Proctitis cystica profunda diagnosed by endoscopic submucosal dissection: use of a therapeutic procedure for diagnosis

A 54-year-old woman was referred to our gastroenterology unit after discovery of an atypical submucosal rectal lesion. She had developed massive hematochezia 1 month previously. Colonoscopy revealed 3-cm discrete submucosal lesions with superficial mucosal inflammation (Fig. 1). Pathologic analysis of a superficial biopsy specimen was inconclusive. Two successive rectal endoscopic ultrasonographic examinations were performed with a 1-month interval, and hypoechoic cystic lesions of the submucosal layer (third hyperechoic layer) were discovered (Fig. 2). All submucosal lesions had the same ultrasonographic features, but one exhibited a hyperechoic area with acoustic shadowing typical of calcification (Fig. 3). Endoscopic biopsies were again inconclusive, and endoscopic ultrasound guided-fine needle aspiration (EUS-FNA) was not performed because of the high risk of infection. Instead, we performed endoscopic submucosal dissection of one lesion to obtain a complete pathological analysis (Video 1, Fig. 4). A diagnosis of proctitis cystica profunda was made based on typical pathological features (Fig. 5 and Fig. 6).
Submucosal lesions are less frequent in the rectum than in the upper gastrointestinal tract, and both diagnosis and treatment can be challenging. EUS plays a key role in such situations [1], particularly for lesions with an intramural origin such as lipomas, leiomyomas, gastrointestinal stromal tumors, and neuroendocrine tumors. Intramural cystic lesions are more challenging to diagnose because of the risk of infection associated with transrectal EUS-FNA, and pathological analysis remains the gold standard. Proctitis cystica profunda is a rare condition that presents as a localized lesion or, diffusely, as multiple lesions [2, 3]. Cysts develop in the submucosa or muscularis mucosa. Older cysts often lack an epithelial lining and are surrounded by fibrous tissue and/or a polymorphic inflammatory infiltrate or hemosiderin. The material in the cysts may calcify [4]. Fibrosis is usually moderate, but can be significant in some cases. The presence of these lesions within the rectum is often associated with prolapse and solitary ulcer formation. Differential diagnoses include invasive mucinous carcinoma and, in women, endometriosis. EUS may be diagnostic for submucosal cysts associated with calcification. In summary, we have reported the first case involving one-stage endoscopic submucosal dissection for both pathological diagnosis and resection of proctitis cystica profunda, thus avoiding riskier surgery.

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References

Bibliography
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Fig. 6 Pathological analysis of submucosal cysts from the rectum of a 54-year-old woman. The material in the cysts may calcify. (hematoxylin erythrosine saffron stain, magnification ×25).