

Case report

Clinical course of a tailgut cyst transformed to retrorectal mucinous adenocarcinoma: A case report

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ABSTRACT

Introduction and importance: Tailgut cysts (TGC) are rare retrorectal lesions originating from embryological remnants of the hindgut. Malignant transition is rare; treating it with an unplanned diagnostic algorithm can complicate the clinical course.

Case presentation: We report a case of retrorectal mucinous adenocarcinoma arising from a TGC in a 63-year-old female. She had experienced constipation, recurrent urinary tract infections, and difficulty voiding for three months. A total hysterectomy was performed elsewhere for a presumed leiomyoma. A transrectal biopsy was negative, but partial cystectomy was attempted. Histology showed a benign lesion. Postoperatively, she developed pelvic sepsis and urinary/fecal incontinence and was referred to our clinic. MRI revealed a 16 × 12 cm pericoccygeal cyst compressing the rectum and bladder. The cyst, rectum, and coccyx were resected en bloc. Pathology showed mucinous adenocarcinoma arising from TGC with positive radial margins. Adjuvant chemoradiotherapy was recommended.

Clinical discussion: This case highlights diagnostic and management challenges in retrorectal lesions. Inadequate interventions such as transrectal biopsy or partial resection may result in sepsis and tumor spillage. A multidisciplinary and experienced team is essential for optimal outcomes.

Conclusion: Management of retrorectal lesions should be tailored by a multidisciplinary team. The index surgical approach determines the ultimate outcome. Transrectal biopsies and partial resections should be avoided due to the high risk of pelvic sepsis and tumor dissemination.

1. Introduction

Tailgut cysts (TGC) are rare cysts arising from embryonic hindgut remnants. They are mainly located in retrorectal space. Retrorectal, also referred to as presacral space, is a potential space demarcated by presacral fascia in the posterior, fascia propria of the rectum, and mesorectum in the anterior, levator ani muscle in the inferior and peritoneal reflection superiorly [1]. TGC is heterogeneous, and the decision for surgical approach is challenging. Magnetic resonance imaging (MRI) is crucial for identifying TGC characteristics and optimizing diagnostic and therapeutic approaches. Although female predominance is reported in retrospective series, a systematic review has demonstrated higher malignant transformation rates among males (48 % and 27 % for males and females, respectively) with an overall incidence of 26 % [2]. Multidisciplinary tumor (MDT) boards are essential for assessing optimal clinical and surgical approaches due to the risk of malignant transformation.

Herein, we present the complicated clinical course of TGC with mucinous adenocarcinoma managed in an academic university hospital. This case report has been reported in line with the SCARE checklist [3].

2. Case report

A 63-year-old female patient was admitted to our hospital with concerns of constipation, recurring urinary tract infections, and difficulty voiding, which persisted for three months. She had undergone a total hysterectomy for a large leiomyoma at another institution previously. Despite seeking treatment at various hospitals, her symptoms remained unresolved. A transrectal biopsy showed no signs of malignancy, and at a different hospital, a partial cystectomy was performed in an attempt to remove the lesion. Pathological examination revealed no malignancy. Fecal incontinence developed following the pelvic sepsis, which occurred postoperatively and the patient was then referred to our

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clinic with a retrorectal abscess. Abdominal MRI revealed a 16×12 cm peri-coccygeal complicated cyst compressing the rectum and bladder with low signal on T1-weighted and high signal intensity on T2-weighted images. Septal alterations showed significant enhancement after contrast administration (Fig. 1). We proceeded with the complete removal of the remnant cyst with attached rectum plus coccyx, a colorectal anastomosis, and a diverting loop ileostomy were created. The surgery started with the lithotomy position, then we switched to the jackknife position. The remnant cystic lesion was observed as infected and adhered to the intra-pelvic structures intraoperatively. There were no complications or adverse outcomes related to the procedure.

The surgical specimen was thoroughly examined macroscopically as wholmount transverse sections. Microscopic evaluation revealed that the TGC is lined by columnar epithelium containing mucinous epithelium with papillary growth pattern. Cyst epithelium showed extensively malignant transformation to mucinous adenocarcinoma (Fig. 2). Invasive areas consist of poorly differentiated carcinoma cell groups in extracellular mucin secretion. Cyst epithelium and carcinoma cells stained positive with CK7 and negative with CK20. The circumferential radial margin of the resection material was positive. Immunohistochemical analyses exhibited proficient mismatch repair protein (pMMR) (Fig. 3) and negative HER2. In molecular analysis, the tumor was pan-RAS and BRAF wild. The MDT board recommended adjuvant chemoradiotherapy (CRT).

Since there is no standard adjuvant protocol for malignant TGC, she was planned to receive adjuvant treatment similar to adjuvant rectal cancer protocols: radiotherapy (RT) after 2–3 cycles of chemotherapy with CAPEOX (capecitabine and oxaliplatin). However, the patient started to complain about severe neuropathic pain radiating from the sacrum to the lower extremity, gait disturbances due to pain, and weakness in the lower extremities, in addition to urinary retention symptoms after the second cycle. A suspicious destructive lesion in the sacrum was identified in the control pelvic MRI, and a biopsy revealed infiltration of mucinous adenocarcinoma. She was referred for RT for palliation of her symptoms; she received 5×5 gy RT to the sacrum. FOLFIRI (5-Fluorouracil, folinic acid, irinotecan) + cetuximab regimen was initiated as second-line therapy after RT. Her symptoms improved nearly totally after the second cycle of systemic treatment; however, a control MRI after the fifth cycle revealed metastasis in L4–5 vertebrae. A timeline of the patient's course is presented in Fig. 4.

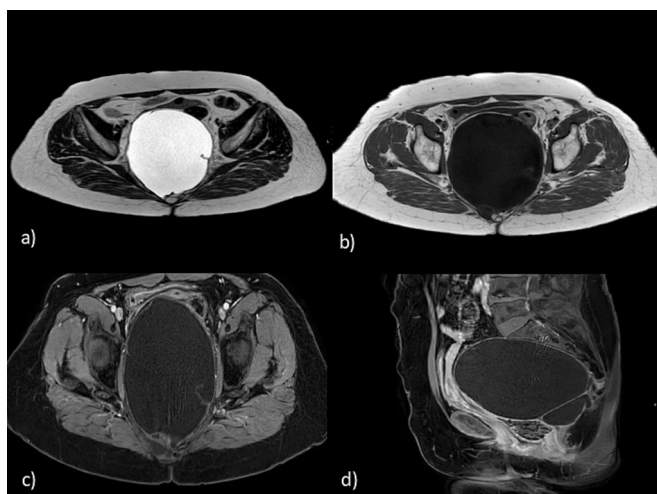


Fig. 1. Displays a detailed view of the axial T1-weighted (a), axial T2-weighted images (b), T1-weighted axial contrasted (c), and sagittal contrasted (d) MRI images of TGC. The images provide a clear visualization of the structure and composition of TGC, allowing for a more comprehensive understanding of the subject.

3. Discussion

This case report highlights the necessity of multidisciplinary diagnostic and therapeutic management of TGCs. Inadequate work-up and unnecessary interventions can lead to a complex situation, resulting in a missed opportunity for a cure, as was the case in our study. A severe symptomatic course is associated with an increased risk of postoperative morbidity and delay in proper clinical approach [4].

In determining the precise histological diagnosis in presacral tumors, preoperative MRI and preoperative biopsy demonstrated a sensitivity of 84 % and 86 %, respectively [5]. The preoperative biopsy may assist in determining the surgical approach and potential postoperative complication management. A study by Sagar et al. found no evidence of preoperative biopsy having a detrimental effect on the clinical and surgical management of TGCs. Meanwhile, the biopsy may indicate the need for neoadjuvant treatment for optimal management [4]. Holmstrom et al. reported that the presence of both benign and malignant tissues, known as tumor heterogeneity, can lead to preoperative misdiagnosis and mislead clinicians [6].

It should be noted that endoscopic biopsy has been associated with the potential risk of eliciting tumor spread, fecal abscess, and fistula [6–8]. Nevertheless, two single-experience case series from the Cleveland Clinic and the Mayo Clinic reported no tumoral inoculation at the biopsy site [1,6]. It is crucial to consider the milieu of the mass when evaluating the appropriate course of action for diagnostic procedures. A review of 42 studies comprising 932 patients revealed that performing a biopsy was the recommended course of action for solid presacral tumors [9]. Kruckemeier et al. have also suggested an alternative to preoperative biopsy, which is an intraoperative frozen section work-up [10]. It is noteworthy that intraoperative biopsy could result in missed opportunities for neoadjuvant therapy. For instance, in a case report by Baverez et al., a preoperative biopsy of TGC revealed mucinous adenocarcinoma, prompting the administration of neoadjuvant CRT (50 Gy in 25 fractions plus concurrent capecitabine 800 mg/m² twice daily) per guidelines for locally advanced rectal adenocarcinoma. This approach can effectively reduce the risk of postoperative recurrence [11]. Another case of TGC diagnosed as mucinous adenocarcinoma after R0 resection received adjuvant chemotherapy with capecitabine 1000 mg/twice per day for 8 cycles without neoadjuvant therapy or adjuvant radiotherapy [12]. There is limited data about the biology and molecular features of TGC transformed into mucinous adenocarcinoma. Generally, the clinical approach is determined by extrapolation from rectal cancer protocols since there is no standardized neoadjuvant and adjuvant treatment approach for retrorectal tumors.

Due to its rarity, limited cases are reported in the literature, which precludes setting a standard of care. Nevertheless, an individualized, patient-centered medical approach is critical. In our case, a previous failed attempt of resection had resulted in severe symptoms and cyst abscess upon initial presentation leading to a late-stage diagnosis. As a result, achieving a clear resection margin and en bloc resection was impossible. To minimize the potential for such complications, engaging in a collaborative decision-making process via MDT boards or referring patients to tertiary centers in the absence of boards is essential. Although rare, MDT should remain vigilant about malignant transformation. Upon this, radiologists included in MDT play a significant role in differential diagnosis and detection of malignant features such as mural nodularity and postcontrast enhancement [13].

Furthermore, close postoperative surveillance is crucial for assessing the need for reoperation in cases with residual macroscopic disease. Accurate preoperative staging, supported by immunohistochemistry and molecular profiling, may guide systemic treatment decisions. In selected cases, this may include immunotherapy or targeted therapy, particularly when histological features such as mucinous differentiation or microsatellite instability are present.

A multidisciplinary, collaborative approach ensures optimal diagnostic accuracy and therapeutic decision-making tailored to each

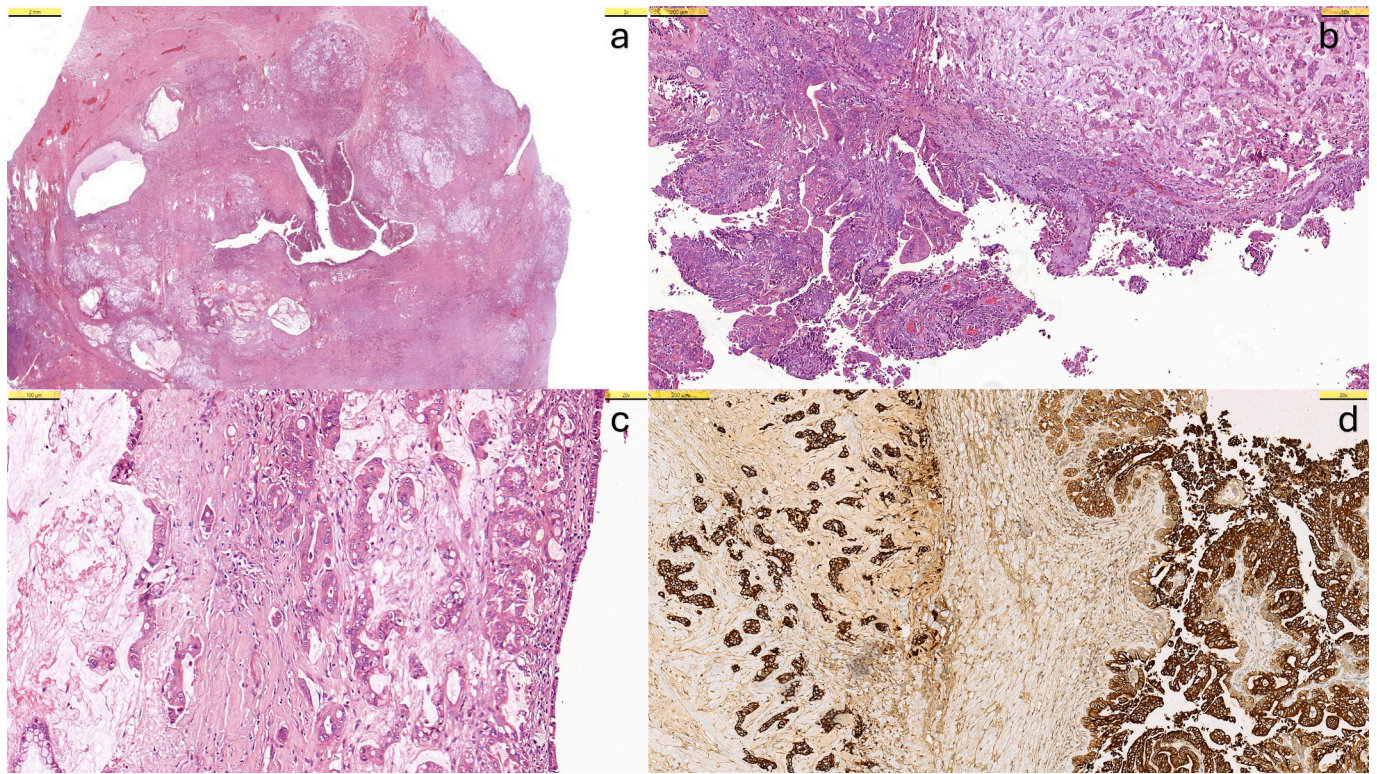


Fig. 2. a) Wholemount section of tailgut cyst wall lined by columnar epithelium containing mucinous epithelium (HE, 2×). b) In higher magnifications columnar epithelium with papillary growth of cyst shows neoplastic transformation to mucinous adenocarcinoma. c) Poorly differentiated cancer groups in extracellular mucin pools. d) Both cyst epithelium and carcinoma cells are stained strongly and diffusely with CK7.

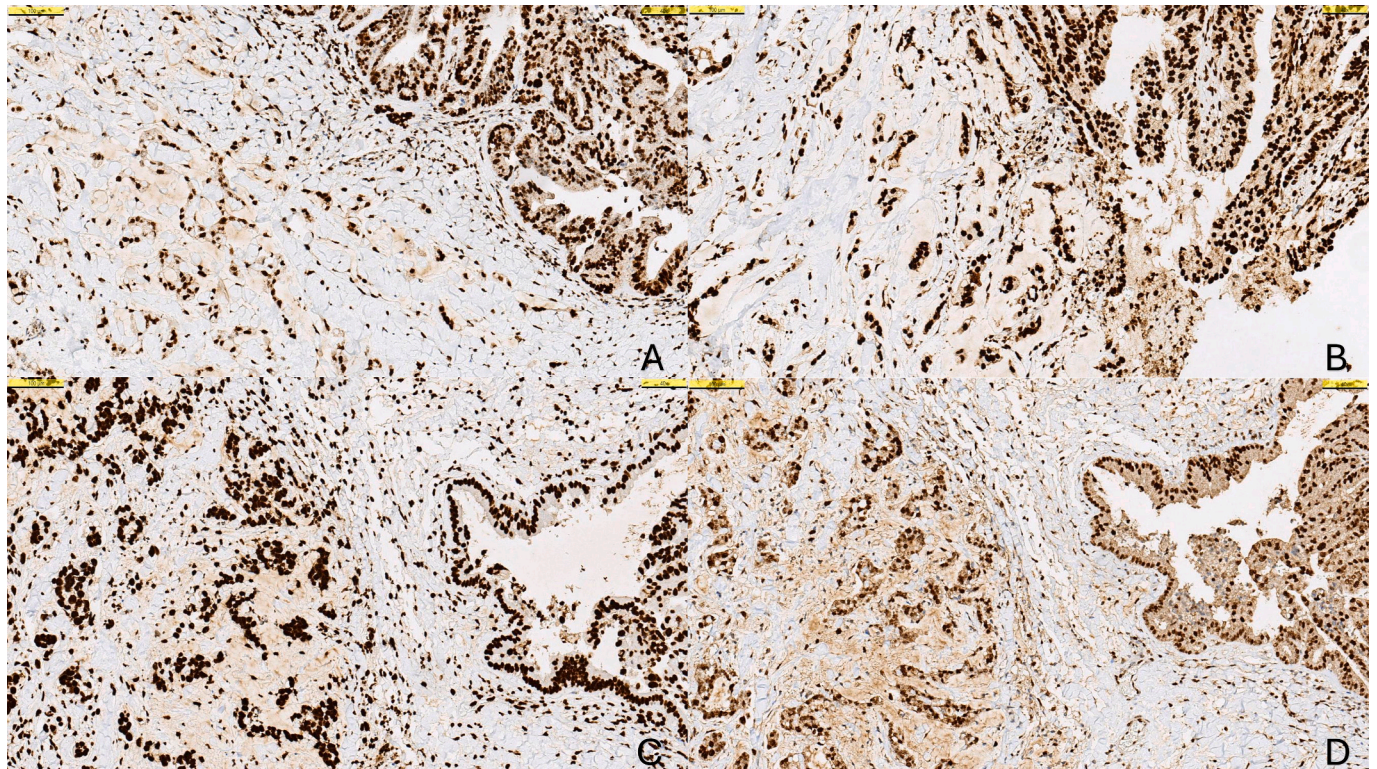


Fig. 3. Immunohistochemistry shows intact nuclear staining in both cyst epithelium and carcinoma cells with MLH1 (A), MSH2 (B), MSH6 (C) and PMS2 (D) (pMMR).

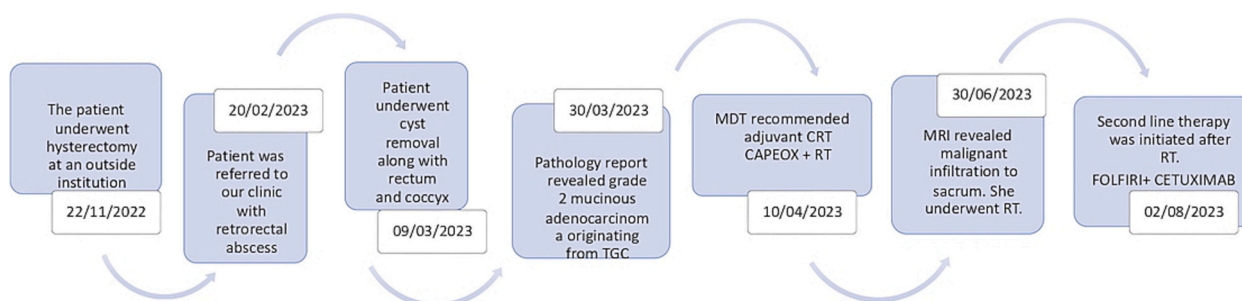


Fig. 4. Timeline of the patient's management.

patient's clinical scenario.

4. Conclusion

The use of MDT boards can improve decision-making, reduce unnecessary interventions, and increase positive outcomes for patients. Therefore, healthcare providers are encouraged to adopt this approach as a fundamental aspect of their practice in TGC management.

Author contribution

Writing original draft: Mirac Ajredini, Erman Aytac, Leyla Ozer. Supervision: Leyla Ozer, Erman Aytac, Sibel Erdamar Cetin. All authors participated in the revision of the manuscript and approved the submitted version.

Consent for publication

Written informed consent was obtained from the patient for the publication of this case report, including accompanying images and details. All efforts were made to ensure patient confidentiality.

Ethical approval

Ethical approval was not required for this case report as it is a retrospective analysis of a single patient, and no experimental interventions were performed. Written informed consent was obtained from the patient for the use of clinical data and images for publication purposes.

Guarantor

Leyla Ozer.

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Conflict of interest statement

The authors declare no conflicts of interest.

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Data availability

Data sharing not applicable to this article as no datasets were generated or analysed during the current study.

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