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Case Report

Hirschsprung's disease in adulthood: a case report

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ABSTRACT

Hirschsprung's disease (HD) is caused by a gut development anomaly that leads to functional constipation. In most cases, HD is diagnosed and treated during childhood. Abdominal pain, distension and chronic constipation are typical symptoms of adult presentation. An adult HD case is presented, using analysis of electronic medical records and literature review for diagnostic and management insights. A man in his early 40s, with a history of chronic constipation since infancy, presented to the emergency department with an intestinal obstruction. Imaging tests revealed pronounced colonic dilation along with faecal loading. Following favourable progress, the patient was referred for outpatient investigation. Endoscopy revealed rectal scars with biopsy consistent with HD. A laparoscopic Duhamel-Martin procedure was performed in two stages. The postoperative period was uneventful. Adult HD is overlooked and misdiagnosed. Therefore, a high level of suspicion is needed to diagnose and avoid urgent surgeries, often with the need of colostomy and its particularities. When feasible, appropriate management by elective surgery should be the gold standard.

Keywords: Chronic constipation, Adult, Hirschsprung's disease, Congenital anomalies, Gastrointestinal surgery, General surgery

INTRODUCTION

Hirschsprung's disease (HD) is the most common congenital motor gut disorder characterised by functional obstruction.[1] While it is a highly heritable disease (>80%), showing incomplete penetrance and variable expressivity, the phenotype may vary considerably.[2,3]

HD has an incidence of about 1:5000 births and occurs with an overall male: female ratio of 3:1 to 4:1.[1,4] Its pathogenesis is explained by a defect in the craniocaudal migration of neuroblasts that originate from the neural crest during the gut development, resulting in an absence of the enteric ganglia in a variable length of the intestine. This segment will fail to relax and thus leads to abnormal emptying.[4-6]

Most HD cases are diagnosed during the neonatal period. Although uncommon, HD can be identified later in adulthood. This delay in diagnosis is a result of mild presentation, usually with symptoms of abdominal distension and a long-standing history of constipation managed with laxatives and enemas.[7,8] When the dilated colon cannot compensate for the non-functional segment, rapid worsening of constipation, faecal retention and megacolon can occur, and an urgent surgical intervention may be needed.[6,9,10]

Most HD cases require surgical management. The aims of surgery include resection or bypass of the aganglionic segment and bringing the proximal innervated segment down to the anus, allowing the construction of an adequate anastomosis.[9,11] It is very important that injuries of the anal canal and internal anal sphincter, and autonomic innervation of the small pelvis are prevented, thereby reducing the risk of urinary or sexual disorders.[11-13] Because HD is usually a disease of infancy, recommendations in the literature are lacking concerning the surgical management of this condition in the adult population.

We report the case of a male patient who presented to the emergency department (ED) complaining of abdominal pain and intestinal obstruction. The initial approach was conservative and led to clinical improvement. Further evaluation was performed to exclude malignancy, the results of which were consistent with adult HD. Accordingly, a two-step elective surgical procedure was proposed to the patient and successfully performed.

CASE REPORT

An independent man in his early 40s presented at the ED complaining of crampy abdominal pain, mainly in the lower quadrants, and over the past four days had not been able to pass gas or faeces. Former similar episodes had occurred but were resolved with laxatives. He denied fever, loss of appetite and vomiting, and there were no signs of intestinal bleeding. The patient had a past medical history of inguinal hernia and lumbar hernia surgery.

An examination revealed that the abdomen: was distended; had few sounds; was tympanic to percussion; was tender; and had a palpable mass in the right lower quadrant. No guarding or peritoneal irritation signals were identified. A digital rectal exam showed a dilated rectal ampulla with touchable feces and mucosal fold hypertrophy; however, no palpable lesions or blood in the stool were noted.

A plain abdominal radiograph revealed extensive faecal retention causing colonic dilation, but no air-fluid levels or a 'stop' transition point were identified. No significant changes were evident from the laboratory tests.

After an unsuccessful first attempt of treatment with analgesia and enemas, the patient was submitted to an abdominal and pelvic computed tomography (CT). This revealed faecal content in the small intestine and confirmed the colonic dilation along with faecal loading, mainly in the sigmoid segment. This was found to be redundant, with a maximum calibre of 9 cm in the transition zone to the rectum (Figures 1 and 2). The rectum wall appeared to be thickened in a 3.5 cm extension.

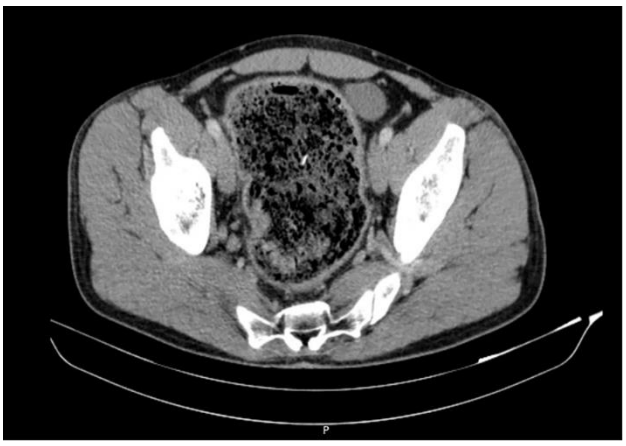

Figure 1: Abdominal and pelvic CT showing sigmoid colon dilation, along with faecal loading.

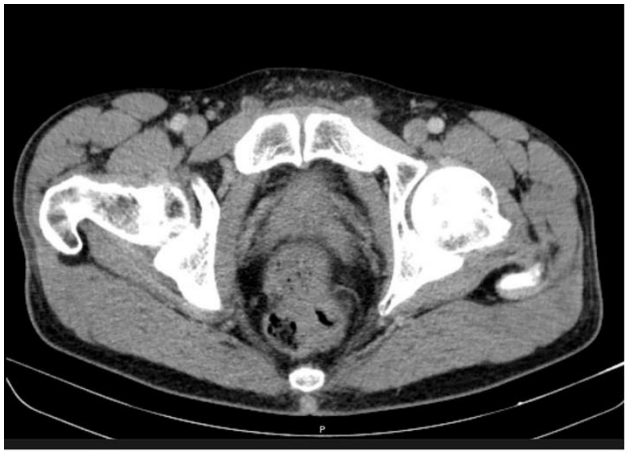

Figure 2: Abdominal and pelvic CT showing rectal wall thickening and the difference between the calibre of the rectum and the dilated sigmoid colon shown in Figure 1.

During the workup, the evacuation of faeces was achieved followed by abdominal pain control. The patient was discharged from the ED and further evaluation was conducted in an outpatient clinic.

Upon re-evaluation, the patient reported a history of long-standing constipation since infancy, sometimes requiring enemas and digitation. The patient had one to two bowel movements per week (type 2 on Bristol scale), with a maximum of two weeks without passing stools. Later in adulthood, painless rectal bleeding related to the defecatory effort, abundant flatus and tenesmus had developed. Weight loss, soiling or faecal incontinence were absent.

Total colonoscopy revealed a dilated sigmoid colon and scar tissue in the distal rectal mucosae, but lesions suggesting malignancy were not found. 'Bite-on-bite' rectal biopsies were taken proximal to the dentate line and showed an absence of ganglion cells even after immunostaining for calretinin and S100 protein. This suggested HD.

A barium enema confirmed sigmoid dilation, but the precise transition zone could not be identified. An anorectal manometry was performed, but this was also inconclusive. For that reason, and due to the concern regarding ultrashort-segment disease (USSHD) we performed other 'bite-on-bite' biopsies to limit the aganglionic segment, adding biopsies just proximal to the dentate line and 5 cm above the internal sphincter (Figure 3). The histologic report remained consistent with HD and ruled out USSHD.

The patient's clinical condition slowly worsened with sustained abdominal pain and distension, increased defecatory effort and constipation, which required the patient to regularly use medication to pass stools.

The patient was admitted to the hospital four days before surgery for nutritional supplementation and to accomplish intestinal preparation.

A laparoscopic Duhamel–Martin procedure was performed.[14-16] A redundant and distended sigmoid colon and a thickened rectum were identified intraoperatively (Figure 4). Dissection was performed in the same way as an upper rectum anterior resection with mobilisation of the descending colon. Rectal transection was obtained with a linear stapler followed by retro-rectal, trans-rectal then trans-anal lowering of the left colon (Figure 5). Then, the upstream colon was sectioned in a zone of normal calibre (well-vascularised and well-prepared, with peristalsis responding well to stimulation), and was fixed and opened in the buttocks.

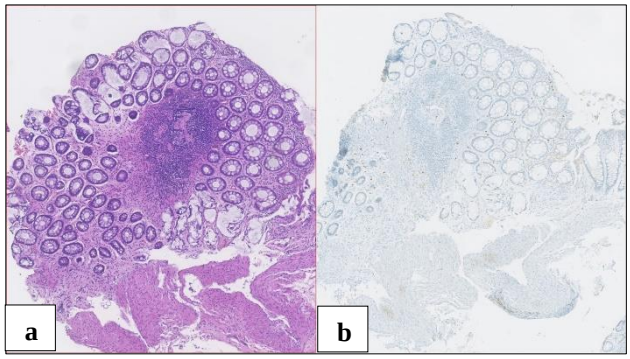

Figure 3: Histologic exam showing (a) intestinal mucosa on haematoxylin and eosin staining, and (b) immunohistochemical staining for calretinin, which only stains rare mast cells dispersed in the mucosal chorion.

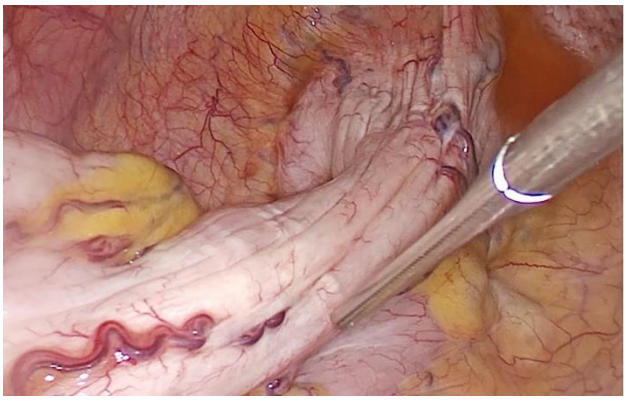

Figure 4: Intraoperative finding: wall thickening of the rectum. Rectal wall stimulation revealed an absence of peristalsis.

The evolution was good; the patient was able to maintain bowel movements through the pulled colon with anal continence (Figure 6a-d).

On the twelfth day of lowering, the second procedure was carried out: resecting the remaining exteriorised colon (Figure 6d) plus colorectal latero-lateral anastomosis construction with a linear stapler to create a reservoir. Coloanal anastomosis (posterior hemi-circumference) was established with separate resorbable sutures. A transrectal tube was placed and removed at 24 hours postoperatively.

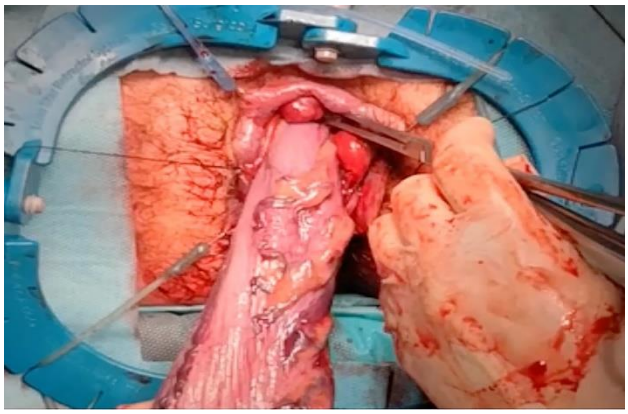

Figure 5: Trans-rectal and trans-anal lowering of the left colon.

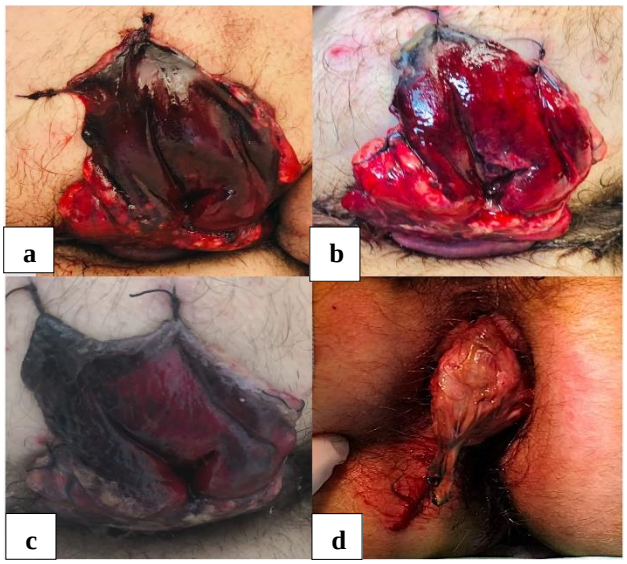

Figure 6: Photographic evolution of exteriorized remaining colon (a) postoperative (PO) day 1, (b) PO day 3, (c) PO day 5, and (d) PO day 12, right before the second procedure.

No major complications occurred during the postoperative period. The patient was discharge at postoperative (second procedure) day 5.

The anatomopathological analysis revealed a regular sigmoid colon with the presence of nerve plexus, as expected, given that in the Duhamel operation the aganglionic segment remains in place.

Evaluation at postoperative months 1, 3 and 6 revealed an uneventful recovery. Bowel movement frequency was two to three times per day with well-shaped faeces and normal continence. There has been no evidence of sexual

dysfunction. A digital rectal exam showed a permeable colorectal anastomosis.

DISCUSSION

The first case of adult HD was described in 1950 by Rosin et al.[17] That report was about a 54-year-old male patient. Since then, other authors have reported cases of adult HD, using that term whenever the patient was older than 10 years of age at the time of diagnosis.[2]

Miyamoto et al reviewed the literature over a 50-year period beginning in 1950, and reported a male predominance (133 versus 42 female cases), and an average age of 24.1 years (range 10–73).[2]

In most cases, HD is diagnosed during the neonatal period, occurring in approximately 1:5000 live births.[1,18] However, for the adult population, its incidence is unknown given that HD is frequently overlooked and misdiagnosed.[1,2,18]

HD is a heritable disease in more than 80% of cases, and can be syndromic or, as in most cases, non-syndromic.[2,3] It is known that some genetic mutations (e.g. RET proto-oncogene) and chromosomal anomalies are associated with HD.[4] Syndromic HD is associated with these chromosomal anomalies, and for non-syndromic HD the transmission can occur by autosomal dominant inheritance (usually in long-segment disease) or by autosomal recessive or multifactorial inheritance (mainly for short-segment disease).[19]

Such anomalies lead to a migration defect of the neural crest cells to the hindgut, resulting in a total absence of ganglion cells in the submucosa and muscle layer over a variable length of the intestine, and evolving into functional obstruction.[20] In addition, defects in the differentiation and destruction of ganglion cells have already been identified as pathogenic mechanisms.[4]

The aganglionic segment is mostly limited to the rectosigmoid. The short-segment disease, the most common variant, involves aganglionosis limited to the rectum or both the rectum and sigmoid colon, accounting for approximately 80% of cases.[19] When the diseased segment extends proximally beyond the sigmoid colon, it is considered long-segment disease (15–20%).[19] Total colonic aganglionosis, affecting the entire colon, accounts for 5% of cases.[19] Rarely, aganglionosis extends into the small bowel (total intestinal aganglionosis).[19] Furthermore, there is a controversial variant – the ultrashort-segment disease (USSHD) –characterised by a segment of aganglionosis extending 1–4 cm just above the dentate line.[21]

Late diagnosis of HD can be explained by milder forms of the disease (as short- or ultrashort-segment disease), which can be well-managed with symptomatic medication while the non-diseased proximal colon is able to play a compensatory role.[10,22]

Miyamoto et al reported that symptoms of longstanding refractory constipation, since birth, infancy or childhood, occurred in most cases.[2] Abdominal distention and pain were common (83–86%) and frequently associated with palpable faecal masses (50–53%) or faecal impaction (25–36%); cathartics and/or enemas were used regularly to achieve bowel movements in most cases (73–92%).

Gamez et al reviewed the literature and reported that chronic constipation was present in 100% of the 36 patients; intestinal obstruction with abdominal distention occurred in 97.2% of cases; and abdominal pain in 12 cases (33%).[23] Half of the patients had tried laxatives, home remedies or enemas.

The above-mentioned clinical features are quite similar to those reported in the present case: a male patient in his early 40s with abdominal pain and distension and chronic refractory constipation, despite the regular use of laxatives and enemas.

Abdominal and pelvic CT is helpful in excluding other causes of constipation and may show the dilated colon and the transition zone.[22] The diagnosis of HD is suggested by performing a barium enema, an anorectal manometry and a rectal biopsy.[9-11,20,22] The characteristic rectal narrowing seen in the barium enema was not detected in approximately 20% of the patients studied in the literature review by Miyamoto et al which was attributed to short- or ultrashort-segment disease.[2] Anorectal manometry can be very useful in the diagnosis mainly when it reveals lack of relaxation of the internal anal sphincter with balloon rectal distension.[5,10] Vorobyov et al reported that the recto-anal inhibitory reflex (RAIR) was absent in 51 of the 80 patients who underwent anorectal physiology (63.8%), but in the remaining 36.2%, the RAIR, although weaker, was present and cast doubt on the diagnosis.[10]

Rectal biopsy is the gold standard for the diagnosis when the absence of ganglion cells can be documented.[20] Other suggestive findings include abundant hypertrophic submucosal nerves, increased acetylcholinesterase activity and decreased or absent calretinin-immunoreactive mucosal innervation.[24] In the present case, the barium enema did not show the transition zone, and the anorectal manometry was inconclusive. The rectal biopsies performed revealed an absence of ganglion cells on haematoxylin and eosin staining, and the absence of calretinin-immunoreactive fibres. These findings were present in both biopsies allowing the conclusion that this was a short-segment HD.

The mainstay of HD treatment is surgery. Vorobyov et al observed that failure of conservative management had resulted in urgent surgical treatment in 30% of patients (27/90).[10] Indeed, patients of this type necessitate surgical intervention due to the irreversibility of their functional

obstruction.[13] In the early phases, these patients can obtain some symptom relief with a low-residue diet, laxatives, irrigations and antispasmodics; however, their clinical condition will get worse with progressive dilation of the upstream colon and the worsening of constipation.[22] Consequently, serious complications – such as acute intestinal obstruction or perforation, respiratory insufficiency due to abdominal distention or necrotising enterocolitis – may occur.[6,8,13,22]

The aims of surgery include resection or bypass of the aganglionic segment and bringing the proximal innervated segment down to the anus, allowing the construction of adequate anastomosis.[8,11] The Swenson, Duhamel, Soave and Lynn procedures are the principal surgical techniques described and studied in the treatment of this condition.[2,11] The literature data seem to favour the Duhamel procedure as the preferred choice in surgical management.[2,11] By combining the various modifications of the Duhamel procedure (by Martin, Soper, Miller and others), avoidance of problems such as stump impaction, pelvic dissection and neuromuscular injury was achieved.[25] When compared with the Swenson or Soave techniques, the Duhamel procedure seems to be superior in terms of avoiding impotence and anastomotic dehiscence, and minimising soiling.[10] In fact, this procedure appears to be associated with fewer postoperative major complications, and a higher rate of very satisfactory long-term outcomes (including continence and fewer bouts of constipation) when compared with the other surgical techniques.[2,11,25]

In the present case, a laparoscopic Martin modification of the Duhamel procedure was conducted in two stages. First, the colon extending beyond the anal margin was opened to ensure the evacuation of gases and materials during the first postoperative days, thus avoiding the creation of an upstream bypass stoma. Second, 12 days later, the colorectal and coloanal anastomosis were constructed. The patient experienced uneventful recoveries from both procedures.

No major complications occurred at the first month, third month and sixth month after surgery. The patient now has a bowel movement frequency of two to three times per day, with normal continence, and has no evidence of sexual dysfunction. Such outcomes reinforce the excellent results of the modified Duhamel procedure. A longer follow-up period will provide additional insight on this patient's quality of life.

CONCLUSION

Adult HD is uncommon, overlooked and misdiagnosed. Typical symptoms of adult presentation include abdominal pain and distention, and a long-standing history of constipation well-managed with cathartics or enemas. Rectal biopsy is the gold standard exam for the diagnosis.

The mainstay of treatment is surgery. The modified Duhamel procedure is a safe and feasible surgical option, showing superiority regarding anastomotic dehiscence, minimisation of soiling and no sexual dysfunction.

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