



Cardiac Metastasis of Medullary Phenotype Unclassified Renal Cell Carcinoma: A Case Report

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Abstract

Renal medullary carcinoma is an uncommon and aggressive variant of renal cell carcinoma (RCC), accounting for less than 0.5% of all RCC cases. It predominantly affects young individuals with sickle cell hemoglobinopathies. However, as seen in this case, unclassified medullary phenotypic RCC without hemoglobinopathy can also occur. This case report discusses a patient diagnosed with unclassified RCC of the medullary phenotype, exhibiting metastases to the left ventricle and liver. Diagnosis was confirmed through percutaneous biopsy, revealing loss of SMARCB1 expression along with positive vimentin and OCT3/4 staining. Treatment included chemotherapy with carboplatin and paclitaxel, followed by second-line therapy with cabozantinib. Left ventricular metastasis and cardiac lesions are rare but poor prognostic features of RCC. This case, one of the few documented in the literature, underscores the importance of a multidisciplinary approach to diagnosis and treatment.

Keywords: Medullary carcinoma, cardiac metastasis, medullary phenotype unclassified renal cell carcinoma, case report

Introduction

Renal cell carcinoma (RCC) represents the most common type of primary kidney cancer, accounting for approximately 80-85% of all renal neoplasms. The incidence is 4-7 per 100,000 individuals. Approximately 25-30% of people exhibit metastatic illness upon diagnosis, with the majority of instances being incidentally identified (1).

Renal medullary carcinoma (RMC) is an extremely rare and severe variant, comprising less than 0.5% of RCCs. It predominantly impacts young people with sickle cell hemoglobinopathies (2). Tumors that resemble RMC but arise in the absence of hemoglobinopathy have historically been classified as unclassified RCC with a medullary phenotype (3). Metastases from these tumors predominantly affect the lungs, liver, and lymph nodes, although cardiac metastases are exceptionally rare, with only a limited number of cases documented (4). This case report describes a patient diagnosed with unclassified RCC of the medullary phenotype, with metastases to the liver and left ventricle.

Case Report

A 26-year-old male presented to the emergency department with a 17-kg weight loss over the past several months, accompanied by fever, night sweats, tachycardia, nausea, and vomiting in the preceding month. His medical history included cleft lip repair and schizoaffective disorder. The physical examination showed no costovertebral angle tenderness or palpable abdominal mass. In contrast to the majority of RMC cases linked to sickle cell trait (5), our patient revealed no hemoglobinopathy. Transthoracic echocardiography identified a pericardial effusion and a solid-cystic mass measuring 7×3.5×8.5 cm on the left ventricular wall. Cardiac magnetic resonance imaging revealed a malignant lesion consistent with metastasis (Figure 1). Contrast-enhanced abdominal computed tomography (CT) revealed a heterogeneous tumor in the left kidney measuring 7.8×6.2×6.2 cm (Figure 2), as well as metastatic lesions in the liver (Figure 3) and retroperitoneal lymphadenopathy. The percutaneous tru-cut biopsy of the renal tumor confirmed a diagnosis of undifferentiated RCC with a medullary phenotype.

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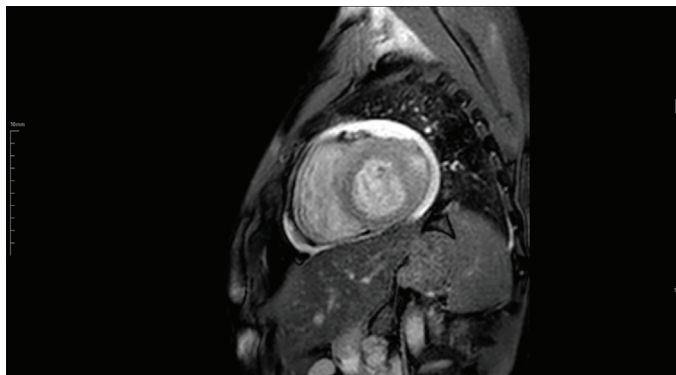


Figure 1. Metastatic lesion in the left ventricular wall on T1-weighted image in cardiac contrast-enhanced magnetic resonance imaging



Figure 2. Contrast-enhanced abdominal computed tomography showing the left renal tumor

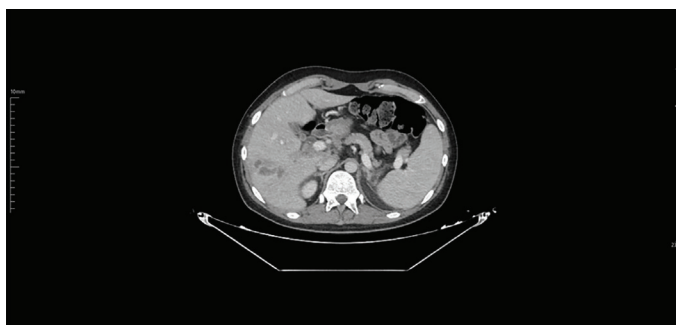


Figure 3. Contrast-enhanced abdominal computed tomography showing metastatic liver lesions

Histopathological analysis demonstrated nuclear SMARCB1 (INI1) expression and positive immunostaining for vimentin and OCT3/4. Karyotype analysis was advised for the patient. Molecular analysis indicated the absence of an *SMARCB1* gene mutation. Tissue-based karyotype analysis could not be conducted due to technical limitations. For advanced staging, the patient underwent fluorodeoxyglucose (FDG) PET imaging, which revealed that the primary renal mass had metastasized to the liver, abdominal lymph nodes, and heart. According to the IMDC risk assessment, the patient would benefit from cytoreductive nephrectomy to reduce tumor burden before systemic therapy; however, the patient declined the procedure. Following a medical oncology consultation,

chemotherapy with carboplatin and paclitaxel was commenced. The patient underwent four cycles of carboplatin-paclitaxel treatment. After completion of chemotherapy, an 18F-FDG PET/CT restaging scan was performed, revealing extensive progressive disease with a mixed metabolic response. Subsequently, cabozantinib monotherapy was commenced as a second-line treatment. While undergoing cabozantinib treatment, the patient presented with a necrotic lesion on the right foot, necessitating a tru-cut biopsy. Histopathological assessment indicated metastatic disease. The immunohistochemical examination of the biopsy samples revealed PD-L1 expression, resulting in cessation of the current treatment. The patient then commenced third-line therapy with nivolumab. A restaging imaging evaluation was scheduled following the third cycle. Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

Discussion

RMC is an uncommon but highly malignant tumor. The majority of patients present at an advanced stage with extensive metastatic disease. This case is remarkable because of the absence of an underlying hemoglobinopathy, despite most cases being linked to sickle cell hemoglobinopathy. RMC has been observed more commonly in males, predominantly originates from the right kidney (2). The presence of left renal involvement alongside a cardiac metastasis in the left ventricle is a rare, although clinically important, observation reported in the literature.

The diagnosis of RMC is strongly associated with loss of nuclear SMARCB1 (INI1) expression (6). This feature is also characteristic of SMARCB1/INI1-deficient medullary-like renal cell carcinoma, historically referred to as unclassified RCC with a medullary phenotype (3,6). The absence of SMARCB1 expression, coupled with positivity for OCT3/4 and vimentin, supported medullary differentiation in this patient. The literature indicates a very poor prognosis for RMC, particularly in patients presenting with metastatic disease (7).

Cardiac metastases originating from RCC are extremely uncommon, with left ventricular involvement regarded as extraordinary. RCC may invade the right atrium by direct extension through the inferior vena cava. The occurrence of left ventricular metastases, as observed in this case, may indicate hematogenous or lymphatic spread and is typically associated with poor prognosis (5).

Cahill et al. (5) reviewed RCC with cardiac metastases and highlighted the diagnostic and therapeutic challenges associated with this uncommon presentation. Cardiac metastases may present significant challenges to clinicians in both diagnostic and therapeutic settings.

Cardiac metastases are primarily documented at autopsy, whereas intraventricular involvement in living individuals is exceptionally uncommon. Despite advances in imaging techniques that improve identification of atypical metastatic locations, the literature remains sparse regarding cases of renal medullary cancer with both left-heart involvement and synchronous multiorgan metastases (5).

Davis et al. (4) provided the seminal description linking RMC with sickle cell nephropathy. Subsequent studies have confirmed the strong association between RMC and sickle cell trait while also characterizing its distinctive molecular and immunohistochemical profile (2,6).

Platinum-based chemotherapy remains an important systemic treatment approach for RMC, particularly in metastatic disease (2,7). However, as demonstrated in the current case, clinical responses may be limited. Targeted therapies such as cabozantinib have been used in individual patients, although evidence remains limited for this rare tumor (2).

The role of nephrectomy in RCC remains controversial and needs to be considered in the context of performance status, disease burden, and response to systemic therapy. In the present case, the patient refused surgery, and despite systemic treatment, the development of cardiac and peripheral metastases was noted.

Unclassified RCC with a medullary phenotype constitutes an uncommon and aggressive tumor characterized by significant metastatic potential. Rare metastatic sites, such as cardiac involvement, emphasize the systemic nature and poor prognosis of this condition, underscoring the need for a multidisciplinary approach to diagnosis and treatment. In contrast to the few occurrences documented in the literature, the present case is notable for its involvement of the left heart and for the lack of an underlying hemoglobinopathy. Documenting and molecularly characterizing these cases will be essential for improving understanding and therapeutic approaches to this complex disease.

Ethics

Informed Consent: Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

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Publication: The results of the study were not published in full or in part in form of abstracts.

Contribution: There is not any contributors who may not be listed as authors.

Footnotes

Authorship Contributions

Concept: E.K., F.S., Design: T.S., Data Collection or Processing: E.K., Analysis or Interpretation: M.Y.A., F.S., Literature Search: M.Y.A., T.S., Writing: M.Y.A.

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