Malignant *TFE3*-rearranged perivascular epithelioid cell neoplasm (PEComa) presenting as a subcutaneous mass

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Abstract

Perivascular epithelioid cell neoplasms (PEComas) are a group of mesenchymal tumors with concurrent melanocytic and myogenic differentiation. Although many cases are sporadic, PEComas can be associated with tuberous sclerosis. A distinct subset of visceral and deep-seated PEComas has been shown to carry *TFE3* fusions. Herein, we describe a rare case of malignant PEComa with *TFE3* gene rearrangement in a 32-year-old woman without tuberous sclerosis. The patient presented with a 12-year progressive subcutaneous mass in the left thigh. Grossly, it demonstrated an 8.5 cm lobulated tan mass with focal hemorrhage. Microscopically, the tumor was composed of mainly epithelioid cells with abundant clear and eosinophilic cytoplasm, arranged in infiltrative nested growth pattern. Many areas also showed scattered mitotic figures and marked necrosis. Immunohistochemistry revealed positive staining for S-100, SOX-10, Melan-A, HMB45, and TFE3. However, the tumor cells were negative for SMA, desmin, and MITF. By fluorescence *in situ* hybridization, the presence of *TFE3* gene rearrangement was demonstrated. Despite the wide surgical resection and adjuvant radiation therapy, the patient subsequently developed a pulmonary metastasis. Malignant PEComa can be present as a primary subcutaneous mass and should be considered in the differential diagnosis of tumors with (myo)melanocytic differentiation. To our knowledge, this is the first reported case of primary subcutaneous malignant PEComa with molecular confirmation of *TFE3* gene rearrangement.

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