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

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Tuberous sclerosis
T1 sagittal demonstrates a partially cystic mass in the area of the 3rd 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 lymphangiomatosis/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

• American Journal of Roentgenology. 2015;204: 933-943. 10.2214/AJR.13.12235

  https://link.springer.com/article/10.1007%2Fs00247-008-0832-y