

Case report: sigmoid volvulus in a young adult, a manifestation of Hirschsprung disease

Reporte de caso: vólvulo de sigmoides en un adulto joven, una manifestación de enfermedad de Hirschsprung

Claudio D. Rojas-Gutiérrez^{1*}, Jorge S. Haro-Cruz¹, Diego F. Cabrera-Eraso¹, Víctor M. Torres-García², Juan C. Salas-Álvarez², and Javier O. Valencia-Jiménez³

¹Gastrosurgery Department, Specialist Hospital "Dr. Bernardo Sepúlveda", of the XXI Century National Medical Center, Mexican Institute of Social Security (IMSS); ²Undergraduate Department, National Autonomous University of Mexico; ³Surgery Department, General Hospital of Zone 1-A "Dr. Rodolfo Antonio de Mucha Macías", IMSS. Ciudad de México, México

Abstract

Background: Hirschsprung's disease is characterized by the absence of enteric ganglia in the distal digestive tract; being usually diagnosed in neonates, it rarely debuts during adulthood, where it presents as chronic constipation refractory to treatment. **Case Report:** 24-year-old female patient with Hirschsprung's disease who arrives to the emergency room with a sigmoid volvulus requiring urgent surgical therapy. **Conclusions:** Hirschsprung's disease doesn't frequently debut in adulthood, presenting a diagnostic and therapeutic challenge for the general surgeon, especially when dealing with its complications.

Keywords: Young adult. Hirschsprung disease. Intestinal volvulus. Intestinal obstruction.

Resumen

Antecedentes: La enfermedad de Hirschsprung se caracteriza por un defecto de las células nerviosas en el intestino distal. Suele diagnosticarse durante los primeros años de vida, rara vez debutando en adultos, en quienes se manifiesta como una constipación crónica refractaria al tratamiento. **Caso Clínico:** Mujer de 24 años con enfermedad de Hirschsprung no diagnosticada, quien acude a urgencias por un cuadro de vólvulo de sigmoides que requiere intervención quirúrgica. **Conclusiones:** La enfermedad de Hirschsprung rara vez debuta en la edad adulta y generalmente se diagnostica secundaria a sus complicaciones, por lo que supone un reto diagnóstico y terapéutico para el cirujano general.

Palabras clave: Adulto joven. Enfermedad de Hirschsprung. Vólvulo intestinal. Obstrucción intestinal.

*Correspondence:

Claudio E. Rojas-Gutiérrez
E-mail: claudioprojas@hotmail.com

Date of reception: 09-07-2021

Date of acceptance: 05-08-2021

DOI: 10.24875/CIRUE.M21000494

Cir Cir (Eng). 2022;90(6):828-833

Contents available at PubMed

www.cirugiaycirujanos.com

2444-0507/© 2021 Academia Mexicana de Cirugía. Published by Permanyer. This is an open access article under the terms of the CC BY-NC-ND license (<http://creativecommons.org/licenses/by-nc-nd/4.0/>).

Introduction

The Hirschsprung disease or aganglionic megacolon is characterized by the absence of the enteric ganglions in the submucosal and myenteric plexus. It tends to manifest during the first years of life, with an incidence of approximately 1 out of every 5,000 live births, appearing in the neonatal period as a delayed state in the passing of meconium, followed by abdominal distention, biliary vomiting, or both^{1,2}. Its occurrence in adulthood is extremely rare, and only approximately 500 cases have been reported in the medical literature, so its real incidence is unknown, but it is estimated that it could be close to 2% of the total of the cases of chronic constipation in adults³.

These patients have a more larvate form of the disease due to the hypertrophy of the proximal segment, which compensates for the functional secondary obstruction to the lack of distal innervation in the aganglionic segment^{4,5}. In adults, the disease tends to manifest as a chronic refractory constipated state in the traditional way. The verifying diagnosis can be carried out in an outpatient visit by means of anorectal manometry, barium enemas, or full thickness biopsies of the damaged segment⁶. A small proportion of diagnosed cases in adulthood is the result of incidental secondary findings to the complications that this condition may cause, making this disease a diagnostic and therapeutic challenge for the general surgeon^{3,4}.

We present the case of a 24-year-old patient with Hirschsprung's disease without prior diagnosis who went to the emergency room of a second-level hospital during the COVID-19 pandemic with an intestinal obstruction condition secondary to a sigmoid volvulus requiring urgent surgical therapy with the execution of a total colectomy and a terminal ileostomy. This case reflects the clinical larvate pattern of this disease, its atypical appearance and the need for, high clinical suspicion in young adults that manifest a colonic volvulus in the emergency room.

Clinical case

A 24-year-old Hispanic patient is admitted in the emergency room, undergraduate student, without chronic-degenerative pathological history, hospitalizations or previous surgical history, who reports recurring bouts of abdominal pain, constipation and distension, of approximately 5 years of development, which have increased in intensity and frequency in the last 6

months, being significantly exacerbated in the last 48 hours, also accompanied by absence of stool and inability to pass gas. She mentions regular use of nonsteroidal anti-inflammatories and antispasmodics, without having to use laxatives or enemas, with spontaneous remission of the previous states.

In the physical examination we found an ectomorphic patient with appearance of being in pain, conscious, neurologically intact, with an attack to the general state and a fever, tachycardic in the heart auscultation. The pulmonary fields appear to be properly ventilated. In the abdominal examination a significant distention can be observed, predominantly in the upper left quadrant, where we can appreciate a palpable mass with tympanism to thoracic percussion, mobile, not attached to the deep layers, as well as data of peritoneal irritation focused on that area. The rectal digital examination shows a normal sphincter tone with adequate puborectal muscle descent, without palpable masses, hematic rests or stool.

Laboratory tests are made, including a complete blood count, kidney, and liver function tests, serum electrolytes, clotting times, and arterial blood gases, reporting leukocytosis with neutrophilia ($23 \times 10^3/\mu\text{l}$, 90% neutrophils), as well as data of metabolic acidosis, and incipient acute renal injury. An abdomen radiography in bipedestation upon her admission shows a significant colon dilatation up to 20 cm in its maximum diameter with the image in the "coffee bean" (Fig. 2). An abdomen and pelvis computerized tomography is performed and it verifies a significant dilatation of the transverse colon, descending, and sigmoid with mesentery alterations (Fig. 2).

Given the lack of the availability of an endoscopy service to try the conservative management, it is decided to carry out an emergency laparotomy under general anesthesia, with the patient in supine position, finding a panacolonial dilatation, from the cecum to the sigmoid, with volvulus at the level of the mesosigmoids, so a total colectomy with distal closure in the Hartmann's pouch in two planes using absorbable suture and terminal ileostomy with Brooke technique in the right flank. The surgical sample is sent for its histopathological study (Fig. 3). The procedure was carried out by a general surgeon assigned to the service and two resident physicians of third and second year. The patient presents an adequate postsurgical evolution, remaining under supervision for 72 hours, with beginning of the enteral route in the first 24 hours and being released without complications. Consequently, she is sent to the colon



Figure 1. Simple abdomen radiography in bipedestation, of anteroposterior view, in which it can be observed the typical image in "coffee bean", with significant dilatation of the involved intestine, as well as an absence of gas in rectal ampulla.

and rectum surgery office for a follow-up and the following protocol of restitution of the gastrointestinal tract, where at the 3-month follow-up no complications are reported, with adequate wound healing and stable waste by means of the ileostomy.

The histopathological report of the sample confirms the presence of an enteric consistent neuropathy with adult aganglionosis, as well as enterocolitis with mixed inflammatory infiltrate, lymphoid follicular hyperplasia, and areas of epithelial atrophy with predominance in the sigmoid. The Masson's trichrome staining evidences fibrosis in the submucosa, perivascular, and in the Auerbach's plexus, with negative immunohistochemistry for calretinin, confirming the diagnosis of Hirschprung's disease (Fig. 4).



Figure 2. Simple tomography of the abdomen, axial section at the level of the umbilical scar, documenting the place of the torsion of the vascular axis of the mesenteric vessels, also known as the "swirl sign" (arrow).

Discussion

The strengths of the case presented, despite being an infrequent pathology and not having all the diagnostic-therapeutic tools, lies in that an etiological diagnosis was properly achieved and an adequate emergency surgical resolution was provided with a favorable prognosis for the life and function of the patient. Similarly, the weakness of our case lies in those deficiencies, since, due to the manner of presentation, it was impossible to carry out the diagnostic approach proposed by the literature, in order to try a traditional management, escalating itself according to the therapeutic answer of the patient.

Hirschprung's disease is a diagnosed pathology in the neonatal period or during childhood up to 95% of cases, manifesting as a delay in meconium passing or persistent states of constipation, which becomes a key symptom in the adult form of the disease⁷. Despite the fact that its physiopathology has not been fully discerned, the main defect is aganglionosis of the plexus submucosus (Meissner) and plexus myentericus (Auerbach), which may affect any segment of the digestive tract, but it shows a predominance in the distal segment thereof, generating an inability to relax the damaged segment. In the population whose diagnosis remains elusive until adulthood there is a tendency to have great tolerance to the obstruction conditions, as there are two concomitant conditions: short segments of aganglionosis (< 10 cm) and an adequate driving force of the unaffected segments³. This condition may go unnoticed with the use of

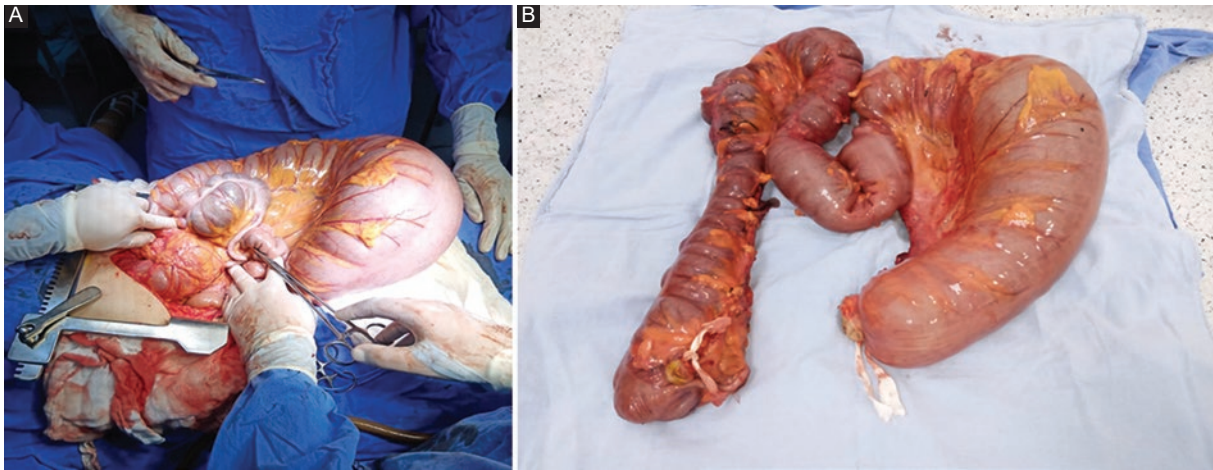


Figure 3. Transoperative findings. **A:** large intestine with a significant dilatation from the cecum to the sigmoid, with predominantly distal illness and **B:** large intestine with significant noticeable dilatation in the image of the following piece to the colectomy. The length of the piece becomes suggestive of a megadolichocolon, a variant of the development characterized by the redundancy of the large intestine.

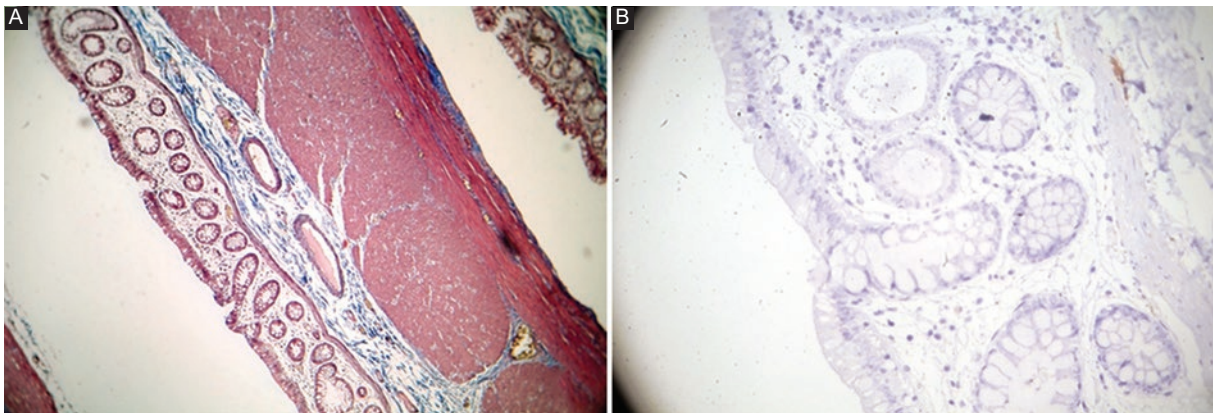


Figure 4. Colon histological sections **A:** brightfield photomicrography in 10x dyed with Masson's trichrome, in which it can be observed a clear division between the mucosa with abundance of glands and the cylindrical simple epithelium accompanied by some calyceal cells, submucosa, muscle layer and serosa. It can also be observed areas of fibrosis in the submucosa, perivascular and Auerbach's plexus. **B:** brightfield photomicrography in 40x with immunohistochemistry for calretinin, showing a ganglionic cell staining practically non-existent in the Auerbach's plexus.

symptomatic treatments for chronic constipation, delaying its detection and making it a challenge, both diagnostic and therapeutic, for the general surgeon, especially outside specialized centers or third-level care^{6,8}.

This disease is classified according the extension of the aganglionosis. The internal anal sphincter is usually defined as the lower limit of the aganglionosis, while the upper limit without aganglionosis is used to define the spreading of the aganglionosis as an ultrashort segment, short-segment, long-segment, and total colonic aganglionosis (Table 1). The most common type is the short-segment type, present in approximately 80% of

cases and affecting from the rectum to the distal portion of the sigmoid colon. The next most commonly found type is the long-segment type, affecting 15-20% of the patients with Hirschprung's disease, while the total colonic aganglionosis is the most uncommon type of manifestation, which occurs in less than 5% of cases⁹⁻¹¹.

The diagnosis of this disease is usually made with the combination of functional studies; nevertheless, the histopathological examination of full-thickness biopsy is still the preferred method. In this context, the suggestive findings include the absence of ganglion cells, the presence of hypertrophic nerve fibers, the increase of the acetylcholinesterase activity, and the

Table 1. Classification of the Hirschprung disease according to the anatomical segments damaged by the aganglionosis

Type	Damaged segment
Ultra-short segment	Anal canal above the pectinate line
Short segment	Distal position of the sigmoid and rectal colon
Long segment	Rectum, sigmoid colon and colon to the splenic flexure
Total colonic aganglionosis	Aganglionosis that spreads proximal to the splenic flexure

absence of fibers positive to calretinin in the lamina propria. The hypertrophia of nerve fibers may not appear in breastfed babies of less than 8 weeks, so its detection in adults is not usually a problem. On the other hand, in case of ultra-short segment disease it's important to clarify that a higher probability of false secondary negatives to the site of the sample collection has been reported¹². In our case, that diagnosis was made after the findings during the transoperative and the sample shipment for its final study.

In literature, previously reported cases tend to be patients in an outpatient context, in examination for chronic constipation states. There are few articles and reports that recount the complications of Hirschprung's disease not previously detected^{4,7,12}. While some authors suggest the diagnostic certainty prior to consider a definitive management with some of the described procedures (Duhamel, Swenson, Soave), the rarity of this type of manifestations has not allowed to standardize an ideal management, so the therapeutical plan must be individualized according to the clinical setting^{6,13}.

The current surgical approach depends in turn on the length of the damaged segment, the degree of reversibility of the colonic dilatation, the length of the achalasia area, the nutritional status of the patient and as with the rest of surgical approaches, experience and the skill of the surgeon in charge. In the ultra-short disease, one of the main options is the transanal myectomy, with the objective of eliminating the spasm of the aganglionic zone. The Swenson procedure consists in the removal of the aganglionic rectum and the distal colon irrevocably stretched, with coloanal anastomosis. The Duhamel technique, described in an era in which the morbidity of a rectal dissection surpassed the one involved in this procedure, involves the descent of the healthy colon behind the damaged rectum, which is left in place, creating a short-circuit effect.

Considering the advances in current surgery, this technique has become obsolete, but it remains as part of the historical context of the surgical management of Hirschsprung's. The Soave procedure, in the same way, aims to preserve the distal rectum, due to the morbidity related to the aforementioned procedure in the historical moment of the development of this technique, only resecting the colon and the upper part of the damaged rectum, and then making lowering the healthy colon through the remnant rectum once its mucosa has been eliminated^{3,14}.

Another factor that must be taken into account in analyzing the pathology sample is the suggestion of a concomitant condition of the megadolichocolon, which has been described as a variant in the development of the large intestine, characterized by its redundancy, and described for the first time in 1820. This profuse prolongation of the intestine is usually followed by alterations in the motility of the ascending and descending segments. Epidemiologically, it is not usually associated with higher frequency in a specific gender or age, but if it is symptomatic, the symptoms tend to manifest in early stages of life¹⁵.

Conclusions

Hirschprung's disease or aganglionic megacolon in adults is an uncommon condition whose manifestation can be a challenge for the general surgeon, requiring a high degree of diagnostic suspicion. Therefore, it is relevant to use an adequate approach in young adults with a masked state of chronic constipation of difficult management. Further evidence is required to issue recommendations about the emergency surgical management that improves the prognosis of these patients.

Acknowledgements

The authors acknowledge the pathology department of the General Hospital of Zone 1-A for their assistance and cooperation in making the report of this case possible and contributing to the scientific community of their country, despite the limitations that have appeared due to the COVID-19 pandemic.

Funding

The authors declare that they have not received any type of funding for this article.

Conflicts of interest

The authors state they have no conflicts of interest.

Ethical disclosures

Protection of people and animals. The authors declare that no experiments were carried out on humans or animals for this research.

Data confidentiality. The authors declare that they have followed their center's protocols on the publication of patient data.

Right to privacy and informed consent. The authors have obtained the informed consent of the patients and/or subjects referred to in the article. This document is in the possession of the corresponding author.

References

1. Heuckeroth RO. Hirschsprung disease — integrating basic science and clinical medicine to improve outcomes. *Nat Rev Gastroenterol Hepatol*. 2018;15:152-67.
2. Drissi F, Meurette G, Baayen C, Wyart V, Cretolle C, Guinot A, et al. Long-term outcome of Hirschsprung disease: impact on quality of life and social condition at adult age. *Dis Colon Rectum*. 2019;62:727-32.
3. Lupón E, Labbe F, Nini E, Sondji S. Hirschsprung disease in an adult with intestinal malrotation and volvulus: an exceptional association. *J Med Case Rep*. 2019;13:124.
4. López Ruiz JA, Tallón Aguilar L, Sánchez Moreno L, López Pérez J, Pareja Ciuró F, Oliva Mompeán F, et al. Hirschsprung disease with debut in adult age as acute intestinal obstruction: case report. *Rev Esp Enferm Dig*. 2016;108:742-6.
5. Zeng M, Amodio J, Schwarz S, Garrow E, Xu J, Rabinowitz SS. Hirschsprung disease presenting as sigmoid volvulus: a case report and review of the literature. *J Pediatr Surg*. 2013;48:243-6.
6. Shitta AH, Ugwu BT, Peter SD, Ozoilo KN, Adighije PF, Omolabake BI. Hirschsprung's disease in an adult: a case report. *J West Afr Coll Surg*. 2014;4:121-6.
7. Qiu J, Shi Y, Hu L, Fang L, Wang H, Zhang M. Case report. Adult Hirschsprung's disease: report of four cases. *Int J Clin Exp Pathol*. 2013;6:1624-30.
8. Reategui CO, Spears CA, Allred GA. Adults Hirschsprung's disease, a call for awareness. A case report and review of the literature. *Int J Surg Case Rep*. 2021;79:496-502.
9. Estopiñán Rebollar R, Estopiñán Cánovas R, Pila Peláez R. Hirschsprung's disease in an adult patient. *Rev Colomb Gastroenterol*. 2016;31:52-5.
10. Szyłberg Ł, Marszałek A. Diagnosis of Hirschsprung's disease with particular emphasis on histopathology. A systematic review of current literature. *Gastroenterol Rev*. 2014;5:264-9.
11. Uylas U, Gunes O, Kayaalp C. Hirschsprung's disease complicated by sigmoid volvulus: a systematic review. *Balkan Med J*. 2021;38:1-6.
12. Arencibia Pérez B, Delgado Sánchez JJ, Marchena Gómez J. Aganglionic megacolon in the adult. Urgent and surprising cause of intestinal occlusion. *Rev Esp Enferm Dig*. 2019;111:405.
13. Wu S, Sun X, Yu Y, Shen Y. Hirschsprung's disease-related giant sigmoid volvulus complicated by refractory hypertension in an elderly man. *Am J Case Rep*. 2018;19:467-71.
14. Needham V, Huang R, Levine R. Management of colonic volvulus. En: Cameron JL, Cameron AM, editores. *Current surgical therapy*. 13th ed. Baltimore: Elsevier; 2020. p. 215-9.
15. Mera-Martínez PF, Rivas-Goyes JA, Portillo-Miño JD. Megadolicocolon de presentación poco usual en un paciente de edad avanzada: a propósito de un caso. *Rev Colomb Cir*. 2020;35:695-8.