

Endoscopic ultrasonography and rectal duplication cyst in an adult

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ABSTRACT

Rectal duplication cysts account for 4% of all duplications of the alimentary tract. Presentation in adulthood is rare. An asymptomatic 54-year-old man was referred for endoscopic colorectal cancer screening. A bulging mass covered by normal mucosa was identified in the rectum. Endoscopic ultrasonography (EUS) with fine needle aspiration (FNA) was made for a diagnosis of rectal duplication cyst. The patient was operated and the diagnosis was confirmed. The diagnosis of the rectal duplication cyst is a challenge. EUS may have a singular role when identifying a muscular layer, because this is the only absolutely necessary criterion for the diagnosis. FNA by EUS may eventually identify colorectal and/or heterotypic epithelium that are the other diagnostic criteria of the duplication cyst.

Key words: Endoscopic ultrasonography (EUS), fine needle aspiration (FNA), rectal duplication cyst

INTRODUCTION

Duplications of the alimentary tract are rare congenital malformations. Rectal duplication cysts account for 4% of all duplications. They are seen in childhood more frequently and presentation in adulthood is rare. The diagnosis can be a challenge.

CASE REPORT

An asymptomatic 54-year-old man was referred for endoscopic colorectal cancer screening. A bulging mass covered by normal mucosa was identified in the rectum [Figure 1].

An endoscopic ultrasonography (EUS) with fine needle aspiration (FNA) was performed [Figure 2]; between 5 cm and 8 cm from the anal verge, it was found to be an extrinsic compression of the rectal wall in relation with a heterogeneous lesion, 43 mm × 35 mm, cystic in the major part with various anechoic cavities separated by septa of different thickness; some of them was very thick that mimicked echogenic solid components; at the luminal border some parts of the muscular layer of the rectal wall were involved, and it was possible to identify

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that this layer was duplicated as well and involved the lesion partially; the submucosa and mucosa of the rectal wall were preserved. The lesion was well-delimited, although, parts of the contralateral borders were irregular. It did not involve any perirectal structure. No adenopathies were identified.

Our first diagnostic possibility was a rectal duplication cyst; but we could not exclude an eventual malignant degeneration because of the existence of solid components, that is, thick septa. Consequently, we performed FNA by EUS with a 22-gauge needle (30 min after the administration of 200 mg ciprofloxacin intravenously). The aspirated material was white colored and thick. Cytological examination revealed the presence of mucus, containing isolated cells with vacuolated macrophage-type cytoplasm and groups of cylindrical epithelial cells without features of malignancy compatible with colorectal mucosal cells [Figure 3].

The patient was submitted to surgery. We found that the lesion was involving the lateral wall of the rectum and an *en bloc* excision of the lesion and the lateral wall of the rectum was performed. The patient recovered without complications.

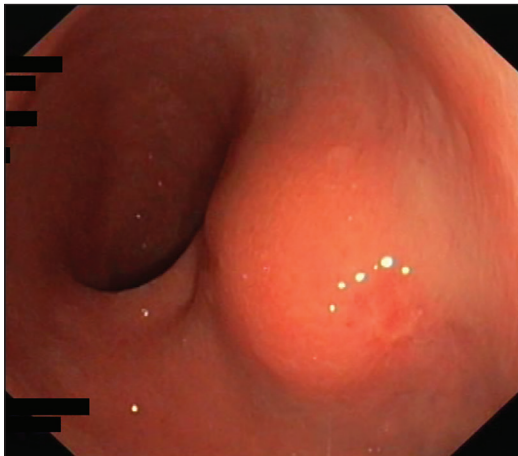


Figure 1. Colonoscopy. Bulging mass covered by normal mucosa

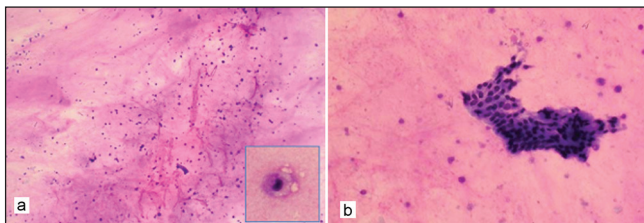


Figure 3. Cytological examination. (a) Mucus with multiple isolated cells (Giemsa stain, 100×) – macrophages [right inferior corner (Giemsa stain, 400×)] (b) Group of cylindrical epithelial cells compatible with colorectal mucosal cells (Giemsa stain, 200×)

The macroscopic examination of the excised specimen, with 5 cm × 5 cm × 2 cm, showed the presence of the rectal mucosa, in a small area of its external surface; dissection revealed an irregular cavity with mucus-type material; no communication between the external surface and the cavity was found.

The histological examination revealed [Figure 4] the presence of mucus and, in part of the internal surface, a colorectal mucosal lining, which lay on a smooth and well-defined muscular layer. This layer was in contiguity with the rectum muscular layer, only separated by a thin connective tissue. Externally, the rectal mucosa was identified. In

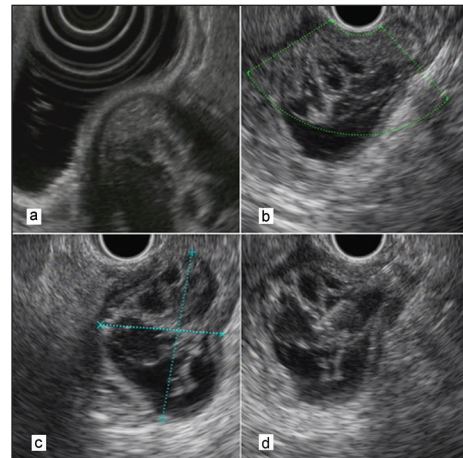


Figure 2. Endoscopic ultrasonography. (a) Extrinsic compression of the rectal wall with preservation of the submucosa and mucosa (b) Duplication of the muscular layer (c) Anechoic cavities separated by different septa of different thickness (d) Fine needle aspiration (FNA)

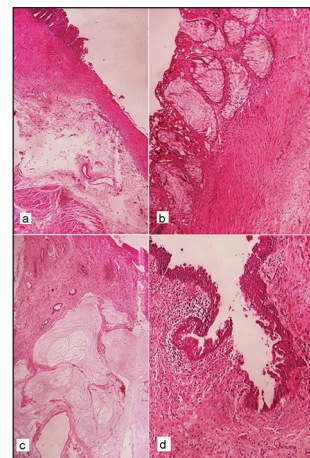


Figure 4. Histological examination. (a) Colorectal mucosal lining that lays on a smooth muscular layer in contiguity with the rectum muscular layer (HE stain, 40×) (b) Colorectal mucosal lining and muscular layer of the cystic wall (HE stain, 100×) (c) Free mucus in the connective tissue (HE stain, 40×) (d) Stratified epithelium in the cystic lining (HE stain, 100×)

another part of the lesion's internal surface, we found a stratified epithelium and some acinar glandular structures that could be considered heterotypic finding. There was no evidence of malignancy. The diagnosis of rectal duplication was made.

DISCUSSION

Rectal duplication cysts are extremely rare. In most cases, they occur in childhood, with rectal bleeding, rectal pain, painful defecation, tenesmus, constipation, prolapse, urinary retention, fistulization, or infection.^[1] The clinical presentation in adulthood is variable, depending on its size and consequent mass effect or developed complications.^[2] They can be asymptomatic as well and be accidentally found during digital rectal examination such as a hard and smooth mass bulging into the rectal lumen.

In our case, the identification of this lesion was an incidental finding during a colonoscopic examination performed by cancer screening. The diagnosis of rectal duplication cyst is classically considered a difficult one. There are various reasons for this fact. Clinically, as mentioned before, they can be asymptomatic or manifested through different clinical ways. There are no imaging means that provide specific images and there are different diagnostic possibilities, such as: Teratoma, meningocele, leiomyosarcoma, retrorectal pyogenic abscess, anal duct or gland cyst, and subperitoneal pelvic cyst lymphangioma.^[2,3]

Among the auxiliary means of diagnosis, we have found some references to the use of endorectal ultrasound in the diagnosis of rectal cystic lesions.^[4-7] In none of these cases did we find references to an eventual identification of duplication of the *muscularis propria* or to the fact that the lesion shared the muscular layer of the rectal wall. In our case, it was possible to identify these two circumstances, which allowed a strong diagnostic possibility of the rectal duplication cyst to be considered. In spite of these findings, we chose to perform FNA by EUS, because we could not exclude an eventual malignant component in relation with the thick septum. In literature, we have found three cases of performed FNA, two of them guided under computed tomography (CT) visualization^[8,9] and one under endorectal ultrasound.^[3] In two cases the procedure was useful, because in both cases there was a strong

suspicion of neoplastic lesions: Carcinoid^[8] and mucinous.^[9] In our case, the cytology showed mucus and benign epithelial cells; these features supporting a cystic nature of the mass.

As a general rule in those situations, the patient was operated.^[2,3] Due to the cytological information of the absence of malignancy, only the excision of the lesion was performed, because if malignant degeneration was suspected, total excision, including the normal rectum, should be considered. The surgery confirmed the finding of EUS that the lesion shared a small part of *muscularis propria* of the rectal wall that did not allow its preservation and that small part was, therefore, resected.

CONCLUSION

In summary, the diagnosis of rectal duplication cyst is still a challenge, but we believe that EUS, with or without FNA, may have a single role in this diagnosis when identifying a muscular layer, due to the fact that this is the only absolutely necessary criterion for the diagnosis.^[10] We think FNA by EUS that can be performed during the same procedure may be an important step because it may identify colorectal and/or heterotypic epithelium that are the other diagnostic criteria of the duplication cyst. Furthermore, if it does not show malignancy, it can influence the surgical option, allowing a more conservative surgery, with rectal preservation, such as in our case.

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Conflicts of interest

There are no conflicts of interest.

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