Big Baby Bellies-

The distended neonatal abdomen

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[childrensmercy.org/colorectal]
Characteristic of normal anal opening

- ARM or imperforate anus
- Congenital
- 1:5,000 live births
- Anus, rectum and nerves to not develop properly during fetal growth

How to examine newborn female infant
How to examine newborn female infant

What is the significance of this scrotal finding?
What is the significance of this scrotal finding?

PERINEAL FISTULA

- Bladder
- Urethra
- Rectum
- Perineal fistula
- Sphincter complex

COMPREHENSIVE COLORECTAL CENTER
What is the likely type of malformation here?

[Image of a medical condition]

What is the likely type of malformation here?

[Diagram labeled 'RECTOURETHRAL FISTULA']

- Bladder
- Rectum connected to urethra
- Urethra
- No anal opening

[Image of a medical condition]
How would you describe this perineum?

- A. Flat bottom
- B. Good buttock crease

What is the likely type of malformation here?
Female newborn #perineal openings

<table>
<thead>
<tr>
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<tbody>
<tr>
<td></td>
<td>Perineal fistula</td>
<td>Urogenital sinus</td>
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<td></td>
<td>Vestibular fistula</td>
<td>Rectovaginal fistula</td>
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<td></td>
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<td>Vaginal atresia</td>
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<td></td>
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<td>No fistula</td>
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<td></td>
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<td>Cloaca</td>
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What is the likely type of malformation here?
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What is the likely type of malformation here?
**Cloaca**

- Uterus
- Bladder
- Urethra
- Vagina
- Common channel
- Rectum

**Associated anomalies**

**VACTERL Screening**

- Physical Exam
- Thorax/abdomen X-ray with feeding tube
- AP/Lateral sacral X-ray
- Ultrasound
  - Renal
  - Pelvic
  - Spine
- Echocardiogram
Predictors of Continence in ARM

A - Anorectal type
S - Sacrum
S - Spine
Sacral ratio
A = Superior border of iliac bone
B = Sacro-iliac joint
C = Inferior part of sacrum/coccyx
ARM continence predictor index

<table>
<thead>
<tr>
<th>ARM TYPE</th>
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<tbody>
<tr>
<td>Perineal Fistula</td>
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<tr>
<td>Anal Stenosis</td>
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<td>Rectal Atresia</td>
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<td>Rectovestibular Fistula</td>
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<tr>
<td>Rectourethral Fistula</td>
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<td>ARM without Fistula</td>
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<td>Cloaca &lt; 3 cm Common Channel</td>
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<td>Rectourethral Fistula</td>
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<tr>
<td>Rectovaginal Fistula</td>
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<tr>
<td>Rectoblaaderneck Fistula</td>
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<td>Cloaca &gt; 3 cm Common Channel</td>
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<td>Cloacal Exstrophy</td>
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<tr>
<td>Normal flum appearance</td>
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<td>Abnormally low termination of the conus (below L3)</td>
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<td>Abnormal fatty thickening of flum</td>
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<td>Myelomeningocele</td>
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<th>SACRUM</th>
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<tr>
<td>Sacral Ratio less than 0.69 or greater than 0.4</td>
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<tr>
<td>Hemieunum</td>
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<tr>
<td>Sacral hemivertebrae</td>
<td>2</td>
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<tr>
<td>Presacral mass</td>
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<tr>
<td>Sacral Ratio less than 0.4</td>
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TOTAL NUMBER
- 3-4 = Good Potential for Continence
- 5-6 = Fair Potential for Continence
- 7-9 = Poor Potential for Continence
ARM Management

Infant

VACTERL workup

Perineal Stool

Primary PSARP or home with dilations

NO perineal stool

Colostomy

Colostomy to PSARP

1. Colostomy
Urology

- VUR (vesicoureteral reflux)
- Neurogenic bladder
- Hypospadias
- Horseshoe kidney
- Solitary kidney
- Hydronephrosis
- Bladder abnormalities
- Ectopic Ureter

Gynecology

- Hydrocolpos
  - Urine filled vagina
  - Cloacal malformations
- Duplicated systems
  - Uterus
  - Cervix
- Vaginal septum
- Atresia
- Bicornuate system (Uterus)
Early post-surgery considerations

- Diet
- Wound care
- Foley
- Dilations
- When can we do the colostomy closure?
- What happens after colostomy closure?


Long Term Follow Up

Colorectal
- 1 month, 3 months post-op, 6 months
- Annually

Renal
- Based on urological status
- VCUG 3 months after tethered cord release
- Annual renal US until puberty

GYN
- Pelvic ultrasound 6 months after breast budding

BMP (bowel management program)
Hirschsprung disease

- The nerves (ganglion cells) contract and relax the colon
- Incidence: 1 in 5000 live births
Presentation

• Newborn:
  • No meconium in 48 hours of birth
  • Enterocolitis
  • Failure to thrive (<5%)

• Older children:
  • Unmanageable chronic constipation
  • Delayed growth

Associated Diagnoses

• Trisomy 21 (10% of Hirschsprung cases)
• Central Hypoventilation Syndrome
• MEN (multiple endocrine neoplasia) Type 2
• Mowat Wilson syndrome
• Waardenburg syndrome
  • Blonde/gray hair, deaf, translucent appearing eyes,
    sideways displacement of the inner angles of the eyes

Newborn with no meconium passage >24h and obstructive symptoms
Differential diagnosis?

1. Hirschsprung Disease
2. Small Bowel atresia
3. Anorectal malformation
4. Meconium Plug (10% HD)
5. Meconium ileus (CF)
6. Colonic atresia (5% HD)
7. Allergic proctocolitis (Milk protein allergy)
8. Magnesium sulfate intrapartum
9. Diabetic mother (small left colon)
10. History of opioids
11. Hypothyroidism
Rectal biopsy method

• Suction rectal biopsy (done with suction gun, awake)
• Full thickness biopsy (done in OR)

Rectal biopsy processing

• Frozen section
  • Cannot diagnose HD, but can rule out HD

• Permanent section
  • Final read to diagnosis and determine transition zone
  • 100 slices of sample evaluated
Basic criteria for pathological diagnosis of HD

- Lack of ganglion cells in the submucosal ± the intramuscular nerve plexus
- Hypertrophied nerve fibers and nerve trunks (>40 microns)

Additional staining

- **Calretinin** adds diagnostic value when inadequate submucosa or rare ganglia present (+ in normal bowel)

- **AChe** - only for specimens in distal parts of the colon (+ in abnormal bowel)
Rectal biopsy interpretation

No ganglion cells and no presence of hypertrophic nerves?
Biopsy too low!

No ganglion cells and presence of hypertrophic nerves

Classification of Hirschsprung Disease

- Short segment = Left colon and below (80%)
- Long segment = splenic flexure and above (15%)
Case

- A 22-hour old male presents with emesis. Contrast enema performed. What would you do after this study?

A. Repeat the study
B. Perform an upper GI study

Repeat contrast enema
Contrast study - where is the transition zone?

- **Inverse rectosigmoid ratio** on *lateral view*
- Avoid CE during enterocolitis

- *Accuracy of newborn CE*: No transition zone in 11%, TZ R/S 8% had long-segment. **Biopsy first**

Proctor et al. JPS 2013
**Rectal Irrigation**

- Large Foley catheter 22-24G
- 20mls saline aliquots until clear and abdomen decompressed
- Irrigation ≠ Enema

A child with Hirschsprung disease becomes distended with fever and decreased appetite. What would you do?

A. Glycerin suppository  
B. Enema  
C. Irrigation  
D. All the above
Treatment of HAEC

- Rectal irrigations
- Antibiotics – Flagyl
- IV hydration
- Can occur even if diverted


Early post-surgical considerations

**MEDICAL ALERT**
This patient has Hirschsprung Disease and is at risk of Hirschsprung's Enterocolitis

Symptoms include: lethargy, fever, vomiting, diarrhea, and abdominal distention.

Immediate treatment: Stop oral feeds. Give IV antibiotics (Ciprin/Flagyl), IV fluids, NG tube, rectal irrigation (washout). Do not give enema.

Date:

This child is a patient of Dr. Brennan and Children's Mercy. Please contact the on-call Pediatric Surgeon at (1)(800) GO-MERCY immediately upon arrival to your facility.
Early post-surgical considerations

**MEDICAL ALERT**

This patient has Hirschsprung Disease and is at risk of Hirschsprung’s Enterocolitis.

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**Immediate Treatment:** Stop oral feeds. Give IV antibiotics (Cipro/Flagyl), IV fluids, NG tube, rectal irrigation (washout). Do not give enema.

This child is a patient of Dr. Kastenberg and Children’s Mercy. Please call Children’s Mercy Pediatrics, 800-345-2222.

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**Options**

- Ostomy with future pull-through
- Primary pull-through
- Irrigations with future pull-through

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**Timing of primary pull-through**


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Pull-through surgery for Hirschsprung disease
Hirschsprung patients postoperatively

- Nothing per rectum x 2 weeks postoperatively
- Families learn irrigations after birth in the NICU
- Irrigation learning refreshed after surgery
- After 1 month postop, if HAEC-->> irrigation
- Patients do not get routine dilations
- Botox

Summary

- ARM and HD incidence 1/5000
- Responsible for distal bowel obstruction (big belly baby)
- Workup begins with good history/physical
- Treatment determines long term outcomes