Adult Pseudo-Hirschsprung’s Disease: An Unusual Cause of Chronic Constipation In An Adult.

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Abstract

Constipation is a common reason for medical consultation, and it arises by a wide range of causes. Hirschsprung’s disease and Pseudo-Hirschsprung’s disease, are unusual causes of constipation for adults. They always require surgical treatment. The following is a report of a case of hypoganglionosis of an adult without bowel movement for more than 4 months.

KEYWORDS: Chronic Constipation, Hipoganglionosis, Hirschsprung’s disease
INTRODUCTION
Constipation is a common reason for medical consultation, and it arises by a wide variety of causes. Hirschsprung’s disease and Pseudo-Hirschsprung’s, are unusual causes of constipation for adults. They always require surgical treatment. The following is a report of a case of hypoganglionosis of the colon in an adult without bowel movement for more than 4 months.

CASE REPORT
A 20 year old male suffered constipation from birth. The use of suppositories and enemas were the common solution to constipation during childhood until discomfort and loss of efficacy lead to cessation of use. When the heavy stool load was incapacitating, he usually attended a medical unit for a digital extraction of feces.

He arrived to a specialized center for treatment 127 days after his last visit to the medical unit as he had not evacuated stool during this time. Clinical evaluation showed no respiratory compromise, abdominal rigidity without peritoneal irritation, partially open anus with decreased sphincter tone, and hard fecal material in the rectal vault. Due to the high risk of complications he was hospitalized. Laboratory and thyroid function tests demonstrated no alteration. Anti-Chagas antibodies were also negative. Treatment with IV fluids, prokinetic medications, laxatives and enemas was unsuccessful.

Large amounts of fecal material (11 Kg, 8 Kg, and 7 Kg) were extracted under anesthesia in the operating room, on three occasions in order to disimpact the colon. Anorectal manometry established Hirschsprung’s disease diagnosis by absence of the recto-anal inhibitor reflex. Upper and lower gastrointestinal endoscopy were normal. Barium enema showed great dilatation of colon and rectum but a narrow anus, suggesting ultra short Hirschsprung’s disease (Figure 1).

Trans-anal myectomy surgery was performed as treatment. Histology and Immunohistochemistry revealed a decrease number of nodes (Figure 2) confirming the diagnosis of Pseudo-Hirschsprung’s disease. After 2 years of follow-up, the patient has daily evacuations with the help of a stool softener and shows no symptoms of recurrence.

DISCUSSION
In this special case of chronic constipation, both the diagnosis and the disease etiology went unsuspected for 20 years, delaying surgical treatment. The delay in diagnosis might be due to the fact that hypoganglionosis (Pseudo-Hirschsprung’s disease) represents 5% of the neural malformation of intestine (1) and it affects only one of every 5000 live births (2).

Symptoms of this disease, and those affecting an ultra-short segment of rectum, vary from complete intestinal obstruction to severe constipation, which can be refractory and children who suffer from them can reach adulthood undiagnosed. In some cases, the diagnosis is made when a patient presents with an abdominal complication that requires...
surgery and histological findings or autopsy confirm the condition (2, 3, 4). Our literature review could not find reports of patients who have not evacuated stool for such a long period of time without complications.

The first challenge faced in this patient’s treatment was to keep the colon free of stool, so regular bowel preparation and enemas were initiated. The result was not as expected, forcing us to carry out a digital extraction of the stool under sedation in the operating room, three different times. Once the colon was free of fecal material, anorectal manometry was performed which proved the absence of a recto-anal inhibitory reflex and the diagnosis of Hirschsprung’s disease was established (2).

Colon masses were not found on colonoscopy or barium enema, and the latter revealed a great dilation of the colon with an abrupt caliber decrease reaching the anus. These findings confirmed the previous diagnosis of Hirschsprung’s disease, ultra-short segment variant (2). Nevertheless, a histological test was required to establish a definitive diagnosis.

Manometry and imaging diagnosis lead to the question of which surgical approach would be best, trans-anal or trans-abdominal? Abdominal approaches present increased morbidity compared to the trans-anal approach. On the other hand, the gold standard diagnosis for this condition is a muscle biopsy demonstrating the absence of ganglia. Thus, we decided to perform a biopsy as our first step for histological and immunohistochemistry diagnosis, which also provided a solution to this disease (2, 5).

REFERENCES


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Figure 1. Barium enema which suggest ultra-short Hirschsprung’s disease
Figure 2. Immunohistochemistry reveals just one ganglion.