

Late Recurrence of Non-Seminomatous Germ Cell Tumor via Somatic Transformation to Adenocarcinoma: A 30-Year Diagnostic Challenge

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Abstract

Importance

Late recurrence (>2 years) of germ cell tumors (GCT) is rare, with this case being one of the longest late recurrences reported in literature.

Case Presentation

A 53-year-old male presented with a pelvic mass 30 years after initial treatment for testicular cancer. While initial pathology and elevated carcinoembryonic antigen (CEA) suggested a colorectal primary, fluorescence in situ hybridization (FISH) for isochromosome 12p confirmed somatic transformation of a GCT.

Conclusions

This case highlights the necessity of maintaining suspicion for GCT recurrence regardless of the latency period and underscores the utility of cytogenetic testing in discordant diagnostic presentations.

Introduction

Late relapse of GCT is defined as a recurrence occurring more than 2 years after a complete response to initial therapy. In rare instances, these tumors undergo somatic transformation, wherein teratoma components evolve into histologies typically resistant to standard platinum-based GCT regimens, such as sarcomas or adenocarcinomas. Because adenocarcinoma transformation can mimic gastrointestinal (GI) malignancies, it can lead to significant diagnostic delays.

Case Presentation

History

A 53-year-old male presented in May 2020 with a pulmonary embolism, followed by hematuria in June. His history was significant for right-sided testicular cancer in the 1990s, treated with orchiectomy and retroperitoneal lymph node dissection (RPLND) (the histopathological evaluation from the RPLND confirmed mature teratoma with extensive necrosis in the pelvic soft tissue, alongside metastatic teratoma identified in two out of ten (2/10) retroperitoneal lymph nodes). The operative note mentioned a tumor in the right obturator fossa that was unresectable due to proximity to the nerve. The patient had been lost to follow-up since 2003.

Diagnostic Workup

Imaging revealed a large right pelvic mass causing hydronephrosis and iliac vein occlusion. A biopsy of the pelvic sidewall mass showed adenocarcinoma of probable colorectal origin with intense CDX2 and CK20 staining. However, a subsequent colonoscopy was negative, conflicting with the biopsy findings. Laboratory studies showed a significantly elevated CEA (21.1 ng/mL, later rising to 475 ng/mL), while β -hCG and AFP remained near normal.

Management and Outcome

The patient received neoadjuvant capecitabine and intensity-modulated radiation therapy (IMRT), followed by extensive surgical resection. The turning point occurred during pathology review of the resected mass. Although the specimen showed metastatic adenocarcinoma, the patient's history of prior pelvic teratoma prompted FISH testing.

This confirmed the presence of isochromosome 12p, establishing that the mass originated from his previous germ cell tumor (GCT). Postoperatively, the patient developed peritoneal carcinomatosis and pulmonary nodules. Despite initiating FOLFOX, his condition declined, and he entered hospice in March 2021.

Discussion

This case illustrates a 30-year latency period. While most late relapses occur at a median of 5 to 10 years,¹ and the majority within 20 years,² a thirty-year dormancy is rare. Similar therapeutic and diagnostic dilemmas have been documented even at shorter intervals; for instance, Coca et al. reported a case of metastatic adenocarcinomatous deposits presenting 12 years after initial treatment for an immature testicular teratoma, which similarly failed local radiation and required palliative chemotherapy.³ The presentation—characterized by adenocarcinoma histology, elevated CEA, and near-normal markers—could have led to a misdiagnosis of a primary GI cancer. The diagnosis was confirmed by FISH for i(12p). This marker is present in virtually all GCTs and up to 89% of somatic-type malignancies arising from them.⁴⁻⁵ Somatic-type malignancies do not respond to cisplatin-based GCT regimens, and NCCN guidelines recommend that for teratoma with somatic-type malignancy, histology-directed therapy should be considered.⁶

Conclusion

- Late-onset pelvic masses in patients with a history of GCT should be considered recurrence until proven otherwise.
- Cytogenetic testing for i(12p) is a critical diagnostic tool when histology and clinical history are discordant.
- Long-term oncologic surveillance beyond the standard 10-year window may be considered in high-risk patients.

Conflict(s) of Interest

The authors declare no conflicts of interest.

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Ethical Statements

This case report has no prior research intent, and the University of Utah IRB does not require review if the report does not meet the regulatory definition of research.

Informed Consent

Consent obtained from patient and family.

Data Availability Statement

This article does not contain original data.

Declaration of AI Use in Scientific Writing

Not applicable. AI tools were not used to generate scientific content, interpret data, or influence the conclusions of this review.

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Editor Disclaimer

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