Gastrointestinal Stromal Tumor (GIST): An Update on Its Uncommon and Complex Presentations

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ABSTRACT

Gastrointestinal stromal tumor (GIST) is recently diagnosed as a tumor entity. In the past, these tumors were classified as leiomyomas, leiomyosarcomas and leiomyoblastomas. GISTs are the rare soft tissue sarcomas of the gastrointestinal tract (GIT), accounting for 0.1-3% of all GIT tumors. The most common site of GIST is the stomach (60-70%) followed by small intestine (20-30%) and less common than 5% in colon, esophagus, omentum and mesentery. The epidemiology of GIST is not known completely. Most often these cases were detected on endoscopy, on imaging of the abdomen, at surgery for the other condition or at autopsy. The varied clinical presentations depend on the size of the tumor. Small GIST (less than 2cms) are asymptomatic and incidentally detected at endoscopy or at laparotomy. The large tumors give rise to vague abdominal discomfort or pain (20-50%), acute or chronic GI bleeding, intestinal obstruction (20%) or altered bowel habits and about 35% are detected incidentally. As per literature, the most common (50%) presentation is bleeding. Very large or malignant GIST presented as the exophytic palpable mass. Regarding unusual and complex presentations of GIST, we encountered two cases of GISTs in the twelve years of histopathology practice.

Keywords: Gastrointestinal Stromal Tumor, GIT, UNUSUAL, C-KIT

Introduction

Gastrointestinal stromal tumor (GIST) is recently diagnosed as a tumor entity. In the past, these tumors were classified as leiomyomas, leiomyosarcomas and leiomyoblastomas.¹ GISTs are the rare soft tissue sarcomas of the gastrointestinal tract (GIT), accounting for 0.1-3% of all GIT tumors.²

The most common site of GIST is the stomach (60-70%) followed by small intestine (20-30%) and less common than 5% in colon, esophagus, omentum and mesentery.³ The epidemiology of GIST is not known completely. Most often these cases were detected on endoscopy, on imaging of the abdomen, at surgery for the other condition or at autopsy.¹

In view of its complex and unusual presentations over the period of twelve years, we encountered two cases in sigmoid colon and jejunum respectively.

Discussion

The varied clinical presentations depend on the size of the tumor. Small GIST (less than 2cms) are asymptomatic and incidentally detected at endoscopy or at laparotomy.¹ The large tumors give rise to vague abdominal discomfort or pain (20-50%), acute or chronic GI bleeding, intestinal obstruction (20%) or altered bowel habits and about 35% are detected incidentally. As per literature, the most common (50%) presentation is bleeding. Very large or

malignant GIST presented as the exophytic palpable mass.¹

Regarding unusual and complex presentations of GIST, we encountered two cases of GISTs in the twelve years of histopathology practice. The first case was presented with perforation and peritonitis in sigmoid colon in a 70 year old male⁴. We reported this case in view of its rare site that is sigmoid colon and its unusual presentation as perforation, because colonic GIST rarely perforates. To the best of our knowledge, this was the second documented case of malignant GIST after Hwango Y et al⁵. The patient presented to surgery with pain in abdomen, on basis of clinical and radiological examination, emergency laparotomy revealed perforation of sigmoid colon and the mass protrudes through the defect. Spindle cell morphology and C-Kit(CD-117) positivity on immunohistochemistry (IHC) confirms the malignant GIST.⁴

Second case was very interesting in view of its mysterious presentation. A 78 year old male presented with per rectal bleeding and hypotension. The investigations revealed no specific pathology. In view of no relieved in symptoms, emergency life saving laparotomy was performed. The intraoperative findings showed jejunal mass protruding through wall with blackish areas of hemorrhage with multiple nodules on peritoneal suggestive of metastasis. Histopathologically revealed spindle cell morphology with moderate mitotic figures favored malignant GIST it was confirmed by CD-117.⁶

Light microscopically majority of the GIST have spindle cell morphology (80%) with fascicular or storiform pattern of growth.⁷ Rest (20%) are epithelioid cells with abnormal eosinophilic to clear cytoplasm.⁷ Surgical resection with negative surgical margin is the treatment of choice in small intestinal GIST and Imatinib mesylate is the standard regimen for inoperable and malignant/metastatic masses.¹

Recently, the new group of GIST was added is "SDH resistant GIST" by WHO in 2013, that is succinate dehydrogenase GIST. Cytogenetically mutation of KIT or PDGFRA occurs in micro GIST (1-10mm).⁸ GISTs are positive for CD-117 /c-KIT by IHC in 95% cases. New markers DOG1 and PKC-theta are expressed in KIT negative cases. SDH deficient GIST always occur in stomach and pediatric age group with indolent behavior even in presence of metastasis.⁸

Conclusion

The clinicians needs to aware of these varied ,unusual and mysterious presentations of GIST due to avoid misdiagnosis in view of its aggressive, fatal and malignant nature.

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Financial or other Competing Interests: None.

Date of Submission : 08.02.2017

Date of Acceptance : 16.02.2017

Date of Publication : 21.02.2017