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Jejunal Extragonadal Germ Cell Tumor Presenting as an Abdominal Mass: A Case Report

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Abstract

Extragonadal germ cell tumors (EGGCTs) are rare malignancies that arise outside the gonads and occur infrequently in the gastrointestinal tract. Their presentation often mimics that of other abdominal neoplasms, posing diagnostic challenges. Histopathological and immunohistochemical confirmation, along with tumor marker evaluation, is essential for diagnosis. Early diagnosis and platinum-based chemotherapy can significantly improve patient outcomes. A 25-year-old male presented with abdominal pain, a palpable mass in the umbilical region, and subacute intestinal obstruction. Imaging revealed a heterogeneously enhancing lesion in the mesentery and a subcapsular lesion in the left kidney. Emergency debulking surgery was then performed. Histopathology and immunohistochemistry confirmed an EGGCT favoring a yolk sac tumor. Serum marker levels were elevated with AFP (alpha-fetoprotein) of 854 IU/mL, 23 mIU/mL of β-human chorionic gonadotrophin (β-hCG); 277 U/L of LDH. Ultrasonography of the testes revealed microcalcifications in the left testis, without overt malignancy. The patient was diagnosed with a Stage IIIC extragonadal non-seminomatous germ cell tumor. He was started on an EP regimen owing to poor pulmonary function, followed by BEP, and was scheduled for a high inguinal orchidectomy. This case highlights the importance of including EGGCTs in the differential diagnosis of abdominal masses in young males. A multidisciplinary approach with timely histological and oncological assessments is vital. Early intervention with platinum-based chemotherapy offers a favorable prognosis, even in advanced stages. Given the rarity of this presentation, further studies are needed to refine diagnostic and therapeutic strategies.

Keywords

- extragonadal germ cell tumor
- yolk sac tumor
- abdominal mass
- non-seminomatous germ cell tumor
- young adult

Introduction

Extragonadal germ cell tumors (EGGCTs) are a rare subset of germ cell neoplasms, accounting for 2 to 5% of all germ cell tumors. These tumors typically arise in midline structures such as the mediastinum and retroperitoneum, with gastrointestinal involvement being extremely uncommon. Its origin is thought to stem from the aberrant migration of primordial germ cells during embryogenesis.[1]

Clinical manifestations depend on tumor location, but intra-abdominal cases often present with nonspecific symptoms, such as abdominal pain, mass effect, or intestinal obstruction. Given their rarity, EGGCTs in the gastrointestinal tract are frequently misdiagnosed or initially mistaken

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for more common pathologies such as gastrointestinal stromal tumors or lymphomas.[2]

Diagnosis requires a high index of suspicion and relies on a combination of histopathology, immunohistochemistry, and serum tumor markers. Yolk sac tumors, a frequent histological variant, characteristically express alpha-fetoprotein (AFP), glypican-3, and SALL4.[3] Elevated AFP and β-human chorionic gonadotropin (β-hCG) levels are important for both diagnosis and treatment response monitoring.

The standard treatment for non-seminomatous EGGCTs involves platinum-based chemotherapy, primarily the bleomycin, etoposide, and cisplatin (BEP) regimen. Surgery is often reserved for residual disease or complications, such as obstruction or bleeding.[4] Surveillance of the testes is essential to exclude metachronous or occult gonadal diseases.[5] Given the infrequent occurrence of gastrointestinal EGGCTs, further case reporting and literature synthesis are crucial to guide early diagnosis and evidence-based treatment.

Germ cell tumors are neoplasms derived from primordial germ cells that are typically found in the gonads.[6,7] However, these tumors can also occur at extragonadal sites because of aberrant migration of primordial germ cells during embryogenesis.[8] The most common locations for EGGCTs are the mediastinum, retroperitoneum, and pineal gland.[9,10] The diagnosis of EGGCTs can be challenging because of their rarity and the wide range of differential diagnoses that must be considered. Symptoms vary considerably depending on the size and location of the tumor, and a significant number of individuals remain asymptomatic.[9] Gastrointestinal stromal tumors are the most common mesenchymal tumors of the gastrointestinal tract, and may be benign or malignant.[11] However, EGGCTs presenting as primary intestinal masses are exceedingly rare, with only a handful of cases reported in the literature. These tumors pose a diagnostic challenge for clinicians because of their unusual presentation and the need to differentiate them from other, more common intestinal malignancies.

This case highlights the exceptional presentation of an EGGCT localized to the jejunum, a seldom-involved site. Documenting such rare clinical entities adds to the current understanding, raises diagnostic awareness, and supports timely and accurate management in similar future cases.

Case Report

A 25-year-old male patient presented to a tertiary oncology care center with complaints of abdominal pain, abdominal mass, and subacute intestinal obstruction for 1 month. Vital signs on presentation were a blood pressure of 112/70 mm Hg and a pulse rate of 90 beats per minute. Upon clinical examination, the mass in the abdomen was palpable at approximately 8×9 cm in the umbilical region of the abdomen. No other physical findings were noted. The patient underwent an emergency debulking surgery.

The complete blood count revealed total leucocyte count of $9.1 \times 10^3/\mu L$, absolute neutrophil count of $5.6 \times 10^3/\mu L$, and platelet count of $422 \times 10^3/\mu L$. No abnormalities were observed in liver function, renal function, or serum electrolytes. Histopathology (jejunal mass debulking) was suggestive of poorly differentiated neoplasm (►**Fig. 1**). Immunohistochemistry (jejunal mass debulking) was positive for SALL4, CK, patchy PLAP, and AFP, suggesting an EGGCT favoring a yolk sac tumor (►**Fig. 2**). Serum markers analysis demonstrated a β-hCG of 23 mIU/mL, AFP of 854 IU/mL, and LDH of 277 U/L. Postoperative computed tomography scan of thorax, abdomen, pelvis was done, which showed an ill-defined heterogeneously enhancing lesion. The lesion was epicentered at the root of the mesentery and was abutting anterior abdominal wall measuring 4.2 cm $\times$ 5.9 cm $\times$ 9 cm (AP $\times$ TR $\times$ CC), right kidney was normal, and left kidney was measuring 7.7 cm $\times$ 8.9 cm $\times$ 12.5 cm (AP $\times$ TR $\times$ CC) with few heterogeneously enhancing lesion with hypoenhancing areas noted in the subcapsular region of left kidney measuring 3.9 cm $\times$ 2.8 cm. The retroperitoneum and thorax were normal (►**Fig. 3**). Ultrasonography of the bilateral testes showed a normal right testis and left testis with microcalcification (►**Fig. 4**). The diagnosis of stage IIIC poor-risk non-seminomatous germ cell tumor was made according to the IGCCCG (International Germ Cell Cancer Collaborative Group) classification.

Treatment

The patient underwent a first-cycle EP because of an ECOG performance status of 2, and PFT showed a restrictive pattern due to poor effort post-surgery, with a percentage predicted DLCO of 45 and a percentage predicted FEV1 of

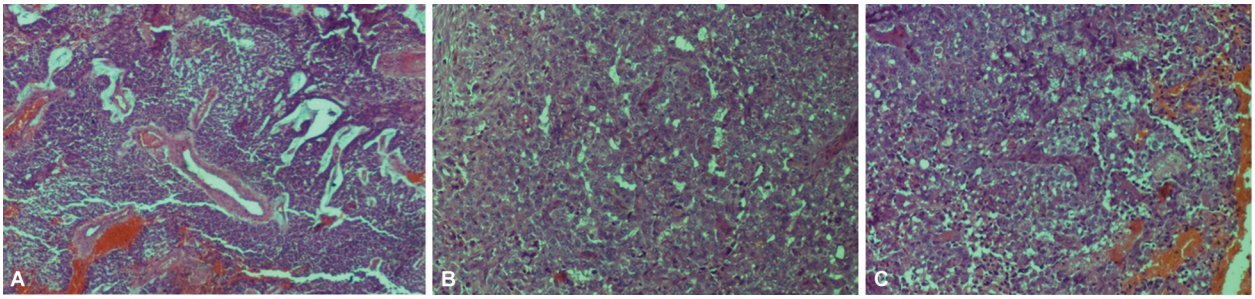

Fig. 1 (**A**) Histopathological image of a yolk sac tumor demonstrates a reticular (microcystic) growth pattern, characteristic of this malignant germ cell tumor. (**B**) Histopathological image of yolk sac tumor; hyaline globules are seen. (**C**) Histopathological image of yolk sac tumor hyaline globule and Schiller-Duval bodies.

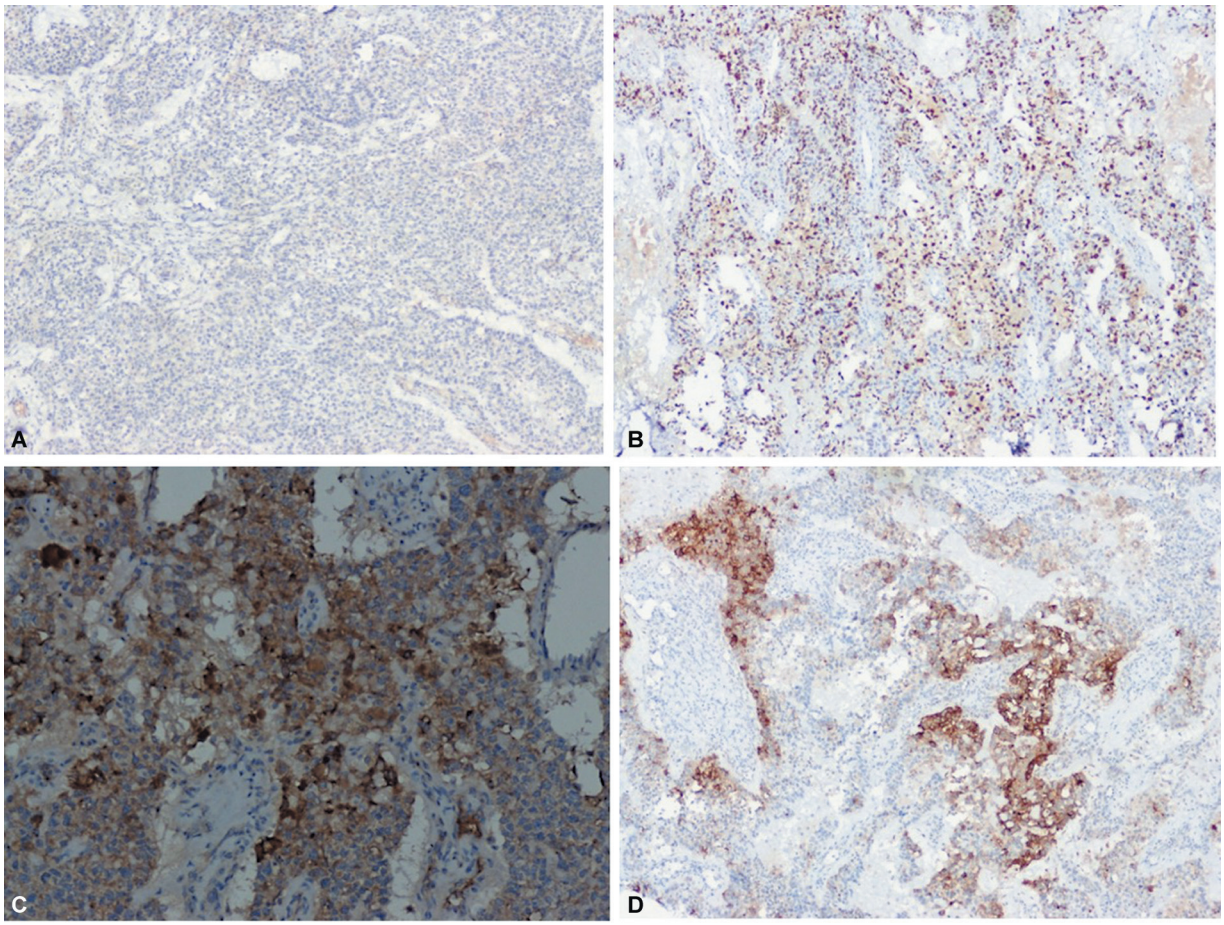

Fig. 2 Immunohistochemistry (**A**) negative for CK7 and CK20 (**B**) diffuse membranous and cytoplasmic positive for CK (**C**) diffuse strong nuclear positivity for SALL4 (**D**) patchy positivity for AFP and PLAP.

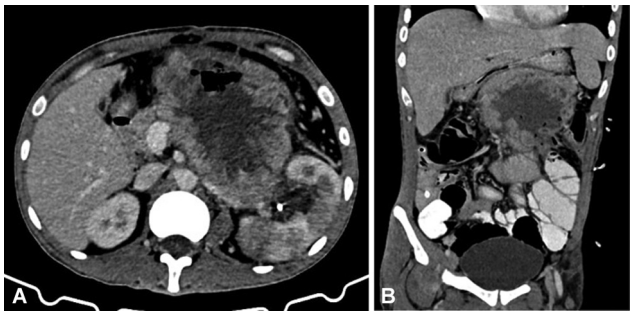

Fig. 3 (**A**) CT image with cross-sectional view of jejunal mass. (**B**) CT image with coronal view showing jejunal mass.

62, FVC of 58, and FEV1/FVC of 108. After the first cycle of chemotherapy, serum marker levels showed a decreasing trend. PFT and DLCO were repeated, which showed an improved report with a percentage predicted DLCO of 76. The patient was started on the BEP regimen, and was scheduled for high inguinal orchidectomy on a later date. Currently, the patient is undergoing treatment and has completed one cycle of EP and three cycles of BEP with residual jejunal disease of more than 1 cm postchemotherapy. Patient is asymptomatic and planned for surgical resection of the residual abdominal mass along with a left high inguinal orchidectomy in view of microlithiasis noted on USG, Suggestive of a burnt-out primary tumor. Addressing the burnt-out primary is essential, as leaving it untreated increases the risk of relapse.

Discussion

This case highlights the atypical presentation of an EGGCT in a young male with complaints of abdominal pain and subacute intestinal obstruction. The absence of gonadal involvement and localization to the jejunum highlights an unusual clinical scenario. Diagnosis was established through a combination of histopathology, immunohistochemistry, and markedly elevated serum tumor markers (AFP and β-hCG).[1,3] The patient responded well to platinum-based chemotherapy (EP/BEP) and was scheduled for surgical intervention, indicating a favorable trajectory despite poor-risk staging.[4]

EGGCTs are primarily present in the mediastinum and retroperitoneum; gastrointestinal tract involvement remains exceedingly rare. According to recent reviews, only a handful of jejunal germ cell tumors have been reported, and most are diagnosed postsurgically because of their overlapping presentations with GISTs or lymphomas.[9] Similar to the findings of Dieckmann and Oechsle[1] and Park et al,[2] our case highlights the difficulty in preoperative diagnosis and the crucial role of

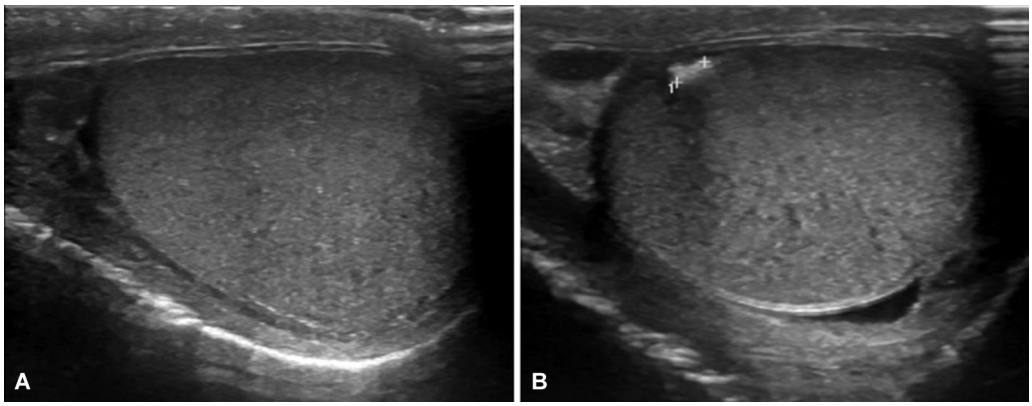

Fig. 4 Ultrasound of bilateral testes showing (**A**) normal right testis and (**B**) left testis with microcalcification.

immunohistochemistry in establishing the tumor lineage. The diagnostic challenge in this case stemmed from the rare anatomical location and nonspecific nature of gastrointestinal symptoms. A definitive diagnosis requires high clinical suspicion, detailed histopathological assessment, and immunohistochemical staining for markers, such as SALL4, AFP, and PLAP.[8] Imaging and tumor marker profiles further supported the extragonadal origin of the tumor. Differential diagnosis included lymphoma, adenocarcinoma, and GIST.[10] This case reinforces the importance of considering EGGCTs in the differential diagnosis of abdominal masses in young adults, even in the absence of testicular lesions. Early diagnosis and initiation of systemic chemotherapy can improve outcomes significantly.[4] EGGCT is classified under poor risk, stating poor outcomes as compared to gonadal GCT.[4] Reporting such rare presentations will aid in refining diagnostic algorithms and contribute to evidence-based management protocols for EGGCTs at atypical sites.[12]

Conclusion

This is a case report of an EGGCT that presented as an abdominal mass in a young male. Histopathological confirmation of the diagnosis is of utmost importance. Testicular assessment should always be performed in cases of EGGCT and monitoring of the normal testes. This strategy helps decrease the risk of metachronous testicular tumors. The early diagnosis of EGGCT can be life-saving and has a good prognosis. The treatment paradigms include platinum-based chemotherapy and surgery.

Patients' Consent

The authors certify that they have obtained all appropriate patient consent forms. In this form, the patient(s) has/have given their consent for his/her/their images and other clinical information to be reported in the journal. Patients understand that their names and initials will not be published, and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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None.

Conflict of Interest

None declared.

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