

AMSER Case of the Month

October 2026

An Ocular Window into Multisystem Disease

A 13-year-old female presents with headache and
painful vision loss

Gregory Owendoff – Cleveland Clinic Lerner College of Medicine

Brendan McCleary, MD Pediatric Radiologist , Cleveland Clinic

Ruchi Yadav, MD, Thoracic Radiologist, Cleveland Clinic

Patient Presentation

- 13-year-old female
- History of retinal capillary hemangioma
 - Treated over the last year with anti-VEGF and photodynamic therapy
- Presents to ophthalmology with:
 - Persistent headache
 - Chronic left eye pain
 - Variable vision loss

Pertinent Clinical Findings

- **Vitals:** BP:121/76, HR: 113, T: 36.9C, SpO2: 98%, RR: 18
- **Fundoscopy Exam**
 - Raised 3 x 3.5 mm salmon-colored retinal lesion
 - Overlying subretinal fluid + surrounding exudate
 - Lesion adjacent to optic nerve and fovea
- **Clinical concern:** Retinal hemangioblastoma
- **Next step:** *Evaluation* for hereditary tumor predisposition syndrome

Continued Workup

GENE	VARIANT	ZYGOSITY	VARIANT CLASSIFICATION
VHL	c.470C>T (p.Thr157Ile)	heterozygous	PATHOGENIC

- **Diagnosis:** Autosomal dominant von Hippel-Lindau syndrome
- Referred to pediatric hematology/oncology
- Requires evaluation for additional VHL-related neoplasm

What imaging study should be ordered in newly diagnosed VHL syndrome for CNS tumor screening?

Variant: 1 Adult. Primary brain tumor screening. Genetic risk factors.

Procedure	Appropriateness Category	Relative Radiation Level
MRI head without and with IV contrast	Usually Appropriate	○
MRI complete spine without and with IV contrast	May Be Appropriate	○
MRI head without IV contrast	May Be Appropriate	○
MR spectroscopy head without IV contrast	Usually Not Appropriate	○
MRI complete spine with IV contrast	Usually Not Appropriate	○
MRI complete spine without IV contrast	Usually Not Appropriate	○
MRI functional (fMRI) head without IV contrast	Usually Not Appropriate	○
MRI head perfusion with IV contrast	Usually Not Appropriate	○
MRI head perfusion without IV contrast	Usually Not Appropriate	○
MRI head with IV contrast	Usually Not Appropriate	○
MRI head without IV contrast with DTI	Usually Not Appropriate	○
CT head with IV contrast	Usually Not Appropriate	☢☢☢
CT head without and with IV contrast	Usually Not Appropriate	☢☢☢
CT head without IV contrast	Usually Not Appropriate	☢☢☢
DOTATATE PET/CT brain	Usually Not Appropriate	☢☢☢
DOTATATE PET/MRI brain	Usually Not Appropriate	☢☢☢
FDG-PET/CT brain	Usually Not Appropriate	☢☢☢
FDG-PET/MRI brain	Usually Not Appropriate	☢☢☢
Fluciclovine PET/MRI brain	Usually Not Appropriate	☢☢☢
Fluciclovine PET/CT brain	Usually Not Appropriate	☢☢☢☢

Variant: 1 Adult. Primary brain tumor screening. Genetic risk factors.

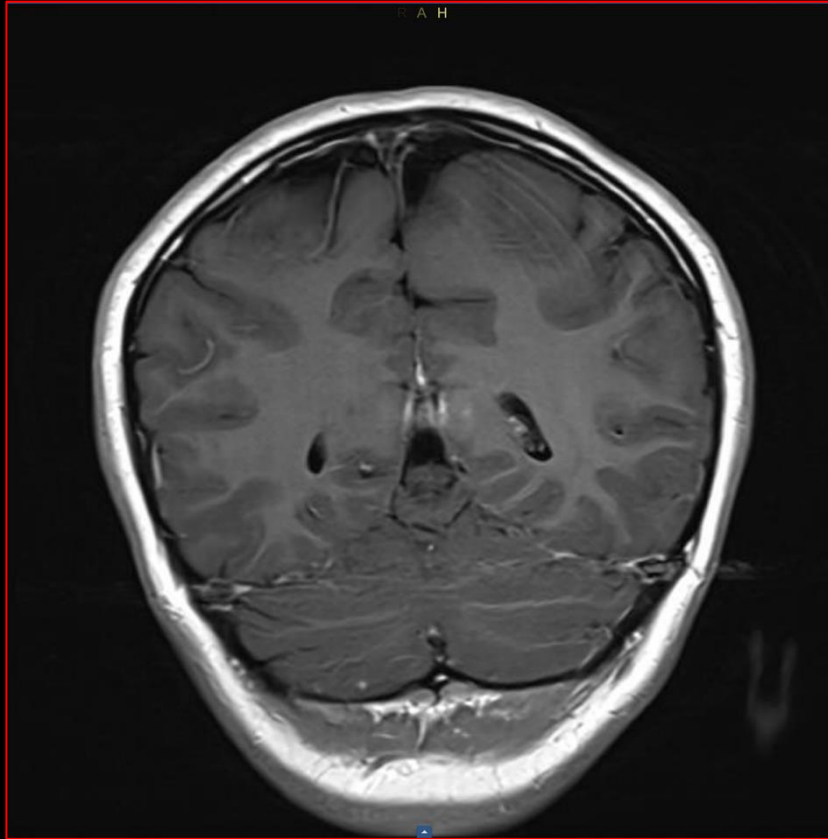
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Ordered by
hematology/oncology

Variant: 1

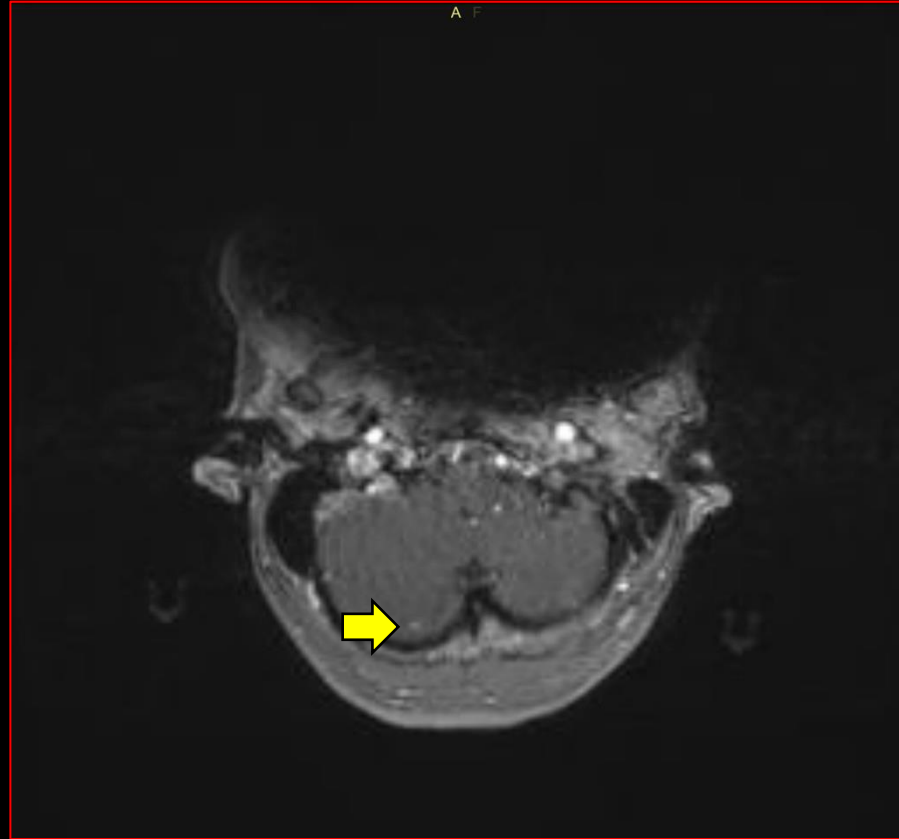
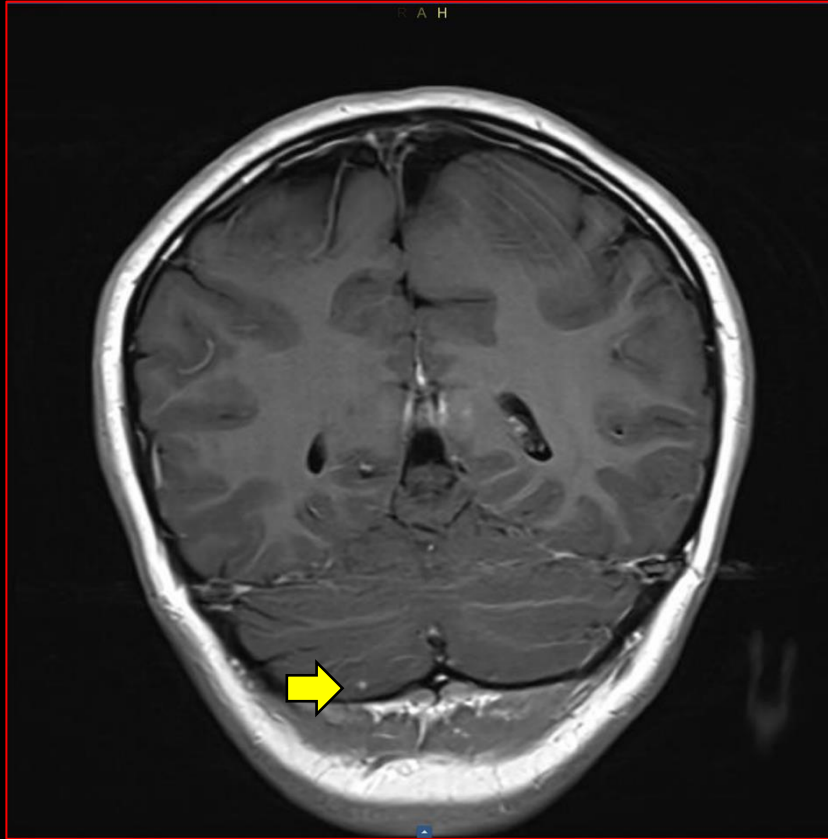
“...For VHL, biannual MRI of the brain and spine is recommended starting at age 10 years to screen for hemangioblastoma...”

MRI Findings (unlabeled)



T1 w contrast

MRI Findings (labeled)



Enhancing right cerebellar lesion

- Compatible with hemangioblastoma
- Characteristic CNS manifestation in the setting of VHL

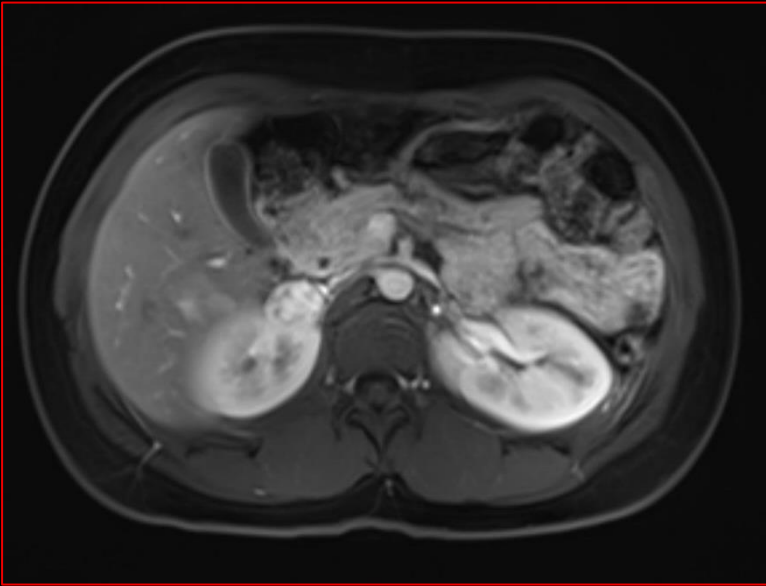
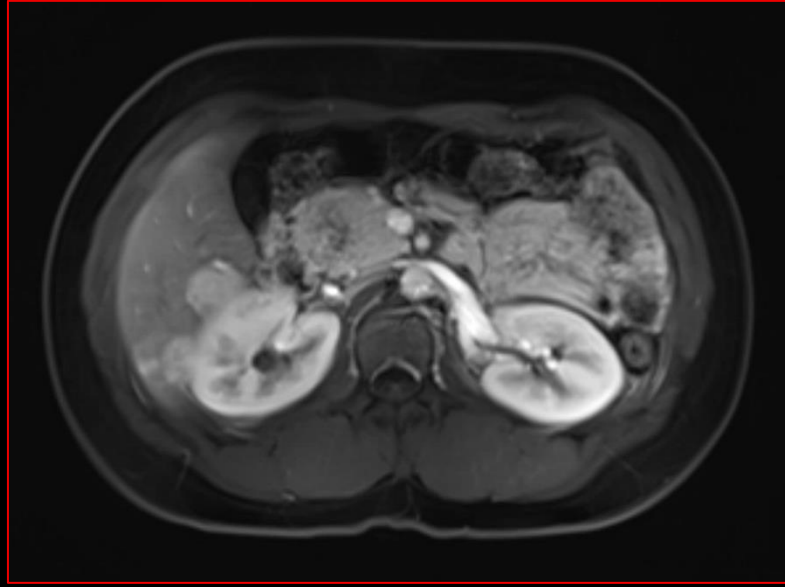
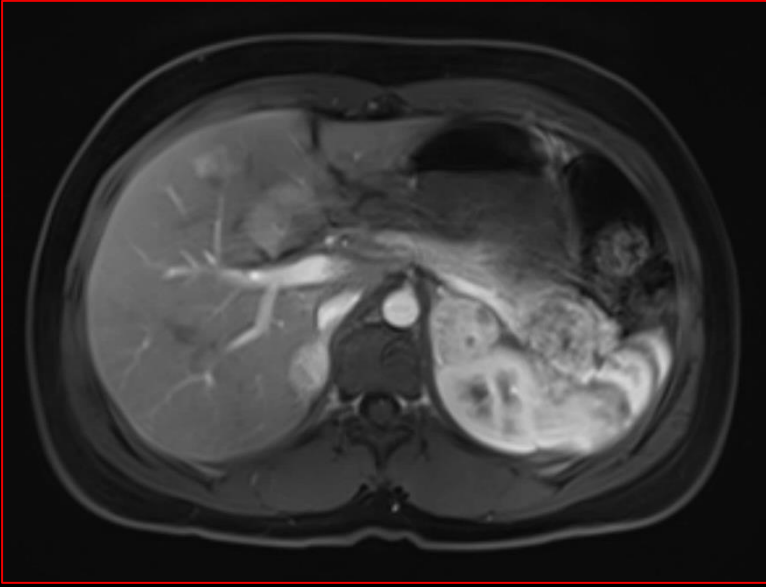
Continued Workup

- Plasma metanephrines: **markedly elevated**
 - Normetanephrine: **4940 pg/mL** (ref range: 22 - 83)
- Combined with CNS MRI findings, prompted further systemic imaging

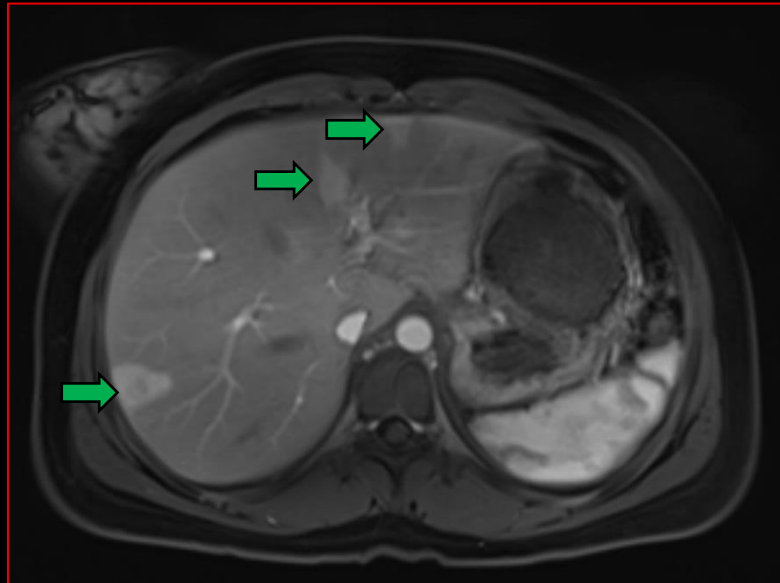
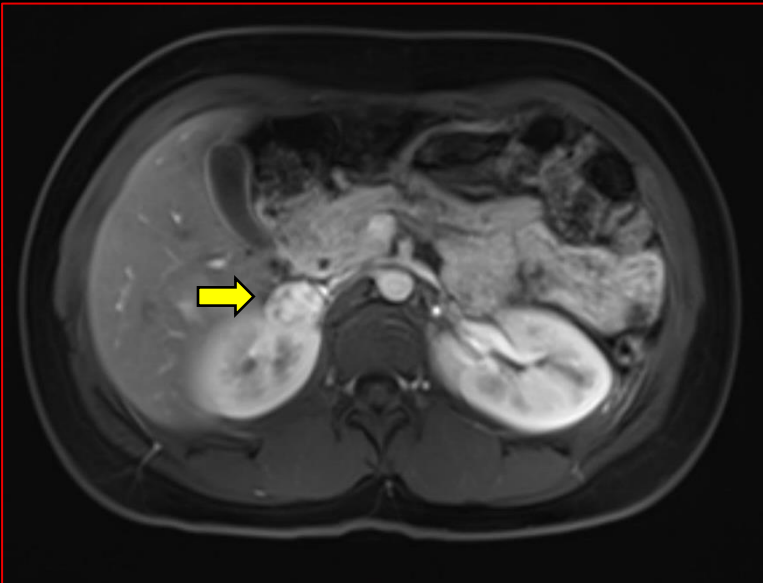
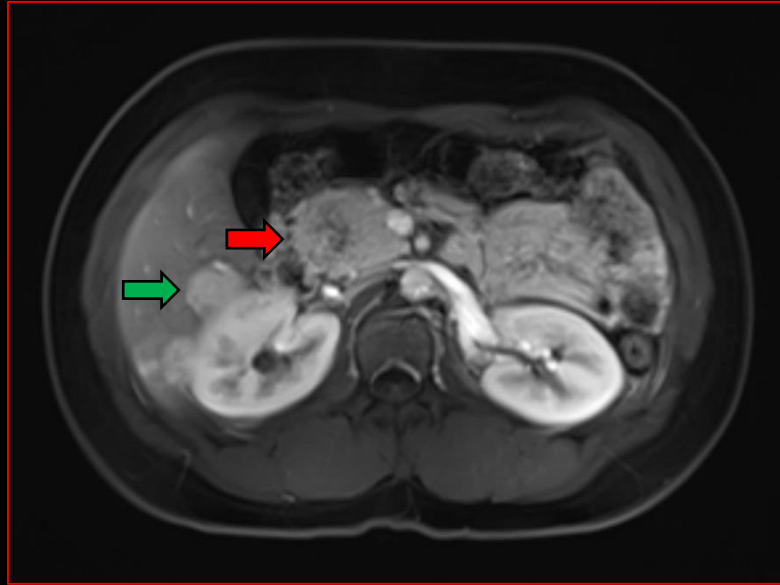
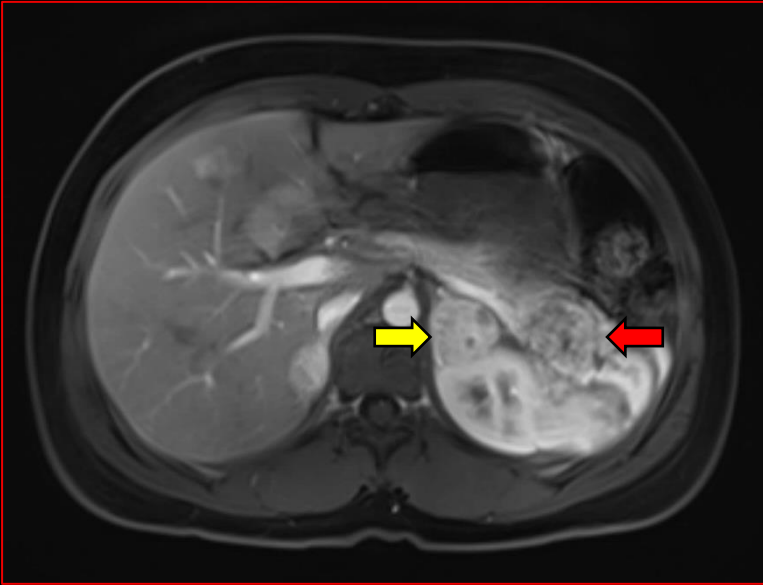
Variant 3. Adult. Known or suspected pheochromocytoma. Initial staging.

Procedure	Appropriateness Category	Relative Radiation Level
MRI abdomen and pelvis without and with IV contrast	Usually Appropriate	0

MRI (T1 post-contrast) unlabeled



MRI (T1 post-contrast) labeled



- Bilateral adrenal masses (yellow arrows): suspect pheochromocytomas
- Hyper-enhancing pancreatic head and tail lesions (red arrows): suspect neuroendocrine tumors
- Multiple enhancing hepatic lesions (green arrows): concerning for neuroendocrine metastasis

What is the best next imaging to evaluate suspected pancreatic neuroendocrine metastasis?

Variant: 2 Adult. Staging of pancreatic neuroendocrine tumor. Evaluation for metastatic disease.

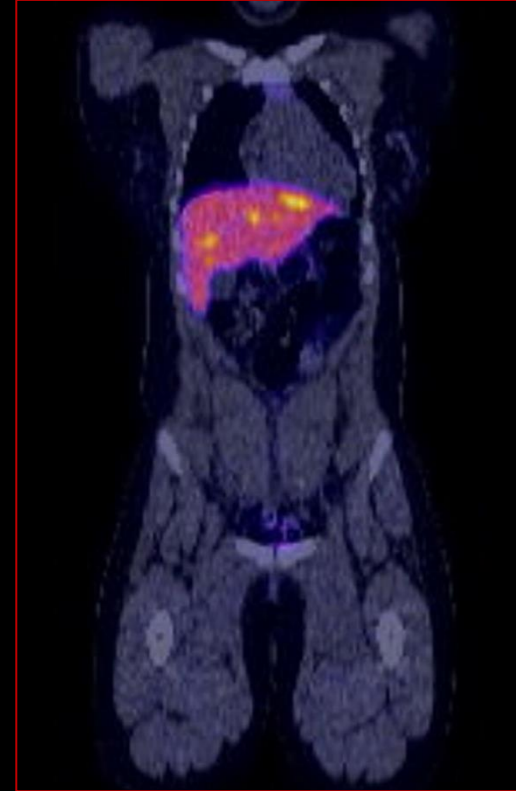
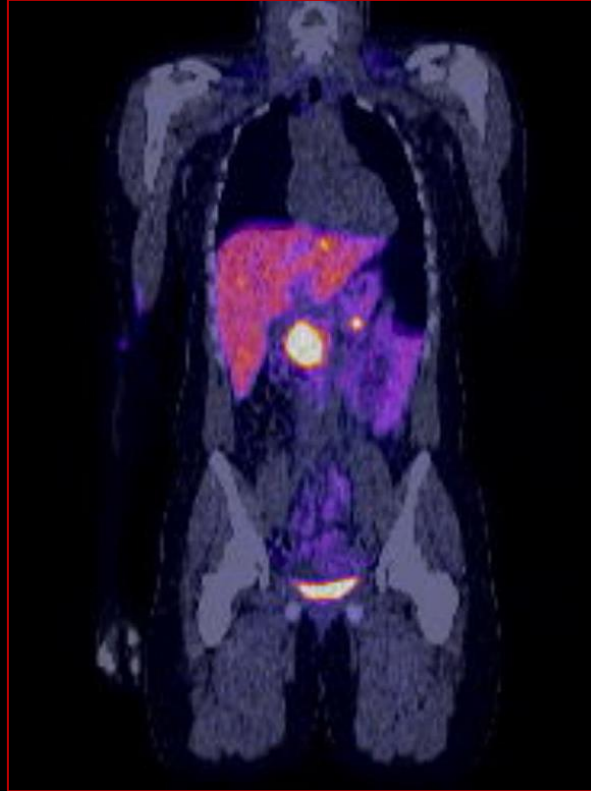
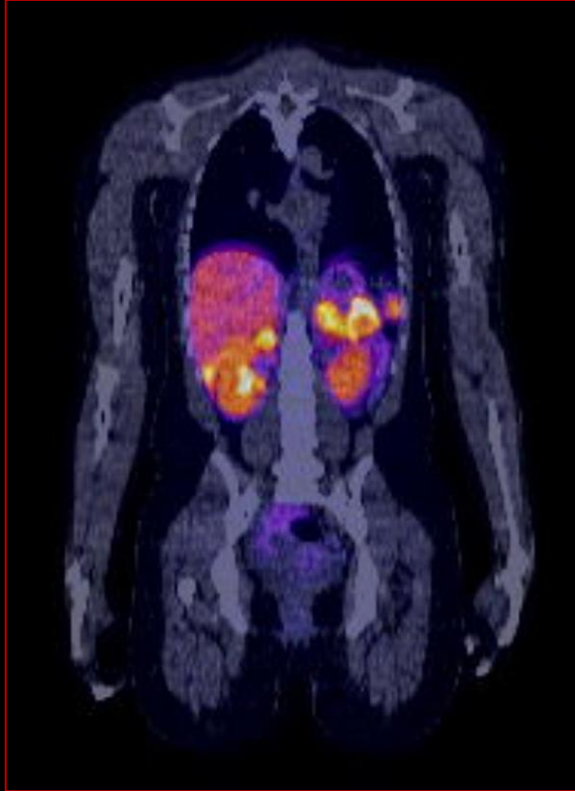
Procedure	Appropriateness Category	Relative Radiation Level
MRI abdomen and pelvis without and with IV contrast	Usually Appropriate	○
CT abdomen and pelvis with IV contrast	Usually Appropriate	☢☢☢
DOTATATE PET/CT skull base to mid-thigh	Usually Appropriate	☢☢☢
CT chest abdomen pelvis with IV contrast	Usually Appropriate	☢☢☢☢
MRI abdomen and pelvis without IV contrast	May Be Appropriate	○
CT abdomen and pelvis without and with IV contrast	May Be Appropriate	☢☢☢☢
CT chest abdomen pelvis without and with IV contrast	May Be Appropriate	☢☢☢☢
FDG-PET/CT skull base to mid-thigh	May Be Appropriate	☢☢☢☢
US abdomen endoscopic	Usually Not Appropriate	○
CT abdomen and pelvis without IV contrast	Usually Not Appropriate	☢☢☢
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Variant: 2 Adult. Staging of pancreatic neuroendocrine tumor. Evaluation for metastatic disease.

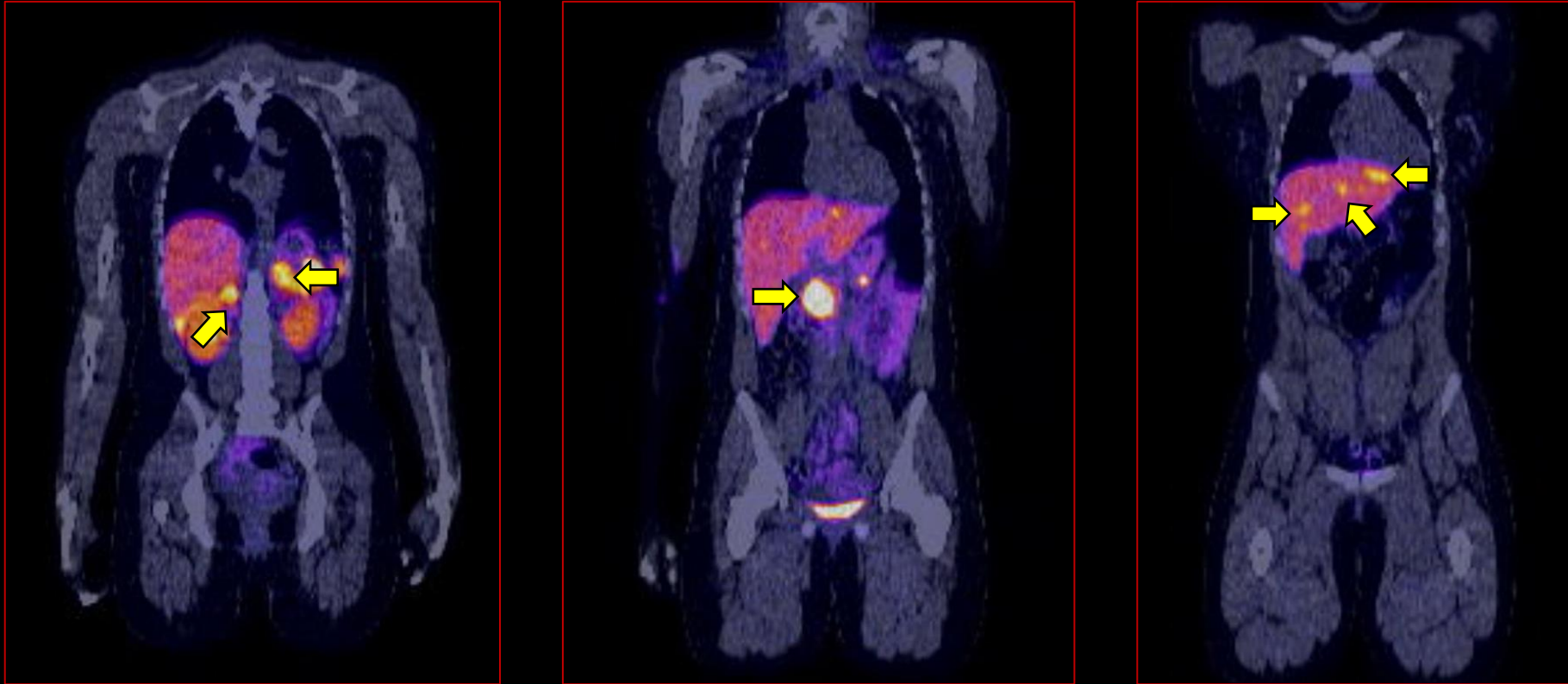
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^{68}Ga -DOTATATE: targets cell surface type 2 somatostatin receptors

PET CT (unlabeled)



PET CT (labeled)



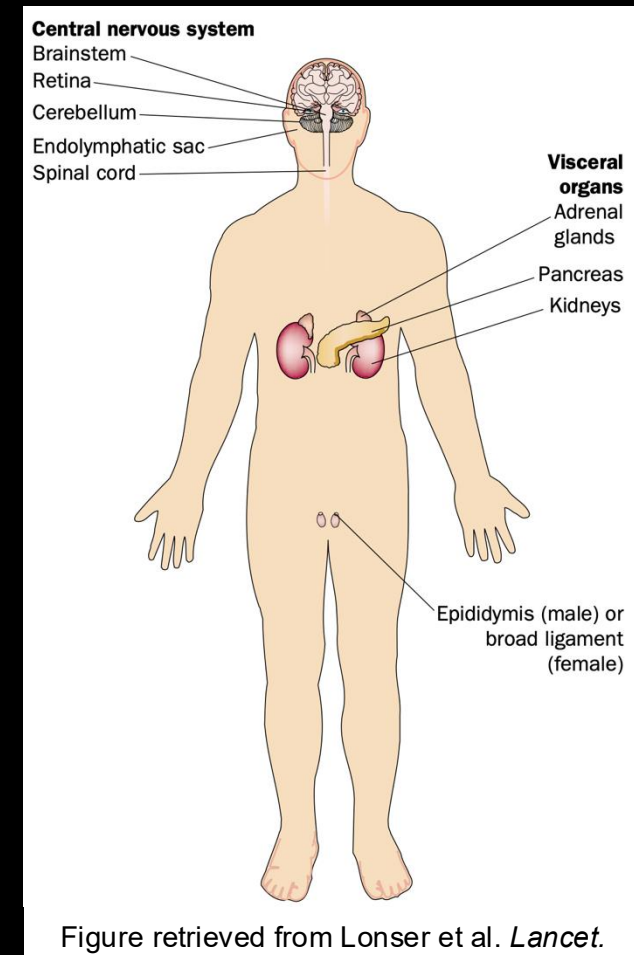
- Highly avid pancreatic and bilateral adrenal lesions
- Approximately 10 bilobar SSTR2 expressing hepatic lesions: **metastasis**

Discussion

- **Final diagnosis:** VHL syndrome with multisystem neoplasia
- Germline mutation of VHL tumor suppressor gene (chromosome 3)⁴
 - Highly penetrant autosomal dominance
- 1 in 36,000 live births

Role of imaging

- VHL surveillance → CNS hemangioblastoma
- Lesion characterization → adrenal + pancreas + liver
- SSTR-targeted staging⁵
 - ⁶⁸Ga-DOTATATE binds SSTR2 expressed on differentiated NETs
 - Offers receptor-targeted staging and guides SSTR-directed therapy

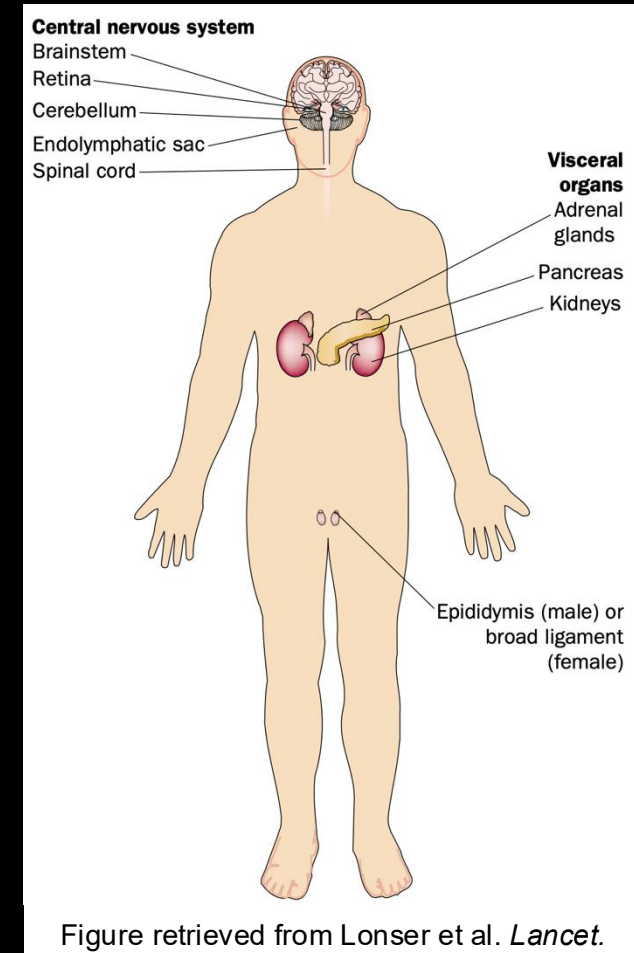


VHL: Affected Organs

Discussion

This case had several hallmark findings of VHL:

- Retinal hemangioblastoma
- Cerebellar hemangioblastoma
- Bilateral adrenal tumors, likely pheochromocytoma⁶
 - Classically T2 hyperintense “light-bulb” appearance
 - T1 iso- or hypo-intense with avid contrast enhancement
 - Larger lesions → heterogenous due to necrosis, cystic change, or hemorrhage
- Multifocal pancreatic neuroendocrine tumors
- SSTR-positive hepatic metastasis of neuroendocrine tumors



VHL: Affected Organs

References:

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3. Yacoub JH, Yano M, Chang KJ, et al. ACR Appropriateness Criteria® Staging and Follow-Up of Pancreatic Neuroendocrine Tumors. *Journal of the American College of Radiology*. 2026;23(7):1445-1461. doi:10.1016/j.jacr.2026.02.012
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