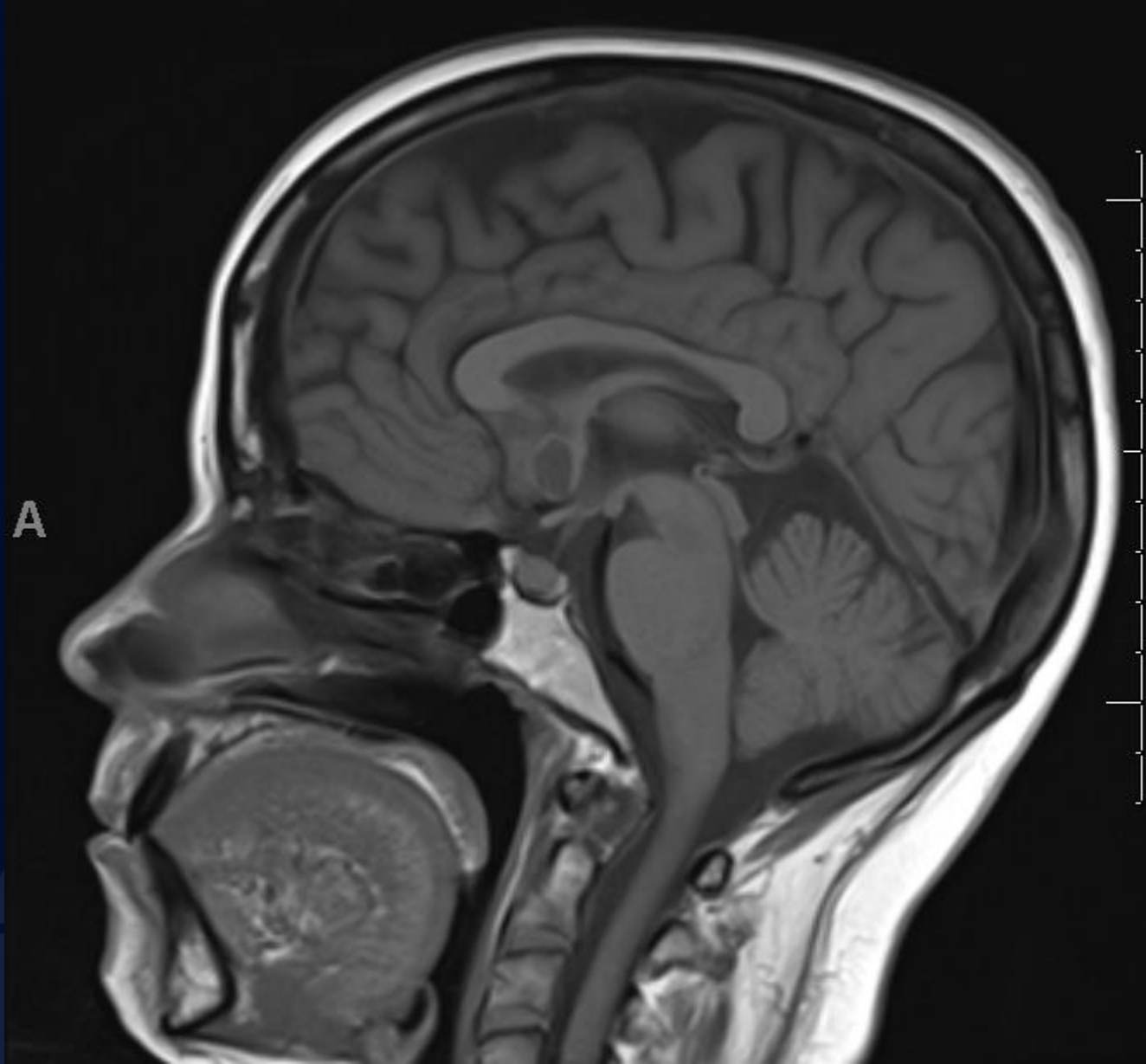
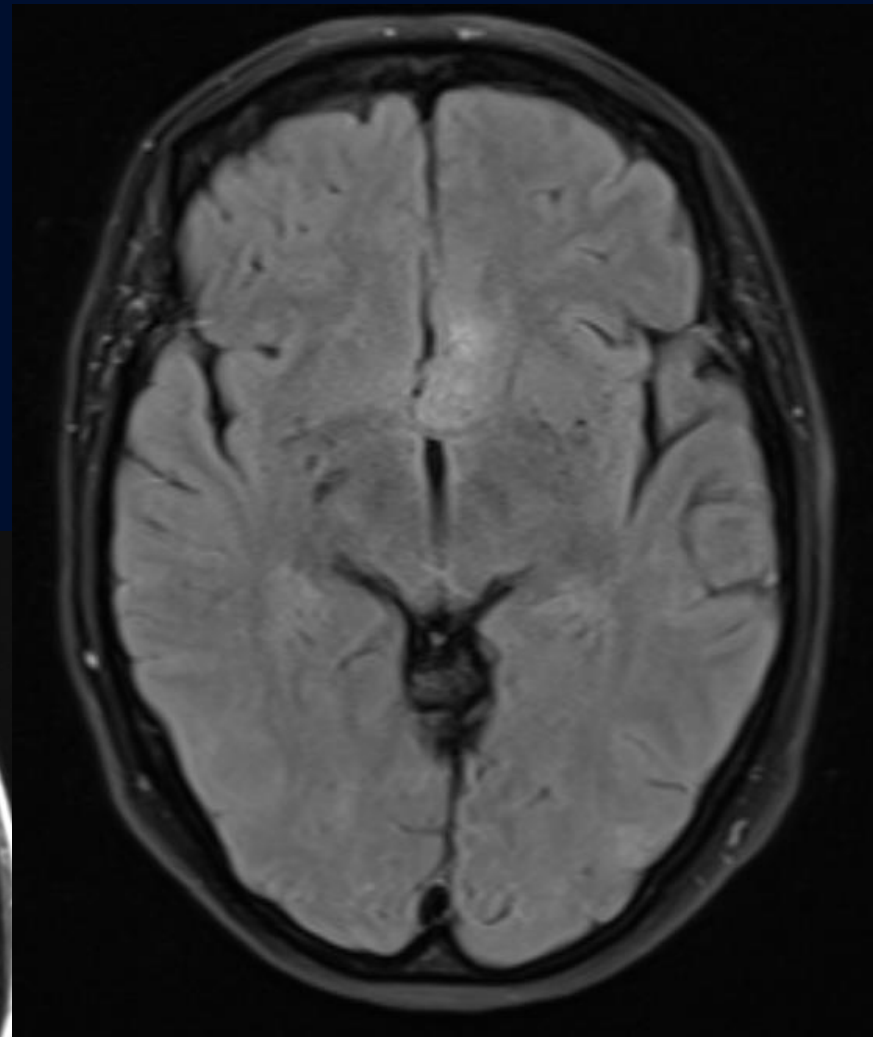
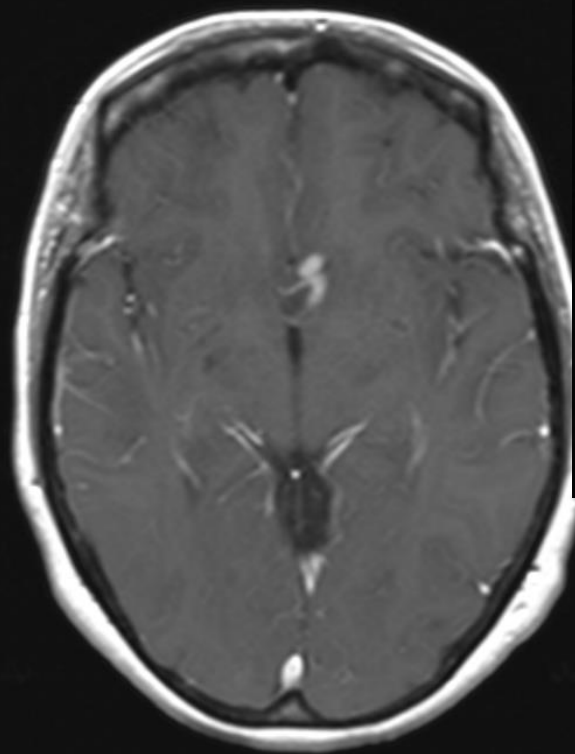
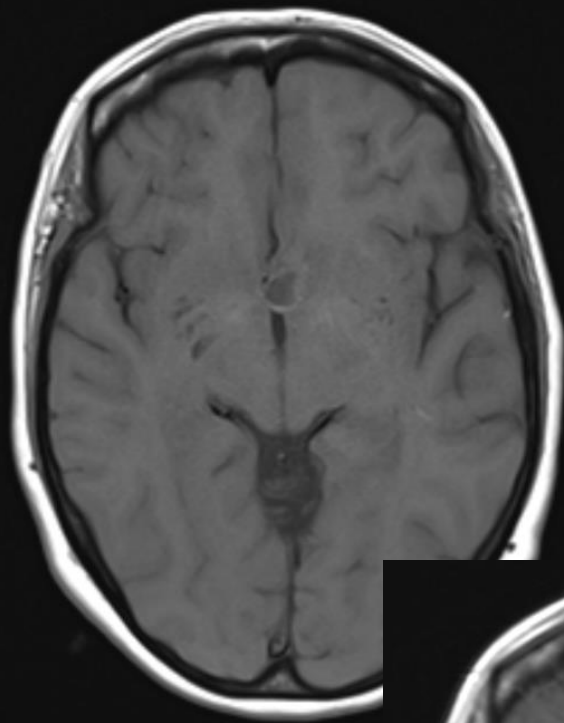
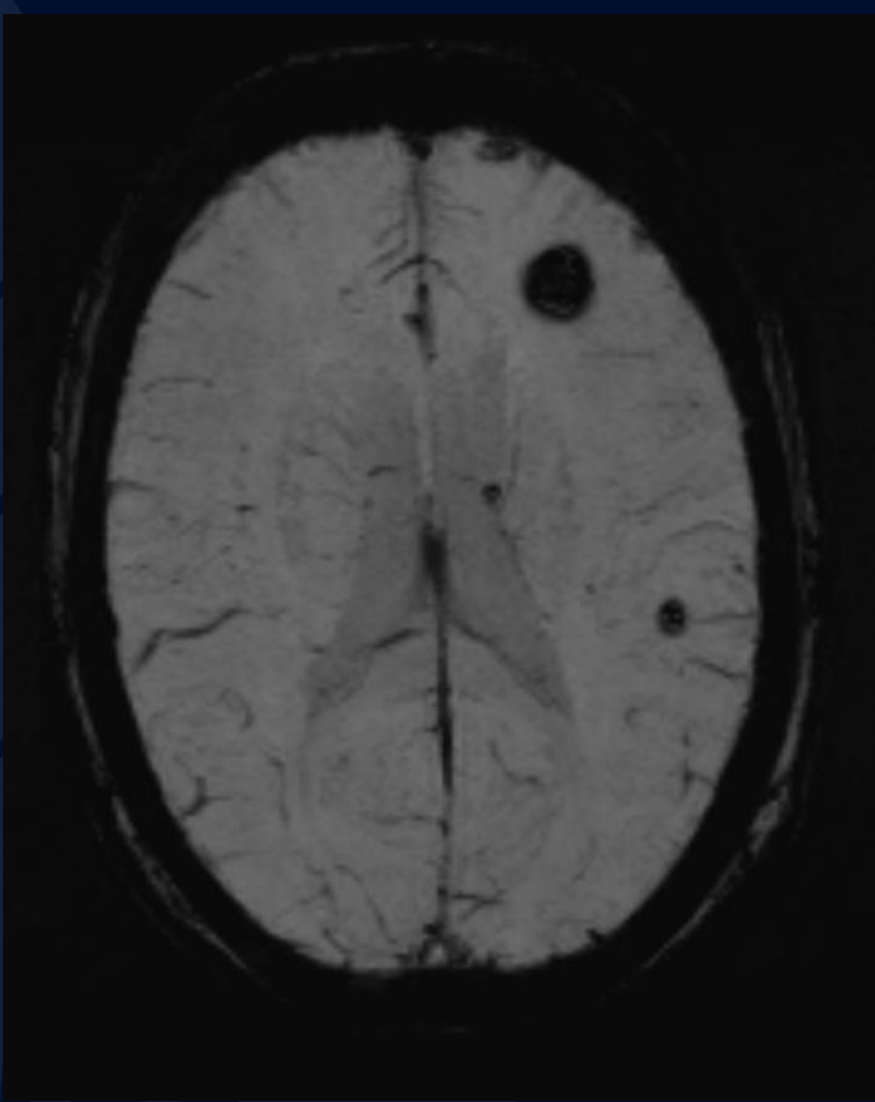


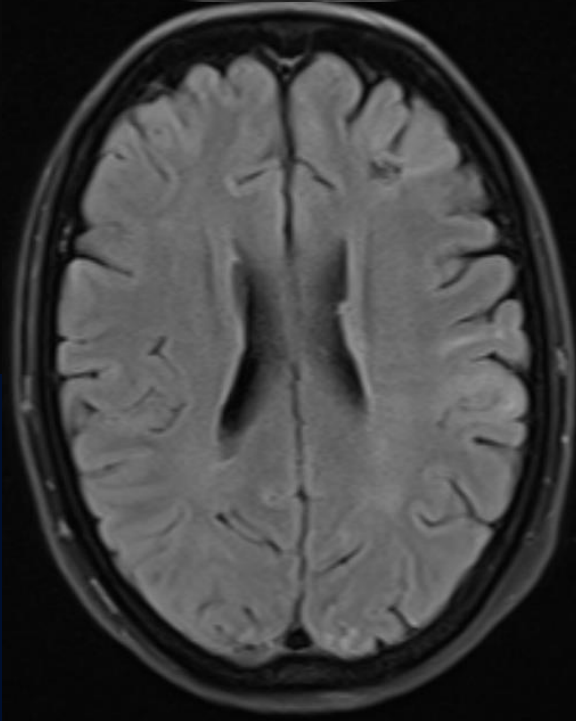
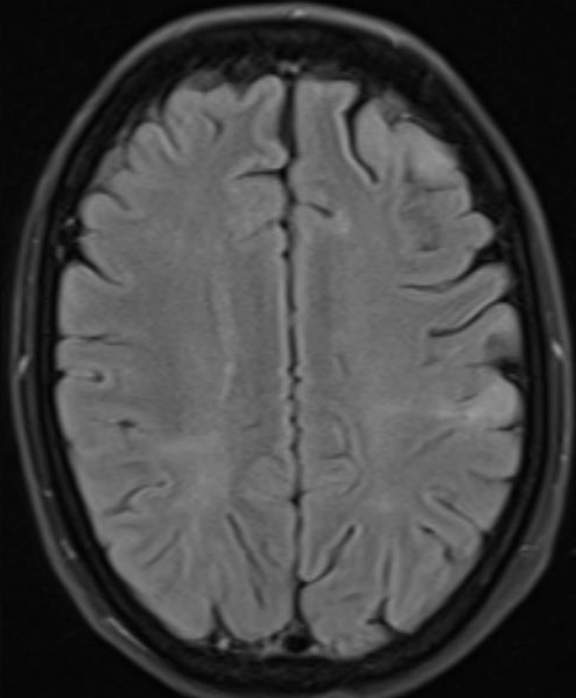
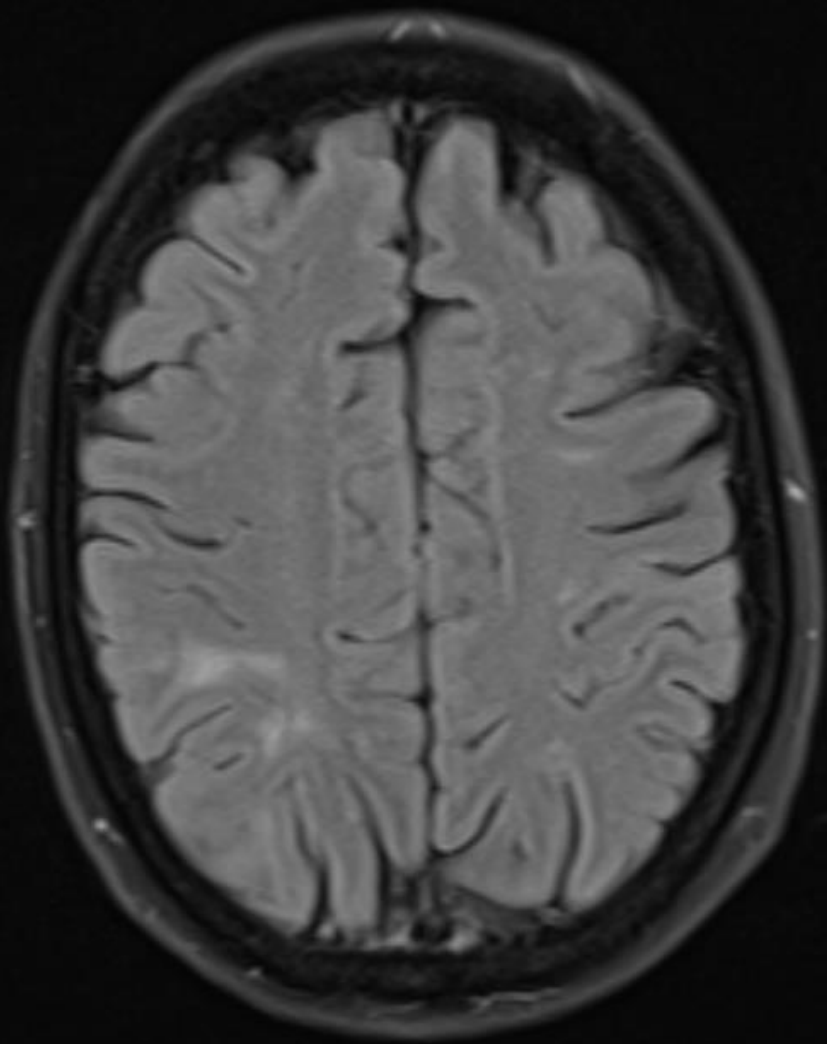
# 17-year-old male presents for routine follow-up, history of childhood seizures and developmental delay

John J. DeBevits IV, MD











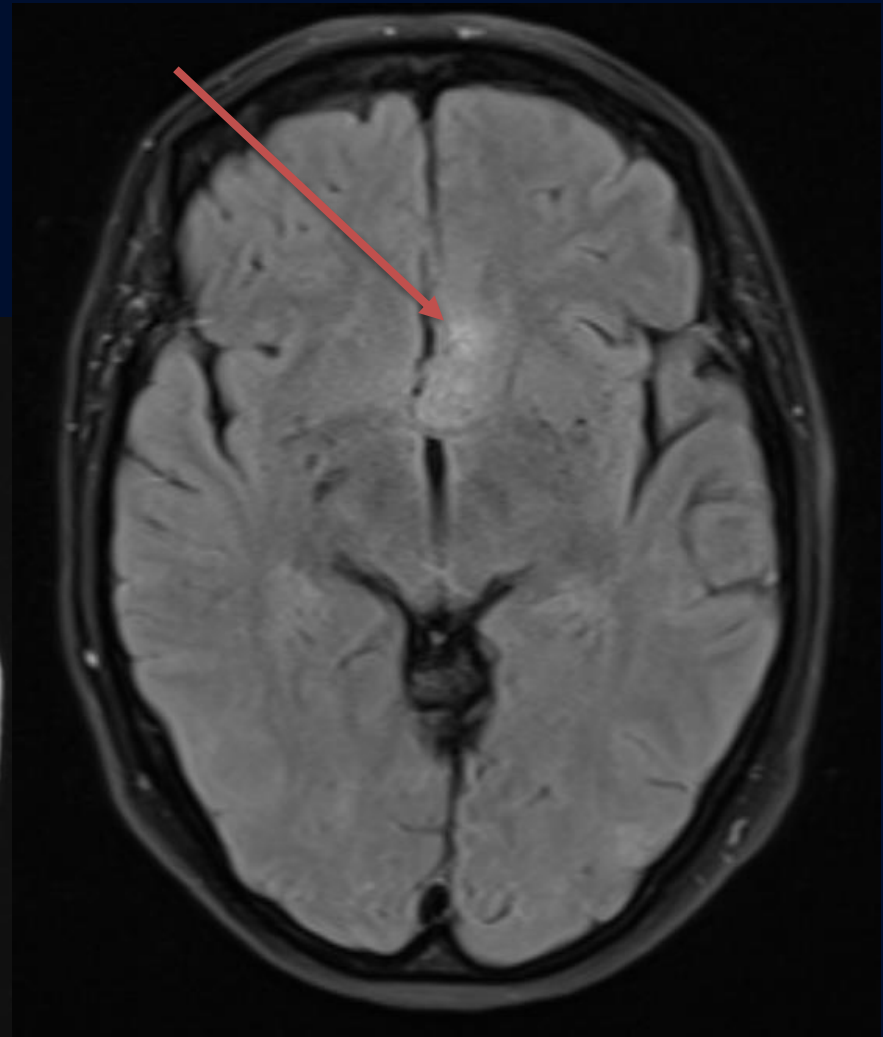
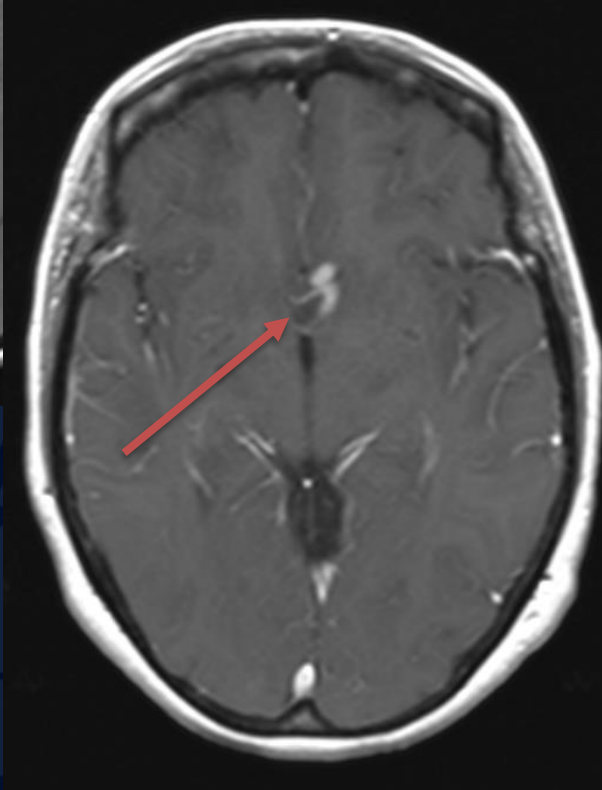
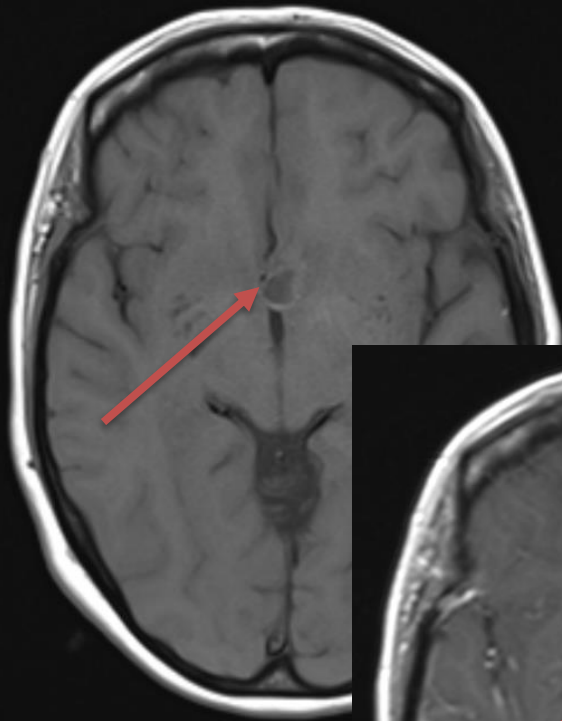
A large, stylized graphic of an oak leaf in a dark blue color, positioned on the left side of the slide. The leaf has a prominent central vein and several smaller veins branching off, with a wavy, lobed edge.

# Tuberous sclerosis

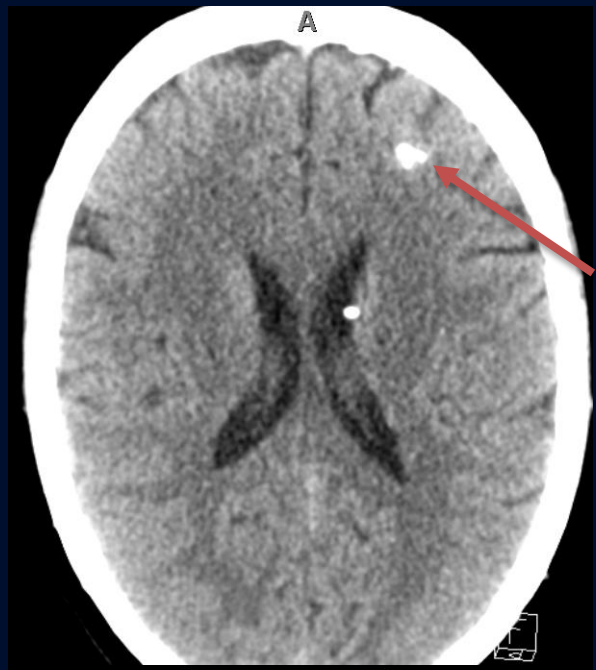
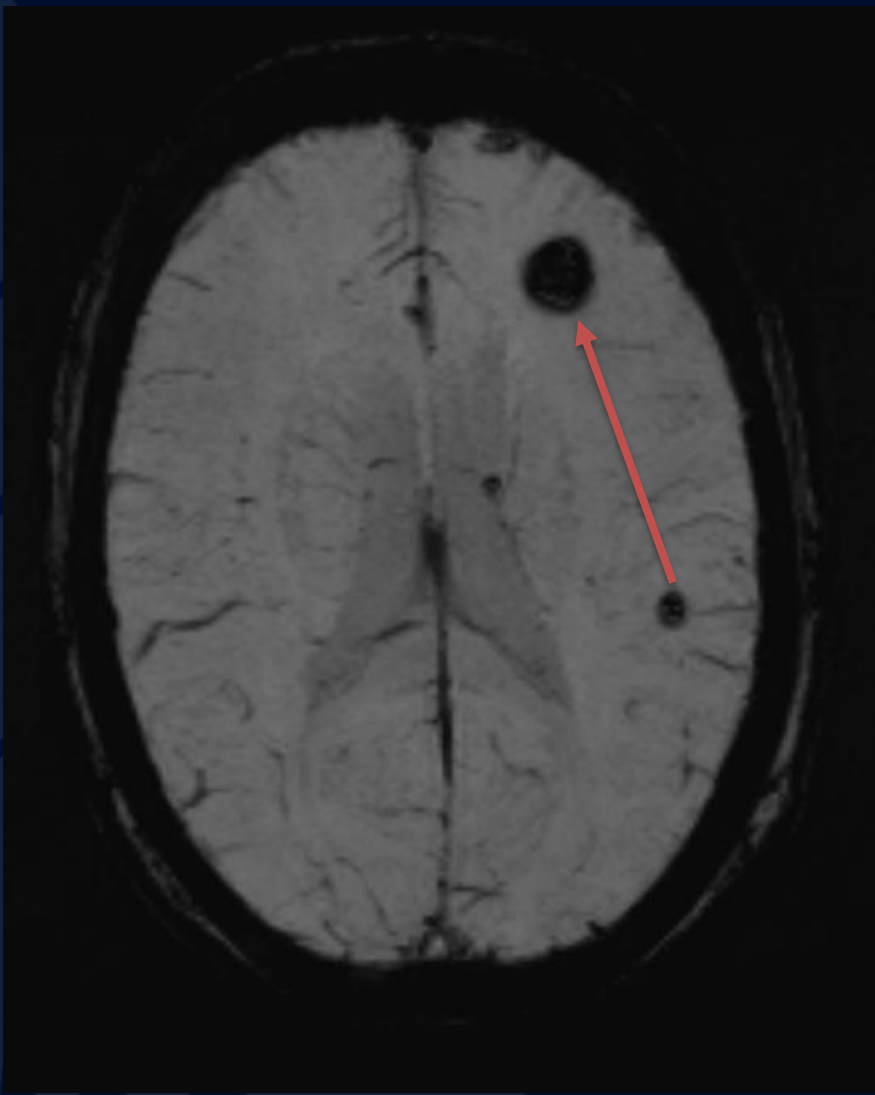


T1 sagittal demonstrates a partially cystic mass in the area of the 3<sup>rd</sup> ventricle

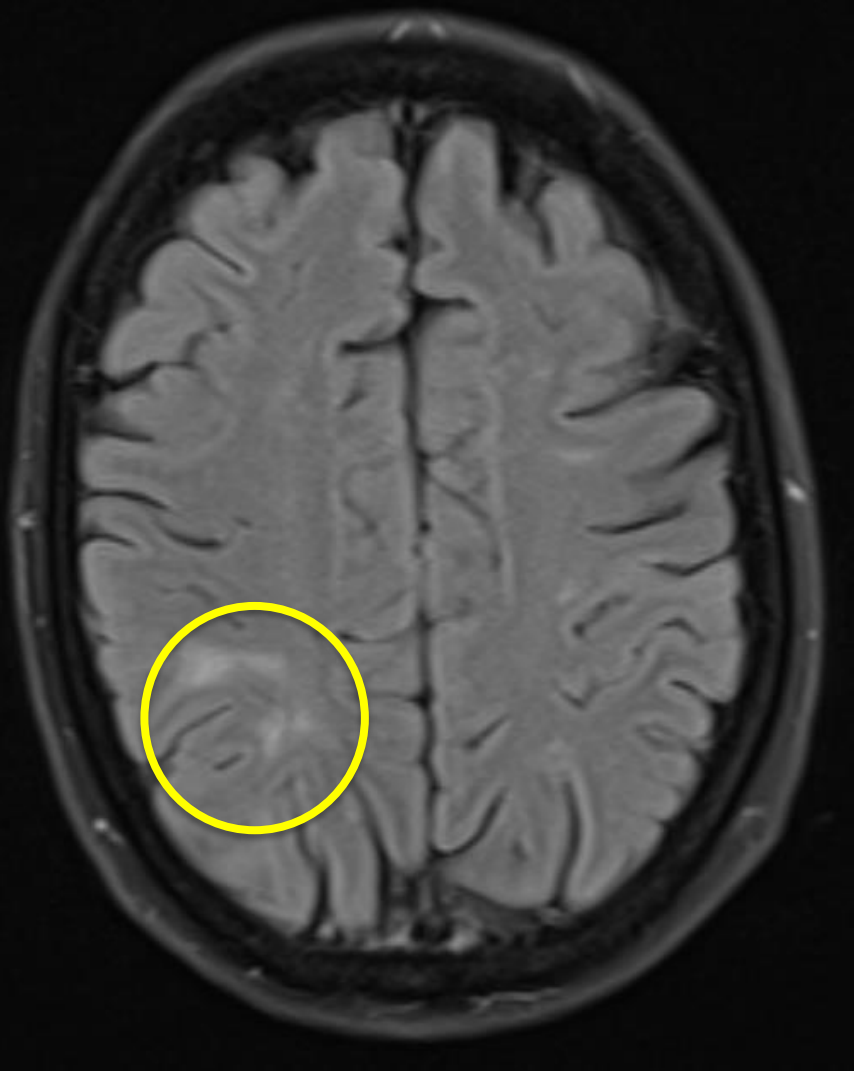




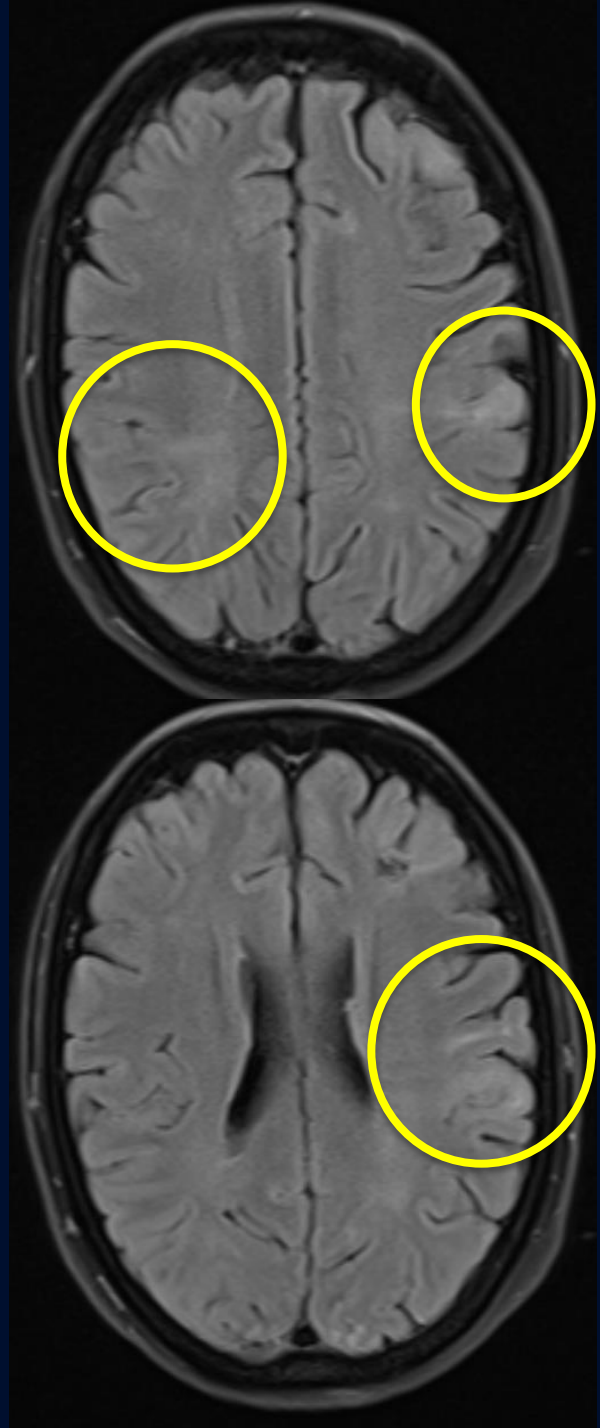
T1: partially cystic mass adjacent to the foramen of Monro on the left  
T1C+: solid component enhances strongly (most sensitive along with FLAIR)  
T2 FLAIR: Most sensitive sequence, will be hyperintense



GRE sequence (left) and noncontrast CT head (right) demonstrate calcified rounded nodules in the subcortical white matter



T2 FLAIR: multiple cortical and subcortical hyperintense lesions disrupting normal grey-white differentiation of the cortex



# Tuberous sclerosis

- Multisystem genetic disorder with epilepsy, multiorgan tumors, and hamartomas
- Mutation in TSC1 or TSC 2 gene
- 1:6,000 live births
- CNS spectrum of hamartomas with dysplastic neurons and giant (balloon) cells
- Most common presentation is neurologic
  - Seizure or infantile spasm if very young, MR, developmental delay

# Imaging findings

- Calcified subependymal nodules (SEN)
- Subependymal giant cell astrocytoma (SEGA – 15%)
  - WHO Grade I lesion
  - Most common location = foramen of Monro
  - Yearly surveillance imaging if SEGA is incompletely calcified or enhances
    - Assess for rapid growth → **obstructive hydrocephalus!**
- Cortical-subcortical tubers (95%)
- White matter radial migration lines
- Cyst-like white matter lesions
- SEN enhancement on MR > CT

# Associated abnormalities

- Renal: **Angiomyolipoma** and cysts (40-80%)
- Cardiac: **Rhabdomyoma** (50-65%)
  - Majority involute over time
- Lung: Cystic lymphangiomyomatosis/fibrosis
- Solid organs: adenomas, leiomyomas
- Skin: Ash leaf spots, facial angiofibromas, shagreen patches
- Extremities: Subungual fibromas (15-20%), cystic bone lesions, undulating periosteal new bone formation
- Ocular: “Giant drusen” (50%), retinal astrocytomas
- Dental pitting of permanent teeth in adults

# References

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- Baskin, H.J. Pediatr Radiol (2008) 38: 936.  
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