CASE REPORT

Case Report: Germ cell tumor presenting as cecal mass [version 1; peer review: awaiting peer review]

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Abstract

Extra gonadal germ cell tumors most frequently occur in the anterior mediastinum, retro-peritoneum, and pineal and suprasellar regions. The infrequency of its occurrence inside gastrointestinal tract makes it an arduous diagnostic challenge.

A 23 year old male with no significant past medical history presented to the emergency department with increasing abdominal pain, diarrhea, episodic vomiting for 3 weeks. Review of systems was positive for melena and shortness of breath on exertion. Fullness and irregularity along with tenderness was noted around the right iliac region.

CT scan (computed tomography) of the abdomen revealed a cecal mass with multiple metastases to liver, lungs and abdominal lymph nodes. Colonic endoscope was performed but it could not be advanced beyond the cecal mass. Biopsies from the mass were reported as poorly differentiated metastatic carcinoma. During the course of hospitalization, he developed symptomatic small bowel obstruction with perforation. Colonic resection was performed and histology showed Germ-Cell Tumor. Beta HCG level was 118789 IU/L suggestive of a non-seminomatous germ cell tumor. Ultrasound of the scrotum, MRI brain (magnetic resonance imaging) and CT scan of the chest did not reveal a primary tumor. Chemotherapy was started with Bleomycin, Etoposide and Cisplatin after which beta human chorionic gonadotropin (HCG) levels dropped dramatically. His hospital course got complicated with neutropenic sepsis with shock which progressed to multi-organ dysfunction and unfortunately, he succumbed to the disease burden.

This case demonstrates one of the rare presentations of extragonadal germ cell tumors and the diagnostic challenges associated with it. Very few cases have been reported in the literature, and none of them presented as a cecal mass. Early recognition of this presentation will help in reducing the tumor burden and the mortality associated with it, as germ cell tumors are highly susceptible to chemotherapy.

Keywords

germ cell tumor, cecal mass, chemotherapy
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Introduction
Extra gonadal germ cell tumors (EGCTs) are rare and account for 1–3% of all the gonadal tumors \(^1\)–\(^3\). Several mechanisms have been described, including possible migration of pluripotent stem cells, which convert to germ cells during embryonic development. The usual sites of EGCTs are mediastinum, retro-peritoneal organs, and pineal gland\(^1\)–\(^4\). Involvement of gastrointestinal tract is rare and is very unlikely in the absence of a primary elsewhere. Shogbesan et al. reported an EGCT presenting as duodenal mass with primary tumor identified in the testis. There are a few cases reported in literature without an identifiable primary in the gonads; none of which presented as a cecal mass\(^1\)–\(^2\).

Case presentation
A 23 year old Caucasian male who worked part-time as a waiter at a restaurant, with no significant past medical history presented during the fall of 2018 to the emergency department with increasing abdominal pain, diarrhea, and episodic vomiting for 3 weeks. Review of systems was positive for melena and shortness of breath on exertion. Family history was only relevant for an aunt who died from lung cancer. He had a five pack year smoking history and smokes marijuana socially. On admission, he was tachycardic and febrile, and exam revealed cachexia with bitemporal wasting. Fullness and irregularity in the right ileac fossa with associated tenderness was noted on palpation of the abdomen. See Figure 1 for full timeline.

Investigations
Computed tomography (CT) of the abdomen revealed a cecal mass with multiple metastases to liver, lungs and abdominal lymph nodes (Figure 2, Figure 3). Colonoscopy was performed, but the endoscope was unable to be advanced beyond the cecal mass. The tumor was poorly differentiated, and urothelial carcinoma was in the differential due to the positive GATA3 immunohistochemical staining. Liver biopsy reported an undifferentiated metastatic carcinoma. Beta HCG (human chorionic gonadotropin) level was 118,789 IU/L. After the elevated beta HCG level was detected, the colonic specimen was stained with the markers for germ cell tumors (beta HCG and PLAP) which was positive. Ultrasound of the scrotum was done to look for a primary tumor, which was negative. Magnetic resonance imaging (MRI) of the brain and a CT scan of the chest did not reveal any growth in the pineal gland or mediastinum, respectively.

Treatment
Chemotherapy was started with Bleomycin (30 units/wk IV on days 1, 8 and 15 regimen), Etoposide (100 mg/m\(^2\)/day IV) and Cisplatin (20 mg/m\(^2\)/day IV on days 1–5); given the high Beta HCG levels and deteriorating clinical status. Within 7 days of starting this regimen, beta HCG levels dropped dramatically to 31,163 IU/L. During the course of hospitalization the patient developed symptomatic small bowel obstruction with perforation, which was confirmed with a CT scan. Colonic resection was performed. The resected mass was identified to be germ cell tumor as it picked up beta HCG, SALL4, GATA3, OCT 4 as well as PLAP stains on histo-path exam (Figure 4, Figure 5).

Outcome and follow-up
The patient responded to chemotherapy initially with a drop in HCG levels; however, hospital course was complicated with tumor lysis syndrome and neutropenic sepsis, which progressed to multi-organ dysfunction. He required mechanical ventilatory support along with vasopressors for blood pressure support. As a result of deterioration, the family opted for comfort care measures. The patient expired 36 hours later. Even though his cancer burden was responding to chemotherapy, he unfortunately succumbed from complications of the treatment.

Discussion
EGCTs are considered metastatic from an occult or “burned out” gonadal cancer if a primary testicular tumor is not apparent. An ultrasound of the scrotum should always be performed to rule out a testicular tumor. A gonadal biopsy to rule out such tumor is not recommended\(^5\). The patient in the present study...
was found to have an EGCT in the cecum and colon. Similar to GCTs, EGCTs are sensitive to radiotherapy and chemotherapy.

Currently, common drugs used for chemotherapy include cisplatin, etoposide and bleomycin. Our patient received these medications along with surgery which helped in reducing his tumor burden. The International Germ Cell Cancer Collaboration Group (IGCCCG) places the patients into poor prognosis category if they are found to have a primary tumor or visceral metastases elsewhere apart from the lungs and retro peritoneum. The 5-year survival rate in such patients is 48%.

This case demonstrates one of the rare presentations of extra-gonadal germ cell tumors and the diagnostic challenges associated with it. Early diagnosis entails high index of suspicion, especially in young males presenting with tumor of unknown origin. Early recognition of this presentation will help in reducing the tumor burden and the mortality associated with it. It also allow for early treatment, which usually entails surgery (for accessible tumors) along with chemotherapy. However, our knowledge about these tumors and their presentations is still limited, and as seen in this patient, treatment can result in complications well. This limitation warrants the need for studies on EGCTs and their management.

Conclusion/learning points/take home messages

- Gonadal tumors should be among the differentials in patients presenting with abdominal mass, especially in younger adults.

- The importance of this case is to identify a constellation of symptoms, obscurities, diagnostic difficulties, adverse effects of treatment of an extra gonadal germ cell tumor.

- The authors would encourage testing with Beta HCG in young males presenting with a tumor of unknown origin.
• The authors would also encourage a multidisciplinary approach with surgery and early initiation of chemotherapy in such patients to control the disease burden.

• This report also increases awareness that one should be watchful for complications after implementing treatment with chemotherapy, as early detection of complication may help prevent negative outcomes.

Consent
Written informed consent for publication of their clinical details and clinical images was obtained from the parent of the patient.

Data availability
Underlying data
All data underlying the results are available as part of the article and no additional source data are required.

References
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