Case Report: Metastatic Mucinous Carcinoma of the Appendix in a 33-year-old Female

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Abstract

Background. Primary cancers of the appendix are rare, with an incidence of approximately 1.2 cases per 100,000 people per year and this tumor is difficult to diagnose preoperatively. The purpose of this paper is to present a rare case of metastatic mucinous carcinoma of the appendix and to provide a high index of suspicion to patients presenting with the same history, signs, and symptoms.

Case Summary. We present a case of a 33-year-old Filipina who reported abdominal pain and right lower quadrant mass. Following several preoperative diagnostic tests, a colonoscopy revealed synchronous tumors in various locations, prompting the need for an exploratory laparotomy to evaluate the abdomen. Histopathological examination was performed to confirm the final diagnosis which revealed primary mucinous carcinoma of the appendix. The tumor had extended into adjacent structures, including the cecal colon, ileum, and right ureter. Metastatic lesions were also identified in the descending and sigmoid colon. The disease was classified as stage IVC (T4b, N1c, M1c), indicating advanced progression with both extensive local invasion and distant metastasis.

Conclusion. Histopathology remains the gold standard for cancer diagnosis. Given the rarity and complexity of appendiceal mucinous carcinoma, a multidisciplinary approach is also essential. This collaborative strategy from various specialties is vital not only for achieving an accurate diagnosis but also for developing and implementing an effective, individualized treatment plan that addresses the distinct challenges of this uncommon malignancy.

Key words. *Appendiceal tumors, Metastatic mucinous carcinoma of the appendix, Rare gastrointestinal tumors*

Introduction

Primary cancers of the appendix are rare, with an incidence of approximately 1.2 cases per 100,000 people annually. Appendiceal adenocarcinomas typically affect patients in their 5th to 7th decade of life.[11] According to the SEER program database and National Cancer Database, there is a slight female predominance, approximately 53-57%. Patients are often asymptomatic or have nonspecific symptoms wherein the most frequent symptom is acute or chronic right lower quadrant abdominal pain. These tumors are challenging to diagnose preoperatively due to its nonspecific symptoms.[13]

This report aims to raise awareness for patients presenting with similar history, signs, and symptoms as in this case. By doing so, the need for extensive preoperative diagnostic tests can be reduced. Additionally, it highlights the critical role of histopathology in confirming a cancer diagnosis.

Case Report

A 33-year-old Filipino female from Nueva Vizcaya, with a history of polycystic ovary syndrome (PCOS) and a family history of renal cell carcinoma, presented with a month-long history of intermittent generalized abdominal pain (5/10) and frequent soft to watery stools without blood, fever, or weight loss. Abdominal ultrasound showed a 5.91cm x 5.52cm x 4.33cm suspicious mass, moderate hydronephrosis of the right kidney, and a dilated pelvicalyceal system. CT urogram revealed a right lower quadrant complex cystic mass (6.5cm x 4.4cm x 5.3cm) compressing the right ureter, leading to severe hydronephrosis hence, double J stent was placed.

A video colonoscopy *(Figure 1)* was performed to investigate a right lower quadrant mass but was unsuccessful due to a sigmoid obstruction. Because of this, CT virtual colonoscopy *(Figure 2)* was done and identified multiple masses: a cystic ileocecal mass involving the right ureter, adnexal masses near the

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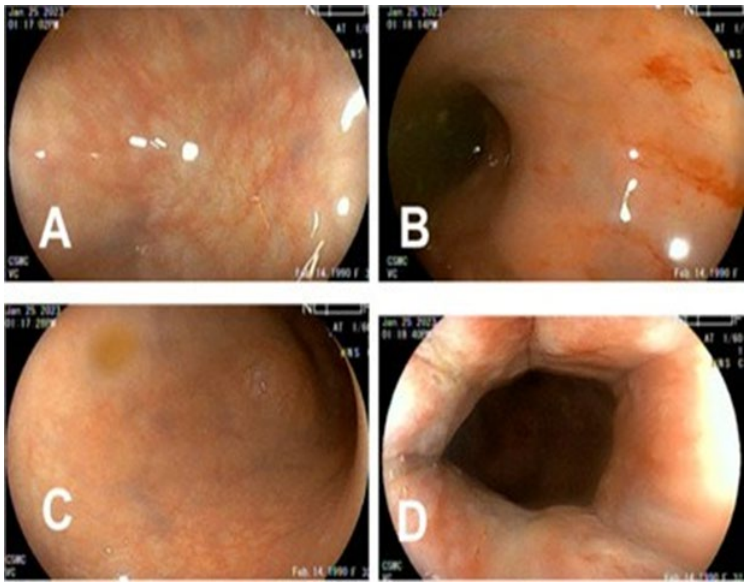

Figure 1. Video Colonoscopy. B. Acute bend noted 30 cms level from the anal verge. Bypassing the area was unsuccessful.

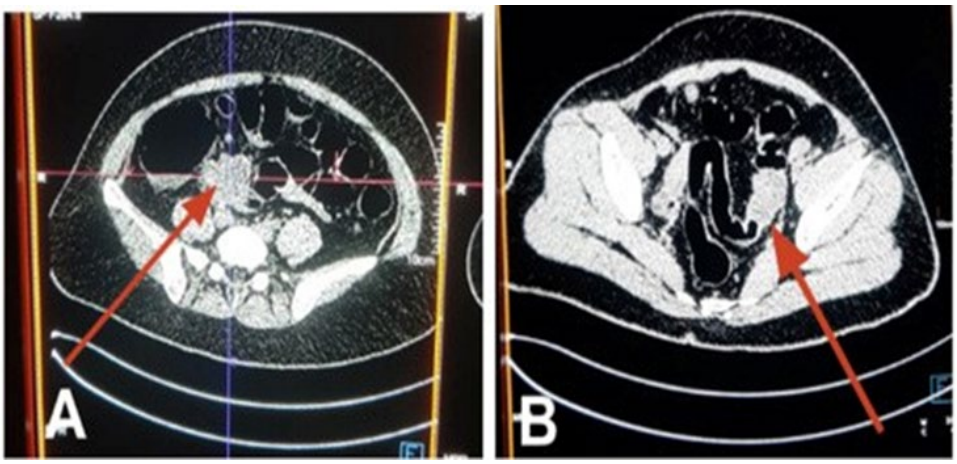

Figure 2. Abdominal CT scan (virtual colonoscopy). [A] Hypodense structure with cystic component and associated rim calcifications involving the ileocecal region measuring 6.6cm x 4.7cm x 4.3cm, remains intimately related to the right ureter. [B] Irregular circumferential wall thickening in the sigmoid colon with heterogeneously hypodense attenuation and associated with almost complete luminal obstruction

fallopian tubes, and a nearly obstructive sigmoid mass extending toward the uterus.

The patient underwent exploratory laparotomy *(Figures 3 and 4)*, right hemicolectomy with anastomosis, right segmental ureterectomy, infracolic omentectomy, and sigmoidectomy with colostomy. Intraoperative findings included an ileocecal mass (5cm x 8 cm) adherent to the ureter, a sigmoid mass (2cm x 3 cm), omental and lymph node nodules, and a hepatic nodule (1cm x 2 cm). Official histopathology *(Figures 5-8)* was released showing mucinous carcinoma with focal signet ring cell features of the cecal area with ileal extension, descending colon and sigmoid, and mucinous carcinoma of right ureter, which are all suggestive of an appendiceal primary, with full thickness infiltration and widespread metastasis. Cytoreductive surgery (CRS) with hyperthermic intraperitoneal chemoperfusion (HIPEC) was planned.

Discussion

One of the main diagnostic challenges of this case is the difficulty to diagnose primary appendiceal tumors using laboratory testing and imagining, hence histopathological examination is needed. Intraoperatively, appendix may appear grossly unremarkable or demonstrate dilatation as a result of abnormal accumulation of mucin. Gross perforation with mucin extrusion may be evident. The wall may be attenuated, or markedly thickened and partially

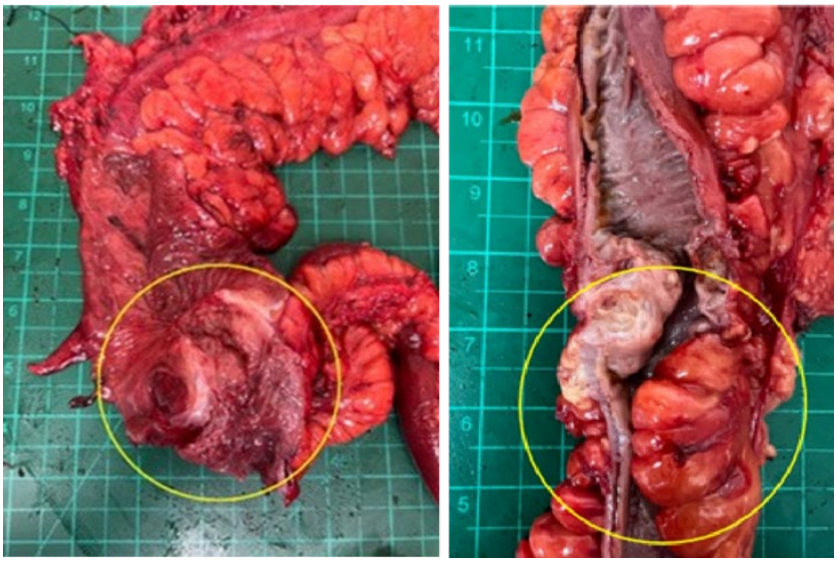

Figure 3. [A]. Ileocecal mass and the appendix are not visualized. [B]. Sigmoid mass with palpable mesenteric nodes

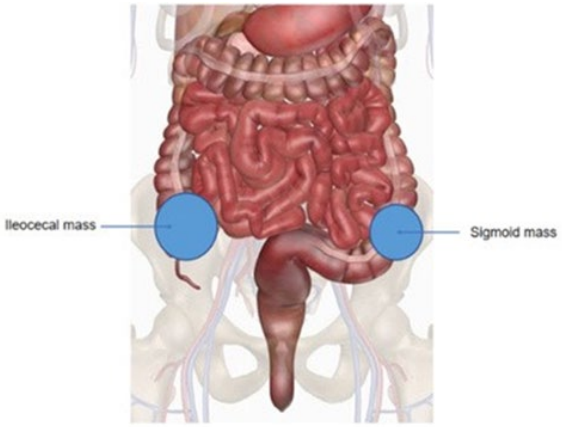

Figure 4. The locations of the masses: Ileocecal and Sigmoid Areas

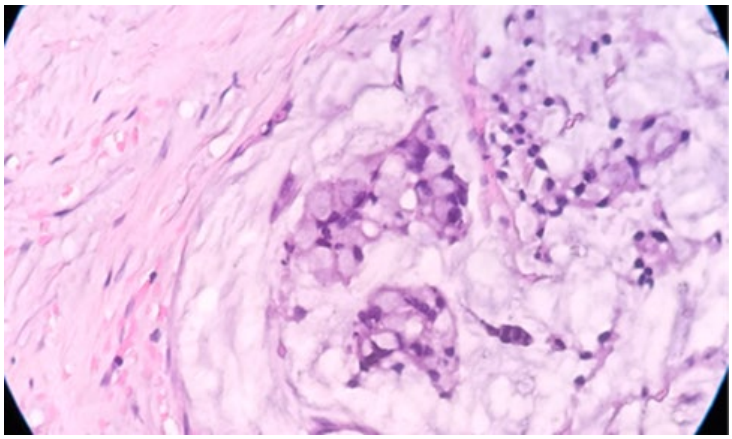

Figure 5. Appendiceal Area. The tumor cells have glandular formation in pools of mucin. Tumor cells resembling signet ring cells are also noted.

calcified. As seen in this rare case, the appendix was not visualized and solid since it might be ruptured. A prospective study entitled histopathological spectrum of appendectomy specimens showed that histopathological examination is still the gold standard of choice for confirmation of any appendectomy specimens since there will be instances of incidental diagnosis of carcinoma.[19]

Primary cancer of the appendix is a rare malignancy of the gastrointestinal tract and most cases were diagnosed after histopathological examination. It is uncommon to the fact that its prevalence in histopathological examinations was only 0.2 to 0.3% of appendectomy specimens.[8]

Furthermore, intraoperative histopathology is needed because an adenoma–carcinoma sequence exists in the appendix that is similar to the one described in the colorectum. These are similar in that they all define benign neoplastic adenoma as confined to the mucosa, without mucin or cells penetrating the muscularis mucosa or evidence of perforation. Invasive adenocarcinoma is a frankly invasive neoplastic lesion with cellular invasion beyond the muscularis mucosa.[8,20]

The American Joint Committee on Cancer 8th Cancer Staging Manual also classified the appendiceal mucinous tumors as a three-tier grading system: Grade 1 (well-differentiated) are low grade appendiceal mucinous neoplasm which is low-grade cytology, usually lacking infiltrative invasion, high grade appendiceal mucinous neoplasm and the conventional mucinous adenocarcinomas qualify as Grade 2 (moderately differentiated) and tumors with unequivocal signet-ring cells and signet ring cell carcinoma are graded as Grade 3 (poorly differentiated). Mucinous adenocarcinoma with signet ring cells comprises less than 50% of

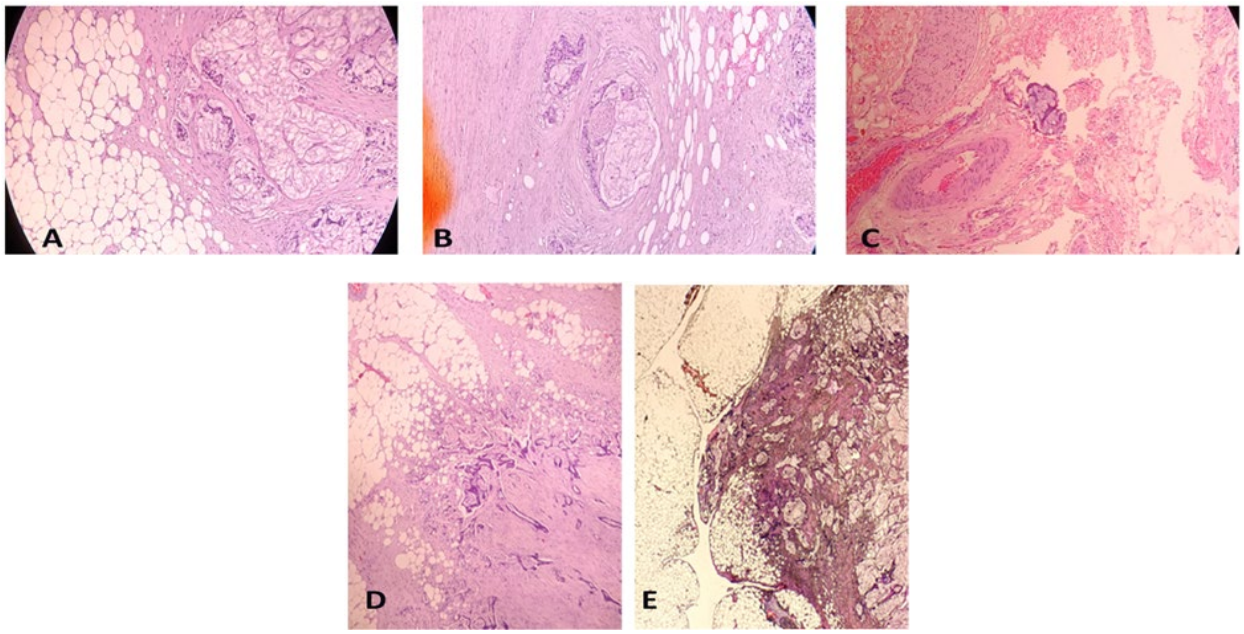

Figure 6. Ileum to Proximal Transverse Colon. [A]. Malignant neoplasm with transmural invasion into the pericolic fatty tissue up to the serosal surface. [B] Perineural invasion. No definite lymphovascular invasion. [C] Acellular mucin seen in the mesenteric margin. The posterior and distal margins are negative for tumor. [D] All 16 lymph nodes were negative for metastasis. However, two tumor deposits are seen. [E]. The omentum is also involved by tumor

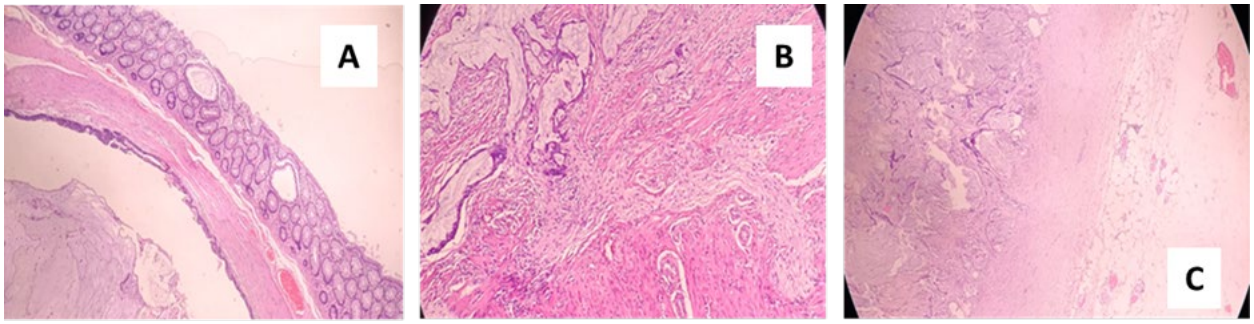

Figure 7. Descending Colon to Sigmoid. [A] Mass involves the muscularis propria and serosa, without involvement of the mucosa. [B] Perineural invasion is present. No definite lymphovascular invasion is seen. [C] All four regional lymph nodes were negative for metastasis. However, 3 tumor deposits were seen.

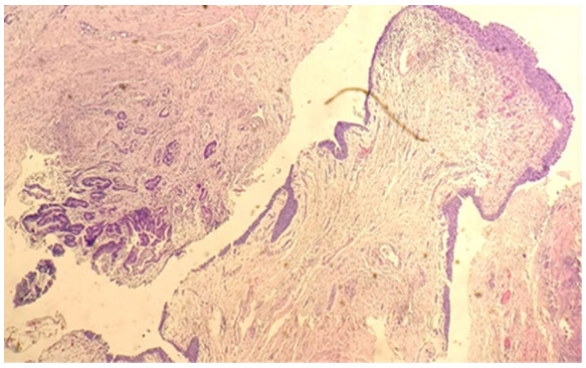

Figure 8. Ureter. Positive for carcinoma

signet ring cells while signet ring carcinoma has more than 50% signet ring cells. As seen in the histopathological slides of the patient, the tumor cells have glandular formation of pools of mucin and the

signet ring cells were evident in the appendiceal area hence, it is high grade.[2]

Epithelial appendiceal neoplasms demonstrate a propensity for peritoneal metastasis and metastasize outside the peritoneal cavity less frequently, hence, they are amenable to regional therapies. The most commonly used technique for therapy is cytoreductive surgery (CRS) with hyperthermic intraperitoneal chemoperfusion (HIPEC). Cytoreductive surgery refers to surgical removal of all visible (macroscopic) intra-abdominal tumors, whereas HIPEC aims to eradicate microscopic tumor deposits as a locoregional treatment. It is achieved by reaching intra-peritoneal concentration of cytotoxic drugs approximately 20 times more than systemic concentrations (intravenous). The most commonly used drugs for HIPEC are FOLFOX: cisplatin, mitomycin-C (MMC), oxaliplatin (Oxali), and 5-fluorouracil (5-FU). For patients with unresectable disease, systemic chemotherapy can provide disease control in more than 50% of cases.[16]

Histology has an important role in determining the clinical course of the disease. The prognosis still varies according to histologic stage and grade. Patients with mucinous adenocarcinomas have a better outcome than those with non-mucinous adenocarcinomas but grades 2 and 3 have significantly worse prognosis than Grade 1.[6,7]

Conclusion

Primary appendiceal carcinoma is a rare and often overlooked diagnosis, particularly in young patients with vague abdominal symptoms. This case highlights the importance of maintaining a high index of suspicion, as early diagnosis is frequently delayed due to its nonspecific presentation. Histopathological examination confirmed mucinous carcinoma of the appendix with extensive local invasion and distant metastases, classifying it as stage IVC (T4b, N1c, M1c). As the gold standard for diagnosis, histopathology is essential for guiding appropriate treatment. Given the rarity and complexity of appendiceal mucinous carcinoma, a multidisciplinary approach is essential. Collaborative efforts across various medical specialties are crucial—not only for accurate and timely diagnosis but also for creating and executing an individualized, effective treatment plan. This case report emphasizes the need for greater clinical awareness and diagnostic vigilance in managing patients with similar presentations.

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