

Rare Variant Hirschsprung Disease Presenting as Large Bowel Obstruction in Adulthood: Hypoganglionosis in the Anorectal Canal

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ABSTRACT

Hirschsprung disease (HD) is a congenital bowel innervation disorder characterized by the absence of ganglion cells in the neural plexus of the colorectal wall. Variant HD describes a heterogeneous group of intestinal innervation disorders in which clinical presentation resembles HD despite the presence of ganglion cells seen in rectal biopsies. We present the first reported case of a rare variant HD, hypoganglionosis isolated in the anorectal canal, diagnosed in an adult who presented with a long history of constipation and treated with proctosigmoidectomy with coloanal anastomosis. Histopathology showed rare ganglion cells isolated in the anorectal canal.

INTRODUCTION

Hirschsprung disease (HD) is a congenital bowel innervation disorder characterized by the absence of ganglion cells in myenteric and submucosal plexus in the distal colon.¹ The diagnosis of HD can be confirmed by histological evaluation demonstrating the absence of ganglion cells in the submucosa.² The absence of ganglion cells begins from the anal region and can extend to a variable length as high as the duodenum.³ The term “variant Hirschsprung disease” has been used to describe a heterogeneous group of intestinal innervation disorders in patients with a clinical presentation that resembles HD despite the presence of ganglionic cells seen in rectal biopsy.² Isolated hypoganglionosis is a rare variant HD with resected bowel histopathological findings of reduction in the number of nerve cells in the myenteric ganglia.²

CASE REPORT

We present a case report of a 19-year-old man diagnosed with isolated hypoganglionosis in the anorectal canal after a long history of fecal impaction and constipation since childhood. The patient passed his meconium on the day of birth, but by 1 week of age, he experienced projectile vomiting and constipation, which was relieved by a lactose-free formula. At 2 years of age, he suffered severe constipation and was prescribed magnesium hydroxide. At 5 years of age, he was diagnosed with encopresis and was prescribed magnesium hydroxide, polyethylene glycol, and enemas. At 12 years of age, he continued to be symptomatic and treated intermittently with fecal disimpaction. At 15 years of age, he endured daily episodes of fecal incontinence, alternating between constipation and diarrhea.

Because of changes in health insurance coverage, the patient was lost to follow-up. His next medical encounter occurred at 19 years of age when he presented to our institution with complaints of distended abdomen. Initial vitals included temperature 37°C, heart rate 70, respiratory rate 18, blood pressure 121/79, and oxygen saturation 99% on room air. Physical examination demonstrated a distended abdomen, absence of abdominal surgical scars, intact rectal tone, and hard stool palpated in the rectal vault. Complete blood count and comprehensive metabolic panel were within normal limits. Computed tomography imaging showed a massively distended rectosigmoid colon filled with abundant fecal material (Figure 1). Because of his inability to tolerate a bedside digital rectal examination,

he was brought to the operating room to receive disimpaction under general anesthesia. The procedure note indicated the removal of “a very large cannon ball-sized fecaloma” after digital fragmentation and pulse irrigation.

During the outpatient follow-up, an abdominal series showed stool scattered throughout his colon and a dilated, saccular appearing rectum. A water-soluble contrast enema revealed a short segment (approximately 8 cm) narrowing of bowel in the region of the rectum with dilated upstream sigmoid colon (Figure 2). The remainder of the visualized bowel, including proximal sigmoid, descending, transverse, ascending colon, and cecum, were of normal caliber. The colorectal surgeon considered the patient’s history and imaging pattern to be sufficient for diagnosis and treatment of HD. The patient was scheduled for laparoscopic-assisted proctosigmoidectomy with a stapled end-to-end coloanal anastomosis without further diagnostic testing. During the surgery, there was significant difficulty with circumferential dissection of the rectum because of the chronic thickness and size of the rectum; therefore, diverting loop ileostomy was elected.

Although the operative report noted that the colon and omentum were adherent to the spleen, there was no other mention of adhesions. The surgical pathology demonstrated the presence of ganglion cells and hypertrophy of the smooth muscle layer in the sigmoid colon and rectum, but the anorectal canal showed the presence of rare ganglion cells with active inflammation and surface erosion (Figure 3). On follow-up, the patient underwent another water-soluble contrast enema that confirmed an intact anastomosis, and he subsequently underwent successful ileostomy closure 2 months later. One week after closure, the patient had recovered well, reporting daily bowel movements with good evacuation control and no fecal incontinence.

DISCUSSION

HD affects about 1 in 5,000 live births and usually presents in the neonatal period.⁴ A small number of cases with less severe

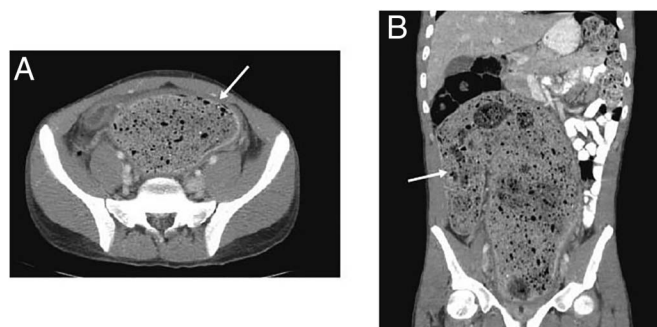


Figure 1. Abdominal and pelvic computed tomography with intravenous and oral contrast was performed on admission with axial images and coronal reformations (A and B, respectively). These images demonstrated marked sigmoid colonic fecal loading and dilation (white arrows), compatible with a chronic obstructive process, such as Hirschsprung disease.

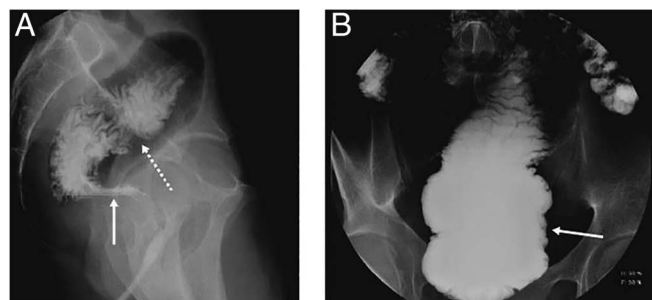


Figure 2. Water-soluble enema study showing (A) a short segment (8 cm) narrowing of the distal rectum (white solid arrow) with upstream dilatation of the proximal rectum (white dashed arrow) and (B) the anteroposterior view demonstrates massive upstream dilatation of the rectum and sigmoid colon.

disease are frequently misdiagnosed as chronic constipation, and HD may not be recognized until adulthood.⁵ The diagnosis of HD is usually much more challenging in adults than in infants because of the relatively benign course from a higher incidence of a short or ultra-short segment of aganglionosis in the former.⁶ Review of the National Surgical Quality Improvement Program database from 2012 to 2016 identified 35 adult patients diagnosed with HD.⁷ In retrospect, HD could have been suspected on the basis of our patient’s long-standing history of constipation since childhood, managed with laxatives, motility agents, irrigation, and manual disimpaction.

Our case report demonstrates the importance of performing a thorough investigation in adults with chronic refractory constipation. According to the American Gastroenterological Association’s technical review on constipation, evaluation should begin with history and physical examination. Patients without an identifiable cause of constipation should have a colonic structural inspection, which can be accomplished with colonoscopy, flexible sigmoidoscopy, contrast enema, or computed tomographic colonography.⁸ Because constipated patients often have difficulty with achieving good quality of colon

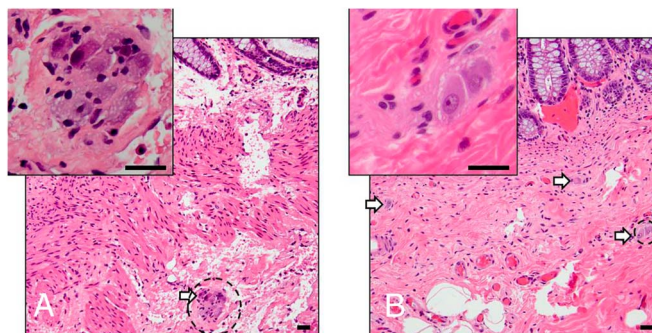


Figure 3. Hematoxylin and eosin stain of (A) the anorectal canal and (B) the sigmoid rectal area. Arrows indicate ganglion cells, which were rare in the anorectal canal region. Insets show the higher magnification of the circled area.

cleansing required for a colonoscopy, a contrast enema may be a preferable option for assessment of colonic structure. If these measures above are unrevealing, a colonic transit study can help distinguish between slow colonic transit and outlet dysfunction.⁹ In any instance in which an obstructive process may be in play, barium is contraindicated and, for the most part, is not used all that much anymore except for nonemergent outpatient examination.

The hallmark feature of HD on contrast enema is the exhibition of a luminal caliber change, demonstrated by the presence of a narrow zone in the distal part of the colon with a proximal cone-shaped segment.¹⁰ The so-called “transition zone” describes a colonic segment where a properly innervated intestine (widened) descends into the aganglionic segment (narrowed).¹⁰ Differentiation of HD and its variants requires rectal biopsy and histological evaluation. To the best of our knowledge, this is the first case of adult HD variant, intestinal hypoganglionosis, with histology demonstrating hypoganglionosis isolated in the anorectal canal and no evidence of a ganglionsis in the rectum and sigmoid colon.

DISCLOSURES

Author contributions: All authors contributed equally to this article. JH Birkholz is the article guarantor.

Financial disclosure: None to report.

Previous presentation: This case was presented at the Pennsylvania Radiological Society Annual Meeting, September 25-26, 2020; Virtual.

Informed consent could not be obtained from the patient despite several attempts. All identifying information has been removed from this case report to protect patient privacy.

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