Tuberous Sclerosis

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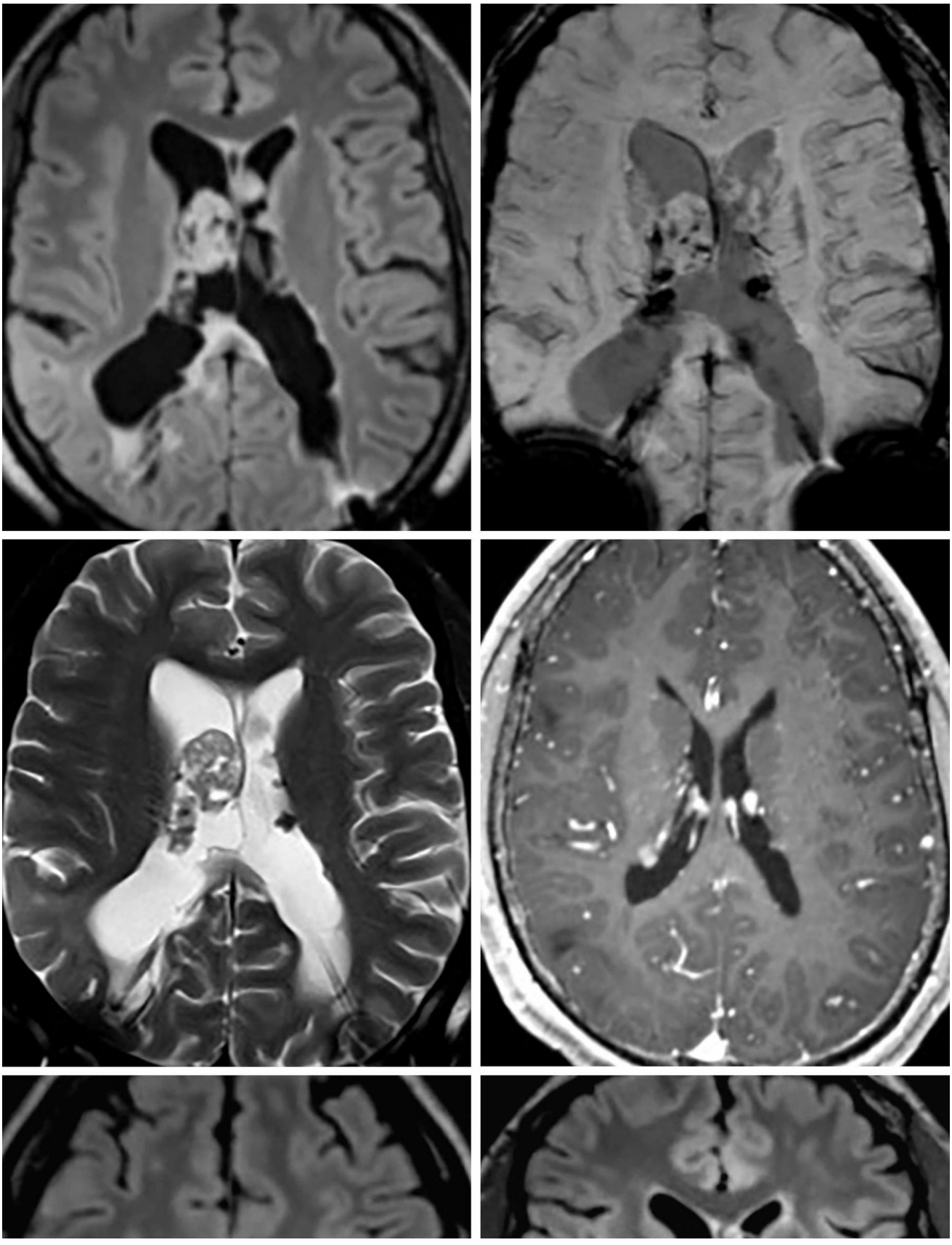

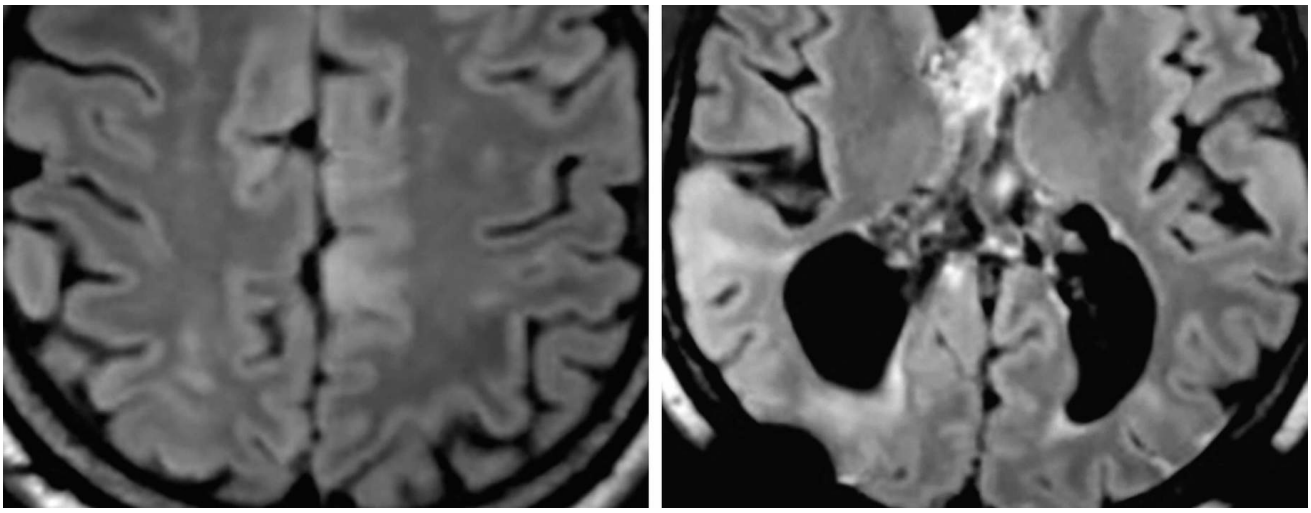

Figure 1: Multiple FLAIR-hyperintense (top left) subependymal nodules with corresponding dark susceptibility artifact (top right) that reflects calcification. (Middle Left) These nodules are also clearly visible in a T2-weighted image against the bright cerebrospinal fluid. (Middle Right) A few of the nodules demonstrate moderate enhancement. Note also the much larger heterogeneous lesion along the right lateral ventricle at the level of the caudothalamic groove compatible with a SEGA. In addition, there are numerous regions of cortical expansion with T2/FLAIR-hyperintense signal (middle left and bottom left) and associated, increased linear signal radiating from the periventricular white matter to the subcortical regions (bottom right), known as the radial band sign.

Description

- Multisystem, neurocutaneous disorder characterized by development of multiple benign tumors of ectodermal origin

Pathology

- Caused by mutation in the *TSC1* or *TSC2* gene
- Mutations lead to abnormal cellular differentiation and proliferation

Clinical Features

- Symptoms
    - Most commonly has neurologic manifestations
    - Classic triad is actually uncommon (~30%)

        - Facial angiofibromas, seizures, and mental retardation
- Age
    - Highly variable age at presentation due to variable penetrance
- Gender
    - No predilection

Imaging

- General
    - Calcified subependymal nodules, subependymal giant cell astrocytomas (SEGAs), and cortical and subcortical tubers
- Modality specific
    - CT
        - Tubers: hypodense to isodense, although often difficult to appreciate with CT
        - Subependymal nodules
            - Calcified and noncalcified subependymal nodules
            - Presence of enhancement raises the concern of developing SEGAs
    - MRI
        - T1WI
            - Tubers: hypointense to isointense, sometimes hyperintense
            - Subependymal nodules: often hyperintense
            - SEGAs: hypointense to isointense
        - T2WI
            - Tubers: isointense to slightly hyperintense
            - Subependymal nodules: isointense to hyperintense
            - SEGAs: heterogeneous isointense to hyperintense
        - DWI
            - No restriction
        - T2*
            - Subependymal nodules and SEGAs: hypointense calcifications

        - Contrast
            - Tubers: rare enhancement
            - Subependymal nodules: may have enhancement
            - SEGAs: avid enhancement
- Imaging recommendations
    - MRI with contrast
- Mimic
    - Superficial tubers can mimic gliomas but are usually multifocal. Subependymal nodules may have the appearance of intraventricular ependymal tumor spread. However, calcifications of these nodules in CT imaging is virtually pathognomonic for tuberous sclerosis.

For more information, please see the corresponding chapter in Radiopaedia.

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