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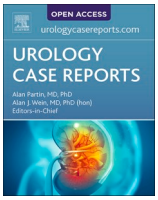

Oncology

A rare case of collecting duct renal cell carcinoma☆

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ABSTRACT

Collecting duct carcinoma is an exceptionally rare and aggressive form of renal cell carcinoma (RCC), representing between 0.4 and 1.8% of RCC cases. The most commonly affected demographic are young African-American males. Here, we present a rare case of collecting duct RCC in a 22 year-old Caucasian female with final pathological staging of pT1aN1 who underwent robot-assisted right radical nephrectomy, with peri-hilar and para-aortic lymph node dissection. Given her node-positivity, adjunctive treatment is discussed.

1. Introduction

Collecting duct carcinoma is an exceptionally rare and aggressive form of renal cell carcinoma (RCC), representing between 0.4 and 1.8% of RCC cases.[1] Given its rarity, information pertaining to its presentation, treatment, and outcomes is limited. However, it has been noted to present at a higher stage (T3/4) than clear cell RCC with more frequent nodal involvement and metastasis.[1] Despite surgical management, mortality risk has been found to be 2.72-fold higher for patients with collecting duct RCC than those with clear cell RCC.[1] Based on a SEER database analysis, three-year disease-specific survival for collecting duct RCC was found to be 58%.[1] Here we present a rare case of collecting duct RCC in a 22 year-old with final pathological staging of pT1aN1.

2. Case presentation

A 22-year-old Caucasian woman with history notable for anemia presented to the emergency department reporting a one-week history of right lower quadrant pain. CT obtained on the day of initial presentation showed a poorly organized fluid collection in the inferior right renal pole that measured approximately 2.4 x 2 × 2.1 cm (Fig. 1).

The patient ultimately underwent a renal biopsy by Interventional Radiology. The biopsy pathology results returned as collecting duct carcinoma, strongly positive for *CK8/18* and *PAX8*, and positive for *AE1/3* and *EMA*. Additional staining showed loss of *INI1*.

Three days later, she underwent a robot-assisted right radical nephrectomy, with peri-hilar and para-aortic lymph node dissection. Of note, the paracaval lymph nodes were enlarged. Postoperative course was uneventful, and she was discharged postoperative day one. On the same day, a CT was completed which showed no evidence of metastatic disease. Furthermore, a MRI of the brain was performed, negative for intracranial metastasis.

Pathologic review of the kidney again revealed collecting duct carcinoma, approximately 2.5 cm in its greatest dimension, with lymphovascular invasion and negative margins. Two paracaval nodes were found to be positive for metastasis, rendering a pathologic stage of pT1aN1 (Fig. 2)

Lymph node specimen was sent for next-generation sequencing (NGS) and found to be negative for *NTRK1/2/3, TMB, MSI, RET* fusions, *BRAF V600E*, and absent of microsatellite instability. However, mutations in *IGF2R, ARID1A, DOTL1* and *MUTYH* genes were identified. No variants with therapeutic significance were found, and thus the patient was not eligible for any targeted therapies or related clinical trials.

3. Discussion

This case represents a unique presentation of a rare form of renal cell carcinoma given that our patient was 22 years old at the time of diagnosis. This case is particularly unique because of the patient's age, sex, and ethnicity. Median age at diagnosis of collecting duct RCC has been cited as mid-sixties, which is similar to that for clear cell RCC,[1,2] and fully four decades later in life than our patient's age at diagnosis.

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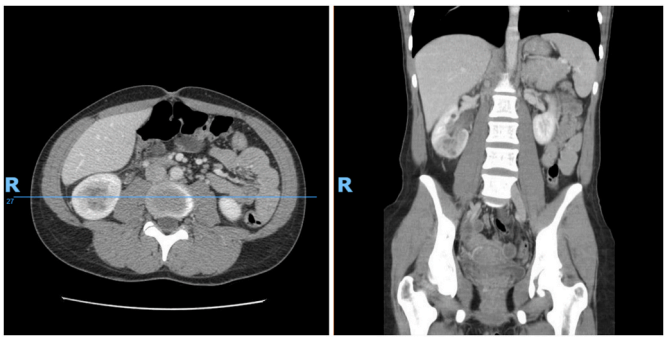

Fig. 1. CT abdomen and pelvis with contrast showing a 2.4 cm lower pole lesion in the right kidney.

Additionally, collecting duct RCC occurs more commonly in men than women (75.6% versus 24.4%, respectively)[2] and African-Americans than in Caucasians,[1] with our patient being female and Caucasian.

Originally believed to arise from collecting ducts of the renal medullary pyramid, new evidence has pointed to an origin in the distal convoluted tubule.[3] However, collecting duct RCC is distinct from the more common clear cell subtype of RCC, which originates in the proximal convoluted tubule.[1,3] Collecting duct RCC is also distinct from renal medullary carcinoma (RMC), which originates in the medullary pyramid collecting ducts and has very high comorbidity with sickle cell trait.[4]

Collecting duct RCC presents most commonly (61.7–80.5%)[2,5] as stage T3 or T4, with nodal metastasis present in about half of cases (48.8%).[2] A 2007 case series of 41 patients with collecting duct RCC found median survival to be 4.9 years,[2] whereas a 2017 retrospective cohort study looking 577 cases of collecting duct RCC from the National Cancer Database found median survival to be 13.2 months.[6] Even more recently, a 2022 retrospective review of 74 patients from two institutions reported a median survival of 24.0 months.[7]

This poor survivability is compounded by the lack of efficacious adjuvant therapies, with >90% of patients being treated with surgery.[1,5] At the time of writing, our patient has not yet begun adjuvant treatment. Currently, the recommended first-line chemotherapy regimen is platinum plus gemcitabine, which has shown a median overall survival of only 10.5 months.[4] In recent years, cabozantinib has also shown promise as a first line therapy with a median progression-free survival of 6 months.[4] However, given the generally poor response rates of collecting duct RCC to chemotherapy, novel therapies, including tyrosine kinase inhibitors and checkpoint inhibitors have been explored.

A 2022 retrospective review of patients with either collecting duct RCC or RMC found that patients who went on after first-line treatment to receive subsequent therapy with cytotoxic agents had an objective response rate (ORR, representing the proportion of patients with either complete or partial response) of 12%.[4] The authors also looked at the ORR for patients undergoing subsequent therapy with either an immune-checkpoint inhibitor (ICI) or tyrosine-kinase inhibitor (TKI) and found the rates to be 10% and 8%, respectively.[4] Thus, they concluded that therapies following the primary treatment have limited efficacy independent of drug class.[4]

That said, when including patients who showed stabilization of their disease because of the subsequent therapy, those receiving TKIs fared the best with 50% achieving disease control. Unfortunately, the durability of any of these therapies was quite poor, with the response only lasting approximately two months.[4]

Given these generally unfavorable options for adjuvant treatment, early and aggressive surgery appears to be essential for providing the best prognosis. In fact, surgery appears to be the only definitive treatment available with adjuvant therapies showing no-to-minimal effect on survival.[3] The rarity of this disease poses a challenge for completing randomized clinical trials; however, further research is needed to develop more efficacious treatment modalities.

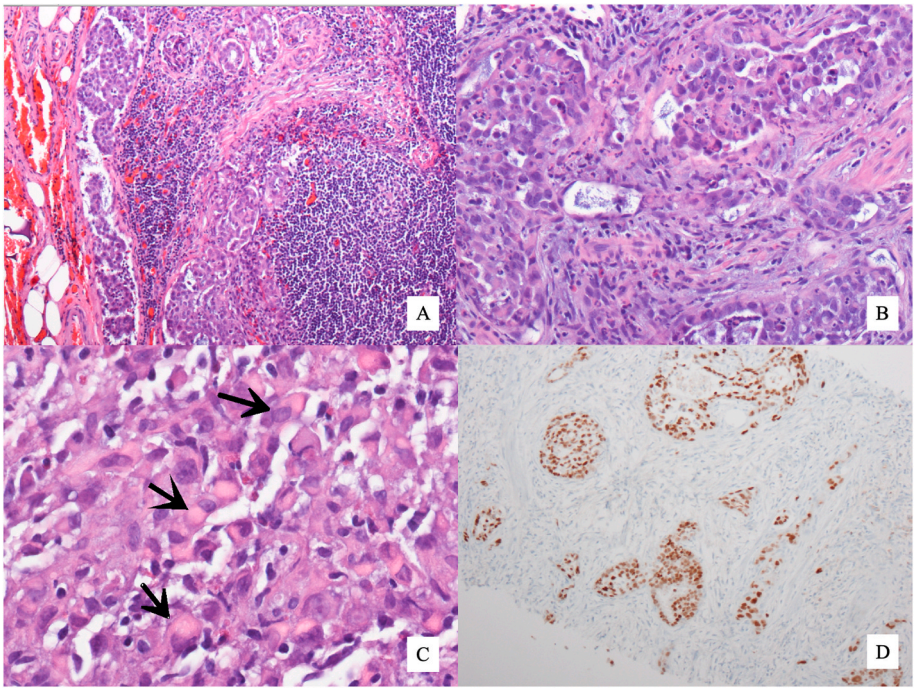

Fig. 2. Histology revealing a diffusely infiltrative neoplasm with poorly differentiated solid, tubular, and papillary appearances with some rhabdoid morphology. A) Tumor in lymph node, B) tubular and papillary areas with brisk acute inflammation, C) poorly differentiated tumor with rhabdoid cells (arrows), and D) tumor cells positive for PAX8.

ABBREVIATIONS: Renal cell carcinoma (RCC), next-generation sequencing (NGS), renal medullary carcinoma (RMC), immune-checkpoint inhibitor (ICI), tyrosine-kinase inhibitor (TKI).

4. Conclusion

We present an extremely rare case of collecting duct RCC, staging pT1aN1 with staining positive for loss of INI1 in a young Caucasian female, who was treated with radical nephrectomy and limited-template lymph node dissection.

Declarations of interest

none.

CRediT authorship contribution statement

Vance Gentry: Writing – review & editing, Writing – original draft, Conceptualization. **Sriharsha Talluri:** Writing – review & editing, Writing – original draft, Supervision, Conceptualization. **Richard Hessler:** Writing – original draft, Visualization, Investigation. **Benjamin Waldorf:** Writing – review & editing, Supervision, Methodology, Investigation, Conceptualization.

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