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A 40-Year-Old Woman With an Incidental Congenital Retrorectal Epidermoid Cyst Presenting as an Asymptomatic Presacral Mass: A Rare Case Report

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Data Interpretation D
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Patient: Female, 26-year-old
Final Diagnosis: Retrorectal epidermoid cyst
Symptoms: Asymptomatic
Clinical Procedure: —
Specialty: Surgery

Objective: Rare disease

Background: Retrorectal epidermoid cysts are uncommon developmental lesions that may remain asymptomatic for prolonged periods and can be difficult to distinguish from other presacral masses on clinical grounds. Magnetic resonance imaging is particularly useful for defining the lesion and its relationship to the rectum and surrounding pelvic structures. Complete surgical excision provides definitive histopathological diagnosis and treatment. This report describes a 40-year-old woman with a gradually enlarging, incidentally detected retrorectal epidermoid cyst followed radiologically for 13 years.

Case Report: A 40-year-old woman had an incidentally detected cyst on routine ultrasound in 2012 and remained asymptomatic for more than a decade. A 13-year follow-up with serial imaging demonstrated gradual enlargement of the lesion from 20 mm to 70 mm, prompting surgical intervention. The patient subsequently underwent successful laparoscopic cyst excision. Histopathology demonstrated a cyst lined by stratified squamous epithelium with a prominent granular layer and lamellated keratin, consistent with an epidermal inclusion cyst. Estimated blood loss was 300 mL, and no transfusion was required. The patient was discharged on postoperative day 7 and remained asymptomatic at 3-month telephone follow-up.

Conclusions: A slowly enlarging asymptomatic retrorectal epidermoid cyst can remain clinically silent for many years. Progressive enlargement, increasing mass effect, diagnostic uncertainty, or concern for complications can justify definitive excision even in an asymptomatic patient. Careful preoperative imaging and meticulous dissection are important when a large cyst is closely related to the rectum and pelvic structures. Histopathological examination remains essential for definitive diagnosis.

Keywords: Case Reports • Laparoscopy • Rectum • Incidental Findings • Congenital Abnormalities
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Introduction

Retrorectal or presacral tumors arise in the retrorectal space, which is the potential space between the rectum and sacrum and is formed superiorly by the peritoneal reflection, anteriorly by the fascia propria, posteriorly by Waldeyer's fascia, inferiorly by the levator ani muscles, and laterally by the ureter and iliac vessels [1]. These tumors can be congenital or developmental, inflammatory, neurogenic, osseous, or miscellaneous on the basis of origin [2]. Retrorectal tumors have an incidence of 1 in 40 000 in hospital admissions. Female predominance is associated with congenital cysts (15: 1) and male predominance with chordomas (5: 1) [3,4]. Clinical presentation is usually nonspecific, with the majority of cases presenting with dull aching pain. These lesions pose a diagnostic challenge and high rate of misdiagnosis due to vague symptoms. Most symptoms can be attributed to compression or invasion of the pelvic viscera or nerves [5]. Most lesions are benign, but malignant neoplasms are not uncommon. Malignancy is more common in the paediatric population than in adults, and solid lesions are more likely to be malignant than are cystic lesions [6,7]. Malignant transformation risk in retrorectal tumors varies from 1% to 12%, and the probability is even rarer in epidermoid cysts, at 0.011% to 2.2% [8,9].

Congenital or developmental tumors form the most common type of retrorectal tumors, which are further classified into epidermoid cysts, dermoid cysts, and tailgut cysts. Epidermoid cysts are lined by stratified squamous epithelium and contain keratinous material, whereas dermoid cysts contain skin appendages [1,3]. Magnetic resonance imaging (MRI) is particularly valuable in evaluating presacral lesions because it defines the cystic or solid nature of a lesion, its relationship to the rectum and pelvic structures, and features that may raise concern for malignancy.

Considering the rare occurrence, there are no established guidelines to follow for management but complete excision is recommended to prevent malignant transformation [10]. The optimal operative route depends on lesion level, size, relationship to adjacent structures, and suspicion of malignancy.

This report describes the clinical course, imaging characteristics, operative management, and histopathological findings of a 40-year-old woman with an incidentally detected congenital retrorectal epidermoid cyst that was followed for 13 years. The present case is notable for the prolonged 13-year radiological history, progressive enlargement from 20 mm to approximately 70 to 80 mm, and successful laparoscopic excision despite the lesion's low pelvic location.

Case Report

Initial Presentation and Surveillance

A 40-year-old woman with hypothyroidism (on tab thyroxine 100 µg once daily) and a previous history of gestational diabetes mellitus, was found to have a pelvic cyst measuring 20 × 20 mm on routine ultrasound during pregnancy in 2012. She was asymptomatic, with no obstipation, tenesmus, weight loss, or pain. Serum carcinoembryonic antigen, carbohydrate antigen 19-9, and cancer antigen 125 were within normal limits. A serum squamous cell carcinoma antigen test was not done. Because she had no symptoms, she declined surgical intervention and remained under radiological surveillance.

Serial Imaging

MRI of the pelvis done in 2019 showed a well-marginated cystic lesion in the retro-rectal space measuring 52 × 50 × 40 mm, displacing the rectum anterolaterally to the right side. No lymphadenopathy was seen. The lesion demonstrated heterogeneous T2-weighted signal intensity and hyperintensity on diffusion-weighted imaging, with restricted diffusion. Internal debris, suggestive of red blood cells, was noted.

Repeat MRI of the pelvis in 2025 showed further enlargement to 74 × 70 × 64 mm. The lesion continued to displace the rectum toward the right side and caused mild compression of adjacent pelvic structures, as shown in **Figures 1 and 2**.

CT angiography of the abdomen showed a cystic lesion measuring 79 × 77 × 78 mm in size closely abutting the rectum pushing it toward the right side. It was also abutting the anal canal and vaginal wall anteriorly. No enhancing septa, mural nodules, or calcifications were identified. There was no evidence of sacral erosion (**Figure 3**).

Indication for Surgery

Although the patient remained asymptomatic, the lesion had increased substantially in size over serial imaging and was producing mass effect on adjacent pelvic structures. After multidisciplinary assessment and discussion with the patient, surgical excision was selected for definitive diagnosis and treatment.

Operative Findings and Procedure

The patient underwent laparoscopy surgery. The rectum was seen displaced to the right side. The distal sigmoid colon and recto-sigmoid junction were mobilized to facilitate access to the retrorectal space. The peritoneum was opened above the sacrum, and the plane between the rectal fascia and presacral fascia was developed. A cystic mass measuring 70 × 65 mm was

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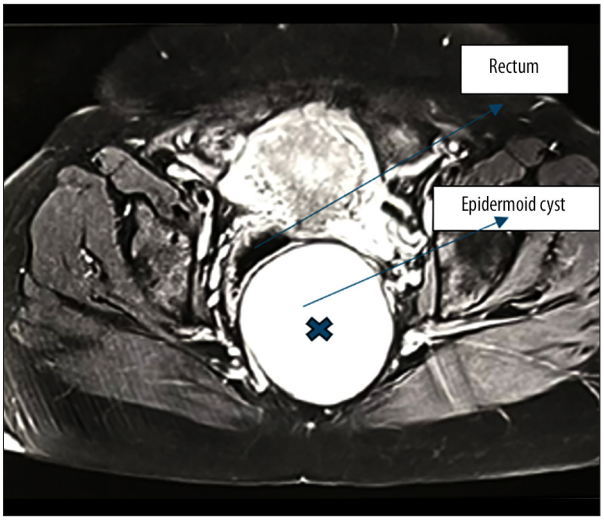


Figure 1. MRI Axial section showing hyperintense cyst on T2-weighted imaging with the rectum displaced anterolaterally.

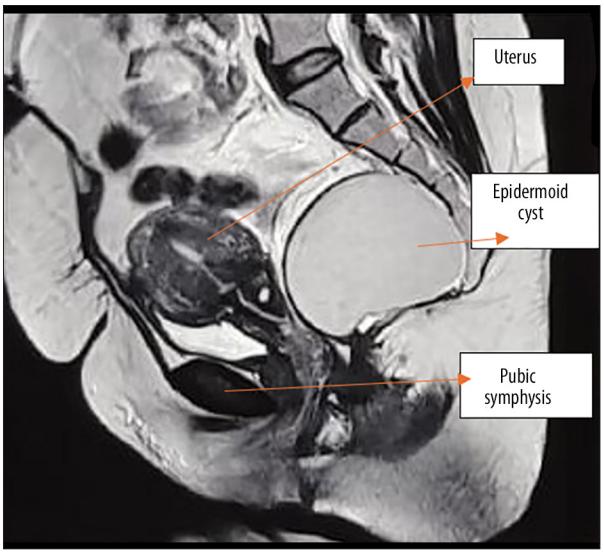


Figure 2. Saggital magnetic resonance imaging section showing level of cyst in rectouterine pouch with upper extent at S1-S2 level and its correlation with surrounding structures.

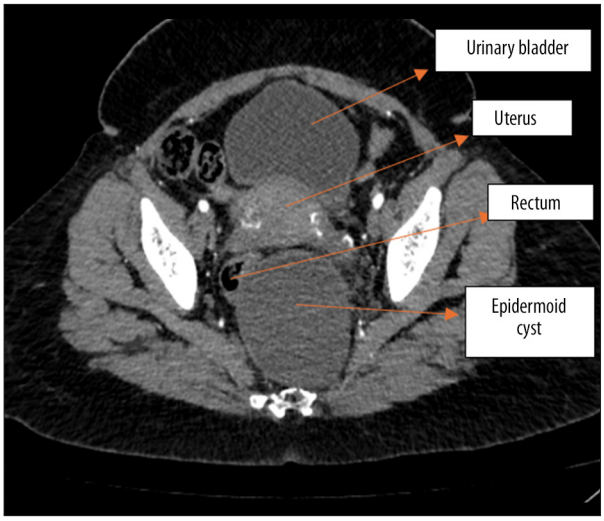


Figure 3. Computed tomography angiography axial image showing arterial phase showing hypo-enhancing cyst with no arterial supply. The uterus and urinary bladder are seen anteriorly as marked. The rectum is displaced anterolaterally towards right side.

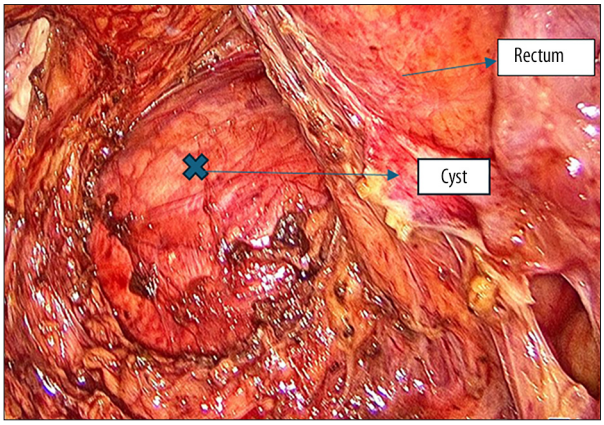


Figure 4. Intraoperative image showing the cyst as shown by cross in pelvis with surrounding adhesions. The plane between the rectal fascia and presacral fascia was developed. The rectum was pushed toward the right and anterioly.

noted, with its uppermost extent at approximately S1-S2 level. Dense adhesions were present between the cyst wall and the surrounding tissue, as shown in **Figure 4**. Careful circumferential dissection was performed to avoid injury to adjacent structures. Thick, cheesy material was aspirated, and the cyst wall was completely removed in an endobag (**Figures 5 and 6**).

Postoperative Course and Follow-Up

Intraoperative blood loss was 300 mL; no transfusion was required. The patient was monitored in the intensive care unit until postoperative day 1. The nasogastric tube was removed on postoperative day 1, and soft diet was allowed on postoperative day 3. Computed tomography with oral contrast performed on postoperative day 5 showed contrast reaching the rectum without evidence of an anastomotic or bowel leak. She was discharged on postoperative day 7 in stable condition. Postoperative follow-up was on postoperative day 14, during which staples were removed. At 3-month telephonic follow-up, she remained asymptomatic. No postoperative surveillance imaging had been performed since then.

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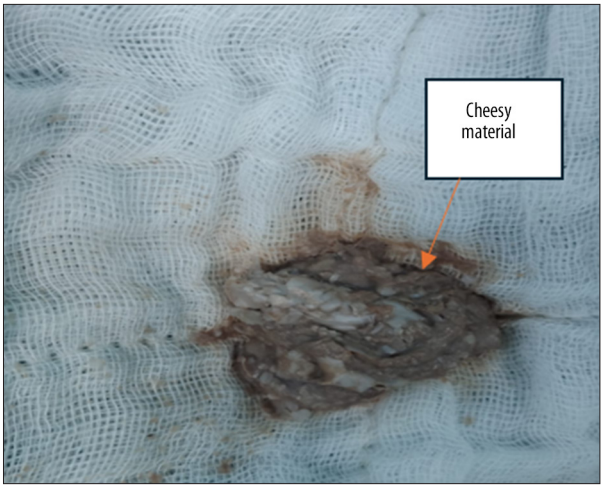


Figure 5. Caseous cheesy material aspirated from the cyst is placed on a sponge.



Figure 6. The cyst wall is extracted in an endobag placed on a green towel outside.

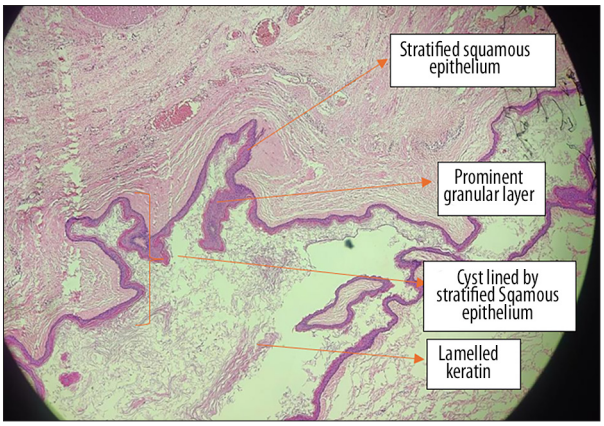


Figure 7. Histopathology image showing cyst lined by stratified squamous epithelium with a prominent granular layer and consisting of lamellated keratin.

Histopathology

Histopathological gross examination revealed a cystic tissue piece measuring 50 × 40 × 50 mm. Both the outer and inner surfaces were congested. Wall thickness ranged from 2 to 9 mm. Microscopic examination showed a cyst lined by stratified squamous epithelium, with a prominent granular layer, and consisting of lamellated keratin (**Figure 7**). No granulomas or fungus were seen. No evidence of malignancy was seen, and a diagnosis of epidermal cyst was made.

Discussion

The principal lesson from this case is that a retrorectal epidermoid cyst can remain completely asymptomatic for many years despite substantial enlargement. In this patient, a 20-mm lesion detected incidentally in 2012 increased to more than 70 mm on subsequent imaging and eventually produced displacement and mild compression of adjacent pelvic structures. The prolonged asymptomatic course illustrates why clinical symptoms alone may not reflect the size or potential future consequences of a presacral lesion.

Congenital or developmental retrorectal cysts are the most common type of rectrorectal tumors [11]. They are most commonly seen in female patients. Jao et al [3], in a study conducted between 1960 and 1979 on 120 patients with retrorectal tumors, found a female predominance (15: 1) in congenital cysts. A similar female predominance was seen in a study of 53 cases by Hjermstad et al, in which 7% were reported to be malignant. Congenital or developmental retrorectal cysts are further subdivided into 3 types, epidermoid cysts, dermoid cysts, and tailgut cysts, and may present incidentally or with nonspecific symptoms caused by mass effect or infection. Tailgut cysts are lined by variable cells: cuboidal, columnar, or transitional cells. Epidermoid cysts contain keratinous material and are characterized histologically by a stratified squamous epithelial lining without cutaneous appendages. Dermoid cysts can be differentiated from epidermoid cysts by the presence of skin appendages, such as hair follicles, sweat glands, or teeth [1,3,4]. The incidence of malignant transformation in epidermoid cyst is reported to be 0.011% to 2.2% [8,12] and the cause for malignant transformation is postulated to be due to the chronic inflammatory response [13]. Only 3 cases of malignant transformation of retrorectal epidermoid cysts have been reported, with squamous cell carcinoma seen in all [14-16].

Epidermoid cysts originate from remnants of ectodermal tissue [4]. Symptoms are mostly due to mass effect of tumor on surrounding structures such as the rectum and urinary bladder (constipation and increased frequency). Pain can occur in case of infection or malignant transformation. The source of

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infection is postulated to be due to trauma or malignant transformation. Based on the location these can be easily confused with gynecological pathology [4].

Abdominal X-ray can sometimes detect solid tumors compressing the sacrum [17]. Computed tomography (CT) can distinguish solid from cystic lesions and also invasion into the surrounding structures and vascular involvement. MRI is the preferred imaging modality for characterization and surgical planning of presacral lesions. Epidermoid cysts may show diffusion restriction because of their keratin content and appear hypointense or mildly hyperintense on T1-weighted imaging and hyperintense on T2-weighted imaging. Features such as a solid or enhancing mural component, irregular wall, nodularity, invasive behavior, or adjacent bone destruction should increase concern for malignant transformation and influence operative planning. MRI better delineates perineal anatomy, especially for assessing neural involvement. Needle biopsy, which can identify solid tumors with sensitivity of 96% and specificity of 100%, can also be used in cases of inconclusive results. Needle biopsy is not used in cystic lesions because of low negative predictive value.

The differential diagnosis of a retrorectal cystic mass includes developmental cysts (epidermoid, dermoid, or tailgut cysts), anterior meningocele, cystic degeneration within a neoplasm, and other pelvic cystic lesions. Imaging can narrow the differential, but histopathological examination is required for definitive classification. In particular, the presence of skin appendages favors a dermoid cyst, whereas an epidermoid cyst is lined by stratified squamous epithelium and contains keratin without skin appendages.

Management of an asymptomatic retrorectal cyst is individualized. Observation may be considered in selected patients with small, confidently characterized, asymptomatic lesions when the risks of intervention outweigh the expected benefit; however, there is no universally accepted surveillance interval for all retrorectal cysts. Because these lesions are rare and heterogeneous, published management is largely based on retrospective series and case reports. In the present patient, continued observation was initially chosen because she was asymptomatic, but progressive enlargement over serial imaging ultimately prompted definitive excision.

The decision to operate in an asymptomatic patient should be individualized. Practical triggers include documented progressive enlargement, development of symptoms, increasing mass effect on the rectum or adjacent organs, recurrent infection, diagnostic uncertainty, or imaging features suspicious for malignancy. The present case supports the value of serial imaging when surveillance is selected, while emphasizing that the surveillance interval should be individualized because evidence for a standardized schedule is limited.

Surgical excision followed by histopathological examination is the gold standard for diagnosis as well as treatment in all solid or cystic lesions to avoid infection, recurrence, or malignant transformation. Preoperative planning regarding the choice of approach plays a crucial role. Choice of approach (abdominal, perineal, transrectal, or transvaginal) depends upon the location of tumor. In a case series, Menteş et al described that if the tumor is above the level of S3, an abdominal approach is preferred, while in cases in which the tumor extends below S3, a perineal approach should be preferred. In case of very low-lying tumors, an intersphincteric posterior approach for sphincter function preservation can be attempted [17]. The abdominal approach is performed when there is risk of malignancy or the tumor is very large. Laparoscopy offers magnified visualization and access to pelvic planes but can be technically demanding when a large lesion extends low in the pelvis or is densely adherent to surrounding structures. In the perineal approach, the tumor is accessed directly, but there is risk of injury to surrounding nerves and vessels, so delicate dissection is required [18,19].

In our case, we preferred the transabdominal via laparoscopic approach even though the lesion was extending below S3. It permitted controlled mobilization of the rectosigmoid colon, direct visualization of the retrorectal plane, and careful dissection of the cyst wall. A combined approach can be used in extensive lesions with the aim of complete excision to prevent the risk of recurrence and complications. Congenital cysts carry an excellent prognosis after complete excision.

Chung et al published a case series on outcomes of the laparoscopic transabdominal approach for retrorectal tumors between 2017 and 2020 including 5 patients in whom the median tumor size was 50 × 40 mm. There was a surgical site infection in 1 case and recurrence was seen in 2 cases due to incomplete tumor resection because of the possibility of damage to nerves and vessels [20]. This series highlighted the laparoscopic approach as a better option and the importance of complete resection. Histopathological examination is necessary to exclude unexpected malignant change.

This case has several limitations. It is a single-patient report and therefore cannot establish a preferred surveillance interval or a universal indication for surgery in asymptomatic retrorectal epidermoid cysts. The patient had only 3 months of documented postoperative follow-up, and no postoperative surveillance imaging had been performed at the time of reporting. In addition, serum squamous cell carcinoma antigen was not obtained before surgery. These limitations should be considered when interpreting the clinical course.

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Conclusions

Retrorectal epidermoid cysts can remain asymptomatic for many years despite progressive enlargement. In an asymptomatic patient, documented growth, increasing mass effect, diagnostic uncertainty, or suspicious imaging features may support a decision for surgical excision. MRI is valuable for diagnosis and operative planning, while complete excision and histopathological examination provide definitive treatment and diagnosis. Laparoscopic excision can be feasible even for a large low pelvic lesion when careful dissection and appropriate preoperative planning are possible. Because evidence is limited to case reports and small series, individualized surveillance and treatment decisions remain necessary.

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Informed Consent

Informed consent was obtained for the publication of this report.

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