CASE REPORT

Jejunal GIST with extramural hemangiomatous component presenting as an obscure lower GI bleed : a case report

Devang J. Desai · M. D. Kamath · P. J. Haldar

Received: 15 April 2006 / Accepted: 29 October 2007

Abstract GISTS (Gastrointestinal stromal tumours) are malignant form of sarcoma of the digestive tract. Various unusual presentations and morphological variants of GIST have been described, including extramural components of GIST. The literature has no reference regarding an extramural hemangiomatous component of a GIST. Here we present a case of 54-year-old woman with this unusual morphological condition presenting as an obscure lower GI bleed. Diagnosis in this case was made by subjecting the resected specimen to immunohistochemical analysis.

Keywords Gastrointestinal stromal tumours (GISTS) · Extramural hemangiomatous component · CD 117 · Imatinib mesylate

Introduction

GISTS are rare form of soft tissue sarcoma of the digestive tract – commonly occurring in the stomach, but can crop up anywhere in the digestive tract. Usually they are asymptomatic but can present as abdominal pain, bleeding or mechanical obstruction. Exophytic growths of these tumours have been noted in 18–30 % of cases [1]. Here we present a case of a jejunal GIST with an extramural hemangiomatous component presenting as obscure lower GI bleed.

Case report

A 54-year-old woman presented to us with complaints of weakness, breathlessness on exertion since 1998 and recurrent episodes of melena since 2002 – for which patient received 17 blood transfusions in the past. She had history of hematochezia at presentation to our hospital. There was no history of upper GI complaints, hematemesis, alteration in bowel habits or history of abdominal lump or distension. On examination patient was pale but general examination including vital parameters were within normal limits. Systemic examination including abdominal examination, PR and PV examinations were essentially normal. She had normocytic hypochromic anemia with Hb 9.2, USG abdomen was normal, barium studies showed rapid transit time but no lesion. Upper and lower GI endoscopies were normal. She had undergone a RBC ⁹⁹Tc scan before presenting to us showing bleeding from branches of the superior mesenteric artery. She was subjected to a MR Angiography (Fig. 1), which showed a hypervascular mass in the distal jejunum and the proximal ileum with a luminal narrowing and an exophytic component with AV shunting, abutting against sigmoid colon, but fat planes were maintained. We undertook an exploratory laprotomy where we resected a mass from the jejunum showing both an intra and an extra

D. J. Desai · M. D. Kamath · P. J. Haldar
Department of Surgery and Gastrointestinal Surgery,
Jagjivan Ram Hospital,
Mumbai - 400 008,
India

D. J. Desai (✉)
e-mail: docdevang@gmail.com
Tel: +91 / 22 / 23877513

mural component (Figs. 2 and 3). On subjecting it to histopathological and immunohistochemical analysis we found a jejunal GIST with an intramural component attached to an extramural hemangiomatous component. It was CD 117 and CD 34 positive, with mitotic figures of 3/50 hpf. The patient had an uneventful recovery and is asymptomatic with no recurrence detected on investigations at a one-year follow up.

Discussion

GISTs are rare nonepithelial primitive digestive tumours, first identified by Mazur and Clark in 1983, arising from the connective tissue of the digestive tract with an incidence of 0.1–1 % of gastrointestinal malignancies.

Primary mesenchymal tumours of the digestive tract can be divided into following major categories according to their phenotypic features (1) Tumours with differentiation towards smooth muscle cells [I.H.C. – (alpha) smooth muscle actin and desmin] (2) Tumours with neural differentiation (I.H.C. – S 100 protein) (3) Tumours with no differentiation – GIST [2]. GISTs are believed to arise from the interstitial cells of Cajal (pacemaker cells of GIT) and hence were termed in the past as GANT (Gastrointestinal Autonomic Nerve Tumours). Age of onset is between 50–70 years without any sex predilection, 25 % of GISTs are malignant. GISTs have been associated with pulmonary chondroma and extra adrenal paraganglionoma (Carney's triad), with HIV infection, in von Recklinghausen's disease and bilateral pheochromocytoma [3], in MEN – 1 duodenopancreatic gastrinoma, pituitary adenoma, adrenal adenoma, parathyroid hyperplasia and lipomas [4].

The clinical manifestations depend upon the site and size of the GIST. About 5 % are oesophageal, 50–70 % are in the stomach, 25–40 % in the small intestine (10–20 % in the duodenum, 27–37 % in jejunum, 27–53 % in ileum) and < 10

% are colorectal (50 % colonic 50 % rectal). 60 % are submucosal, 30 % are subserosal and some are intramural [5].

Most GISTs are clinically silent till they grow large, bleed or rupture. They may be detected incidentally on imaging, endoscopy or surgery [5]. Some of the presentations include primary mass, abdominal pain, GI bleed or as metastasis. Ulceration of the mucosa over a GIST may cause bleeding. Exophytic and extramural GISTs have been noted in 18–30 % of cases [1]. Various unusual presentations reported are as – gastroduodenal GIST presenting as gastric outlet obstruction [6], GIST found in Meckel's diverticulum [7] and hemoperitoneum due to rupture of GIST [8].

Small bowel GISTs have higher propensity to have malignant behaviour than gastric GISTs [5]. Malignant

Fig. 2 MR Angiography showing hypervascular mass being supplied by branches of Sup. Mesenteric Artery A-V shunting present

Fig. 1 Extra-mural hemangiomatous component of Jejunal Gist. Intra-mural component of Jejunal Gist.

Fig. 3 Extra-mural hemangiomatous component of Jejunal Gist

GISTS recur locally and spread mainly to adjacent organs, omentum, mesentery, retroperitoneum, liver, and to lymph nodes, extra abdominally to lung and rarely to bone. Upto 30 % of GISTS are metastatic on presentation. Prognostic criteria laid down (Berman 2001; Fletcher et al 2002) are as follows (Table 1)

Other proposed prognostic factors are mucosal and muscularis propria invasion, ploidy, proliferation markers (PCNA, Ki 67), necrosis, hemorrhage, degeneration and calcification (which can be found in benign GISTS) [5], cellularity, pleomorphism and immunophenotype. No single factor is predictive enough to be used exclusively.

It has been shown that the c-kit protooncogene located on chromosome 4q11-21, encodes a type III receptor tyrosine kinase protein (CD117) is expressed in 81–100 % of benign and malignant GISTS. CD 117 positivity has become definitional in the diagnosis of GISTS. Metastasis may lose CD 117 positivity. CD 34 positive GISTS have a more favourable prognosis.

Preoperative diagnosis of GIST is difficult. CT and MR Angiography are helpful in clinching preoperative diagnosis. US endoscopy and FNA with subsequent immunohistochemical analysis along with c-kit gene mutation study offers the best diagnostic accuracy [9]. RT-PCR and cDNA array hybridization studies of target genes have been the latest modalities of distinction between benign and malignant GISTS.

Resection offers the best treatment option. Unresectable and/or metastatic GISTS (CD 117 +ve) are treated with Imatinib mesylate (tyrosine kinase receptor inhibitor). Doxorubicin based chemotherapy have shown good results [5].

Follow up of patients is lifelong as recurrences are noted even after 30 years of primary resection.

Our patient presented as an obscure lower GI bleed due to an extramural hemangiomatic component of an intramural jejunal GIST, CD 117 and CD 34 positive, with mitotic figures of 3/50 hpf, without any evidence of metastasis. The review of literature points to an observation in which distinctive hemangioma-like, sometimes glomerular, vascular proliferation between sheets of tumour cells was seen in 37 cases [10], but in these cases there wasn't any

gross hemangiomatic component as seen in our case. In one case, the vascular proliferation contained a focus indistinguishable from vascular invasion [10], while in our case the extramural component contained malformed blood vessels with A-V shunting.

In general gastric GISTS are spindle celled in 70 to 80 % of cases and epitheloid in 20–30 %, while small intestinal GISTS are usually spindle celled, usually the benign ones. Esophageal and rectal GISTS are usually malignant and predominately spindle celled [5]. In our case the jejunal GIST was benign but had spindle cell histology.

Conclusion

GISTS are rare tumours of GIT, which are difficult to diagnose preoperatively. Surgery offers definitive treatment and regular follow up is required lifelong post resection.

Their unusual presentations must be borne in mind. An unusual extramural hemangiomatic component of an intramural jejunal GIST, presenting as an obscure lower GI bleed is presented in this case report.

References

1. L. Giglio, G. Andreoli, V. Cantisani, et al (2001) GIST: evaluation by Spiral CT and Angiography. *Eur J Radiol* 3(6):250–261
2. Miettinen M, Virolainen M, Sarlomo-Rikala M (1995) Gastrointestinal stromal tumours – Value of CD34 antigen in their identification and separation from true leiomyomas and schwannomas. *Am J Surg Pathol* 19:207–16
3. Rizzo S, Bonomo S, Moser A, et al (2001) Bilateral pheochromocytoma associated with duodeno-jejunal GIST in patient with von Recklinghausen's disease: report of a clinical case. *Chir Ital* 53(2):243–46
4. Pappillon E, Rolachon A, Calender A, et al (2001) A malignant GIST in a patient with multiple endocrine neoplasia type I. *Eur J Gastroenterol Hepatol* 13(2):207–211
5. Mukhopadhyay S, Gupta SD (2002) Gastrointestinal Stromal Tumours: Bench-to-bedside review. *GI Surgery Annual* 9: 101–148
6. Crowther KS, Wlyd L, Yamani Q, Jacob G (2002) Case report: gastroduodenal intussusception of a GIST. *BJR* 75(900):987–988
7. Johnston AT, Khan AL, Beakney R, Keenan RA (2001) Stromal tumour within a Meckel's Diverticulum: CT and ultrasound findings. *BJR* 74(888): 1142–1144
8. Dubenec SR, Dawes-Higgs RJ, Truskett PG (2001) Haemoperitoneum caused by spontaneous rupture of a GIST. *ANZ J Surg* 71(10):612–614
9. Lattarulo S, Ugenti I, Ferrarese F, Fabiano G (2003) GIST : current issues. *Chir Ital* 55(2):219–226
10. Miettinen M, Kopeczynski J, Makhlof HR, et al (2003) Gastrointestinal stromal tumours, Intramural leiomyomas, and leiomyosarcomas in the duodenum. *Am J Surg Pathol* 27(5):625–641

Table 1 Prognostic Criteria for GIST (Berman 2001 ; Fletcher et al 2002)

	Size in cm.	Mitotic count/50 hpf
Very low risk	< 2	< 5
Low risk	2–5	< 5
Intermediate risk	< 5	6–10
	5–10	< 5
High risk	> 5	> 5
	> 10	any
	any	> 10