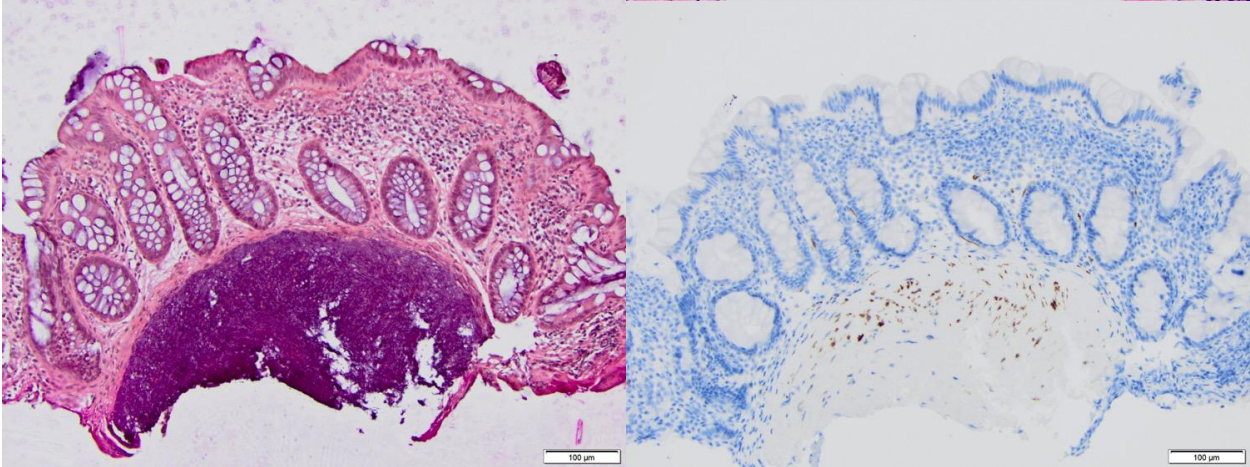
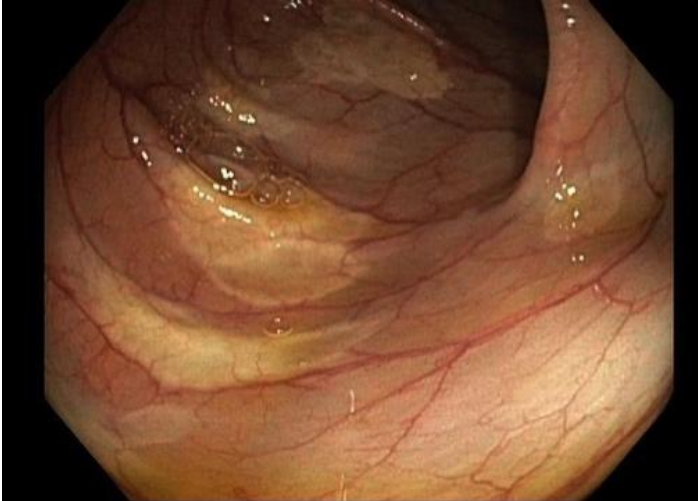
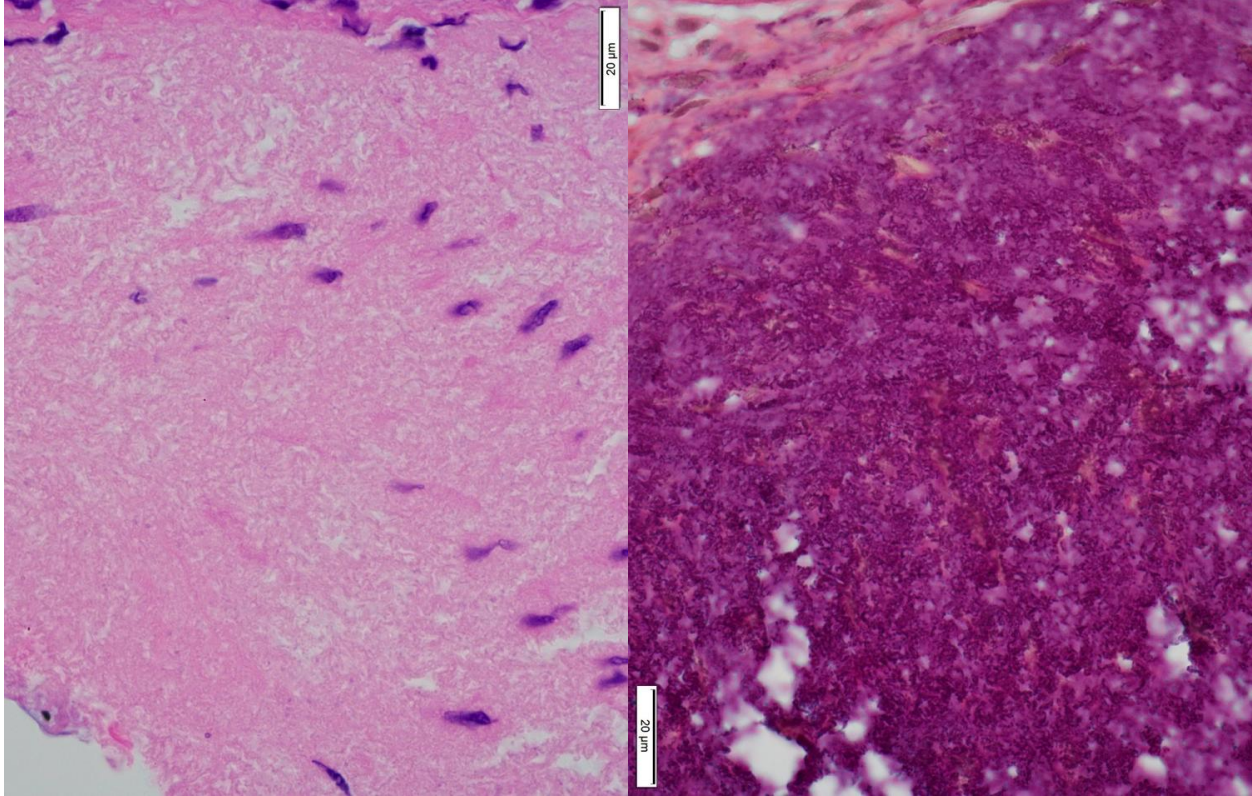


A 50 year old female with no significant past medical history presented for a routine screening colonoscopy. The patient reported only symptoms of reflux. She had no family history of colorectal cancer or genetic syndromes. On colonoscopy, multiple submucosal white plaques measuring 5-15mm were seen in the descending colon and biopsies were taken. Representative endoscopic images, H&E, Verhoeff-Van Gieson and a desmin immunostain are provided for your review.





What is the diagnosis?

- A. Amyloidosis
- B. Caseating/necrotizing granuloma
- C. Elastofibromatous change
- D. Granular cell tumor
- E. Leiomyoma

ANSWER AND DISCUSSION ON NEXT PAGE

Answer and discussion:

Elastofibromatous change (choice C) is the correct answer: The muscularis mucosae appeared thickened and expanded by a fibrillary and granular, pale eosinophilic material. Elastica van Gieson stains demonstrated accumulation of degenerated elastic fibers in a dense and haphazard distribution within the muscularis mucosae; immunostains for desmin and smooth muscle actin highlighted degenerated/splayed smooth muscle fibers surrounding the elastic fibers, confirming the diagnosis of fibroelastomatous change.

Elastofibromatous change (also described as elastosis, elastofibromatous change, elastofibromatous polyp, or perivascular fibrotic lesion) of the GI tract is a rare, benign lesion that is usually identified incidentally or during screening colonoscopy. It can occur anywhere along the GI tract from the esophagus to anus but is usually reported as a single polyp in the colon; in a few cases, it can be multifocal or have a non-descript appearance. Male and female patients are affected in near equal proportions, with a median age of 61 (range 47-83). Histologic changes can be seen in the lamina propria, muscularis mucosae and/or submucosa, with the latter being more common. In some cases, submucosal vessels can be seen associated with the lesion at its center or at the border. The cause of elastofibromatous change is uncertain but may be related to previous injury, as 9% are associated with prior intervention (such as polypectomy, GI surgery) and 7% have a history of radiation therapy. To our knowledge, there is no evidence that elastofibromatous change of the GI tract is caused by systemic connective tissue disorders; there is only one report of elastofibromatous change of the GI tract (stomach) in a patient who also had elastofibroma dorsi.

On endoscopy, amyloidosis (Choice A) can look normal or have a nonspecific appearance, including erythema, erosions, friability, or thickened, plaque-like mucosa, similar to the findings on colonoscopy in this patient. On microscopic exam, amyloid can be seen as extracellular deposits in any layer of the bowel, from the mucosa to muscularis propria and often involves vessel walls. Amyloid usually has a homogeneous, dense, "waxy," eosinophilic appearance that often shows slit-like spaces ("cracking" artifact from processing) and are positive for Congo red as well as other histochemical stains (thioflavine, crystal violet, toluidine blue). In contrast, elastofibromatous change shows an expansion of the muscularis mucosae or submucosa by fine, fibrillary eosinophilic material (that represent degenerated muscle fibers). A Congo red was negative in this case.

Caseating/necrotizing granulomas in the GI tract (Choice B) are associated with infections including *Mycobacterium tuberculosis*, *Mycobacterium avium-intracellulare* and *Yersinia* infection, and can involve any layer of the bowel wall. At low power magnification, the fibrillary material of elastofibromatous change may mimic central necrosis in a granuloma. However, caseating granulomas are surrounded by palisading histiocytes and frequently accompanied by a prominent lymphoid cuff, neither of which are seen in elastofibromatous change. Furthermore, inflammatory cell infiltration in elastofibromatous change tends to be scant in the muscularis mucosae and submucosa, as in our case.

Granular cell tumor (GCT) of the GI tract (Choice D) makes up 4-6% of all GCTs. The colon is the second most common site for GCTs in the GI tract and tumors appear as yellowish submucosal nodules. Most

tumors are well-circumscribed but 20% can have an infiltrative growth pattern on histology, with the latter pattern more common in GCTs of the colon. Lymphoid cuffs at the edge of GCTs are also common, particularly in colorectal GCTs. Accompanying peripheral inflammation is not a feature described in elastofibromatous change. While GCTs can involve the mucosa as well as the submucosa, can display more spindled morphology, and can have subtle, small nuclei, the eosinophilic material is granular and present within lysosomes. The degenerated fibers in elastofibromatous change can appear granular but tends to be more fibrillary and the lesion is paucicellular. GCTs are positive for S-100, CD68, CD56, SOX-10, NSE, synaptophysin by immunohistochemistry.

On colonoscopy, leiomyomas (Choice E) generally appear as small polypoid or larger nodular lesions, but not as an ill-defined plaques. Colonic leiomyomas generally arise from the muscularis mucosae and form sharply demarcated nodules. The tumor displays low to moderate cellularity and is composed of fascicles of bland smooth muscle cells arranged in a somewhat haphazard fashion. In contrast, elastofibromatous change can appear endoscopically as a polyp-forming lesion, area suspicious for inflammation or have no specific gross alterations. Elastofibromatous change can involve the muscularis mucosae but unlike in leiomyomas, desmin immunostains highlight smooth muscle bands around and between collections of degenerated elastic fibers but do not stain the elastofibromatous material.

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