

Synchronous sporadic gastrointestinal stromal tumors (GISTs) of the colon

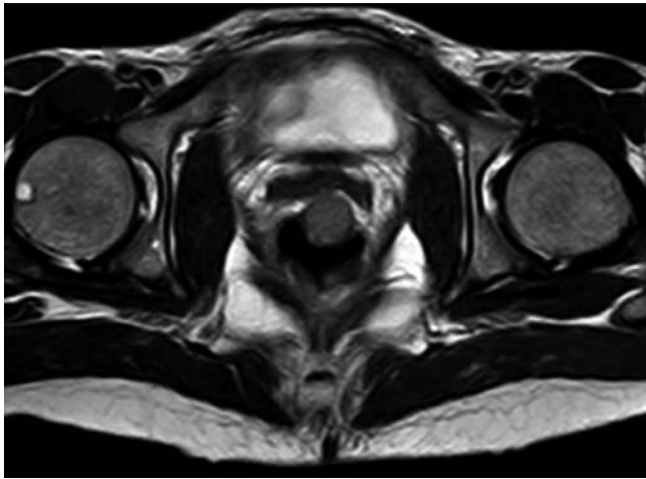


Fig. 1 Magnetic resonance imaging (MRI) scan showing a rectal gastrointestinal stromal tumor (GIST).



Fig. 2 Colonoscopy views showing: **a** a 2-cm gastrointestinal stromal tumor (GIST) in the rectum; **b** a 2.5-cm GIST in the transverse colon.



Gastrointestinal stromal tumors (GISTs) are rare mesenchymal tumors of the gastrointestinal tract originating from the cell of Cajal [1]. Up to 75% of cases are driven by constitutional activation of the proto-oncogene *cKIT*; 10% by a mutation

of the gene encoding for the platelet-derived growth factor receptor (*PDGFRA*); 12% are wild-type and their pathogenesis has been related to mutations of the succinate dehydrogenase complex, *BRAF* or *NF1* genes. Commonly GISTs arise as a

solitary lesion; multiple spread is extremely rare and usually associated with familial GIST, type 1 neurofibromatosis (NF-1), or Carney's triad [2].

In July 2012, a 49-year-old healthy woman underwent a medical consultation after an episode of rectal bleeding. On examination she was found to have external hemorrhoids; digital rectal examination revealed an incidental finding of a 2-cm nodule in the rectovaginal area, which was later confirmed by a pelvic magnetic resonance imaging (MRI) scan (Fig. 1). A colonoscopy showed a 2-cm solid lesion at 6 cm from the anal margin (Fig. 2a) and a second solid lesion of 2.5 cm in the transverse colon (Fig. 2b), both of which were covered with normal mucosa. An abdominal computed tomography (CT) scan confirmed the colonic lesion (Fig. 3).

The patient underwent resection of the transverse colon and enucleation of the rectal lesion. Histopathological examination of the colonic specimen showed a GIST of 1.5 cm, with 1 mitosis per 50 high power fields (HPFs). Molecular analysis revealed a mutation in exon 11 of the *cKIT* gene. The rectal lesion was a GIST of 2 cm with 2 mitoses per 50 HPFs and a mutation in exon 9 of *cKIT*. Both GISTs were low risk for recurrence so no adjuvant therapy was given. The patient was started on clinical and radiological follow-up and is free from disease more than 1 year after her surgery.

Multiple sporadic GISTs not related to familial or NF-1 syndromes are a rare but recognized clinical presentation of GISTs and can occur as synchronous or metachronous spread. After the first publication, which reported five cases of multiple sporadic GISTs [2], other groups reported their experience, which showed the interest of experts in gaining a better understanding of the pathogenesis and clinical outcome of this peculiar presentation [3–6]. Synchronous second tumors have been reported in 13% of patient with GISTs and consideration of a differential diagnosis is mandatory for a correct therapeutic approach [7]. A molecular analysis is needed to distinguish between multiple primary GISTs and multiple recurrence or metastatic GISTs [7, 8]. To our knowledge this is the first report of sporadic synchronous GISTs originating in the colorectal tract.

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Fig. 3 Computed tomography (CT) scan showing the gastrointestimal stromal tumor (GIST) in the transverse colon (red arrow).

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