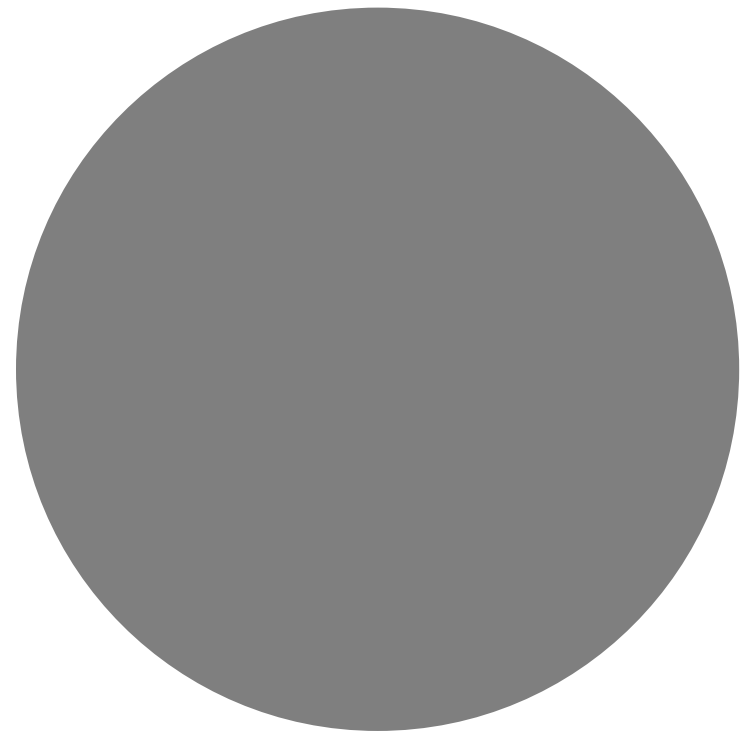
HIRSCHSPRUNG'S DISEASE



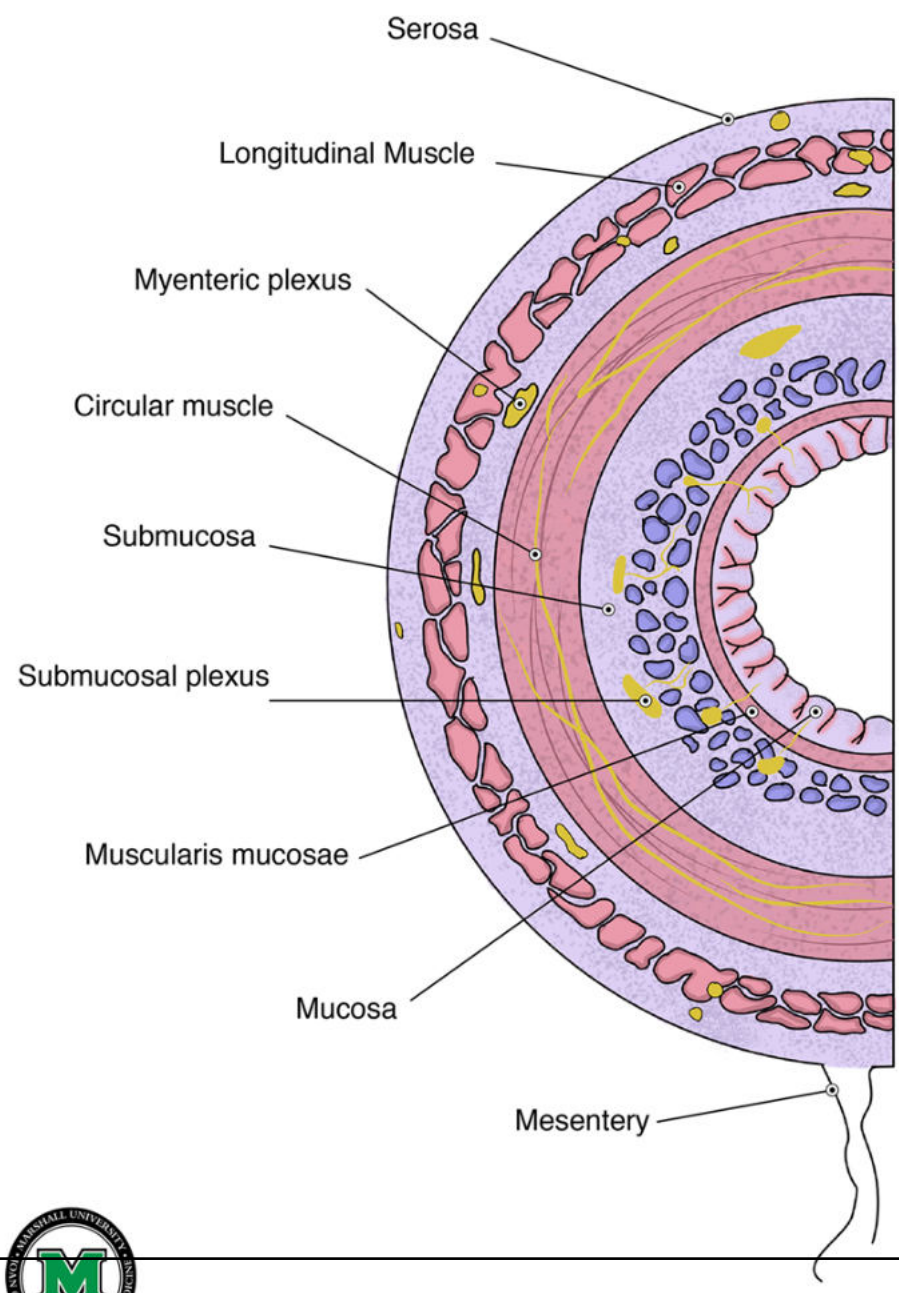
- Introduction
- Incidence and spectrum of disease
- Etiology and genetic basis of disease
- Clinical Presentation and diagnosis
- Pre-operative preparation
- Surgical management
- Post-operative management
- Long term outcomes

Introduction

Also known as congenital megacolon.

Characterised by absence of ganglion cells in the myenteric (Auerbach) and submucosal (Meissner)plexus of the intestine.

In 1887, Harald Hirschsprung, a paediatrician from Copenhagen, described two cases of the condition that ultimately bore his name.



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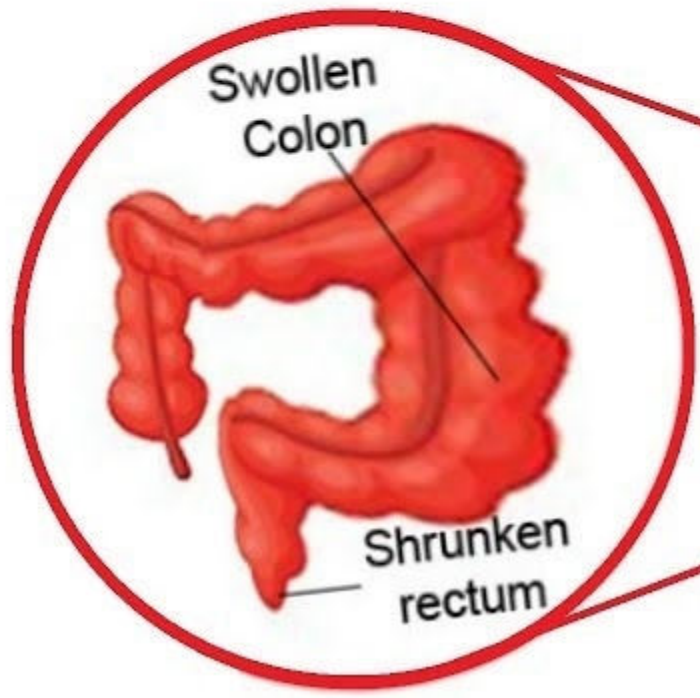
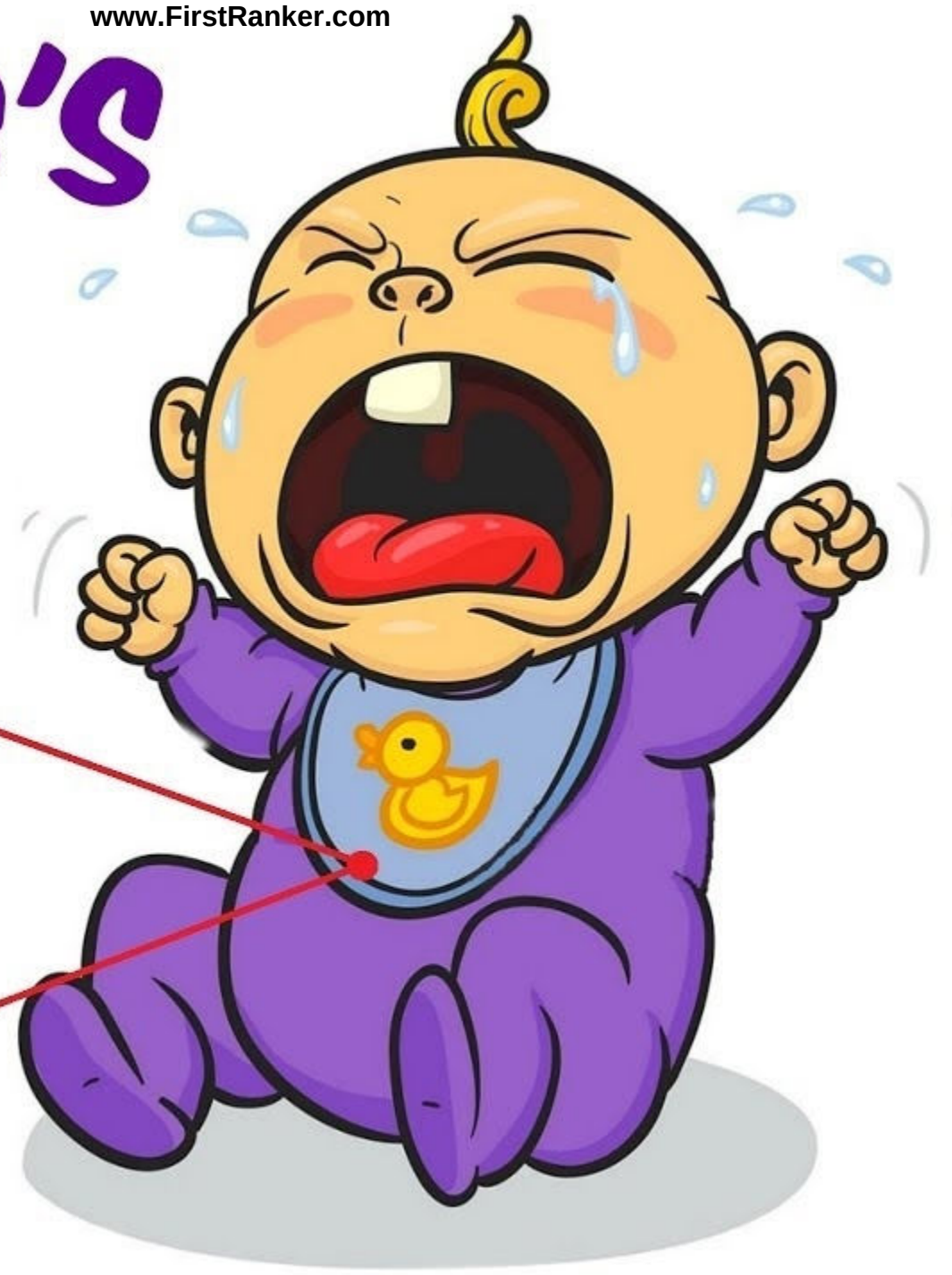




Incidence and spectrum of disease

Occurs in approx. 1 in 5000 live births.

Most commonly extent of involvement is rectosigmoid- 80%

Proximal colon involvement- 10%

Total colon involvement with part of distal ileal involvement- 5-10%

Etiology and genetic basis of disease

Male to female ratio= 4:1

For total colonic aganglionosis, male to female ratio= 1:1

Ganglion cells are derived from the neural crest.

By 13 weeks post-conception, the neural crest cells have migrated from proximal to distal through the gastrointestinal tract

Differentiation into mature ganglion cells.

Two theories

Early maturation of neural crest cells before reaching distal part of colon and rectum.

Inhospitable micro-environment for neural crest cells in the distal bowel leading failure of survival of neural crest cells.

Genetics

- Most common gene involved is RET proto-oncogene. (Probably early cell death)
- Other genes are:
  - SOX-10 genes (probably early maturation)
  - S1P1
  - Phox 2B
  - Hedgehog notch complex.

Associated syndromes and conditions

- Trisomy 21
- Opitz syndrome
- Congenital central hypoventilation syndrome
- Neurofibromatosis
- Neuroblastoma

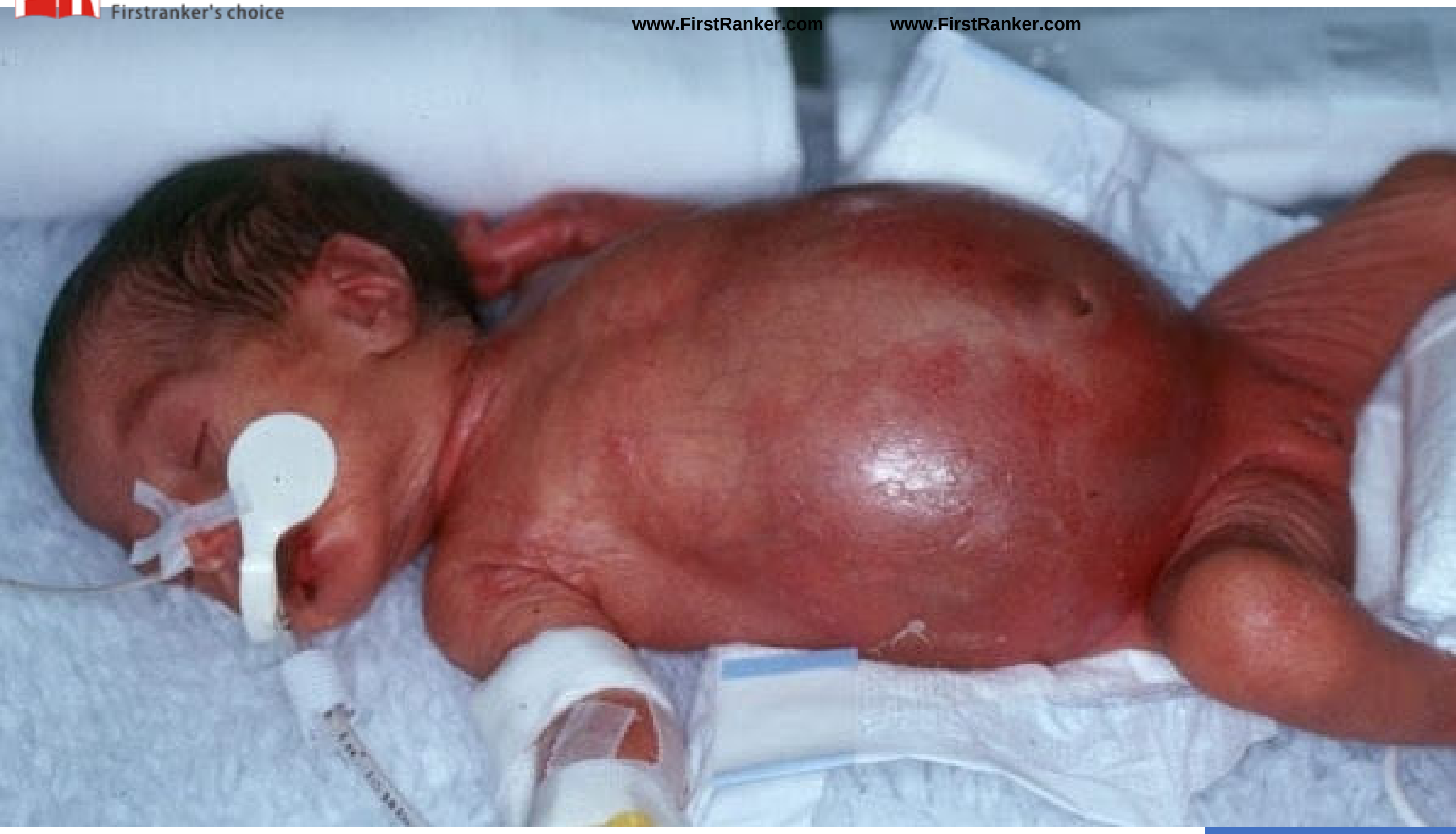
CLINICAL PRESENTATION AND DIAGNOSIS

Prenatal diagnosis is rare.

Most affected patients present in the neonatal period

- Abdominal distension
- Bilious vomiting
- Feeding intolerance
- Delayed passage of meconium beyond the first 24hours (90%)
- Prolonged passage of meconium

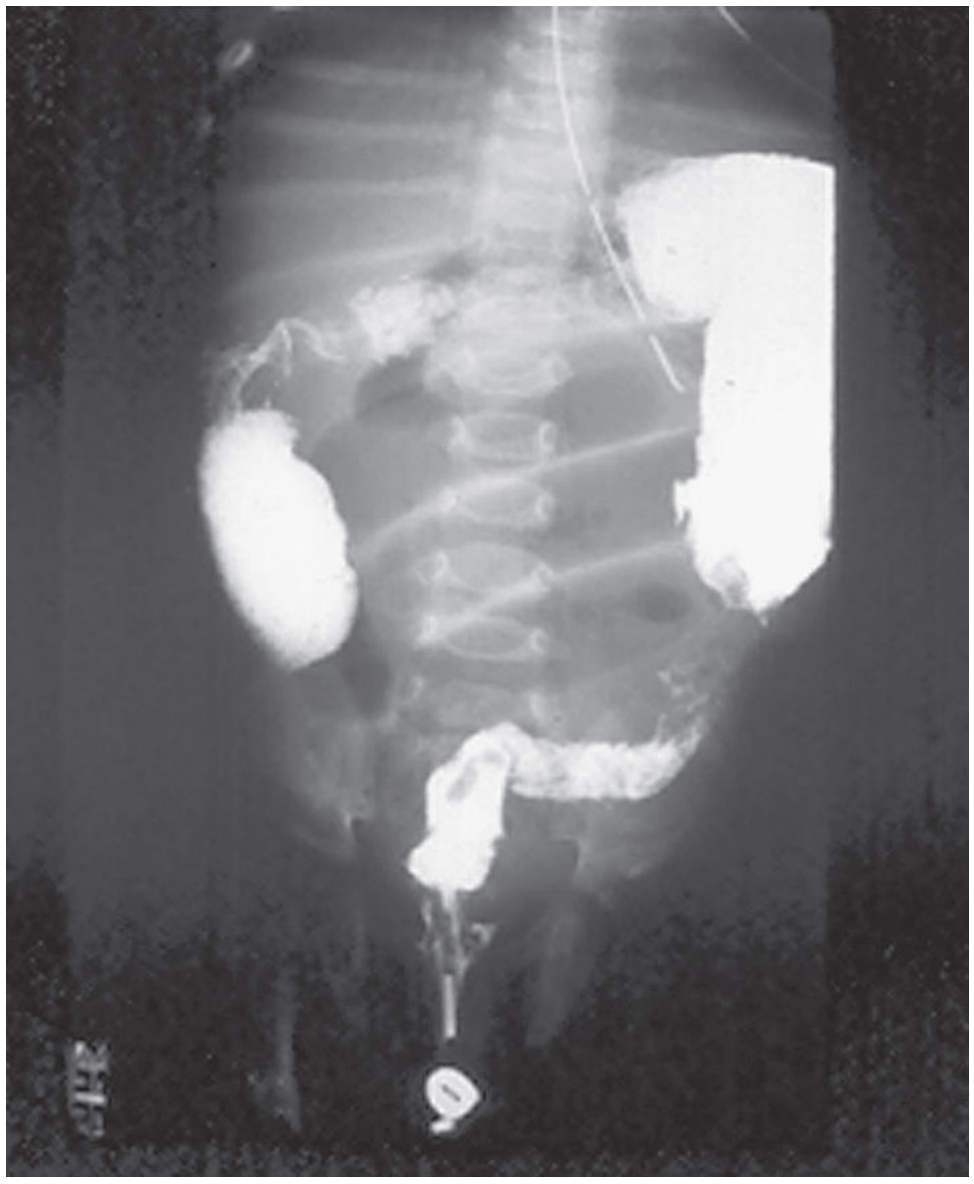
Occasional caecal or appendiceal perforation.

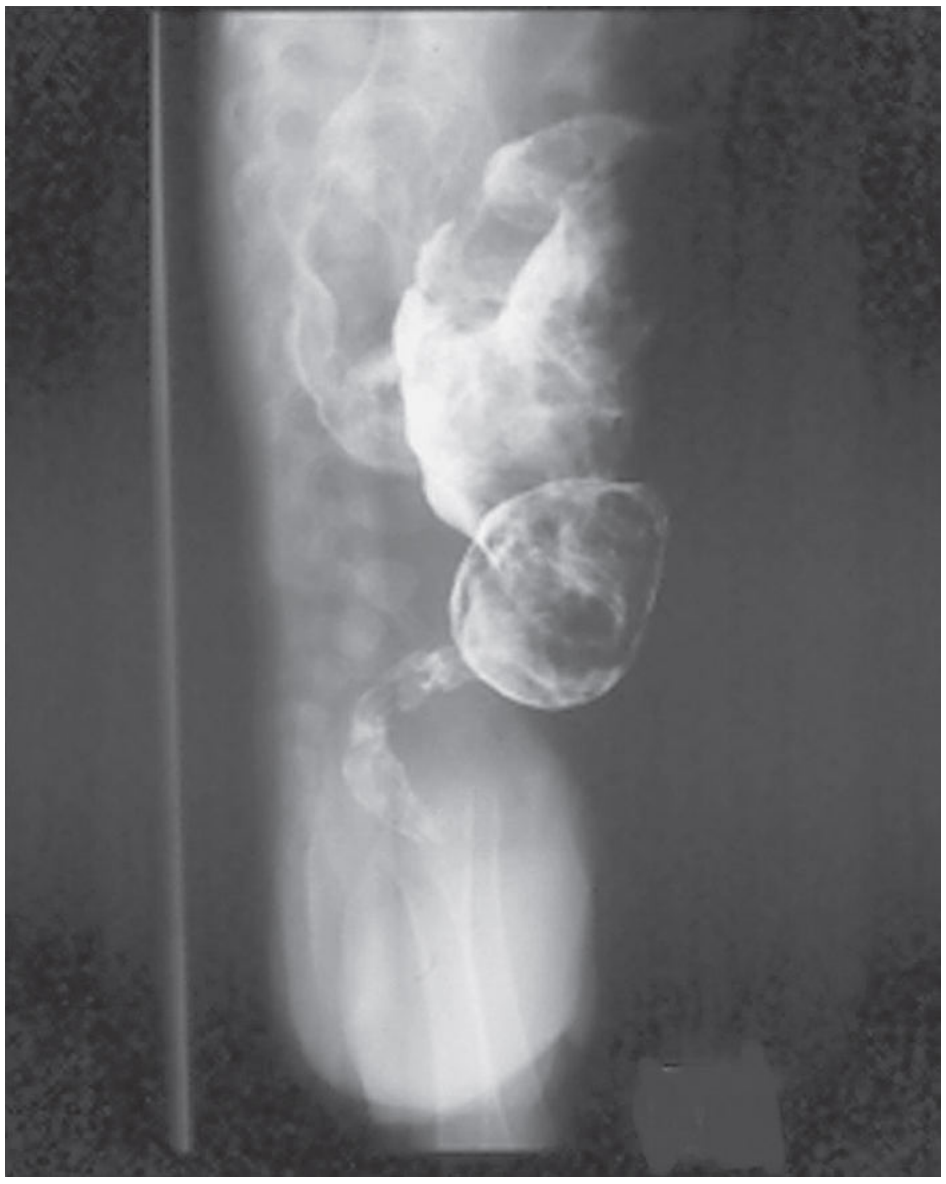


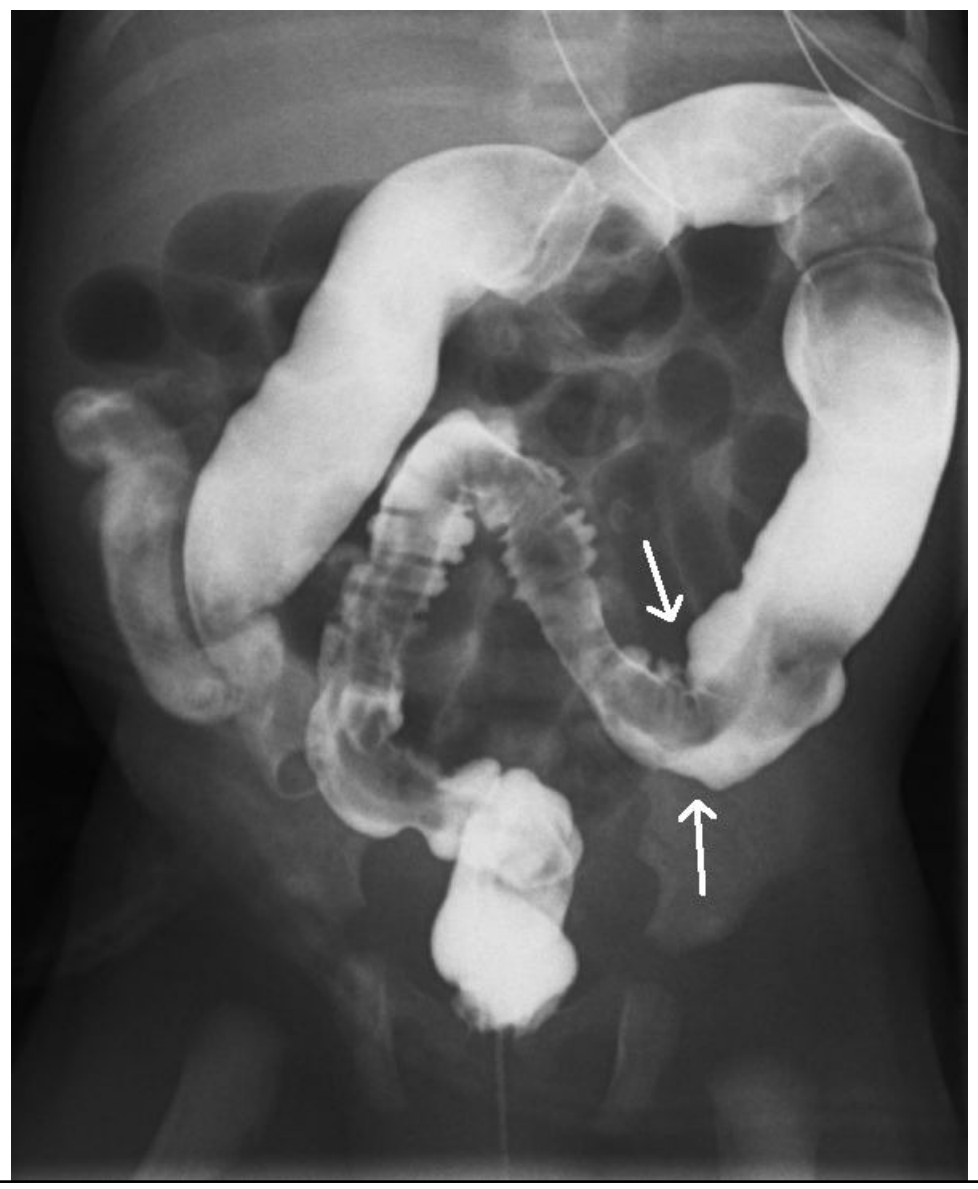
Radiology

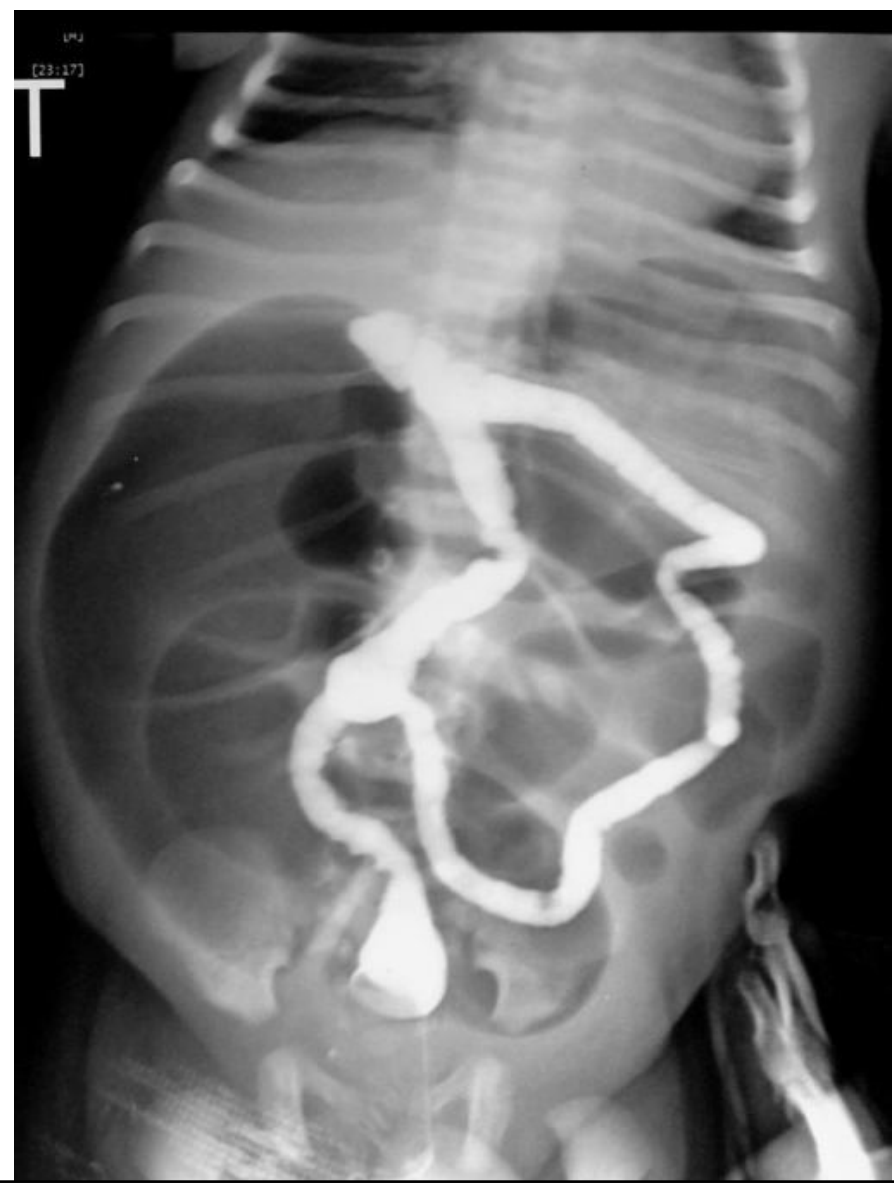
- Plain abdominal X-rays- Dilated loops of bowel and absence of air in the pelvic region.

- Contrast enema or Barium enema-
  - Presence of transition zone between normal and aganglionic bowel
  - Retention of the contrast beyond 24hours









Presentation in late infancy

Rectal biopsy using suction technique and histopathology showing absence of the ganglion cells is pathognomic.

Most patients will also have evidence of hypertrophied nerve trunks

Chronic constipation with presence of meconium history

Usually on and off constipation is present till the child is on breast feeds but, it becomes profound once weaning is started.

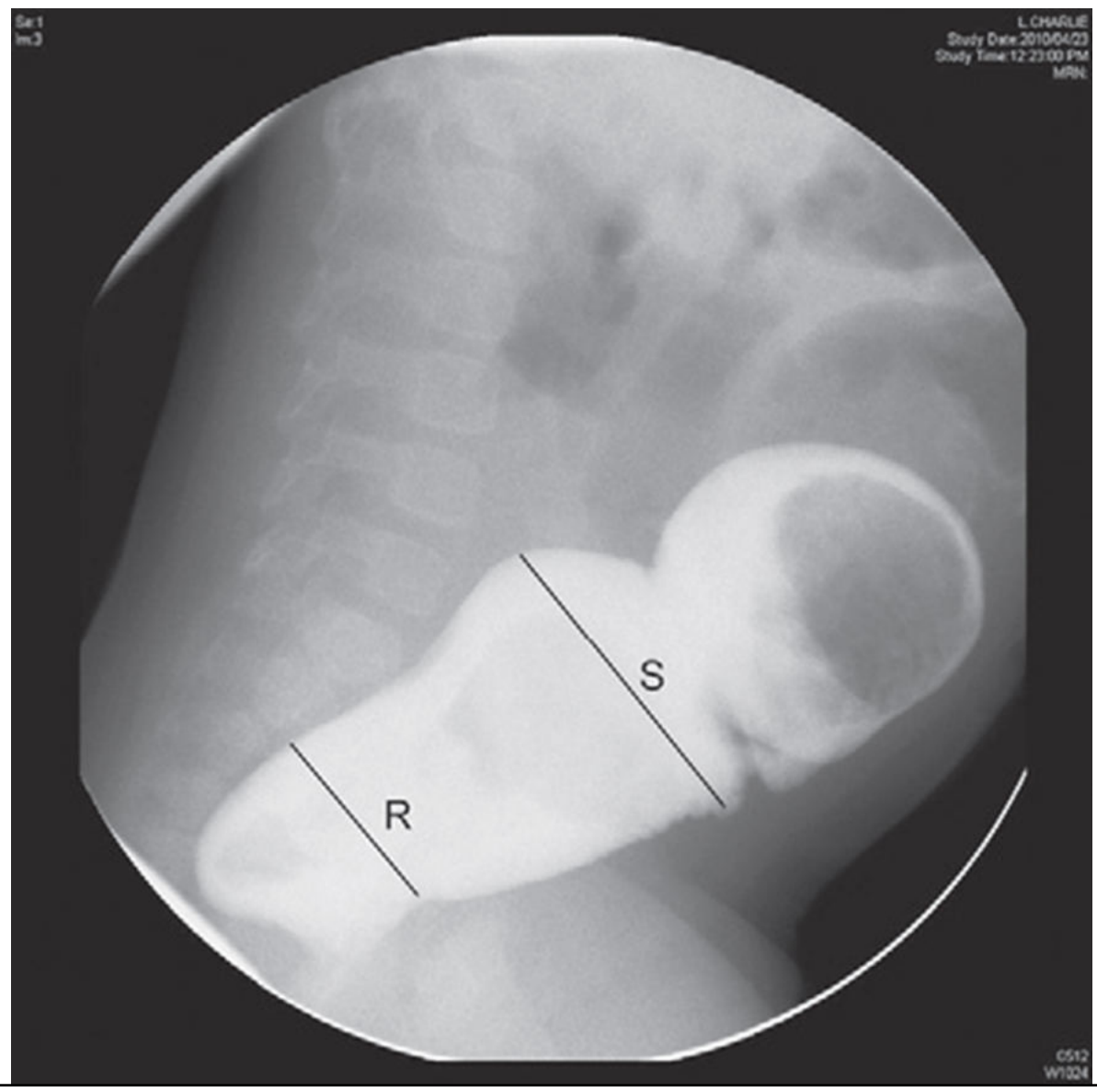
Failure to thrive

Dependence on laxatives and enemas for constipation

X-rays abdomen

Distended bowel loops

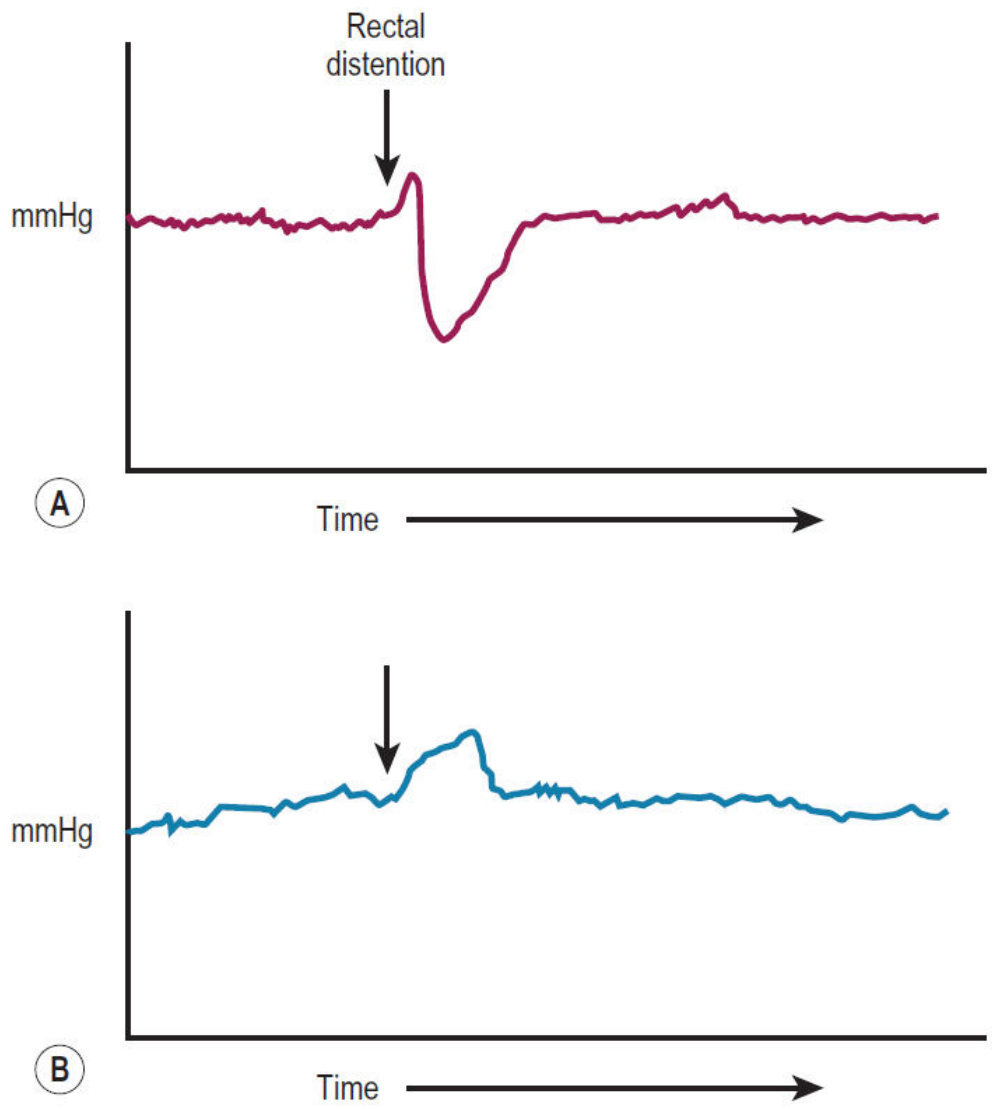
Reversal of rectosigmoid ratio and retention of contrast on a 24hours post evacuation film.



Anorectal manometry

The recto-anal inhibitory reflex (RAIR) is defined as reflex relaxation of the internal anal sphincter in response to rectal distension and is present in normal children but absent in children with Hirschsprung disease.

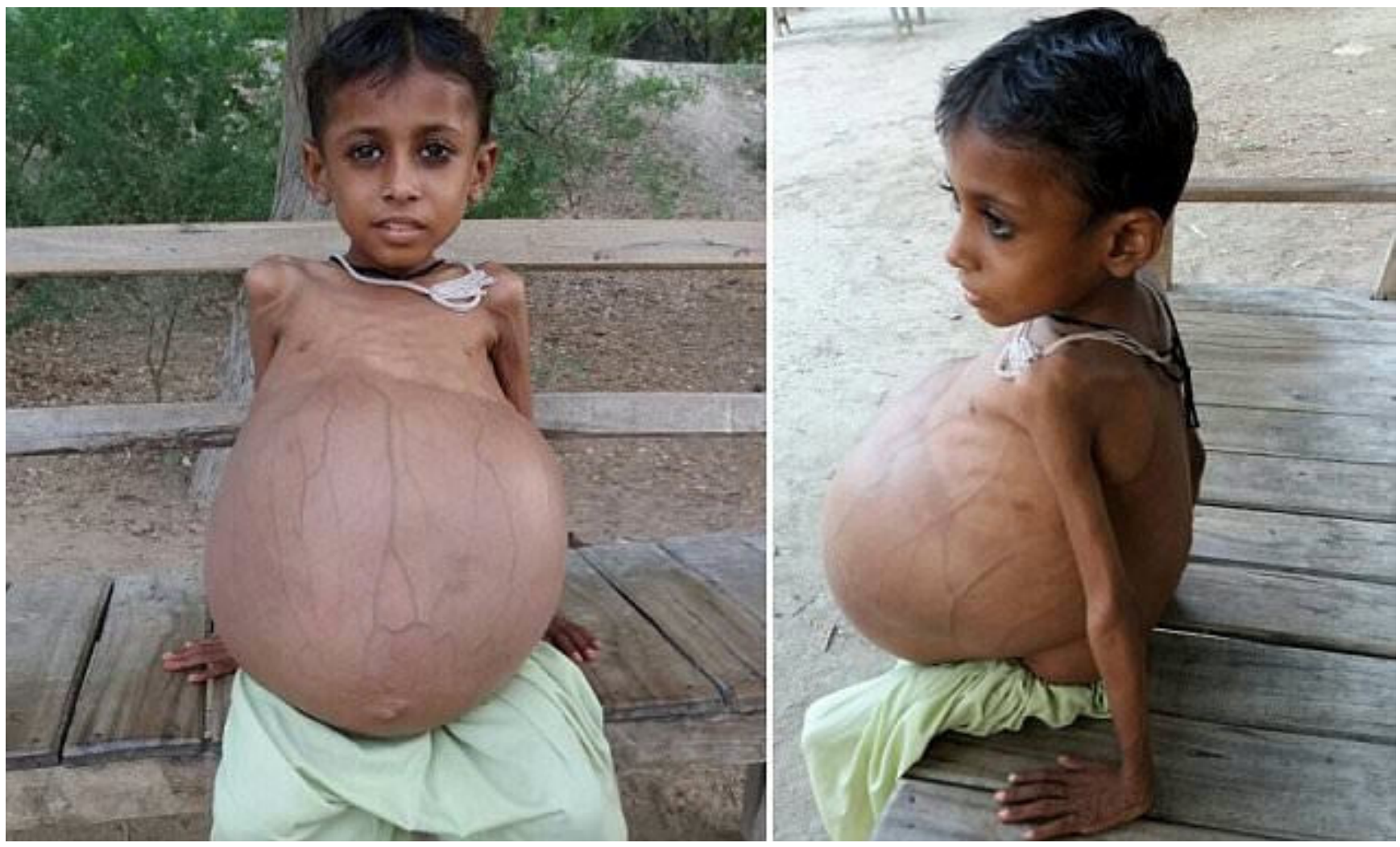
The RAIR can be documented using anorectal manometry by inflating a balloon in the rectum while simultaneously measuring the internal sphincter pressure.



A- Normal child

B- Hirschsprung's disease

Older children



In older children, the suction rectal biopsy may be less reliable because of a higher risk of sampling error.

Full-thickness biopsies, usually under general anesthesia, may be necessary in these patients.

Hirschsprung-associated enterocolitis (HAEC)

Approximately 10% of neonates present with fever, abdominal distention, and diarrhea due to Hirschsprung -associated enterocolitis (HAEC)- can be life-threatening.

Since HD characteristically causes constipation rather than diarrhea, this presentation may be confusing and the diagnosis may not be considered.

A careful history, including a history of delayed passage of meconium and intermittent stooling, should lead to an investigation for HD.

Aggressive treatment with IV fluids and IV antibiotics is done.

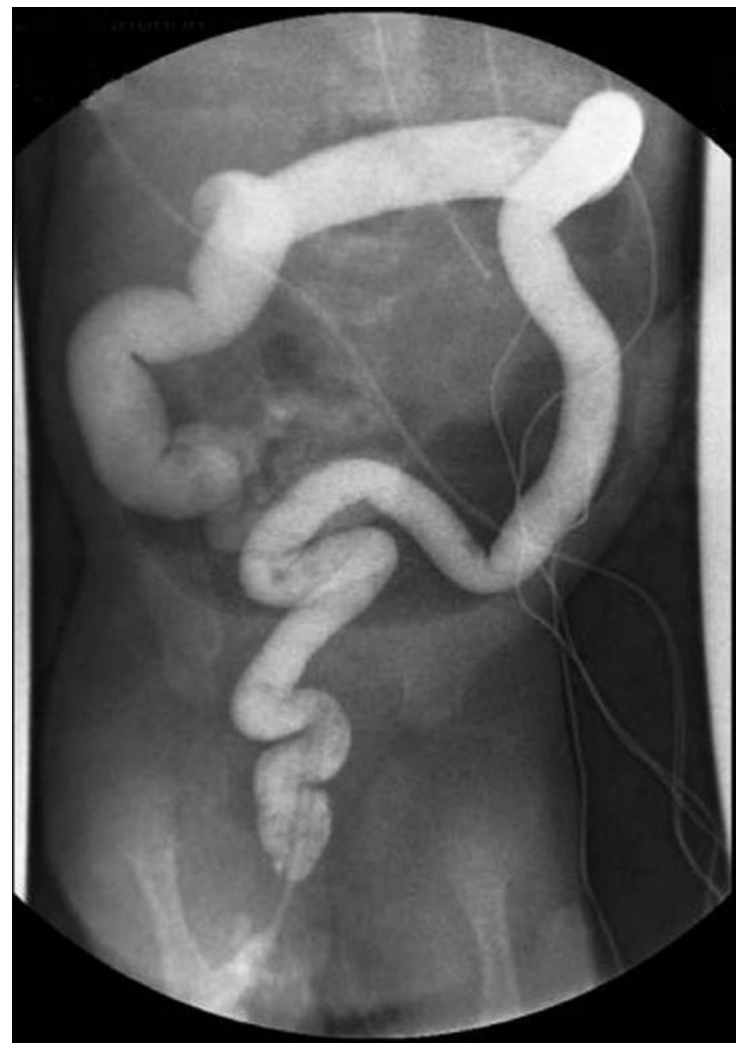
Long segment involvement

Rectal irrigations do not help.

Needs a diverting colostomy or ileostomy to relieve obstruction.

Barium enema shows- Question Mark sign.

Question mark sign



Hematoxylin and eosin

Stains for Acetylcholinesterase

Immunological staining with calretinin

Histopathology

The gold standard for the diagnosis is the absence of ganglion cells in the submucosal and myenteric plexuses on histological examination.

Most patients will also have evidence of hypertrophied nerve trunks.

PREOPERATIVE PREPARATION

- Once the diagnosis of HD is made, the child should be
  - appropriately resuscitated with intravenous fluids
  - treated with broad-spectrum antibiotics
  - Nasogastric drainage
  - rectal decompression using rectal stimulation and/or irrigation.

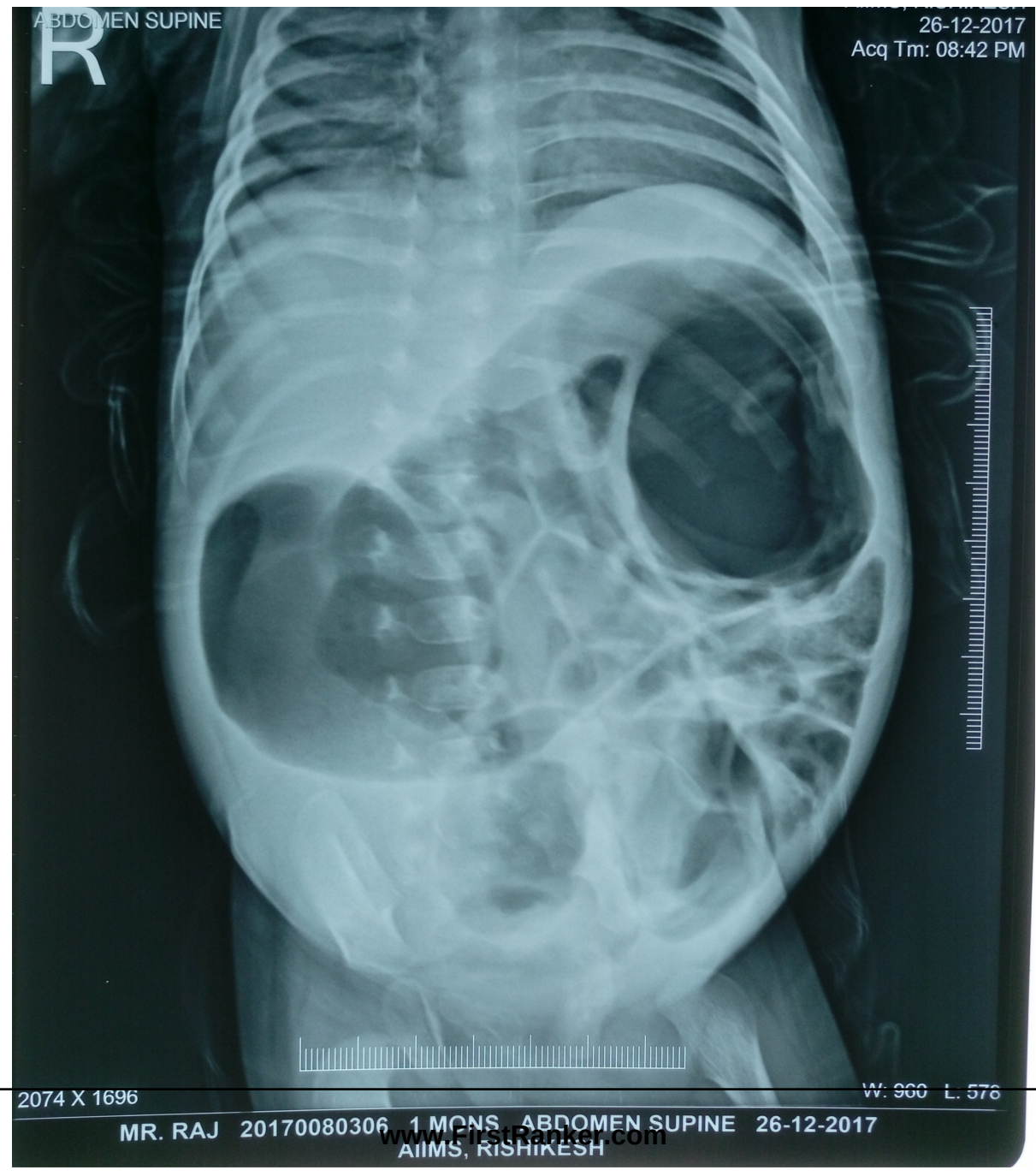
Once the infant or child has been resuscitated and stabilized, the operation can be done semi-electively.

While waiting, many infants can be discharged home on breast milk or an elemental formula, in combination with rectal stimulations or irrigations.

In the older child with an extremely dilated colon, pull-through should be delayed until the diameter of the colon has decreased sufficiently to perform a safe procedure.

Weeks or months of irrigations may occasionally be needed.

Some of these children may need an initial colostomy to adequately decompress the dilated colon.



SURGICAL MANAGEMENT

Goals:

- Remove the aganglionic bowel
- Reconstruct the intestinal tract by bringing the normally innervated bowel down to the anus
- Preserve normal sphincter function.

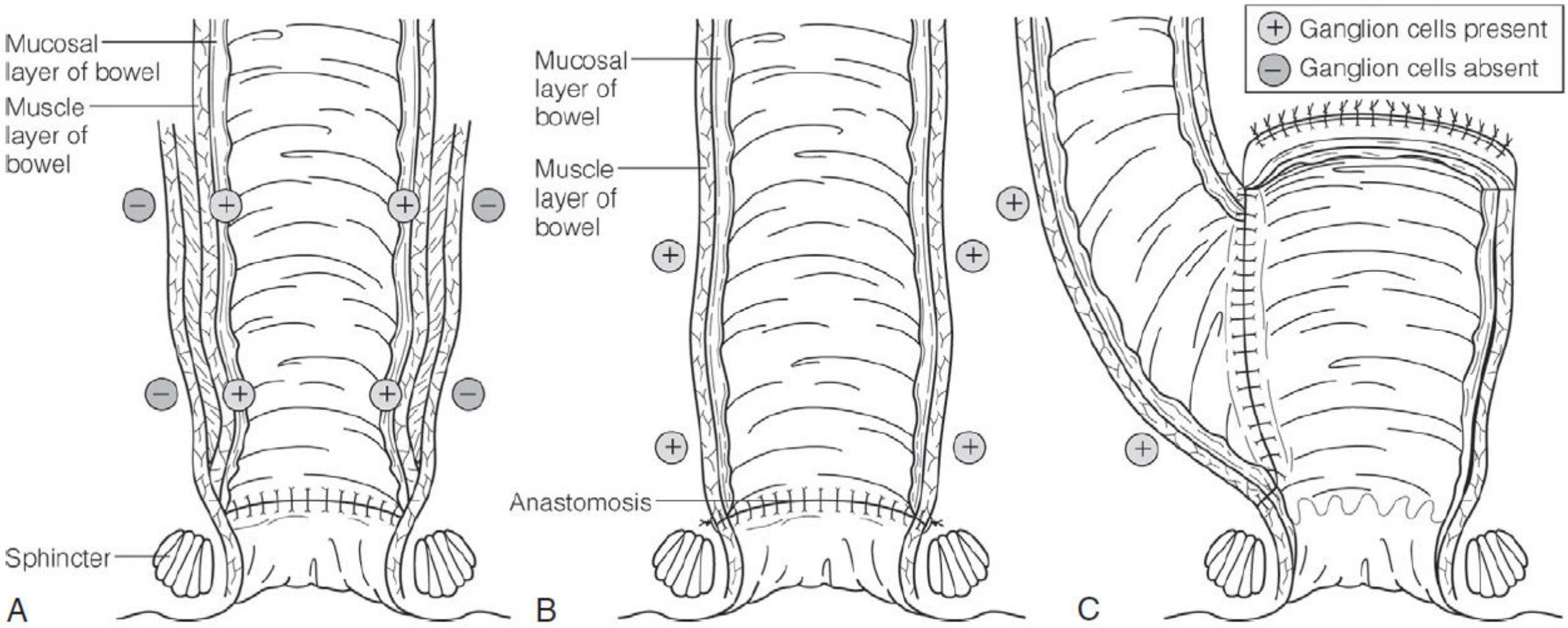
Name of surgery: Pull through procedure

Stages:

- Single stage
- Three stages: Levelling colostomy+ Pull through procedure + Colostomy closure

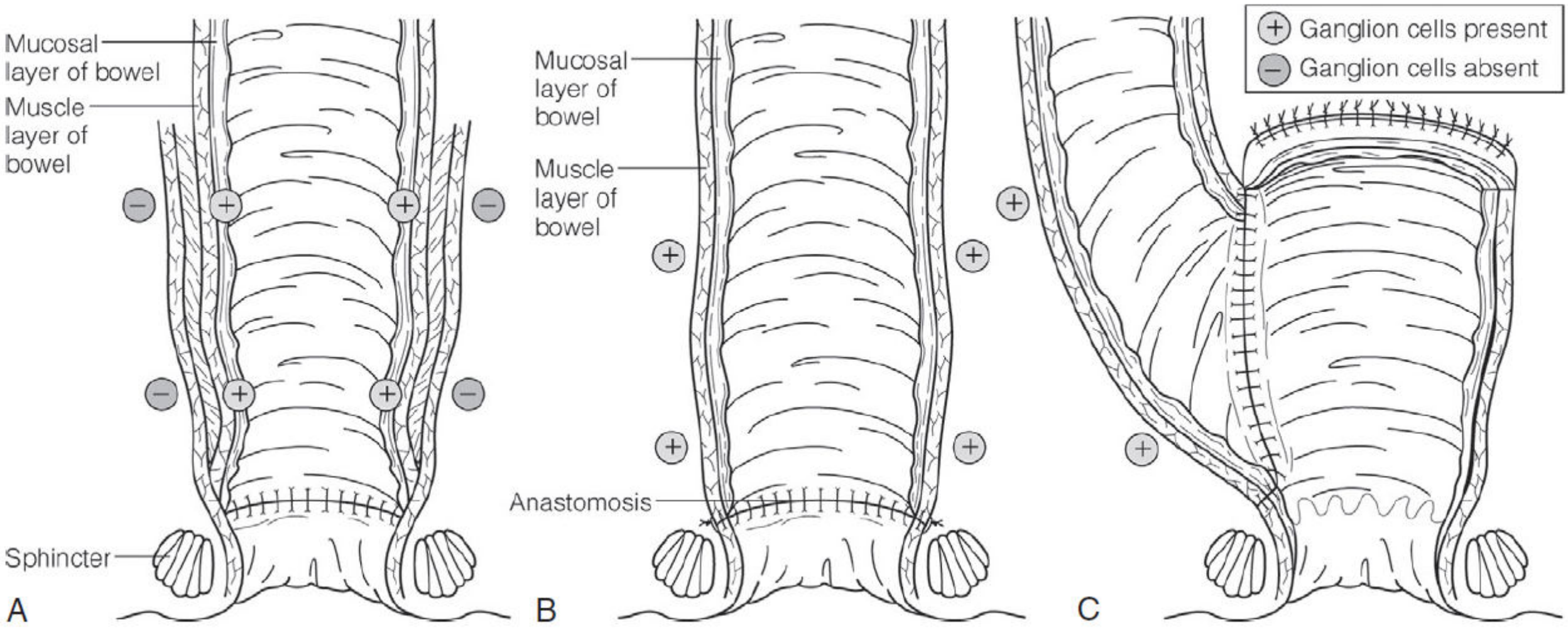
Swenson Procedure

- The goal of the Swenson pull-through is to remove the entire aganglionic colon, with an end-to-end anastomosis above the anal sphincter.



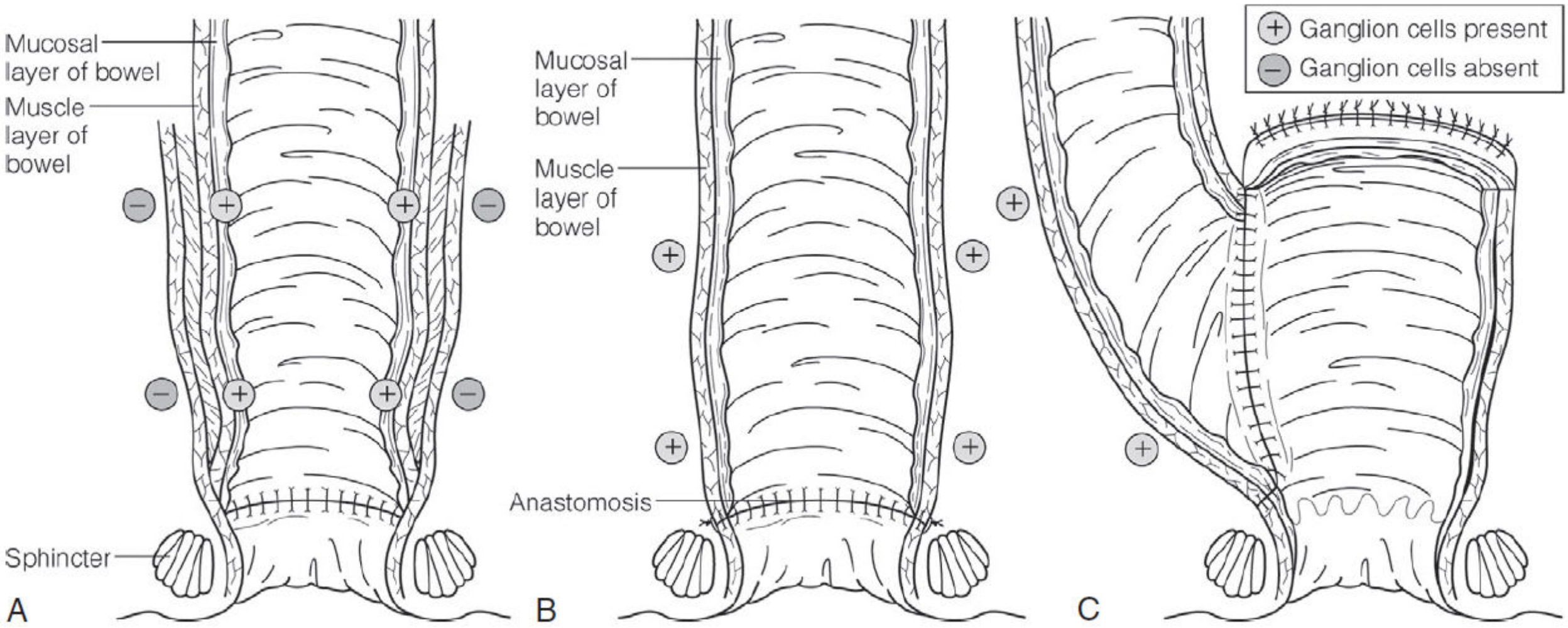
Soave Procedure

- Designed to avoid the risk of injury to important pelvic structures by performing a submucosal endorectal dissection and positioning the pull-through bowel within an aganglionic muscular ‘cuff’



Duhamel 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 which is aganglionic anteriorly and normally innervated posteriorly.



Other approaches

- Transanal pull through
- Laparoscopic pull through

Complications

- Anastomotic leaks are the most common early complication of the Duhamel and Swenson techniques, at a rate of up to 11 per cent for the Swenson technique.
  - The management of a leak is usually to form an enterostomy and then allow the anastomosis to heal.
  - Depending on the degree of anastomotic disruption, and subsequent stricturing, redo pull-through ultimately may be required.

- Cuff abscess or retraction of the proximal bowel may occur with the Soave technique and increase the chance of long-term stricturing.

- Voiding dysfunction may occur postoperatively with any of these operations.
  - Bladder innervation may be disrupted partly at the time of surgery, rather than it being a congenital problem.
  - This is more common in older children and with the extensive pelvic dissection of the Swenson technique

Long term complications

- Long-term complications include
  - anastomotic stricture
  - recurrent enterocolitis
  - constipation
  - faecal incontinence

- Anal dilatation may be required in Soave's procedure and Swenson's procedure post-operatively for avoiding anastomotic stricture

Anastomotic stricture

- Stricture is very rare after the Duhamel operation, where, alternatively, a rectal spur or diverticulum may be left or may develop with time.
  - Presents as recurrent enterocolitis, constipation.
  - Division of the spur is required- Usually done endoanal approach.

Enterocolitis

Enterocolitis may occur at any time.

Infectious agents such as *Clostridium dificile* or rotavirus have been postulated as being causative.

The incidence does decrease with age.

Maturation of the gut mucosal immune defences may account for this decrease.

Needs aggressive treatment.

Constipation

- Constipations occurs most commonly because of disordered motility in the residual colon or small bowel- Use of laxatives and prokinetic agents
- internal sphincter achalasia- Internal sphincterotomy, local application of botulinum toxin or nitroglycerine application
- functional megacolon caused by stool-holding behaviour- Bowel management by behaviour modification

Faecal soiling/ incontinence

- Abnormal sphincter function may be due to sphincter injury during the pull-through.
- There are two forms of abnormal sensation.
  - The first is lack of sensation of a full rectum.
  - Inability to detect the difference between gas and stool
- Biofeedback training, dietary modifications are used for preventing faecal soiling.

- Overall survival has improved.
- Late deaths are due to:
  - Recurrent enterocolitis
  - Associated comorbidities