

## Tailgut cyst adenocarcinoma

Pedro Martins<sup>a</sup> , Rita Canotilho<sup>a</sup> , Mariana Peyroteo<sup>a</sup> , Mariana Afonso<sup>b</sup> ,  
Augusto Moreira<sup>a</sup> , Abreu de Sousa<sup>a</sup> 

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### ABSTRACT

Tailgut cysts (TGCs) are rare congenital entities arising from remnants of the embryological postanal primitive gut. Malignancy in TGCs is rare, with the majority being adenocarcinomas and carcinoid tumors. A search of the published literature yielded only 27 cases of adenocarcinoma developing in TGCs. We described the case of a 54-year-old female who presented with complaints of pelvic and perineal pain of several weeks. After the initial work-up, a mass in the right presacral location was diagnosed. She underwent radical resection of the tumor, using a posterior approach. The lesion was removed en bloc with the middle rectum, coccyx, and sacrum (S4–S5). The histopathologic examination revealed an adenocarcinoma arising in a TGC, and the patient received adjuvant chemoradiotherapy. Our case underlines that diagnosing a TGC is difficult as it is a rare congenital lesion. Clinical examination may be challenging as TGCs present with various symptoms, which can mimic other commonly proctologic disorders. Patients should be referred to a tertiary center with experience in pelvic surgery and must be managed by a multidisciplinary approach to maximize successful treatment. The recommended treatment is surgical excision given the malignant potential of TGCs and their risk of causing local complications.

### Keywords

Cysts; Adenocarcinoma; Congenital Abnormalities; Pelvic Neoplasms

### INTRODUCTION

Tailgut cysts (TGCs) are rare congenital entities arising from remnants of the embryological postanal primitive gut. The majority of TGCs are benign lesions located in the retrorectal space. This space is defined anteriorly by the rectum, posteriorly by the sacrum, superiorly by the peritoneal reflection, inferiorly by the levator ani and coccygeus muscle, and laterally by the ureter and iliac vessels. Malignancy in TGCs is rare, with the majority being adenocarcinomas and carcinoid tumors. A search of the published literature yielded only 27 cases of adenocarcinoma developing in TGCs.<sup>1–22</sup> The reported cases were identified using the

electronic database search on PubMed (January 1970 to July 2018). The following free text terms were used: “tailgut cyst”, “retrorectal”, and “adenocarcinoma”. The reference lists of published studies were also reviewed to find additional cases.

### CASE REPORT

A 54-year-old female presented with complaints of pelvic and perineal pain of several weeks' duration. No history of urinary complaints or difficulties in

<sup>a</sup> Instituto Português de Oncologia do Porto Francisco Gentil, Entidade Pública Empresarial, Surgical Oncology Service. Porto, Portugal

<sup>b</sup> Instituto Português de Oncologia do Porto Francisco Gentil, Entidade Pública Empresarial, Anatomic Pathology Service. Porto, Portugal

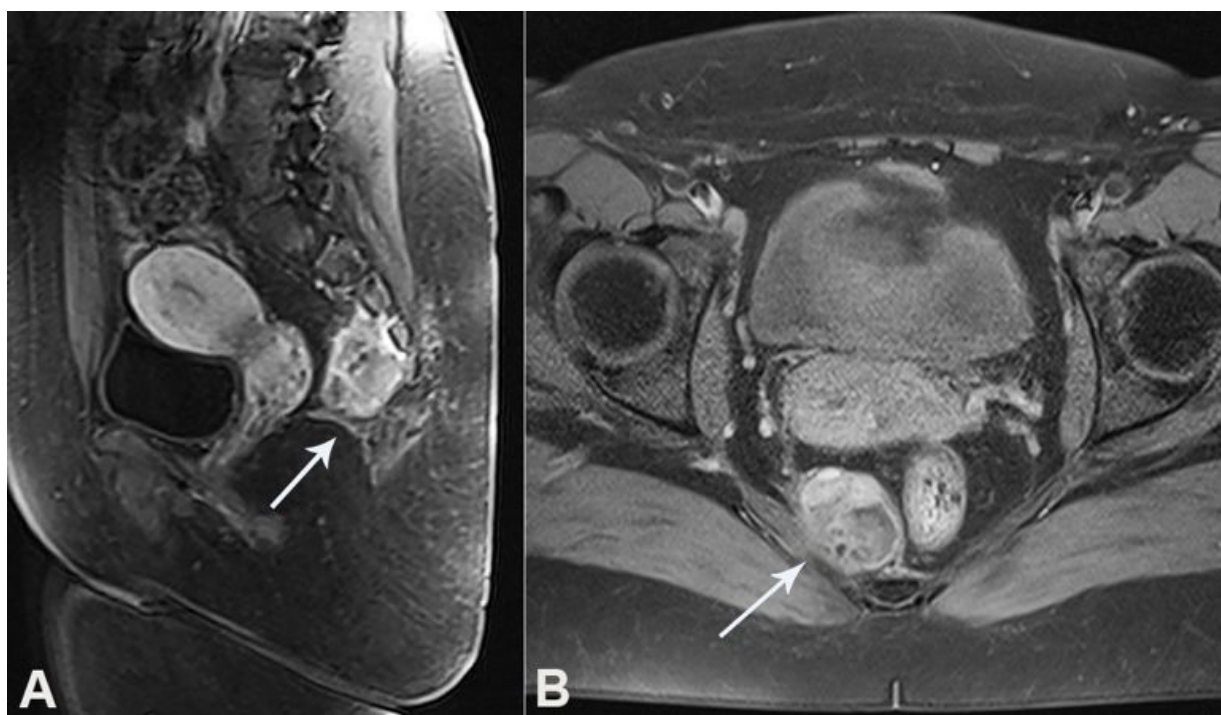


defecation were reported. On physical examination, there was no abnormality. Proctosigmoidoscopy revealed a bulging of the rectal wall in the middle rectum, 7 cm from the anal margin, with suprajacent normal mucosa. Further work-up included a pelvic magnetic resonance imaging (MRI), which revealed a mass in the right presacral space, with lobulated contours and soft tissue density (Figure 1).

The mass measured 5 × 3 × 3.5 cm (longitudinal, transverse, and antero-posterior axis, respectively) and exhibited a heterogeneous signal intensity. After administration of intravenous contrast, a heterogeneous enhancement was observed, which persisted in the late phase. The neoplasm had characteristics of aggressiveness, with infiltration of the adjacent sacrum. However, the rectal mucosa was found to be intact and the fat plane was preserved within the rectal ampulla. Computed tomography (CT)-guided biopsy (18G) revealed fibrous tissue of desmoplastic aspect, in which intestinal-like adenocarcinoma structures were identified. A staging CT scan did not show any evidence of distant metastases. The patient underwent en bloc resection of the tumor using a posterior approach (Kraske procedure). During surgery, we found a mass present in the retrorectal space. It was adherent to and not easily separated from the rectum and the perirectal fat. The mass was carefully dissected and removed

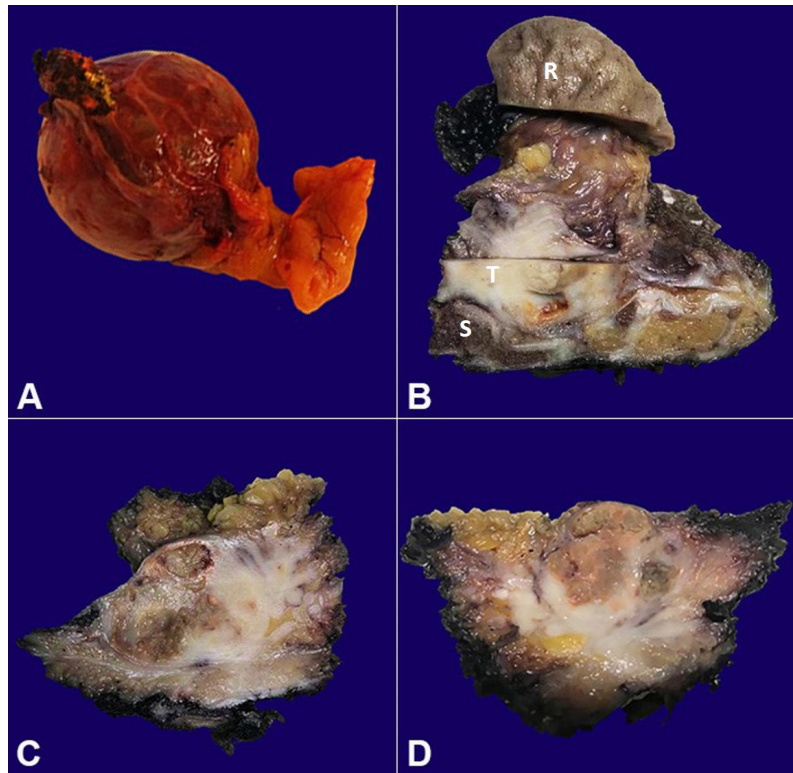
intact in a block with the middle rectum, coccyx, and sacrum to the level of S4. On gross examination, the resected specimen measured 8.8 cm × 7.5 cm × 8.5 cm, and included a 4.9 cm × 4 cm × 3 cm whitish and hardened neoplasia (Figure 2).

It contained a multiloculated cystic area, with brownish content. The histopathologic evaluation revealed the presence of a malignant neoplasm with a predominantly intestinal pattern of adenocarcinoma (Figure 3A and 3B). This neoplasm coexists with a multiloculated cystic lesion, covered by a columnar-type epithelium, focally sketching micropapillae with areas of low- and high-grade dysplasia (Figure 3D). It had an infiltrative growth pattern and invaded the adjacent soft tissues (skeletal muscle), and focally, the sacrum-but did not reach the rectal wall. It showed vascular and perineural invasion. The margins of resection were free of the carcinoma with exception to the proximal margin (upper pre-sacral soft tissue), which was focally involved. An immunohistochemical study showed diffuse positivity for CAM 5.2 and CDX2; multifocal positivity for CK20; and focal positivity for CK7 (Figure 4). Combined with clinical symptoms and imaging, a histopathologic diagnosis of adenocarcinoma arising in a TGC was established.

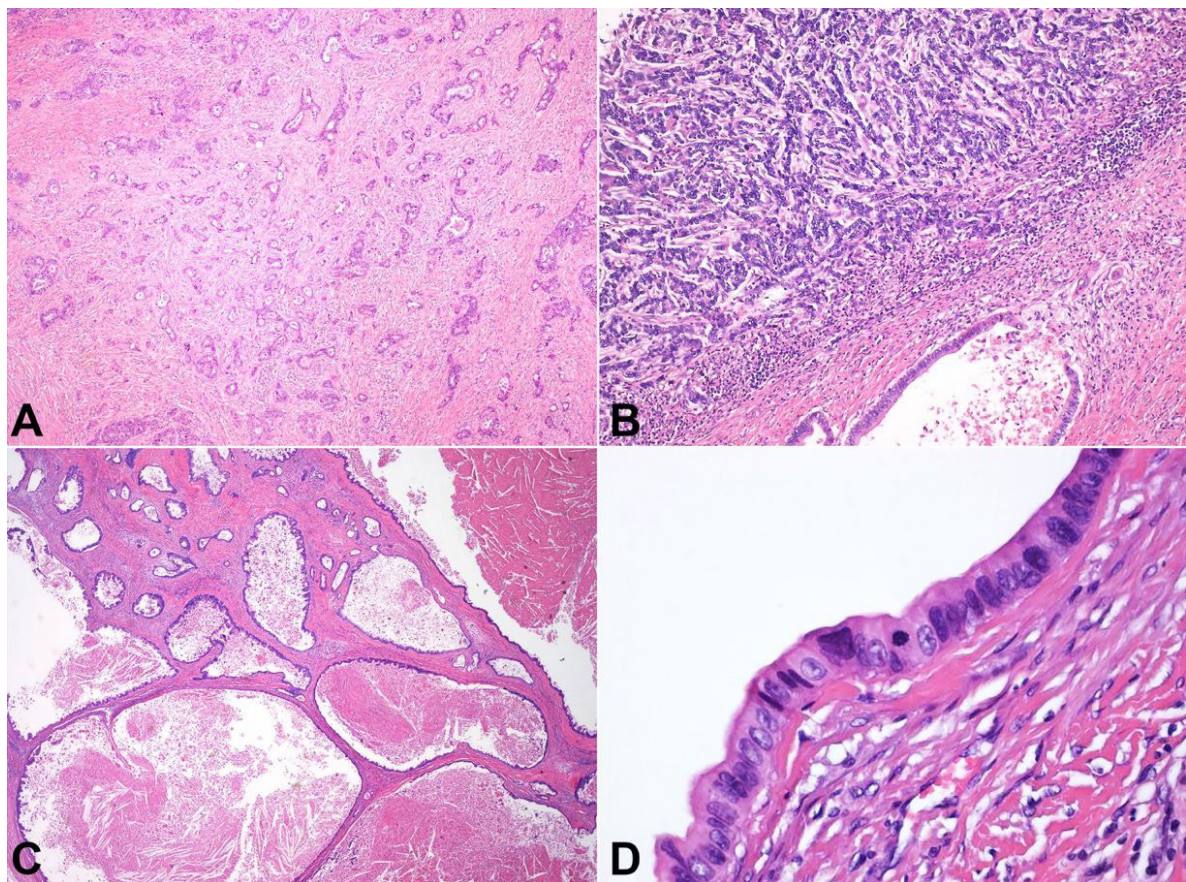


**Figure 1.** Sagittal (A) and axial (B) section of the pelvic MRI showing the tailgut cyst (arrows). MRI = magnetic resonance imaging.



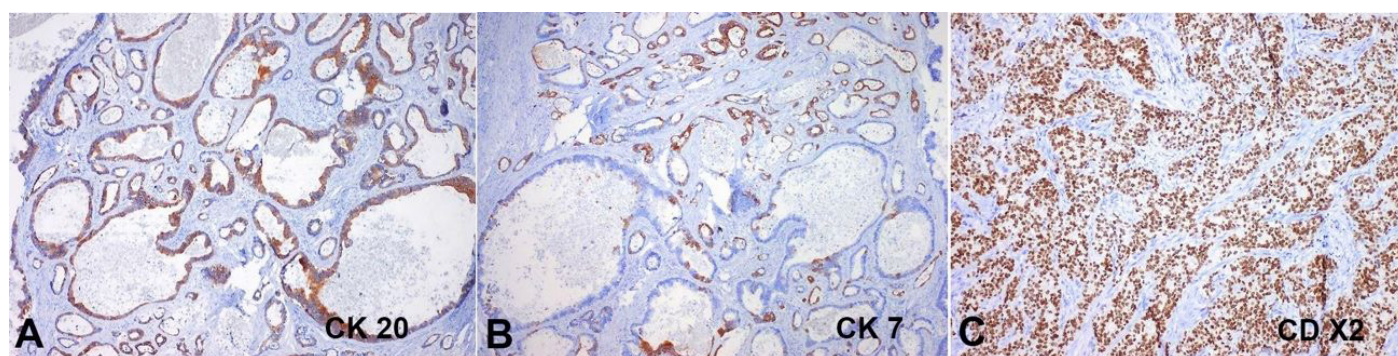


**Figure 2.** Specimen after surgical excision (A). Gross pathology of the resected specimen on cross sectioning showing the tumor and its relationships with adjacent tissues (B) R = Rectum; S = Sacral bone; T = Tumor. Macroscopic appearance of tumor within the tail gut cyst (C). Extensive infiltration of pre-sacral soft tissues (D).



**Figure 3.** Photomicrographs of the tumor showing the morphology of the adenocarcinoma arising within the tailgut cyst (A and B). Multiloculated, cystic areas (C). Cyst wall showing the uniform lining of the luminal surface by columnar type epithelium with areas of low- and high-grade dysplasia (D).





**Figure 4.** Photomicrographs of the tumor. Immunohistochemical staining: positivity of CK20 (A), CK7 (B), and CDX2 (C).

The patient's postoperative recovery was complicated with a wound infection, which was treated with antibiotics. After evaluation, the tumor board decided to advise her for adjuvant chemoradiotherapy. She received a dose of 54 Gy/30 fractions to the pelvis (including sacrum), according to the intensity-modulated radiotherapy technique for 6 weeks, associated with oral capecitabine. Since then, the patient has had regular visits to our department and remains without relapse, 11 months after surgery.

## DISCUSSION

The retrorectal (or presacral) space is an area of growth of multiple embryologic structures that involute during embryonic development. Consequently, this space can harbor a heterogeneous group of tumors (both benign and malignant) originating from the embryologic remnants.

Retrorectal tumors are rare and their incidence in the general population is unknown, generally because most cases are concentrated in tertiary centers. Hobson et al.<sup>23</sup> reported that a surgeon practicing outside the setting of a major referral center can expect to see, on average, at least one patient with a presacral tumor during the course of a typical career.

The classification system proposed by Dozois et al.<sup>24</sup> from the Mayo Clinic is the most commonly used. According to this classification, tumors are divided into five categories: congenital, neurogenic, osseous, inflammatory, or miscellaneous, which are further grouped into benign and malignant.

The most frequent lesions in children are teratomas, while in adults they are chordoma and

developmental cysts.<sup>3,25</sup> Retrorectal tumors are most commonly congenital and benign. Congenital tumors have a female predominance, while the other disease groups had a similar distribution between the two genders. However, malignant tumors occur more frequently in men.<sup>26</sup>

The tailgut is a post-cloacal extension of the embryonic gut that normally, by the eighth week of embryogenesis, atrophies and the tail involutes.<sup>16</sup> TGC is a rare congenital retrorectal lesion believed to derive from vestiges of the embryonic post-anal gut (tailgut) that fails to regress completely.

Malignancy in TGCs is a rare phenomenon. Malignant neoplasms that have been reported within TGCs include adenocarcinomas, carcinoid tumors, neuroendocrine carcinomas, endometrioid carcinomas, adenosquamous carcinomas, squamous cell carcinomas, and sarcomas. The first case of a TGC with malignant transformation was described by Ballantine in 1932;<sup>1</sup> since then, only 27 cases of adenocarcinoma developing in TGCs were described in the literature.<sup>1-22</sup>

Symptoms may be due to mass effects, causing lower abdominal and back pain, rectal bleeding, tenesmus, urinary frequency, rectal fullness, and constipation.<sup>10</sup> A careful rectal examination is essential to the diagnosis in more than 90% of patients.<sup>23</sup> Flexible sigmoidoscopy can determine the involvement of the rectal mucosa. Our patient had pelvic pain, a symptomatology that is most frequently associated with other gynecological and digestive disorders.

Contrast enhanced pelvic MRI is the gold standard of the imaging modalities currently available. MRI appears to have an advantage over

the CT scan because of superior soft-tissue contrast resolution, which provides improved delineation of the anatomic extent of the tumor and superior tissue characterization.<sup>27</sup> However, the accuracy of an MRI and a CT scan for a specific histologic retrorectal tumor type was only 28% and 18%, respectively, and some authors have advocated that it is not advisable to avoid resection based solely on non-invasive studies.<sup>28</sup> In this case, the MRI allowed us to decide the surgical approach and to be alert to the potential risk of sacrum involvement.

Biopsy of a retrorectal mass is controversial. On the one hand, given the limitations of imaging to make a definitive diagnosis, percutaneous biopsy of solid or heterogeneous presacral tumors could be obtained preoperatively to facilitate decision making for the use of neoadjuvant therapies and for optimizing surgical planning.<sup>29</sup> On the other hand, there is the fear that it can lead to contamination or tumor spread. Some authors only recommend a biopsy to be performed if the lesion appears to be unresectable and if a tissue diagnosis is required to guide neoadjuvant therapy.<sup>26</sup> In a comprehensive review, 27% of the patients underwent biopsy to confirm the diagnosis. Of these patients, incorrect diagnoses occurred in 44%.<sup>26</sup>

This patient had a biopsy before the evaluation in our center. In this case, we did not think that the biopsy was necessary since it was a clearly resectable lesion. The result of the biopsy would not change the future management and eventually could increase the risk of infection.

There is no consensus on management for TGC adenocarcinoma because of the very low incidence rate. Surgery is the mainstay of treatment. Early surgical excision is recommended for two reasons: (i) to enable a diagnose of malignancy; and (ii) to allow definitive treatment and elimination of the risk of complications.<sup>8</sup>

The common surgical approaches include the anterior transabdominal approach, the posterior approach (perineal, trans-sacral), and the combined approach. Low-lying lesions can be removed via the posterior approach. Tumors above the level of S3 can be resected via the anterior trans-abdominal or the combined approach. The rate of postoperative complications after resection of a retrorectal tumor is estimated to be 45%.<sup>30</sup>

A significant number of cases described in the literature had a poor prognosis due to local recurrence

and distant metastasis. This fact has favored the aggressive surgical treatment of TGCs. For the same reason, in some cases, adjuvant radiation therapy with or without chemotherapy has been employed with good outcomes.<sup>8,17</sup>

This case highlights the importance of TGC as a differential diagnosis of presacral masses, underscoring that malignant transformation can occur and may result in mortality and morbidity if surgery is not carried out.

Diagnosing TGC can be challenging because of the lack of familiarity with this entity due to its rarity. The clinical features do not follow a standard presentation and can mimic other commonly occurring proctologic disorders. MRI is indispensable in the evaluation of this lesion, showing the size, extension, and invasion of adjacent organs, which helps the clinician to choose the best surgical approach.

Patients should be referred to a tertiary center with experience in pelvic surgery and must be managed by a multidisciplinary team to maximize successful diagnosis and treatment.

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### **Correspondence**

Pedro Martins

Instituto Português de Oncologia do Porto FG - EPE - Serviço de Oncologia Cirúrgica

Rua Júlio Ramos, 4, 3F, Porto – Portugal

4200-072

Phone: +35 1914402724

pedrocm05@gmail.com