Beyond Acute Appendicitis: Mucinous Neoplasm Presenting as Appendiceal Mucocele: A Case Study

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Abstract

Mucocele of the appendix is an uncommon disorder characterized by dilation of the appendiceal lumen due to accumulation of mucinous substance. It has no typical clinical presentation and is considered a potentially premalignant condition, making it challenging yet important to be diagnosed and treated early. If untreated, one type of mucocele may rupture resulting in a potentially fatal condition called pseudomyxoma peritonei. In this case study, a 55-year-old-femal with a large appendiceal mucocele is suspected of malignancy. A right hemicolectomy was performed, and grossing revealed a large cystic lesion protruding into the mesoappendix. Histological evidence provided the diagnosis of low-grade appendiceal mucinous neoplasm (LAMN) with focal extension beyond muscularis propria, ruling out the risk of pseudomyxoma peritonei. Due to the tumor's location in the ileocecal region, the pathologists' assistant played a critical role in determining its relationship to all adjacent structures by comprehensive sampling, together with a thorough and meticulous gross examination and description.

Introduction

While acute appendicitis is a common pathology of the appendix, numerous other disease processes including a variety of neoplasms can mimic the clinical and imaging findings of acute appendicitis. One such presentation is appendiceal mucocele (AM), which is characterized by accumulation of intraluminal mucin. Clinically, it can be confused with acute appendicitis. AM is not a histological diagnosis, but a gross description with underlying causes, both benign and malignant. Because of lack of specific signs or due to its sometime quiet presentation, this condition is frequently found incidentally, or diagnosed at an advanced stage. Although rare with incidence of 0.2%-0.7% of all appendectomied specimens [1], AM is considered potentially premalignant, making it challenging, yet important to be diagnosed early. If misdiagnosed, one type of mucocele may rupture resulting in a spillage of malignant cells into the abdominal cavity, a potentially fatal condition called pseudomyxoma peritonei. Mucocele of the appendix is a heterogenous group and presents with a wide spectrum of histopathological alterations. Literature review suggests 4 causal pathologic conditions depending on the different histological subtypes and the extent of dissemination determine treatment options and prognostic information, with benign cases having 91%-100% 5-year survival rate, and 25% for malignant types [2]. This stud explores the case of a 55-year-old female presenting with an appendiceal mucocele, suspected of malignancy.

Pertinent Patient History

A 55-year-old lady was initially presented to the emergency department after being seen by her family doctor for right lower quadrant pain. Her abdomen is soft and distended, and displayed tenderness in the right lower quadrant to palpation. No peritonitis was found. Her family doctor suspected appendicitis and directed her to either go to the emergency department or have an outpatient ultrasound. At the emergency department she underwent a CT scan, which findings (Fig. 1) were suggestive of appendiceal mucocele. Her colonoscopy revealed an abnormal bulky and erythematous appendiceal orifice, although with maintained central opening, and not centrally protuberant. There was no evidence of abnormal surface mucosa. It also showed mild sigmoid diverticulosis. Biopsies were submitted and results confirmed the CT findings. Given the size of the mucocele (4 cm), it was concerning that it could be malignancy. Imaging showed no evidence of obvious nodal or distant spread. Preoperative diagnosis was appendiceal mucocele based on imaging. Acute surgical team performed the open right hemicolectomy procedure on the patient.

Pathologic Findings - Gross

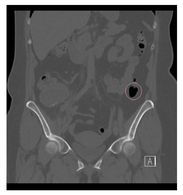

Fig. 1. Patient's pelvic CT scan demonstrating a mass in the right lower quadrant

The specimen contained 8.0 cm long terminal ileum, 4.0 cm long, 1.3 cm diameter appendix, a portion of 13.0 cm ascending colon, and attached 15.0 x 4.5 x 3.5 cm pre-colonic adipose tissue. The approximal margin was stapled and measured 2.0 cm in diameter. The mesenteric radial margin around the ileum was inked green. Within the appendix there was a cystic nodule that protruded into the meso-appendix in a diverticular fashion. The nodule measured 5.5 x 4.5 x 0.7 cm, and was located 0.6 cm from the radial margin, 11 cm from the distal margin, and 3.2 cm from the proximal margin. The nodule was found to be dilated and filled with tan-yellow gelatinous mucoid material. It was partially covered by retracted serosa and predominately covered by roughened, soft tissue that was combined with the mesentery, surrounding the cecum. The whole area was inked black (Fig. 2). The remaining ileum had pink-tan and shining mucosa, with no other lesion noted. The entire cystic nodule was serially sectioned along the short axis with a section of the adjacent ileum, appendix, and cecum, respectively, into 14 slices (Fig. 3). The extent of invasion was difficult to identify grossly, and sections were submitted for microscopic assessment. Examination of the peri-colonic adipose tissue reveled multiple lymph nodes, ranging 0.2 cm to 1.0 cm in greatest dimension. Due to the nodule's location in the ileocecal region, it was important to sample heavily, and submit sections to demonstrate the relationships of the appendiceal lesion to all adjacent structures, to margins, and to evaluate the deepest invasion (Fig. 4). A total of 44 cassettes were submitted. Sections were taken from each slice including: lesion with soft tissue margin, lesion with appendix and cecum, with retracted serosa, with appendix, and with appendix and ileum. Tan-white solid areas surrounding the lesion were also collected. Additionally, the proximal and distal margins were submitted en face, respectively. Finally, lymph nodes search was performed by thoroughly palpating the peri-colonic adipose tissues and identified 27 lymph nodes, which were bisected or serially sectioned and submitted entirely. All slices are photographed, and the sections taken were mapped on the digital image.

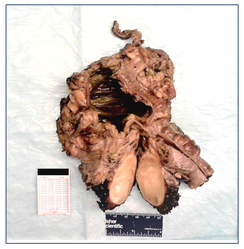

Figure 2. Right hemicolectomy specimen with appendiceal mucocele in a divertucular fashion.

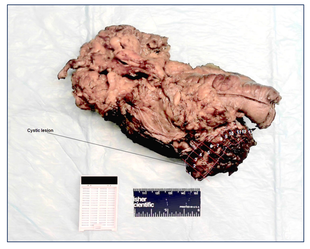

Figure 3. Cystic lesion (mucocele) is sectioned into 14 slices as labelled.

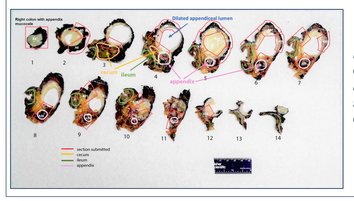

Figure 4. Mapping on the slices demonstrating submitted sections that include tumor with surrounding structures to demonstrate relationships and involvement.

Pathologic Findings - Microscopic

Microscopic sections of the thinned appendiceal wall revealed an attenuated flat-to-filiform mucinous epithelium with broad/pushing expansion associated with obliteration of the underlying muscularis mucosa and loss of submucosal lymphoid tissue (Fig. 5) . It showed that the tumor invaded through muscularis propria into subserosa or meso-appendix but did not extend to serosal surface. The diagnosis came to low-grade appendiceal mucinous neoplasm (LAMN) (Fig. 6). Patches of the neoplastic epithelium rest directly on fibrous tissue. Mucinous epithelium and pools of acellular mucin very focally extend beyond the muscularis proria (pT3) (Fig. 7). Twenty-seven regional lymph nodes were examined, and all showed negative for malignancy, indicating pN0. All Resection margins were negative of non-invasive tumor. This case was reviewed at intradepartmental rounds, and a gastrointestinal pathologist was consulted who confirmed the diagnosis. It was further noted that the risk of pseudomyxoma peritonei was minimal to zero, as all of the acellular mucin and neoplastic epithelium were confined within the diverticular outpouching of the appendix and surrounded by a thick fibrous capsule and there was no evidence of perforation of acellular mucin on the appendiceal surface.

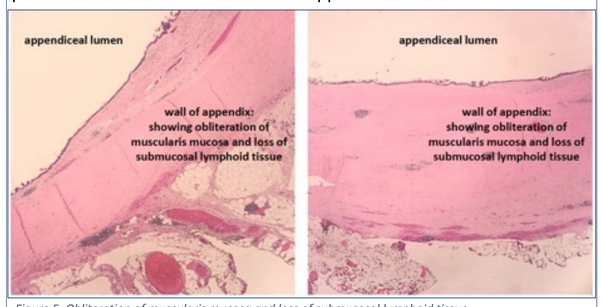

Figure 5. Obliteration of muscularis mucosa and loss of submucosal lymphoid tissue.

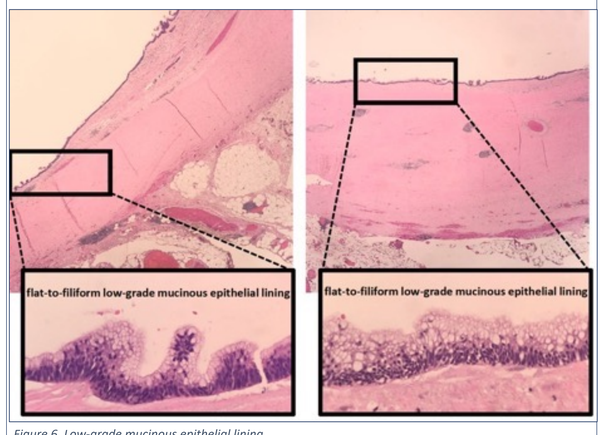

Figure 6. Low-grade mucinous epithelial lining

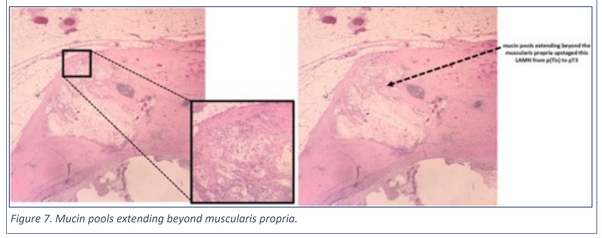

Figure 7. Mucin pools extending beyond muscularis propria.

Discussion

As previously discussed, mucocele of the appendix is a gross description and the underlying pathology needs to be determined by histological examination. In this case study, the mucocele was caused by low-grade appendiceal mucinous neoplasm (LAMN). Appendiceal mucinous neoplasms is a complex, diverse group that often presents as cystic dilation of the appendix due to accumulation of gelatinous material, morphologically referred to as mucocele [3]. The WHO 5th Digestive System Digestive System Tumors defined the AMN as a mucinous epithelial proliferation with extracellular mucin and pushing invasion pattern or pushing tumor margins [4]. Most AMNs are developed in middle-aged or elderly patients and present with non-specific symptoms or signs [5]. The AJCC 8th Cancer Staging Manual defined a LAMN as a mucinous neoplasm with low-grade cytology associated with the obliteration of the muscularis mucosae without overt features of the invasion. The lamina propria is frequently effaced, and mucosal lymphoid tissues are decreased or absent. The essential histological finding of LAMN is low-grade cytology with pushing invasion, but with no destructive invasion in the appendiceal wall [5]. Typical LAMNs usually have thin fibrotic walls and abundant intraluminal mucin, and less commonly, calcification of the wall. Thus it was imperative to assess the depth of invasion for histological examination by meticulous sampling of tumor with respect to surrounding tissue. A thorough lymph node search also contributed in determining the pN category that concluded negative for malignancy. Since it was determined that the neoplastic epithelium was confined within the diverticular appendix, and that no other gastrointestinal structures or locations were involved, the final diagnostic was able to confidently rule out pseudomyxoma peritonei (PMP). PMP is the most significant complication of appendiceal tumors, which is characterized by the grossly persistent accumulation of mucinous ascites in the peritoneal cavity. The expansion of mucin within the abdominal cavity results from mucus following the normal flow of peritoneal fluid, redistribution of the mucin, and neoplastic cells. Based on clinical and immunohistochemical data, most cases are due to the perforation of AMNs [5]. Patients with appendiceal neoplasms should be under high surveillance for PMPs to restrict the spread of cancerous cells in the peritoneal cavity and ultimately remote metastases. Due to the close anatomic relationship of the appendix to a few adjacent structures, it is worth noting that the comprehensive sampling of the mass to all adjacent structures performed by the pathologists' assistant played a critical role in deriving to the final diagnosis in a confident and efficient manner. Finally, the pathologist assistant's digital photographs and detailed mapping further assisted in the visual representation and clear communication of this complex specimen during the specialist consultation.

References

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