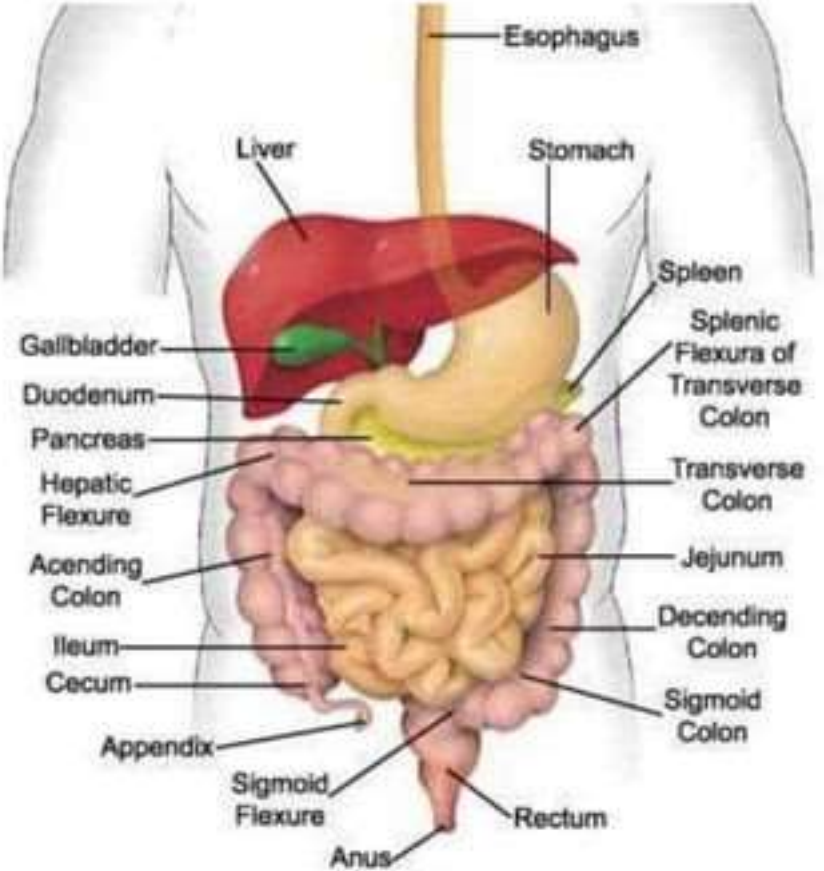
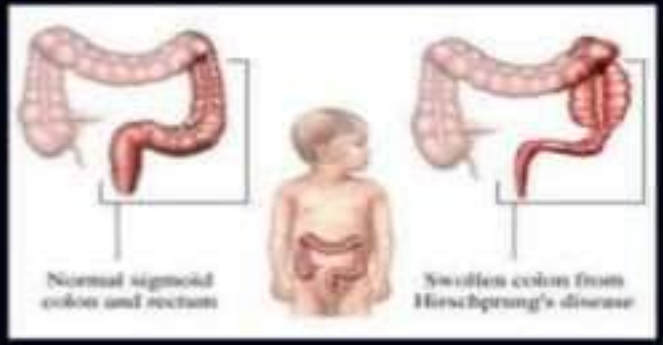
HIRSCHSPRUNG'S DISEASE/CONGENITAL MEGACOLON

Mr.S.Aravindakumar M.Sc (N).,
Assist Professor,
Dept of Paediatric Nursing,
SSCON


Esophagus
Liver
Stomach
Spleen
Splenic Flexura of Transverse Colon
Gallbladder
Duodenum
Pancreas
Hepatic Flexure
Transverse Colon
Acending Colon
Jejunum
Decending Colon
Ileum
Cecum
Sigmoid Colon
Appendix
Sigmoid Flexure
Rectum
Anus

Introduction

- Hirschprung's Disease, aslo known as Congenital Megacolon or Aganglionic Megacolon an abnormality in which certain nerve fibres are absent in segments of the bowel, resulting in severe bowel obstruction.
- Although this condition was first described by Ruysch in 1691, it was later identified and popularized in 1886 by Dr. Harold Hirschprung.

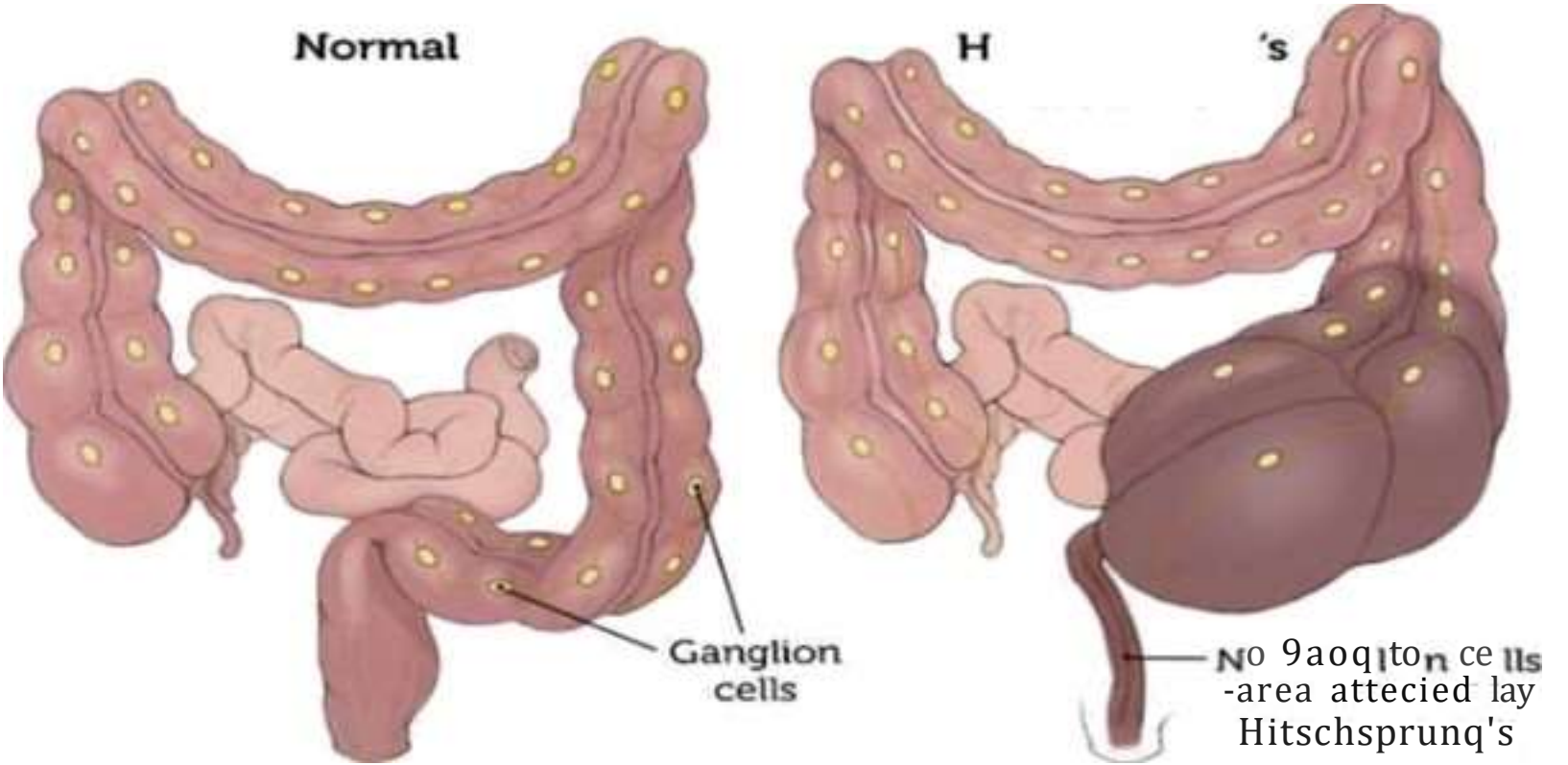


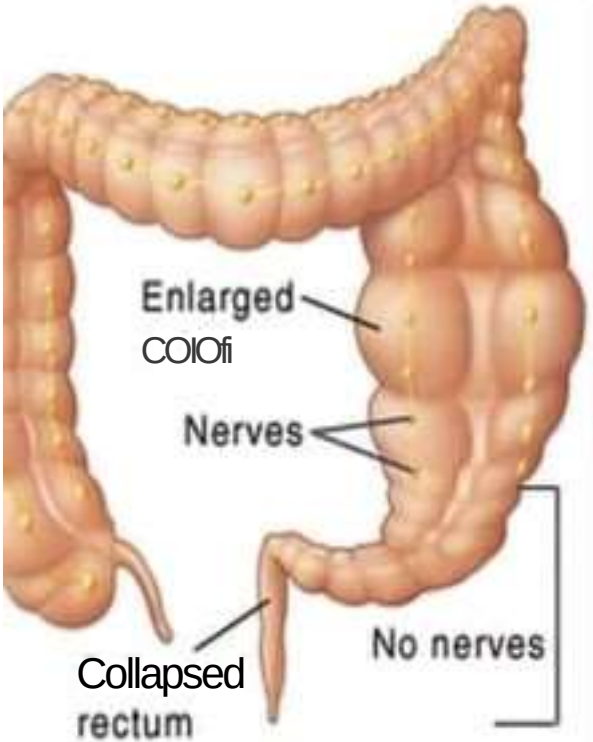
What is Congenital Megacolon?

Children suffering from congenital megacolon are devoid of the nerves that exist in the large intestine. As a result, the waste of the digestive system cannot seep through the colon.

HIRSCHSPRUNG'S DISEASE/CONGENITAL MEGACOLON

- It is a disorder of the gut caused due to congenital absence of gangalion cells in the submucosal and myentric plexus of intestine.This disease is also known as megacDlon or cDngenital Agang1ion!c megacolon.


Normal
H
's
Ganglion cells
No 9aoqlton ce lls
-area attecied lay
Hitschsprunq's

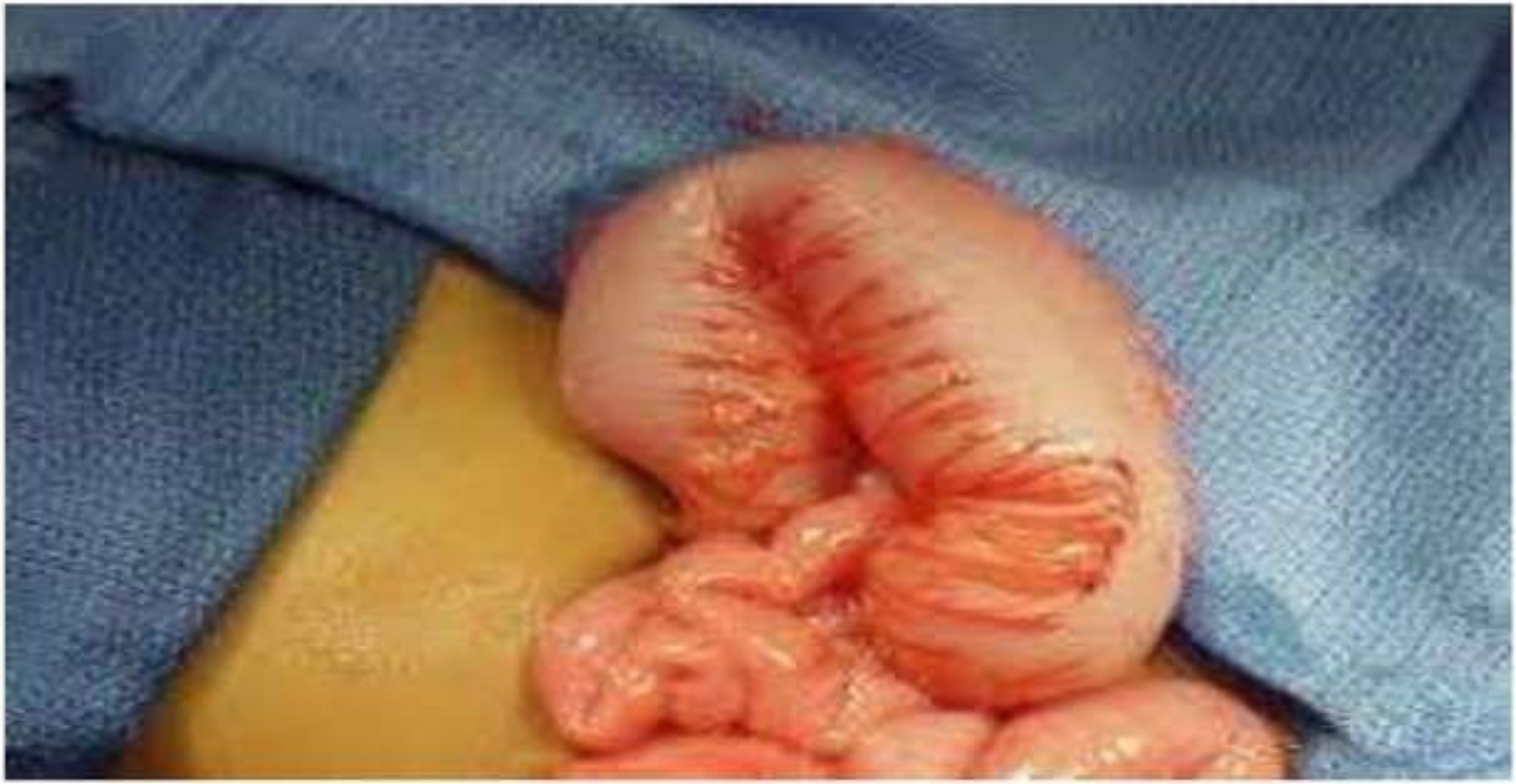


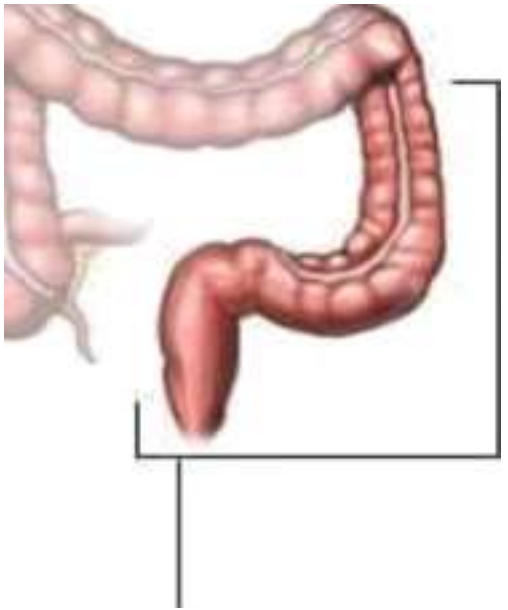
healthy
section ”-- ---

diseased
section
(without
nerve cells)

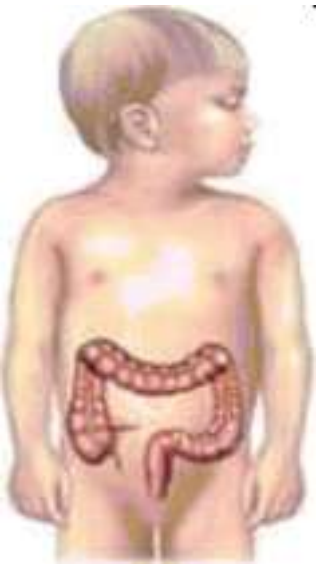
rectum

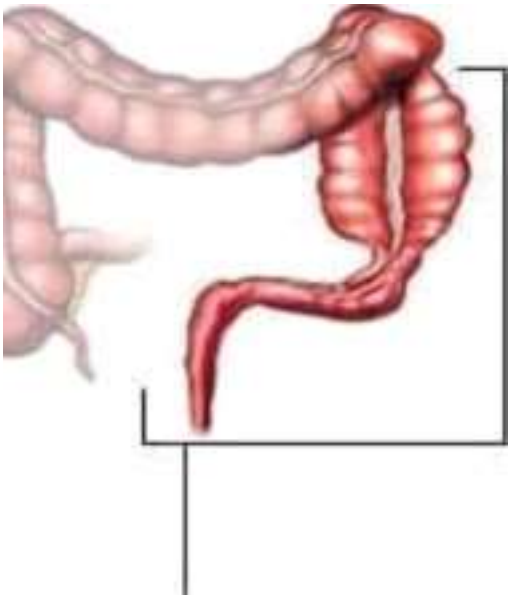
—anus





NonnøJ sigmoid colon ønd rectum



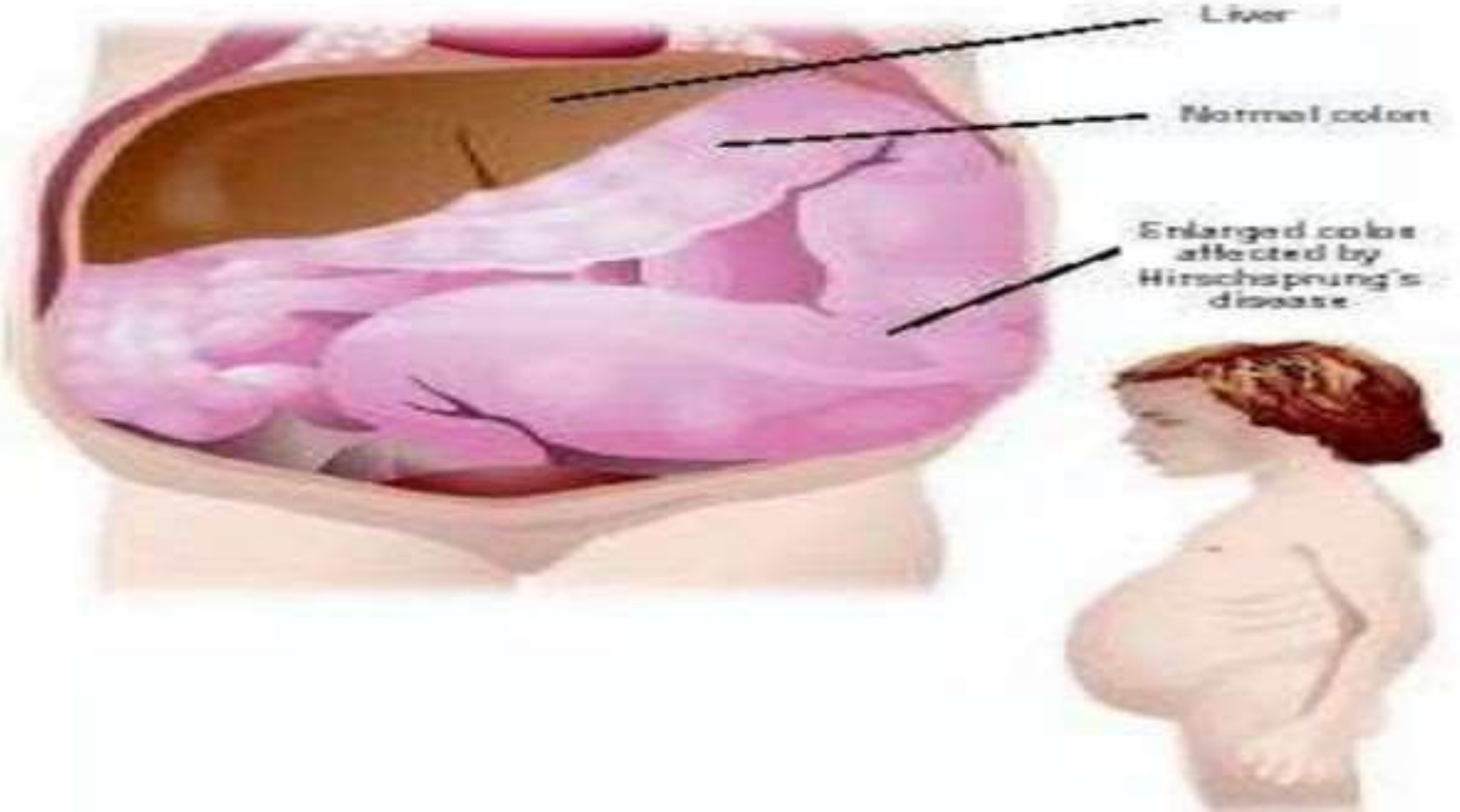


Swoümi aeon Earn Hinæhpœngs Œæææ

EPIDEMIOLOGY

500O live birth newborn

- 70-80% is boys. (M / F. 4: 1)
- Łscs common in blacks.


Liver
Normal colon
Enlarged colon affected by Hirschsprung's disease

TYPES

T. Cang&nitaT : This type is the corrsnonest one .

a o if zhe disease is still onkno we. bu Gcoetle factors are n ow

°/elO pf cases have farnllialhistory, especiafty th ose w it h 1snd segment dtseas e.

* Acquired :

Degeneration of the ganglions mny occur due to:

Vosculor co•'- like after pluTIthrough proceclure due to isclnemia & tension.

- **Non vascular causes** like

Tryp anosorria (chas•'s Qiseasc).

vic e• def.

Chronic •ntection t TB.}.

ASSOCIATED ANOMALY

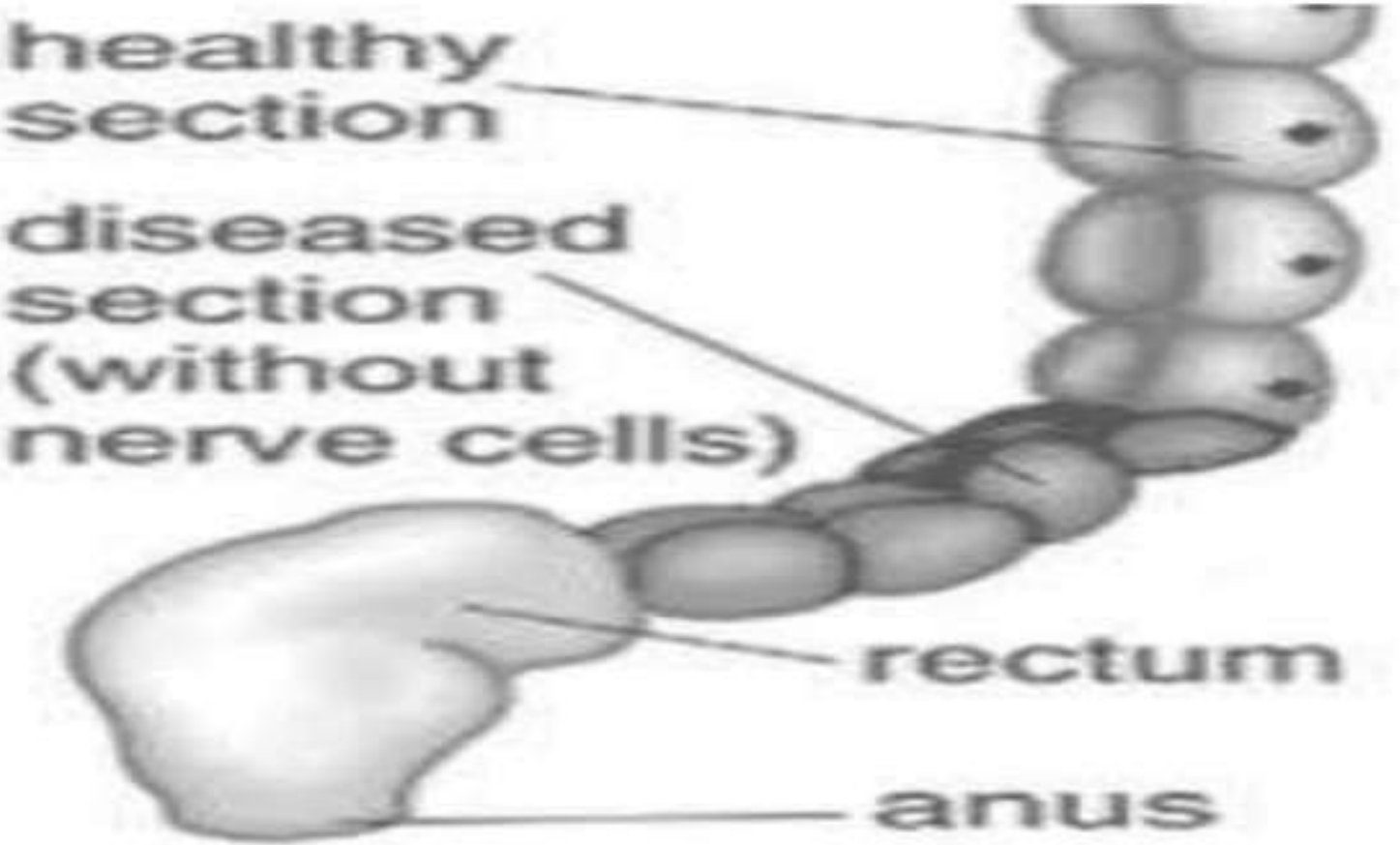
- HD is usually a ***solitary anomaly*** in a full term, otherwise healthy infant
- Associated anomalies do occur in nearly **20%** of cases
  - urogenital system (11%)
  - cardiovascular system (6%)
  - gastrointestinal system (6%),
  - with 8% having various other malformations

- **Prematurity** is reported in as many as **10%** of those children with HD
- **Trisomy 21** occurs in approximately **5%** of cases

Embryology and Etiology

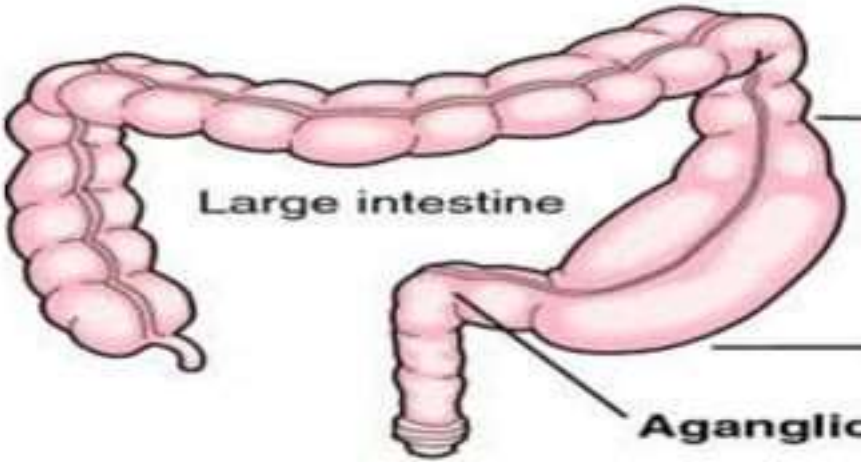
- Neuroenteric cells migrate from neural crest to upper end of alimentary tract by the 5th week of gestation ,and proceed towards intestine by 7th week.

- By 12th week : migration to distal colon occurs– first into myenteric (Auerbach's plexus) then into submucosal (Meissner's plexus) plexus.


healthy section
diseased section (without nerve cells)
rectum
anus

- Failure of neural crest migration.
- Mutations in the genes RET EDNRB, GDNF, NRTN, ET3, ZFHX1B,PHOX2B, SOX10, and SHH have been found in 50% patients in a 2013 survey.
- Absence or dysfunction of fibronectin, laminin, neural cell adhesion molecule (NCAM), and other neurotrophic factors.

- It is caused by congenital absence of autonomic parasympathetic ganglion cells in the submucosal and myenteric plexus of the intestine



Megacolon

Dilated with feces and gas

Ganglion cells present (normal in function)

Aganglionic region

Tonic contraction and inadequate peristalsis

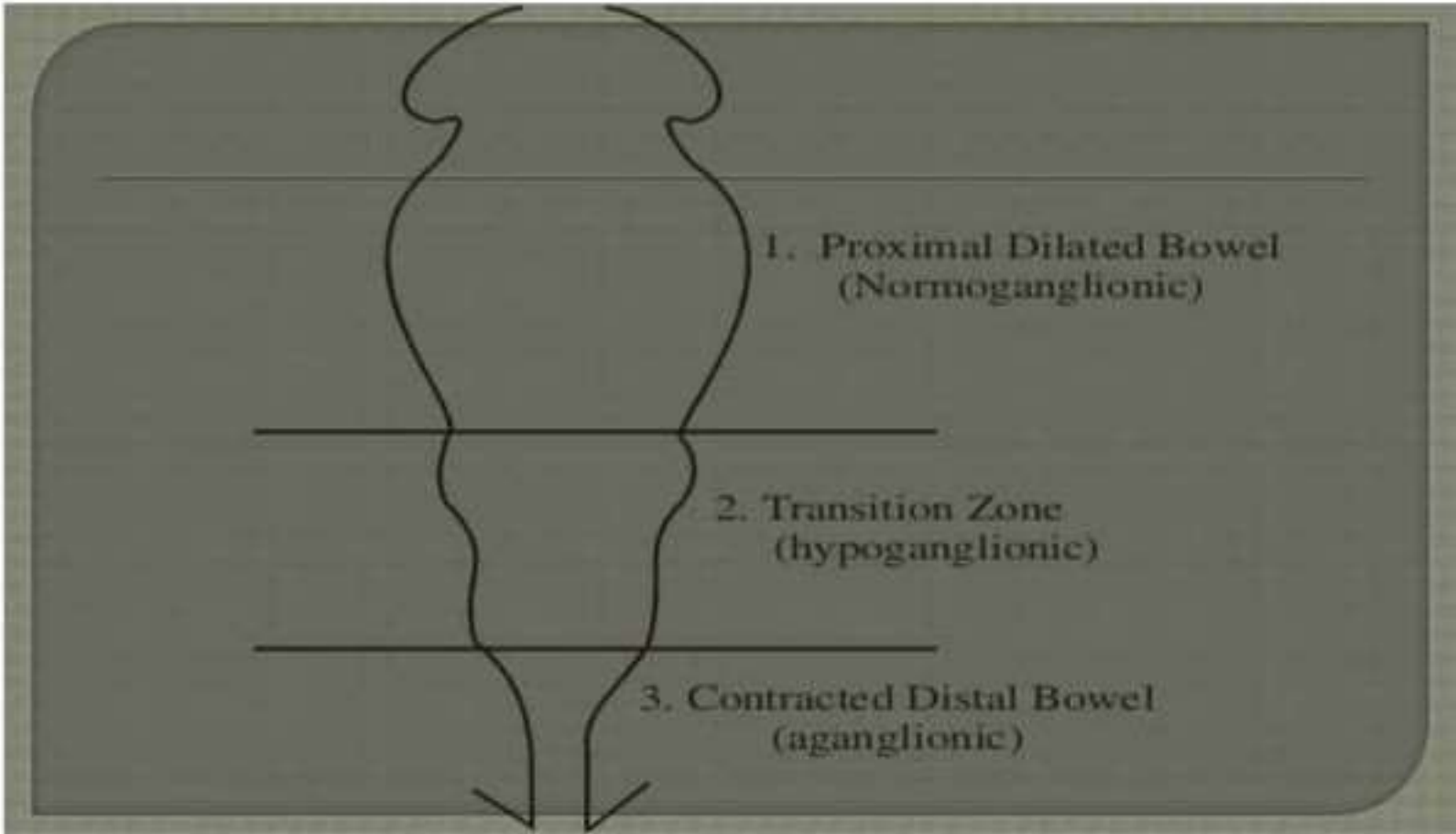
Partial obstruction

Chronic constipation

Enlargement of proximal segment

Complete bowel obstruction

Enterocolitis


1. Proximal Dilated Bowel
(Normoganglionic)
2. Transition Zone
(hypoganglionic)
3. Contracted Distal Bowel
(aganglionic)

Treatment:

Decompression: introduce a rectal tube and irrigation

Chronic constipation :

- laxativ e
- saHne enenna.
- Walk up to estabHsh the cliagnosis
- + then th e deflnitive treatment wifi be planned

There are m any surgic al optfon s for F'uII-through operation. All aiming at resection of agangfionic segment ancl anastomosing the two n oiwial ganglionic ends. They give exceil ent result in go:a.

a. swenson.
b. soave.
c. Rehbeln.
d. Duhame).
e. Boley's

Hirschsprung's disease

Treatment

Transanal Endorectal P.ul(-lrough

- It can be performed safely in infant as well
- Generally one-stage surgery
- No abdominal phase
- The anastomosis is happening ina „safe" place at the pectinate line

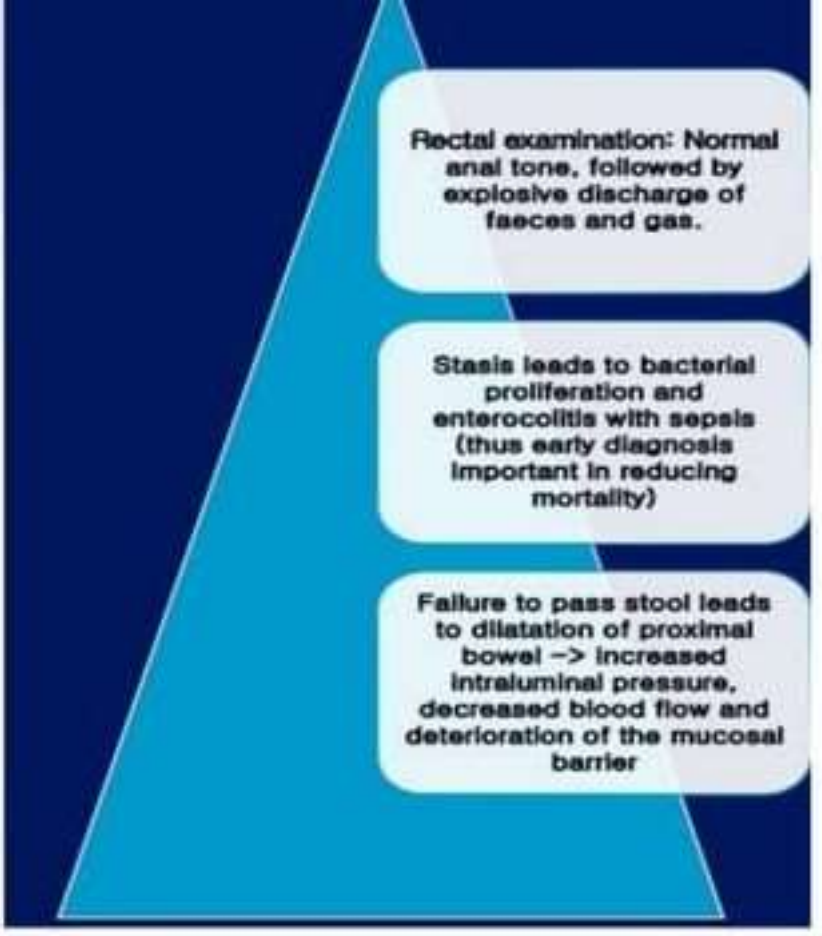
COMPLICATION

anastornotic 1ea k.
stricture
retraction of th e colon.

4. fecal incontinen ce (snili rig or encopresis).
5. persTstant constlpatlon

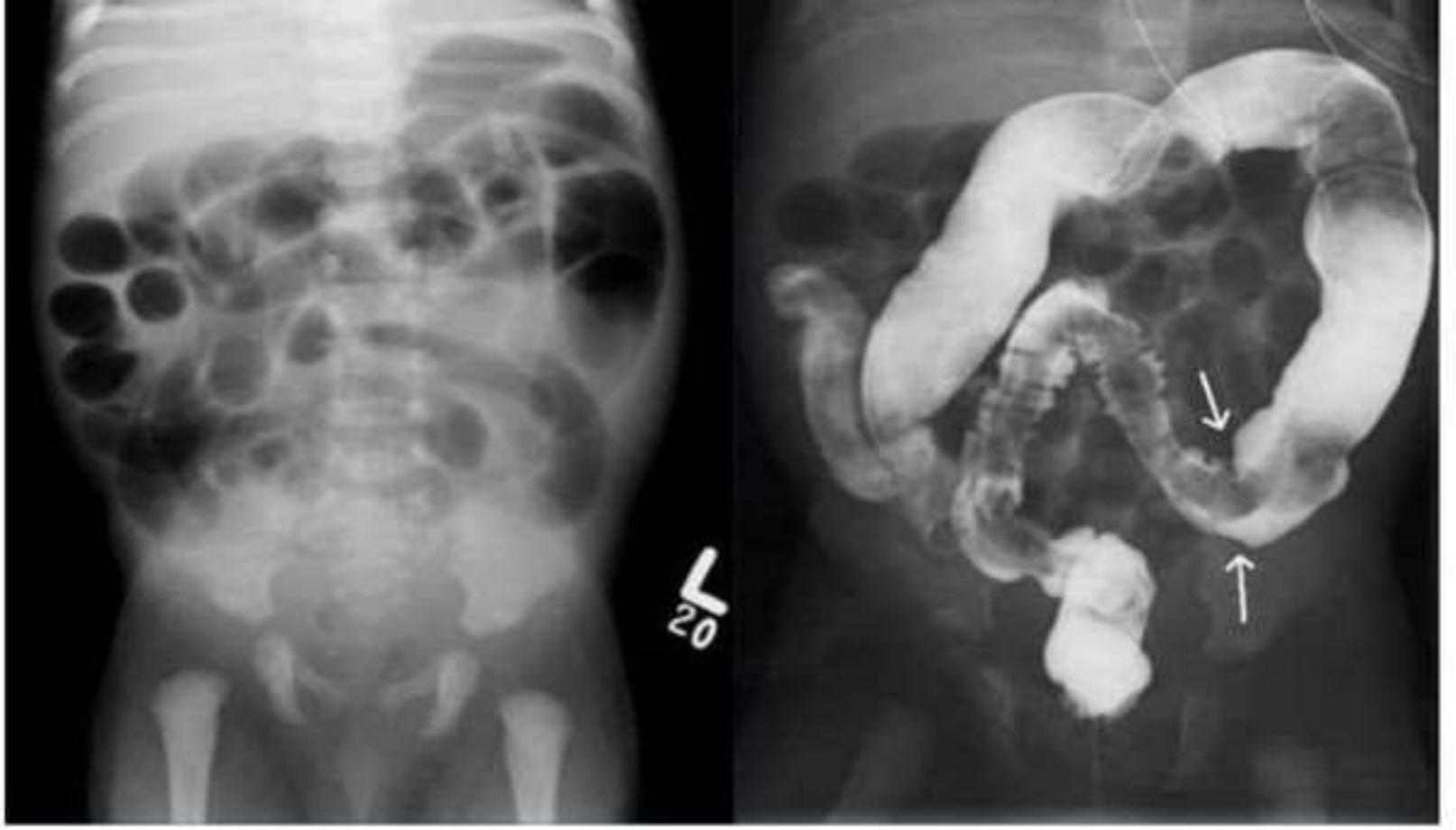
Clinical Symptoms

- Hirschprung's Disease should be considered in any child who has history of constipation dating to newborn period, 90% of cases are diagnosed in newborn period
- Most common presentation in newborns: delayed stool passage within first 48 hours of life
- Constipation, abdominal distension, poor feeding and vomiting;
- Constipation followed by explosive diarrhea, failure to thrive;
- In older children, large fecal mass palpable in left lower quadrant while the rectum is empty
- Stool Character: small pellets, ribbon-like, fluid consistency


Rectal examination: Normal anal tone, followed by explosive discharge of faeces and gas.
Stasis leads to bacterial proliferation and enterocolitis with sepsis (thus early diagnosis important in reducing mortality)
Failure to pass stool leads to dilatation of proximal bowel -> increased intraluminal pressure, decreased blood flow and deterioration of the mucosal barrier

Diagnosis I

- Abdominal Straight X-rays: Air fluid levels in colon and distended loops of intestine.
- Barium contrast enema: narrow distal segment and dilated proximal intestine; funnel-shaped transition zone between these 2 segments (diagnostic accuracy 80-90%)
- Significant barium remaining in colon in 24-hr delayed film helpful in determining level of Aganglionosis.


L
20

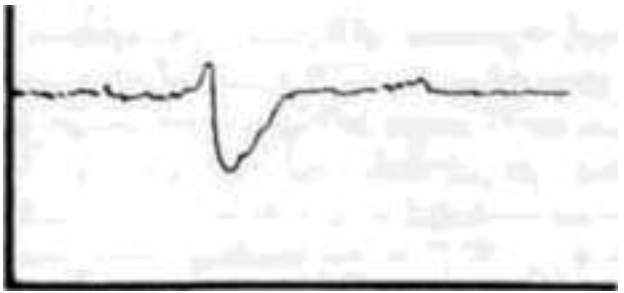
Diagnosis II

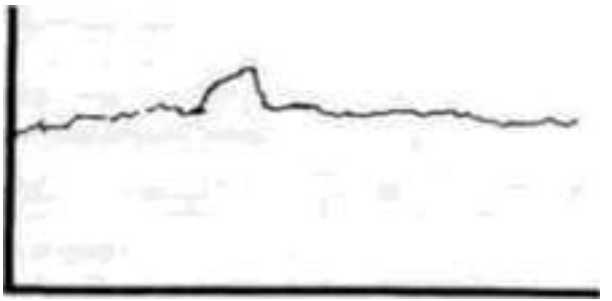
- Anorectal manometry: Shows absence of relaxation reflex after distension of balloon in rectum. Diagnostic accuracy 85%.
- May be done at bedside or as OPD procedure without complications
- Unreliable in cases where gestational age plus age after birth is less than 39 weeks and weight is less than 2.7kg.

N 0 RM1L

uiR s Cn spR urge' s

ANAL CANAL





RE CT AL Dł SY EN TîON

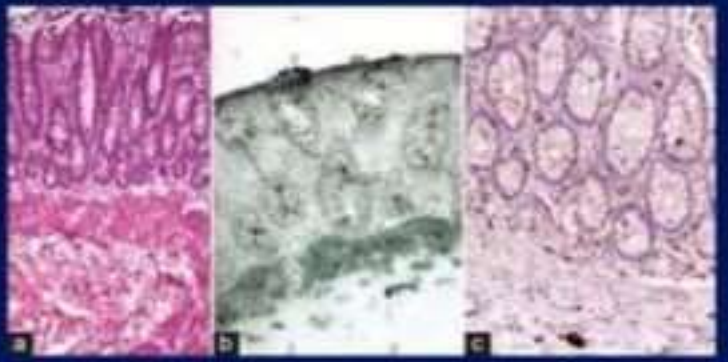
Diagnosis III

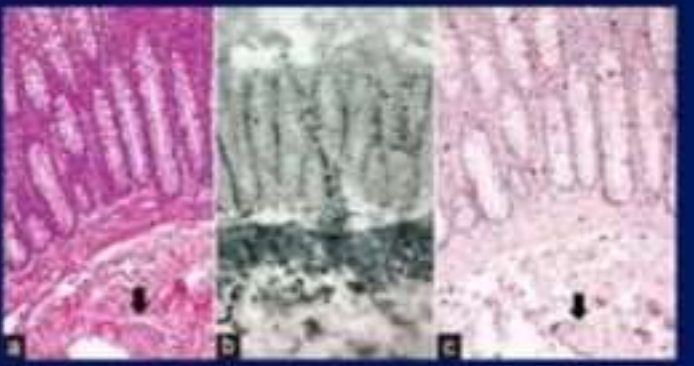
- Rectal suction biopsy: Gold standard for Hirschprung's Disease
- It can be performed at bedside without general anesthesia.
- Biopsy taken at 2 cm, 3 cm, 5 cm above dentate line.
- Diagnostic accuracy: 99.7%

Suction Biopsy

Non–Hirschsprung disease: Normally innervated rectal mucosa (a) showing no increase in Acetylcholinesterase activity (b), and calretinin staining positive fibers in the mucosa and muscularis mucosa

Hirschsprung disease: Rectal mucosa (a) showing hypertrophic nerve bundle in the submucosa. Increase in Acetylcholinesterase activity (b) of pattern A and negative staining with calretinin (c) note the negative staining of hypertrophic nerve fibre (arrow) with calretinin





Differential diagnosis

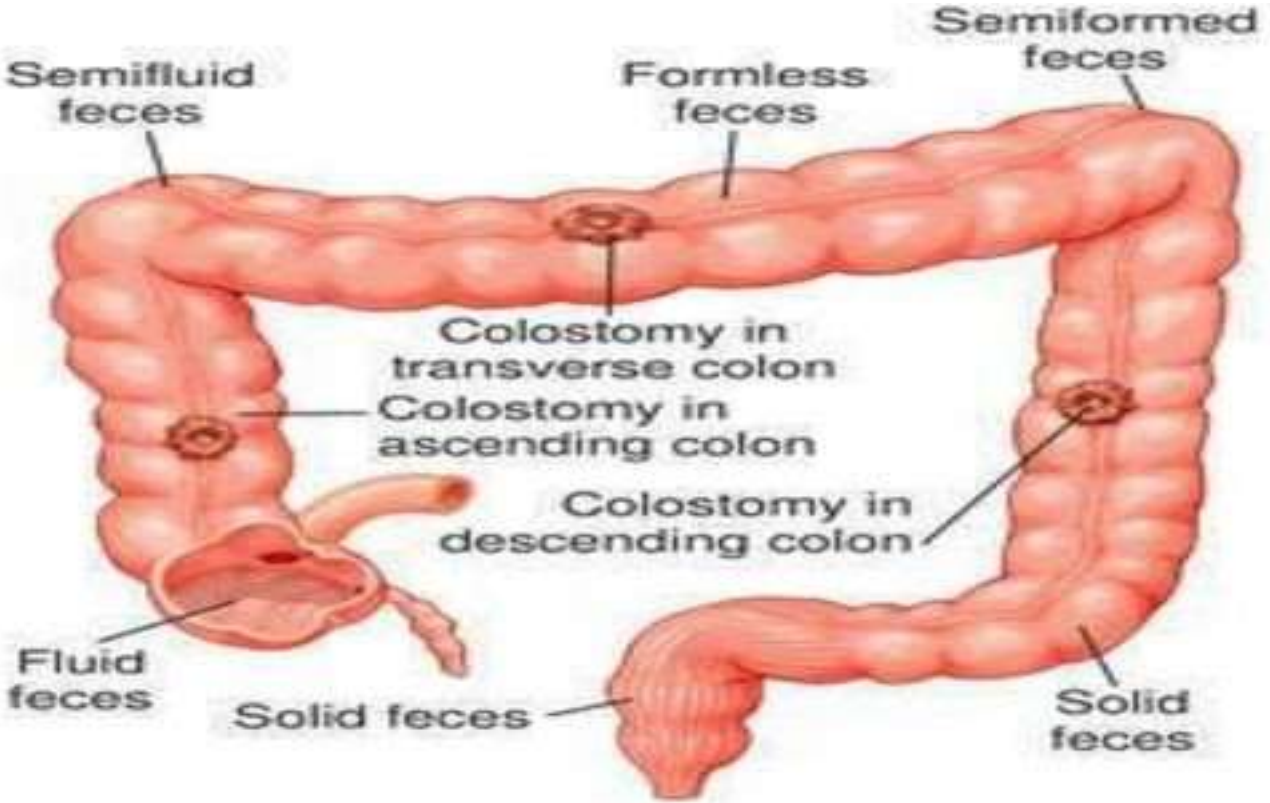
- Meconium plug syndrome
- Small left colon syndrome
- Distal ileal atresia
- Cystic fibrosis (meconium ileus)
- Low imperforate anus
- Prematurity
- Hypothyroidism
- Brain injury
- Neonatal Sepsis

MEDICAL MANAGEMENT

- Administration of isotonic enema
- Administration of stool softeners
- Law rRsidue diet

Surgical management

- The aim of the surgery is to remove the aganglionic bowel followed by anastomosis of the remaining portion.
- The surgery involves twD steps
- In first stage
- A temporary colostomy is done above the transition zone of ganglionic and aganglionic bowel in the sigmoid or transverse colon.This enables the normal distal bDwel to return to its orginal tone and size.
- Second stage involves definitive surgery ,which is done when the child weight and condition is appropriate.


Semifluid feces
Formless feces
Semiformed feces
Colostomy in transverse colon
Colostomy in ascending colon
Colostomy in descending colon
Fluid feces
Solid feces
Solid feces

° Pul! through procedure; Excision of aganglionic segment enabling an anastomosis to be done by corrective Surgeries

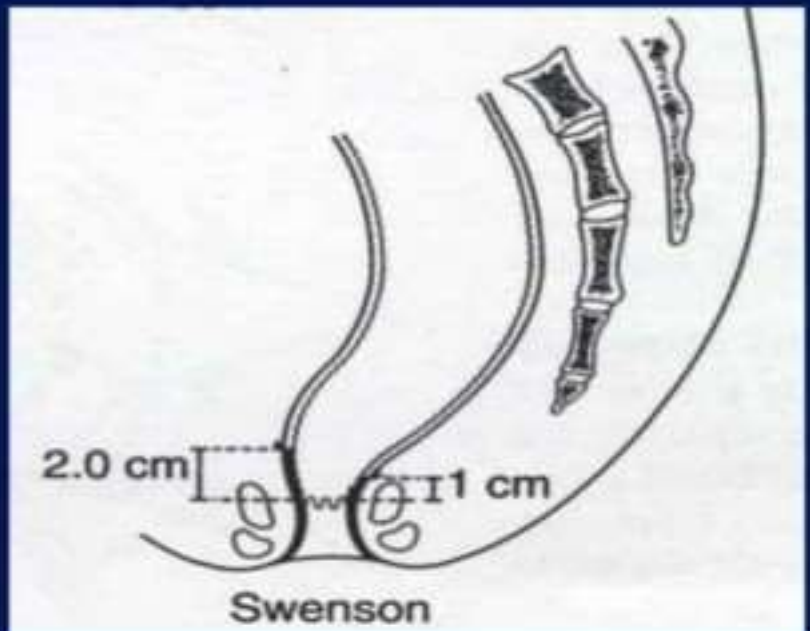
Treatment

- Decompression
- Surgery done only after diagnosis is complete.
- 3 basic surgical approaches:
- Swenson's, Duhamel's & Soave's

Swenson's

Excision of aganglionic segment and anastomosis of the normal proximal bowel with the rectum 1–2 cm above the dentate line.

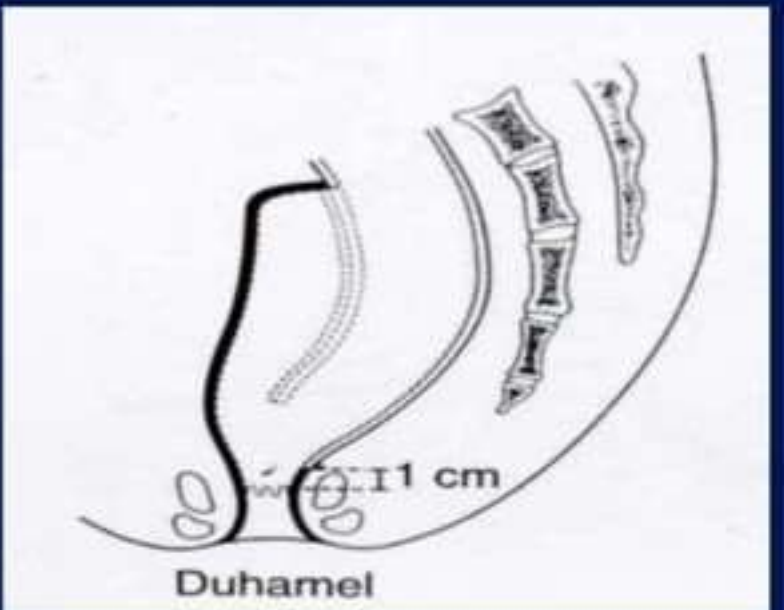
Complication: Enterocolitis

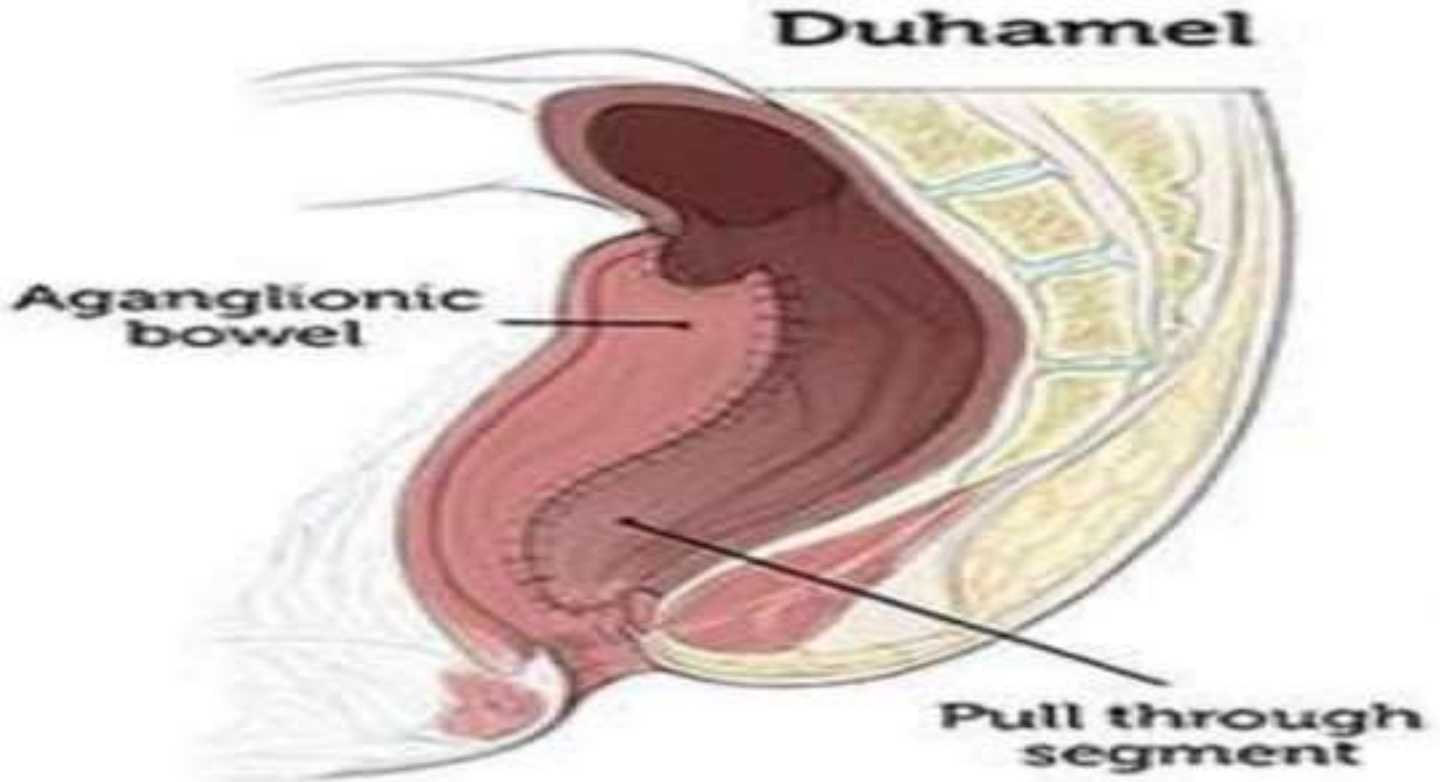


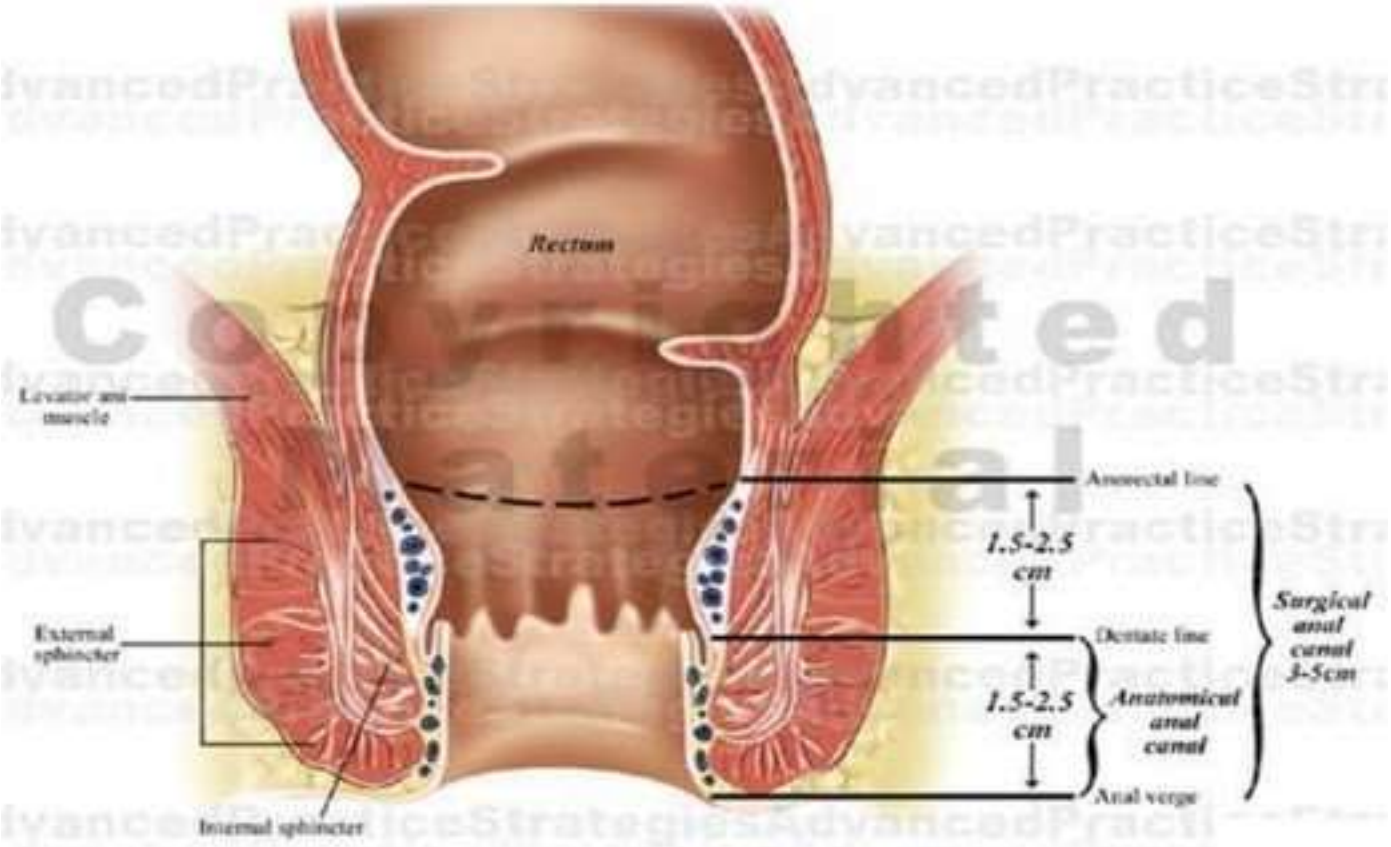
Duhamel's

Neorectum created-> Normally innervated bowel is brought down behind aganglionic rectum (anterior aganglionic half with normal sensation and posterior half ganglionic with normal propulsion)

Complication: Constipation



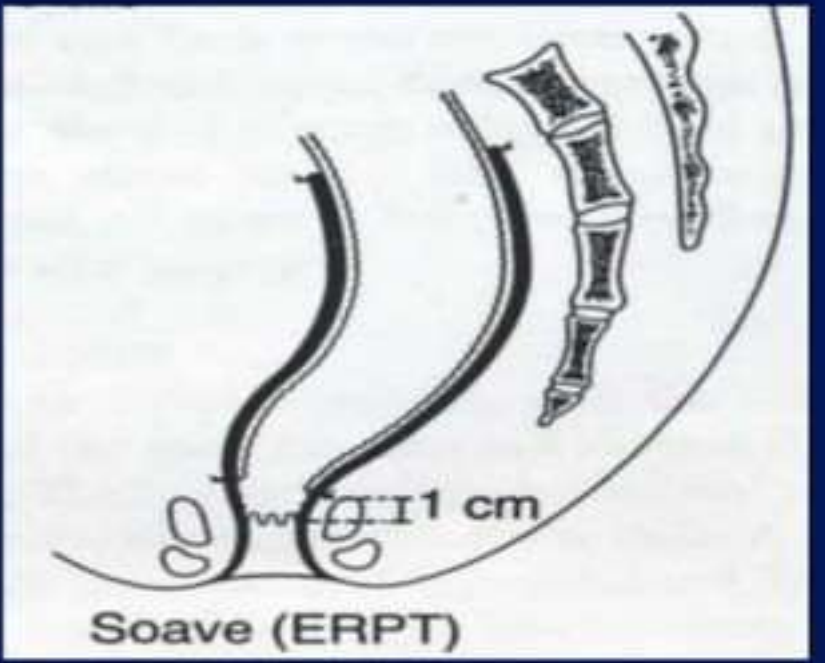

Duhamel
Aganglionic bowel
Pull through segment


Rectum
Levator ani muscle
External sphincter
Internal sphincter
Anorectal line
1.5-2.5 cm
Dentate line
1.5-2.5 cm
Anatomical anal canal
Anal verge
Surgical anal canal 3-5cm

Soave's

Endorectal pull-through procedure -> Mucosa stripped from the aganglionic rectum and normally innervated colon is brought through the residual muscular cuff.

Complication: Diarrhoea and incontinence



Complications

- Early complications: anastomotic strictures (15%), wound infections (11%), anastomotic leaks (7%)
- Late complications: chronic constipation, enterocolitis, chronic obstruction, encoporesis, recto vesical fistula
- Good prognosis: more than 90% of children achieve normal bowel movement.

THANK YOU