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Management of a Primary Retroperitoneal Yolk Sac Tumor

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Patient: Male, 31-year-old
Final Diagnosis: Primary retroperitoneal yolk sac tumor
Symptoms: Lower abdominal pain
Medication: —
Clinical Procedure: —
Specialty: Oncology • Surgery

Objective: Rare disease

Background: Existing literature has detailed occurrences of retroperitoneal yolk sac tumors (YSTs) as the result of metastasis from a primary gonadal site. However, primary retroperitoneal YSTs are extremely rare, thus remaining a challenge to diagnose and treat. We present a complex case of a large primary retroperitoneal YST in a man treated with neoadjuvant chemotherapy followed by surgical resection.

Case Report: A 31-year-old man presented with a chief symptom of severe lower abdominal pain. Diagnostic imaging revealed a large, rapidly progressing neoplasm in the retroperitoneal region, initially thought to be a sarcoma. However, the pathological results from further biopsies found the mass to be a retroperitoneal YST, which was tethered to a large portion of the small bowel. A testicular ultrasound was used to confirm that the mass was a primary tumor with no origins in the gonads. The tumor progressed to involve several fistulas connected to the small intestine and anterior abdominal wall. The patient was treated with 3 cycles of bleomycin, etoposide, and cisplatin, followed by surgical excision of the residual mass. The patient retained normal gastrointestinal functions, and subsequent imaging revealed no evidence of recurrence 2.5 years following resection.

Conclusions: Owing to the rarity of extragonadal primary YSTs, diagnostic and treatment standards have not yet been sufficiently explored. Our case demonstrates that a combination of chemotherapy and surgical resection should be considered for select patients with primary YST in the retroperitoneal region.

Keywords: General Surgery • Neoadjuvant Therapy • Neoplasms, Unknown Primary • Retroperitoneal Neoplasms • Yolk Sac • Male Germ Cell Tumor

Full-text PDF: https://www.amjcaserep.com/abstract/index/idArt/933258

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Background

A germ cell tumor (GCT) is a malignant germ cell neoplasm typically occurring in the gonads. While the exact pathogenesis of such GCTs is not yet fully understood, the literature is currently divided between 2 possible explanations. While some reports propose that extragonadal GCTs arise from primordial cells that have failed to complete their normal migration to the urogenital ridge during development, others posit that the gonadal germ cells in their correct location then undergo a process of "reverse migration" [1,2]. A yolk sac tumor (YST), in particular, is a rare type of nonseminomatous germ cell tumor. Extragonadal YST occurrences make up only 5.7% of GCTs among male patients in the United States, with mediastinal sites being the most common, followed by the pineal gland and retroperitoneum [3].

The rarity of primary extragonadal GCTs, in addition to their frequently mixed histologic presentation, often make them a challenge to diagnose early [4]. Furthermore, YSTs are difficult to identify, as they commonly present themselves in combination with other types of GCTs, such as embryonic carcinoma and teratoma [5]. In almost all cases of YST, the results of immunohistochemical staining reveal high serum levels of alpha-fetoprotein. However, this alone cannot be used as a unique biomarker, as alpha-fetoprotein is also produced by non-GCTs, including ovarian and liver cancers [6]. In terms of clinical presentation, patients with retroperitoneal YSTs most commonly present with abdominal and back pain as primary symptoms [7]. Following diagnosis, extragonadal YSTs are most commonly treated using a combination of chemotherapy and surgical resection [4,6,7].

The existing literature has detailed several occurrences of retroperitoneal YST as a result of metastasis from a primary gonadal site. However, primary retroperitoneal YSTs with no evidence of an original gonadal mass are extremely rare. To the best of our knowledge, fewer than 20 cases have been reported to date. We report a case of primary retroperitoneal YST in an adult male patient, which was successfully managed with neoadjuvant chemotherapy followed by surgical resection of the residual mass.

Case Report

A 31-year-old man presented to the Mayo Clinic Jacksonville with severe lower abdominal pain. Abdominal and pelvic magnetic resonance imaging revealed a retroperitoneal mass sized 20×10×14 cm, which had doubled in diameter over a period of approximately 5 weeks (**Figure 1**). Following computed tomography-guided core needle biopsies, the mass was determined to be a high-grade malignant neoplasm and was initially

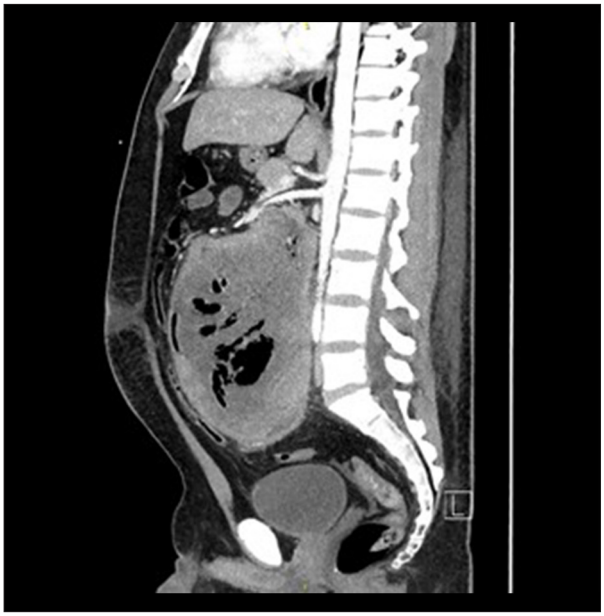

Figure 1. Sagittal view of primary retroperitoneal yolk sac tumor at initial presentation.

suspected to be a large retroperitoneal sarcoma, a class of cancer originating in the bone or connective tissue. Within 1 month of initial consultation, the patient's symptoms drastically progressed to include hemorrhage and superinfection of the mass. This precluded the patient from undergoing radiation therapy prior to surgery. An exploratory laparotomy revealed the surface of the tumor to be tethered to the majority of the small intestine as well as the sigmoid colon and right and left colon mesentery. Due to the need for significant bowel resection, the mass was deemed unresectable. A follow-up esophagogastroduodenoscopy showed 2 sites of small bowel fistulization to the third portion of the duodenum as well as the jejunum, which were suspected to be a source of slow intra-luminal blood loss. A substance resembling coffee grounds was also identified in the stomach. The patient then underwent a pigtail G-tube placement by the interventional radiology team for decompression. In an effort to reduce active bleeding, the patient was administered 6 days of palliative radiation therapy to the abdomen, which was tolerated well. The bleeding appeared to slow following treatment, as indicated by a stabilizing hemoglobin level.

Pathology results from the aforementioned biopsies were diagnostic of a YST. Thus, the patient underwent a testicular ultrasound which showed no indication of a primary gonadal mass. The patient consented to proceed with chemotherapy consisting of 4 cycles of bleomycin, etoposide, and cisplatin (BEP therapy). Following 3 cycles of chemotherapy, the mass was visibly reduced to 6×6×5 cm, with significantly decreased association with the small bowel (**Figure 2**). The patient underwent a diagnostic laparoscopy, which revealed a new fistula

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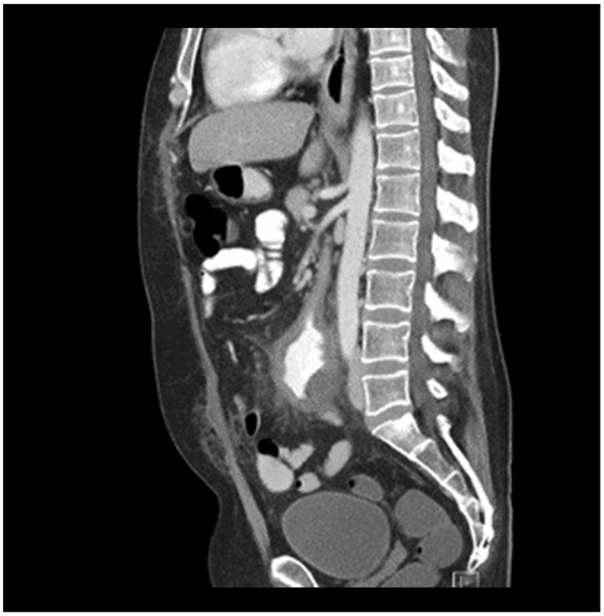

Figure 2. Sagittal view of shrunken primary retroperitoneal yolk sac tumor following 3 cycles of chemotherapy.

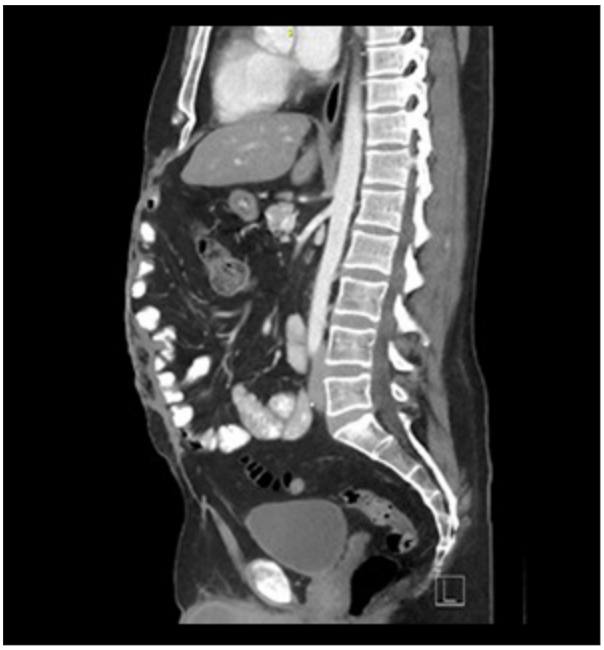

Figure 3. Sagittal view of abdomen 2.5 years following resection, with no evidence of recurrence.

from the tumor to the anterior abdominal wall. We proceeded with surgical resection of the residual mass. Upon entry into the abdomen, an extensive desmoplastic reaction was evident surrounding the tumor and its adjacent structures. Owing to the observed fistulization, a partial small bowel resection was performed, which included the third and fourth portions of the duodenum, the proximal jejunum, a short segment of mid jejunum, and a short segment of ileum. The appendix was also removed. Approximately 260 cm of small bowel remained, and the resections were consistent with an oncologic R0 resection, indicating negative gross and microscopic margins. Aside from the primary tumor mass, there was no evidence of metastasis within the abdomen. Total operative time was 557 min, with an estimated 300 mL of blood loss.

The patient recovered well and was discharged 26 days after surgery. Subsequent imaging performed up to 2.5 years following surgery did not show any evidence of recurrence, and the patient continued to retain normal gastrointestinal functions (**Figure 3**).

Discussion

A germ cell tumor with no identifiable origin in the ovary or testes is classified as a primary extragonadal GCT. Although varying with age, these tumors typically arise in midline locations, consistent with what was found in the present case. Adults most frequently present with GCTs in the mediastinum, while children more commonly develop masses in the sacrococcygeal or intracranial regions [7].

Following diagnosis, our patient was initially treated with 3 rounds of cisplatin-based chemotherapy to reduce the significant size of the tumor. In select patients, chemotherapy alone can serve as an effective treatment. A similar case presented by Hong et al detailed the use of BEP therapy to successfully treat a primary retroperitoneal YST in an adult male patient [8]. This strategy has also been reportedly successful in the management of similar presentations in children. For instance, a report by Murat et al describes a case in which a 28-month-old boy received 4 cycles of BEP therapy to treat a primary retroperitoneal YST, with no evidence of recurrence [9].

However, in instances such as ours, surgical resection of a residual mass may also be warranted. In this case, extensive small bowel resection was necessary owing to the multiple fistulized tracts between the primary mass and surrounding structures. The intense desmoplastic reaction also caused some significant difficulty in differentiating between the YST mass and surrounding necrosis. This was overcome by samples from suspicious areas being sent for pathologic confirmation during surgery, thus decreasing the risk of unnecessary resection close to vital structures, such as the abdominal aorta and inferior vena cava. The decision to perform an appendectomy was also necessary owing to the increased mobility of the colon following resection, giving it a tendency to migrate into the left upper quadrant.

Although the literature on primary retroperitoneal YSTs is limited, a combination of BEP therapy and surgical resection

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appears to be an effective treatment. Guo et al described the management of a primary YST in the retroperitoneal region of a 19-year-old woman using surgical resection as well as neoadjuvant and adjuvant BEP therapy [10]. Similarly, Wada et al performed an incomplete resection followed by multiple cycles of chemotherapy to manage a primary retroperitoneal YST in an adult male patient [11].

According to the published literature, the 5-year progression-free rate for patients with this type of tumor was 42%, with an overall survival rate of 65% [7]. In cases of recurrence, studies have shown that salvage therapy with cisplatin-based chemotherapy still affords patients long-term disease-free survival in 30% of cases [12]. Fortunately, the present patient showed no sign of tumor recurrence following his initial surgery and continues to do well 2.5 years later.

Our patient was thoroughly evaluated with testicular ultrasound to confirm the absence of a testicular primary mass and verify that this was in fact a true primary retroperitoneal GCT. However, there have been some reported cases where the primary gonadal tumor had regressed, making identification of a testicular primary difficult [13,14]. Given the rarity of this type of tumor and its relatively good prognosis with the established treatment, patients can benefit greatly from increased awareness leading to the earlier identification of primary extragonadal YST.

Conclusions

While uncommon, primary retroperitoneal YSTs progress rapidly and require quick diagnosis as well as aggressive treatment. As demonstrated by this case, the use of chemotherapy followed by surgical resection can be effective in the management of large retroperitoneal masses. Further studies are warranted to establish standards in the identification and management of primary retroperitoneal YSTs.

Declaration of Figures' Authenticity

All figures submitted have been created by the authors who confirm that the images are original with no duplication and have not been previously published in whole or in part.

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