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Case report

PEComa in the rectum: A case report and review of the literature on epithelioid angiomyolipoma

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ABSTRACT

Introduction and importance: Perivascular epithelioid cell tumor (PEComa), especially angiomyolipoma (AML) is a rare mesenchymal tumor in wide array of anatomic locations such as the kidney, lung, uterus, and gastrointestinal tract. AML is commonly found in the kidneys and classified as a classic AML or epithelioid AML. We report a case of epithelioid AML diagnosed in the rectum, treated by robot assisted low anterior resection.

Presentation of case: A 45-year-old woman was referred to our hospital because when an intramural rectal mass was detected on a colonoscopic examination performed during a regular health checkup. Colonoscopic examination revealed an intramural mass, 2 cm in diameter, bulging in the rectal wall, 6 cm from the anal verge. Based on abdominal and pelvic computed tomography images and magnetic resonance imaging findings, the patient was suspected of having gastrointestinal stromal tumor of the rectum. The patient was treated by robot assisted low anterior resection under the diagnosis of GIST. The patient improved without any postoperative complication and was diagnosed as epithelioid AML, a type of PEComa.

Discussion: AML diagnosed in gastrointestinal tract is very rare and among them, epithelioid AML has possibility of malignancy. However, confirmed diagnosis before surgical resection is difficult because PEComa shows nonspecific imaging characteristics. Treatment of choice of epithelioid angiomyolipoma is surgical resection.

Conclusion: Because epithelioid AML has the potential for malignancy, clinicians must be aware of the knowledge of the characteristics and natural history of epithelioid AML.

1. Introduction and importance

Perivascular epithelioid cell tumor (PEComa) is a mesenchymal tumor and has characteristic perivascular epithelioid cells. PEComa is a rare tumor occurred anywhere [1]. Family of PEComas includes angiomyolipoma (AML), clear-cell "sugar" tumor (CCST), lymphangioleiomyomatosis (LAM), clear-cell myomelanocytic tumor of the falciform ligament/ligamentum teres (CCMMT) and rare clear-cell tumors of wide array of anatomic locations such as the uterus, colon, and soft tissues [2]. AML is the most common type of PEComa and the kidneys are one of the most common locations where AML is diagnosed. AML is categorized into two types, mainly the classic type and approximately 7% of AML cases, epithelioid type [3]. We report a case of epithelioid AML arising in the rectum that was clinically diagnosed clinically as a gastrointestinal stromal tumor (GIST). The report has been arranged in line with the Updating Consensus Surgical Case Report (SCARE 2020) guidelines [4].

2. Case presentation

A 45-year-old woman was referred to our hospital when an intramural rectal mass was detected on a colonoscopic examination performed during a regular health checkup. She had diabetes mellitus and was taking tamoxifen for left breast cancer after breast-conserving surgery and radiation therapy. There were no family and psychosocial history. The patient was not present any symptoms and abnormality on physical examinations. Colonoscopic examination revealed an intramural mass, 2 cm in diameter, bulging in the rectal wall, 6 cm from the anal verge (Fig. 1). The mass was well-defined and homogeneous on abdominal and pelvis computed tomography (Fig. 2). Magnetic resonance imaging in our hospital revealed a well-demarcated, homogeneous mass in the left lateral rectal wall (Fig. 3). Tumor markers, such as carcinoembryonic antigen (CEA) and CA19-9, were normal. The patient underwent robot-assisted low anterior resection by specialist of

Abbreviations: CT, computed tomography; GIST, gastrointestinal stromal tumor; PEComa, perivascular epithelioid cell tumor; AML, angiomyolipoma.

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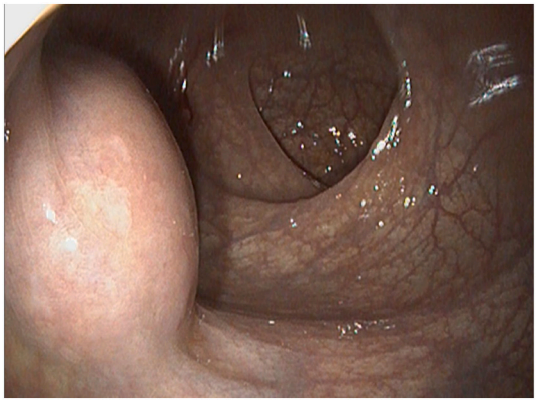

Fig. 1. Pre-operative colonoscopic imaging. 2 cm-sized intramural mass, bulging in the rectal wall.

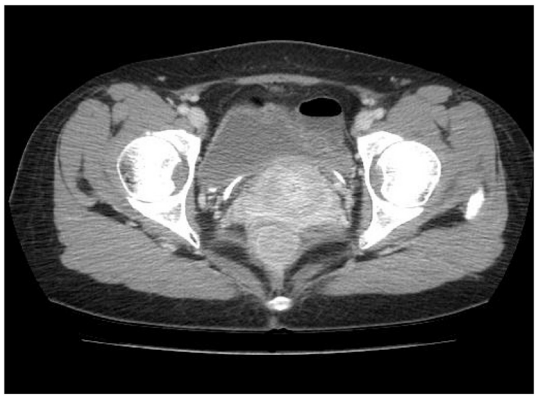

Fig. 2. Pre-operative CT imaging. Well-defined and homogenous mass in rectum.

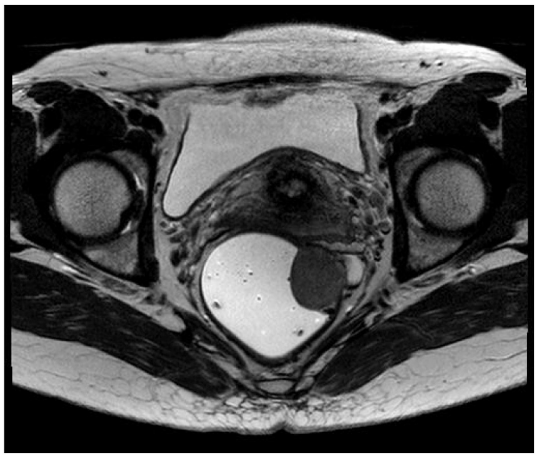

Fig. 3. Pre-operative MRI imaging. Well-defined and Homogenous mass in the left lateral rectal wall.

colorectal surgery for a suspected diagnosis of GIST of rectum. Operation time was 265 min and estimated blood loss was 30 cc. One drainage tube was inserted and removed 5 days after surgery. The patient improved without any postoperative complication and discharged to home at postoperative day 7. No metastasis and recurrence were not observed on abdominal pelvic CT taken 2 months, 8 months and 15 months after surgery.

On macroscopic examination, a well-circumscribed protruding mass was found occupying the submucosa. Microscopic examination revealed perivascular epithelioid cells were attached to the tortuous, irregular and thick-walled blood vessels. The epithelioid cells had abundant cytoplasm and prominent nuclei. Immunohistochemistry showed a diffuse positive reaction for HMB45 (Fig. 4). Therefore, diagnosis of the patient was corrected with perivascular epithelioid cell tumor, especially epithelioid angiomyolipoma.

The patient has been followed up for 2 years using abdomen & pelvis CT and recurrence or metastasis has not found.

3. Discussion

PEComa is rare mesenchymal tumor that has been reported in several organs such as the kidney, lung, and uterus [5]. PEComa rarely occurs in the gastrointestinal tract and commonly present with non-specific clinical signs [2,6]. Depending on the organ and the size of the tumor, clinical presentations include abdominal pain, melena, rectal bleeding, obstruction and sometimes a patient can also be asymptomatic [2]. PEComa can also results in intussusception [8]. A diagnosis of PEComa should be differentiated into malignant GIST, malignant melanomas, sarcomas, and paragangliomas [7]. However, differential diagnosis before surgical resection is difficult because PEComa shows nonspecific imaging characteristics [2]. CT is an effective tools of imaging, and the tumor appears as a mass with well-defined margin and heterogeneous or homogeneous enhancement [2,7]. On magnetic resonance imaging (MRI), the lesions show bright on T1-weighted images and dark on fat-suppressed images [2]. Ultrasonography may represent a well-defined hyperechoic, heterogeneous mass [2,7].

Angiomyolipoma(AML) is the most common type of PEComa. Renal AML is common, and a small number of AML findings in other locations have also been described. AML is categorized into two types: classic AML and epithelioid AML. Classic AML present tortuous and thick-walled blood vessels, and perivascular epithelioid cells have abundant lipid resembling fat. It is presented that smooth muscle-like spindle cells are irregularly arranged and a clear eosinophilic component arranges around blood vessels [3]. Epithelioid AML is composed partly or entirely of atypical large epithelioid cells with abundant cytoplasm, vesicular nuclei and prominent nucleoli. It has immunohistochemical characteristics that are positive for melanocytic markers such as HMB-45, HMB-50, and smooth muscle markers due to the presence of epithelioid cells [8]. Epithelioid AML is more frequently malignant and metastasis within the abdomen or lung than classic AML [5]. Therefore, all patients diagnosed with eithelioid AML should be closely monitored using imaging tools [3]. The criteria that can distinguish malignant AML are as follows: (1) 70% or more of atypical epithelioid cells, (2) two or more mitotic figures per 10 high-power fields (HPF), (3) atypical mitotic figures, and (4) necrosis [3].

Surgical resection is the treatment of choice for AML [8,9]. However, insufficient resection may result in rapid local recurrence [9]. Most patients undergo laparotomy or laparoscopic surgical procedures [2]. However, surgery might not be adequate in some of patients with local recurrence and distal metastases [8]. While chemotherapy could be used in these cases, the effectiveness remains unclear [2]. There were a small number of case reports of administration of interferon-alpha for PEComa in colon and bladder, but its effectiveness has not been clearly demonstrated [10,11]. The patient in this study did not receive chemotherapy.

PEComa and AML are difficult to diagnose clinically as they do not have distinctive imaging and clinical characteristics. Therefore, surgical removal and biopsy are the only methods of confirming PEComa and AML [8]. We recommend that clinicians must keep in mind of the characteristics of PEComa and AML, and also about the potential for malignancy of these patients. Clinical progress in such patients should be carefully monitored.

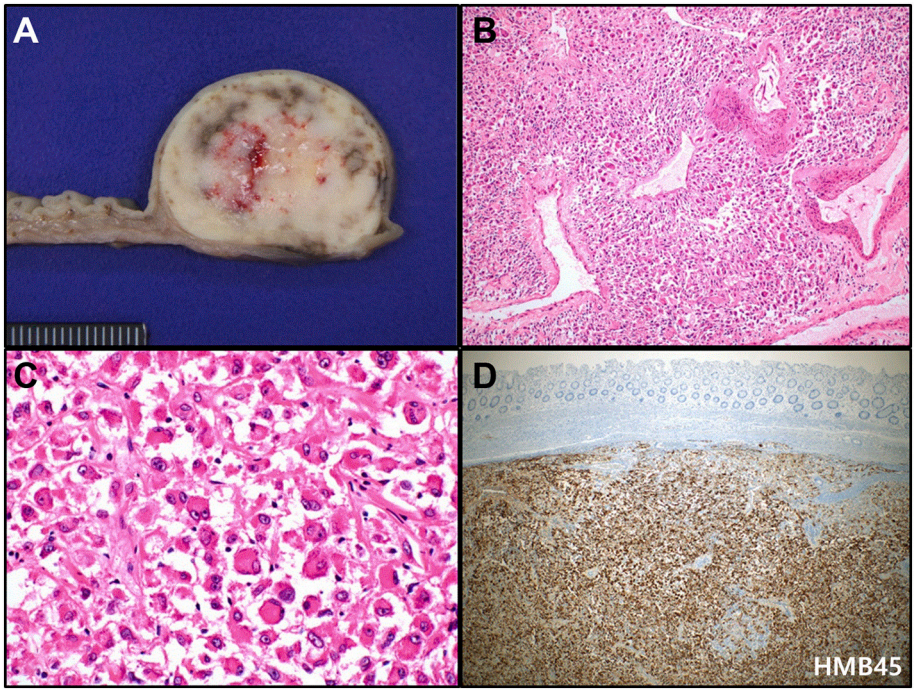

Fig. 4. Postoperative pathologic imaging. (A) Well-circumscribed protruding mass occupying the submucosa. (B) Perivascular epithelioid cells attached to the tortuous, irregular and thick-walled blood vessels. (C) Epithelioid cell having abundant cytoplasm and prominent nuclei. (D) Diffuse positive reaction for HMB45 in immunohistochemistry.

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None.

Ethical approval

Ethical approval was done at IRB (Institutional review board).
IRB file number is 2021-04-020.

Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. No information that would enable his identification has been provided.

Author's contribution

Hee Jin Yeon, Nak Song Sung, Won Jun Choi and Seung Jae Roh designed the study. Nak Song Sung performed the surgery. Hee Jin Yeon and Seung Jae Roh performed data and evidence collection. Yong Wook Park reviewed the pathology specimens.

Research registration

Not applicable.

Guarantor

Nak Song Sung.

Provenance and peer review

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Declaration of competing interest

None.

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