Isolated Intestinal Ganglioneuroma Mimicking Small Bowel Crohn’s Disease

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ABSTRACT

Ganglioneuromas (GNs) are hamartomatous tumors derived from the autonomic nervous system. GNs are frequently associated with neurofibromatosis-1 and multiple endocrine neoplasia type 2b and commonly present with constipation, abdominal pain, weight loss, obstruction, and gastrointestinal bleeding. We report a 40-year-old man with symptoms of chronic abdominal pain, weight loss, and diarrhea for 1 year. Imaging was suggestive of thickening of ileal wall with a stricture, and subsequent biopsy revealed intestinal GN. To our knowledge, this is the first case report of an isolated intestinal GN masquerading as Crohn’s disease.

INTRODUCTION

Ganglioneuromas (GNs) are usually associated with neurofibromatosis-1 and multiple endocrine neoplasia 2b (MEN 2b) syndromes but can present in a sporadic and isolated form. They are rarely seen in the gastrointestinal tract and can present either as a solitary lesion or as multiple polyps in the colon and/or terminal ileum. We describe a patient with chronic abdominal pain and intermittent diarrhea who was found to have a small bowel stricture and ulceration due to isolated GN in the absence of neurofibromatosis-1 or MEN 2b syndrome.

CASE REPORT

A 40-year-old man with a history of abdominal pain associated with chronic diarrhea was admitted to the hospital because of persistent and severe symptoms. An esophagogastroduodenoscopy done 1 year before this presentation showed Cytomegalovirus-esophagitis without evidence of Helicobacter pylori gastritis. Duodenal examination and biopsies were negative for celiac disease, inflammatory bowel disease, and lymphoma. Colonoscopy done at the same time with intubation of the terminal ileum showed normal mucosal examination, and random biopsies showed normal colonic mucosa. Laboratory workup came back positive for human immunodeficiency virus infection. The patient was started on antiretroviral therapy and ganciclovir for Cytomegalovirus.

A follow-up EGD with biopsy 6 months later showed resolution of the esophagitis, and with treatment, human immunodeficiency virus ribonucleic acid levels became undetectable. However, the patient continued to experience recurrent attacks of acute abdominal pain and watery diarrhea, with a significant weight loss of around 15 lbs. There was no vomiting, heartburn, or hematemesis. The patient denied the use of alcohol, cocaine, or nonsteroidal anti-inflammatory drugs. Physical examination revealed an afebrile, chronically ill, thin man in no acute distress. Abdominal examination revealed periumbilical tenderness without guarding, rebound, or rigidity. Laboratory workup showed mild chronic normocytic anemia and normal basic metabolic panel including electrolytes, calcium, and kidney function, without elevation in serum inflammatory markers. Stool was negative for white blood cells; ova and parasites were not detected and stool cultures showed normal flora.

Computed tomography scan of the abdomen with intravenous and oral contrast showed diffuse small bowel thickening. Capsule endoscopy was planned but could not be performed because the patient had a spinal stimulator in situ for chronic low back pain. Hence, a single balloon enteroscopy to the mid jejunum was performed with no evidence of ulcers, strictures, nodules, or mass lesions to explain...
the abdominal pain. A few weeks later, a computed tomography abdomen was repeated, which showed thickening from the terminal jejunum to the mid ileum with luminal narrowing (Figure 1). The pancreas and adrenal glands were unremarkable.

The working diagnosis was Crohn’s disease and the patient was referred for a diagnostic laparoscopy, which revealed thickening of mid to distal ileum. There was no lymphadenopathy. Forty centimeters of the ileum was resected and primary entero-enteral anastomosis was performed. The gross specimen showed diffuse edema, wall thickening, and areas of luminal narrowing and stricturing. Histology showed ileal mucosal ulcerations with ill-defined proliferation of ganglion cells and benign spindle-shaped Schwann cells with a significant eosinophilic infiltrate extending to the margin. Cells were positive for S100 and calretinin, findings consistent with a diagnosis of GN (Figure 2). Postoperatively, the patient recovered uneventfully. At a follow-up visit 6 months later, abdominal pain and diarrhea had resolved.

**DISCUSSION**

Intestinal GNs are encountered very rarely. They can be seen either in association with syndromes such as familial adenomatosis polyposis, tuberous sclerosis, Cowden syndrome, Von Recklinghausen syndrome, and MEN 2b, or they can be isolated. Isolated intestinal GN is much rarer, especially in the absence of systemic syndromes. Most reported cases involve the colon and rectum, and less commonly, the ileum and appendix.

Gastrointestinal GNs are categorized into 3 groups. Solitary GN is the most common form and can be found as sessile or pedunculated colon polyps, <2 cm in size, and not associated with systemic or genetic disorders. Multiple (>20 polyps), less than or equal to 2 cm in size, and diffuse GNs, which are nodular and large (up to 17 cm), involve the myenteric plexus.

Clinical presentation can be asymptomatic, but depending on the size, location, and degree of involvement of the affected segment, patients can present with abdominal pain, weight...
loss, obstruction, and bleeding. Most commonly, these are encountered incidentally during colonoscopy as polyps. Diagnosis is usually confirmed on histology. These tumors can also secrete catecholamines and vasoactive intestinal polypeptides, leading to diarrhea. Ulceration and development of strictures in isolated GN as in our patient is extremely rare and more typical of diffuse ganglioneuromatosis.

Endoscopic polypectomy of incidentally found single or multiple GN polyps is curative. There is a paucity of data on association between solitary GN and colon cancer. Follow-up surveillance-colonoscopy to ensure complete resection is probably beneficial because a few cases of coexisting colon cancer have been reported. For diffuse GN polyposis, colectomy may be necessary. Patients should also be screened for related genetic syndromes and tumors at other sites like thyroid, colon, breast, and uterus.

The differential diagnosis for benign small bowel strictures includes Crohn’s disease, chronic nonsteroidal anti-inflammatory drug use, ischemia, infections caused by parasites such as ascaris and bacteria such as tuberculosis. There are 2 cases in the literature of diffuse intestinal GN, mimicking Crohn’s. To our knowledge, this is the first case of isolated GN presenting with symptomatic small bowel stricture.

DISCLOSURES

Author contributions: M. Badrinath analyzed the data, wrote and edited the manuscript, and is the article guarantor. D. Lowe, D. Manocha, and T. Achufusi revised the manuscript.

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Informed consent was obtained for this case report.

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