International Journal of Innovative Research in Medical Science (IJIRMS)
Volume 07, Issue 10, October 2022,
https://doi.org/10.23958/ijirms/vol07-i10/1529

Case report

Anorectal Gastrointestinal Stromal Tumor: About a Rare Case Report and Review of Literature

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Received 29 September 2022; Accepted 25 October 2022; Published 28 October 2022

Abstract

Gastrointestinal stromal tumor's are the most common mesenchymal tumor of the gastrointestinal tract in adults; stromal tumors in the rectum are extremely rare. We report the case of a 64 years old woman admitted for admitted for etiological assessment of low-grade rectal bleeding and terminal appearance constipation, whose paraclinical investigations showed the presence of a GIST without secondary lesions with a high risk of recurrence and whose therapeutic decision was to start imatinib as a neoadjuvant treatment and then schedule her for a subsequent resection.

Keywords: ***Gastrointestinal stromal tumors, constipation, rectal syndrome, Immunohistochemistry, Imatinib.***

Introduction

The term of Gastrointestinal Stromal Tumor (GIST) appeared for the first time back in 1983 [1]. They are rare tumors with a specific KIT-positive proto-oncogene (CD117) and are the most frequent gastrointestinal mesenchymal neoplasms [2]. The common sites of GIST are the stomach (50-60%), small intestine (30-40%), colon (7%) and esophagus (1%) [3]. Anorectal GIST comprises 2-5% of all cases [3-5].

Case report

Patient information

A 64-year-old patient, who's known to be diabetic type 2 on glimepiride, she had no comorbidities and denied any family history of cancer. was admitted for exploration chronic constipation and rectal syndrome associated to rectal bleeding of medium abundance, which had appeared six months earlier. No other functional gastro-intestinal or extra-digestive signs were reported by the patient and a good general state.

Clinical findings

The clinical exam was unremarkable except for the presence of a firm, circumferential with a regular surface and non-stenosing mass located at 3 cm from the anal margin with a good sphincter tone. Biological assessment showed a hemoglobin at 9,8 g/dl, white blood cells at 4850/uL, platelet count at 401,000/uL, PCR negative at 3.32 mg/L, tumor markers were negative.

Diagnostic assessment

Colonoscopy was performed showing a mass at 3 cm from the anal margin with a submucosal appearance, budding in the rectal lumen, ulcerated on the surface, with an acute connection angle, extending upwards for about 5 cm. (**Figures 1,2,3**) Biopsie was performed and the anatomopathology was in favor of spindle cell tumor proliferation in favor of a stromal tumor with an immunohistochemical study confirming GIST (positive anti CD34, anti CD117, anti Ki67 and anti DDG1) with a high risk of postoperative recurrence according to the histopathological score of Miettinen.

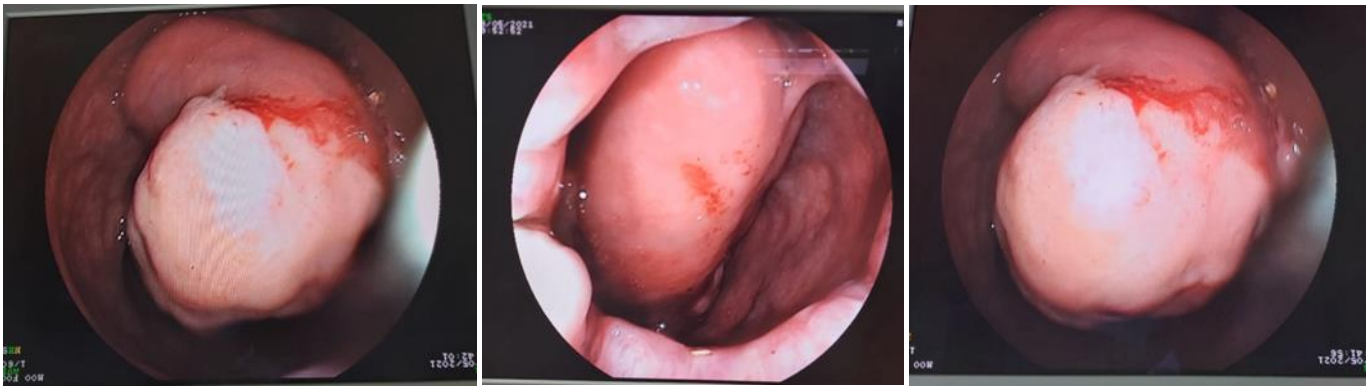
A thoraco-abdomino-pelvic CT scans was performed for staging and provided an objective view of the lesion, showing the presence of a budding endoluminal and exophytic growth tumor of 69.2 mm x 107.4 mm in contact with the uterus and the right tube with loss of separation line, invading the mesorectum on the right side with the presence of a left mesorectal lymph node of 6.2 mm of small axis with no other secondary localizations. (**Figures 4,5**)

Therapeutic intervention

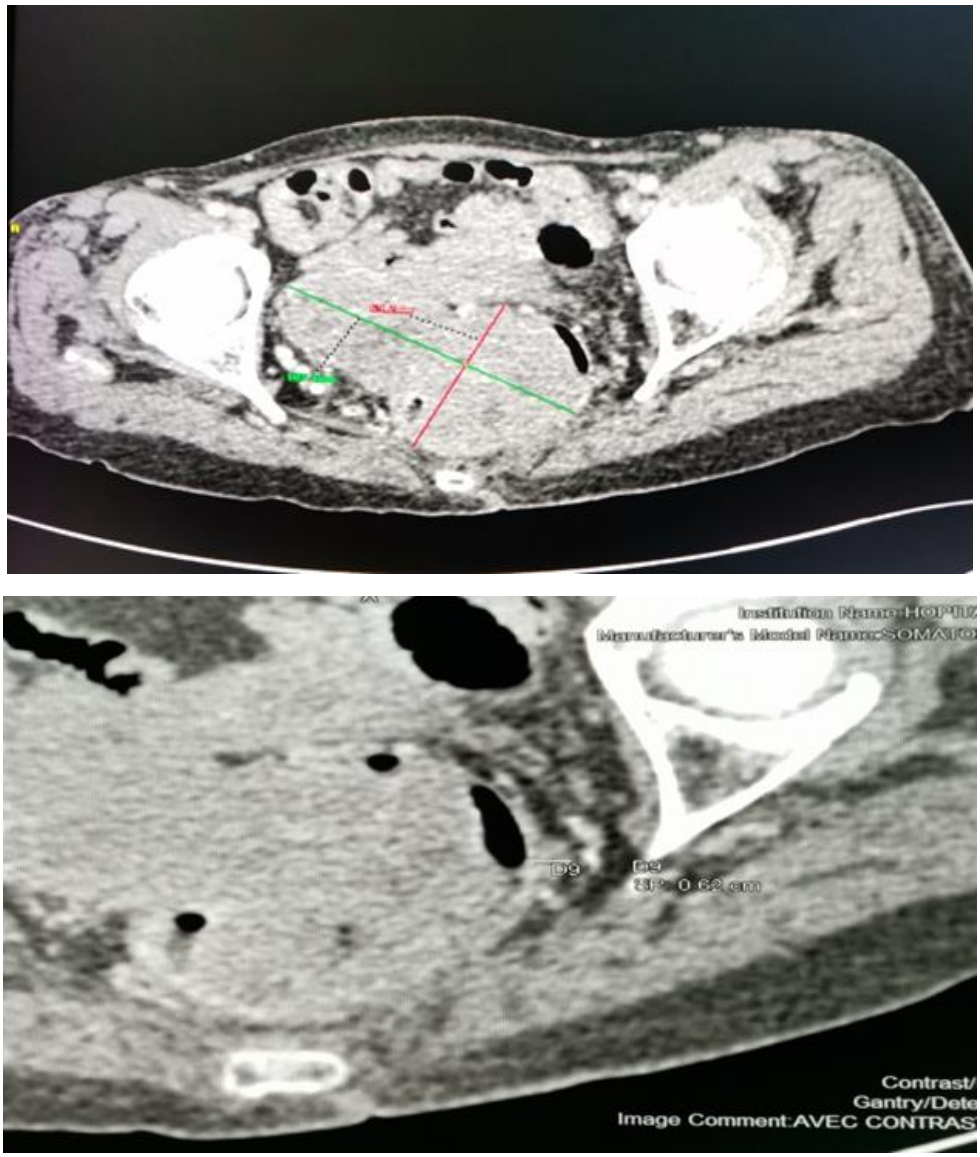
The decision was to perform a neoadjuvant treatment with Imatinib in front of the potential malignant aspect of the tumor and the unrespectability.

Follow up and outcomes

The patient died within 3 months of diagnosis



Figures 1,2,3: Showing an intra-rectal mass



Figures 4,5: Abdomino-pelvic CT scan showing an endoluminal and exophytic growth tumor of 69.2 mm x 107.4 mm locally advanced

Discussion

Gastrointestinal stromal tumors (GIST) are mostly located in the stomach (50-70%) followed by the small intestine in (20-30%). The rectal location is extremely rare and represents only 5% [6]. If the majority of GISTs have a benign evolution, they can rarely present an aggressive evolution. This is the case for our patient. The prognosis on the aggressiveness of a GIST tumor can usually be made on the basis of the data of the tumor size examination (> 2cm) and histology (Miettinen score) [7]. Rectal GIST occur in the majority of cases sporadically, even if some familial cases have been described [8].

The clinical manifestations are very variable and not very specific, and do not differ from those of other rectal tumors. Usually they are asymptomatic; Sometimes, they are associated with non-specific symptoms and in the case of large rectal GISTs, they can be associated with abdominal pain or constipation. The majority of rectal GISTs presents as a small and hard nodule. Large rectal GISTs can ulcerate and imitate a rectal adenocarcinoma with rectal bleeding, abdominal discomfort and constipation [9,10].

Digital examination of the rectum, recto-sigmoidoscopy, colonoscopy are essential for diagnosis, together they represents the reference examination that allows to visualize the tumor which

forms a rounded formation bulging under a normal or ulcerated mucosa.

The rectal-ultrasound (UES) allows to have a prognostic idea of the tumor: by evaluating the masse size, its contours, the existence of a loco-regional infiltration: Appearance of GIST is often typical of tumors developing in the smooth muscle. It appears as a hypoechoic lesion, often oval and homogeneous, with regular boundaries, developing from the fourth hypoechoic layer, which corresponds to the muscularis. The presence of criterias such as the presence of a tumor mass greater than 10 cm, locoregional invasion, the existence of central necrosis, poorly bounded contours, and intra-tumoral cystic areas are factors probably associated with a higher potentiaĺ of malignancý of GISTs. UES provides a probabilistic diagnosis of GIST with high sensitivitý and specificitý but not perfectly determined [10,11].

All these diagnostics assessments provides to perform biopsies for anatomopathological confirmation which is the gold standard in the diagnosis of GIST, since it provides information on the immunohistochemical features and mitotic count for a prognostic evaluation of the risk of recurrence thanks to the Miettinen classification. GIST's typically expresses CD117, often CD34 and sometimes SMA and S-100, but its expressions vary depending on different sites [12].

Logo-regional extension and staging of the tumor are usually identified by cross-sectional CT or MRI imaging. CT injection is a key examination for the diagnosis and staging of GIST and provides comprehensive information including detecting the presence of metastatic spread, GISTs typically appear as a mass with exoluminal development and clear boundaries. Variable size: from 4 to 30 cm. Heterogeneous tissue density [13,14]. On MRI, the tumor is usually hyposignal in T1 with well-defined boundaries and iso-signal in T2 [15].

Surgical resection in mono-block is the standard curative treatment for GIST but it remains limited to localized forms. This complete resection with negative margins on pathological examination remains the reference treatment and can be accomplished in 40-60% of all GIST patients. As GISTs can occur anywhere in the gastrointestinal tract, the therapeutic approach and surgical approach differs, notably the trans-anal approach is the preferred one for GISTs of the lower rectum. On the other hand, the treatment of advanced rectal GIST is controversial and imatinib (a tyrosine kinase inhibitor) is the medical treatment of choice for advanced or metastatic GIST. It initially allows a reduction in tumor volume to increase the chances of a monobloc resection during surgery, but also reduces the morbidity of the surgical procedure and improves the patient's quality of life [16].

The 5-year overall survival of patients with rectal GIST varies in the literature between 20% to 60 %. If the tumors have a high metotic index (>5) and mesures more than 10 cm it has a high risk of recurrence [10].

Conclusion

GIST of the lower rectum is an extremely rare entity requiring multidisciplinary management. The positive diagnosis is mainly immunohistochemical. Surgical resection with an R0 resection margin and neoadjuvant Imatinib is the treatment of choice for large and high-risk tumors.

Authors' Contributions

All authors participated in the conception, drafting the work, critically revised the manuscript, approved the final version to be published, and agree to be accountable for all aspects of the work.

Funding

The author(s) received no financial support for the research, authorship, and/or publication of this article.

Declaration of conflicting interests

The author(s) declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

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