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Case Report

Adult Hirschsprung's disease: report of four cases

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Abstract: Adult Hirschsprung's disease (HD) is a rare motor disorder of the gut that is frequently misdiagnosed as refractory constipation. The primary pathogenic defect in adult HD is identical to that seen in infancy or childhood, and is characterized by the total absence of intramural ganglion cells of the submucosal (Meissner) and myenteric (Auerbach) neural plexuses in the affected segment of the bowel. Ninety-four percent of HD cases are diagnosed before the patient reaches 5 years of age, however, on rare occasion, mild cases of HD may go undiagnosed until he or she reaches adulthood. In this study, we describe four cases of adult HD with a history of longstanding recurrent constipation, relieved by laxatives, and presenting to the Department of Gastrointestinal Surgery with progressive abdominal distention, colicky pain or acute intestinal obstruction. Barium enema or computed tomography revealed a grossly distended proximal large colon with fecal retention. Intraoperative frozen section biopsy was performed in all cases and showed aganglionosis of the stenotic segment and a normal distal rectum. In all cases, patient symptoms were completely resolved and there were no complications arising immediately post-surgery or at one-year follow-up. Adult HD should be considered in the differential diagnosis of cases where adult patients present with chronic constipation or even acute intestinal obstruction. The modified one-stage Martin-Duhamel or Rehbein's procedure is a feasible surgical option for treating cases of adult HD involving a segment or the entire bowel.

Keywords: Adult, constipation, treatment, Hirschsprung's disease

Introduction

Hirschsprung's disease (HD) occurs in approximately one in 5000 live births with an overall male to female ratio of 3:1 to 4:1 [1, 2]. The cause of HD is most commonly attributed to defective craniocaudal migration of neuroblasts originating from the neural crest during the first twelve weeks of gestation, resulting in functional intestinal obstruction [3, 4]. HD is not commonly seen in adults as most patients are diagnosed early in life and are treated surgically. However, some patients with mild symptoms may go undiagnosed into adulthood, likely because the colonic region proximal to the distally obstructed segment assumes a compensatory role [5, 6]. Such cases, specifically those in which the diagnosis is established when the patient is over ten years of age, are termed "adult HD" [7, 8]. Generally, patients can manage this condition through the use of cathartic agents. However, at some point, the dilated proximal colonic segment may decompensate secondary to the distal obstruction and patients may experience rapidly worsening constipation or even acute obstruction. Herein, we report four cases of adult HD and describe our experience in diagnosing and treating this rare disease.

Case report

Case 1

A 67-year-old woman presented with chronic symptoms of recurrent constipation and abdominal distention which she had been experiencing over 5 years. The constipation had improved over the years through use of an enema. However, during the last six months, her constipation had worsened and she had also suffered abdominal distention associated with colicky pain. She had no hematochezia, had not undergone any previous surgery and had no family history of HD. Clinical examination showed no abdominal mass on palpation but the abdomen was grossly distended with visible peristalsis. Digital rectal examination

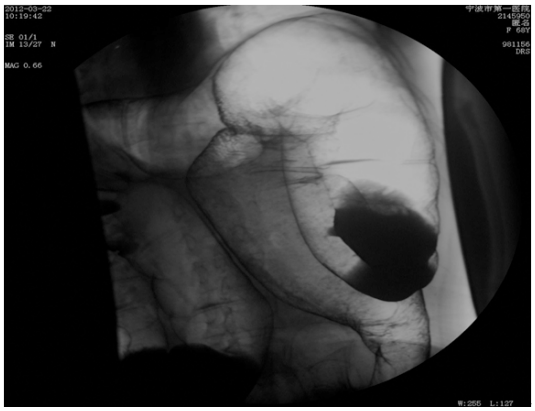

Figure 1. Barium enema revealing an extremely dilated sigmoid colon with a corn shaped transition zone proximal to a narrow segment.

revealed good anal sphincteric tone with a high rectal fecal load. Routine laboratory evaluation, including a complete blood count, urinalysis, and blood chemistry testing, were all within normal limits. The barium enema showed a grossly dilated, large colon, with a transition zone located at the middle one-third of the rectum (**Figure 1**). Computed tomography (CT) with three-dimensional reconstruction showed a dilated sigmoid colon and descending colon (**Figure 2**). A clinical diagnosis of adult HD was made. Seven days after bowel preparation, the patient underwent low anterior resection with colorectal anastomosis (Rehbein procedure). Histological staining showed an aganglionic narrowed segment with a normal distal rectum (**Figure 3**). The patient followed up for one and half years without any constipation or other complications.

Case 2

A 46-year-old female was admitted in March 2012 requesting an intestinal stoma closure. She had been experiencing recurrent episodes of constipation and abdominal distention since early adulthood before acute abdominal pain, progressive abdominal distention, and severe vomiting presented last year. An exploratory laparotomy had been carried out at another hospital. Nothing abnormal was observed aside from a dilated ascending and transverse colon; therefore, a right hemicolectomy and ileostomy was performed. Postoperative histological assessment demonstrated a lack of ganglion cells in the colon wall, strongly suggestive of a diagnosis of adult HD. Five days following admission, the left dilated descending colon and the sigmoid colon including the stenosed region were resected and ileorectal anastomosis was performed using a modified one-stage Martin-Duhamel procedure. Histological examination of the stenosed segment revealed rudimentary aganglionic cells consistent with the diagnosis of adult HD (**Figure 4**).

Case 3

A 52-year-old woman was admitted to Emergency suffering from recurrent abdominal pain and distention over a period of 6 months. Over the last three days, these symptoms worsened and were accompanied by constipation. Clinical examination showed a markedly distended abdomen with visible peristalsis. The patient also presented with abdominal tenderness and Blumberg's sign was mildly positive. CT showed an extremely dilated colon containing feces (**Figure 5**). The patient was diagnosed with adult HD and an emergency total colectomy with ileorectal anastomosis (modified Martin-Duhamel procedure) was performed. Consistent with the CT results, surgery revealed a massively dilated colon with a narrow transition zone at the distal rectum (**Figure 6**). Intraoperative frozen section biopsy and postoperative histological assessment confirmed the diagnosis of the adult HD (**Figure 7**).

Case 4

A 55-year-old female was admitted to Emergency with symptoms of increasing abdominal distention and pain in May 2012. The patient had a history of chronic constipation requiring daily enemas since early adulthood. Three years ago, an exploratory laparotomy was performed in another hospital for symptoms of acute abdominal pain and progressive abdominal distention. Surgery revealed a dilated sigmoid colon thus a sigmoid resection was carried out. Histological evaluation showed a lack of ganglion cells in the excised sigmoid colon wall. Furthermore, the symptoms of chronic constipation and abdominal distention persisted after the operation. Clinical examination presented a moderately distended abdomen with an incision scar 20 cm in length. CT showed a highly dilated colon

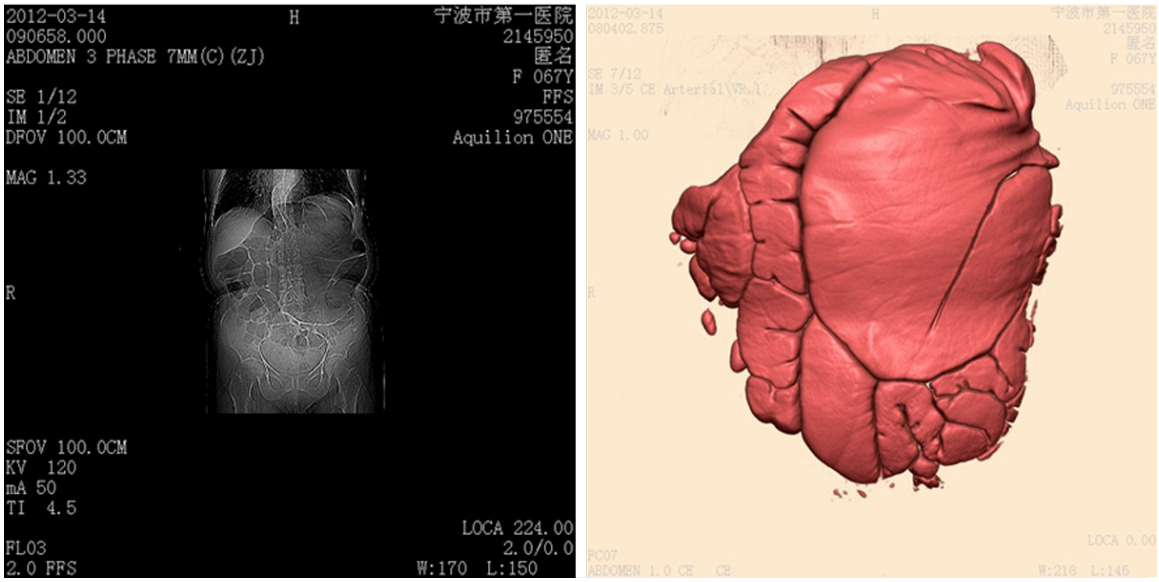

Figure 2. Computed tomography with three-dimensional reconstruction showing a dilated sigmoid colon and descending colon.

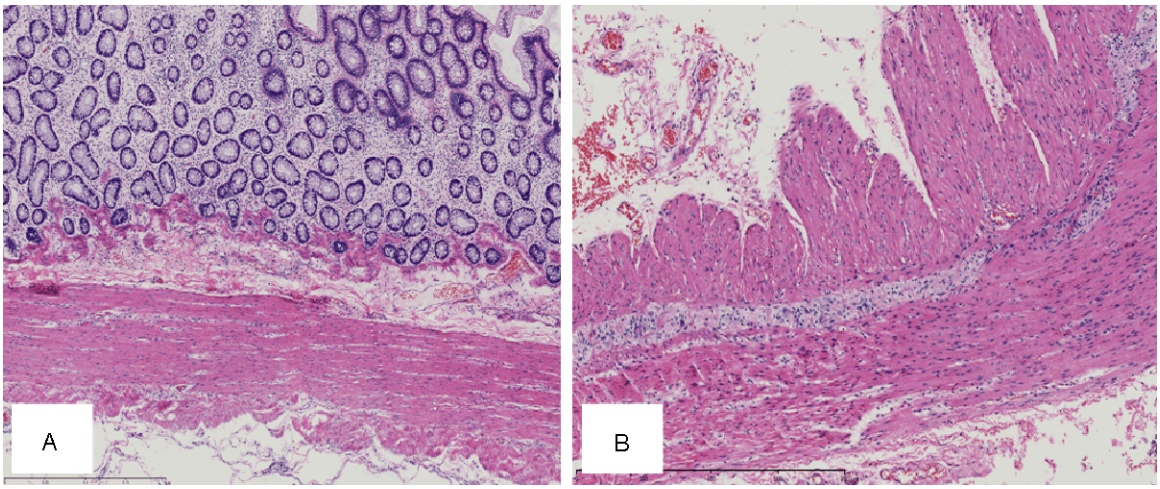

Figure 3. Microphotography showing an absence of ganglion cells in the myenteric plexus (A) and a normal distribution of ganglion cells in the distal rectum (B) (H&E staining ×10).

containing feces (**Figure 8**). A diagnosis of adult HD was established based on the patient's history and the clinical data. Five days after admission, a modified Martin-Duhamel procedure was performed and postoperative recovery was deemed satisfactory. Since this definitive surgery, the patient's symptoms have completely subsided and bowel movements occur three to four times a day. Histological examination of the stenosed segment revealed aganglionic cells consistent with the diagnosis of adult HD (**Figure 9**).

Discussion

The first documented case of adult HD was described by Rosin et al. in 1950 [9]. The incidence rate of adult HD at present is not known, primarily because this disease is frequently overlooked in the adult population. According to the detailed literature review performed by Miyamoto et al., adult HD is more commonly seen in men than women with a male to female ratio of 133 to 42 [10]. However, in our study, all four cases of adult HD happened to be female.

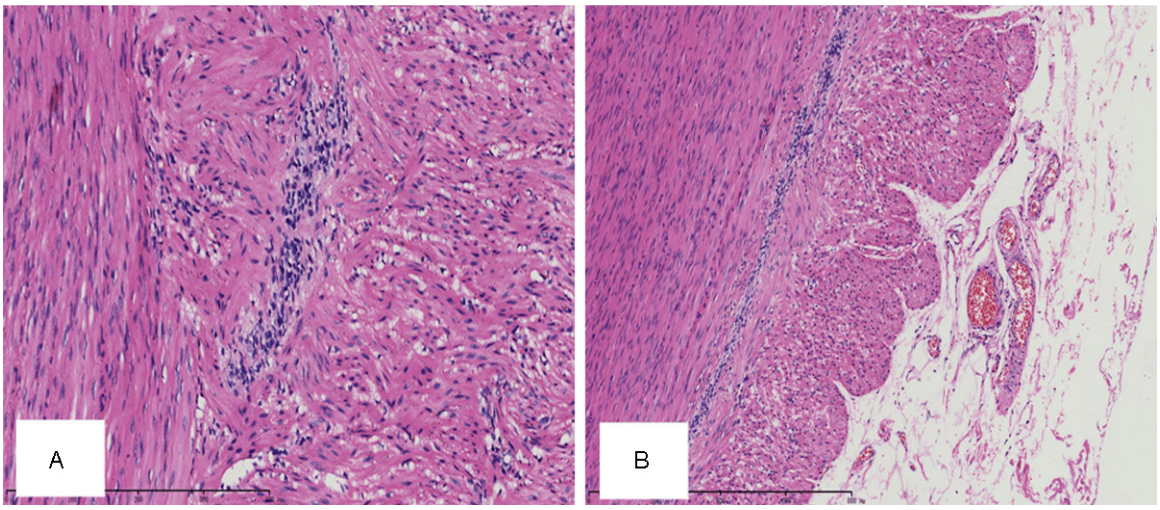

Figure 4. Microphotography showing an absence of ganglion cells in the myenteric plexus (A) and a normal distribution of ganglion cells in the distal rectum (B) (H&E staining ×10).

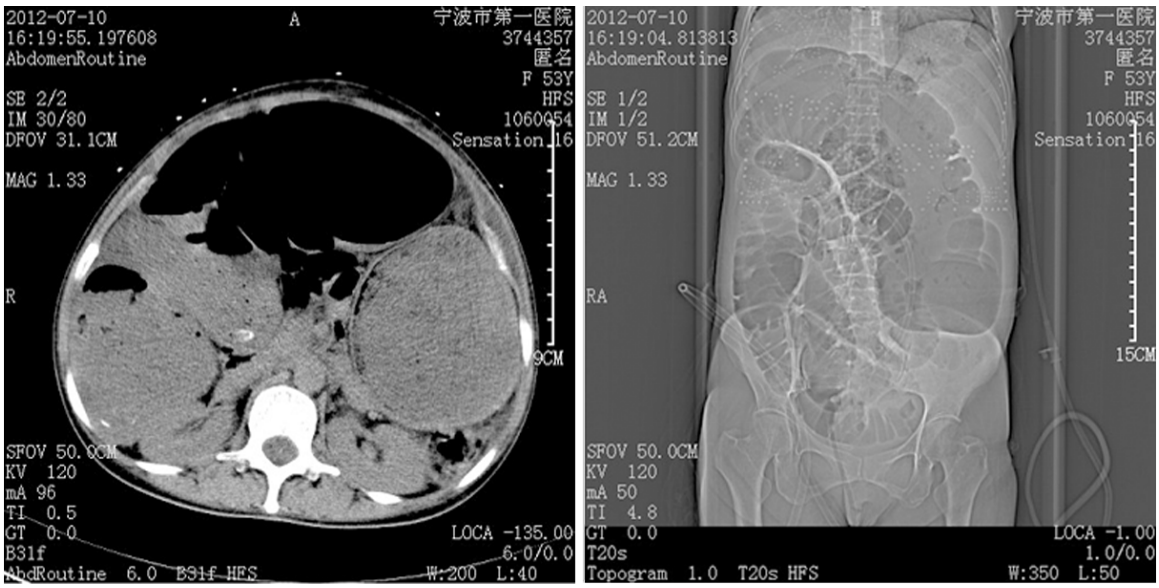

Figure 5. Computed tomography showing a total dilated colon containing feces.

Symptoms of adult HD consist of longstanding recurrent constipation and varying degrees of abdominal distention and pain. Most patients require regular use of cathartics in order to relieve their symptoms. On clinical examination, abdominal distention and tenderness are common and usually associated with palpable fecal masses. Radiographs of the abdomen typically show massive distention of the proximal region of the colon, with a small narrowed distal segment. CT is a useful imaging tool providing the opportunity to not only view the dilated colon and the transition zones but also to definitively exclude other diseases which can also cause chronic constipation in adults, such as colorectal cancer. An accurate diagnosis of adult HD is based on the collective assessment of medical history, barium enema test results, and most importantly, full-thickness rectal biopsy findings.

The surgical procedure developed to treat HD in children can be applied to adults with some modifications. Specifically, a one-stage

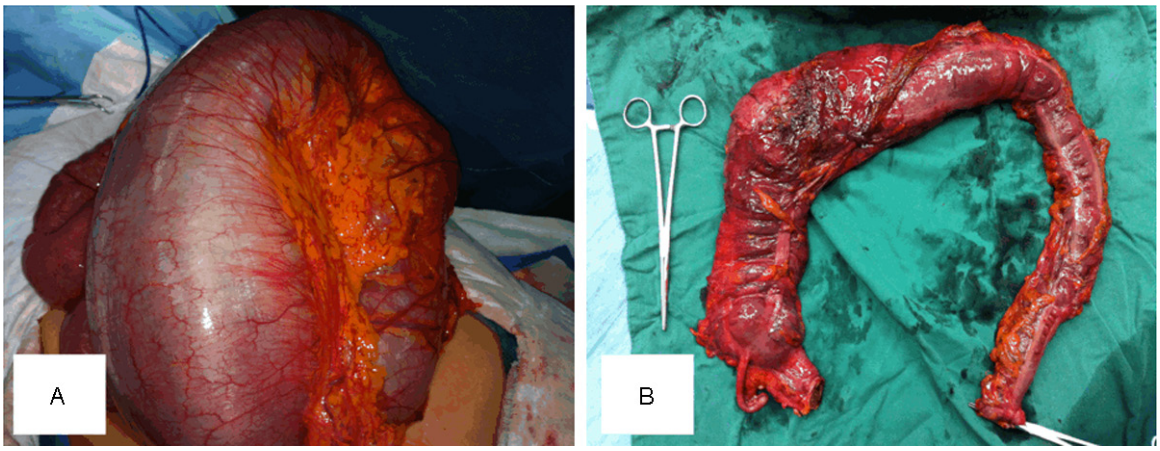

Figure 6. An extremely dilated colon (A) with a narrow transition zone at the distal rectum (B).

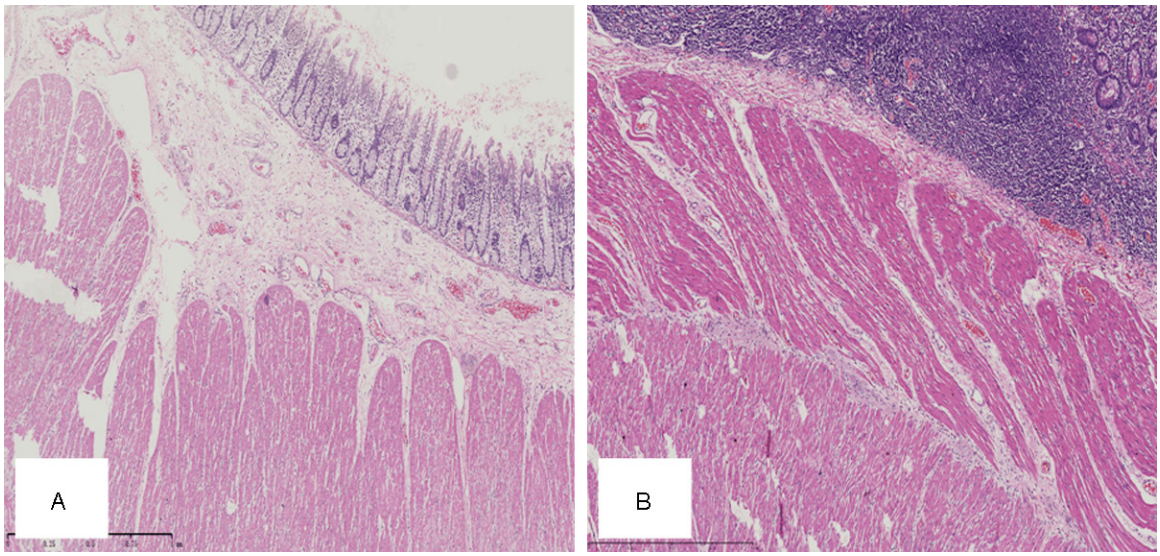

Figure 7. Microphotography demonstrating a lack of ganglion cells in the myenteric plexus (A) and a normal distribution of ganglion cells in the distal rectum (B) (H&E staining ×10).

approach is more feasible when treating adult HD due to the relatively healthier nutritional status of adult HD patients and the ability to use a GIA stapling device. In the four cases presented in this study, rectocolectomy with colorectal anastomosis (Rehbein procedure) or total colectomy with ileorectal anastomosis (modified Martin-Duhamel procedure) were carried out according to the extent of involved colorectal segment, without performing a temporary protecting colostomy. Intraoperative frozen section biopsy was required to identify the distal margin of the bowel with ganglion cells. The postoperative results were rated satisfactory without any complications. All four patients were healthy with complete resolution of symptoms, and were having bowel movements three to four times a day at 10 to 18 months' follow-up.

It is worth mentioning that on rare occasion, patients with adult HD present with acute intestinal obstruction. In these cases, a laparotomy may be required. In our study, case 2 and case 4 patients had both previously undergone emergent laparotomies, which had not resolved their condition. This is due in part to the atypical clinical features at presentation, but, more importantly, to limited awareness of this disease as it is not a common cause of acute

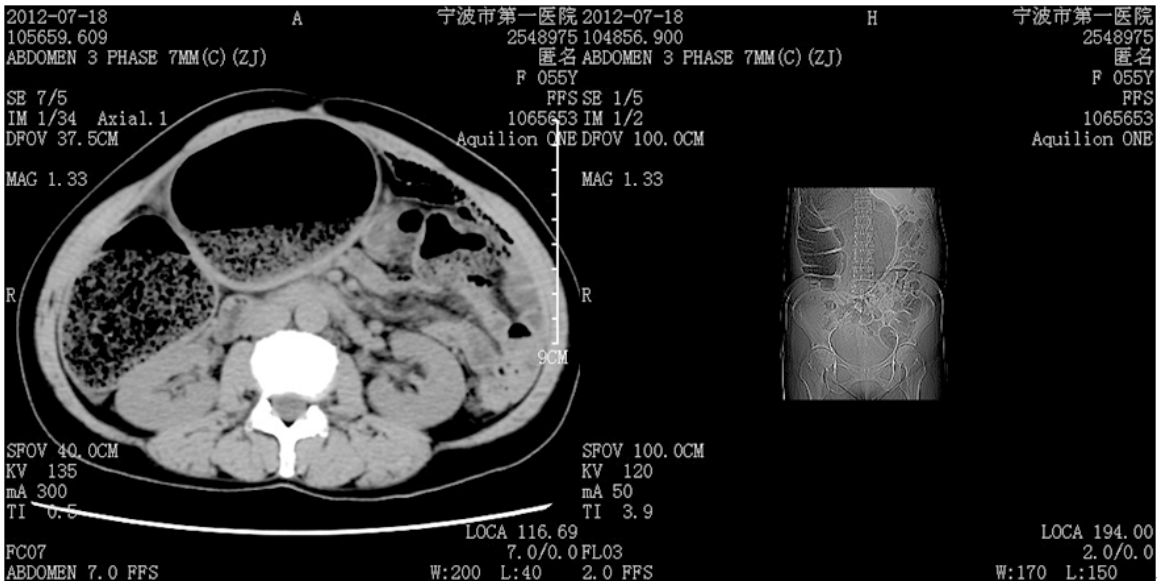

Figure 8. Computed tomography showing a dilated ascending colon and transverse colon.

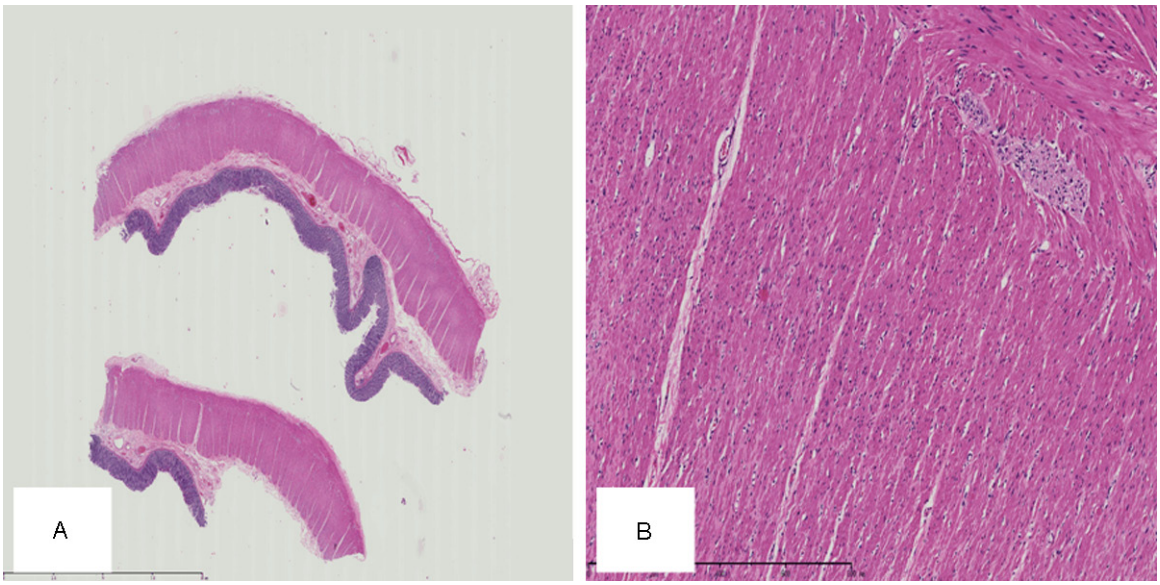

Figure 9. Microphotography showing the absence of ganglion cells in the myenteric plexus (A) and a normal distribution of ganglion cells in the distal rectum (B) (H&E staining ×10).

intestinal obstruction in adults [11]. However, in order to avoid such errors in the diagnosis and treatment of this condition, a possible diagnosis of adult HD should be considered when patients, such as those discussed herein, first present with a history of chronic constipation. In addition, intraoperative frozen section biopsy of the colorectum may be helpful when a patient has undergone an emergent laparotomy and a dilated colon is observed; but there is no obstructive cause identified.

In conclusion, adult HD should be considered in the differential diagnosis of patients that present with a markedly dilated feces-filled proximal colon with a transition zone and a narrowed distal colonic segment without any other obstructive cause, combined with a history of chronic

refractory constipation. A modified one-stage Martin-Duhamel or Rehbein's procedure would be a viable surgical option for adult HD.

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Disclosure of conflict of interest

None.

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References

[1] Chen F, Winston JH 3rd, Jain SK, Frankel WL. Hirschprung's disease in a young adult: Report of a case and review of the literature. Ann Diagn Pathol 2006; 10: 347-351.

[2] Suita S, Taguchi T, Ieiri S, Nakatsuji T. Hirschsprung's disease in Japan: Analysis of 3852 patients based on a nationwide survey in 30 years. J Pediatr Surg 2005; 40: 197-201.

[3] Fu CG, Muto T, Masaki T, Nagawa H. Zonal adult Hirschsprung's disease. Gut 1996; 39: 765-767.

[4] Bakari AA, Gali BM, Lbrahim AG, Nggada HA, Ali N, Dogo D, Abubakar AM. Congenital aganglionic megacolon in Nigerian adults: Two case reports and review of the literature. Niger J Clin Pract 2011; 14: 249-252.

[5] Natsikas NB, Sbarounis CN. Adult Hirschsprung's disease: an experience with the Duhamel-Martin procedure with special reference to obstructed patients. Dis Colon Rectum 1987; 30: 204-206.

[6] Kim HJ, Kim AY, Lee CW, Yu CS, Kim JS, Kim PN, Lee MG, Ha HK. Hirschsprung Disease and Hypoganglionosis in Adults: Radiologic findings and differentiation. Radiology 2008; 247: 428-434.

[7] Fairgrieve J. Hirschprung's disease in adults. Br J Surg 1963; 50: 506-514.

[8] Barnes PR, Lennard-Jones JE, Howley PR, Todd IP. Hirschprung's disease and idiopathic megacolon in adults and adolescents. Gut 1986; 27: 534-541.

[9] Rosin JD, Bargen JA, Wargh JM. Congenital megacolon of a man 54 years of age. Report of a case. Mayo Chin Proc 1950; 25: 710-715.

[10] Miyamoto M, Egami K, Maeda S, Ohkawa K, Tanaka N, Uchida E, Tajiri T. Hirschprung's disease in adults: Report of a case and review of the literature. J Nippon Med Sch 2005; 72: 113-120.

[11] Yüksel I, Ataseven H, Ertuğrul I, Başar O, Saşmaz N. Adult segmental Hirschsprung disease. South Med J 2009; 102: 184-185.