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A Rare Case of Early Gastrointestinal Stromal Tumor in 34-Year-Old Woman: A Caution for Adnexal Tumor Mimicry

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ABSTRACT:

Introduction: Gastrointestinal Stromal Tumors (GISTs) are the leading type of mesenchymal tumor in the gastrointestinal (GI) tract, constituting 80% of these tumors and 0.1% to 3% of all gastrointestinal malignancies. They arise from spindle-shaped mesenchymal cells known as interstitial cells of Cajal. Its onset can be at any age, with the peak occurring in the sixth decade.

Case Presentation: We report the case of a 34-year-old woman with a complaint about a lump in her lower abdomen that has been progressively enlarging over the past year without any significant symptoms following the lump. Findings in the ultrasound suggest an ovarian mass. Laparotomy was carried out, and while no ovarian mass was found, instead a mass was identified in the mesentery of the small bowel. As a result, a complete excision of the tumor was performed, with histopathologic and immunohistochemical results confirming a GIST diagnosis.

Conclusion: Representing a tiny percentage of gastrointestinal tumors, GISTs are rare. GISTs may be asymptomatic or present without any typical symptoms. Sole reliance on clinical and radiological investigations can cause them to be easily overlooked. Awareness of GIST is critical and care must be taken in diagnosis since other tumor can mimic GISTs in endoscopic, imaging, and histological findings.

KEYWORDS: Gastrointestinal Stromal Tumors, GIST, Extragastrointestinal Stromal Tumors, EGIST, CD117

INTRODUCTION

Gastrointestinal Stromal Tumors (GISTs) are the leading type of mesenchymal tumor in the gastrointestinal (GI) tract, constituting 80% of these tumors and 0.1% to 3% of all gastrointestinal malignancies.[1] The primary sites for GISTs are the stomach (50%-70%) and small intestine (20%-30%), specifically the duodenum, jejunum, and ileum, with rarer occurences in the large intestine (5%) and esophagus (2%-5%).[2,3] A limited number of GISTs are found outside the gastrointestinal tract. These are termed extragastrointestinal GISTs (EGISTs) and involve the omentum, mesentery, retroperitoneum, and perineum.[4] The median diagnostic age of GIST is generally in the mid-60s across various studies, with equal representation of gender distribution.[5]

CASE PRESENTATION

A 34-year-old woman came to the obstetrician-gynecologist outpatient clinic with a concern about a lump in her lower abdomen that has progressively grown over the past year. There were no remarkable symptoms following the lump. On physical examination, a solid, immobile mass measuring about 10x8 cm was discovered. The patient then underwent an ultrasound examination. The exam revealed a solid heteroechoic mass measuring 8.7 x 7.6 cm in the left iliac region of the abdominal cavity with the leading differential diagnosis being an ovarian mass by the radiologist

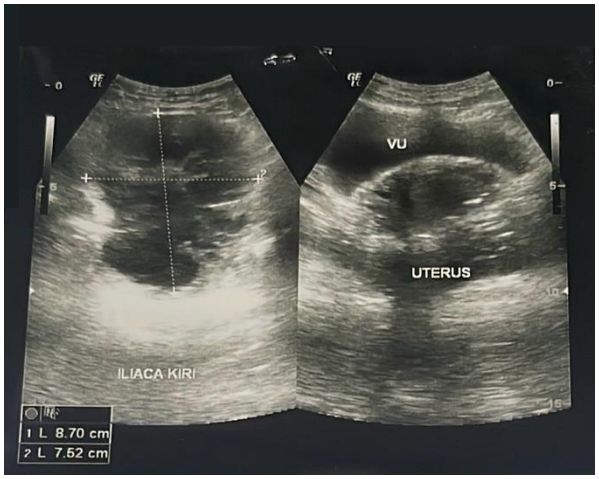

Figure 1. Abdominal USG showing a heteroechoic solid mass in the left iliac region of the abdominal cavity.

The obstetric and gynecologist subsequently performed an elective laparotomy through infraumbilical transverse incision and no adnexal mass was discovered as the ultrasound findings, instead, a mass was found around the small bowel. Consequently, a surgeon was brought in for consultation during the procedure.

The surgeon made an incision along the midline. Identification from the ligamentum of treitz to the ileocaecal junction was carried out during the surgery. A mass was observed in the jejunum, 35 cm from the ligamentum of treitz, located in the mesentery. End-to-end anastomosis resection was performed with macroscopic tumor-free margins. A firm mass with an irregular surface measuring 11 x 7.5 x 7 cm was found.

Postoperatively, the patient had a smooth recovery, showing no sign of anastomotic leakage. For the first two days, the patient received total parenteral nutrition. A residual test was carried out on the third day prior to beginning a liquid diet via nasogastric tube. Removal of the nasogastric tube and drain took place on the fifth day, followed by the initiation of a soft diet. The patient was discharged from the hospital after a week-long stay.

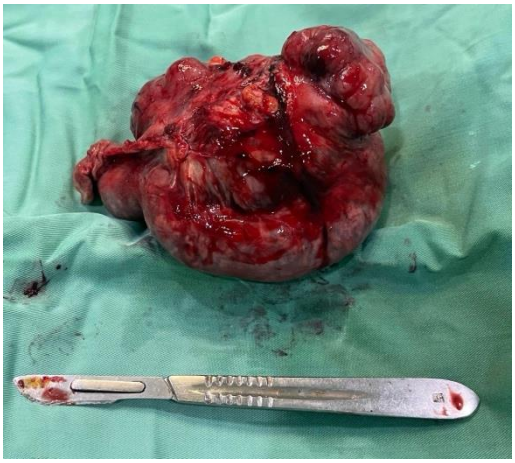

Figure 2. Excised tumor.

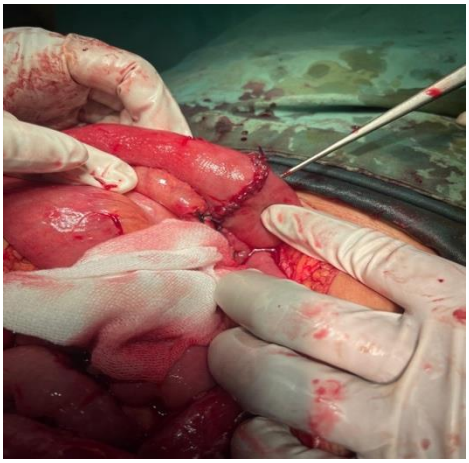

Figure 3. End-to-end anastomosis resection (right).

The histopathology results showed that the tumor mass was composed of sheets, nets, and solids, some were syncytial and diffuse. Tumor cells are spindle-shaped, with elongated nuclei, coarse chromatin, and partially prominent nuclei with slightly eosinophilic cytoplasm. In the focus, atypical epithelioid cells were seen, round and oval in shape, coarse chromatin, pale eosinophilic cytoplasm. Mitosis was found (>5/50 HPT). According to the results, a high-risk malignant gastrointestinal tumor was identified.

Due to the limited additional examination and treatment resources in which the case took place, the patient was referred for further immunohistochemistry tests and therapy planning. The CD117

Immunohistochemistry test results were positive.

DISCUSSION

Gastrointestinal stromal tumors (GISTs) are the prevailing soft tissue sarcoma observed in the GI tract with an anticipated yearly occurrence of 0.68 to 0.78 per 100,000 individuals in the United States.[6,7] These cells are derived from mesenchymal cells that have

spindle shape, termed interstitial cells of Cajal (ICCs) or their progenitor stem cells. Interstitial cells of Cajal are specialized nerve cells serving as pacemaker cells, located within the muscularis propria and around the myenteric plexus.[8]

Extragastrointestinal Stromal Tumor (EGISTs) are GISTs that develop outside the GI tract.[9] Constituting a mere 5% to 10% of all GISTs, EGISTs are extremely rare and primarily occur in the omentum or mesentery.[10] Therefore, it is indicated that this mesenteric EGIST case is considered to be particularly rare.

The symptoms associated with GISTs vary by their size and location. Smaller GISTs are usually symptom-free, but larger ones can lead to abdominal pain, digestive discomfort, and a feeling of fullness in the abdomen.[1] Typically, EGISTs do not cause symptoms unless they compress surrounding tissues.[11]

Radiological tests may not be sufficient for locating GISTs. Localization can be achieved using barium contrast studies, endoscopy, and CT scans, which also help determine the size and secondary localization like hepatic metastases. PET scans can be useful for secondary localization, but histological and immunohistochemical confirmation are essential and consider as the gold standard of diagnosis.[12,13]

The main histological classification of GISTs include spindle cell (70%), epitheloid cell (20%), and mixed type (10%).[14] In immunohistochemistry (IHC) studies, positive CD117 (c-KIT) staining is essential for diagnosing GISTs. Additional markers include BCL-2 (80%), CD34 (70%), SMA (30%), desmin (5%), and DOG1.[15] About 95% of GISTs test positive for KIT staining.[14] The tumor presented in our case was positive for c-KIT.

The recommended treatment of GISTs is radical resection, and for high-risk tumors, adjuvant imatinib is prescribed for at least 3 years after surgery.[16] This approach is necessary because determining whether a GIST is benign or malignant is challenging even with pathological examination. Obtaining clear margins during excision is crucial, as it significantly impacts the prognosis.[17] There is still possibility of metastasis or recurrence in patient who have undergone full resection with no rupture and clean margins. Thus, early referral to medical oncologist is needed for multidisciplinary care. Eligibility for adjuvant therapy ought to be decided based on risk evaluation. For advanced disease patients, assessing the tumor's mutational status initially is vital for targeted chemotherapy, given the correlation between tumor genotype and tyrosine kinase inhibitor response.[18,19,20]

CONCLUSION

Representing a tiny percentage of gastrointestinal tumors, GISTs are rare. GISTs may be asymptomatic or present without any typical symptoms. Sole reliance on clinical and radiological investigations can cause them to be easily overlooked. Awareness of GIST as a unique GI tract lesion is critical in managing these rare and aggressive tumors, which have significant likelihood of recurrence and malignant behavior. As awareness of GISTs increases, care must be taken in diagnosis since other tumor can mimic GISTs in endoscopic, imaging, and histological findings. In this case, identifying mesenteric GISTs is challenging as they don't present with clear clinical or radiological signs. Therefore, it's important to consider GISTs in the present of large painless abdominal mass.

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