Hirschsprung Disease

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Synonyms and related keywords: Hirschsprung's disease, ultrashort-segment Hirschsprung disease, congenital megacolon, congenital dilation of the colon, congenital megacolon, aganglionic megacolon, meconium, anorectal reflex enterocolitis, trisomy 21, Down syndrome, bowel obstruction, bilious vomiting, abdominal distention, poor feeding, faecal incontinence, Auerbach plexus, submucosal plexus, Meissner plexus, colonic lavage, full-thickness rectal biopsy, simple anorectal manometry, diverting colostomy, Swenson procedure, Duhamel procedure, Soave procedure, anorectal myomectomy

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Disclosure

INTRODUCTION
**Background:** Hirschsprung disease is a developmental disorder of the enteric nervous system by an absence of ganglion cells in the distal colon resulting in a functional obstruction. Although described by Ruysch in 1691 and popularized by Hirschsprung in 1886, the pathophysiology was determined until the middle of the 20th century when Whitehouse and Kernohan described the distal intestine as the cause of obstruction in their series of patients (Whitehouse, 1948). In 1949, Swenson described the first consistent definitive procedure for Hirschsprung disease, rectosigmoidectomy with coloanal anastomosis. Since then, other operations have been described, including the Duhamel and Soave techniques. More recently, advances in surgical technique, including minimally invasive procedures, and early diagnosis have resulted in decreased morbidity and mortality for patients with Hirschsprung disease.

Most cases are now diagnosed in the newborn period. Hirschsprung disease should be considered in any newborn who fails to pass meconium within 24-48 hours after birth. Although contrast enema is useful in diagnosis, full-thickness rectal biopsy remains the criterion standard. Once the diagnosis is confirmed, the basic treatment is to remove the poorly functioning aganglionic bowel and create an anastomosis to the healthy innervated bowel (with or without an initial diversion).

**Pathophysiology:** Congenital aganglionosis of the distal bowel defines Hirschsprung disease with the anus, which is always involved, and continues proximally for a variable distance. Both (Auerbach) and submucosal (Meissner) plexus are absent, resulting in reduced bowel peristalsis. The precise mechanism underlying the development of Hirschsprung disease is unknown.

Enteric ganglion cells are derived from the neural crest. During normal development, neuroblasts migrate to the small intestine by the 7th week of gestation and will reach the colon by the 12th week of gestation. One possible etiology for Hirschsprung disease is a defect in the migration of these neuroblasts to the distal intestine. Alternatively, normal migration may occur with a failure of neuroblasts to differentiate in the distal aganglionic segment. Abnormal distribution in affected intestine of components required for neuronal growth and development, such as fibronectin, laminin, neural cell adhesion molecule, and neurotrophic factors, may be responsible for this theory (Gaillard, 1982; Langer, 1994; Tosney, 1986).

Additionally, the observation that the smooth muscle cells of aganglionic colon are electrically undergoing electrophysiologic studies also points to a myogenic component in the development of Hirschsprung disease (Kubota, 2002). Finally, abnormalities in the interstitial cells of Cajal, pacemaker cells, and intestinal smooth muscle have also been postulated as an important contributing factor (Vanderwinden, 1996).

Three neuronal plexus innervate the intestine: the submucosal (ie, Meissner) plexus, the intermuscular (ie, Auerbach) plexus, and the smaller mucosal plexus. All of these plexus are finely integrated aspects of bowel function, including absorption, secretion, motility, and blood flow.

Normal motility is primarily under the control of intrinsic neurons. Bowel function is adequate, with extrinsic innervation. These ganglia control both contraction and relaxation of smooth muscle, predominating. Extrinsic control is mainly through the cholinergic and adrenergic fibers. The cholinergic system, and the adrenergic fibers mainly cause inhibition.

In patients with Hirschsprung disease, ganglion cells are absent, leading to a marked increase in innervation. The innervation of both the cholinergic and adrenergic systems is 2-3 times that of normal bowel. The adrenergic (excitatory) system is thought to predominate over the cholinergic (inhibitory) system.
increase in smooth muscle tone. With the loss of the intrinsic enteric inhibitory nerves, the increase in smooth muscle tone is unopposed and leads to an imbalance of smooth muscle contractility, uncoordinated peristalsis, and a functional obstruction.

Frequency:

- **In the US:** Hirschsprung disease occurs at an approximate rate of 1 case per 5400-7200 newborns.
- **Internationally:** The exact worldwide frequency is unknown, although international studies have reported rates ranging from approximately 1 case per 1500 newborns to 1 case per 7000 newborns (Meza Russell, 1994).

Mortality/Morbidity:

- Approximately 20% of infants will have one or more associated abnormality involving the cardiovascular, urological, or gastrointestinal system (Ryan, 1992). Hirschsprung disease is associated with the following:
  
  o Down syndrome
  o Neurocristopathy syndromes
  o Waardenburg-Shah syndrome
  o Yemenite deaf-blind syndrome
  o Piebaldism
  o Goldberg-Shprintzen syndrome
  o Multiple endocrine neoplasia type II
  o Congenital central hypoventilation syndrome

- Untreated aganglionic megacolon in infancy may result in a mortality rate of as much as 80%. Operative mortality rates for any of the interventional procedures are very low. Even in cases of treated Hirschsprung disease, the mortality rate may be as high as 30% as a result of enterocolitis.

- Possible complications of surgery include anastomotic leak (5%), anastomotic stricture (1%), obstruction (5%), pelvic abscess (5%), and wound infection (10%). Long-term complications include obstructive symptoms, incontinence, and enterocolitis. Although many patients will encounter these problems postoperatively, long-term follow-up studies have shown that most children make significant improvement and will do relatively well (Yanchar, 1999). Patients with an associated syndrome and those with long-segment disease have been found to have poorer outcomes (De la Torre 1990; Hackam, 2003).

Race: The disease has no racial predilection.

Sex: Hirschsprung disease occurs more often in males than females, with a male-to-female ratio of 17:1.
4:1. However, with long-segment disease, the incidence increases in females.

**Age:** Hirschsprung disease is uncommon in premature infants.

- The age at which Hirschsprung disease is diagnosed has progressively decreased over the past century. In the early 1900s, the median age at diagnosis was 2-3 years; from the 1950s to 1970s, the median age was 2 months.
- Currently, approximately 90% of patients with Hirschsprung disease are diagnosed in the newborn period.

**History:**

- Approximately 10% of patients have a positive family history. This is more common in patients with longer segment disease.
- Hirschsprung disease should be considered in any newborn with delayed passage of meconium or in any child with a history of chronic constipation since birth. Other symptoms include bowel obstruction, abdominal distention, poor feeding, and failure to thrive.
- Prenatal ultrasound demonstrating bowel obstruction is rare except in cases of total colonic atresia (Belin, 1995).
- Older children with Hirschsprung disease have usually had chronic constipation since birth and show evidence of poor weight gain.
- Older presentation is more common in breastfed infants who will typically develop constipation around the time of weaning.
- Despite significant constipation and abdominal distension, children with Hirschsprung disease rarely develop encopresis. In contrast, children with functional constipation or stool-withholding behaviors more commonly develop encopresis.
- About 10% of children may present with diarrhea caused by enterocolitis, which is thought to be related to stasis and bacterial overgrowth. This may progress to colonic perforation, causing life-threatening sepsis (Teitelbaum, 1989).

**Physical:**

- Physical examination in the newborn period is usually not diagnostic, but it may reveal a distended abdomen and/or spasm of the anus.
- A low imperforate anus with a perineal opening may have a similar presentation to that of an acquired imperforate anus.
Hirschsprung disease. Careful physical examination differentiates the two.

- In older children, however, a distended abdomen resulting from an inability to release flatus

**Causes:** See Pathophysiology.

**Differentials**

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<td>Megacolon, Chronic</td>
<td>Megacolon, Toxic</td>
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**Other Problems to be Considered:**

- Intestinal atresias or stenosis
- Small left colon syndrome
- Meconium blockage syndrome
- Anorectal malformations
- Prematurity
- Intestinal malrotation
- Neuronal intestinal dysplasia

**Workup**

**Lab Studies:**

- Chemistry panel: For most patients, electrolyte and renal panel findings are within reference ranges. Children presenting with diarrhea may have findings consistent with dehydration. Test results may aid in directing fluid and electrolyte management.

- CBC count: This test is obtained to ensure that the preoperative hematocrit and platelet counts are within reference ranges.

- Coagulation studies: These studies are obtained to ensure that clotting disorders are corrected before surgery. Values are expected to be within reference ranges.

**Imaging Studies:**

- Plain abdominal radiographs may show distended bowel loops with a paucity of air in the rectum.
- **Barium enema**
  - Avoid washing out the distal colon with enemas before obtaining the contrast enema zone.
  - The catheter is placed just inside the anus, without inflation of the balloon, to avoid risk of perforation.
  - Radiographs are taken immediately after hand injection of contrast and again 24 hours later.
  - A narrowed distal colon with proximal dilation is the classic finding of Hirschsprung disease. Findings in neonates (ie, babies aged <1 mo) are difficult to interpret and will fail to demonstrate this transition zone approximately 25% of the time (Smith, 1991).
  - Another radiographic finding suggestive of Hirschsprung disease is the retention of barium enema has been performed.

**Other Tests:**

- **Anorectal manometry**
  - Anorectal manometry detects the relaxation reflex of the internal sphincter after distension of the rectal lumen. This normal inhibitory reflex is thought to be absent in patients with Hirschsprung disease (Pensabene, 2003).
  - Swenson initially used this test. In the 1960s, it was refined but has recently fallen into disfavor because of its many limitations. A normal physiological state is required, and sedation is also usually necessary. At useful, false-positive results have been reported in up to 62% of cases, and false-negative results have been reported in up to 24% of cases.
  - Because of these limitations and questionable reliability, anorectal manometry is not commonly used in the United States.

- Because cardiac malformation (2-5%) and trisomy 21 (5-15%) are associated with congenital aganglionosis, cardiac evaluation and genetic testing may be warranted.

**Procedures:**

- **Rectal biopsy**
  - The definitive diagnosis of Hirschsprung disease is confirmed by rectal biopsy, ie, findings that indicate an absence of ganglion cells.
  - The definitive method for obtaining tissue for pathologic examination is by a full-thickness rectal biopsy. The specimen must be obtained at least 1.5 cm above the dentate line because ag ganglionosis may normally be present below this level.
  - Disadvantages include the potential for bleeding and scarring and the usual need for general anesthesia during full biopsy procedures.
Simple suction rectal biopsy

- More recently, simple suction rectal biopsy has been used to obtain tissue for histologic examination.
- Rectal mucosa and submucosa are sucked into the suction device, and a self-contained cylindrical knife cuts off the tissue.
- The distinct advantage of the suction biopsy is that it can be easily performed at the bedside.
- However, pathologically diagnosing Hirschsprung disease from samples obtained by suction biopsies is considerably more difficult than pathologically diagnosing Hirschsprung disease from samples obtained by other methods.
- Ease of diagnosis has been improved with the use of acetylcholinesterase staining, which intensely stains the hypertrophied nerve fibers throughout the lamina propria and muscularis propria.

**Histologic Findings:** Both the myenteric (Auerbach) and submucosal (Meissner) plexus are absent from the muscular layer of the bowel wall. Hypertrophied nerve trunks enhanced with acetylcholinesterase stain are also observed in the muscularis propria.

**Medical Care:** The general goals of medical care are 3-fold: (1) to treat the complications of unrecognized or untreated Hirschsprung disease, (2) to institute temporary measures until definitive reconstructive surgery can take place, and (3) to manage bowel function after reconstructive surgery.

- Management of complications of recognized aganglionosis is directed toward reestablishing normal fluid and electrolyte balance, preventing bowel overdistension (with possible perforation), and managing complications such as sepsis. Intravenous hydration, nasogastric decompression, and, as indicated, administration of intravenous antibiotics remain the cornerstones of initial medical management.

- Because cardiac malformation (2-5%) and trisomy 21 (5-15%) are associated with congenital aganglionosis, cardiac evaluation and genetic testing may be warranted.

- Colonic lavage, consisting of mechanical irrigation with a large-bore rectal tube and large volumes of irrigant, may be required.

- Balanced salt solutions may help prevent electrolyte imbalances.

- Nasogastric decompression, intravenous fluids, antibiotics, and colonic lavage may also be used in patients who develop enterocolitis as a complication. Sodium cromoglycate, a mast cell stabilizer, has been reported to be of benefit in these patients as well (Rintala, 2001).

- Routine colonic irrigation and prophylactic antibiotic therapy have been proposed as a means of decreasing the risk of enterocolitis (Marty, 1995; Elhalaby, 1995).

- Injecting the nonrelaxing internal sphincter mechanism with botulinum toxin (BOTOX®) has recently been shown to induce normal patterns of bowel movements in postoperative patients with enterocolitis.

**Surgical Care:** Surgical management of Hirschsprung disease begins with the initial diagnosis.
biopsy. Traditionally, treatment also includes creating a diverting colostomy at the time of diagnosis, and, once the child grows and weighs more than 10 kg, the definitive repair is performed.

This standard of treatment was developed in the 1950s after reports of relatively high leak and stricture rates with the single stage procedure were initially described by Swenson. However, with the advent of safer anesthesia and more advanced hemodynamic monitoring, a primary pull-through procedure without a diverting colostomy is increasingly being performed. Contraindications to a one stage procedure include massively dilated proximal bowel, severe enterocolitis, perforation, or inability to accurately determine the transition zone by frozen section.

For neonates who are first treated with a diverting colostomy, the transition zone is identified and the colostomy is placed proximal to this area. The presence of ganglion cells at the colostomy site must be unequivocally confirmed by frozen section before an end stoma is appropriate, usually based on the surgeon’s preference.

A number of definitive procedures have been used, all of which have demonstrated excellent results in experienced hands. The 3 most commonly performed repairs are the Swenson, Duhamel, and Soave procedures. Regardless of the procedure, cleaning the colon prior to definitive repair is necessary.

- **Swenson procedure**
  - The Swenson procedure was the original pull-through procedure used to treat Hirschsprung disease.
  - The aganglionic segment is resected down to the sigmoid colon and the remaining rectum, and an oblique anastomosis is performed between the normal colon and the low rectum.

- **Duhamel procedure**
  - The Duhamel procedure was first described in 1956 as a modification to the Swenson procedure.
  - Key points are that a retrorectal approach is used and a significant portion of aganglionic rectum is retained.
  - The aganglionic bowel is resected down to the rectum, and the rectum is oversewn. The proximal bowel is then brought through the retrorectal space (between the rectum and sacrum), and an end-to-side anastomosis is performed on the remaining rectum.

- **Soave (endorectal) procedure**
  - The Soave procedure was introduced in the 1960s and consists of removing the mucosa and submucosa of the rectum and pulling the ganglionic bowel through the aganglionic muscular cuff of the rectum.
  - The original operation did not include a formal anastomosis, relying on scar tissue formation between the pull through and the surrounding aganglionic bowel. The procedure has since been modified by Boley to include a primary anastomosis at the anus.

- **Anorectal myomectomy**
  - For children (and occasionally adults) with ultrashort-segment Hirschsprung disease, removing a strip of posterior midline rectal wall is an alternative surgical option.
  - The procedure removes a 1-cm wide strip of extramucosal rectal wall beginning immediately proximal to the dentate line.
extending to the normal ganglionic rectum proximally.

- The mucosa and submucosa are preserved and closed.

- Procedures for long-segment Hirschsprung disease
  - Patients with total colonic involvement require modified procedures to bypass the absorptive surface area and allow for proper growth and nutritional support.
  - Most procedures include a side-to-side anastomosis of the ganglionic/propulsive or aganglionic/absorptive colon.
  - Whether a short right 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).
  - Long-segment anastomoses, such as the Martin procedure, are no longer advocated.
  - A laparoscopic approach to the surgical treatment of Hirschsprung disease was first described in 1999 by Georgeson. The transition zone is first identified laparoscopically, followed by mobilization of the rectum below the peritoneal reflection. A transanal mucosal dissection is performed, followed by prolapsing of the rectum through the anus and anastomosis. Functional outcomes appear to be equivalent to open techniques based on short-term results (Georgeson, 1999; de Lagausie, 1999; Curran, 1996).
  - Transanal pull-through in which no intra-abdominal dissection is performed has also been described (Langer, 1999; De La Torre Mondregan, 1998). The entire procedure is performed from below in a manner similar to perineal rectosigmoidectomy. The transition zone is identified and anastomosis is performed. Similar to the laparoscopic approach, one-stage approaches with the benefits of minimal analgesia and shortened hospital stays (Langer, 2000; De La Torre, 2000; Langer, 2003).

Consultations:

- Pediatric surgeons
- Pediatric gastroenterologists
- Geneticists (if trisomy 21 is present)

Diet:

- The patient should have nothing by mouth before the operation.
- Institute tube feeding or formula/breast milk once bowel function resumes.
- High-fiber diets and diets containing fresh fruits and vegetables may optimize postoperative bowel function.

Activity: Limit physical activity for about 6 weeks to allow the wound to heal properly (applies
The goals of pharmacotherapy are to eradicate infection, reduce morbidity, and prevent complications.

Drug Category: Antibiotics -- Empiric antimicrobial therapy must be comprehensive and cover all likely pathogens in the context of this clinical setting. Antibiotic selection should be guided by blood culture sensitivity whenever feasible.

### Drug Name

- **Ampicillin (Marcillin, Omnipen, Principen)** -- Bactericidal activity against susceptible organisms. Alternative to amoxicillin when unable to take medication orally.

### Adult Dose

- 1-2 g IV q6h

### Pediatric Dose

- 25 mg/kg IV q6h

### Contraindications

- Documented hypersensitivity

### Interactions

- Probencid and disulfiram elevate levels; allopurinol decreases levels and has additive effects on ampicillin rash; may decrease effectiveness of oral contraceptives

### Pregnancy

- B - Usually safe but benefits must outweigh the risks.

### Precautions

- Adjust dose in renal failure; evaluate rash and differentiate from hypersensitivity reaction

- **Gentamicin (Garamycin, Jenamicin)** -- Aminoglycoside antibiotic for gram-negative coverage. Used in combination with both an agent against gram-positive organisms and one that covers anaerobic bacteria.

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The first course examines the case of John B—, a 69-year-old man who received a diagnosis of type 2 diabetes more than 20 years ago. His most recent glycosylated hemoglobin (HbA1c) concentration was 9.5%, prompting his physician to keep a diary of self-monitored fasting plasma glucose (FPG) levels. His 2-hour postprandial glucose (PPG) levels for 2 weeks were 220 mg/dL and 300 mg/dL, respectively. (This article is for AMA PRA Category 1 Credit™.)
### Drug Name: **Toxins**

Induce more normal patterns of bowel movements in postoperative patients with enterocolitis.

<table>
<thead>
<tr>
<th>Drug Name</th>
<th>Adult Dose</th>
<th>Pediatric Dose</th>
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<th>Interactions</th>
<th>Pregnancy</th>
<th>Precautions</th>
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<tr>
<td><strong>Cephalosporins</strong></td>
<td>5-7 mg/kg/d IV</td>
<td>2.5 mg/kg IV q8h; check peak and trough levels after third dose</td>
<td>Documented hypersensitivity; non–dialysis-dependent renal insufficiency</td>
<td>Coadministration with other aminoglycosides, cephalosporins, penicillins, and amphotericin B may increase nephrotoxicity; aminoglycosides enhance effects of neuromuscular blocking agents; prolonged respiratory depression may occur; coadministration with loop diuretics may increase auditory toxicity of aminoglycosides; possible irreversible hearing loss of varying degrees may occur (monitor regularly)</td>
<td>C - Safety for use during pregnancy has not been established.</td>
<td>Narrow therapeutic index (not intended for long-term therapy); in renal failure (not on dialysis), myasthenia gravis, hypocalcemia, and conditions that depress neuromuscular transmission; adjust dose in renal impairment</td>
</tr>
<tr>
<td><strong>Metronidazole (Flagyl)</strong></td>
<td>500 mg PO/IV q6-8h</td>
<td>7.5 mg/kg IV q6h</td>
<td>Documented hypersensitivity</td>
<td>May increase toxicity of anticoagulants, lithium, and phenytoin; cimetidine may increase toxicity; disulfiramlike reaction may occur with orally ingested ethanol</td>
<td>B - Usually safe but benefits must outweigh the risks.</td>
<td>Adjust dose in hepatic disease; monitor for seizures and development of peripheral neuropathy</td>
</tr>
<tr>
<td><strong>Botulinum toxin type A (BOTOX®)</strong></td>
<td>1.25-2.5 U IM; may repeat q3-4mo</td>
<td>&gt;12 years: Administer as in adults &lt;12 years: 0.25-1 U IM; may repeat q3-4mo</td>
<td>Documented hypersensitivity</td>
<td>Aminoglycosides or drugs that interfere with neuromuscular transmission</td>
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Further Inpatient Care:

- If a diverting colostomy is created in a newborn, he or she must remain in the hospital until the ostomy is functioning and feeding goals are obtained. Feedings are usually initiated 24-48 hours after the creation of the colostomy.

- After the definitive pull-through procedure is performed, the patient is hospitalized until full feedings are possible and evidence of the return of bowel function is obtained. Patients are to take nothing by mouth, with intravenous fluid hydration until they pass flatus or have a bowel movement. Once this occurs, clear liquids may be started, and the diet may be advanced until feeding goals are obtained. Intravenous antibiotics are also continued until evidence of proper bowel function is observed.

Further Outpatient Care:

- After a definitive pull-through procedure is performed, normal growth and development should ensue.

- Patients should be monitored for normal bowel habits. Patients with no other underlying disorders and no postoperative complications should develop normal bowel habits. However, such habits may not develop until the patient is older.

In/Out Patient Meds:

- Immediately after the diverting colostomy is created or a definitive pull-through procedure is performed, patients should usually remain on broad-spectrum intravenous antibiotics (eg, ampicillin, gentamicin, metronidazole) until bowel function has returned and feeding goals are achieved.

- Once a definitive pull-through procedure is performed and normal bowel function is obtained, no additional medication is required.

Transfer:

- Neonates and older children thought to have Hirschsprung disease should be treated in a center where pediatric specialists are available to make the diagnosis and provide definitive care.

Deterrence/Prevention:

- Hirschsprung disease cannot be prevented; however, heightened clinical awareness prevents a delay in diagnosis.

Complications:

- Potential complications for the complex operations associated with Hirschsprung disease include surgical complications.
Although the incidence rates of these complications are roughly the same when surgeons with experience perform the procedures versus when surgeons with less experience perform them, each procedure has been associated with a specific level of difficulty.

Complications may include an increased incidence of postoperative enterocolitis with the Swenson procedure, constipation following the Duhamel repair, and diarrhea and incontinence with the Soave pull-through.

In general, the complications are anastomotic leak (5%), anastomotic stricture (5-10%), intestinal obstruction (5%), pelvic abscess (5%), and wound infection (10%).

- Later complications associated with surgical management of Hirschsprung disease include symptoms, and incontinence.

- Enterocolitis accounts for significant morbidity and mortality in patients with Hirschsprung disease.

  - Enterocolitis results from an inflammatory process of the mucosa of the colon or small intestine. As the disease progresses, the lumen of the intestine becomes filled with fibrinous exudate and is at increased risk for perforation. This process may occur in both the aganglionic and ganglionic portion of the bowel.

  - Patients typically present with explosive diarrhea, abdominal distention, fever, vomiting, and lethargy.

  - Approximately 10-30% of patients with Hirschsprung disease develop enterocolitis. Moreover, the risk of developing enterocolitis remains despite surgical correction.

  - Treatment consists of intravenous antibiotics and aggressive colonic irrigations. Some authorities advocate decompression of the bowel, especially in patients with long-segment disease, with an enterostomy placed proximally to the transition zone.

- Patients may present postoperatively with abdominal distension, vomiting, or constipation (Dasgupta, 2004).

  - Mechanical obstruction can be easily diagnosed with digital rectal exam and contrast of the pull-through may be required (Langer, 1996).

  - Persistent aganglionosis occurs rarely and may be due to pathologic error, inadequacy of the pull-through. If a rectal biopsy does not show ganglion cells, revision of the pull-through may be necessary (Schmittenbecher, 1993; Langer, 1999; Teitelbaum, 2003).

  - Motility disorders may be associated with Hirschsprung disease. Workup may include contrast studies, manometry, and biopsy to evaluate for intestinal neuronal dysplasia (Schmittenbecher, 1999; Di Lorenzo, 2000).

  - Internal sphincter achalasia may result in persistent obstruction. This can be treated with intrasphincteric botulinum toxin, or nitroglycerin paste. Most cases will resolve by the age of 5 years (Minkes, 2000; Millar, 2002).

  - Functional megacolon may be present due to stool-holding behavior. Bowel management regimens may include cecostomy and antegrade enemas reserved for refractory cases (Chait, 2003).

  - Incontinence may be the result of abnormal sphincter function, decreased sensation, or constipation (Dasgupta, 2003). In general, anorectal manometry and ultrasound should aid in differentiating between these causes.

http://www.emedicine.com/med/topic1016.htm
diagnoses.

Prognosis:

- The long-term outcome after definitive repair of Hirschsprung disease is difficult to determine because of conflicting reports in the literature. Some investigators report a high degree of satisfaction, while others report a significant incidence of constipation and incontinence.

- Unfortunately, approximately 1% of patients with Hirschsprung disease require a permanent colostomy to correct incontinence.

- As expected, patients with associated trisomy 21 tend to have poorer clinical outcomes.

- In general, more than 90% of patients with Hirschsprung disease have satisfactory outcomes.

Medical/Legal Pitfalls:

- Potential medicolegal pitfalls may be related to the diagnosis and treatment of children with this disorder. Children with Hirschsprung disease should generally be cared for by pediatric surgeons and gastroenterologists.

- Any child with a history of constipation since birth requires an extensive workup and a trial of medical management should not be initiated until Hirschsprung disease has been appropriately excluded.

Special Concerns:

- Ultrashort-segment Hirschsprung disease
  - Ultrashort-segment Hirschsprung disease is characterized by a few centimeters of aganglionic bowel in the rectum, adjacent to the anus.
  - Recognizing this condition can be very difficult. These patients are not typically diagnosed until they are older.
  - The principal symptom is severe constipation that usually begins between the ages of 6 and older.
  - Barium enemas tend not to demonstrate a transition zone.
  - Anorectal manometry is useful in the workup of these patients and demonstrates an absent anorectal reflex, but the definitive diagnosis is made by rectal biopsy.
  - A definitive pull-through procedure is usually unnecessary because most patients are satisfactorily treated with a surgical myomectomy. This involves resecting a longitudinal strip of the posterior muscular wall of the rectum.

- Total colonic aganglionosis
Total colonic aganglionosis is a more severe form of Hirschsprung disease in which small intestine is aganglionic. It occurs in 3-12% of cases and extends to the terminal ileum in about 20% of cases, and to the jejunum in 5% of cases (Bickler, 1992).

These patients tend to have more severe signs and symptoms than those who have been found to have increased morbidity and mortality (Anderson, 1986; Bickler, 1992; Ikeda, 1986).

Diagnosis may prove to be difficult with radiographic studies being diagnostic in only 20% made at the time of laparotomy or leveling colostomy. Frozen section of the appendix confirms the diagnosis (Coran, 2000).

Any of the 3 standard repairs may be used to treat patients with total colonic aganglionosis, although primary pull-through in the newborn period is controversial since results in this subgroup of patients is poorer than in those with rectosigmoid disease (Coran, 2000).

Specific modifications have been made to these repairs, with the goal of increased fluid and electrolyte absorption.

**PICTURES**

**Caption:** Picture 1. Hirschsprung disease. Contrast enema demonstrating transition zone in the rectosigmoid region.

**Picture Type:** X-RAY

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NOTE:
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