Colitis cystica profunda: a two-patient case series

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Abstract

Colitis cystica profunda (CCP) is a rare benign disease characterized by mucus-filled cysts in the submucosa, sometimes similar to malignant tumors. Endoscopic, radiological and even histological examinations are not highly specific, which can easily lead to missed diagnosis and misdiagnosis, resulting in unnecessary radical surgical resection. In this report, we present

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CONSENT

Written informed consent was obtained from the patient to publish this report in accordance with the journal's patient consent policy

CONFLICT OF INTEREST

The authors have no pertinent conflict of interest to report for this manuscript.

Abstract

Colitis cystica profunda (CCP) is a rare benign disease characterized by mucus-filled cysts in the submucosa, sometimes similar to malignant tumors. Endoscopic, radiological and even histological examinations are not highly specific, which can easily lead to missed diagnosis and misdiagnosis, resulting in unnecessary radical surgical resection. In this report, we present two cases of CCP with their clinical and imaging features.

Keywords

Rectum;Colitis cystica profunda;Endorectal ultrasound

INTRODUCTION

Submucosal cysts in the colon was first reported by Stark in 1766 during the autopsy of two specimens of patients with chronic dysentery. In 1863, Virchow named it colitis cystica polyposa for multiple polypoid

cystic submucosal lesions. In 1957, Goodall and Sinclair renamed colitis cystica polyposa to colitis cystica profunda (CCP), giving the name and detailed description of the disease firstly [1, 2]. CCP is a rare benign disease occurring in the rectum or colon, most commonly in the anterior wall of the rectum. The main pathological characteristics of CCP are mucus-containing cysts reaching deep into the submucosa, muscular layer or even the serous layer. The cysts are filled with large amounts of mucus, and floating exfoliated epithelial cells visible in the mucus, without the characteristics of malignant tumors[3]. There are few case reports concerning endorectal ultrasonography of CCP in previous literatures. We report two cases of CCP with the purpose of better awareness of its clinical and imaging features.

CASE REPORT

CASE 1

A 34-year-old male with defecation characteristics changed for more than 6 years, mainly manifested as mucous defecation, no increase in defecation frequency, no urgency, no abdominal pain and other discomfort. No special treatment was given, and now he came to the hospital for diagnosis and treatment.

Digital rectal examination (DRE): 5cm into the finger can reach the tough mass, smooth surface, movable, no blood staining in the receding finger sleeve. Laboratory examinations: Blood routine, liver and kidney function, tumor markers are all in normal range.

Endorectal ultrasound (ERUS): a cystic mass was observed at 70 mm from the anal verge, at the 12-2 o'clock direction in the lithotomy position, in the submucosal layer and intrinsic muscular layer of the rectum, with a size of 26 mm in diameter, clear boundary, regular morphology, protruding into the lumen, with poor internal transmission and heterogeneous echogenicity, and continuous echogenicity in the mucosal and serous layers of the rectum. Color Doppler flow imaging (CDFI): A few blood flow signals were observed in the periphery, but no obvious blood flow signals were observed in the interior of lesion (Figure 1).

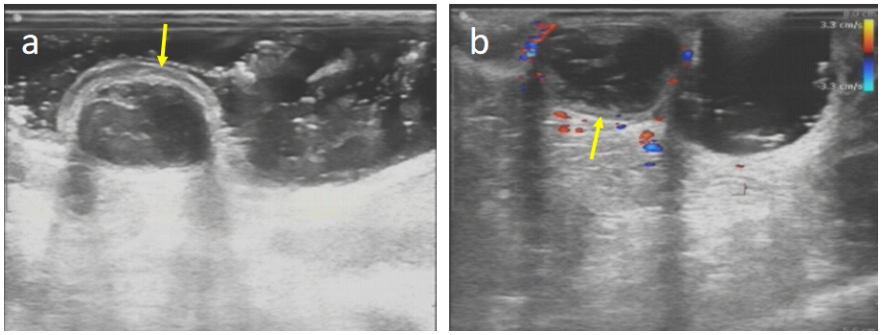

Fig. 1 Endorectal ultrasound a, Gray scale ultrasound showed a cystic mass in the submucosa, with clear boundary, poor internal transmission, and continuous echogenicity in the mucosal layer (yellow arrow). b, Color Doppler ultrasound showed a small amount of blood flow signal around the cystic mass, no obvious blood flow signal inside, and continuous echogenicity in the serous layer of the intestinal wall (yellow arrow).

Contrast-enhanced computed tomography (CECT): A round-like low-density lesion with a size of 3.1x2.4x2.2cm was observed in the submucosa of the anterior wall of the middle rectum, with calcification on the edge and no obvious enhancement. The rectal mucosa was smooth, the rectal wall was intact, the surrounding fatty space was clear, and no enlarged lymph nodes were seen (Figure 2).

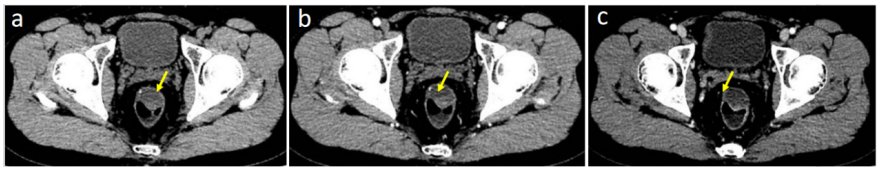

Fig. 2 Contrast enhanced CT. a, A quasi-circular low-density foci on plain scan, with smooth rectal mucosa and intact intestinal wall. b-c, No enhancement was seen in the arterial and venous phases of the lesion.

Contrast-enhanced magnetic resonance imaging (CEMRI): a round-like signal foci with a size of about 2.3x1.8x1.7cm was seen in the submucosa of the anterior wall of the middle rectum, T1 showed low signal, T2 showed high signal, DWI showed high signal, ADC showed no reduction, no obvious enhancement of the foci in contrast enhanced scanning. The rectal mucosa was smooth, the rectal wall was intact, the surrounding fatty space was clear, and no obvious enlarged lymph nodes were seen in the pelvic cavity (Figure 3).

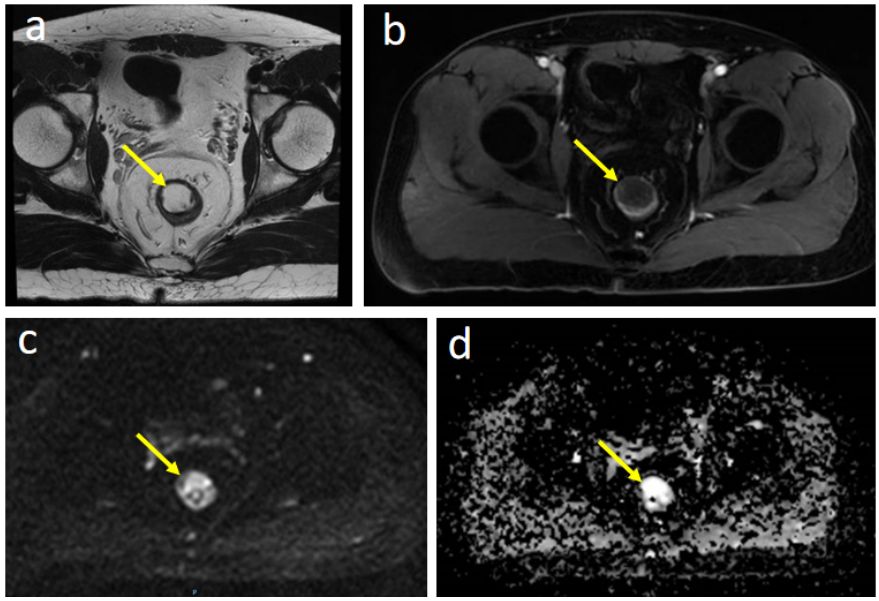

Fig. 3 Contrast-enhanced MRI. a, T2WI showed high signal. b, enhancement showed slight enhancement in the periphery of the lesion and no significant enhancement in the interior. c, DWI showed high signal. d, ADC showed high signal.

Finally, the patient underwent transanal endoscopic resection of rectal mass, and the postoperative pathology was as follows: multiple mucous lakes could be seen in the rectal intestinal wall, locally covered by intestinal epithelium, no atypia, and the laminar propria interstitials could be seen, which was consistent with CCP (Figure 4).

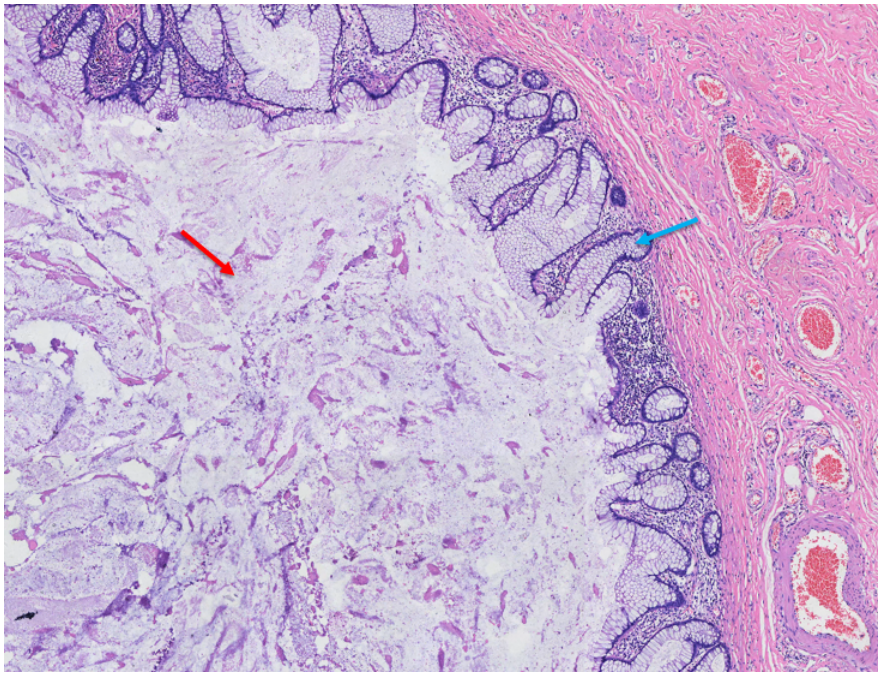

Fig. 4 Pathology. Multifocal mucus paste (red arrows) visible in the intestinal wall, locally covered with intestinal epithelium without heterogeneous hyperplasia (blue arrows).

CASE 2

A 27 years old female patient had a change in stool habits and traits for more than 2 years, which showed that the frequency of defecation increased to 5-6 times a day, and the stool became slightly thinner, accompanied by a feeling of urgency and incompleteness of the stool, and mucus on the surface of the stool. Colonoscopy was performed at a local hospital, which showed that a mucosal eminence of about 2 × 2.5cm in the rectum 2-4cm from the anus. Biopsy under colonoscopy showed chronic inflammation of the mucosa with erosion. No further treatment was performed and no follow-up examination was conducted. Recently, the stool surface was mixed with blood, so he came to the hospital for diagnosis and treatment.

DRE: A tough mass about 2-3cm in diameter could be detected on the anterior wall of the rectum about 6cm from the anus, with moderate mobility, slight tenderness, and no blood staining after exiting the finger sleeve. Laboratory examination: Blood routine, liver and kidney function, tumor markers were all in normal range.

ERUS: a cystic mass was seen in the submucosal layer of the rectum at 10-12 o'clock in the lithotomy position about 56 mm from the anal verge, with size of 26 mm in diameter, clear borders, regular morphology, intact mucosal layer, intact and continuous intrinsic muscular layer and serous layer. CDFI: a small amount of blood flow signal was seen in the periphery, and no significant blood flow signal was seen inside (Figure 5).

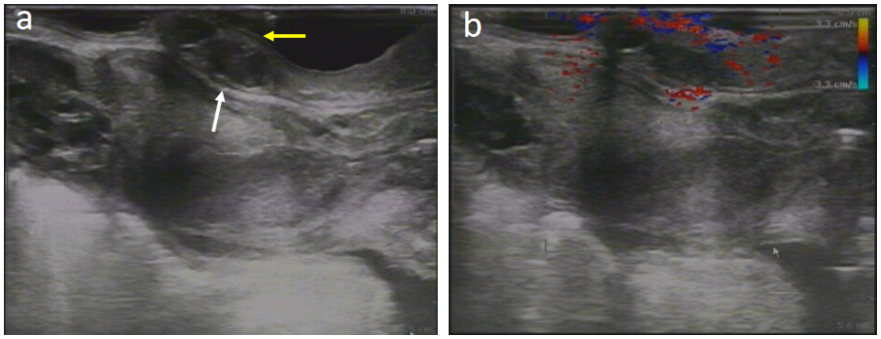

Fig. 5 Endorectal ultrasound. a, Gray scale ultrasound showed a cystic mass in the submucosal layer with clear borders and poor internal transmission, and continuous echogenicity in the mucosal layer (yellow arrow) and serous layer (white arrow). b, CDFI showed a small amount of blood flow signal around the cystic mass, but no obvious blood flow signal inside.

CECT: The anterior wall of the lower part of the rectum at about 50mm from the anal margin, presented a local protuberant change, with a local thickness of about 11mm, and the mucosal surface was smooth. The enhancement of the local submucosal area was not obvious, and a patchy dense shadow was still visible. The perienteric fatty space was clear (Figure 6).

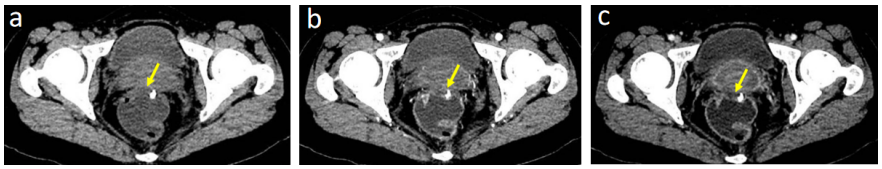

Fig. 6 Contrast-enhanced CT. a, Localized thickening and eminence of the anterior wall of the lower rectum in plain scan, with visible calcification. b-c,No significant enhancement was seen in the arterial and venous phase.

CEMRI: An oval abnormal signal foci was seen in the submucosa of the anterior wall of the lower segment of the rectum about 46 mm away from the anal verge, with a size of 17 mm×18 mm×20 mm; T2WI showed high signal and multiple low signal separations within it; No obvious limited diffusion was observed on DWI and ADC images; separation enhancement was observed on enhanced scan, and no clear enhancement was observed in the remaining lesions; the boundary of the lesion was clear, and the outer wall of the lesion was smooth (Figure 7).

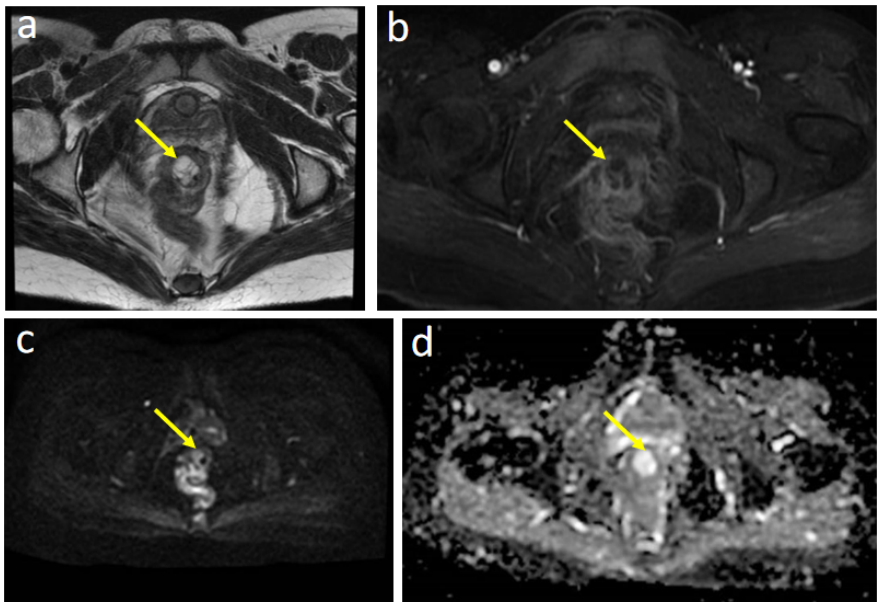

Fig. 7 Contrast-enhanced MRI. a, T2WI showed high signal intensity and multiple low signal separations. b, separate enhancement was presented in the lesion. C,DWI showed high signal (low signal area was calcification). d,ADC showed high signal without any decrease.

Colonoscopy: A size of 20 mm × 18mm protuberant mass was seen on the anterior wall about 4cm from the anal verge, with superficial ulcer on the surface. Endoscopic resection of rectal submucosal mass was performed, and the submucosal incision showed gelatinous appearance (Figure 8). Postoperative pathology: colitis cystica profunda of the rectum with calcification (Figure 9).

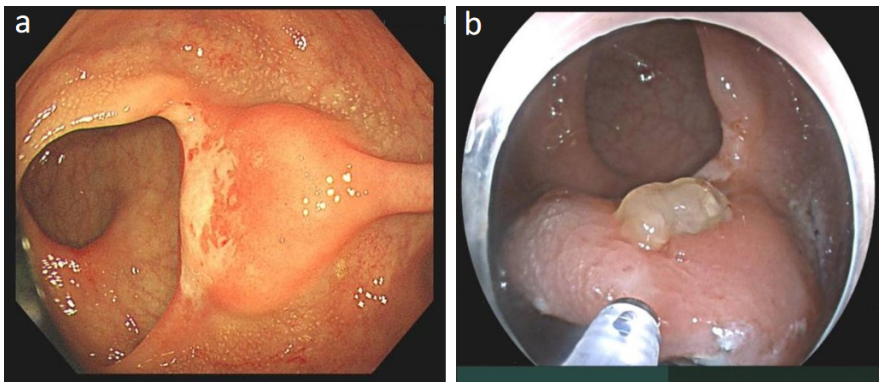

Fig. 8 Colonoscopy. a, A protuberant mass showing the formation of superficial ulcers. b, gelatinous mucus can be seen by incision of submucosa.

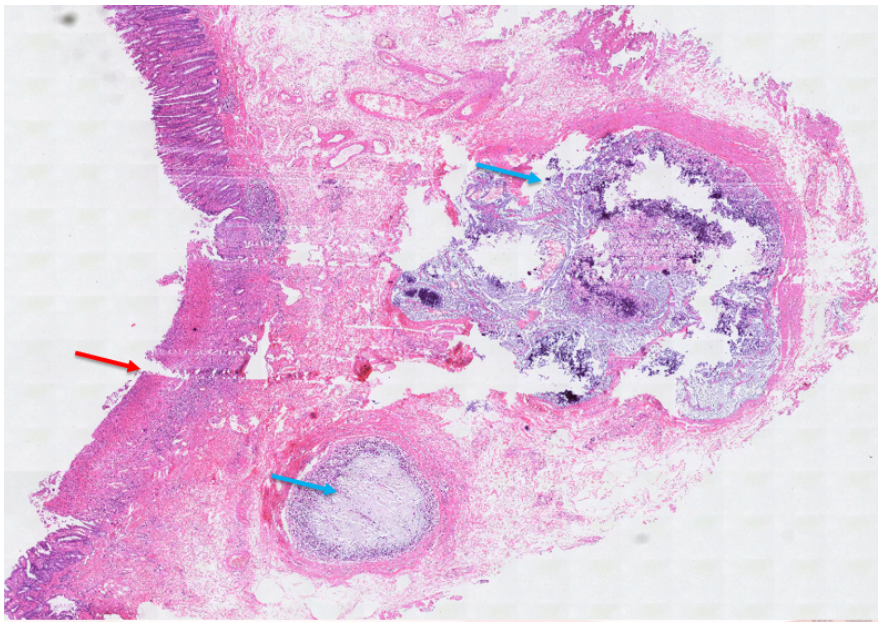

Fig. 9 Pathology. local erosion of intestinal mucosa, partly with ulcerative repair changes (red arrow), and a focal cystic cavity containing a large amount of mucus in the submucosa (blue arrow).

DISCUSSION

Cystica profunda is a rare benign non-neoplastic disease that can occur throughout the gastrointestinal tract, mostly in the rectum, followed by the colon, and more rarely in the stomach and small intestine. It is also known as gastritis cystica profunda (GCP), enteritis cystica profunda (ECP), and colitis cystica profunda (CCP)[4]. CCP is a rare benign disease occurring in the rectum or colon, most commonly in the anterior wall of the rectum. The pathogenesis of CCP is unknown and is thought to be due to congenital or acquired mucosal muscle weakness caused by inflammation, infection, trauma or ischemia resulting in embedding of the mucosal epithelium in the submucosa [5]. The main histological features are the presence of multiple cysts of varying size and morphology in the submucosa lined with a single layer of flattened or columnar epithelial cells, which are filled with mucus, and the disappearance of some of the covered epithelium of the cyst wall, forming a mucus lake[6].

CCP can occur at any age, most common in young and middle-aged people aged 30-40 years, and is more common in men. The clinical manifestations of CCP are diverse and nonspecific, mainly including abdominal pain, diarrhea, constipation, blood in stool, mucus stool, urgency, rectal pain, change in stool habit, internal rotation, abdominal mass and intestinal obstruction [7-9]. According to the extent of invasion, there are diffuse and local types[10]. The diffuse type involves the entire colon with villous or tipped polypoid lesions or even ulcers, mostly due to intestinal inflammation and ulceration. It is associated with Crohn's disease, ulcerative colitis, infectious colitis and radiation enteritis[11,12]. The local type is mostly seen in the anterior wall of the rectal 5-12 cm from the anal verge, presenting as nodules or polyps, which is associated with rectal prolapse and isolated rectal ulcer syndrome. The local type is the most common, while the diffuse type accounts for less than 15% of cases reported in the literature [1,3,13,14]. Both of the two cases in this paper were young and middle-aged patients with clinical characteristics of mucous excretion. The locations of all cases were located in the anterior rectal wall of 5-12cm from the anal verge, which was consistent with the location reported in the literature. Both cases showed nodularity and belonged to the local type.

The imaging findings of CCP are characteristic. X-rays are normal or nonspecific in the early stage, or show luminal narrowing and irregularity due to multiple submucosal cystic structures. Barium enema shows single or multiple filling defects or shows thickened folds. CT images show a non-infiltrating submucosal mass with

well-defined borders and an unenhanced cystic lumen of variable sizes. The perirectal adipose tissue was absent and the levator anus muscle was thickened[2,15]. MRI showed a homogeneous low signal on T1WI and a submucosal high signal nodule on T2WI, with no significant enhancement on contrast-enhanced images, high signal on DWI, no diffusion restriction on ADC, and significant high signal on T2WI suggesting that the presence of mucin in the lesion[4,16]. In both cases in this paper, CT showed non-enhancing hypodense lesions with clear borders, calcifications at the margins, smooth mucosa, intact rectal wall, clear surrounding fatty spaces, and no enlarged lymph nodes. MRI presentations were also consistent with the above imaging manifestations. However, the thickening of the levator anus muscle described in the literature was not present. In addition, the presence of calcification of the cyst wall was described in our case, which suggests a chronic benign disease and not associated with rectal cancer where calcification usually inside the tumor [17].

Colonoscopy in CCP shows tipped or villous polypoid lesions covered by normal, edematous or ulcerated mucosa, or shows polypoid or nodular mucosal thickening with or without ulceration, which can be easily confused with colorectal cancer [8]. In contrast, colonoscopy allows biopsy and therefore has the dual advantage of obtaining both imaging and pathology at the same time. However, due to insufficient depth of biopsy and limited sampling, it is easy to missed diagnosis and misdiagnosis. In case 2, the results of the first external colonoscopy biopsy only showed chronic inflammation of the mucosa with erosion, which may be related to the shallow and limited sampling.

ERUS showed multiple hypoechoic or anechoic cysts in the submucosal layer of the rectum with a small amount of internal echogenicity, no involvement of the mucosal layer, no lymphadenopathy, and no penetration or invasion of the muscular layer beyond the submucosal layer[4,9]. In this paper, ERUS showed cystic lesions in the submucosal layer in both cases, with the mucosal and serous layers intact. The difference between the two cases is that the lesion was located in the submucosal layer and the intrinsic muscular layer, with invasion of the muscular layer in case 1. While in case 2, the lesion was located only in the submucosa, and the intrinsic muscle layer was not invaded outside the submucosa. ERUS can clearly localize the different layers of the intestinal wall and can assess whether the mucosal layer, intrinsic muscular layer, and serous layer of the intestinal wall are intact. Although ERUS cannot confirm the diagnosis, it is of great importance to exclude malignant tumors in the deeper layers of the intestinal wall. Moreover, ERUS is safe, noninvasive, nonradioactive, easy to perform, well tolerated, and can be repeated as a follow-up examination.

CCP is similar to benign and malignant colorectal tumors and inflammatory bowel disease, and the diagnosis of CCP also requires the identification of any colon or rectal polypoid or intramural benign or malignant masses (adenomatous polyps, polypoid inflammatory granulomas, leiomyomas, lipomas, adenocarcinomas, mucinous carcinomas, sarcomas), inflammatory lesions (ulcerative colitis, Crohn's disease, and ischemic colitis or proctitis), and endometriosis [10].

The goal of treatment for CCP is to prevent or reduce symptoms. Depending on the severity of symptoms, treatment can be either conservative medical or surgical. Patient education and behavior modification are the primary treatments for patients with CCP. Conservative treatment includes bowel habits, postural correction, avoidance of stress and strain, and increased dietary fiber can alleviate symptoms in the majority of patients. Conservative treatment is ineffective and surgical resection is required when symptoms are persistent or severe, such as bowel obstruction, bleeding, or rectal prolapse[18,19]. In recent years, endoscopic submucosal dissection (ESD), with the advantages of rapid recovery, minimal trauma, and preservation of colonic integrity, has been widely used in the treatment of CCP [20].

In conclusion, CCP is a rare disease that is difficult to diagnose and easily misdiagnosed as other occupying lesions of the intestine because of its low incidence, lack of specificity in clinical and endoscopic manifestations, and lack of awareness among physicians in various disciplines. Once misdiagnosed, it not only brings unnecessary painful surgical resection to patients, but also poses risks to physicians. In contrast, imaging study plays a crucial role in both disease detection and differentiation from malignant diseases, helping to initially determine the benignity and malignancy of the lesion and avoiding erroneous radical resection surgery.

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CONFLICT OF INTEREST

The authors have no pertinent conflict of interest to report for this manuscript.

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