Incidental Gastrointestinal Stromal Tumor of the Rectum found during Malignant Melanoma follow-up

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Objectives

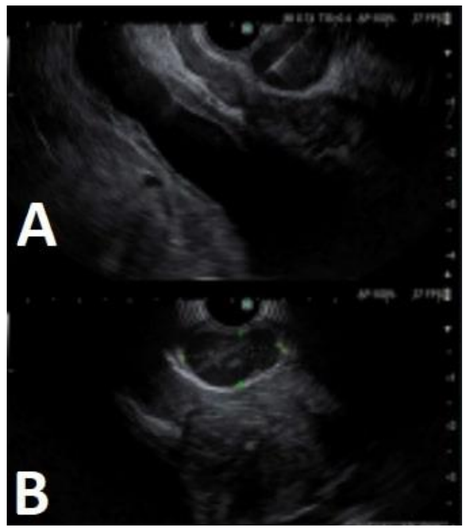
- To document a case of an uncommon GI tract tumor.
- To review pathophysiology of gastrointestinal stromal tumor (GIST).
- To review diagnostic and treatment modalities available for GISTs.

Introduction

- GISTs of the rectum are an infrequently reported phenomenon.
- The majority of GISTs occur in the stomach or the small intestine.
- Rectal GISTs encompass 5% of all GISTs and 0.1% of all rectal neoplasms, and generally occur in patients over age 50.
- We present a rare case of rectal GIST occurring in a 48-year-old male.

Imaging

Figure 1. Endoscopic ultrasound with fine needle aspiration of rectal lesion.



Patient Case

HPI:

- A 48-year-old male presented to the office with 1.5cm anterior rectal mass discovered on positive emission tomography (PET) scan.

ROS

- Positive: None.
- Negative: abdominal pain, change in bowel habits, nausea, vomiting.

Past Medical History: malignant melanoma

Past Surgical History: None

Physical exam:

- Digital rectal exam revealed firm anterior rectal mass, three to four cm from the anal verge.

Imaging

- Colonoscopy with endoscopic ultrasound demonstrated a 2.0cm submucosal nodule in the rectum and biopsied with cold forceps.
- The recto-sigmoid colon, hepatic flexure, ascending colon and cecum appeared normal.
- Sonography demonstrated a hypoechoic, non-circumferential lesion predominantly on the left anterior rectal wall.
- The origin appeared within the intramural wall, but the exact layer could not be determined.
- The lesion measured 1.9cm in thickness.
- An intact interface was seen between the lesion and the superficial mucosa suggesting lack of invasion.

Histology

- Histologic examination revealed spindle cell proliferation consistent with gastrointestinal stromal tumor.
- Pankeratin, SMA, desmin and S100 were negative.

Discussion

- GIST of the rectum is a rare malady.
- The majority of GISTs occur sporadically, although some have been associated with genetic syndromes.
- The exact etiology of GISTs has yet to be determined, but thought to be related to interstitial cells of Cajal and overexpression of KIT tyrosine kinase.
- Most GISTs originate from the muscularis propria and occasionally from the muscularis mucosa.
- Diagnosis of GISTs requires tissue analysis with the typical morphological features of spindle cell, epithelioid or pleomorphic mesenchymal tumors.
- CD117 immunohistochemical reaction supports diagnosis of GIST and predicts efficacy of tyrosine kinase inhibitor therapy.
- GISTs have also been associated with DOG-1, CD34, desmin, S100, alpha smooth muscle actin expression.
- CT scan with contrast is the gold standard for detection of GISTs, and guidelines for the treatment of GISTs have not yet been established due to the rarity of the disease.
- The current standard of care for treatment includes surgery for primary disease and additionally Imatinib for metastatic disease.

Conclusion

- While there is limited information regarding rectal GISTs, this case illustrates symptomatology and progression of this rare malady.
- Histology and immunohistochemical staining are key for diagnosis.
- Surgical resection is the mainstay of treatment.

References

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