UNUSUAL TUMOURS OF THE KIDNEY AND URINARY TRACT IN CHILDREN AND ADULTS

Other sarcomas of the kidney

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Mesenchymal tumors mostly **PEDIATRIC:**

- CLEAR CELL SARCOMA
- RHABDOID TUMOR
- CONGENITAL MESOBLASTIC NEPHROMA
- OSSIFYING RENAL TUMOR OF INFANCY

Mesenchymal tumors mostly **ADULT:**

- LEIOMYOSARCOMA
- ANGIOSARCOMA
- RHABDOMYOSARCOMA
- SYNOVIAL SARCOMA
- RENAL EWING SARCOMA
- SOLITARY FIBROUS TUMOR
- ANGIOMYOLIPOMA
- LEIOMIOMA, HEMANGIOMA, HEMANGIOBLASTOMA...
INTRATUMORAL HETEROGENEITY
CLEAR CELL SARCOMA OF THE KIDNEY
WILMS T.
Vs
NEUROBLASTOMA

23 cm
<table>
<thead>
<tr>
<th>TUMOR TYPE</th>
<th>CK</th>
<th>DES</th>
<th>MIOG</th>
<th>CD99</th>
<th>WT1</th>
<th>NKX2-2</th>
<th>ETV4</th>
<th>BCOR</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ewing sarcoma</td>
<td>+</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>++</td>
<td>-</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>CIC-DUX4 sarcoma</td>
<td>+</td>
<td>-</td>
<td>-</td>
<td>+</td>
<td>++</td>
<td>-</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>CCSK</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>++</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>PD.Synovial</td>
<td>+</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>RMS</td>
<td>+</td>
<td></td>
<td>+</td>
<td>-</td>
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<tr>
<td>DSRCT</td>
<td>++</td>
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<td>++</td>
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<td>-</td>
<td>-</td>
</tr>
<tr>
<td>Blastemal Wilms</td>
<td>/+</td>
<td></td>
<td>-</td>
<td>+</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>-</td>
</tr>
</tbody>
</table>
**IMMUNOHISTOCHEMICAL PANEL**

- Vimentin, BCL2, TLE1, SATB2...
- Cyclin D1 (biphasic pattern)
- BCOR (biphasic pattern)
MOLECULAR ALTERATIONS IN CCSK

ITD BCOR

YWHAE – NUTM2 B

Double negative
BCOR family of neoplasms

- **URCS/PMMTI**
- **HG-ESSs**
- **CCSK**
- Renal sarcoma *BCOR-CCNB3*
- Bone/soft tissue sarcoma *BCOR-CCNB3*
- Neuroepit Tumor. CNS
Primary Renal Sarcomas With BCOR-CCNB3 Gene Fusion
A Report of 2 Cases Showing Histologic Overlap With Clear Cell Sarcoma of Kidney, Suggesting Further Link Between BCOR-related Sarcomas of the Kidney and Soft Tissues

Age: 11-12 yr
Renal mass: 12-23 cm
Solid-cystic
Abdominal relapse

BCOR-CCNB3

Am J Surg Pathol, vol 41, 12, Dec 2017
## NEOPLASMS RELATED TO BCOR OR SYNOVIAL SARCOMA

<table>
<thead>
<tr>
<th></th>
<th>CCSK</th>
<th>BCOR-CCNB3 Renal Sarcoma</th>
<th>URCS/PMMTI</th>
<th>Bone/soft tissue sarcoma BCOR-CCNB3</th>
<th>Renal synovial sarcoma</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Age (yr)</strong></td>
<td>3</td>
<td>11.5</td>
<td>&lt;1</td>
<td>Adolescence</td>
<td>43</td>
</tr>
<tr>
<td><strong>Morphology</strong></td>
<td>![Image]</td>
<td>![Image]</td>
<td>![Image]</td>
<td>![Image]</td>
<td>![Image]</td>
</tr>
<tr>
<td><strong>Genetics</strong></td>
<td>BCOR ITD (90%) YWHAE-NUTM2 (&lt;10%)</td>
<td>BCOR-CCNB3 fusion</td>
<td>BCOR ITD</td>
<td>BCOR-CCNB3 fusion</td>
<td>SS18-SSX fusion</td>
</tr>
</tbody>
</table>
## INMUNOHISTOCHEMICAL PANEL

<table>
<thead>
<tr>
<th>Antibody</th>
<th>CCSK</th>
<th>BCOR-CCNB3</th>
<th>URCS/PMMTI</th>
<th>Bone/soft tissue sarcoma</th>
<th>Renal synovial sarcoma</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>Renal Sarcoma</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>PAX8</td>
<td>20%</td>
<td>50%</td>
<td>N/A</td>
<td>50%</td>
<td>NEGATIVO</td>
</tr>
<tr>
<td>BCOR</td>
<td>POSITIVE</td>
<td>POSITIVE</td>
<td>POSITIVE</td>
<td>POSITIVE</td>
<td>50%</td>
</tr>
<tr>
<td>CYCLIN D1</td>
<td>POSITIVE</td>
<td>POSITIVE</td>
<td>POSITIVE</td>
<td>POSITIVE</td>
<td>60%</td>
</tr>
<tr>
<td>SATB2</td>
<td>POSTIVE (42%)</td>
<td>POSITIVE</td>
<td>POSITIVE</td>
<td>POSITIVE</td>
<td>POSTIVE (60%)</td>
</tr>
<tr>
<td>TLE1</td>
<td>POSTIVE (80%)</td>
<td>POSITIVE</td>
<td>POSITIVE</td>
<td>POSITIVE</td>
<td>POSITIVE</td>
</tr>
</tbody>
</table>
ITD BCOR
YWHAE-NUTM2

CCSK

BCOR-CCNB3
RENAL SARCOMA

Histological images of CCSK and BCOR-CCNB3-related renal sarcoma.
MOLECULAR ALTERATIONS IN CCSK

ITD BCOR

YWHAE – NUTM2 B

Double negative → BCOR-CCNB3
Case 1:
- Female, 72
- Cystic mass
- 4 cm

Caso 2:
- Male, 21
- Solid mass
- 15 cm

MEIS1-NCOA2
PRIMITIVE SPINDLE CELL RENAL SARCOMA MEIS1-NCOA2
PRIMITIVE SPINDLE CELL RENAL SARCOMA

MEIS1-NCOA2

SPINDLE CELL PATTERN WITH PRIMITIVE ROUND CELLS

NONSPECIFIC IMMUNOHISTOCHEMICAL PANEL
(Ciclina D1, BCI2, CD99, WT1, CD57, TLE1, BCOR...)

MEIS1-NCOA2
Mesenchymal chondrosarcoma (HEY1-NCOA2)

Angiofibroma (AHRR-NCOA2)

Spindle cell rhabdomyosarcoma (NCOA2-MEIS1)

Alveolar Rhabdomyosarcoma (NCOA2-PAX3)
CCSK

METANEFRIC STROMAL TUMOR

SYNOVIAL SARCOMA

CIC-fused SARCOMA

MEIS1-NCOA2

PRIMITIVE SPINDLE CELL SARCOMA

SPINDLE CELL MORPHOLOGY

YWHA2-NUTM2

ITD BCOR

BCOR-CCNB3

SS18-SSX

CD34 +

MEIS1-BCOR
Male, 27. Backache

Painful abdominal mass

ECO: Mass at left hypochondrium

CT-scan

8x11x10 cm
Diagnostic possibilities

- CCSK with BCOR-CCNB3
- CCSK (ITD-BCOR)
- CCSK (YWHAE-NUTM2)
- Renal sarcoma MEIS1-NCOA2

MDM2 NEGATIVE
BCL2 focal
Cyclin D1: focal
CD99: focal

FISH
Sequencing
BCOR Overexpression in Renal Malignant Solitary Fibrous Tumors
A Close Mimic of Clear Cell Sarcoma of Kidney

Pedram Argani, MD,* Yu-Chien Kao, MD,† Lei Zhang, MD,‡ Yun-Shao Sung, MS,§
Rita Alaggio, MD,§ David Swanson, BSc,|| Andres Matoso, MD,* Brendan C. Dickson, MD,||
and Cristina R. Antonescu, MD,‡

- 5 cases (age 30-60)
- D/d CCSK
- BCOR ++++
- CD34 negative or focal
- STAT6 ++++

Am J Surg Pathol 2019, Mar
Malignant solitary fibrous tumor
NGS (Archer Sarcoma Panel)
Take home messages:

- Many soft tissue sarcomas can have a renal involvement
- There are primary renal sarcomas without a soft-tissue counterpart
- Intratumoural heterogeneity
- Considerable overlapping in morphology and immunohistochemistry
- BCOR, NCOA2….fashionable!
- BCOR expression: CCSK, mSFT, primitive MEIS1-NCOA2 sarcoma
- Renal and extrarenal SFT express BCOR