CASE REPORT

TAILGUT CYST (RETRORECTAL CYSTIC HAMARTOMA): A CASE REPORT WITH RADIOLOGICAL-PATHOLOGICAL CORRELATION

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ABSTRACT

Tailgut cysts (retrorectal cystic hamartomas) are rare congenital lesions arising from remnants of the embryonic hindgut. They are typically located in the presacral space and often present with nonspecific symptoms or are incidentally discovered. We report a case of a 42-year-old female presenting with chronic perianal pain and constipation. Imaging, particularly MRI, revealed a well-defined multiloculated cystic lesion in the retrorectal space without solid enhancing components. Complete surgical excision was performed, and histopathology confirmed the diagnosis of a tailgut cyst. This case highlights the importance of imaging in diagnosis, differentiation from other retrorectal lesions, and guiding surgical management due to the potential risk of malignant transformation.

Keywords: Tailgut cyst, Retrorectal cystic hamartoma, Presacral mass, MRI pelvis, Congenital lesions

Introduction

The retrorectal or presacral space is an anatomically complex region containing embryologic remnants from multiple tissue planes, making it a potential site for a diverse range of congenital and acquired lesions.[1] Developmental cysts represent the most common benign presacral masses in adults and include tailgut cysts, epidermoid cysts, dermoid cysts, rectal duplication cysts, and anterior sacral meningoceles.[1,2]

Tailgut cysts (TGC), also known as retrorectal cystic hamartomas, these are a rare class of congenital developmental lesions, which originate from the remnants of the embryonic postanal gut, a structure that normally regresses during the eighth week of gestation.[3,4] While they are present from birth, they often remain dormant for decades, typically coming to clinical attention in middle-aged women, at a ratio of 3-4:1.[5]

As these cysts are located deep within the retrorectal (presacral) space, they are anatomically sequestered between the rectum and the sacrum, making them difficult to detect through routine physical examination alone. The clinical presentation of a TGC is very diverse; around 26-50% of the patients remain entirely asymptomatic,[2] with the overall incidence of 1 in 40,000 estimated.[6] The lesion is often discovered incidentally during pelvic imaging for unrelated issues. However, when symptoms do arise, they are often the result of the mass effect on adjacent pelvic organs, leading to chronic constipation, tenesmus, urinary issues, pilonidal cysts, abscesses or lower back pain.[2,6,7]

The diagnostic journey for these patients is frequently complicated by the nonspecific nature of their symptoms. Complications can further obscure the clinical picture, as these cysts are prone to recurrent infections and fistula formation, which may lead to unsuccessful repeated surgeries if the underlying cyst is not identified.[8,9]

Of greatest clinical concern is the potential for malignant transformation within the cyst wall. Although rare (14%), various cancers can arise from the epithelial lining; the

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most common are adenocarcinoma or neuroendocrine tumors.[5] Consequently, imaging plays a crucial role in the modern management of these lesions. MRI, in particular, is the investigation of choice,[7] providing the high-resolution soft-tissue contrast necessary to visualize internal septations and evaluate the cyst's relationship to the pelvic nerves and musculature. Surgical excision is the recommended treatment of TGC. The literature quotes various surgical approaches, including posterior, trans-abdominal, and combined approaches,[10] the posterior approach (Kraske's procedure) is particularly successful for the cyst excision.[11,12]

Case Presentation

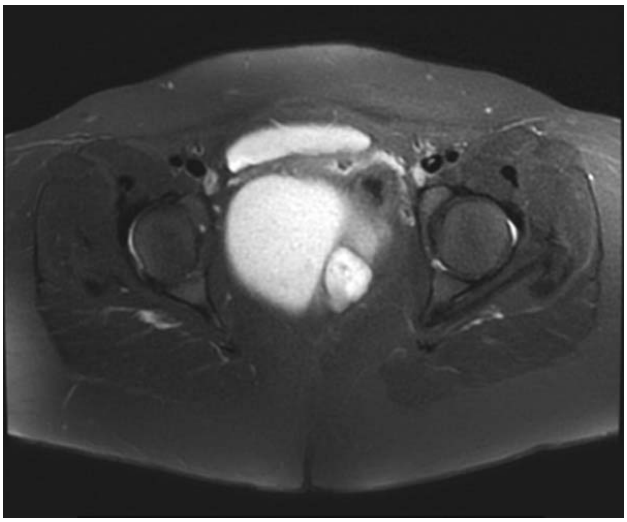
A 42-year-old female without any known comorbidities came with complaints of chronic perianal pain and constipation. She initially presented to general surgery, where her initial workup was done for abdominal pain, and no cause could be identified. An MRI pelvis with contrast study was carried out, which showed a multiloculated cyst in the presacral region.

She underwent a surgical procedure for the excision of that cyst through laparotomy, but the procedure was unsuccessful, and the patient was referred to neurosurgery. On presentation, she had no neurological deficit, having intact power in the lower extremities and intact sensations of sacral distribution. She also had intact bowel and bladder function.

MRI demonstrated a large predominantly cystic lesion occupying the presacral space and measuring approximately 12x11x12 cm in anteroposterior, transverse, and craniocaudal dimensions, respectively.

The lesion demonstrated multiple loculated cystic components along its inferior aspect. A focal low-signal-intensity component was identified along the anteriosuperior wall. Post-contrast images demonstrated enhancement of the cyst wall and internal loculated regions. Diffusion-weighted imaging revealed restricted diffusion within one of the inferior loculated cystic components. (Fig.1&2)

Based on MRI findings, the lesion was considered most suggestive of an epidermoid cyst because of focal diffusion restriction. However, the multiloculated morphology and retrorectal location also raised significant consideration for a tailgut cyst. After obtaining informed



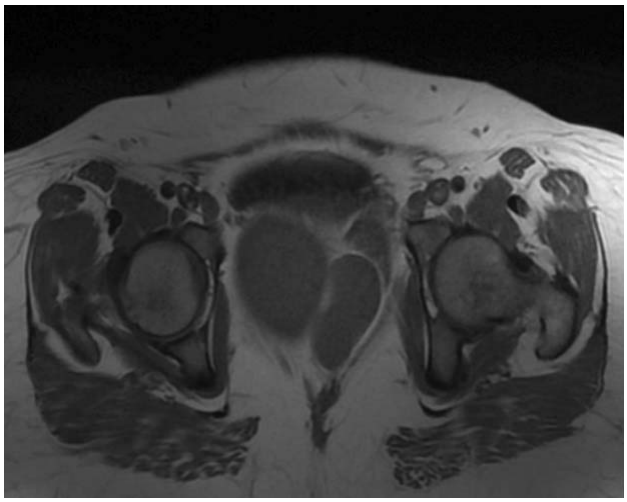
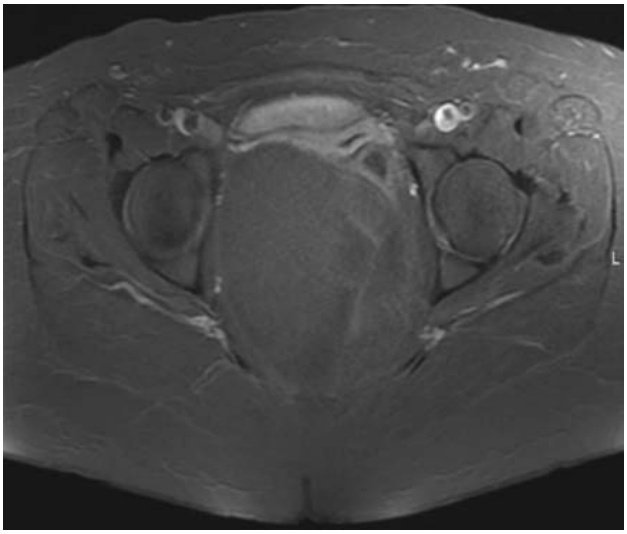


Figure 1: Axial T2 & T1 weighted MRI showing a hyperintense multiloculated cystic lesion in the retrorectal space with variable signal intensity within the lesion.



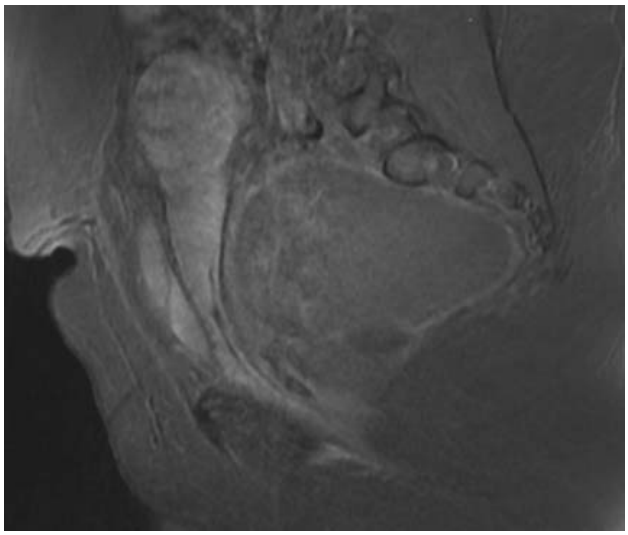


Figure 2: Post-contrast images showing absence of enhancing solid components.

consent, the patient underwent complete surgical excision of the cyst via a posterior approach under general anesthesia. The patient was placed prone on the operating table with full abdominal and pelvic support. A midline incision was performed over the coccyx, and the muscles were separated and retracted laterally to expose the coccyx bone completely. After coccygectomy was completed, the lesion was visualized clearly, a plane was created all around it and it was separated from the surrounding structures. The cyst was opened and partially decompressed. There was a thick, yellow colored fluid filled cyst in the presacral region, which was excised completely and sent for histopathology, there was no major blood loss encountered during the procedure.

Surrounding structures were inspected for any evidence of injury. After complete inspection, the wound was closed in layers. Patient was discharged on the first post-operative day, with instructions to use air ring to prevent wound dehiscence, and regular sitz bath for local hygiene. Biopsy revealed a multiloculated cyst lined by columnar, squamous, and transitional epithelium with no evidence of dysplasia or malignancy. The postoperative course was uneventful. (Fig.3)

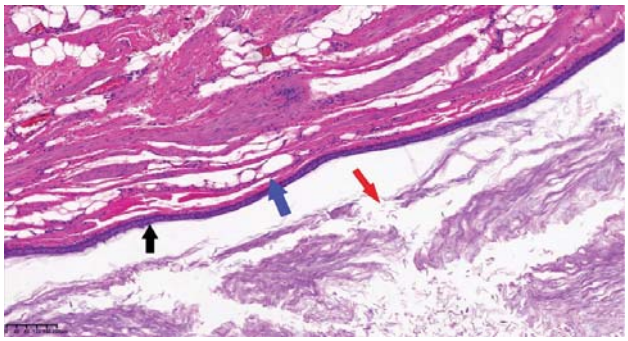


Figure 3: Magnification of 5.4x, showing stratified squamous epithelium along with keratin pearls and smooth muscle bundles Black arrow shows keratinized squamous epithelium, Blue arrow shows smooth muscle Red arrow shows keratin pearls

Discussion

Presacral lesions remain diagnostically challenging because of their rarity and the broad spectrum of pathologies that may arise within the retrorectal space.[1,13] Developmental cysts account for a substantial proportion of benign presacral masses, with tailgut cysts and epidermoid cysts among the most frequently encountered entities.[2]

The clinical profile of a tailgut cyst is characterized by its ambiguity. Because of their deep pelvic location, they often lack external physical markers.[14] Approximately half of all patients are asymptomatic, with the lesion discovered incidentally during routine gynecological exams or imaging for unrelated complaints.[5,14]

When symptoms occur, they are typically secondary to the mass effect. As the cyst expands, it may compress the rectum, leading to chronic constipation and tenesmus, pelvic or lower back pain due to pressure on the sacral nerves, urinary frequency if the cyst exerts pressure on the bladder.[15]

One of the most significant challenges in managing tailgut cysts is the high rate of misdiagnosis. Because they can become infected, they are frequently mistaken for more common perianal conditions, such as epidermoid cyst, pilonidal disease or recurrent perianal abscesses. Patients may undergo multiple unsuccessful drainage procedures before the underlying cystic hamartoma is identified as the true source of the pathology.[8,9]

Latif et al. present a similar case where a middle-aged lady presented with perianal pain, without other associated symptoms. The MRI showed a multiloculated cystic lesion in the presacral region. Upon reviewing the literature, it has become evident that most of the TGC cases present with lower abdominal or perianal pain without other associations like bleeding PR, urinary urgency or alterations in bowel habit.[16]

The lesion in this case exerted considerable mass effect upon the rectum, sigmoid colon, and uterus without causing bowel obstruction. Similar compressive symptoms have been described in large retrorectal developmental cysts because patients may remain asymptomatic until lesions attain substantial size.[17] Although overt malignant imaging features were absent, the possibility of malignant transformation remains clinically relevant in tailgut cysts, particularly in lesions demonstrating mural irregularity or enhancement.[7]

Given the complexity of the pelvic anatomy, Magnetic Resonance Imaging (MRI) has emerged as the gold standard for diagnostic evaluation. MRI provides superior soft-tissue contrast, allowing clinicians to distinguish between the multilocular nature of tailgut cysts and other presacral masses, such as chordomas or epidermoid cysts.[8]

On MRI, these lesions typically appear as well-circumscribed, multi-septated cystic masses. The presence of T1-weighted hyperintensity often suggests the presence of mucin or haemorrhage, while T2-weighted imaging provides a clear cyst's relationship to the pelvic floor muscles and the sacral bone. This anatomical clarity is indispensable for preoperative planning, as it dictates the surgical approach-whether transsacral, abdominal, or a combined procedure.

Kobayashi et al. argue that histopathological examination remains the gold standard for confirming the diagnosis. 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 keratinising or non-keratinising squamous, columnar, ciliated and transitional epithelium. These features are in line with our case, which shows keratinised squamous epithelium with abundant keratin flakes and focal mucinous epithelium.[18]

While tailgut cysts are benign, they harbour a silent danger: the risk of malignant transformation. The epithelial lining-which can be squamous, columnar, or transitional-can undergo neoplastic changes. The most common malignancies arising within these cysts are adenocarcinomas and neuroendocrine tumours (carcinoids). Because it is impossible to definitively rule out malignancy or prevent future infection through imaging alone (6). Surgical excision is the recommended treatment for both symptomatic and asymptomatic cysts.[15]

Total resection is the only way to prevent recurrence and eliminate the risk of cancer. Surgeons must be meticulous in removing the entire cyst wall; leaving even a small remnant can lead to a high rate of local recurrence and persistent symptomatic disease. With modern surgical techniques and early detection through MRI, the long-term prognosis for patients with tailgut cysts remains excellent.[2]

Conclusion

Tailgut cysts are rare but important differential considerations in presacral masses. MRI plays a pivotal role in diagnosis and surgical planning. Often these lesions are misdiagnosed and the patient often undergoes the unnecessary surgery.

CONFLICT OF INTEREST: No

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