

UIC Department of
UNIVERSITY OF ILLINOIS Pathology
AT CHICAGO *COLLEGE OF MEDICINE*

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Illinois Registry of Anatomic Pathology

Case histories and diagnoses
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Case 1: Tropical Spastic Paraparesis/HTLV-1 Associated Myelopathy (HAM)

Daniel S. Massi, MD; Manuel F. Utset, MD, PhD

Clinical History:

A 57 year old black female of Caribbean descent had a 14-year history of progressive upper and lower extremity weakness. In the last year, she became bed-bound requiring nursing home placement. She presented with decubitus ulcers and sepsis and expired a week later.

Diagnosis: Tropical Spastic Paraparesis/HTLV-1 Associated Myelopathy (HAM)

Differential Diagnosis: Is broad and include these categorical disorders

- Degenerative
- Demyelinating
- Toxic/Metabolic
- Paraneoplastic
- Infectious
- Neoplastic

Key Microscopic Features:

- Infiltration of T-lymphocytes and microglia with CD4 predominance early in the disease, switching to CD8 in chronic disease (> 5 years)
- Increase in foamy macrophages as total percentage of inflammatory cells over time
- Lateral and anterior column degeneration with axonal damage and demyelination
- Spinal cord atrophy
- Leptomeningeal fibrosis

Immunohistochemical and special stains: Immunohistochemical (Neurofilament, CD68, CD20, and CD3) and special stain (Myelin) workup revealed loss of axons and myelin in the lateral corticospinal tracts and anterior columns with an inflammatory infiltrate of microglia, T-lymphocytes, and foamy macrophages.

Discussion:

- HTLV-1 is endemic in the Caribbean, South America, Africa, and Japan.
- The endemic rate in some areas is up to 30% with around 1% of infected individuals developing disease.
- Modes of transmission include sexual contact, blood products, and vertical transmission (mainly breast feeding).
- The virus is largely non-infectious in the cell-free state and is transferred by cell-to-cell contact mediated by a virological synapse, orchestrated by the viral Tax protein.

References:

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Case 2: Urothelial Carcinoma of the Renal Pelvis

Jennifer Prather, MD; Andre Balla, MD; Suman Setty, MD

Clinical history:

The patient, a 77 year-old man with hypertension, diabetes mellitus, and COPD, presented with a small bowel obstruction and a recent history of urinary tract infection with hematuria. CT scan revealed a 7 cm renal mass involving the entire right kidney.

Diagnosis: Urothelial Carcinoma of the Renal Pelvis

Differential Diagnosis:

- Renal Cell Carcinoma, Clear Cell Type
- Collecting Duct Carcinoma

Key Morphologic Features:

- Gross: Solid tumor with infiltrating pattern, obliteration of pelvis and calyces
- Microscopic: Sheets of infiltrating, high-grade, pleomorphic cells, some with abundant cytoplasm and nests of cells with clear cytoplasm; urothelial carcinoma *in situ* present
- Immunohistochemical stains: Positive for CK 19, HMWK, CEA, and p63. Negative for RCC, vimentin, and mucicarmine
- Cytogenetics: Polysomic for chromosomes 3, 7 and 17; lacks del 3p

Discussion:

- 7-8% of renal malignancies are urothelial in origin
- 2 gross presentations: papillary mass in renal pelvis, solid mass infiltrating renal parenchyma
- Microscopically, tumor cells are usually high grade with a wide variety of growth patterns and cytologic features
- Immunohistochemical stains for CK19, HMWK, CEA and p63 are positive in urothelial carcinoma, while vimentin is negative.
- Polysomy for 3, 7, and 17 are pathognomonic for urothelial carcinoma.
- These patients need to be investigated and monitored for a concurrent or a future urinary tract malignancy

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1. Dabbs, D. Diagnostic Immunohistochemistry. New York: Churchill Livingstone, 2002.
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Case 3: Localized Tubulo-glomerulocystic Change associated with Tuberous Sclerosis

Marta Helenowski, MD; Michael Pins, MD

Clinical History:

A 4 month old female was born at 34 5/7 weeks gestation by emergency cesarean section secondary to a fetal arrhythmia. Upon delivery she presented with rhabdomyomas in the ventricular wall of the heart, subependymal nodules in the brain, and a lower pole kidney mass. Her family history did not reveal any genetic abnormalities. In conclusion, she was diagnosed clinically with tuberous sclerosis.

Diagnosis: Localized Tubulo-glomerulocystic Change associated with Tuberous Sclerosis

Differential Diagnosis:

- Segmental renal dysplasia
- Glomerulocystic kidney disease
- Angiomyolipoma with epithelial cysts
- Tubulo-cystic carcinoma of the kidney
- Tubulo-glomerulocystic change

Key Morphologic Features:

- Two components: tubulocystic and glomerulocystic
- Characteristic hyperplastic epithelium with eosinophilic cytoplasm

Discussion:

- Tuberous Sclerosis Complex often presents with a variety of cystic and/or neoplastic renal manifestations
- Tubulo-glomerulocystic change associated with Tuberous Sclerosis
- Unilateral, segmental presentation very rare

References:

1. Bisceglia M, Galliani CA, Senger C, Stallone C, Sessa A. Renal cystic diseases: A review. *Adv Anat Pathol.* 13: 26-56, 2006.
2. Martignoni G, Pea M, Cossu Rocca P, Bonetti F. Renal pathology in the tuberous sclerosis complex. *Pathology.* 35: 505-512, 2003.
3. Weber M, Risdon R, Malone M, Duffy P, Sebire N. Isolated unilateral tuberous sclerosis-associated renal cystic disease in a neonate. *Fetal and Pediatric Pathology.* 24:267-275, 2005.

Case 4: Congenital gastric teratoma, mature

Seung Park, MD; Steven Garzón, MD

Clinical History:

A newborn male born to a 20 year old G1P0 with no significant past medical or obstetric history at 40 6/7 weeks via Caesarian section, was found to have a large abdominal mass. Disseminated intravascular coagulopathy ensued; a presumptive diagnosis of neuroblastoma was made, and the patient was given empiric chemotherapy. The mass increased in size as a result; therefore, an exploratory laparotomy with partial gastrectomy was performed.

Diagnosis: Congenital gastric teratoma, mature

Differential Diagnosis:

- Immature teratoma
- Immature or mature teratoma with yolk sac tumor elements

Key Morphologic Features:

- Grade does not predict behavior
- Tissue representing each of the three germ cell layers
- Grade 0 (mature): mature tissue only
- Grade 1 (immature): abundant mature tissue, few foci of embryonic appearing neuroglial or neuroepithelial components (less than one low power field)
- Grade 2 (immature): embryonic appearing neuroglial or neuroepithelial components, 1 < 4 low power fields
- Grade 3 (immature): numerous neuroglial or neuroepithelial components present occupying 4+ low power fields

Discussion:

- Rare location for a common childhood tumor
- Disseminated intravascular coagulopathy is a very rare complication
- Resection with negative margins is generally curative regardless of grade, except when yolk sac tumor elements are present
- Higher grade tumors are associated with increasing incidence of yolk sac elements
- Increased serum AFP is correlated with the presence of yolk sac tumor elements

References:

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Case 5: Clear Cell Renal Cell Carcinoma with Extensive Rhabdoid Differentiation

Yi Zhou, MD, PhD; Carey August, MD

Clinical History:

A 46 year old male presented to his primary care physician with left flank pain. Both CT scan and MRI showed an 11.0 cm mass in the left kidney. The patient underwent left radical nephrectomy.

Diagnosis: Clear Cell Renal Cell Carcinoma with Extensive Rhabdoid Differentiation

Differential Diagnosis:

- Malignant rhabdoid tumor
- Renal medullary carcinoma
- Clear cell renal cell carcinoma
- Unclassified renal cell carcinoma
- Collecting duct carcinoma
- Urothelial carcinoma
- Primary renal synovial sarcoma
- Epithelioid angiomyolipoma
- Renal rhabdomyosarcoma
- Renal lymphoma

Key Morphologic Features:

- **Gross:** The tumor appears to involve renal sinus, renal pelvis, ureter, and renal vein. Sectioned tumor reveals tan/white, hemorrhagic and multicystic cut surface with focal tan/bright-yellow areas.
- **Microscopic:** Predominantly rhabdoid cells characterized by abundant eosinophilic cytoplasm with an irregular eccentric nucleus and a rounded eosinophilic cytoplasmic inclusion. Transition between rhabdoid cells and conventional clear cell type of renal cell carcinoma is present. Tumor cells are immunoreactive for vimentin, AE1+3, MAK-6, and CD10.

Discussion:

- Studies showed that clear cell renal cell carcinoma (RCC) with rhabdoid component represent 4.5% of all RCCs.
- Identification of the rhabdoid phenotype in adult RCC has important diagnostic and prognostic value.

References:

1. Gökden N, Nappi O, Swanson PE, Pfeifer JD, Vollmer RT, Wick MR, Humphrey PA. Renal cell carcinoma with rhabdoid features. *Am J Surg Pathol.* 24:1329-38, 2000.
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Case 6: Extranodal, Peripheral T-cell Lymphoma, (NOS); most likely immunodeficiency related (EBV positive)

Liaqat Ali MD, Sujata Gaitonde MD, Frederick Behm MD

Clinical History:

A 51 y.o. male was transferred to UIC for further management of metastatic disease. There was no past medical history until 6 months ago when he had a car accident. He experienced leg pain and enlarging leg masses attributed to a hematoma from trauma. Lower extremity CT scan revealed bilateral masses with osseous invasion. Imaging also revealed metastasis.

Diagnosis: Extranodal, Peripheral T-cell Lymphoma, not otherwise specified; most likely immunodeficiency related (EBV positive).

Differential Diagnosis:

- Extranodal NK/T cell lymphoma, nasal type
- Angioimmunoblastic T cell Lymphoma
- Peripheral T-cell lymphoma, NOS

Key Morphologic Features:

- Monomorphous cellular infiltrate with extensive necrosis
- Medium to large sized cells, pleomorphic nuclei with irregular nuclear contours, prominent nucleoli and mitotic figures

Immunohistochemistry:

Positive: CD3, CD5, CD7, CD8, partial CD2, Granzyme B, TIA, vimentin.

Negative: CD45, CD20, CD4, CD56, AE1/3, S100, Melan-A, chromogranin & HMB-45.

Immunophenotype by flow cytometry:

Positive: CD45, CD3, CD8, CD2, CD5, TCR $\alpha\beta$.

Negative: CD4, , CD7, CD56, CD1a, CD19.

Discussion:

- HIV Related T cell Lymphoma categories include:
 - Enteropathy Type T cell Lymphoma.
 - Angio-immunoblastic T cell Lymphoma
 - Peripheral T cell Lymphoma, NOS
 - HTLV-1 associated Adult T cell Lymphoma/Leukemia
- HIV related PTCLs are uncommon, biologically diverse and have a poor prognosis
- Treatment of T- cell lymphomas lag behind that of B-cell NHL due to their rarity and biological heterogeneity
- Majority of the patients present with advanced lymphoma (stage III or IV)
- Involved sites can be nodal (17%) or extra nodal (83%)
- Treatment includes chemotherapy or chemotherapy plus radiation

References:

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