A case of Hereditary Phakomatosis

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Patient Presentation

- 26 y/o 5’5” 135lbs female.
- Patient is seen for follow-up of a hereditary congenital disorder.
- Patient has a family history of multiple pancreatic and kidney cysts.
- Patient is asymptomatic.
Radiologic study performed

- CT scan of Head, Abdomen, Pelvis without (C-) and with (C+) contrast.
- Indication for study:
  - Follow-up evaluation of multiple lesions in kidneys and pancreas.
- Axial images were obtained.
Liver

Pre contrast (C-)

Multiple low attenuation rounded lesions in pancreas

Pancreas

Kidney?

Post contrast (C+)

Multiple non-enhancing Cystic lesions in pancreas

Portal Vein

Aorta

IVC

Spleen

Multiple hypodense lesions in left kidney
Multiple hypodense lesions in both kidneys

Multiple non-enhancing lesions in both kidneys

HU = Hounsfield Unit
What is a Hounsfield Unit?

- Measure of the relative density of a structure.
- Water has 0 HU.
- Other examples:
  - Air = -1500
  - Fat = -40
  - Soft tissue = 80
  - Calcium = 400
- A difference of >20 HU between Pre and Post Contrast means that there is enhancement.
- If enhancement is seen it would possibly be due to the presence of a neoplasm.
- Measure of >20 means the cyst probably contains proteinaceous or hemorrhagic material
- Difference of 1.7 HU between pre and post contrast images.
- Non-enhancing kidney cyst.
Head CT: axial views

- Distortion of posterior fossa anatomy.
- Poorly defined lesion

Well defined enhancing mass at posterior fossa
Poorly defined mass at cervicomedullary junction

Intensely enhancing mass at cervicomedullary junction
Head MRI was performed to obtain a better definition of this lesion.
Head MRI: T1 weighted, sagittal view

Intensely enhancing mass at cervicomedullary junction

Mass distorting cervicomedullary junction

PACS BIDMC
**Head MRI: FLAIR, axial view**

**FLAIR:**
- Sensitive to edema
  - Fluid is dark
  - Edema is bright

-Bright region at cervicomedullary area.
- Hemangioblastoma causing edema of nervous tissue.

http://www.amershamhealth.com/medcyclopaedia/
Which hereditary syndrome presents with retinal and CNS hemangioblastomas and multiple cysts in pancreas and kidneys?
Von Hippel-Lindau (VHL)
Von Hippel-Lindau

- Named after Eugen von Hippel (ophthalmologist) and Arvid Lindau (pathologist).
- Transmitted as an autosomal dominant disorder with variable penetrance.

Hallmarks of the condition

- Retinal Angiomas.
- Hemangioblastomas of cerebellum and spinal cord.
- Renal cysts and renal cell carcinoma.
- Pancreatic cysts and tumors.
- Pheochromocytomas.

Diagnosis

- The patient has more than one hemangioblastoma in the CNS.
- One hemangioblastoma with visceral manifestations of the disease.
- One manifestation of the disease with a positive family history.

Classification of VHL

- **Type I:**
  - Most common.
  - Retinal and CNS hemangioblastomas, renal cysts and carcinoma, pancreatic cysts, **but no pheochromocytoma.**

- **Type IIA:**
  - Retinal and CNS hemangioblastomas, pheochromocytoma, pancreatic islet cell tumors, **but no renal or pancreatic cysts.**

- **Type IIB:**
  - Retinal and CNS hemangioblastomas, pheochromocytoma, renal and pancreatic disease.

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Clinical Manifestations

- Characterized by development of benign and malignant tumors in multiple organ systems:
  - Brain
  - Retina
  - Pancreas
  - Kidney
  - Adrenal gland
  - Labyrinth
  - Epididymis
Retinal Hemangioblastoma of right eye

Hemangioblastoma in right Cerebellar hemisphere

http://www.amershamhealth.com/medcyclopaedia/
Abdominal CT: axial view

Large contrast-enhanced mass in pancreatic head.

http://www.ajronline.org/cgi/content/full/181/4/1049
Abdominal CT:
axial view

Large left adrenal pheochromocytoma

http://www.amershamhealth.com/medcyclopaedia/
Our patient’s CT demonstrated an enhancing left renal mass.
Pathology revealed…

renal cell carcinoma

http://www-medlib.med.utah.edu/WebPath/RENAHTML/RENAL055.html
Treatment

**Radiofrequency thermal ablation:**

- Patient in prone position to access retroperitoneum.
- A needle was inserted between the ribs to thermally ablate with radiofrequency (RF ablation) the left kidney mass.
Differential Diagnosis of multiple renal cysts

- Autosomal dominant polycystic kidney disease.
- Autosomal recessive polycystic kidney disease.
- Von Hippel-Lindau.
- Tuberous sclerosis.
- Acquired cystic disease (eg, from dialysis).
- Hepatic fibrosis-renal cystic disease.

Radiological Screening in VHL

- CT and MRI are preferred for intracranial and spinal cord lesions.\(^5\)
- Retinal tumors visualized best on sonograms.\(^5\)
- CT yearly or every 2 years (beginning at 20 years old) is recommended for abdominal lesions.\(^6\)

Summary

- VHL is an autosomal dominant disease associated to a defect in chromosome 3.
- Hallmarks of the disease are cerebellar and retinal hemangiomas associated with multiple renal cysts.
- Patients are screened yearly for potential malignant evolution of these lesions.
- Tumor recurrence and frequent surgical intervention are the most common cause of morbidity and mortality.
- Renal cell carcinoma is the second most common cause of death.
References:

- [http://moon.ouhsc.edu/jspitz/4802images/mlpel.htm](http://moon.ouhsc.edu/jspitz/4802images/mlpel.htm)
- [http://www.cristalp.es/docencia/c0305.jpg](http://www.cristalp.es/docencia/c0305.jpg)
- [http://www-medlib.med.utah.edu/WebPath/RENAHTML/RENAL055.html](http://www-medlib.med.utah.edu/WebPath/RENAHTML/RENAL055.html)
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The Cambridge protocol for screening patients with VHL

- Annual physical exam and urine test,
- Annual ophthalmologic exam.
- Annual angiography.
- Annual ultrasonographic examination.
- MRI or CT scan of brain every 3 years.
- Abdominal CT scan every 3 years.
- Annual 24-hour urine collection of vanillylmandelic (VMA) levels.
Endolymphatic sac tumor compressing the cerebellopontine angle and extending into the mastoid

http://www.amershamhealth.com/medcyclopaedia/
Testicular Ultrasound

Epididymal cysts

http://moon.ouhsc.edu/jspitz/4802images/mlpel.htm