HIRSCHSPRUNG DISEASE

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OBJECTIVES

- Understand the epidemiology and pathogenesis Hirschsprung
- Recognize the classical clinical presentation
- Discuss the testing and evaluation modalities, and management

PATHOGENESIS

- Epidemiology
  - 1 in 5000 live births, overall male:female ratio of 3-4:1
- Cause due to defect in craniocaudal migration of neuroblasts between fourth week to seventh week of gestation in the myenteric and submucosal plexus of colon
  - Cells fail to reach distal colon leading to aganglionic and nonfunctional colon
  - Two types: short segment and long segment Hirschsprung
    - Short: aganglionic segment distal to splenic flexure
    - Long: aganglionic segment proximal to splenic flexure
- Associated syndromes: Down, familial dysautonomia, MEN2

CLINICAL PRESENTATION

- Neonatal
  - Bilious emesis
  - Abdominal distension
  - Failure to pass meconium or stool within 48 hours
  - Enterocolitis

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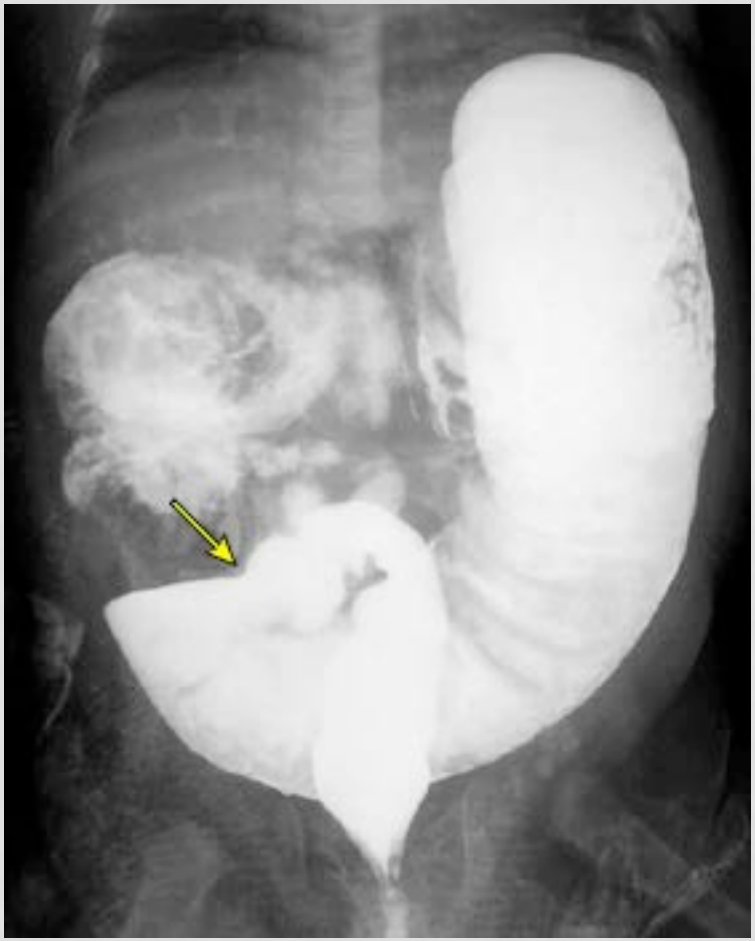
  - Tight anal sphincter or “squirt sign” on rectal exam
  - Enterocolitis
    - Fever
    - Hemodynamic instability
    - Diarrhea
    - May progress to toxic megacolon

CLINICAL PRESENTATION

- Postnatal
  - Refractory constipation with no stool in rectal vault
  - Failure to thrive
  - Tight anal sphincter or “squirt sign” on rectal exam
- Associated congenital anomalies
  - CAKUT – congenital anomalies of the kidney and urinary tract
  - Congenital heart disease (usually with Down)
  - Anorectal malformations

DIAGNOSTIC TESTING

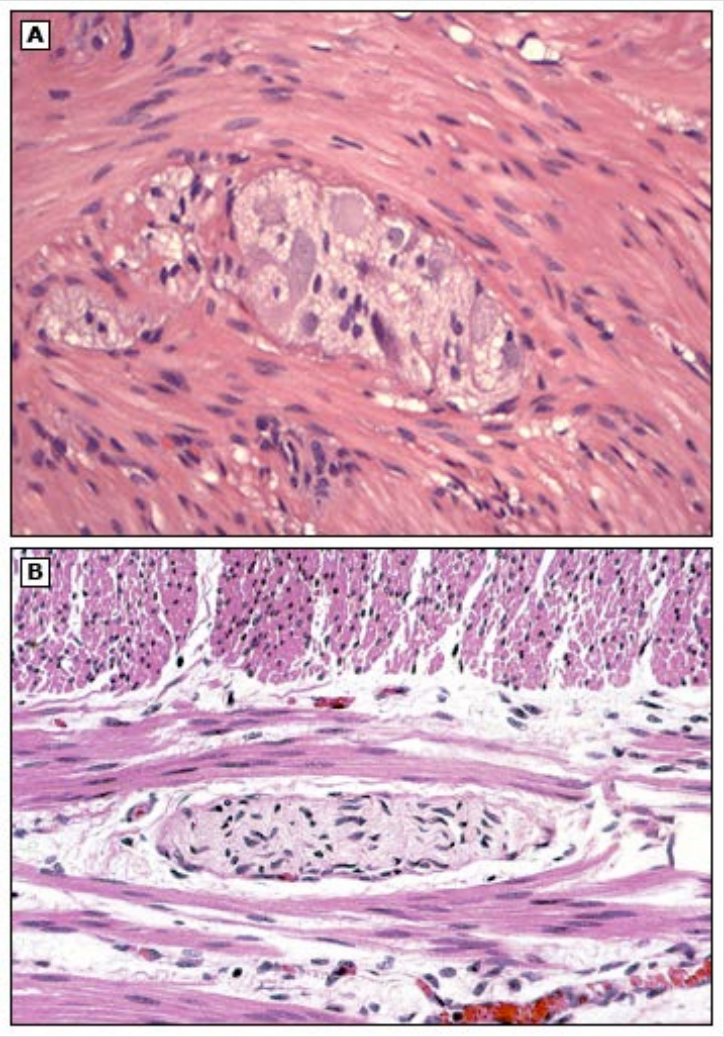
- Contrast enema: "transition zone" pathognomonic sign
  - Limited by false-negative results
- Anorectal manometry: useful in ultrashort-segment
  - PPV 75 to 95% but less accurate in infants <1 month age, and those with longstanding constipation



Source: Update, Congenital aganglionic megacolon (Hirschsprung disease), retrieved Apr 2020

DIAGNOSTIC TESTING

- Rectal biopsy is the gold standard
  - Pathognomic if ganglion cells are absent
  - Supportive findings
    - Presence of hypertrophic nerve fibers
    - Increased acetylcholinesterase activity or staining in muscularis mucosae
    - Decreased/absent calretinin-immunoreactive fibers in lamina propria



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MANAGEMENT

- Treatment: surgery
  - Removal of all of aganglionic segment with anastomosis of normal proximal bowel to distal rectum or anal canal
  - Goal: establish regular and spontaneous defecation, maintain continence
- Outcomes generally good but common complications include:
  - Constipation
  - Enterocolitis - usually occurs in first year after surgery
  - Incontinence
  - Urologic/sexual complications (no more common than in matched controls but something to consider)

REFERENCES

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- Wesson DE, Lopez ME. Congenital aganglionic megacolon (Hirschsprung disease). In: UpToDate, Singer JI, LI BU(Eds), UpToDate, Waltham, MA, 2020.