

# Presacral tailgut cyst with complex perianal fistula: A case report and a brief review of the literature

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**Abstract.** Tailgut cysts (TGCs) are rare congenital cystic lesions that pose diagnostic challenges due to varied manifestations, anatomic locations and a tendency to mimic or associate with other anorectal or pelvic pathologies. The present case report describes a challenging case of TGC associated with a complex perianal fistula. A 41-year-old female patient presented with painful defecation. A digital rectal examination revealed a deep, soft, ill-defined fullness palpable in the presacral region. There were no palpable masses, secondary tracts, abscess cavities, or signs of purulent discharge. Magnetic resonance imaging revealed a multiloculated cystic lesion in the presacral region associated with a complex perianal fistula. The cyst and its associated fistulous tract were removed by en bloc excision. A histopathological examination of the cyst confirmed a TGC. In addition, in reviewing 13 recently reported cases, the common clinical presentations included lower abdominal or pelvic pain and constipation, while approximately a quarter of the cases were incidentally found. Malignant transformation was documented in 5 cases, most commonly into adenocarcinoma, with metastases in 3 cases. Of the cases with malignant transformation, 3 cases succumbed to disease. All patients underwent surgical excision, with the open approach used most frequently (~76.9%).

Outcomes were excellent among patients with benign lesions, with no recurrence during follow-up, whereas those with malignant transformation had a poorer prognosis. TGCs may be associated with or masked by other perianal conditions, such as fistulas, necessitating proper investigation to avoid misdiagnosis or improper management.

## Introduction

Tailgut cysts (TGCs), also known as retrorectal cystic hamartomas, are rare congenital cystic lesions originating from remnants of the embryonic hindgut (1,2). They are typically located in the retrorectal or presacral space, a potential anatomical area bounded anteriorly by the fascia propria of the rectum and mesorectum, posteriorly by the presacral fascia, inferiorly by the levator ani muscle and superiorly by the peritoneal reflection (1). The disease was first recognized in the literature as developmental anomalies arising from the incomplete involution of the postcloacal gut during embryogenesis (3,4). Normally, the tailgut undergoes regression by the 6th week of gestation; failure of this process results in the formation of these cystic lesions (3). A number of patients remain asymptomatic, and lesions may be discovered incidentally (5,6). However, symptomatic cases can present with nonspecific complaints, including lower abdominal or perineal pain, constipation, dyschezia, urinary dysfunction, or neurological symptoms (2,4,7). This anomaly predominantly affects middle-aged women, with reported female-to-male ratios ranging from 3:1 to as high as 9:1, although malignant transformation appears to occur more frequently in males (1,3,4). Despite the female predominance, the overall incidence is extremely low, estimated at ~1 in 40,000 individuals, highlighting the rarity of this condition (6,8). The diagnosis of TGCs remains challenging due to their rarity, nonspecific symptoms and anatomic location, which may mimic or be associated with

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other anorectal or pelvic pathologies (6). In addition, given the potential for neoplastic transformation, early recognition and management are deemed necessary (2,5). The present case report describes a case of TGC associated with a complex perianal fistula in a middle-aged patient.

### Case report

*Patient information.* In July 2024, a 41-year-old female patient presented to Smart Health Tower (Sulaymaniyah, Iraq) with severe anal pain during defecation, without radiating to other regions. The patient denied bleeding per rectum, urinary symptoms (including dysuria, frequency, or urgency) and any signs of genitourinary disease, and reported regular bowel habits. She had no history of anal or perineal discharge, local trauma, systemic symptoms such as fever, chills, weight loss, or anorexia, nor any chronic illnesses, including diabetes mellitus, inflammatory bowel disease, or malignancy. The family history for gastrointestinal, colorectal, or congenital conditions was negative. Her menstrual and gynecologic history was normal, with regular cycles and no reported complications. She had undergone three prior cesarean sections and a laminectomy ~2 months prior to presentation. There was no history of immunosuppressive therapy, radiation, or previous pelvic interventions.

*Clinical findings.* Upon a physical examination, the patient appeared comfortable, afebrile and hemodynamically stable. The abdomen was soft, with no rebound tenderness, guarding, palpable masses, organomegaly, or ascites. Bowel sounds were normal and audible. A perineal inspection identified a small external wound at the 6 o'clock position relative to the anal verge, with no active discharge or signs of local cellulitis. There were no signs of perianal abscess, skin breakdown in the anal verge, or external hemorrhoids. A digital rectal examination revealed a normal resting tone with pinpoint tenderness in the presacral region and localized tenderness along the posterior rectal wall. There was a deep, soft, ill-defined fullness palpable in the presacral region, while no palpable masses, secondary tracts, abscess cavities, or signs of purulent discharge were noted. The overlying rectal mucosa was smooth, and there was no intraluminal bulge or mass effect. A neurological examination revealed normal lower limb strength and reflexes. Anal reflex and voluntary contraction were intact.

*Diagnostic assessments.* Blood investigations revealed a normal complete blood count. Stool calprotectin was measured at 20.2  $\mu\text{g/g}$  (normal range, <50  $\mu\text{g/g}$ ), excluding active inflammatory bowel disease. Trans-perineal ultrasonography identified a single external opening at the 6 o'clock position at the anal verge, from which a tract extended into the intersphincteric plane. A multiloculated, thin-walled cystic lesion, measuring 25x16x10 mm, was identified posterior to the anal canal. The absence of direct communication with the rectal or anal canal lumen suggested an extra-luminal origin. A magnetic resonance imaging (MRI) was conducted and confirmed the presence of a multiloculated cystic lesion, measuring 30x16x15 mm, situated in the presacral region, anterior to the distal rectum, and extending towards the anorectal junction. The cyst was thin-walled and lacked solid

components, with no connection to the rectal lumen (Fig. 1). Furthermore, the MRI revealed an associated complex perianal fistula, classified as grade II according to the St. James's Hospital Classification. An ileo-colonoscopy revealed patchy areas of colitis, and multiple biopsies were obtained. A histopathological analysis was performed on 5- $\mu\text{m}$ -thick paraffin-embedded tissue sections. The sections were fixed with 10% neutral buffered formalin at room temperature for 24 h and then stained with hematoxylin and eosin (H&E; Bio Optica Co.) for 1-2 min at room temperature. The sections were then examined under a light microscope (Leica Microsystems GmbH). The histopathological examination revealed no notable abnormalities, effectively ruling out inflammatory or neoplastic conditions (data not shown).

*Therapeutic intervention.* Under the jackknife position, the patient underwent a fistulectomy. The external opening was identified, and the fistulous tract was meticulously followed using electrocautery, ensuring controlled dissection in the intersphincteric area. This dissection led to the identification of the underlying multiloculated cystic lesion. The cyst and its associated fistulous tract were removed by en bloc excision, ensuring the complete removal of both the pathological cyst and the communicating fistula. Following hemostasis, the surgical defect was intentionally left open to heal by secondary intention. The procedure was completed without any intraoperative complications. A histopathological examination of the cyst was performed on 5- $\mu\text{m}$ -thick paraffin-embedded tissue sections. The sections were fixed with 10% neutral buffered formalin at room temperature for 24 h and then stained with hematoxylin and eosin (H&E; Bio Optica Co.) for 1-2 min at room temperature. The sections were then examined under a light microscope (Leica Microsystems GmbH). This confirmed a TGC (Fig. 2).

*Follow-up and outcome.* The postoperative course was uneventful. Standard analgesic therapy was administered, and routine wound care was provided to facilitate healing. The perineal wound exhibited healthy granulation tissue formation and progressive contraction, achieving complete closure within 6 weeks of the procedure. During a follow-up period exceeding 12 months, there was no evidence of recurrence of either the fistulous disease or the cystic lesion, and the patient remained asymptomatic.

### Discussion

Tumors and cystic lesions in the presacral or retrorectal space are exceedingly rare (5,9). Owing to the intricate embryologic development of the region, the spectrum of presacral lesions includes inflammatory conditions such as fistulas, abscesses and granulomas, as well as neoplastic lesions. The neoplastic group encompasses osseous and mesenchymal tumors (including osteoma, osteosarcoma, Ewing sarcoma, lipoma, liposarcoma, fibrosarcoma, leiomyoma and leiomyosarcoma), neurogenic tumors (neurofibroma, neurofibrosarcoma, and ganglioneuroma) and congenital lesions such as teratoma, TGC, and rectal duplication cyst. Developmental cysts account for more than half of congenital presacral lesions, and TGCs are among the rarest congenital types derived from primitive

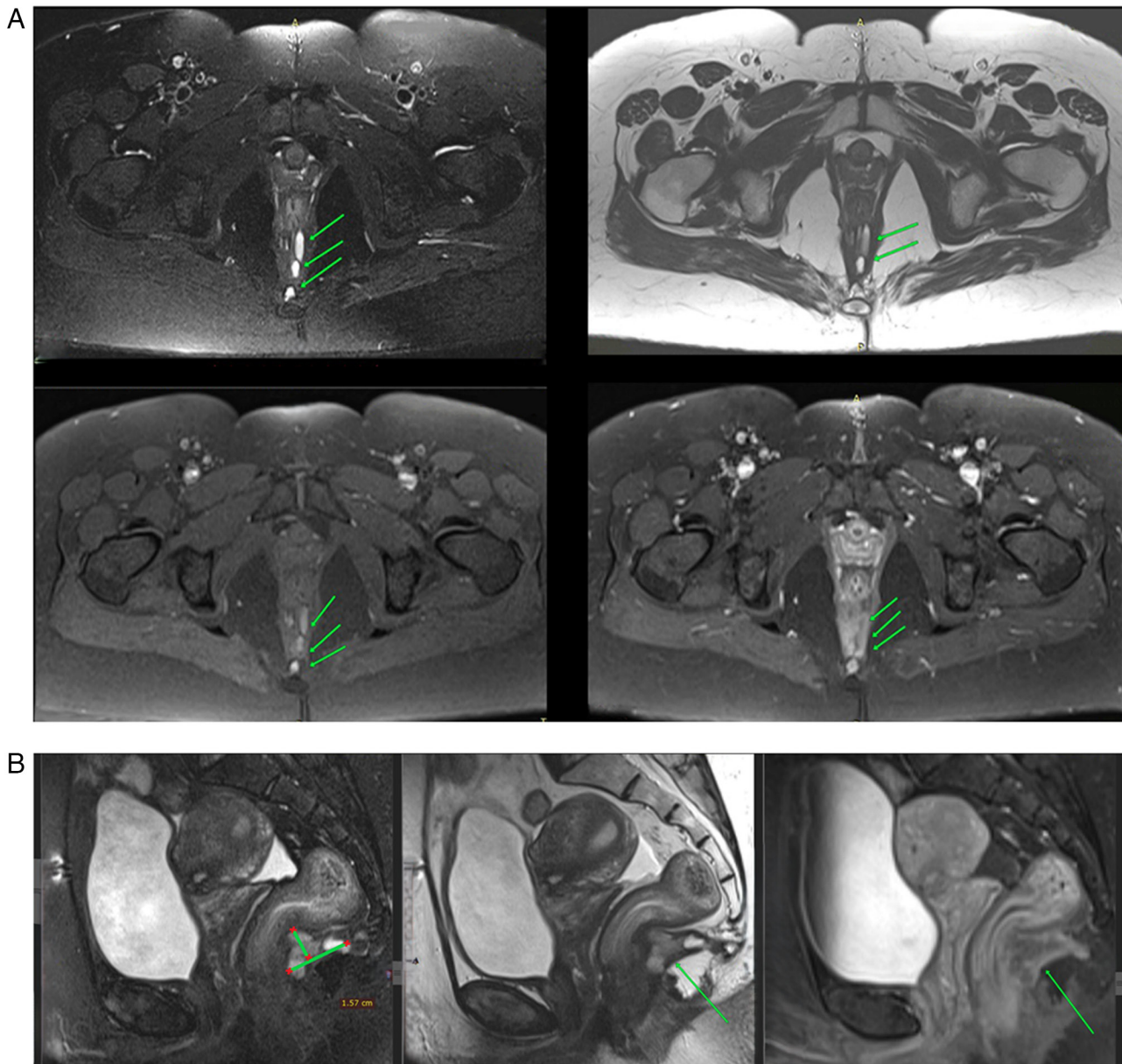


Figure 1. (A) Axial section images [images on the upper panel (left panel, T2-weighted fat-saturated; right panel, T2-weighted); images on the lower panel (left panel, T1 fat-saturated pre-contrast; right panel, T1 fat-saturated post-contrast)] illustrating an elongated multiloculated thin-walled cystic lesion in the presacral space (green arrows), extending from the posterior aspect of the rectum toward the sacrococcygeal region while rectum and vertebrae are normal with no surrounding soft tissue edema. (B) Sagittal plane images (left panel, T2-weighted fat-saturated; middle panel, T2-weighted; right panel, T1 fat-saturated post-contrast) illustrating a presacral space lesion (green arrows), which is above the pelvic diaphragm and extending toward the anorectal junction.

hindgut remnants (4,5,9). Embryologically, during the 4th week of intrauterine life, the embryo folds inward to enclose the primitive gut, and the cloacal membrane (containing endoderm below Hensen's node) becomes positioned ventrally to enclose the distal portion of the hindgut, forming the tailgut. Normally, the tailgut regresses by the sixth week of gestation. Failure of this regression results in a mucus-secreting cystic lesion known as a TGC (10).

Clinically, TGCs exhibit a broad spectrum of presentations, ranging from incidental findings to symptomatic lesions. Symptoms typically result from the mass effect of the lesion on adjacent structures, leading to lower abdominal pain, pelvic or sacral discomfort, constipation, rectal fullness, dysuria, pollakiuria, and urinary retention (6,10). In some cases,

infection or inflammation of the cyst can cause acute pain or lead to the formation of a sinus tract that drains externally, often misdiagnosed as a simple abscess or perianal fistula (11). Furthermore, malignant transformation, although rare, has been reported in up to 13% of cases, resulting in adenocarcinomas or carcinoid tumors (3). Due to their deep pelvic location and non-specific symptoms, retrorectal masses are often misdiagnosed. Half of all cases are discovered incidentally, and there is disagreement regarding the effectiveness of digital rectal examination in their detection (4). When palpable, they are often described as extrinsic, fluctuant masses during digital rectal evaluation, raising suspicion for a retrorectal lesion (6). In the present study, a literature research on Google Scholar was conducted using the key word 'tail gut cyst', and some of

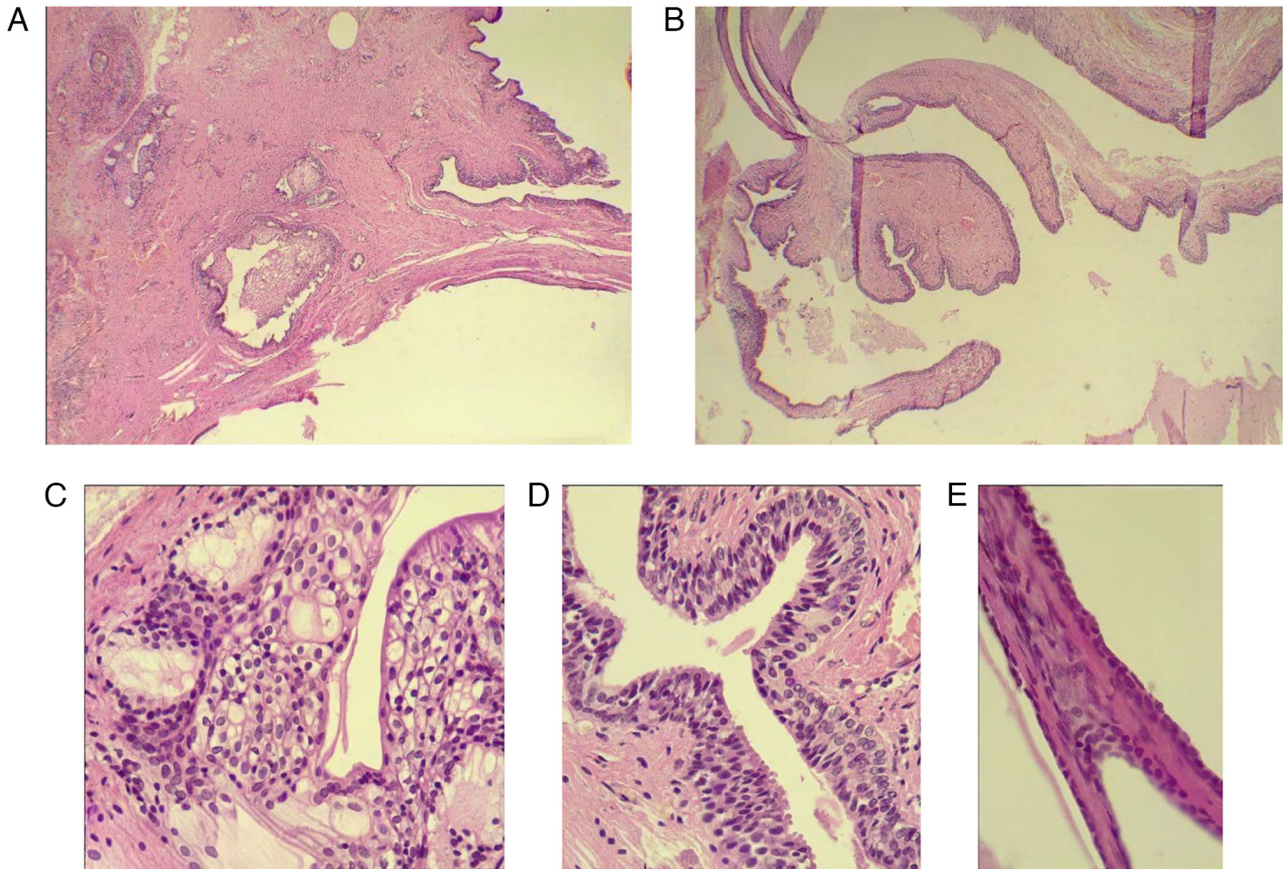


Figure 2. (A) The lesion is cystic and contains numerous broad fibrous papillae that are lined by epithelium. There is debris in the lumen of the cyst. (B) There are various types of epithelium in the lining and wall of the cyst, including transitional, stratified squamous, mucinous columnar, and flat to low cuboidal epithelium. (C) The transitional epithelium is stratified and composed of tall cells with indistinct cytoplasmic borders, a moderate amount of lightly eosinophilic cytoplasm, and oval nuclei with fine chromatin. It differs from the transitional epithelium of the urothelial tract and does not belong to an otherwise specific category of epithelium in the body. (D) The stratified squamous epithelium is non-keratinizing and is composed of cells with distinct cytoplasmic borders, a moderate amount of clear to lightly eosinophilic cytoplasm, and round nuclei with fine chromatin. The mucinous glands are lined by tall columnar cells with abundant cytoplasmic mucin and small, round nuclei with fine chromatin. (E) The flat to low cuboidal epithelium lines some areas of the cyst and is composed of cells with minimal eosinophilic cytoplasm and oval nuclei with fine chromatin. Images illustrate hematoxylin and eosin staining; (A and B) original magnification, x40; (C-E) original magnification, x400.

the recently published cases were reviewed. In reviewing 13 reported cases (1-6,8-10,12-15), there was a marked female predominance (84.6%), and an age range spanning from the neonatal period to 71 years. The common clinical presentations included lower abdominal or pelvic pain, constipation, urinary retention, and perianal swelling, while approximately a quarter of the cases were incidentally discovered. In total, 2 cases were initially misdiagnosed as ovarian or dermoid cysts. Malignant transformation was documented in five cases (~38.5%), most commonly into adenocarcinoma, followed by squamous cell carcinoma, with metastases observed in 3 cases (Table I). The patient in the present case report complained of severe anal pain during defecation without any associated symptoms or significant medical history. Upon a physical examination, there was only a small external wound in the anal verge, without discharge or local cellulitis. A digital rectal examination revealed a deep, soft, ill-defined fullness palpable in the presacral region with no palpable masses or signs of abscess.

The heterogeneous morphology of these cysts contributes to radiological ambiguity, leading to misdiagnosis in >50% of cases (4). The pre-operative diagnosis of TGCs relies heavily on imaging. MRI and computed tomography (CT) are

considered the modalities of choice. On MRI, TGCs are generally hypointense on T1-weighted images and hyperintense on T2-weighted images. Still, these characteristics can vary with cyst content. CT scans usually demonstrate well-defined presacral cystic lesions with central fluid attenuation, peripheral soft-tissue enhancement, and occasional rim calcification (3). Transrectal ultrasound can further define the association of the cyst with the rectal wall and determine whether the lesion is cystic or solid (10). Pre-operative MRI and biopsy have exhibited a sensitivity of 84 and 86%, respectively, for presacral tumors (1). While a biopsy may assist in determining the surgical plan and in identifying malignant transformation, its use is controversial. Biopsy is discouraged in resectable cases as it is associated with the risk of infection or malignant seeding along the biopsy tract (11). However, in unresectable cases, biopsy may be justified to guide neoadjuvant therapy (1). In the case presented herein, trans-perineal ultrasonography identified a single external opening in the anal verge, from which a tract extended into the intersphincteric plane. In addition, a multiloculated, thin-walled cystic lesion was identified posterior to the anal canal, with no direct communication with the rectal or anal canal lumen, suggesting an extra-luminal

Table I. Summary of 13 recently reported cases of tailgut cyst identified in the literature.

Authors, year of publication	Age/sex	Presentation/ symptoms	Duration of symptoms	PMSH	Imaging findings	Size of cyst (cm)	Misdiagnosis/ associated pathology	Malignancy transformation	Resection status of the cyst/ technique	Outcome and recurrence (Refs.)
Malutan <i>et al</i> , 2025	30/F	Low abdominal pain	3 years	Three surgical procedures for presumed left ovarian cysts without relieving symptoms	Presacral tumor with right pararectal development and mass effect on the rectum	2.88	Initially misdiagnosed as a left ovarian cyst	None	Complete/Open (Pfannenstiel approach)	Symptom-free and no recurrence after 1 year of follow-up
Rakia <i>et al</i> , 2025	34/M	Asymptomatic (incidental finding)	N/A	None	Bilobed retrorectal cysts displacing the rectum	4 & 7	None	None	Complete/ laparoscopy	Symptom-free and no recurrence at the 3-month follow-up
Achugatla <i>et al</i> , 2025	Neonate/F	Swelling over mid-gluteal region above anal opening	Since birth	None	Well-defined solid-to-cystic lesion in precoccygeal space initially suggestive of a dermoid cyst	3.64	Suspected as dermoid cyst on imaging	None	Complete/open (Kraske approach)	Symptom-free and no recurrence after 1 year of follow-up
Ajredini <i>et al</i> , 2025	63/F	Constipation, recurrent UTI, difficulty voiding	3 months	Total hysterectomy for a large leiomyoma and partial cystectomy	Pericoccygeal complicated cyst compressing rectum and bladder, later MRI showed a destructive sacral lesion and L4-L5 vertebral metastasis	13.8	None	Transformed into mucinous adenocarcinoma with metastasis	Complete/Open	Symptoms partially improved with progressive metastatic disease
Al Jada <i>et al</i> , 2025	Early 50s/F	Progressive lower abdominal pain, urinary	2 months	None	Well-defined presacral cystic lesion displacing	10.9	None	Transformed into mucinous adenocarcinoma with metastases	Incomplete/ Open (exploratory laparotomy)	Recurrence after 6 months with metastases and

Table I. Continued.

Authors, year of publication	Age/sex	Presentation/ symptoms	Duration of symptoms	PMSH	Imaging findings	Size of cyst (cm)	Misdiagnosis/ associated pathology	Malignancy transformation	Resection status of the cyst/ technique	Outcome and recurrence	(Refs.)
Kitazawa <i>et al</i> , 2025	40/F	retention, and tenesmus	N/A	Uterine polyp treated with endometrial curettage	rectum, uterus, and bladder; follow-up CT/MRI: Recurrent presacral cyst with liver and osseous metastases	1.2	None	None	Complete/ laparoscopy	Symptom-free	(2)
De Crombrugge <i>et al</i> , 2024	59/M	Severe coccygeal pain exacerbated by sitting	N/A	Thrombophlebitis	Large ovoid cystic lesion in anal canal posterior wall	3	None	None	Complete/open	Symptom-free and no recurrence	(8)
Haval <i>et al</i> , 2024	18/F	Intermittent abdominal pain and constipation	6 months	None	Multiloculated cystic mass in presacral/ precoccygeal space displacing rectum	5.4	None	None	Complete/open (transabdominal approach)	Symptom-free	(10)
Kiosov <i>et al</i> , 2024	45/F	Asymptomatic (incidental finding)	N/A	None	Oval hypoechoic inhomogeneous lesion adjacent to posterior rectal wall and connected to its muscle layer	2.5	None	None	Complete/ endoscopy	Symptom-free	(5)

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Table I. Continued.

Authors, year of publication	Age/sex	Presentation/ symptoms	Duration of symptoms	PMSH	Imaging findings	Size of cyst (cm)	Misdiagnosis/ associated pathology	Malignancy transformation	Resection status of the cyst/ technique	Outcome and recurrence (Refs.)	
Manikandan <i>et al</i> , 2024	23/F	Vaginal discharge and lower abdominal pain	N/A	Exploratory laparotomy with marsupialization of cyst	Thick-walled hypodense lesion with septations and calcifications displacing rectum	8.9	None	Transformed into moderately differentiated adenocarcinoma	Complete/open (exploratory laparotomy)	PET-CECT showed residual disease. chemotherapy was planned but patient lost to follow-up	(14)
Moshtaghian <i>et al</i> , 2023	51/F	Pelvic pain, constipation, and fecal urgency	N/A	None	Hypodense cystic lesion in left retrorectal region causing rectal deviation	2.3	None	Transformed into squamous cell carcinoma	Incomplete/ Open	Recurrence and metastasis; succumbed after 1 year	(9)
Russo <i>et al</i> , 2023	31/F	Asymptomatic (incidental finding)	N/A	None	Retrorectal heterogeneous cystic mass located inferior to gravid uterus	7.2	None	None	Complete/open (sub-umbilical median laparotomy)	Symptom-free	(15)
Atiya <i>et al</i> , 2023	71/F	Increasing tailbone/pelvic pain	Several months	Prior mass (unknown detail) in childhood	Large multiloculated cyst with septal calcifications	12.4	None	Transformed into adenocarcinoma	Complete/open	Succumbed to disease	(4)

M, male; F, female; PMSH, past medical and surgical history; CT, computed tomography; MRI, magnetic resonance imaging; UTI, urinary tract infection; N/A, not available; PET-CECT, positron emission tomography-computed tomography.

origin. An MRI confirmed a multiloculated cystic lesion associated with a perianal fistula without discharge.

Johnson *et al* (11) described a 16-year-old girl with persistent pelvic pain who had undergone several surgeries for presumed recurrent perianal fistulas and abscesses. Upon re-evaluation, a presacral cystic mass was identified, leading to the diagnosis of a TGC. The authors of that study highlighted that TGCs should be considered in patients presenting with recurrent presacral abscesses or anal fistulas, as a misdiagnosis may result in delayed recognition and inappropriate management (11). Similarly, Sauer *et al* (16) reported the case of a 58-year-old woman initially treated for a presumed presacral abscess who developed recurrent perianal fistulas despite drainage procedures. Subsequent abdominoperineal resection revealed an adenocarcinoma arising from a TGC, emphasizing that persistent or recurrent fistulas may conceal benign or malignant cystic lesions (16). In a Korean series of 24 cases, 42% of TGCs were infected, with few presenting as perianal swelling or discharging sinuses (17). Consistently, Malutan *et al* (6) observed that up to half of TGCs become clinically apparent only after infection or fistula formation. These findings may explain the case in the present study, in which a perianal fistula developed without an abscess or discharge, and the definitive diagnosis could have been easily overlooked without appropriate diagnostic evaluation.

A histopathological examination remains the gold standard for confirming the diagnosis. Microscopically, TGCs are typically multilocular and filled with mucinous material. Their walls contain irregular bundles of smooth muscle and are lined by a mixture of epithelial cell types, including keratinizing or non-keratinizing squamous, columnar, ciliated and transitional epithelium. These features distinguish TGCs from other cystic lesions, such as epidermoid cysts or teratomas (7).

Complete surgical excision is the treatment of choice for both symptomatic and asymptomatic TGCs (3). The surgical approach depends mainly on the location and extent of the lesion. Lesions above the S3 vertebral level are typically approached anteriorly via a transabdominal route, while those below are managed through a posterior route, such as parasacral, transsacral, trans-coccygeal, or intersphincteric approaches. Combined abdominosacral approaches are reserved for extensive lesions (6,17). Each technique has distinct advantages: The anterior route offers direct visualization of pelvic structures, such as the ureters and iliac vessels, whereas the posterior approach provides easier access to distal lesions but carries a higher risk of nerve injury (6). Ensuring complete excision with negative margins is critical, as incomplete resection can lead to recurrence, infection, or delayed malignant transformation. Post-operative follow-up is recommended with periodic digital rectal examinations and imaging analyses, such as CT scans (3). In addition to open surgery, endoscopic removal has also been performed in some cases for small, well-defined lesions adjacent to the rectal wall with no major vessels in proximity, and acceptable outcomes have been achieved (5).

A multidisciplinary approach is essential to ensure an accurate diagnosis and optimal management. Inadequate pre- and peri-operative work-up may result in recurrence or a missed opportunity for cure (1). A previous case report assumed that recurrent lesions decades after incomplete excision may

sometimes be associated with malignant change (4). This highlights the necessity for complete primary excision and vigilant long-term follow-up (4). Among the cases reviewed herein, mortality occurred in 3 cases due to malignant transformation. All patients underwent surgical excision, with the open approach used most frequently (~76.9%). In 2 cases which were malignant, the excision was incomplete and ended in metastasis and mortality. Outcomes were excellent among patients with benign lesions, with no recurrence during follow-up, whereas those with malignant transformation had a poorer prognosis. The patient in the present case report underwent the en bloc excision of both the TGC and its associated fistula without any intra- or post-operative complications. Over 1-year follow-up, the patient remained asymptomatic and had no recurrence. The limitation of this report include the inability to retrieve the histopathological image from the initial ileo-colonoscopy biopsy.

In conclusion, TGCs, as a congenital anomaly, may present with non-specific symptoms and may be associated with or masked by other perianal conditions, such as fistulas, necessitating appropriate investigation to avoid misdiagnosis or inappropriate management.

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#### **Availability of data and materials**

The data generated in the present study may be requested from the corresponding author.

#### **Authors' contributions**

SL, OHG, KAA and SHS were involved in the design and conception of the study, in the literature review, and in managing the case. SMF was the radiologist who performed the radiological examination and prepared the related figures. RMA was the pathologist who performed the histopathological examination of the case and also critically revised the manuscript. MMA, HOA, KFHH and FHK were involved in the literature review, in the drafting of the manuscript, in critical revision, and in the design of the study. All authors have read and approved the final manuscript. SL and KAA confirm the authenticity of all the raw data.

#### **Ethics approval and consent to participate**

Written informed consent was obtained from the patient for participation in the present case report.

#### **Patient consent for publication**

Written informed consent was obtained from the patient for the publication of the present case report and any accompanying images.

## Competing interests

The authors declare that they have no competing interests.

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