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Genetic and Developmental Disorders

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Cannonball Opacities in the Emergency Department: A Case of Recurrent Malignant Schwannoma in a Long-Term Survivor

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INTRODUCTION: Malignant peripheral nerve sheath tumours (MPNSTs), or malignant schwannomas, are extremely uncommon soft-tissue sarcomas of neural origin. They behave aggressively, are rapidly progressive and typically portend a poor prognosis.

CASE PRESENTATION: A 50-year-old lady with neurofibromatosis type 1 (NF-1), smoker (19 pack-year history), presented to the emergency department (ED) due to difficulty breathing at rest with progressive worsening over the previous few weeks. The patient had recently (two weeks earlier) received her third course of chemotherapy (CYVADIC protocol: cyclophosphamide, vincristine, doxorubicin, and dacarbazine) for recurrence of MPNST. Urgent chest X-ray in the ED revealed cannonball opacities in both lungs, predominantly in the bases. These radiological changes were consistent with pulmonary metastases from the MPNST, but mimicked those seen in primary metastatic lung cancer. The patient was in the terminal stage of disease and succumbed a week later.

DISCUSSION: Von Recklinghausen disease, or NF-1, is among the most common genetic disorders.¹ About half of the cases of MPNSTs occur in the context of NF-1.² This is a unique case of NF-1 and recurrence of MPNST in a long-term survivor. Approximately 17 years earlier, the patient had undergone tumour reduction and adjuvant radiotherapy for MPNST of the left forearm. Seven months prior to this ED presentation, recurrence of MPNST in the same forearm required repeat surgical reduction. Three months later, her left arm was amputated at the middle of the humeral shaft due to disease relapse and chemotherapy was subsequently initiated.

CONCLUSIONS: This report serves to: 1) raise awareness about this exceptionally rare pathology, 2) illuminate the achievement of long-term survival in a NF-1 patient and her dramatic ED presentation due to lung metastases and 3) highlight the importance of maintaining regular follow-up visits to ensure timely intervention for recurrent and potentially metastatic disease.

Reference #1: Farid M, Demicco EG, Garcia R, Ahn L, Merola PR, Cioffi A, Maki RG. Malignant peripheral nerve sheath tumors. *Oncologist* 2014 Feb;19(2):193-201. doi: 10.1634/theoncologist.2013-0328.

Reference #2: Ducatman BS, Scheithauer BW, Piepgras DG, Reiman HM, Ilstrup DM. Malignant peripheral nerve sheath tumors. A clinicopathologic study of 120 cases. *Cancer* 1986;57: 2006-21.

DISCLOSURE: The following authors have nothing to disclose: Katherina Sreter, Gorana Mirosevic
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