

HIRSCHSPRUNG'S DISEASE

Etiology

- motor disorder of the colon
- most accepted theory is due to a defect in the craniocaudal migration of neuroblasts originating from the neural crest that occurs during the first 12 weeks of gestation (colonic development)
- these neuroblasts are the precursors of colonic ganglion cells
- affected segment of the colon fails to relax, causing a functional obstruction.
- Affected region: 75% rectosigmoid, approx 10% entire colon
- M : F ratio of 3-4 : 1
- Predominant affected gene is Ret-proto oncogene; affects 50% of familial; 20% of sporadic cases
- associated with other chromosomal abnormalities and syndromes: Trisomy 21, cardiac disease (especially septal defects), MEN2

Clinical Features

- bilious emesis, abdominal distension, and failure to pass stool are common symptoms of intestinal obstruction diagnosed in the neonatal period
- The diagnosis can be suggested by a delay in passage of the first meconium (greater than 48 hours of age).
- Blast sign: an explosive expulsion of gas and stool after the rectal examination which temporarily may provide some relief of the obstruction
- Emergency presentation may include enterocolitis; patients have a sepsis-like picture with fever, vomiting, and diarrhea which can progress to toxic megacolon. May require IV antibiotics, IV fluid resuscitation.
- patients with less severe disease may not be diagnosed until later in childhood; these patients commonly have a history of chronic constipation and failure to thrive

Diagnosis

- Rectal biopsy
 - Gold standard for diagnosis
 - Performed using a mucosal suction technique
 - absence of ganglion cells is diagnostic
 - additional findings include the presence of hypertrophic nerve fibers and elevated acetylcholinesterase activity
- Barium enema
 - A cone-shaped transition zone or caliber change can be seen, (usually in the rectosigmoid area) which represents the change from the normal caliber/narrowed rectum (aganglionic segment) to the dilated colon proximal to the aganglionic region
- Anorectal Manometry
 - Lack of relaxation of the internal anal sphincter with balloon rectal distension is suggestive of HD.
 - A study showing normal relaxation of the internal anal sphincter with distension of the rectum excludes the diagnosis of HD.

- Comparison of diagnostic procedures

Comparison of Sensitivity and Specificity for Rectal Suction Biopsy, Contrast Enema, and Anorectal Manometry in the Diagnosis of Hirschsprung's Disease in Children^{†*}

Test	Sensitivity (95% CI)	Specificity (95% CI)
Rectal suction biopsy	93 percent (77 - 98 percent)	100 percent (96 - 100 percent)
Contrast enema	76 percent (57 - 89 percent)	97 percent (91 - 99 percent)
Anorectal manometry	83 percent (63 - 93 percent)	93 percent (85 - 97 percent)

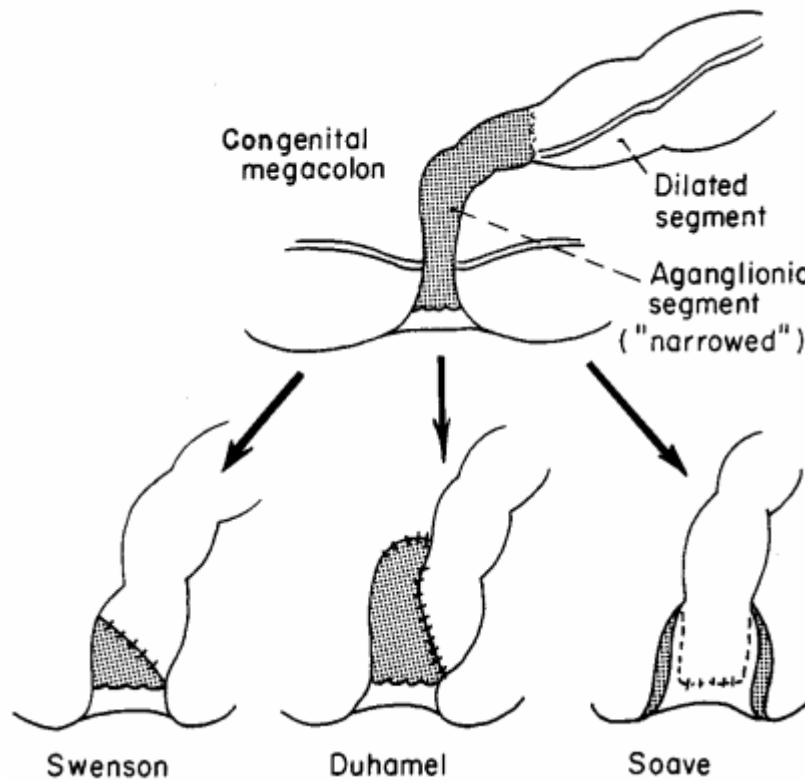
* Diagnosis was confirmed by full-thickness rectal biopsy and excluded by clinical follow-up for at least six months
[†] Data from: De Lorijn, F, Reitsma, JB, Voskuil, WP, et al. Diagnosis of Hirschsprung's disease: a prospective, comparative accuracy study of common tests. J Pediatr 2005; 146:787.

Treatment:

- Medical: goal is to institute temporary measures until definitive reconstructive surgery can take place; IVF and electrolyte management
- Surgical: goal is to resect the affected segment and bring the normal ganglionic bowel down close to the anus and preserve sphincter function
- Traditionally, a two stage operation is performed.
 - First stage involves a laparotomy in the LLQ; the location of the TZ is identified;
 - a diverting colostomy is then performed in the region of normal ganglionated bowel
 - a definite procedure is performed at a later time.
- 3 main definite procedures:
 - Swenson procedure: the aganglionic bowel is removed down to the level of the internal sphincters and a coloanal anastomosis is performed
 - Duhamel procedure: the ganglionated normal colon is brought posterior to the aganglionic rectal stump. A GIA stapler is then inserted through the anus with one arm within the normal, ganglionated bowel posteriorly and the other in the aganglionic rectum anteriorly. Firing of the stapler therefore results in formation of a neorectum that empties normally, due to the posterior patch of ganglionated bowel.
 - Soave procedure: involves an endorectal mucosal dissection within the aganglionic distal rectum. The normally ganglionated colon is then pulled through the remnant muscular cuff and a coloanal anastomosis is performed.
 - Recently, this procedure has been performed in the newborn primarily; without an initial ostomy
- Alternate surgical procedures
 - Anorectal myomectomy: for patients with ultrashort-segment Hirschsprung disease. The procedure removes a 1-cm wide strip of extramucosal rectal wall beginning immediately proximal to the dentate line and extending to the normal ganglionic rectum proximally. The mucosa and submucosa are

preserved and closed.

- Long segment Hirschsprung disease: Most procedures include a side-to-side anastomosis of the ganglionic/propulsive small bowel to a short segment of the aganglionic/absorptive colon. Whether a short colonic patch or a small bowel-to-rectal wall Duhamel anastomosis is created is perhaps less important than maintaining a short patch length (<10 cm).



References:

- ACS surgery: principles and practice
www.acssurgery.com
- Townsend: Sabiston Textbook, of Surgery, 17th ed., Copyright © 2004

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