THE RADIOLOGY OF HIRSCHSPRUNG DISEASE

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Constipation is a very common problem in childhood, accounting for 6% of pediatric outpatient visits.[1] It is also one of the most frequent reasons for a barium enema examination in pediatric patients.[2] Constipated children are frequently referred to a radiologist for a barium enema to exclude Hirschsprung disease, which is one of the most serious identifiable causes of constipation.[1] The aim of this article is to review the pathogenesis, clinical manifestations, preparation of patients for barium enema studies, barium enema technique, imaging findings seen in Hirschsprung disease, and differential diagnosis.

PATHOGENESIS OF HIRSCHSPRUNG DISEASE

Hirschsprung disease or congenital aganglionic megacolon is now recognized as one of several disorders of intestinal innervation. Collectively known as "dysganglionoses", these disorders include aganglionosis of the bowel wall, hypoganglionosis, neuronal dysplasia (hyperganglionosis), absence of intrinsic innervation of the colon, and absence of innervation of the intestine.[3]

Hirschsprung disease is characterized by a congenital absence or deficiency of the ganglion cells in the myenteric (Auerbach's) and submucosal (Meissner's) plexuses of the rectum and the distal part of the colon, together with hyperplasia of cholinergic nerve fibers in the circular muscle layer, muscularis mucosae, and mucosa with a high activity of acetylcholinesterase.[4] This is due to a lack of the normal caudal migration of the neurons of the neural crest along the intestinal branches of the vagus nerve in utero. The region of aganglionosis is almost always continuous; the existence of "skip areas" is extraordinarily rare.[5] Absence of intramural ganglion cells interferes with the normal relaxation of peristalsis in the bowel wall and the internal anal sphincter. The aganglionic segment of the colon becomes hypertonic and behaves as a functional stenosis leading to partial or complete colonic obstruction. Immediately proximal to the aganglionic segment the intestine becomes markedly dilated with feces and gas.[4]

The rectosigmoid region is the most common transition site from normal to abnormal bowel. The transition zone is located in this region in 75% of patients. It is in the left colon in 12% of patients, splenic flexure in 4%, transverse colon in 4% and right colon in 3%.[3,6] There is total colonic aganglionosis with or without small intestinal involvement in 2% of patients. When the transition zone is in the rectosigmoid region, it is called short segment disease. When it is proximal to the sigmoid, it is called long segment disease. Ultrashort segment disease with aganglionosis limited to the region of the internal sphincter is very rare, as is aganglionosis involving the entire alimentary tract.[6,7]

CLINICAL MANIFESTATIONS

Hirschsprung disease occurs in one of 5,000 to 8,000 live births.[3] According to the American Academy of Pediatrics Surgical Section Survey the ratio of male to female patients with short segment Hirschsprung disease is 3.8 to 1. The ratio is 2.8 to 1 in long segment disease and 2.2 to 1 in total colonic aganglionosis.[6] Familial incidence is significant only in very long

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segment disease of the colon.

70-80% of patients with Hirschsprung disease become symptomatic within the first week of life. Most affected children are term neonates.[8] They usually present with distal bowel obstruction. Hirschsprung disease is responsible for approximately 15% to 20% of cases of neonatal bowel obstruction.[9] Ninety percent or more of normal neonates pass meconium in the first 24 hours of life, and 99% pass meconium by 48 hours of life. Any infant who fails to pass meconium by 48 hours of life should be considered as having Hirschsprung disease until proven otherwise.[10] A minority of patients with Hirschsprung disease are found later in infancy or childhood, usually with chronic, severe constipation.

Up to a third of patients with Hirschsprung disease have or develop enterocolitis, either as a presenting feature, before, or after surgery.[11] Enterocolitis is most frequently seen during the first 3 months of life.[6]

Hirschsprung disease has been associated with various congenital defects, such as Down syndrome, cardiac anomaly, genitourinary anomaly, gastrointestinal anomaly, and Ondine's curse (congenital hypoventilation syndrome).[3,5,8] Anorectal malformation has reported as an association.[12]

On examination, a child with Hirschsprung disease usually has a distended abdomen and may have palpable solid stool in the colon. Rectal examination may result in the explosion of foul-smelling liquid, virtually pathognomonic for Hirschsprung disease with early enterocolitis. Diagnosis before the onset of enterocolitis is critical in reducing mortality.[11]

IMAGING FINDINGS

1. Plain abdominal films

Plain abdominal films in neonates with Hirschsprung disease show a distal bowel obstruction, often with markedly dilated loops of bowel[5,9,10] (Figs. 1, 2). There is little or no rectal gas. A prone lateral cross table film can be taken to assess the amount of air in the rectum.[13] In the older child, plain abdominal films may show stool in an obvious megacolon. A lack of air in the distal colon and rectum is usually seen. Newman et al. have found that 4.4% of infants less than 1 year old with Hirschsprung disease present with pneumoperitoneum secondary to perforation.[14] Most of these patients have total colonic aganglionosis. Calcification in the lumen of distal small bowel has also been found on plain abdominal films of patients with total colonic aganglionosis.[15] Pneumatosis intestinalis or an abnormal mucosal pattern is sometimes seen on plain abdominal films of patients with Hirschsprung disease and enterocolitis.[8,10]

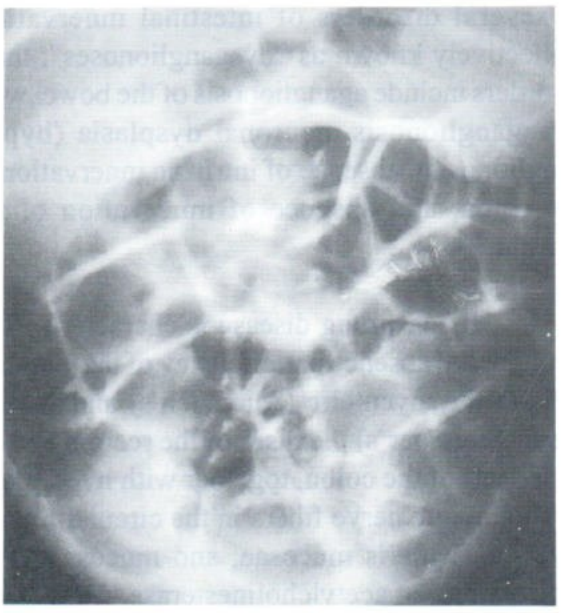

Fig. 1. Plain abdominal film shows dilatation of multiple bowel loops with no rectal gas.

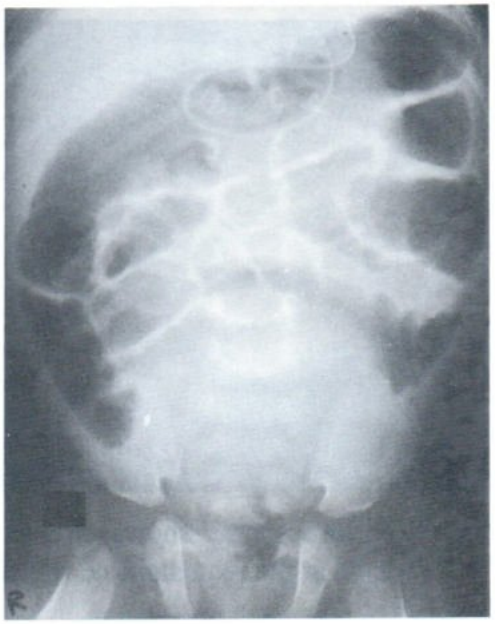

Fig. 2. Plain abdominal film shows dilatation of multiple bowel loops with a little rectal gas.

2. Contrast Enema

Of the available methods for diagnosing Hirschsprung disease, a contrast enema is used to determine the proximal extent of aganglionosis, to exclude other diseases, and to alert the surgeon if the patient has developed enterocolitis.[8,16] The other methods are rectal biopsy and rectal manometric examination. Rectal biopsy is the gold standard for diagnosis of Hirschsprung disease. Rectal manometric examination has been most useful in differentiating ultrashort segment Hirschsprung disease from psychogenic constipation.[8]

Barium enema is less accurate in cases of Hirschsprung disease with total colonic aganglionosis, very-short segment aganglionosis, in infants younger than one month of age, and in patients who have had a colostomy.[3] Overall, barium enema has a false-negative rate of 5% to 40% and a false-positive rate of up to 27%, depending on which combination of radiographic signs is present.[3]

Barium enema is contraindicated if plain films reveal pneumoperitoneum, pneumatosis intestinalis, or an abnormal mucosal pattern of enterocolitis.[8] Administering an enema to a patient with active colitis may precipitate sepsis and secondary circulatory collapse.

PATIENT PREPARATION

Although Rosenfield et al. have found that rectal examination and a cleansing enema prior to the study appear not to obscure visualization of a transition zone,[17] no cleansing enemas should be performed for several days before the contrast enema. Bowel cleansing prior to contrast enema is contraindicated because it makes the transition zone less obvious, and more importantly, bowel evacuants may precipitate fulminant colitis. The tendency of affected children to retain cleansing enemas may also cause serious electrolyte imbalance.[8]

IMAGING TECHNIQUE

O'Donovan et al. have found that a water-soluble contrast enema has a sensitivity and specificity equal to those of a barium enema for the detection of Hirschsprung disease and avoids the problems associated with barium spillage into the peritoneal cavity with perforation.[18] In addition, other causes of low intestinal obstruction such as meconium plug and meconium ileus are better studied with water-soluble contrast media than with barium.[10] At our hospital, barium sulfate is still used for a contrast enema because it is much cheaper than water-soluble contrast media.

Balloon catheters should never be used because of the likelihood of rectal trauma in a spastic and narrowed rectum, and they may obscure the aganglionic segment and the transition zone.[4,5,8] Only the tip of a soft catheter should be inserted into the anus to insure complete visualization of the contrast-filled

rectum.

The examination is begun with the lateral projection to detect a distal transition zone. Barium is introduced slowly to best demonstrate denervation hyperspasticity.[4,5,8] Rapid overdistention of the colon with barium is the most common reason for missing both the spasm and the transition zone.[8] The procedure has to be done slowly to leave enough time for the bowel to adapt to the volume of barium. Some time is needed for the aganglionic segment to contract to its former diameter.[4] When the transition zone has been identified, the proximal colon does not need to be examined because no additional information is likely to be obtained and because the predictably poor ability of the colon to empty will leave it needlessly filled with barium, which may complicate the procedure for the surgeon performing a diverting colostomy.[8]

If initial films of the colon show no spastic segments or transition zone, delayed lateral and frontal views of 24 or 48 hours may reveal the transition zone.

Abnormalities seen on barium enema

1. Transition zone between the normal or slightly small distal aganglionic bowel and the dilated ganglionic proximal bowel (Figs. 3, 4). Presence of a transition zone in the rectosigmoid is a highly reliable diagnostic feature of Hirschsprung disease; however absence of this sign does not rule it out.[17] The transition zone may be abrupt or gradual (Fig. 5) and is mostly at the rectosigmoid junction. This finding is present in approximately 65% of neonates.[17] A transition zone and a megacolon can be seen usually after 4-6 weeks of age.[4]

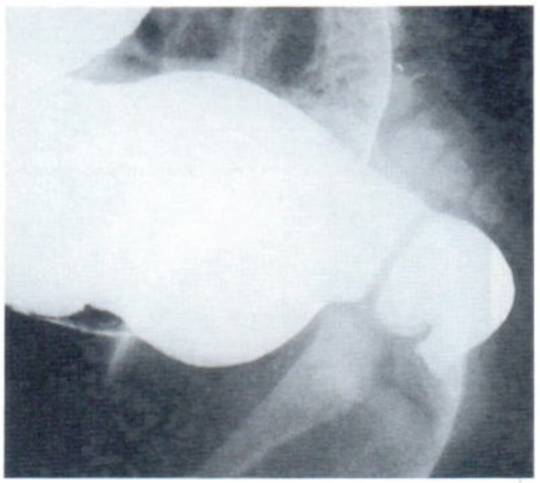

Fig. 3. Barium enema shows a transition zone at the rectosigmoid region.

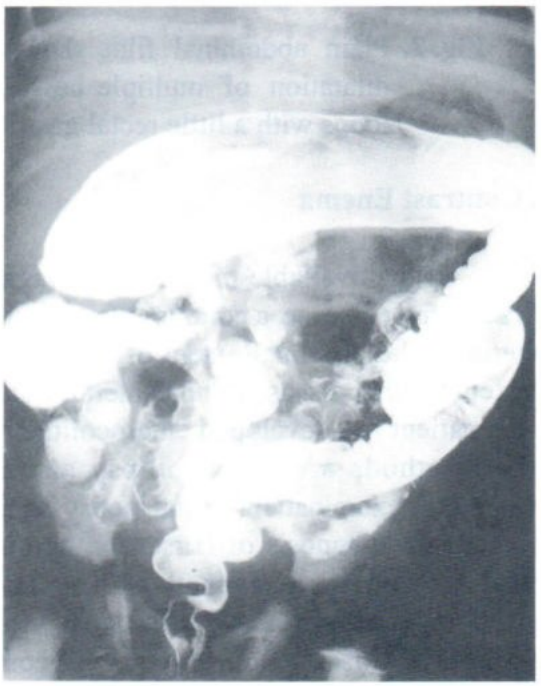

Fig. 4. Barium enema shows a transition zone at the splenic flexure.

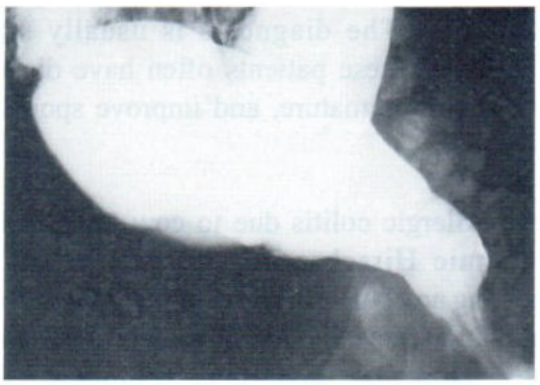

Fig. 5. Barium enema shows a gradual transition zone at the rectosigmoid region. The rectosigmoid index is 0.5

2. Abnormal contractions and peristaltic activity with pointed serrations or "saw-tooth" appearance of the aganglionic segment (Fig. 6). This is infrequently seen in the aganglionic segment but is a reliable sign of Hirschsprung disease. It has been reported in 26% of barium enemas of infants with Hirschsprung disease.[17]

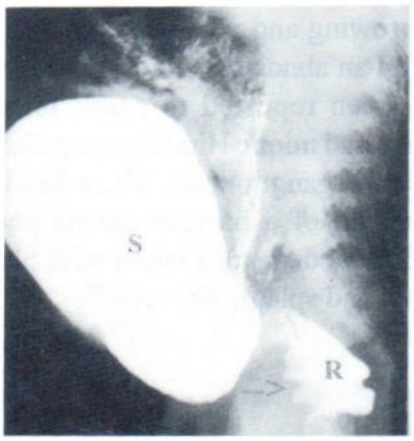

Fig. 6. Barium enema shows a transition zone in the rectosigmoid region and abnormal contraction (arrow) of the rectum (R). S = sigmoid.

3. Retention of barium proximal to the aganglionic segment at 24 hours after the examination. This is the most sensitive sign, presenting in over 80% of neonates with Hirschsprung disease. However it is less specific, because it can be seen in patients without Hirschsprung disease,[17,19] and a good evacuation does not rule out Hirschsprung disease.

A pattern of barium mixed with stool is infrequently seen on a 24-hour delayed film but has been reported as a highly reliable sign of Hirschsprung disease.[17]

4. Rectosigmoid index. This is obtained by dividing the widest diameter of the rectum by the widest diameter of the sigmoid colon when the colon is fully distended. The widest rectal diameter is measured at any point below the third sacral vertebra.[20]

In normal newborn infants, as well as cases of meconium plug syndrome and other causes of low intestinal obstruction, the rectum is commonly found to be wider than the sigmoid loop. The rectosigmoid index is normally greater than or equal to 1.0. In Hirschsprung disease the widest rectal diameter is usually less than the widest diameter of the sigmoid loop, even in the absence of a transition zone. A rectosigmoid index of less than 1.0 is suggestive of Hirschsprung disease. It is obviously unnecessary when a transition segment is clearly evident.[20,21] It is not a useful measurement in long segment or total colonic aganglionosis.[21]

5. Work hypertrophy (jejunization). In a child older than 3 months, the proximal colon develops hypertrophy of the circular and longitudinal muscle fibers in response to obstruction of the aganglionic segment. This causes a prominence of the muscle fibers that radiographically resembles the plicae circulares of the jejunum[8] (Fig. 7). This jejunization is a useful supportive

sign of aganglionosis.

6. In children with Hirschsprung disease and colitis, the enema may show **mucosal nodularity, mucosal irregularity,** and **spiculation**[17] (Fig. 7). Clearly, enemas should not be performed in newborns who are critically ill with colitis.

7. The radiologic diagnosis of total colonic Hirschsprung disease is very difficult. Findings include a normal barium enema, **a short colon of normal caliber**, a **microcolon**, and a **transition zone in the ileum**[22,23] (Fig. 8). Additional findings include **easy, extensive reflux far back into small bowel**[24] and a **pseudotransition zone in the colon.**[22] Bowel shortening is seen as loss of the redundancy of the sigmoid colon that is seen in normal infants. The colon has been described as resembling a question mark.[8] There is also prolonged retention of barium.[23]

DIFFERENTIAL DIAGNOSIS

The most important disease to be considered in differential diagnosis is functional immaturity of the colon (meconium plug syndrome or neonatal small left colon syndrome). In contrast to Hirschsprung disease, in the first days of life the barium enema shows a marked narrowing of the distal colon up to the left colonic flexure with an abrupt transitional zone in the area of the left colonic flexure,[4] and in a patient with functional immaturity of the colon, the rectum is usually quite distensible.[4] The diagnosis is usually made clinically, as these patients often have diabetic mothers, are premature, and improve spontaneously.[10]

Allergic colitis due to cow milk protein can mimic Hirschsprung disease; it causes narrowing and mucosal irregularity of the rectum, an abnormal rectosigmoid index, and a transition zone.[25] However, pronounced mucosal fold thickening and irregularity are constant findings in allergic colitis, reflecting the inflammatory response of the rectum to the antigenic proteins. In comparison, the rectal mucosa in Hirschsprung disease is usually intact, with the serrated appearance of the contrast column due to intermittent spasm.[25] In addition, the location of the mucosal abnormality is atypical for Hirschsprung enterocolitis, in which mucosal abnormality is seen predominantly above the level of the transition zone.[11]

Salmonella colitis has been reported to cause narrowing and mucosal irregularity of the rectum and an abnormal rectosigmoid index.[26] It has also been reported to cause large bowel obstruction and mimic Hirschsprung disease with total colonic aganglionosis. There has been one case reported of a barium enema showing a narrowed and edematous colon with blunting of the hepatic and splenic flexures.[27]

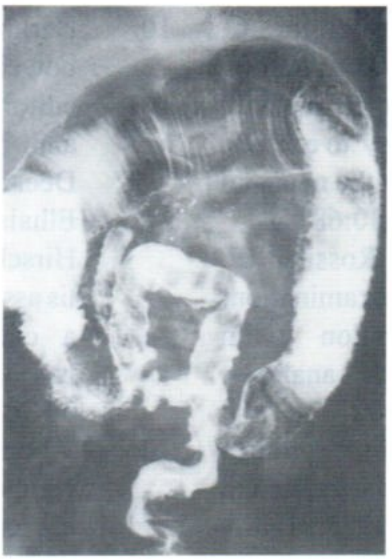

Fig. 7. Barium enema shows long segment Hirschsprung disease. The transition zone is at the junction of the distal descending colon and sigmoid. Note mucosal irregularity of the descending colon from colitis and jejunization at the transverse colon.

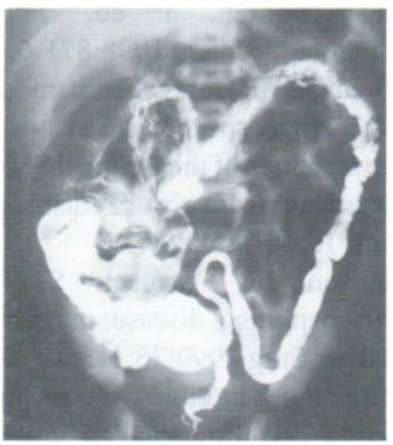

Fig. 8. Barium enema (left) shows narrowing of the rectum, sigmoid, descending colon, and transverse colon; and reflux of barium into the small bowel. A 24-hour delayed film (right) shows diffuse narrowing of the colon and dilatation of the distal small bowel. At surgery total colonic aganglionosis was found.

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