RADY 403 Case Presentation

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UNC SCHOOL OF MEDICINE
Radiology

Focused patient history and workup

- Baby boy J is a 35w6d male born to a G3P1112 via SVD. Pregnancy was complicated by AMA and pre-eclampsia with severe features requiring magnesium, labetalol, and nifedipine. Pregnancy was also complicated by GDMA2. Baby boy received prenatal diagnoses of Trisomy 13, Dandy Walker malformation, an abdominal wall defect, and ambiguous genitalia.

[1]

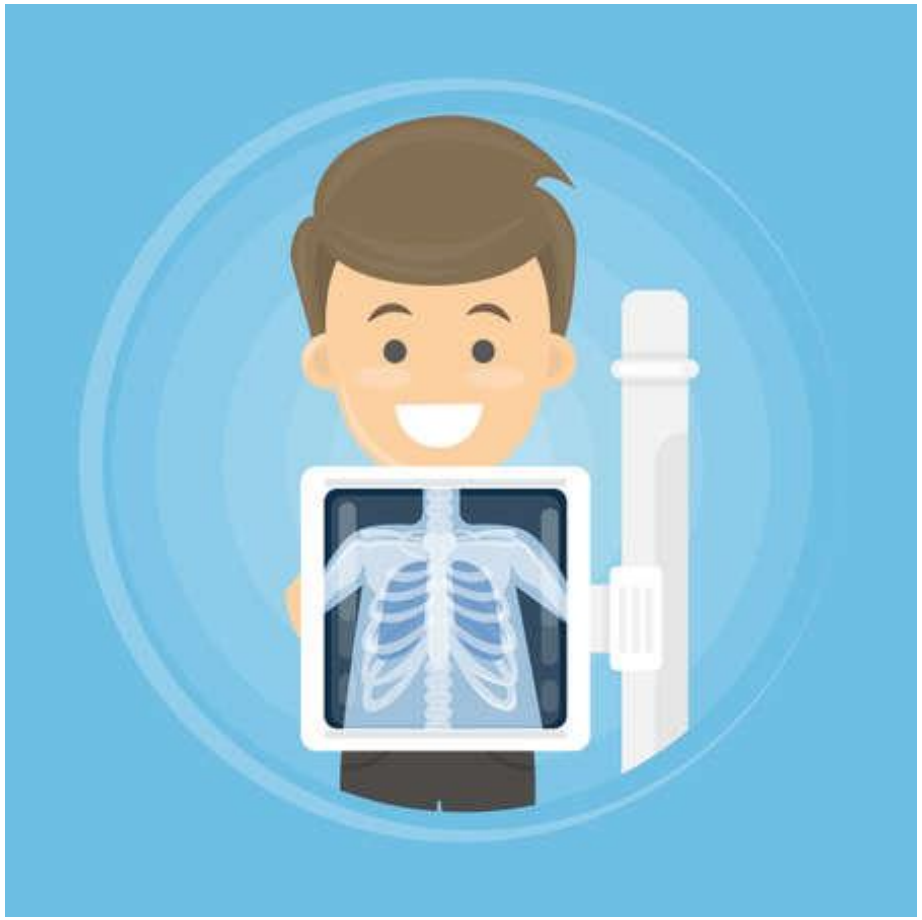
- On exam after birth, he had a cleft palate, extra-abdominal loops of bowel, 6th digits on both hands and feet, and non-palpable testes.
- Imaging:
  - Echocardiogram, US Scrotum, US Neonatal Head, US Renal Complete, MRI Brain w/o contrast
- Consults: Genetics, ENT, Cardiology, Peds Surgery, Dietician, others

List of pertinent imaging studies

1) X-Ray Abdomen Portable

2) X-Ray Abdomen (2 Views + 1 View)

3) Additional Diagnostic Imaging performed, discussed later



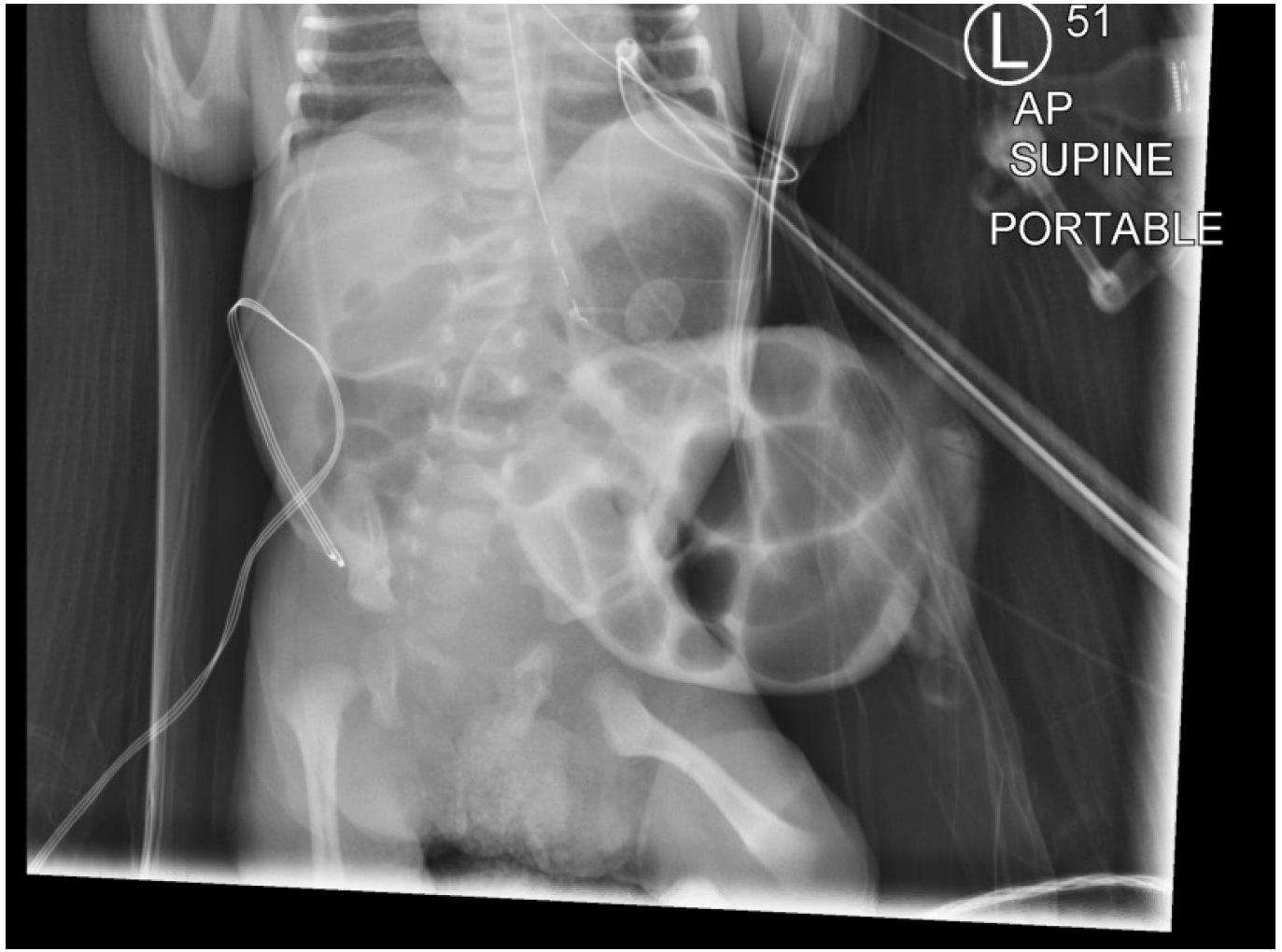
[2]

X-Ray Abdomen Portable

Indication: Vomiting (1 days old)

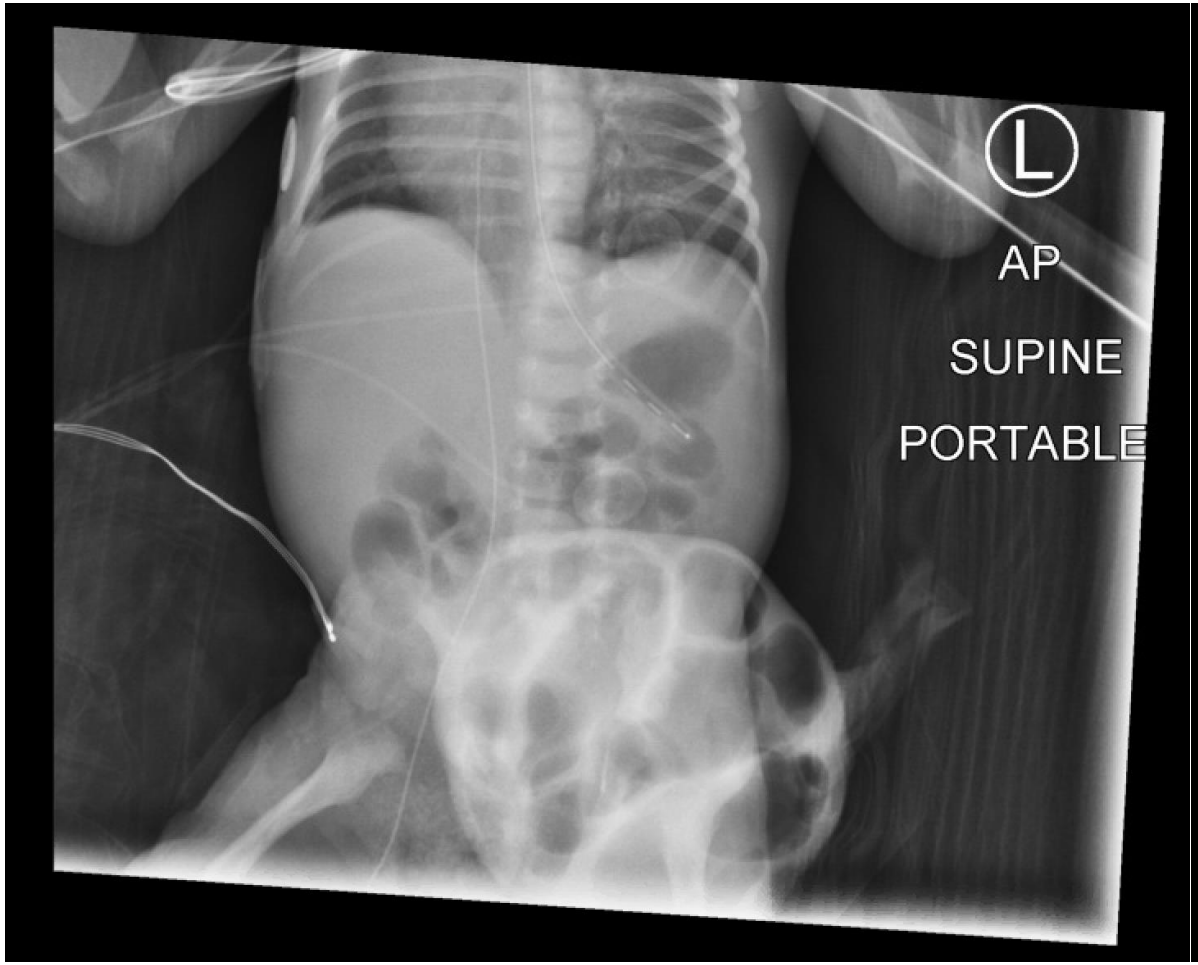
Findings:

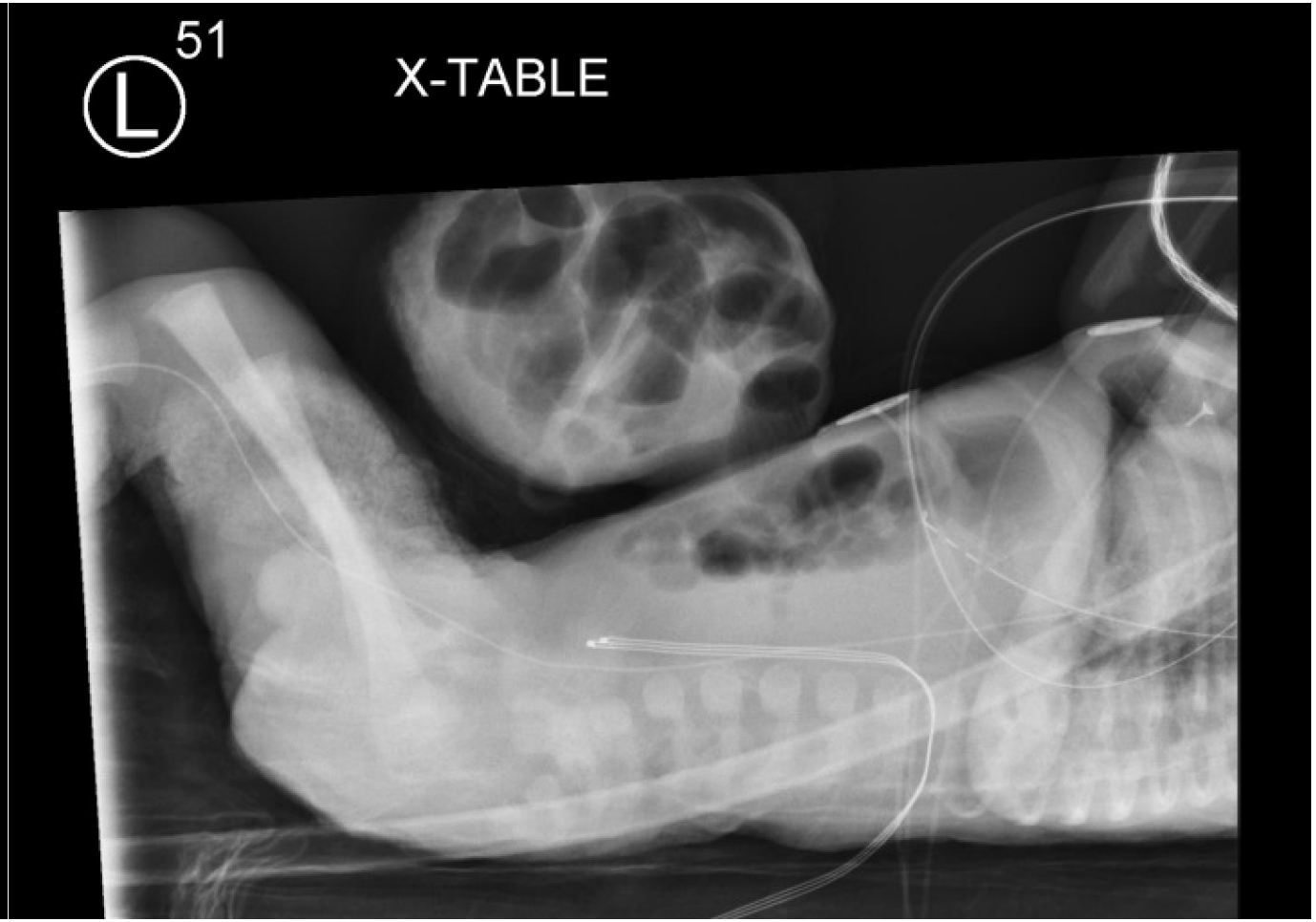
- Radiographic findings consistent with omphalocele with gas and distended bowel loops throughout.
- Enteric tube tip in the left upper quadrant, presumably within the stomach, with proximal side hole at the GE junction. Gaseous distention of the stomach.
- No definite findings to suggest free air.
- No pneumatosis or portal venous gas seen.



X-Ray Abdomen 2 View

Indication: Line Check (3 days old)





Treatment Plan

- Findings:
  - Omphalocele with sac intact.
- Plan:
  - Keep omphalocele wrapped with gauze and moist with saline inside a bag
  - Surgical team's plan was to allow his omphalocele to epithelialize, giving time for his abdominal cavity to grow prior possible reduction closer to 1 year of age if desired by family.
  - Await consults by other medical specialties

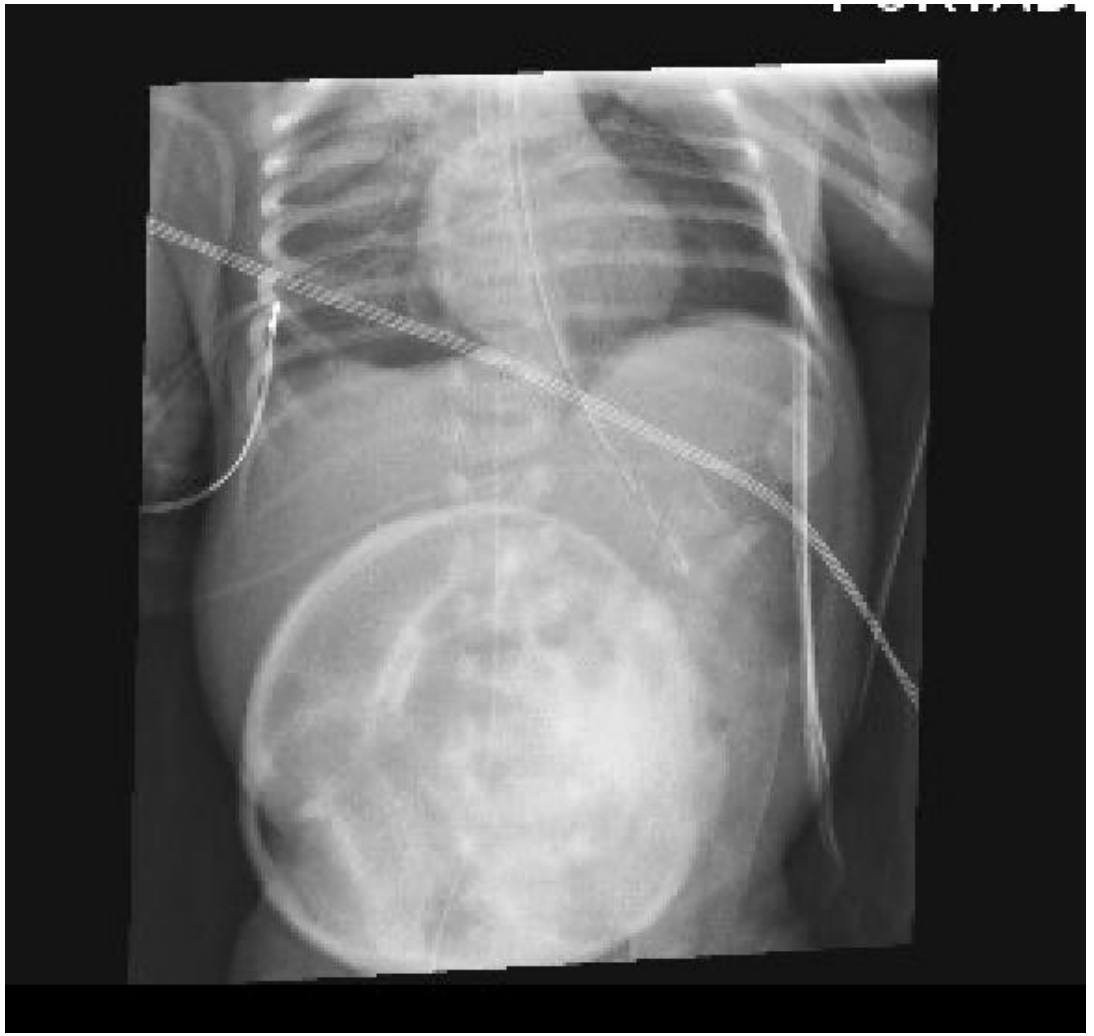
[3]

X-Ray Abdomen 1 View (Supine)

Clinical Progression:
Baby boy J, now 5 days old, developed concern for obstruction following bilious vomiting, considered likely to obstruction in omphalocele. Imaging was ordered to evaluate bowel gas pattern

X-Ray Abdomen (1 View)

Findings: Omphalocele containing multiple gas distended loops of bowel, similar to prior exam.



Additional Imaging Performed

- **Echocardiogram**: PDA, small L->R atrial shunt, mildly dilated RV, normal RV systolic function, mildly diminished LV systolic function
- **Renal US**: echogenic kidneys (consistent with prematurity) and no evidence of urinary tract dilation.
- **Head US**: no evidence of IVH, nonspecific right peri-atrial white matter, which could reflect ischemic white matter injury, and bilateral lenticulostriate vasculopathy (nonspecific finding that can be seen in the setting of trisomy 13)
- **MRI Brain w/o Contrast**: Hemorrhage, white matter lesions, ocular defects, others

Patient Outcome

- Omphalocele kept wrapped with saline-moistened gauze and placed inside bag to prevent insensible water loss.
  - Pediatric Surgery recommended correction at 9-12 months of life, if family chooses.

- Omphalocele developed defect concerning for fistula with meconium output.
  - Fistula was treated as ostomy

- Height of pandemic: mom wanted father to meet baby alive and breathing before discharge
- Intubated throughout hospital stay, multiple failed extubations
- Outcome was considered poor. Parents decided to care for patient at home.

- **Baby boy J discharged on CPAP to home hospice**

[4]

Prenatal Screening: ACR Appropriateness Criteria

New 2020

American College of Radiology
ACR Appropriateness Criteria®
Second and Third Trimester Screening for Fetal Anomaly

Variant 1: **Second and third trimester screening for fetal anomaly. Low-risk pregnancy. Initial imaging.**

Procedure	Appropriateness Category	Relative Radiation Level
US pregnant uterus transabdominal anatomy scan	Usually Appropriate	O
US pregnant uterus transabdominal detailed scan	Usually Not Appropriate	O
US echocardiography fetal	Usually Not Appropriate	O
MRI fetal without and with IV contrast	Usually Not Appropriate	O
MRI fetal without IV contrast	Usually Not Appropriate	O

Variant 2: **Second and third trimester screening for fetal anomaly. High-risk pregnancy. Initial imaging.**

Procedure	Appropriateness Category	Relative Radiation Level
US pregnant uterus transabdominal detailed scan	Usually Appropriate	O
US echocardiography fetal	May Be Appropriate	O
MRI fetal without IV contrast	May Be Appropriate (Disagreement)	O
US pregnant uterus transabdominal anatomy scan	May Be Appropriate (Disagreement)	O
MRI fetal without and with IV contrast	Usually Not Appropriate	O

Variant 3: **Second and third trimester screening for abnormal finding on ultrasound: soft markers. Next imaging study.**

Procedure	Appropriateness Category	Relative Radiation Level
US pregnant uterus transabdominal detailed scan	Usually Appropriate	O
US pregnant uterus transabdominal follow-up	Usually Appropriate	O
US echocardiography fetal	May Be Appropriate	O
MRI fetal without IV contrast	Usually Not Appropriate	O
MRI fetal without and with IV contrast	Usually Not Appropriate	O

Variant 4: **Second and third trimester screening for abnormal finding on ultrasound: major anomalies. Next imaging study.**

Procedure	Appropriateness Category	Relative Radiation Level
US pregnant uterus transabdominal detailed scan	Usually Appropriate	O
MRI fetal without IV contrast	Usually Appropriate	O
US echocardiography fetal	Usually Appropriate	O
US pregnant uterus transabdominal follow-up	Usually Appropriate	O
MRI fetal without and with IV contrast	Usually Not Appropriate	O

[5]

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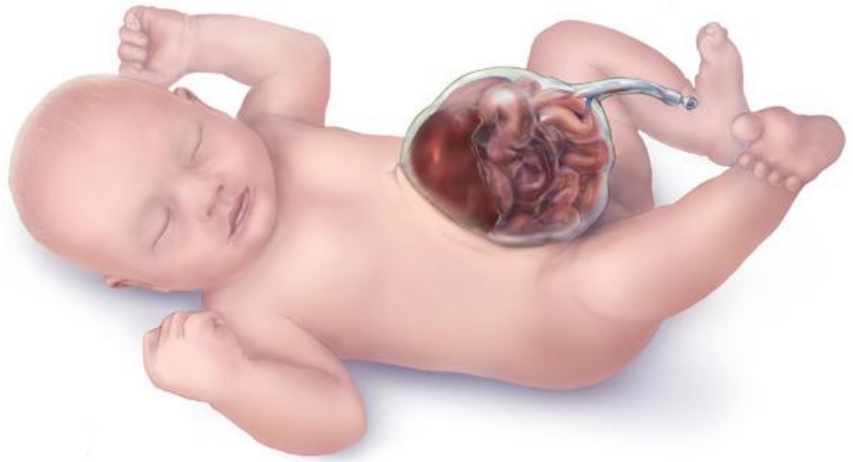
Omphalocele: What is it?

- Prevalence: 2 in 100,000 births [7]
- Majority are diagnosed prenatally

- Congenital midline abdominal wall defect comprised of herniation of **internal organs** and **membranous sac** at th umbilicus (most common)
  - Can be subdivided into liver (80%) and non-liver containing
  - Can be small or large
- Sac comprised of amnion, Wharton's jelly (from umbilical cord), and peritoneum

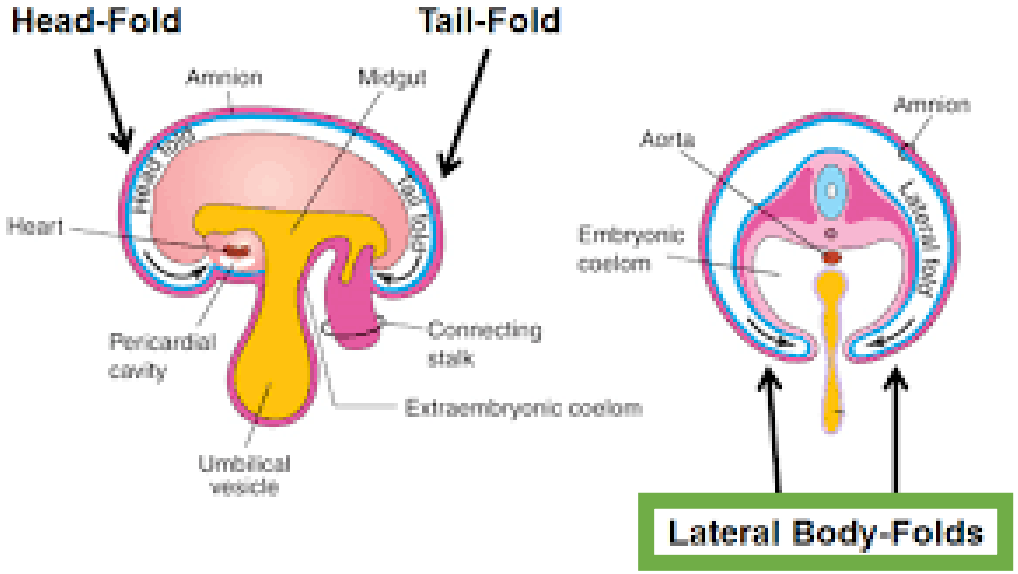


[6]

- Majority of infants (~55%) have **concurrent congenital and/or genetic abnormalities** (Beckwith-Wiedemann Syndrome, Trisomy's, etc.), discussed separately
  - Pushes mortality anywhere between 80-100% (~10% if in isolation)

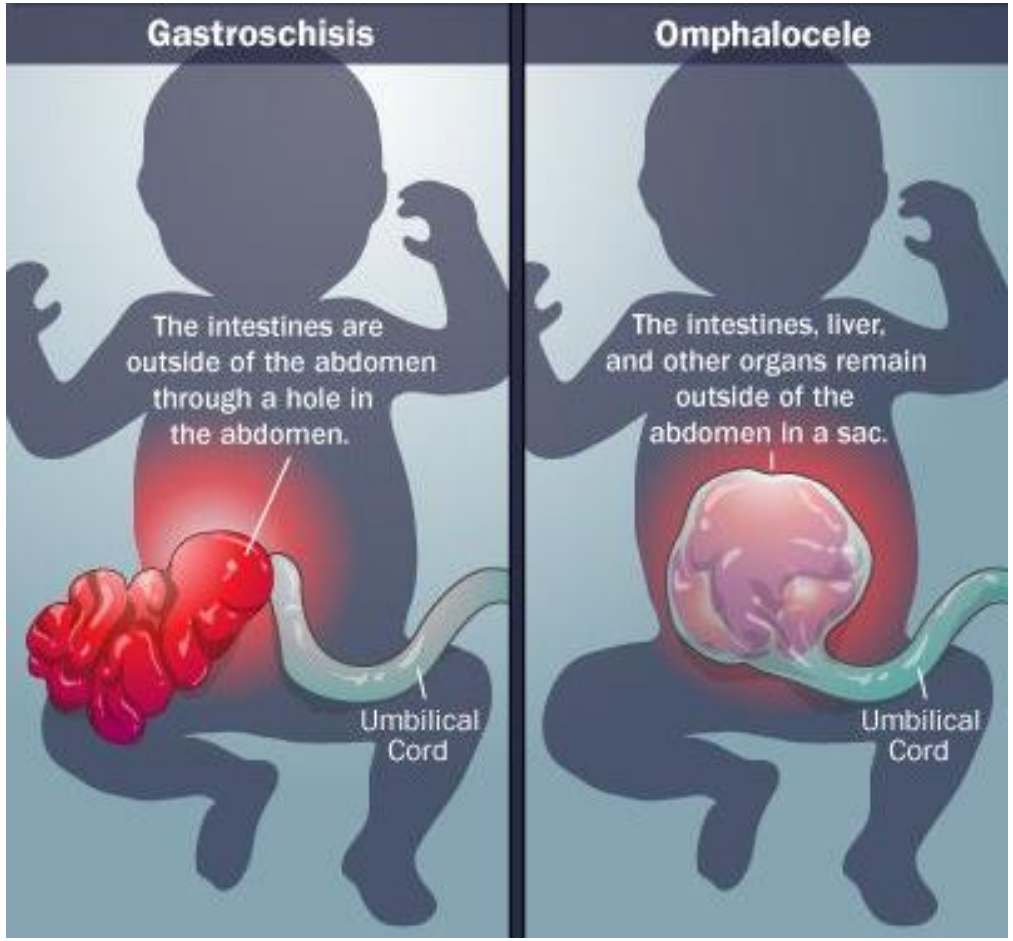
Pathogenesis

- During normal development, at approximately week 6, bowel contents herniate through the umbilicus into surrounding coelom
  - Umbilicus formed at union of lateral folds of embryologic disc
  - Bowel undergoes rotation and returns to abdominal cavity
- Several theories regarding error:
  - **Failed Rotation** and re-entry [9]
  - **Failed Union** of lateral embryologic folds, which form the umbilical ring [9]



[8]

Omphalocele vs. Gastroschisis



[10]

Gastroschisis	Omphalocele
Location: right side	Location: center
Content not covered by membranes	Presence of peritoneum-amniotic membrane
No umbilical cord	Umbilical cord inserted in caudal area of the hernial sac
Content: intestine (100%), colon, bladder, gonads (occasionally)	Content: intestine, liver (in most cases); spleen, colon, bladder (occasionally)
Rarely associated with other congenital anomalies (15%)	Frequently associated with other congenital anomalies (40-80%)

[11]

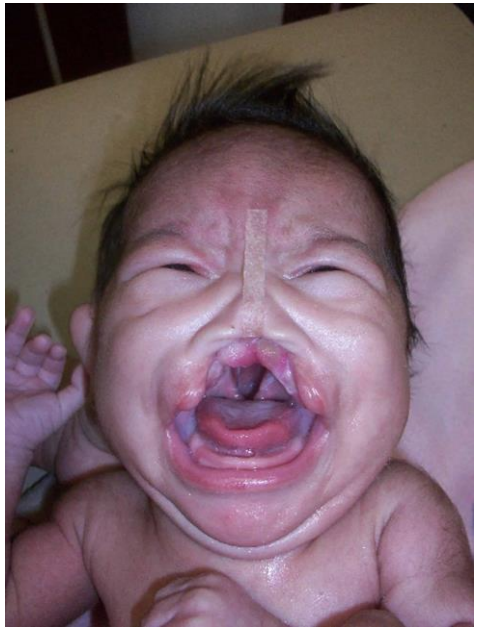
Associated Abnormalities

- Genetic
  - Omphaloceles with only small bowel associated with aneuploidy, particularly ***Trisomy 13 (Patau Syndrome)*** (our patient) and ***Trisomy 18 (Edwards Syndrome)*** [12]

- Syndromes
  - **Beckwith-Wiedemann** – omphalocele, macroglossia, gigantism
  - **Turner Syndrome**

- Structural
  - **GI Abnormalities** (e.g., Malrotation, Intestinal atresia)
  - **Cardiac anomalies** (e.g., VSD)
  - **Genitourinary** (e.g., Renal agenesis, polycystic kidney)
  - **Cleft lip/palate**
    - Workup involves many radiographic studies, as in our patient

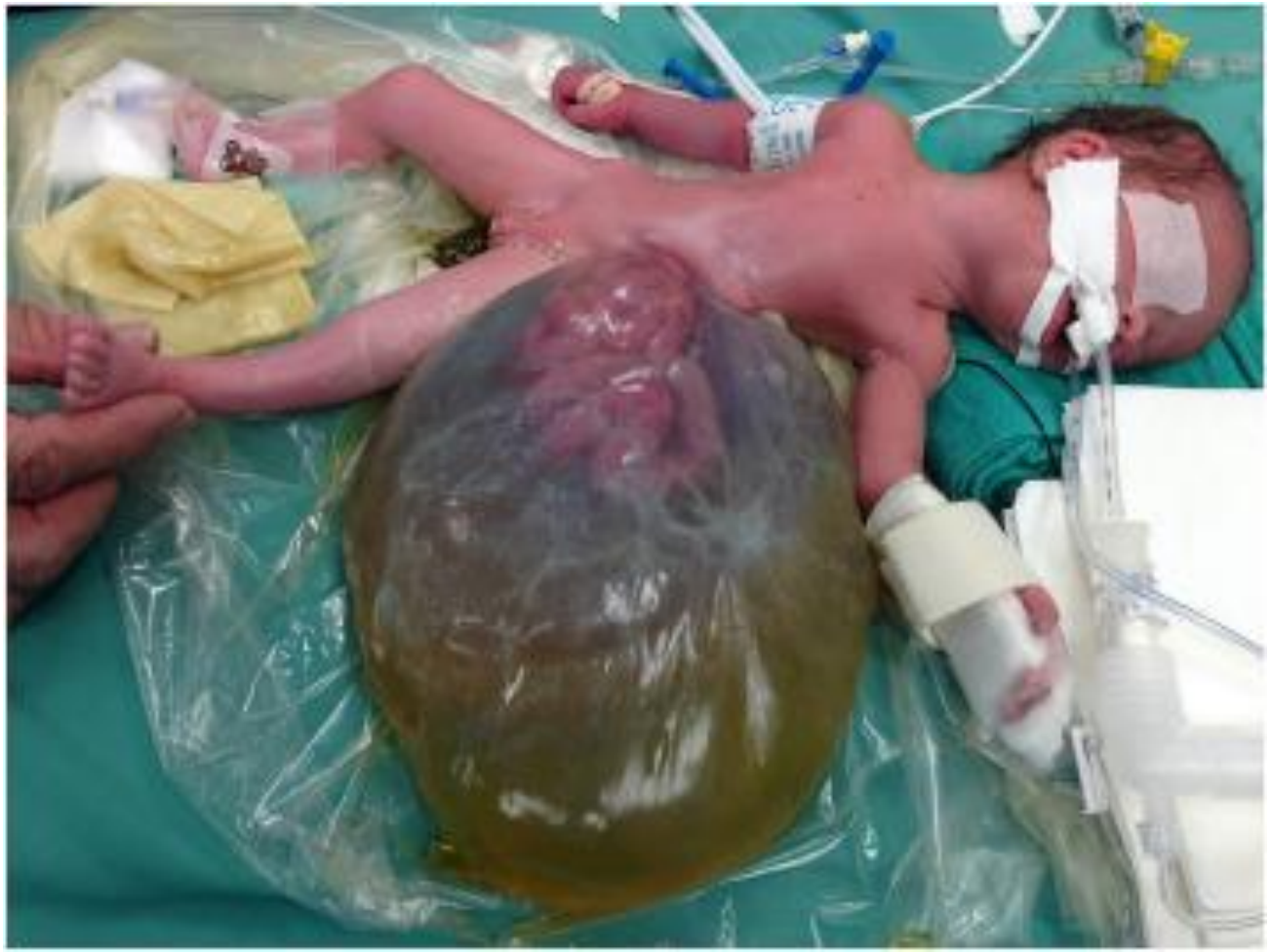
[13]



[14]

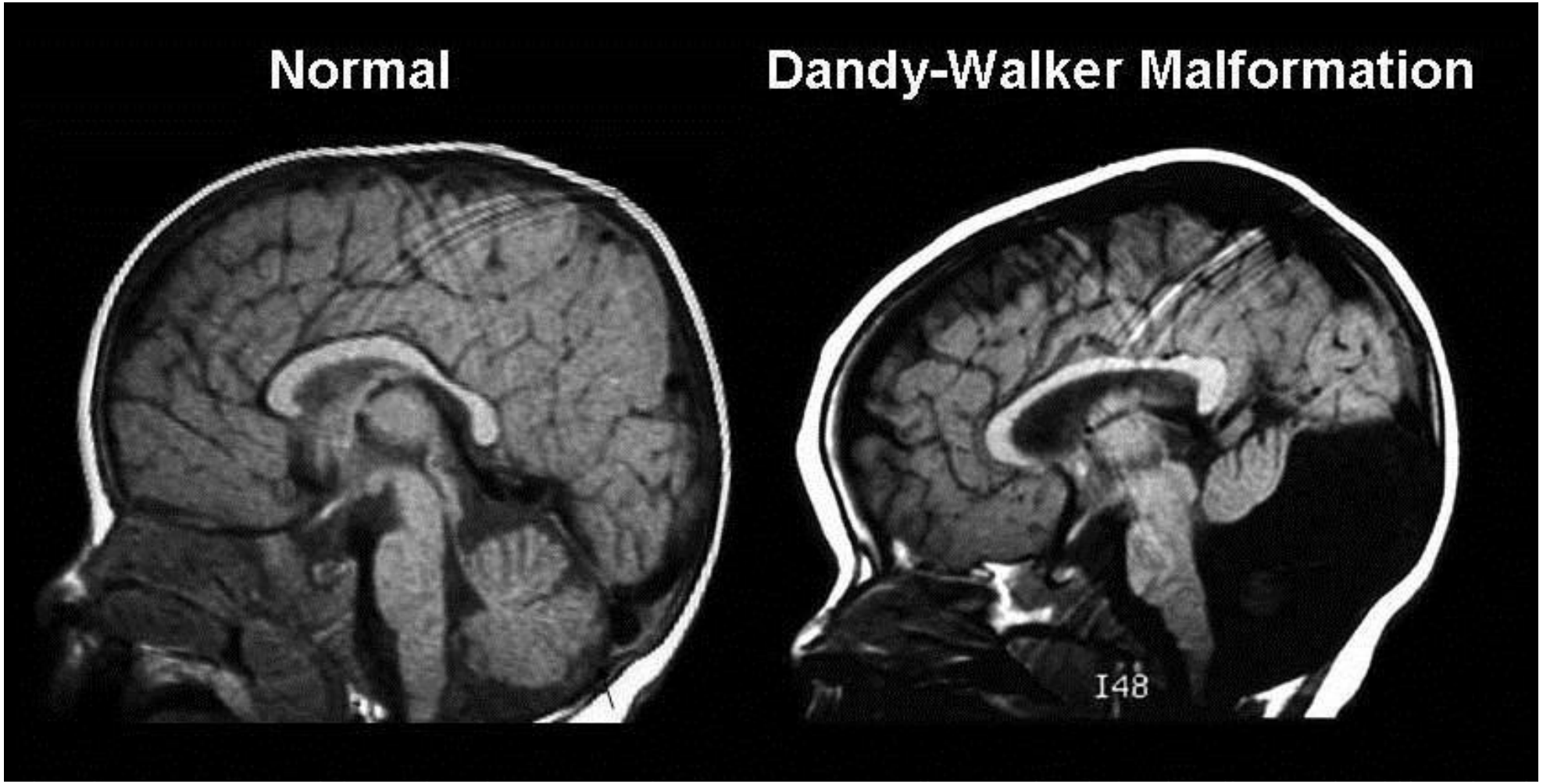
Giant Omphalocele

- Rare condition (incidence is 1 in 6,000 live births) [16]
- No consensus definition, but many consider **>5 cm** in diameter and contains all or most of the liver [17]
- Makes both delivering the fetus and surgical management quite complex
- Associated with **significant neurodevelopmental delay** [17]



[15]

Dandy-Walker Malformation



[18]

Hypoplasia or agenesis of cerebellar vermis

When to Image with Omphalocele?

- In general, **no direct postnatal imaging** of omphalocele is required!
- Imaging indications include, among others:
  - **Signs of obstruction**
    - Ex: bilious vomiting, abdominal distention, inability to void
  - **Signs of infection**
    - Septic criteria met, looking for source (e.g., bowel perforation, free air)
- What modality?
  - **Abdominal radiograph** is a good first-line option (CT backup, consider US (though not well-studied), MRI case reports (not as helpful))
- **Screening/Workup imaging** (Renal US, Scrotal US, Head MRI, Echo, etc.) **should be performed** in patients with omphalocele given risk of other genetic and structural abnormalities

[19]

Wrap Up

- Omphalocele is a congenital midline abdominal wall defect comprised of herniation of internal organs and membranous sac at the umbilicus
- Most cases are diagnosed prenatally with transabdominal fetal ultrasound, and ACR guidelines are employed to pick optimal investigative imaging modality
- Omphalocele is associated with significant genetic and anatomical abnormalities, so extensive radiographic and genetic workup is advised

[20]

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