A Rare Case of Appendiceal Schwann Cell Hamartoma

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Introduction

- Mucosal Schwann cell hamartoma (MSCH) is a rare, benign, neurogenic tumor characterized by a disorganized proliferation of S100-positive Schwann cells in the lamina propria, predominantly in the rectosigmoid colon.[1]
- Most often, it is an incidental finding in a routine colonoscopy. Here we present an infrequently encountered case of MSCH at the appendiceal orifice.

Case Description

- A 53-year-old male with a past medical history of hypertension, obstructive sleep apnea, obesity, depression, and osteoarthritis of the knee presented to the clinic for surveillance colonoscopy.
- On colonoscopy three years ago, the patient had two sessile polyps removed and interval colonoscopy at three years was recommended due to poor preparation.
- At this visit, he reported heartburn but denied hematochezia, melena, or bowel pattern changes.
- Given the upper GI symptoms, he underwent both esophagogastroduodenoscopy (EGD) and colonoscopy. EGD showed a 3cm hiatal hernia and erythematous mucosa in the stomach.
- Colonoscopy showed a solitary 4 mm polyp at the appendiceal orifice (Fig 1).
- Biopsy of the polyp showed spindle cell proliferation of Schwann cell phenotype located in the lamina propria without nuclear atypia, pleomorphism, or mitoses (Fig. 2).
- Immunohistochemical stains showed positive S100 and negative Desmin, SMM-HC, CD117, and CD34 (Fig. 3 & 4).

Discussion

- Benign nerve cell tumors are commonly described in the skin and soft tissue, and involvement of the gastrointestinal tract has been increasingly identified in the last decade.[2]

Images

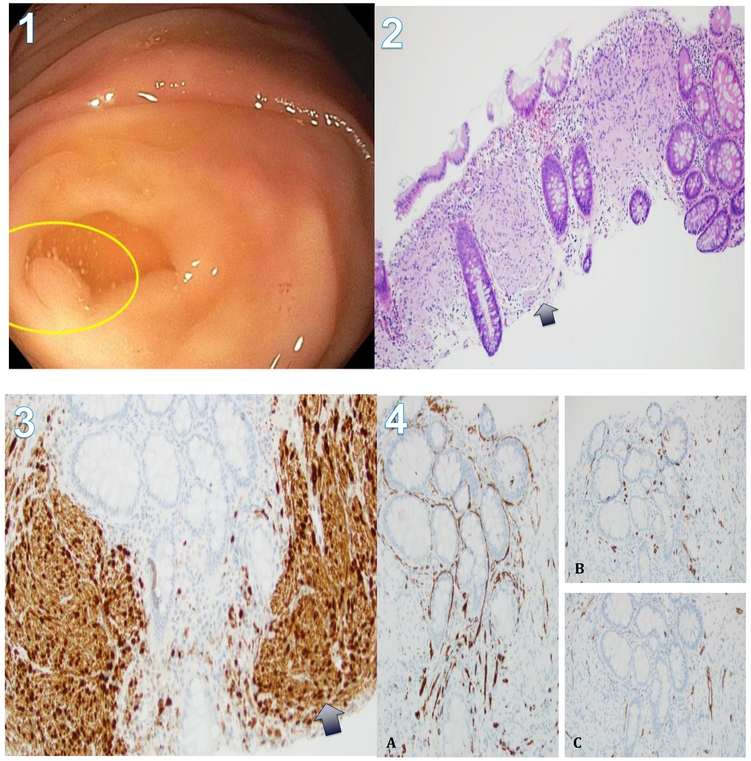

Fig 1: *Polyp at appendiceal orifice*
Fig 2: *Spindle cell proliferation separating the crypts*
Fig 3: *Spindle cell staining positive for S100.*
Fig 4: *Spindle cells are non-reactive to other markers employed*
A. Smooth muscle myosin
B. CD117
C. CD34

- It is a rare disease of the colonic mucosa, often diagnosed during screening colonoscopy.[3]
- They have been described as polyps usually < 1cm, predominantly located on the left side of the colon and seen in middle-aged female patients.[4]
- Very few cases of appendiceal MSCH and gallbladder MSCH have been reported.[4,5]
- Gibson and Hornick coined the term MSCH in 2009 to distinguish it from true “neuromas” and “neurofibromas”.[6]
- It is essential to diagnose and distinguish it from other neuronal polyps accurately - GIST, colorectal neurofibroma, mucosal neuromas, GI ganglioneuromas, mucosal perineurioma, inflammatory fibroid polyps, as some of these are associated with familial syndromes with worse outcomes and different management than MSCH.[3]
- More studies are required to evaluate recurrences and long-term prognosis for MSCH.

Conclusion

- No association between MSCH and inherited syndromes or malignancies has been established.
- However, it should be considered an important differential diagnosis of incidental GI polyps and encourage clinicians to test for specific markers to rule out other causes and prevent aggressive or unnecessary treatments,[6] thus reducing the burden on the health care resources.

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