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Squamous cell carcinoma arising in a tailgut cyst: role of radiotherapy

Diego Aldave ,[1] Ana Teijo ,[2,3] Claudia Abril,[4] Laura Cerezo[1]

[1]Radiation Oncology, Hospital Universitario de la Princesa, Madrid, Spain
[2]Pathology, Hospital Universitario 12 de Octubre, Madrid, Spain
[3]Pathology, MD Anderson Madrid, Madrid, Spain
[4]Pathology, Hospital Virgen de la Salud, Toledo, Spain

Correspondence to
Dr Laura Cerezo;
lcerezo@salud.madrid.org

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SUMMARY

Tailgut cysts (TGCs) are rare tumours which can undergo malignant transformation. The gold standard of treatment is complete surgical excision. Multidisciplinary assessment is advisable in order to apply adjuvant treatment. Postoperative radiotherapy should be given in case of compromised surgical margins or other factors such as high histological grade or perineural and lymphovascular invasion. Here, we present a case of a squamous cell carcinoma arising from a retrorectal TGC treated with surgery and postoperative radiotherapy and review the main indications and techniques of this therapy.

BACKGROUND

Retrorectal cystic hamartomas or tailgut cysts (TGCs) are rare congenital lesions that arise from embryonic hindgut remnants when incomplete regression occurs during embryogenesis. Also, they have been related to meningothelial multiplication and benign thyroid tissue with oncocytic transition. TGC develops in the retrorectal space, defined anteriorly by the rectum, posteriorly by the lower sacrum, superiorly by the peritoneal reflection, inferiorly by the levator ani muscle and pelvic floor, and laterally by the iliac vessels and ureters.[1]

These lesions are uncommon and can present diagnostic challenges as symptoms and signs are not specific. The most common complaints include pain in the buttock/lower back and constipation.[2] As a result, there is a high misdiagnosis rate and delays.

They are more common in women, with a woman-to-man ratio of 3:1, and the average age at diagnosis is 35 years. Patients of male gender and older age are more likely to have a malignant tumour.[3]

Although the majority of TGC are benign lesions, the gold standard of treatment is complete surgical excision to avoid complications such as malignant transformation, infection or perianal fistula formation. When symptomatic, these lesions tend to be associated with malignant transformation.[4]

There are multiple approaches to resection of retrorectal tumours that are tailored to the clinical and anatomical considerations of each patient.[1 5–9]

The role of radiation therapy as an adjunct to surgery is not well known, as very few cases treated with radiotherapy have been reported in the literature; therefore, it is important to publish a new case and review the indications and technical aspects of this treatment.

CASE PRESENTATION

A woman presented with pelvic and coccygeal pain of 4 years duration, which had worsened in the past 6 months. Also, in the last weeks, she noticed a lump in the upper part of her gluteal fold. No history of urinary or defaecation difficulties was reported.

On physical examination, a soft and fluctuating subcutaneous lesion was palpated without inflammatory signs. The pelvic MRI revealed a bilobed lesion surrounding the second and third coccygeal bodies of 29 mm transverse × 49 mm anteroposterior × 50 mm longitudinal diameter, hyperintense in T1 and T2, with two solid hypercaptant poles in the interior cavity, one in the left inferolateral region, measuring 6 mm × 9 mm × 20 mm, and another in the upper pole, measuring 7 mm × 7 mm × 16 mm. The mass displaces the pelvic floor muscles superiorly, without spreading towards the intrapelvic region. The rest of the intrapelvic structures were within normal limits (figure 1). A staging CT scan did not show evidence of distant metastases. Preoperative CEA (Carcinoembrionary Antigen) and CA 19.9 (Carcinoembrionary Antigen 19.9) markers were within normal limits.

The patient underwent en bloc resection of the tumour using a posterior approach (razor position) with a sacrococcygeal spindle incision. During surgery, a 5 cm polylobulated cystic tumour was found, with the larger component posterior to the coccyx and a 2 cm precoccygeal lobulation encompassing the last coccygeal vertebra and extending superiorly to the second coccygeal vertebra. Both parts of the lesion communicate through the anococcygeal raphe. The lesion was removed from the pars sacrorectalis fibres of the levator, some of which were attached to the wall of the cyst and were sacrificed in order to avoid the opening of the cyst.

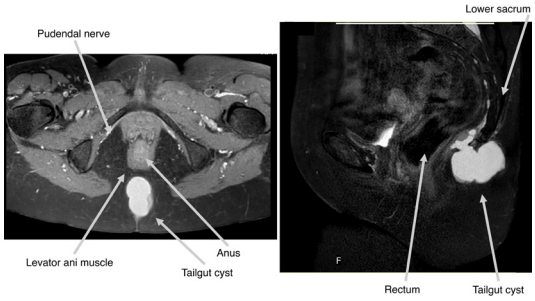

Figure 1 Sagittal and axial views of the diagnostic MRI showing the retrorectal cyst and their anatomical relations.

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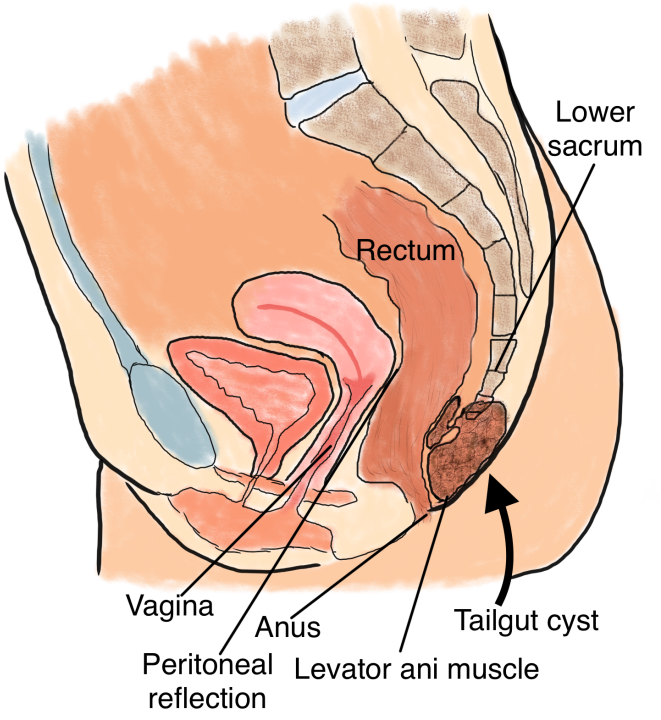

Figure 2 Diagram of retrorectal cyst and their anatomical relations. This figure was entirely illustrated by D Aldave, co-author of the article.

The last coccygeal vertebra was resected en bloc with the cyst to gain access to the anterior part of the cyst, located in the precoccygeal-presacral space. The resection was completed with an intact cyst. The integrity of the rectal wall was checked with methylene blue.

Total excision surgery is the gold standard, although this is not always possible since TGCs are sometimes close to important structures. These anatomical relations are shown in figure 2. Thus, the risk of the surgery would be to damage the posterior wall of the rectum, the pudendal nerves and the fibres of the levator ani muscle, but all these structures were well preserved in our case, as described above.

The patient recovered from surgery uneventfully, except for a small dehiscence in the intergluteal surgical wound, which finally healed with local cures at the outpatient clinic. The main complications of this surgery include: asthenia (immediate), dehiscence of surgical wound (early) and fibrosis (late).

INVESTIGATIONS

Gross appearance of the surgical specimen is shown in figure 3. Microscopic analysis (figure 4) showed a multiloculated cystic lesion lined with a squamous epithelium with a smooth muscle wall with disorganised bundles and in which a gigantocellular foreign body reaction appears with expumose histiocytes, acute and chronic inflammation and calcified areas. The tumour was located at 2 mm from the most proximal radial margin. Focally, coinciding with the most solid areas, high-grade intraepithelial dysplasia and solid nests are evidenced infiltrating the thickness of the cystic wall without reaching the fat or touching the surgical margin. These nests are made up of pleomorphic cells with extensive oeosinophilic cytoplasm and prominent nucleolus. Frequent figures of mitosis are evident, many of them atypical, with keratinising areas and necrosis. Occasional images of perineural invasion are identified, with no evidence of lymphovascular invasion. Surgical margins are free of dysplasia. Immunohistochemical study: p63+, CK5-6 and p53. p16 negative. Ki 67: 70%. In summary, these findings indicate the presence of squamous cell carcinoma, moderately differentiated on a retrorectal cystic hamartoma (TGC).

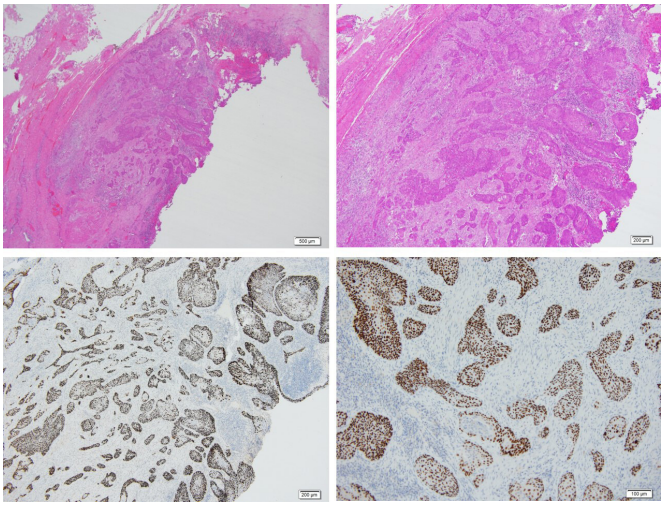

Figure 4 Microscopic study: H&E stain and immunohistochemical study (CK5-6 and p63).

DIFFERENTIAL DIAGNOSIS

Combined with clinical symptoms and imaging, a histopathological diagnosis of adenocarcinoma arising in a TGC was established.

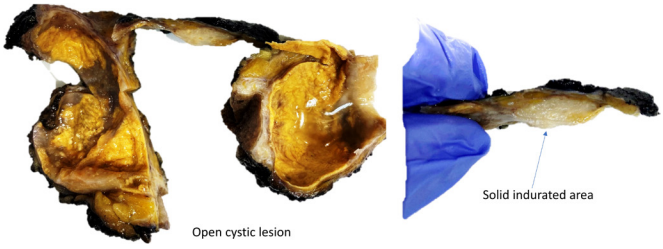

Figure 3 Gross appearance of the resection specimen: the lesion has been opened to show the cystic cavity and the solid indurated area in the wall.

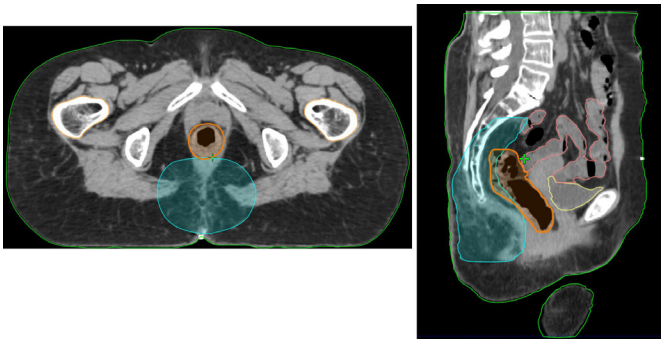

Figure 5 Planning target volume for radiotherapy in blue and organs to be spared: rectum (orange), bladder (yellow) and small intestine (brown).

TREATMENT

The case was discussed in the tumour board, and a final decision was made about giving postoperative radiotherapy. She was evaluated in our clinic for the adjuvant treatment.

In view of the histological findings of perineural invasion and elevated Ki 67 (70%) and the close surgical margin (2 mm in one point), we prescribed postoperative RT (Radiotherapy) to the surgical bed plus a safety margin in order to reduce the risk of local relapse. The target volume was designed on a planning CT, with an opaque marker placed on the intergluteal scar. The preoperative RM (Magnetic Resonance) diagnostic images and the description of the surgical findings were used to delimitate the target volume. The coccyx, the ischiorectal fossa and the presacral space until the level of the first sacral vertebra were included. Three-dimensional conformal radiation therapy (3DCRT) was used, with a posterior field and two lateral fields of 15 MV photons, with wedges, as shown in figure 5. The patient received a total dose of 50 Gy in 25 fractions, 5 days a week.

A cone beam CT was taken weekly, plus KV (Kilovoltage) X-rays daily to ensure accurate patient position setup, to improve the precision and accuracy of treatment delivery.

Tolerance of the treatment was good. The only complaint was increasing bowel movements of two or three times per day and slight asthenia during the last week of the treatment. Inspection of the skin showed a faint erythema in the intergluteal fold.

OUTCOME AND FOLLOW-UP

At last follow-up 12 months after treatment, the patient continues well, without evidence of disease recurrence or sequel from the treatments. The frequency of defaecation has changed from one time per day to two times per day.

DISCUSSION

TGCs are rare and relative unknown tumours that arise in the presacral space. The present case was the first TGC treated in our department in the last 30 years at least. Management of TGC is based on surgery, but radiotherapy or chemoradiotherapy (CRT) can be given to decrease the risk of recurrence in cases of malignant transformation.

Although it is not always possible, an early diagnosis is desirable for an adequate management of these cysts. CT and MRI tests are helpful. In MRI, TGCs usually have low signal intensity on T1-weighted images and high signal intensity on T2-weighted images.[10] MRI is more sensitive than CT for differentiation of unilocular from multilocular masses. CT and MRI can help to detect malignant transformation areas as they show suspicious features such as irregular mass contours, contrast enhancement within the cyst or nodular wall thickening as in our case.[11] TGC should be distinguished from other lesions which may occur in the retrorectal space including teratomas, epidermal cysts, anal gland cysts and chordomas.

Biopsy is not usually recommended due to the risk of infection and tumour spreading of the mucous contain into the pelvic cavity. Biopsy should be performed only in unresectable cases.[12] Surgical extirpation will provide an adequate specimen for definite histopathological diagnosis and will serve as definitive treatment in most cases.[13–15]

Malignant transformation of a TGC is not very common, ranging from 2% to 13% of the cases.[16] In a recent series of 52 retrorectal tumours, 56% were TGCs, and only four were malignant (Carpelan-Holmström 2020). Histologies of malignant transformation of TGC include adenocarcinoma, neuroendocrine carcinoma, endometrioid carcinoma, squamous carcinoma and sarcoma. Squamous cell carcinoma was reported previously in only two cases, so this would be the third case published in literature. One of the previously reported cases was synchronous with a rectal adenocarcinoma.[17]

It has been suggested that hormones, like ghrelin and oestrogen, might be important in the development of malignant transformation of TGCs.[8] In fact, TGC is more frequent in women than in men. However, the clear pathogenesis of the malignant transformation of TGC remains unknown.

Treatment of TGC is based on surgery guided by preoperative imaging. Limited reports exist on recurrence rates after TGC resection. Some reported a range of 0%–16%, usually related to incomplete excision or poor histological prognostic factors.[9]

The publications about the use of radiotherapy before or after surgical intervention are sparse. We carry out a search in PubMed with the keywords: 'retrorectal cystic hamartomas OR tailgut cysts AND malignant transformation AND radiotherapy'. Most of them were adenocarcinomas (44%) and in most cases surgery was the only treatment (86%). In only 6 of the 40 patients with malignant transformation and adjuvant treatment was administered, being CRT in 3, postoperative radiotherapy in 1 and chemotherapy in 2 patients (table 1).

In table 1, we summarise the published cases of malignant transformation TC, their histology and the treatment applied.

Table 1 Published cases of TC (Computed Tomography) treated with RT

Author, year	Histology	Indications for RT	Dose, fractionation and volume treatment
Baverez *et al*[18] 2021	Adenocarcinoma	Invasion of anal sphincters and perianal skin.	Neoadjuvant. 50 Gy in 25 fractions + oral capecitabine.
Wang *et al*[22] 2020	Adenocarcinoma	Uterus and rectum were compressed.	–
Tay and Azhar[19] 2020	Squamous cell carcinoma	Invasion of skin surface and bowel.	–
Martins *et al*[23] 2019	Adenocarcinoma	Vascular and perineural invasion. Proximal margin focally involved (upper presacral soft tissue).	Adjuvant. 54 Gy in 30 fractions to the pelvis (including sacrum), IMRT + oral capecitabine.
Nuno André Almeida Costa *et al*[11] 2018	Adenocarcinoma	Focal invasion of the removed sacrum (S4 and S5) and perineural and vascular permeation.	Adjuvant radiotherapy and chemotherapy.
Demirel *et al*[24] 2018	Squamous cell carcinoma	Invasion of rectum, bladder, right ischiorectal and ischioanal fossa.	Adjuvant. 66 Gy in 30 fractions to the operative and residual sites of the tumour, IMRT.
Mitsuyama *et al*[25] 2015	Neuroendocrine	Invasion of the lymph nodes.	Adjuvant. 59.4 Gy in 37 fractions to the presacral lymph nodes.
Jarboui *et al*[20] 2008	Adenocarcinoma	Focus of adenocarcinoma in the wall.	Adjuvant. 45 Gy in 25 fractions + intravenous 5-fluorouracil and folinic acid.

IMRT, intensity modulated radiation therapy.

The rationale for giving adjuvant radiation therapy is to reduce the local recurrence rate. As in other malignant tumours, the factors that can influence the incidence of the recurrence are the tumour histology, grade of the tumour, the proliferation index (Ki 67), the clearance of the surgical margins and the presence of lymphovascular and/or perineural invasion.

The volume, dose and technique are not well established since the experience with these tumours is limited. As TGC arise in the retrorectal space, the target volume should encompass the surgical bed, the ischiorectal fossa and the entire presacral space, as in figure 5. The elective irradiation of inguinal lymph nodes, as in anal cancer, is not clear but would be justified if positive lymph nodes are found. Conformal techniques, such as intensity modulated radiation therapy or 3DCRT should be used in order to spare the rectum, bladder and small intestine. Standard doses of 50–54 Gy at 2 Gy per fraction are normally given[11 18] if surgical margins are clear. A boost might be considered in case of residual disease.

Preoperative radiotherapy or CRT can be given if the tumour affects critical structures, such as the anal sphincter, similar to the approach used in locally advanced rectal adenocarcinomas.[18] Adjuvant chemotherapy can be used to decrease the incidence of distant metastases, as there are reports in literature of systemic dissemination.[19–21] The time of the diagnosis, the radical nature of surgical resection and a histopathological diagnosis are the most important prognostic factors.[21]

Learning points

- Retrorectal cystic hamartomas or tailgut cysts are rare congenital tumours which can undergo malignant transformation.
- The gold standard of treatment is complete surgical excision.
- Postoperative radiotherapy should be given in case of compromised surgical margins or other factors such as high histological grade, the proliferation index (Ki 67) or perineural/lymphovascular invasion in order to reduce the local recurrence rate.

Contributors DA performmed the literature search, made the table, has entirely illustrated figure 2 and wrote parts of the manuscript. AT performed the pathology study and made the pictures of the pathological specimen. CA helped with the manuscript redaction and did the interpretation of the pathological findings. LC treated the patient and supervised the elaboration of the case report.

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Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

ORCID iDs
Diego Aldave http://orcid.org/0000-0002-5956-3081
Ana Teijo http://orcid.org/0000-0002-8280-8446

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