

CASE REPORT

Retrorectal Cystic Lesion Presenting As Subcutaneous Swelling: A Case Report

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INTRODUCTION

The retro rectal space, a potential area, manifests when a mass develops within it¹. Derived from embryonic neuroectoderm, notochord, and hindgut, this region houses a diverse range of both benign and malignant lesions². About two-thirds of presacral tumours are congenital, with over 60% being developmental cysts³. In adults, developmental cysts, including epidermoid cysts, dermoid cysts, enteric cysts, and neurenteric cysts, are the most prevalent retrorectal cystic lesions, particularly in middle-aged women⁴. Here, we present a rare case of a perianal subcutaneous swelling that, upon evaluation, revealed a dumbbell-shaped retrorectal cystic lesion, ultimately diagnosed as an epidermoid cyst.

CASE DESCRIPTION

A 28-year-old male, previously enjoying good health, presented with a progressive increasing swelling in his perianal region persisting for the last two years. Remarkably, he denied any concurrent local pain, fever, constipation, tenesmus, or alterations in bowel habits. During the physical examination, around 4×5 cm painless, well-circumscribed cystic swelling was observed in the region between the anus and coccyx. Notably, there were no signs of local inflammation.

During the digital rectal examination, there was a soft, round, nontender mass with a regular shape on the posterior wall. Laboratory haematological investigations were normal. On pelvic MRI, a dumbbell-shaped cystic lesion

measuring (5.2×4.6×6) cm was revealed, situated posteriorly to the anorectal canal. The lesion appeared hypointense on T1-weighted images and hyperintense on T2-weighted images with discernible hypointensity foci on T2-weighted images. Importantly, the MRI findings ruled out involvement with other pelvic organs such as the bladder, urethra, seminal vesicles, prostate, and rectum. The lesions appeared distinctly separate from these structures. The differential diagnosis based on the MRI results included an epidermoid cyst, tailgut cyst or a congenital cyst.

Consequently, based on clinical and imaging findings, patient was taken up for surgical intervention. Posterior approach was used for the surgical excision of the mass through a 4cm midline perineal incision, with the patient in jackknife position. The surgical procedure involved careful dissection of the mass from the anterior rectal wall and the posterior coccyx curve. The mass was completely excised without any rupture.

The diagnosis of an epidermoid cyst was confirmed through histological analysis of the excised tissue. Following a smooth postoperative course, the patient was discharged on the fourth postoperative day, attesting to the success of the intervention.

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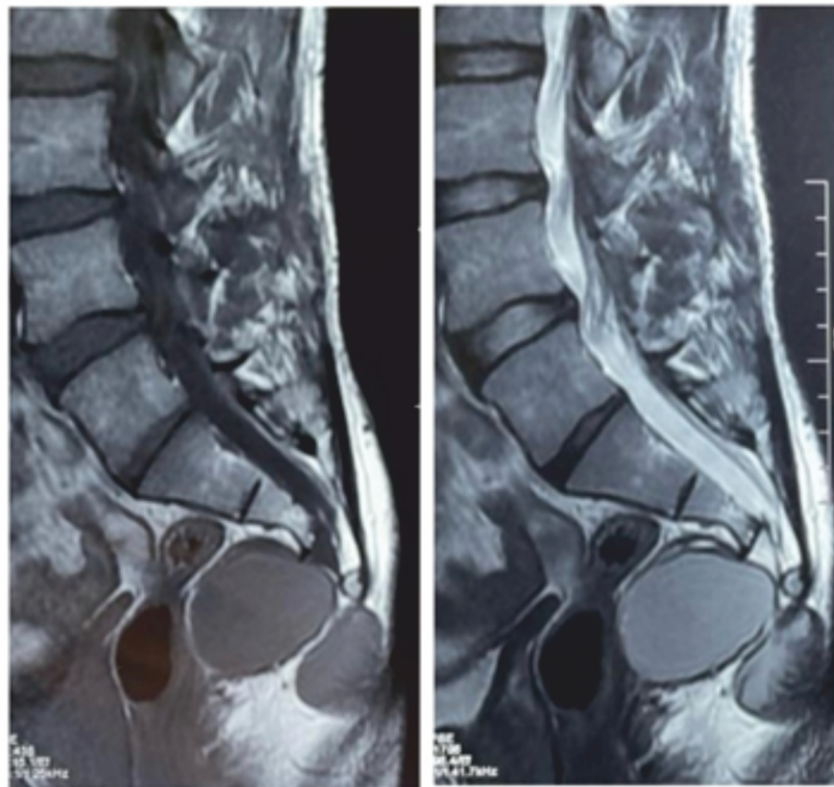


Fig. 1 The lesion appeared hypointense on T1-weighted images and hyperintense on T2-weighted images with discernible hypointensity foci on T2 weighted images



Fig.2. a) Intraoperative picture of cyst, located between the anus and coccyx
b) Excision of cyst. A posterior transperineal approach was used to excise the cystic mass

DISCUSSION

Primary tumors in the retrorectal or presacral space are exceedingly rare, constituting a mere 1 in 40,000 hospital admissions according to extensive data from the Mayo Clinic⁵. This distinctive group primarily encompasses congenital developmental cysts, such as dermoid cysts, epidermoid cysts, enteric duplication cysts, and neuroenteric cysts⁵. Notably, tail gut cysts, also recognized as retro rectal cystic hemartomas, fall under this classification⁶. Predominantly benign, retrorectal masses present a diagnostic challenge, requiring consideration of diverse possibilities including cystic teratoma, epidermal cyst, neuroblastoma, pyogenic abscess, adnexal mass, and infrequently necrotic rectal leiomyosarcoma⁷. Presacral tumours are further classified into inflammatory, congenital, neurogenic, osseous, and miscellaneous categories, distinguished as congenital or acquired⁸.

Within the realm of acquired lesions, there exists a subset featuring rare malignant transformations in tailgut cysts, comprising carcinoid tumours, endometrioid carcinoma, adenosquamous carcinoma, and sarcoma. Epidermoid cysts located in the retrorectal region are notable for their origin from remnants of ectodermal tissue and having distinctive features. These cysts have a thin wall lined with stratified squamous epithelium, a granular cell layer, and a unique content mix of desquamated debris, keratin, cholesterol, and water⁹. Interestingly, data from a particular study suggests that retrorectal epidermoid cysts are associated with one in every 40,000 admissions¹⁰.

Clinical manifestations of retrorectal and presacral masses are contingent upon factors such as lesion size, location, and the presence of infection or malignancy¹¹. Identifying these cysts poses a challenge, as symptomatic cases manifest noticeable symptoms that can be easily confused with anorectal abscesses, complicated fistulas, and pilonidal sinuses. Typical symptoms include anal pain, perianal discomfort, tenesmus, defecation difficulties, bleeding, thin stool, changes in bowel habits, incontinence, and the presence of a sacrococcygeal mass¹².

Diagnosis is a multifaceted process encompassing physical and rectal examinations, supplemented by proctoscopy and radiological imaging. A nuanced understanding of pelvic anatomy, patient presentation, imaging nuances, and histologic findings is imperative for accurate diagnosis and

tailored treatment. Magnetic Resonance Imaging (MRI) is the favored imaging method for retrorectal lesions. In T1-weighted images, epidermoid cysts exhibit hypointensity, while in T2-weighted images, they appear hyperintense, with restricted diffusion on diffusion-weighted imaging. The importance of incorporating diffusion-weighted imaging in MRI is underscored¹³. Nevertheless, opting for a preoperative biopsy remains a topic of controversy due to the potential risks of cyst infection and tumour spillage. Surgical resection, particularly for resectable lesions, stands out as the optimal diagnostic and therapeutic avenue, considering the risk of secondary infection and the rare potential for malignant degeneration¹⁴. Even for asymptomatic lesions, surgery is recommended for various reasons. Firstly, around 50% of developmental cysts may experience spontaneous infections. Secondly, although uncommon, developmental cysts carry the risk of malignant transformation¹⁵. This occurrence has been documented for enteric cysts and tailgut cysts. Lastly, in women of childbearing age, a presacral mass might complicate vaginal delivery, potentially causing dystocia¹⁶.

The crux of treatment lies in complete surgical excision, offering a definitive solution. Surgical approaches, including laparoscopic, anterior, posterior, or combined, are selected based on intricate considerations of size, location, and potential invasion of surrounding structures. The anterior approach is favoured for tumours located above the mid-body of S3 or S4 vertebrae, whereas the posterior approach is suitable for those below. A combined approach is advised for tumours spanning both areas or showing potential invasion of surrounding structures. In instances of observed invasion, a multidisciplinary approach may be required. Ensuring the meticulous excision of the cyst wall is crucial to prevent recurrence and minimize complications¹⁵.

CONCLUSION

Uncommon and diverse, retro rectal tumours are seldom encountered and frequently subject to misdiagnosis due to their typically vague and nonspecific symptoms. Utilizing radiologic imaging, particularly MRI, is crucial for surgical preparation, and caution should be exercised against resorting to biopsy. Optimal treatment for diagnosed presacral lesions involves complete resection, even in asymptomatic cases.

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