EJCRIM
European Journal
of Case Reports
in Internal Medicine

MULTIFOCAL COLONIC MASTOCYTOSIS MIMICKING ISCHAEMIC COLITIS: A RARE CAUSE OF LOWER GASTROINTESTINAL BLEEDING

Karam Karam[1], Kim Charro[1], Aurélien Morini[2], Emilien Brousset[1], Christophe Locher[1]

[1] Department of Gastroenterology & Hepatology, Grand Hôpital de l'Est Francilien (GHEF), Meaux, France
[2] Department of Anatomical Pathology, Grand Hôpital de l'Est Francilien (GHEF), Meaux, France

Corresponding author's e-mail: karamek7@gmail.com

Received: 05/03/2026
Accepted: 16/03/2026
Published: 28/05/2026

Conflicts of Interests: The Authors declare that there are no competing interests.
Patient Consent: A signed informed consent was obtained.
This article is licensed under a CC Attribution-NonCommercial-NoDerivatives 4.0 License

How to cite this article: Karam K, Charro K, Morini A, Brousset E, Locher C. Multifocal colonic mastocytosis mimicking ischaemic colitis: a rare cause of lower gastrointestinal bleeding. *EJCRIM* 2026;**13**:doi:10.12890/2026_006523

ABSTRACT

Introduction: Mastocytosis is a rare clonal disorder characterised by abnormal proliferation and accumulation of mast cells in one or more organs. Although gastrointestinal symptoms are common in systemic mastocytosis, direct mucosal infiltration by neoplastic mast cells is uncommon and remains poorly characterised. Isolated involvement of the colon is exceptionally rare and may present with non-specific clinical and endoscopic findings.

Case description: We report the case of a 59-year-old male presenting with recurrent rectorrhagia without associated gastrointestinal symptoms or relevant medical history. Colonoscopic examination revealed mucosal changes suggestive of ischaemic colitis. Histopathological evaluation of colonic biopsies demonstrated dense aggregates of mast cells within the lamina propria and muscularis mucosae. The cells exhibited characteristic morphology with oval to spindle-shaped forms, abundant cytoplasm containing basophilic granules and centrally located hyperchromatic nuclei. Immunohistochemical analysis showed strong positivity for CD117 (KIT) and tryptase, while CD25 and CD138 were negative. No significant increase in eosinophils or lymphocytic infiltrates was observed in adjacent mucosa. These findings were consistent with multifocal colonic mastocytosis in the absence of evidence of systemic disease.

Conclusion: Colonic mastocytosis is an exceedingly rare entity that may mimic other gastrointestinal disorders both clinically and endoscopically. Histological examination with immunohistochemical confirmation is essential for accurate diagnosis. Increased awareness of this rare presentation is important for appropriate evaluation and follow up, given the potential risk of progression to systemic mastocytosis.

KEYWORDS

Mastocytosis, colonic mastocytosis, immunohistochemistry, rectorrhagia

LEARNING POINTS

- Gastrointestinal mastocytosis is rare and may present without systemic manifestations.
- Colonic involvement is exceptionally uncommon and may mimic ischaemic or inflammatory colitis on endoscopy.
- Histology with immunohistochemical staining (CD117 and tryptase) is essential for diagnosis.
- Long-term follow up is recommended to monitor for potential progression to systemic mastocytosis.

EFIM
European Federation of Internal Medicine

Publisher
smc media srl

INTRODUCTION

Mastocytosis is a rare clonal disorder characterised by the proliferation and accumulation of neoplastic mast cells in various tissues. In systemic mastocytosis, the bone marrow is almost invariably involved, and cutaneous manifestations occur in approximately 80% of cases. Gastrointestinal symptoms are common and are primarily mediated by the release of mast cell-derived mediators such as histamine, resulting in manifestations including abdominal pain, diarrhoea, nausea and vomiting.

Infiltration of the gastrointestinal tract by neoplastic mast cells is uncommon and most frequently affects the terminal ileum and duodenum. In contrast, primary involvement of the colon in the absence of systemic mastocytosis is exceedingly rare.

We report the case of a 59-year-old male presenting with recurrent rectorrhagia in whom colonic biopsies revealed aggregates of mast cells with characteristic morphological features, including oval to spindle-shaped cells with abundant cytoplasm, hyperchromatic nuclei and basophilic cytoplasmic granules. Immunohistochemical analysis demonstrated positivity for CD117 and tryptase and negativity for CD25 and CD138. No significant increase in eosinophils or lymphocytes was observed in the surrounding mucosa.

This case highlights the diagnostic importance of histological and immunohistochemical evaluation in identifying gastrointestinal mastocytosis and underscores the need for increased clinical awareness of this rare presentation.

CASE DESCRIPTION

A 59-year-old male was referred to the gastroenterology clinic for evaluation of recurrent rectorrhagia. The patient had no significant past medical history and no family or personal history of gastrointestinal malignancy or inflammatory bowel disease. He denied any changes in bowel habits, abdominal pain, nausea, vomiting or diarrhoea.

Routine laboratory investigations were within normal limits, and physical examination was unremarkable.

Colonoscopy revealed circumferential interspersed non-necrotic ischaemic ulcers with oedematous mucosa and lumen narrowing suggestive of ischaemic colitis *(Fig. 1)*. Random colonic biopsies were obtained for histopathological evaluation.

Microscopic examination demonstrated sheets and aggregates of mast cells within the colonic mucosa. The cells exhibited a characteristic 'fried-egg' appearance and were round to oval in shape with abundant pale eosinophilic cytoplasm containing fine basophilic granules. The nuclei were centrally located and hyperchromatic and showed prominent nucleoli. The mast cells displayed distinct cytoplasmic borders and infiltrated both the lamina propria and the muscularis mucosae *(Fig. 2A and B)*. Notably, there was no associated increase in eosinophils or lymphocytic inflammatory infiltrates in the adjacent mucosa. Immunohistochemical analysis demonstrated strong membranous and cytoplasmic positivity for CD117 and diffuse cytoplasmic positivity for tryptase. The mast cells were negative for CD25 and CD138 *(Fig. 2C and D)*. Based on the histopathological and immunohistochemical findings, a diagnosis of colonic mastocytosis was established.

A workup for systemic mastocytosis was performed, including clinical evaluation for cutaneous and extracutaneous involvement. No evidence of mast cell infiltration in other organs was identified. The patient received antihistamines loratadine and famotidine (H1+H2 blockade) along with cromolyn sodium, a mast cell stabiliser. Targeted oral therapy with avapritinib (25 mg once daily) was started with subsequent complete clinical improvement. At the most recent follow up, the patient remained clinically stable without new lesions or systemic symptoms.

DISCUSSION

Mastocytosis is a rare clonal myeloproliferative disorder characterised by the abnormal proliferation and accumulation of mast cells in one or more organs. The disease encompasses a heterogeneous spectrum ranging from indolent cutaneous forms to aggressive systemic variants, including mast cell leukaemia. Systemic mastocytosis is the most frequent form diagnosed in adults and is characterised by extracutaneous involvement, most commonly affecting the bone marrow, liver, spleen, lymph nodes and gastrointestinal tract[1].

Gastrointestinal involvement represents one of the most common extracutaneous manifestations of mastocytosis. Reported prevalence varies widely, with gastrointestinal symptoms occurring in approximately 14–85% of patients, reflecting the heterogeneity of disease expression[2]. Symptoms are generally non-specific and include abdominal pain, diarrhoea, nausea, vomiting and malabsorption. These manifestations are largely mediated by the systemic release

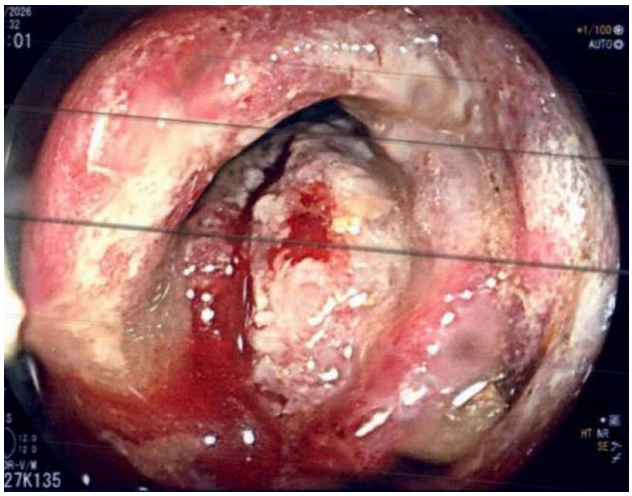


Figure 1. *Colonoscopic findings revealing circumferential interspersed non-necrotic ischaemic ulcers with oedematous mucosa suggestive of ischaemic colitis.*

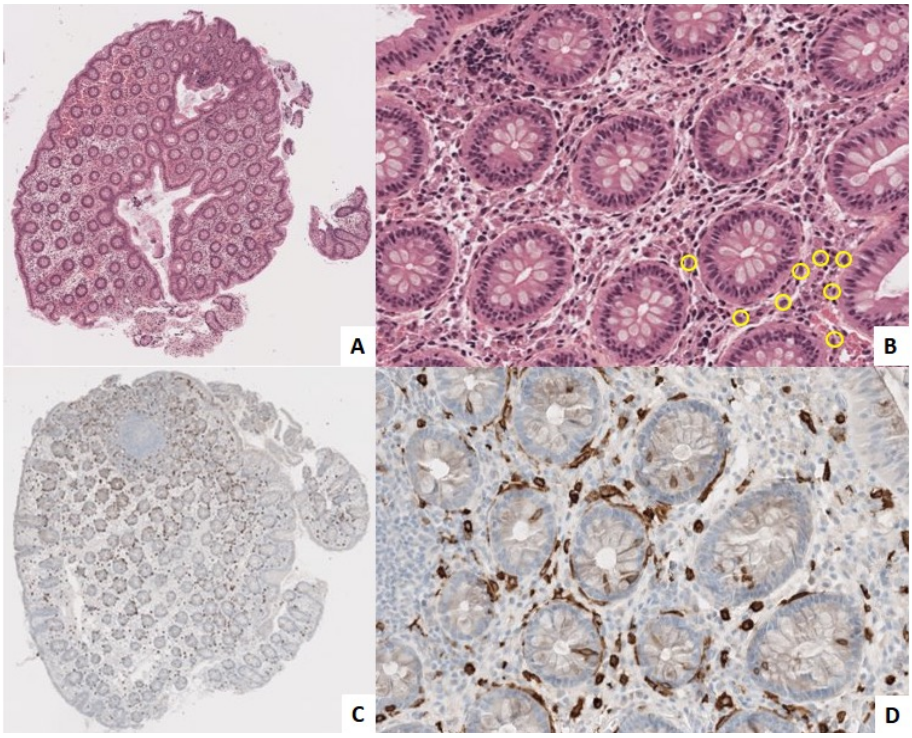


Figure 2. A) Haematoxylin and eosin staining (H&E) (×5), B) H&E (×30). The yellow circles correspond to mast cells. C) c-Kit (×5), D) c-Kit (×30).

of mast cell mediators such as histamine, leukotrienes, prostaglandins and proteases, which affect gastrointestinal motility, mucosal permeability and inflammatory pathways[1].

Despite the high frequency of gastrointestinal symptoms in systemic mastocytosis, direct infiltration of the gastrointestinal mucosa by neoplastic mast cells is relatively uncommon and remains poorly characterised. Most reported cases involve the small intestine, particularly the duodenum and terminal ileum, while isolated colonic involvement is rarely described[3]. Moreover, gastrointestinal mastocytosis may present with non-specific endoscopic findings, including mucosal erythema, nodularity, polypoid lesions or subtle inflammatory changes, which can mimic inflammatory bowel disease, ischaemic colitis or other inflammatory conditions[4].

Histopathological examination remains the cornerstone of diagnosis. The key diagnostic feature is the presence of multifocal dense aggregates of mast cells within the lamina propria, often composed of spindle-shaped or oval cells with abundant cytoplasm containing metachromatic granules. Immunohistochemical staining is essential to confirm the mast cell lineage, with neoplastic mast cells typically expressing CD117 (KIT) and tryptase, two highly sensitive markers for mast cell identification. Aberrant expression of CD25 or CD2 may suggest systemic mastocytosis and is incorporated into the current diagnostic criteria[1].

The differential diagnosis includes inflammatory bowel disease, eosinophilic colitis and reactive mast cell hyperplasia, conditions in which mast cells may be increased but generally do not form dense aggregates. Recognition of these histological patterns is critical, as gastrointestinal mastocytosis may be overlooked in routine biopsies, particularly in the absence of classic cutaneous manifestations. Several studies have demonstrated that up to 40–50% of patients with systemic mastocytosis may lack skin lesions at the time of diagnosis, further complicating clinical recognition[3].

In this case, the patient presented with recurrent rectorrhagia, an unusual manifestation of mastocytosis. Most reported gastrointestinal presentations involve diarrhoea, abdominal pain or dyspeptic symptoms, while lower gastrointestinal bleeding is rarely described in the literature. The endoscopic findings initially suggested ischaemic colitis, highlighting the potential for mastocytosis to mimic other gastrointestinal disorders and emphasising the importance of histological assessment.

Our case also underscores the need for systematic clinical evaluation to exclude systemic mastocytosis, including serum tryptase measurement, molecular testing for KIT mutations (particularly KIT D816V) and bone marrow examination when indicated. The diagnosis of systemic mastocytosis is established according to World Health Organization criteria, which require the presence of multifocal mast cell aggregates in extracutaneous tissues in combination with specific minor criteria such as aberrant immunophenotype or KIT mutation[1].

Although isolated gastrointestinal mastocytosis appears to follow a more indolent clinical course, long-term surveillance remains essential. Patients initially presenting with localised disease may subsequently develop systemic involvement over time. Therefore, multidisciplinary collaboration between gastroenterologists, pathologists and haematologists is crucial to ensure accurate diagnosis, appropriate staging and optimal follow up.

This report highlights a rare presentation of multifocal colonic mastocytosis manifesting as recurrent rectorrhagia, emphasising the importance of considering mast cell

disorders in the differential diagnosis of unexplained gastrointestinal bleeding. Increased awareness among clinicians and pathologists is essential to avoid diagnostic delay and to facilitate appropriate management.

REFERENCES

1. Nouri A, Rajab I, Ayaseh QZ, Hussein A, Farokhian A, Cavanagh, Y. Unmasking systemic mastocytosis: a case of gastrointestinal involvement misdiagnosed as achalasia. *J Surg Case Rep* 2025;**2025**:rjaf590. doi: 10.1093/jscr/rjaf5908
2. Patel HN, Hudey SN, Pepper AN. A case of systemic mastocytosis with predominately gastrointestinal manifestations 1845. *Am J Gastroenterol* 2018;**113**:S1048–S1049.
3. Kirsch R, Geboes K, Shepherd NA, de Hertogh G, Di Nicola N, Lebel S, et al. Systemic mastocytosis involving the gastrointestinal tract: clinicopathologic and molecular study of five cases. *Mod Pathol* 2008;**21**:1508–1516. doi: 10.1038/modpathol.2008.158
4. Bedeir A, Jukic DM, Wang L, Mullady DK, Regueiro M, Krasinskas AM. Systemic mastocytosis mimicking inflammatory bowel disease: a case report and discussion of gastrointestinal pathology in systemic mastocytosis. *Am J Surg Pathol* 2006;**30**:1478–1482. doi: 10.1097/01.pas.0000213310.51553.d7