CASE REPORTS & REVIEWS
https://doi.org/10.63838/001c.154978 ISSN 2819-2842

Metastatic PEComa successfully treated with pembrolizumab: A case report and review of the literature

Authors:
John Loggie[1] MD, MSc; Mathieu Castonguay[1] MD; Kelly Dakin Haché[1] MD, PhD; Bruce Colwell[2] MD; Michael Carter[1] MD, PhD.

Affiliations:
[1]Department of Pathology, Dalhousie University, Halifax, NS, Canada.
[2]Department of Medicine (Oncology), Dalhousie University, Halifax, NS, Canada.
*Author for correspondence (john.loggie@nshealth.ca).

The authors declare that there are no undisclosed conflicts of interest regarding the publication of this paper. All authors have provided Clockwork Communications Inc. with non-exclusive rights to publish and otherwise deal with or make use of this article, and any photographs/images contained in it, in Canada and all other countries of the world.

Keywords:
Immunotherapy, microsatellite instability, Perivascular Epithelioid Cell Neoplasms, biomarkers, DNA mismatch repair.

ABSTRACT

Perivascular epithelioid cell tumours (PEComas) are rare mesenchymal neoplasms with the potential for malignant biological behavior. Patients with metastatic PEComas may benefit from effective biomarker-driven treatments. Genetic alterations associated with mismatch repair deficiency (MMRd) and microsatellite instability-high (MSI-high) are rarely identified in PEComas but have emerged as successful predictors of response to pembrolizumab in a variety of solid tumours. Here, we report a case of a 29-year-old female who presented with an enlarging lesion in her left femoral neck, which was resected and diagnosed as malignant PEComa. Two metastatic pulmonary nodules then appeared and were resected. After another large metastatic lesion appeared in her left lung, the patient elected to start systemic therapy. Next-generation sequencing performed on one of the metastatic tumours did not identify actionable genetic variants, however several microsatellite sites were noted to be unstable, manifesting as small duplications and deletions within mononucleotide repeats. Subsequent immunohistochemical evaluation for MMR expression was performed, showing subtotal expression loss of MLH1 and PMS2, and MSI testing revealed the tumour to be MSI-high. These findings suggested the use of the anti-PD-1 agent pembrolizumab in a patient who would otherwise not have been able to access this medication in Canada at the time of her diagnosis. The patient was referred to medical genetics where she was found to not have Lynch syndrome. Sixteen months after initiating pembrolizumab, the patient continues to show reduction in the size of her metastatic lung lesion. This case is the first to our knowledge to describe the successful use of immunotherapy for metastatic MSI-high PEComa arising in a patient without Lynch syndrome.