

A Rare Case of Schwann Cell Hamartoma in the Duodenum

Kasopefoluwa Y. Oguntuyo, PhD¹, Lauren L. Donnangelo, MD², Guangjing Zhu, MD, PhD³, Stephen Ward, MD, PhD³, and Abhik Bhattacharaya, MD²

¹Icahn School of Medicine at Mount Sinai, New York, NY

²Division of Gastroenterology, Icahn School of Medicine at Mount Sinai, New York, NY

³Department of Pathology, Icahn School of Medicine at Mount Sinai, New York, NY

Mucosal Schwann cell hamartomas (MSCHs) are benign neural lesions that are not associated with inherited syndromes and are primarily found in the distal colon. We report the first case of an MSCH in the duodenum. This case highlights the expansive nature of MSCHs and discusses the implications of this finding in the duodenum and in the context of a hematologic malignancy.

INTRODUCTION

In their 2009 landmark study, Gibson and Hornick described primarily distal colon lesions from 26 patients, many of whom were undergoing screening colonoscopies. These lesions were solitary with a sessile morphology and 1–6 mm in size with histologic features of spindle cells, wavy nuclei, eosinophilic cytoplasm, and no mitotic activity. Further characterization with immunohistochemistry was notable for S-100 protein positivity without reactivity to markers for other nonadenomatous polyps such as gastrointestinal stromal tumors (KIT), leiomyomas (smooth muscle actin), neurofibromas (CD34), and perineuromas (epithelial membrane antigen or claudin-1). Given this distinct morphological and histological phenotype, Gibson and Hornick proposed the name mucosal Schwann cell hamartomas (MSCHs). This nomenclature also served the additional purpose of distinguishing these benign lesions from malignant lesions and other gastrointestinal neural lesions associated with inherited syndromes such as NF1, MEN2B, or Cowden syndrome.¹ Since this original case

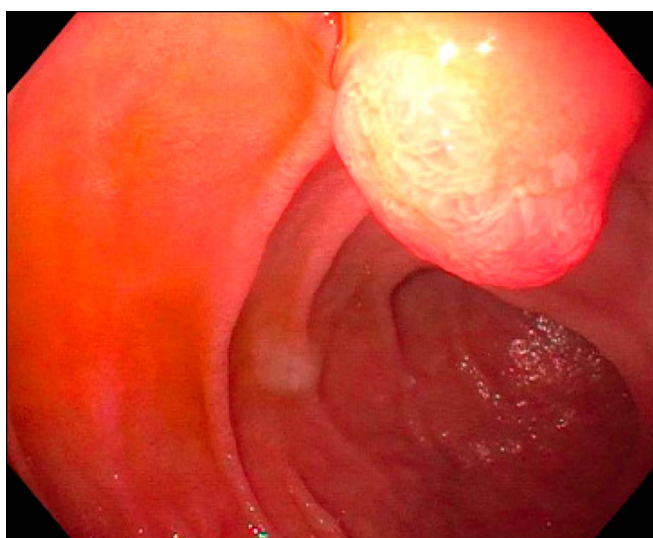


Figure 1. Endoscopic imaging of a 15 mm pedunculated polyp in the first portion of the duodenum.

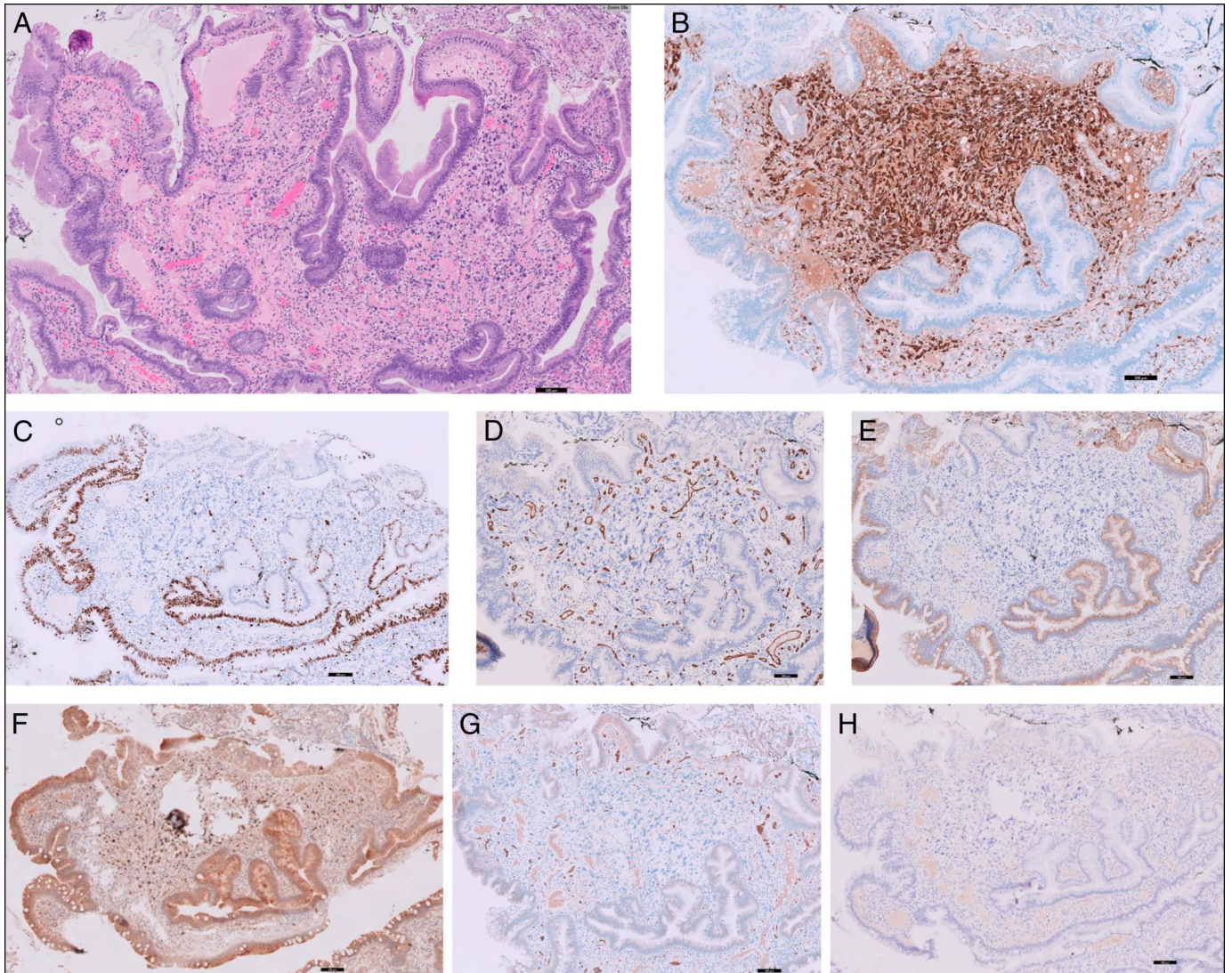


Figure 2. Histological findings from duodenal lesion is consistent with mucosal Schwann cell hamartoma. Hematoxylin and eosin stain with low power magnification (A). Immunohistochemistry for S-100 (B), MIB1/Ki67 (C), CD34 (D), epithelial membrane antigen (E), SOX10 (F), Glut1 (G), and HMB45 (H).

series, many groups have reported cases with colonic MSCH lesions and also in the gastroesophageal junction (GEJ), gastric antrum, gallbladder, and cecum.²⁻¹¹ We present the first MSCH lesion in the duodenum that was incidentally found in a patient with leukemia.

CASE REPORT

A 37-year-old woman with a new diagnosis of acute myeloid leukemia with central nervous system involvement and status post leukapheresis, 7 days of cytarabine with short infusions of idarubicin for the first 3 days, and intrathecal chemotherapy and the course complicated by stroke, seizures, and COVID-related respiratory failure status post tracheostomy on a ventilator presented for percutaneous endoscopic gastrostomy tube placement. Upper endoscopy was notable for a 15 mm pedunculated

polyp in the first portion of the duodenum (Figure 1). The polyp was removed completely by hot snare polypectomy.

Immunohistochemical staining of the lesion was positive for S-100 and SOX10 and negative for MIB1/Ki67, CD34, epithelial membrane antigen, Glut1, and HMB45 (Figure 2). This profile led to the pathologic diagnosis of duodenal mucosa with Schwann cell hamartoma with degenerative changes. Interestingly, this is the first MSCH case report to show positive staining of SOX10, consistent with its role as a transcription factor in Schwann cells.

Of note, the patient did not have physical examination findings of axillary freckling, inguinal freckling, or café au lait spots that would be suggestive of NF1. In addition, there was no family history of colorectal cancer.

DISCUSSION

Our incidental finding of an MSCH in the duodenum of a patient undergoing PEG placement for the management of their leukemia extends the gastrointestinal regions susceptible to developing MSCHs. Although the cause of MSCHs has not been determined, it is hypothesized that these lesions may be the result of a reactive process in areas prone to mucosal injury.⁶ This hypothesis is supported by the predominance of MSCHs at the rectosigmoid region and MSCHs at the GEJ⁶ and a retrospective study of 500 cholecystectomies that identified MSCHs in 4% of cases with histological evidence of regional hypertrophy and chronic inflammation.^{1–6,10} Notably, MSCHs found within the GEJ and the gallbladder often lacked the typical polyploid structure found elsewhere in the gastrointestinal tract like the colon or duodenum.^{6,10} This phenotypic difference may suggest either a distinct morphology or perhaps a spectrum of morphologies for MSCHs in different gastrointestinal contexts. Specifically for the duodenum, this polyploid MSCH may be the result of constant exposure to pancreatic digestive enzymes and bile acids.

Moreover, it is uncertain what role the patient's leukemia may have played in the development of the duodenal MSCH. Interestingly, there is one case report of a patient with lymphoma who was found to have a 3 mm MSCH in the transverse colon.⁵ Given the immune system's role in regulating uncontrolled proliferation of cells, it is feasible that the loss of this regulation in both patients allowed for the development of MSCHs. In addition, one case report of a patient with ulcerative colitis identified a 3 mm MSCH in the sigmoid colon, further suggesting that dysregulation of the immune system may play a role in the development of MSCHs.⁴

MSCHs are largely asymptomatic and benign, but there are cases of patients presenting with gastrointestinal/rectal bleeding, loose stools, abdominal pain with intermittent tenesmus, and epigastric pain.^{1,7,12,13} Most of these cases received polypectomies, but one patient had a subtotal gastrectomy with lymph node dissection due to features originally concerning for a gastrointestinal stromal tumor.⁷ As a result, there is a need to consider MSCHs as a diagnosis for polyploid and nonpolyploid lesions throughout the gastrointestinal tract. For the latter, Okamoto et al used narrow band imaging and endoscopic ultrasound to characterize MSCHs that presented atypically as skip-like lesions in the sigmoid colon. With narrow band imaging, granular white opacities were highlighted on the mucosal surface and endoscopic ultrasound highlighted that the lesion was present only in the superficial mucosa.¹⁴ Although there have been extensive case series and reports characterizing MSCHs, it is still unknown what the true prevalence of these lesions are, what risk factors contribute to the development of these lesions, what mutations drive the proliferation of these cells, and what are the long-term consequences of these lesions.

DISCLOSURES

Author contributions: KY Oguntuyo wrote the manuscript and is the author guarantor. LL Donnangelo edited the manuscript. G. Zhu provided pathology slides and interpretation of the findings. S. Ward supervised and reviewed the pathological findings. A. Bhattacharaya edited the manuscript and revised the manuscript for intellectual content.

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Informed consent could not be obtained from the family of the deceased. All identifying information has been removed from this case report to protect patient privacy.

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