Case report

Total colonic aganglionosis with ileum extension Hirschsprung's disease in an adult: A case report

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ABSTRACT

Introduction: Hirschsprung's disease is a gastrointestinal anomaly affecting neuronal development and function. The primary abnormality is the absent ganglionic cells in the submucosal and myenteric neural plexuses. Disease presentation can vary, and patients may present with delayed meconium passage or late in adulthood. Total colonic aganglionosis is considered a rare subtype and even rarer to extend proximally into the small bowel. Therefore, adult Hirschsprung disease is frequently misdiagnosed as chronic constipation until comorbid events such as volvulus or perforation occur.

Case presentation: A 34 years South-Sudanese male presented with chronic constipation since early childhood, abdominal discomfort, pain, and distension. His bowel habits were infrequent, requiring occasional enemas. A strong family history of Hirschsprung disease was identified. He underwent total abdominal colectomy and end ileostomy. Postoperative diagnosis of TCA and small bowel extension of more than 50 cm was confirmed. The patient was discharged home after ileostomy education and possible future reconstruction.

Clinical discussion: Hirschsprung's ileal extension resembles a risk of morbidities and mortality. It increases as the segment extends proximally. No international consensus on surgical management. It should be tailored to patient condition, diseased segment length, and the possible definitive primary reconstruction. Our patient's surgical diversion considered the patient's long-standing constipation history and dilated fecal-loaded ileum.

Conclusion: TCA requires a high index suspension during the workup of chronic constipation patients. There is no reported superior surgical approach, and it depends on the institution's experience and surgeon's expertise. Primary reconstruction can be achieved, but we recommend an ileostomy for such cases.

1. Introduction

Hirschsprung's disease is a congenital gastrointestinal anomaly that affects neuronal development and function. The primary abnormality is the absence of ganglionic cells in the submucosal and myenteric neural plexuses of the GI tract. Disease presentation can vary in time and complaint, as some patients present with delayed passage of meconium as neonates and others present late in adulthood.

Reported Hirschsprung's disease (HSD) incidence of 1 in 3000 to 5000 living deliveries with male predominance [1]. According to aganglionic segment length, Hirschsprung's disease is classified into ultrashort, short-segment, and long-segment. In addition, the long-segment disease is subdivided into long-segment colonic, total colonic aganglionosis, and total colonic aganglionosis with small bowel extension.

Total colonic aganglionosis (TCA) was initially described by Zuelzer & Wilson and diagnosed in less than 8% of the disease population. It is considered a rare subtype of HSD and rarely extends proximally to affect the ileum and jejunum. Adult Hirschsprung disease is frequently misdiagnosed as chronic constipation until comorbid events such as volvulus or perforation occur. It may mandate extensive workup & radiological investigations to reach the ultimate diagnosis.

This work has been described according to the SCARE criteria [2].

2. Case presentation

A 34 years old South-Sudanese male presented with chronic constipation since early childhood long-standing history of abdominal
discomfort, abdominal pain, and distension. His abdominal pain was colicky, mild to moderate in severity, increased with distension, and partially relieved after defecation. His bowel habits were infrequent, with the passage of small hard feces every 4 to 7 days, occasionally requiring rectal enemas for defecation. No nausea, vomiting, or fever was reported. His initial management was an emergency surgical exploration four months prior to presentation for evacuation of fecaloma at a local hospital. The surgery was complicated by laparotomy wound infection and partial wound dehiscence. A strong family history of a similar illness was identified in his elder brother, who died of the same condition, and two siblings. One of his siblings was a girl of five years who recently died of enterocolitis of the same condition. He was referred to our gastrointestinal surgery unit at our tertiary care center for further investigations & management.

The patient's clinical examination revealed normal vital signs, apart from 110 beats/min tachycardia. Visible distension of the abdomen, an evident dilated bowel loop in the lower abdomen, and visible peristalsis at the periumbilical region. Moderate diffuse abdominal tenderness, but no guarding or rigidity. No deep masses or organomegaly were noticed. Bowel sounds were high-pitched with average frequency. Digital rectal examination (DRE) showed normal sphincteric tone, roomy rectum with hard feces, but no anorectal masses or blood on the gloved finger. Laboratory blood investigations and urinalysis were of average normal values.

Initial radiological assessment by abdominal erect and supine X-rays showed grossly dilated, fecal loaded large and small bowel, and fecal matter shadowing and air reaching the rectum (Fig. 1). In addition, a CT abdomen was utilized to exclude mechanical obstruction & other surgical pathologies, and confirmation of diffuse colonic dilatation with fecal loading was achieved.

![Fig. 1. Erect & supine abdominal X-rays, and dilated terminal ileum (red lines). (For interpretation of the references to colour in this figure legend, the reader is referred to the web version of this article.)](image-url)
The patient underwent a leveling decompressive, left-sided loop sigmoid colostomy with a full-thickness bowel wall and a full-thickness rectal biopsy to assess the presence of ganglion cells. Both biopsies revealed the absence of ganglion cells and hypertrophy of nerve endings, consistent with Hirschsprung’s long-segment disease (Fig. 2). However, the colostomy was not functioning well postoperatively, despite per colostomy enemas and laxatives. In addition, the patient was reporting the same colicky abdominal pain and lower abdominal distension without any marked improvement.

The consultant colorectal and general surgeons planned an exploratory laparotomy for total colectomy and end ileostomy. Exploration with a midline laparotomy incision and formal laparotomy done. Gross dilatation of the ascending colon & distal ileum identified with fecal loading reaching up to 70 cm proximal to the ileocecal valve.

The proximal ileum & jejunum were healthy, with multiple benign-looking mesenteric lymph nodes. Open total abdominal colectomy with ileum resection of 5 cm and end ileostomy was done (Figs. 3 & 4). The rectal stump was closed and left in situ if a future restorative procedure is attempted. Histopathological exam of the specimen revealed no ganglion cells in the colon, appendix, and resected ileum margin of 5 cm & associated hypertrophy of the nerve ends. The leveling full-thickness bowel wall biopsy was 50 cm proximal from the ileocecal valve and was proven aganglionic as well.

The patient postoperative recovery was uneventful. End ileostomy started to function on the second postoperative day. The patient was discharged home on day five after ileostomy education and counselling about future options for reconstruction. A regular follow-up up to twelve weeks continued, and the patient was satisfied with a fully functioning end ileostomy. He started to put in some weight and looked for his restorative procedure and bowel reconstruction.

A final diagnosis of total colonic aganglionosis with small bowel/ileum extension was confirmed. Unfortunately, the exact length of the aganglionic ileum was not reached because of our center’s unavailability of the frozen section histopathological examination services and consideration of bowel length preservation.

3. Discussion

Diagnosis of Hirschsprung’s disease (HSD) can be challenging, especially in older children and adult patients who present late, and the course of the disease is rather protracted. The total colonic disease variant is rare in adults and even rarer when ileal extension of aganglionosis is present [3,4]. Adults presenting long-standing chronic constipation usually require a broad set of tests and radiological examinations to reach a definitive diagnosis. The differential diagnosis includes obstructive pathologies, HSD, and other motility disorders.
affecting the bowel.

S. Ieiri et al. reported an association between distance from the ligament of Treitz and mortality rate. A 75 cm distance distal to the Treitz ligament and duodenoejunal junction is the predictive point for the HSD’s mortality with small bowel extension. Therefore, segment length and the risk of morbidities and mortalities were strongly related and snowballed as the segment got more proximally [5].

Definitive management entails surgical excision of the diseased segment and restoration of the bowel continuity. As reported by some centers, the Duhamel procedure is considered the gold standard with the most favorable outcomes in adult HSD patients. However, no international consensus on the optimal surgical treatment. Therefore, the management plan should be tailored according to patient condition, length of diseased segment, comorbidities, and the possibility of performing definitive reconstruction at the primary surgery [6–8].

In a recent review of literature by C. Gamez, T.O. de Boer, N. Saca, et al., they reported 36 patients of adult HSD, with a mean age of 30.8 years and male predominance of almost two thirds. The cardinal feature and complaints were chronic constipation & abdominal distension with or without intestinal obstruction. In addition, their systematic review reported a comparable outcome of the different surgical operations of Duhamel, Swenson, and Soave. Nevertheless, their review did not include the segment length or the disease extent [4,9].

Our patient had a long-standing history of chronic constipation since early childhood, which worsened over time. He has a strong family history of a similar condition with his elder brother and daughter. His brother died at 20 years of age of the same condition, and recently his daughter died of enterocolitis. His family history strongly suggests an HSD family cluster and genetic mutation, but yet to be confirmed. His clinical presentation, course of the disease, post-diversion non-functional aganglionic sigmoid colostomy, and development of severe abdominal colicky pain directed us toward the possibility of long-segment disease & total colonic aganglionosis.

This led us to plan the surgery of total colectomy and end ileostomy and possible delayed restorative procedure. The creation of an end ileostomy in patients with TCA was first described in babies as a temporary measure till future reconstruction [10]. In our case, it was planned to be an end ileostomy with the possibility of reversal and reconstruction if the patient wishes to resume the bowel continuity. The question is to resect or not at reconstruction & restorative procedure. The possibility of short bowel syndrome, technical difficulty, and increased morbidity if a long segment of the ileum and small bowel is to be resected.

Our case addressed an adult total colonic aganglionosis extending into the ileum for more than 50 cm proximal to the ileocecal valve and a possible Hirschsprung disease family cluster. The challenge will be the extent of resection and postoperative functional outcome after the restorative procedure.

4. Conclusion

In adults, total colonic aganglionosis with small bowel proximal extension (TCAS) is sporadic. It requires a high index of suspicion during the workup of patients presenting with long-standing constipation, especially if the small bowel is loaded with feces during imaging studies. Routinely, clinicians reach the diagnosis postoperatively, and HSD/TCA should be part of differentials. A non-functioning left-sided stoma and persistent symptoms should alert the treating surgeon about the possibility of TCA. The surgical approach and operation depend on the patient's clinical condition, institution experience, and surgeon's expertise.

The surgical approach should consider the perioperative hospitalization, associated enterocolitis, functional clinical outcomes, and possible mortality. No superior surgical technique was reported. However, primary reconstruction of the bowel can be achieved immediately after resection. We recommend a temporary ileostomy in such cases to work as a bridge in the management and activate the ileal brake mechanism to enhance the absorptive capability of the small bowel.

**Abbreviations**

HSD Hirschsprung’s disease
DRE digital rectal examination
CT computed tomography
TCA total colonic aganglionosis
TCAS total colonic aganglionosis with small bowel extension

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N/A.

**Ethical approval**

The hospital ethical committee obtained ethical approval for publishing this case report.

**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.

**CRediT authorship contribution statement**

**Study conception and design**: Mohamed Eltayeb Abdelrahman Naiem, Suliman Hussein Suliman. **Acquisition of data**: Mohamed Eltayeb Abdelrahman Naiem. **Analysis and interpretation of data**: Mohamed Eltayeb Abdelrahman Naiem, Suliman Hussein Suliman, Mohamed Elfatih A. Elgurashi. **Drafting of the manuscript**: Mohamed Eltayeb Abdelrahman Naiem, Suliman Hussein Suliman, Mohamed Elfatih A. Elgurashi, Nadir Ali Hilal. All authors read and approved the final manuscript to submit.

**Research registration**

N/A.

**Guarantor**

Mohamed Eltayeb Abdelrahman Naiem.

**Declaration of competing interest**

All authors declare that they have no competing interests.

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