中文題目：原發性腎臟滑膜肉瘤--兩件病例報告

英文題目：Primary renal synovial sarcoma: 2 cases report in a single medical center

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Introduction

Synovial sarcoma is a rare neoplasm. The annual incidence rate in adults was 1.42 per million and median age of diagnosis was 34 years[1]. The most common sites of primary tumor location were extremities (65.7%), followed by trunk (18.7%) and head/neck area (8.5%)[2]. Primary renal synovial sarcoma accounted for less than 1% of all synovial sarcoma and only approximately 150 cases were reported to date[3]. Herein we shared the experience of 2 patients with primary renal synovial sarcoma and review of medical literature.

Case Presentation

Case 1: A 24-year-old healthy man presented with sudden onset of left flank pain without gross hematuria in 1 month ago. He visited local hospital and underwent renal ultrasonography which didn't reveal any renal stone, but a heterogeneous mass in the left upper pole of kidney was detected. The Computed Tomography (CT) of abdomen with contrast showed a 6.1 cm tumor extending into the left renal pelvis with tumor thrombus inside left renal vein, highly suspected renal cell carcinoma. Based on a clinical diagnosis of clinical T3 (AJCC 8th) renal cell carcinoma, the patient underwent left radial nephrectomy. Histopathological examination revealed ovoid to spindle neoplastic cells with hyperchromatic nuclei, with positive of CD99, Vimentin and TLE-1 in immunohistochemistry staining. Fluorescence in situ Hybridization (FISH) study revealed split signal of SS18 gene which was consistent with the diagnosis of poorly differentiated synovial sarcoma of the kidney. The patient received adjuvant chemotherapy with doxorubicin and ifosfomide.

Case 2: A 40-year-old woman with no known medical history, presented to our gynecology clinic with right flank pain and painless hematuria for 6 months. Pelvic exam revealed a huge abdominal mass, and the gynecological

sonography showed a 13.30 x 9.30 cm pelvic heterogeneous mass. CT of the abdomen revealed a huge right renal mass occupied nearly the whole kidney with tumor thrombus invading to right renal vein and inferior vena cava (IVC), and multiple enlarged retroperitoneal lymph nodes and lung metastases. The patient underwent CT-guided biopsy, and it showed relatively monotonus polygonal small blue cells with negative of AE1/AE3, CD10, WT-1 and positive CD99 and Vimentin, in favor of a primitive neuroectodermal tumor (PNET). The patient received 2 cycles of vincristine, doxorubicin and cyclophosphamide (VAC) with partial response. However, the histopathologic diagnosis was argued with no split signal of EWSR1 by FISH assay which is typical genetic feature of PNET. By contrast, the SSL18-SSX2 fusion gene copy was detected by reverse transcription-PCR that implicated is as a synovial sarcoma. We shifted chemotherapy regimen to doxorubicin, ifosfamide and mesna (AIM) accordingly.

The patient got good partial response on 2 cycles of AIM, and underwent subsequently cytoreductive right nephrectomy. The pathology confirmed the diagnosis of primary renal synovial sarcoma with monophasic cell type. Postoperative chemotherapy with 6 cycles of ifosfamide, carboplatin and etoposide (ICE) followed by 6 cycles of mesna, doxorubicin, ifosfamide and dacarbazine (MAID) was administered to maintain the maximal tumor control. However, CT images revealed progressive lung metastasis and a new mass at right adrenal region. She had been treated on pazopanib for 7 months. The patient eventually died because of pneumonia, 36 months after diagnosis.

Discussion

Synovial sarcoma is a mesenchymal neoplasm with unknown cell origin. It could be further divided into various morphologic subtypes, including monophasic, biphasic spindle cell types and poorly differentiated types. The differential diagnosis of synovial sarcoma is wide by its similar morphological patterns. SS18-SSX fusion oncogene is highly specific while no other neoplasm has shown such unique gene expression[4]. Both FISH and RT-PCR are optimal and convenient diagnostic tools for detecting translocated genes.

The typical feature of renal synovial sarcoma in CT imaging often appears a huge, heterogeneous, cystic mass (>3 cm) with enhancing septa. Unlike angiomyolipoma, the most common benign neoplasm of the kidney that is fat-predominant at image, renal synovial sarcoma has soft-tissue attenuation and could extend into the renal pelvis or even the renal vein and

inferior vena cava[5]. It could be further differentiated from a large renal cell carcinoma (RCC) by the absence of central necrosis. However, because the differential diagnosis of large renal mass is broad and easily misleading, physicians should never make the diagnosis solely based on images and it often requires percutaneous biopsy.

El Chediak A et al. reported the characteristics of 114 primary renal synovial sarcomas in 2018[3]. The median age was 40.5 years old, and the most common symptoms were hematuria (38%) and flank/lumbar pain (24.4%). Available data for metastasis were observed in 27.1% (19/70) patients. The prognosis of primary renal synovial sarcoma was generally poor regardless of all kinds of treatment. On the contrary, the negative predictors for synovial sarcoma in trunk and extremities were old age, inoperable disease, big tumor size and high histologic grade on outcomes[6].

We reported two cases of rare primary renal synovial sarcoma occurring in young and middle-aged adults with an insidious clinical presentation and poor prognosis. CT image could help us to differentiate the renal mass initially but the diagnosis is based on the presence of SS18-SSX fusion oncogene, which is exclusively shown in synovial sarcoma. The future research on early diagnosis and intervention is crucial to improve the clinical outcome.

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