

Hirschsprung's Disease in Adulthood: Case Report and Review of the Literature

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Abstract

Case Report

Hirschsprung's disease (HD) in adults is rare and often undiagnosed or misdiagnosed. We report a case of HD in a 35-year-old woman who had a history of chronic constipation that required laxatives and enemas since early childhood. Symptoms had worsened in recent months, prompting her to seek the evaluation of a proctologist. A computed tomographic scan confirmed significant fecal loading of the colon and rectum. An anal manometry revealed lack of normal rectoanal inhibitory reflex. A rectal biopsy showed aganglionic anorectum. A diagnosis of adult HD was made and treated. Hirschsprung's disease should be considered in adults who have long-standing and refractory constipation.

Keywords: Hirschsprung disease - Adult case – Constipation – Megacolon.

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INTRODUCTION

Hirschsprung's disease (HD) is a congenital malformation of the distal end of the gastrointestinal tract characterized by an absence of neuronal ganglion cells of the Meissner (submucosal) and Auerbach (muscular) nerve plexuses over a segment of variable length [1–4]. It is a rectosigmoid lesion in 80% of cases [4]. This rare disease (1/5000 live births) results to a permanent spasm of the aganglionic segment and leads to progressive dilation of the upstream colon [1, 2, 4]. It is a condition traditionally diagnosed and treated in infancy, but diagnosis of HD in adulthood is rare [2, 3]. Diagnosis of HD is delayed in some settings by ignorance of the patient, lack of knowledge and habitual long-term treatment of chronic constipation [2, 3, 5]. Stubborn constipation is the main circumstance of discovery, more rarely an acute intestinal obstruction [2, 3, 5, 6]. Numerous surgical procedures have been described, the best known of which are Swenson, Duhamel, Soave and Lynn [2,7-10]. The goal of surgical treatment is to resect or exclude the aganglionic segment and to lower the healthy colon, normally innervated at the anus while preserving the sphincter function [1,2]. Modifications of these techniques have been proposed in a minimally invasive approach [2, 3, 11, 12]. We report a case of adult Hirschsprung's disease treated by the rectosigmoid resection with a transanal colonic pullthrough procedure followed by a delayed coloanal anastomosis.

CASE REPORT

A 35-year-old woman with an unremarkable past medical history. She had previously been hospitalized and evaluated multiple times due to clinical symptoms characterized by chronic constipation since infancy that did not respond to treatments with laxatives or enemas. Symptoms had worsened in recent months, She was admitted to the gastrointestinal medicine service for evaluation and treatment.

Physical examination revealed a significant abdominal distension and tympany to palpation, There was evidence of chronic distension of the abdominal skin. Without any other relevant alterations. No abnormalities resulted from the rectal examination. General laboratory tests were performed (complete blood count, blood chemistry, and TSH) and the results were within the normal ranges. Bowel rest, nasogastric tube decompression, and daily enemas were effective for several days. Additional studies were then performed. Flexible sigmoidoscopy revealed a markedly distended colon. Plain films of the abdomen showed the characteristic shadow of a mass of feces occupying especially the rectum and the sigmoid colon which are very distended, but there were no air-fluid levels in the intestine (Figure 1). Severe abdominal distention and tenderness at this presentation prompted diagnostic testing with a contrast enhanced computed tomography (CT) scan. The distal colon and recto-

sigmoid were markedly dilated with impacted stool and there was no obstructive lesion identified. (Figures 2a-2b). The anorectal manometry study revealed an anal canal with normal length, baseline and phasic canal pressures within normal values, adequate rectal sensitivity and capacity, the Valsalva maneuver with no escape, inhibitory rectoanal reflex absent up to the maximum tolerable rectal capacity (100 cc), and positive balloon expulsion maneuver. The results were consistent with Hirschsprung's disease (figure 3). Full-thickness rectal biopsies demonstrated a paucity of ganglion cells, suggestive of HD.

She underwent a total proctocolectomy and ileal pouch anal anastomosis with diverting loop ileostomy. The lumen of the entire colon was packed with firm solid fecal material. The colon was massively dilated, worse in the distal portion. No ganglion cells were identified in the submucosa or between the muscular layers in the distal rectum. The rest of the colon and small bowel showed normal distribution of the ganglion cells.

After the operation she reported dramatic improvement in her bowel function and gained weight rapidly. Her diverting ileostomy was taken down and she reports markedly improved bowel function and quality of life.



Fig-1: Abdominal X-ray showing distended rectum and sigmoid colon

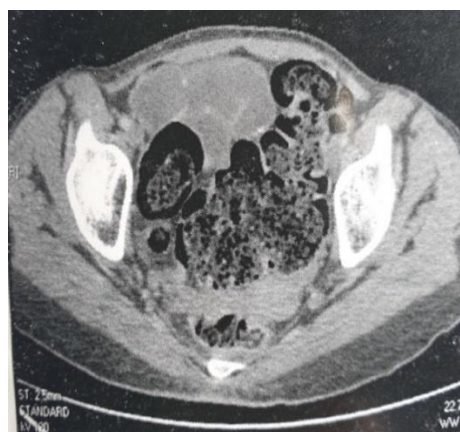


Fig-2a: Abdominopelvic computed tomography (axial plane) showing fecal impaction with distended sigmoid colon



Fig-2b: Abdominopelvic computed tomography (coronal plane) showing fecal impaction with distended sigmoid colon

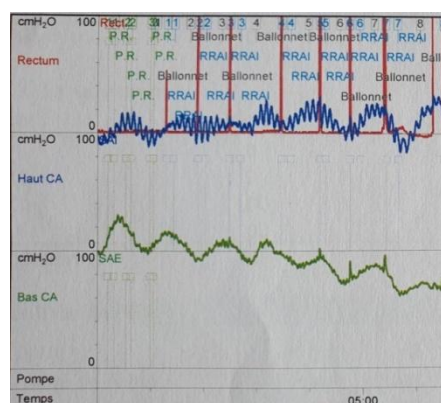


Fig-3: Conventional anorectal manometry trace showing the absence of the inhibitory rectoanal reflex with the maximum tolerable rectal capacity

DISCUSSION

The first recorded observation of HD is credited to Frederick Ruysch [13], who published an autopsy report be more useful for better anatomic delineation. Anorectal manometry typically demonstrates no internal anal sphincter relaxation in response to rectal distension. The diagnosis is confirmed by rectal biopsy. A biopsy from the narrowed segment shows absence of ganglion cells, hyperplasia, and hypertrophy of nerve fibers, and an increased level of the enzyme acetylcholinesterase.

While a vast majority of patients with Hirschsprung's disease are diagnosed in the neonatal period, Hirschsprung himself acknowledged this was a disease of all ages [14,23], whereas others have defined adult HD as cases in which the diagnosis was made after age 18 or 19 years [16]. Although case reviews in the English literature based on different diagnostic criteria have given different numbers of total cases, the clinical presentation, radiological features, and pathologic findings have been similar among the various reports. The first well documented case of adult HD was described in 1950 by Rosin *et al.* in a 54-year-old physician with a short aganglionic segment [23]. Thereafter, occasional case reports appeared in the

literature [14-17, 24-27]. Nearly 300 cases with at least some features of adult or adolescent HD have been documented, some of which were diagnosed by rectal biopsy or resection.

Fairgrieve documented HD in 7 men whose ages varied from 17 to 34 years. All of the patients had short-segment aganglionosis [14]. Two of their patients had megarectum with no demonstrable stenotic segment on the barium enema films. Anuras *et al.* in 1984 [16] reported 4 cases of adult patients with HD in whom the diagnosis was confirmed by rectal biopsy. Three of them showed pancolonic dilatation similar to our patient, but only 2 of the cases had rectal narrowing.

Most recently, Miyamoto *et al.* [17] reported a 23-year-old man who had a history of chronic constipation that required daily enemas since early infancy. The patient had remained in good health until he experienced severe intestinal obstruction for which a subemergency colostomy was performed.

The typical adult patient with HD has a history of longstanding constipation since infancy or early childhood; the male to female ratio is approximately 4:1. Patient age ranges from 10 to 73 years, and the average age is 24.1 years. Half of the patients are younger than 30 years [14-17, 23-26]. Other symptoms include abdominal discomfort, distension, and abdominal pain. Physical examination frequently reveals palpable fecal masses. The patients tend to use cathartics, suppositories, and enemas chronically to achieve bowel movements. Fecal incontinence is not a feature of the adult patient in contrast to infants. Rectal narrowing on barium enema is seen in three quarters of the patients. However, in about 20% of the patients with adult HD, a dilated colon without characteristic rectal narrowing, as seen in our patient, is demonstrated. This finding may be due to a short, or more commonly, an ultrashort diseased segment. The exact incidence of adult HD is unknown because those cases are frequently misdiagnosed or undiagnosed.

The diagnosis of HD in the adult can be much more difficult than the diagnosis in early infancy. This is due to the rarity of the disease in adults and the higher incidence of short or ultrashort segment aganglionosis with relative mild symptoms during the early stage of the disease. As in the newborns, diagnosis is based on the combination of anorectal physiology studies (sensitivity 91%, specificity 94%) and the characteristic finding is absence of the inhibitory rectoanal reflex. Baritaded imaging studies (sensitivity 70%, specificity 83%) can identify the so-called «transition zone» and inversion of the rectosigmoid index. It should be emphasized that the aganglionic segment is strictured. Full-thickness biopsies of the wall of the rectum are considered the gold standard (100% sensitivity and specificity), in which the absence of ganglion cells in the submucosa

and the myenteric plexus is pathognomonic [27]. Rectal biopsy should be performed only after more common causes of constipation and megacolon have been ruled out. Constipation and acquired megacolon in adults may be due to neoplasm, volvulus, stricture, slow colonic motility, Chagas disease, anatomical or functional outlet obstruction, or idiopathic (non-Hirschsprung's) megacolon. Causes from external factors include dietary factors, medications, psychologic factors, and systemic diseases. Whenever there is a reasonable doubt, manometric studies and biopsies are warranted.

The treatment of choice is surgical and aganglionic segment resection and anastomosis of the healthy colonic segment to the anal canal can be performed laparoscopically [28]. In adults, the treatment of choice is the Duhamel operation, which results in reduced fecal impaction after the procedure [30]. There are few reports on this disease in stages after infancy in the national and international literature [28-31]. It is important to consider the presence of this rare pathologic process in young adult patients with treatment refractory chronic constipation, in whom other more common diseases have been ruled out through easily accessed studies, such as imaging or anorectal physiology studies.

CONCLUSION

We report a case of HD in an adult and emphasize the typical clinical presentation and massive dilatation of the entire colon without rectal narrowing. The diagnosis may be easily overlooked or misdiagnosed in adult patients with chronic constipation, particularly in the rare individual with ultrashort HD such as our patient. Therefore, all patients who have severe chronic constipation since birth or childhood need a thorough evaluation to rule out HD. The diagnosis can be suspected by barium enema and manometry, and confirmed by rectal biopsy. Making the diagnosis of HD is extremely important because surgical management is effective with satisfactory long-term functional results and significantly improved quality of life [17,32].

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