Synchronous sporadic gastrointestinal stromal tumors (GISTs) of the colon

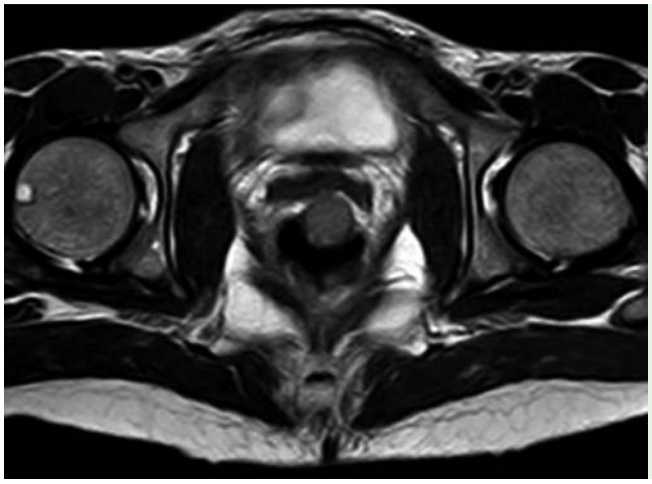

Fig. 1 Magnetic resonance imaging (MRI) scan showing a rectal gastrointestinal stromal tumor (GIST).

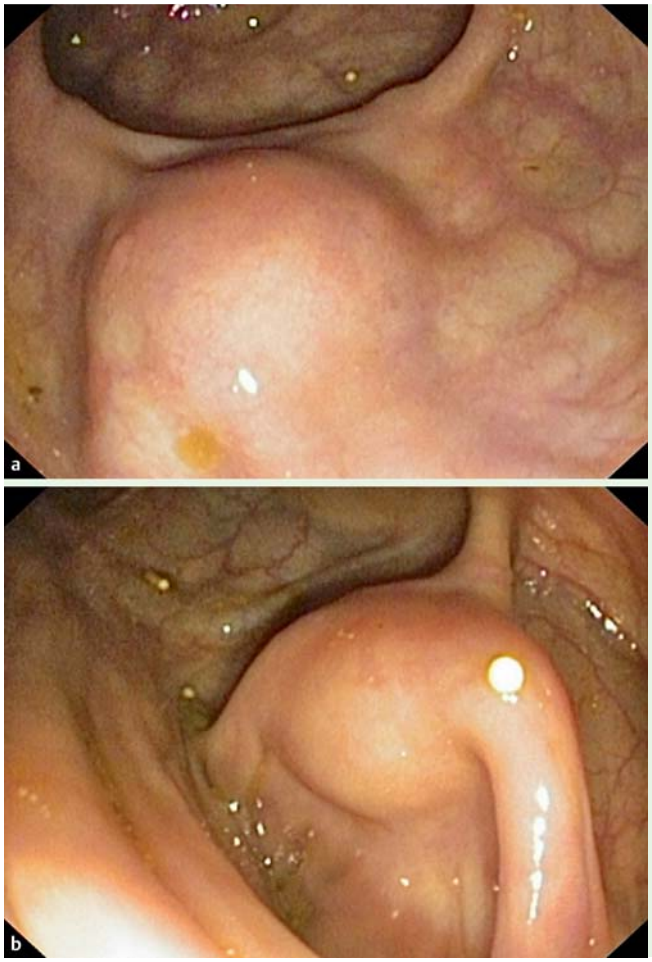

Fig. 2 Colonoscopy views showing: **a** a 2-cm gastrointestinal stromal tumor (GIST) in the rectum; **b** a 2.5-cm GIST in the transverse colon.

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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Competing interests: None

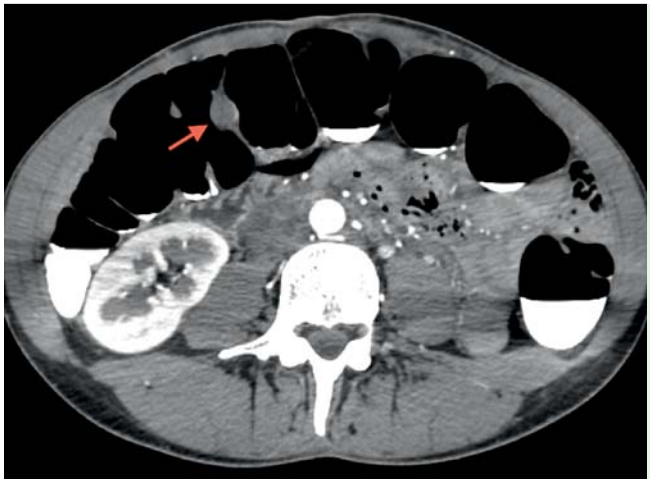

Fig. 3 Computed tomography (CT) scan showing the gastrointestinal stromal tumor (GIST) in the transverse colon (red arrow).

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