Hirschprung Disease

HOLCOMB AND ASHCRAFT'S PEDIATRIC SURGERY

Definition

- characterized by the absence of ganglion cells in the myenteric and submucosal plexuses of the intestine. This cell are responsible for normal peristalsis, patients will present with functional intestinal obstruction at level of obstruction.
- Incidence 1:5000 live births.
- 80% have a transitional zone in rectum or rectosigmoid colon.
- 10% more proximal colonic involvement.
- 5-10% total colonic aganglionosis with variable involvement of small intestine.
- Rarely near total intestine aganglionosis.

Etiology

- Ganglion cells are derived from the neural crest.
- By 13 weeks postconception, the neural crest cells have migrated from proximal to distal through the gastrointestinal tract, after which they differentiate into mature ganglion cells.
- **First theory,** possibility that the neural crest cells never reach the distal intestine due to early maturation or differentiation into ganglion cells.
- **Second theory**, possibility that the neural crest reach their destination, but fail to survive or differentiate into ganglion cells.

Etiology

- HD is a heterogenous group of diseases with multiple genetic causes and etiologies. The gene identified is
  - RET proto-oncogene(early neuronal cell death),
  - endothelin-3 receptor and endothelin-B receptor(abnormalities of neural crest-derived tissues, eg Shah-Waardenberg syndrome, central hypoventilation syndrome),
  - SOX-10 genes(early maturation),
  - S1P1(now known as ZFHX1B)
  - Phox2B

Box 34.1 Congenital Anomalies and Conditions Commonly Associated With Hirschsprung Disease

Down syndrome (trisomy 21)
Neurocristopathy syndromes
- Waardenberg–Shah syndrome
- Yemenite deaf-blind-hypopigmentation
- Piebaldism
- Other hypopigmentation syndromes

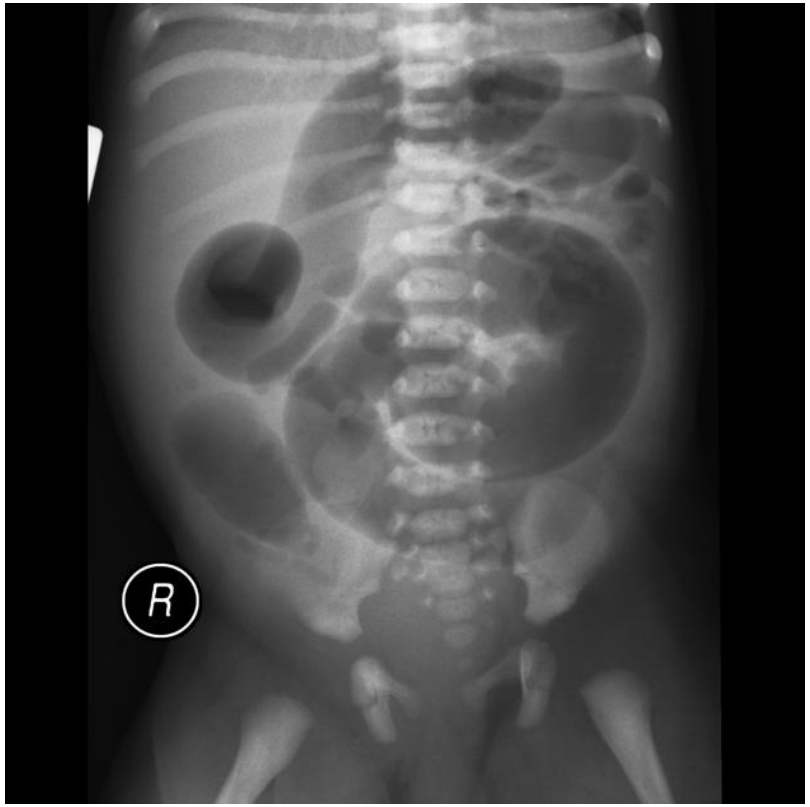
Goldberg–Shprintzen syndrome
Smith–Lemli–Opitz syndrome
Multiple endocrine neoplasia 2
Congenital central hypoventilation syndrome (Ondine's curse)
Isolated congenital anomalies
- Congenital heart disease
- Malrotation
- Urinary tract anomalies
- Central nervous system anomalies
- Other

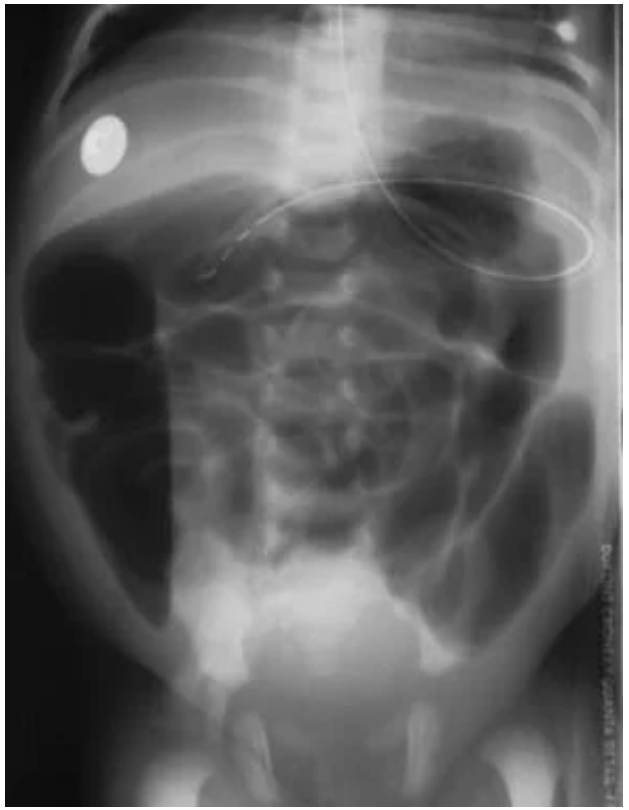
Clinical presentation

- **Neonatal period (50-90%)**
  - Delay passage of meconium beyond the first 24 hours. (90%)
  - Abdominal distention
  - Bilious vomiting
  - Feeding intolerance
  - Cecal or appendiceal perforation
  - Hirschsprung-associated enterocolitis(HAEC) : fever, abdominal distention, diarrhrea (10%)
- **Childhood period**
  - Severe chronic constipation
  - Delayed passage meconium at birth 48 hrs
  - Failure to thrive
  - Abdominal distention
  - Dependence on enema w/o significant encopresis
  - enterocolitis

Imaging : Plain radiograph

- Dilated bowel loops throughout the abdomen.





Imaging : Contrast enema

- First step : water-soluble contrast enema
- Pathognomonic finding is a **transitional zone** between the normal and aganglionic bowel.
- 10% may not seen TZ, false-negative
- Plain radiograph 24hr later, retention of enema maybe suggestion HD.

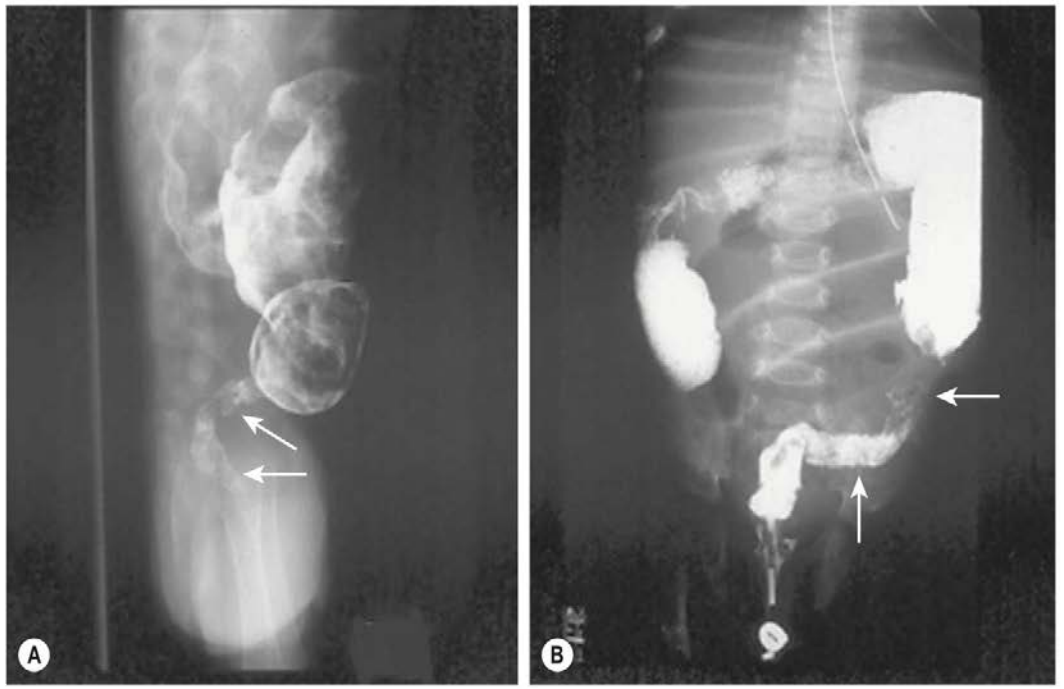


Fig. 34.1 **(A)** and **(B)** represent contrast enema examinations in different infants demonstrate Hirschsprung disease. The aganglionic rectum (arrows in both studies) is small and contracted. The proximal ganglionic colon is dilated. A transition zone between the aganglionic and ganglionic colon is nicely seen in both studies.

Imaging : Contrast enema

- Reverse of rectosigmoid ratio

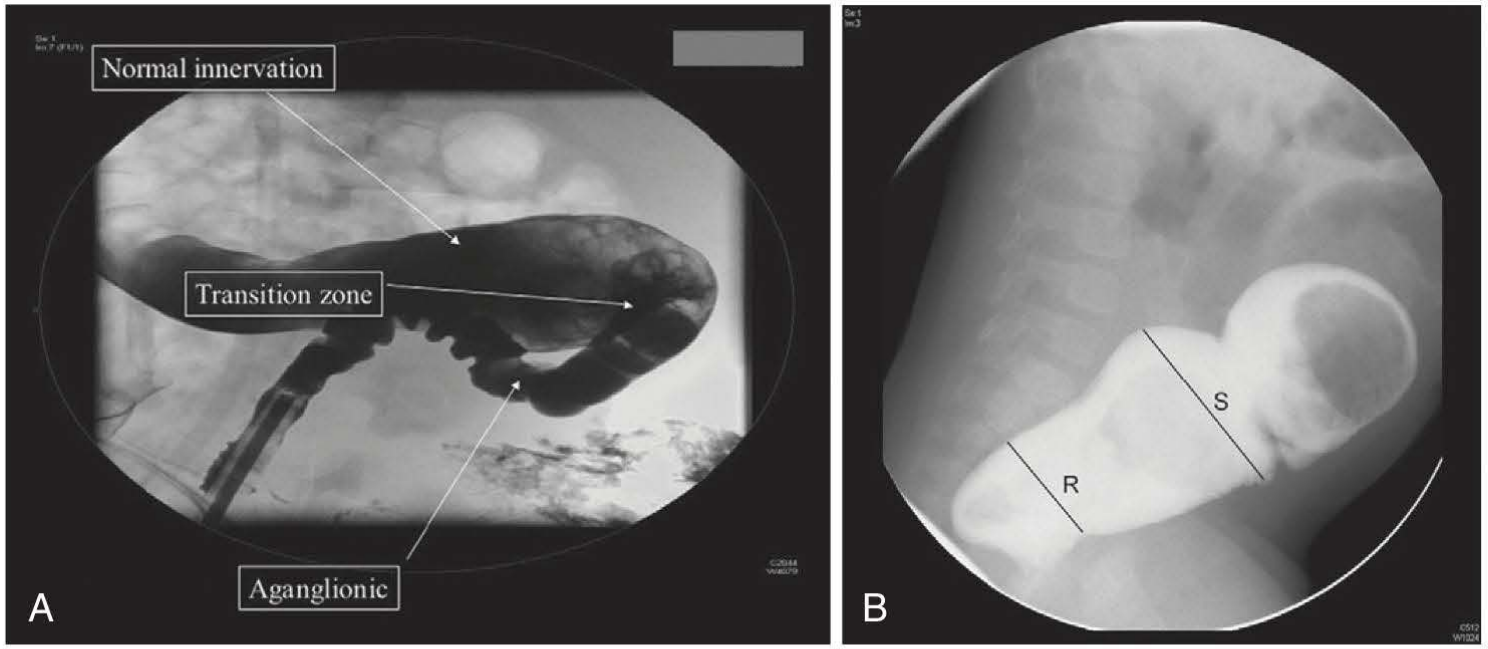


FIGURE 101-1 **A,** Water-soluble contrast enema demonstrating a transition zone at the splenic flexure. **B,** The lateral view is the most important one to identify a low transition zone. In this case the recto-sigmoid index, consisting of the ratio of rectal diameter (*R*) to sigmoid diameter (*S*), is less than 1.0. D. Retention of contrast on a 24-hour postevacuation film.

Diagnosis : Anorectal manometry

- Screening technique
- **Presence of a recto-anal inhibitory reflex(RAIR)**, consisting of reflex relaxation of the internal anal sphincter in response to balloon dilation of rectum, essentially rule out HD.
- Absence of RAIR may represent a false-positive, rectal biopsy should be followed.
- Not widely available for neonates and is often operator dependent, is easily in older children p/w chronic constipation.

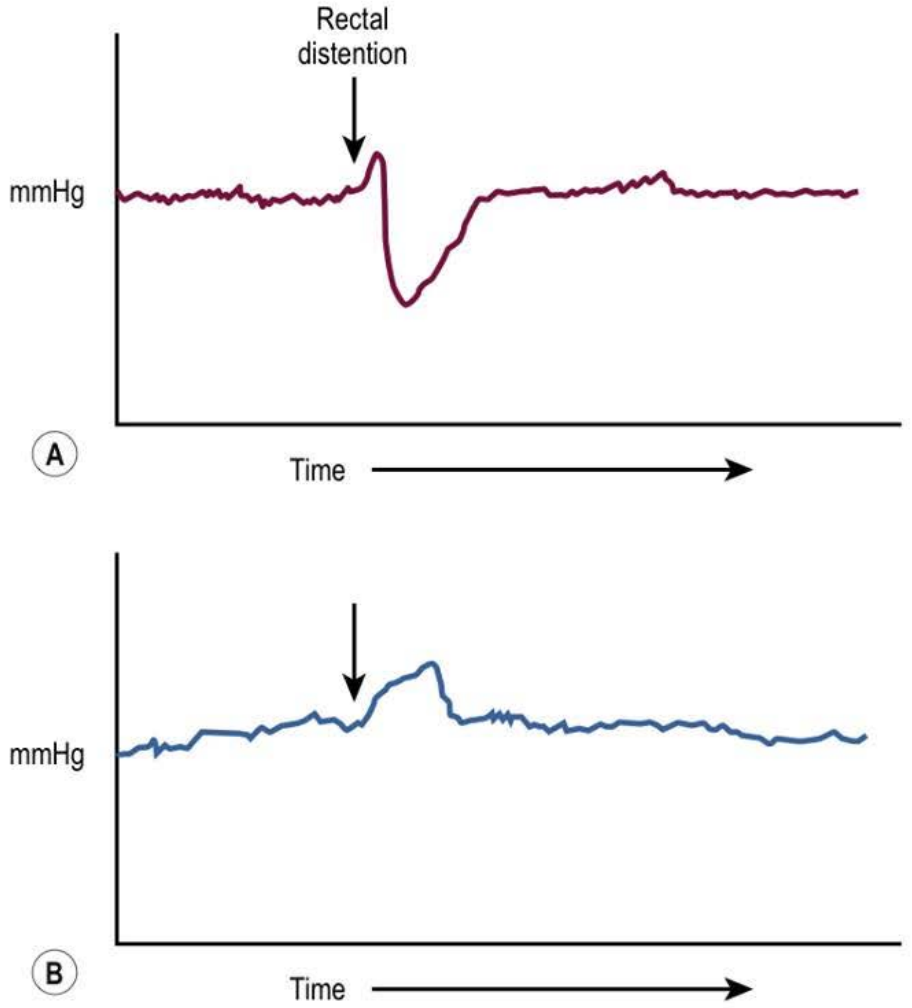


Fig. 34.3 (A) In the child without Hirschsprung disease undergoing anorectal manometry, the recto-anal inhibitory reflex is normal. Note the drop in the internal sphincter pressure with rectal distention. **(B)** A child with Hirschsprung disease is seen to have abnormally increased contraction of the anal canal and no relaxation of the internal sphincter with rectal distention. (The arrow points to the initiation of rectal distention in both **A** and **B**.)

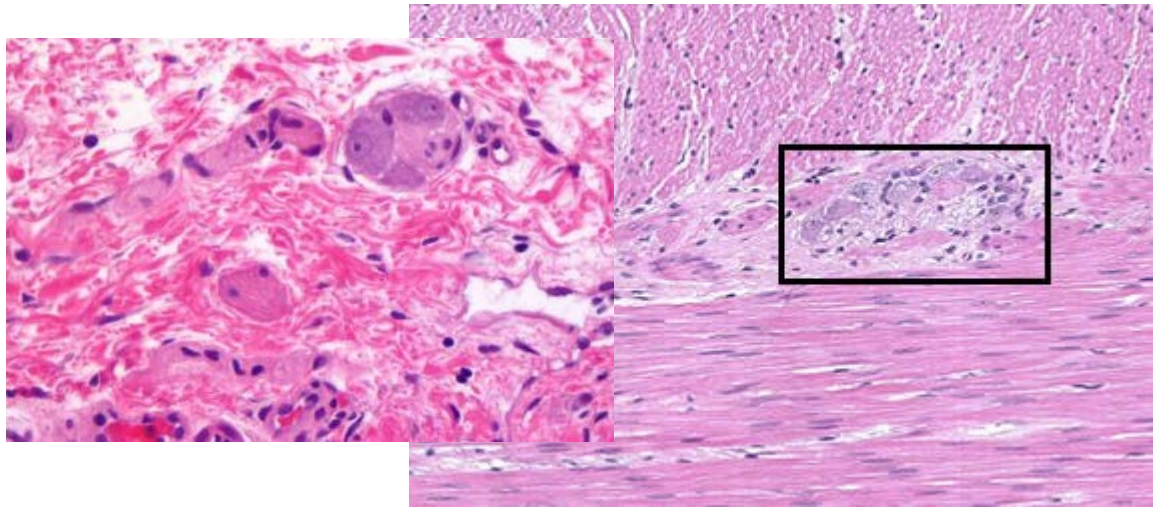
Diagnosis : Rectal biopsy

- Goal standard : absence of ganglion cells in the submucosal and myenteric plexuses on histological examination
- Rectal biopsy
  - Suction rectal biopsy :
    - low risk of perforation or bleeding,
    - less liable in older children due to sampling error cause the mucosa is too thick.
  - Full-thickness rectal biopsy :
    - Provide more tissue and deeper levels.
- Normally there is a paucity of ganglion cells in the area 0.5-1.0 cm above dentate line, the rectal biopsy should be taken >= 1.0-1.5 cm above it.
- Early rectal biopsy in preterm is not recommended due to :
  - The pathologist may have difficult recognizing ganglion cell due to their maturity.
  - May be difficult to obtain enough tissue without increasing rick of complication.

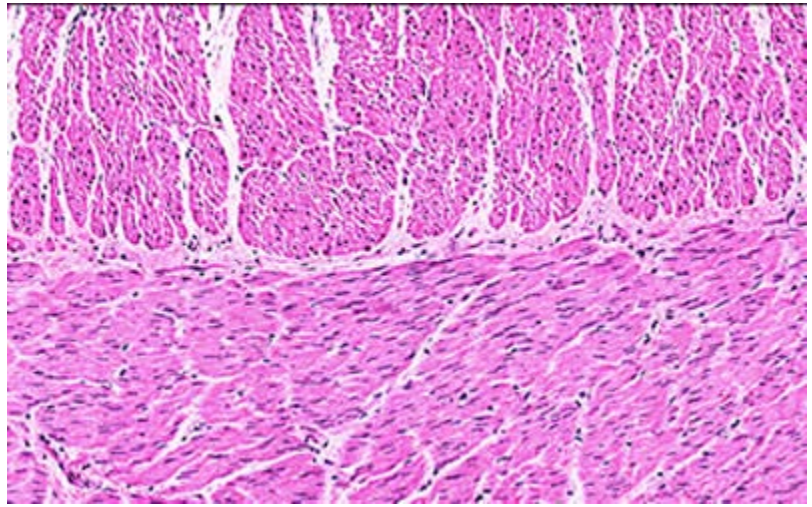
Histology

- **Absence of ganglion cells** in the submucosal and myenteric plexuses on histological examination

normal ganglion cell

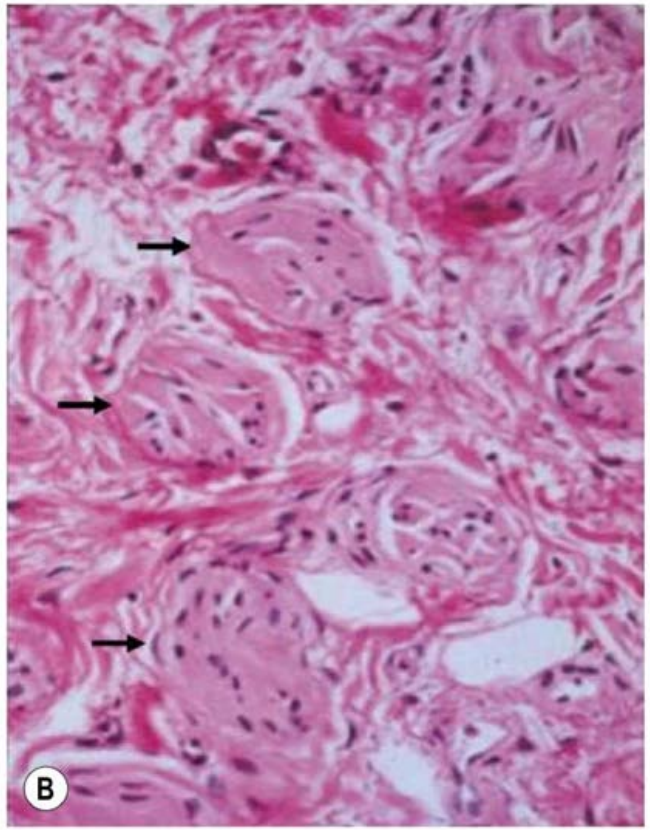


aganglion cell



Histology

- Evidence of hypertrophied nerve trunks.
  - Total colonic disease
  - Short segment



Histology

- H&E stain : ***stain for acetylcholinesterase***, which has a characteristic pattern in the submucosa and mucosa in children with HD.

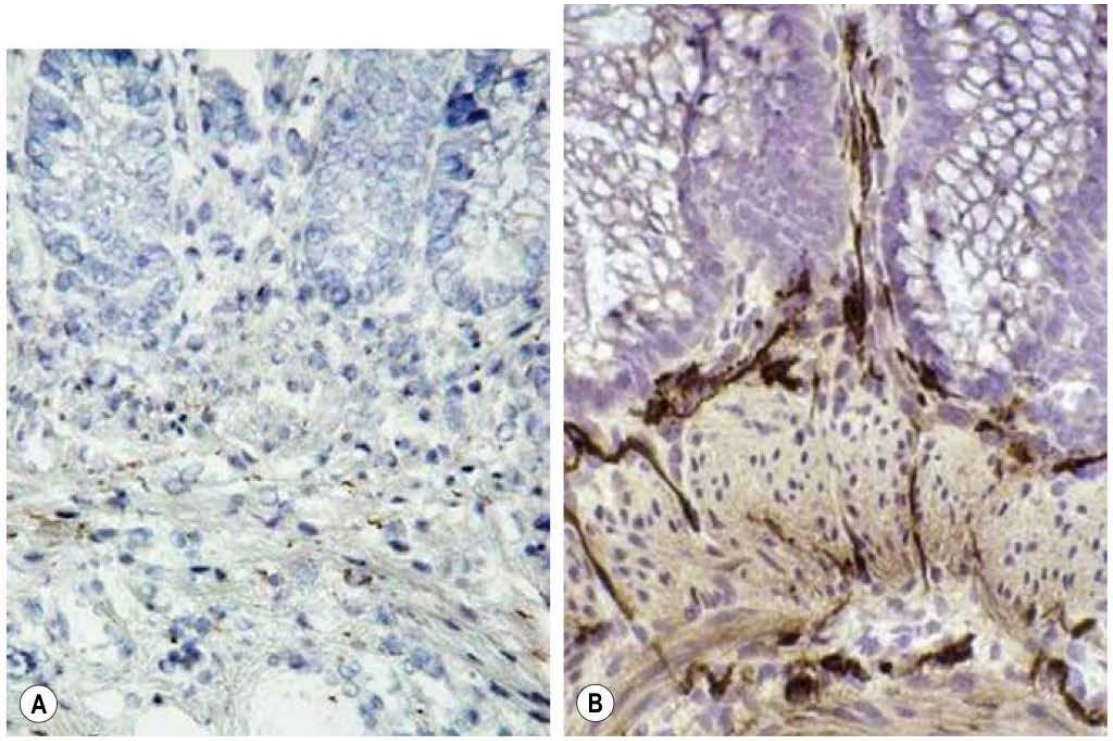


Fig. 34.5 Cholinesterase staining in **(A)** normal colon and **(B)** colon affected by Hirschsprung disease.

Histology

- ***Absent stain for calretinin***, which is almost always absent in patient with HD.

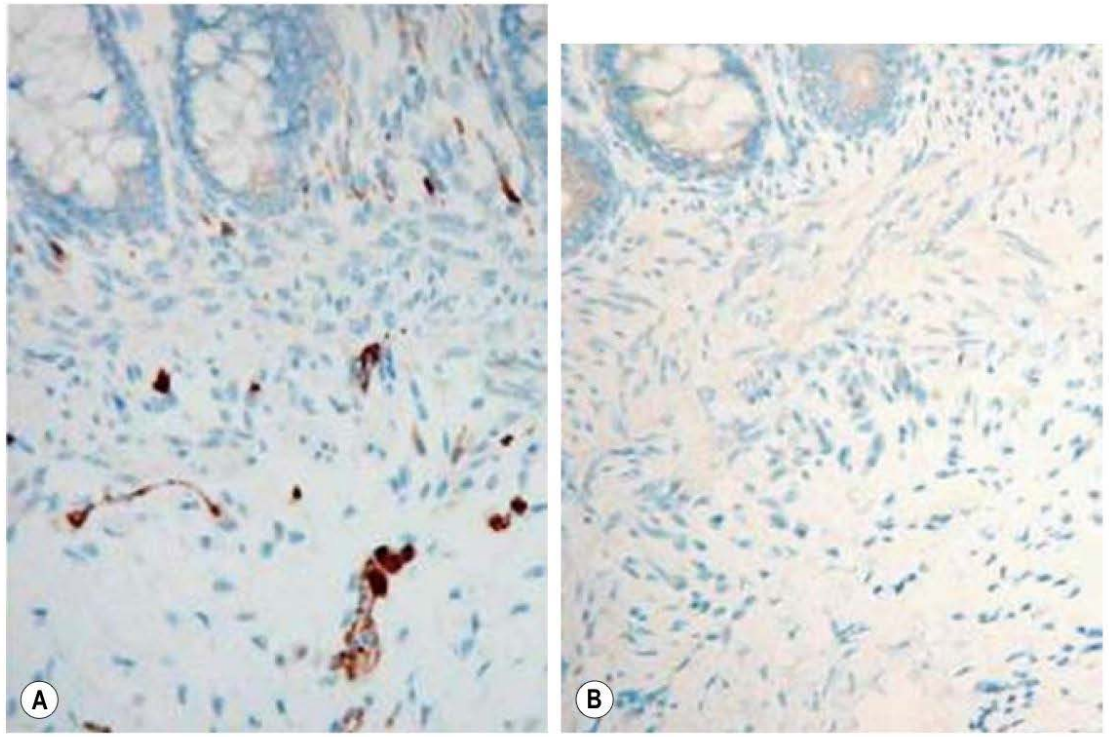


Fig. 34.6 Calretinin staining is seen in **(A)** normal colon but not in colon **(B)** affected by Hirschsprung disease.

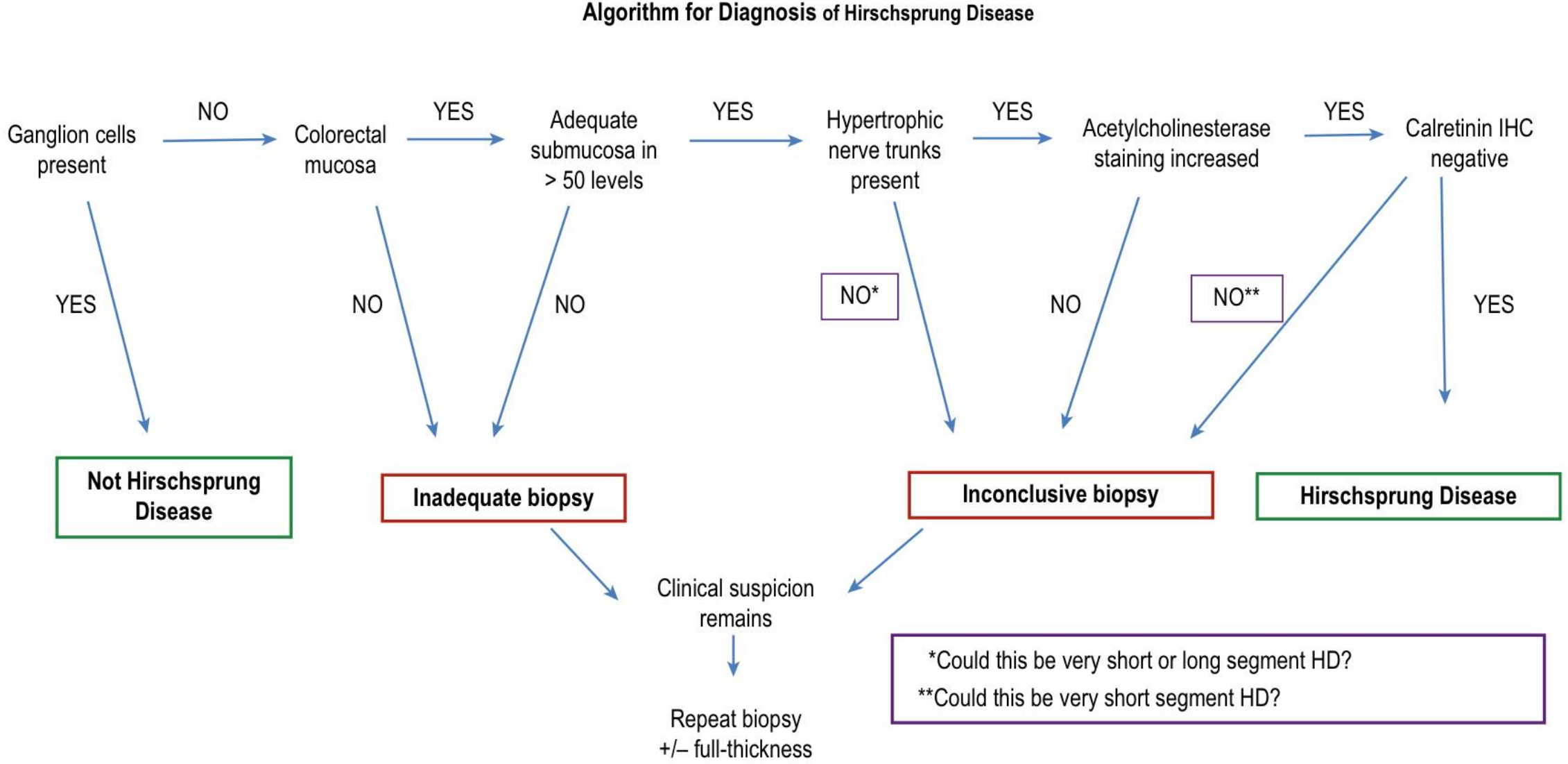


Fig. 34.7 Algorithm for the pathologic diagnosis of Hirschsprung disease from a rectal biopsy. (Courtesy Dr. Gino Somers and Dr. Glenn Taylor, Department of Pathology, Hospital for Sick Children, Toronto.)

Preoperative Preparation

- Once diagnosis HD
  - Appropriately resuscitated with IV fluid
  - Treated with broad-spectrum ATB
  - Nasogastric drainage
  - Rectal decompression : rectal stimulation or irrigation
  - Evaluate other Co-underlying disease
- The Operation can be done semi-electively
- While waiting infant can be discharged home with rectal irrigation.
- Older children with an extremely dilated colon, pull-through should be delayed until the diameter has decrease, some may need colostomy to adequate decompress dilated colon.

Preoperative Preparation

- Short segment : some physicians have advocated
  - nonoperative long-term using enema and laxative
  - Simple myomectomy
- But these approach not provide good quality if life so most pediatric surgeons recommended a pull-through procedure.

Surgical management

- The goal are to **remove the aganglionic bowel** and reconstruct the intestinal tract, that normally innervated bowel down to the anus while preserving normal sphincter tone.
- Nowaday, we prefer one-stage operation as it was safe, avoided the morbidity of stomas in children and was more cost effective .
- However, a stoma may still be needed for some patient with severe enterocolitis, perforation, malnutrition or massively dilated proximal bowel and in situations when it is not possible to reliably identify the transition zone on frozen section.

Swenson procedure

- To remove the entire aganglionic colon with an end to end anastomosis above the anal sphincter.

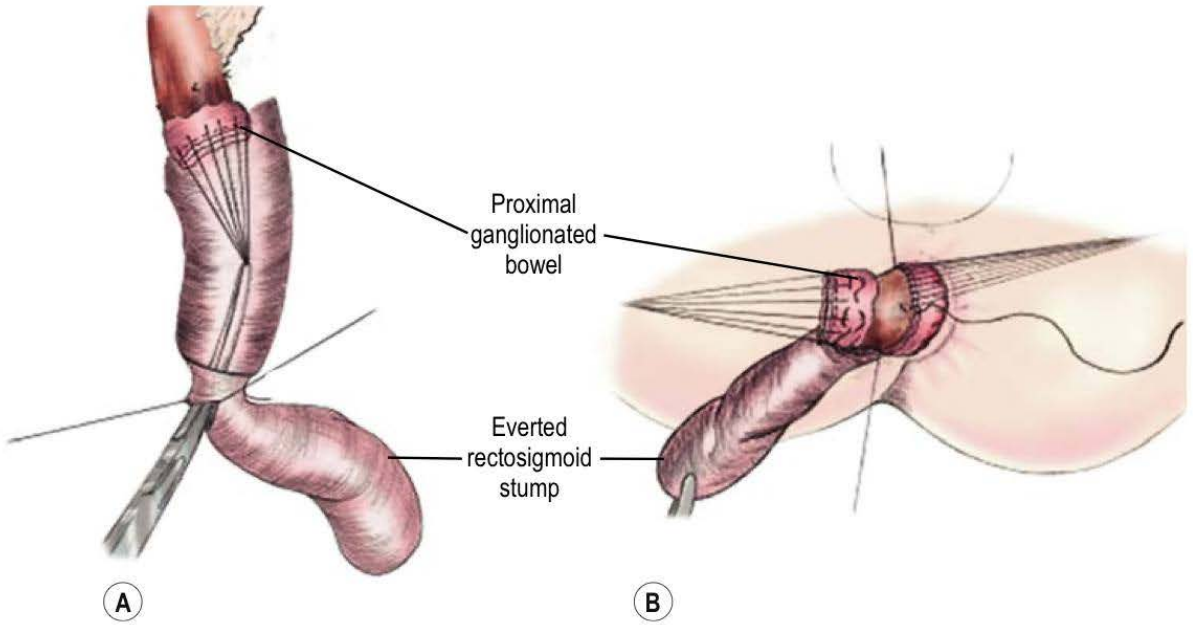


Fig. 34.8 The principles of the Swenson pull-through procedure are seen in these drawings. **(A)** The proximal ganglionated bowel is grasped through an incision in the prolapsed rectosigmoid stump. **(B)** The ganglionated bowel is then sewn to the anus.

Soave procedure

- Was designed to avoid the risk of injury to important pelvic structure by performing a submucosal endorectal dissection and positioning the pull-through bowel withing an aganglionic muscular cuff.
- Less pelvic dissection.
- Late follow up reported similar outcomes between the Soave and Swenson procedure.

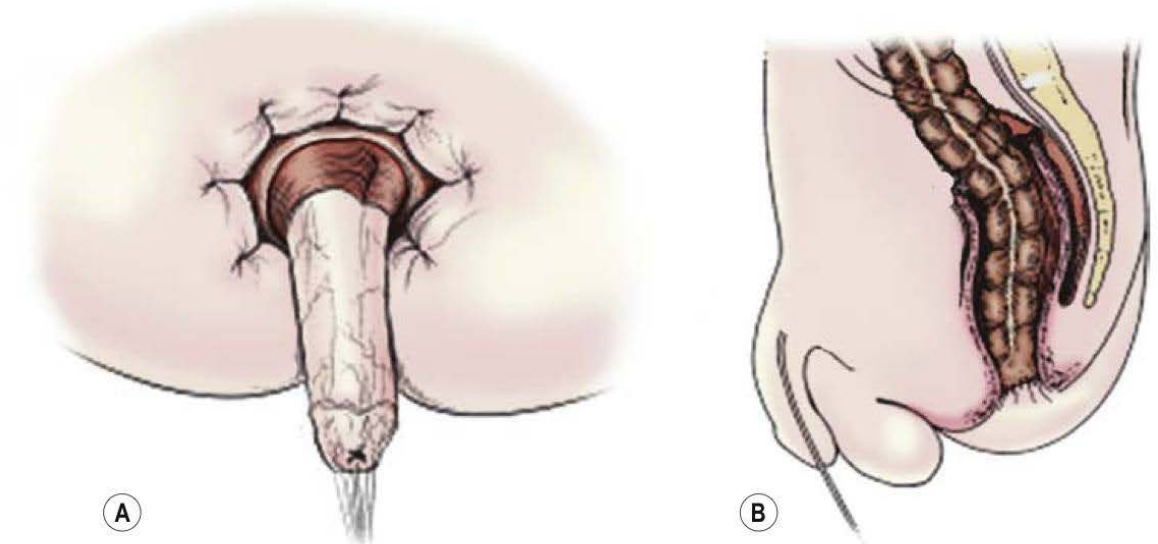


Fig. 34.9 **(A)** For the Soave operation, there is extramucosal dissection of the rectum after circumferential incision of the rectal mucosa. **(B)** The ganglionated colon is pulled through the aganglionic rectal cuff, and a coloanal anastomosis is performed.

Duhamel procedure

- Procedure involves bringing the normal colon down through the bloodless plane between the rectum and sacrum, and joining the two walls with a linear stapler to create a new lumen that is *aganglionic anteriorly* and *normally innervated posteriorly.*
- Less pelvic dissection.
- It results in a very large anastomosis, which reduces the risk of stricture.
- Reported long term results have been similar to other two operations.

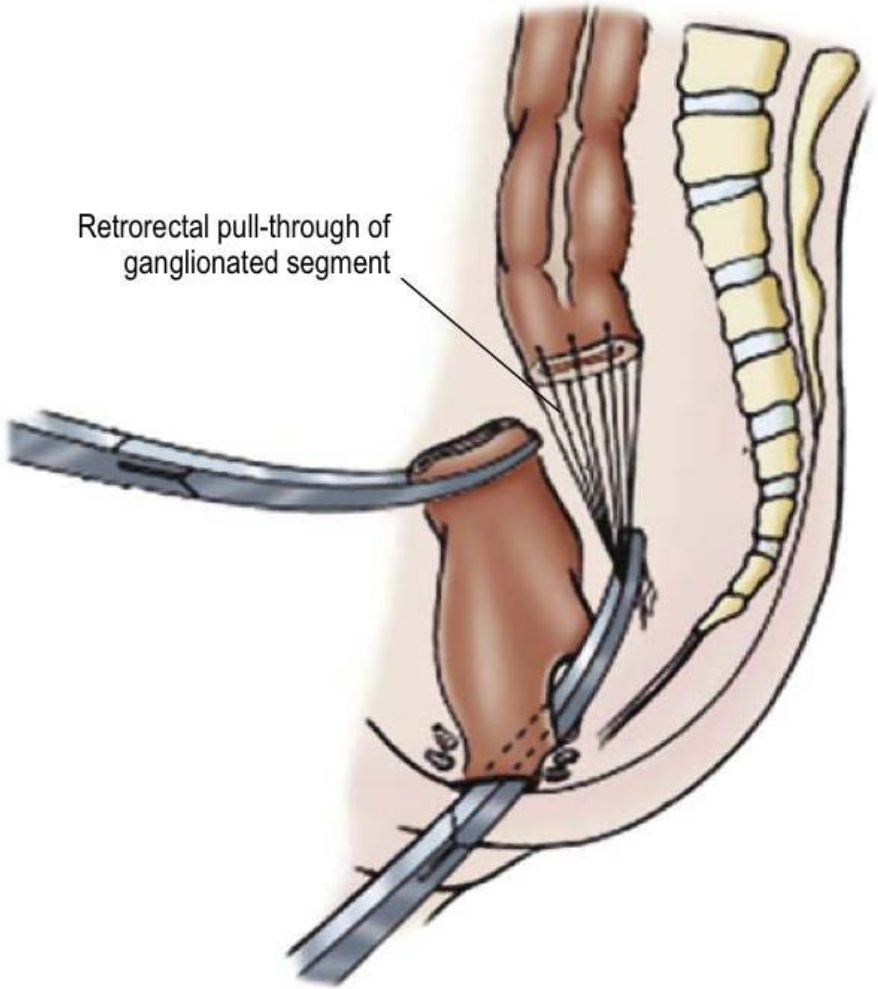
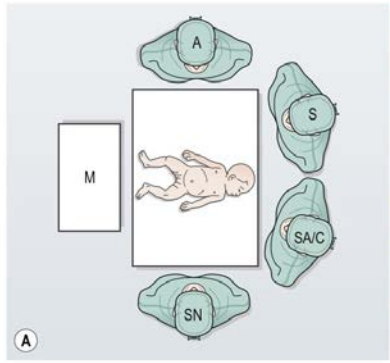
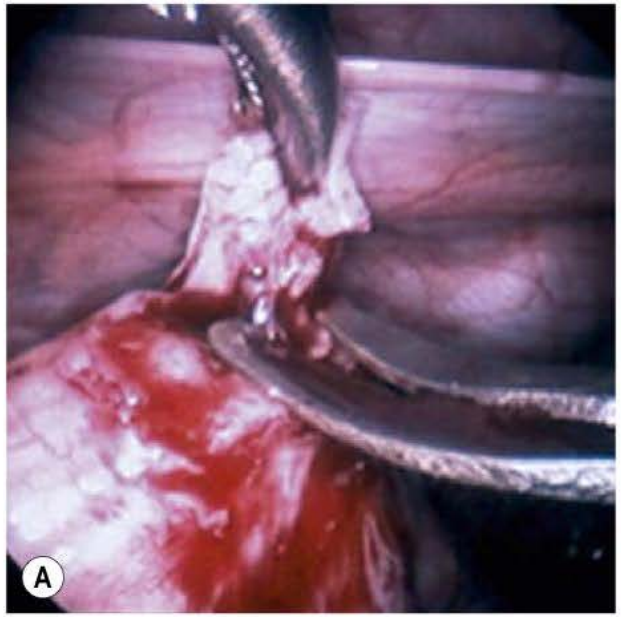


Fig. 34.10 With the Duhamel technique, the ganglionated bowel is delivered through an incision in the posterior aspect of the native aganglionic rectum and sewn to the anus. The septum between the ganglionated pull-through colon and the aganglionic native rectum is then divided using a stapler.

Laparoscopic pull-through

- With this technique, a biopsy is initially performed to identify the TZ.





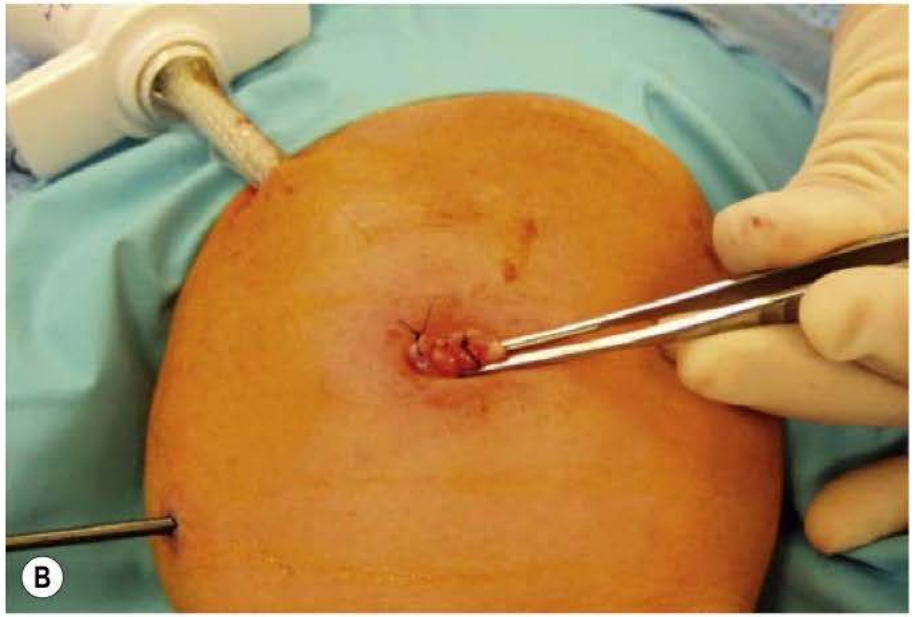
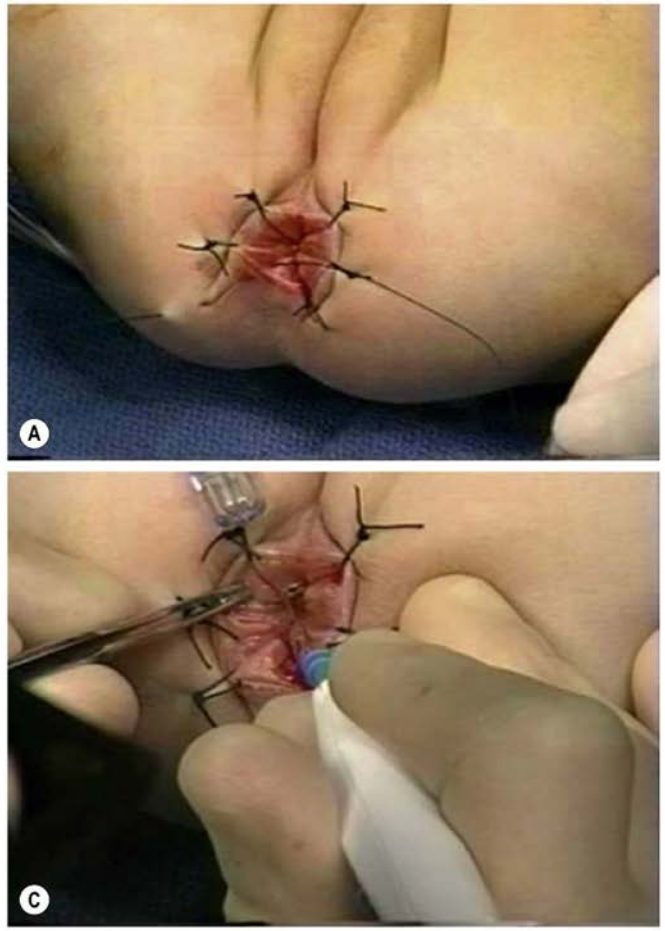


Fig. 34.12 (A) An intracorporeal biopsy is being performed on the sigmoid colon. A fine-tipped grasping forceps has been used to grasp the biopsy site, and Metzenbaum scissors are used to obtain the biopsy specimen. **(B)** This biopsy was performed through the umbilical incision. One port and another instrument have been introduced through the infant's abdominal wall. A site on the colon for the biopsy was visualized and delivered just under the umbilical cannula. The umbilical cannula was removed, and this portion of the colon was grasped and exteriorized. An extracorporeal biopsy was obtained, and the biopsy site was closed. **This is an alternative means for obtaining the biopsy.** (From Morowitz MJ, Georgeson KE. Laparoscopic assisted pull-through for Hirschsprung's disease. In: Holcomb GW, Georgeson KE, Rothenberg SS, eds. Atlas of Pediatric Laparoscopy and Thoracoscopy. Philadelphia: Elsevier; 2008, p. 101–108. Reprinted with permission.)

Laparoscopic pull-through

- The rectum is mobilized below the peritoneum reflection.
- Perineal dissection begin with the placement of circumferential 2-0 silk traction from dentate line to perineum 2-3 cm outward anus.
- A circumferntial mucosal incision is made 0.5 cm proximal to anal column.



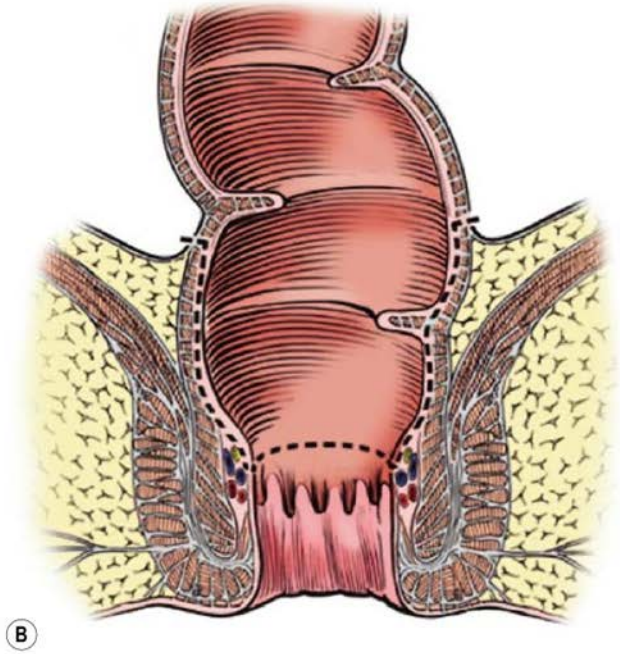


Fig. 34.14 **(A)** The perineal dissection begins with the placement of circumferential 2-0 silk traction sutures from the dentate line to the perineum 2–3 cm outward from the anus. **(B, C)** A needle-tipped electrocautery is used to circumferentially incise the rectal mucosa approximately 5 mm proximal to the anal columns. Fine silk traction sutures are then placed in the rectal mucosa to help retract the mucosa during circumferential dissection. (From Morowitz MJ, Georgeson KE. Laparoscopic assisted pull-through for Hirschsprung's disease. In: Holcomb GW, Georgeson KE, Rothenberg SS, eds. Atlas of Pediatric Laparoscopy and Thoracoscopy. Philadelphia: Elsevier; 2008, p. 101–108. Reprinted with permission.)

Laparoscopic pull-through

- The mucosa dissection is performed by perineal approach and has been exteriorized.
- The anastomosis performed above the biopsy site and is being performed with interrupted 4-0 absorbable sutures.

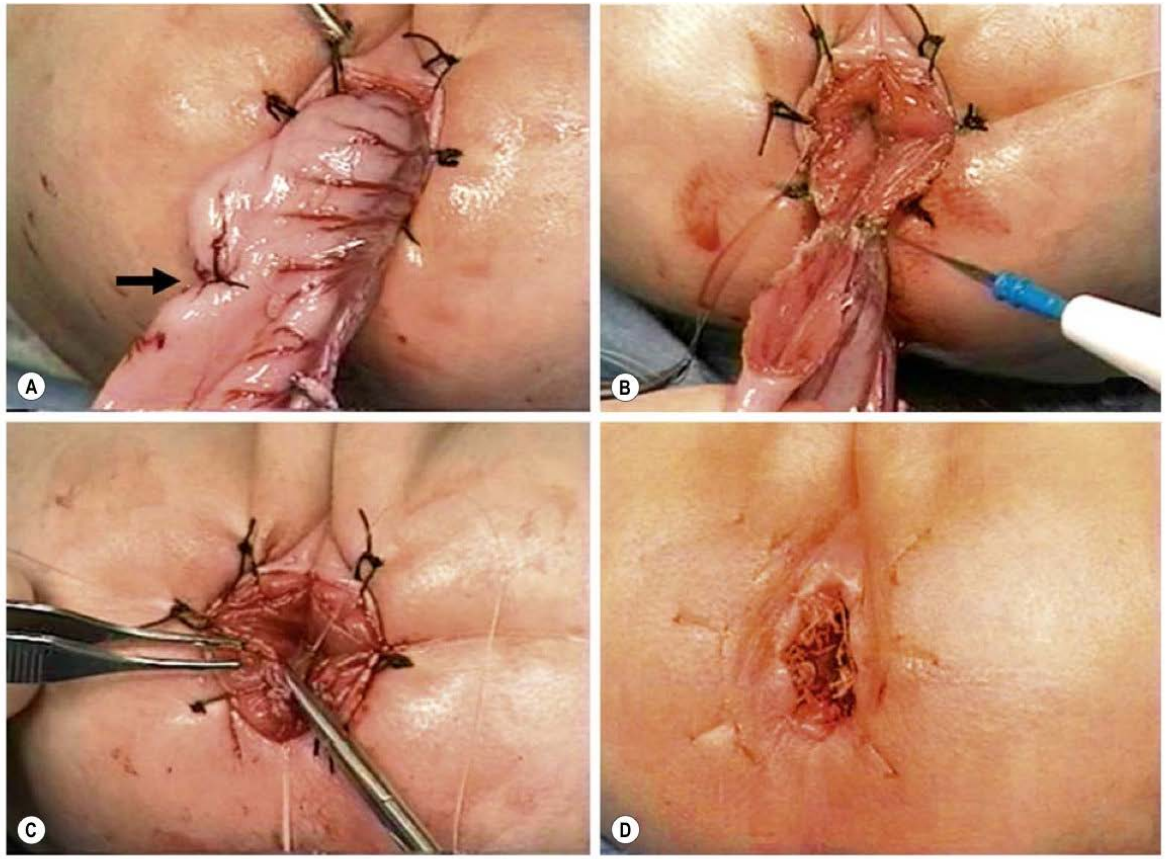


Fig. 34.15 (A) The muscular cuff of the rectum has been divided, and the ganglionic colon has been exteriorized through the anal canal. Note that the anastomosis will be performed proximal to the biopsy site (arrow). **(B)** The pull-through colon is being completely transected above the biopsy site and made ready for the coloanal anastomosis. **(C)** The anastomosis is being performed with interrupted 4-0 absorbable sutures. **(D)** The everting stay sutures have been cut, allowing the anastomosis to retract cephalad. (From Morowitz MJ, Georgeson KE. Laparoscopic assisted pull-through for Hirschsprung's disease. In: Holcomb GW, Georgeson KE, Rothenberg SS, eds. Atlas of Pediatric Laparoscopy and Thoracoscopy. Philadelphia: Elsevier; 2008, p.

Transanal(perineal) pull-through

- The operation can be performed in the prone or lithotomy positions.
- A mucosal incision is made 0.5-1.0 cm above the dentate line.
- The mucosal is stripped from the underlying muscle as in the Soave operation. The length of cuff is varies due to surgeon, a short cuff may be associated with a lower incidence of enterocolitis and stricture.
- Some surgeon prefer to do Swenson operation.
- In pt with more proximal TZ, laparoscopic or a small umbilical incision is needed to mobilized the left colon.

Transanal(perineal) pull-through

- There are controversy whether biopsy of TZ should be done before the operation on the fact that 8-10% of children who have a rectosigmoid TZ on contrast study have a more proximal histologic TZ.
- A prelim biopsy does not have a deleterious effect on postoperative outcome as time to feeding, pain, or LOS.
- Advantage
  - Low complication rate
  - Requires minimal postoperative analgesia
  - Early feeding
  - Early discharge

Long-segment aganglionosis

- Although there is no formal system for classifying length of aganlionic bowel.
- Long-segment HD is usually defined as a transition zone that is proximal to the mid-transverse colon.
- The most common form is total colonic aganglionosis (TCA), which usually also includes some of distal ileum.
- In rare cases, near-total intestinal aganglionosis, most of small bowel.
- Most neonates present with a distal small bowel obstruction, although children occasionally may not present after weaning from breast milk.
- Long segment disease is more likely to be associated with a positive family history.

Long-segment aganglionosis

- The contrast enema typically shows a shortened, relatively narrow question mark colon.
- There may also be a transition zone in small bowel.

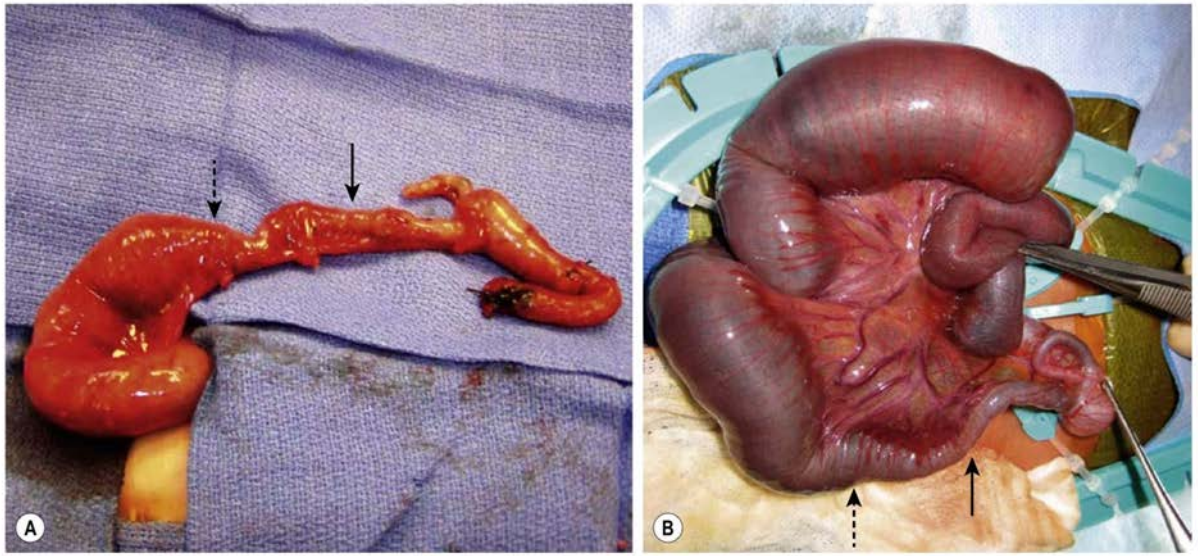


Fig. 34.17 These two photographs (**A, B**) depict neonates with total colon aganglionosis. In each photograph, the solid arrow marks the contracted aganglionic terminal ileum, and the transition zone to ganglionated ileum is marked by the dotted arrow. In **(B)**, the small bowel proximal to the transition zone is especially dilated.

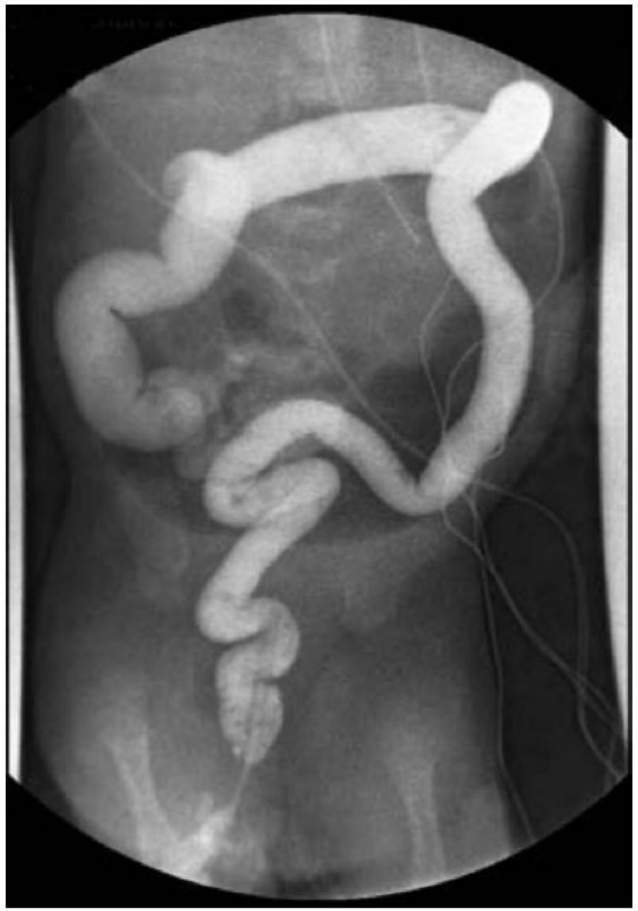


Fig. 34.18 This contrast enema was performed in a child with total colonic aganglionosis. There is no transition zone in the colon, and the colon is foreshortened with a "question mark" configuration.

Long-segment aganglionosis

- The initial operative approach involves sequential colonic biopsies looking for ganglion cells on frozen section.
- A biopsy of cecum is preferable and sequential colonic biopsies.
- Once the level of aganglionosis is identified, most surgeons create a stoma, wait for permanent sections and perform the definitive reconstruction procedure later, between 6 months or several years of life as the outcome of pull-through are better once the stool has thickened.

Long-segment aganglionosis

Reconsrtuction Operations

1. Straight pull-through : bringing normally innervated ileum to just above the anal sphincter.
2. Colon patch :using left colon(Martin) or right colon(Kimura) – decrease stool output but the aganglionic colon tend to dilate and cause enterocolitis which required surgical removal.
3. Ileal J-pouch

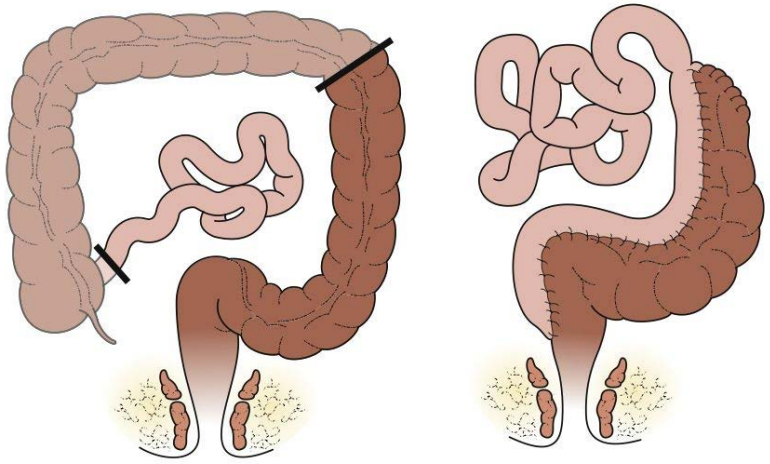


Fig. 34.19 This diagram depicts the Martin procedure for total colon aganglionosis.

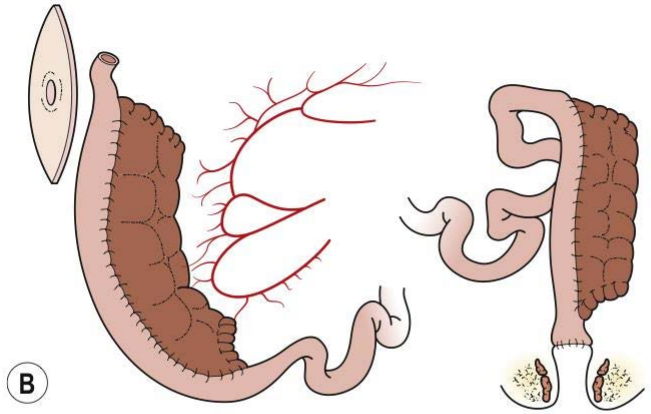
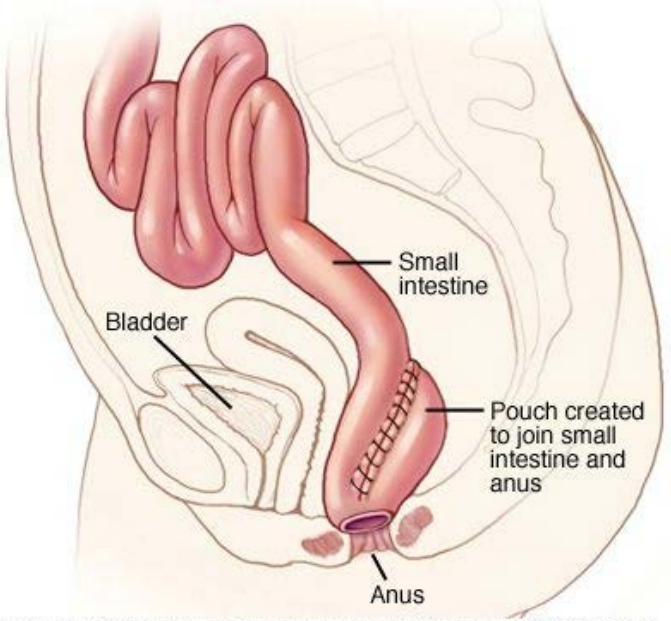


Fig. 34.20 **(A)** The ganglionic small bowel is anastomosed side-to-side to the right colon and brought out as a stoma. **(B)** After several months, when the right colon has acquired a blood supply from the small bowel, the ileo-colon is brought down to the anus as a pull-through.

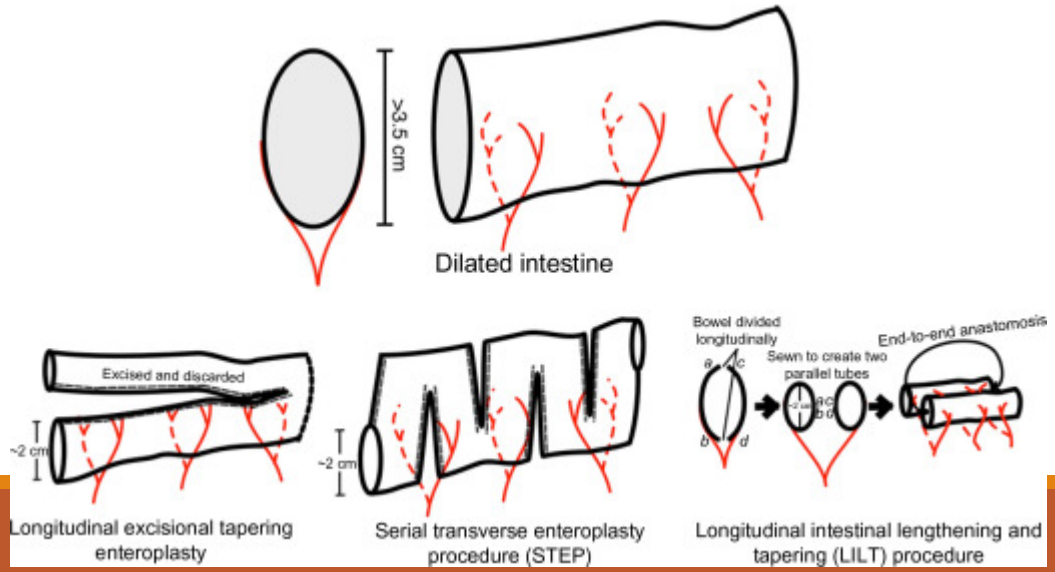


Long-segment aganglionosis

- Rarely, almost the entire intestinal tract is aganglionic usually leaving 10-40 cm of normally innervated jejunum.
- These patient require TPN from birth, most case there are not enough functional small bowel to support enteral nutrition.
- The goal is to determine the extend of aganglionosis and to create stoma at the most distal point with ganglionic bowel.
- Gastrotomy should be considered for continuous feeding of breast milk or elemental formula.
- A multidisciplinary group focused on intestinal failure; strict attention to prevent of sepsis, treatment of bacterial overgrowth, use of trophic feedings, prevention of TPN-related cholestasis.

Long-segment aganglionosis

- Surgical management depend on individual length of normally innervated bowel and pt status.
- Significant dilatation of normally innervated bowel maybe tapering, imbrication, or bowel lengthening procedures such as the Bianchi(LILT) or serial transverse enteroplasty(STEP) procedure, or myectomy-myotomy procedure(of a aganglionic small bowel distal to TZ).
- For some of these patient, small bowel or combined small bowel-liver transplantation may offer for long term survival.



Postoperative management

- Patient undergoing a laparoscopic or transanal pull-through can be fed immediatedly and discharged within 24-48 hrs.
- The anastomosis is calibrated with an appropriately sized dilator or finger 1-2 week after procedure and routinely dilatation.
- Barrier cream to prevent perineal skin breakdown.
- Educated family about sign and symptoms of postoperative enterocolitis.
- Complication :
  - Wound infection
  - Intra-abdominal bleeding
  - Anastomosis leakage, stricture
  - Intestinal perforation
  - Bowel obstruction
  - fistula

Long-term outcomes

Obstructive symptoms

- Symptoms : abdominal distension, bloating, vomiting, or ongoing severe constipation.
- 5 major causes
  - Mechanical obstruction
  - Recurrent or acquired aganglionosis
  - Disorder of motility in the residual colon or small bowel
  - Internal sphincter achalasia
  - Function megacolon caused by stool-holding behavior

Long-term outcomes

Mechanical obstruction

The most common is stricture.

- Obstruction after a Swenson or Soave operation
- A retained spur consisting of aganglionic bowel, which fill with stool and obstruct
- A twist in the pull-through bowel
- narrowing due to long muscular cuff in Soave operation

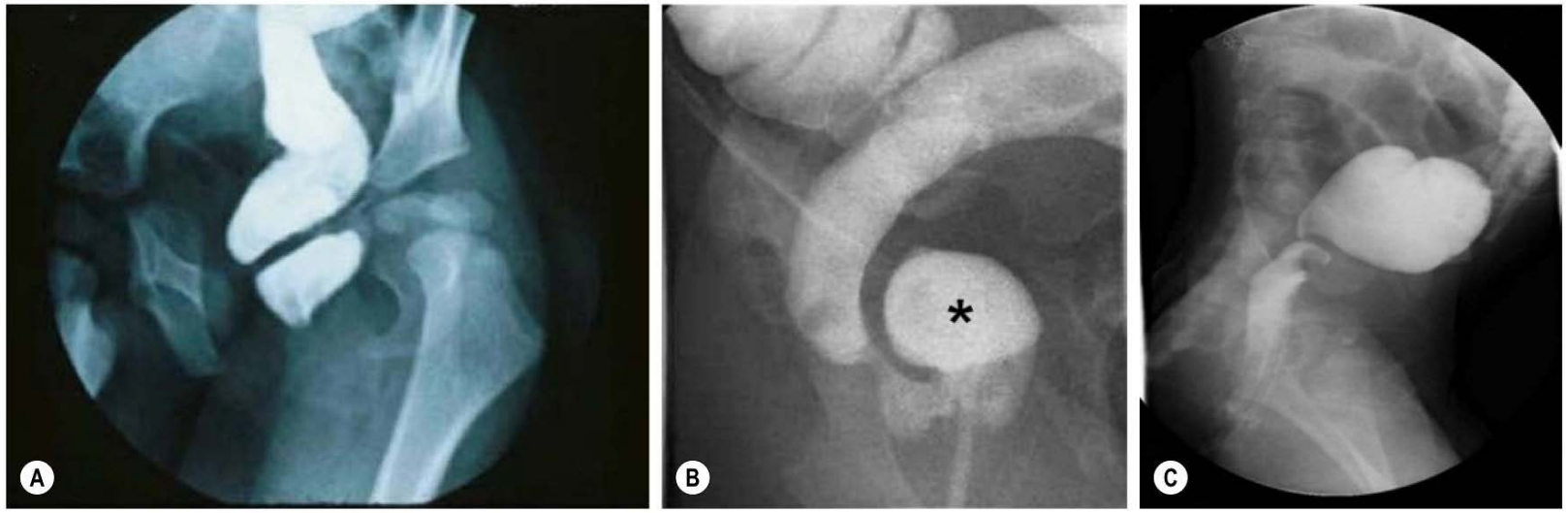


Fig. 34.22 Causes of mechanical obstruction after a pull-through are shown. **(A)** Stricture following a Soave procedure. **(B)** Anterior aganglionic "spur" (asterisk) following a Duhamel procedure. **(C)** Twisted transanal pull-through.

Long-term outcomes

Mechanical obstruction

- An anastomosis stricture can be treat be *repeated dilatation*.
- Recalcitrant strictures : *antegrade dilation, intralesion steroid, topical mitomycin*.
- Retain aganglionic segment, Duhamel spur, twisted pull-through require re-operation.

Long-term outcomes

Persistent or acquired aganglionosis or TZ pull-through

- This problem may be due to an error in histologic analysis, a transition zone pull-through, or loss of ganglion cells.
- Can be diagnosed by biopsy.
- Tx : a redo-pull-through

Long-term outcomes

Motility disorder

- Children with HD often have abnormal motility throughout the intestinal tract, including GERD and delayed gastric emptying, small bowel dysmotility and disordered colonic motility.
- Diagnosing
  - A radiologic shape study
  - Radionuclide colon transit study
  - Colonic manometry
  - Laparoscopic biopsy for intestinal neuronal dysplasia(IND)
- These abnormality can be
  - focal, usually the left colon => redo-pullthrough
  - Generalized => bowel management, use of prokinetic agents.

Long-term outcomes

Internal sphincter achalasia

- Caused by lack of a normal RAIR
- Most children eventually grow out of this problem overtime, usually age of 5 years and may *resolve on its own*.
- A *Diagnosis of exclusion* after rule out mechanical obstruction, aganglionosis and dysmotility.
- Confirm diagnosis by *clinical response of intrasphicteric botulinum toxin.*
- The traditional operative approach is internal sphinterotomy or myectomy, but there is concern about continence, we prefer botulinum injection.
- Repeated *injection of botulinum toxin*, *nitroglycerine paste* or *topical nifedipine* are necessary while waiting for resolution of the problem.

Long-term outcomes

Functional megacolon

- The result of stool-holding behaviour.
- Tx : bowel management consist of laxatives, enemas and behavior modification strategies.
- In severe cases may requires antegrade enemas or even proximal stoma.

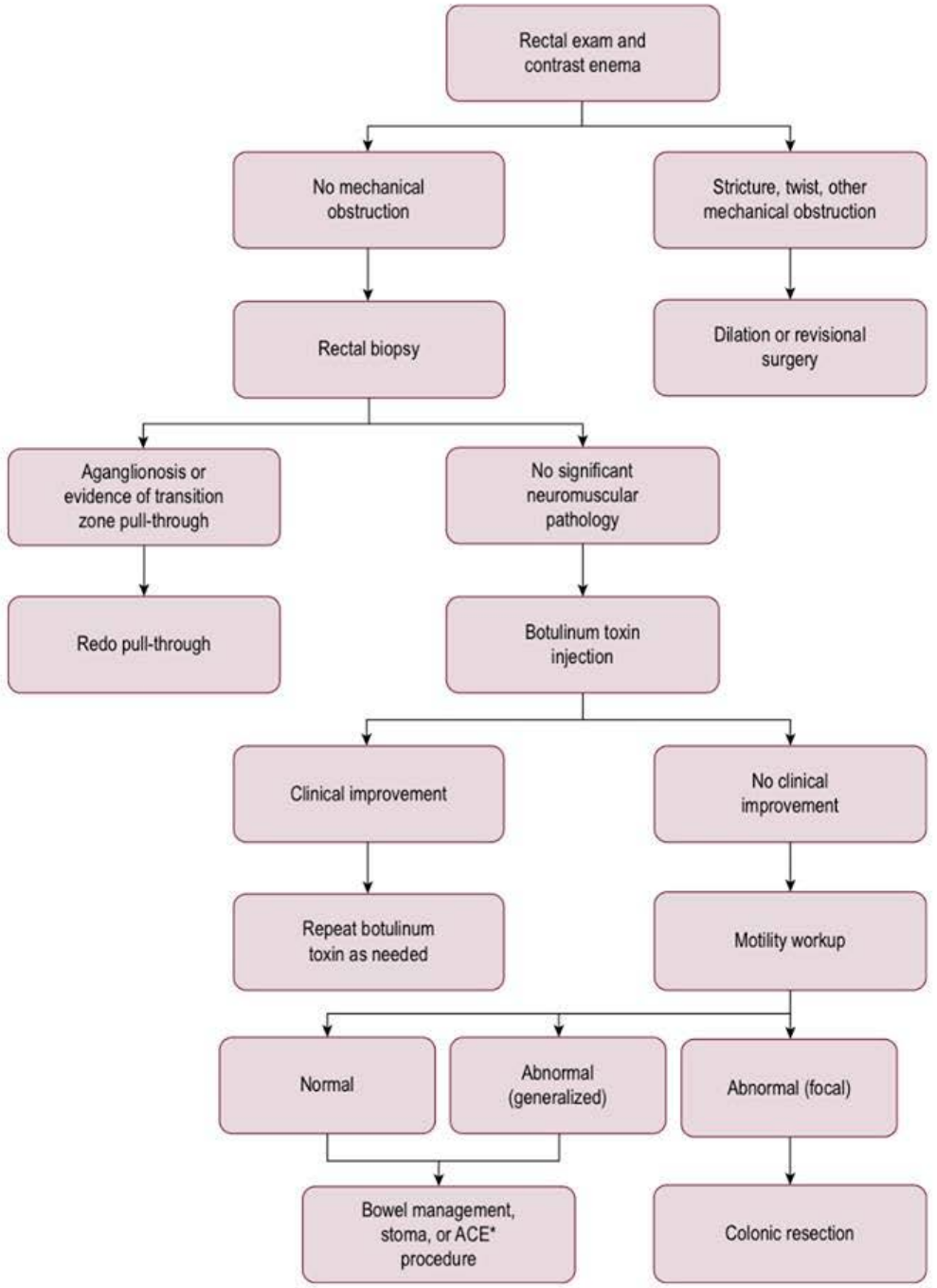


Fig. 34.21 Algorithm for the investigation and management of the child with obstructive symptoms following a pull-through. ACE, antegrade colonic enema.

Long-term outcomes

Fecal soiling

Causes

- Abnormal sphincter function
- Abnormal sensation
- Pseudo-incontinence

Long-term outcomes

Abnormal sphincter

- May be due to sphincter injury during the pull-through or to a previous myectomy or sphincterotomy
- Identifed using anorectal manometry or endorectal US.

Abnormal sensation

1. Lack of sensation of full rectum : identified by using anorectal manometry
2. Inability to detect the difference between gas and stool : due to loss of transitional epithelium because the anastomosis was performed below the dentate line.

- Mx : bowel routine, constipating diet, stimulant laxatives, rectal or antegrade enemas, biofeedback training, colostomy

Long-term outcomes

Pseudo-incontinence

- Caused by severe obstipation with a massively distended rectum and overflow of liquid stool.
- Other patient leak small amount of stool though the day.
- Other can suffer from hyperperistalsis of the pull-through bowel, which result inability of the anal sphincter to achieve control of normal sphincter.
- Diag :
  - contrast enema
  - Colonic motility studies

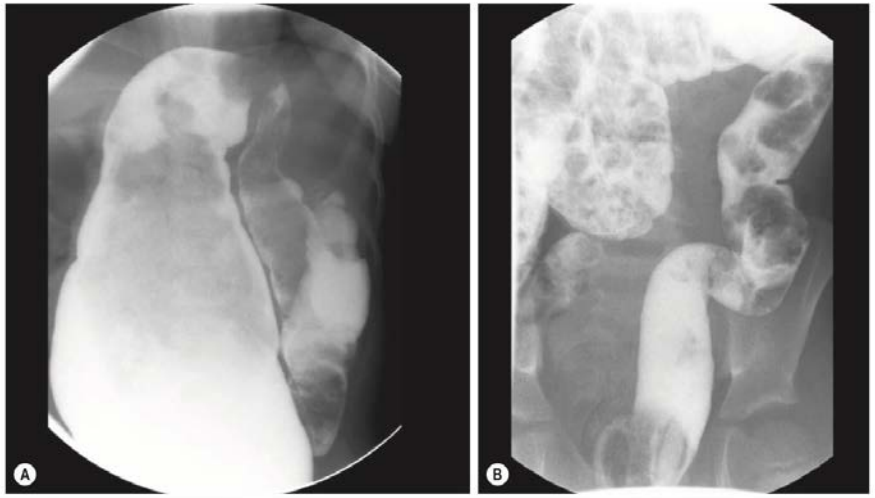


Fig. 34.24 The difference between obstipation and hyperperistalsis forms of pseudo-incontinence can be demonstrated on contrast enema. **(A)** With obstipation, the rectum is extremely dilated and the sigmoid colon may be very redundant. **(B)** With hyperperistalsis, the caliber of the sigmoid and rectum is more normal.

Long-term outcomes

Treatment fecal soiling

- Severe constipation : passive laxative therapy ; lactulose, PEG
- Abnormal sphincter and/or sensation : stimulant laxative ; senna, bisacodyl
- Inadequate in achieving rectal emptying : require retrograde or antegrade enemas.
- Children with stool-holding : behavior change, high fiber diet, passive laxative.
- Hyperperistalsis : consipating diet and medication such as loperamide.

Long-term outcomes

Enterocolitis

- The etiology of **HAEC** is unknown and is probably multifactorial
- Stasis caused by functional obstruction permits bacterial overgrowth with secondary infection.
- Infectious agent such as *Clostridium difficle* or *rotavirus* have been postulated as being causative.
- Enterocolitis may be present both before and after operative correction, and can range in severity from mild to life threatening.
- More common in younger children, longer segment disease, and trisomy21.
- Clinical presentation : Fever, Abdominal distention, Diarrhea, Elevated leukocyte count, Evidence of intestinal edema on abdominal film

Table 34.1 Hirschsprung-Associated Enterocolitis (HAEC) Score

	Score
CRITERIA HISTORY	
Diarrhea with explosive stool	2
Diarrhea with foul-smelling stool	2
Diarrhea with bloody stool	1
Previous history of enterocolitis	1
PHYSICAL EXAMINATION	
Explosive discharge of gas and stool on rectal exam	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 rectosigmoid with absence of distal air	1
Pneumatosis	1
LABORATORY	
Leukocytosis	1
Shift to left	1
TOTAL	20

A score of 10 or higher was associated with a positive diagnosis of HAEC by an international panel of experts.[108]

Long-term outcomes

Enterocolitis

Treatment

- Nasogastric drainage
- IV fluid
- Broad spectrum ATB
- Decompression if rectum and colon using rectal stimulation or irrigations.

Prevention : reduce in higher risk patient

- Routine irrigation
- Administration of metronidazole or probiotic agents

Long-term outcomes

HD-associated inflammatory bowel disease

- There is small group of children with HD who develop a condition resembling Crohn disease when they get older.
- This consists of chronic inflammation and in some cases fistula formation.
- Risk factor : long-segment disease and trisomy 21
- It is unclear whether this condition is a chronic form of HAEC or has a separate etiology.
- In many cases, the use if biologic and other anti-inflammatory medications that are commonly used to treat Crohn disease are ineffective, some may require stoma.

Variant Hirschsprung disease

Intestinal neuronal dysplasia(IND)

- Type A :
  - diminished or absent sympathetic innervation of the myenteric and submucosal plexuses, along with hyperplasia of the myenteric plexus.
  - less common
- Type B :
  - dysplasia of the submucosal plexus with thickened nerve fibers and giant ganglia, increase acetylcholinesterase staining, and identification of ectopic ganglion cells in the lamina propria.
  - Can independently or concomitant with HD.
- May be diffuse or local

Variant Hirschsprung disease

Hypoganglionosis

- Is characterized by *spare and small ganglia*, usually in distal bowel
- Often associated with abnormalities in acetylcholinesterase distribution.
- Tx : resection of abnormal colon
- This condition must be differentiated from immature ganglia, which are seen in preterm children and should not be treated surgically.

Variant Hirschsprung disease

Internal sphincter achalasia

- There are some children who have normal ganglion cells on rectal biopsy, but who lack the RAIR on anorectal manometry and develop symptom of HD
- Usually improve over the first five year of life
- TX
  - Bowel management : diet, laxative, enema and irrigations
  - botulinum toxin,
  - nitroglycerine paste
  - topical nifedipine
  - Anal sphincter myectomy

Variant Hirschsprung disease

Ultra-short segment Hirschsprung disease

- Children who document aganglionic segment <3-4 cm.
- The finding of hypertropic nerves and abnormal cholinesterase may be absent.
- Treatment is controversial
- Simple anal sphincter myectomy VS aganglionic segment pull-through

Variant Hirschsprung disease

Desmosis coli

- Rare condition characterized by chronic constipation associated with total or a focal lack of connective tissue net in circular and longitudinal muscles as well as the connective tissue layer of myenteric plexus without abnormalities in the enteric nervous system.

Thank you