CASE REPORT

PEComa (Pure Epithelioid Angiomyolipoma) of kidney

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ABSTRACT

A 60-year-old male patient presented with complaints of loss of appetite and bloated abdomen of two weeks duration. The radiological examination revealed a large mass involving left kidney. With a clinical and radiological diagnosis of renal cell carcinoma, radical nephrectomy was done. Histopathological examination revealed a neoplasm composed purely of epithelioid cells. Based on histopathological and immunohistochemical findings a diagnosis of Epithelioid Angiomyolipoma of kidney was made. We are reporting this case because of the rarity of this variant of angiomyolipoma and to highlight the diagnostic difficulties of this entity when compared to the more common classic angiomyolipoma.

Key words: PEComa, kidney, epithelioid, angiomyolipoma

INTRODUCTION

Perivascular epithelioid cell tumors (PEComas) are rare potentially malignant mesenchymal neoplasms composed of histologically and immunohistochemically distinctive perivascular epithelioid cells.1 PEComas express melanocytic and myogenic markers such as HMB-45 and Actin.2 Patients presenting with PEComas may have associated tuberous sclerosis complex, an autosomal dominant genetic disease characterized by loss of TSC1 (9q34) or TSC2 (16p13.3) genes. Tuberous sclerosis presents with mental retardation, seizures, subependymal giant cell astrocytoma, angiomyolipoma, cutaneous angiofibroma, cardiac rhabdomyoma. The common PEComas include Angiomyolipoma (AML), Clear Cell Sugar Tumor (CCST) of lung and extra pulmonary sites and Lymphangioleiomyomatosis. Epithelioid angiomyolipoma is a variant of AML composed purely of epithelioid cells.

CASE REPORT

A 60-year-old male presented with complaints of loss of appetite and bloated abdomen of fifteen days duration. Radiological examination showed left renal mass measuring 7x5 cm confined to the kidney involving upper and mid poles. No calcification was noted within the mass. There was extensive tumor thrombus in left renal vein and inferior vena cava with extension up to right atrium. With a clinical and radiological diagnosis of renal cell carcinoma patient was taken up for surgery. A left radical nephrectomy with thrombectomy was performed. The radical nephrectomy specimen measured 10x7x8 cm. Cut section showed a large grey-white, brownish growth measuring 8x5 cm with areas of hemorrhage and necrosis involving mainly upper pole of kidney. Normal renal tissue was identified at lower pole. (Fig. 1)

Figure 1, Cut surface of kidney showing large grey white growth with hemorrhagic and necrotic areas.
Microscopy showed an infiltrating neoplasm composed purely of sheets of large epithelioid cells with abundant eosinophilic cytoplasm and vesicular nucleus with prominent nucleoli (Fig. 2). Multinucleated cells and large cells with vacuolated cytoplasm resembling ganglion cells were also noted. Areas of necrosis were also seen. The tumor cells were positive for HMB-45, SMA and S100 and negative for cytokeratin, vimentin, desmin, CD10 and MUM-1. Based on histopathological and immunohistochemical findings a diagnosis of epithelioid angiomyolipoma was made.

**DISCUSSION**

Epithelioid angiomyolipoma is a sub type of AML composed purely of epithelioid cells. Epithelioid AMLs were described by Pea et al in 1998 and are characterized by the absence of adipocytes and abnormal vessels seen in the classical AML. This subtype of AML is potentially malignant and may present with recurrences and metastasis. Epithelioid AML is thus a neoplasm composed of purely epithelioid cells arranged in sheets, without adipocytes and abnormal blood vessels. The co-expression for melanocytic and muscle markers along with histopathological features help in reaching a diagnosis. The sclerosing variant of epithelioid angiomyolipoma was described by Matsuyama et al. The main differential diagnoses to be considered histopathologically are malignant melanoma and variants of renal cell carcinoma especially rhabdoid variant. PEComas and malignant melanoma share common morphological, immunohistochemical, and ultra structural features, such as epithelioid cell morphology and melanocytic immunophenotype. Melanocytic markers commonly expressed in PEC tumors include HMB-45, Melan-A/MART-1, tyrosinase, microphthalmia transcription factor (MITF) and occasionally S100. Given this morphological and immunophenotypical overlap, the differential diagnosis between a PEComa and malignant melanoma can present a challenge. Recent studies have shown that MUM-1, a known lymphocyte marker shows positive immunostaining in nevi and melanomas. Its expression in PEComas and clear cell sarcoma was studied by Ferenczi K et al, which showed negative staining in AMLs. Our case also showed negativity for MUM-1, thus ruling out the possibility of melanoma. In our case there was positive staining with melanocytic markers, HMB-45 and S100 and muscle marker SMA.

The renal cell carcinomas with rhabdoid features which histologically presents as sheets and clusters of variably cohesive large epithelioid cells with vesicular nuclei and prominent nucleoli is another close differential to be considered. Negative staining with vimentin, cytokeratin and CD10 helped to rule out renal cell carcinoma in our case.

In a study of 41 cases of epithelioid AMLs by Nese et al clinicopathologic parameters associated with disease progression (recurrence, metastasis, or death due to disease) included associated tuberous sclerosis complex, necrosis, metastasis at diagnosis, tumor size more than 7 cm, extra renal extension and/or renal vein involvement and carcinoma-like growth pattern. Tumors with less than two adverse prognostic parameters were considered to be low risk for progression tumor, with only 15% having disease progression. Tumors with two to three adverse prognostic parameters were considered to be “intermediate risk,” with 64% having disease progression. Tumors with more than four or more adverse prognostic parameters were considered to be high risk, with all patients having disease progression. Of tumors with three or more adverse prognostic parameters, 80% had disease progression. An exact logistic regression analytic model showed that only carcinoma-like growth pattern and extra renal extension and/or renal vein involvement were significant predictors of outcome. The extension of tumor into IVC as seen in our case was also observed in a previous report by Park et al.

There has been previous case reports of epithelioid angiomyolipoma presenting with metastasis to lymph nodes, liver, bone, adrenal etc. However Meng et al is of the opinion that lymph node involvement and tumor emboli in renal vein represent multifocal tumor. They are also of the opinion that definite evidence of malignancy requires demonstration of distant metastasis. According to the clinicopathological parameters pro-
posed by Nese et al., our patient at the time of diagnosis had several adverse prognostic factors like involvement of renal vein, tumor size more than seven centimeters, presence of necrosis etc. Our patient presented with disease progression and metastasis to liver and lung within six months of surgery. Metastatic disease is associated with a bad prognosis.

REFERENCES