Atypical presentation of renal medullary carcinoma: A case report and review of the literature

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Introduction

Renal medullary carcinoma (RMC) is a rare renal neoplasm and highly aggressive. It was initially described by Davis et al., in 1995 and revealed almost exclusively found in young black individuals with sickle cell trait. It has an extremely poor prognosis with most cases diagnosed later in the disease course, with metastasis often present at the time of initial presentation. Median time of survival is 4 months following surgery.1

The reported patients were associated with sickle cell hemoglobinopathy, mainly with sickle cell trait and less frequently without sickle cell disease. The prognosis is very poor because the tumor is very aggressive and resistant to conventional chemotherapy.

The histopathological features of RMC include epithelial cells with reticular, adenoid cystic aplasia, and prominent inflammation.

Pathologically, RMC arises from the renal medulla, grows rapidly in an infiltrative pattern, and invades the renal sinuses. Previous studies on RMC have documented the pathological and clinical features of this rare form of renal carcinoma. However, there are limited studies on RMC focusing on computed tomography (CT) imaging findings.2

Case report

A 68-year-old white male, known case of diabetes, hypertension and no past medical history of sickle cell trait presented with Right-sided loin pain, without hematuria. Ultrasound examination showed a right renal mass. Abdominal computed tomography (CT) scan confirmed the sonographic findings demonstrating a 9 cm mass in the lower pole of the right kidney, with enlargement of regional lymph-nodes and multiple bilateral lung metastases (Fig. 1). His hemoglobin was 14 g/dl, white blood cell count 7.5103/ml and creatinine 1mg/dl. Serum electrolytes and liver function test values were within normal. After the diagnosis, electrophoresis was done for the diagnosis of sickle cell hemoglobinopathy. The result was negative for sickle cell hemoglobinopathy. Physical examination was unremarkable. Ultrasound-guided true cut biopsy was performed for the renal mass, which microscopically was consistent with renal medullary carcinoma in which tumor cells are positive for PAX8, cytokeratin AE1/AE3, and cytokeratin 7. They are negative for RCC immunostain BAF47 (INI1) (Fig. 2).

The patient was started on Sutent treatment. A repeat CT scans during one-year follow up showed that the bilateral pulmonary nodules had partial regression and stable right renal mass.

Discussion

What renders our case unique is the occurrence of this condition in a 68-year-old white man, without sickle cell trait, making the diagnosis of RMC less likely in our differential.

RMC is exceedingly rare and accounts for less than 1% of all renal cancers. It typically presents in young patients and the male-to-female ratio is 2:1.3 The median age for conventional renal cell carcinoma is 64 years of age, according to the National Cancer Institute. Review of the Literature by Madhumati et al. found 262 cases (including case reports and series). This occurs almost exclusively in patients of African origin and 88–98% of cases possess sickle cell trait.4 The median age of the reported cases in the Davis study was 21 years (range 11–39 years

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RMC has been reported in Hispanics/Brazilians and a few Caucasians and nearly all RMC patients have sickle cell trait or, rarely, sickle cell disease unlike our patient who does not have either sickle cell trait or sickle cell disease in addition to advanced age at presentation. In adults, a 2:1 male-to-female ratio has been observed in patients with RMC. In children, the male predominance is even greater. Initial clinical presentation varies, but most patients present with symptoms; pain and hematuria are the most common. The right kidney is the affected kidney in more than 75% of cases. Metastasis at presentation is extremely common. The most common sites of metastases are the regional lymph nodes, adrenal glands, lung, liver, inferior vena cava, and peritoneum.

RMC tumors usually express cytokeratin AE1/AE3, low molecular weight cytokeratin, vimentin, hypoxia-inducible factors (HIF), and vascular endothelial growth factor (VEGF). Variability in the expression rate of high molecular weight cytokeratin is common and the histopathology of our patient tumor was compatible with these markers.
RMC is a highly aggressive cancer with an extremely poor prognosis. Mean survival time of less than 1 year is seen in most cases. Treatment is difficult. Neither chemotherapy nor radiation therapy has been found to be particularly efficacious.

Despite the lack of available prospective evidence, Our patient was treated with trial of a VEGF-targeted tyrosine kinase inhibitor (sunitinib) and at one year follow up still alive with significant clinical and radiological response.

Statement of ethics

The authors have no ethical conflicts to disclose.

The patient provided permission to publish these features of his case and the identity of the patient has been protected.

Disclosure statement

The authors declare that they have no relevant financial interests.

Appendix A. Supplementary data

Supplementary data to this article can be found online at https://doi.org/10.1016/j.eucr.2018.09.023.

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