AMSER Case of the Month September 2023

A pediatric patient with epileptic seizures since infancy

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Patient Presentation

- HPI: Pediatric patient with a history of epilepsy with medically difficult to control seizures since infancy and a significant developmental delay with behavioral issues. Breakthrough seizure
- PMHx: Epilepsy, myotonic muscular dystrophy and autism
- FMHx: Father with Tuberous Sclerosis

What Imaging Should We Order?

Select the applicable ACR Appropriateness Criteria[1]

Variant 1: **New-onset seizure. Unrelated to trauma. Initial imaging.**

Procedure	Appropriateness Category	Relative Radiation Level
CT head without IV contrast	Usually Appropriate	☢☢☢
MRI head without IV contrast	Usually Appropriate	O
MRI head without and with IV contrast	May Be Appropriate	O
CT head with IV contrast	Usually Not Appropriate	☢☢☢
CT head without and with IV contrast	Usually Not Appropriate	☢☢☢
FDG-PET/CT brain	Usually Not Appropriate	☢☢☢
MEG	Usually Not Appropriate	O
MRI functional (fMRI) head without IV contrast	Usually Not Appropriate	O
HMPAO SPECT or SPECT/CT brain ictal and interictal	Usually Not Appropriate	☢☢☢

This imaging modality was ordered first

Followed by MRI head with contrast and CT abdomen pelvis with contrast to look for additional abnormalities

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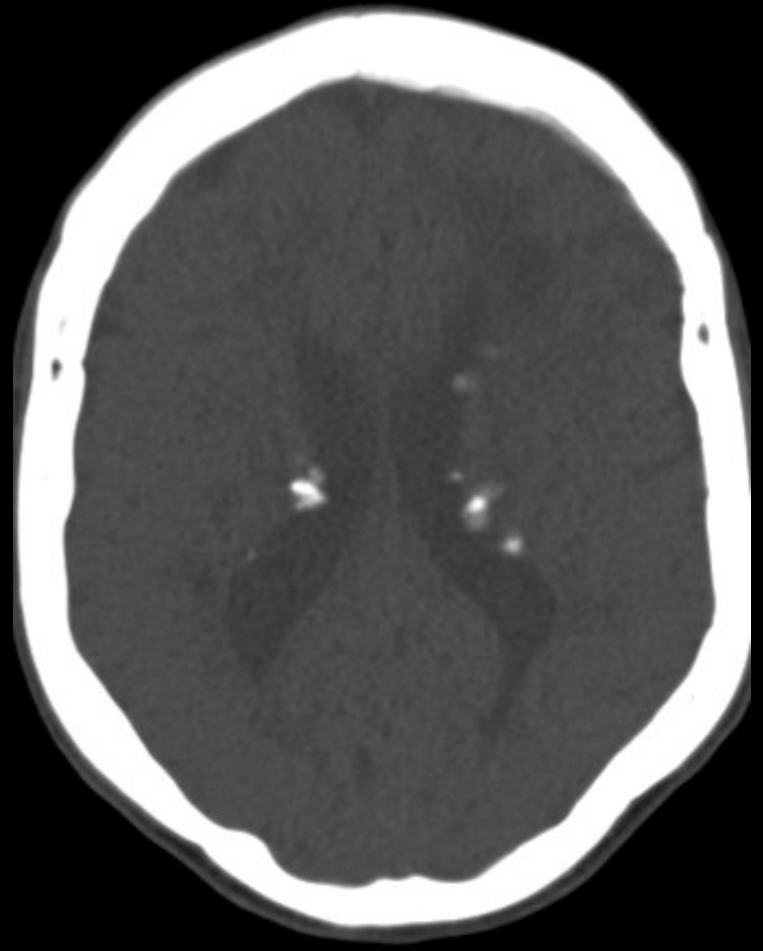
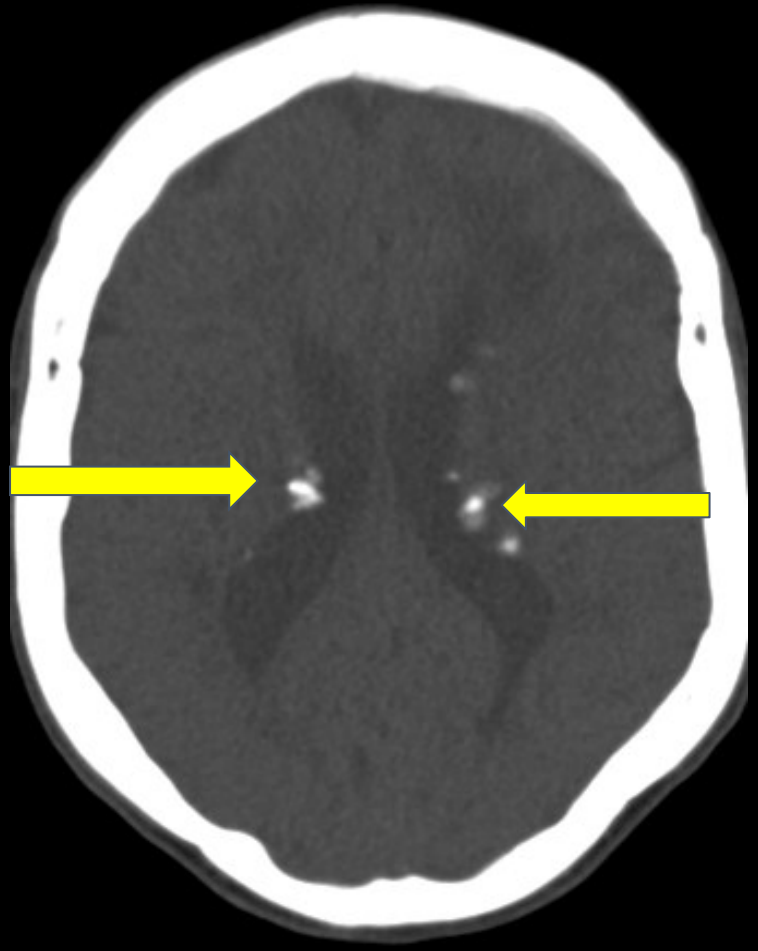
Head non-contrast CT Findings (unlabeled)

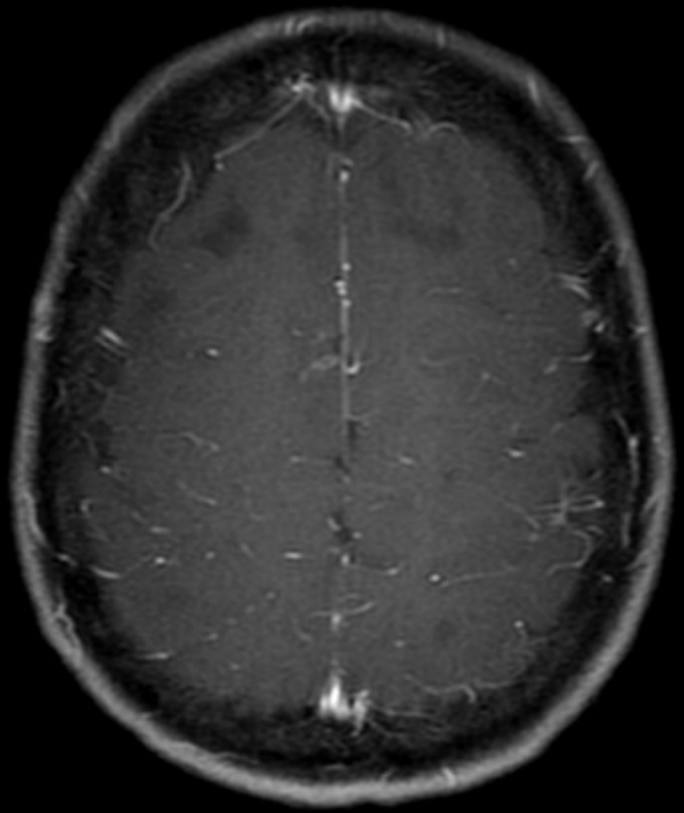


Head non-contrast CT Findings (labeled)

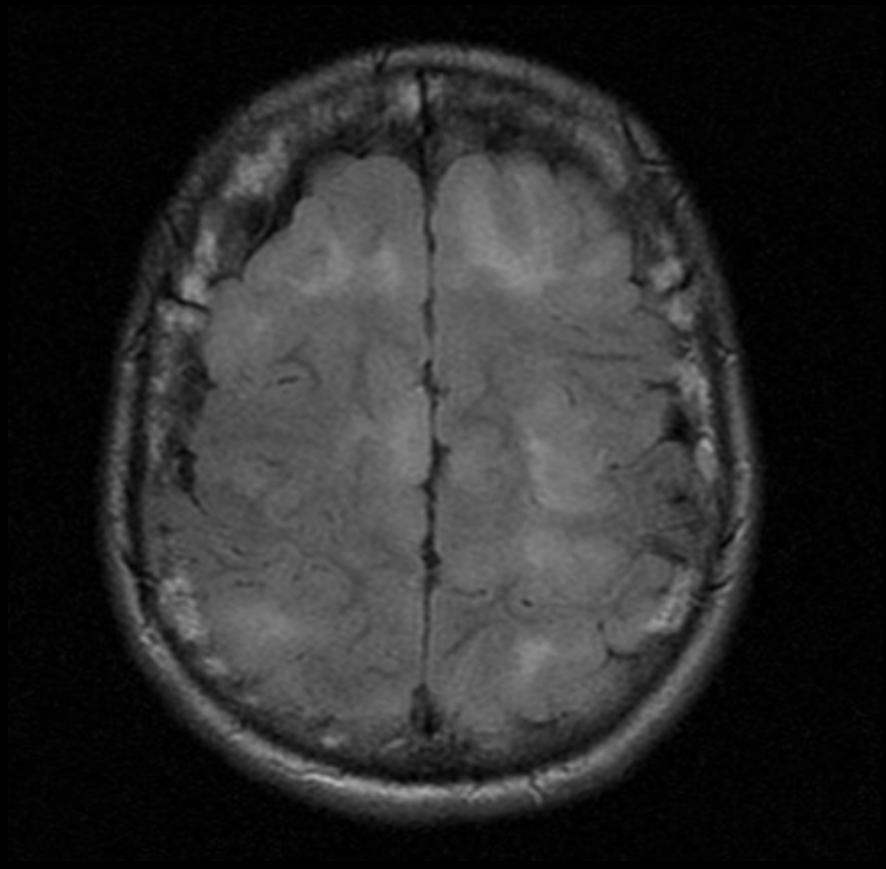


Yellow Arrows:
Calcified subependymal nodules

Head MRI Findings: (unlabeled)

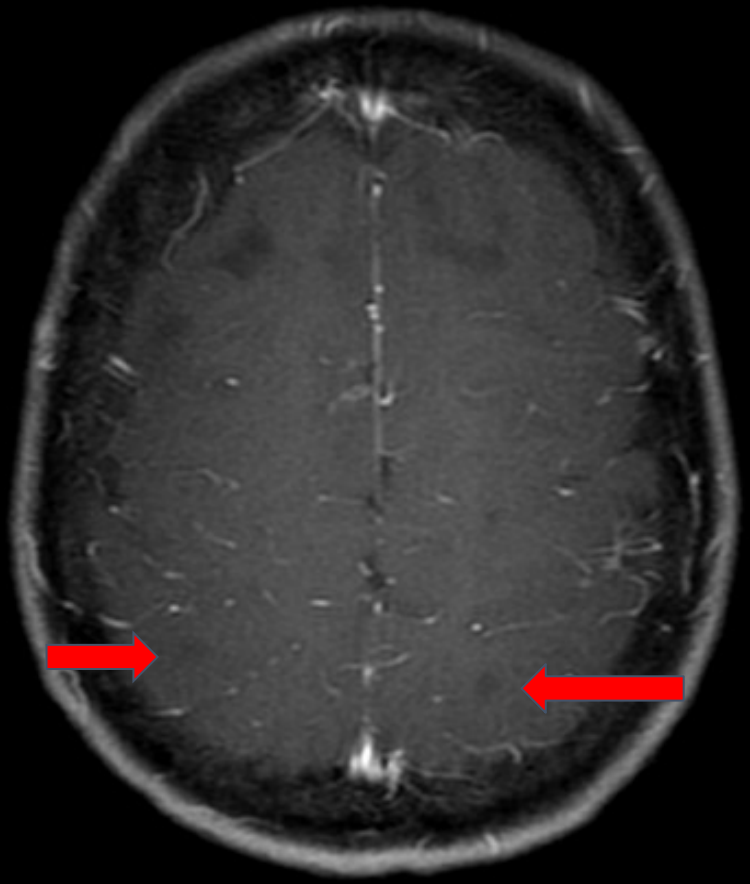


Ax T1 C+

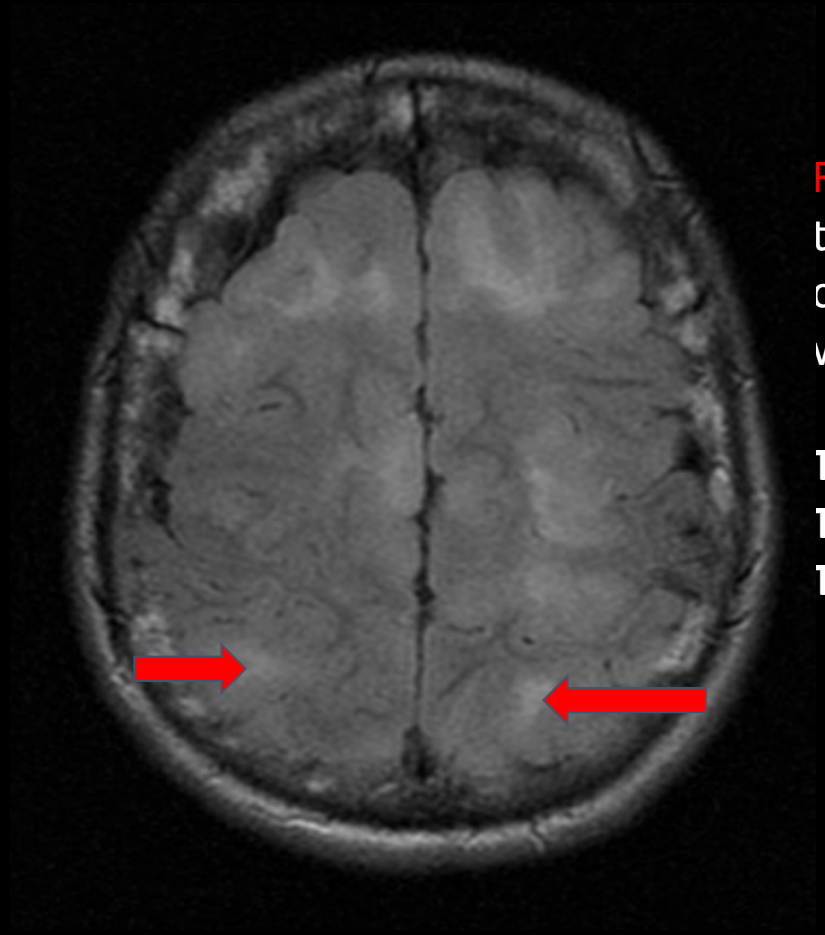


Ax T2 FLAIR

Head MRI Findings: (labeled)



Ax T1 C+



Ax T2 FLAIR

Red Arrows: Cortical tubers are triangular-shaped lesions at cortex/juxtacortical location, with apex toward the ventricles

T1: low signal
T2/FLAIR: high signal
T1 C+ (Gd): lack of enhancement

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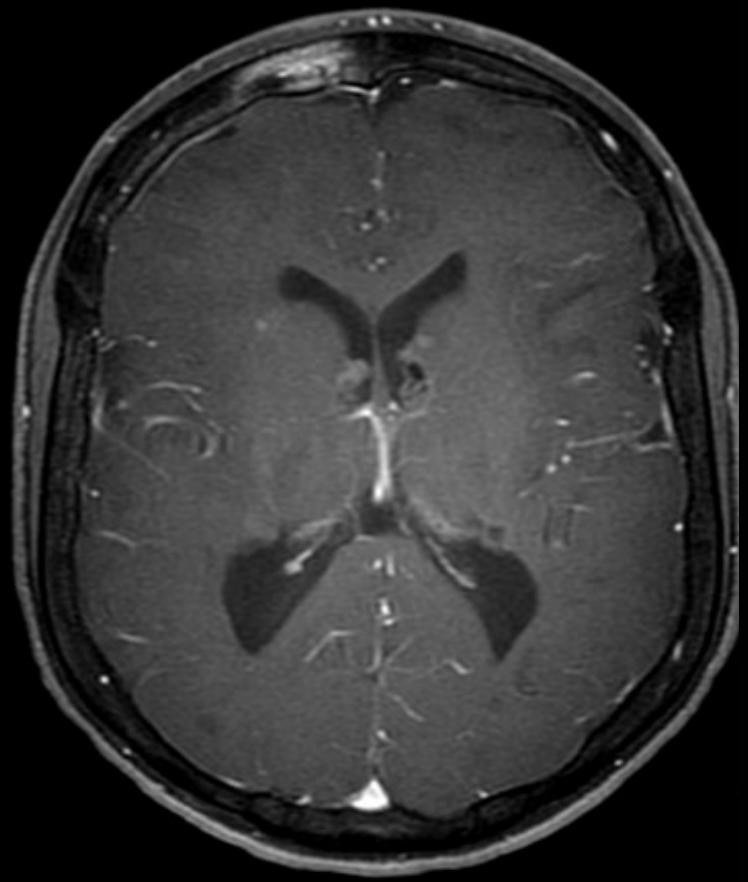
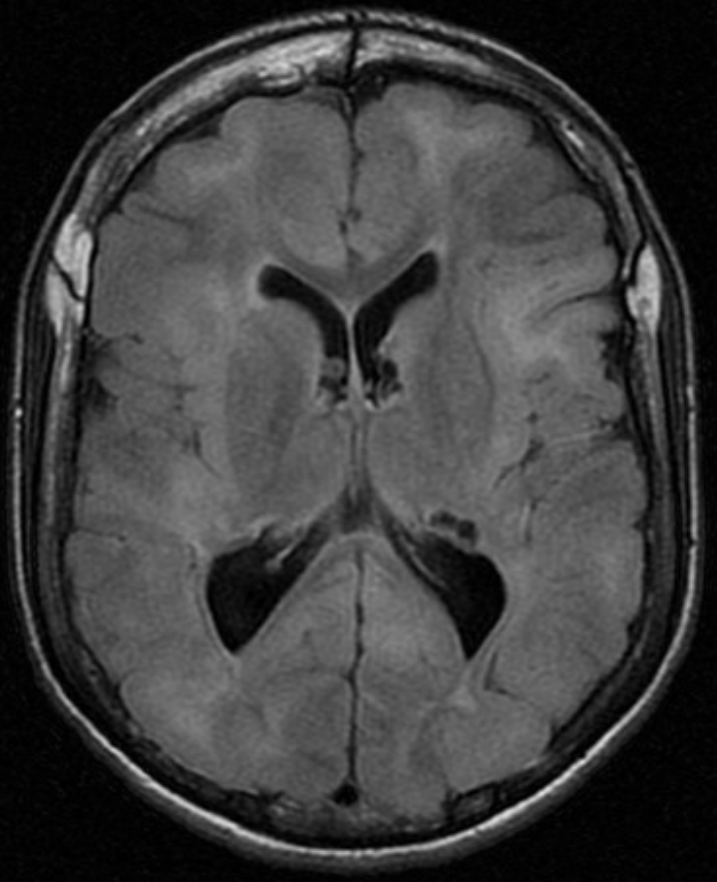
Head MRI Findings: (unlabeled)



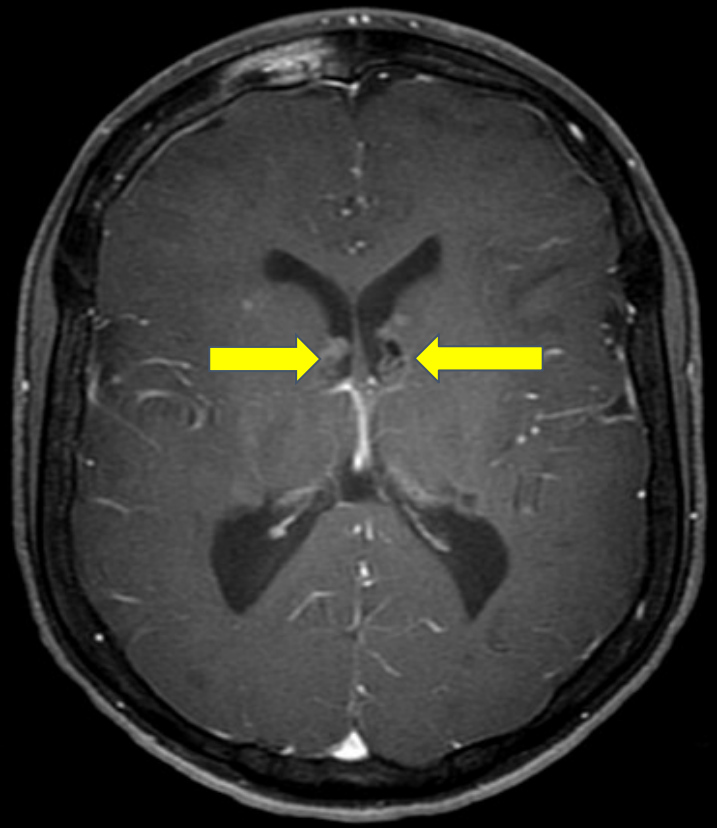
Ax T1 C+



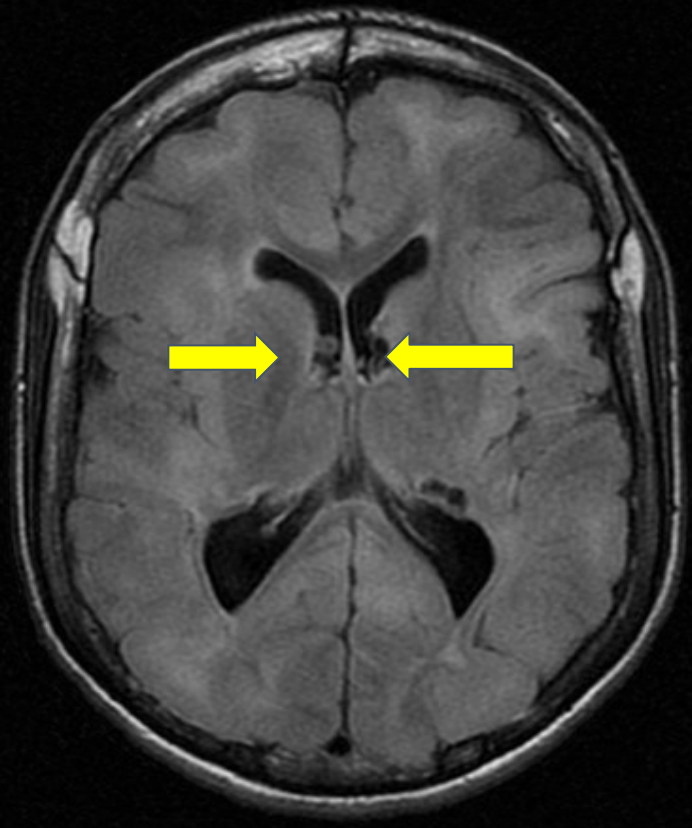
Ax T2 FLAIR

Head MRI Findings: (labeled)

Yellow arrows:
Subependymal hamartomas at the Foramen of Monro



Ax T1 C+



Ax T2 FLAIR

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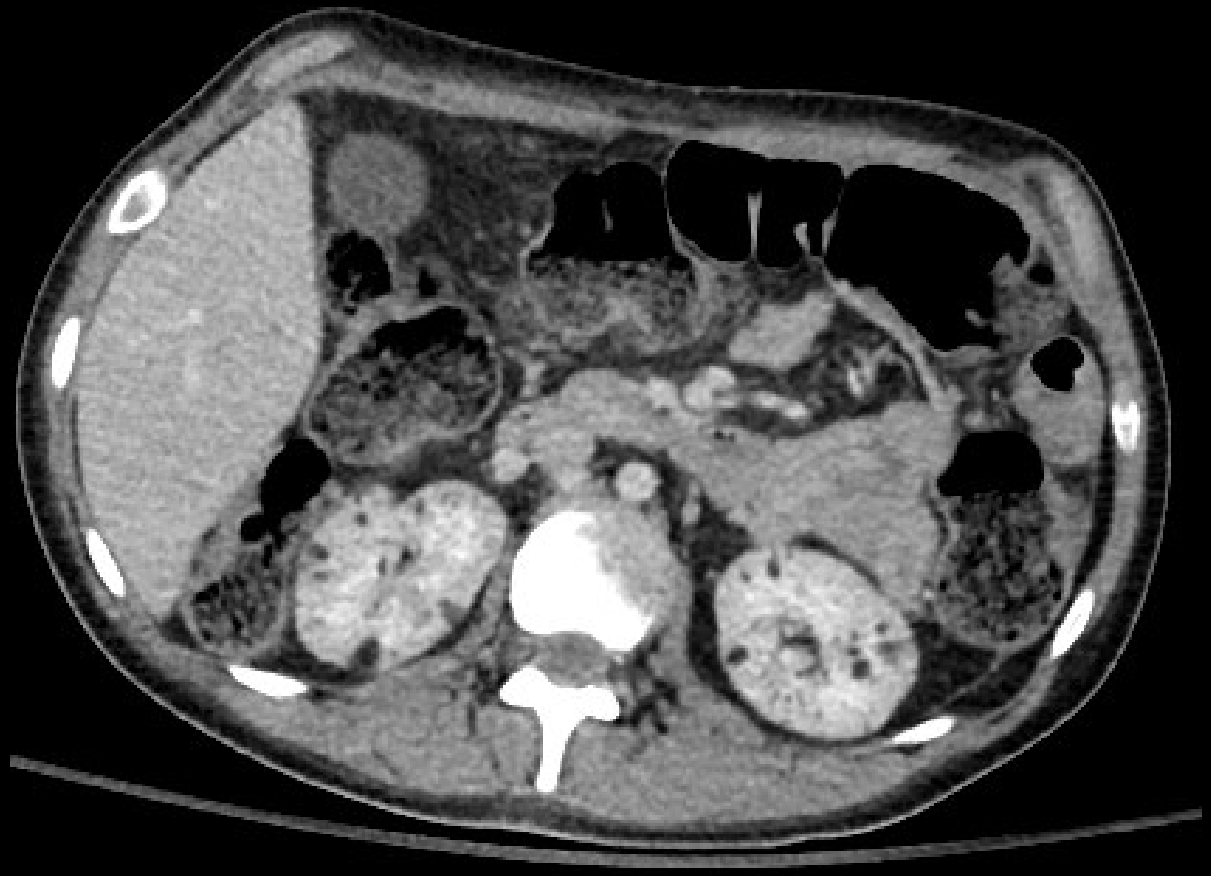
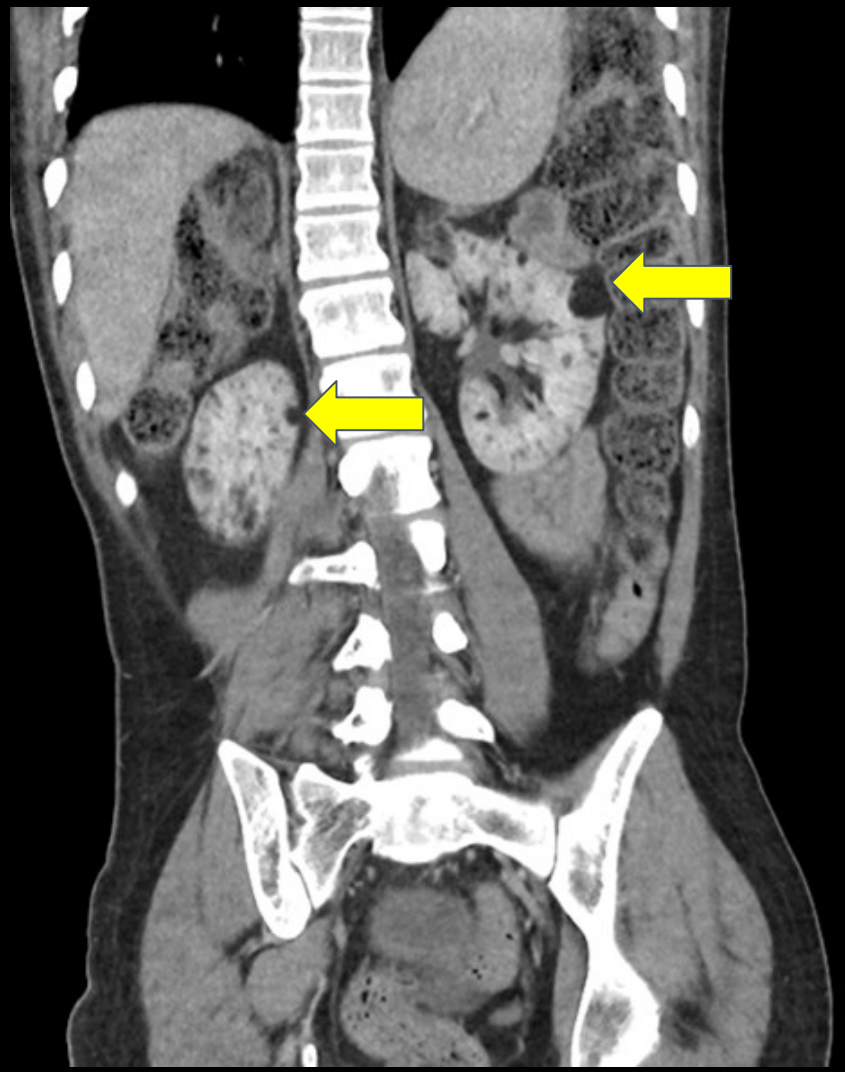
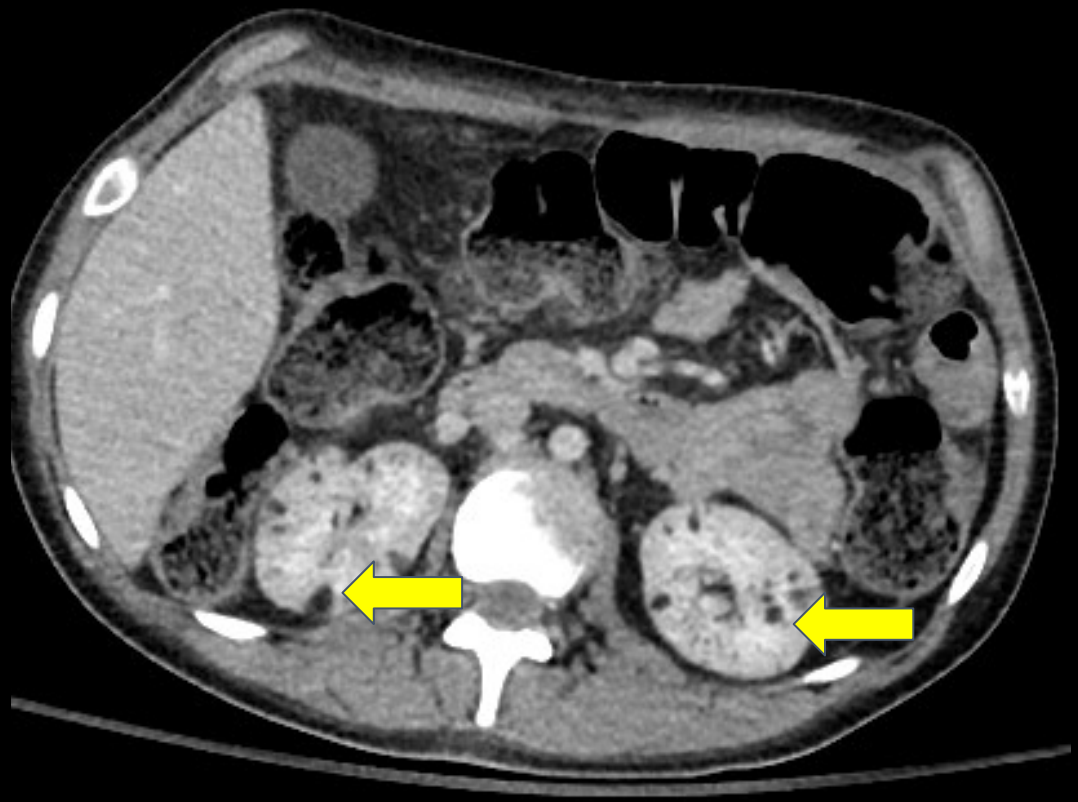
Abdomen and Pelvis CT Findings (unlabeled)



Abdomen and Pelvis CT Findings: (labeled)





Yellow arrows:
Multiple fat-attenuation renal masses consistent with bilateral angiomyolipomas

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Final Diagnosis:

Tuberous Sclerosis

Case Discussion (1-3 slides)

- Presentation
  - Intellectual disability, infantile spasms, seizures, brain tumors, cardiac rhabdomyoma, and renal disease. [2,4]
  - Skin/cutaneous [2,4]
    - Adenoma sebaceum (facial angiofibromas)
    - ash leaf spots
    - shagreen patches
    - Ungual fibromas
- Diagnostic Studies
  - EEG[2,4]
    - Seizure activity
  - Genetic testing [2,4]
    - Autosomal dominant mutation in tumor suppressor genes TSC1 or TSC2 gene

Adenoma sebaceum (left)[2], shagreen patches (middle)[2] ungual fibroma (right)[2]

Case Discussion

- Diagnostic Imaging findings:
  - Neurologic [3]
    - **Cortical/subcortical tubers** (50% in frontal lobe)
    - **Subependymal hamartomas** (88% with calcification)
    - **Giant cell astrocytoma**
    - **White matter abnormalities** such as radial bands
  - Abdominal [3]
    - **Renal cysts** (18-53% of patients with tuberous sclerosis)
    - **Angiomyolipoma** (55-75% of patients with tuberous sclerosis)
    - **Renal cell carcinoma** (similar prevalence as general population but occur at younger age with tuberous sclerosis).
    - **Pancreatic neuroendocrine tumors**
    - **Hepatic angiomyolipoma**

Case Discussion

- Diagnostic imaging findings continued:
  - Thoracic [3]
    - **Cardiac rhabdomyoma** (seen in 50-65% of patients with tuberous sclerosis, can cause ventricular hypertrophy and arrhythmias)
  - Musculoskeletal [3]
    - **Sclerotic bone lesions** (seen in 40-66% of patients with tuberous sclerosis)
    - **Scoliosis**
    - **Bone cysts**
- Management
  - Anti-seizure medications for epilepsy. [5]
  - mTOR inhibitors for renal angiomyolipoma and inoperable astrocytoma. [5]
  - Surgery for obstructive hydrocephalus and drug resistant seizures [5]
  - Renal disease and epilepsy are most common cause of death [5]

References:

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3. Yap J. Tuberous sclerosis: Radiology reference article. Radiopaedia Blog RSS. January 6, 2023. Accessed June 11, 2023. https://radiopaedia.org/articles/tuberous-sclerosis?lang=us.
4. 1. Randle S. Tuberous sclerosis complex: Genetics, clinical features, and diagnosis. UpToDate. Accessed June 13, 2023. https://www.uptodate.com/contents/tuberous-sclerosis-complex-genetics-clinical-features-and-diagnosis?search=tuberous+sclerosis&source=search_result&selectedTitle=1~94&usage_type=default&display_rank=1.
5. 1. Randle S. Tuberous sclerosis complex: Management and prognosis. UpToDate. Accessed June 13, 2023. https://www.uptodate.com/contents/tuberous-sclerosis-complex-management-and-prognosis?search=tuberous+sclerosis&source=search_result&selectedTitle=2~94&usage_type=default&display_rank=2.

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