Case Report

Rectal gastrointestinal stromal tumor as an incidental finding in a patient with rectal polyps

Yong Zhou1, Xu-Dong Wu2, Ren-Gen Fan1, Wen-Zhang zha1, Yong-Hua Xu1, Cheng-Lin Qing1, Jing Jia3

1Department of General Surgery, 2Department of Gastroenterology, 3Department of Nephrology, Yancheng City First People’s Hospital, Yancheng 224005, China

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Abstract: A patient who was diagnosed as rectal polyps in the local hospital went to our hospital for surgical treatment. Abdominal CT demonstrated a large irregular extra-luminal tumor of at least 5 cm cross-section on the ventral side of the lower rectal wall. Intraoperatively, a large irregular extra-luminal tumor (about 5×4.5×4 cm) was found. Anterior resection with end colostomy and rectal stump (Hartmann’s procedure) was performed. Postoperative histological examination showed simultaneous development of rectal GIST and polyps.

Keywords: Rectal polyps, GISTs, simultaneous development, KIT mutation

Introduction

Rectal gastrointestinal stromal tumors (GISTs) and polyps are distinct neoplasms originating from different cell layers. Although rectal polyps constitute the most common type of rectal benign tumor, cases of simultaneous development of a GIST are rare because of the low incidence of rectal GISTs. Here, we present and discuss a case of rectal gastrointestinal stromal tumor as an incidental finding in a patient with rectal polyps.

Case report

A 66-year-old man without significant medical history presented to our hospital with occasional rectal bleeding for thirteen months. A proctoscopy made in the local hospital revealed rectal polyps (the largest 2.5×2 cm) (Figure 1). Histopathologic examination revealed tubular adenoma with lower-grade dysplasia. Digital investigation of the rectum revealed a mass of approximately 2.5 cm in diameter on the left lateral rectal wall at about 4 cm above the dentate line. The mass was soft, elastic and mobile. Rectal wall below the mass was hard, with an irregular surface. Furthermore, blood was found on the exploratory finger.

Abdominal CT was performed, which demonstrated a large irregular extra-luminal tumor of at least 5 cm cross-section on the ventral side of the lower rectal wall (Figure 2).

Intraoperatively, a polyp (about 2.5×2 cm) and some small polyps attached to the rectal wall were detected. Furthermore, a large irregular extra-luminal tumor (about 5×4.5×4 cm) was found arising from the ventral side of the rectal wall located about 5 cm from the anal margin with few enlarged regional lymph nodes present. The patient was diagnosed presumptively with simultaneous development of rectal cancer and rectal polyps.

Anterior resection with end colostomy and rectal stump (Hartmann’s procedure) was performed.

Pathology examination confirmed the presence of a tubular adenoma with lower-grade dysplasia measuring 2.5×2.0 cm in diameter (Figure 3). The second lesion, however, was a 5×4.5 cm rectum GIST with high mitotic index (>10 mitoses/50 high-power fields). There was no lymph node metastasis. Immunohistochemistry indicated strong staining for c-Kit/CD117 and Dog-1, while expression of S-100 and CD34 was negative (Figure 3). KIT exon 11 mutation
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(W557G) was detected. He was diagnosed as high grade rectum GIST due to large tumor size, and high mitotic index.

The patient accepted adjuvant therapy with imatinib (400 mg/day). Postoperative CT scan performed 1 year later showed tumor recurrence in the patient. Then the patient died of subsequent emaciation 15 months after surgery.

Discussion

GIST is an uncommon mesenchymal tumor of the gastrointestinal tract and expresses CD117, a tyrosine-kinase growth factor receptor and the most important GIST marker [1]. Primary GISTs arise most commonly in the stomach (50-70%), followed by the small intestine (25-35%), only about 5% of all GISTs originate in the rectum [2].

Recent years, cases of simultaneous development of a GIST and another neoplasm with different histotypes originating from the same organ have been reported more frequently [3-5]. Various hypotheses, such as gene mutations, expression of metallothioneins (MT) and influenced neighboring tissues by the same carcinogen have been proposed regarding the coexistence of a GIST and other carcinomas [6, 7]. However, no data are available to support such hypotheses until now. Furthermore, because of the low incidence of rectal GISTs, cases of simultaneous development of a GIST and another neoplasm in rectum are rare. As for our case, simple coincidence could be the most reasonable explanation.

Surgery remains the mainstay of treatment in GIST patients with resectable GIST. Recently, imatinib is indicated for the first-line treatment of metastatic and unresectable GIST, which is particularly attractive for patients with large or poorly localized primary GISTs that would otherwise require extensive surgery or sacrifice of a large amount of normal tissue [8].

More recent studies of GISTs have focused on the effect of mutational status on response to imatinib or other tyrosine kinase inhibitors. The presence and type of KIT mutations have been found to predict response to tyrosine kinase inhibitors [9]. However, PDGFRA mutations predominantly occur in gastric GIST often leading to primary imatinib resistance [10]. Patients with exon 11 mutations have better objective response rate (63%-83.5%) and increased progression free survival than those with exon 9 mutations (34%-47.8%) or wild-type mutations (0%-44.6%) [9-11]. Furthermore, patients with KIT mutations involving codons 557-558 indicate an unfavorable prognosis [12].

As shown in our case, anterior resection with end colostomy and rectal stump (Hartmann’s procedure) was performed. Our case had the W557G mutation at position 557 in KIT from an amino acid tryptophan (W) to glycine (G). Large tumor size, high mitotic index and codons 557 mutation proposed to be predictors of the unexpected poor prognosis. Then the patient accepted adjuvant therapy with imatinib (400 mg/day). However, treatment failed to provide a good long time outcome.

Disclosure of conflict of interest

None.

Address correspondence to: Dr. Xu-Dong Wu, Department of Gastroenterology, Yancheng City No. 1 People’s Hospital, 16 Yuehe Road, Yancheng, Jiangsu Province, China. Tel: +86-15861974719; E-mail: hnjsycwx@163.com

References

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Figure 2. CT scans showed a rectal tumor and the relationship with adjacent organs. A: rectal tumor and seminal vesicles. B: rectal tumor and bladder. C, D: rectal tumor and prostate. B: Bladder; t: Rectal tumor; p: prostate; →: seminal vesicles.
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Figure 3. A: Microscopic image of tubular adenoma with lower-grade dysplasia (×10 magnification). B: Microscopic image of GIST (×10 magnification). The immunohistochemistry indicated strong staining for c-Kit/CD117 (C: ×10 magnification), Dog-1 (D: ×40 magnification).