Internal Medicine
The Japanese Society of Internal Medicine
doi: 10.2169/internalmedicine.4664-24
Intern Med 64: 2155-2159, 2025
http://internmed.jp

【 CASE REPORT 】

Myeloid Sarcoma in the Small Intestine

Masaya Iwamuro[1], Tomohiro Kamio[1], Shoichiro Hirata[1], Katsunori Matsueda[1], Daisuke Kametaka[1], Takehiro Tanaka[2], Seiji Kawano[1] and Motoyuki Otsuka[1]

Abstract:
Myeloid sarcoma is a rare extramedullary tumor of immature myeloid cells that is often associated with acute myeloid leukemia (AML). We herein report an 81-year-old man who presented with intestinal obstruction due to myeloid sarcoma of the small intestine. Diagnostic challenges were overcome using double-balloon enteroscopy and a biopsy, which confirmed the diagnosis of myeloid sarcoma. The patient subsequently developed AML but responded well to chemotherapy. This case underscores the importance of considering myeloid sarcoma in the differential diagnosis of small-bowel tumors. Highlighting the significance of a histological analysis, even in patients presenting with small bowel obstruction, the early diagnosis and treatment are crucial for improving outcomes, particularly in patients without a history of hematologic malignancies.

Key words: acute myeloid leukemia, double-balloon enteroscopy, myeloid sarcoma, small intestine

(Intern Med 64: 2155-2159, 2025)
(DOI: 10.2169/internalmedicine.4664-24)

Introduction

Myeloid sarcoma, also known as granulocytic sarcoma or chloroma, is a rare extramedullary manifestation of acute myeloid leukemia (AML) and other myeloproliferative disorders, characterized by the proliferation of immature myeloid cells outside the bone marrow (1-5). It can be present at various anatomical sites, most commonly the skin, lymph nodes, and soft tissues. However, it can also affect the gastrointestinal tract, bones, and central nervous system. Myeloid sarcoma is reported in approximately 3-9% of patients with AML; it may occur concomitantly with AML, precede AML, or present as an isolated lesion without bone marrow involvement (6, 7).

We herein report a case of myeloid sarcoma with small-bowel infiltration, detailing its clinical presentation and diagnostic challenges. This case underscores the importance of considering myeloid sarcoma in the differential diagnosis of small-bowel tumors in patients without a history of hematological malignancies.

Case Report

An 81-year-old Japanese man presented to his primary care physician with abdominal distension and constipation that had persisted for three months. He did not exhibit symptoms suggestive of AML, such as fatigue due to anemia, palpitations, shortness of breath, infection-related signs, such as a fever due to a decreased immune function, or bleeding symptoms associated with thrombocytopenia. The patient was diagnosed with small intestinal obstruction, and a nasointestinal tube was inserted. Subsequently, he was transferred to our hospital for a further evaluation.

The patient had no history of hematological disease or medication use. The family history was negative for any blood disorders. Blood tests at the time of transfer revealed anemia with a red blood cell count of 4.00×10^6/μL, hemoglobin level of 12.8 g/dL, and hematocrit value of 20.8%. The white blood cell count was 5,520/μL, with normal proportions of neutrophils (50.1%), lymphocytes (40.0%), monocytes (8.4%), eosinophils (1.4%) and basophils (0.2%). No atypical myeloid or lymphoid cells were observed in the peripheral blood. The patient exhibited hypoproteinemia,

[1]Department of Gastroenterology and Hepatology, Okayama University Graduate School of Medicine, Dentistry, and Pharmaceutical Sciences, Japan and [2]Department of Pathology, Okayama University Graduate School of Medicine, Dentistry, and Pharmaceutical Sciences, Japan
Received: September 5, 2024; Accepted: October 25, 2024; Advance Publication by J-STAGE: December 19, 2024
Correspondence to Dr. Masaya Iwamuro, iwamuromasaya@yahoo.co.jp

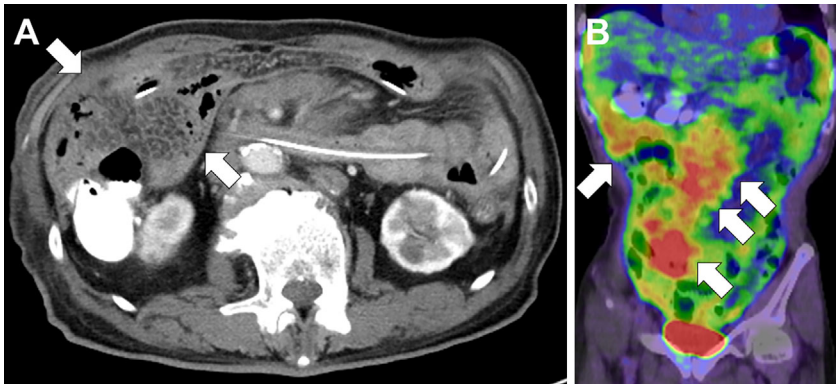

Figure 1. Radiology images. Abdominal CT shows increased density of the greater omentum fat (A, arrows). Positron emission tomography-CT demonstrates a marked tracer uptake in the small intestine and mesenteric lesions, suggesting disease infiltration (B, arrows). CT: computed tomography

with a total protein level of 5.8 g/dL and an albumin level of 2.8 g/dL. Elevated levels of alkaline phosphatase [361 U/L (reference range: 106-322 U/L)], lactate dehydrogenase [343 U/L (124-222 U/L), creatinine (1.27 mg/dL), amylase (164 U/L (44-132 U/L)], C-reactive protein (2.51 mg/dL), beta-2 microglobulin (7.17 mg/L), and soluble interleukin-2 receptor (1,908 U/mL) were also noted.

Computed tomography (CT) revealed thickening of the small intestinal wall, multiple mesenteric lymphadenopathies, and an increased density of the greater omentum fat (Fig. 1A). Positron emission tomography-CT revealed a marked tracer accumulation in the small intestine and mesenteric lesions, indicating spread of the disease within the abdominal cavity (Fig. 1B, arrows). Lymphadenopathy with the uptake of tracer was observed in the mediastinal, hilar, parasternal, and right supraclavicular brain regions.

Video capsule endoscopy was not performed because of small intestinal obstruction; therefore, we performed peroral double-balloon enteroscopy. A contrast study using double-balloon enteroscopy revealed luminal narrowing of the ileum (Fig. 2A, arrow) and a circumferential ulcerative tumor with a granular surface as the cause of the intestinal obstruction (Fig. 2B-D). The endoscope could not pass through the stricture, which was thought to be tumor-related. Multiple elevated lesions were observed in the duodenum, jejunum, and ileum (Fig. 2E). An endoscopic biopsy revealed neoplastic cells with dispersed chromatin and high nuclear-to-cytoplasmic ratio (Fig. 3A, B). The cells were positive for myeloperoxidase (Fig. 3C).

Although no atypical cells were observed in the peripheral blood on admission, myeloblasts were detected in 6% of the peripheral blood samples on day 4 of hospitalization. A bone marrow examination showed that 30-40% of the cells were blasts, characterized by a relatively large size, high nucleus-to-cytoplasm ratio, and basophilic cytoplasm with occasional vacuoles. Scatter eosinophils with basophilic granules were also observed. No dysplastic changes were observed in the background cells. A flow cytometric analysis revealed that blasts were positive for CD33, CD34, and HLA-DR. A chromosomal analysis revealed inv(16)(p13.1q22). These findings led to a diagnosis of acute myelomonocytic leukemia with eosinophilia. The intestinal lesions were identified as myeloid sarcomas.

The patient responded to induction chemotherapy for AML and later achieved molecular remission.

Discussion

Myeloid sarcoma is a rare extramedullary manifestation of AML and other myeloproliferative disorders that can present at various anatomical sites. Although myeloid sarcomas within the gastrointestinal tract are relatively rare, the small intestine is considered the most common site of occurrence (8-10). According to a review of myeloid sarcoma cases involving the gastrointestinal system, the small intestine accounts for approximately 78% (11/14) of cases (11). Further studies have confirmed that the small intestine is the most frequent site of occurrence of myeloid sarcoma within the intestinal tract, with the ileum being the most commonly affected part, accounting for approximately 65% of cases (12). However, the absolute incidence of myeloid sarcoma is low, and its proportion among small intestinal tumors remains unknown.

A PubMed search using the keywords ‘myeloid sarcoma’ and ‘balloon’ identified five reported cases in which balloon-assisted enteroscopy was employed to examine small-intestinal lesions of myeloid sarcoma (13-17). Table summarizes the six cases, including the present case. Among these, five were men and one was a woman, with ages ranging from 33 to 81 years old. Although the specific location of the small intestinal lesion was not detailed in one case, three cases involved the ileum, and one involved the jejunum. In one case, balloon-assisted enteroscopy could not detect small intestinal lesions, whereas in another case, it revealed multiple shallow ulcers with irregular margins. In contrast, in four cases, including the present case, myeloid sarcoma appeared as a mass lesion with a granular surface attributed to enlarged villi. Despite the limited number of reports that pose challenges to generalization, findings from previous and current cases suggest that the typical endo-

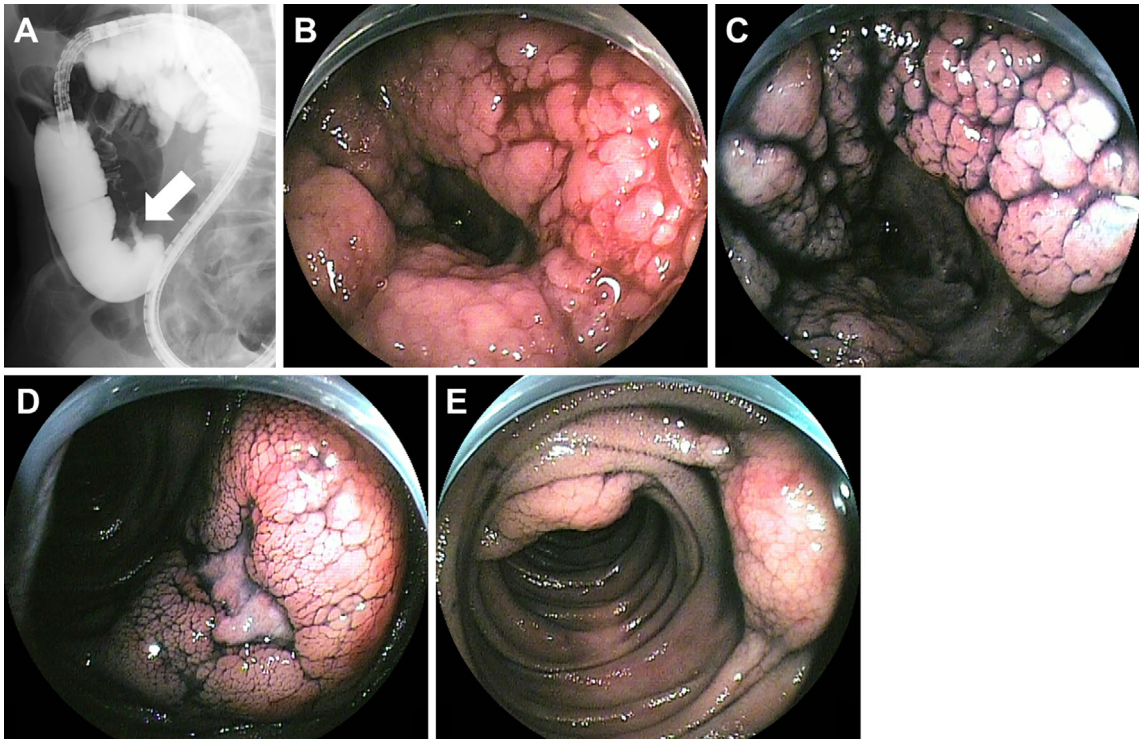

Figure 2. **Endoscopy examination images. A contrast study using double-balloon enteroscopy shows luminal narrowing in the ileum (A, arrow). Double-balloon enteroscopy reveals a circumferential ulcerative tumor with a granular surface in the ileum (B, white light; C, after indigo carmine spraying). Ulcers are noted on the tumor (D). Multiple elevated lesions are observed, consistent with disseminated myeloid sarcoma (E).**

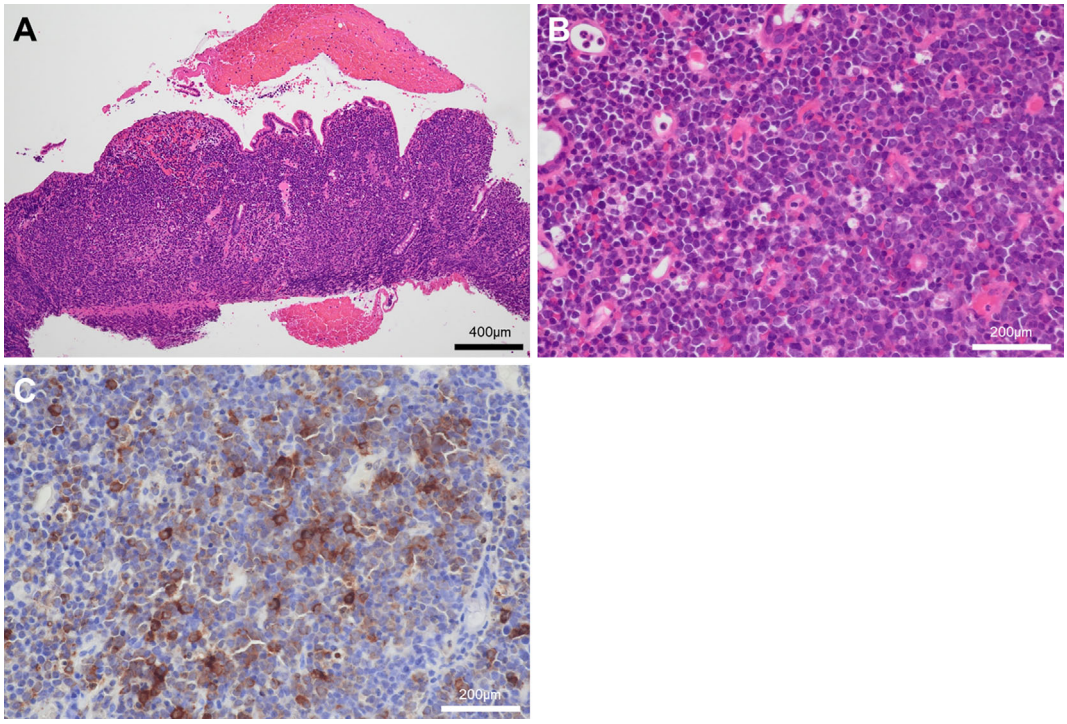

Figure 3. **Histopathological findings of endoscopic biopsy specimens from the small intestine. Neoplastic cells diffusely infiltrate the small intestinal mucosa [A, Hematoxylin and Eosin (H&E) staining, scale bar=400 μm]. High-power view showing neoplastic cells with dispersed chromatin and a high nuclear-to-cytoplasmic ratio (B, H&E staining, scale bar=200 μm). Immunohistochemical staining shows positivity for myeloperoxidase, confirming the diagnosis of myeloid sarcoma (C, scale bar=200 μm).**

Table. Reported Cases in which Balloon-assisted Enteroscopy was Employed to Examine Small Intestinal Lesions of Myeloid Sarcoma.

Reference no.	Year	Age	Sex	Involved organ(s)	Observations of double-balloon enteroscopy
13	2013	56	M	Jejunum	A tumorous lesion with swollen villi of different sizes
14	2014	50	M	Ileum	Diffuse thickening with swollen villi of different sizes
15	2020	51	M	Not specified	Multiple tumors with red and swollen villi
16	2022	40	M	Ileum	Multiple shallow ulcers with irregular margins
17	2024	33	F	Ileum	Not detected
Present case	-	81	M	Duodenum, jejunum, and ileum	Tumors with a granular surface, with or without ulceration

scopic appearance of small-intestinal myeloid sarcoma is a single or multiple tumor with a granular surface due to villous enlargement. Small intestinal lesions with this appearance may include subepithelial proliferative neoplastic diseases, such as lymphoma and neuroendocrine tumors, or subepithelial deposition diseases, such as amyloidosis. Therefore, a histopathological evaluation through a biopsy is essential for a definitive diagnosis, underscoring the importance of a balloon-assisted endoscopic evaluation and a biopsy for an accurate diagnosis.

Causes of mechanical intestinal obstruction include tumors, foreign bodies, postoperative adhesions, intussusception, volvulus, and hernias. Previous studies have demonstrated that balloon-assisted enteroscopy is effective for diagnosing intestinal obstruction (18-21). Although surgical intervention is often required, obtaining an accurate preoperative diagnosis enables the selection of the most appropriate surgical procedure.

In our case, myeloid sarcoma lesions in the small intestine appeared before leukemic cells were detected in the peripheral blood. This observation underscores the fact that myeloid sarcomas generally pose a risk for progression to AML. If left untreated, they are reported to progress to AML within an average of 10-12 months (2, 22, 23). Several studies have suggested that patients with myeloid sarcomas may have better treatment outcomes than those with AML when treated with AML-type therapy (24). In a study by Tsimberidou et al., the event-free and overall survival rates in patients with myeloid sarcoma were superior to those of matched patients with AML (25). This higher treatment responsiveness in myeloid sarcoma could be attributed to the relatively lower tumor burden and absence of systemic leukemic involvement at the time of the diagnosis. Therefore, recognizing myeloid sarcoma as a distinct clinical entity and its early diagnosis are crucial for optimizing patient outcomes. Accurate differentiation between this disease and other forms of gastrointestinal malignancies is vital, as it directly influences the treatment strategy and prognosis.

Myeloid sarcoma of the small intestine is a rare but clinically significant entity that presents unique diagnostic and therapeutic challenges. The early and accurate diagnosis of myeloid sarcoma is crucial, given its favorable response to AML-type chemotherapy compared to AML itself. This case emphasizes the importance of a histological analysis even in patients presenting with small bowel obstruction. Clinicians should consider myeloid sarcoma in the differential diagnosis of small-bowel tumors with a granular surface due to villous enlargement, even in patients without a history of hematologic malignancies, to ensure timely and appropriate management.

The authors state that they have no Conflict of Interest (COI).

References

1. Loscocco GG, Vannucchi AM. Myeloid sarcoma: more and less than a distinct entity. Ann Hematol **102**: 1973-1984, 2023.
2. Zhao H, Dong Z, Wan D, et al. Clinical characteristics, treatment, and prognosis of 118 cases of myeloid sarcoma. Sci Rep **12**: 6752, 2022.
3. Yilmaz AF, Saydam G, Sahin F, et al. Granulocytic sarcoma: a systematic review. Am J Blood Res **3**: 265-270, 2013.
4. Neiman RS, Barcos M, Berard C, et al. Granulocytic sarcoma: a clinicopathologic study of 61 biopsied cases. Cancer **48**: 1426-1437, 1981.
5. Pileri SA, Ascani S, Cox MC, et al. Myeloid sarcoma: clinicopathologic, phenotypic and cytogenetic analysis of 92 adult patients. Leukemia **21**: 340-350, 2007.
6. Eckardt JN, Stölzel F, Kunadt D, et al. Molecular profiling and clinical implications of patients with acute myeloid leukemia and extramedullary manifestations. J Hematol Oncol **15**: 60, 2022.
7. Bakst RL, Tallman MS, Douer D, et al. How I treat extramedullary acute myeloid leukemia. Blood **118**: 3785-3793, 2011.
8. Nemésio RA, Costa B, Abrantes C, et al. Myeloid sarcoma of the small intestine in a patient without overt acute myeloid leukaemia: a challenging diagnosis of a rare condition. BMJ Case Rep **2018**: bcr2017222718, 2018.
9. Cicilet S, Tom FK, Philip B, et al. Primary myeloid sarcoma of small bowel. BMJ Case Rep **2017**: bcr2017220503, 2017.
10. Wang P, Li Q, Zhang L, et al. A myeloid sarcoma involving the small intestine, kidneys, mesentery, and mesenteric lymph nodes: a case report and literature review. Medicine (Baltim) **96**: e7934, 2017.
11. Yamauchi K, Yasuda M. Comparison in treatments of nonleukemic granulocytic sarcoma: report of two cases and a review of 72 cases in the literature. Cancer **94**: 1739-1746, 2002.
12. Kheirkhah P, Avila-Rodriguez AM, Radzik B, et al. Gastrointestinal myeloid sarcoma: a case presentation and review of the literature. Mediterr J Hematol Infect Dis **13**: e2021067, 2021.
13. Hotta K, Kunieda K. Granulocytic sarcoma of the jejunum diagnosed by biopsies during double-balloon endoscopy before treatment (with video). Dig Endosc **25**: 468, 2013.
14. Hotta K, Kunieda K. Granulocytic sarcoma of the ileum observed by double-balloon endoscopy before treatment (with video). Gastrointest Endosc **79**: 166, 2014.
15. Ono K, Mikami Y, Hosoe N. Primary granulocytic sarcoma of the

small intestine diagnosed by single-balloon enteroscopy: a case report. Dig Endosc **32**: 436, 2020.
16. Nakanishi R, Henmi S, Marusawa H. Small bowel bleeding caused by myeloid sarcoma. Intern Med **61**: 123-124, 2022.
17. Minagi H, Kanaya N, Kondo Y, et al. Primary ileal myeloid sarcoma presenting with bowel obstruction: a case report. Surg Case Rep **10**: 229, 2024.
18. Soga K, Mukai H, Kitae H. Management of shiitake mushroom-induced ileus using balloon enteroscopy. Dig Endosc **34**: e58-e59, 2022.
19. Koshikawa Y, Nakase H, Matsuura M, et al. Ischemic enteritis with intestinal stenosis. Intest Res **14**: 89-95, 2016.
20. Ishioka M, Jin M, Matsuhashi T, et al. True primary enterolith treated by balloon-assisted enteroscopy. Intern Med **54**: 2439-2442, 2015.
21. Shozushima M, Ohata K, Nonaka K, Matsuhashi N. Shiitake mushroom-induced ileus managed using double-balloon enteroscopy. Endoscopy **45** Suppl 2 UCTN: E437, 2013.
22. Lim SW, Lee HL, Lee KN, et al. A case of myeloid sarcoma of intestine. Korean J Gastroenterol **68**: 148-151, 2016.
23. Kosti J, Mervak T, Terebelo H. Extramedullary myeloid leukemia in the setting of a myeloproliferative neoplasm. J Med Cases **13**: 561-568, 2022.
24. Avni B, Koren-Michowitz M. Myeloid sarcoma: current approach and therapeutic options. Ther Adv Hematol **2**: 309-316, 2011.
25. Tsimberidou AM, Kantarjian HM, Wen S, et al. Myeloid sarcoma is associated with superior event-free survival and overall survival compared with acute myeloid leukemia. Cancer **113**: 1370-1378, 2008.