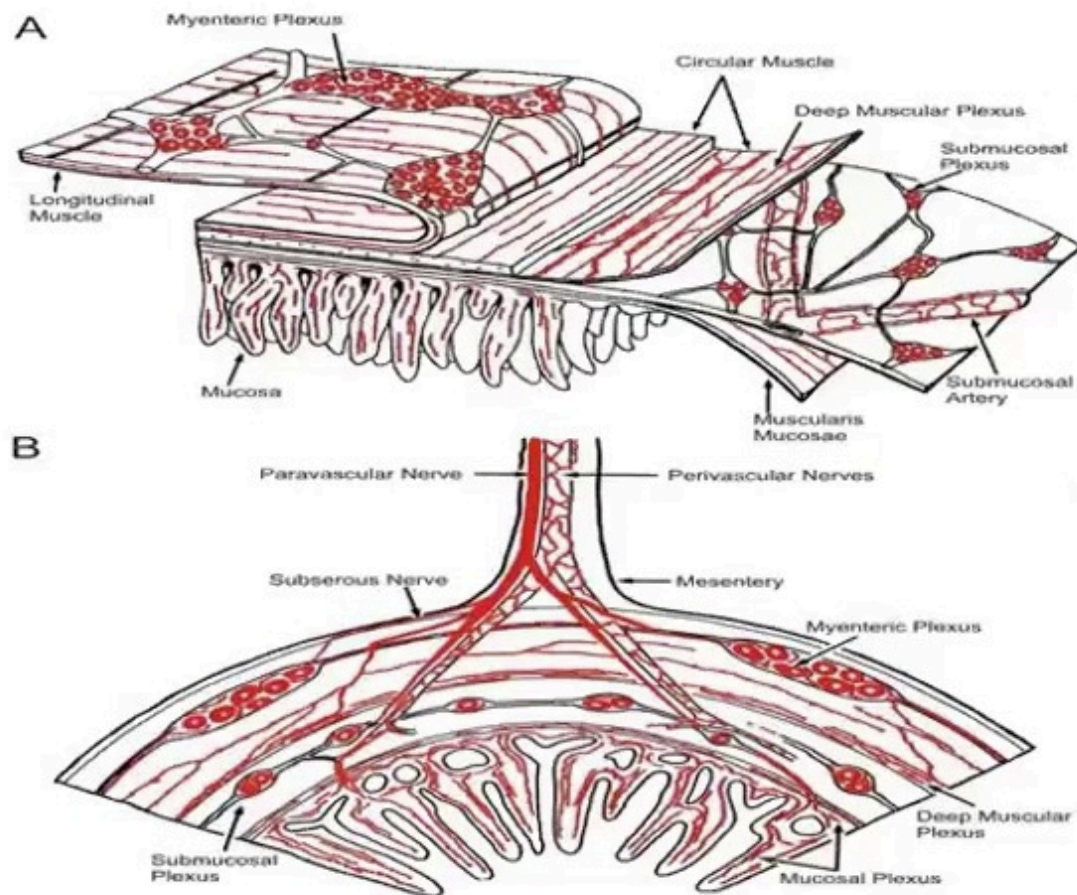


CONGENITAL AGANGLIONIC MEGACOLON (HIRSCHSPRUNG'S DISEASE)



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Prof. Harald Hirschsprung



1830 - 1916

- ✓ Native of Copenhagen.
- ✓ Major interest on rare diseases of GIT. His doctoral thesis was on atresia of esophagus and small bowel: 1861
- ✓ He became the first Danish pediatrician in 1870 and worked at Queen Louise Children's' Hospital
- ✓ In 1904, when he was 74 years old, he was forced to resign from his practice due to cerebral sclerosis

- ✓ In 1886, at a Berlin Congress for Children's Diseases, he spoke of **2 infants** who had died from a disease characterised by constipation associated with dilatation and hypertrophy of the colon.
- ✓ He believed the proximal dilated bowel was the pathology.
- ✓ The absence of ganglion cells in the distal colon of a child with Hirschsprung disease was first noted by Tittel in 1901.
- ✓ In 1949 Swenson published a paper in the ***New England Journal of Medicine*** recommending rectosigmoidectomy with preservation of the sphincters as the optimal treatment

INTRODUCTION

- ❑ Hirschsprung's disease is a developmental disorder of the intrinsic component of the enteric nervous system
- ❑ Characterized by the absence of parasympathetic ganglion cells in the myenteric and submucosal plexuses of the distal intestine.
- ❑ The resulting aganglionic segment of the colon fails to relax, causing a functional obstruction.

Epidemiology

- ❖ Incidence – 1/5000 live births
- ❖ Short segment(rectosigmoid)
 - ✓ M:F = 4:1
- ❖ Longer segment
 - ✓ Decreased sex ratio, approaches 1:1 in total colonic aganglionosis
- ❖ A proportion of cases are familial (20%), but HSCR most commonly presents with a sporadic occurrence

Aetiology

- ❑ The most accepted theory is that there is a defect in the craniocaudal migration of neuroblasts originating from the neural crest that occurs during the first 12 weeks of gestation.
- ❑ Differentiation of neuroblasts into ganglion cells and accelerated ganglion cell destruction within the intestine may also contribute to the disorder
- ❑ The enteric microenvironment may become inhospitable for colonization by crest-derived cells if EDN3 is deficient or if the EDNRB is lacking

Pathophysiology: Neural crest migration

- ✓ NC-derived cells first appear in the developing esophagus at the 5th week of gestation
- ✓ Craniocaudal migration to the rectum by 12th week of gestation
- ✓ NC cells first form the myenteric plexus
- ✓ After the craniocaudal migration has ended, the submucous plexus is formed by the neuroblasts which migrate from the myenteric plexus across the circular muscle layer and into the submucosa
- ✓ This progresses in a craniocaudal direction during the 12th to 16th week of gestation

- ✓ The absence of ganglion cells in Hirschsprung's disease has been attributed to a failure of migration of vagal NC cells.
- ✓ The earlier the arrest of migration, the longer the aganglionic segment is.
- ✓ The colonization of the gut by sacral NC-derived cells and the contribution of the cells to the development of the ENS is controversial
- ✓ Fujimoto et al. studied NC cell migration in the developing gut in the human embryo using antineurofilament protein triplet antibody and found that enteric ganglia originated from a single vagal NC source.

Pathology: CLASSIFICATION

1. Ultra-short segment
2. Short segment. confined below the rectosigmoid junction (70%)
3. long segment- below the splenic flexure(20%)
4. Total colonic aganglionosis (9%) – appendix is involved.
5. Total intestinal involvement(2-5%)

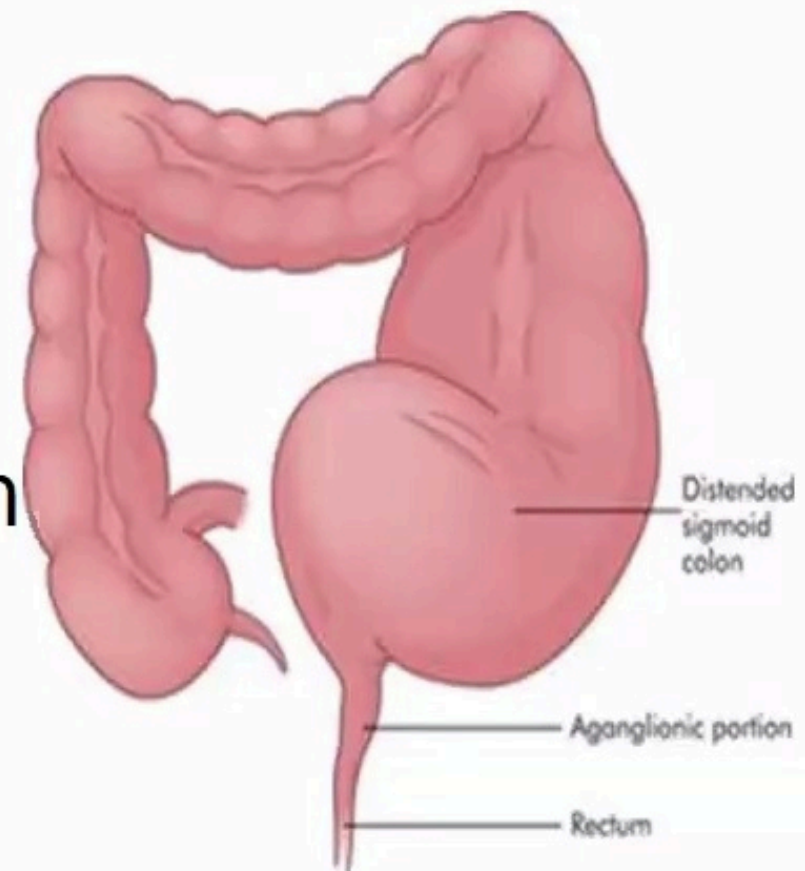
The functional abnormality always includes the internal anal sphincter

Macroscopic features

✓ Narrow aganglionic segm

✓ A transitional zone

✓ Dilated proximal portion with a thickened bowel wall as a result of hypertrophy of the muscular wall of the intestine



Clinical presentation

- ✓ Depends on extent and age of patient
- ✓ DRE important to rule out anorectal malformations
- ✓ Delayed meconium passage
 - 95% of normal children pass meconium within 24 hours
 - 10% of children with Hirschsprung's pass meconium within 24 hours

Clinical presentation: infants

- ✓ Decreased defecation frequency: constipation
- ✓ Requires digital stimulation/suppositories
- ✓ Decreased appetite
- ✓ FTT
- ✓ Intermittent episodes of obstruction and diarrhoea(enterocolitis)

Older patient

- ✓ long history of constipation
- ✓ Abdominal distention
- ✓ Malnutrition/FTT
- ✓ Large fecal masses
- ✓ Do not bleed or have anal pain – more consistent with functional constipation
- ✓ Often have extensive enema programs

Diagnosis

1. History

2. Clinical examinations

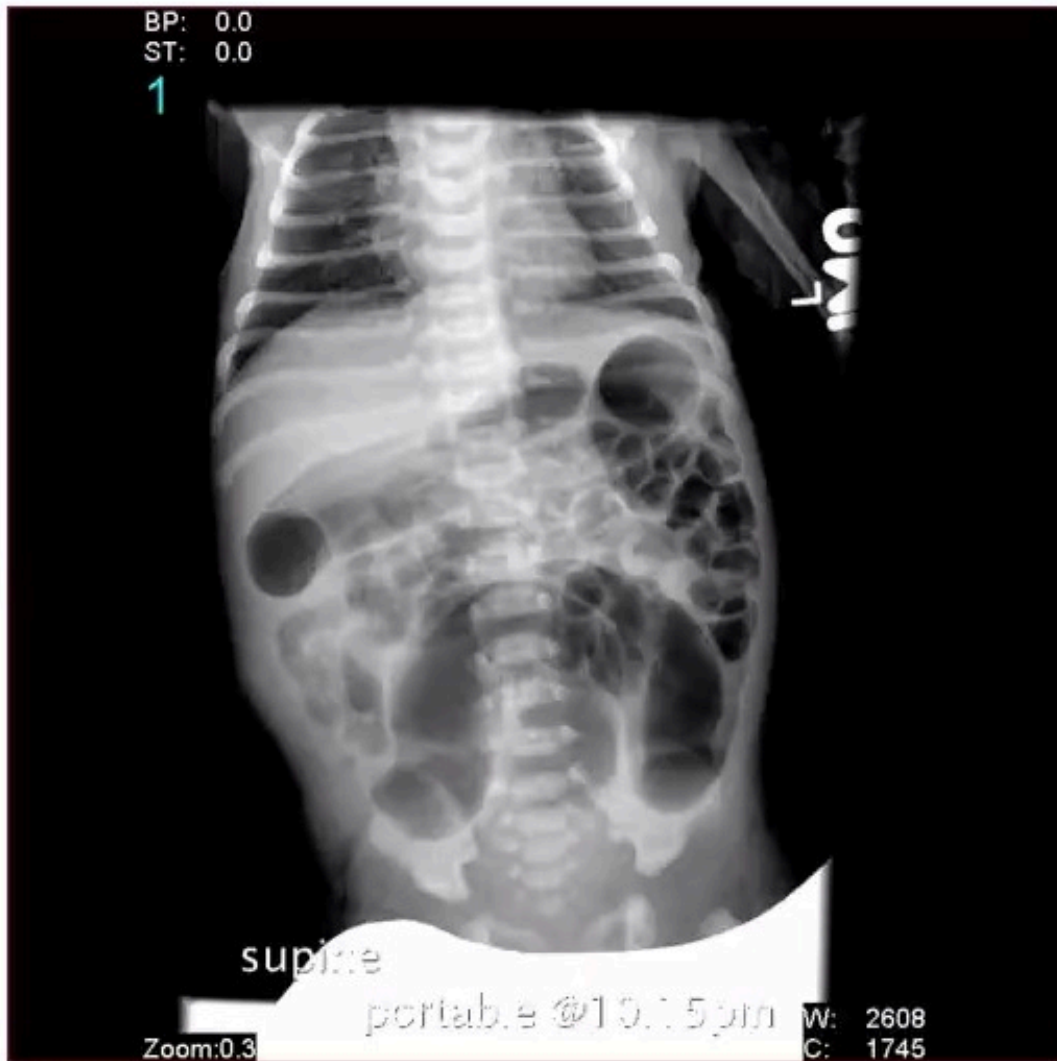
3. Radiology

4. Histopathology

Abdominal x-ray

The diagnostic accuracy of abdominal x-rays is 52%.

1. Look for signs of low intestinal obstruction and distended bowel
2. loops of different calibres
3. Erect plates can demonstrate in fluid levels.
4. A lateral view may demonstrate the narrow rectum



Frontal abdominal radiograph showing marked dilatation of the bowel with no gas in the rectum.

Diagnosis

1. Barium enema

- ❖ most important view is the lateral projection, in which a rectal transition zone will be most evident
 - Rule out obstructing lesion e.g meconium plugs in neonate
 - Rectal calibre $\leq$ calibre of rest of colon
 - Hallmark – conical transition from proximal dilated to distal narrowed colon
 - approximately 10% of neonates with Hirschsprung disease may not have a demonstrable radiologic transition zone
 - Small colonic calibre with rounded flexures suggests aganglionosis

- ❑ Irrigation of the colon before the contrast must not be performed as it may result in possible decompression of the megacolon or the distended bowel



Lateral view from a barium enema examination depicting the reduced diameter of the rectum and sigmoid





- ❑ The aganglionic segment may be irregular, demonstrating a sawtooth appearance, probably as a result of muscular fasciculations.
- ❑ A delay in the clearing of contrast (barium sulphate) within 24 hours is also a reliable sign, and a follow-up x-ray should be performed the following day.

Ano-rectal manometry

Investigations

- Rectal manometry:
 - Helpful In older children who present with chronic constipation and an atypical history.
 - Children with HD fail to demonstrate reflex relaxation of the internal anal sphincter in response to inflation of a rectal balloon.



Rectal biopsy

- Full thickness – gold standard
- Looks at the plexi
- Important to get biopsy 2-3cm(3-5 in older children) above dentate line
- Disadvantage: GA, presacral abscess
- Alternatives – punch biopsy, suction biopsy
- stains:

- Hematoxylin&Eosin** - identify ganglion cells
 - difficulties in identifying immature ganglion cells from plasma cells or lymphocytes.
- Ach esterase staining** - show an increased activity in the parasympathetic nerves of the affected zone as well as neurofibrils within the lamina propria and muscularis mucosa

Histological findings in HSD

1. Absence of ganglion cells in the submucosal layer and intermyenteric(Auerbach's) plexus
2. Presence of the enlarged peripheral nerve trunks in the submucosa
3. Increased AChE staining—proliferation of neurofibrils in the lamina propria and the muscularis mucosa (absent in normally innervated intestine)

Differential Diagnosis

Neonates

- Small left colon
- Meconium plug
- Meconium ileus
- Megacystis-megacolon-intestinal hypoperistalsis syndrome
- Intestinal dysmotility
- Hollow viscous myopathy
- Chronic intestinal pseudo-obstruction

Infants and children

- trauma, spina bifida
- Anal pathology, fissure
- IND
- endocrine. DM, hypothyroidism
- Psycosocial

management

1. The initial aim of management is to confirm the diagnosis
2. The second aim is to correct the problem (i.e., relieve the intestinal obstruction) by means of definitive surgery.

Principles of Surgical Management

- ☐ A defunctioning colostomy followed by a definitive pull-through procedure
- ☐ Bowel irrigation techniques allows for early one-age surgery to be performed at a much earlier stage (mostly still within the neonatal period).
- ☐ Colostomy may still be necessary if nursing care is unreliable

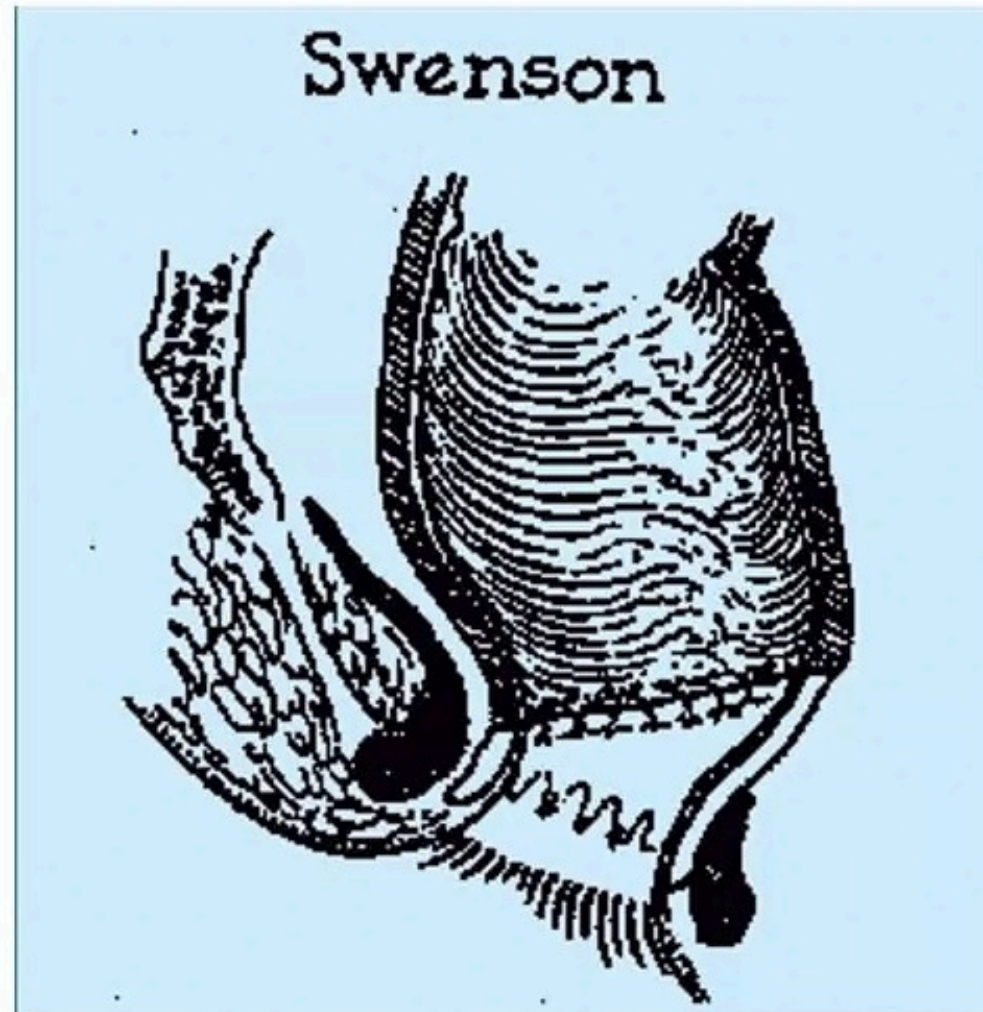
SURGICAL PROCEDURES

1. SWENSON PROCEDURE
2. DUHAMEL PROCEDURE
3. SOAVE PROCEDURE
4. LAPAROSCOPIC PULL-THROUGH
5. TRANSANAL (PERINEAL) PULL-THROUGH
6. POSTERIOR MYOECTOMY with or without lateral sphincterotomy

Swenson Procedure

- ✓ Sharp extrarectal dissection down to just above the anal canal
- ✓ Aganglionic colonic segment resected
- ✓ End-to-end anastomosis of normal proximal colon to anal canal
- ✓ Completely removes defective aganglionic colon

Swenson Procedure



Operative complications

- ❑ Leak at anastamosis: 5-7%
- ❑ Post-op Enterocolitis: 19-27%
- ❑ Constipation
- ❑ Stricture Formation
- ❑ Incontinence

THANK YOU

