**The University of Illinois Hospital & Health Sciences System Combined Residency Program**

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Illinois Registry of Anatomic Pathology (IRAP)

Case Summaries

09/25/2017

# Case 1: Mesenchymal chondrosarcoma

**Presenter: Erica Vormittag, MD; Attending: Grace Guzman, MD; and Steven A. Garzon, MD**

**Clinical History:**

A 16-year-old African American male with no significant past medical history presented with an enlarging anterior mandibular mass for six weeks. A maxillofacial computed tomography (CT) scan showed a vascular lesion at the bony mandibular protuberance with extensive midline bony destruction and extension to the buccal surface, 5.3 cm in greatest dimension.  By imaging the lesion was presumed to be a vascular malformation and enucleation of the lesion was performed.

**Diagnosis:**

* Mesenchymal chondrosarcoma

**Differential Diagnosis:**

* Ewing sarcoma
* Small cell osteosarcoma
* Poorly-differentiated synovial sarcoma

**Key Microscopic Features:**

* Biphasic tumor with atypical cartilage interspersed with primitive small round blue cell component
* Prominent vasculature with primitive cells having a hemangiopericytoma-like pattern

**Immunohistochemical stains:**

* Positive: CD-99, SOX9, S100 in cartilaginous component
* Negative: FLI-1 and epithelial markers

**Molecular studies:**

* FISH or RT-PCR for *HEY1-NCOA2* fusion protein product.
* This is a result of an inversion on Chromosome 8.

**Discussion:**

* Rare high-grade cartilaginous sarcoma that can occur at any age
* Most commonly seen in axial skeleton with predilection for jaw bones
* Biphasic tumor with atypical cartilage islands adjacent to primitive component with prominent vasculature and hemangiopericytoma-like pattern
* Associated with characteristic fusion genes involving a chromosome 8 inversion causing *HEY1-NCOA2* fusion protein
* Surgical resection and chemoradiation therapy is essential, but many have recurrence and poor long-term prognosis

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# Case 2: Pulmonary capillary hemangiomatosis

**Presenter: Khin Su Mon, MBBS; Attending: Marin Sekosan, MD**

**Clinical History:**

A 34-year-old African American female with a past medical history of pneumonia presented with a chronic cough, exertional dyspnea, and chest pain. High-resolution computed tomography (HRCT) revealed bilateral peri-lymphatic nodular opacities and bronchiectasis with mediastinal and hilar lymphadenopathy. Wedge biopsies were taken from the right middle and right lower lobes of the lung.

**Diagnosis:**

* Pulmonary capillary hemangiomatosis (PCH)

**Differential Diagnosis:**

* Normal lung parenchyma
* Capillary hemangioma
* Pulmonary veno-occlusive disease (PVOD)

**Key microscopic features:**

* Diffuse proliferation of thin-walled capillary-sized vascular channels both within the peribronchiolar interstitium and alveoli
* No areas of necrosis, cytological atypia, or mitotic activity are seen

**Immunohistochemical stains:**

* Proliferating cells stained positive for CD31 and CD34

**Take Home Points:**

* PCH is a benign condition characterized by diffuse proliferation of small, thin-walled capillaries in the alveoli and interstitium
* Recent literatures suggest *EIF2AK4* mutation as a novel genetic cause of PCH
* Though PCH may have an insidious clinical onset, it is rapidly followed by a detrimental clinical course with median survival of only 3 years following diagnosis
* Lung transplantation is the definitive treatment in medication non-responders
* Vasodilators should be used cautiously in PCH as it can have a devastating outcome

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# Case 3: Sebaceous lymphadenoma of parotid gland

**Presenter: Suzanne Iwaz, MD; Attending: John V. Groth, MD and Steven A. Garzon, MD**

**Clinical History:**

A 71-year-old Hispanic male with no significant past medical history presented with a slowly growing, painless, left neck mass for over a year. A computed tomography (CT) scan revealed a 5.5 x 4.1 x 3.9 cm well-circumscribed parotid mass with mixed solid and cystic components. An ultrasound-guided fine needle aspiration (FNA) was performed, followed by a superficial parotidectomy.

**Diagnosis:**

* Sebaceous lymphadenoma of parotid gland

**Differential Diagnosis:**

* Pleomorphic adenoma
* Warthin tumor
* Mucoepidermoid carcinoma
* Sebaceous neoplasms
* Metastatic carcinoma

**Key microscopic features:**

* Clusters of sebaceous glands which appear as large polygonal cells with abundant uniform vacuolated cytoplasm
* Clusters of benign squamous cells and basaloid cells
* Abundant lymphoid background

**Take Home Points:**

* Sebaceous neoplasms of the parotid are rare
* Primary sebaceous lesions
* Other neoplasms with sebaceous differentiation
* On cytology sebaceous neoplasms are most commonly initially diagnosed as other salivary glandular lesions and in particular mimic mucoepidermoid carcinoma, although in sebaceous neoplasms assessment for the (MECT1-MAML2) gene fusion due to a t(11;19)(q21;p13) translocation (typical of mucoepidermoidc arcinoma), will not be indentified
* While challenging, cytology specimens may be suggestive of sebaceous differentiation, although the characteristic sebocytes may be rare

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# Case 4: Metanephric Stromal Tumor with Sarcomatoid dedifferentiation

**Presenter: Anastasia Sorokina, MD; Attending: Michael R. Pins MD**

**Clinical History:**

A 2-year-old Caucasian female with no significant past medical history presented with gross hematuria and an abdominal mass was found via ultrasound. Abdominal computed tomography (CT) scan further characterized it as a 12.0 x 9.0 x 9.0 cm complex, enhancing solid and cystic mass with a single internal calcification arising from the right kidney. Subsequently, the patient underwent a right nephrectomy.

**Diagnosis:**

* Metanephric stromal tumor with sarcomatoid dedifferentiation

**Differential Diagnosis:**

* Anaplastic Wilms tumor
* Congenital mesoblastic nephroma
* Anaplastic renal sarcoma
* *DICER-1*-associated cystic nephroma

**Key microscopic features:**

* Metanephric stromal tumor component: spindle cell stroma with cuffing around the renal tubules, formation of collarets and angiodysplasia
* Sarcomatoid component: larger spindle to ovoid cells with more abundant eosinophilic cytoplasm and features of anaplasia such as nuclear pleomorphism, hyperchromasia and atypical mitoses, and foci of geographic necrosis

**Immunohistochemical stains:**

* Sarcomatoid component stained strongly for p53, WT-1 and Bcl2 compared to the metanephric stromal component

**Molecular Studies:**

* *BRAFV600E* mutation not detected
* FISH for *SS18* and *ETV6* rearrangements were not detected
* Karyotype analysis showed non-specific findings

**Take Home Points:**

* Metanephric stromal tumor is a benign tumor of childhood with classic morphology and excellent prognosis, frequently associated with *BRAFV600E* mutation
* Sarcomatoid transformation can occur in a setting of childhood cystic nephroma and is linked to pleuropulmonary blastoma as a part of *DICER1* germline mutation
* Anaplastic Wilms tumor should require further work up as a large part of these tumors can be subclassified and have been diagnosed as a large variety of pathologic entities
* Anaplastic transformation infrequently occurs in renal stromal tumors and implies a possibility of metastasis and worse prognosis without treatment
* Sarcomatoid transformation of metanephric stromal tumor should be considered as a possibility in the setting of anaplastic changes in the stromal component, strong p53 expression and aberrant WT-1 expression, and should be followed and treated as a renal sarcoma

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# Case 5: Synchronous endometrioid carcinomas of uterine corpus and ovary

**Presenter: Nasma K. Majeed, MD; Attending: John V. Groth, MD**

**Clinical History:**

A 22-year-old female with no significant past medical history presented to an outside hospital with lower abdominal pain suspicious for pelvic inflammatory disease. Ultrasound imaging, following antibiotic treatment, revealed a left ovarian mass prompting a transfer to our institution. Further imaging by computed tomography (CT) scan showed an 11.0 x 6.9 cm thick-walled, multi-loculated left ovary with dilated fallopian tubes and a thickened endometrium, with magnetic resonance imaging (MRI), suggestive of deep myometrial invasion. The patient underwent a left salpingo-oophorectomy and endometrial curettage followed by total abdominal hysterectomy, a month later.

**Diagnosis:**

* Synchronous endometrioid carcinoma of uterine corpus and ovary

**Differential Diagnosis:**

* Two independent primary endometrioid carcinomas
* Primary endometrial endometrioid cancer with metastases to ovary
* Primary ovarian endometrioid carcinoma with metastases to endometrium

**Key microscopic features:**

* The ovarian mass showed papillae, cribriform glands, cysts and irregular nests of cells surrounded by variable amounts of edematous, myxoid and fibrous stroma
* The uterus was composed of complex epithelial growth pattern with little intervening stroma, with budding and branching of large glands and papillae
* The neoplastic cells in both lesions demonstrated mild to moderate atypia

**Immunohistochemical stains:**

* Positive for CK7, ER, EMA, CA125, CD10 highlights periglandular stroma in the ovary
* Negative for WT1, CK20 and CD10 in glands
* p53 displayed patchy focal weak to moderate expression in the epithelial cells (< 20%).
* Vimentin was strongly and diffusely positive in the glandular cells, and largely negative in the squamoid cells.
* MLH1, PMS2, MSH2 and MSH6 were positive diffusely in both lesional and nonlesional cells

**Molecular Studies:**

* Molecular analysis by next generation sequencing identified identical *CTNNB1*, *PIK3CA*, *PTEN, APC, PRSS8, ASXL1, BRCA2, ESR1, FANCE,* and *MDM2* mutations in both tumors
* Additional *MLL3* and *EPHB1* mutations were identified only in the uterine endometrioid tumor

**Take Home Points:**

* Coexisting endometriod carcinoma lesions in the ovary and uterus require application of morphologic and clinical and to determine site of origination
* Molecular findings may not solve difficult cases
* Low-grade endometrioid carcinomas confined to a single ovary and the endometrium, have a good prognosis and should be considered clinically to behave as FIGO stage I independent tumors

**References:**

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