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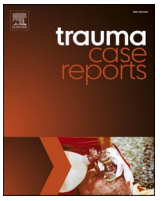

Case Report

Suspicion diagnostic of Hirschsprung's disease in an adult intraoperatively: A case report

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ABSTRACT

Background: Hirschsprung disease, a developmental disorder affecting the neuronal ganglion cells in distal colon, is the leading cause of intestinal obstruction in newborns, predominantly males, although the diagnosis can be made lately in rare cases. We describe an adult Hirschsprung disease case found intraoperatively.

Case description: He is a 20-year-old male patient with past medical history of epilepsy, psychomotor delay and recently a perineal perforating injury, admitted in the emergency room with initial diagnosis of peritonitis by perforated viscus with Frankel grad B spinal cord injury. Among his initial signs and symptoms were abdominal distension, rebound tenderness, decreased bowel sounds and diffuse pain. Laboratory tests revealed increased Hb, decreased white cells count and increased creatinine level. Initial management included: fluid resuscitation, analgesics, antibiotics and laxatives. After becoming hemodynamically unstable, the patient was introduced to the operating room where a highly dilated rectum with areas of necrosis at the base was discovered after access to the abdominal cavity. While doing the intervention, the patient experienced 3 cardiac arrests following by successful ressucitation and blood transfusion; leading to the decision to delay the definitive closure using the Bottega technique. The surgical course was unfavorable as the patient died around 4 hour post-surgery.

Conclusion: Hirschsprung disease in adult, due to its rarity and its overlapping features with many other conditions that can affect the GI system, can be misdiagnosed or discovered lately. A thorough evaluation by an appropriate specialist is essential for adequate diagnosis and management.

Background

Hirschsprung's disease is the leading cause of intestinal innervation disorder and the most common cause of intestinal obstruction in newborns [1,2]. First described by Professor Hirschsprung, it has an incidence rate ranging from 1 over 4500 to 5500 at birth and is 3 times more common in males [1–3,5]. While the diagnosis is generally made at birth, rare cases have been reported in adults [1,6]. Since the first case identified in an adult [7,8] in 1950, it is estimated that there have been approximately 550 cases observed in adults [8], accounting for 2 % of adults with chronic or refractory constipation [5,6]. The term "adult" in the context of Hirschsprung's disease refers to patients over 10 years old; it is estimated that in 94 % of cases, the diagnosis is made before the age of 5 [8]. Cases found in adults are usually due to patient ignorance, lack of knowledge, and sometimes long-term treatment of chronic constipation

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[9]. Generally, they present with fecal incontinence and obstructive symptoms, including intestinal obstruction [4,9] sometimes complicated by volvulus affecting the prognosis [1,10]. We present a case of unusual Hirschsprung's disease discovered intra-operatively in a 20-year-old patient with acute abdomen.

Case description

A 20-year-old man with psychomotor delay and a history of epilepsy was admitted to the emergency department of the State University Hospital in Haiti for abdominal distension, rigidity, generalized pain, absence of bowel movements and gas, and a history of vomiting. He had a history of falling on a sharp object that pierced his perineum 6 days prior, with the onset of symptoms 3 days later. Vital signs on admission were blood pressure 104/58 mmHg, heart rate 138 beats/min, respiratory rate 20 breaths/min, temperature 36.2 degrees Celsius, and oxygen saturation 98 % on room air. Initial physical examination revealed rebound tenderness, decreased bowel sounds, and diffuse pain. Rectal examination showed a clean anal margin, tonically competent anal sphincter, and a rectal ampulla filled with stool. He had a functional impairment of grade B according to the Fränkel classification, with a lesion level at L2/L3. The initial diagnosis on admission was peritonitis due to hollow organ perforation with a Fränkel grade B spinal cord injury. Initial treatment included ceftriaxone 2 g IV, metronidazole 500 mg IV, tramadol 100 mg, omeprazole 40 mg IV, dexamethasone 8 mg IV, tetanus serum 5000 U/ml, intra-rectal fleet enema, fluid resuscitation with Ringer lactate, and placement of a Foley catheter for diuresis monitoring. Laboratory tests showed a hemoglobin level of 16 g/dL, white blood cell count of 3250, urea level of 25 mg/dL, creatinine level of 6.9 mg/dL, and normal electrolyte levels. No imaging studies were available at the center. After anesthetic evaluation, the decision was made to proceed to the operating room, because he becomes hemodynamically unstable with a blood pressure of 90/50 mmHg, heart rate of 148, oxygen saturation of 94 %, and a urinary drainage bag containing 400 cc of highly colored urine was emptied.

In the operating room, the abdomen was accessed through a supra umbilical-pubic incision that was later extended. After resecting various anatomical planes, the abdominal cavity was opened, revealing a highly dilated rectum with areas of extensive necrosis at the base (Fig. 1).

The colon was dissected, revealing dilation of the entire colonic frame (Fig. 2) using Mattox's maneuver to explore the retroperitoneal zones 1 and 2. Three staged biopsies were performed (diseased zone, transition zone, and healthy zone) (Fig. 3).

A double-barreled sigmoid colostomy was created, followed by irrigation of collected materials (Fig. 3). Approximately 300 cc of reactive fluid was aspirated. During the procedure, the patient experienced three cardiac arrests but was successfully resuscitated with the transfusion of two units of compatible blood. Due to the patient's inability to tolerate the rest of the procedure, closure was done using the Bogota technique to delay definitive closure.

The surgery lasted about 2 h and 40 min, and the patient was transferred to the postoperative care unit with a diagnosis of Hirschsprung's disease complicated by severe acidosis and acute renal failure. The patient left the operating room in a relatively stable

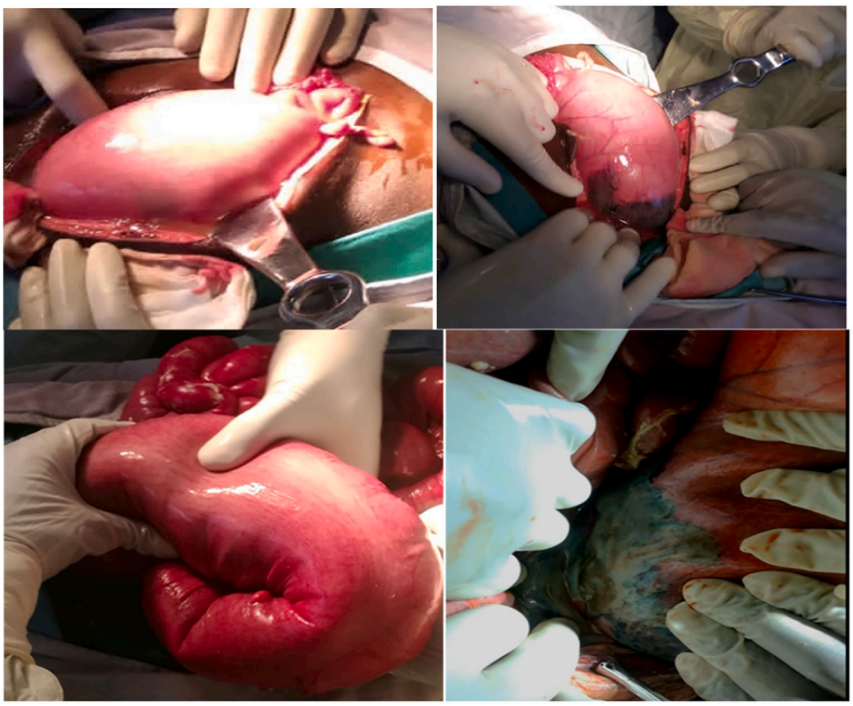

Fig. 1. Intraoperative description of the dilated rectum and extensive necrotic areas on its base.

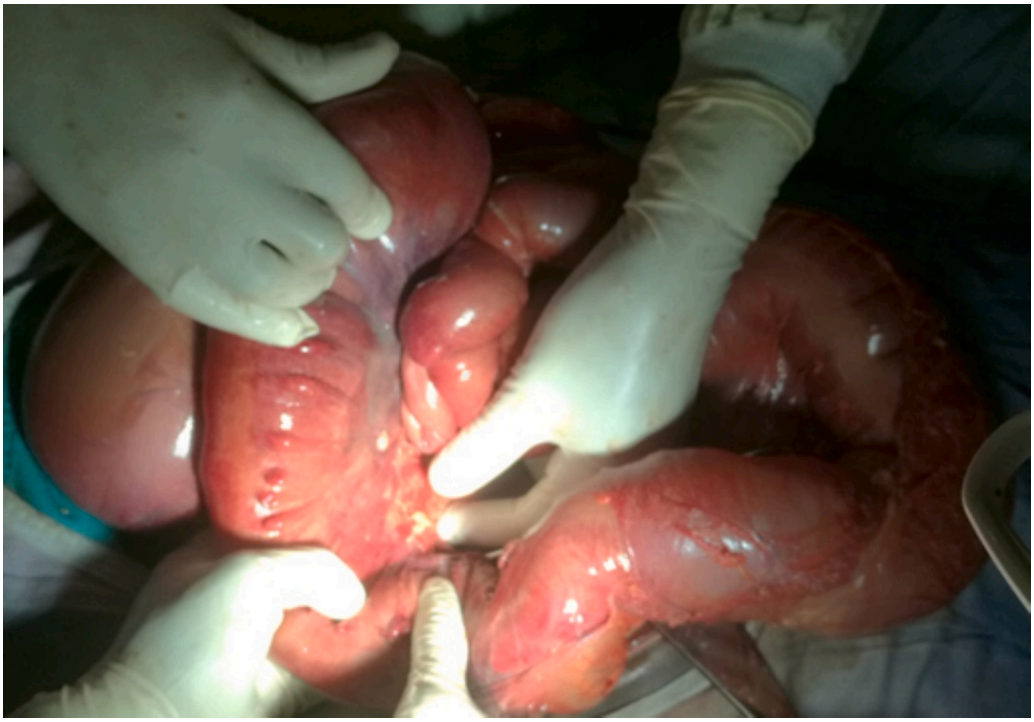

Fig. 2. Mattox maneuver.

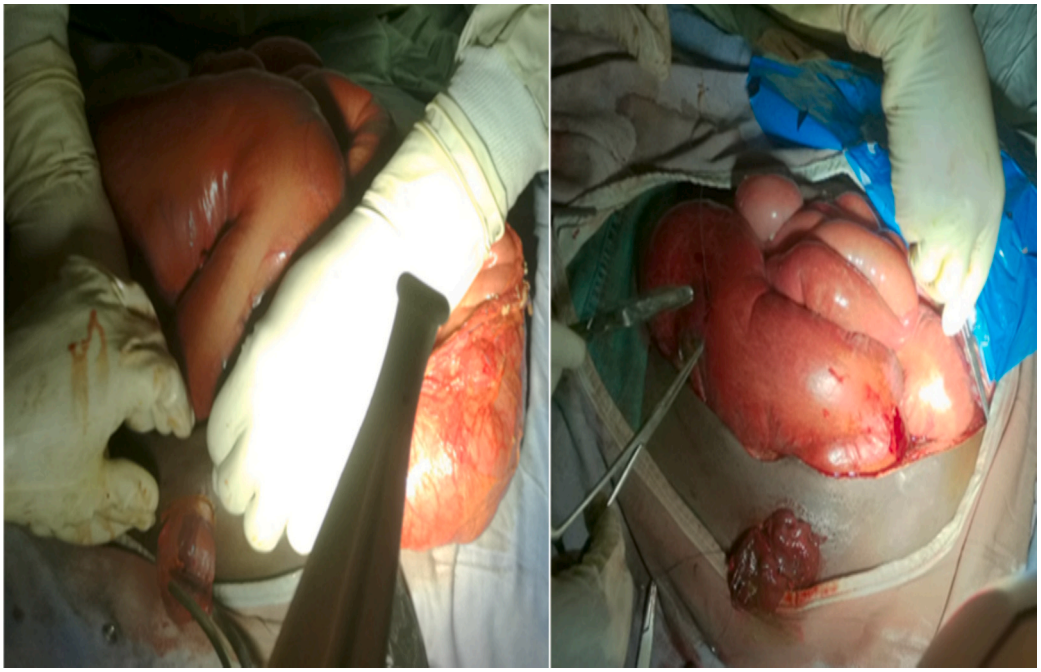

Fig. 3. Double-Barrel colostomy and Bogota technique.

condition with the following vital signs: blood pressure 130/60 mmHg, heart rate 70 beats/min, and oxygen saturation 93 % on room air. However, approximately 15 min later in the postoperative period, the patient's condition deteriorated with blood pressure dropping to 63/39 mmHg, heart rate increasing to 154 beats/min, respiratory rate of 44 breaths/min, temperature of 39 degrees Celsius, and oxygen saturation of 70 % with supplemental oxygen. About 4 h later, the patient passed away.

Discussion

Hirschsprung disease is characterized by a congenital absence of neuronal ganglion cells in the submucous membrane in the Meissner plexus and in the muscularis in the Auerbach plexus [6,20]. While typically diagnosed in the neonatal period, cases in adulthood, termed adult Hirschsprung disease, have been usually in patients older than ten years [11,15,21]. Often mistaken for chronic constipation, further investigations reveal a clinical correlation in about 2 % of adult patients with chronic refractory constipation [6]. This form of the disease may be explained by a shorter aganglionic zone and propulsive forces from the upstream colon countering distal obstruction [12,6]. Males are predominantly affected [13,17,21], as reflected in our study where we reported a case involving a 20-year-old man.

Symptoms include chronic constipation, abdominal distension, pain [19], nausea, vomiting [17], and palpable fecal masses. History may include delayed meconium passage and use of procedures like fecal decompaction with enemas or cathartics [19]. Complications can include intestinal obstruction, sigmoid volvulus [12,16], and malnourishment [17]. The literature highlights an association between Hirschsprung disease and central nervous system anomalies, such as congenital brain anomalies, mental retardation, and developmental deficits [14]. Syndromes like Down syndrome, Haddad syndrome, Goldberg-Shprintzen syndrome, and Mowat-Wilson syndrome further underscore this association [14]. Psychiatric disorders, including borderline cognitive abilities and attention-deficit disorders, can also occur [14].

Our patient was presented with abdominal distension, pain, vomiting, obstipation, and lack of flatus following a perineal perforating injury. The diagnosis of Hirschsprung disease was delayed due to emergent clinical signs consistent with peritonitis by perforated viscus associated with medullary lesion Frankel grade B. However, intraoperative findings revealed dilated rectum and colon with necrotic patches consistent with reported cases in the literature [19]. Treatment involved intravenous fluid resuscitation, decompression methods (rectal washouts) [20], and **Double-barrel transverse colostomy.**

Despite resuscitative efforts, the patient experienced three cardiac arrests and received 3 units of blood. The Bogata technique was utilized to delay definitive treatment due to hemodynamic instability. However, three specimens were obtained for diagnostic purposes. Postoperatively, the patient was diagnosed with adult Hirschsprung disease complicated by severe acidosis and acute kidney failure).

The surgical approach depends on biopsy results indicating the length of the achalasia zone and the reversibility of colonic dilation. For a short aganglionic zone, transanal myomectomy is performed; for longer zones, sigmoid rectal resection with colon-anal anastomosis and Swenson's procedure are preferred. Other techniques include the Duhamel technique and Soave's procedure. Unfortunately, the patient passed away 4 h after postoperative.

Given the rarity of the diagnosis, a thorough differential is crucial. The disease involves considering various conditions that present with similar symptoms and may affect the colon and gastrointestinal tract. These diseases include chronic constipation, inflammatory bowel disease, obstructive defecation syndrome, pelvic floor dysfunction, organic stenosis, metabolic disorders, colon disease, anorectal disorders, and innervation disorders such as intestinal neuronal dysplasia, hypoganglionosis, ganglioneuromatosis, and chronic intestinal pseudo-obstruction [2,18].

Distinguishing among these disorders can be challenging due to overlapping clinical and histopathological features but a comprehensive evaluation, including medical history, physical examination, imaging studies (e.g., contrast enema, barium studies), anorectal manometry, and rectal biopsy help differentiate them. Psychological conditions such as depression or anxiety can sometimes manifest with gastrointestinal symptoms resembling those of Hirschsprung's disease. Consulting with a gastroenterologist or colorectal surgeon is essential for accurate diagnosis and appropriate management.

Ethical approval

Our study received approval from the ethical and bioethical axis of LABMES. Subsequently, the head of the surgery department granted us access to patient recordupon request of the vice-dean of the Faculty of Medicine and Pharmacy of UEH.

Consent

Next of skin of the patient was given us verbal consent to write the case.

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Guarantor

Axler Jean Paul.

Research Registration Unique Identifying Number (UIN)

N/A

Provenance and peer review

No, our paper was not invited and no one peer reviewed it before the submission.

CRediT authorship contribution statement

Thaimye Joseph: Resources, Writing – original draft, Writing – review & editing. **Axler Jean Paul:** Conceptualization, Investigation, Methodology, Supervision, Writing – original draft, Writing – review & editing. **Abigaël Francis:** Validation, Writing – original draft, Writing – review & editing. **Olnick Joseph:** Conceptualization, Supervision, Validation, Writing – review & editing.

Declaration of competing interest

There is not any source of conflict.

Data availability

Data generated during the student is available upon reasonable request.

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