Rush University Medical Center

Department of Pathology



Illinois Registry of Anatomic Pathology

April 26, 2021

**CASE #1**

**Presenter:** Matthew Vega, MD

**Attending:** Xinhai R. Zhang, MD, PhD

**CASE HISTORY**: The patient is a 41-year old male whose past medical history is significant for that of bipolar disorder and tobacco use disorder. The patient presents for management of a hypomanic episode that has been occurring for approximately five to six months, syncopal events, and new onset seizures. The patient believes that this persistent hypomanic episode was triggered to an electrocution event at work. Imaging demonstrated a left temporal lesion with cerebral edema and areas of hemorrhage and necrosis. A left craniotomy was performed for the resection of the left enhancing lesion.

**DIAGNOSIS**: Diffuse midline glioma, H3 K27M-mutant, WHO grade 4

**DIFFERENTIAL DIAGNOSIS:**

* Lymphoma
* Anaplastic oligodendroglioma WHO grade 3
* Glioblastoma, IDH-wildtype, WHO grade 4
* Astrocytoma, IDH-mutant, WHO grade 4
* Diffuse midline glioma, H3 K27M-mutant, WHO grade 4

**DISCUSSION:**

* Diffuse midline glioma is mainly in midline structures, but can be cortically-based. The final diagnosis is based on H3 K27M mutation confirmed by immunohistochemistry or genetic sequencing
* Possible difference among prognoses as aggressive surgical intervention is likely more plausible
* Immunohistochemical stains for H3 K27M is standard for suspicious midline tumors but is not protocol for more cortically-based lesions

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**CASE #2**

**Presenter:** Natalia Lashmanova, MD

**Attending:** Ira Miller, MD, PhD

**CASE HISTORY** A previously healthy 11-year-old boy presented with a soft tissue mass in the left lateral ankle causing some symptoms of discomfort. The tumor was present with minimal growth for several years. The past medical and family history are otherwise unremarkable.

Physical exam showed a nontender subcutaneous 4 cm x 7 cm x 3 cm mobile mass over the anterolateral left distal tibia. There was full range of motion and no sensory loss.

MRI showed a T1 hyperintense lesion measuring 2.9 x 1.4 cm in the anterolateral aspect of the left ankle which appeared somewhat encapsulated and caused mild mass effect on the adjacent extensor tendons.

During the resection procedure the surgeon was unable to separate the superficial peroneal nerve from the tumor, and the tumor was felt to be more firm than typical lipomas. The nerve was sacrificed.

**DIAGNOSIS**: Lipomatosis of nerve

**DIFFERENTIAL DIAGNOSIS of Lipomatosis of Nerve**

* Traumatic neuroma
* Diffuse lipomatosis
* Lipoblastoma

**DISCUSSION:**

* Lipomatous tumors account for less than 10% of all soft tissue lesions in pediatric patients
* The epidemiology and the prevalence of histological types of adipose tissue tumors differ in the pediatric population compared with adults
* Chromosomal and genetic rearrangements are similar to those of adult cases in each histological type
* The identification of characteristic molecular alterations for many tumor types has been central to classification and has helpful diagnostic applications
* Awareness of the various syndromic associations for lipomatous neoplasms is also important when encountering fatty tumors in pediatric patients

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**CASE #3**

**Presenter:** Joanna Solarewicz, DO

**Attending:** Indu Agarwal, MD

**CASE HISTORY**: The patient is a 28-year-old male with a past medical history of psoriatic arthritis (on Humira) and pilonidal cyst status post excision, who presented with a “lump” in the left scrotum that has been present for about a year and is now causing him significant discomfort. This prompted an evaluation by general surgery for a possible hernia. Physical examination revealed that the mass was more likely to be testicular in origin, and as a result, the patient was referred to the urologist’s office for further evaluation. A scrotal ultrasound was ordered and showed a solid-appearing, hypoechoic, left intratesticular lesion measuring approximately 1.6 x 1.4 x 0.6 cm with internal flow on color Doppler imaging, concerning for a testicular neoplasm. The lesion did not appear to invade the tunica albuginea. Tumor markers were also ordered: AFP 2.15 ng/mL (range: 0.00-10.00 ng/mL) and β-hCG <1 mIU/mL (range: 0-4 mIU/mL). The patient agreed to undergo a left radical orchiectomy.

**DIAGNOSIS**: Sex-cord stromal tumor-mixed type involving testis

**DIFFERENTIAL DIAGNOSIS:**

* Mixed Germ Cell Tumor
* Sertoli Cell Tumor
* Leydig Cell Tumor
* Granulosa Cell Tumor, adult type
* Carcinoid Tumor
* Mixed Sex Cord-Stromal Tumor
* Adenomatoid Tumor
* Myoid Gonadal Stromal Tumor

**DISCUSSION:**

* Granulosa and Leydig cell tumors are both frequently associated with gynecomastia.
* Mixed sex cord stromal tumor is a rare tumor of descended testis and constitutes less than 1% of testicular tumors.
* Sex cord stromal tumors usually present with painless testicular swelling.
* Usual germ cell markers, including beta-hCG and AFP, are found within normal limits in patients with sex cord stromal tumors.
* Granulosa cell tumors grossly show a gray-yellow to tan nodule with solid and cystic areas.
* Leydig cell tumors grossly show a variably yellow-brown or tan, well-circumscribed or lobulated intratesticular nodule.
* The standard treatment for sex cord stromal tumors is radical inguinal orchiectomy.
* CT of the chest and abdomen should be performed after determining a pathologic diagnosis, because about 20% of sex cord stromal tumors are metastatic at initial diagnosis.
* Retroperitoneal lymph node dissection is more important for staging purposes than therapeutic intervention.

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**CASE #4**

**Presenter:** Abdullah Almajnooni, MBBS

**Attending:** Nicolas Lopez-Hisijos, MD

**CASE HISTORY**: This case is of a 39-year-old woman with Down syndrome who presented to an outside institution with a mass in the right upper arm noticed by her mother. On physical examination, the mass was large and warmth to touch. MRI showed a 6.1 x 9.4 x 17 cm lobulated mass centered in the triceps muscle, containing a cystic component and peripheral solid components with mass effect. She was referred to RUMC for further diagnostic work-up and management.

**DIAGNOSIS**: Dedifferentiated liposarcoma

**DIFFERENTIAL DIAGNOSIS**:

* Anaplastic lymphoma and Extranodal Hodgkin disease
* Myxoinflammatory fibroblastic sarcoma
* Epithelioid sarcoma
* Undifferentiated pleomorphic sarcoma
* Abscess

**DISCUSSION:**

* Sarcoma can present with extensive inflammation mimicking abscess
* 60 reported cases of sarcoma mimicking abscess clinically, radiographically and morphologically
* 40% of reported cases experienced a delay in diagnosis
* A negative biopsy should not be assumed as benign. Excision is warranted if clinically indicated, not to delay treatment

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**CASE #5**

**Presenter:** Dr. Subramanya S Mallikarjunappa

**Attending:** Dr. Ram Al-Sabti

**CASE HISTORY:** A 68-year old male with a past medical history of right renal cell carcinoma 12 years ago status-post partial nephrectomy now presents with renal colic. CT scan showed a renal mass at the hilum measuring 2.8 x 1.8 cm. Clinically, the presentation was suspicious for recurrence of renal cell carcinoma/possible metastasis to the hilar lymph node. The patient underwent total nephrectomy.

**DIAGNOSIS:** Pleomorphic hyalinizing angiectatic tumor (PHAT)

**DIFFERENTIAL DIAGNOSIS:**

* Recurrence of renal clear cell carcinoma
* Schwannoma
* Pleomorphic hyalinizing angiectatic tumor (PHAT)
* Hemosiderotic Fibrolipomatous tumor (HFLT)
* Dedifferentiated liposarcoma
* Undifferentiated pleomorphic sarcoma
* Myxoinflammatory Fibroblastic sarcoma (MIFS)

**DISCUSSION:**

*Histology*: Variably cellular proliferation of spindled cells (nuclear hyperchromasia and pleomorphism), ectatic thin-walled blood vessels with fibrionoid material. Intranuclear pseudoinclusions, hyalinized to myxoid stroma. Rare mitosis with no necrosis.

*IHC:* Positive for CD34 and negative for S100, CK, desmin

*Molecular:* TGFBR3 and MGEA5 gene rearrangements

*Pathophysiology:*

* Possible pathogenetic relationships exists with other entities – Hemosiderotic fibrolipomatous tumor (HFLT) and myxoinflammatory fibroblastic sarcoma. Not completely understood.
* PHAT shows areas like HFLT in the periphery in many cases
* PHAT and HFLT are locally aggressive/local recurrence
* Recurrent TGFBR3 and/or MGEA5 rearrangements seen in both
* Some HFLT shows progression to MIFS and both have shown recurrent TGFBR3 and/or MGEA5 rearrangements (Hybrid HFLT/MIFS)
* Classic MIFS lacks TGFBR3 and /or MGEA5 rearrangements
* Hybrid HFLT/MIFS represents a form of malignant progression within HFLT rather than a strict lesion related to classic MIFS.

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**CASE #6**

**Presenter:** Fernando Alekos Ocampo Gonzalez, MD

**Attending**: Mary K. Allen-Proctor, MD

**CASE HISTORY**: This is a 56-year-old male, a prior smoker, with no other relevant prior history who presents with a 1-year history of unintentional weight loss and 1 week of dysphagia. Laryngoscopy performed at ENT clinic shows a large supraglottic mass obstructing the vocal cords as well as post-cricoid area. Tracheostomy, placement of PEG tube, and biopsy of the mass are performed. A section from the subsequent resection specimen is provided.

**DIAGNOSIS**: Inflammatory myofibroblastic tumor

**DIFFERENTIAL DIAGNOSIS:** Of inflammatory myofibroblastic tumor

* Contact ulcer
* Pyogenic granuloma
* Spindle-cell squamous cell carcinoma
* Low-grade fibromyxoid sarcoma
* Nodular fasciitis
* Solitary fibrous tumor

**DISCUSSION:**

* Typically a diagnosis of exclusion
* IHC pattern includes strong reaction for vimentin, variable staining for desmin and smooth muscle actin, and negativity for CD34, CD117, S100, cytokeratins.
* ALK rearrangements correlate with ALK immunohistochemistry, and are less common in patients >40 years old

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