HIRSCHSPRUNG'S DISEASE

Definition: Aganglionosis of distal colon and rectum

Embryology:

Neural crest cells migrate into GI tract (5th - 10th week of gestation) from cranial to caudal in myenteric (Auerbach) and sub mucosal (Meissner) plexus.

***Can not** develop Hirschsprung's disease – it is present at birth*

Pathophysiology:

Aganglionic distal bowel interferes with bowel motility
Hypertrophy of extrinsic innervation results in increased muscle tone of anal sphincter – cannot pass flatus or stools

Incidence

1 : 5000 births
Short segment (90%) of cases. Only rectum and part of sigmoid colon involved
Long segment (10%) of cases. Rectum, sigmoid and part of colon involved
Can be genetic (family history) but mostly random.
Associated anomalies possible: cardiac, urogenital, anorectal malformation, Down's syndrome

Clinical presentation in infants

- No meconium passed within 24 hours of life
- Resonant abdominal distension
- Explosive stool discharge on rectal examination
- Poor feeding, vomiting late symptom
- Enterocolitis due to stasis in proximal dilated bowel – septicaemia can be fatal
- Perforation (3%)

Diagnosis

- *History* of no meconium / constipation since birth / never passes flatus
- *Clinical:* Very distended abdomen
  Explosive stools on rectal examination (pr)
- *AXR:* Dilated bowel loops filled with air and NO air-fluid levels
- *Contrast study:* Narrow distal segment, dilated proximal bowel, retention of contrast >24h
- *Definitive diagnosis with histology:* No ganglion cells in full thickness rectal biopsy

Emergency management

Bowel obstruction: Nasogastric tube on free drainage and npo
IV fluids: 5% Dextrose water in new-born
Paediatric surgeon to do colostomy

Enterocolitis Nasogastric tube on free drainage and npo
IV fluids: Bolus Ringers (20ml/kg), then maintenance
Saline enemas to empty colon from debris, until fluid returns clear
Antibiotics

Surgical management in 3 phases:

Diverting colostomy to relieve obstruction
Full thickness rectal biopsy to establish diagnosis
When recovered from acute disease: aganglionic bowel resected and normal bowel anastomosed to anus
Resection and colo-anal anastomosis can be done in one stage without colostomy

Clinical presentation in older children

- Severe constipation – not passing stools for 1-2 weeks!
- Problem since birth
- Never passing flatus
- Massive abdominal distension (does not fit in clothes for age-group)
- Malnutrition
- Rectum empty (on pr)

- Enterocolitis (rare)

DEGENERATIVE VISCERAL MYOPATHY

Clinical picture similar to Hirschsprung's disease in older child

BUT: This is progressive disease, normal at birth and constipation as toddler
Affects muscle of urinary tract as well
Only in black Africans
Positive family history of early death due to bowel problem
Rectal biopsy: normal ganglion cells present, need to ask for evaluation of muscle layer
No cure, die as teenagers (due to kidney failure)

Conclusion

Be suspicious
Close follow up of new-born with constipation / failed to pass meconium in 24-48h after birth
Remember 5 causes of not passing meconium:
ARM
Hirschsprung's
Small bowel atresia
Malrotation and volvulus
Meconium abnormal: cystic fibrosis/meconium ileus/sticky meconium
Better to refer patient to rule out, than to miss Hirschsprung's disease in new-born
Always take proper history of constipated child
Refer to specialist if atypical presentation of constipation / not responding to treatment