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CASE REPORT | COLON

# Anal Mucinous Adenocarcinoma Presenting as a Superficial Lesion Without Luminal Involvement

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## ABSTRACT

Anal mucinous adenocarcinoma arises from mucin-secreting columnar epithelium within anal glands and is extremely rare, comprising 2%–3% of all gastrointestinal malignancies. We present a unique case of 65-year-old developmentally disabled man with complaint of rectal pain. Examination showed an excoriated erythematous perianal region with mucinous film and subdermal nodularity. Surgical pathology of the lesion revealed poorly differentiated mucinous adenocarcinoma of intestinal type. Subsequent colonoscopy was without findings of intraluminal lesions. He established with oncology and later underwent a positron emission tomography scan that showed extensive metastasis. This case highlights a unique presentation of *de novo* mucinous adenocarcinoma with luminal sparing.

**KEYWORDS:** mucinous adenocarcinoma; anorectal cancer; gastrointestinal malignancy; perianal pain

## INTRODUCTION

Adenocarcinoma of the anal canal accounts for 3%–19% of all anorectal cancers and confers a poor prognosis compared with epidermoid cancers.[1,2] The type of anal neoplasia that develops is typically dependent on the site of origin. Mucinous adenocarcinoma arises within the anal mucosa from a proliferation of mucin-secreting columnar epithelium deep within the anal glands. Consequently, this adenocarcinoma develops a mucinous characteristic and is extremely rare, comprising 2%–3% of gastrointestinal malignancies.[3] The anal mucosa lies approximately 1 cm distal to the dentate line and proceeds proximally into the anal canal. Therefore, mucinous adenocarcinoma usually exists within the anal canal. Mucinous adenocarcinoma is often diagnosed at a late stage, but metastasis is rare.[4] Symptoms include bleeding on defecation, perianal pain, or pruritus.[5] We present a unique case of a 65-year-old man with mucinous adenocarcinoma presenting as a superficial lesion of the gluteal crease without luminal involvement.

## CASE REPORT

A 65-year-old developmentally disabled man with a history of colonic polyps presented to the hospital with rectal pain. At the age of 51 years, the patient underwent a screening colonoscopy and was found to have a lesion in the ascending colon. Pathology showed adenocarcinoma. His healthcare proxy elected for a laparoscopic right hemicolectomy with removal of the 3-cm adenocarcinoma. Six years later, he was found to have a 1-cm semi-pedunculated tubulovillous adenoma in the descending colon, which was resected during colonoscopy. Interval endoscopic evaluations, with the most recent colonoscopy 3 years before presentation, were documented as unremarkable. Physical inspection of the rectum was deferred during annual physical examination which occurred 2 years before presentation.

Approximately 6 months before presentation, the patient went to the hospital with complaint of rectal pain. He was evaluated by general surgery, who noted an irregular anal lesion with a high suspicion for malignancy based off appearance. He was recommended to follow-up with a colorectal surgeon for further workup. However, follow-up was never completed.

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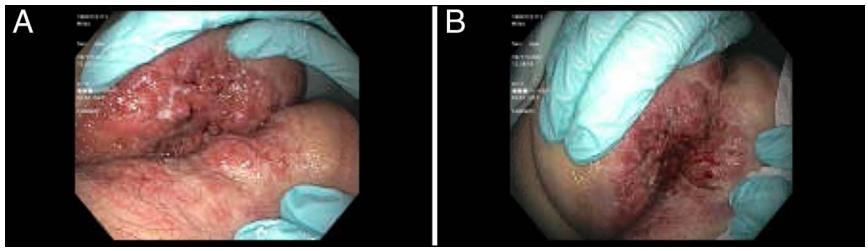


**Figure 1.** (A) Superficial examination showing excoriated erythematous perianal region with glossy mucinous film. (B) extension of subdermal nodularity 6–8 cm circumferentially from the anus.

Six months later, he presented to the hospital again for rectal pain. Vital signs were notable for elevated blood pressure and heart rate in the setting of pain. Laboratory work revealed mild normocytic anemia. Examination showed a severely excoriated, erythematous perianal region with a glossy mucinous film, and subdermal nodularity extending 6–8 cm circumferentially from the anus. Computed tomography scan of the abdomen and pelvis demonstrated a mass-like lesion of the rectum with extension into the perirectal soft tissues and left inner buttock. In addition, local lymphadenopathy and lesions of the liver thought to be hemangiomas were present. The primary site of malignancy was determined to be the anal canal.

During his hospitalization, flexible sigmoidoscopy showed a normal rectum, sigmoid, and recto-sigmoid colon. He also underwent colonoscopy, which did not demonstrate any intraluminal lesions. Surgical pathology of the superficial rectal lesion revealed poorly differentiated mucinous adenocarcinoma, intestinal type. Immunohistochemistry showed positivity for cytokeratin 20 (CK20), caudal type homeobox 2 (CDX2), and special AT-rich sequence-binding protein 2 (SATB2), with focal positivity for cytokeratin 7 (CK7).

The patient established care with oncology outpatient, who discussed potential treatment options including chemotherapy with a guarded prognosis. He later underwent a positron emission tomography scan, which showed high uptake in the rectum, perirectal lymph nodes, and multiple areas of lymphadenopathy throughout the abdomen, pelvis, and mediastinum, indicating widespread cancer. Given the patient's comorbidities, the patient's healthcare proxy elected not to proceed with further life-prolonging care, and the patient was transitioned to a palliative care approach only (Figures 1 and 2).

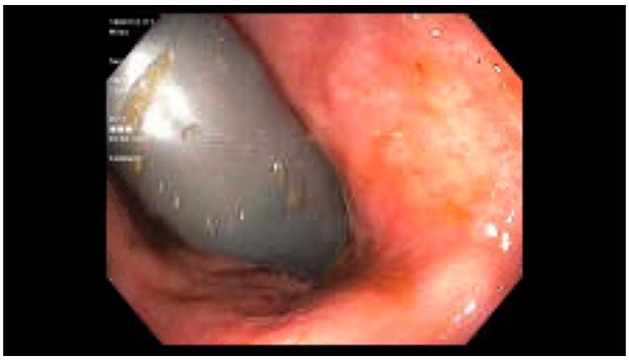

**Figure 2.** Flexible sigmoidoscopy demonstrating rectal sparing.

## DISCUSSION

Squamous cell carcinoma represents 85% of all anal canal tumors.[2] By contrast, adenocarcinoma of the anal canal accounts for a small fraction of all anorectal malignancies and confers a poor prognosis compared with epidermoid cancers.[4] Mucinous adenocarcinoma is an extremely rare subtype, comprising 2%–3% of all gastrointestinal malignancies and 3%–11% of all perianal cancers.[3] As a result of this low prevalence, there are only a few large-scaled trials studying this disease, resulting in a diagnostic and therapeutic dilemma.

The exact pathophysiology of mucinous adenocarcinoma remains unclear. A mechanism has been proposed in which inflammatory reactions transform the glands into mucinous adenocarcinoma.[3] This transformation could occur in the setting of perianal Crohn's disease, perianal abscesses, lymphogranuloma venereum, or fistula-in-ano.[3,6] Anal fistulas result in repeated friction, inflammatory reactions, and scarring, which likely predispose the area to neoplasia. An association with mucinous adenocarcinoma has been well documented.[3,7] Uniquely, the case presented above had no known fistulizing disease and seemingly arose *de novo*.

Patients diagnosed with rectal adenocarcinoma most commonly report anal pain, followed by rectal bleeding.[5] The presentation of this disease is usually late, with the average age of diagnosis being 59 years, possibly due to the nonspecific symptoms associated.[7] As a result of late diagnosis, anal adenocarcinomas generally have a poor prognosis.[7] The association of rectal cancer with inflammatory conditions such as Crohn's disease makes early diagnosis more difficult and may contribute to a later-stage diagnosis.[6] Tumor size appears to be the most important prognostic factor for mucinous adenocarcinoma, which is aggressive with a high rate of recurrence, if untreated, 18-month mortality is as high as 95%.[1,8] Advanced disease with local lymphadenopathy suggests a poor prognosis due to limited ability to perform sphincter-preserving surgery with negative margins.[3] Some cases describe patients with advanced disease who present with inguinal lymphadenopathy, mesenteric, pelvic, or hepatic metastasis, as was the case with our patient. No known relationship exists between mucinous

adenocarcinoma and risk factors such as smoking, obesity, or alcohol consumption. Some reports suggest a 2:1 male-to-female predominance, while other reports suggest no gender correlation.[1,3,7]

When the diagnosis of mucinous carcinoma is made, a complete evaluation of the gastrointestinal tract by both an upper endoscopy and colonoscopy should be performed to confirm the diagnosis.[4] Additional imaging, such as computed tomography scan, magnetic resonance imaging, or endoscopic ultrasound, can help determine the extent of disease. Positron emission tomography scans can aid in metastatic mapping, although anal malignancies rarely metastasize. The most common areas of rectal metastasis reported are the liver, lung, and mesentery.[4]

Owing to the limited prevalence of this disease, treatment regimens have not been subjected to randomized double-blind prospective analysis. Treatment options include surgery, chemotherapy, and radiotherapy. Some studies suggest that multimodal treatment offers the best chance of survival; however, studies are conflicting.[1] One study of 30 patients treated with neoadjuvant therapy showed a 30% survival at 3 years, while another longitudinal study reported only 5% survival at 5 years.[5,9]

This case underscores the significance of a fundamental yet sometimes overlooked skill in medicine: the thorough physical examination. When coupled with the patient's clinical history, this approach ensures that malignancy remains at the forefront of diagnostic considerations. Based on the information available in this case, we do not have enough data to extrapolate a revision of current screening guidelines. We recommend future studies consider a comprehensive analysis of similar cases published in the literature. Once a sufficient body of evidence is available, experts in the field can consider potential changes to guidelines.

Mucinous adenocarcinoma is an extremely rare malignancy with unclear etiopathogenesis. It has been documented that mucinous adenocarcinoma may arise from chronic inflammation of the anorectal region in individuals with anal fistula or *de novo* from anal glands. This case is unique in that the endoscopic examination was benign, but superficial inspection revealed a large malignancy. Our case was also rare in that the patient had no known history of anal fistula or inflammatory conditions. Finally, the disease burden at the time of diagnosis in this case seems to be underrepresented in the literature. We want to draw attention to an uncommon presentation of a rare tumor and the diagnostic and therapeutic challenges it imposes.

## DISCLOSURES

Author contributions: All authors meet the ICMJE criteria for authorship by substantially contributing to the work, drafting, reviewing and confirming final version for publication. D. Wozny is the article guarantor.

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