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CASE DECISION

# A case of extensive colonic dilation complicated by ileus due to Hirschsprung's disease in adults: An easily missed diagnosis

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**CASE SUMMARY**
Hirschsprung's disease is a congenital condition characterized by the absence of ganglion cells in the colon. While approximately 90% of cases are diagnosed before the age of five, the occurrence of Hirschsprung's disease in adults is rare and frequently overlooked. This report presents the case of a 61-year-old male patient who was admitted to the hospital with symptoms indicative of intestinal obstruction caused by dilatation of the entire colon. A comprehensive medical history combined with computed tomography findings suggested Hirschsprung's disease. The patient subsequently underwent a subtotal colorectal resection with ileorectal anastomosis. A postoperative follow-up at five months indicated favorable outcomes.

***Keywords:*** Hirschsprung's disease, Intestinal obstruction, Surgical outcome

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## INTRODUCTION

Hirschsprung disease (also known as congenital aganglionic megacolon) is a motility disorder of the gastrointestinal tract caused by failure of neural crest cell migration during fetal development, leading to the absence of ganglion cells in the colon. This results in the affected segment of the colon being unable to relax, which leads to functional intestinal obstruction. It is one of the most common congenital disorders in children, with an incidence of 1:5000 live births, and is more frequently observed in boys. Most patients are diagnosed during the neonatal period, with over 95% diagnosed before one year of age. Hirschsprung disease in adults is very rare and is diagnosed after the age of 10 years. The disease often presents as chronic constipation and can easily be misdiagnosed as other causes, leading to ineffective treatment.

We report a case of Hirschsprung's disease in a 61-year-old male patient. The patient underwent total colectomy with ileorectal anastomosis, and his condition was stabilized postoperatively. Through this clinical case, we aimed to raise awareness among clinicians about this rare condition to avoid missed diagnoses.

## CASE PRESENTATION

A 61-year-old male patient presented to the emergency department of 108 Central Military Hospital with a two-day history of intermittent abdominal cramping, progressively increasing abdominal distension, and constipation. He still passed small amounts of gas but had no nausea or vomiting. His childhood developmental history was normal. For the past two years, he had experienced chronic constipation,

typically having bowel movement only once every 10-15 days, with hard stools. Despite multiple consultations at various medical facilities and prescription of laxatives and enemas to soften stools, his condition did not improve. He often had to manually evacuate stools during defecation. The family had no notable medical history.

On physical examination, the patient was in a stable condition, with a blood pressure of 132/80 mmHg, pulse of 88 beats per minute, no fever, and normal skin turgor. His abdomen was markedly distended but soft, with prominent bowel loops (+), absent peristaltic waves (−), no localized tenderness, and no abdominal wall rebound tenderness. Digital rectal examination revealed increased anal sphincter tone, with a small amount of hard stool in the rectal vault and no palpable masses.

Blood tests are within normal limits. The CT scan shows significant dilation of the entire colon (Figure 1), with the colonic lumen filled with hard stool; no abnormal wall thickening or tumors are detected.

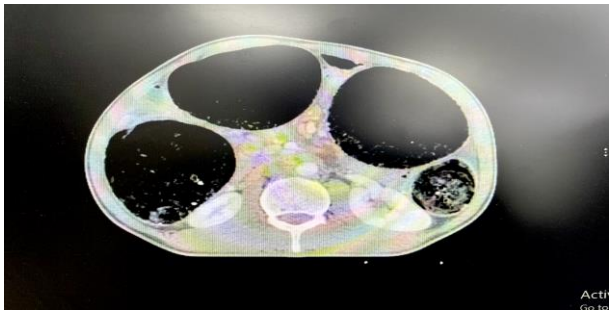
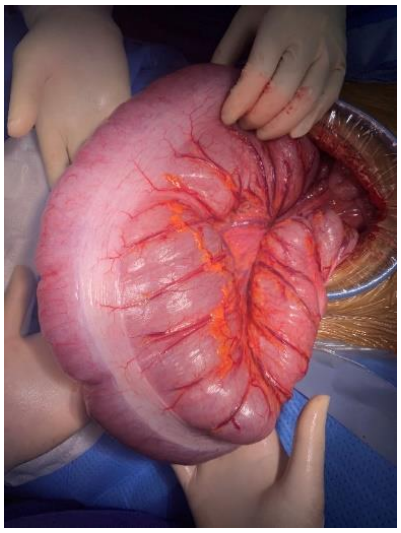


***Figure 1:*** *Image showing dilation of the entire colon on CT scan.*

The patient was provisionally diagnosed with partial intestinal obstruction due to colonic distension with an indication for surgery. However, because of his stable condition and no immediate need for emergency surgery, the patient was admitted for conservative treatment, including fasting, intravenous nutrition, and water enemas, aimed at reducing the stool burden in the colon to facilitate surgery. After four days of medical treatment, the abdominal distension improved, but the daily stool output from the enemas was minimal, leading us to decide on surgical intervention to address the underlying cause.

During the surgery, midline laparotomy was performed, revealing a massively dilated colon from the cecum to the sigmoid colon, with a maximum diameter of approximately 15 cm, containing a large amount of soft stool and showing a lack of peristalsis (Figure 2A). The rectum had a normal diameter with no masses. Owing to the high risk of septic complications from the long-standing stool burden, we decided to perform a near-total colectomy and rectal resection (Figure 2B), restoring gastrointestinal continuity with an ileorectal anastomosis using a J-pouch configuration and an EEA 28 stapler. Intraoperative frozen section biopsy confirmed the diagnosis (consistent with Hirschsprung disease) and determined the resection margins to ensure complete removal of the affected tissue.



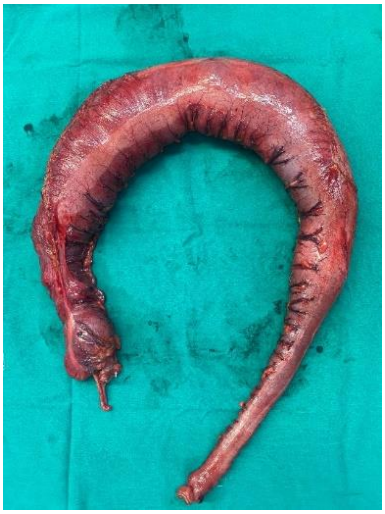


***Figure 2:*** *Gross intraoperative images. [A] Large dilation of the entire colon from the cecum to the upper rectum, with a maximum diameter of approximately 15 cm. [B] Surgical specimen after near-total resection of the colon and rectum.*

Postoperatively, the patient's condition remained stable. He started passing gas on the third postoperative day (POD3) and had bowel movements by POD4, with soft yellow stools occurring 3-4 times a day. The patient was discharged on POD7 after successfully tolerating oral nutrition and achieving adequate gastrointestinal function. At the 5-month follow-up, the patient recovered well, with an average of two soft stools per day and a return to normal daily activities.

## DISCUSSION

Hirschsprung disease in adults is rare and is typically diagnosed during infancy. The first case was reported in the 1880s by Dr. Harald Hirschsprung, and the first adult case was published in 1950. Its pathogenesis includes the complete absence of ganglion cells in the myenteric and submucosal plexuses of the colonic wall, starting from the upper border of the internal sphincter, and abnormal hypertonicity of the internal sphincter. These factors result in the loss of the ability of the affected colonic segment to relax, causing stool accumulation in the proximal segment and leading to functional intestinal obstruction. The incidence of Hirschsprung disease in adults is difficult to determine due to frequent misdiagnosis; all reports are case studies, primarily in patients aged 20-30 years. Cases involving middle-aged patients, such as ours, are rare.

Miyamoto et al. suggested that up to 2% of adults with chronic constipation might have Hirschsprung disease, with most of these patients having the short-segment type (pathological segment limited to the rectosigmoid colon). Adults with chronic constipation are often initially managed by primary care providers, highlighting the need for increased awareness of this condition to ensure appropriate specialist referrals.

Hirschsprung disease is classified based on the length of the aganglionic segment as follows: 1) The most common form involves short-segment aganglionosis, seen in 75-80% of cases, with the aganglionic segment located in the distal sigmoid colon and rectum. 2)The second type, long-segment aganglionosis, accounts for 10% of cases, in which the aganglionic segment extends from the rectum to the splenic flexure through the sigmoid colon. 3) The rarest and most severe form is total colonic aganglionosis, which is observed in 5% of patients, where the entire colon lacks ganglion cells. 4)The final type, ultra-short-segment aganglionosis, involves a very short aganglionic segment in the anal canal, just above the dentate line.

Understanding these classifications and the clinical presentation of Hirschsprung’s disease is crucial for timely diagnosis and management.

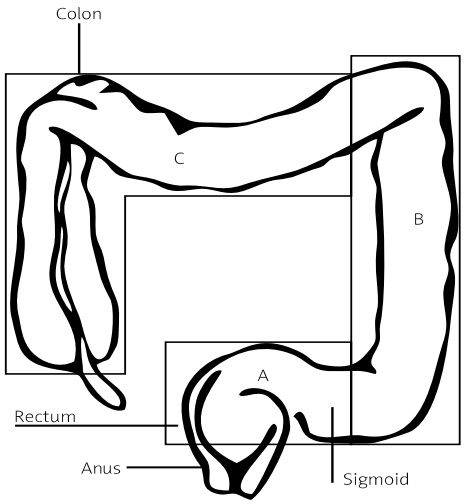



***Figure 3:*** *Classification of Hirschsprung's disease based on the length of the aganglionic segment: A – Short aganglionic segment; A and B – Long aganglionic segment; A, B, and C – Total aganglionic segment of the entire colon [9].*

Once Hirschsprung's disease is clinically suspected, appropriate diagnostic tests should be performed, including imaging (X-ray, CT scan), anorectal manometry, and rectal wall biopsy. Approximately 80% of Hirschsprung cases are of the short-segment type [1]; therefore, CT scans or contrast X-rays of the colon can identify the transition zone between the narrow pathological segment and dilated normal colon. However, in our patient's case, prolonged fecal stasis caused significant colonic dilation, making the transition zone undetectable on CT scans, and it was also impossible to administer a contrast enema due to obstruction by a large fecal mass. Anorectal manometry, used to identify the absence of the rectoanal inhibitory reflex, is a valuable noninvasive test with a sensitivity of up to 97% [10]. However, only a few medical facilities in Vietnam use this equipment. Finally, rectal wall biopsy is considered the gold standard for diagnosing Hirschsprung disease. Biopsies can be performed using suction biopsy with specialized equipment, endoscopic biopsy, or surgical biopsy. In our patient, because of the ineffectiveness of conservative medical treatment, surgery was clearly indicated. Therefore, we decided to perform an intraoperative biopsy, which revealed an absence of ganglion cells.

Surgery is the definitive treatment for Hirschsprung disease. The goal of surgery is to remove the pathological segment of the intestine and restore digestive continuity between the segments of the intestine and normal ganglion cells. Surgery can be performed in a single stage, including resection and immediate anastomosis, or in two stages, with the initial creation of a temporary stoma followed by definitive surgery later [11]. One of the most important principles of surgery is to preserve as much of the internal anal sphincter as possible to ensure bowel function, especially because most patients are children or young adults who need a good quality of life. Surgery can be performed laparoscopically or open via the abdominal or anal route depending on the patient's condition. Due to less significant fecal stasis in children, laparoscopic or transanal resection can be performed [11]. In adults with Hirschsprung's disease, due to prolonged fecal stasis, large, hard fecal masses, and significant bowel distention, open surgery through the midline is often required [12-14].

The surgical method depends on the type of disease and must be individualized for each patient. For congenital megacolon, the narrow segment, transition zone, and dilated segment were resected, and frozen section biopsy of the rectal margin was performed to determine the resection margin. An ileostomy is created if the anastomosis is low and closed after 3-6 months. Total colectomy with ileorectal or colorectal anastomosis can be performed for idiopathic megacolon in

adults. In emergency cases, a temporary stoma is initially created followed by definitive surgery. For megacolons with ganglion cell deficiency, total colectomy may be performed because the rectum is less affected, allowing an ileorectal or ascending colonic-rectal anastomosis. For toxic megacolon, segmental colectomy can be performed in mild cases, while total colectomy is performed for severe infections. For treatment-induced megacolon, resecting the narrowed segment with colonic-rectal anastomosis is an option [15].

Classic anastomosis techniques include end-to-end anastomosis of the proximal end to the rectum (Swenson technique), side-to-side anastomosis (Duhamel technique), or anastomosis of the proximal end to the rectum with mucosal and submucosal resection (Soave technique) [11]. Compared to Swenson and Duhamel, Soave requires less deep dissection in the mid-to-lower rectum, reducing the risk of pelvic nerve and vascular injuries.

In this clinical case, without an emergency surgical indication, we initially managed the patient medically to reduce fecal volume and abdominal distention to facilitate laparoscopic surgery. However, due to prolonged fecal stasis, the hard fecal mass responded poorly to conservative treatment, necessitating an open midline approach. In our patient, the entire colon was dilated with feces, making it difficult to identify the transitional zone. We performed an intraoperative biopsy to diagnose and assess the colonic damage. Due to total colectomy, we preserved a portion of the rectum (the lower two-thirds) to improve postoperative bowel function despite biopsy findings indicating residual rectal pathology. We used J-pouch ileorectal anastomosis to create a new rectal reservoir to improve postoperative bowel function and quality of life [16]. Five months after surgery, the patient reported good bowel function, averaging two bowel movements per day.

A study by Shengzhe Ma and colleagues on 89 Hirschsprung patients, aged 16-73 years (mean age 43.61 ± 15.79), found that 46.1% (41 patients) had congenital megacolon. Of these, 29 underwent low anterior resection with ileostomy and 12 underwent low anterior resection without ileostomy. Idiopathic adult megacolon was observed in 39.3% (35 patients) of patients, and 68.6% (24 patients) underwent total colectomy with ileorectal anastomosis. Of these, 31.4% (11 patients) presented with bowel obstruction, five received medical treatment, and six underwent emergency surgery with initial stoma creation, followed by total colectomy. Two patients (2.25%) were diagnosed with ganglion cell deficiency after surgery, with a narrowed segment primarily in the descending and sigmoid colon and less involvement of the rectum. The surgical treatment involved segmental colectomy and ileorectal or colorectal anastomosis. Toxic megacolon was seen in 3.38% (three patients), treated with total or segmental colectomy to prevent systemic infection. Megacolon due to treatment was seen in 7.87% (eight patients), with six developing rectal strictures post-radiation therapy for cancer, managed initially with decompressive rectal tubes, but optimally with stoma creation. All patients recovered and were discharged without fatalities. The mean hospital stay was 19.5 days. Complications occurred in 10 cases (11.24%), including postoperative bowel obstruction (3.37%), anemia (2.25%), abdominal fluid accumulation (1.12%), wound infection (1.12%), and one case of anastomotic leak treated conservatively [15].

Comparing our patient outcomes with this study highlights the importance of accurate diagnosis and appropriate treatment.

## CONCLUSION

Hirschsprung's disease in adults is rare and often overlooked, leading to delayed treatment and increased difficulty in management. Clinicians should be vigilant, especially in patients with prolonged, unexplained constipation. Surgery is the only definitive treatment that significantly improves patients' quality of life. Laparoscopic surgery offers aesthetic and functional benefits with proper patient preparation, whereas open surgery is reserved for patients with significant colonic dilation, complicating laparoscopic procedures.

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**Conflict of interests**

The authors declare that there is no conflict of interest regarding the publication of this article.

**Sources of funding**

None.

**Consent**

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

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