Primary synovial sarcoma of the kidney with unusual follow up findings

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We present a case report of a 17-year-old patient with a large renal mass that was detected on a computed tomography scan during investigation for secondary hypertension. Radical nephrectomy was performed and the morphologic and immunocytochemical findings were compatible with a diagnosis of monophasic synovial sarcoma of the kidney. A cytogenetic search for t(X;18) translocation was performed, which was negative. The patient underwent an ifosfamide-based chemotherapy regimen. During follow up, a positron emission tomography scan showed increased 18F-fluorodeoxyglucose metabolism at the right femur. Although cancer cells were expected in the biopsy specimen, only fibrous dysplasia of the bone was found. The patient was disease free at his 29 month follow up check up.

Key Words: kidney, positron emission tomography, renal tumor, synovial sarcoma, *SS18 (SYT)/SSX*

Introduction

Synovial sarcoma is a soft-tissue tumor that develops mainly in the limbs, but which has also been reported in many locations including the kidney.[1] Although sarcomas account for about 1%-3% of malignant kidney neoplasms, synovial sarcoma of the kidney is exceedingly rare. To the best of our knowledge, about 40 cases have been reported in the literature. Young adults are more commonly affected,[2] although reported age at diagnosis ranged from 15 to 59 years. The tumors usually present as large masses and recurrence more frequently affects the lungs, although liver and bone metastases have also been described.[3]

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Although synovial sarcoma of the kidney is virtually indistinguishable from other kidney tumors—grossly or on computed tomography (CT) scans—its histologic and cytogenetic features may suggest the correct diagnosis.

The presence of a unique chromosomal translocation, t(X;18)(p11.2;q11.2), which results in the fusion of the *SS18* gene (previously known as the *SYT* gene[4]) on chromosome 18 with an *SSX*-family gene (*SSX1*, *SSX2*, or *SSX4*) on chromosome X is the main diagnostic hallmark of this tumor.[5] However, this chromosomal rearrangement may be absent in up to 10% of cases,[6] and in those cases, microscopic features and positive immunohistochemistry results for selected markers play a major role in diagnosis.

Due to the rarity of this tumor, no treatment guidelines are available; however, adjuvant therapies are generally used to treat these patients postoperatively based on this tumor's clinically aggressive behavior.[3]

Case report

A search for secondary causes of severe hypertension was performed on a 17-year-old patient who was being treated with nifedipine and nebivolol. A contrast-material–enhanced CT scan showed a large (10.6 cm x 8.9 cm x 16 cm), heterogeneous, expansive lesion originating in the left kidney, which was mainly cystic with solid areas.

An open radical left nephrectomy was performed. Microscopic examination of the biopsy specimen revealed a moderately cellular tumor composed of monomorphic, plump spindle cells growing in short, intersecting fascicles, Figure 1 and Figure 2. The cells had scant cytoplasm and were ill-defined. Extensive necrosis and areas with a hemangiopericytoma-like vascular pattern were found. Peripherally, there were cysts lined with mitotically inactive epithelium with eosinophilic cytoplasm and a hobnail appearance. Immunohistochemistry tests were positive for bcl-2, vimentin, CD56, CD99 (focally), epithelial membrane antigen (EMA; weak and focally) and negative for AE1/AE3, CK7, GFAP, WT1, CD34, pS100, desmin, and smooth muscle actin. These pathological findings were compatible with a diagnosis of a monophasic synovial sarcoma of the left kidney. Hilar and preaortic lymph nodes that were macroscopically suspicious of being invaded and thus resected during surgery were negative for neoplasm cells. Based on these results, a cytogenetic analysis was performed to search for t(x;18) translocation using fluorescence in situ hybridization (FISH) and probing for translocation of the *SS18* gene. No rearrangement of this gene was found.

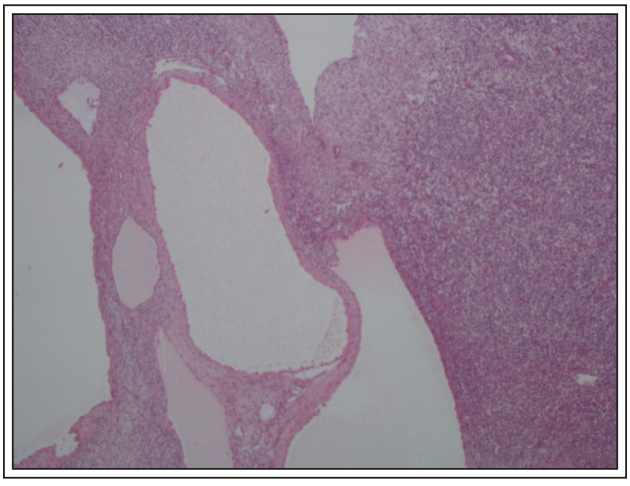

Figure 1. Microscopic image showing a solid tumor with cystic spaces (original magnification x10, hematoxylin-eosin stain).

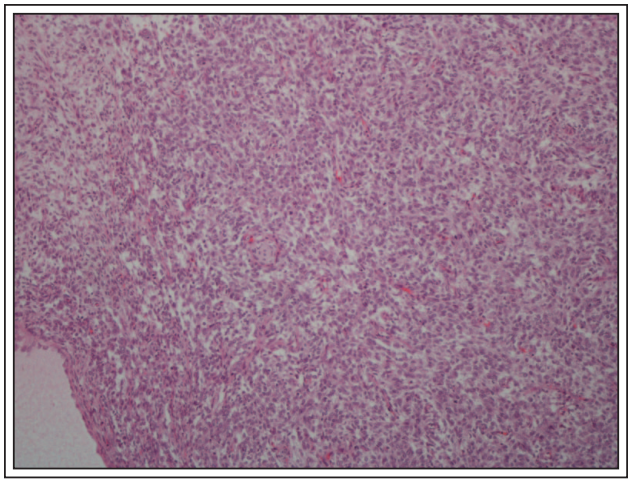

Figure 2. Microscopic image confirming a tumor composed of spindle-shaped cells arranged in interlacing short fascicles (original magnification x20, hematoxylin-eosin stain).

The patient was referred to the oncology department and started on adjuvant doxorubicin-ifosfamide chemotherapy.

One month after surgery, 18F-fluorodeoxyglucose (^{18}F-FDG) positron emission tomography (PET) imaging and CT imaging were performed to detect any metastasis. Three intramedullary lesions with increased FDG metabolism were found in the right femur. The largest measured 37 mm and had a maximum standardized uptake value (SUV) of ^{18}F-FDG of 6.80. A magnetic resonance imaging (MRI) test was performed to better assess these lesions, but it was inconclusive. Shortly afterwards, the patient had incapacitating pain in his right knee, and he received opioids and needed auxiliary equipment to be able to walk. The patient was referred to an oncological orthopedic surgeon. A biopsy of the lesions was performed to make a definite assessment of their etiology. The patient missed a follow up appointment because he suffered a pathological fracture 4 days after the biopsy. The pathology report revealed the presence of fibrous dysplasia with no malignant tissue.

The patient showed no evidence of recurrence of tumors at his 29 month follow up check up.

Discussion

Even though an *SS18* gene rearrangement was not found in this patient, microscopic and immunocytochemical findings strongly suggested a diagnosis of a synovial sarcoma of the kidney. Large-volume masses with or without secondary lesions at the time of diagnosis

of a synovial sarcoma of the kidney and a high recurrence rate of this tumor suggest that this tumor behaves aggressively. Most clinicians agree that adjuvant therapy should be considered. However, evidence-based treatment guidelines have not yet been developed, since only a few cases have been reported. Since the current patient had no lymph node involvement and had negative preoperative staging for distant metastases, he was given an ifosfamide-based regimen. After more than 2 years of follow up, there was no recurrence of cancer.

The PET and CT false positive results were unexpected. The three bone lesions had high values for maximum SUV of ^{18}F-FDG (the lowest maximum SUV was 3.15), which was highly suggestive of metastatic cancer. Additional ways to increase ^{18}FDG-PET/CT scan specificity for bone tumors have been proposed, since the usual cut off of a maximum SUV > 2 for increased FDG metabolism seems to frequently result in suspicion being raised for benign conditions.[7] Our patient's biopsy revealed fibrous dysplasia of the bone, which is a genetic-based, nonhereditary bone disorder that is difficult to differentiate from other types of bone tumors, including metastatic lesions.[8] □

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