ORIGINAL RESEARCH PAPER

Radio-Diagnosis

CASE REPORT: A RARE ENCOUNTER – UNMASKING A RETROPERITONEAL GASTROINTESTINAL STROMAL TUMOR (GIST) IN A 37-YEAR-OLD MALE

KEY WORDS: Retroperitoneal Gist, Paraganglioma, Radiological Diagnosis, Histopathological Examination, Immunohistochemistry

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ABSTRACT

Gastrointestinal stromal tumors (GISTs) are the most prevalent mesenchymal tumors of the gastrointestinal system. They arise from progenitor cells known as interstitial cells of Cajal, which are mainly found in the small intestine and stomach. Though they are far less common, extra-gastrointestinal stromal tumors (EGISTs) can develop in other areas, such as the retroperitoneum, mesentery and omentum. We present a rare case of a 37-year-old male with a retroperitoneal gastrointestinal stromal tumor (GIST), initially misdiagnosed as a retroperitoneal paraganglioma based on radiological findings. The patient presented with right-sided abdominal pain and distension. A contrast-enhanced CT (CECT) of the abdomen revealed a lobulated, hypervascular retroperitoneal mass with central necrosis and hemorrhage, suggestive of a paraganglioma. The patient underwent an excision biopsy of the lesion, and histopathological examination with immunohistochemistry confirmed the diagnosis of GIST, with positivity for CD117 and DOG1. The patient was treated with Imatinib postoperatively and showed clinical improvement. This case highlights the diagnostic challenges posed by retroperitoneal masses and the importance of corroborative histopathological and immunohistochemical analysis in establishing a definitive diagnosis.

INTRODUCTION

Gastrointestinal stromal tumors (GISTs) are the most common mesenchymal neoplasms of the gastrointestinal tract, typically originating in the stomach or small intestine. However, extra-gastrointestinal stromal tumors (EGISTs), such as those occurring in the retroperitoneum, are exceedingly rare. Due to their atypical location, these tumors often pose diagnostic challenges and can mimic other retroperitoneal masses, such as paragangliomas, retroperitoneal sarcomas. This case highlights the diagnostic dilemma posed by a retroperitoneal GIST initially suspected to be a paraganglioma based on imaging findings.

Case Presentation

A 37-year-old male presented with right-sided abdominal pain for 25 days, which had worsened over the past 3 days. He also reported abdominal distension. The patient had no known comorbidities, with a history of hemorrhoidectomy 9 years prior. Physical examination revealed rigidity, severe right upper and lower quadrant tenderness without any palpable masses. Baseline blood investigations were within normal limits, apart from slight elevation in the liver enzymes- SGOT/SGPT- 58/76.

Radiological Investigation

A contrast-enhanced CT (CECT) of the abdomen revealed a solid, thick-walled, lobulated lesion measuring 11.4 x 8.3 x 11.0 cm in the paraaortic region of the retroperitoneum, likely arising from the Organ of Zuckerkandl. The lesion exhibited peripheral rim hyperenhancement during the arterial phase, with central areas of necrosis and hemorrhage.

Key findings included:

Displacement of adjacent structures: The mass displaced the third part of the duodenum anteriorly and compressed the inferior vena cava (IVC) significantly.

Extensions: The mass extended from the inferior end plate of L1 to L4 vertebra and was closely related to the uncinate process of the pancreas and the abdominal aorta.

No invasion: Despite the close proximity to major vessels and organs, there was no evidence of infiltration.

Imaging features, including the hyperenhancing nature of the mass and its vascularity, strongly suggested a retroperitoneal paraganglioma.

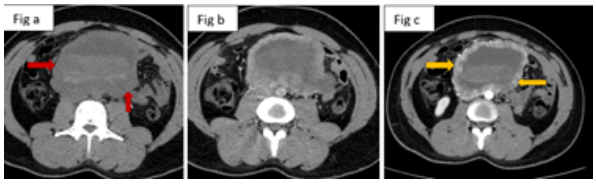

Figure 1: CECT Abdomen axial plain (a), venous (b) and arterial (c) sections of the abdomen showing a peripherally enhancing (yellow arrows), solid, lobulated lesion with central areas of necrosis and hemorrhage (red arrows) in the paraaortic region of retroperitoneum.

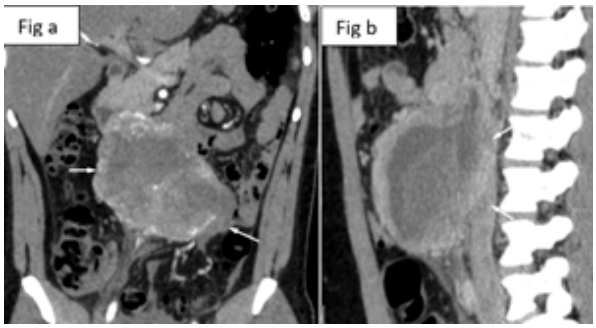

Figure 2: Coronal (a) and sagittal (b) venous images of the lesion showing its extent and relation with the adjacent structures.

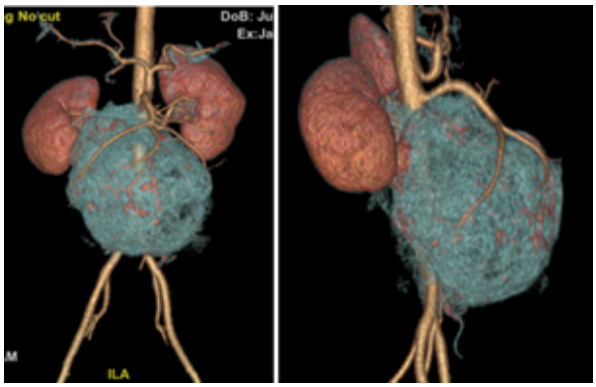

Figure 3: 3-D volume rendered imaging of the lesion

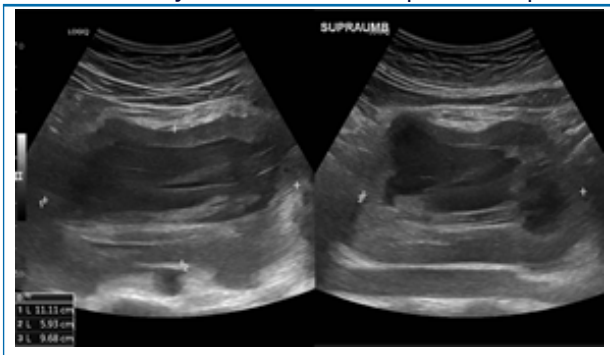

Figure 4: Ultrasound image of the retroperitoneal lesion extending upto the supraumbilical region confirming the central areas of necrosis.

Differential Diagnosis

The initial radiological impression suggested a retroperitoneal paraganglioma, but the following differential diagnoses were considered:

- **Paraganglioma**: Arising from the Organ of Zuckerkandl, paragangliomas are neuroendocrine tumors that exhibit hypervascularity and can present with necrosis and hemorrhage.
- **Retroperitoneal Sarcoma**: Sarcomas, such as leiomyosarcomas and liposarcomas, are common retroperitoneal masses, though the imaging features in this case were not entirely consistent with sarcoma.
- **Lymphoma**: Though retroperitoneal lymphomas are often large and painless, the hypervascular nature of this lesion made lymphoma less likely.
- **Gastrointestinal Stromal Tumor (GIST)**: While rare in the retroperitoneum, GISTs are a consideration when a hyperenhancing mass with spindle cells is encountered.

Surgical Intervention

Given the diagnostic uncertainty, the patient underwent excision biopsy of the retroperitoneal tumor under general anesthesia. Intraoperatively, the patient experienced significant blood loss and required transfusion of 3 units of packed red blood cells (PRBC) and 2 units of fresh frozen plasma (FFP). Vasopressor support with noradrenaline was initiated due to intraoperative hypotension.

Postoperatively:

The patient was shifted to the surgical ICU (SICU). Noradrenaline was tapered off by postoperative day (POD) 1 as the patient stabilized.

A nasogastric tube was reinserted on POD 2 due to abdominal distension. On POD 3, the surgical wound was found to be healthy, and the epidural catheter was removed.

Histopathological And Immunohistochemistry Findings

The histopathological examination (HPE) of the excised tumor revealed:

Spindle-shaped tumor cells arranged in fascicles and bundles within a fibrous stroma, with mild atypia.

Brisk mitotic activity (7 mitoses per 10 high-power fields) and areas of hemorrhage.

No necrosis was noted in the multiple sections studied.

An initial impression of a spindle cell neoplasm was made, and immunohistochemistry (IHC) was advised for confirmation.

The IHC panel showed:

- **CD117 (KIT)**: Positive
- **DOG1**: Positive
- **Desmin and SOX10**: Negative
- **Ki67 labelling index**: 30%, indicating high proliferative activity.

These findings confirmed the diagnosis of a gastrointestinal stromal tumor (GIST). Due to the fragmented nature of the specimen, grading was not possible, but the elevated Ki67 index suggested the need for close follow-up.

Treatment And Outcome

Following the histopathological diagnosis, the patient was started on Imatinib (400 mg/day), a tyrosine kinase inhibitor that targets KIT-positive GISTs. The patient's symptoms of abdominal pain and distension improved postoperatively.

The patient remains on Imatinib and continues to be monitored regularly for recurrence or progression. Follow up imaging after 6 months with CECT Abdomen revealed no residual/ recurrent lesion.

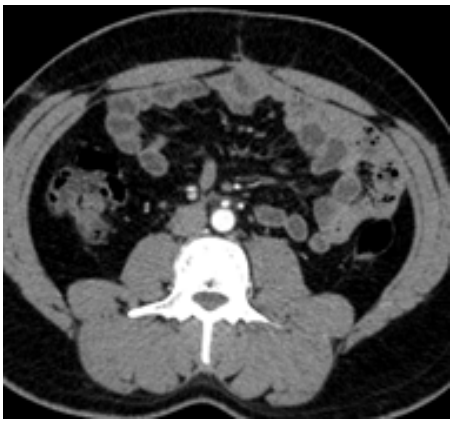

Figure 5: 6 months post operative imaging- CECT axial arterial section showing no residual lesion.

DISCUSSION

Gastrointestinal stromal tumors (GISTs) are the most common mesenchymal tumors of the gastrointestinal tract, with an estimated incidence of 10-20 cases per million people per year. They primarily arise from interstitial cells of Cajal or precursor cells, most commonly located in the stomach (approximately 60% of cases) and the small intestine (about 30%). However, extra-gastrointestinal stromal tumors (EGISTs) can occur in other locations, including the retroperitoneum, although they are much rarer. The estimated incidence of retroperitoneal GISTs accounts for approximately 5-10% of all GIST cases, representing a significant diagnostic challenge due to their atypical location and presentation.

Radiologically, GISTs are characterized by considerable variability in size, often presenting as well-defined, lobulated masses that can exceed 20 cm. On contrast-enhanced CT, they typically exhibit heterogeneous enhancement due to necrosis and hemorrhage, with peripheral rim enhancement correlating with their vascular supply. Central necrosis and hemorrhage are common, complicating differentiation from other tumors such as paragangliomas. GISTs can displace surrounding structures while maintaining fat planes, which is crucial for assessing organ infiltration. Internal calcification is uncommon, and lymphatic spread is rare, with predominant hematogenous spread to the liver and peritoneum. Accurate diagnosis relies on histopathological examination and immunohistochemistry, with GISTs showing positivity for CD117 (KIT) and DOG1. A Ki67 proliferation index reflecting high mitotic activity, such as the 30% seen in this case, indicates the need for close postoperative monitoring.

The case presented herein exemplifies the diagnostic difficulties associated with retroperitoneal GISTs, as imaging findings initially suggested a paraganglioma. Paragangliomas often present as hypervascular tumors, and their imaging characteristics can overlap with those of GISTs.

Given the rarity of retroperitoneal GISTs, the differential diagnosis must include not only paragangliomas but also other sarcomas, lymphomas, and neurogenic tumors.

CONCLUSION

This case highlights the rare presentation of a retroperitoneal GIST, which initially mimicked a paraganglioma based on imaging. It emphasizes the importance of histopathology and immunohistochemistry in the definitive diagnosis of retroperitoneal masses. Early recognition and treatment with Imatinib significantly improve outcomes in GIST patients, even when the tumor arises in atypical locations.

Learning Points

- **Atypical Location**: GISTs can occur in rare locations such as the retroperitoneum, mimicking other tumors like paragangliomas.
- **Imaging Limitations**: Radiological findings, while suggestive, may not always be sufficient to differentiate between tumors in the retroperitoneum.
- **Role of Immunohistochemistry**: Immunohistochemical markers such as CD117 and DOG1 are essential for confirming the diagnosis of GIST.
- **Treatment and Follow-Up**: Imatinib is an effective treatment for GIST, and close follow-up is necessary in cases with high mitotic activity and Ki67 index.

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