Hirschsprung disease

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Introduction

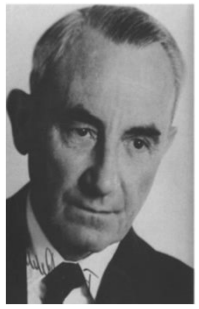
- Also known as **congenital megacolon**
- **common** condition in pediatric surgery (1:5000)
- **Enterocolitis** is the most feared complication
- basic principles of the surgical treatment established by Dr. Swenson are still timely but **different modalities** of have been described
- Despite the relatively common occurrence of postoperative constipation, soiling, and enterocolitis, **most resolve in first 5 years** of life.

Introduction

1887, **Hirschsprung** = described cases of HD

1949, **Swenson** = first pullthrough

1956, **Duhamel** = anastomose Ganglionic to post. rectum



1958, **Rehbein** = low anterior resection for HD

1964, **Soave** = submucosal dissection

1995, **Georgeson** = laparascopic pullthrough

1998, **De la Torre** = transanal pullthrough in rabits

1999, **Langer** = transanal one stage pullhtrough

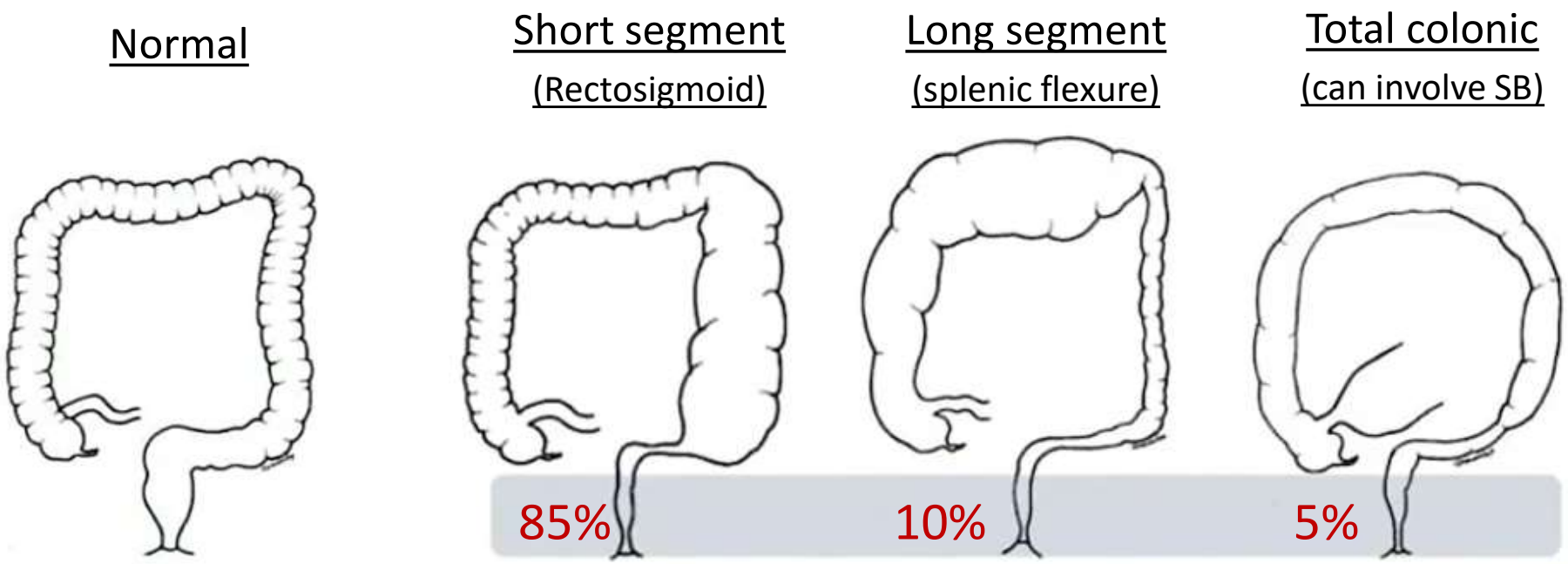
Outline

1. Basic science
2. Patient evaluation
3. Diagnosis
4. Preoperative considerations
5. Definitive Surgery
6. Post pullthrough problems
7. Variant Hirschsprung Disease

1. Basic science

- Spectrum
- Etiology
- Genetics
- pathogenesis

Spectrum of HD



- ***Existence of ultrashort segment is debated****. (difficult to d/t from idiopathic constipation and internal sphincter achalasia)*

Etiology

- 13 wk embryo - neural crest cells migrated through the GI tract (from proximal to distal)
  - *A disorder of target cell **migration** from the neural crest during embryonic development*
- Subsequently - Neural crest cells differentiate into mature ganglion cells
  - *Abnormal **differentiation** of cells in an altered microenvironment*

Genetics

- Not genetic disease but **associated genes** identified
  - RET mutation (tyrosine kinase) in 50 % of familial and 15-20% sporadic HD
  - ENDRB mutations (endotheine receptor B) in 5%
  - EDN3 (endothelin 3)
- Presence of **family history** (overall recurrence risk in siblings is 4%)
- **Chromosomal abnormalities** are reported in 12% (5% of have downs syndrome)
- Up to 18% have an **associated syndrome (**MEN 2A, Shah–Waardenburg, central hypoventilation)

Genetics

Sex and length of aganglionosis in proband	Risk of recurrence in a brother (%)	Risk of recurrence in a sister (%)
Male + short segment	5	1
Male + long segment	17	13
Female + short segment	5	3
Female + long segment	33	9

- **Carter's paradox** (The highest risk is for the brother of a girl with long segment HD). length of the aganglionosis and sex affects the risk of recurrence. (M:F = 4:1 but in long segment 1.5:1 and in familial 1:3)

Pathogenesis

- **lack of ganglion cells in the bowel wall plexuses**
  - Superficial submucosa (meisners)
  - Deep submucosa (henle)
  - Musculais (auerbach)
- **Colonic obstruction**
  - Spasm
  - lack of propulsive peristalsis
  - lack of relaxation of the internal sphincter.
- **overgrow abnormal bacteria** (C.difficle) b/c HD patients don't tolerate fecal stasis like normal individuals
  - Enterocolitis
  - Endotoxemia
  - Hypokalemia
  - Perforation
  - pericolic abscesses

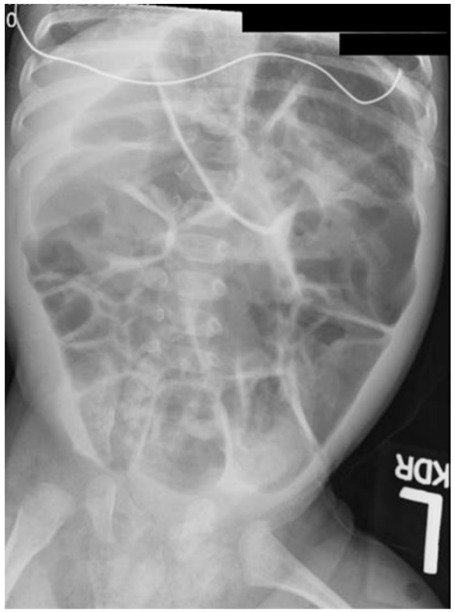
2. Patient Evaluation

- Neonatal presentation
- Later presentation

Neonatal presentation

- Delayed meconium passage after 24hr (90% of HD)
- Abdominal distention
- Vomiting
- cecal or appendiceal perforation may be the initial event
- rectal stimulation results in an explosive and massively deflating passage of liquid stool and gas
- enterocolitis (fever, fetid diarrhea, distention) in 10%

Neonatal presentation



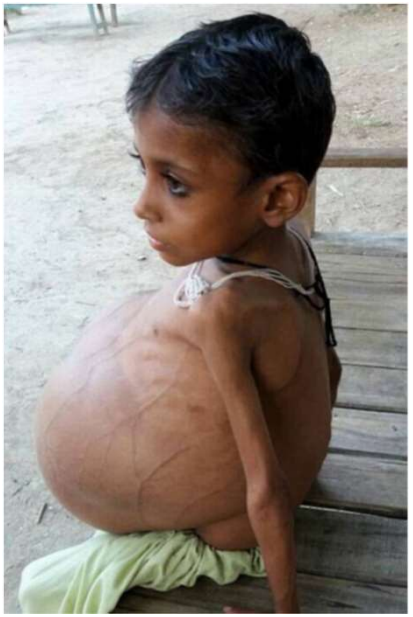
- It is extremely difficult to differentiate in a plain abdominal film in a newborn baby a distended colon from a distended small bowel. But there are few neonatal conditions that give rise to such enormously dilated bowel. Next step is next step is a water-soluble contrast enema.

Differential diagnosis for neonatal HD

1. **Meconium Plug** (10% HD)
2. Meconium ileus (CF)
3. Hypomotile colon (Hypothyroidism, History of MgSo4, Opioids)
4. Allergic proctocolitis (Milk protein allergy)
5. Small left colon syndrome (History of diabetic mother)
6. Colonic atresia (5% HD)
7. Small Bowel atresia
8. Anorectal malformation

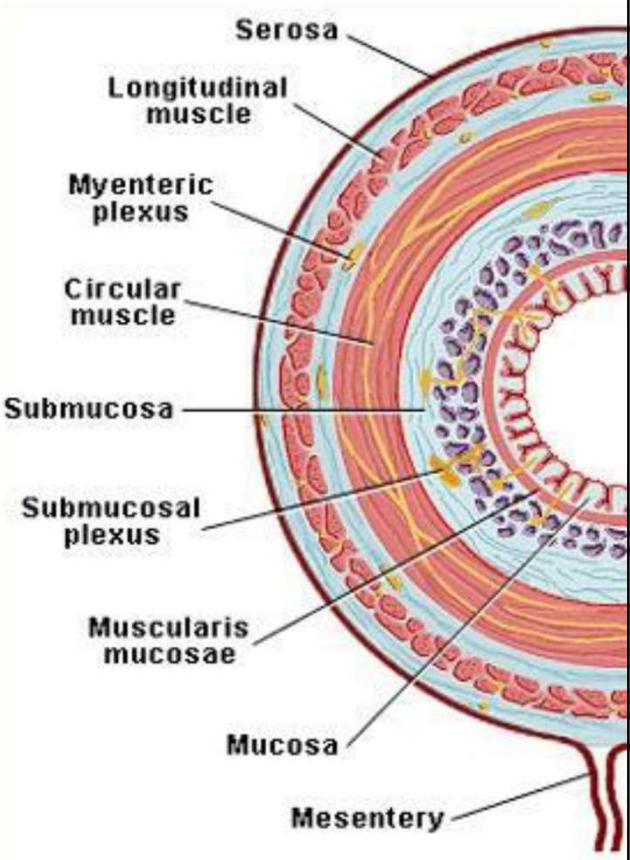
Late presentation

- chronic abdominal distention
- Poor growth (malnutrition)
- dependence on enemas without significant encopresis
- Enterocolitis
- Volvulus (undiagnosed short segment HD)



3. Diagnosis

- contrast Enema
- Anorectal manometry
- Rectal Biopsy

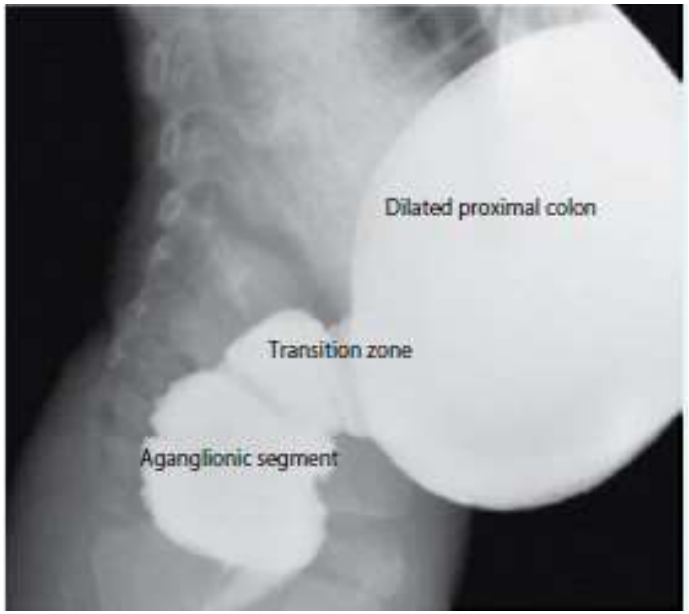


2.1. Contrast Enema

Contrast enema reinforces diagnosis (80% sensitivity)

- small-caliber catheter is placed in the rectum as close to the external sphincter as possible
- Iso-osmolar Water soluble contrast preferred in neonate to r/o other diagnosis
- critical imaging is the *initial lateral view* as contrast enters the colon.
- contrast enema is terminated when a zone of transition is identified.

2.1. Contrast Enema



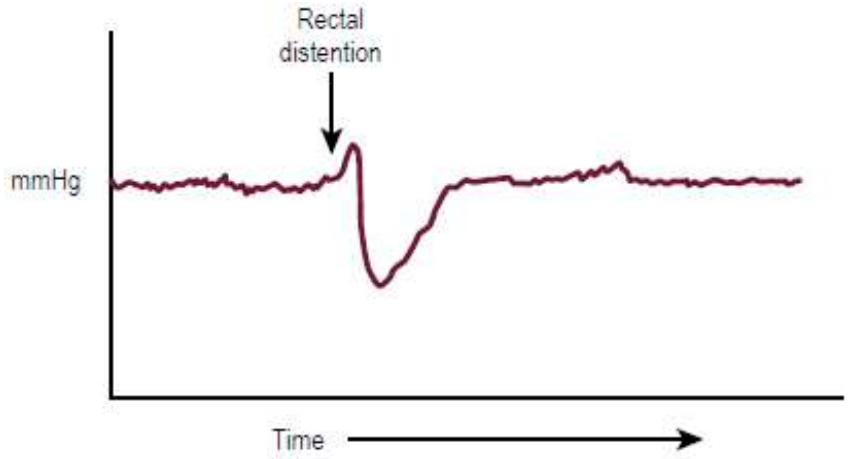
- aganglionic colon is often normal in caliber with an abrupt **transition** to dilated proximal colon. serrated in appearance due to aperistaltic contractions. May not be very obvious in the newborn period (10%). Abnormal **rectosigmoid ratio.** may be misleading in very short segment and in more extensive aganglionosis. Another feature is on plain xray after 24 hrs with **failure to expel contrast.**

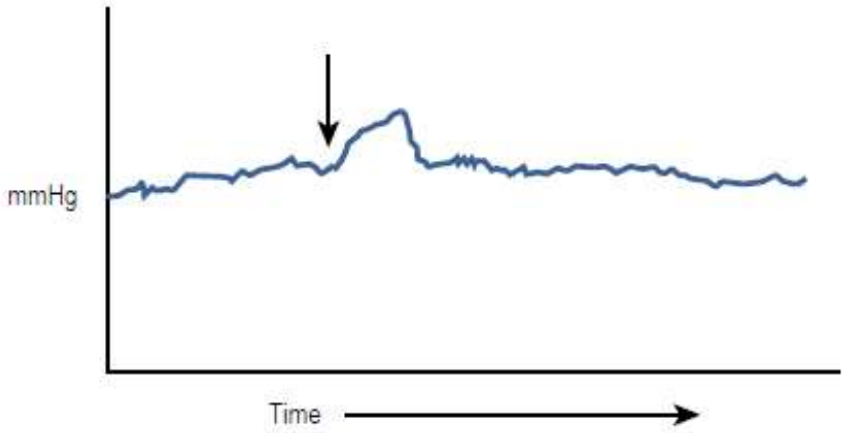
2.2. Anorectal manometry

Presence of rectoanal inhibitory reflex rules out HD

- Use limited to children >1yr
- better initial diagnostic pathway for older children (b/c suction rectal biopsy is less reliable)

2.2. Anorectal manometry





- In contrast to a normal manometry, there is no reflex relaxation (drop in the pressure) in response to balloon distention of the rectum

2.3 Rectal biopsy

Biopsy confirms diagnosis

- Indication
  - Rectal biopsy is mandatory if contrast enema is suggestive of HD
  - Total colonic mapping is needed for transition proximal to splenic flexure on contrast enema
- accuracy depends on …
  - Site of biopsy (>1 cm above dentate) to avoid anatomic area of hypoganglionosis (but still no hypertrophic nerve)
  - Adequate specimen
  - Skills in processing of the specimen
  - Correct interpretation by the pathologist
  - Accurate reporting and interpretation by surgeon

2.3 Rectal biopsy

	Suction biopsy	**Open biopsy**
Indication	Gold standard	After 2 inadequate suction biopsies
contents	Submucosa only (less rich in ganglion cells)	Full layer (both plexus)
Pathology interpretation	More difficult	Easier
Other	Not for age >3 yr	Scar problematic for subsequent pullthrough

2.3 Rectal biopsy

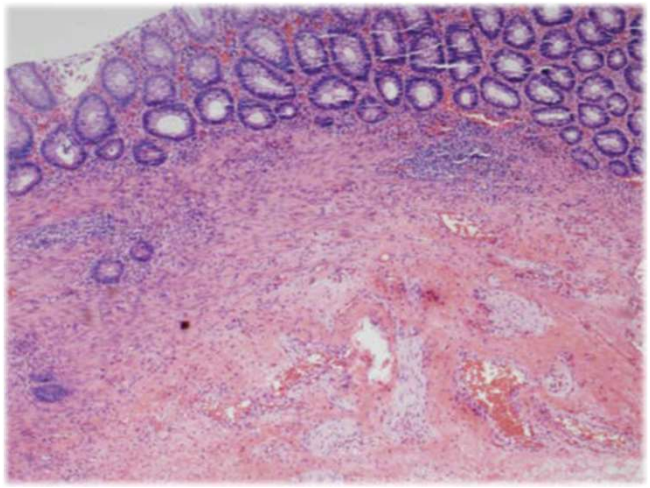
- **Positive biopsy**
  - Absence of ganglion cells in submucosa (± muscularis)
  - Presence of hypertrophied nerves (>40 µm for infants)- -(found in submucosal layer)
- **Inconclusive biopsy**
  - Absence of ganglion cells but hypertrophic nerves absent and AChE staining not increased

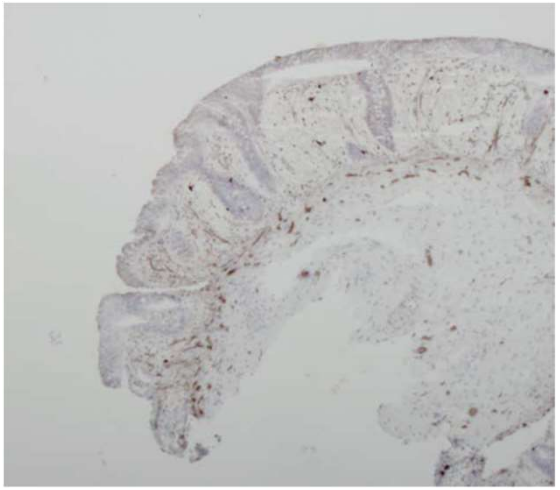
  **repeat biopsy if clinical suspicion remains (could be very short or long segment HD)
- **Inadequate biopsy**
  - No adequate submucosa in >50 levels.
  - Squamous epithelium

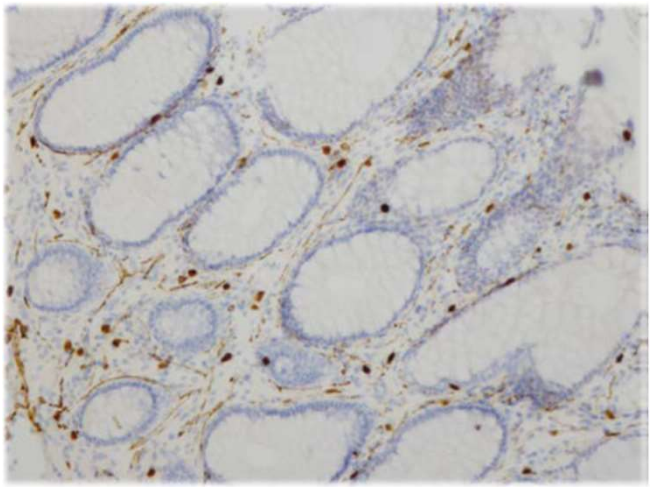
Histopathologic processing

- **H + E stain**
  - *preferred choice for identifying ganglion cells*
  - Requires only fixation and standard processing
  - If ganglion cells seen, one can reject the diagnosis of HD.
  - If ganglion cells are not seen, further tests are required to prove HD
- **AChE hyperactivty**
  - *thickened mucosal and submucosal nerve fibers*
  - Requires additional processing (should be sent lab fresh- without formalin)
  - Inadequate sensitivity (~85%)- superficial biopsy (no muscularis), immature enzyme (<2yr)
  - If characteristic findings are not met, should consider additional specimens
- **Calretnin immunohistochemistry**
  - *calretinin-positive fibrils are absent in aganglionic ganglionic bowel*
  - Used if problem with ganglion identification on H&E, specimens with inadequate submucosa

Histopathologic features



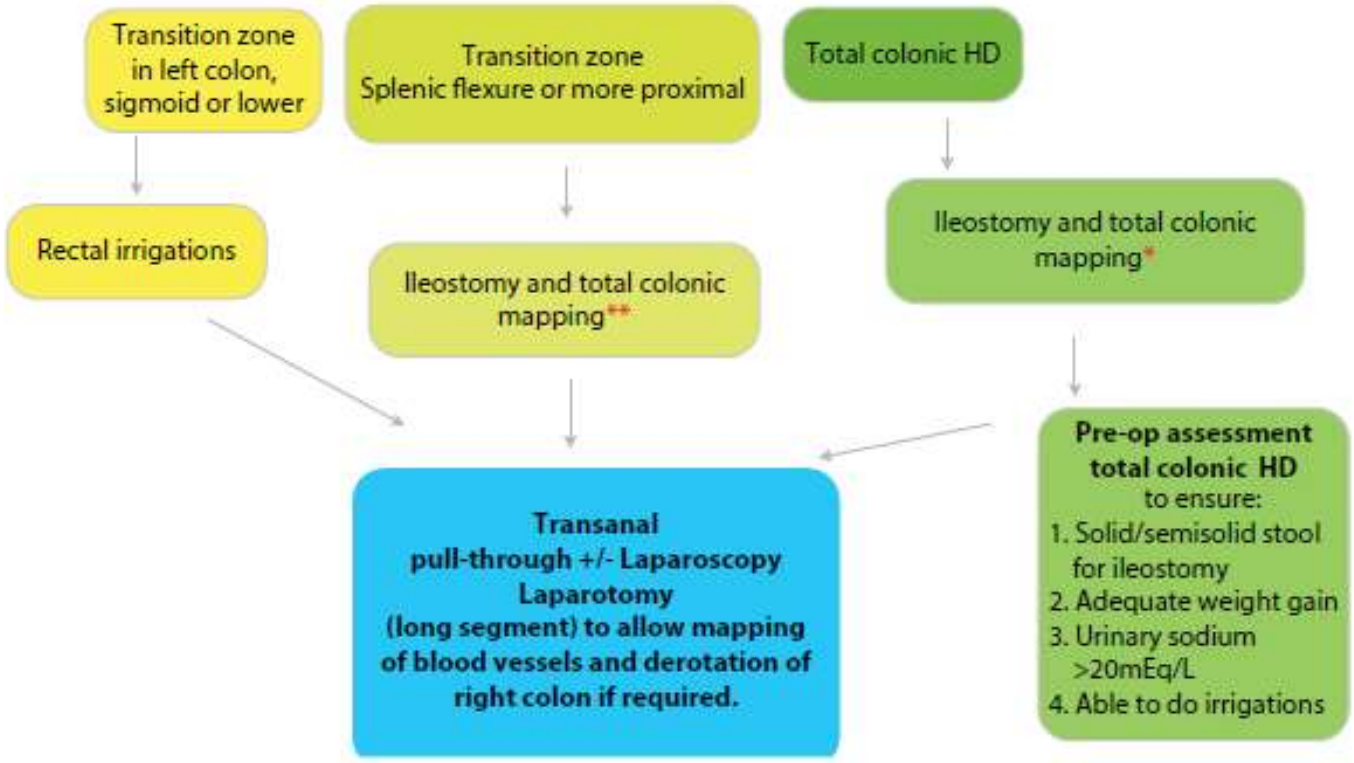




- H&E = absence of ganglion cells, presence of hypertrophic nerve trunks
- AChE = increased staining
- calretnin = negative

4. Pre-op considerations

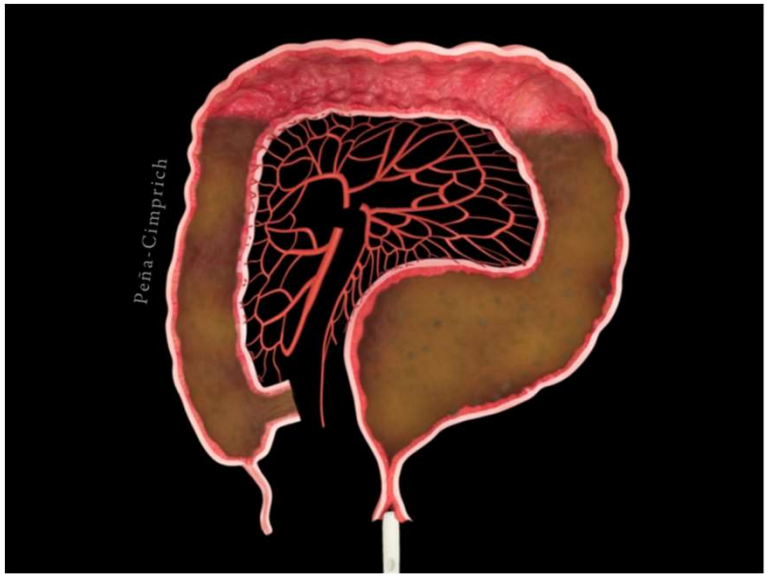
Management approach



Considerations in neonates

- **Stabilization**
  - IV fluid
  - Parenteral nutrition
  - IV metronidazole
  - Colonic irrigation (use is limited if transition is above rectosigmoid)
- Can be discharged home and **pullthrough semi-electively**.
  - with Breast milk/ elemental formula
  - Parents should be educated and equipped to perform Rectal irrigations.
- **Primary pullthrough in newborn** has many requirements
  - experience, sophisticated surgical environment
  - good anesthesia, good intensive care
  - good pathology
  - possibilities to provide parenteral nutrition to the baby.

Considerations in neonates



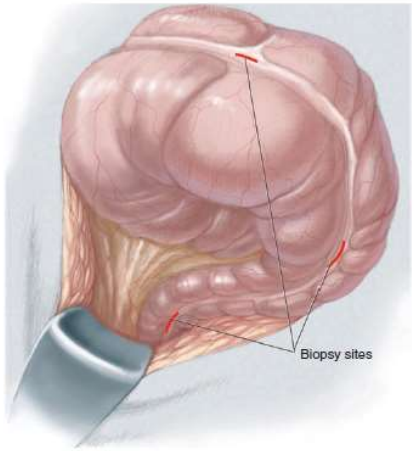
- **Play Video =>** **Colonic irrigation** = Decompress colon with large (20-24 Fr) foley catheter inserted through the rectum past the transition zone. irrigate with 30-50 mL saline at a time (no more than 20ml/kg should remain in colon). Liquid stool comes out through the lumen of the tube- keep moving the tube back and forth and rotating it, trying to get into the pockets of gas and liquid stool. Goal is to keep abdominal wall flat and may be repeated several times during the day as needed (at least 3x/day). *Do NOT delegate rectal irrigations to the nurse right away.* Irrigation should be continued until definitive surgery. If a child deteriorates under irrigations, order another AXR to check on colonic dilation and enterocolitis .

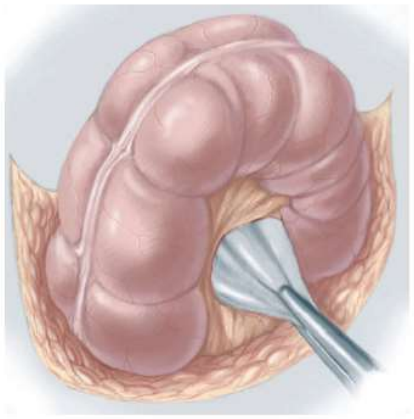
Considerations in later presenters

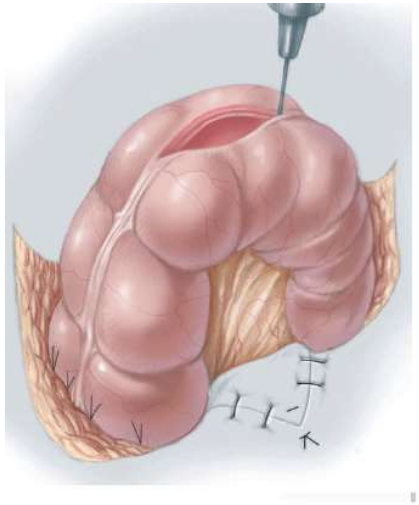
- May have an extremely dilated colon (evaluated on contrast enema), and pull-through should be delayed until the diameter has decreased sufficiently
    - weeks to months of **irrigation**

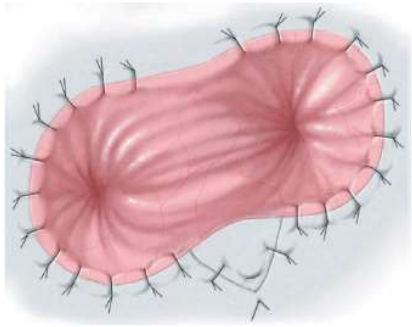
      or
    - **Leveling colostomy** (The stoma end may need to be tapered if extremely dilated
- Older children require open rectal biopsy
- Older children tend to bleed more during pullthrough (prepare blood)

Considerations in late presnters









- **Leveling colosotomy =** stoma in ganglionated bowel as determined by intraoperative frozen section. In the usual case of rectosigmoid aganglionosis, three seromuscular biopsies are taken along the antimesenteric surface without entering the lumen. One biopsy is taken from the narrowed segment of bowel, a second biopsy from the transition zone and a third biopsy from the dilated portion above the transition zone.

Considerations in Typical HD

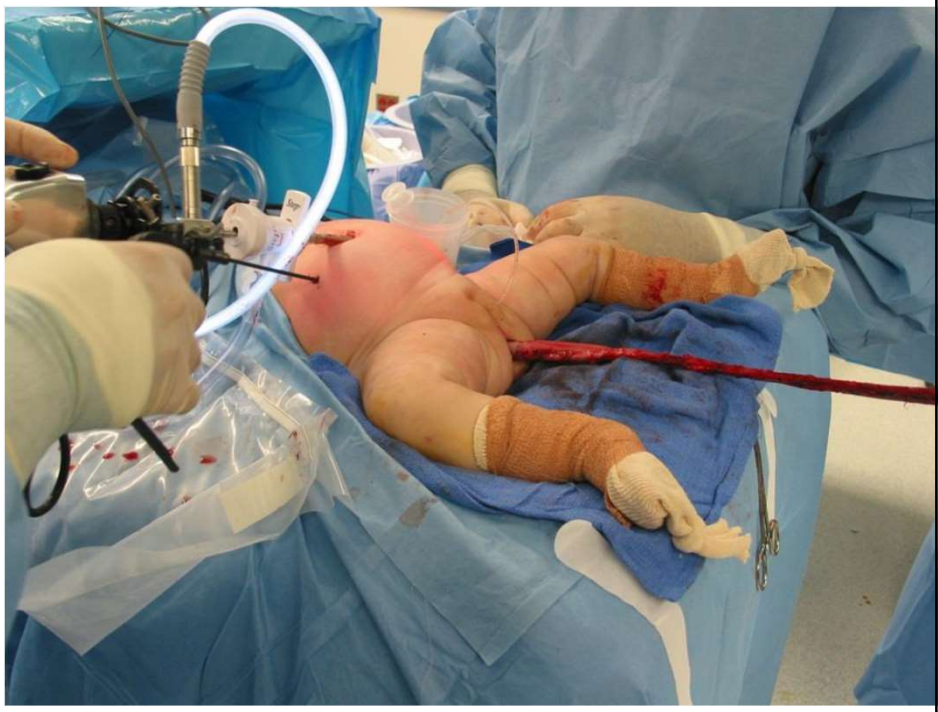
- **Colostomy** was advocated after the initial Swenson observed to have high leak and stricture
- **Primary pullthrough** was later proven to be safe and efficient even in small infants.
- However, there are still indications for colostomy
  - Neonatal obstruction **not responding to irrigations**
  - severe **enterocolitis**
  - **Perforation**
  - **Malnutrition**
  - Older child with **massively dilated** proximal bowel (will normalize after stoma)
  - Situations where it is not possible to identify transition zone with **frozen section**

Considerations in Typical HD

- Total transanal approach might suffice
- usually a sigmoid transition zone easily decompresses with irrigation
- **If bowel not decompressed on x-ray**, consider long segment (abdominal approach)

5. Definitive Surgery

- Types
- Approach
- Technique

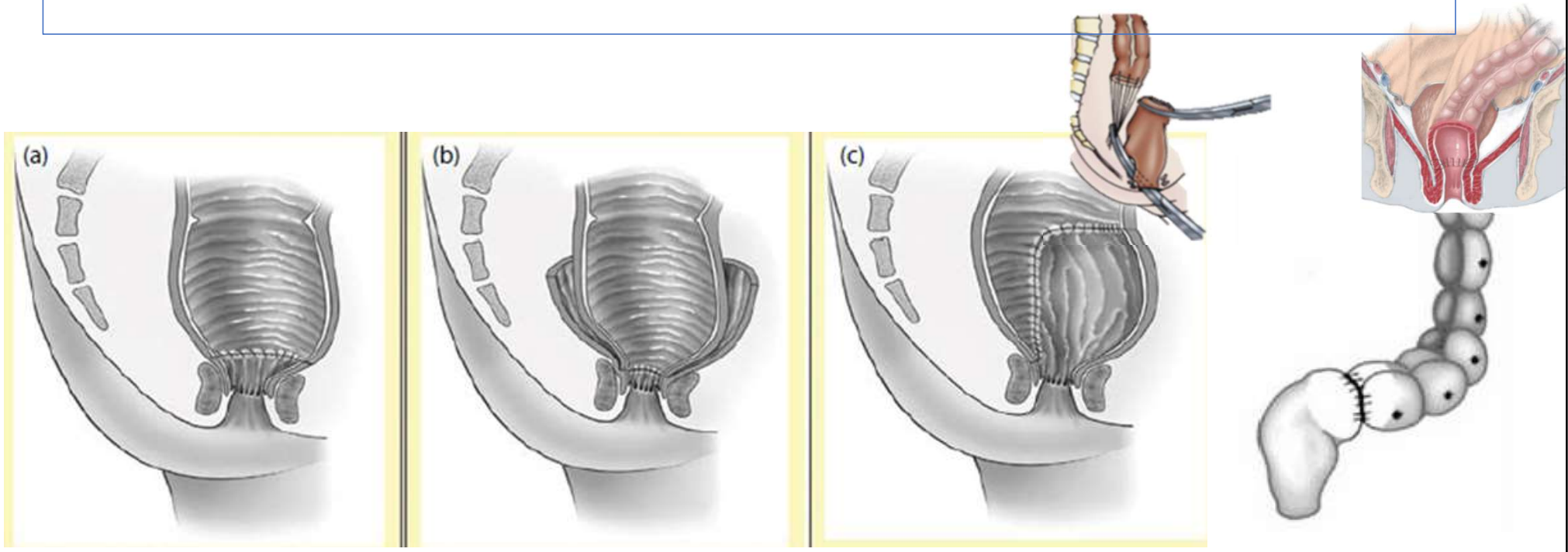


Choosing type of pullthrough

As there are very few prospective studies comparing operations, the best surgery is the one that the surgeon has been trained to do and does frequently.

- **Swenson** = Risk of damage to adjacent structures
- **Soave** = **less damage** to adjacent structures but risk of **cuff obstruction**
  - Initially Submucosal dissection extended above peritoneal reflection but this resulted in rolling back and obstruction even if cuff longitudinally divided
  - length of submucosal dissection varies according to surgeon, although trend is toward a shorter cuff
- **Duhamel** = **less damage** to other structures **and stenosis** but risk of feces **accumulating in pouch**
  - * recent studies suggest Duhamel is inferior
- **Rehbein** = **increased anastomotic leak** and **obstruction** but still practiced in German countries
  - deep anterior resection with anastomosis 3-4 cm above dentate (not widely practiced)

Choosing type of pullthrough

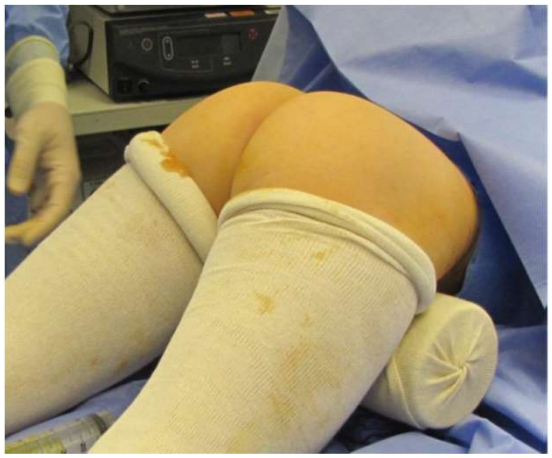


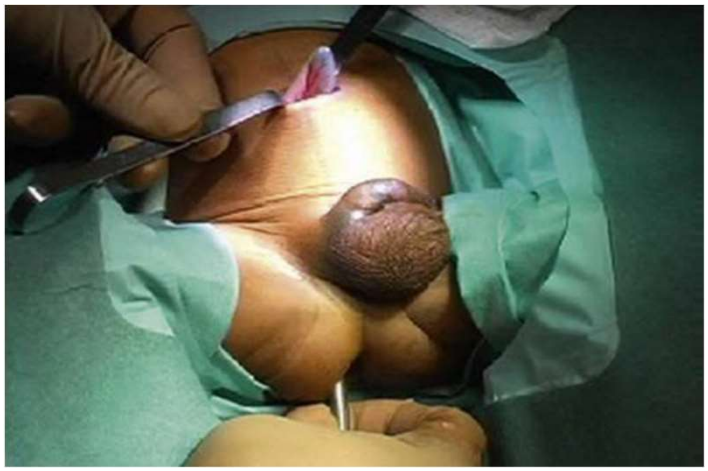
- **Swenson:** no submucosal dissection (no remnant aganglionic), **Soave:** submucosal dissection to peritoneal reflection (remanat aganglionosis circumfrentally), **Duhamel:** rectum divided and closed at peritoneal reflection and anastomosed posteriorly (remnant aganglionosis anteriorly), **Rhebein:** low anterior resection (remnant aganglionic distally)

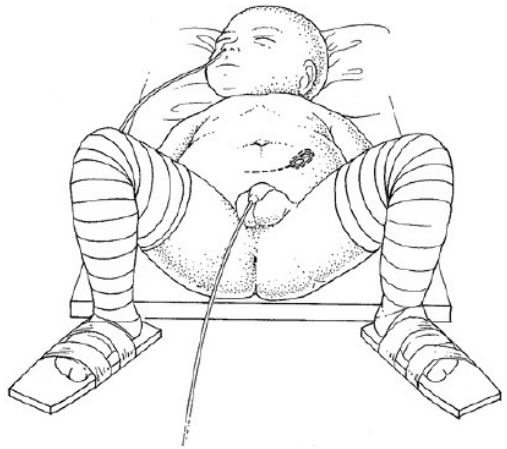
Approach to pullthrough

- **Transanal only** (swenson/soave)
  - Option for those with rectosigmoid HD (80%)
  - some argue transitional zone biopsy should be done before beginning dissection
  - Can be utilized in a child with pre-existing colostomy using mini-laparotomy
  - Less pain, early feeding, short hospital stay, better cosmesis, ? Less adhesion
- **laparoscopy assisted**
  - Higher transition zone (above mid sigmoid)
  - To mobilize left colon/splenic flexure
  - To take leveling biopsy
- **Laparotomy**
  - Bowel to distended for laparoscopic mobilization

Technique: positioning & incision

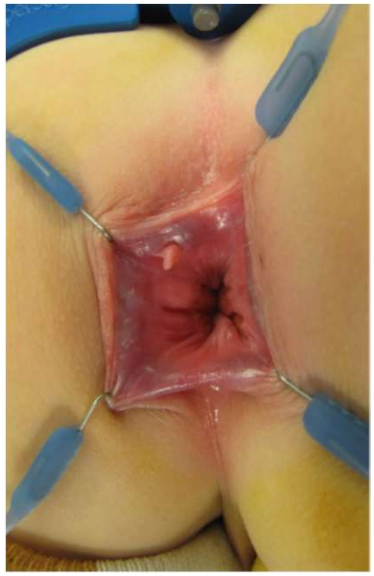


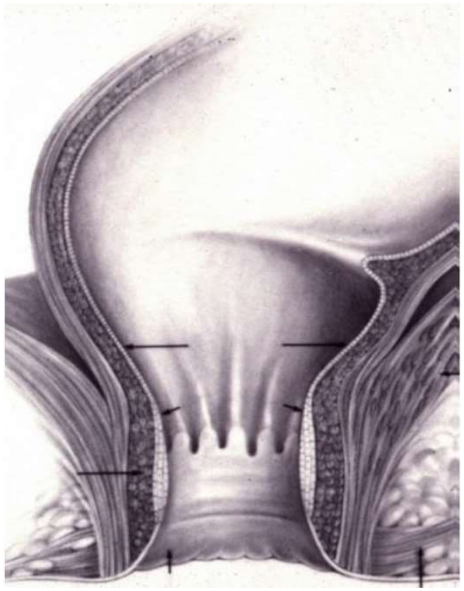


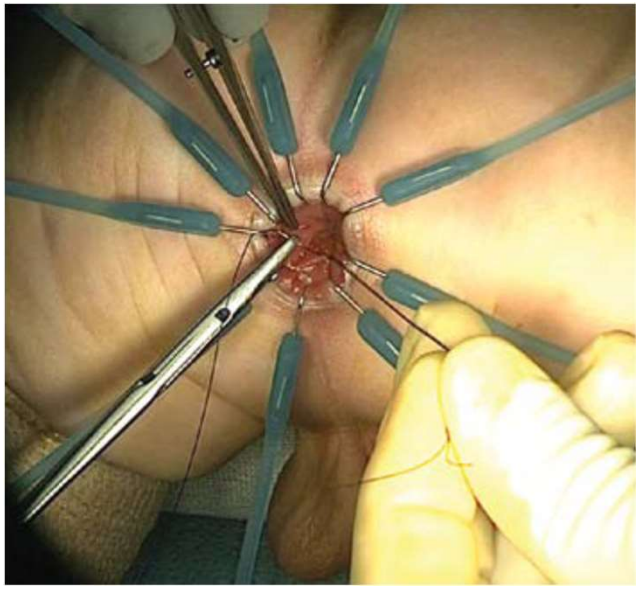


- Some prefer **prone** position with total transanal approach b/c straighter and also more familiar to the pediatric surgeon. Some argue **laparoscopy** and transition zone biopsy should be done prior to anal dissection b/c **20%** of neonates have **no radiographic transition** zone and **8–10% of** rectosigmoid transition on contrast have **more proximal histologic transition**. **Small Umbilical incision** can also be used and hegar dilator can push sigmoid into the incision for the biopsy. In children with colostomy an **oblique (hockey-stick) incision incorporating the stoma** can be used for intrabdominal mobilization .

Technique: Identifying dentate

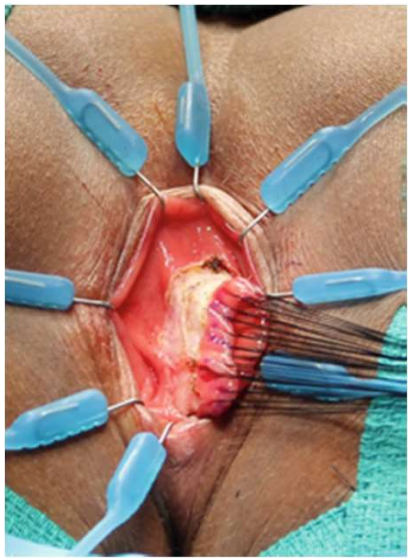


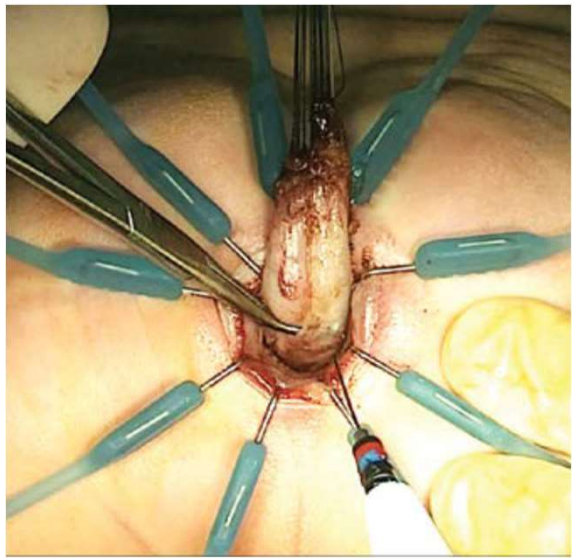


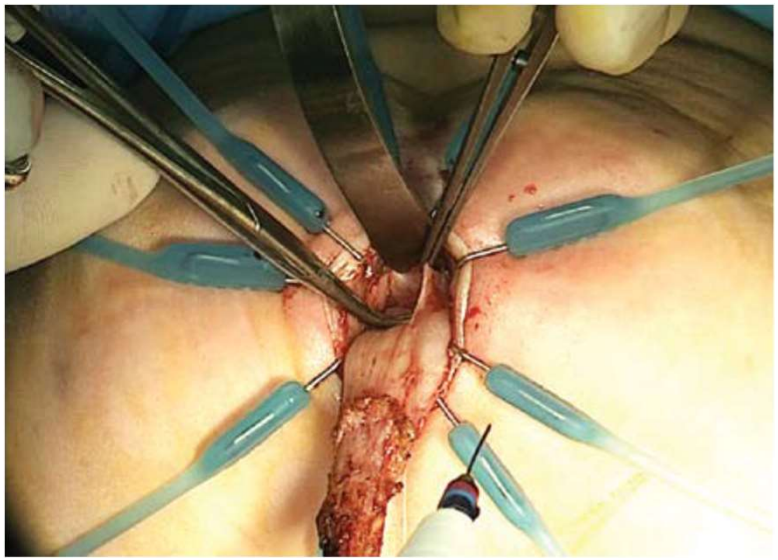


- place the 8 hooks of the lone retractor are placed to visualize dentate. dentate line is above the columns. Then hooks are placed 1cm above dentate to protect anal canal. Multiple 5-0/6-0 silk sutures are inserted circumferentially 1 cm above the dentate line (5mm for neonate) to mark the line of initial dissection.

Technique: Transanal disssection

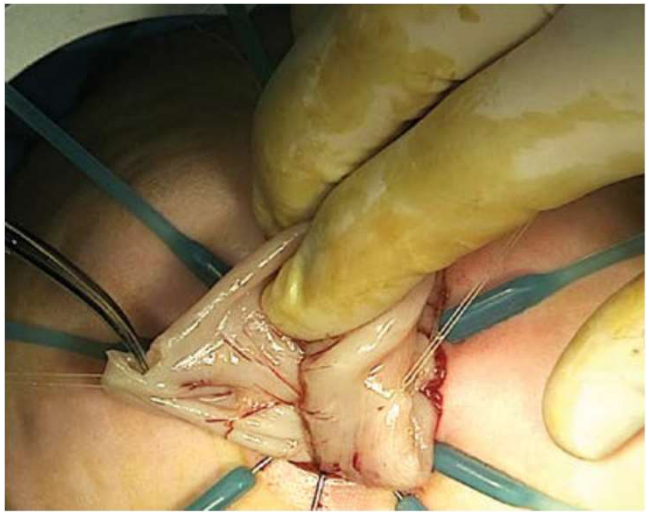


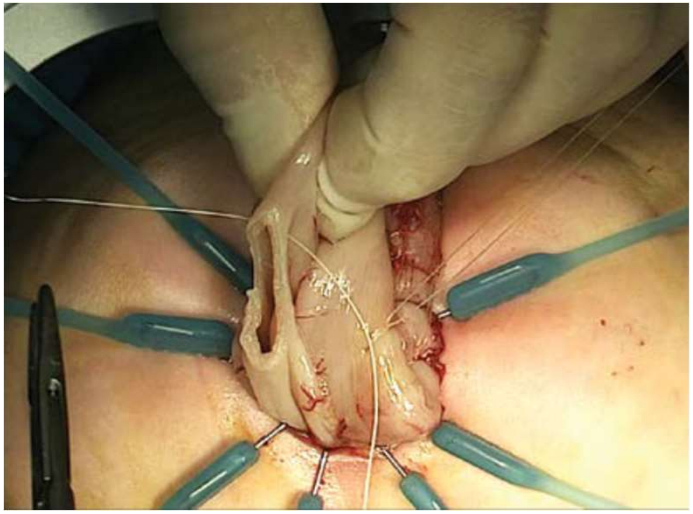


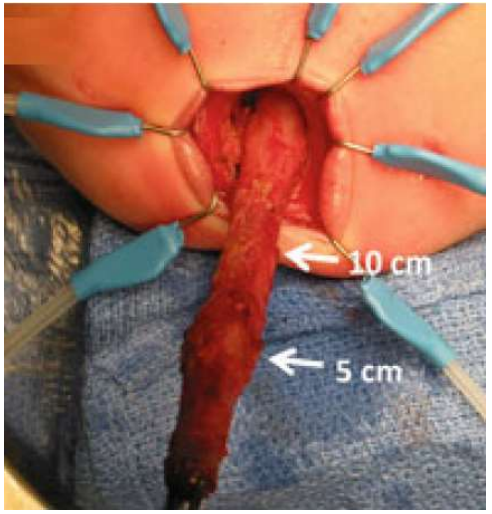


- Use silk sutures for traction. Use needle tip cautery to make full-thickness incision peripheral to sutures for the Swenson plane or initial 2-3 cm submucosal dissection for soave. first anteriorly plane then posterior. Dissection should be as close as possible to the bowel wall without injuring any nerves and/or pelvic organs. Should not violate the skeletal muscle surrounding rectum to go into ischorectal fat pad. long, narrow Deaver retractor may be used

Technique: leveling biopsy

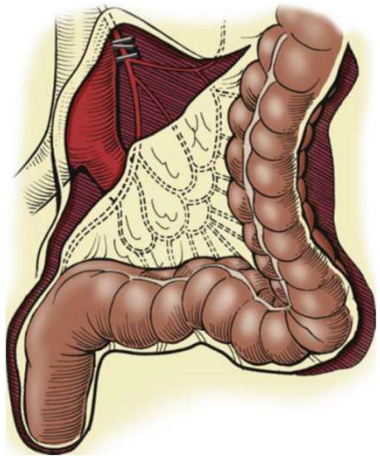
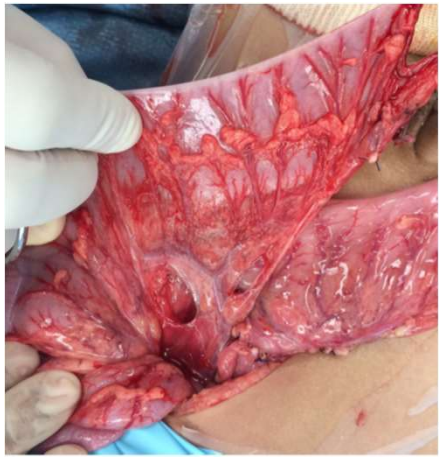
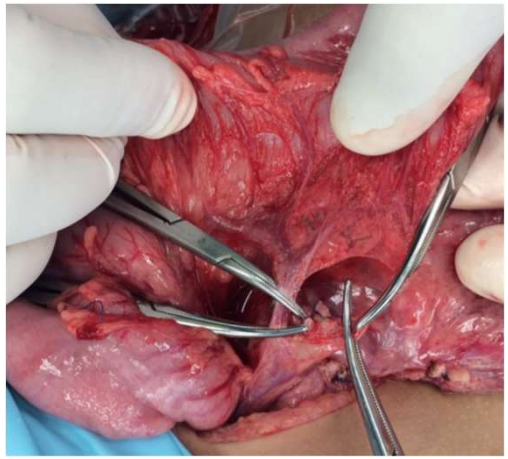
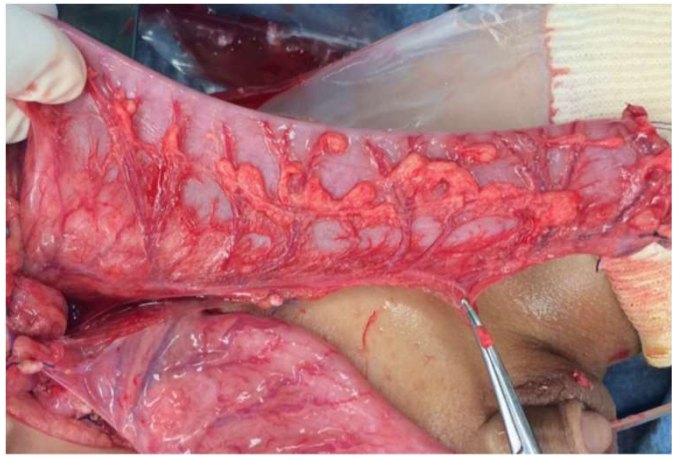






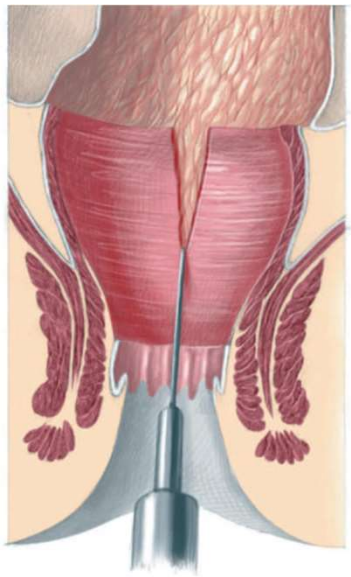
- dissection above peritoneal reflection to deliver sigmoid and transition zone biopsy taken. sigmoid is then further mobilized. take full-thickness biopsies every 5 cm until we reach the normoganglionic bowel. Then go 5 more cm (at least 2 cm). If the normal ganglionic bowel is very dilated, we continue pulling down even more colon.

Technique: intra-abdominal mobilization

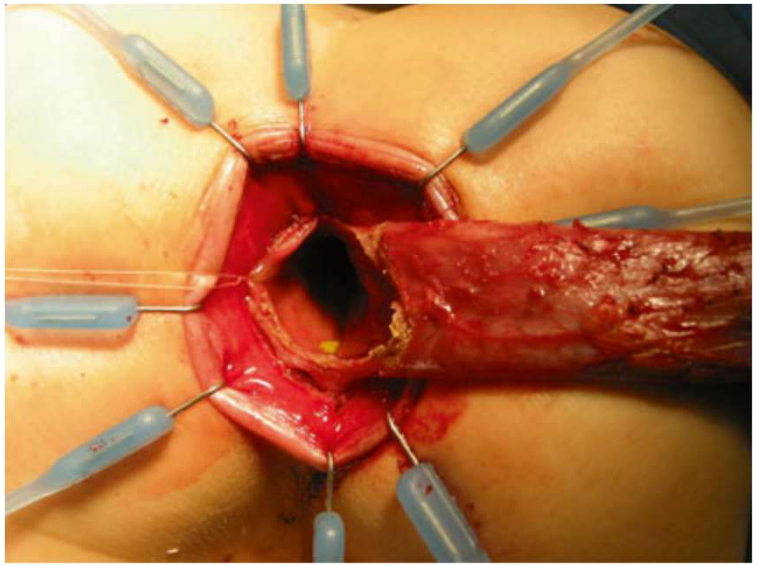
- In 20% normo-ganglionic bowel not reached through rectum. dissection becomes more difficult and risky trying, to cauterize mesenteric intraperitoneal vessels that may retract and bleed into the peritoneal cavity. In these cases as well as in patients with colostomy, laparoscopy/laparotomy is done. Descending colon and splenic flexure are detached from their attachments. It may also be necessary to ligate and **divide either the IMA** just distal to its origin **or the left colic artery** just at its origin sparing the arcades (marginal artery). colon must reach just below the level of the perineum when drawn over the child's pubic symphysis with only modest tension.

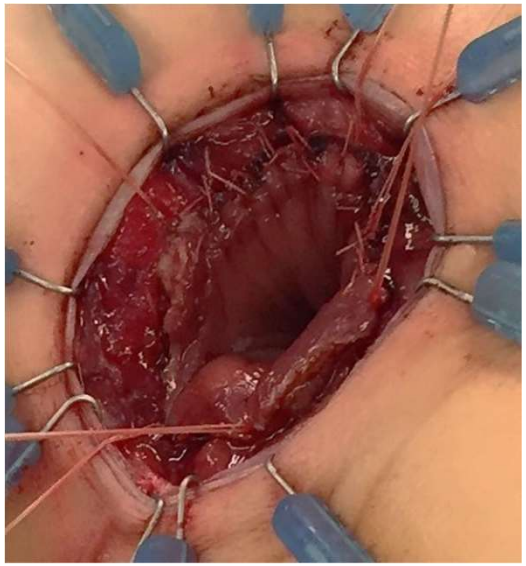
Technique: splitting rectal cuff



- The length of the submucosal dissection varies according to surgeon. If submucosal rectal dissection is short (1-2 cm) before transitioning through the muscle into the “Swenson” plane splitting rectal cuff is unnecessary (Langer). However, Long soave rectal muscular cuff (peritoneal reflection) needs to be split longitudinally, either anteriorly or posteriorly

Technique: Anastomosis





- Bowel trimmed and anastomosed circumferentially in two-layers 2 cm above the pectinate line. First layer (serosal) is anastomosed above edge of the resection of rectal mucosa before resection of colon. In second inner layer mucosal edges brought together with 4-0 vircyl. In case of dilated proximal bowel with size discrepancy, better to hold stiches at 4 cardinal points with ends connected to mosquito to pull them out (this will straighten the bowel in between). Continue anastomosis bisecting stiches and pulling them out. If stiches are too deep it will pick up sphincter behind (will cause pain on defecation).

6. Post-pullthrough complications

Post op care

- Post op **Foley**, **analgesia**, IV **antibiotics**
- **protect the buttocks** with barrier cream
- **Educate about enterocolitis** (rapid illness and death)
- **Feeding**
  - transanal pull-through can be fed immediately and discharged within 24–48 hours
  - Milk substitute– most are lactose intolerant. And may lead to enterocoitis
  - if there is distention, x-ray dilated colon with gas and liquid = surgeon has to perform colonic irrigation
  - If child continues not to toerate dec irrigation for months consider redo and resecting more normoganglionici bowel
- **Dilation**
  - rectal examination done at 1 mo to check if anastomosis is narrow (pena)
  - calibrated with an appropriat size dilator or finger at 1–2 weeks (langer).
  - weekly calibration by the surgeon for 4-6 weeks

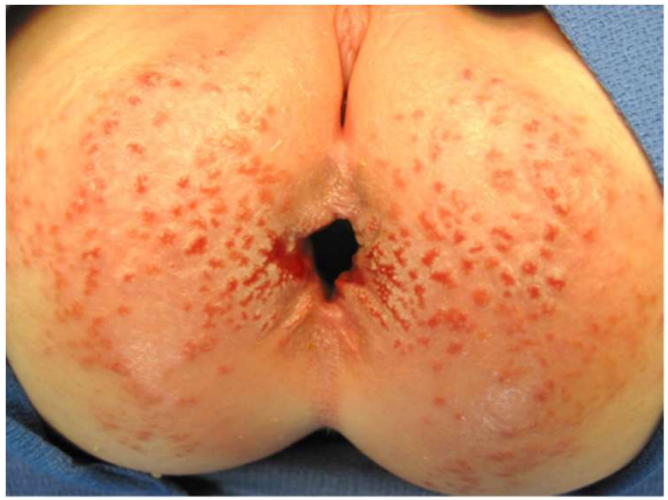
Early complications

- Bleeding
- Injuries
  - neurogenic bladder, impotence
  - rectourethral and rectovaginal fistulas

    * can lead to chronic abscess and fistula
- Perineal Excoriation
- Small bowel Obstruction
  - Adhesion
  - Internal hernia

    **prevent by securing mesentery of pulled-through segment to the retroperitoneum*
  - Intussusception

Early complications



- Perineal excoriation: can occur due to Hypermotile colon. Common but usually resolves after 2-3 mo. Skin will heal with resolution of postop diarrhea. Use of barrier cream starting 1st post op day prevent this complication.

Early complications

- **Wound infection**

- **Cuff abscess / Pelvic abscess** (<7%)
  - ischemia, retained rectal mucosa, bleeding, pelvic contamination, and tension.
  - Treatment ranges from antibiotic and percutaneous drainage to diverting colostomy

- **Anastomotic leak** (1-10%) - Most serious early postop complication
  - tension, ischemia, technical, poor nutritional status, residual aganglionosis/distal obstruction.
  - A sign should be posted on bedside prohibiting any rectal manipulation (thermometer, medication)
  - Can lead to **Pullthrough dehiscence and retraction**
  - Leads to **localized abscess** or **free peritoneal leakage** or may be subclinical and result in **stricture**

Early complications

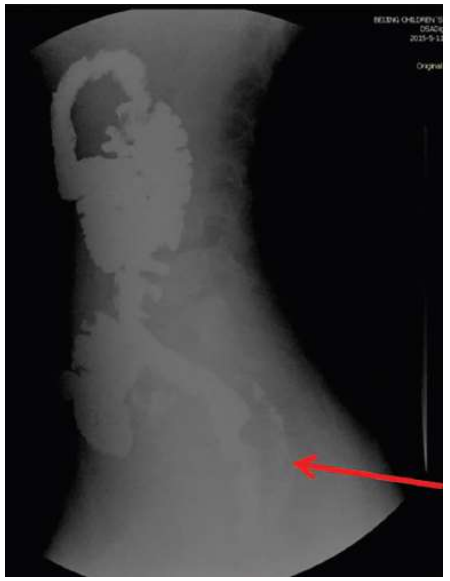
Observational Study

Medicine®

OPEN

STROBE-anastomotic leakage after pull-through procedure for Hirschsprung disease

Chun-Hui Peng, MM, Ya-Jun Chen, MD*, Wen-Bo Pang, MM, Ting-Chong Zhang, MD, Zeng-Meng Wang, MM, Dong-Yang Wu, MM, Kai Wang, MM



- **Anastomotic leak**:. Observational study has suggested older children (>3yr) may be prone to leak. Diagnosis can be made with rectal exam and ultrasound. Contrast study can show contrast extravasation into presacral space. E**arly ileostomy and re-suturing anastomosis** can be used to treat leak. Examination during re-anastomosis revealed dehiscence occurred at 6 o'clock position. Most of these patients didn't require repeat pullthrough.

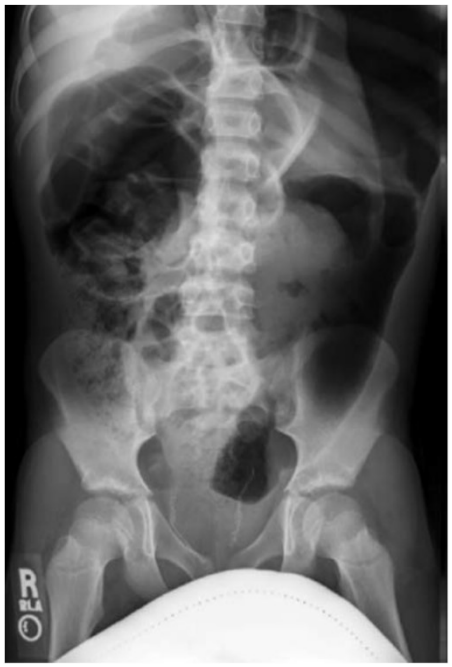
Late Complications

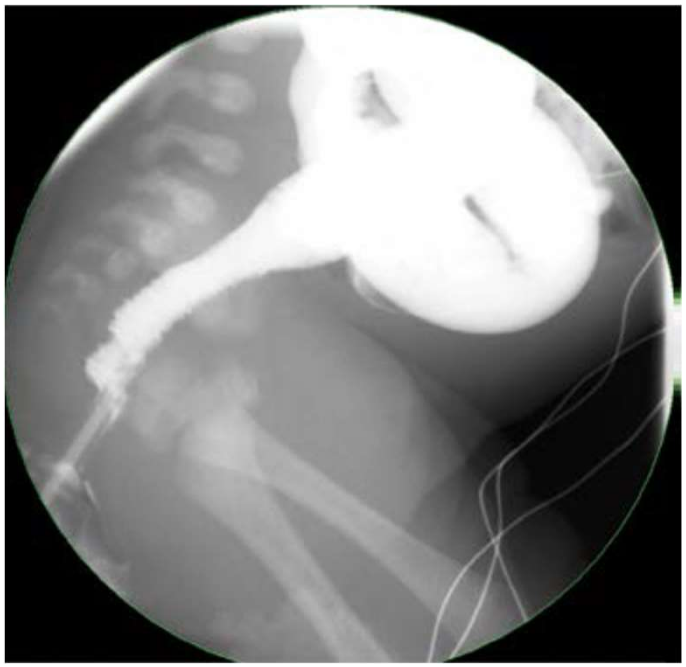
- Most children thrive after pullthrough
- Subset of patients who are not doing well can be grouped into 3. (groups often overlap)
  - Soiling
  - Obstructive symptoms
  - enterocolitis
- Most of these complications **resolve after 5 years** of life
  - *Exception - children with long segment disease and children with Down's syndrome (increased enterocolitis and incontinence)
- **Mortality is low** (under 2%) and mostly associated with HAEC
- candid **discussion with parents**
  - Realistic expectations
  - need for close surveillance for late complications

6.1. Enterocolitis

- most severe and **potentially lethal** complication of HD
- non predictable, **non preventable** complication
- Because there is **overlap between gastroenteritis**, the true incidence is unknown.
- Can occur preop or postoperatively
- Pathophysiology
  - **Fecal stasis** (obstruction) is the most important predisposing factor
  - alterations in **intestinal mucin** production & **mucosal immunogbulin** prod. In HD
  - **No specific pathogen** but Clostridium difficile or rotavirus have been postulated

6.1. Enterocolitis





- A combination of **cut-off sign** (absence of air in distal rectosigmoid) and at least 2 air fluid levels has been strongly associated with HD. **Sawtooth appearance** of rectum on contrast enema consistent with HAEC. A contrast enema should **not** be performed if HAEC is suspected because of the risk of intestinal perforation.

6.1. Enterocolitis

- Risk factors
  - **Pre-op**
    - **delayed** presentation (>1wk),
    - **longer segment** disease,
    - **trisomy 21**
  - **Postop**
    - young age at resection (<6mo),
    - post op **obstruction**,
    - history of **HAEC preop**

6.1. Enterocolitis

- **Prevention** (high risk)
  - **irrigations** (teach parents)
  - chronic administration of **metronidazole**
- **Preop** =
  - IV antibiotic & irrigation
  - colostomy indicated if fail to improve or ischemia/perforation,
- **Postop** = Can occur in 30% without obstruction. Symptoms may start later (after 1 year of age)
  - One attack may be normal if not severe
  - Repeated attacks should raise suspicion of obstruction
  - Consider long term temporary stoma in ongoing chronic entercolitis (persistent, severe)

6.1. Enterocolitis

Criteria	Score
History	
Diarrhea with explosive stool	2
Diarrhea with foul smelling stool	2
Diarrhea with bloody stool	1
Previous history of HAEC	1
Physical examination	
Explosive discharge of gas and stool on rectal examination	2
Distended abdomen	2
Decreased peripheral perfusion	1
Lethargy	1
Fever	1
Radiology	
Multiple air fluid levels	1
Dilated loops of bowel	1
Sawtooth appearance with irregular mucosal lining	1
Cut-off sign in the rectosigmoid with absence of distal air	1
Pneumatosis	1
Laboratory	
Leukocytosis	1
Shift to the left	1
Total	**20**
HAEC	**≥10**

Source: Adapted from Pastor AC et al. *J Ped Surg* 2009; 44: 251–256.

- Langer: HAEC score (2009)

6.1. Enterocolitis

APSA categorization	Typical presenting symptoms	Typical radiographic features	Recommended treatment (do all of the following)	Possible additional measures
Possible HAEC (Grade I)	Anorexia, diarrhea, mild abdominal distention	Normal radiograph, or mild signs of ileus	▪ Oral hydration ▪ Oral metronidazole	▪ Rectal irrigations
Definite HAEC (Grade II)	One or more of the following:* ▪ Explosive diarrhea ▪ Fever, tachycardia, or lethargy ▪ Moderate abdominal distention and/or tenderness ▪ Explosive gas/stool on rectal examination	May include: ▪ Signs of ileus, including air-fluid levels and dilated loops of bowel ▪ Distension of the proximal colon, with rectosigmoid cutoff¶	▪ Clear liquids or hold feeds ▪ IV hydration ▪ Metronidazole (oral or IV) ▪ Broad spectrum antibiotic coverage△ (in addition to metronidazole) ▪ Rectal irrigations	▪ Nasogastric decompression◇
Severe HAEC (Grade III)	Symptoms of Grade II (above), PLUS: ▪ Obstipation ▪ Poor perfusion ▪ Hypotension ▪ Altered consciousness/mentation ▪ Marked abdominal distension ▪ Signs of peritonitis	Signs of Grade II (above), PLUS possible: ▪ Pneumatosis intestinalis ▪ Pneumoperitoneum (rare)	▪ Hold feeds ▪ Metronidazole (IV), AND ▪ Broad-spectrum IV antibiotics△ ▪ Rectal irrigations	▪ Nasogastric decompression◇ ▪ Possible surgical intervention§

Adapted from Gosain A, Frykman PK, Cowles RA, et al. Guidelines for the diagnosis and management of Hirschsprung-associated enterocolitis. Pediatr Surg Int 2017; 33:517.

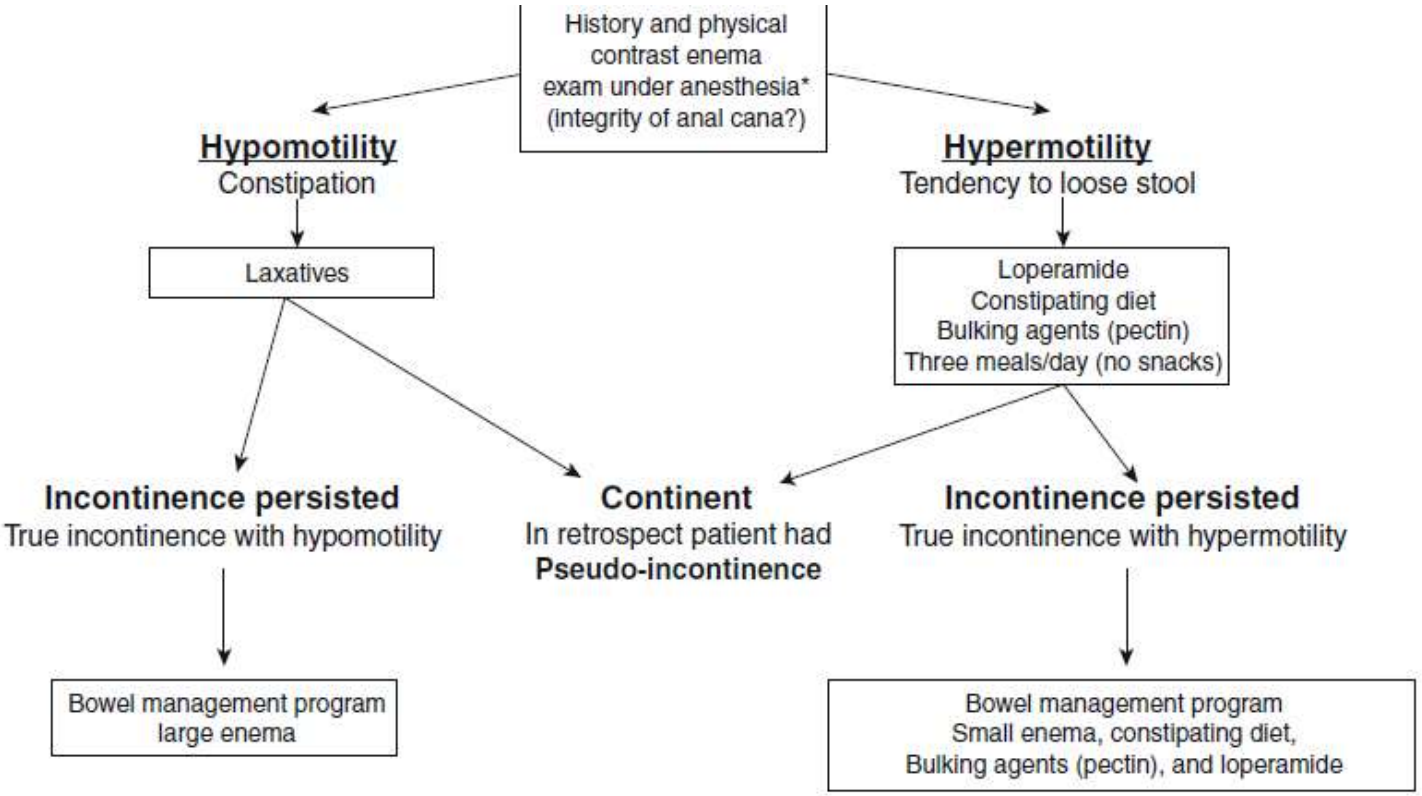
- APSA: HAEC guideline (2017)

6.2. Soiling

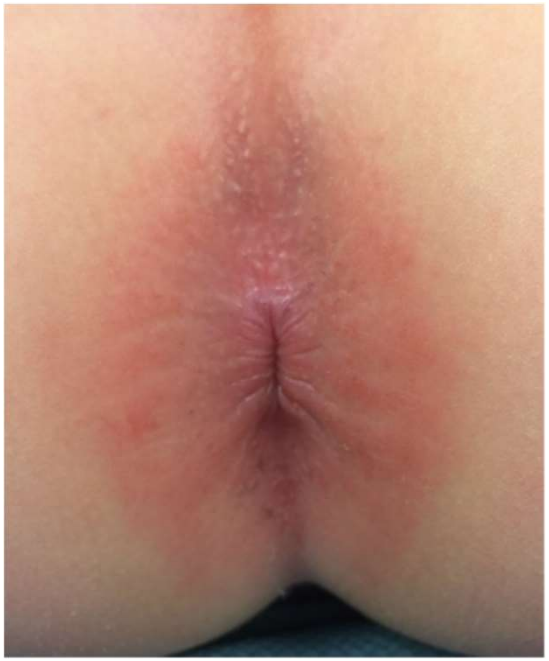
Most children suffer from overflow pseudo-incontinence

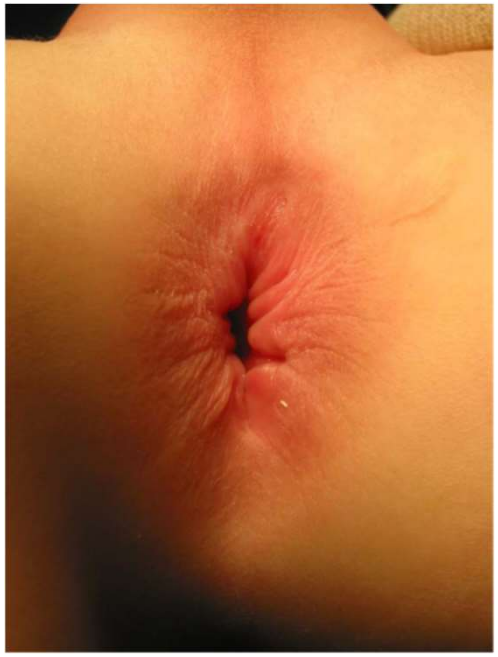
- **Examination under anesthesia**
    - asses sphincter tone
    - asses weather the anus/dentate is intact or not (failure to detect d/f b/n gas and stool)
- **Contrast enema**
    - Hypermotile colon
        - b/c rectosigmoid reservoir is resected (More in soave)
    - Hypomotile colon
        - if dilated normoganglioonic bowel is left (More with Duhamel)
- **Anorectal manometry**
    - lack of sensation of a full rectum (in overflow pseudoincontinence)
- **Colonic motility studies**

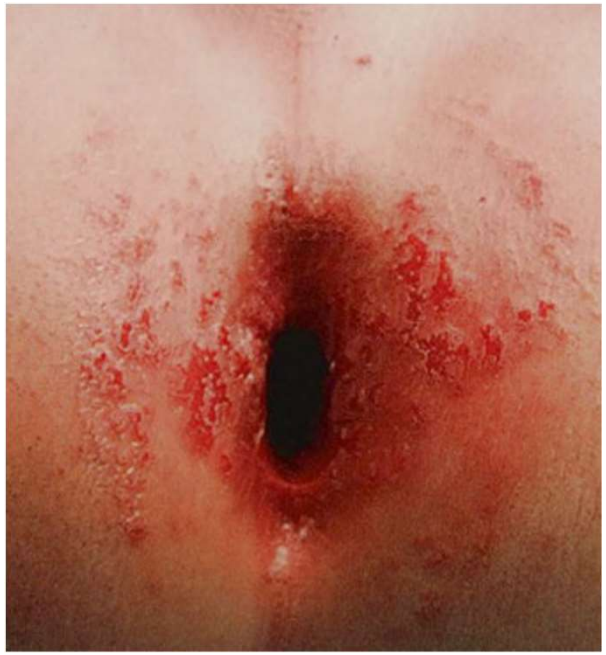
6.2. Soiling


History and physical
contrast enema
exam under anesthesia*
(integrity of anal cana?)
Hypomotility
Constipation
Laxatives
Hypermotility
Tendency to loose stool
Loperamide
Constipating diet
Bulking agents (pectin)
Three meals/day (no snacks)
Incontinence persisted
True incontinence with hypomotility
Continent
In retrospect patient had
Pseudo-incontinence
Incontinence persisted
True incontinence with hypermotility
Bowel management program
large enema
Bowel management program
Small enema, constipating diet,
Bulking agents (pectin), and loperamide

6.2. Soiling

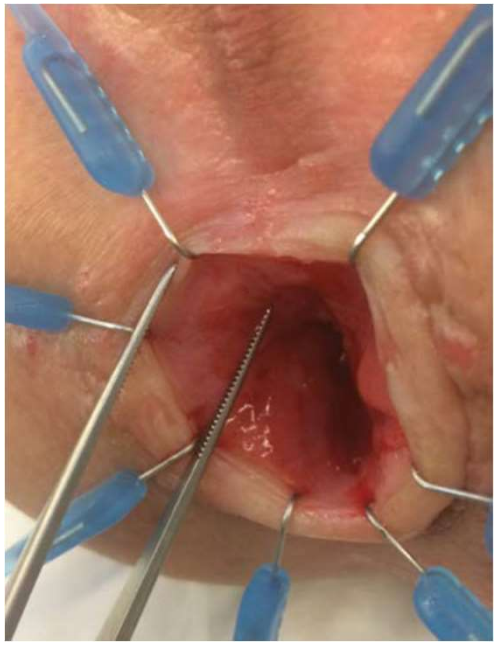


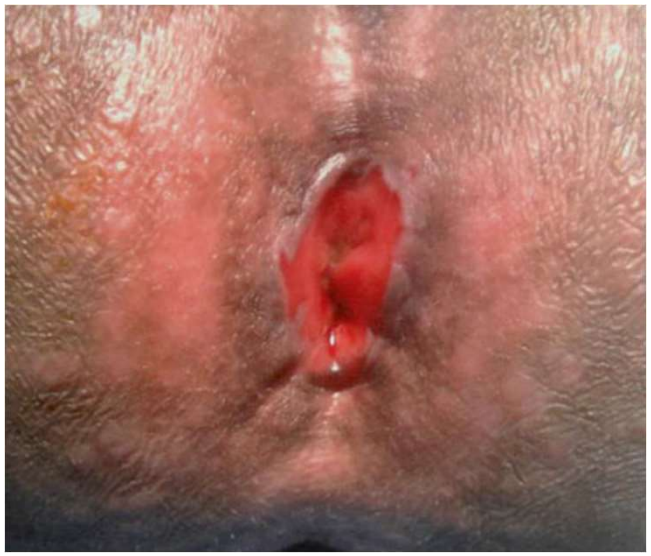




- EUA and evaluation of Sphincter tone: very patulous anus can occur and is indicative of overstreching of the sphincter duiring pulthrough.

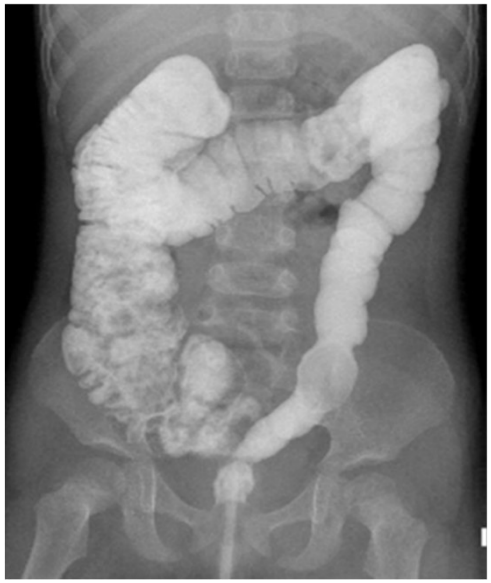
6.2. Soiling

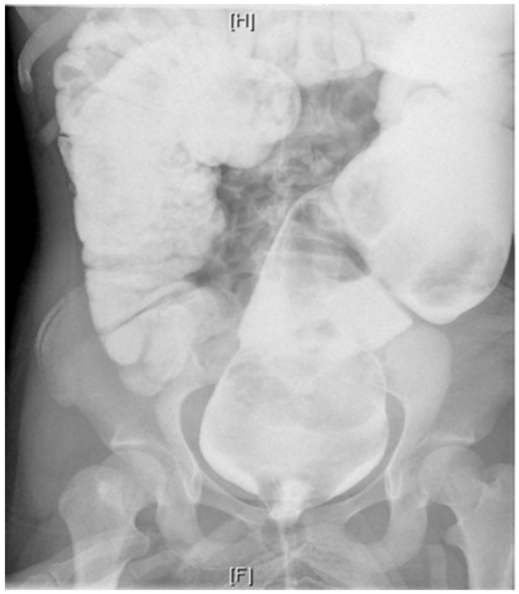




- **EUA and evaluation of dentate line:** partially intact dentate as indicated by the forceps but dentate line is absent at the 6 o'clock position. This might be enough to allow for normal anal canal sensation. Totally absent dentate line with bowel skin anastomosis will render patient incontinent.

6.2. Soiling



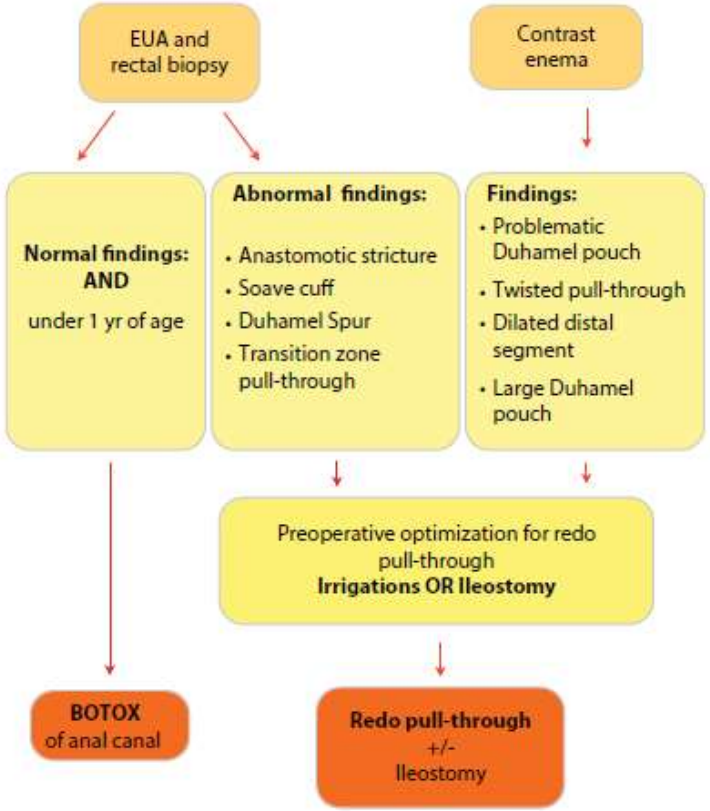


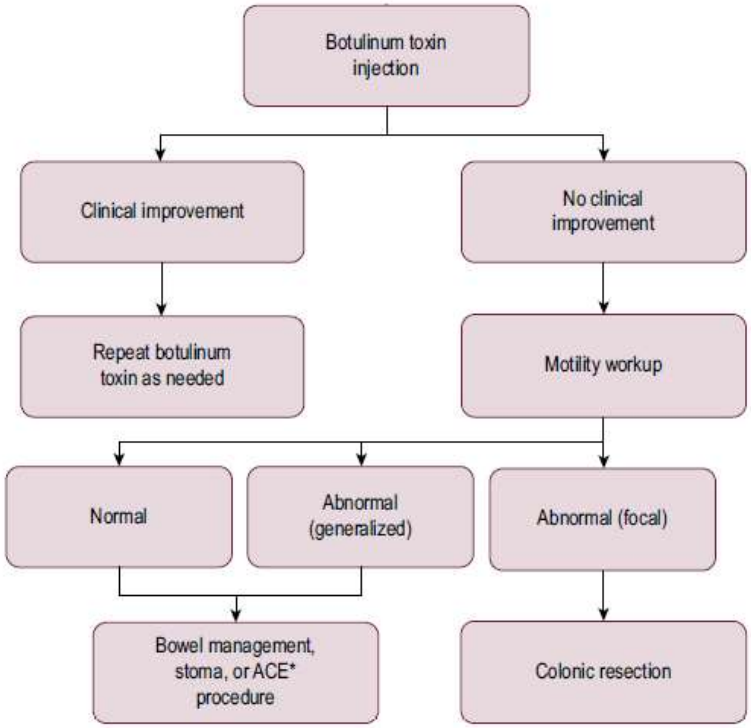
- **Contrast enema**: Hypermotile and Hypermotile colon need to be differentiated and management can be commenced. Sometimes children with constant soiling and severe excoriation which is unresponsive to bowel management may need colostomy which will be closed after age of 5 yr.

6.3. Obstruction

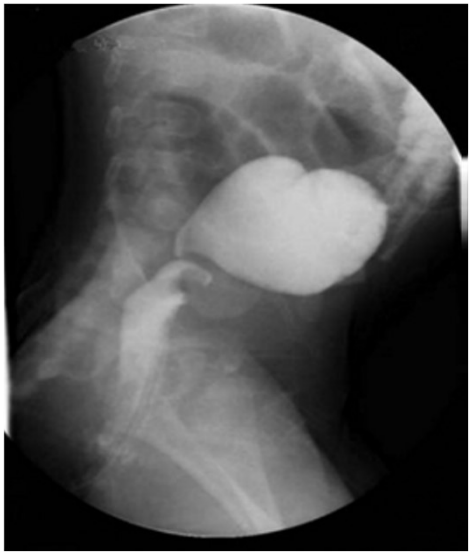
- Children with HD should be able to evacuate their bowels after pull through
- Present with **constipation, abdominal distention, FTT**
- recurrent **enterocolitis** should raise concern with the pull-through.
- Requires investigation as per the HD algorithm
  - contrast enema
  - EUA
  - Rectal biopsy
  - Botox trial

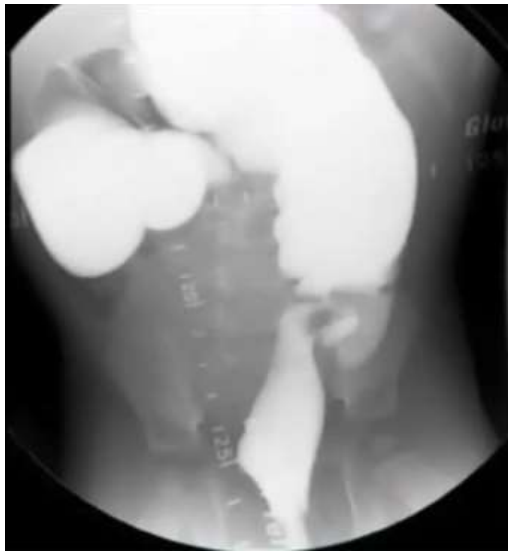
6.3. Obstruction





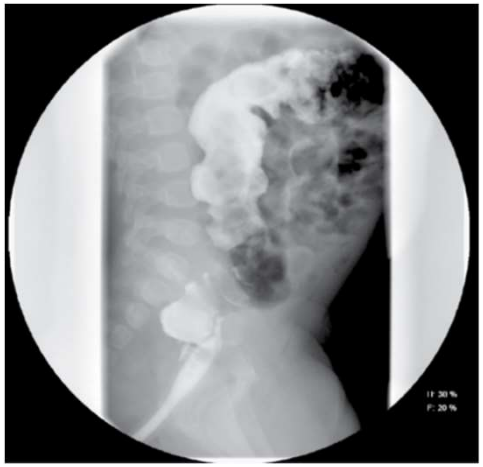
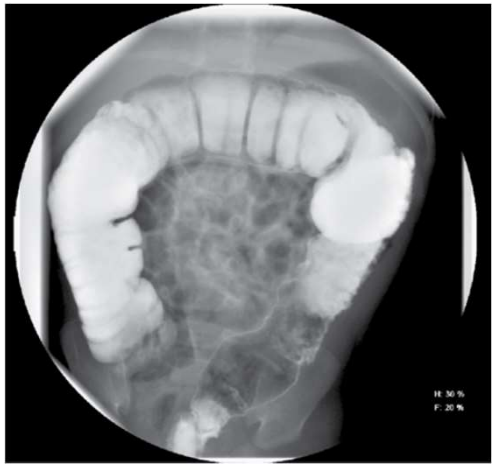
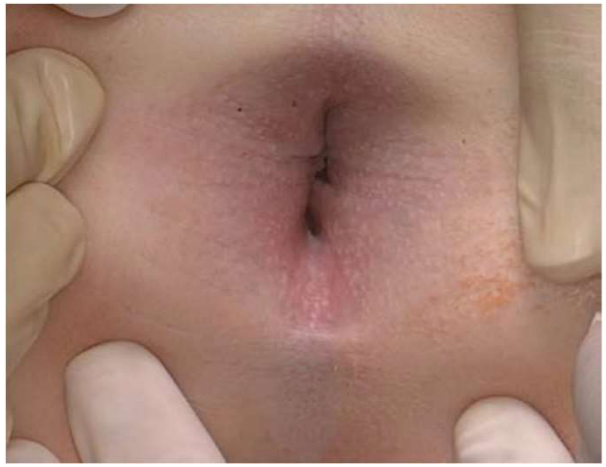
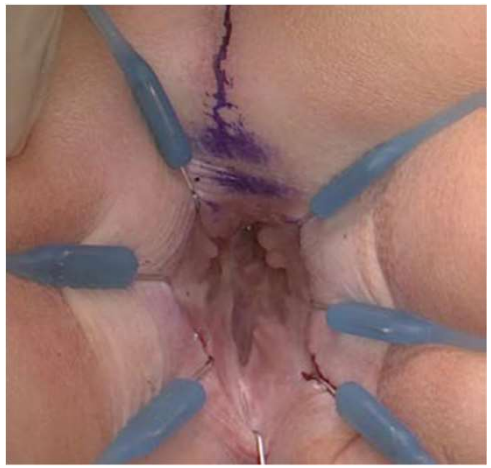
6.3.1. Mechanical Obstruction





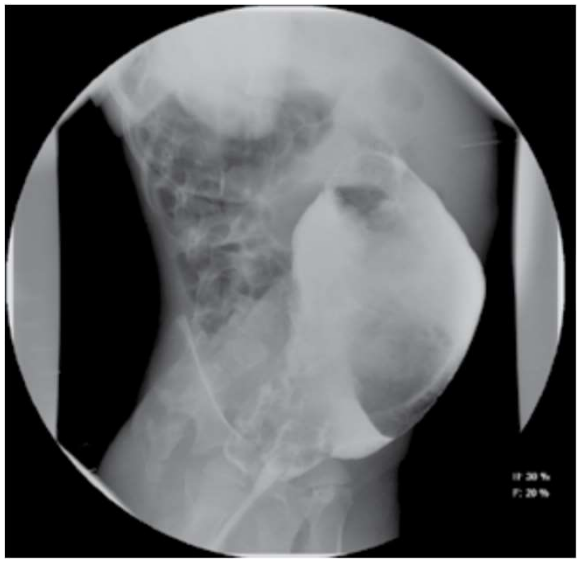
- **Twisted pullthrough**: Is especially a problem in **transanal** pullthrough. To prevent this complication a distinct stich should be used to mark either 12 or 6 o'clock once rectum is detached. A hegar can also be used at the end of procedure to check weather it passes easily. Diagnosing twist on contrast enema might be difficult if it is not complete (<360 degree) and the obstruction may be intermittent. May be detected on PR with bimanual palpation.

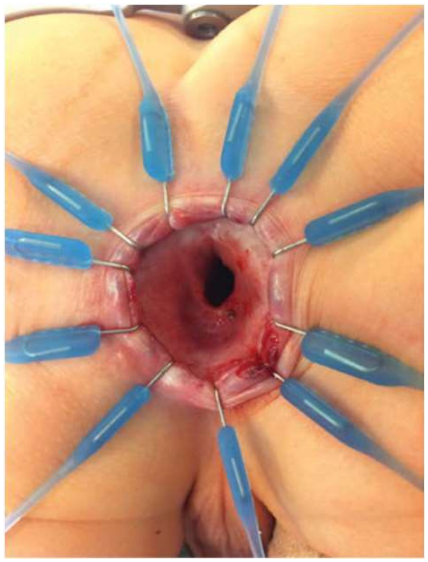
6.3.1. Mechanical Obstruction

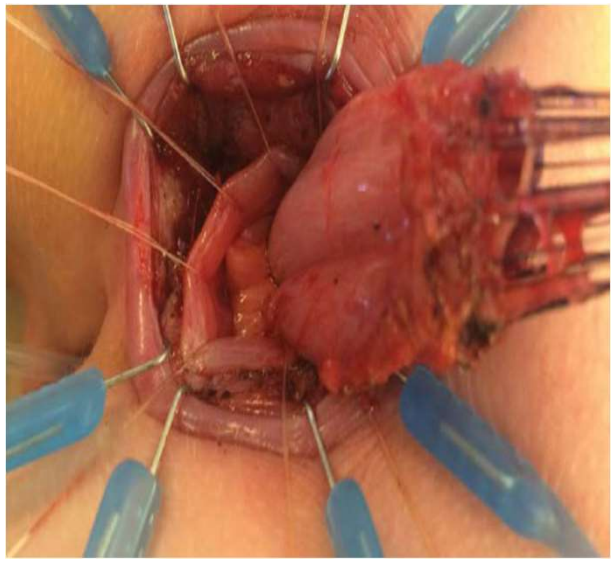





- **Anastomotic stricture** (8-24%) Most common complication after **swenson**/soave. Narrowing of the distal segment with proximal dilatation suggest stricture. irregularity of the left colon suggest enterocolitis. Intra-operative image showing extensive stricture. Stricture is a result of poor surgical technique and frequently ischemia of the bowel or excessive tension. Repeated dilatation using a finger, dilator, or balloon. Intralesional steroid, or topical mitomycin C can be tried. Posterior sagittal incision and even laparotomy may be required to resect scarred rectum with redo pullthrough.

6.3.1. Mechanical Obstruction

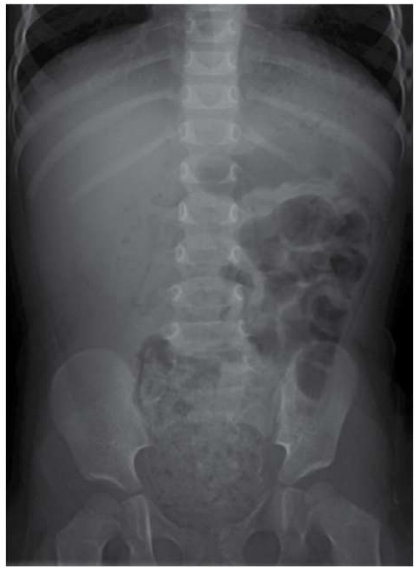


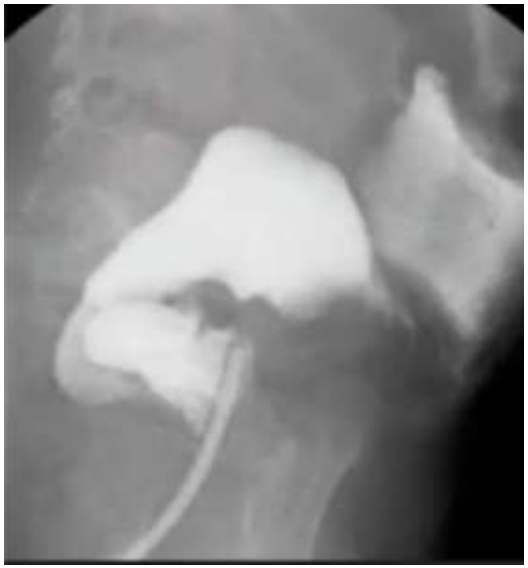


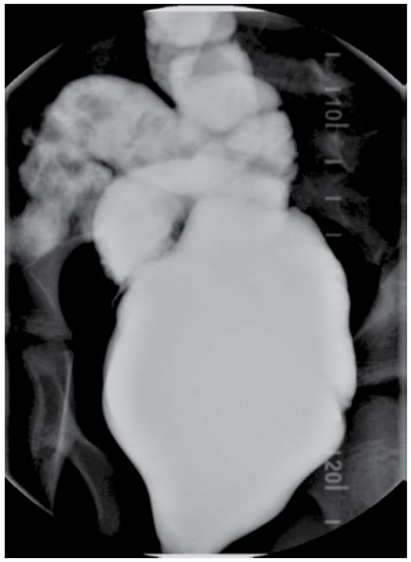


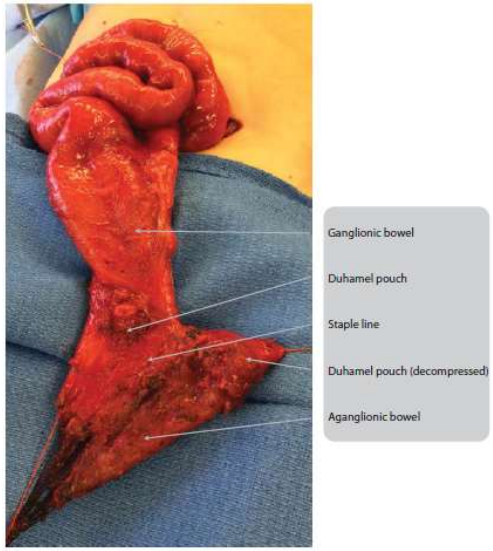
- **Retained Soave cuff (long/narrow)**: Lateral view of contrast image shows excess widening of the pre-sacral space. On digital examination, the boy was found to have a circumferential rubbery ring along the hollow of the sacrum consistent. Cuff can be divided via laparoscopy occasionally but a redo pullthrough is usually required. Transanal view showing extrinsic compression from Soave cuff. The soave cuff was identified and removed. pullthrough then sutured in place.

6.3.1. Mechanical Obstruction



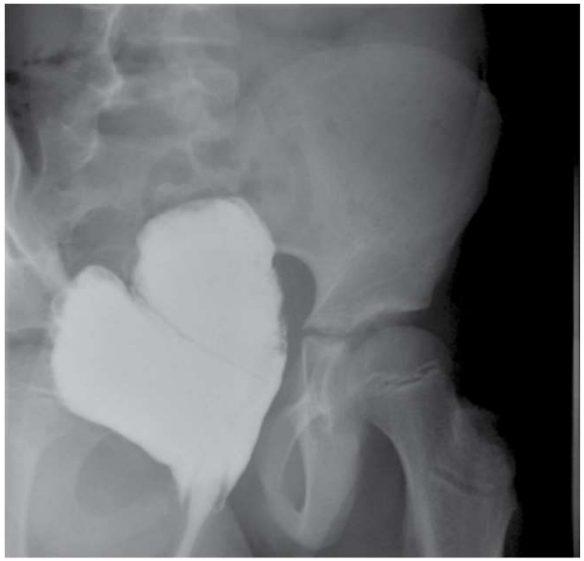


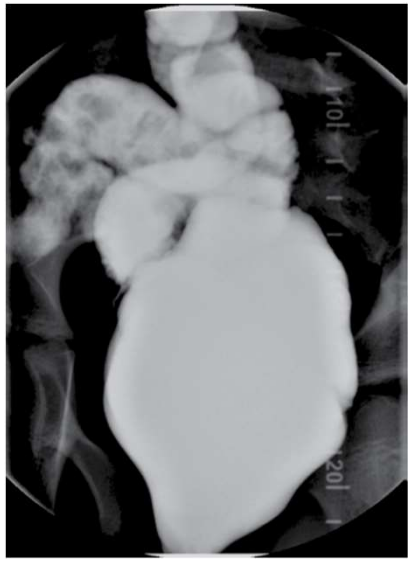


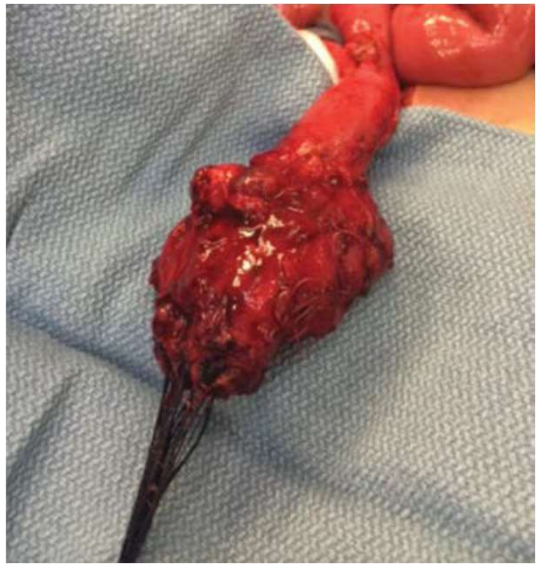


- **Duhamel Spur**: Duhamel pullthrough demonstrated in plain film (staple line in pelvis) and contrast enema (two rectal compartments). If the native rectum is not in continuity with pullthrough rectum, stool accumulates anteriorly and pushes on the pullthrough which is failing to empty. Early spur occurs if rectum above staple line hasn't been resected adequately or if there is fusion of two staple lines. Spurs can occur later if constipation occurs with mega Duhamel pouch. Excision of the spur (laparascopy/transanal-stapler) or removal of the pouch (redo pullthrough- difficult) are two options for treatment.

6.3.1. Mechanical Obstruction

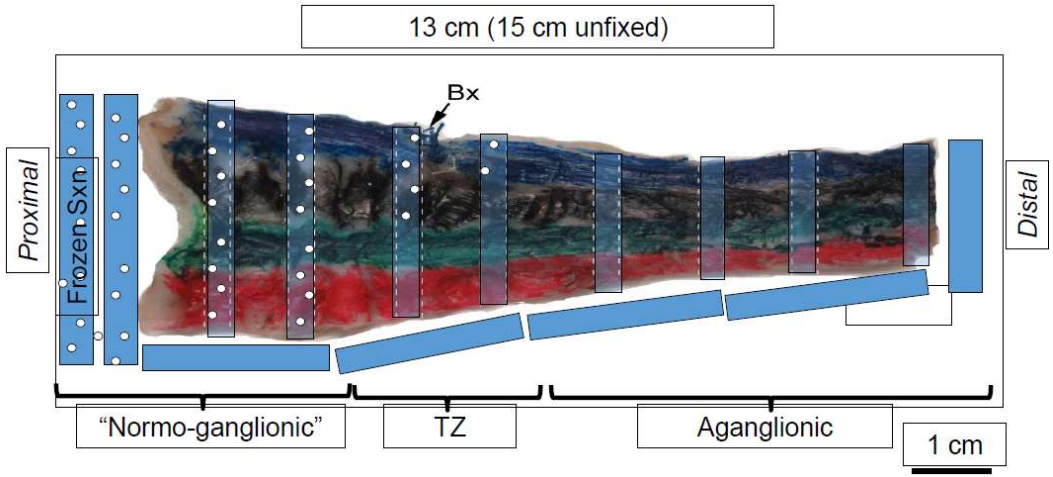






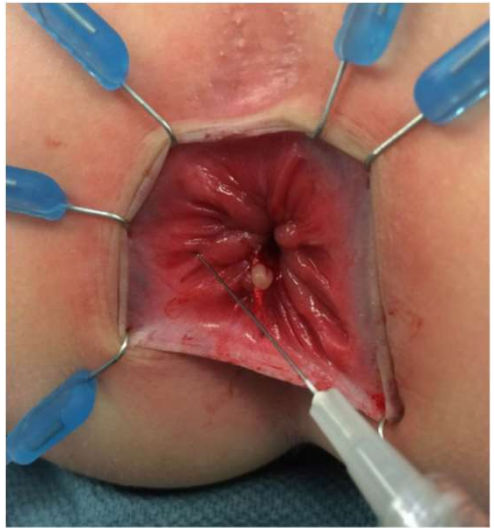
- **Mega Duhamel pouch**: No spur was identified. We felt the pouch itself was the cause of the obstruction. This his child underwent a redo pull-through procedure involving resection of the Duhamel pouch.

6.3.2. Aganglionic/Transition zone pullthrough



- **Rectal biopsy**: If mechanical obstruction has been ruled out the next step is to take biopsy above colo-colic anastomosis. The specimen from the original operation should also be reviewed. Transition zone pullthrough can occur because difficult to diagnose(circumferential distribution of ganglion cells in TZ is uneven). Some surgeons specify for pathologists to examine 4 quadrants from the proximal end. An error in histologic analysis can also miss aganglionic bowel. Another possible cause is acquired aganglionosis. Most require a redo-pullthrough.

6.3.2. Internal sphincter Achalasia



- **Botox trial**: All children with HD have achalasia. Botox should be tried if mechanical obstruction and residual aganglionosis ruled out. Inject into the external sphincter to help reduce anal spasms, which are thought to contribute to failure to evacuate stool in those under 2 years of age. Inject circumferentially at the dentate but avoid anterior part not to go too deep into periurethral tissue. If patient responds, repeat sessions are needed as they wear off (3-6mo) or application of nitroglycerine paste/topical nifedipine. Most resolve by 5 yr. If no response, repeat session may be attempted before ruling out achalasia.

6.3.4. Motility disorder

Most children with HD have abnormal GI motility including GER and delayed gastric emptying

- Work up should be done if not responding to botox injection.
  - radiologic shape study
  - radionuclide colon transit study
  - colonic manometry
  - Laparoscopic biopsies (for IND)
- **Focal dysmotility (Lt colon)** resection with repeat pull-through using normal bowel is needed.
- **Diffuse dysmotility** is best treated with bowel management

6.3.5. Functional constipation (megacolon)

- consider stool withholding after all causes ruled out
- Start bowel management (laxative, fiber, preferably without enema)
- In severe cases, consider long term temporary stoma (? 5yrs, ? adolescence)

7. Variant HD

- Internal neuronal dysplasia
- Hypoganglionosis
- Internal sphincter achalasia
- Ultra-short segment HD
- Desmosis Coli

Variant HD

- clinical picture suggestive of HD, but with ganglion cells on rectal biopsy.
- controversy surrounding the definitions and existence of many of these conditions

7.1. Intestinal neuronal ysplasia

- Type A= **diminished or absent sympathetic innervation** of the myenteric and submucosal plexuses, along with hyperplasia of the myenteric plexus.

- Type B = **dysplasia of the submucous plexus** with thickened nerve fibers and giant ganglia, increased AChE staining, and identification of ectopic ganglion cells in the lamina propria

- Debate weather histologic changes are secondary to chronic obstruction or the cause of it

7.2. Hypoganglionosis

- sparse and small ganglia, often associated with abnormal acetylcholinesterase distribution
- must be differentiated from immature ganglia, which are seen in preterm children
- Treatment is pullthrough

7.3. Internal sphincter Achalasia

- Normal ganglion cells on rectal biopsy, but have absence of rectoanal inhibitory reflex
- constipation associated with this condition usually improves over the first five years
- First step is diet, laxatives, and enemas or irrigations.
- chemical sphincterotomy (botulinum, nitroglycerine or topical nifedipine)
- Some advocate anal sphincter myectomy. (concern of sphincter damage)

7.4. Ultrashort segment HD

- documented aganglionic segment of <3–4 cm.
- Findings of hypertrophic nerves and abnormal cholinesterase staining may be absent.
- Some use the term interchangeably with internal sphincter achalasia
- Treatment is also controversial
  - ? nonoperative long-term management (enemas and laxatives).
  - ? simple myectomy (risks include incontinence, fistula and abscess)
  - pull-through (recommended by most)

7.4. Desmosis coli

- Rare
- total or a focal lack of the connective tissue in bowel wall, without any abnormalities in the enteric nervous system.

References

- Jacob Langer, **Hollcomb and Aschcraft's pediatric surgery**, 2020
- Victoria Lane, Richard Wood, **Pediatric colorectal and pelvic surgery**, 2017
- Alberto Pena, **Surgical treatment of colorectal problems in children**, 2016