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GASTROINTESTINAL STROMAL TUMOR MIMICKING GYNECOLOGICAL PATHOLOGY: A CASE REPORT

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Summary

Diagnosis of gastrointestinal stromal tumors (GISTs) of the terminal part of the small intestine on ultrasound examination can be difficult because of their similarity in appearance to gynecological tumors. We present a case of a 49-year-old asymptomatic female patient with GIST of the small intestine, which presented as a pelvic mass, mimicking an ovarian tumor. Tumor was diagnosed during the control check up and ultrasound gynecological examination. Computed tomography (CT) showed tumor mass in the pelvis on the right and free fluid in the lesser pelvis. During the surgery, exploration of the abdominal cavity displayed tumor of the terminal part of the small intestine (ileum). The uterus and both adnexes were normal. The patient was treated by resection of the terminal part of the small intestine and termino-terminal anastomosis. Immunohistochemical evaluation demonstrated positive vimentin, positive CD117, and negative CD 34. In the presence of a pelvic mass, especially if other unusual anamnestic data are present, the possibility of other than a gynecologic tumor has to be considered.

KEYWORDS: *gastrointestinal stromal tumor, asymptomatic, immunohistochemia, transvaginal ultrasound.*

GASTROINTESTINALNI STROMALNI TUMOR KOJI OPONAŠA GINEKOLOŠKU PATOLOGIJU: PRIKAZ SLUČAJA

Sažetak

Dijagnozu gastrointestinalnih stromalnih tumora (GIST) završnog dijela tankog crijeva teško je postaviti ultrazvučnim pregledom jer su izgledom nalik ginekološkim tumorima. Opisujemo slučaj 49-godišnje asimptomatične bolesnice s GIST-om tankog crijeva koji se prikazuje kao tvorba u zdjelici i oponaša tumor jajnika. Tumor je dijagnosticiran na kontrolnom i ultrazvučnom ginekološkom pregledu. Kompjutorizirana tomografija (CT) otkrila je tumorsku tvorbu u zdjelici i slobodnu tekućinu maloj zdjelici. Pretraživanjem trbušne šupljine tijekom operacije otkriven je tumor završnog dijela tankog crijeva (ileuma). Maternica i adneksi bili su zdravi. Bolesnici je napravljena resekcija završnog dijela tankog crijeva i termino-terminalna anastomoza. Imunohistokemijska analiza pokazala je pozitivnu reakciju na vimentin i CD117, te negativnu na CD 34. Kad je u zdjelici prisutna tvorba, osobito ako su i drugi anamnestički podaci neuobičajeni, treba uzeti u obzir mogućnost da je riječ o nekom drugom, a ne ginekološkom tumoru.

KLJUČNE RIJEČI: *gastrointestinalni stromalni tumor, asimptomatičan, immunohistokemija, transvaginalni ultrazvuk*

INTRODUCTION

Gastrointestinal stormal tumors (GISTs) are rare diseases (10-20 / 1 000 000) (1) and constitute less than 1% of all digestive tumors (2). These tumors usually occur in the stomach (70%), small intestine (20-25%), large intestine and rectum (5%) (1), and rarely in the esophagus (<5%), retroperitoneum, mesentery and omentum. It can appear in almost every age, although are much more common in people older than 50, equally in both sexes (2-4). GISTs are among the most common

mesenchymal tumors of the gastrointestinal (GI) tract. Diagnosis of GIST is based on pathohistological analysis (spindle-shaped cells are present in 70% of all cases, with a 20% of those dominated by epithelioid cells) as well as immunohistochemical features (vimentin positive in 98% of the cases, the expression of CD 117 (c-kit protein) in 95% of all cases, often in combination with expression of CD 34 which makes 60 - 70% of all cases) (5). Positive CD 117 is considered a key marker in the differentiation of GIST from other mesenchymal tumors of the GI tract. The genetic analysis has determined the correlation of the GIST and the two genetic mutations: KIT (codes kit protein) and PDGFRA (codes A chain of PDGF receptors) (5, 6). The demonstration of mutations in target genes is required only in cases that are histologically suggestive but CD 117 (c-kit) negative; beyond this indication, this is only undertaken in research protocols (5). According to the Fletcher's classification, these tumors may be of very low, low, medium and high-level of malignancy, depending on the size of tumors and number of mitosis (7). Only surgical intervention provides an opportunity for cure. Imatinib mesylate, a potent inhibitor of KIT activity, is now a standard front-line therapy for advanced GIST (unresectable and/or metastatic malignant CD 117 (c - kit)-positive GISTs) (8). With the introduction of imatinib, there have been dramatic improvements in response rates, time to progression, and survival. Unfortunately, many patients eventually recur or progress during imatinib therapy (9). Before imatinib mesylate was discovered, unresectable and/or metastatic GIST had been a malignant illness with rapid death outcome (9). The average survival period of patients with unresectable malignant GIST, without the therapy of imatinib mesylate is 10-23 months and in a GIST with diagnosed metastases 12-19 months from the moment of establishing the diagnosis of metastases. Advanced malignant GIST responds very poorly to radiotherapy (9). The 5-year overall survival ranges from 21% to 88% in different series, depending of the risk level and completeness of the surgical resection (10-12).

THE CASE

A 49-year-old patient was hospitalized for a right adnexal tumor and planned surgical procedure. In her family anamnesis there were no cases of GIST, no malignant diseases and no neurofibromatosis type I. The patient had no symptoms, and the tumor was diagnosed during the control check up and ultrasound gynecological examination. The laboratory tests were normal (WBC 6.75 x 10^9 /L, RBC 4.38 x 10^{12} /L, HGB 131 g/L, HCT 0.410 L/L, MCV 93.7 fL, MCH 29.9 pg, MCHC 319 g/L, PLT 297 x 10^9 /L, urine normal). Gynecological examination and transvaginal color doppler ultrasound showed a formation on the right, above the uterus, 6 x 10 cm, without a detectable flow, unhomogenic (Figures 1 and 2).

Tumor markers were also determined, and the values were within the limits of normal (CEA - 1.9 ug / L, CA-125 - 18 kui / L, CA 15-3 - 16 kui / L, CA 19-9 - 31.2 kui / L).

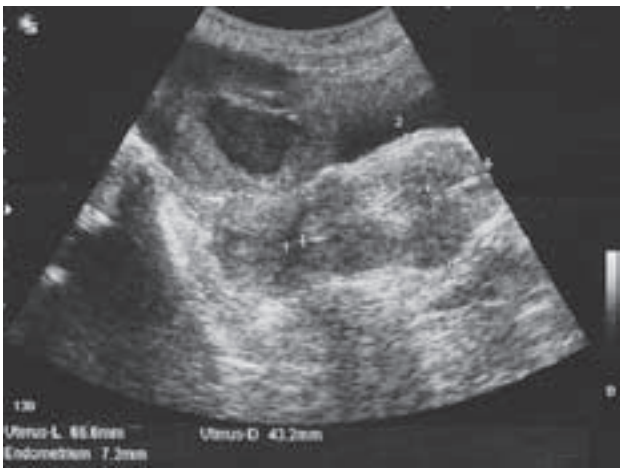

Figure 1. Normal ultrasound uterine scan and a suspicious adnexal mass on the right

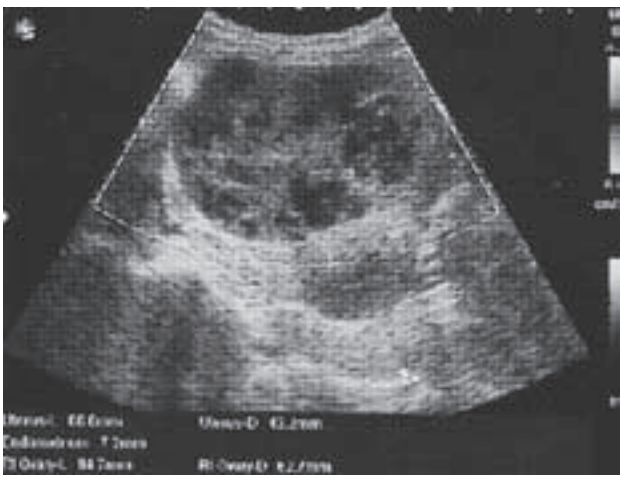

Figure 2. Suspicious ovary tumor on the right

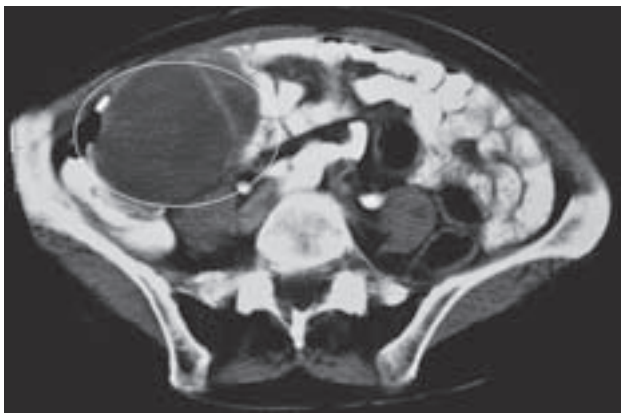

Figure 3. Tumor mass in the pelvis leaning on the right adnexa

Computed tomography (CT) findings (Figure 3) showed a tumor mass on the right and free fluid in the lesser pelvis. The liver was of normal size, homogeneous structure, with normal coefficient of absorption, and without any suspicious changes. There were no enlarged lymph nodes.

Intravenous urography was done as a part of the additional examination and it showed an impression of the bladder. During the surgical procedure (exploratory laparotomy) a tumor of the terminal part of the small intestine (ileum) was found, near the right adnexis. The tumor was removed entirely, along with resection of the terminal part of the small intestine and termino-terminal anastomosis. At the end of surgery, exploration of the abdominal cavity displayed the normal uterus and both adnexes and there were no more tumor masses in abdomen. The material obtained was sent for pathohistological and immunohistochemical analysis. After the completion of surgery, the patient was transferred to the post-anesthesia care unit and closely monitored. The post-operative period passed without any complications. Pathohistological and immunohistochemical analysis confirmed diagnosis of a low-level malignant GIST (the tumor tissue built of a thick mass of spindle-shaped cells with eosinophilic cytoplasm and oval nucleus that show polymorphism, hiperchromasis and mitosis, <10 pathological mitotic shapes in 10 HPF, positive smooth-muscle aktin, CD117 positive, vimetin positive, and CD 34 negative). Positron emission tomography/computed tomography (PET/CT) check up after the surgery did not show signs of residual disease. The patient did not receive imatinib mesylate therapy. She undergoes regular checks up.

DISCUSSION

Sporadic cases of GIST (as in our patient) are much more frequent, but some familiar GIST syndromes are described. Familial GISTs are inherited in an autosomal dominant manner, with incomplete penetrance in the same genes as sporadic GISTs, alone or as a component of a syndrome associated with other tumors (like neurofibromatosis type 1) or GISTs associated with paragangliomas by allelic loss in the genes coding for succinate dehydrogenase subunits (13, 14).

Clinical presentation is heterogeneous: GISTs can present as small, incidentally found nodules to large aggressive tumors (15) with nonspecific clinical symptoms (cramps or diffuse abdominal pain, melena, abdominal mass). Localization and size of GIST in the digestive tract are the two most important factors that clinical features of illness depend on. This variability is represented in the literature with a high percentage of asymptomatic incidental findings (3), as in our patient, versus other published series with a majority of symptomatic patients (16, 17). Smaller tumors are usually asymptomatic. Because of their variable clinical presentation there is no standard diagnostic protocol, so all imaging or endoscopic techniques may be used in the detection and location of these tumors (1). In an asymptomatic patient with a suspicious mass in the pelvis on ultrasound scan, in the area of adnexis, the first and usually correct diagnosis is an adnexal tumor. To determine the localization of the tumor, other imaging techniques may be useful, but exact diagnosis is often made during the surgical procedure, as reported in our case.

A major difficulty with these malignant tumors is their diagnosis and subsequent prognosis, due to their rare and nonspecific symptoms. Diagnosis and treatment of GIST require a multidisciplinary approach, given the combination of pathologic and radiographic evaluation, surgical treatment, and oncology care required to successfully treat patients with GIST, as shown in our case. Only entire surgical removal of the tumor provides an opportunity for healing and long-term survival.

CONCLUSION

There are two reasons why our case report is an interesting one: this tumor was primary diag-

nosed as a gynecological tumor and the presented case is an exception when compared with the literature worldwide, where in a majority of cases patients are over 50 years of age and with a gastric tumor.

GISTs, especially of the small intestine, as reported by others (18-23) may simulate a gynecological tumor in women. In such circumstances they are first noticed by gynecologists and are often diagnosed, prior to surgery, as ovarian tumors. Only at surgery their nature is fully understood. Therefore, in the presence of a pelvic mass, especially if other unusual anamnestic data are present too, the possibility of other than a gynecologic tumor has to be considered.

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