



Case Report

Adult Hirschprung's disease diagnosed postoperatively: A case report

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ABSTRACT

Background: Hirschprung's Disease (HD) is a congenital disease where the ganglion cells that innervate the colon fail to migrate. Most cases are diagnosed during childhood, however, in rare cases it can go unnoticed until adulthood.

Case presentation: We present a case of a 40-year-old-man who had been managing his chronic constipation with an atypical diet, until he was diagnosed with HD following an emergent abdominal surgery due to unresolved constipation. His diagnosis was delayed mainly out of fear of medical procedures. The surgery was later complicated and followed by a second and a final third and definitive surgery, suitable for the diagnosis of HD.

Conclusion: HD should be included in the differential diagnosis of constipation even in this age. Also, patient awareness should be increased to ensure better quality of life.

1. Introduction

Hirschprung's disease is caused by the absence of ganglion cells that innervate the musculature of the lower gastrointestinal tract [1]. It mostly affects children, with the majority of cases diagnosed before the first year of life [2]. Symptoms include constipation and abdominal pain, which may be tolerable enough to delay the diagnosis for many years [3]. We present a case of a 40-year-old man who was diagnosed with HD after an emergent surgery to treat obstruction.

Our work has been reported in line with the SCARE 2020 criteria [4].

2. Case Presentation

A 40-year-old man presented to Aleppo University Hospital, a major district general hospital, complaining of chronic constipation, abdominal pain and constant flatulence. A detailed history revealed that the patient had been suffering of constipation since he was a child; he became accustomed to having a single bowel movement in an entire week, sometimes even in two weeks. Out of fear of the medical community and the prospect of needing surgical treatment, he seldom consulted doctors, and when he did so, he refused to undergo endoscopy to get a biopsy as recommended by the physicians. Instead, he opted for a personalized treatment, where he only eats the foods that he can bear, as most foods caused him to have severe flatulence and abdominal pain.

Some of these episodes obliged him to visit an ER on some rare occasions. He found out that legumes and vegetables were the most harmful to him, while meats were tolerable. So, quite astonishingly, our patient coped with his chronic constipation eating a high protein and fat and low fiber diet; processed deli meats and fried chicken being his favorite meals. It's worth noting that the patient has no other medical problems, no allergies, takes no medications, and his family history revealed that his brother has mild constipation that does not interfere with his life whatsoever.

However, one of those episodes of severe abdominal pain didn't resolve as usual, which made him present to us. His complete blood count showed mild anemia (hemoglobin 11.6 g/dl, hematocrit 35.8%). The remainder of the lab tests, including liver and kidney functions and biochemistry were within normal limits, except for a slightly decreased serum albumin (3.1 g/dl) and an INR of 1.4. Abdominal ultrasound was inconclusive due to abdominal gas accumulation, which made it impossible to clearly identify any organs. An abdominal plain radiograph (Fig. 1) showed multiple air-fluid levels. He was diagnosed with intestinal subocclusion; an emergent colectomy was made by a senior general surgeon at AUH and the specimens were sent to pathology. Upon macroscopic examination, the report stated the presence of a colonic loop with a length of 130 cm, attach to it an 8-cm-long ileal loop. The colonic loop showed notable narrowing in the distal 5 cm, with the widest part being 4.5 cm. Proximal to it, there was a remarkable dilation

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Fig. 1. Abdominal XRayShowing air-fluid levels.

for 75 cm, with the widest part measuring nearly 40 cm, in addition to effaced mucosa due to the dilation. The cecum, the attached ileal part and the appendix were normal.

Microscopically, the specimen was studied in 40 slides and. There was an absence of nerve cells in the submucosa and muscularis layers of the distal 35 cm, with normal continuation proximally. Therefore, the diagnosis of Hirshprung's disease (HD) was established.

Following the pathology report, the surgeon recommended an elective resectomy to be made in order to avoid future complications, however, the patient refused and was assured by the relative improvement in his bowel movements.

Unfortunately, the patient presented a month later with the feared complication. He showed signs of peritonitis and it was quickly deduced that there was intestinal perforation secondary to intestinal obstruction. As a consequence, the patient was suffering of disseminated Intravascular Coagulation (DIC), basilar pneumonia and an overall toxic state. It was imperative to conduct a second surgery where the adhesions were released. However, due to the patient's greatly deteriorating toxic state and the unexpected severity of adhesions, saline lavage was made, followed by an ileostomy and the surgery was concluded, with plans to do a resectomy when the state improves. The patient received a thorough post-operative treatment, where he was kept on high-protein total parenteral nutrition, insuring adequate fluid and electrolyte intake.

After six months, resectomy was successfully made by the same senior surgeon. Also, an ileal pouch was made with an anal anastomosis, a protective ileostomy was kept for 2 months and eventually was closed.

In a follow-up after 6 months, the patient's overall state improved, his constipation resolved; in fact, he was complaining of fecal incontinence as he had diarrhea 7–10 times per day. He also complained of moderate abdominal pain that was responsive to analgesics. Other than that, the patient was satisfied with his improved quality of life and the functional tests showed that the patient is tolerating his surgical modifications well.

3. Discussion

Hirschsprung's disease (HD) is a disease characterized by the loss of ganglion cells in the submucosa (Meissner's plexus) and the muscularis layers (Auerbach's plexus) of the lower gastrointestinal system [1]. It is generally diagnosed in early childhood, with the vast majority of cases revealed before the first year of life. However, the diagnosis of HD can be delayed until later in childhood or even in adults. The first case of adult HD was reported in 1950 by Rosin et al. [2,5]. HD is more common in males than females, with an approximate ratio of 4:1 [5].

Symptoms include constipation, abdominal distension and pain, and excessive flatulence, which usually appear in infancy and persist until diagnosis and treatment [3]. Telborn et al. emphasized in their study that patients with HD face significant dietary challenges, and the entire family's lifestyle may be altered in order to find the most suitable options for the patients; something that was confirmed by our patient [6]. However, the psychosocial aspect cannot be neglected, as the impaired bowel activity produces sensations of being different, isolation and embarrassment, all of which exacerbate the problem and may indirectly discourage from seeking professional care [7]. It is well documented that patients could tolerate their symptoms for a long time by using conservative treatments like cathartic drugs or daily enemas [8,9]. In contrast, our patient did not use any medications or enemas. Instead, he has been following a special diet since he was five-year-old, which efficiently managed his symptoms. Interestingly, our patient didn't opt for the traditional laxative foods such as dairy products, raw vegetables, beans, legumes, and fiber-rich options [9]. On the contrary, he found out that consumption of beans and vegetables exacerbates his constipation, so he gradually took note of the foods that helped him most, which included lean deli meats and fried chicken, which are low in fiber.

Computed tomography and double-contrast barium enema can suggest the diagnosis by detecting the transition zone between the proximal dilated segment of the colon and the distal narrowed aganglionic segment. Kim et al. in their review, found that the transition zone existed in the descending or sigmoid colon. Anal manometry can reveal an increased tone of the rectum, which is beneficial in emphasizing the diagnosis, especially when the aganglionic segment is located at the anorectal junction. The final diagnosis of HD can be made based on the result of the full-thickness biopsy, which demonstrates the absence of ganglion cells [10,11]. Surgery is the definitive treatment of HD. Surgical procedures used in the treatment of HD aimed to remove the aganglionic segment of the bowel, pulling through the normal bowel and connecting it to the anus. Several types of pull-through procedures exist, including the Soave, Swenson, and Duhamel [11,12].

In our report, the patient presented with a history of lifelong constipation and frequent symptoms of obstruction. It is quite understandable that HD was not at the top of the differential diagnosis when this patient presented, primarily due to his age. It's intuitive to assume that HD severe enough to cause such pain and obstruction must have been diagnosed much earlier in the life of the patient. Therefore, the surgical decision was made to address the problem of chronic functional constipation, after which the pathologic study revealed the diagnosis of HD.

This surgical decision wouldn't have been sound if HD was strongly suspected, because it is associated with some well-known complications, of which our patient was not exempt. He did suffer of a life-threatening leakage from the anastomosis area obliging the surgeons to do another operation to conduct a resectomy and construct a J-pouch with an ileoanal anastomosis. As we've mentioned in the presentation, the patient still had some medical concerns after the second operation, the most important being fecal incontinence and soiling, which is also a well-documented and expected complication of HD surgeries. [13].

We recommend that physicians lower the threshold of suspicion to HD as old age is not always a sufficient reason to exclude the diagnosis. In such emergent cases, radical surgeries must be delayed for as long as possible until HD is definitely excluded.

However, our case did not only demonstrate the dire need of a higher

suspicion of HD, but also the need for better education of patients suffering from constipation regardless of its etiologies. Patients and their families must be educated by the physicians on how to cope with the condition, recommending certain lifestyles, food protocols, laxative medications, and possibly enemas. Our patient was not educated on laxatives and enemas, partly because of his highly hesitant stance towards visiting healthcare providers and applying their advice. Therefore, patient education must be on the priority list in every encounter with a patient suffering from chronic constipation.

4. Conclusion

HD is a disease mostly diagnosed in children but it needs to be higher in the differential diagnosis of adults presenting with chronic constipation. HD must be excluded before any radical surgeries are made. Patient education is a vital step in ensuring an adequate increase in the quality of life of such patients.

Authors' contribution

Conception and design: RA, AJ. Analysis and interpretation of Data: AA, SR, ZZ, FA. Writing of the manuscript: AA, SR, ZZ, FA. Critical Revision of the paper: RA, AJ, NM. All authors read and approved the final manuscript.

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Ethical approval

We declare we received ethical approval from Aleppo University Hospital Ethical Board.

Consent

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Declaration of competing interest

We declare that there is no conflict of interest.

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Appendix A. Supplementary data

Supplementary data to this article can be found online at <https://doi.org/10.1016/j.amsu.2022.103512>.

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