

## Case Report

# Primary synovial sarcoma of the kidney-A case report and literature review

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**Abstract:** Primary synovial sarcoma of the kidney is a very rare spindle cell neoplasm that occasionally displays epithelial differentiation. It occurs between 15-60 years of age with a mean of 35 years and a slight male predilection. Most of synovial sarcomas appear as relatively nonspecific soft tissue masses involving the kidney. This rare entity has some overlapping morphologic and immunohistochemical characteristics with other more common renal spindle cell neoplasms. Molecular tools add valuable diagnostic confirmation. We report a 56 year old male who presented to the emergency department with hematuria and abdominal pain. He had an abdominal CT-scan which showed a 6.6 cm enhancing right renal mass. Morphologic and immunohistochemical studies were directed towards synovial sarcoma with confirmation by SYT-SSX gene fusion using RT-PCR molecular technique. We reviewed the literature on the epidemiologic, histologic spectrum, immunophenotypic, clinical significance and prognosis and therapy.

**Keywords:** Primary, renal, synovial sarcoma, spindle cell neoplasm, biphasic

## Introduction

Primary synovial sarcoma of the kidney is a very rare spindle cell malignant neoplasm that occasionally displays epithelial differentiation and is characterized by specific translocation t(X:18). This rare tumor occurs between 15-60 years with mean of 35 years and slight male predilection [1]. This entity accounts for 5-10% of adult soft tissue sarcomas. Primary renal synovial sarcoma was first described in 1999 by Argani *et al* [2]. Clinically, patients with primary renal synovial sarcoma present with abdominal or flank pain which could be accompanied with abdominal distension [3]. Histopathologic diagnosis is difficult, and usually necessitates immunohistochemical staining and molecular evaluation. Molecular studies have demonstrated the presence of the chromosomal translocation t(X;18)(p11;q11) in over 90% of cases of synovial sarcoma. This anomaly leads to a hybrid creation which involves gene SYT on 18p11 and one gene of SSX family on chromosome X, mostly SSX-1, less frequently SSX-2, and rarely SSX-4 [4]. Differential diagnoses of this predominantly spindle cell tumor include sarcomatoid renal cell carcinoma, malignant

melanoma, malignant solitary fibrous tumor, nephroblastoma of the adult type, dedifferentiated liposarcoma, rhabdomyosarcoma, leiomyosarcoma and angioliipoma. We describe a case of primary synovial sarcoma of the kidney, and present a review of the relevant literature.

## Case report

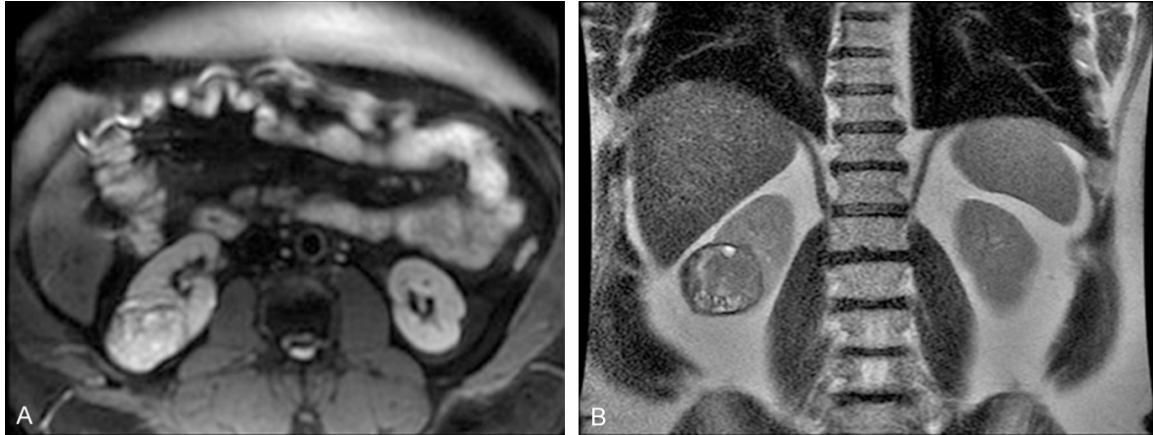
### Subject

A 56 year old male presented to the emergency room with a recent history of hematuria and abdominal pain. He is non-smoker with no significant past medical history. His blood workup was nonspecific.

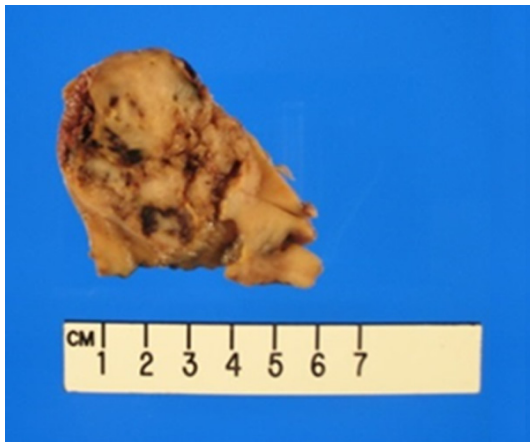
### Radiology

Abdominal and pelvic CT-scans and MRI showed a 6.6 cm enhancing right renal mass with high suspicion for malignancy (**Figure 1**).

The patient underwent a partial nephrectomy for which intraoperative consultation for diagnosis was provided. The intraoperative consultation was "High grade malignant neoplasm with spindle cell features". The surgical margin was free of tumor.



**Figure 1.** Cross and coronal abdominal MRI images show an enhancing right renal mass.



**Figure 2.** Gross picture of the right partial nephrectomy shows a tan/pale tumor with focal cystic and hemorrhagic areas.

#### *Gross examination*

Gross examination of the partial nephrectomy specimen showed a 175 gm, 8.5 × 7.0 × 4.5 cm partial nephrectomy with moderate attached perinephric fat. Serial sectioning was remarkable for a 6.6 × 5.0 × 4.5 cm tan/white partially necrotic and focally hemorrhagic mass. Multiple cystic areas with clear contents were identified. The remainder of the kidney parenchyma was tan/brown with a well-defined corticomedullary junction (**Figure 2**).

#### *Microscopic examination*

Microscopically, the tumor was mitotically active, with monomorphic plump spindle cells and distinct cell borders growing in short, intersecting fascicles. Cysts were lined by mitotically inactive polygonal eosinophilic cells with “hobnailed” epithelium (**Figure 3**).

#### *Immunohistochemistry*

Properly controlled immunohistochemical stains were performed to detect the nature of the tumor cells. Differential diagnoses of this predominantly spindle cell tumor include sarcomatoid renal cell carcinoma, malignant melanoma, malignant solitary fibrous tumor, nephroblastoma of the adult type, dedifferentiated liposarcoma, rhabdomyosarcoma, leiomyosarcoma and angioliipoma. The presented case tumor cells were positive for vimentin, CD99, Bcl-2 and focally positive for cytokeratin (AE1/AE3), synaptophysin, and EMA. Tumor cells were negative for CEA, desmin, CD34, S-100, CD-10, PAX-8, and HMB45. The cystic epithelium was focally positive for cytokeratin (AE1/AE3) and EMA (**Figure 4**).

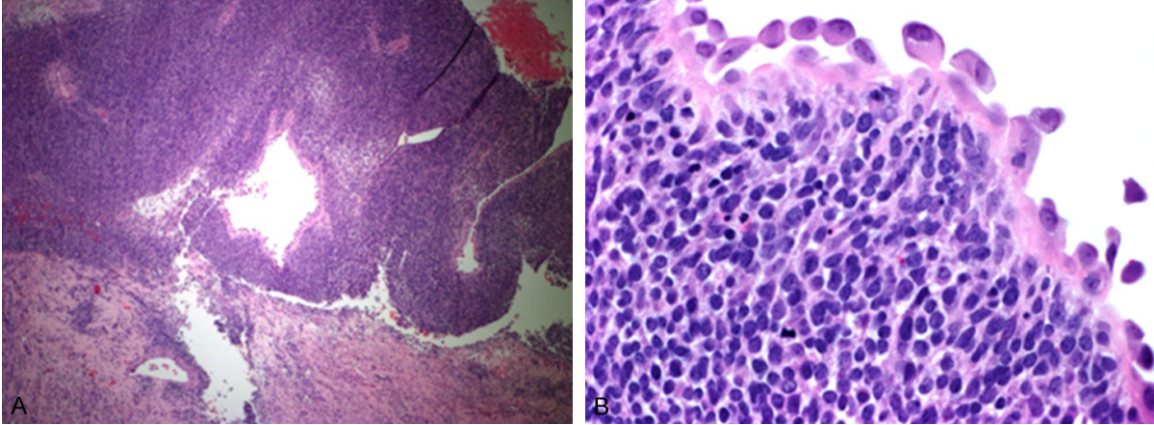
#### *Molecular study*

The current diagnostic gold standard for synovial sarcoma is to demonstrate the fusion of the SYT (Synonyms: SS18-synovial sarcoma translocation, chromosome 18) gene on chromosome 18 to either SSX1 (synovial sarcoma, X breakpoint 1) or SSX2 (synovial sarcoma, X breakpoint 2) gene on chromosome Xp11. RT-PCR study of SYT-SSX genes 1 and 2 fusions were detected by work of Dr. Sarah Kerr and her group at Mayo Clinic Rochester, USA (**Figure 5**).

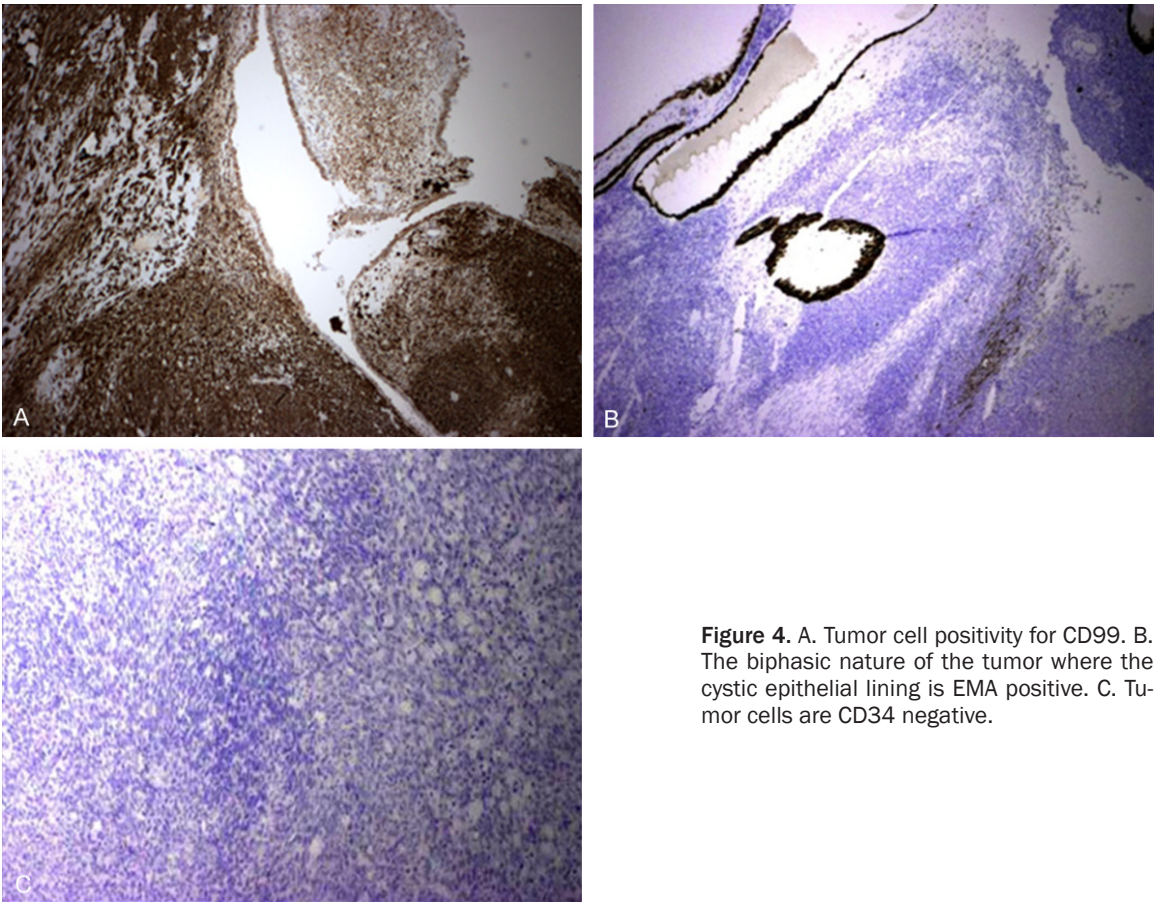
#### **Discussion**

Primary synovial sarcoma of the kidney is a very rare soft tissue tumor. This rare renal tumor typically affects younger patients of both genders, with a slight predominance in males [5].

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**Figure 3.** A. Low power view of the tumor shows monomorphic spindle cell proliferation and focal cystic areas. B. High power view of the tumor shows cystic areas with "hobnailed" epithelial lining.

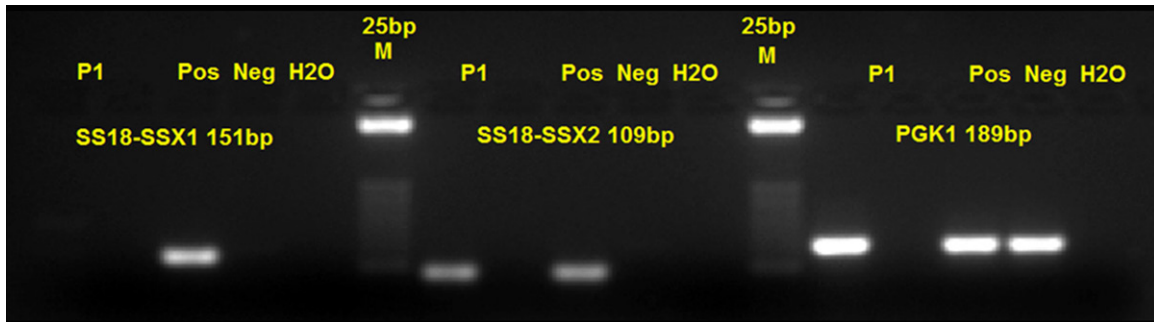


**Figure 4.** A. Tumor cell positivity for CD99. B. The biphasic nature of the tumor where the cystic epithelial lining is EMA positive. C. Tumor cells are CD34 negative.

For the pathologist, it may be challenging to differentiate synovial sarcoma from other, more common forms of spindle tumors arising from the kidney, particularly sarcomatoid renal cell carcinoma, metastatic sarcoma, solitary fibrous tumors and retroperitoneal sarcomas involving the kidney [6].

There are no specific clinical or imaging characteristics for synovial sarcoma: clinical symptoms and CT images do not differ from other malignant renal tumors [7].

Primary renal synovial sarcoma consists of sheets of undifferentiated monotonic spindle



**Figure 5.** RT-PCR study shows that tumor cells are SS18-SSX 1 and 2 genes fusion.

cells with hyperchromatic nuclei and frequent mitoses [8]. To the best of our knowledge, approximately only 34 cases have been reported till date [11]. Biphasic synovial sarcoma can be diagnosed with the existence of epithelial and spindle cell elements. However, monophasic synovial sarcoma may be difficult to differentiate from other spindle cell sarcomas. The cystic areas are lined by epithelial cells which are focally positive for cytokeratin (AE1/AE3) and for EMA which may represent entrapped tubules.

Literature review showed that the spindle cells in the synovial sarcoma are Bcl-2, vimentin, CD99, EMA, CD56 positive, while they show no focal reactivity with S100, desmin, SMA, CD34, AE1/AE3, and WT-1.

From the molecular point of view, the current diagnostic gold standard for synovial sarcoma is to demonstrate the fusion of the SYT (Synonyms: SS18-synovial sarcoma translocation, chromosome 18) gene on chromosome 18 to either SSX1 (synovial sarcoma, X breakpoint 1) or SSX2 (synovial sarcoma, X breakpoint 2) gene on chromosome Xp11 [9].

The overall clinical, histopathological, immunohistochemical and molecular results directed toward primary synovial sarcoma of the kidney than the other differential diagnosis of leiomyosarcoma, sarcomatoid renal cell carcinoma, malignant solitary fibrous tumor, and dedifferentiated liposarcoma.

Primary renal synovial sarcomas are treated with complete surgical resection (Nephrectomy) with negative margins. The role of adjuvant therapy can be applied as preoperative irradiation for large or initially unresectable primary

tumors and chemotherapy for disseminated disease. Local recurrence in synovial sarcomas is common, especially following incomplete resection with metastatic rate up to 50% of cases [10]. The most common site of metastasis is lung where late metastases can appear after many years. Synovial sarcomas have a 5-year survival rate of 50-85% [7]. Tumors smaller than 5 cm that occur in childhood have favorable prognosis [8]. Biphasic versus monophasic morphology has no prognostic relevance.

### Conclusion

Primary renal synovial sarcoma is a very rare tumor with a non-specific presentation. Clinicians should consider it among the differential diagnosis of renal masses composed of spindle cells. Since morphological demarcation from other tumors may be complicated, additional diagnostic techniques like immunohistochemistry, cytogenetics, and advanced molecular analyses need to be applied.

### Acknowledgements

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### Disclosure of conflict of interest

None.

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