Clinics of Oncology[R]

ISSN 2640-1037 Volume 9

Case Report Open Access

Diffuse Non-Polypoid Intestinal Ganglioneuromatosis: An Uncommon Cause of Acute Abdomen. A Case Report

Fernández Bellone[1*], María Luz[1], Viscido Germán[1], Paganini, Virginia[1], Mandojana Facundo[1] and Doniquian Alejandro[1]

Coloproctology Surgery Section, General Surgery Department; Clínica Universitaria Reina Fabiola, Universidad Católica de Córdoba.

***Corresponding author:**

Fernández Bellone,
María Luz; General Surgery Department, Clínica Universitaria Reina Fabiola, Universidad 'Católica de Córdoba, Oncativo 1248, Córdoba, Argentina

Received: 03 Jan 2026
Accepted: 18 Jan 2026
Published: 4 Feb 2026
J Short Name: COO

Keywords:

Vólvulo Intestinal; Ganglioneuromatosis; Tumor Intestinal; Dolor Abdominal; Dolicolon

Abbreviations:

DGN: Diffuse Ganglioneuromatosis; DIGN: Diffuse form of Intestinal Ganglioneuromatosis;
GNP: Ganglioneuromatous Polyposis; GIST: Gastrointestinal Stromal Tumors; IGN: Intestinal Ganglioneuromatosis; MEN 2B: Multiple Endocrine Neoplasia type 2B; NF1: Neurofibromatosis type 1; PGN: Polypoid Ganglioneuromas

Citation:

Fernández Bellone, Diffuse Non-Polypoid Intestinal Ganglioneuromatosis: An Uncommon Cause of Acute Abdomen. A Case Report. Clinics of Oncology® 2025; V9(1): 1-4

1. Abstract

1.1. Introduction

Intestinal Ganglioneuromatosis (IGN) is an extremely rare benign neoplasm of the enteric nervous system, characterized by localized or diffuse proliferation of ganglion cells, nerve fibers, and Schwann cells within the intestinal wall. The diffuse form of Intestinal Ganglioneuromatosis (DIGN) is associated with genetic syndromes such as Neurofibromatosis type 1 (NF1) and Multiple Endocrine Neoplasia type 2B (MEN 2B).

1.2. Discussion

The diffuse form of Intestinal Ganglioneuromatosis presents clinically with nonspecific symptoms, including altered bowel transit, gastrointestinal bleeding, and occasionally intestinal obstruction or acute colonic volvulus.

Conclusion: Surgical intervention is the treatment of choice in symptomatic or complicated cases.

1.3. Presentation of The Case

We present the case of a 49-year-old female patient with a history of NF1, who presented with abdominal pain and symptoms consistent with intestinal obstruction. Her clinical presentation corresponded to a volvulus of the left colon, requiring emergency surgery with bowel resection and colostomy. Histopathological analysis confirmed diffuse form in the surgical specimen.

2. Introduction

Intestinal ganglioneuromatosis (IGN) is a rare, histologically benign tumor condition characterized by hyperplasia of the intramural plexuses of the gastrointestinal tract and enteric nerve fibers (Schwann cells). These neoplasms are histologically benign and derived from the sympathetic nervous system. Fewer than 100 cases have been reported to date [1-3]. Within the gastrointestinal tract, IGN is classified based on the pattern and extent of the lesion into three subtypes: polypoid ganglioneuromas (PGN), ganglioneuromatous polyposis (GNP), and diffuse ganglioneuromatosis (DGN) [3]. IGN accounts for approximately 0.1% of all intestinal disorders and 0.1% of motility disorders, following Hirschsprung disease and intestinal neuronal dysplasia [4]. Epidemiologically, there is no gender predilection, and it is more commonly diagnosed in childhood, being uncommon in adults [5]. DGN is associated with multiple endocrine neoplasia type 2B (MEN 2B), and less frequently with neurofibromatosis type 1 (NF1) or juvenile polyposis. This subtype frequently affects the colon, terminal ileum, and cecal appendix [5,6]. Clinical manifestations in the gastrointestinal tract depend on the location, focality or diffuseness, and size of the lesion. While often asymptomatic, patients may present with nonspecific symptoms such as altered bowel habits, intestinal obstruction, or gastrointestinal bleeding, among others [7,8]. Depending on the type, location, and clinical presentation, treatment may involve periodic surveillance, endoscopic resection, or surgical management, with most authors recommending surgery in cases of DGN [3,6,9]. We present the case of a female patient with NF1 and diffuse intestinal ganglioneuromatosis who presented with acute intestinal obstruction due to colonic volvulus requiring emergency surgery.

3. Case Presentation

A 49-year-old female with a history of neurofibromatosis type 1 presented with cutaneous neurofibromas on the face, popliteal fossae, lower extremities, and skull base, previously treated with radiotherapy. She was admitted with a 10-day history of severe (10/10 intensity) diffuse abdominal pain, accompanied by nausea, vomiting, abdominal distension, and three days without passage of flatus or bowel movements. On physical examination, the abdomen was distended with multiple cutaneous neurofibromas (Figure 1), absent bowel sounds, and pain on both superficial and deep palpation. Vital signs were abnormal: heart rate of 115 bpm and a blood pressure of 90/50 mmHg. Laboratory results showed marked leukocytosis of 31,100 cells/μL (reference range: 3.8–10 $\times 10^3$/μL), with 91% segmented neutrophils, elevated C-reactive protein at 70 mg/L (normal: <5 mg/L), and lactate at 3.41 mmol/L (normal: 0.5–2.2 mmol/L). With suspicion of bowel obstruction, an abdominal-pelvic CT scan was performed (Figure 2), revealing severe colonic dilatation, especially in the left-transverse colon with a transverse diameter of approximately 17 cm. A mesenteric twist was noted in the left flank and hypochondrium, consistent with colonic volvulus. Moderate free fluid was also observed in the pelvic cavity. An emergency exploratory laparotomy was indicated due to acute obstructive abdomen secondary to sigmoid volvulus. A midline supra- and infraumbilical incision was performed, revealing a dolichosigmoid colon with a lax mesentery and volvulus, involving approximately 35 cm of the descending/sigmoid colon with ischemic changes. A segmental colectomy was performed with creation of a terminal left transverse colostomy due to the patient's hemodynamic instability, acute presentation, and the severe colonic distension that precluded primary anastomosis. Postoperative recovery was favorable. The patient was managed in the intensive care unit and experienced return of bowel function on postoperative day 3. She was discharged on postoperative day 5. Histopathology revealed sloughed colonic epithelium with submucosal and muscular (and focally mucosal) proliferation of ganglion cells of the enteric plexus, occasionally forming ganglia. There was also hyperplasia and hypertrophy of nerve fibers in all four intestinal wall layers. These findings were consistent with non-polypoid diffuse ganglioneuromatosis. Immunohistochemistry was positive for S-100 protein.

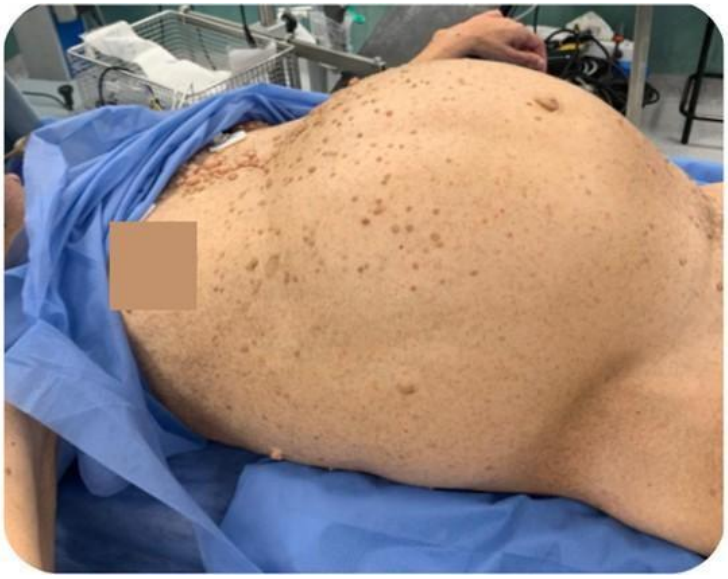

Figure 1: Neurofibromas cutaneos y gran distension abdominal.

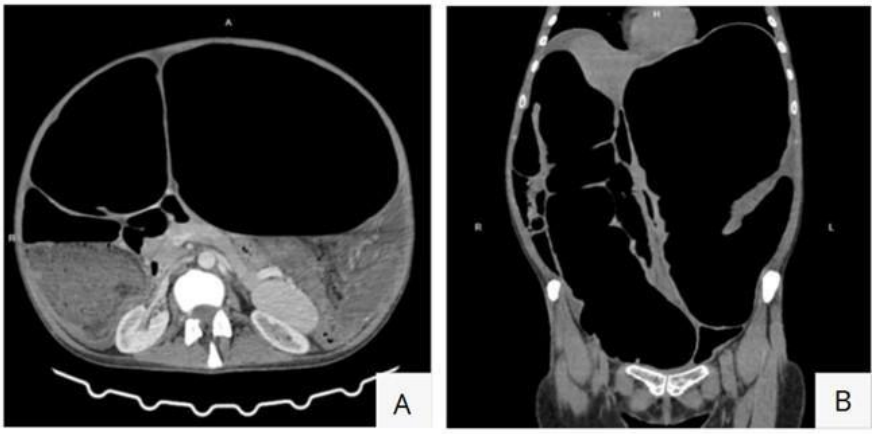

Figure 2: Corte axial (A) y coronal (B) de TAC abdomen-pelvis con contraste, en el cual se objetiva gran distension de asas intstinales y volvulo de sigmoides.

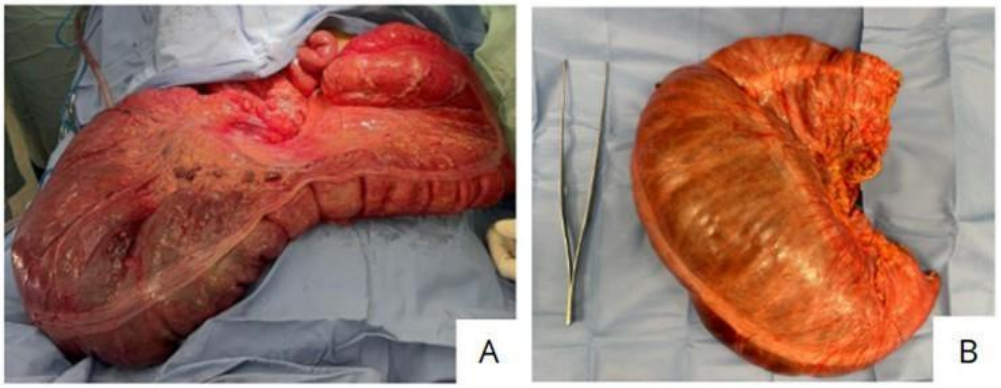

Figure 3: Pieza quirugica tras colectomia segmentaria: (A) Dolicocolon. (B) Colon sigmoides resecado.

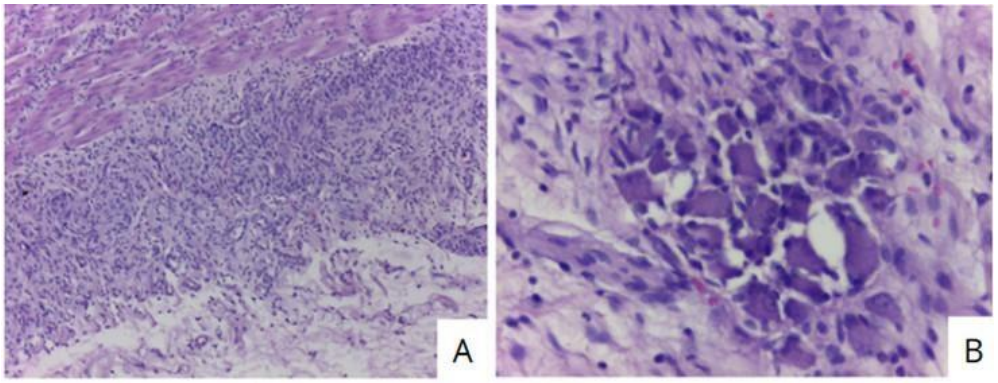

Figure 4: Hallazgos patologicos del colon: (A) Colon con tinccion HE a 10x, donde se ven filetes neriosos hipertroficos y mega ganglios del plexo de plexo de meisser en un fondo neurofibrilar. (B) Colon con tincion HE a 40x, donde se visualiza un mega ganglio nervioso (>10 celulas ganglionares).

4. Discussion

IGN is a rare neoplastic condition characterized by a prominent proliferation of ganglion cells, Schwannian nerve fibers, and hyperplasia of the intramural plexuses of the gastrointestinal tract [1,2]. Based on lesion pattern and extension, it is classified into three types: polypoid ganglioneuromas, ganglioneuromatous polyposis, and diffuse ganglioneuromatosis [3]. The most common subtype is the polypoid form (PGN), consisting of single or multiple mucosal and submucosal polyps resembling adenomas, typically found in the colon. GNP is defined by multiple sessile or pedunculated mucosal polyps in the colon or terminal ileum. DGN involves any intestinal wall layer but predominantly affects the Auerbach and submucosal plexuses, as in our case [3,4,10]. Morphologically, DGN may present as submucosal nodules or diffuse bowel wall thickening with or without associated stenosis [10]. In our case, approximately 35 cm of bowel showed severe dilatation, ischemia, and submucosal and muscular hypertrophy. The most common sites of involvement in DGN are the colon, cecal appendix, and terminal ileum [5]. DGN may present sporadically or, more commonly, in association with syndromes such as Neurofibromatosis Type I—as in our case (café-au-lait spots, cutaneous neurofibromas on the face, popliteal region, lower limbs, and skull base)-as well as Multiple Endocrine Neoplasia type 2B and Cowden syndrome [3]. While often asymptomatic, DGN may present with nonspecific gastrointestinal symptoms such as constipation, diarrhea, vomiting, episodic abdominal pain, or more severe complications such as megacolon, bowel obstruction, colonic volvulus, or gastrointestinal bleeding. Rare presentations include mucosal ulceration, anemia, intestinal perforation, steatorrhea, pancreatitis, obstructive jaundice, or colorectal carcinoma [2,6,9,11]. In our case, the patient presented with acute intestinal obstruction due to colonic volvulus with marked clinical and laboratory inflammatory response. Imaging findings depend on the clinical presentation. In our case, CT imaging demonstrated signs of colonic obstruction with volvulus. No specific radiologic findings for DGN have been described. Imaging may be normal or show nonspecific features such as segmental wall thickening, diffuse intestinal involvement mimicking Crohn’s disease, solitary or multiple wall or mesenteric lesions, or abscesses [4,6,7,9,12]. The endoscopic presentation is contingent upon the morphological distribution of the lesions, varying according to whether they exhibit a focal or diffuse pattern. Subepithelial masses or polyps of varying size and distribution may be seen. In some cases, numerous small lesions may line the gastrointestinal tract; in others, a dominant lesion may cause significant luminal narrowing or act as a lead point for intussusception. Multiple colonic ulcers or erosions may also be present. Endoscopic biopsy is critical for diagnosis, although deep lesions may not be adequately sampled, potentially yielding only normal or non-diagnostic superficial tissue [7,9,12]. The differential diagnosis includes Crohn’s disease, cytomegalovirus infection, intestinal tuberculosis, amyloidosis, and spindle-cell tumors such as gastrointestinal stromal tumors (GIST), leiomyomas, schwannomas, neurofibromas,

and mesenteric fibromatosis [3,9]. In endemic regions, Chagas disease-related intestinal involvement should be considered. Definitive diagnosis is histopathologic. Characteristic findings include band-like thickening or nodules composed of nerve fibers, Schwann cells, and ganglion cells of the enteric plexus, along with benign-appearing spindle cells with scant cytoplasm, rare or absent mitoses, and nuclear pleomorphism. These are interspersed with collagen fibers and varying numbers of mature ganglion cells [5,6,10]. Hematoxylin and eosin staining is typically sufficient for diagnosis in most cases. Immunohistochemical staining (positive for S-100, glial fibrillary acidic protein, vimentin, NSE, and synaptophysin) confirms the neural origin of the cellular proliferation [3,6,7,9]. Reported treatments include conservative management and continuous rifaximin therapy to alleviate symptoms such as abdominal pain and diarrhea due to bacterial overgrowth, with some clinical success [2,3]. Only one previously reported case described DGN presenting with intermittent volvulus requiring initial endoscopic and later surgical management [6]. In our case, surgery was immediately indicated based on clinical, laboratory, and imaging findings. Segmental resection and colostomy were performed. In DGN, conservative management generally fails, and surgical resection is considered the treatment of choice [2-4,6,9,12,13,14]. The relationship between intestinal ganglioneuromatosis and malignancy is not well established. Some studies report an association between GNP or DGN and adenomatous polyps or colorectal carcinoma, but the incidence is low, and the literature is insufficient to establish a definitive link [11,15].

5. Conclusion

Diffuse ganglioneuromatosis is a rare pathological condition that requires a high index of suspicion, especially in patients with a personal history of NF1. Surgical intervention remains the mainstay of treatment. Further case reporting is essential to develop standardized guidelines for diagnosis, management, and long-term follow-up of this uncommon disease.

References

1. Una patología muy infrecuente: ganglioneuromatosis intestinal. Reporte de un caso. Revista ACTA. 2020.
2. Charagundla SR, Levine MS, Torigian DA, Campbell MS, Furth EE J. Diffuse intestinal ganglioneuromatosis mimicking Crohn's disease. AJR Am J Roentgenol. 2004; 182: 1166-8.
3. Bahmad HF, Trinh S, Qian L, Terp K, Alloush F. Colonic Ganglioneuroma: A Combined Single-Institution Experience and Review of the Literature of Forty-Three Patients. Diseases. 2023; 11(2): 69.
4. Hoerschgen K, Muirhead D. Diffuse Ganglioneuromatosis of the Colon in a Patient with Neurofibromatosis 1. S D Med. 2022; 75: 392-4.
5. Al Samaan F, Alhimyar M, Alhalbouni M, Aldirani A, Assad L. Isolated diffuse intestinal ganglioneuromatosis presented as a redundant sigmoid colon: a case report. Ann Med Surg (Lond). 2023; 85: 219-24.
6. Hussein K, Josiane M, Layla Abou K, Rosy F, Georges A. Diffuse intestinal ganglioneuromatosis: A case report with review of literature. Int J Pathol Clin Res. 2022; 8.
7. Williams AJ, Doherty ES, Hart MH, Grider DJ. Diffuse Gastric Ganglioneuromatosis: Novel Presentation of Hamartoma Syndrome-Case Report and Review of Gastric Ganglioneuromatous Proliferations and a Novel Gene Mutation. Case Rep Med. 2018; 2018: 4319818.
8. Mauro A, Zenzeri L, Esposito F, Gaglione G, Strisciuglio C. Isolated intestinal Ganglioneuromatosis: case report and literature review. Ital J Pediatr. 2021; 47: 80.
9. Fernandes A, Ferreira AM, Serra P, Carvalho L. Intestinal ganglioneuromatosis: an unusual aetiology for occult gastrointestinal bleeding. BMJ Case Rep. 2015; 2015.
10. Iwamuro M, Omote R, Tanaka T, Sunada N, Nada T, Kondo Y. Diffuse Intestinal Ganglioneuromatosis Showing Multiple Large Bowel Ulcers in a Patient with Neurofibromatosis Type 1. Intern Med. 2017; 56: 3287-91.
11. Oh JS, Hong SW, Noh JH, Yoon J, Kang HJ, Park YS. Development of colon cancer in a patient with longstanding colonic diffuse ganglioneuromatosis: a case report. Clin Endosc. 2022; 55: 452-7.
12. Carter J-E, Laurini J-A. Isolated intestinal neurofibromatous proliferations in the absence of associated systemic syndromes. World J Gastroenterol. 2008; 14: 6569-71.
13. Aihole JS. A case of ganglioneuromatosis in a child and its outcome. International Journal of Surgery Case Reports. 2023; 107: 108335.
14. Lu C, Qiu Y, Lu X, Li G. Synchronous diffuse ganglioneuromatosis and multiple schwannomas of the colon: A case report and literature review. Experimental and Therapeutic Medicine. 2015; 9(3): 733-6.
15. Shousha S, Smith PA. Colonic ganglioneuroma. Report of a case in a patient with neurofibromatosis, multiple colonic adenomas and adenocarcinoma. Virchows Archiv A, Pathological anatomy and histology. 1981; 392(1): 105-9.