Unexpected Diagnosis of Worsening Abdominal Distension

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INTRODUCTION

- Gastrointestinal stromal tumors (GISTs) constitute about 1% of primary gastrointestinal cancers.
- Primary omental GIST without gastrointestinal involvement is extremely rare.
- We present case of a primary stromal tumor of the omentum with peritoneal metastases (extra-gastrointestinal stromal tumor or EGIST) and partial response to Imatinib.

CASE PRESENTATION

- A 68-year-old Caucasian male with a history of coronary artery disease and chronic obstructive lung disease presented with progressively worsening abdominal distension and shortness of breath for several months.
- Labs were significant for hemoglobin of 10.8 g/dL and positive fecal occult blood test.
- CT abdomen and pelvis revealed innumerable masses in the peritoneum and omentum. There was no evidence of solid visceral abnormality (Figure 1A).
- The patient underwent CT-guided core biopsy of the omental mass, which revealed cluster spindle cells. The cells were diffusely positive for c-kit (CD117) and CD34; negative for cytokeratin 5/6, D2-40, calretinin, cytokeratin AE1/3, Melan-A, S-100, and actin. These were consistent with EGIST (Figure 2).
- Subsequent esophagogastroduodenoscopy and colonoscopy did not reveal any neoplastic lesion.
- Imatinib was initiated, and his symptoms dramatically improved. In 3 months, follow-up CT scan showed partial remission, with the largest nodule measuring 3.8 x 2.4 cm (Figure 1B).

IMAGES AND LABORATORY INVESTIGATIONS

- Primary omental stromal tumors are extremely rare. The tumors without GI tract involvement or attachment are even rarer and only account for about 1/5 of the total primary omental tumors.
- Classic GISTs originate from the interstitial cells of Cajal (ICCs) or their stem cells. EGISTs share the same immunophenotype despite arising outside of GI tract. Evidence suggests the tumors may simply lose the original connection with GI tract. However, EGISTs can originate from ICC-like or precursor cells which accidentally dispersed during fetal development.
- The most common symptoms associated with omental stromal tumors are abdominal distension and pain. The tumors tend not to cause obstruction at earlier stages and are often found as large masses when symptoms develop.
- CT is the primary imaging modality for identification of the primary tumors and metastases. Endoscopy can be used to obtain additional information and biopsy.
- Due to the rarity of the condition, there are no established treatment guidelines for EGIST, and the guidelines developed for GIST are typically used for management of EGIST. Surgery is the modality of choice for localized disease. Imatinib was initiated in our case because of numerous non-resectable masses, but only partial response was observed. More studies are needed to better understand the pathophysiology and evaluate treatment options for EGIST.

CONCLUSIONS

- Primary omental stromal tumors are extremely rare. The tumors without GI tract involvement or attachment are even rarer and only account for about 1/5 of the total primary omental tumors.
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REFERENCES