DISCUSSION

TOPICS

* EPIDEMIOLOGY
* MALIGNANT TRANSFORMATION
* DIAGNOSIS WORK-UP
* HISTOLOGY
* TREATMENT MODALITIES AND PROGNOSIS

Epidemiology

Lesions in the retrorectal space are rare and affects a heterogeneous group, and the literature have focused predominantly on the surgical approach and in the histologic classification of those pathologies. The reports estimate the incidence of those lesions at 1 of 40.000 to 63.000 in hospital admissions (1, 2). Despite those numbers, the true prevalence of the retrorectal lesions remains unknown since it has misdiagnosis and asymptomatic cases. Particularly in tailgut cysts, like chronic perirectal and perianal symptoms is often unrecognized or underrated, or even misdiagnosed as duplication cysts and endometriotic cysts which maybe leads to a decreased of the incidence. Among retrorectal lesions, tailgut cysts is classified as congenital and arise from residual fragments of the embryonic hindgut and mainly located on the presacral space. This space is defined posteriorly by Waldeyer’s fascia, anteriorly by the fascia propria of the rectum, inferiorly by the coccygeus and elevator ani muscles, superiorly by the peritoneal reflection between the second and third sacral segments and laterally by the ureters and iliac vessels (3). Some cases have described other places than the presacral region, like a tailgut cyst located anterior to the rectum, perianally or in the perirenal region (4, 5, 6).

Tailgut cysts can be presented by any age but have a predominance in middle-aged woman. The prevalence with a female to male ratio is 3 : 1 to 9 : 1 (7, 8). In a single-center experience (South Korea) of 24 cases of tailgut cyst, a retrospective analysis was made between 2007-1018, and the median age of the patients was 51.5 years (range, 21-68 years) and 18 patients were females (75%), so the female to male ratio was 3 : 1 (9). The malignant transformation of tailgut cysts have different incidence depending on the study. In 1988, a large series in the literature reported that only 2% rate of malignant transformation among 53 cases, but more recent studies reported 13 to 40% rate (10, 11, 12). A systematic review that used 144 articles, including case reports and case series, reported that 32.1% of cases were malignant degeneration of tailgut cysts and symptomatic cases tend to be associates with this malignant transformation (13, 14).

Malignant Transformation

The first case ever described of a tailgut cyst with malignant transformation was publishes in 1932 by Ballantine (1). Malignant transformation of tailgut cysts includes neuroendocrine carcinoma, endometrioid carcinoma, adenocarcinoma, squamous carcinoma, and sarcoma. Among those, well-differentiated neuroendocrine tumors, adenocarcinoma, and squamous carcinoma are the most common (2). It´s interesting to consider that some cases report in the literature described the diagnosis of sacrococcygeal teratoma-associated carcinoids only based in CT and MRI imagens, so the presence of a tailgut cyst could not be totally excluded since the pathological examination was not made, indicating there may have been an underestimated recognition of tailgut cysts (3). The majority of presacral carcinoids is immunohistochemically investigated using antibodies against general neuroendocrine markers such as chromogranin A, synaptophysin, and NSE. The clear pathogenesis of the malignant transformation in tailgut cysts remains unknown, but a study published in 2010 proved that ghrelin and estrogen are important in the origination and development of the malignant transition of tailgut cysts (4). Before this study, neoplasms exclusively composed of ghrelin-producing cells and associated with high levels of circulating ghrelin only have been described in stomach and in the pancreas (5). So, the expression of ghrelin founded in the tailgut cyst that suffer malignant transformation as well-differentiated neuroendocrine tumors may be hypothetically explained by the possible activation of the ghrelin gene, since this hormone is not usually found in endocrine cells of hindgut derivation. However, this expression may suggest a foregut phenotype of the tumor as well, but it´s important to consider that hormones might also be responsible for the malignant transformation in tailgut cysts.

For the identification of a malignant transformation of tailgut cyst, a define diagnosis can only be achieved by surgical exploration and a histological examination, although CT as MRI are vital for identifying the tailgut cysts (6). The biopsy alone is not the best choice, because specimens obtained from the biopsy usually don´t have epithelial tissues or malignant foci, and another concern is the possibility of malignant cells leaking into the peritoneal cavity during the process of biopsy. In MRI, malignant cysts usually appear heterogeneous with solid and cyst content, and irregular borders, although have some cases in the literature that those characteristics also appear this way in beging tailgut cysts (7, 8). The presence pf calcium in tailgut cysts may help to identify malignant transformation, but it´s not easy to identify this characteristic on MRI. Adenocarcinoma is present as multilocular cystic lesions with well-demarcated hypodense areas with some soft-tissues density in CT imagens. On MRI, tailgut cysts as a neuroendocrine tumor appears to have an intensity to muscles on T1 images and medial signal on T2 images but have some cases in the literature that this type of malignant transformation produced high signals on T2 imagens (9, 10). In the literature, squamous carcinoma associated with tailgut cyst is not common, and one case the patient suffered bilateral deep vein thrombosis of the lower extremities, and when the physicians investigated the cause, a subcutaneous mass was identified over the sacrum and coccyx, revealing a squamous carcinoma associated with tailgut. So, MRI and CT characteristics for this type of malignant transformation needs further study (11).

Diagnostic work-up

For retrorectal cysts, different diagnosis should be considered, like dermoid, epidermoid cyst, rectal duplication cyst, neurogenetic cyst, teratoma, anterior sacral meningocele, leiomyosarcoma, cystic lymphangioma, pyogenic abscess, neurogenic cyst, sacral chordoma, and tailgut cyst. In the literature, some cases of patients that went under unnecessary operations before the correct diagnosis is reported, and a large series of 53 cases collected over 35 tears period by Hjermstad and Helwig found that 51 out of those 53 cases were not given the correct initial diagnosis of tailgut cysts (1).

The gold standard of the imaging modalities currently available is the pelvic MRI for detecting unicolar, multicolar and small peripheral cysts which typically demonstrates low signal intensity on T1-weighted images and high signal intensity on T2-weighted images (2). On CT, tailgut cysts present as thin walled, unilocular or multiocular, non-enhancing lesions, low density in the retro-rectal space, and this imaging modality helps as well see if the cysts involved the sacrum or adjacent structures. MRI appears to have an advantage over CT since the contrast resolution is superior for soft tissue, which provides a good delineation of the anatomic extent of the tumor and superior tissue characterization. So, this imaging type correlate slightly more with the final histology of the tailgut cyst than CT scan can result. With the image, the physicians estimate the extent of the lesion, the characterizing, the risk of malignancy and distinguishing organ-confined disease from tumor spread into adjacent structures, which is important to decide the surgical approach and to be aware for a sacrum involvement. For example, MRI will help to define the margins of resection of the cyst, and if the tumor is positioned below of S3, a perineal approach is considered, or if the tumor extends above S3, an abdominal or a combined approach is regarded (3). As said, it´s important to say that MRI and CT alone might lead to misdiagnosis of tailgut cysts with malignant transformation and have cases in the literature that a tailgut cyst was diagnosed as endometrioma (4). As it said, the final diagnosis remains histopathologic, but biopsy is not an advertised method and have controversies among physicians since it carries the risk of seeding tumor cell, but for cysts with mixed solid and cystic components, a percutaneous preoperative parasacral biopsy should be made to determine the surgical strategy and to know if postoperative adjuvant therapy is needed for the patient (5).

The clinical presentation of tailgut cysts is usually nonspecific, and up to 50% of the patients are asymptomatic, and the cysts probably are identified accidentally by MRI and CT (6). For the symptomatic cases, the symptoms are usually because of the mass effect, and they are constipation, obstructed defecation, lower abdominal pain, back pain, rectal pain, tenesmus, painless rectal bleeding, dysuria, urinary frequency, and even lower limb neurological problems. More rare complications are bleeding, infection, and malignant transformation, and since the broad spectrum of symptoms, the diagnosis of tailgut cysts is complicated (7).

Histology

Tailgut cyst is a congenital lesion located in the retrorectal space and have an embryologic origin. Those cysts are not diagnosed using immunohistochemistry, since the components are relatively simple, and the clinical significance is not important. They are predominantly multicystic, the smooth muscle fibers within the cyst wall are disorganized and do not contain a neural plexus (1).

Tailgut cysts that suffer malignant transformation as neuroendocrine tumors appears to have soft, gray-white, solid and cystic appearance, and histologically, the cyst consists of columnar epithelial cell with clear (2).

**I will do the discussion of this topic based on the clinical cases.**

Treatment modalities and prognosis

For retro-rectal tumors, the complete surgical excision of the mass is the best treatment, since the risk of recurrent, hemorrhage, chronic infection or malignant transformation exists. The most three most common surgical approaches are anterior (trans-abdominal), posterior (inter-sphincteric, trans-sphincteric parasacrococcygeal, trans-sacral, transsacrococcygeal, trans-anorectal and trans-vaginal) or a combined anterior and posterior approach (1). The choice for surgical approach depends on some factors, like the size and the extension of the cyst, presence of bleeding or infection, adherence to neighboring structures and presence of absence of malignant transformation. Some studies shows that the posterior approach is required when the tailgut cyst is below S3 or the sacral promontory, and the anterior approach is better for higher lesions above S3 or the sacral promontory (2). The management of presacral masses are difficult since the most common strategies have limitations since anterior approach have limited access to the caudal component of a mass and the posterior approach is complicated because of the vascular control (3). A study that analyzed the indices from the Mayo Clinic medical centers in all adults with at least 18 years old who needed the surgical extraction of tailgut cysts between 1985 and 2008 showed that complete removal of the cyst was achieved, where of 28 patients, 20 have the posterior approach, 9 anterior approach and 2 have the combined approach. 8 patients have postoperative complications, but the 30-day mortality rate was zero. Some long-term complications included delayed wound healing (2 patients), pelvic floor dysfunction (2 patients) and one case of sexual disfunction. As said, the most common surgical approach on the study was the posterior parasacral incision, which have the goal of the complete removal of the cyst, and the decision between the different techniques was based on the proximal extension of the cyst, the adherent surrounding structures and if it suffers malignant transformation and required “en bloc” resection. (4)

Cocccygectomy is controversial for benign congenital presacral cysts, and some past studies argue that the removal of the coccyx may decrease the possibility of the recurrence of the cyst since it can remain totipotent cellular remnants. The incision is usually made at the sacrococcygeal junction close to the coccyx, or if the mass is large, the procedure is associated with vascular isolation plus extraperitoneal iliac incision (5). However, more recent studies shows that surgeons preferred to maintain the coccyx unless the tailgut cyst suffered malignant transformation and is attached to the coccyx (6).

A case that involved transanal minimally invasive surgery, as an alternative to a posterior parasacrococcygeal approach, have some benefits like decreased risk for sacral neurologic injury, minimal discomfort for the patient and good visualization and exposure of the cephalad extent of the cyst, and in this case had no postoperative complications (7).

Limited repots exist on recurrence after the surgical extraction of the tailgut cyst, and the literature repost a range of 0%-16% for complete excision (8). A study made in a single center experience that involved 24 patients with a pathologically confirmed tailgut cyst in South Korea had no cases of recurrence after a median follow-up of 12 months (9). Another study that investigated all adult patients who underwent surgical excision of tailgut cyst from the Mayo Clinic between 1985 to 2008 followed 31 patients for more than one year to analyze the risk of recurrence. The 30-day mortality rate was zero, and recurrence of a begin cyst was detected in one patient after 5 years after an anterior resection (10). So, the chance of the recurrence of the cyst is rare, and patients are seen mostly annually for a digital rectal examination.

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