STEPS

STANDARIZED TOOLBOX OF EDUCATION FOR PEDIATRIC SURGERY

Hirschsprung Disease

APSA Education Committee
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Pathophysiology

- Congenital defect of parasympathetic ganglia
- Ganglia fail to migrate to distal bowel
- Distal bowel fails to relax
- Leads to **functional, distal bowel obstruction**

Distribution of Disease

- All patients with Hirschsprung Disease have absence of ganglia in rectum
- Proximal distribution of aganglionosis varies:
  1. Short-segment (rectum only)
  2. Rectosigmoid (~75% of all patients)
  3. Long-segment (proximal to sigmoid, although true definition variable)
  4. Total colonic
  5. All of colon and some or all of small bowel (rare)

Epidemiology

- 1 in 5000 live births
- 4:1 male-to-female ratio
- ~10% have **Trisomy 21**
- Most patients present during the neonatal period
- Some patients may present in toddler years or older with failure to thrive, chronic constipation, and/or enterocolitis

History

- **Delayed passage of meconium** (>24 hours)
- Abdominal distension
- Poor feeding
- Poor weight gain
- Enterocolitis (explosive, foul-smelling stools)
- May or may not develop bilious vomiting

Physical Exam

- May appear dehydrated or lethargic
- Fever
- Tachycardia
- Abdominal distension
- “Squirt” sign with rectal examination or stimulation

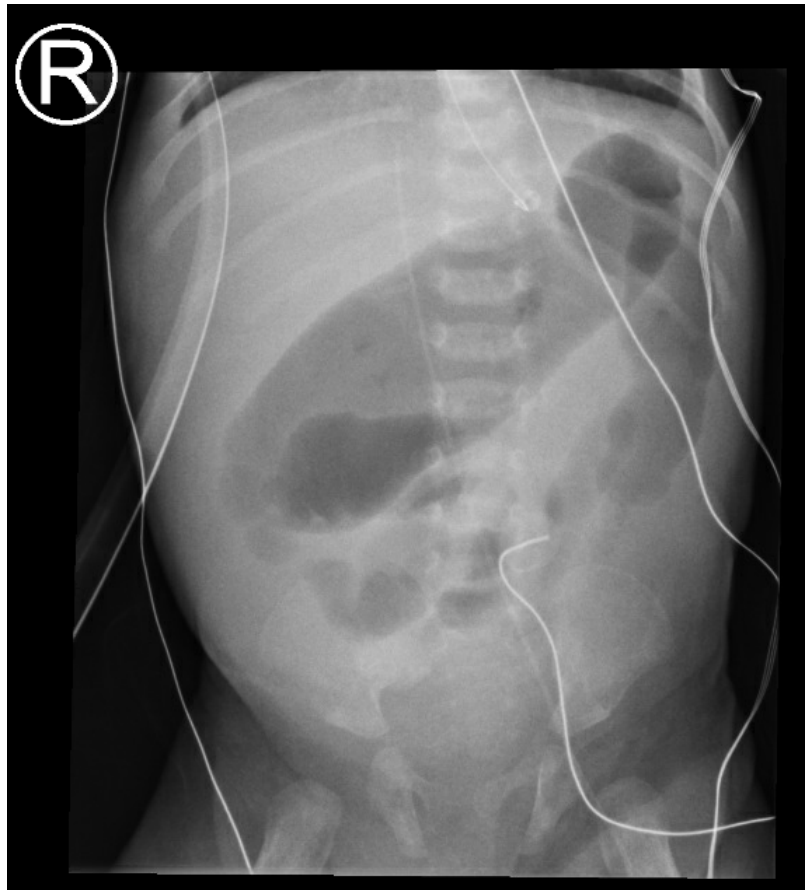
Pitfalls

- “Constipation” in a young patient with Trisomy 21 is Hirschsprung Disease until proven otherwise
- Enterocolitis is the most common cause of mortality
- Enterocolitis may occur before or **after** surgery

Studies (Labs, Imaging)

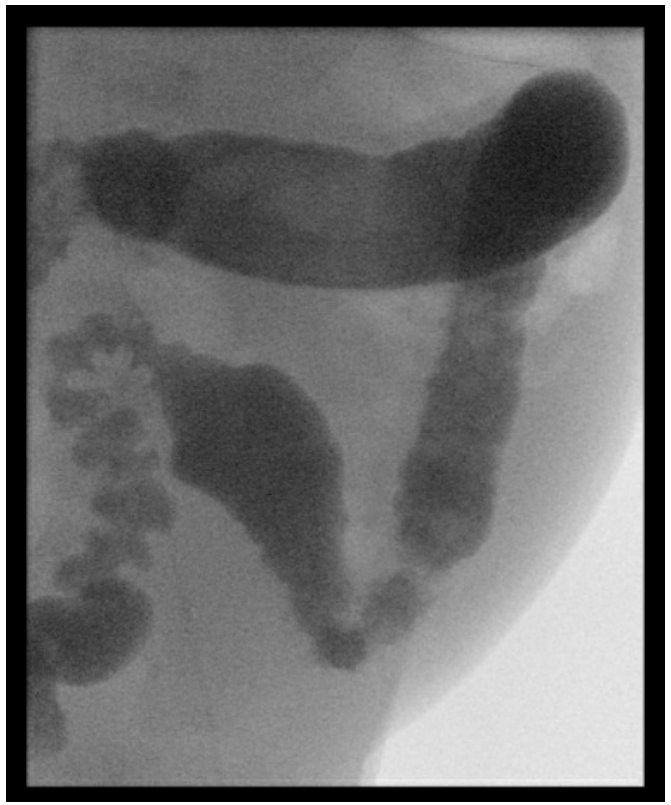
- Abdominal x-ray
- Contrast enema (not “barium enema”)
- Rectal biopsy
- +/- Anorectal manometry

Abdominal x-ray



- Dilated bowel loops
- No air in rectum
- No pneumatosis
- No free air

Contrast enema



- **Reversal of recto-sigmoid ratio** (rectum smaller than sigmoid)
- Transition zone
- Spastic-appearing distal bowel

Rectal biopsy

- Look for absence of ganglia +/- nerve hypertrophy
- Tissue should be sent to Pathology fresh (not in formalin)
- Stained for Acetylcholinesterase historically (positive in hypertrophic nerves) and more commonly, Calretinin (negative in aganglionosis)

Rectal biopsy

- **Suction** rectal biopsy
  - Preferred method in infants
  - No anesthesia required
  - Performed on ward or in clinic
  - Use “biopsy gun”
  - Biopsy of submucosa

Rectal biopsy

- **Full-thickness** rectal biopsy
  - Preferred method in older children
  - General anesthesia required
  - Performed in operating room
  - Use retractors, scissors, and sutures
  - Biopsy of submucosa +/- muscularis

Case Presentation

- 3 day-old term infant with abdominal distension
- First passed meconium at 36 hours of age
- Poor feeding
- Now febrile and tachycardic
- Abdominal x-ray shows dilated bowel loops

Initial Management

- Admission to Neonatal Intensive Care Unit
- Bloodwork and septic workup by Neonatology team
- NPO
- IV access
- Saline fluid bolus (20 ml/kg)
- Antibiotics with anaerobic coverage (e.g., Zosyn)

Surgical Consult

- Review history
- Careful physical examination (i.e., rule out incarcerated inguinal hernia, imperforate anus, etc.)
- Start rectal **irrigations** (not enemas) every 6 hours (administer 20 ml/kg of warm saline via catheter into rectum and allow drainage)
- Contrast enema

Further Management

- Re-introduce feeds once distension has resolved
- Suction rectal biopsy
- Hold irrigations for 24 hours after suction rectal biopsy
- Teach parents how to do irrigations
- Pullthrough surgery once patient is thriving (either on index admission or at approximately one month of age)

Alternatives to Surgery

- Rectal irrigations (not a good long-term solution!)
- Levelling ostomy (i.e., no pullthrough surgery)
  - Laparoscopic or open approach
  - Intra-operative fresh frozen biopsy of bowel to identify ganglia
  - Colostomy or ileostomy at “level” where ganglia are present

Operation (pullthrough)

- Surgical approach:
  - Laparoscopic-assisted
  - Open-assisted
  - Transanal only
- Anastomosis type:
  - Swenson (end-to-end)
  - Soave (submucosal cuff)
  - Duhamel (stapled pouch)*

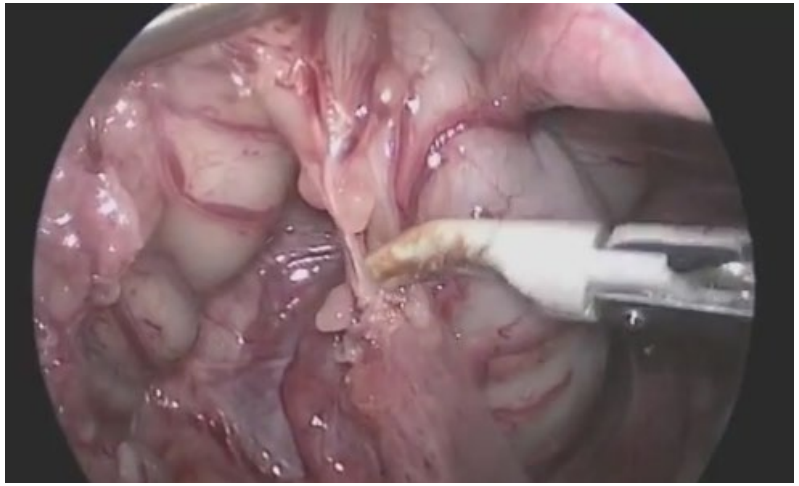
* Generally used only for total colonic disease

Operation (pullthrough)

- Foley catheter insertion
- Laparoscopic or open colonic biopsy above transition zone
- Await for confirmation of ganglia on fresh frozen biopsy
- If ganglia not present, repeat biopsy more proximally

Operation (pullthrough)

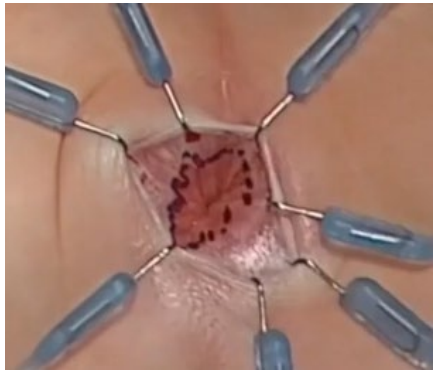
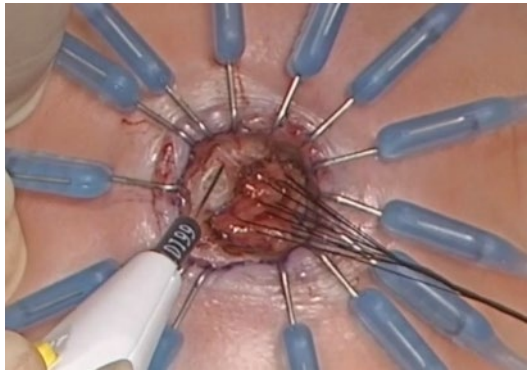
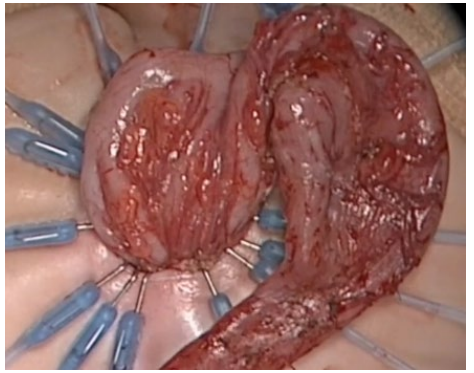
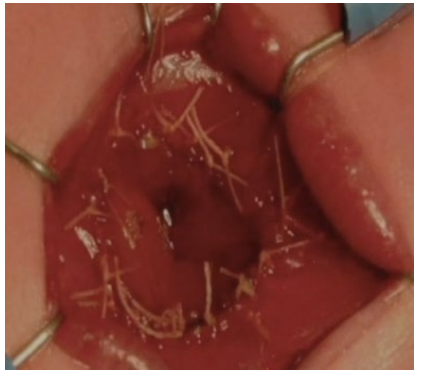
- If normal ganglia are present, proceed with pullthrough to that level
- Mobilize distal colon
- Divide mesentery (blood supply)



Images courtesy of Marc Levitt

Operation (pullthrough)

- Start transanal dissection ~1 cm above dentate line
- “Pull” the divided bowel “through” the anal canal
- Complete anastomosis with simple interrupted stitches

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Post-Op Management (inpatient)

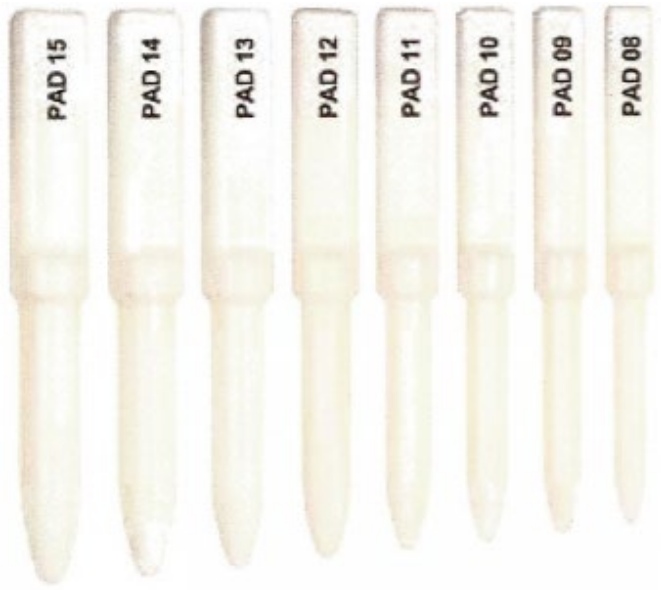
- Administer Tylenol for pain control
- Remove Foley catheter next day after surgery
- Introduce diet once patient starts stooling
- Apply perianal barrier cream to prevent diaper rash

Complications (inpatient)

- Diaper rash (common)
- Ileus
- Bowel obstruction
- Wound infection
- Anastomotic leak (rare)

Post-Op Management (outpatient)

- Start anal dilatations ~2 weeks post-op
- Teach parents how to do this at home
- May need to start with smaller size
- Gradually upsize to 12 mm for term infant then stop



- Some surgeons avoid anal dilatations altogether

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Post-Op Management (outpatient)

- Teach parents signs and symptoms of enterocolitis:
  - Abdominal distension
  - Foul-smelling, explosive stools
  - Poor feeding
  - Lethargy

Long-term complications (**enterocolitis**)

- Enterocolitis is more common in patients with Trisomy 21, young age, and long-segment disease
- Inpatient management:
  - Bowel rest
  - IV fluids
  - Antibiotics
  - Rectal irrigations
- Consider Botox injection, oral Flagyl, and/or probiotics for recurrent enterocolitis

Long-term complications (**obstruction**)

- Stricture
- Twisted anastomosis
- Transition zone anastomosis (i.e., retained aganglionosis)
- Internal anal sphincter achalasia
- Dysmotility
- Obstructing “cuff” post-Soave
- Dilated pouch post-Duhamel

Long-term complications (**fecal soiling**)

- Iatrogenic removal of dentate line (anastomosis too low)
- Iatrogenic overstretching of anal sphincter
- Normal dentate line but poor anal sensation
- Overflow fecal incontinence from constipation
- Colonic hypermotility

Questions

- Which of the following syndromes are associated with Hirschsprung Disease:

- VACTERL
- CHARGE syndrome
- Trisomy 21
- Gastroschisis
- Omphalocele

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Questions

- What is the most common distribution of disease in patients with Hirschsprung Disease?

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- Rectosigmoid
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Final Discussion/Review

- Hirschsprung Disease presents in neonates with delayed passage of meconium, abdominal distension, and/or enterocolitis
- Workup includes contrast enema and rectal biopsy
- Definitive procedure is pullthrough surgery
- Long-term complications include enterocolitis, obstruction, and fecal soiling

References

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- Saadai P, Trappey AF, Goldstein AM, et al.; American Pediatric Surgical Association Hirschsprung Disease Interest Group. (2019) Guidelines for the management of postoperative soiling in children with Hirschsprung disease. *Pediatr Surg Int* 35:829-34. PMID: 31201486.

Acknowledgement Slide

The preceding educational materials were made available through the
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