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Mucosal Schwann cell hamartoma: a benign and little-known entity

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Dear Editor,

A 50-year-old female with a personal history of mutation of the BRCA1 gene and previous prophylactic double anexectomy consulted for rectal bleeding without pain during two weeks. A blood test was performed, with hemoglobin levels of 13.1 g/dl and no iron deficiency. There were neither external hemorrhoids nor anal fistulas in the anal inspection, so a colonoscopy was requested. All the colon mucosa was normal in the colonoscopy. However, internal engorged hemorrhoids were found in the rectal retroflexion, and indurated mucosa was found surrounding the 50 % of the anal opening an erythematous (Fig. 1). Biopsies were taken and the pathology report described proliferation of spindle-shaped cells exclusively in the lamina propria, with eosinophilic cytoplasm and unclear cell borders (Fig. 2). No nuclear atypia or mitotic activity were observed. On immunohistochemistry, S-100 protein was strongly positive (Fig. 3) and CD34, SMA, EMA and c-kit were negative. These results were concordant with the diagnosis of Schwann cells in the context of a mucosal Schwann cell hamartoma (MSCH). Given that these lesions do not seem to have malignant potential,

the patient was discharged without control colonoscopies. The episodes of rectorrhagia were attributed to the presence of internal hemorrhoids.

Discussion

MSCH are benign and intramucosal tumors with a mesenchymal origin (1). They are most commonly located in the distal colon, but they were also found in the gallbladder, the esophagogastric union and in the antrum. They are most frequently observed in middle aged women (around 60 years-old) and are generally asymptomatic. They present as polyps between 1 and 6 mm, but in other cases they appear as small whitish nodules, protruding lesions with normal superficial mucosa or even they were found in random biopsies of the colon (2).

The MSCH are a rare entity with an unknown prevalence. Less than 100 cases are described in the literature. The differentiation between this entity and the schwannomas or the gastrointestinal stromal tumors (GIST) is essential. Schwannomas are rare in the colon, they are well circumscribed (in contrast with the MSCH) and they are not limited to the lamina propria. GISTs are more frequently located in the stomach and they are positive for c-kit (3,4). MSCH are not associated with hereditary syndromes such as neurofibromatosis and, in contrast with schwannomas or GIST, they do not require surveillance because they are benign.

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Fig. 1. Endoscopic image of mucosal Schwann cell hamartoma.

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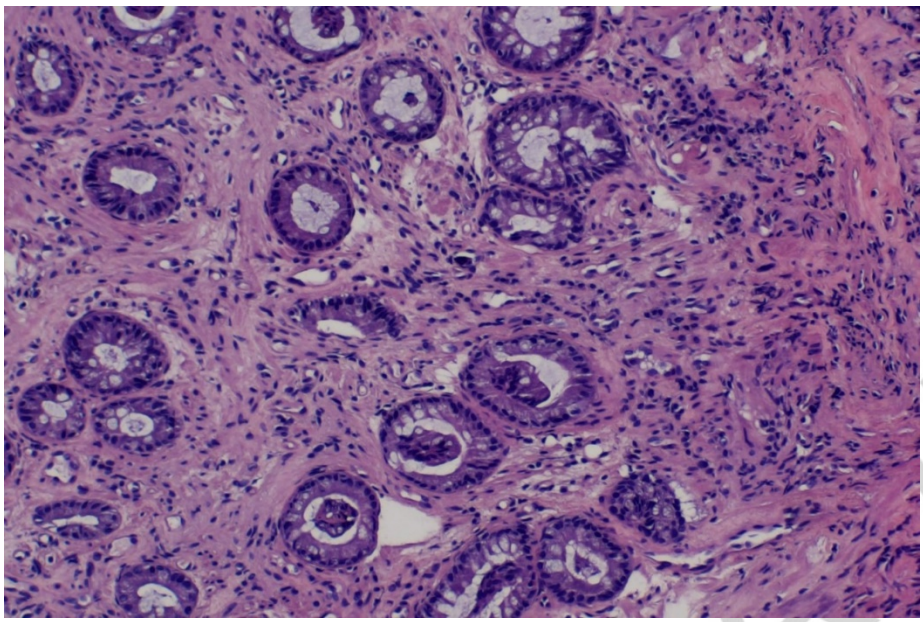

Fig. 2. Hematoxylin and eosin stain.

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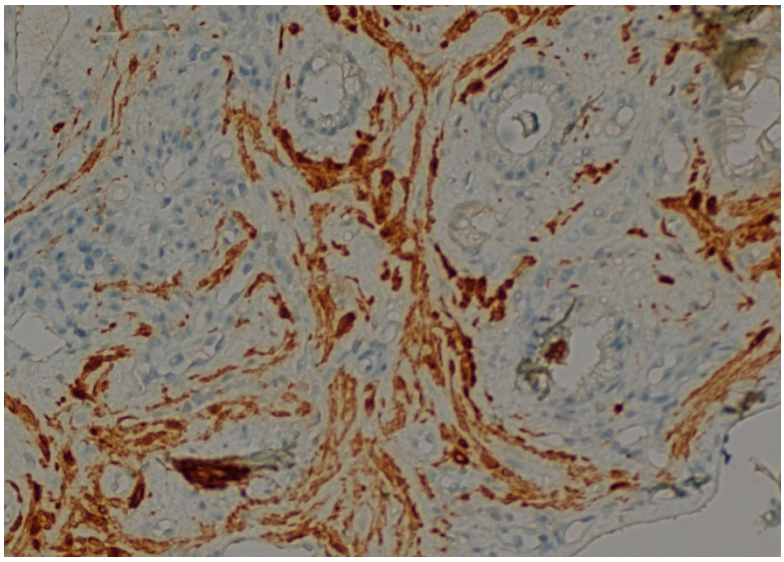

Fig. 3. Immunohistochemical positivity of S-100 protein.