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CASE REPORT | COLON

Appendiceal Elastofibroma Requiring Ileocecectomy

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ABSTRACT

Elastrofibromas are rare lesions characterized by collagen and elastic fiber deposition. They are generally found in soft tissues and throughout the gastrointestinal tract. The pathogenesis of this lesion is still uncertain, but it is hypothesized to be a reactive process. We present a case of an appendiceal elastofibroma, a unique anatomic location that necessitated surgical removal, with pathologic findings suggestive of an inciting event from a sclerosed vascular abnormality.

KEYWORDS: colon; appendix; elastofibroma; colonoscopy; ileocecectomy resection

INTRODUCTION

Within the gastrointestinal tract, the submucosa and muscularis mucosae have been found to occasionally exhibit focal or diffuse increase in elastin fibers.[1] Previous reports have variably referred to these as elastomas, elastofibromas, or elastofibromatous changes depending on the presence of accompanying fibrous tissue and whether the excess elastic fibers form a polypoid shape. Previous literature has demonstrated elastofibromas throughout the gastrointestinal tract including gastric, ileal, and colonic lesions.[1–9] To date, none have demonstrated such a finding at the appendiceal orifice nor a significant association with vascular abnormalities. We describe a rare example of an appendiceal gastrointestinal elastofibroma successfully removed by surgical appendectomy.

CASE REPORT

A 59-year-old Hispanic man was referred to gastroenterology for screening colonoscopy as part of renal transplant evaluation. He reported intermittent abdominal pain without changes in bowel movements or weight loss. Medical history included end-stage renal disease requiring hemodialysis, type 2 diabetes mellitus, and hypertension. Surgical history included arteriovenous fistula creation for hemodialysis. Colonoscopy was performed under monitored anesthesia care. Boston Bowel Preparation Scale score was 9. The examination was notable for a localized area of thickened mucosal folds at the appendiceal orifice (Figure 1). Biopsies were taken for pathologic evaluation. Additional findings included two 4-mm tubular adenomas and a few small-mouthed sigmoid diverticula. There were no postprocedural complications. A survey of family history did not include other elastofibroma or colonic masses. Abdominal and pelvic computed tomography scan with IV contrast was ordered and found mild thickening of the appendix and enlargement up to 1 cm proximally without periappendiceal stranding, luminal distention, or bowel obstruction (Figure 2). There was also notable atherosclerosis.

The case was discussed at pathology conference to review the biopsy findings. Initial hematoxylin and eosin sections demonstrated colonic mucosa with a large lesion characterized by elastosis/elastofibromatous features, felt to be compatible with elastofibroma (Figure 3). A large sclerotic vessel was also noted within the lesion, seen on hematoxylin and eosin staining (Figure 3). Further staining performed included Maison trichome and Elastic van Gieson (EVG), as well as immunostaining with smooth muscle myosin (SMMS-1) to evaluate for malignant muscle proliferation, discovered on GIST-1 (DOG-1) and tyrosine-protein kinase Kit (c-KIT) for gastrointestinal stromal tumor, and S-100 for nerve sheath involvement. Trichome staining demonstrated a significant amount of collagen deposition throughout the lesion and the embedded sclerotic vessel (Figure 3). EVG straining highlighted the elastic components of the sclerotic vessel (Figure 3). Immunostaining was negative for SMMS-1, DOG-1, c-KIT, and S-100.

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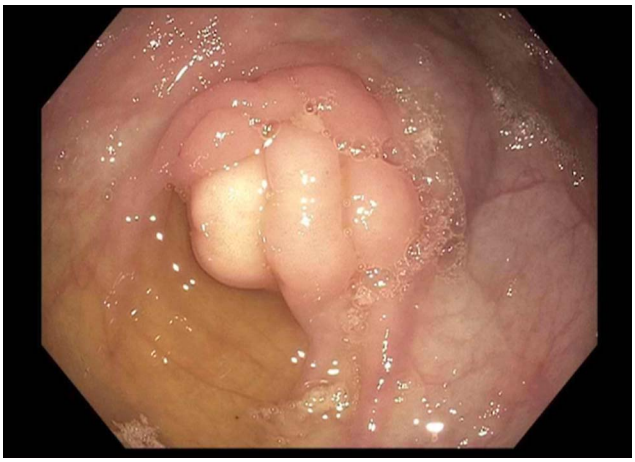

Figure 1. Endoscopic image of the thickened appendiceal orifice.

Based on these findings, imaging results, and concern for potential obstruction of the appendiceal orifice the patient was referred to a colorectal surgeon. They recommended he undergo robotic-assisted appendectomy. During the surgery, dense adhesions were found between the small bowel and the right lateral wall, and the omentum. The appendix appeared distended. The decision was made to proceed with appendectomy and partial cecectomy with a robotic stapler, with care given to preserve the ileocecal valve.

Gross evaluation of the surgical specimen demonstrated an appendix with a bland-appearing submucosal lesion (1 cm in greatest dimension), characterized by elastosis, vessels, and muscle components. The lesion was negative for dysplasia. Akin to the pathology from colonic biopsies, histopathologic and immunochemical staining demonstrated that the submucosal lesion was characterized by elastosis with significant collagen composition, a large sclerosed vessel, vascular proliferation, and components of muscle tissue. Trichrome straining again highlighted the collagen components of the sclerosed vessel (Figure 3), with elastin noted on EVG, and CD34 stains returning positive. To evaluate the etiology of this tumor, immunostains were again performed for SMMS-1, S-100, c-KIT, and DOG-1, all of which were negative. Outpatient surgical follow-up demonstrated no surgical complications, with well-healed incision sites and no further abdominal pain.

DISCUSSION

In this report, we describe the unique finding of a gastrointestinal elastofibroma at the appendiceal orifice, a site not previously demonstrated in previous literature. Previous published cases of elastofibromas were primarily either polypoid lesions or ulcerated gastric mucosa, unlike this lesion that appeared as mucosal thickening.[1–4,6–8] In addition, the location of this lesion introduced the anatomical concern of potential obstruction of the appendix and risk of appendicitis. Curiously, although the computed tomography scan demonstrated mucosal thickening without luminal distention or fat-stranding, at the time of surgery, there was distention and thick adhesions that necessitated modification to the planned resection, despite no previous abdominal surgery. Although elastofibromas are considered benign, the local effects demonstrate that it was appropriate to remove it before further complications.

Pathologic examination demonstrated characteristics of elastofibromatous changes previously seen in literature including $CD34^{+}$ cells, elastin + staining, and components of endothelium with collage deposition.[1,2,4–8,10] In terms of previous vascular findings, most previous histologic samples demonstrated normal vasculature, although one did show early elastotic changes and another degeneration of microvessels.[1,2,5,7,8] With the finding of peripheral inflammatory changes and the large sclerosed vessel coupled with smaller abnormal vessels and even calcium depositions, these findings raised the possibility of a collapsed arteriovenous malformation with sclerosis and elastofibromatous changes as sequelae.

This points to a reactive process, without a clear initial precipitant. However, because of the rare nature of these lesions, we ultimately still cannot completely describe their pathogenesis. We did demonstrate the clinical importance of ruling out malignancy and resecting this potentially obstructive inflammatory reactive process. Whether this began as a sclerosed arteriovenous malformation that led to local alterations and

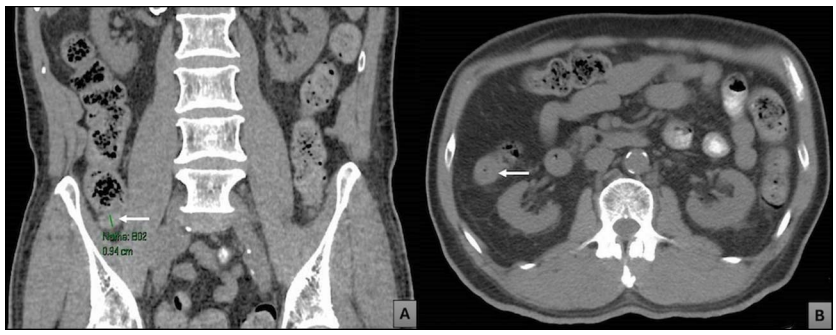

Figure 2. Abdominal computed tomography imaging to evaluate the appendiceal thickening in (A) coronal and (B) axial views. Arrows indicate areas of appendiceal thickening.

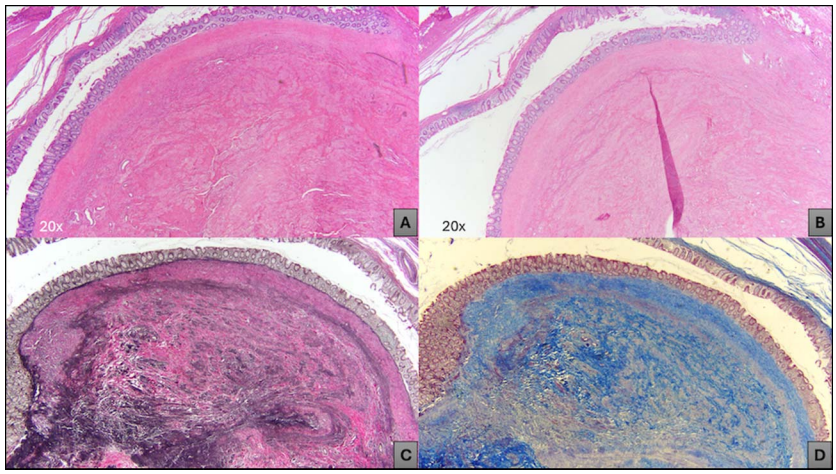

Figure 3. Hematoxylin and eosin sections showing separate sites of the lesion at ×20 with the (A) elastofibroma primary portion and (B) sclerosed vessel portion featured. Elastin staining (C) with Elastic van Gieson staining highlighting elastic fibers in the fibroma and from the sclerotic vessel. A trichrome (D) section displaying significant fibrotic tissue.

elastofibromatous changes or was an independent process is uncertain, but it suggests a possible pathway for the origin of this rare lesion.

DISCLOSURES

Author contributions: JM Luevano: aided in conception and creation of manuscript, drafting and revisions of the manuscript, final approval, and agreement of accountability for all aspects of the work. C. Bowman and R. Garcia-Carrasquillo: aided in conception of the manuscript, drafting of the manuscript, review of revisions, final approval, and agreement of accountability for all aspects of the work. R. Garcia-Carrasquillo is the guarantor of the article.

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