diagnosis may influence timing and location of delivery
  - Neonatal resuscitation focused on normothermia, fluid resuscitation, GI decompression, and ruling out associated anomalies
  - Surgical management – primary vs. staged closure
    - Goal is to minimize damage to bowel and obtain cosmetically acceptable closure

- **Prognosis**
  - Omphalocele – associated anomalies
  - Gastroschisis – degree of bowel injury
REFERENCES

- Contemporary postnatal surgical management strategies for congenital abdominal wall defects
- Neonatal abdominal wall defects
  Christison-Lagay ER, et al. Division of General and Thoracic Surgery, Hospital for Sick Children, 555 University Ave, Toronto, Ontario M5G 1X8, Canada, Department of Pediatric Surgery, Massachusetts General Hospital for Children, Boston, MA, USA
- Review of the evidence on the closure of abdominal wall defects
- Etiology of intestinal damage in gastroschisis. II. Timing and reversibility of histologic changes, mucosal function, and contractility.
THANK YOU