Retrorectal tumors: A challenge for the surgeons

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Abstract

Retrorectal or presacral tumors are rare lesions located in the presacral area and considered as being derived from multiple embryological remnants. These tumors are classified as congenital, neurogenic, osseous, inflammatory, or miscellaneous. The most common among these are congenital benign lesions that present with non-specific symptoms, such as lower back pain and change in bowel habit. Although congenital and developmental tumors occur in younger patients, the median age of presentation is reported to be 45 years. Magnetic resonance imaging plays a crucial role in treatment management through accurate diagnosis of the lesion, the evaluation of invasion to adjacent structures, and the decision of appropriate surgical approach. The usefulness of preoperative biopsy is still debated; currently, it is only indicated for solid or heterogeneous tumors if it will alter the treatment management. Surgical resection with clear margins is considered the optimal treatment; described approaches are transabdominal, perineal, combined abdominoperineal, and minimally invasive. Benign retrorectal tumors have favorable long-term outcomes with a low incidence of recurrence, whereas malignant tumors have a potential for distant organ metastasis in addition to local recurrence.

Key Words: Retrorectal tumors; Congenital cystic lesions; Teratomas; Perineal approach; Transabdominal approach, Combined abdominoperineal approach

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Retrorectal tumors are usually asymptomatic lesions (26%-50% of cases), being discovered incidentally on routine digital rectal examination. Symptoms such as sacral pain, constipation, incontinence, and pencil-thin stools usually indicate tumor invasion to adjacent structures [1]. Patients may present with lower back pain that worsens with sitting and is alleviated by walking and standing [4]. Patients who present with recurrent anal fistula and perirectal abscesses should be suspected of retrorectal tumor and subjected to additional imaging studies [9].

Retrorectal tumors are classified based on their origin, namely congenital, neurogenic, osseous, inflammatory, or miscellaneous. Moreover, these tumors can be divided according to the lesions’ histopathology, as benign congenital, malignant congenital, benign acquired, and malignant acquired (Table 1) [10,11].

**Congenital lesions**

The most common type of retrorectal tumor is congenital, of which two-thirds are cystic lesions, such as tail-gut, epidermoid and dermoid cysts [1,4,10]. The incidence of those developmental cysts tends to be higher in females. Although many of them are benign lesions, malignant transformation of tail-gut cysts has also been reported by tertiary centers [12,13]. Epidermoid and dermoid cysts can communicate with skin and present as postanal dimple or sinus, which can be easily misdiagnosed as pilonidal sinus or perirectal abscess (Figures 1 and 2) [14,15].

The risk of malignancy is higher for solid retrorectal tumors, the most common of which are the chordomas [16]. These slow-growing tumors arise from the fetal notochord’s vestiges, usually from within the vertebral bodies. Unlike developmental cysts, chordomas are more common in males. Patients with chordomas usually present with urinary or gas incontinence and intensive sacral or perineal pain due to invasion of the adjacent structures. Radical resection is usually required because of the
### Table 1 The classification of retrorectal tumors[10,11]

| Benign | Malignant |
|--------|-----------|
| Congenital | Developmental cysts (Tail-gut, epidermoid, dermoid, teratoma) | Chordoma |
| | Anterior sacral meningocele | Teratocarcinoma |
| | Adrenal rest tumor | |
| Neurogenic | Schwannoma | Neuroblastoma |
| | Neurofibroma | Malignant nerve sheath tumors |
| | Ganglioneuroma | Ganglioneuroblastoma Ependymoma |
| Osseous | Giant-cell tumor | Osteogenic sarcoma |
| | Osteoblastoma | Ewing sarcoma |
| | Aneurysmal bone cyst | Chondrosarcoma |
| | | Myeloma |
| Inflammatory | Abscess/hematoma | |
| Miscellaneous | Lipoma | Liposarcoma |
| | Fibroma | Fibrosarcoma |
| | Hemangioma | Hemangiopericytoma |
| | Endothelioma | Leiomyosarcoma |
| | Leiomyoma | Metastatic carcinoma |

**Figure 1** A patient presented with complaints of recurrent fistula, which was ultimately diagnosed as epidermoid cyst.

Relatively higher recurrence rates of this type of congenital lesion[17,18]. Teratomas are true neoplasms, that include all three germ layers. They can be solid or cystic, and often contain both components. They are also more common in females and associated with a 40%-50% risk of malignant degeneration in the adult population [19]. In the absence of malignancy, they rarely adhere to the rectum or other adjacent viscera[4].
Neurogenic lesions
Neurogenic tumors are the second most common retrorectal tumors, after congenital lesions. These are slow-growing tumors that typically arise from peripheral nerves, and 85% of them are benign, consisting of neurofibromas and schwannomas[20].

Osseous lesions
Osseous tumors account for 10% of retrorectal tumors and have a high risk of recurrence. These include benign tumors (i.e. osteoblastoma, giant-cell tumor) and malignant tumors (i.e. Ewing sarcoma, chondrosarcoma, osteogenic sarcoma), which arise from bone, cartilage, fibrous tissue, and marrow[4,21].

Inflammatory lesions
Inflammatory tumors are less common than congenital lesions and are considered secondary reactions to foreign substances left in the body from previous surgeries[22]. It has been reported that they can also result from an extension of infection from either the perirectal space or abdomen[2,6].

Miscellaneous lesions
Miscellaneous tumors account for 10%-25% of all retrorectal tumors, including lipoma, fibroma, hemangioma, leiomyoma, and liposarcoma[21]. These lesions can also be a metastasis from primary rectal cancer.

DIAGNOSIS
A careful rectal examination carries the utmost importance for making a diagnosis, accounting for diagnosis in 90% of cases. Unfortunately, unless the physician has a high index of suspicion, these soft and compressible lesions can easily be missed[4]. As such, magnetic resonance imaging (MRI) in conjunction with computed tomography (CT) has emerged as the diagnostic tool of choice (Figure 3)[5]. CT is useful for demonstrating the nature of the lesion (cystic-solid) and bone destruction, whereas MRI is more advanced in evaluating soft tissue and adjacent structures’ involvement (Figures 4 and 5)[23]. On MRI, based on the lesion’s internal signal characteristics, the lesion is diagnosed as a cystic tumor when it displays cystic elements comprising greater than 80% of the lesion and a solid tumor when the lesion shows solid elements in greater than 80%; the remainder are classified as heterogeneous[6]. Radiological features that indicate malignant lesions are heterogeneous signal intensity, irregular infiltrative margin, sacral destruction or remodeling, and enhancement[24]. MRI also enables the surgical care team to plan for extent of resection (local vs en bloc) and
Figure 3 Flow diagram for the management of retrorectal tumors. MRI: Magnetic resonance imaging; GIST: Gastrointestinal stromal tumor; TRUS: Transrectal ultrasonography.

Figure 4 Sagittal and axial magnetic resonance images showing a cystic teratoma localized in the retrorectal area.

surgical approach (anterior vs posterior vs combined) in a preoperative setting[22].

Other applicable imaging modalities are flexible sigmoidoscopy, transrectal ultrasonography (TRUS), and fistulograms. The flexible sigmoidoscopy is a newly established option to demonstrate rectal mucosa involvement or exclude a primary rectal cancer, whereas TRUS provides detailed information on the size, consistency of the tumor, and evidence of local invasion[2,5]. Fistulograms can be preferred in patients with a chronically draining sinus, to evaluate underlying pathology such as developmental cyst[21].

Preoperative biopsy has been controversial for retrorectal tumors, according to the potential risk of secondary infection and seeding of the tumor[1,8,21]. With the
advances in imaging modalities and improved neoadjuvant therapy options that have become available in recent years, it has become a feasible technique \cite{25}. On the other hand, a preoperative biopsy may lead to misdiagnosis, with a reported rate of incorrect diagnosis as high as 44\% \cite{8}. Nevertheless, studies have demonstrated preoperative biopsy to have better diagnostic accuracy in solid or heterogeneous tumors and to affect treatment management \cite{26, 27}.

Neoadjuvant chemotherapy is essential for some retrorectal tumors, such as Ewing sarcoma and osteogenic sarcoma, or metastatic chordoma, and tyrosine kinase inhibitors have been shown effective in progression-free survival \cite{28-30}.

In our clinical practice, if preoperative imaging modalities provide sufficient information regarding the nature of the lesion and if the treatment management will not change according to additional findings, we do not advocate performing a preoperative biopsy. It should be emphasized that if it is indicated, performing biopsies by an experienced radiologist and choosing the appropriate transperineal or parasacral approach have been recommended. However, transperitoneal, transretroperitoneal, transvaginal, and transrectal biopsies should be avoided, and the biopsy tract must be removed en bloc \cite{21}.

### SURGICAL APPROACH

The optimal management of retrorectal tumors is surgical resection, including of benign tumors, given the potential for developing symptoms and malignancy \cite{2, 31, 32}. The morphology of tumor determines the level of extension of surgery. Complete gross resection is recommended for benign tumors, whereas radical resection or en bloc resection of involved adjacent organs is required for malignant tumors \cite{21}. Surgical approaches include those from the anterior (transabdominal), the combined abdominoperineal, and the posterior (perineal). A general consideration is that an anterior or combined approach is preferred for tumors above the level of S3 and a posterior approach for lesions below the level of S3.

**Anterior (transabdominal) approach**

The anterior approach is recommended for tumors located above S3 or which show sign(s) of pelvic wall involvement in the preoperative investigation. If the tumor cells have invaded into adjacent structures or an en bloc resection for malignant lesions
Figure 6  Perineal approach via parasagittal incision in a patient with a tail-gut cyst.

Figure 7  Perineal approach via parasagittal incision in a patient with an epidermoid cyst.
(such as sacrectomy) is required, this approach is more feasible[33,34]. For such, the patient is placed in the modified Lloyd-Davis position. The dissection starts with the opening of the pelvic peritoneum and continues to the posterior of the rectum. After the anterior margins of the tumor are dissected from the mesorectum, it is separated from the presacral fascia. Since the arterial supply of the tumor can originate from the middle sacral artery, it is crucial to identify and ligate the tumor's vascular structures first[25,35].

**Posterior (perineal) approach**
The posterior approach is indicated for tumors below S3 without any involvement of the sacrum and other pelvic organs (Figures 6-8). The patient is placed in the prone jack-knife position, and a midline or parasagittal incision is performed. Excision or elevation of the coccyx can be necessary for better exposure to the retrorectal space [23]. The division of the levator muscles follows, to enable access into the retrorectal space. Abdominoperineal resection may be required in patients with malignant tumors, as part of the *en bloc* resection. It has been reported that the posterior approach is preferred over the combined abdominoperineal approach, due to its lower morbidity rate than the latter, which has the highest recurrence and complication rate of all approaches[8,30-38].

**Combined abdominoperineal approach**
The combined abdominoperineal approach is recommended for malignant lesions, invading adjacent structures and obscuring normal surgical planes. The patient is placed in a modified Lloyd-Davies position, in order to access both areas[23]. If an extended soft tissue resection is required to achieve clear surgical margins, simultaneous or staged pedicle or free flap transfers can be used to prevent chronic sinus formation and fistulation[39,40]. Permacol mesh can also be applied for the reconstruction of the pelvic wall.

**Minimally invasive surgery**
Although it has not been reported whether laparotomy or laparoscopy has better long-term results, it is known that the laparoscopic approach provides an enhanced visualization of pelvic structures and facilitates precise dissection of the tumor from adjacent structures[8]. The laparoscopic approach has been demonstrated as a safe and feasible technique for treating retrorectal tumors[41,42]. There have also been case series reporting that the robotic approach can be chosen for large tumors, offering the benefits of shorter operation time and shorter length of hospitalization compared to laparotomy[43,44].

Transanal endoscopic microsurgery (TEMS) is also newly being applied to retrorectal tumors; however, with this approach, following oncological principles for malignant tumors is difficult[45,46]. Thus, it is recommended that malignancy should be excluded before TEMS is performed[23].

**FOLLOW-UP AND SURVEILLANCE**
Long-term results depend on the type of tumor and the successful surgical resection with clear margins achieved in the first operation. Although many authors have reported that benign retrorectal tumors have 100% overall survival rates with no recurrences[2,11], the patients should be followed-up for potential risk of local recurrence. Benign local recurrences' have been shown to have a good prognosis, even after repeated resection[47]. In contrast, malignant tumors can metastasize to the liver, lung, and brain, which are all associated with significantly worse prognosis[48-50].

**CONCLUSION**
Retrorectal tumors are uncommon lesions occurring in the retrorectal space. The most common retrorectal tumors are congenital benign tumors. The diagnostic algorithm starts with suspicion by a physician who carries out a thorough physical examination. MRI is the chosen imaging modality, with or without CT and TRUS. The preoperative biopsy is highly recommended for solid or heterogeneous tumors, although it is contraindicated for pure cystic lesions. The posterior approach is the preferred surgical method for most retrorectal tumors, producing lower morbidity rates. A multidiscipli-
linary team is usually required, since these complex tumors have a potential risk of invading adjacent structures and necessitating an *en bloc* resection.

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