Case Report

# Renal medullary carcinoma with an ophthalmic metastasis

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## Abstract

Renal medullary carcinoma (RMC) is a rare, aggressive primary renal malignancy that classically occurs in adolescent males with sickle cell trait and universally presents with metastatic disease at presentation. We report a case of medullary carcinoma in a young man with likely ophthalmic metastasis. We also review relevant literature available to date. The patient is a 20-year-old African-American male with a past medical history significant to for sickle cell trait who presented to the University Medical Center with cough and the right eye pain for 1 month as well as painless gross hematuria for 1 week. A chest and abdominal computed tomography showed a 7 cm hypodense right renal mass with bilateral hilar adenopathy, and multiple bilateral pulmonary nodules. A renal biopsy was performed and showed RMC. Ophthalmic exam revealed the right retinal hemorrhage concerning for a metastatic lesion. Palliative chemotherapy was offered to the patient, however, he and his family chose to enroll in hospice care considering his poor prognosis. He subsequently passed away 33 days after presentation. To our knowledge, there is only one other case of ophthalmic metastasis in a patient with metastatic RMC. Thus, we present this case to contribute to current literature regarding orbital metastasis in this largely fatal disease.

**Key Words:** Ophthalmic metastasis, renal medullary carcinoma, sickle cell trait

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## INTRODUCTION

Since its description by Davis *et al.* in 1995 renal medullary carcinoma (RMC) has remained a highly aggressive tumor that almost exclusively plagues young individuals who carry the sickle cell trait. The mean age at diagnosis among these patients is 22 with ages ranging between 11 and 39, however, two cases have been reported in an 8- and 69-year-old.[1,2] The survival rate ranges between 3 and 52 weeks after diagnosis.[1] Patients generally report gross hematuria with flank pain at presentation.[1-4] With the exception of two reported cases patients uniformly have regional lymphatic or distant metastasis upon presentation.[3,5,6] The most common sites of distant metastasis include liver, lungs, bones, inferior vena cava, and omentum.[2,3,6,7] To our knowledge, there is only one case of metastasis to the orbit in a patient with metastatic RMC.[7] We present a case of a young man with RMC, who was found to have metastasis to the right orbit.

## CASE REPORT

The patient is a 20-year-old African-American man with a past medical history significant for sickle cell trait who presented

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to University Medical Center complaining of blurred vision with pain at his right eye as well as a dry cough with worsening dyspnea for 1 month. More recently, the patient reported multiple episodes of gross painless hematuria for 1 week.

On examination, the patient's lungs were clear to auscultation bilaterally, he had neither costovertebral angle tenderness nor abdominal pain upon palpation. Ophthalmic examination revealed visual acuity of 20/20 in the left eye and 20/200 in the right. The optic disc was found to be obscured by exudative retinal detachment with yellow turbid subretinal fluid at the periphery concerning for mass. The remainder of his examination was benign. His laboratory values were significant only for a white blood cell count of 11.59.

A computed tomography (CT) scan of the abdomen revealed a single poorly circumscribed centrally located hypodense right renal mass, measuring 5.6 cm × 6.8 cm [Figure 1]. CT scan of the chest showed diffuse, bilateral pulmonary nodules with extensive bilateral hilar, and anterior mediastinal adenopathy.

Ultrasound-guided core needle renal biopsy of the mass was performed. Five cores were obtained, and light microscopy showed poorly differentiated epithelioid cells forming nests, columns, and gland-like spaces, with surrounding reactive fibrotic tissue consistent with desmoplastic stroma. Cytoplasm ranged between clear, eosinophilic, and vacuoles [Figure 2]. Pleomorphic nuclei with prominent nucleoli and sickled erythrocytes were present. Areas of necrosis were visualized along with scattered neutrophils among the tumor cells. Immunohistochemistry analysis showed positive reactivity to epithelial membrane antigen, low molecular weight cytokeratin 7 (CK 7), CK AE1/AE3, and vimentin. The tumor was also focally positive in cytoplasmic vacuoles for mucicarmine stain. Subsequent magnetic resonance imaging (MRI) of the head showed a 1 cm mass with the abnormal signal at the posterior right globe that enhanced with gadolinium [Figures 3 and 4].

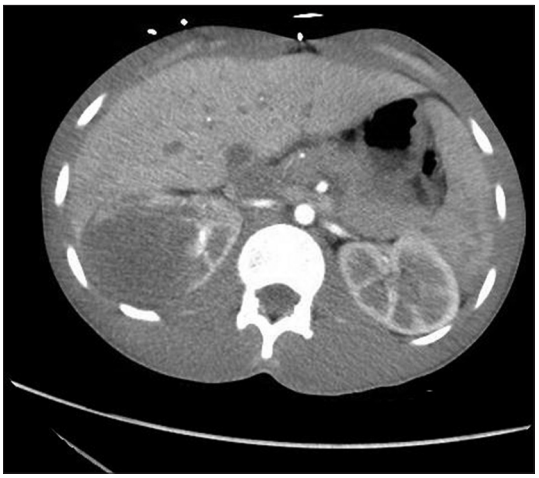

**Figure 1:** Computed tomography scan of abdomen with contrast showing the right renal mass

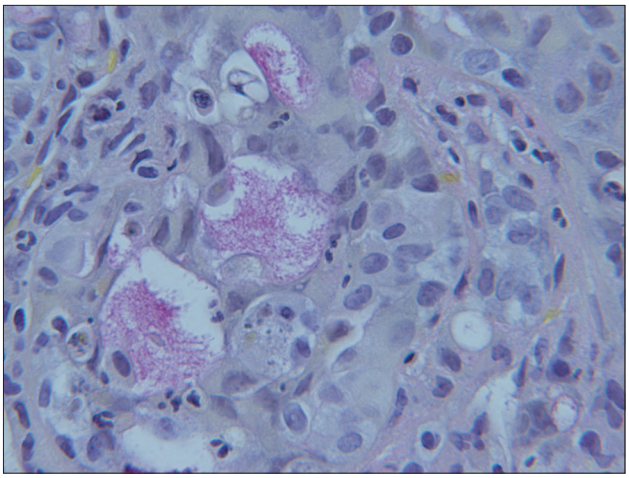

**Figure 2:** High-powered magnification of pathologic slide from the right renal mass biopsy

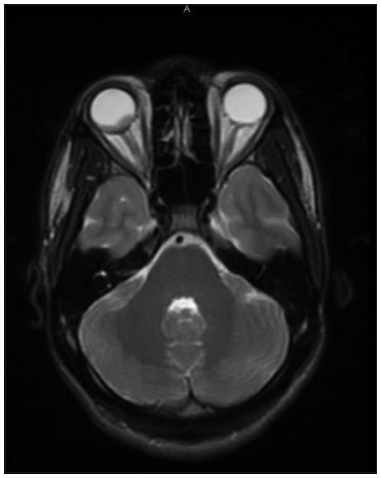

**Figure 3:** Axial view of head magnetic resonance imaging demonstrating a mass at the posterior right globe

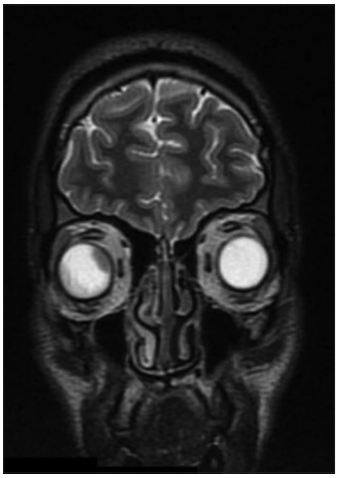

**Figure 4:** Coronal view of head magnetic resonance imaging showing right globe mass

After the prognosis, risks and benefits of nephrectomy, and chemotherapy with carboplatin and gemcitabine were explained to the patient, he elected to receive palliative care only. Fifteen days after presentation, the patient developed worsening cough and dyspnea and underwent palliative thoracentesis. Thirty-one days after presentation, he was readmitted to the hospital for shortness of breath and cough. Chest X-ray at that time showed severe pulmonary edema with bilateral pulmonary effusions. He then underwent palliative thoracocentesis and 33 days after presentation expired secondary to respiratory failure.

## DISCUSSION

Although rare, metastases to the orbit have been reported in cases of metastatic renal cell carcinoma with ocular symptoms, at times, being the initial manifestation of carcinoma.[7,8] To our knowledge, there is only one other case of metastasis to the orbit in a patient with RMC.[7]

In that case, the patient was a 39-year-old African-American male with sickle cell trait who presented with a 4–6 weeks history of painful left eye proptosis initially diagnosed as a cavernous hemangioma. He later developed the right flank pain and hematuria 10 days after his initial presentation. Abdominal CT showed a large right renal mass. Upon transconjunctival medial orbitotomy, the lesion was found to be an infiltrative hard mass. Biopsy results of both the orbital lesion and the renal mass showed histologic features consistent with RMC.[7] Upon metastatic workup, that patient was found to have metastatic lesions in the lungs, liver, bone, and lymph nodes. He then underwent an attempted radical nephrectomy which was terminated due to tumor extension into critical structures and subsequently offered palliative radiation to the orbit which he declined and was begun on interleukin-2 and exhibited a poor response. He then passed away from hypoxia secondary to a pulmonary embolism 4 months after presenting to the ophthalmologist and 6 months after the onset of his ocular symptoms.[7]

Similarly, the patient presented here experienced ocular symptoms prior to the development of flank pain and hematuria. Moreover, although biopsy of this patient's orbital mass was not performed, given the clinical setting, pathologic evaluation of the mass would likely be consistent with RMC. It has been documented in the literature that RMC appears as a mass with low attenuation centrally located in the renal medulla on CT with MRI showing a mixed signal tumor secondary to areas of necrosis.[9] Radiologic characteristics of metastatic lesions have largely been described to have features identical to that of the primary tumor; the case of the ocular lesion depicted a hypodense low attenuation infiltrating mass on maxillofacial CT.[7,9] Although the patient presented here initially had a noncontrasted head CT at an outside facility that described the orbital lesion as a hyperattenuated lesion consistent with retinal hemorrhage, the head MRI at our facility showed a retinal mass that enhances with Gadolinium. Thus, we assert that our patient's orbital metastasis likely had a hemorrhagic component that was seen best on head CT while the mass itself was best visualized with MRI with gadolinium. To our knowledge, there are no reports of the radiographic characteristics of an orbital metastasis on MRI in the setting RMC. Thus, we suggest MRI imaging as an additional modality for the evaluation of ocular symptoms in patients with metastatic RMC.

Due to its resistance to both chemotherapy and radiation regimens, there is no standard treatment regimen that has been shown to improve survival. Regimens that have been employed are used based on their efficacy in genitourinary malignancies such as cisplatin, cyclophosphamide, alpha-interferon, vinblastine, methotrexate, and doxorubicin.[10] The use of topoisomerase II inhibitors and high-dose MVAC therapy have been documented to improve survival, although by a small margin, in patients with metastatic RMC.[11,12] Currently, it is unknown whether or not orbital metastasis portends a worse survival. Neither our patient presented here nor the patient in the Zdinak *et al*. case received chemotherapy, and thus there are no reports to our knowledge regarding the effect of chemotherapeutic regimens on survival in patients with orbital metastasis.

## CONCLUSION

We present the case of a patient with RMC and an orbital metastasis to contribute to current literature regarding orbital metastasis and to discuss the evaluation of ocular metastatic lesion using MRI imaging.

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### Conflicts of interest
There are no conflicts of interest.

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