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

John J. DeBevits IV, MD

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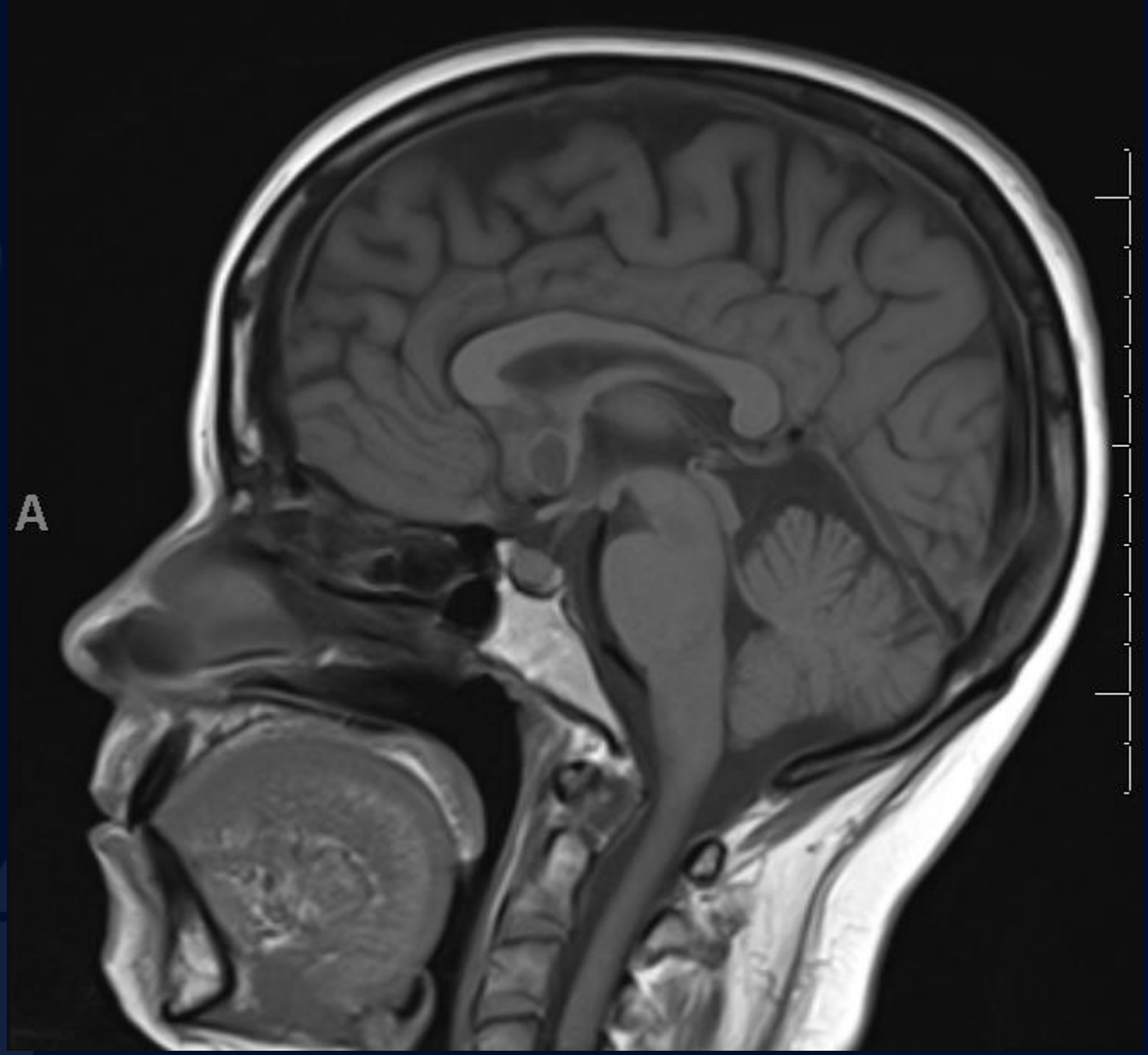

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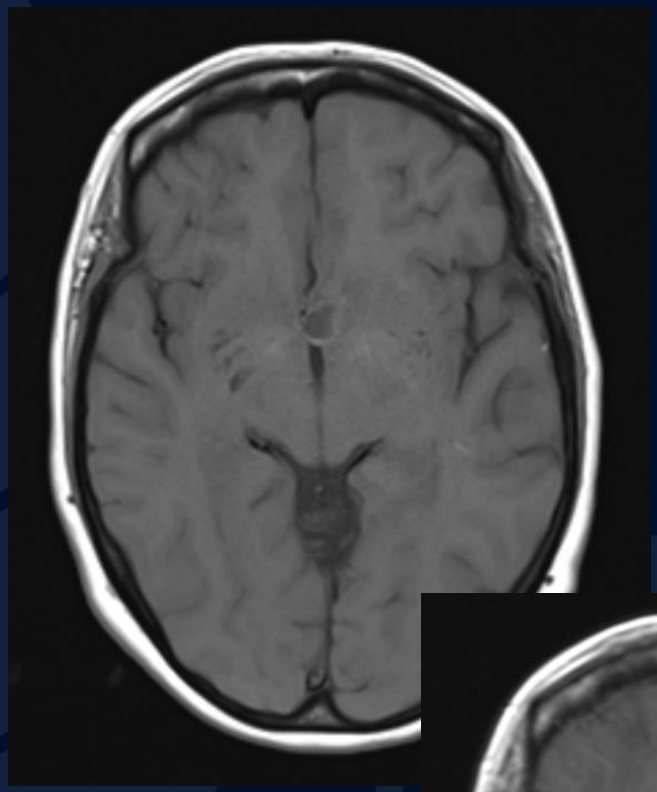

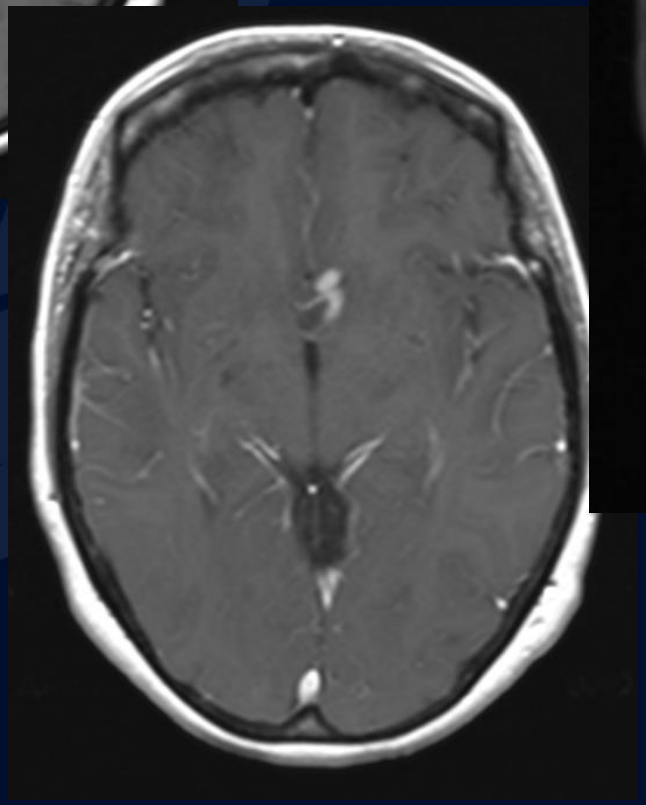

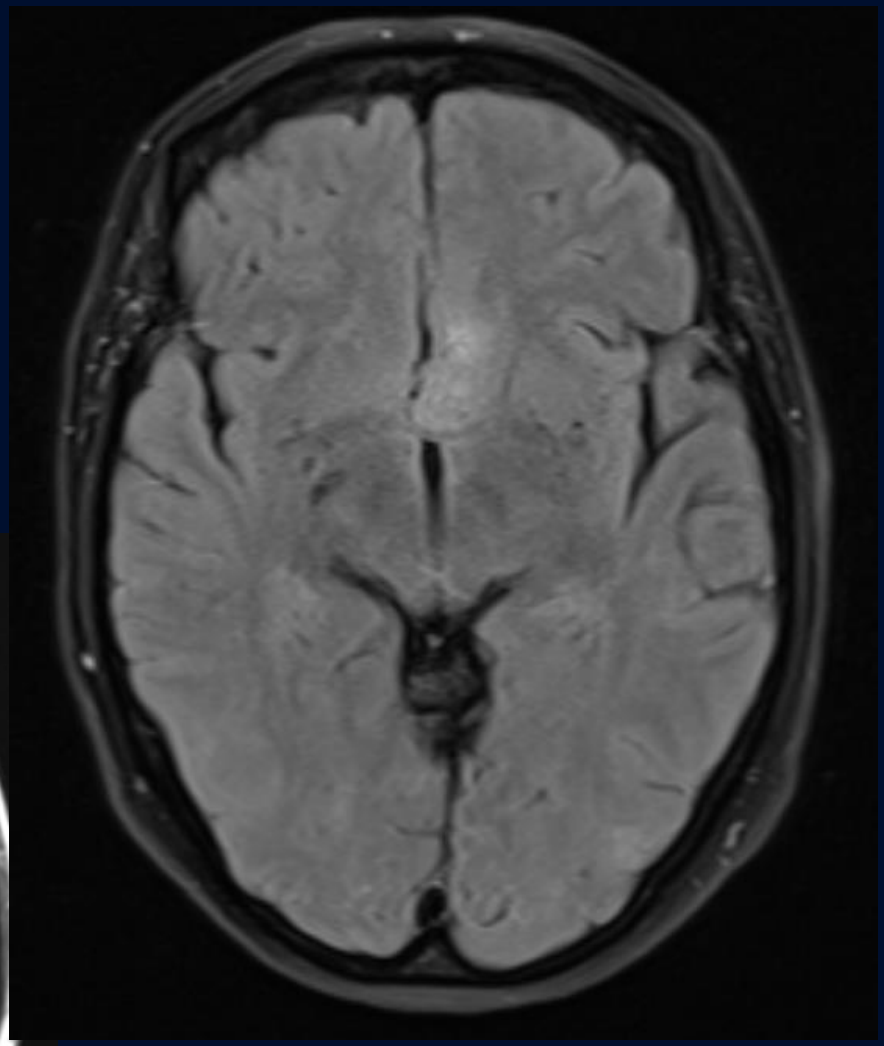

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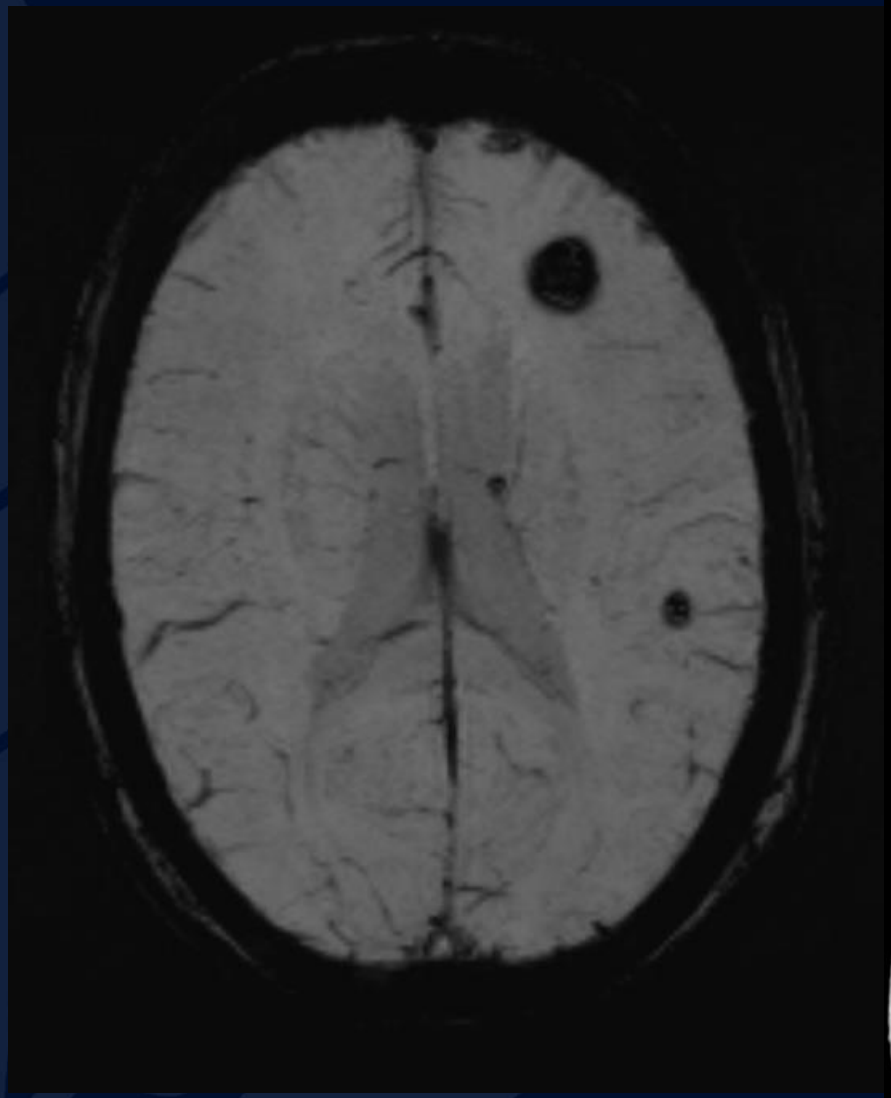

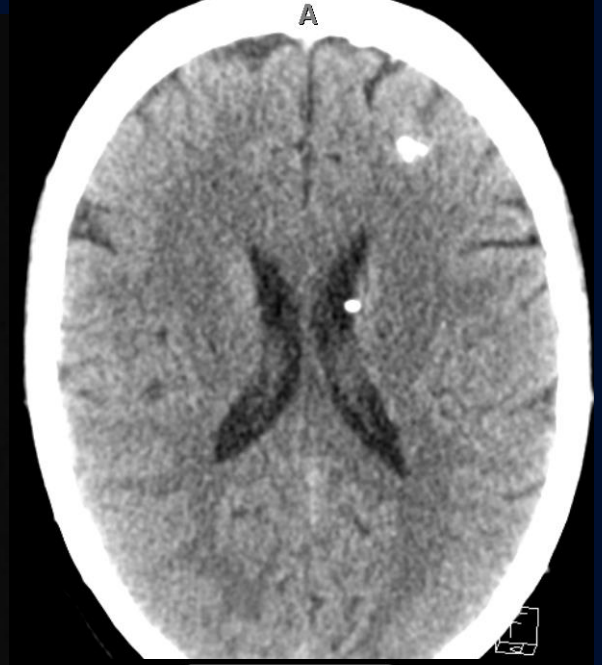

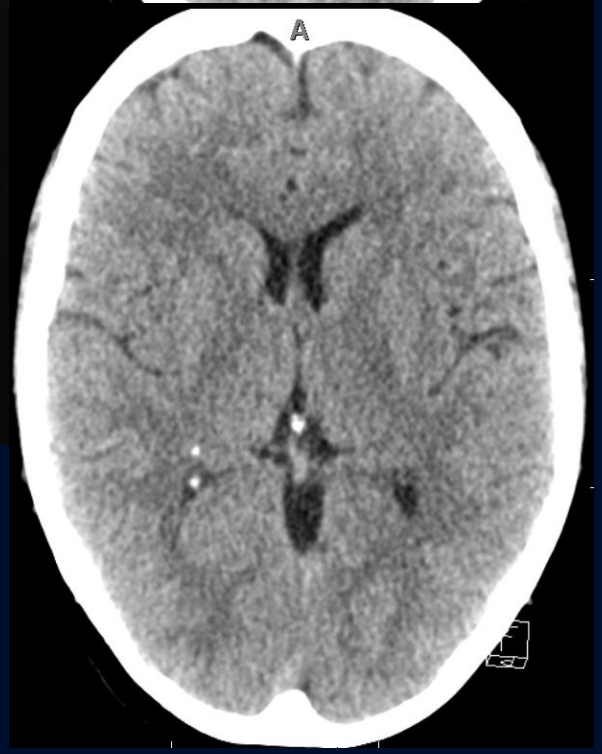

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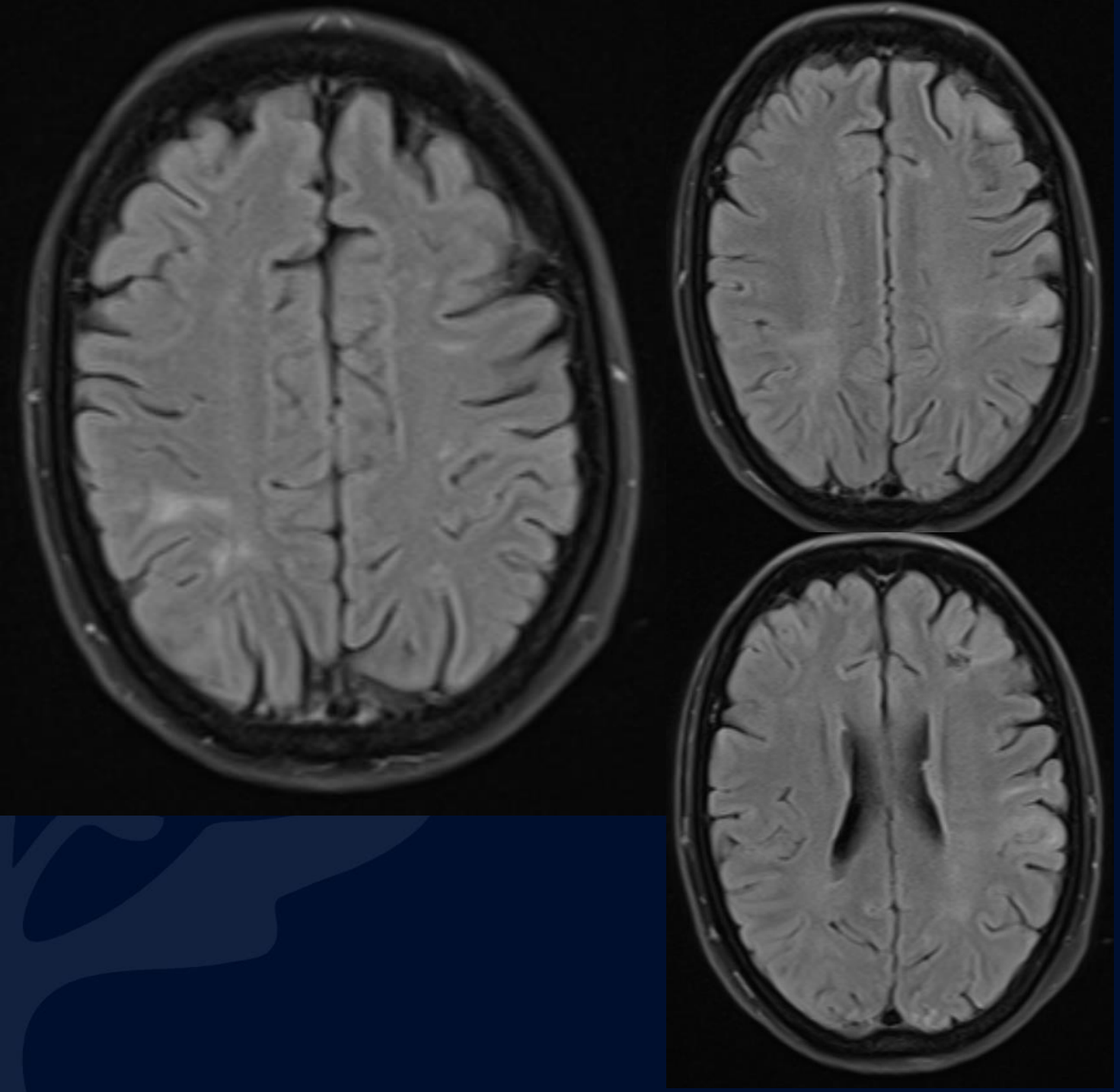

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Tuberous sclerosis

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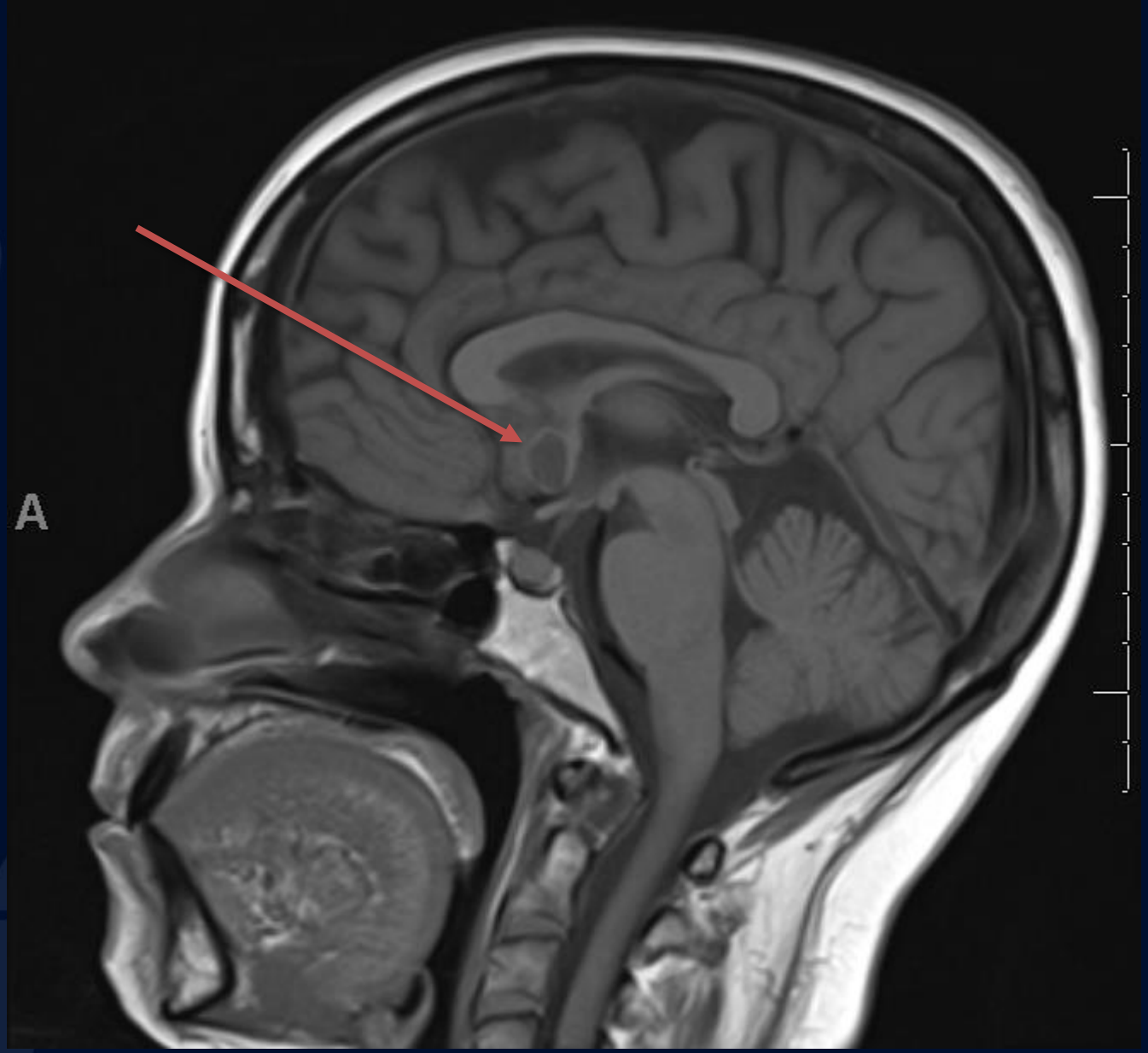

T1 sagittal demonstrates a partially cystic mass in the area of the 3rd ventricle

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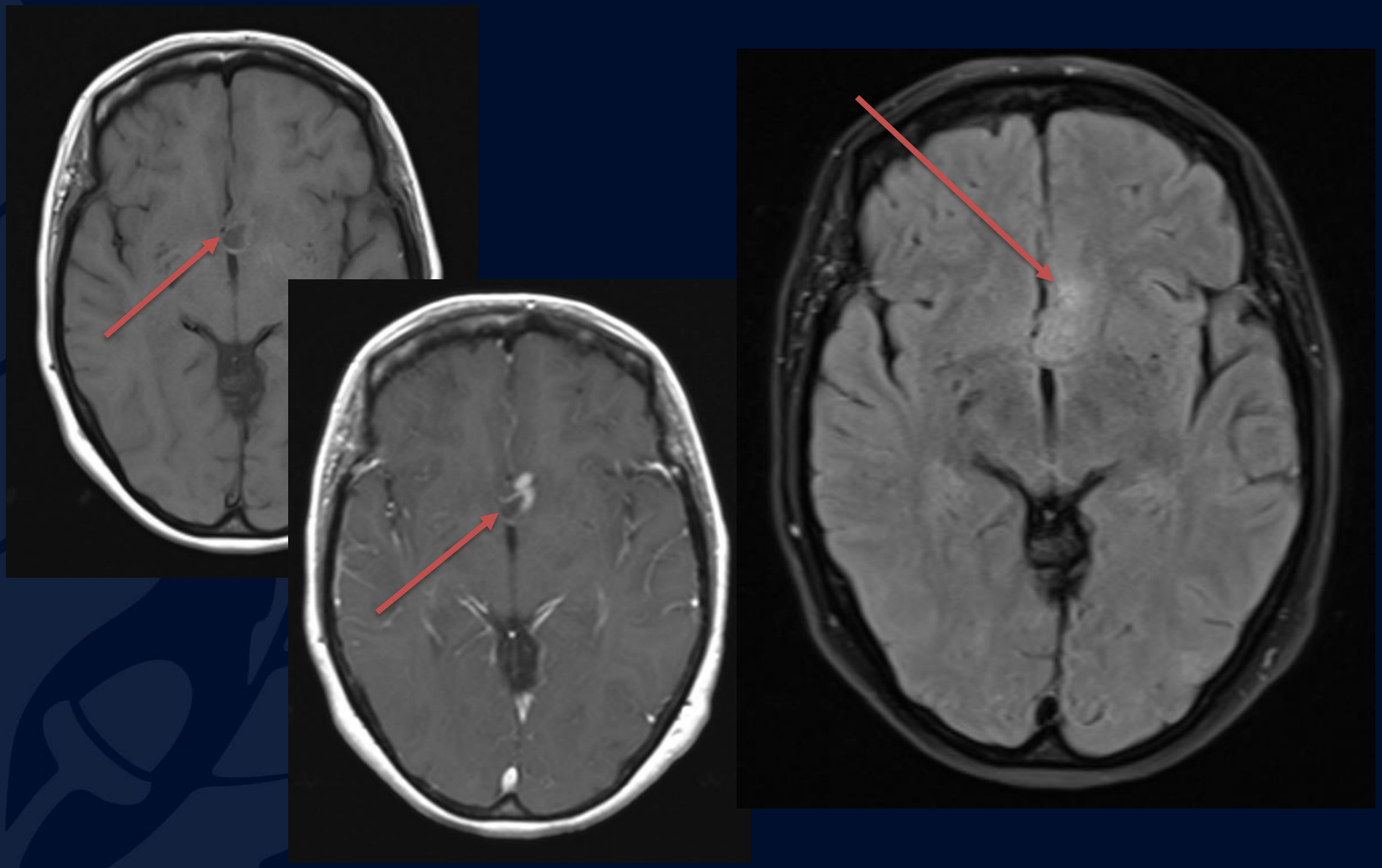

T1: partially cystic mass adjacent to the foramen of Monro on the left
T1C+: solid component enhances strongly (most sensitive along with FLAIR)
T2 FLAIR: Most sensitive sequence, will be hyperintense

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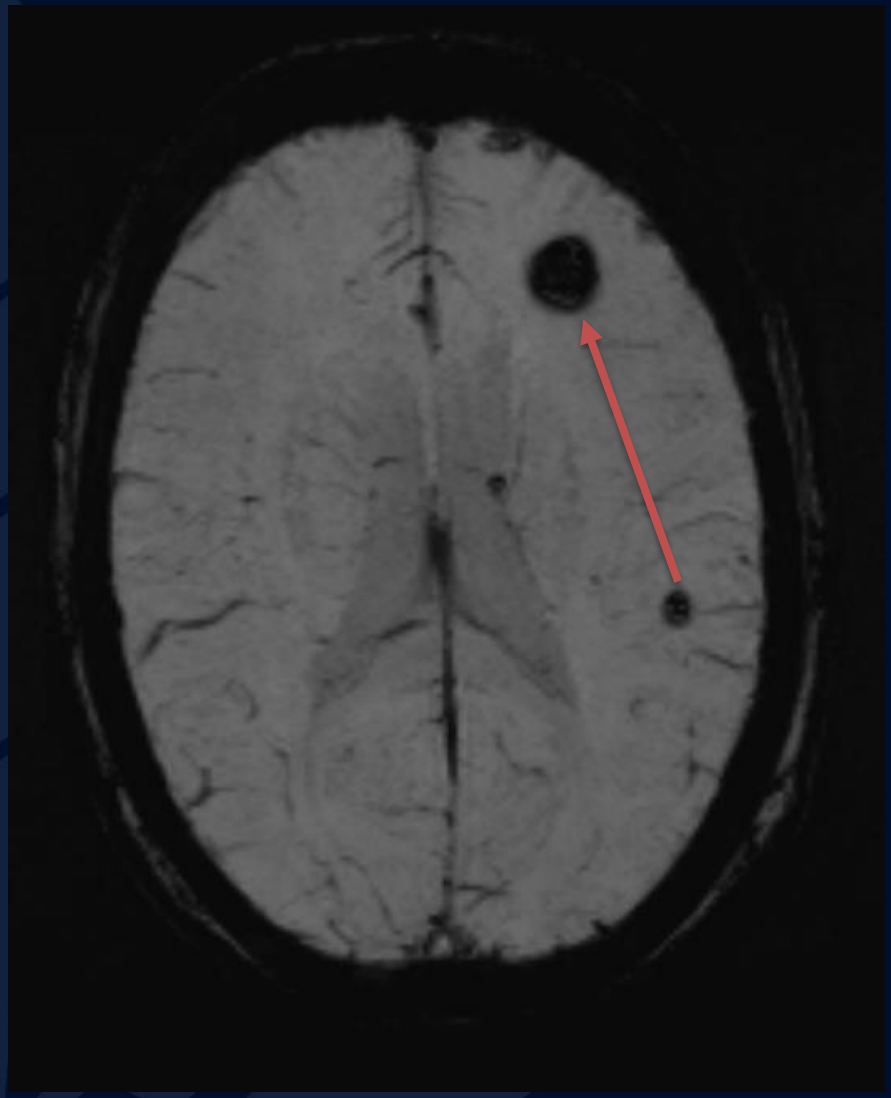
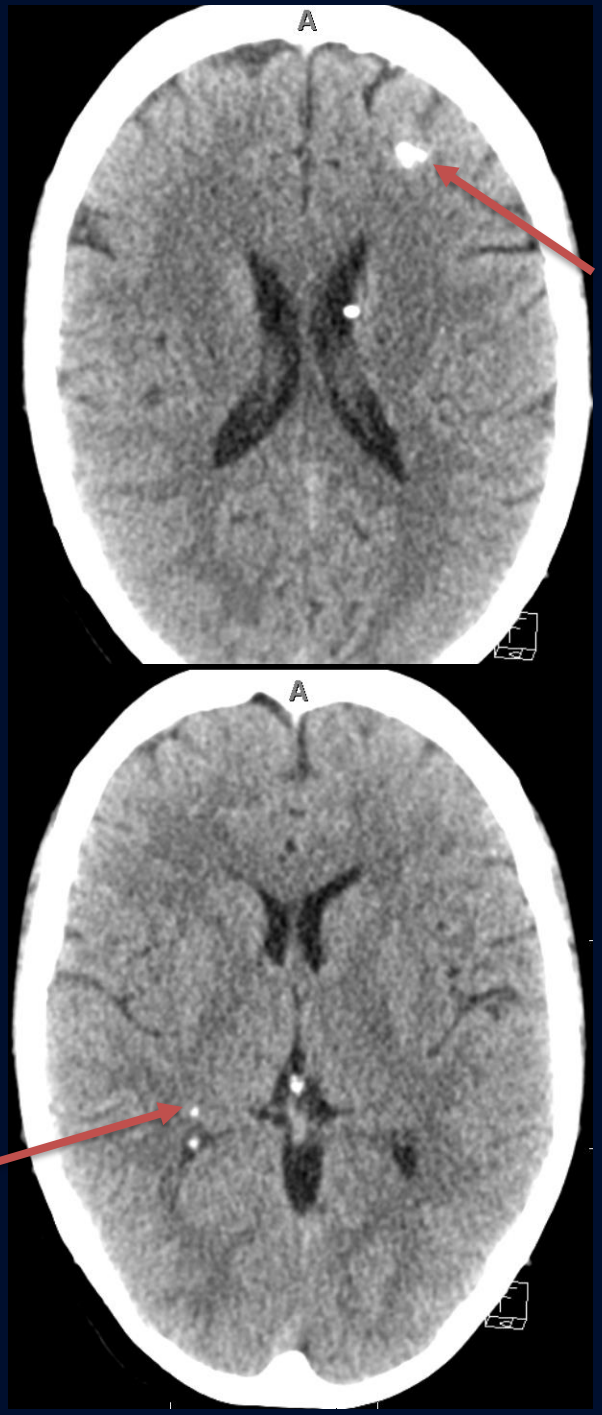
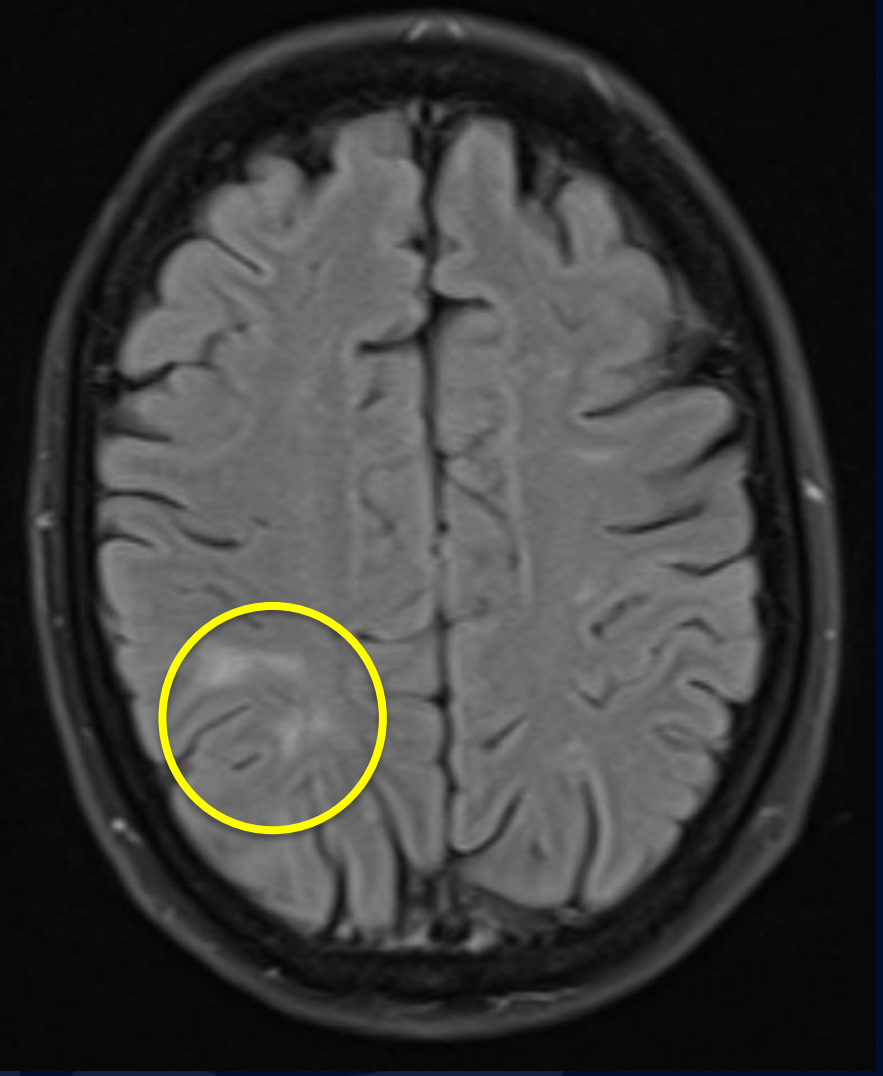
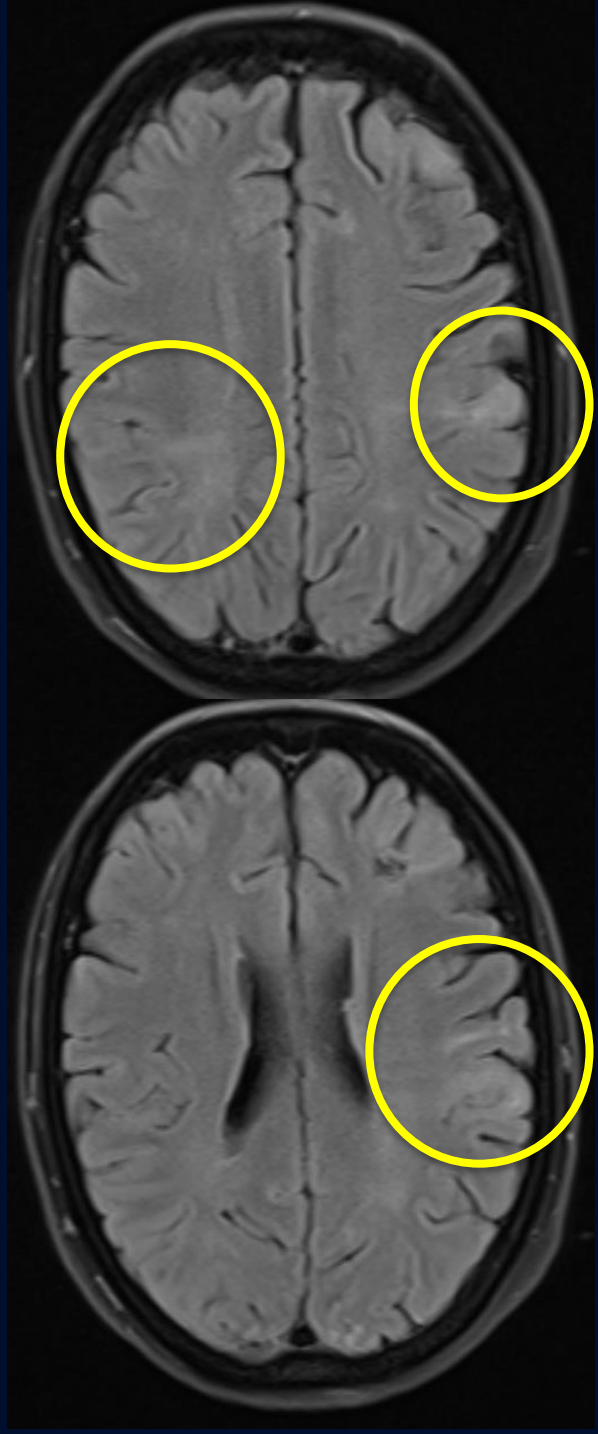




GRE sequence (left) ad noncontrast CT head (right) demonstrate calcified rounded nodules in the subcortical white matter





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

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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: Subungal 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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