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Mucinous adenocarcinoma arising from a tailgut cyst: a case report

Mucinous adenocarcinoma arising from a TC

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Abstract

BACKGROUND

Retrorectal hamartomas or tailgut cysts (TCs) are very rare entities. In most cases they are asymptomatic and benign in nature but rarely, they undergo malignant transformation, mainly in the form of adenocarcinoma.

CASE SUMMARY

A 55-year-old female patient presented with lower back pain. On Magnetic Resonance Imaging (MRI) a large pelvic mass was found, which was located on the right of the ischiorectal fossa and extending to the minor pelvis. The patient underwent extensive surgical resection of the lesion through the right buttock. Histologic examination established the diagnosis of a retrorectal mucinous adenocarcinoma originating from a tailgut cyst. Surgical resection of the tumor was complete and the patient recovered without any complication. A pilonidal sinus was also excised. One year later, semiannual PET-CT and MRI scans have not demonstrated any evidence of local recurrence or metastatic disease.

CONCLUSION

Preoperative recognition, histologic diagnosis and treatment of TCs pose a significant challenge. In addition, the possibility of developing invasive mucinous adenocarcinoma, although rare, should be considered.

Key Words: Key words: Retrorectal tumor; Mucinous adenocarcinoma; Tailgut cyst; Mucosal tumor; Pilonidal cyst; Case report

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Core Tip: Retrorectal hamartomas or tailgut cysts (TCs) are extremely rare entities. In certain cases, they undergo malignant transformation, predominantly in the form of adenocarcinoma. Mucinous adenocarcinomas are rare entities of carcinoma arising from TCs, with only 18 cases reported in literature, from to 1988 to 2021. Furthermore, the coexistence of a pilonidal tract is extremely rare, being the second reported case in the literature, to our knowledge. We present the case of a 55-year-old female patient with a large pelvic mass on the right of the ischiorectal fossa and a pilonidal cyst. Surgical resection of the tumor and the cyst, was complete and the patient recovered well.

INTRODUCTION

Retrorectal hamartomas or tailgut cysts (TCs) are very rare entities, with an incidence rate of approximately 1/40,000. (1) TCs are thought to be embryologically derived from a remnant of the posterior intestine. (2) Alternative terminologies have been used in the literature to describe these lesions, including "cyst of postanal intestine", "retrorectal cystic hamartoma", "tailgut vestiges", "myoepithelial hamartoma of the rectum" and "rectal cyst". (3) These tumors are thin-walled, multilayered structures and lined by a variety of glandular or transitional epithelium. (4)

TCs occur more often in middle-aged female patients, whereas they are rare in children. (5) Forty percent (40%) of TCs occurring in children and newborns are teratomas. Indeed, 10% of teratomas coexist with developmental disorders of the midline, such as encephalocele. (2,5) At this age group, tumors could be benign, while malignant tumors are more common in older children. (5) The majority of TCs in adults are benign; however malignant transformation has been reported in the literature, particularly in symptomatic cases. (6)

TCs mostly are asymptomatic prior to clinical recognition. Symptoms are often associated either with the growing tumor mass and may include lower abdominal pain, rectal tenesmus and constipation, or infectious complications, even in the form of fistulas (5).

Considering the rarity of this developmental anomaly, we present an interesting case of an invasive mucinous adenocarcinoma originating from a tailgut cyst associated to pilonidal cyst, that was managed in our department (Emergency Surgical Department of University Hospital).

CASE PRESENTATION

Chief complaints

A 55-year-old female patient presented to the Emergency Surgical Department of the University Hospital with lower back pain.

History of present illness

The patient complain for pain during the last 6 mo.

History of past illness

The patient had a history of ductal breast cancer, which was diagnosed ten years ago and treated with lobectomy and adjuvant therapy. She underwent also hip arthroplasty 1.5 years ago and was under no medication and in good physical condition and nutrition according to her age.

Personal and family history

No pathological conditions were found.

Physical examination

The level of arterial blood pressure was 130/85 mmHg, the temperature was 36,7°C and the oxygen saturation level was 98%. Physical examination revealed a large, palpable gluteal mass.

Laboratory examinations

In her admission to our department, routine laboratory tests and CEA, were within normal limits.

Imaging examinations

On Magnetic resonance imaging (MRI) of the pelvis (Figure 1a,1c) a pelvic mass was found, that was located to the right of the rectus fossa, in contact with the uterus and the rectum, which seems to puts pressure on the adjacent structures and possibly, on the sciatic nerve and extended to the minor pelvis. The dimensions of the mass were 11x10,6x16,2 cm and neural derivation was initially suspected.

Chest X-ray and Computed tomography (CT) scan showed no abnormal findings. Abdomen CT scan revealed a large, multifaceted formation, located on the right side of the rectum, between the urinary bladder and the coccyx, up to the fatty tissue of the buttocks, with enriched diaphragms. Appendix and ovaries were within normal.

FINAL DIAGNOSIS

The CT findings, raised the suspicion of tailgut cyst or cystic teratoma (Figure 1b,1d).

TREATMENT

The patient underwent extensive surgical resection of the lesion through the right buttock (Figure2).

An incision about 20cm long was made and sharp dissection was used to carry the incision down directly in the midline until the presacral fascia was encountered. The medial gluteal fibers were then divided bilaterally to expose the attached mass which was found to push the rectum and uterus away without infiltrating any of these structures. During dissection it was crucial to avoid injury of the rectal wall, vagina, sciatic nerve and urethra. Injury avoidance of the rectum was facilitated with the use of rectoscopy during surgery, together with the preoperative bowel preparation. A foley catheter was used as a guide for the urethra. The lesion resected and the gluteal muscles

were returned to the midline. The remaining layers of the incision were re-approximated and closed. At the same time a pilonidal sinus was found and removed.

Preoperative planning concerned proper positioning of the patient. A lithotomy positioning was preferred, because of the direct approach to the mass, the rectum, the vagina and the potential need for a combined transabdominal incision.

Concerns was also made about the contingent need of other specialties, such as gynecologist and urologist, if the lesion was found to infiltrate the vagina, or the urinary tract. On that ground, these specialties were stand by during operation.

The recuperation of the patient was uneventful, and she was discharged from the hospital on the 7th post-operative day, due to delayed bowel movement and patient's will.

Wound care was as usual and the removal of skin sutures was done two weeks after, without any complications.

Both the mass and the pilonidal sinus were sent separately for histopathologic examination. Grossly, upon sectioning, the large mass was found to be cystic and filled with mucohemorrhagic material. In a peripheral location, two smaller cystic spaces were identified, which were also filled by mucus and an amorphous material. Microscopic examination confirmed the presence of a cystic mass, which comprised thick fibrous bands that divided it into three cystic spaces, the largest of which corresponded to mucinous adenocarcinoma (Figure 3a). The neoplastic cells were medium to large in size, with roundish or irregular, hyperchromatic, atypical nuclei, that were surrounded by eosinophilic or pale cytoplasm (Figure 3b). Few "signet ring" cells were also apparent. Tumor cells were arranged in glandular or cribriform structures, trabeculae, variably sized solid groups, within large "lakes" of mucin. Rarely, isolated neoplastic cells in floating in the mucin were identified. A high number of mitoses was noted. Regions of tumor necrosis, as well as calcification were additionally observed. On immunohistochemical evaluation, the neoplastic cells exhibited the following immunophenotype: CK20+ (Figure 3c), CDX2+, CK7+ (Figure 4d), GATA3-, ER-, PR-, calretinin-.

The majority of the up to date similar published cases reported positivity of tail gut cyst or adenocarcinoma arising on the cyst to CK7 antibody, even partially.

Embryologically, rectum is the last part of the tail gut and both normal rectal mucosa and rectal adenocarcinomas present CK7 positivity in almost one fifth of the cases (7).

HER2 immunostaining showed faint, segmental, membranous positivity in a small number of tumor cells (HER2: 1+). The other two cystic spaces were lined with keratinizing squamous or pseudostratified ciliated columnar or metaplastic squamous epithelium (Figure 3e). The mass was circumscribed with bundles of connective tissue at the periphery and surgical margins were free of tumor. Based on these findings, the diagnosis of an invasive mucinous adenocarcinoma, possibly on the ground of a posterior rectal cyst sinus (tail gut cyst), was established.

Gross examination of the sacrococcygeal pilonidal cyst revealed an elliptical skin excision specimen. On the skin surface, a hole measuring 0.1 cm in greatest diameter was identified, which upon parallel sectioning, was found to be continuous with a sinus tract, which terminated in a brownish grey-colored area. On microscopic examination, the sinus tract was lined mainly by stratified squamous epithelium, and partially by granulation tissue. Hair shafts were also focally identified around the sinus tract (Figure 3f). The latter was extending to the deep tissue resection margin. No communication between the sinus tract and the tailgut cyst was found, albeit multiple sectioning.

OUTCOME AND FOLLOW-UP

The recuperation of the patient was uneventful, and she was discharged from the hospital on the 7th post-operative day. After the histopathology report, the oncology council recommended 22 sessions of radiotherapy, while the patient completed the treatment. Follow up of the patient, after 1 year, with semiannual PET-CT and MRI has not demonstrated any evidence of recurrent disease, locally or metastatic.

DISCUSSION

We present an interesting case of a mucinous adenocarcinoma arising on a TC. Mucinous adenocarcinoma is a rare type of carcinoma occurring on TC with only 18 cases reported in English literature, from to 1988 to 2021. Furthermore, the coexistence of a pilonidal tract is extremely rare, being the second reported case in the literature, to our knowledge. Connection between the pilonidal sinus and tail gut cyst was not be established either on imaging, intraoperatively or on pathology examination.

Primary retrorectal tumors include congenital tumors, which constitute 55–65% of all tumors in this region, neurogenic tumors (10–12%), osteogenic tumors (5–11%), inflammatory tumors (5%) and other types (5–11%). According to their embryonic origin cysts are classified into epidermal, dermal, neural, teratomas, enteric cysts, rectal duplication cysts, mucous secreting cysts, enterogenous cysts, simplex cysts, gland anal cysts, rectal cysts, hamartomas, and tailgut cysts.(5,8) TCs are found in the presacral space and they are typically thin-walled cysts, that may be single or multiloculated, branched, and may contain green opalescent colloid fluid. (2)

In 1885, Middeldorpf *et al* reported the first case of a cystic mass in the retrorectal space in a one-year-old girl, that was most likely a rectal duplication cyst. (9) Hjermstad and Helwig reported the largest series of TCs, which included 53 cases of patients with an age range of 4 days to 73 years and average age of 36 years. (10) Based on the up to date literature, TCs may be asymptomatic or present with non-specific symptoms due to the large size of the pelvic mass. (2,5,8) They can also lead to several complications, including neurogenic bladder, hemorrhage, fecal incontinence, fecal fistula, intestinal obstruction and infections (11,12) or malignant transformation, as happened in this presented case.

Diagnosis of TCs may be delayed due to the absence of typical symptoms. (6) Oftentimes, TCs are discovered incidentally through imaging tests performed during the investigation of other entities. (5) A CT scan typically shows a well-defined homogeneous retrorectal mass, of water to soft-tissue density (12). A more solid appearance could also be described due to the keratinous or inflammatory debris within a cyst. (5,3) Higher resolution scans may identify most TCs as multiloculated cysts. (3)

On T1-weighted images, MRI reveals a hypointense lesion, whereas they are homogeneously hyperintense on T2-weighted images. However, MRI is not the gold standard for discriminating benign from malignant lesions (3).

Definitive diagnosis is set on histopathologic examination. (14) Tail gut cysts are congenital lesions developed from residual posterior remnant of the intestine, which retains its structure and architecture regarding the mature ectodermal, endodermal and mesodermal tissue elements. The lining epithelium may vary, including squamous, ciliated columnar, pseudostratified, columnar, transitional, goblet columnar and cuboidal epithelium. (8,14) Additionally, it is characterized by the presence of a smooth muscle layer and connective tissue, which may be disarrayed and don't encompass nerve plexus or differentiated neuron cells. (14) The immunophenotype of the mucinous adenocarcinoma in this case was that described in previously similar reported cases (15), which is characterized by CK7, CK20 and CDX-2 positivity.

The majority of TCs are benign, nevertheless rare cases of malignancies, have been reported including this presented case. (6) Apart from adenocarcinomas, neuroendocrine carcinomas, endometrioid carcinomas, adenosquamous carcinomas, squamous cell carcinomas and sarcomas have also been described. (13) Although the option of needle biopsy seems attractive, it is not broadly recommended, due to the possibility for false-negative results, and the risk of tumor seeding. (16)

Once a presacral tumor is diagnosed, treatment of choice is extensive surgical removal, due to the possibility of malignant transformation. The surgical approach depends on tumor location. Regarding tumors extending below sacral spinal nerve 4 (S4), complete excision could be achieved with a posterior approach, which is effective at a rate 95%. For tumors that extend above S4, abdominal or abdominal-perineal approach is suggested. (16) If TC turns out to be malignant, many authors suggest that treatment could include adjuvant radiation therapy alone or in combination with chemotherapy. (6,17,18,19) Martins, Pedro *et al*, suggest both radiation and chemotherapy. (20) In a study by Liang, Feng *et al* it is argued that the mainstream treatment method for tailgut

cysts with adenocarcinoma is surgical resection followed by chemotherapy. (6) Baverez M *et al* suggest that neoadjuvant chemoradiotherapy similar to locally advanced rectal adenocarcinoma decreases the risk of postoperative recurrence. (21) Supplemental treatment can be administered as it is believed to contribute to the prevention of tumor recurrence without clear evidence so far that would actually improve the prognosis as there is no general consensus on treatment standard for TC-associated adenocarcinoma because of the very low incidence rate. (22) Factors that determine the prognosis include the stage at diagnosis time, tumor histology and grade and also, completeness of resection. (3) Adenocarcinomas arising from TCs have poorer prognosis than neuroendocrine tumors, (8) and carry a risk of local recurrence and metastases. Follow up of the patient is also recommended, including monitoring for signs of recurrence with periodic PET-CT scans, MRI, in addition to serum CEA levels, which serve as an indicator of the tumor's response to treatment, as well as recurrence. (19) Chhabra, Saurabh *et al* suggest that once a TC malignancy has been diagnosed and is associated with an elevated CEA, following CEA levels may be used as a simple measure to assess the tumor's response to treatment or development of recurrence. (3, 17) In our case, the patient did not have at any time elevated CEA levels, so this measure for monitoring after surgery, was rejected. Di Nuzzo, Maria Michela *et al* point out the use of combined MRI and endoscopy for postoperative follow-up. (23)

CONCLUSION

TCs represent a rare clinical and pathological entity. The additional rarity of this case lies not only in the presence of a mucinous adenocarcinoma arising from the TC, but also in the fact that it is the second reported case of association of the TC with a pilonidal cyst. In general, TCs constitutes both a diagnostic and treatment challenge. Imaging tests may be helpful but definitive diagnosis is usually set after complete surgical excision and histopathologic examination. Guidelines for the appropriate therapeutic management are required for TC-associated adenocarcinomas, although

timely and extensive surgical resection, along with adjuvant radiation therapy, with or without chemotherapy, have been used with good outcomes.

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