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Case Report

Unusual Localization of a Gastrointestinal Stromal Tumor: A Case Report

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Abstract

Gastrointestinal stromal tumors (GISTs) are rare, accounting for less than 3% of all gastrointestinal neoplasms and less than 6% of all sarcomas.

We present the case of a female patient who presented for non-specific symptoms. She was investigated and the CT examination showed an extensive anorectal stenotic tumor mass, invading the uterus and surrounding muscles. Also, there were found regional adenopathies and a single liver metastasis.

After the biopsy a histopathological examination was performed, and then the immunohistochemistry examination concluded that was the case of GIST.

Keywords: *GIST; CT; MRI; Anorectal Tumor*

Abbreviations

CT: Computed Tomography; GIST: Gastrointestinal Stromal Tumor; MRI: Magnetic Resonance Imaging

Introduction

Gastrointestinal stromal tumors (GISTs) are rare, accounting for less than 3% of all gastrointestinal neoplasms and less than 6% of all sarcomas [1-3].

The mean age at presentation is greater than 50 years, according to studies [3-6]. Although some reports in the literature show that GIST has a male predominance [7,8], others show no gender predilection [9-11].

In the past, mesenchymal tumors of the gastrointestinal tract were usually classified as leiomyomas or leiomyosarcomas [12-14], but growing evidence over the last two decades suggests that GISTs are a unique and separate entity.

GISTs are now defined as spindle cell, epithelioid, and occasionally pleomorphic mesenchymal tumors [15-17] of the gastrointestinal tract that express the KIT(CD117, stem cell factor receptor) tyrosine kinase and showing the presence of activating mutations in KIT or PDGFRα (platelet-derived growth factor alpha) detected at immunohistochemistry [17-20].

This specific tests allowed GISTs to be distinguished from true leiomyomas, leiomyosarcomas, neurofibromas or schwannomas in 95% of patients [13,15-17,21].

GISTs can originate in the gastrointestinal tract, mesentery, or omentum [2,13,21].

Case Report

We present the case of a female patient, aged 71 years, who presented in 2014 abdominal-pelvic pain, weight loss in the last year, transit and urination disorders, and metrorrhagia, symptomatology that progressively worsened. In October 2014 she was admitted for investigation and specialized treatment.

The gynecological consultation identified a vaginal stenosis that couldn't allow the examination with speculum. It was observed that the vagina expresses a purulent greenish secretion. At the internal palpation of the vagina, uterine cervix and uterus could not be palpated due to a tumoral process occupying the pelvis; also, the vaginal posterior wall was infiltrated by a possible abscess.

After the gynecological examination a CT was performed and identified a single hepatic metastasis at the fifth segment, of 4.5/3 cm in axial diameters, a few small bladder polyps of 3 - 4 mm diameter each, left adrenal adenoma of 1.5/2 cm, and a coraliform calculus at the left renal pelvis and lower calyx, measuring 3/1cm in diameter. Also, an anorectal stenotic tumor, unobtrusive, without superjacent intestinal dilation, which measures 10/12 cm in axial diameters and 14 cm in craniocaudal diameter, inseparable from the levatorani muscles, with invasion of the posterior uterine wall; the tumor mass is heterogeneously vascularized, with no necrosis zones. The bladder was not invaded by the tumor, but was compressed to anterior. It was found also that the uterus has an irregular interstitium with a maximum thickness of 1.8 cm, and shows a 7/4 cm intracavitary fluid accumulation in axial diameters. There were founda few para-aortic and perirectal lymph nodes below 1 cm.

The rectosigmoidoscopy, which was subsequently done, revealed a circumferential tumor mass approximately 7 - 15 mm from the anus.

In November 2014, at another hospital, is performed a transanal biopsy, sigmoid colostoma and evacuation of the pelvic abscess. The histopathological examination of the biopsy piece identified tumoral malignant proliferation of the sarcomatous type (fusiform tumoral infiltration and storiform pattern).

In December 2014 the immunohistochemistry examination showed VIM+ diffuse, CL 34 beta E12 negative, P63 negative, ki 67+ 30%, CD 117+, HMB 45 negative, CD 34+ diffuse, ACT negative in tumor cells, positive in vessels, S100 negative, MITF negative, with the conclusion of Gastrointestinal stromal tumor.

In March 2015, the patient started the chemotherapy treatment with Imatinib, and in July 2015, has undergone a MRI examination of the abdomen and pelvis that highlighted a 27/24/20 mm septate cystic metastasis in the sixth segment of the liver, a left adrenal nodule of about 21/18 mm, with the lipomatous component suggestive of adenoma. Also there was found an asymmetrical circumferential parietal thickening in the middle and lower rectum (more expressed in the right lateral wall), with polynodular aspect (maximum axial diameters of 60/34 mm), associating the pararectal fascia breach and extension to the pelvic fat. The lesion hadlow signal intensity on T1 weighted sequence, hypersignal intensity on T2 weighted and STIR sequences, showed diffusion restriction, gadolinophila, and was indistinguishable from the vagina and cervix. As well, there were found perirectal adenopathies (max 5 mm) and bilateral obturators adenopathies (max 17/10 mm).

As a result of the imaging aspect, it was decided that the patient would continue chemotherapy treatment and performed a CT exam at 6 months, which revealed slightly dimensional regression of liver cystic metastasis compared to the previous MRI exam (current dimensions being 2/2/2.2 cm), as well as a stationary aspect of the rectal tumor.

In June 2016 an abdomino-pelvic CT scan was performed that revealed a stationary appearance of the liver metastasis, but a dimensional regression of the rectal tumor, which at that time was visualized as an asymmetric parietal thickening with a nodular aspect, with dimensions of 2.7/1.8 cm, with predominantly peripheral contrast enhancement, who infiltrated the posterolateral area of the cervix, but did not extend beyond the pararectal fascia. Also, the size and location of adenopathies remained unchanged.

After another 6 months, and then in September 2017, two other CT examinations were performed, which noted that the asymmetric rectal parietal thickening had unmodified characteristics and dimensions from the previous examination. Also, at the examination in September 2017, the liver metastasis had smaller dimensions, of 1.4 cm.

The last CT was performed in February 2018, which concluded that the imaging aspect of oncological lesions is stationary.

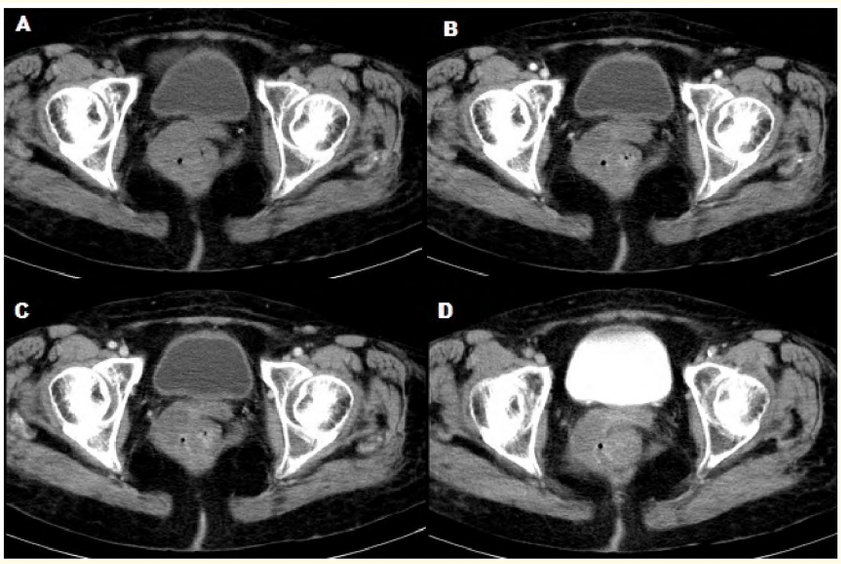

Figure 1: *Abdominal and pelvic CT (February 2018) – A. native examination, B. arterial phase, C. venous phase, D. delayed phase: asymmetric rectal parietal thickening, with a nodular aspect, with predominantly peripheral contrast enhancement, with no necrosis zones, who infiltrated the posterolateral area of the cervix, but did not extend beyond the pararectal fascia.*

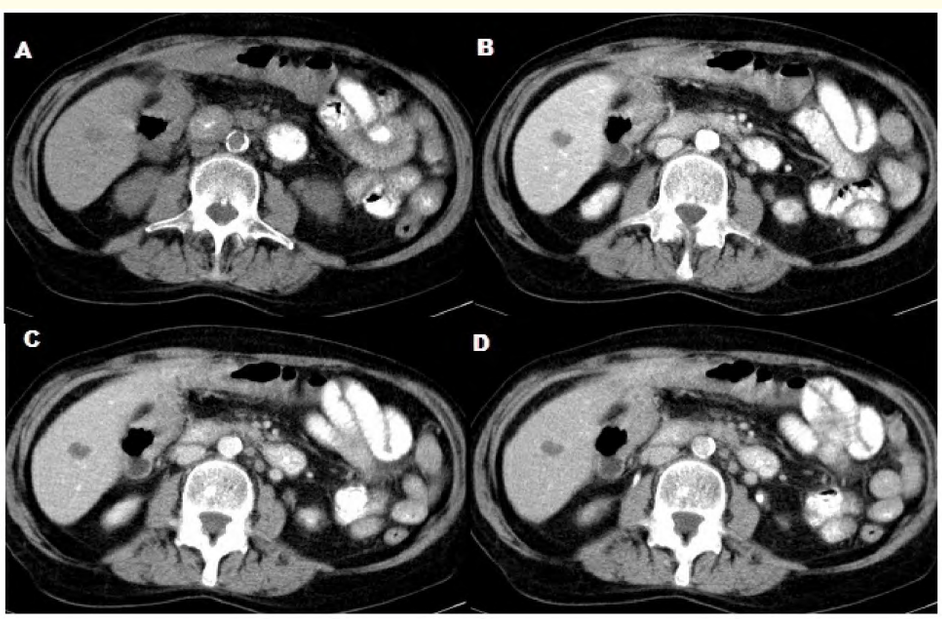

Figure 2: *Abdominal and pelvic CT (February 2018) – A. native phase, B. arterial phase, C. venous phase, D. parenchymal phase: single hepatic cystic metastasis.*

Discussion

Gastrointestinal stromal tumours (GISTs) are fairly rare neoplasms, constituting less than 3% of all gastrointestinal malignancies [22].

GIST is currently thought to originate from interstitial cells of Cajal [19]. The presence of interstitial Cajal-like cells has been reported in several extraintestinal organs including urinary bladder, prostate, gallbladder, omentum, uterus, fallopian tube, and atrial and ventricular myocardium [23,24]. This may explain the development of extraintestinal GIST [25,26].

According to several studies [3-7,18] the stomach and small bowel account for the majority of cases of GIST. In our case the tumor location was anorectal. GIST of anal canal and rectum are often grouped together and account for nearly 5% of all GIST [27]. Six (9%) of the 64 GISTs in the series of Levy., *et al*. [28] were located in the anorectal region. Anorectal GISTs are most commonly mural masses that expand the rectal wall.

In the study done by Yang., *et al*. [14], the primary tumor location was in the colorectum in 10% of patients. Their results were similar to those of previous studies [21,29].

Tumors found incidentally are smaller and have a better prognosis than those that cause symptoms [18]. In one report [4], asymptomatic tumors found incidentally had a mean diameter of only 1.5 cm.

The most common clinical manifestation of a symptomatic GIST in the series of Yang., *et al*. [14] was gastrointestinal bleeding. Patients may present with hematemesis, melena, or signs and symptoms of anemia caused by occult bleeding. This is compatible with previous

In the case we describe, the tumor size at presentation was 10/12/14 cm. These dimensions fit into those presented by Burkill., *et al*. [7] which present a mean primary tumor size of 13 cm ± 6. On the other hand, in Mochizuki's study [31] most tumors reached just 5 cm.

Most GISTs present cystic areas or necrosis according to studies conducted by Mochizuki., *et al*. [31] and Burkill., *et al*. [7] the percentage reaching up to 67%, or in some cases in over 80% [18]. In the study by Tateishi., *et al*. [10] statistically significant CT findings of high-grade tumors included heterogeneous enhancement pattern. The series by Yang [14] and Chun [9] also demonstrated the same result.

In the case presented by us, the tumor mass is heterogeneous in native and post contrast images, but without areas of necrosis, and had infiltrative margins. In the paper presented by Mochizuki., *et al*. [31] only two huge malignant tumors had unclear margins, and in the study conducted by Hersh., *et al*. [18] the tumors had invasive margins in 4 cases from a total of 49 patients analyzed. Consistent with them, Burkill [7] assert that the tumors showed well defined contours in most cases analyzed, namely 86%. Also, he stated that visceral obstruction rarely occur in GIST, even in the presence of extensive peritoneal metastatic disease, as indicated by Hersh., *et al*. also in their study [18].

The incidence of metastases at presentation in the largest clinical series of malignant GISTs approached 50% [3]. The distribution of metastases in some study is similar, with the liver and peritoneum dominating [5,7]. The liver is the most common metastatic site at both presentation and disease relapse [3,7].

In one study [7] metastases were seen in 61% of patients at presentation and in 87% of patients during follow-up. On the other hand other authors [18,19] present in their studies a different frequency of occurrence of metastases, and stated that about 20% of patients will present with or develop distant metastases. Metastatic spread to regional lymph nodes is rare [18] or non-existent [4,7] in GIST according to some studies. In our case, perirectal and bilateral obturators adenopathies were found, but those have remained stationary due to the treatment that the patient received.

Complete surgical excision of the primary tumor offers the best chance of cure [3,4,19], but controversy exists whether abdominoperineal resection (APR) or conservative surgery is the best alternative [32]. However, the high rates of local and distant recurrence indicate the need for effective nonsurgical treatment [18]. Until recently, chemotherapy response rates have been disappointing [3,33,34].

Imatinib mesylate, a tyrosine kinase inhibitor (STI-571, imatinib [Gleevec]; Novartis, Basel, Switzerland), has shown promising results in its management [19,22,35], and altered the clinical approach to GISTs because it has been proven to be effective in the medical treatment of unresectable or metastatic GISTs [35]. Tumor responses to imatinib are seen in 80% of patients [19]. However, kinase inhibition by imatinib is not uniformly successful [19]. It has been suggested that low risk GIST with size < 2 cm and mitosis < 5 per 50 HPF may be considered for local excision if sphincter saving surgery is technically feasible, and more aggressive GIST should be treated with radical excision [32].

In the case presented, the patient was treated with chemotherapy alone, although the lesion had high aggressiveness. The post-treatment imaging results were appreciable, namely the size diminishing of both liver metastasis and tumor, as well as regression of regional tumor invasion.

Conflict of Interest

There is no conflict of interest.

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