Retrorectal cystic hamartoma (tailgut cyst): a rare etiology of anal neoplasia - case report

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ABSTRACT

Background: Retrorectal cystic hamartoma, also known as a tailgut cyst, is a rare type of space-occupying lesion. It is a cystic hamartoma that predominantly forms in the retrorectal region and may serve as a potential etiology for anal neoplasia.

Case Presentation: A 58-year-old female patient was initially treated for a high-grade squamous intraepithelial lesion of the anal canal, which was surgically removed on May 20, 2023, followed by radiotherapy. She also had a known tailgut cyst with a fistula connecting it to the perianal skin at the 6 o'clock position in the lithotomy view. The cyst had been asymptomatic until then. As a prophylactic measure to prevent potential malignant transformation, the decision was made to excise the tailgut cyst. The operation was performed laparoscopically and included perianal mobilization of the fistula. During follow-up, the patient developed perianal pain. A computed tomography scan revealed a fluid collection at the site of the excised tailgut cyst. The collection was surgically drained and identified as a seroma without any signs of infection. The patient was discharged after 7 days and experienced no further long-term complications.

Conclusion: Retrorectal cystic hamartomas are rare and often asymptomatic. When symptomatic, they may present with lower abdominal pain, back pain, obstipation, urinary retention, or complications such as anal abscesses and fistulas. Research to date remains inconclusive regarding the incidence of malignant transformation.

Keywords: Tailgut cyst, retro-rectal cystic hamartoma, anal neoplasia, cystic hamartoma, hindgut remnant.

Type of Article: CASE REPORT **Speciality:** General Surgery

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Background

Retrorectal cystic hamartoma (also referred to as a tailgut cyst) is a rare space-occupying lesion, with an incidence of approximately 1 in 40,000 and a female-to-male ratio of 5:1 [1]. Its etiology is a congenital malformation resulting from remnants of the embryonic hindgut. Diagnosis is often incidental and typically made through imaging studies such as computed tomography (CT) or magnetic resonance imaging (MRI). Recognizing tailgut cysts in routine gastroenterological and colorectal practice is of substantial clinical importance. Although often asymptomatic, these lesions can lead to complications including infection, compression of adjacent structures, and, most notably, malignant transformation into adenocarcinoma, neuroendocrine tumors, or squamous cell carcinoma (SC). Misdiagnosis is common due to the nonspecific presentation and deep pelvic location, with tailgut cysts frequently mistaken for perirectal abscesses, fistulas, or pilonidal disease. Timely and accurate identification facilitates appropriate surgical planning, minimizes the risk of recurrence or incomplete resection, and improves patient outcomes. Furthermore, appropriate recognition allows for symptom relief in cases of obstructive or inflammatory complications and enables multidisciplinary coordination between gastroenterologists, radiologists, and colorectal surgeons. Given these factors, routine consideration of tailgut cysts in the differential diagnosis of presacral masses is essential for optimal patient care. Treatment strategies should be individualized, balancing the risks and benefits of surgical intervention. When feasible, complete surgical excision is recommended to mitigate the potential for malignant transformation and recurrence [2].

Case Presentation

A 58-year-old female patient in good general health, with a history of breast cancer, malignant melanoma, and a known asymptomatic retrorectal cystic hamartoma, was referred to the gastroenterology department by her gynecologist after a digital rectal examination revealed a hemorrhagic space-occupying lesion. Gastroenterological

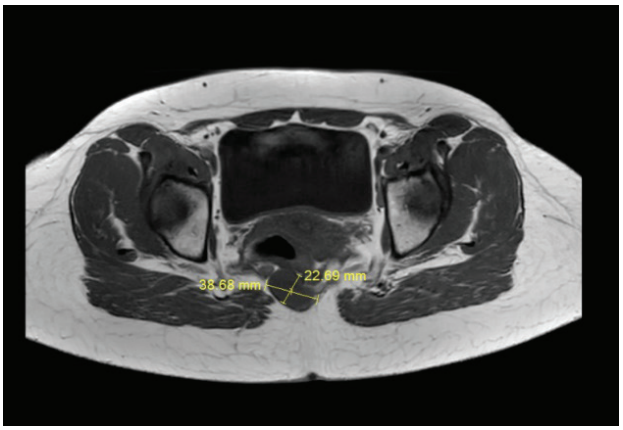

Figure 1. *Axial section of pelvis MRI exhibiting the tailgut cyst (white arrow).*

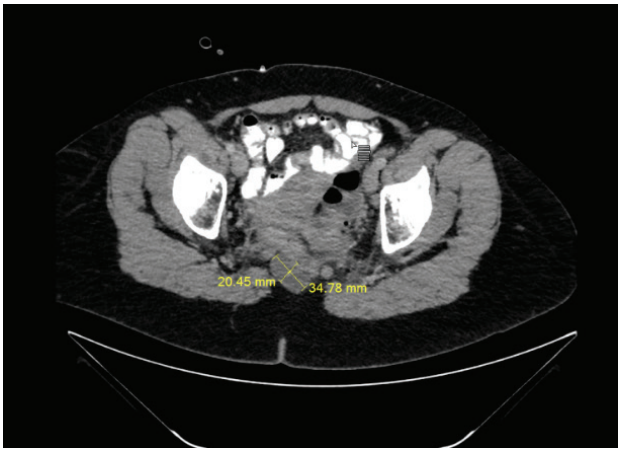

Figure 2. *Axial sections of the abdomino- pelvic CT scan (white arrow) showing the tailgut cyst (white arrow).*

evaluation identified an anal tumor located 2 cm from the anal verge. CT and MRI scans confirmed the presence of the previously diagnosed tailgut cyst as a right paramedian cystic formation measuring 3.8 × 2.3 × 4.1 cm (Figures 1 and 2). Endoscopic ultrasound was also performed and revealed a well-defined, hypoechoic lesion in the retrorectal space, consistent with the imaging characteristics of a tailgut cyst.(Figure 5).

The initial histological analysis of the anal tumor revealed a high-grade squamous intraepithelial lesion (HSIL). Due to the initial uncertainty about malignancy, our interdisciplinary tumor board recommended surgical excision. Transanal submucosal dissection was successfully performed, and the patient was discharged without complications. However, subsequent histopathological analysis identified an invasive SC carcinoma (pT1, pNX, L0, V0, Pn0, R1, G2). Based on these findings, radiochemotherapy was initiated as the recommended treatment.

Given the patient's oncological history and the newly confirmed HSIL, the decision to pursue prophylactic surgical removal of the previously asymptomatic tailgut cyst was made 1 year later in a multidisciplinary setting.

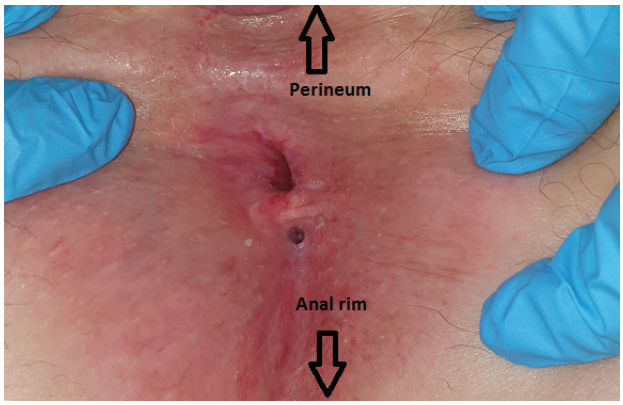

Figure 3. *Picture of the perianal fistula in a lithotomy position.*

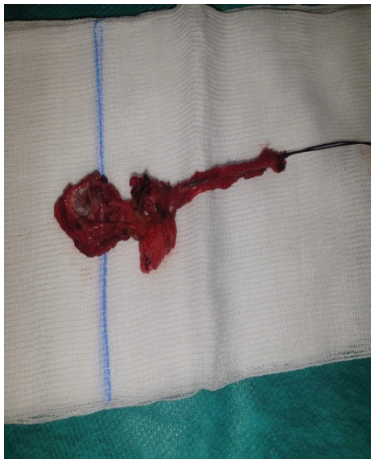

Figure 4. *The sample of the tailgut cyst after resektion.*

Although the cyst had remained stable and symptom-free, the cumulative cancer risk and the documented potential for malignant transformation of tailgut cysts warranted proactive intervention in this context.

The procedure was performed laparoscopically and involved perianal mobilization of the associated fistula, which was located along the external anal sphincter (Figure 3). A combined abdominal - perineal approach was selected to ensure complete visualization and safe dissection of the deep pelvic cyst. The laparoscopic route offered enhanced access to the retrorectal space, minimizing soft tissue trauma, while the perineal component allowed precise mobilization of the fistulous tract and optimal cyst retrieval. Alternative approaches such as open transabdominal or isolated posterior perineal resection, were considered less favorable due to higher morbidity and limited visualization in the setting of prior pelvic radiotherapy.

Surgically, the approach included a retroperitoneal incision on the right side, medial to the common iliac artery. The mesorectum was carefully mobilized from the sacrum, with preservation of the dorsal nerve fibers. Dissection proceeded through the mesorectal fascia until reaching the pelvic floor. The right lateral attachment of

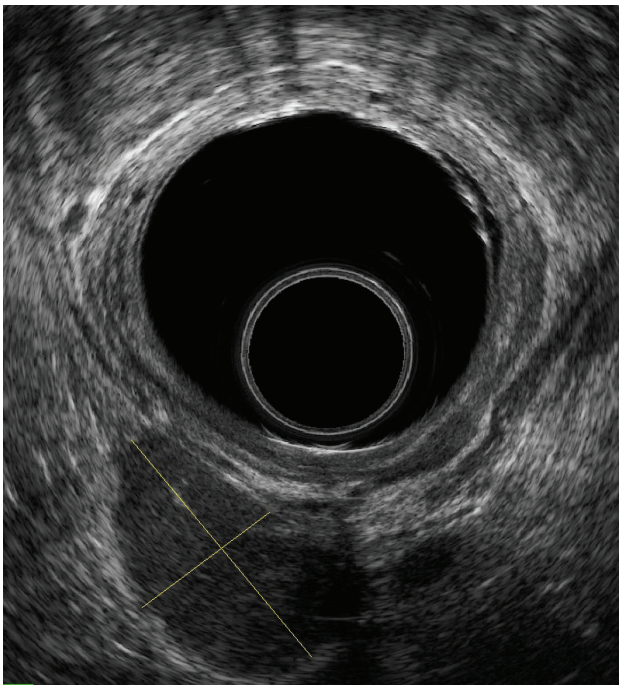

Figure 5. *Endosonography: hypoechoic cystic lesion dorsal to the anal canal.*

the mesentery was divided, and the middle rectal artery was transected to improve visualization. The levator muscle was opened longitudinally to expose the cyst, which was gradually dissected from the surrounding tissues. Complete removal of the cyst was achieved using a rendezvous technique via both abdominal and perineal approaches, with the cyst retrieved through the perineal incision (Figure 4).

The patient was discharged 5 days postoperatively but returned 3 days later with increasing perianal pain. Blood tests revealed a minor elevation in C-reactive protein levels. A CT scan showed a fluid collection in the retrorectal region, which was surgically drained. No evidence of infection or rectal fistula was identified. Postoperatively, the patient received vacuum therapy and transitioned to open wound care on an outpatient basis. She was discharged after 7 days.

During subsequent follow-up visits, the patient reported urinary retention and fecal incontinence, both of which gradually resolved over time.

Discussion

Retrorectal cystic hamartomas are often asymptomatic but may present with vague symptoms resembling other conditions, such as generalized abdominal pain, urinary retention, sciatic radiculopathy, or superimposed infection of the tailgut cyst. In cases of infection, patients may exhibit common symptoms such as fever, malaise, and, in severe cases, sepsis with cardiovascular failure and multiorgan dysfunction. These symptoms most frequently occur between the ages of 30 and 60. The differential diagnosis includes dermoid cysts, ovarian cysts, sacral meningoceles, lymphoma, and retrorectal abscess [3-6]. MRI and CT are the primary diagnostic tools, often identifying the tailgut cyst incidentally. Due to potential complications, including infection or malignant transformation, surgical removal is generally recommended following a personalized benefit-risk assessment. The surgical approach for tailgut cyst removal may be abdominal (via laparoscopy or laparotomy) or posterior (pararectal or transanal). In the presented case, a combined approach was employed due to the presence of a perianal fistula. The fistula's location guided the mobilization of the fistula duct, facilitating laparoscopic mobilization of the tailgut cyst and enabling en bloc resection through the perineal incision. While this combined laparoscopic and perineal approach provided optimal visualization and complete resection, it is not without limitations. The technique is technically demanding and requires careful coordination between the abdominal and perineal teams. There is a risk of injury to pelvic autonomic nerves, which may result in temporary or persistent urinary or fecal dysfunction - as was observed postoperatively in this case. Additionally, the dual-incision strategy may increase the risk of wound-related complications, such as fluid collections or delayed healing, particularly in patients with prior pelvic radiation or multiple surgeries. The necessity for precise anatomical dissection also increases the likelihood of intraoperative bleeding or inadvertent injury to adjacent structures such as the rectum or neurovascular bundles. Therefore, although the combined approach allowed for a tailored and complete excision in this complex anatomical setting, it should be reserved for selected cases in which standard approaches may not ensure complete or safe resection. Thorough preoperative planning and intraoperative adaptability are essential to mitigate these risks.

Conclusion

Further clinical studies are needed to better understand the risk of malignant transformation, as current data remain insufficient. Laparoscopic en bloc resection is a safe and effective method, offering excellent visualization of surrounding structures and a low risk of infection.

What is new?

This case illustrates a rare instance of invasive squamous cell carcinoma occurring concurrently with a diagnosed asymptomatic tailgut cyst, highlighting the importance of proactive management in high-risk patients with significant oncologic history.

A combined laparoscopic–perineal "rendezvous" approach was employed for complete cyst excision in the setting of a perianal fistula and prior pelvic radiotherapy, demonstrating the technique's feasibility and benefits in anatomically complex scenarios

List of Abbreviations

CT Computed tomography
HSIL High-grade squamous intraepithelial lesion
MRI Magnetic resonance imaging

Take home message

Increasing awareness of tailgut cysts among clinicians - gastroenterologists, general surgeons, gynecologists, and urologists - should be a primary goal to facilitate the prompt recognition and diagnosis of this lesion.

Conflict of interest

The authors dedclare that they have no conflict of interest regarding the publication of this case report.

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None.

Consent for publication

Written informed consent for publication was obtained from the patient.

Ethical approval

Written informed consent was obtained from the patient, ensuring anonymity.

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Summary of case

1	Patient (gender, age)	60 years old
2	Final diagnosis	Tailgut cyst
3	Symptoms	High-grade squamous intraepithelial lesion
4	Medications	None
5	Clinical procedure	Laparoscopic removal of the tailgut cyst after transanal mobilization
6	Specialty	General surgery