Incidental subepithelial tumor in the terminal ileum

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Question: A 68-year-old woman was referred to Pusan National University Hospital for evaluation of a suspected subepithelial tumor (SET) in the terminal ileum, which was incidentally detected on screening colonoscopy. The patient had no past medical, surgical, or family history. She denied any GI symptoms, such as nausea, vomiting, diarrhea, constipation, bloody stool, or abdominal pain. Physical examination was unremarkable. Results of initial laboratory tests, including routine blood tests and tests for carcinoembryonic antigen and inflammatory markers, were within the normal range. Colonoscopy revealed a solid, round, yellowish-white SET covered with normal mucosa, measuring about 10 mm in diameter, at the terminal ileum (Fig. A). Endoscopic ultrasonography with a miniature probe showed a heterogeneous, hypoechoic mass, mainly located in the 3rd layer of the colonic wall, confirming that the lesion was SET. A subsequent bite-on-bite biopsy revealed chronic inflammation. Contrast-enhanced abdominal CT showed neither remarkable abnormal findings in the GI tract nor significant lymphadenopathy. We decided to perform endoscopic submucosal dissection (ESD) for accurate diagnosis, and en bloc resection was achieved successfully (Fig. B, yellow arrow: SET). Written informed consent was obtained. Based on above information, what is the most likely diagnosis?
Answer to the Images: Inflammatory Myofibroblastic Tumor of the Terminal Ileum

Histopathological examination of the ESD specimen revealed that the completely resected mass was located in the deep mucosal and submucosal layers and mainly composed of spindle-shaped myofibroblasts with fibrous stroma infiltrated by inflammatory cells, such as plasma cells and lymphocytes (Fig. C, H&E). Immunohistochemical staining was positive for smooth muscle actin but negative for S-100 protein, desmin, CD34, and anaplastic lymphoma kinase (ALK) (Fig. D, × 400). Mutations in c-KIT (exons 11, 9, 13, and 17) or PDGFRα (exons 12 and 18) were not identified. The tumor was finally diagnosed as inflammatory myofibroblastic tumor (IMT). During follow-up at 1 year and 3 years after ESD, the patient was asymptomatic and underwent follow-up colonoscopy and CT with no evidence of recurrence. IMT, previously named inflammatory pseudotumor, is a rare tumor characterized by solid neoplastic mesenchymal proliferation composed of myofibroblastic spindle cells admixed with a varying degree of inflammatory cell infiltrations (plasma cells, lymphocytes, and histiocytes) within a myxoid collagenous stroma. Immunohistochemical staining is positive for smooth muscle actin and vimentin, partially positive for desmin and cytokeratin, and negative for S-100 proteins, CD34, and myoglobin, resembling the characteristics of myofibroblasts. The exact etiology and histogenesis of IMT remain largely unknown. It most commonly affects the lung and abdominal cavity but may occur at any anatomic site. IMTs of the GI tract are seen most frequently in the stomach, followed by the small intestine, colon, and rarely the esophagus. To date, only few reports on IMTs in the terminal ileum have been published in the English literature. All cases presented with an obstructive symptom and surgically resected. However, in this case, the patient had no specific symptoms and was identified after a screening colonoscopy. Although IMTs are a biologically borderline mesenchymal neoplasm owing to a local recurrence and dis-
tant metastasis, they are generally benign, have a good prognosis, and do not require aggressive therapy. Complete resection is the principle treatment, but in unresectable cases, palliative therapy, such as radiotherapy and NSAIDs, can be applied. In the case of ALK rearrangement, crizotinib can be used.

In conclusion, to the best of our knowledge, this is the first case of IMT in the terminal ileum that was successfully treated endoscopically with ESD without need for further surgical intervention. IMTs should be included in the differential diagnosis of SET in the terminal ileum, especially when histological findings show spindle cell proliferation.

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CONFLICT OF INTEREST

No potential conflict of interest relevant to this article was reported.

AUTHOR CONTRIBUTION

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