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Case Report

Integrated Imaging-Pathologic Diagnosis and Treatment Overview of Mucinous Tubular and Spindle Cell Renal Cell Carcinoma: A Case Report and Systematic Review of the Literature

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Keywords
Mucinous tubular and spindle cell carcinoma · Kidney neoplasm · Conservative management · Immunohistochemistry · Cytological analysis · Surgical treatment

Abstract
Introduction: Mucinous tubular and spindle cell carcinoma (MTSCC) of the kidney is a rare, low-grade renal epithelial tumor with a unique histopathological profile. This case report aimed to highlight the clinical, imaging, and immunohistochemical features of MTSCC, facilitating its differentiation from other renal tumors and providing insight into effective management strategies. ***Case Presentation:*** A 52-year-old female presented with left-sided abdominal pain. Imaging studies, including CT and ultrasound, revealed a low-density mass in the left kidney. The patient subsequently underwent laparoscopic radical nephrectomy. Pathological examination identified spindle and cuboidal epithelial cells forming elongated tubules with mild atypia within a mucinous stroma. Immunohistochemical staining showed positivity for CK7, P504S, and PAX8, confirming the diagnosis of MTSCC. Postoperatively, the patient was managed conservatively without chemotherapy or radiotherapy and received only symptomatic treatment. During a 24-month follow-up, she demonstrated favorable recovery with no signs of recurrence or metastasis. ***Conclusion:*** This case underscores the critical role of imaging, histopathology, and immunohistochemistry in accurately diagnosing MTSCC. Early detection and timely surgical intervention can lead to positive outcomes, as illustrated by this patient's recovery. Further research is needed to explore the molecular mechanisms of MTSCC and establish robust guidelines for its management.

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Introduction

Mucinous tubular and spindle cell carcinoma (MTSCC) of the kidney is a rare renal epithelial tumor generally considered to be of low malignant potential. In 2004, the World Health Organization (WHO) classified MTSCC as a low-grade mucinous renal epithelial tumor with features of distal nephron differentiation. Histologically, it is characterized by a combination of tubular structures and spindle cells embedded within a mucinous stroma. MTSCC primarily arises from the renal cortex, with rare cases involving the renal medulla [1]. MTSCC accounts for less than 1% of all primary renal tumors. The patient age distribution ranges from 13 to 81 years, with an average age of 58 years, and it shows a marked female predominance, with a male-to-female ratio of approximately 1:3 [2].

Genomic studies of MTSCC have revealed widespread chromosomal abnormalities, including but not limited to abnormalities in chromosomes 1, 4, 6, 8, 9, 11, 13, 14, 15, 18, 22, and the X chromosome. In cases with classic morphological features, deletions of chromosomes 1, 4, 6, 8, 9, 13, 14, 15, and 22 are commonly observed. Conversely, MTSCC cases exhibiting unfavorable histological features – such as necrosis, solid growth patterns, single-cell infiltration, sarcomatoid transformation, lymphovascular invasion, and high mitotic activity – frequently display deletions in CDKN2A/2B (9p) and other more complex genomic alterations [3].

Clinically and pathologically, MTSCC typically appears as a well-circumscribed lesion within the renal parenchyma without a capsule. Immunohistochemical analysis commonly detects low-molecular-weight cytokeratins (such as CK8/18, CK19, and CK7), AMACR (P504s), E-cadherin, and epithelial membrane antigen (EMA). In addition, high-molecular-weight cytokeratin markers (e.g., 34βE12) and vimentin are often favorable [4]. Surgical resection remains the preferred treatment option for MTSCC [5].

In summary, MTSCC is a rare renal epithelial neoplasm that generally presents as a low-grade malignancy with distinct histological, immunohistochemical, and molecular genetic features. It predominantly occurs in middle-aged to elderly women, though rare cases with high-grade sarcomatoid components have been reported. While the prognosis is generally favorable following surgical excision, a subset of cases with high-grade morphological features may exhibit aggressive clinical behavior. This study aimed to present the clinical and pathological features of a case of MTSCC, focusing on its immunohistochemical characteristics, to clarify diagnostic criteria and guide clinical treatment. The successful management of this case provides valuable experience and a reference for the future diagnosis and treatment of MTSCC.

Materials and Methods

Case

A 52-year-old female patient presented to an external hospital 6 days ago with abdominal pain, where a CT scan revealed a mass in the left kidney. Seeking further diagnosis and treatment, she was referred to the Hangzhou Traditional Chinese Medicine Hospital on October 8, 2021. Throughout her illness, the patient maintained clear consciousness, good mental status, unaffected appetite, undisturbed sleep, normal bowel movements, and no symptoms of urinary frequency, urgency, pain, or hematuria, with no significant weight loss. Physical examination indicated normal skin and conjunctiva, clear lung breath sounds, no tenderness or rebound tenderness in the abdomen, painless percussion over the kidney area, and absence of swelling in both lower limbs. The patient had a history of intestinal obstruction

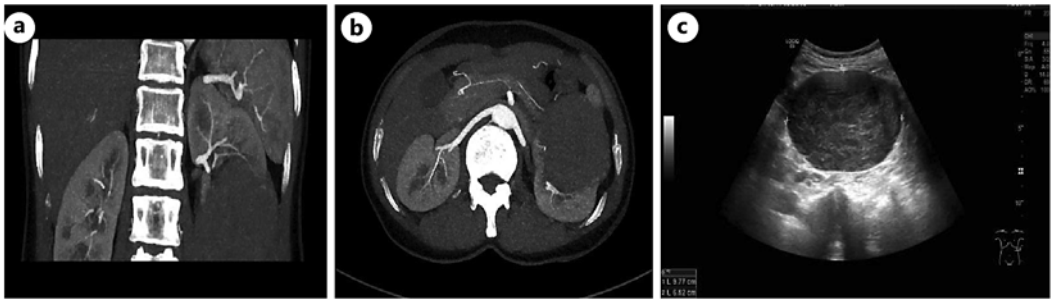

Fig. 1. CT and ultrasound imaging of the kidney reveal a mass lesion in the left kidney. **a** Coronal CT angiography unveils a low-density round lesion in the left kidney. **b** Axial CT angiography displays a low-density lesion with smooth margins in the left kidney. **c** Ultrasound imaging demonstrates a hypoechoic mass lesion in the left kidney.

and had undergone surgical treatment in the past, details of which were unclear, and denied any other surgical or traumatic history.

Upon admission, a CT angiography revealed a low-density round lesion in the left kidney supplied by the left renal artery (Fig. 1a, b). An ultrasound examination also showed a hypoechoic lesion in the left kidney (Fig. 1c). Based on these findings, the patient underwent laparoscopic radical nephrectomy for renal cell carcinoma on October 11, 2021, in the urology department. During surgery, a tumor measuring approximately 10 cm in diameter was found in the mid-lower pole of the left kidney.

Pathological examination of the excised left kidney specimen showed a total size of 16.1 cm × 6.2 cm × 5.3 cm, with the tumor in the mid-lower pole of the left kidney measuring about 10.1 cm × 8.5 cm × 6.6 cm (Fig. 2). The tumor appeared solid with a medium-soft consistency, exhibited a gray-white and pale yellow color on the cut surface, was encapsulated by a 0.1-cm- to 0.2-cm-thick membrane (Fig. 3a), and comprised spindle cells and flattened cuboidal epithelial cells arranged in elongated tubular structures with minimal cellular atypia and varying degrees of mucinous stroma. The tumor had clear margins with surrounding normal tissue, and the remaining kidney tissue showed no significant abnormalities on sectioning (Fig. 3b–d). Immunohistochemical analysis revealed that the tumor cells exhibited partial positivity for CK7 (Fig. 3e), positive expression of P504S (Fig. 3f), positive expression of PAX8V, and Alcian blue staining highlighted the stromal components (Fig. 3h). Based on the morphological characteristics observed with HE staining and the immunohistochemical findings, the pathological diagnosis was confirmed as MTSCC.

Postoperatively, the patient received symptomatic Alexin (5.0 bid) treatment for infection control, spasm relief, and pain management. No radiotherapy or chemotherapy was administered. During 24 months of follow-up, the patient's recovery was favorable, with no signs of recurrence or metastasis.

Immunohistochemistry

In this study, the excised kidneys were initially fixed in 10% formaldehyde, embedded in paraffin, sectioned, and stained with hematoxylin and eosin, Alcian blue, and periodic acid-Schiff. Immunohistochemical studies were performed on dewaxed 4-micron-thick sections using a streptavidin-biotin complex and serum solution for antigen detection (performed using the Ventana ES automated immunostainer system in Strasbourg, France). The antibodies used targeted the following antigens: EMA (E29; 1/25; Dako, Trappes, France), vimentin (V9 1/100; Immunotech, Marseille, France), cytokeratin complex AE1/AE3

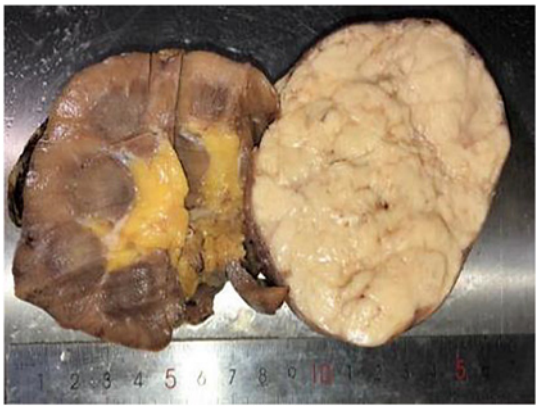

Fig. 2. Gross specimen of the left kidney and pathological examination of the tumor.

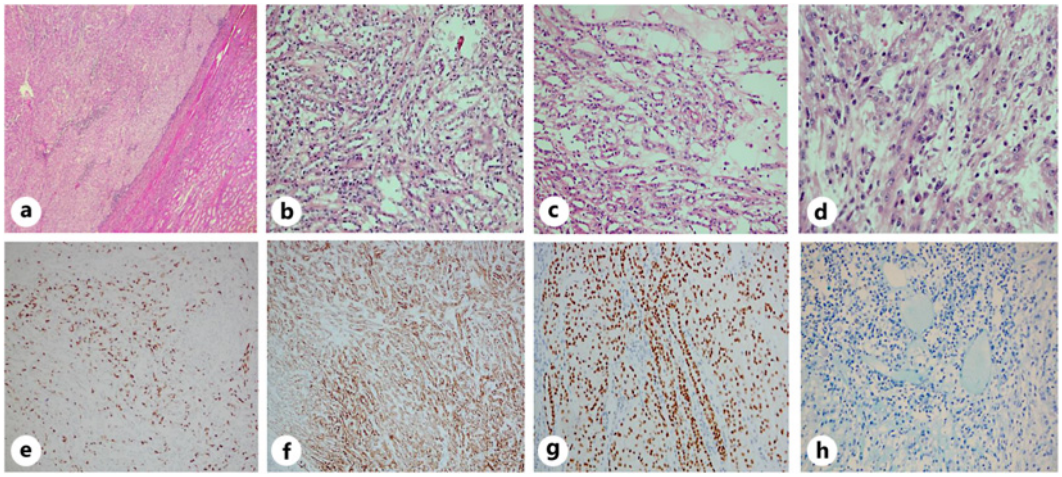

Fig. 3. Histopathological features of mucinous tubular and spindle cell renal cell carcinoma (MTSCC). **a** Well-defined tumor boundary with evident capsule (magnification ×40). **b**, **c** Tumor cells exhibit spindle or tubular structures with mucinous stroma (magnification ×200). **d** High-power view of tumor cells' HE-stained morphology (magnification ×400). **e** Immunohistochemistry shows positive expression of CK7 (magnification ×200). **f** Immunohistochemistry shows positive expression of P504S (magnification ×200). **g** Immunohistochemistry shows positive expression of PAX8 (magnification ×200). **h** Alcian blue (AB) staining highlights the stromal components (magnification ×200).

(prediluted, Ventana, Strasbourg), E-cadherin (4A2C7; 1/100; Zymed, Montluçon, France), cytokeratin 19 (BA-17; 1/50; Dako), cytokeratin 7 (OV-TL 12/30; 1/100; Dako), 34βE12 (1/150; Dako), polyclonal carcinoembryonic antigen (1/1,000; Dako), epithelial cell adhesion molecule-1 (1/100; Dako), CD10 (ALD1; 1/20; Novocastra, Newcastle, UK), CD15 (80H5; 1/25; Immunotech), and KI-67 (MIB-1; 1/50; Immunotech).

Discussion

This study reports the diagnostic and therapeutic process of a 52-year-old female patient (Fig. 4). CT angiography and ultrasound revealed a low-density, round-like lesion in the left kidney, supplied by the left renal artery. A laparoscopic radical nephrectomy was subsequently performed, during which a tumor measuring approximately 10 cm in diameter was identified. Pathological examination provided further details regarding

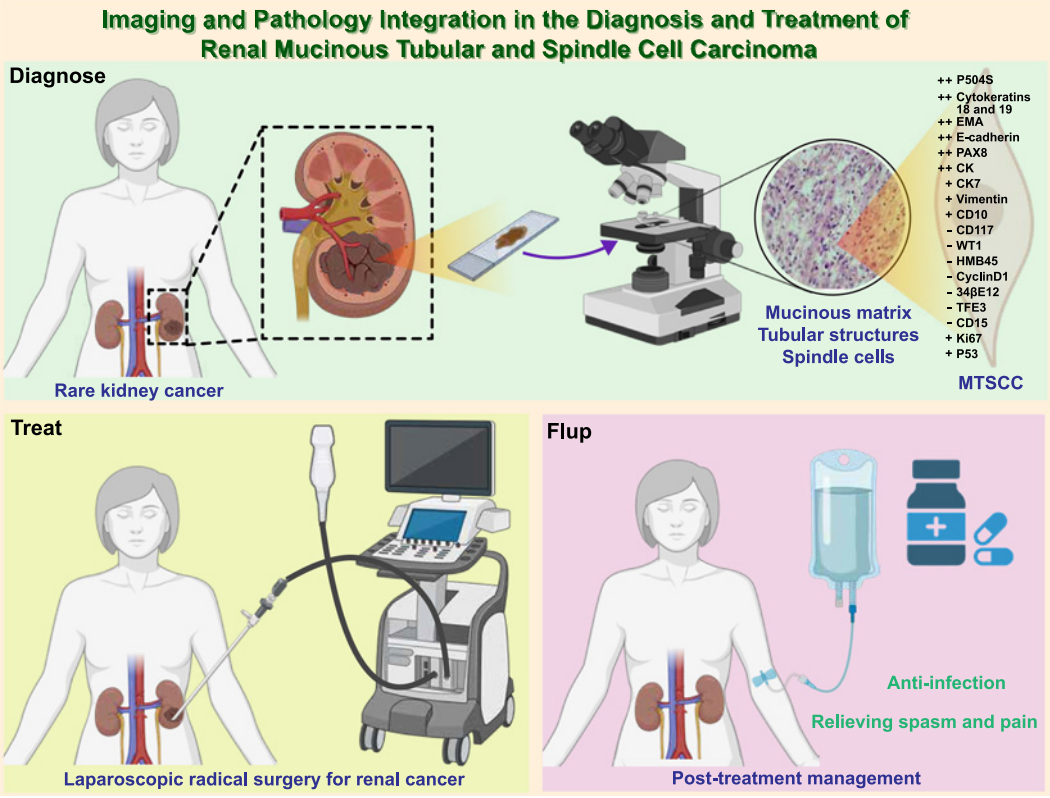

Fig. 4. Overview of imaging-pathological correlation and treatment in collecting duct carcinoma of the kidney with cuboidal cells of the renal tubules with mucin production.

the tumor's size, morphology, and cellular composition, with immunohistochemical staining confirming the expression of specific cellular markers. Based on the morphological and immunohistochemical features, the tumor was ultimately diagnosed as MTSCC.

Following surgery, the patient received symptomatic treatment, including anti-infective and antispasmodic analgesic therapy, without radiation or chemotherapy. A 24-month follow-up revealed good recovery with no evidence of recurrence or metastasis. MTSCC is characterized by distinctive histological features, primarily comprising a mixture of tubular structures and spindle cells within a mucinous stroma [6]. Immunohistochemistry is crucial for diagnosing MTSCC, with positive staining for low-molecular-weight cytokeratins (e.g., CK8/18, CK19, and CK7), AMACR (P504S), E-cadherin, and EMA [4]. The expression patterns of these markers help distinguish MTSCC from other renal cell carcinomas.

MTSCC must be differentiated from other renal tumors in clinical practice as their histological and immunohistochemical features differ. Accurate diagnosis depends on thorough histological evaluation and immunohistochemical analysis. Due to the low incidence of MTSCC, there is limited evidence to guide treatment [7]. Surgical resection, partial or total nephrectomy, is the primary treatment [5]. In line with findings by Ivey et al. [5], this case involved a partial nephrectomy of the left kidney to remove the tumor. Due to MTSCC's low aggressiveness and metastatic potential, patients typically experience favorable long-term survival following surgery. Depending on the patient's condition, adjuvant treatments such as targeted therapy or immunotherapy may be considered, though clinical data on these therapies remain limited [8].

In this case, the patient only received symptomatic treatment post-surgery, including anti-infective therapy with Alexin (5.0 mg, BID) and antispasmodic analgesics without radiation or chemotherapy. During the 24-month follow-up, the patient remained healthy, with no signs of recurrence or metastasis.

Conclusion

This case of MTSCC provides valuable insights, documenting the diagnostic and therapeutic process for this rare renal tumor and highlighting the potential for atypical clinical presentations and favorable postoperative recovery. However, this study has certain limitations. It did not explore the molecular mechanisms underlying the development of MTSCC or investigate its genetic background. Future research should focus on elucidating the molecular pathogenesis of MTSCC and conducting genetic screening to identify potential therapeutic targets. More case studies are needed to validate optimal treatment strategies and improve early diagnostic accuracy. Additionally, long-term follow-up studies are essential to understanding the patterns of recurrence and metastasis and determining overall survival, ultimately contributing to better treatment strategies and improved quality of life for patients. The CARE Checklist has been completed by the authors for this case report, attached as online supplementary material (for all online suppl. material, see https://doi.org/10.1159/000543004).

Statement of Ethics

This study was approved by the Ethics Committee of Hangzhou Hospital of Traditional Chinese Medicine (Approval No. 2022LH001). Relevant guidelines and regulations were performed for all procedures. In addition, the writing and presentation of this case report strictly follow the Case Report (CARE) guidelines to ensure the completeness and transparency of the report. Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

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Author Contributions

Yu Zhang contributed to data acquisition, analysis, and interpretation of imaging and pathological findings. Yu Long was involved in drafting the manuscript, revising it critically for intellectual content, and coordinating the study. Jie Wang provided study design, supervised the project, and approved the final manuscript for submission. All authors read and approved the final version of the manuscript.

Data Availability Statement

The data that support the findings of this study are not publicly available due to privacy considerations for the research participants. However, these data are available from the corresponding author, Jie Wang (email: seaangel303@126.com), upon reasonable request.

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