



- Congenital defect
- Incidence: 1:5000 live births
- M:F = 4:1
- 85% of the time limited to the rectosigmoid segment of the colon

PATHOPHYSIOLOGY

Migration failure of neural crest cells to the distal colon



Absence of ganglion cells in the submucosal and myenteric plexuses



Absence of peristalsis in the aganglionic colon



Functional obstruction



CLINICAL PRESENTATION

NEWBORN

- Failure to pass meconium in first 24 hours after birth
- Bowel obstruction with bilious vomiting



OLDER CHILD

- Severe chronic constipation
- Failure to thrive, malnutrition and feeding problems



PHYSICAL EXAM

- Abdominal distension
- Digital rectal exam:
 - Empty rectum
 - Explosive ejection of stool as the finger is removed ("squirt sign")



INVESTIGATIONS

- TSH (rule out hypothyroidism), electrolytes (rule out hypercalcemia, hypokalemia)
- Abdominal X-Ray → will show dilated loops of intestine
- Contrast enema → will show the "transition zone" between the narrow rectum and distal colon and the dilated proximal colon
 - Contraindicated if enterocolitis is suspected → risk of perforation
 - For infants > 12 months old → anal manometry can show absence of rectoanal inhibitory reflex

DIAGNOSIS:

Rectal biopsy → will show aganglionosis, hypertrophy of nerve trunks, and abnormal calretinin staining

TREATMENT



Surgical "pull-through" procedure: removal of the aganglionic segment and anastomosis between the normally innervated part of the intestine and the anus, preserving the anal sphincter

HIRSCHSPRUNG-ASSOCIATED ENTEROCOLITIS



- Life-threatening complication
- Inflammation of the colon which causes bowel obstruction & sepsis
- Can occur either before or after the pullthrough surgery

Signs and symptoms

- Abdominal distension
- Explosive diarrhea
- Vomiting
- Other: fever, rectal bleeding, shock
- X-ray: megacolon (severely dilated colon)



Treatment

- Broad spectrum antibiotics
- IV fluids
- Labs: CBC, CRP, blood culture
- Nasogastric decompression
- Rectal irrigations
- General surgery consultation

