CASE REPORT

When a polyp is not a polyp: incidental finding of a sigmoid schwannoma at first colonoscopic screening

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SUMMARY

Schwannomas, peripheral nerve sheath tumours arising from Schwann cells, are often associated with inherited disorders such as neurofibromatosis. Gastrointestinal schwannomas, while rare, have been reported in those without personal or family history of neurofibromatosis. Diagnoses of these lesions, however, typically follow evaluations prompted by symptomatic presentations associated with abdominal pain, rectal bleeding, change in bowel habits or positive results on faecal occult blood tests performed for colorectal cancer screening. Further, management of these predominantly benign lesions commonly incorporates surgical resection. We present the case of a sigmoid schwannoma found in an asymptomatic individual on first screening colonoscopy and treated with complete endoscopic polypectomy with anticipated surveillance colonoscopy.

BACKGROUND

We believe this case report offers an account of a rare lesion found in an asymptomatic individual with subsequent treatment by a modality not typically used for this type of lesion and which may be a viable, yet underused, alternative in patients manifesting multiple and complex medical comorbidities.

CASE PRESENTATION

A 70-year-old Caucasian man presented to the endoscopy suite for his first screening colonoscopy. He had no gastrointestinal (GI) symptoms, nor did he exhibit any extraintestinal manifestation of a schwannoma. He had no preceding screening, including faecal occult blood testing, and had elected to directly undergo complete colonoscopy as his first examination for screening of colorectal cancer. The patient had no history of prior malignancy, nor was there a family history of malignancy, either GI or otherwise. Significant medical history included coronary atherosclerosis, hypertension, hypercholesterolaemia, chronic kidney disease, psoriatic arthritis and hypoxemic chronic obstructive lung disease requiring continuous oxygen infusion. Medications included aspirin, amlodipine, lovastatin and albuterol inhaler as needed, with previous use of meloxicam and naproxen. Physical examination, including digital rectal examination, was unremarkable and did not reveal any palpable masses or any other abnormal findings.

Colonoscopy was performed and a 0.5 cm sessile, smooth polypoid lesion was detected in the sigmoid colon. The remainder of the colon was notable only

for two diminutive-to-small-sized polyps in the hepatic flexure and descending colon (respectively), a small area of non-bleeding ulcerated mucosa in the proximity of the ileocaecal valve and moderate diverticulosis. All polyps, including the aforementioned polypoid lesion, were completely removed by polypectomy; the ulcerated area was biopsied. Colonoscopy was completed and the patient was discharged per protocol from the endoscopy suite following recovery from procedural sedation.

Pathology of the polypoid lesion revealed a benign schwannoma, confirmed by immunohistochemical staining with S100, confirming the neural nature of the tumour, and absence of both CD117 and actin expression, ruling out GI stromal tumour (GIST) and leiomyoma, respectively (figure 1), with higher magnification demonstrating typical features of a neural tumour (figure 2). The remaining polypoid lesions were determined to be benign colonic mucosa with underlying nodular lymphoid aggregates. A biopsy of the ulceration demonstrated benign colonic mucosa with ulceration and reactive/regenerative changes, with no malignancy or dysplasia identified.

To optimise ulcer healing and to prevent further ulceration, it was recommended that the patient avoid non-steroidal anti-inflammatory and aspirin-containing agents. Multiple options regarding management of the incidentally found schwannoma were then considered, including whether further evaluation or work-up was indicated. Ultimately, because of the lesion's benign nature, subcentimetre size, location, pathological confirmation of

Figure 1 (A) Well-circumscribed neoplasm composed of spindle cells with a lymphoid aggregate in the centre (white arrow). (B) Absence of CD117 expression. (C) Absence of actin expression. (D) S100 expression (positive staining with brown chromogen); (20× total magnification (A–D)).

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Figure 2 Typical neural histological features of a schwannoma with fibrillar cytoplasm with ill-defined cell borders, vague nuclear palisading and interspersed collagen bands (100× total magnification).

complete removal and absence of any personal or family history of disease, as well as the recognition and consideration of the lack of existence of accepted guidelines for surveillance in this setting, observation with interval surveillance colonoscopy at 5 years was recommended, with further counsel to the patient that colonoscopy should be repeated sooner if the patient exhibits symptoms. At 1 year following endoscopic removal of the schwannoma, the patient is doing well and is without GI-related complaints or issues.

INVESTIGATIONS

Colonoscopy; histopathological evaluation.

DIFFERENTIAL DIAGNOSIS

Colon polyp (tubular adenomatous polyp, tubulovillous polyp, hyperplastic polyp, serrated polyp), GIST, leiomyoma, schwannoma.

TREATMENT

The patient underwent complete polypectomy, with confirmation by histopathological evaluation. He will be observed and undergo repeat colonoscopy in 5 years, with further timing of repeated colonoscopy determined by findings noted at that time, in similar fashion to those guidelines issued by the American College of Gastroenterology regarding interval colonoscopy for surveillance following polypectomy of tubular adenomatous polyps, acknowledging that no current guidelines exist for endoscopic surveillance of small asymptomatic schwannomas such as that described in this case report and taking into account the distinction of histopathology, molecular behaviour, size and inherent low likelihood of malignant transformation.¹

OUTCOME AND FOLLOW-UP

The patient was discharged per protocol from the endoscopy suite following completion of colonoscopy and recovery from procedural sedation. At 1 year following endoscopic removal of the lesion, he is doing well and remains without GI-related complaints or issues.

DISCUSSION

Schwannomas are peripheral nerve sheath tumours that arise from Schwann cells. They are typically well-circumscribed, slow-growing and benign; however, they can cause mass effect if growth is restricted by bony architecture. Schwannomas most commonly develop in the cerebellopontine angle, along the

vestibular branch of the eighth cranial nerve.² It is rare to find a schwannoma in the GI tract, and rarer still, to find one in the sigmoid colon.

Schwannomas comprise only 0.4%–1% of submucosal tumours of the GI tract.³ It has been suggested that these tumours arise from the Auerbach's nerve plexus in the intestinal wall.⁴ GI schwannomas occur most frequently in the stomach and in the small intestine; however, although uncommonly found in the colon, incidences of isolated schwannomas of the caecum, sigmoid, rectosigmoid, transverse colon, descending colon and rectum have been reported.

Men and women are affected equally, with median age at diagnosis of 65 years.⁵ The majority of reported schwannomas are benign, however, they can rarely undergo malignant transformation.^{5–8} Tumour size, high rate of mitoses and atypical mitotic figures of the tumour cells are best predictors of malignancy, with risk of malignant transformation increasing with tumour diameters ≥ 5 cm.^{4,9}

Grossly, GI schwannomas may appear rubbery and glistening on endoscopy, often similar in appearance to soft tissue schwannomas. They may share gross characteristics with GISTs and leiomyomas, tumours existing at a higher prevalence within the GI tract and for which schwannomas may be mistaken. At small and diminutive sizes, they may look extraordinarily like benign-appearing polyps, making the diagnosis impossible to determine strictly on gross appearance. The lesion described in this case report appeared quite polypoid, sessile, with shared features of the surrounding normal colonic mucosa and perceived grossly to be a colon polyp; the similarity in appearance emphasises the fact that the diagnosis of a GI schwannoma is reliant on histopathological evaluation and characterisation.

Schwannomas must be distinguished from other mesenchymal tumours, including GISTs and leiomyomas. Endoscopic ultrasonography (EUS) can be used to attempt diagnosis, however, all three tumour types are hypoechoic, round and found in the submucosa, making differentiation difficult, with GI schwannomas, in particular, lacking pathognomonic endoscopic features.^{3,10} Because EUS is not definitive, evaluation of histopathology remains integral to diagnosis.

Schwannomas follow two histological growth patterns. The Antoni A pattern shows densely packed spindle cells with oval nuclei and cytoplasmic processes (Verocay bodies). The Antoni B pattern demonstrates a loose meshwork of cells, microcysts and myxoid stroma.^{5,11} The Schwann cell is immunoreactive to the S-100 protein and the Leu7 antigen. Schwannomas typically show low affinity for CD34, KIT, cytokeratins AE1 and AE3, alpha smooth muscle actin, desmin and chromogranin. In contrast, GISTs are positive for KIT, and leiomyoma are positive for alpha smooth muscle actin and desmin, with the latter negative for KIT.^{9–11}

Presenting symptoms for a GI schwannoma have been reported to include abdominal pain, intussusception, rectal bleeding, intestinal obstruction and altered bowel habits. Surgical resection is largely accepted as conventional treatment of benign schwannomas, particularly for large lesions, as, though it is unlikely, the rare possibility for malignant transformation exists. Resection has also been described for definitive treatment of large lesions causing obstruction, haemorrhage and risk for intestinal perforation. Endoscopic polypectomy has been considered as a method of excision for small, benign lesions.¹²

While resection appears to be the preferred and most frequently undertaken course of action with respect to definitive treatment of incidentally found schwannomas in the lower GI tract in the literature, our current knowledge of the behaviour

and molecular underpinnings of this disease process, coupled with thorough consideration of the influence of lesion size and histopathological characteristics on risk for malignant transformation, lead us to support the option of observation via surveillance colonoscopy as an adequate and appropriate intervention following complete endoscopic excision of smaller lesions, as in the case described above.

At the time of creation of this case report, a total of 473 publications were generated as a result of a search for the term 'gastrointestinal schwannoma' in the PubMed database. Of these 473 articles concerning GI tract schwannomas, 32 described schwannomas that were colonic in location and diagnosed in either (a) symptomatic individuals presenting with abdominal pain, intestinal obstruction, frank bleeding or change in bowel habits, (b) individuals presenting following positive faecal occult blood testing or (c) individuals with known diagnoses of extraintestinal neurofibromatosis undergoing surveillance endoscopy. Only four colonic schwannomas have been described as incidentally found in asymptomatic individuals presenting specifically and solely for elective colonoscopic screening.^{13–16} Of these four schwannomas in asymptomatic patients, two were treated with segmental resections and two via polypectomy, the latter, in similar fashion to the patient described herein.^{13–16}

Learning points

- ▶ Small schwannomas of the colon may appear grossly similar to more commonly found polypoid lesions during colonoscopy.
- ▶ Definitive diagnosis of a colonic schwannoma found on colonoscopy should be based on histopathological characterisation.
- ▶ Observation following complete polypectomy with potential surveillance colonoscopy might be considered an acceptable treatment option for small, incidentally found colonic schwannomas.

Contributors LPM is the pathologist involved in case; assisted with imaging and discussion regarding histopathology, figures and editing. AL is the medical

student involved who assimilated data, assisted in writing and reviewed literature in PubMed. DMP is the corresponding author and principal provider who directed and supervised the literature review and who is responsible for all writing, editing and creation of the manuscript.

Competing interests None declared.

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