

# **A rare case of incidental mucinous adenocarcinoma with osseous metaplasia associated with cysts of the presacral space**

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Dear Editor,

We present a case of a young woman aged 47 years admitted to our institution for the presence of anal fissure and, for approximately 1 year, feeling of anal heaviness. On macroscopic examination, the resected structure appeared as a cystic neof ormation with interrupted walls due to surgical operations with a fringed external surface of 7 cm as the maximum diameter. When cut, the cyst had fibrotic consistency, with a gnarl on the edge due to the presence of calcium areas. The cutting surface appeared multilocular, with cystic spaces occupied by a thick mucus layer of gelatinous consistency. Histological examination revealed the presence of a malignant intestinal epithelium, with unquestionable invasive areas of poorly differentiated mucinous adenocarcinoma. The carcinoma was present in the cyst wall and had a smooth muscle structure. The leiomyuscular component present in the cyst wall lacked neuroganglionic structures. Some outbreaks of osseous metaplasia were also appreciable. Resection margins were free of neoplasm, so the striated muscle structure of the pelvic floor was resected with the cyst. Immunohistochemical analysis revealed a panel indicative of intestinal epithelial histogenesis with immunoreactivity to CK20 and CDX2 markers. We also found positive staining for CK7 but negative for TTF-1.

Congenital or acquired lesions of the retrorectal space, also called the presacral space, represent a rare type of tumors. The majority of retrorectal tumors are benign (1), and these lesions can be accidentally found in the course of investigations performed for other purposes because they are not often correlated with clinical signs. Pre-operative instrumental and laboratory tests provide useful information for choosing the surgical strategy. The suc-

cess of the treatment of these lesions depends on their histological characterization, and histologic diagnosis is obtained after surgery. Primary carcinomas of the presacral space are notoriously difficult to excise and can be slow growing. When retrorectal tumors are asymptomatic, the clinical diagnosis is delayed, and they may be misdiagnosed (2). Additionally, to avoid infections, fistulas, and malignant transformations, an early complete surgical resection is required.

Different classification schemes have been reported for retrorectal tumors, which are broadly categorized by Singer et al. (2). These lesions result from an abnormal closure of the ectodermal tube or sequestration of the developing hindgut. The former, named dermoid and epidermoid cysts, are always lined by squamous epithelium, with or without skin appendages. The latter, enterogenous cysts, are lined by columnar cuboidal or transitional epithelium. Most often, they are multilocular, and their thin walls are surrounded by smooth muscle fibers. Dermoid and epidermoid cysts are typically benign and diagnosed in females; they do not usually communicate with the rectum. Enterogenous cysts are endodermal in origin, arise from the hindgut, and are lined with columnar mucinous epithelium. Although they are usually benign, some cases of malignant transformation have been reported (3). Among them, there are the mucus-secreting cysts, also called tailgut cysts or cystic hamartomas, that are often lined by transitional, columnar mucinous epithelium and characterized by a multicystic aspect and high frequency in middle-aged women. The most important complications of these cysts are infections with secondary fistulization. Specifically, tailgut cysts do not have myenteric nerve structures in the smooth muscle component present in the wall. This represents an important criterion for a dif-

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ferential diagnosis in case of other cystic lesions of the presacral space.

Although malignant neoplasms of the presacral space represent a rarity, a large variety of histotypes has been described, including those of mesenchymal or neuroendocrine derivation. Thus, the importance of differential diagnosis for the treatment choice and follow-up is only guaranteed by histological examination. There is no defined role of pre-operative biopsy in the diagnosis of these rare lesions, rather the practice is not recommended to avoid dangerous complications that can definitively compromise the success of surgical resection. A complete excision without cyst rupture is required to reduce the risk of local recurrence (4). In our case, the procedure with abdominal access was necessary for extension to the top of the lesion. However, larger lesions are best treated with a combined abdominoperineal approach. In the case reached our observation, the histological diagnosis was easy for the expressed pattern and for the site. The conclusive diagnosis was of a poorly differentiated mucinous adenocarcinoma, intestinal type, arising in the tailgut cyst, with osseous metaplasia. The differential diagnosis was placed with a malignant transformation in a bronchogenic cyst with secondary ossification of the cartilaginous structures. However, when the cystic wall was examined in its entirety, it did not present residual cartilaginous structures.

Osseous metaplasia is a very rare condition in colorectal carcinomas and has been described by Dukes in 1939. It has an estimated incidence of <4% in rectal carcinomas. Bone-forming cells, protein matrix, cytokines, and several growth factors are thought to be involved in this process. Mucin, necrosis, and desmoplastic stroma are also considered to be relevant factors. Immunophenotypical characterization showing CK20 and CDX2 positivity confirmed intestinal histogenesis. The investigation for bronchogenic differentiation with antigen test for TTF1 gave a negative result. CK7 positivity has been extensively described in poorly differentiated rectal carcinomas and allowed the distinction from a metastasis of ovarian cancer due to gonad integrity in laparotomic exploration

and to the instrumental image examinations performed pre-operatively. Despite the breakage during surgical operations, the integrity of the peripheral margin is a prerequisite for a conservative treatment. The effectiveness of an adjuvant treatment is difficult to prove. Radiation therapy may have a role in the palliation of unresectable presacral tumors. Malignant tumors tend to locally recur despite the use of adjuvant radiation.

Recent reports have related the phenomenon of bone metaplasia with KRAS mutation but not with NRAS and BRAF mutations (5). This correlation could have important implications for the therapy. For this reason, a multidisciplinary approach is desirable for a successful treatment and for the reduction of local recurrences of such rare injuries.

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## REFERENCES

1. Glasgow SC, Birnbaum EH, Lowney JK, et al. Retrorectal tumors: a diagnostic and therapeutic challenge. *Dis Colon Rectum* 2005; 48: 1581-7. [\[CrossRef\]](#)
2. Singer MA, Cintron JR, Martz JE, Schoetz DJ, Abcarian H. Retrorectal cyst: a rare tumor frequently misdiagnosed. *J Am Coll Surg* 2003; 196: 880-6. [\[CrossRef\]](#)
3. Michael D, Cohen CR, Northover JM. Adenocarcinoma within a rectal duplication cyst: case report and literature review. *Ann R Coll Surg Engl* 1999; 81: 205-6.
4. Mathis KL, Dozois EJ, Grewal MS, Metzger P, Larson DW, Devine RM. Malignant risk and surgical outcomes of presacral tailgut cysts. *Br J Surg* 2010; 97: 575-9. [\[CrossRef\]](#)
5. Liu X, Xu J, Chen L. Colorectal carcinoma with osseous metaplasia. *Oncotarget* 2017; 8: 65407-13. [\[CrossRef\]](#)