Lower GI bleeding secondary to a stromal rectal tumor
(rectal GIST)

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CASE STUDY

We present the case of a 64-year-old woman without any known allergies to drugs, who had smoked 20 cigarettes a day for more than thirty years, and with no other relevant history, who presented to our Department of Gastroenterology complaining of rectal bleeding and secondary ferropenic anemia with a hemodynamic repercussion. A colonoscopy was made to reveal the presence of a polypoid, submucous, ulcerated lesion in its vertex (8 cm from the anal margin) (Fig. 1).

An endoanal ultrasound scan showed a heterogeneous mass located in the posterior wall of the rectum, approximately 7 cm in size, with no infiltration of perirectal fat (Fig. 2). A biopsy was made with a tru-cut needle, and the pathological study showed a proliferation of fusiform cells, with no mitoses or atypias, and strongly positive for the CD-117 marker; staining by other markers (CD-34, desmine, actine, and S-100) was negative, except for one slight ki-67-related positivity, less than 10%. An abdominal CAT scan revealed no metastases at all levels. With a preoperative diagnosis of rectal stromal tumor, the mass was removed by local excision with preservation of the rectum. The patient is currently in the eighteenth month of follow-up, and has no signs or symptoms of relapse, neither locally nor distally.

DISCUSSION

The incidence of GISTs is greatest in the fifth and sixth decades of life. Histologically they are made up of fusiform, epitheloid, or mixed cells. The diagnosis is confirmed by immuno-histochemical techniques, and by the expression of a
tyrosine-kinase receptor (Kit or CD-117, the product of gene c-kit). The presence of ADN mutations may result in an abnormal receptor that is constantly activated, even in the absence of an appropriate ligand, which results in uncontrolled cellular proliferation. Sixty to seventy percent of GISTs also have concurrent positivity for CD-34. A group of GISTs exist that have a mutation in PDGF-FRA (a receptor derived from platelet growth factor alpha), and these are negative for c-kit (1,2). A differential diagnosis with other mesenchymal tumors is necessary, and is based on negativity for desmine and actine (positive in leiomyomas), and a negative S-100 protein (positive in schwannomas) (3).

Approximately 30% of GISTs are malignant, but clear criteria for malignancy do not exist. Size (> 5 cm) and mitotic index (> 5 mitoses/50 fields) are most useful morphologic features. Other factors for malignancy include location (stomach, better prognosis), the presence of necrosis areas, hemorrhage, hypercellularity, nuclear atypias, etc. The monoclonal antibody Ki-67 is also used as a predictive marker on the basis of its capacity to detect the presence of a nuclear antigen that is only expressed in proliferative cells (if > 10% of cells, it is associated with a worse prognosis). Endoscopic appearance usually includes the presence of a submucosal lesion that is covered with normal mucosa, with an apical ulcerated area. This renders the usefulness of endoscopic biopsies low, and thus frequently insufficient to tell benign from malignant lesions. In echo-endoscopy these lesions appear as hypoechoic areas originating in the muscular layer itself or the muscularis propria. Endoscopic findings associated with a good prognosis include size smaller than 3 cm, lesion homogeneity, and presence of regular contours. Nevertheless, no definitive echo-endoscopic features allowing differentiation between GISTs and leiomyomas are currently available.

Treatment is mainly surgical. Survival at 5 years after surgery ranges between 20 and 80% (4). Relapse is frequent, both local and metastatic, mainly in the liver, followed by the lungs and bones.

The treatment of non-resectable tumors has changed in recent years as we now have a tyrosine-kinase STI-571 inhibitor (imatinib mesylate), which manages to reduce tumor size, or at least to control tumor progression, in 80-90% of cases (5).

In summary, we describe the case of a considerably sized rectal GIST with a low mitotic index, and without associated metastases, which was removed surgically. Clinical follow-up has to be necessarily prolonged in such a case, due to a high risk of relapse even in the long term.

REFERENCES