Retrorectal Cysts and Tumors: From Clinical Presentations and Imaging Techniques to Pathological Diagnoses and Management: A case series and literature review

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Abstract

Background:

Retrorectal cysts and tumors have a variety of etiologies due to embryonic origin. We aim to study clinical presentation, diagnosis, imaging techniques, benefits of each imaging modality, the outcome of histology, and surgical management.

Methods:

We carried out a retrospective case series analysis of patients diagnosed with retrorectal cyst or tumor presenting to a single tertiary-level hospital in Iran from September 2014 to March 2023.

Results:
A total of fourteen females and two males were diagnosed with retrorectal cysts and tumors during this period. Abdominal pain was the most frequent symptom among patients. Two patients had a pathological diagnosis of squamous cell carcinoma and poorly differentiation of carcinoma, two patients had a cloacal cyst, two patients had leiomyoma, two patients had an epidermal cyst, one patient had a tailgut cyst, two patients had high-grade sarcoma, two patients had paraganglioma, two patients had spindle cell carcinoma, one patient had fibrolipoma, and two patients had atypical lipomatous tumor. Moreover, the surgical management of each pathology was discussed.

Conclusion:
Imaging modalities, including MRI, CT scan, sonography, and colonoscopy, have been employed for diagnostic methods. Timely surgical management is required to prevent mass effects, recurrence, bleeding, infection, fistula, and malignant transformations.

Keywords: retrorectal cyst, retrorectal tumor, presacral space, tailgut cyst, surgery

Introduction
Retrorectal cysts are rare cysts in the retrorectal space (also known as presacral space) that are limited by the posterior wall of the rectum anteriorly, the sacrum and coccyx posteriorly, the peritoneal reflection superiorly (at the level of S2, S3), and the pelvic floor muscles inferiorly, and the iliac vessels and ureters laterally. Contrary to retrorectal tumors, which are frequently observed in pediatric populations, retrorectal cysts are more prevalent in adults. Retrorectal cysts are rare lesions with an estimated incidence of 2 to 6 patients annually in tertiary care centers (1), and it is essential to note that most patients fall within the age range of 30 to 60 years old. Women tend to experience the condition more frequently than men, with the likelihood of occurrence being three to five times higher among females (2). Since this space is embryologically derived from hindgut, neuroectoderm, and notochord, a variety of congenital lesions can emerge in this space, such as simple cysts and developmental cysts. Histopathologically, these cysts can transform into developmental cysts such as dermoid cysts, epidermoid cysts, tailgut cysts, neurenteric cysts, rectal duplication, cloacal cysts, and various types of benign or malignant masses (3).

The infrequency of these cysts in adults and the lack of specific clinical indications can lead to misdiagnoses such as fistulae in ano, perianal abscesses, and pilonidal cysts (4). To explore more effective diagnostic methods, imaging techniques like MRI, CT scan, ultrasound, and colonoscopy can detect and show the spatial boundaries of the cysts and masses and their detailed anatomy shape, size, cyst wall thickness, and contents, contrast enhancement, adjacency to the other contents of this space, and adhesions to the surrounding structures. Surgical excision is generally recommended for retrorectal cysts based on current literature. Different surgical approaches are available, including posterior, transabdominal, and combined approaches (5). The posterior approach, precisely the modified Kraske method, is successfully used for cyst excision (6). The transabdominal approach is applied when the cyst is suspected to be malignant, or is located in a higher-up position in the retrorectal space. Transanal excision is not routinely recommended due to the risk of malignant seeding, contamination, recurrence, and infection.

The present case series aims to identify the mode of retrorectal cyst, clinical presentation, diagnosis, imaging techniques, outcome of pathology and histology samples, and surgical intervention and management of all types of cysts.

Methods
This was a retrospective case series analysis of all patients diagnosed with retrorectal cyst or mass presenting to a single tertiary-level hospital in Tehran, Iran, from September 2014 to March 2023. Patient information was recorded from history, examination, imaging techniques, and a confirmed final diagnosis with histology. Hospital electronic records were evaluated to obtain specific clinical details for each patient. We evaluated patient demographics, clinical presentation, imaging techniques such as MRI, CT scan, and colonoscopy, pathological diagnosis, surgical interventions, and management of patients. Patient demographics, such as gender, age, and duration of symptoms were collected in the self-structured questionnaire.
Written informed consent was attained from each patient for their anonymized information to be published in this article. Ethics committee approval had been taken from the institution. Statistical analysis was done using the Statistical Package for Social Sciences (SPSS) version 27.0.1. Categorical values are presented as frequency and percentages, while continuous variables are analyzed via descriptive statistics presented as means and standard deviations (SDs).

Results

Sixteen patients were presented with cysts and masses in the retrorectal space during the study period. All patients were included in the case series. There were fourteen females and two males. The mean age of patients was 40.1 ± 11.6 years. The mean hospital stay was 8 ± 6 days. Nine patients had a diagnosis of retrorectal cysts, and seven patients had a diagnosis of retrorectal masses.

A total of 18 symptoms were reported. Abdominal pain was the most frequent symptom (n=6, frequency: 33.33%). Other common symptoms included pelvic pain (n=5, 27.7%) and feeling a mass (n=4, 22.2%) (Figure 1).

Seven of the retrorectal cysts were detected by MRI. Meanwhile, one of the cysts was detected by colonoscopy. Also, retrorectal tumors and pelvic masses were detected by MRI, CT scan, and transvaginal sonography. After surgical excision and sampling the obtained tissue, two patients had a pathologic diagnosis of carcinoma (squamous cell carcinoma and poorly differentiation of carcinoma), two patients with a pathologic diagnosis of the cloacal cyst, two patients with a pathologic diagnosis of leiomyoma, two patients with a pathologic diagnosis of epidermal cyst, one patient with tailgut cyst, two patients with a pathologic diagnosis of high-grade sarcoma, two patients with a pathologic diagnosis of paraganglioma, two patients with a pathologic diagnosis of spindle cell carcinoma, two patients with a pathologic diagnosis of fibrolipoma, and two patients with a pathologic diagnosis of atypical lipomatous tumor (Table 1) (Figure 2).

Discussion

In this study, 16 patients presented with anal or abdominal pain. A similar female preponderance was reported in the literature for retrorectal cysts and tumors. In a similar case series study, Ramalingam et al. observed 4 patients with retrorectal cysts and found out that all of the patients were female (1). In the present study, patients visited the hospital with chief complaints of abdominal pain, pelvic pain, or anal pain. According to the literature, the most common signs and symptoms are local mass effects, including difficulty in defecation, constipation, rectal fullness, dysuria, lower abdominal pain, discomfort or pain in the pelvic, perianal, sacral area, or lower back (7). Sacral bone defects and calcifications can be rarely associated with developmental cysts (3). The mean age of the sample size was found to be 40.1 ± 11.6 years. The age of patients with retrorectal cysts ranged from 16 to 77 years in the literature, with a mean age of diagnosis of 41, while in our study, the age range of the patients was narrower. In the present study, pathologic diagnosis of the retrorectal cysts demonstrates squamous cell carcinoma, cloacal cyst, leiomyoma, epidermal cyst, tailgut cyst, high-grade sarcoma, paraganglioma, spindle cell carcinoma, fibrolipoma, atypical lipomatous tumor.

A tailgut cyst, or retro-rectal cystic hamartoma, arises when the primitive gut does not involute by the sixth week of gestation. During a rectal exam, tailgut cysts may not be detected because of their asymptomatic essence, resulting in an incorrect diagnosis and a cause of their rarity. Tailgut cysts can be extremely rare in the prerenal and retrovesical regions, described in only four patients, putting pressure on nearby organs such as the rectum, bladder, and ureter, leading to constipation, tenesmus, dyschezia, and polyuria (7). Histologically, tailgut cysts are multilocular cystic masses lined by multiple epithelial cells with mucoid content. It is crucial to ensure complete surgical resection with negative margins to eliminate any chance of recurrence, bleeding, infection, mass effect, and malignant transformations (7). Infection in these cases can cause a perianal fistula or a pelvic abscess (2). Two surgical methods are applied for tailgut cysts, including open surgery and laparoscopic minimally invasive surgery. Patsouras et al. discussed that the laparoscopic method could lead to incomplete resection due to poor exposure of the anterior space of the sacrum (8). Open surgery was a more suitable approach than the laparoscopic method, which could be performed in 3 ways: through the sacrococcygeal region, the abdomen, and the abdomen and sacrococcygeal region (8). Due
to the more hidden sacrococcygeal incision, the better exposed surgical area than the other two methods, the sacrococcygeal region approach benefited the complete resection of the tailgut cysts (9). Kildušis et al. had chosen a trans-rectal access route for surgical excision of tailgut cyst due to the surgical radicality, low incidence of morbidity, and minimal invasiveness for the selected patient, and the patient was symptom-free with no evidence of recurrent or residual disease (10).

Epidermal inclusion cysts, also known as epidermal or epidermoid cysts, are one of the most common retrotrectal cysts that can appear due to the failure of ectoderm closure. Histologically, these cysts are lined by thin-walled stratified squamous epithelium containing characteristic desquamated keratin, cellular debris, cholesterol, and water, often considered benign, but rare cases of malignancy arising within the cyst have been reported. Malignant transformation of an inclusion cyst can be identified by irregular wall-thickening of the cyst on imaging (11). Approximately 1% have been shown to transform into malignancy (12). Epidermoid cysts in the rectovesical pouch and perineum can cause symptoms secondary to mass effect. Regarding the typical radiographic characteristics of an epidermal inclusion cyst on imaging, these cysts should not be biopsied because of the high risk of cyst infection (13). If the epidermal cyst anastomoses with the skin, a post-anal dimple, or sinus, it may be misdiagnosed as perianal fistula/sinus or Pilonidal sinus. CT or MRI is preoperatively essential to find the location of the cysts. With CT imaging, a unilocular, hypodense, thin-walled mass filled with fluid density can be detected. With contrast-enhanced MRI, minimal enhancement of the thin wall with fluid attenuation can be detected, differentiating them from simple cysts and lipomas (14). Since the epidermoid cysts tend to grow over time and get inflamed and infected, complete surgical resection should be done. Three approaches, an anterior, posterior (Kraske) with coccygectomy or anteroposterior, can be performed based on the tumor size and location. The current literature recommends the anteroposterior approach if there is a larger than 5 cm tumor, severe adhesion, or malignancy. An anterior transabdominal approach is recommended if the lower margin is not lower than the fourth sacral level (15). Sasaki et al. presented a patient that the epidermoid cyst was so firmly adhered to the anal sphincter muscle, and the cyst was resected via a posterior approach (16). Kesici et al. discussed a total mass excision with paracoccygeal incision due to the mass extension from the perianal to the retro-coccygeal region (17).

Malignant transformations in retrorectal epidermoid cysts and tailgut cysts are extremely rare despite their malignant potential and can generally form transitional cell carcinoma, adenocarcinoma, and carcinoid tumors. In the present series, squamous cell carcinoma has arisen from a retrorectal cyst. Aldave et al. Ohsawa et al. also discussed the arising of squamous cell carcinoma from a tailgut cyst and an epidermoid cyst, respectively (18, 19). Histologically, squamous cell carcinoma is the nest of malignant squamous epithelial cells with abundant eosinophilic cytoplasm arising from the epidermis extending into the dermis with keratin pearls. Li et al. discussed the arising of adenocarcinoma from tailgut cysts with the CEA and CA 19–9 within normal ranges (16).

Cloacal cysts may arise in the retrorectal space due to the failure of the embryonic anorectal and urogenital channels to divide during the sixth to seventh gestational weeks resulting in the fusion of the rectum, vagina, and urethra. With an estimated incidence of 1 per 50000 live births, this malformation is exclusively seen in females and can be misdiagnosed as extravaginal fistula (20). Warne et al. suggested that any female fetus with a poorly visualized bladder, a cystic lesion arising from the pelvis, and bilateral hydronephrosis cloacal anomalies should be examined precisely for detecting cloacal anomalies, including cloacal cysts (21). MRI can detect the cloacal cysts by defining the anatomy of the cloacal cyst (22). Wael et al. recommended that colostomy be performed first to avoid obstruction complications. Then, reconstructive repair surgery was recommended using the posterior sagittal approach (23).

Cystic rectal duplication, also known as enteric duplication cyst, is commonly found at the mesenteric border of the jejunum or ileum when the developing hindgut sequestrates because of diverticulum formation or caudal twinning. Histologically, these cysts can be lined by squamous, columnar, cuboidal, or transitional epithelium because they arise from the endoderm of the primitive gut. Two layers of muscles with myenteric nerve plexus can also be in the cyst’s wall (1). Considering the anatomical location of duplication cysts, they may cause bowel obstruction, intussusception, volvulus or hemorrhage, or perforation because of gastric
mucosa contents inside. On ultrasonography, round, oval, or tubular cystic structures are detected with defining gut features, including five hyperechoic and hypoechoic layers of mucosa (the echogenic innermost layer), muscularis mucosa, submucosa, muscularis propria, and serosa (the echogenic outermost layer) (24). In both patients with symptomatic and asymptomatic rectal cysts, surgical resection is usually recommended due to the potential for bleeding, perforation, obstruction, and malignant degeneration (7).

The differential diagnosis of a solid retrorectal mass is broad and includes tumors such as neurogenic tumors, miscellaneous, sarcoma, and atypical lipomatous tumors. Neurogenic tumors are the second most common retrorectal masses after developmental cysts. Neurogenic tumors are slow-growing tumors arising from peripheral nerves, including schwannoma, neurofibroma, ganglioneuroma, and paraganglioma (25). On histopathology, schwannomas are fascicles of spindle cells called Schwann cells with a surrounding capsule, while neurofibroma is another benign neurogenic tumor composed of neoplastic Schwann cells, which contains additional non-neoplastic components including fibroblasts, mast cells, perineurial-like cells, and residual axons. Sporadic cutaneous neurofibroma is the most common subtype. Neurofibroma is another benign tumor composed of neoplastic Schwann cells and non-neoplastic components, including fibroblasts, perineurial-like cells, mast cells, and residual axons. For diagnosing and evaluating schwannomas and neurofibroma, MRI can assist in differentiating them from other soft tissue tumors or masses, providing soft tissue resolution and contrast, allowing for detailed visualization of schwannomas and adhesions (26). Miscellaneous tumors include lipoma, fibroma, fibrolipoma, and leiomyoma. Histologically, fibrolipoma is a variant of lipoma characterized by fibrous components with lobules of adipose tissue. Leiomyoma, also known as uterine fibroid, are bundles or fascicles of spindled cells with eosinophilic and possibly fibrillary cytoplasm arising from uterine smooth muscle cells and fibroblasts and surrounded by a thin pseudocapsule of areolar tissue (27). Transabdominal or transvaginal ultrasound is often used as the initial imaging modality for diagnosing leiomyoma and monitoring the growth of fibroids. MRI is considered the most accurate imaging modality for characterizing and assessing leiomyoma, providing detailed imaging of fibroids such as the extent of vascularization or degeneration of the leiomyomas, their size, and location (22). CT scans are not typically the first choice for diagnosing leiomyoma, as they may not provide sufficient soft tissue resolution. However, calcifications may be more visible on CT scans than other diagnostic modalities due to the superior contrast differentiation in CT scanning (28). Hysteroscopy or Laparoscopy can also help visualize the fibroids when tissue sampling is needed (28). Three approaches, abdominal, transsacral, and combined abdominosacral, have been used for the complete resection of retrorectal tumors (29). Tumors not lower than S4 (high lesions) are resected transabdominally by an anterior approach. Lower lesions smaller than 8 cm in diameter are resected transsacrally by the posterior approach. When the upper border of the lesion is palpated on rectal examination, it can be resectable transsacrally. The combined abdominosacral approach is performed for larger lesions or lesions in an intermediate position (29).

Conclusion

Retrorectal cysts are rare cystic lesions in the retrorectal/presacral space. This space is embryologically derived from hindgut, neuroectoderm, and notochord; a variety of congenital lesions can emerge in this space, such as simple cysts and developmental cysts. The infrequency of retrorectal cysts in adults, along with non-specific signs and symptoms, can lead to misdiagnoses. Imaging modalities such as MRI, CT scan, sonography, and colonoscopy have been employed to explore alternative diagnostic methods. Therapeutic approaches using timely surgical management still face challenges for patients affected by retrorectal cysts. Therefore, further research on the aspects of retrorectal cysts should be done to prevent mass effect, recurrence, bleeding, infection, fistula, and malignant transformations.

Author Contribution

Dr. Mehdi Tavallaei provided help during the study design and concept as well as supervising the project. Gita Manzari Tavakoli contributed to the study search, data gathering, reviewing, and writing the introduction section of the manuscript. Ayda Manzari Tavakoli provided help during the study search, data gathering, reviewing, writing the method
and discussion sections of the manuscript, editing, the scientific evaluation and revising the manuscript. Mustafa Ramezanian contributed to the writing the result section of the manuscript, and the scientific evaluation.

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Conflict of Interest
None of the authors have identified a conflict of interest

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Availability of data and material
The data supporting this study’s findings are available on request from the corresponding author. The data are not publicly available due to privacy or ethical restrictions.

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Key Clinical Message
Retrorectal space is embryologically derived from hindgut, neuroectoderm, and notochord; a variety of congenital lesions such as simple cysts and developmental cysts can emerge in this space. Timely diagnosis with appropriate management of retrorectal cysts can prevent mass effect, recurrence, bleeding, infection, fistula, and malignant transformations.

References


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