

# World Journal of *Clinical Cases*

*World J Clin Cases* 2023 October 6; 11(28): 6670-6973



## MINIREVIEWS

- 6670 Neurotransmitters regulate  $\beta$  cells insulin secretion: A neglected factor  
*Kong CC, Cheng JD, Wang W*

## ORIGINAL ARTICLE

## Case Control Study

- 6680 Factors influencing the surveillance of re-emerging intracranial infections in elective neurosurgical patients: A single-center retrospective study  
*Wang JL, Wu XW, Wang SN, Liu X, Xiao B, Wang Y, Yu J*

## Retrospective Study

- 6688 Clinical value of chemiluminescence method for detection of antinuclear antibody profiles  
*Xiang HY, Xiang XY, Ten TB, Ding X, Liu YW, Luo CH*
- 6698 Value of ultrasound guided biopsy combined with Xpert *Mycobacterium tuberculosis*/resistance to rifampin assay in the diagnosis of chest wall tuberculosis  
*Yan QH, Chi JY, Zhang L, Xue F, Cui J, Kong HL*
- 6707 Research on the intelligent internet nursing model based on the child respiratory and asthma control test scale for asthma management of preschool children  
*Pei CF, Zhang L, Xu XY, Qin Z, Liang HM*
- 6715 Effects of different doses of long-acting growth hormone in treating children with growth hormone deficiency  
*Xia W, Wang T, Pan JY*
- 6725 Efficacy and anti-inflammatory analysis of glucocorticoid, antihistamine and leukotriene receptor antagonist in the treatment of allergic rhinitis  
*Qiu C, Feng D*
- 6733 Subchondral fatigue fracture of the femoral head in young military recruits: Potential risk factors  
*Yang JZ, Chen P, Chen BH, Zhao B*
- 6744 Anemia status of infants and young children aged six to thirty-six months in Ma'an Shan City: A retrospective study  
*Wang XM, Wang QY, Huang J*
- Observational Study
- 6754 Impact of coronary artery bypass grafting surgery on the chorioretinal biomicroscopic characteristics  
*Shahriari M, Nikkhah H, Mahjoob MP, Behnaz N, Barkhordari S, Cheraqpour K*

**Prospective Study**

- 6763** Effects of humanized nursing care on negative emotions and complications in patients undergoing hysteromyoma surgery  
*Liu L, Xiao YH, Zhou XH*

**Randomized Controlled Trial**

- 6774** Randomized controlled trial on the efficacy and safety of autologous serum eye drops in dry eye syndrome  
*Zheng N, Zhu SQ*

**SYSTEMATIC REVIEWS**

- 6782** Primary adrenal Ewing sarcoma: A systematic review of the literature  
*Manatakis DK, Tsouknidas I, Mylonakis E, Tasis NP, Antonopoulou MI, Acheimastos V, Mastoropoulou A, Korkolis DP*

**CASE REPORT**

- 6792** Pulmonary artery aneurysm protruding into the bronchus as an endobronchial mass: A case report  
*Li M, Zhu WY, Wu RR, Wang L, Mo MT, Liu SN, Zhu DY, Luo Z*
- 6797** Rare rectal gastrointestinal stromal tumor case: A case report and review of the literature  
*Dong RX, Wang C, Zhou H, Yin HQ, Liu Y, Liang HT, Pan YB, Wang JW, Cao YQ*
- 6806** Bilateral retinal nerve fiber layer thickness reduction in a 9-year-old myopic boy suffering from unilateral optic neuritis: A case report  
*Zhao FF, Yao SQ, Wang Y, Li TP, Yang JF, Pang CP, Cen LP*
- 6812** Application of negative pressure wound therapy after skin grafting in the treatment of skin cancer: A case report  
*Huang GS, Xu KC*
- 6817** Diagnosis and treatment of McCune-Albright syndrome: A case report  
*Lin X, Feng NY, Lei YJ*
- 6823** Paraneoplastic myopathy-related rhabdomyolysis and pancreatic cancer: A case report and review of the literature  
*Costantini A, Moletta L, Pierobon ES, Serafini S, Valmasoni M, Sperti C*
- 6831** Multi-organ hereditary hemorrhagic telangiectasia: A case report  
*Chen YL, Jiang HY, Li DP, Lin J, Chen Y, Xu LL, Gao H*
- 6841** Hyperprogression after anti-programmed death-1 therapy in a patient with urothelial bladder carcinoma: A case report  
*Yang HY, Du YX, Hou YJ, Lu DR, Xue P*
- 6850** Effectiveness of antidepressant repetitive transcranial magnetic stimulation in a patient with refractory psychogenic dysphagia: A case report and review of literature  
*Woo CG, Kim JH, Lee JH, Kim HJ*

- 6857** Entrapment neuropathy of common peroneal nerve by fabella: A case report  
*Lin JC, Tsai MH, Lin WP, Kuan TS, Lien WC*
- 6864** Importance of accurate diagnosis of congenital agenesis of the gallbladder from atypical gallbladder stone presentations: A case report  
*Sun HJ, Ge F, Si Y, Wang Z, Sun HB*
- 6871** Dorsal approach for isolated volar fracture-dislocation of the base of the second metacarpal: A case report  
*Kurozumi T, Saito M, Odachi K, Masui F*
- 6877** Rotationplasty type BIIIb as an effective alternative to limb salvage procedure in adults: Two case reports  
*Chen ZX, Guo XW, Hong HS, Zhang C, Xie W, Sha M, Ding ZQ*
- 6889** Primary cutaneous anaplastic large cell lymphoma with over-expressed Ki-67 transitioning into systemic anaplastic large cell lymphoma: A case report  
*Mu HX, Tang XQ*
- 6895** Confusing finding of quantitative fluorescent polymerase chain reaction analysis in invasive prenatal genetic diagnosis: A case report  
*Chen C, Tang T, Song QL, He YJ, Cai Y*
- 6902** Testicular mixed germ cell tumor: A case report  
*Xiao QF, Li J, Tang B, Zhu YQ*
- 6908** Leukemic transformation during anti-tuberculosis treatment in aplastic anemia-paroxysmal nocturnal hemoglobinuria syndrome: A case report and review of literature  
*Xiu NN, Yang XD, Xu J, Ju B, Sun XY, Zhao XC*
- 6920** Pancreatic arteriovenous malformation treated with transcatheter arterial embolization: Two case reports and review of literature  
*Shin SH, Cho CK, Yu SY*
- 6931** Cecal duplication cyst in an infant presenting as shock: A case report  
*Kim SM, Lee SH, Park GY, Kim SS, Lee CG, Jin SJ*
- 6938** Pulmonary reversed halo cycles and consolidations after immunotherapy: A case report  
*Suo H, Shi YJ, Huang ZD, Xu K, Huang H*
- 6943** Unusual case of emphysematous cystitis mimicking intestinal perforation: A case report  
*Kang HY, Lee DS, Lee D*
- 6949** Malignant proliferative ependymoma of the neck with lymph node metastasis: A case report  
*Wang K, Wen JZ, Zhou SX, Ye LF, Fang C, Chen Y, Wang HX, Luo X*
- 6955** Wandering spleen torsion with portal vein thrombosis: A case report  
*Zhu XY, Ji DX, Shi WZ, Fu YW, Zhang DK*



- 6961** Intracranial infection and sepsis in infants caused by *Salmonella derby*: A case report  
*Yu JL, Jiang LL, Dong R, Liu SY*
- 6967** Large gastric hamartomatous inverted polyp accompanied by advanced gastric cancer: A case report  
*Park G, Kim J, Lee SH, Kim Y*

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## Rare rectal gastrointestinal stromal tumor case: A case report and review of the literature

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### Abstract

#### BACKGROUND

Gastrointestinal stromal tumors (GISTs) are rare tumors of the gastrointestinal tract accounting for less than 1% of all gut tumors. GISTs occurring in the rectum are extremely rare, and these usually present at an advanced stage compared with other sites.

#### CASE SUMMARY

A 60-year-old male who presented with features of sensations of rectal tenesmus was referred to our department with a mass in the lower rectum that was detected during a routine checkup. Colonoscopy, transrectal ultrasound, perianal magnetic resonance imaging and ultrasonic contrast were used to diagnose the rectum GIST, and then the patient underwent complete transanal resection using the ultrasonic scalpel. The patient was discharged ten days after the operation and was defined as low risk. Therefore, he had no need to receive subsequent adjuvant therapies, and he had not suffered any anal dysfunction or had any evidence of recurrence at follow up.

#### CONCLUSION

Surgical resection with histologically negative margins is the standard curative treatment for rectal GISTs. Appropriate surgical techniques based on the location, size, and resectability of the tumor should attract great attention from clinicians.

**Key Words:** Gastrointestinal stromal tumor; Rectum; Ultrasonic scalpel resection; Case report

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**Core Tip:** Gastrointestinal stromal tumors are rare malignancies that rarely occur in the rectum. The diagnosis is established by biopsy and immunohistochemistry. Complete surgical resection with negative margins is the treatment of choice. The appropriate surgical technique should be selected based on the location, size, and resectability of the tumor and the available surgical expertise. This case is reported for its rare location and treatment with ultrasonic scalpel total resection without imatinib treatment.

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## INTRODUCTION

Gastrointestinal stromal tumors (GISTs) are the most common type of mesenchymal tumor involving the gastrointestinal tract. However, GISTs represent a rare entity that accounts for less than 1% of all alimentary tract tumors[1]. The most common sites involved are the stomach (50%–65%), followed by the small intestine (20%–30%)[2]. Esophageal GISTs are rare, but tumors may also arise in extra-gastrointestinal locations, principally in the mesentery, omentum, and retroperitoneum[3]. GISTs less frequently affect the colon and rectum, with an estimated incidence of 5%–15%[4,5]. Rectal GISTs are extremely rare with low incidence, only accounting for merely 0.1% of all kinds of rectal tumors[6]. Different surgical methods have been widely reported, including transanal excision, laparoscopic surgery, transsacral excision, and transanal endoscopic microsurgery[7-10]. Sometimes, rectal GISTs, especially those located in the lower rectum, are detected during daily physical examination.

Here, we present a case of a rectal GIST with complete resection by ultrasonic scalpel to highlight the clinical and surgical features of lower rectal GISTs.

## CASE PRESENTATION

### Chief complaints

A 60-year-old male rectal GIST patient presented with features of sensitization of the rectum with tenesmus.

### History of present illness

The patient was referred to our department with a mass in the lower rectum that was detected during a routine checkup at two months prior. The patient had undergone a colonoscopy two months prior, which incidentally discovered a mass in the rectal submucosa (Figure 1).

### History of past illness

The patient had a clinical history of hypertension and complete right bundle branch block, but ultrasonic cardiogram and blood tests were normal.

### Personal and family history

The patient denied any history of genetic disease or infection.

### Physical examination

Regular physical examination showed no obvious abnormalities, except on the anorectal physical examination (inspection, palpation, digital rectal examination, careful probing, and anoscopy). A submucosal mass was found in the anterosuperior region without signs of macroscopic rectal bleeding.

### Laboratory examinations

Regular preoperative examination, including routine blood work, liver and kidney function tests, coagulation function and electrocardiography were normal, and biotumoural findings such as carcino-embryonic antigen, carbohydrate antigen-724, carbohydrate antigen-199, squamous cell carcinoma, carbohydrate antigen-211, carbohydrate antigen-50, carbohydrate antigen-242, alpha fetoprotein, and neuron-specific enolase were all negative.

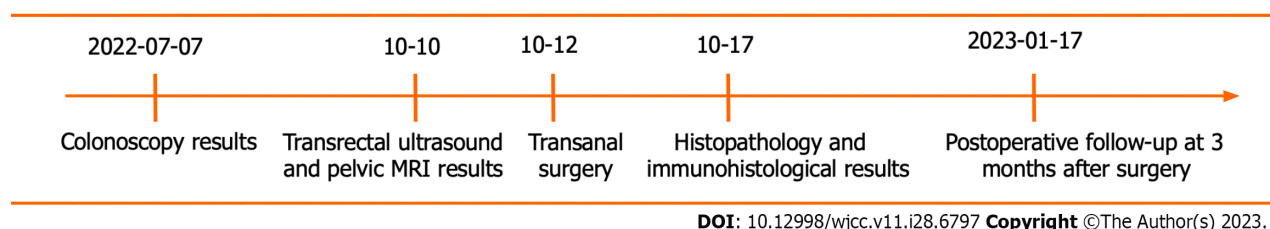


Figure 1 Time axis of the patient's detection of gastrointestinal stromal tumor and surgery treatment and postoperative follow-up.

### Imaging examinations

Colonoscopy (Figure 2A), transrectal ultrasound (Figure 2B), ultrasonic contrast (Figure 3) and perianal magnetic resonance imaging (MRI) (Figure 2C and D) showed that the mass involved the muscularis propria and a characteristic appearance of a well-circumscribed intramural mass involving the anterior and superior sides of the rectum located 3 cm from the anal verge without metastatic disease.

### FINAL DIAGNOSIS

According to the results of colonoscopy, transrectal ultrasound, ultrasonic contrast, MRI and histopathological examination, the patient was eventually diagnosed with a rectal GIST.

### TREATMENT

Based on these examinations and the unusual location of this tumor, we consulted with the radiologist to evaluate whether a transanal resection could intactly excise the tumor with minimal possible surgical complications, such as incontinence of feces or anal sphincter dysfunction. The patient accepted our proposed surgery plan. Under intraspinal anaesthesia, he was required to be in the lithotomy position, and we injected 0.9% saline into the rectum submucosa for full exposure. We used an ultrasonic scalpel to incise the rectum mucosa and then separated the surrounding tissue until the submucosa was reached. During the separation, we were very careful to maintain the tumor capsule integrity. After fully exposing the tumor, we fortunately observed that the rectal GIST was centrally located in the rectum submucosa and not wrapped by the rectal muscle. We incised the rectum mucous membrane and completely separated the tumor from the submucous stratum of the rectum and then resected the intact tumor with an ultrasonic scalpel. The incision in the rectal muscle was designed to be as small as possible to maintain the integrity of the rectal mucosa. Then, we placed mattress sutures within the incision in the muscular layer of the rectum (Figure 4).

The surgical procedure lasted only approximately 20 min, and the blood loss was insignificant. No postoperative complications were observed during hospitalization. A full liquid diet was taken for five days to delay bowel movements and prevent wound infection. The first defecation occurred on the 5 d after the surgery, and the patient was discharged on the tenth postoperative day.

Final pathology reported a tumor of 2 cm × 1.5 cm × 1.5 cm with a complete capsule, fibrous and adipose tissue attached, and tumor-free margins. Histological examination (Figure 5A and B) confirmed a fusocellular subtype of rectal GIST. Its cellularity was low, and its mitotic index [number of mitoses per 50 high-power field (HPF)] was less than 5/50 HPF with areas of necrosis and calcification. Immunohistochemistry (IHC) demonstrated the diagnosis of GIST when the tumor was positive for CD 34 (Figure 5C), CD117 (Figure 5D), Dog-1 (Figure 5E), and Ki67 expressed approximately 3% (Figure 5F) and negative for S-100, HMB 45 (human melanoma black 45), and  $\alpha$ -SMA (smooth muscle actin).

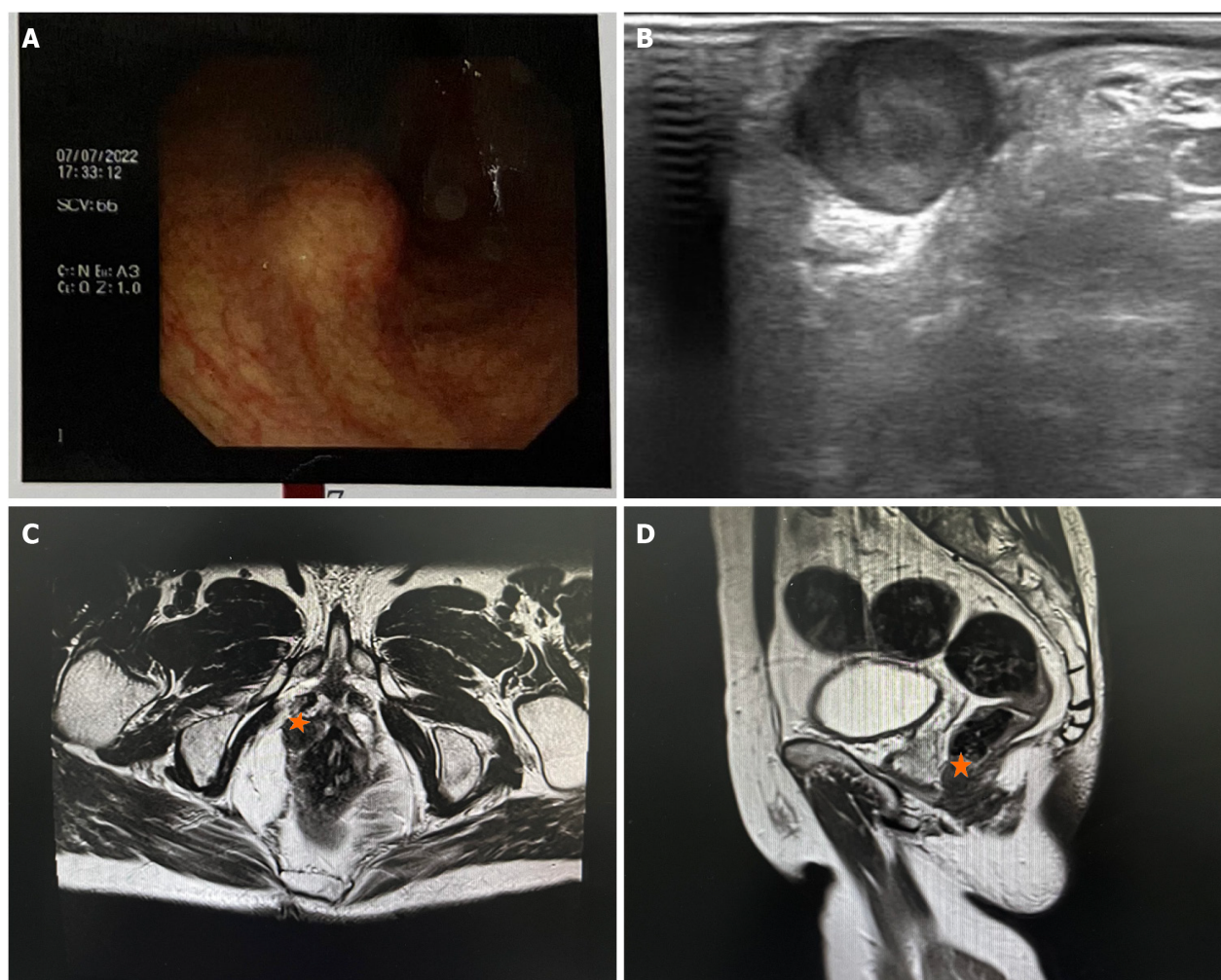
### OUTCOME AND FOLLOW-UP

The recurrence risk assessment system for the primary GIST after complete resection was based on the modified National Institutes of Health classification system (2008). This patient was defined as low risk. Therefore, he had no need to receive subsequent adjuvant therapies such as imatinib treatment after surgery, and he had not suffered any anal dysfunction or evidence of recurrence at follow-up 3 months after surgery.

### DISCUSSION

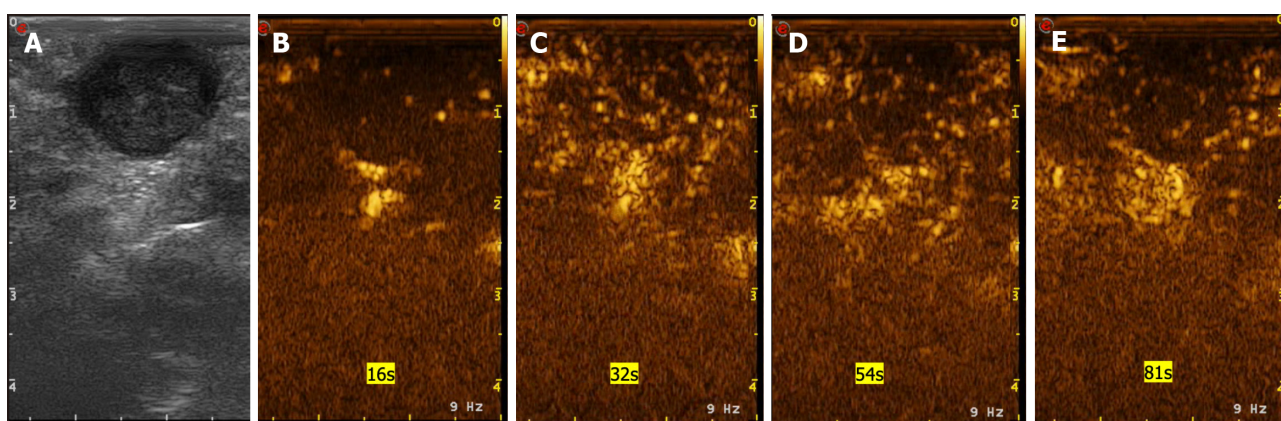
GISTs were first described by Mazur and Clark[11] in 1983. Rectal GISTs are usually found incidentally, either on lower abdominal computed tomography (CT), perianal MRI, colonoscopy, or clinical examination. Low-lying GISTs usually palpate as a smooth, well-demarcated, firm lump during physical examination. For patients with a suspected rectal GIST,





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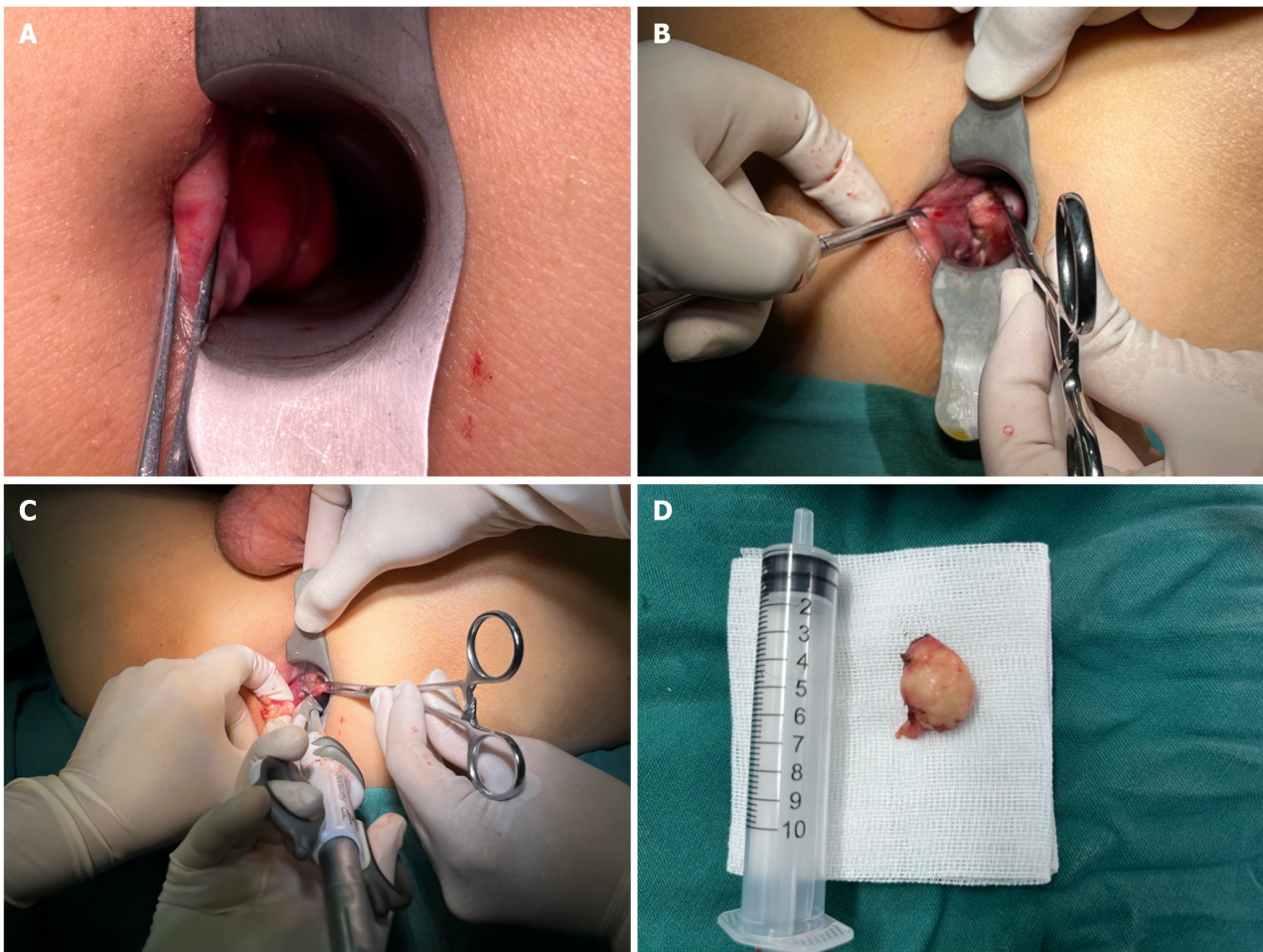
**Figure 2** Imaging examinations show the location of the rectal gastrointestinal stromal tumor. A: The colonoscopy results show that the tumor originated from the rectal dentate line; B: The results of trans-rectal ultrasound show that the tumor protruded from the posterior of the rectum submucosa; C and D: The results of pelvic magnetic resonance imaging revealed that the tumor (pentagram) was located in the rectum submucosa.



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**Figure 3** The presentation of gastrointestinal stromal tumor in transrectal ultrasonography and ultrasonic contrast. A: The presentation of gastrointestinal stromal tumor in transrectal ultrasonography; B: The overall enhancement is low. In the early stage of arterial, 16 S began to enhance; C: 32 S reached the peak and showed uneven low enhancement; D: 54 S began to clear; E: The venous phase showed low enhancement.





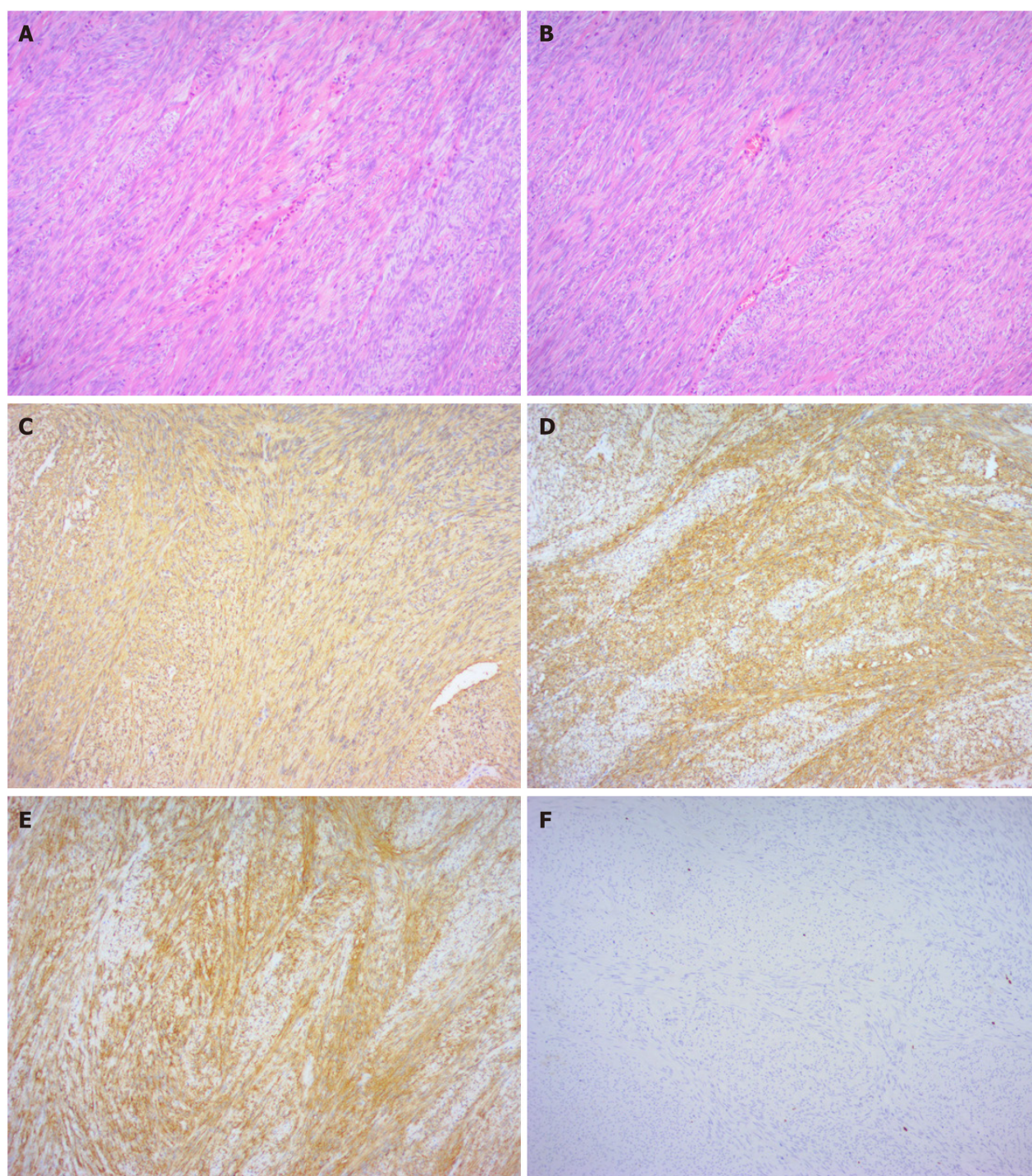
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**Figure 4** Transanal surgery with ultrasonic scalpel. A: Pre-operative patient photos; B: Intraoperative pictures; C: Excision with ultrasonic scalpel; D: Operative specimen.

CT scan with contrast is often the initial imaging modality of choice. It seems to be a reliable technique for the diagnosis and the stage of the disease. GISTs are believed to arise from the interstitial cells of Cajal or other pluripotential mesenchymal stem cells[12]. The morphology of the cells and the findings of IHC are the major factors for the pathological diagnosis of GIST. Regarding microscopic features, there are three types: the spindle cell type (70%), epithelioid cell type (20%), and mixed type (10%). Regarding immunohistochemical findings, 95% of GISTs are positive for CD117 and/or discovered on Dog-1, and 70% are found to be positive for CD34[13]. Although CD117 positivity is a major defining feature for a tumor that has morphological features compatible with GIST, CD117 positivity alone is not an absolute requirement for the diagnosis. When there is CD117 negativity, as in approximately 5% of GISTs, Dog-1 staining, followed by CD34 staining, is considered diagnostic.

The mitotic index, the size of tumor, the location of tumor (gastric *vs.* nongastric), and the tumor rupture are the four independent prognostic factors for GISTs. Additionally, tumor rupture should be considered separately with regard to whether it occurred in preoperative and intraoperative. Surgery is still considered the only treatment modality that can offer a permanent cure for GIST. There are many risk-stratification tools, for example, the 2017 American Joint Committee on Cancer recommendations on staging of rectal GISTs include both tumor size ( $\leq 2$  cm, 2-5 cm, 5-10 cm, and  $>10$  cm) and mitotic rate ( $\leq 5$  mitoses or  $> 5$  mitoses per 50 high-power field) to help determine rates of disease progression[14]. For those GISTs that are inoperable, metastatic, or recurrent, imatinib mesylate was thought to be the first-line standard treatment. After accurate and complete resection, surgery is sufficient for approximately 60% of GIST patients. However, for the remaining 40% of patients with relapses, metastases or local advancement to the point where surgery may not be effective, additional targeted therapy with the tyrosine kinase inhibitor imatinib mesylate may be prescribed[15]. In rectal GISTs, radical resection is thought to be the most important determinant of prognosis[16]. However, since certain anatomical characteristics, such as a narrow, deep pelvis and the vicinity to the internal and external sphincters or invasions of the adjacent organs, there are many difficulties in surgical resection[17]. Although surgical resection without lymph node resection is still the most standard treatment for local rectal GISTs, the selection of both safe and feasible surgical procedures for rectal GISTs remains controversial[18-20]. Local resection, laparoscopic surgery, and endoscopic resection are the three mainstream surgical methods for rectal GISTs[21]. Local resection, including transanal, transsacral, parasacral, perineal, and transvaginal resection, not only achieve a good clinical curative effect but avoid the abnormal appearance and dysfunction of genitourinary and anal functions[22]. However, traditional transanal local excision has





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**Figure 5 Histopathology of the biopsy specimen.** A and B: Histopathology showed that the cells were spindle-shaped and moderately differentiated (magnifications,  $\times 100$ ). C to F: Immunohistochemistry results showed that cluster of differentiation (CD) 34 (C), CD117 (D), Dog-1 (E), and CD34 (F) were all positive, and Ki67 was negative.

limitations. Because of poor visualization, conventional transanal local excision may have difficulty controlling resection limits and allow for complete removal of the rectal wall. The conventional transanal approach is also less oncologically complete, with inherently higher recurrence rates, higher rates of tumor remnants, specimen fragmentation, and inferior overall and disease-free survival compared with radical resection[23]. The choice of rectal GISTs surgical approach should be individualized according to their location and different size of the tumor, range of local invasion, ability for sphincter preservation, and risk of malignant degeneration. The objective of surgical methods for rectal GISTs is not only to improve prognosis but also to preserve the patient's quality of life. Therefore, in this case, we used an ultrasonic scalpel instead of excision.

The ultrasonically activated scalpel, also known as the ultrasonic scalpel, was first introduced in the early 1990s and works by generating the high-frequency harmonic motion of a metallic rod and allows for the denaturation of tissue



proteins, thus producing a protein coagulum that seals coapted vessel walls[24]. These vibrations can simultaneously coagulate and cut tissues[25]. Unlike electrocoagulation, ultrasonic scalpels not only lack eschar formation over the blade and cause minimal collateral thermal damage to the surrounding tissues and less smoke[26] but also provide very effective cutting with less bleeding. There are neither transmission of electricity nor neuromuscular stimulation to patients[27].

The time spent in the operating room is expensive, and this technique can decrease both the operation time and associated costs and seems to reduce hospital costs[28]. Compared with conventional energy systems, multiple studies have demonstrated that there are many superiorities of the ultrasonic scalpel, especially in several surgical subspecialties [29-31]. The most valuable benefits were considered to be a shorter operation time and less intraoperative bleeding[32-33]. Recent studies confirmed that in settings with financial restrictions such as those encountered in low-resource countries, ultrasonic scalpels can be reused safely without any consequences to the patient's condition or postoperative course[34]. Because the ultrasonic scalpel could cut and coagulate effectively, numerous energy exchanges between the scalpel and cautery could be eliminated. Therefore, it is extremely beneficial for surgeries in which required accurate dissection and the avoidance of tissue damage from lateral heat damage, especially in anorectal surgery.

## CONCLUSION

GISTs are rare malignancies that rarely occur in the rectum. The diagnosis of GISTs mainly relies on biopsy and IHC. Complete tumor excision with negative margins is often the ideal method in the clinical treatment of GISTs. Appropriate operative procedures are hard to select because accurate detection based on the location, size, and resectability of the tumor and the available surgical expertise are very difficult. This case is reported for its rare location and treatment with ultrasonic scalpel total resection without imatinib treatment.

## FOOTNOTES

**Author contributions:** Dong RX and Zhou H collected data and drafted the manuscript; Yin HQ provide the transrectal ultrasound and ultrasonic contrast pictures; Liu Y, Pan YB, Wang JW, and Liang HT analyzed the case materials; Wang C and Cao YQ have reviewed the manuscript; all authors have made substantial contributions to the work, and all authors have read and approved the final manuscript.

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