Hirschsprung’s Disease (Congenital Megacolon) — Symptoms and Causes

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Pediatric Hirschsprung Disease occurs when there is an absence of the ganglionic layer in the myenteric layer of the anus and the submucosa of the colon. This leads to the failure of relaxation of the colon, delayed passage of meconium, abdominal distension and constipation. Laboratory investigations are helpful if the patient develops enterocolitis where leukocytosis might be evident. Barium enema studies and histologic examination of biopsies confirm the diagnosis of Hirschsprung disease, and surgical resection of the aganglionic part of the colon is the current treatment of choice.

Definition of Pediatric Hirschsprung Disease

Infants who do not have enteric neurons in the myenteric and submucosal layers of their rectum and colon develop pediatric Hirschsprung disease (PHD).
Epidemiology of Pediatric Hirschsprung Disease

Pediatric Hirschsprung disease has an incidence of about 1 per 5,000 live births, making it a common etiology of delayed passing of meconium. Hirschsprung disease is associated with some life-threatening complications and has a mortality rate of 30%.

Pediatric Hirschsprung disease is more common among males with a 4:1 ratio. By 2 years, all patients with Hirschsprung disease are already diagnosed.

Etiology of Pediatric Hirschsprung Disease

The exact etiology of Hirschsprung disease is unknown, but familial aggregation is evident. Decreased nitric oxide synthase activity, or quantitative deficiency, has been associated with an increased risk of pediatric Hirschsprung disease, pyloric stenosis and other gastrointestinal motility disorders.

Pathophysiology of Pediatric Hirschsprung Disease

Patients with Hirschsprung disease do not have enteric neurons in their rectum and colon which causes a loss of peristaltic waves and constipation. Enteric neurons form from the neural crest and migrate with the vagus nerve down to the intestine. By 8 weeks of gestation, the ganglionic neurons have reached the proximal colon, and by 12 weeks of gestation the rectum. Any arrest in this normal migration process will lead to the absence of enteric neurons in the rectum and distal colon and will result in pediatric Hirschsprung disease.

Clinical Presentation of Pediatric Hirschsprung Disease

Newborns with Hirschsprung disease have a delayed passage of meconium after 48 hours of life and abdominal distension. Family history is positive in one-third of the cases. Patients with Hirschsprung disease do not have soiling or overflow incontinence. Chronic abdominal distension leads to early satiety which can eventually result in malnutrition.

Older infants develop chronic constipation and become dependent on daily enemas for induction of bowel movement. Patients with Hirschsprung disease can develop enterocolitis which presents with abdominal pain, fever and bloody diarrhea. Enterocolitis in this group of patients can be fatal.

Physical examination usually reveals abdominal distension with possible palpation of the colonic loops. Additionally, forceful expulsion of fecal material after the rectal examination is common. The rectum is usually empty on rectal examination.

Diagnostic Work-up for Pediatric Hirschsprung Disease

Laboratory investigations are not helpful in patients with Hirschsprung disease unless they develop a fever or severe abdominal pain; signs of enterocolitis. In enterocolitis,
Patients could have **leukocytosis** or **bacteremia**.

*Image: “A: Plain abdominal radiograph showing a PARTZ at rectosigmoid, arrow. B: Plain abdominal radiograph showing a PARTZ at midsigmoid, arrow. C: Plain abdominal radiograph showing a PARTZ at descending colon, arrow. D: Contrast enema showing a CETZ at rectosigmoid, arrow. E: Contrast enema showing a CETZ at midsigmoid, arrow. F: Contrast enema showing a CETZ at descending colon, arrow. PARTZ, Plain abdominal transition zone; CETZ, Contrast enema transition zone.” by Pratap et al. BMC Pediatrics 2007 7:5 doi:10.1186/1471-2431-7-5. License: CC BY 2.0*

Patients with an uncomplicated presentation usually benefit the most from imaging studies in the diagnosis work-up. As, in any case of bowel obstruction, an **abdominal x-ray** is indicated. Abdominal x-ray shows a **megacolon**, **small bowel obstruction**, and/or **multiple fluid-gas-levels**.

If the patient is not feverish, and they do not have acute abdominal pain, they can undergo a **barium enema study**. Barium enema can easily define the aganglionic from normally innervated colon. The aganglionic part is usually collapsed, while the normal part is dilated and appears to be normal. This investigation is more useful in infants rather than neonates because neonates do not have enough time to develop dilatation of the normal colonic segment above the obstruction level.

On **barium enema imaging**, the rectum is usually nondistensible which is a useful sign for the diagnosis of Hirschsprung disease.

Patients with less severe Hirschsprung disease usually present with **chronic constipation** without a clear image of Hirschsprung disease on imaging. In these patients, **rectal manometry** is useful as it shows the failure of **anorectal sphincter relaxation**.

The gold standard diagnostic test to confirm Hirschsprung disease is a **histologic examination** of a biopsy. **Rectal biopsy** shows the absence of ganglion cells in the submucosa plexus. **Acetylcholinesterase staining** is increased in Hirschsprung disease while carretinin, a stain of enteric ganglionic cells, is usually decreased on histologic examination.

**Treatment of Pediatric Hirschsprung Disease**

Patients with **acute intestinal obstruction** can be **dehydrated** due to third-space fluid loss and should be rehydrated. During fluid replacement therapy, enteric feeding should be discontinued and intestinal and gastric decompression might be indicated.
Intestinal decompression is done by rectal examination or rectal saline irrigation, both results in expulsion of the fecal material. In some patients, gastric decompression might be indicated if the upper gastrointestinal obstruction is suspected. Gastric decompression is achieved by the placement of a nasogastric tube.

If the patient is feverish, have leukocytosis or shows signs of enterocolitis, broad-spectrum antibiotics are indicated.

These measurements are temporary and surgical management for pediatric Hirschsprung disease is the only definitive treatment option.

Several forms of surgery exist for the management of Hirschsprung disease and the choice of the procedure is dependent on the patient’s age, expectations and comorbidities. Additionally, the length of the aganglionic segment is an important factor in defining the type and extent of the surgical intervention.

A single pull-through surgery is preferred in the majority of the patients, especially in neonates. In single pull-through surgery, the aganglionic segment is resected and the normal healthy colon is anastomosed to the anal cavity above the dentate gyrus. This procedure can be performed laparoscopically, or in an open surgery.

The availability of a good pathologist is essential to make sure that a single pull-through surgery is going to work, because multiple biopsies from the anus and rectum need to be examined to define the distal and proximal levels that are normally innervated for the anastomosis to be done at their level.

Patients with severe colonic distension, megacolon or enterocolitis are candidates for a two-step surgery. The first step is to perform a colostomy, which is followed by a pull-through procedure once the patient is stable enough. This picture usually happens when the diagnosis is delayed, which means that this procedure is usually more common among infants and not newborns with Hirschsprung disease.

In the acute phase after surgery and preoperatively, it is recommended for the infant to be breastfed or to at least receive breast milk and not cow milk. It is believed that breast milk decreases the risk of constipation in infants in general, and in patients with Hirschsprung disease in particular.

Patients with a two-step procedure might need to limit their physical activity after the leveled colostomy. On the other hand, patients who undergo a one-step pull-through procedure usually recover faster and can go back to their routine physical activities earlier.

References


Pediatric Hirschsprung Disease via medscape.com
