CASE REPORT

Presacral mass in the setting of an ovarian cyst and abdominal pain

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SUMMARY

Tailgut cysts, also known as retrorectal hamartomas, are very rare neoplasms located in the presacral/retrorectal space that originate from the embryonic hindgut. Although a majority of lesions in this location are benign, 30% of the reported cases in the literature were found to be malignant. This report describes a case of a presacral mass found on CT of a 37-year-old woman who initially presented with worsening abdominal pain and a history of ovarian cyst rupture. This patient's clinical picture was complicated by an enlarging ovarian cyst. The risk of progression to malignancy warranted excision. She recovered well with resolution of her presenting symptoms. We report this case along with a brief review of the literature with a focus on the surgical considerations.

BACKGROUND

Tailgut cysts, also known as retrorectal hamartomas, are very rare neoplasms in the presacral/retrorectal space that originate from the embryonic hindgut. Although a majority of lesions in this location are benign, 30% of the reported cases in the literature were found to be malignant. The pathogenesis of tailgut cysts is thought to be related to incomplete embryogenesis.

Herein, we describe a unique case of a solitary tailgut cyst occurring in a patient who presented with abdominal pain in the setting of a coinciding enlarging ovarian cyst. The presacral mass was successfully removed using a modified York-Mason posterior approach. We also discuss a brief review of the literature with a focus on the surgical considerations of these lesions.

CASE PRESENTATION

A 37-year-old gravida 5, para 1 woman presented to the emergency department (ED) with abdominal pain. She had a history significant for four miscarriages and three ovarian cysts — the most recent cyst ruptured and required emergent surgery.

On presentation to our ED, her pain was similar in nature as prior cystic pain, was colicky, radiating down her right leg, worse with walking and worsening. Of note, the patient had been taking fertility medications for the past several months. She denied any dysuria or haematuria. On exam she was tender to palpation and had a positive obturator sign.

INVESTIGATIONS

Given her history of fertility treatments and prior ovarian pathology, she was first evaluated with ultrasound/Doppler, which revealed a 2 cm simple ovarian cyst. Subsequently, a CT scan was performed to evaluate for appendicitis and other acute causes of her symptoms. This revealed a 4 cm presacral complex mass (figure 1).

Based on history and constellation of findings, an outpatient work-up with MRI abdomen/pelvis and colonoscopy was arranged. The presacral mass was evaluated by MRI (figure 2).

DIFFERENTIAL DIAGNOSIS

The findings of this presacral mass prompted work-up for malignancy, although the suspicion was low given the clinical context. Her lack of an elevated white count and fever made an abscess equally as unlikely.

During this time she returned to the emergency room with worsening abdominal pain and an ovarian cyst that had enlarged to nearly double in size. Our gynaecology colleagues were reassured that this was a physiological cyst in the setting of her fertility treatments and did not require intervention. Confident her pain was not related to her enlarging ovarian cyst, she was discharged home.

TREATMENT

In the clinic, she reported having 'pelvic pressure' and increased pain just prior to bowel movements. Surgery was recommended given her new symptoms and the risk of progression to malignancy. The mass was excised successfully using a modified York-Mason posterior approach.

OUTCOME AND FOLLOW-UP

A week status postdischarge, she developed a fever to 101.8, a white count of 12.8 and was found to have findings on MRI pelvis consistent with an abscess in the retrorectal space. The infected fluid collection was drained via interventional radiology, with no evidence of communication to the rectum. She recovered on a course of intravenous ciprofloxacin and metronidazole and was discharged shortly after. At 2 weeks' follow-up, the patient recovered and the incision had closed nicely. She reported resolution of her 'pelvic pressure' and pain with bowel movements.

Pathology revealed a gross specimen measuring 4×3.5×1.3 cm, multifocal, cystic and ragged nature

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Figure 1 CT with contrast showing a 4 cm mass with thin septations anterior to the tip of the coccyx.

(figure 3). The mass consisted of multiple cystic spaces with abundant mucoid material lined by transitional and glandular epithelium. There was disorganised smooth muscle surrounding the cysts. No malignancy was identified.

Immunohistochemistry stains of lesional cells demonstrated the following: positive for cytokeratin 7, GATA 3, P40 (basal cells) and smooth muscle actin; negative for Keratin 20 20,

Figure 2 MRI pelvis with 4 cm multiloculated cystic lesion with fluid level posterior to the rectum without inflammatory change.

Figure 3 Gross specimen measuring 4x3.5x1.3 cm, multifocal, cystic and ragged nature.

S100 protein, calretinin, Wilms tumor protein-1, Caudal Type Homeobox 2 and Thyroid Transcription Factor 1. Mucicarmine stain was positive in the focus of glandular epithelium. The findings were consistent with a retrorectal cystic hamartoma (figure 4).

DISCUSSION

Tailgut cysts are exceedingly rare congenital lesions of the presacral space. All lesions in this location are estimated at 1 in 40 000 — a recent review detailing only 1708 patient cases in the literature to date.¹ The age range at diagnosis is large (4–73 years), although the average age is 35 years. Interestingly, tailgut cysts are more common in women, with a female-to-male ratio of 3:1. Although women are overwhelming the majority affected, patients of the male gender and older age (53.9 ± 11.5 vs 40.1 ± 12.2 years) were more likely to have a malignant tumour.¹ Benign lesions compose 70% in any category (figure 5). On the other hand, 30% of all lesions in this space are malignant.¹ This

Figure 4 The mass consisted of multiple cystic spaces with abundant mucoid material lined by transitional and glandular epithelium.

Figure 5 Classification of tumours found in the presacral space. Of the benign congenital tumours, tailgut cysts are the most common.

rate may be considerably higher — Mayo Clinic's experience² — with these lesions over the course of 19 years reported at a malignancy rate of 43%.

The origin of presacral masses varies, but roughly 60% are congenital. The rest of the lesions vary pathologically — neurogenic, osseous and carcinoid features have been described.^{1 3} They can often distort surrounding structures and result in bony defects of the lower sacral bone.⁴ Of the congenital subset, tailgut cysts make up about half, followed by teratomas and epidermoid cysts. Tailgut cysts are believed to arise from vestigial remnants of the embryonic hindgut.

Clinical presentation

The anatomical location and slow growth of these tumours are major reasons for the late presentation. The reported symptoms

at time of diagnosis often reflect the growth of the tumour once it begins to affect the surrounding structures, such as the sacrum or rectum (figure 6). The most common symptoms include pain and constipation. Of the patients seen at Memorial Sloan Kettering⁵ over a 28-year period, the major presenting complaint was pain in the buttock/low back, or radicular to the legs. Pain may reflect the mass effect on the surrounding structures.² Rectal fullness and difficulty passing urine or stool are also associated, although not common. Our patient was one of the few who did report these uncommon symptoms that improved following surgery. Low abdominal pain and swelling on the coccyx or gluteal area are sometimes present.

Surgical considerations

Although tailgut cysts are largely benign masses, the gold standard of treatment is complete surgical excision. Hosseini-Nik and colleagues⁶ describe an approach to imaging these tumours that will aid in the selection of the optimal surgical approach. Approaches are based on the lesion extent and relationship to adjacent structures. There are multiple approaches to resection of retrorectal tumours that are tailored to the patient given the clinical and anatomical considerations (figure 7).

Patients have an excellent prognosis with few mortalities reported in the literature. The 5-year survival, although 20% in the malignant cases, was upwards of 80% in the benign group. Postoperative complications did occur in about 40% of patients in one study, which included wound infections, stool incontinence, retrorectal abscesses and a faecal fistula.² Another also reported complications in 15% of their cases, which included bleeding, rectal injuries and infection requiring drainage.⁷

Figure 6 Retrorectal space lies behind the rectum below the pelvic peritoneum and mesorectum.

Figure 7 Considerations for surgical approach of retrorectal tumours based on anatomical location.

Learning points

- Lesions in the presacral space, when large enough, will affect surrounding structures. They commonly lead to presenting symptoms such as pelvic pressure, pain with defecation, low back pain and sacral radiculopathy.
- 30% of all lesions in this space will be malignant — excision is always recommended.
- Although they predominate in middle-aged women as benign lesions, older patients and men are more likely to have a malignant tumour.

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