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Malignant Rhabdoid Tumor of the Kidney in a Young Adult: A Case Report and Review of the Literature

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Abstract

Malignant rhabdoid tumors of the kidney (MRTs) are extremely rare tumors often found in children under 2 years old and even more are in adults with fewer than 10 cases of MRT of the kidney reported in adults. To date, there is no established standard of care for MRTs due to paucity of cases. The present study reports a case of a 21 year-old African American male with a past medical history of sickle cell trait who was found to have MRTs in bilateral kidneys with characteristic clinicopathological features and early onset metastasis. This case of a 21 year-old presents to be the youngest case of MRT present in adults, expanding the previously reported range of 32 to 79 years in adults and reinforcing the importance of recognizing MRTs in a broad range of the adult population.

Introduction

Pediatric primary renal malignancies, aside from the very common Wilms tumor or nephroblastoma, account for less than 1% of all childhood malignancies [1]. Further, the malignant rhabdoid tumor (MRT) is only 1.8% of renal neoplasms [2]. This understudied tumor of the kidney is a rare malignancy that most commonly occurs in children less than two years old, typically ranging between 11 and 18 months [3]. The tumor course is generally aggressive and fatal. Local and distant metastasis occurs early in the disease course and is often resistant to chemotherapy [3-6]. There have been nine adult cases of MRTs reported in the literature [3,5,7–13]. These few reported adult cases suggest an age range of 32-79 years old without gender preference [3,5]. The case of a 21 year-old male with a new diagnosis of MRT presents as a unique challenge and broadens the age range for adult tumors of this kind.

Case Report

A 21 year-old African American male with a past medical history of sickle cell trait presented to the emergency room with a month-long history of nonradiating bilateral flank pain and hematuria. He endorsed weight loss and night sweats, but denied fever, chills, or shortness of breath. On his physical exam, the patient did not have costovertebral angle tenderness or lymphadenopathy. A computed tomography (CT) scan of the abdomen and pelvis without contrast (due to global IV contrast shortage) showed bulky bilateral retroperitoneal adenopathy suspicious for neoplasm including lymphoproliferative and metastatic disease, a 2 mm right kidney stone, and possible 2 mm mass of right kidney in the lower pole. His uric acid level was 3.4 mg/dL, phosphorous was 2.4 mg/dL, and lactate dehydrogenase was mildly elevated at 304 IU/L. Patient elected to leave the emergency department and planned to have outpatient oncology, urology, and surgical oncology.

Renal multiphase CT with contrast performed 2 weeks later showed large multilobular solid masses of right kidney with a dominant conglomerate in the right lower pole measuring 3.5 x 5.4 cm, and small low-attenuation area in the superior pole of the left kidney measuring 2.5 x 2.1 x 1.6 cm with areas of internal nodular enhancement suspicious for mass. CT also showed similar appearance of the suspected 2 mm right kidney stone, though pain was inconsistent with ureteral colic. Positron electron tomography (PET/CT) Whole Body Scan showing bilateral renal masses, left supraclavicular adenopathy, left highest mediastinal adenopathy, and extensive bilateral retroperitoneal and mesenteric adenopathy, results consistent with lymphoma at the time.

Two months after the initial emergency room presentation, the patient presented again to a community hospital emergency room with a one day history of acute respiratory failure, met the criteria for sepsis, and was admitted to the hospital. CT angiography ruled out acute pulmonary embolism. Patient required 2L of oxygen via nasal cannula on and off during the hospitalization.

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Physical exam shows new bilateral cervical lymphadenopathy and no costovertebral tenderness. Of note, this current admission is a day after the patient underwent CT guided right lower pole renal mass and right retroperitoneal lymph node biopsy. Patient was initially started on Zosyn for three days but was discontinued due to infection being less likely. CT abdomen pelvis showed large bilateral pleural effusion with bilateral lower lobe atelectasis. Patient underwent bilateral therapeutic and diagnostic thoracentesis but pleural effusion continuously reaccumulated within a day. Flow cytometry of pleural fluid, in addition to the flow cytometry of renal tissue from biopsy showed no evidence of non-Hodgkin lymphoma. Concurrently, the patient's acute kidney injury progressively worsened throughout the hospitalization stay with creatinine levels up to 2 mg/dL. Renal biopsy from prior finally resulted in a diagnosis of poorly differentiated malignant neoplasm with rhabdoid features with metastasis to lymph nodes.

The microscopic description showed extensive necrosis of a malignant neoplasm comprised of rhabdoid cells with large ovoid nuclei, prominent nucleoli, and eosinophilic cytoplasm. Mitotic activity was brisk with numerous abnormal mitotic figures. Immunohistochemical stains was diffusely positive for Vimentin and Pankeratin and negative for CK7, PAX8, Desmin, Myogenin, PLAP, CD30, Inhibin, hCG, AFP, GATA3, p63, CD45, and CD20. Further there was a diffuse loss of INI expression in the tumor cells. The pathologist highly favored metastatic malignant rhabdoid tumor of the kidney as the final diagnosis.

The patient was then transferred to the larger academic hospital in the area for more access to specialized care and the potential for inpatient chemotherapy. Throughout the course of his stay, the patient was followed by several services. In the setting of his worsening malignant pleural effusions and tense ascites, he required oxygen support and several thoracenteses and paracenteses. Hyperkalemia was noted at a maximum of 6.8 mmol/L and creatinine was 4.73 mg/dL requiring hemodialysis and eventually continuous renal replacement therapy (CRRT). Liver function tests showed moderate elevation including an AST 242 IU/L, ALT 180 IU/L, and total bilirubin 1.9 mg/dL noted to be increasing from previously normal results four days prior. Urologic surgery was consulted for hematuria at presentation, but surgery was not recommended due to hyperkalemia.

Adult oncologic services discussed with pediatric oncology for chemotherapy treatment. Options considered were etoposide, cyclophosphamide, adriamycin, vincristine, and methotrexate. Adriamycin and vincristine would be limited by liver function. Methotrexate was not recommended due to pleural effusions. Thus, the patient was started on a five-day course of ifosfamide with mesna and etoposide. After two days of treatment, the patient's liver function worsened with total bilirubin increasing dramatically to 14.3 mg/dL. A slight elevation in liver function was expected with the chemotherapy regimen, but intervention was considered more beneficial to the patient's overall course. Right upper quadrant ultrasound showed new hypoechoic lesions of 1.3 cm in the right lobe and 1.4 cm in the left lobe not seen in previous PET/CT, concerning for further progression of metastatic disease. On day three of the cycle, the total bilirubin was 17.2 mg/dL, and chemotherapy was held until further improvement.

The hospital course continued to be tenuous for the patient. Recurrent pleural effusions limited breathing. Formation of a reactive ileus required decompression via nasogastric tube. Development of multifactorial shock required increasing vasopressor support. After extensive discussion with the patient, his family, and palliative care, the patient stated he would like to be transitioned to comfort care. He died in the hospital three months after initial symptom presentation at home and 15 days after current hospital admission.

Discussion

Renal MRTs was originally reported to be a rhabdomyosarcomatoid variant of Wilms tumor because of cells' resemblance to rhabdomyoblasts in Beckwith's First National Wilms Tumor Study [14].MRTs of the kidney have only recently been distinguished as a separate malignant renal neoplasm from Wilms tumors after subsequent studies failed to confirm myogenic differentiation [10]. The development of a concise treatment regimen for Wilms tumors demonstrates the advancements made in pediatric oncology, and now has a survival rate of 70% for stage IV disease [1].

Since the identification of MRTs, the tumor has been reported in a number of extrarenal sites including the central nervous system, liver, soft tissues, lungs, skin, heart, orbit, mediastinum, retroperitoneum, pancreas, gastrointestinal tract, and the urogenital system [3,10]. The common presentation among pediatric MRT cases includes a palpable mass, hematuria, abdominal or flank pain [3]. In adults, the presenting symptoms are flank pain and hematuria according to a study by Han et al in 2022 of eight other adult cases of MRTs [5]. Metastatic symptoms are common at diagnosis and mainly affect the lungs, liver, and brain [3]. Other symptoms included constitutional symptoms such as fatigue and weight loss [5]. Confirmation of MRTs can be from nephrectomy, core biopsy, or autopsy specimen [3]. Initial evaluation of suspected MRTs typically rely on clinical features and are necessary for preoperative diagnosis early in disease course. These features include very young age, fast growth, and typically, due to the aggressive nature of the tumor, early metastasis [1,6].

MRTs commonly have similar radiographic findings as that of other renal tumors like Wilms tumors [6]. Additional noted features among MRTs, primarily in pediatric cases, include subcapsular fluid and curvilinear calcifications but these findings are not pathognomonic and can be found in many different malignancies [1]. The only consistent finding across imaging is irregular borders and lymph node metastasis [1]. Radiographically, this case had those findings, including a significant burden on the retroperitoneal and mesenteric lymph nodes as well as the irregular borders of the primary renal tumor. The ambiguity of the radiographic findings can even mislead the diagnosis, such as this case, when the obvious and severe lymph node disease appeared to be consistent with lymphoma.

As stated, pure MRTs are rarely found in the adult population, however, it has been noted in the literature that some renal cell carcinomas can have rhabdoid features. These studies have shown that divergent differentiation of rhabdoid tumors may arise from any subtype of other renal cell carcinoma, often clear cell; however within these studies, histologic features of the originating malignancy were found adjacent to the rhabdoid tumor masses [4,15,16]. Still, the question is raised of whether the presenting case truly had a de novo MRT or if the rhabdoid tumor was a clonal component of a different underlying carcinoma. The two core biopsies from the right kidney were 1.0 cm or less per gross description, thus limiting the true extent of the tumor, and further obscuring the possibility for an adjacent tumor of different features. Renal medullary carcinoma (RMC) was also considered due RMC's common presentation in young patients of African descent with sickle cell trait, similar to the patient's presentation. RMC is one of few other SMARCB1-deficient tumors as signified by loss of INI also including MRT, and atypical teratoid rhabdoid tumor, narrowing the differential substantially [17]. MRT was considered the final diagnosis due to the immunohistochemical staining, which notably was negative for PAX8 and CK7, both of which are commonly positive in RMCs. Despite this and the diffusely rhabdoid morphology, including eosinophilic cytoplasm with prominent nucleoli, there was hesitance in the diagnosis because of the patient's age, representing this unusual presentation.

Some have described MRTs as the most aggressive primary renal neoplasm, due to the practically uniformly fatality despite treatment [1,18]. Typical treatment of pediatric MRTs consists of resection of the primary tumor, chemotherapy and radiation. Chemotherapy regimens often include cyclophosphamide, actinomycin, vincristine, doxorubicin, methotrexate, among others [19] Tomlinson et al showed that in the National Wilms' Tumor Study, there was no difference in survival rate of patients with MRTs who did or did not receive doxorubicin [20]. Chemotherapy regimen for MRTs is still quite variable, but tends to follow Wilms tumor, or even soft tissue sarcomas, chemotherapy regimens [21,22]. For adults, the treatment is much less consistent due to the rarity of disease. From the few previous adult cases in the review by Han et al, treatment included a chemotherapy regimen similar to those above with the addition of interleukin-2 (IL2), which is commonly added to regimens for metastatic renal cell carcinomas [5]. Another case cited using IL-2 along with interferon-alpha (IFN-alpha), used for a comparable purpose as IL-2. Similarly, one of the cases used atixinib, which is typically used for advanced renal cell carcinomas as well.

In the present case, treatment was unable to be completed due to severity of disease burden on the liver. As this further demonstrates that MRTs are not solely pediatric tumors, but those that can also occur in adults, established treatment regimens and early diagnosis are necessary to prolong the course of inevitable mortality. If the tumor can be identified before severe metastatic disease limits surgical intervention, surgery with adjuvant chemotherapy provides the best likely outcome. Addition of IL-2 and IFN-alpha to the chemotherapy regimen in adults has not shown to be beneficial in the review by Han et al as one patient who received this treatment lived over a year after initial diagnosis, while the other died within 5 months [5].

MRT prognosis is abysmal for both pediatric and adult cases as most reported cases of MRTs have short survival times after initial diagnosis along with early-onset local and distant metastasis [3]. The Qureshi group reported that of the patients (n = 14, median age = 11 months) treated with curative intent, 70% of MRT patients developed systemic recurrence and died of progressive disease at a median of 7 months since diagnosis [Qureshi 2020]. In adults, there was a report of a 38-year-old white woman with MRT who died five months after radical nephrectomy and metastasis [10]. Another case report showed that a 65-year-old man with a longstanding history of renal calculi had a month-long history of right flank pain and passed away a few days after MRT diagnosis was established and before any therapeutic decision was made [3].

The difference in regimens likely stems from the age difference. The pediatric Wilms tumor chemotherapy regimens are well studied, and although treatments are usually unsuccessful for MRTs, these therapies are familiar to pediatric oncologists, despite the rarity of the rhabdoid tumor. In children, Tomlinson et al found that age can be considered to be a prognostic factor. Pediatric cases in patients less than one year old have more dismal outcome with early central nervous system metastasis compared to greater than one year old patients with better prognosis [20]. As for adults, the oncologists are faced with an even rarer form of a rare disease. Due to the patient's young age of 21 years-old in this presented case, discussing with pediatric oncology and choosing a regimen common for other pediatric renal malignancies was an appropriate choice, albeit limited by advancing disease. It is unclear whether other regimens used by previous works, including IL-2 or IFN-alpha change prognosis, as one case that used it died over 18 months from diagnosis while the other died within 5 months [5].

The SMARCB1 protein is an abbreviation of its full name: SWI/SNF Related, Matrix Associated, Actin Dependent Regulator of Chromatin, Subfamily B, Member 1, alteration of which disrupts chromatin-remodeling and alters global transcription patterns [17]. Deletion of this IMI-1 gene, a tumor suppressor gene and part of the SWI/SNF chromatin remodeling complex, in knockout mice led to the formation of rhabdoid tumors at an early age, offering an explanation of why these tumors are often lethal in childhood and rarely diagnosed in adulthood [23]. The question still arises, then, why do these cases of late-onset MRTs exist and through what changes do they develop? The possibility still stands that these tumors arise de novo, particularly in those reported in a literature review by Han et al where treatment often included radical nephrectomy, thus eliminating the obscurity of a small sample, as encountered in this case report [5]. Although, slow progression from a different malignancy in the form of rhabdoid transformation is still possible.

Conclusion

Malignant rhabdoid tumors of the kidney in adult patients are extremely rare with no established standard of care due to paucity of cases, resulting in one of the worst prognosis among all renal tumors. Our current case report disputes MRT's bimodal distribution of cases in patients less than 2 year old and adult cases ranging from 32-79 year old [3,5]. In the case of

this young patient, the age range of MRTs has broadened to practically any age, as many would consider 21yearsold as an appropriate age to still include within the realm of pediatrics. Further studies of the pathogenesis and treatment of this rare disease are needed to elucidate its complicated and tremulous course.

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Conflict of Interests

The authors have no conflicts of interests to disclose.

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