Revista Española de Enfermedades Digestivas
The Spanish Journal of Gastroenterology

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DOI: 10.17235/reed.2023.9660/2023
Link: PubMed (Epub ahead of print)

Please cite this article as:
Guerrero Palma Estefanía, Hidalgo Blanco Alba, Benavides de la Fuente Cristian. Diffuse intestinal ganglioneuromatosis. A post mortem diagnosed challenging case. Rev Esp Enferm Dig 2023. doi: 10.17235/reed.2023.9660/2023.

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Diffuse intestinal ganglioneuromatosis. A *post mortem* diagnosed challenging case

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Conflict of interest: the authors declare no conflict of interest.

Keywords: Intestinal ganglioneuromatosis. Benign neoplasm. Enteric plexus hyperplasia.

Dear Editor,

Intestinal ganglioneuromatosis (GN) is a benign disease of the autonomic nervous system characterized by hyperplasia of intramural plexuses of the gastrointestinal tract and enteric nerve fibers. In this article, we present a case of intestinal ganglioneuromatosis that was difficult to diagnose, despite an exhaustive evaluation, for further understanding of the disease.

Case report

A 74-year-old male with cardiovascular risk factors was referred to our Emergency Department due to sudden abdominal pain with increased acute phase reactants, ileitis and secondary acute occlusion signs in an urgent abdominal computed tomography (CT) scan. As infectious origin was first thought, antibiotherapy was started and ileocolonoscopy was performed, without any abnormalities, and biopsies for histology and culture showed no alterations. Infectious serology and stool analysis were negative. Magnetic resonance (MR) enterography showed transmural

involvement of the terminal ileum, so Crohn's disease was suspected and corticotherapy was started for five days without improvement. Ischemic origin and vasculitis were ruled out with CT angiography, showing vascular permeability and autoimmunity profile with negative complement. There were no adenopathies or masses suggestive of intestinal lymphoma in thoraco-abdominal CT. Mantoux and quantiferon were negative, showing clinical progression with abdominal pain resistant to medication, hypoalbuminemia and edema. Gastroscopy with biopsies was performed to rule out protein-losing enteropathy secondary to celiac disease. Videocapsule endoscopy and subsequent enteroscopy showed only a caliber change in jejunoileal tract, without macroscopic alterations. Congo red staining was performed in all biopsies ruling out amyloidosis. Laparotomy with surgical biopsies was proposed but the patient presented progressive clinical deterioration and died. Necropsy showed diffuse ileal ganglioneuromatosis.

Discussion

Intestinal ganglioneuromatosis is a very rare and benign neoplasm characterized by hyperplasia of intramural plexuses of the gastrointestinal tract and enteric nerve fibers. Its presentation is exceptional in adults, affecting mainly children, in whom it is associated with neurofibromatosis type I (NF1) and MEN IIb syndrome.

There is a mucosal and transmural presentation, which can affect any segment of the gastrointestinal tract but occurs more commonly in the ileum, colon and appendix. The clinical manifestations will depend on the affected segment, causing changes in bowel habits, abdominal pain, occlusive symptoms and rarely lower gastrointestinal bleeding secondary to ulceration or erosions of the intestinal mucosa.

The diagnosis requires histopathological findings from endoscopic biopsy, surgical specimen or necropsy. Immunohistochemical examination of the biopsy reveals immunoreactivity to S100. Surgical treatment of the affected segment is the main option. There are no reported data on intestinal transplantation in this entity.

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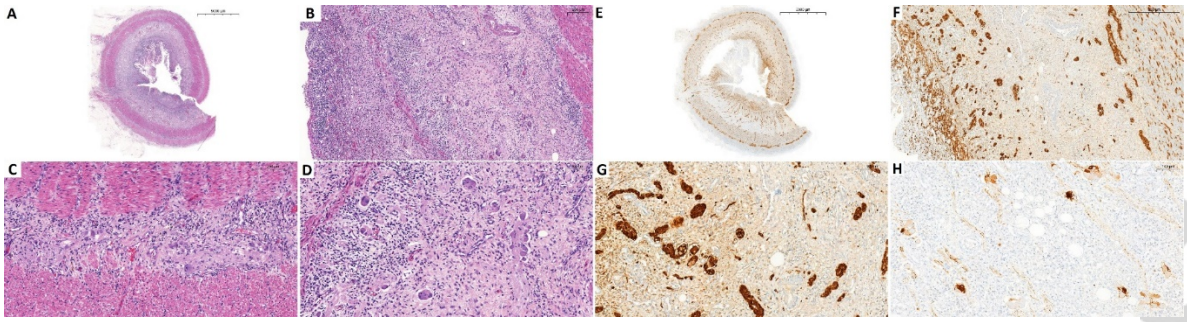

Fig. 1. A. Histopathological study of ileum sections shows focal ulceration of the superficial epithelium with underlying granulation tissue. B-G. In the thickness of the mucosa, submucosa (B) and muscularis propria (C), there is a diffuse proliferation of mature ganglion cells of anomalous distribution expressing positive immunostaining for calretinin (H), which are accompanied by numerous nerve fibers (D) with the presence of positive S100 Schwann spindle cells (E-G) of submucosal and myenteric localization.