35 y/o female with headache and dementia

Jignesh Modi, MD

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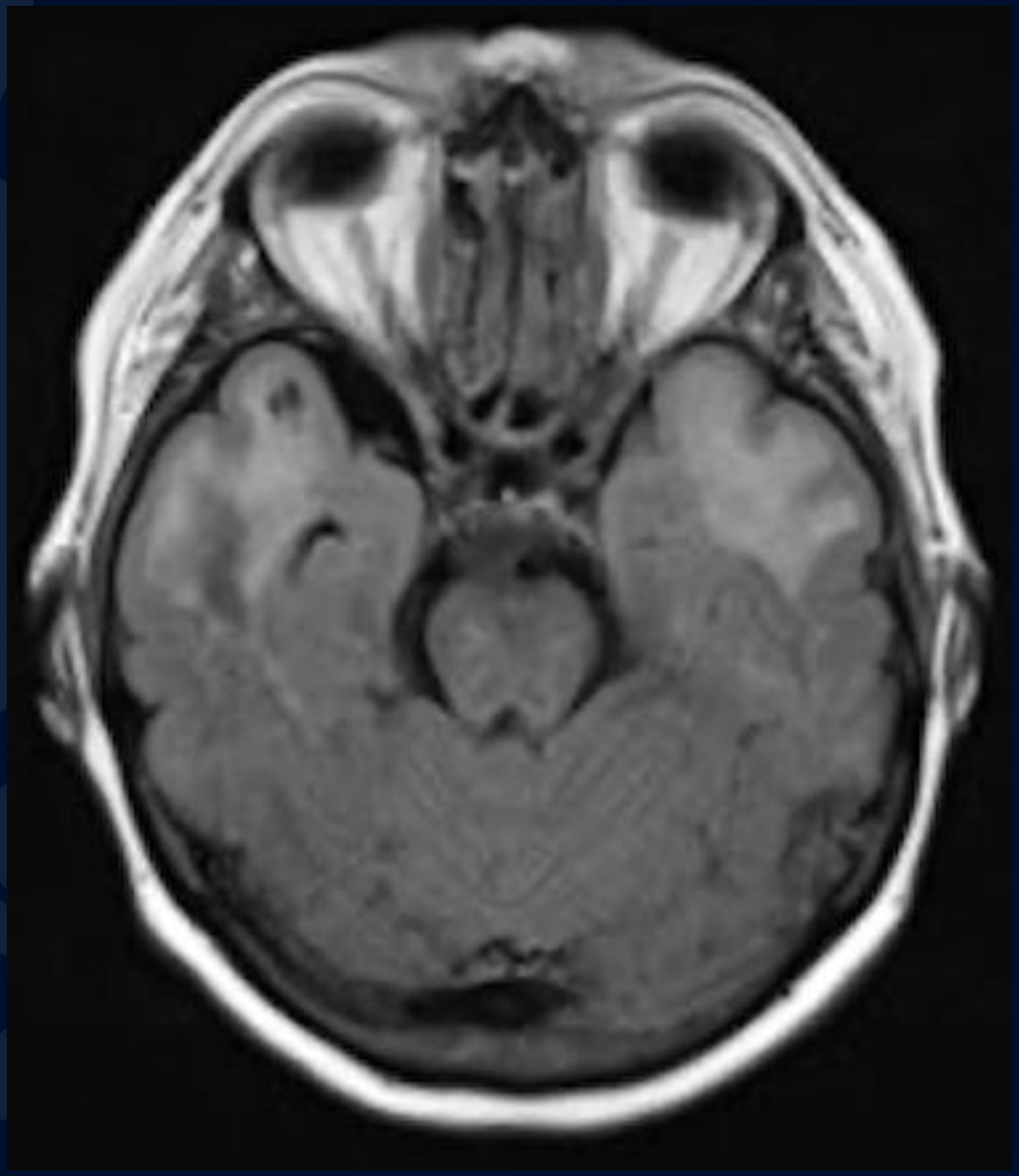

Axial Flair

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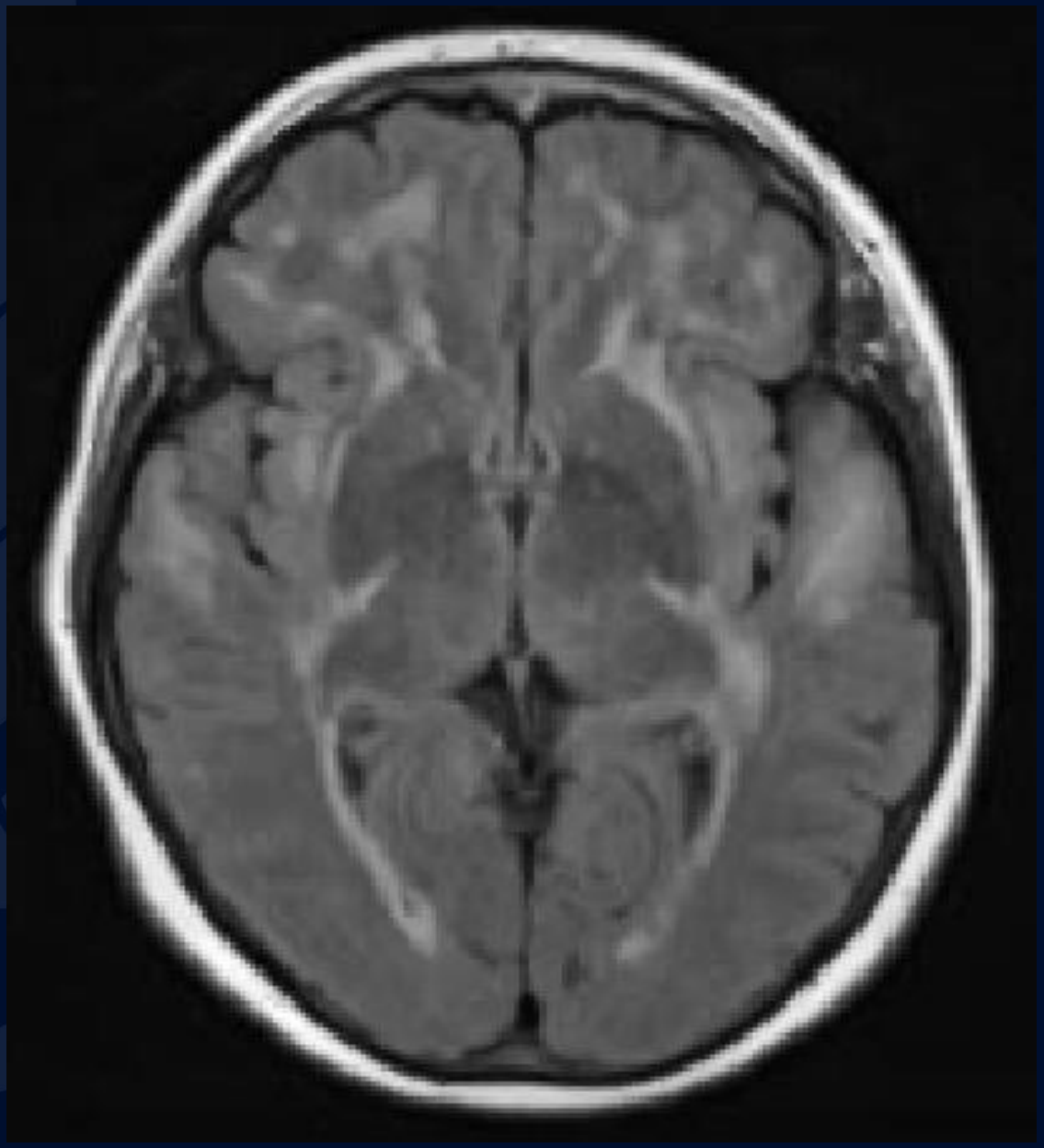

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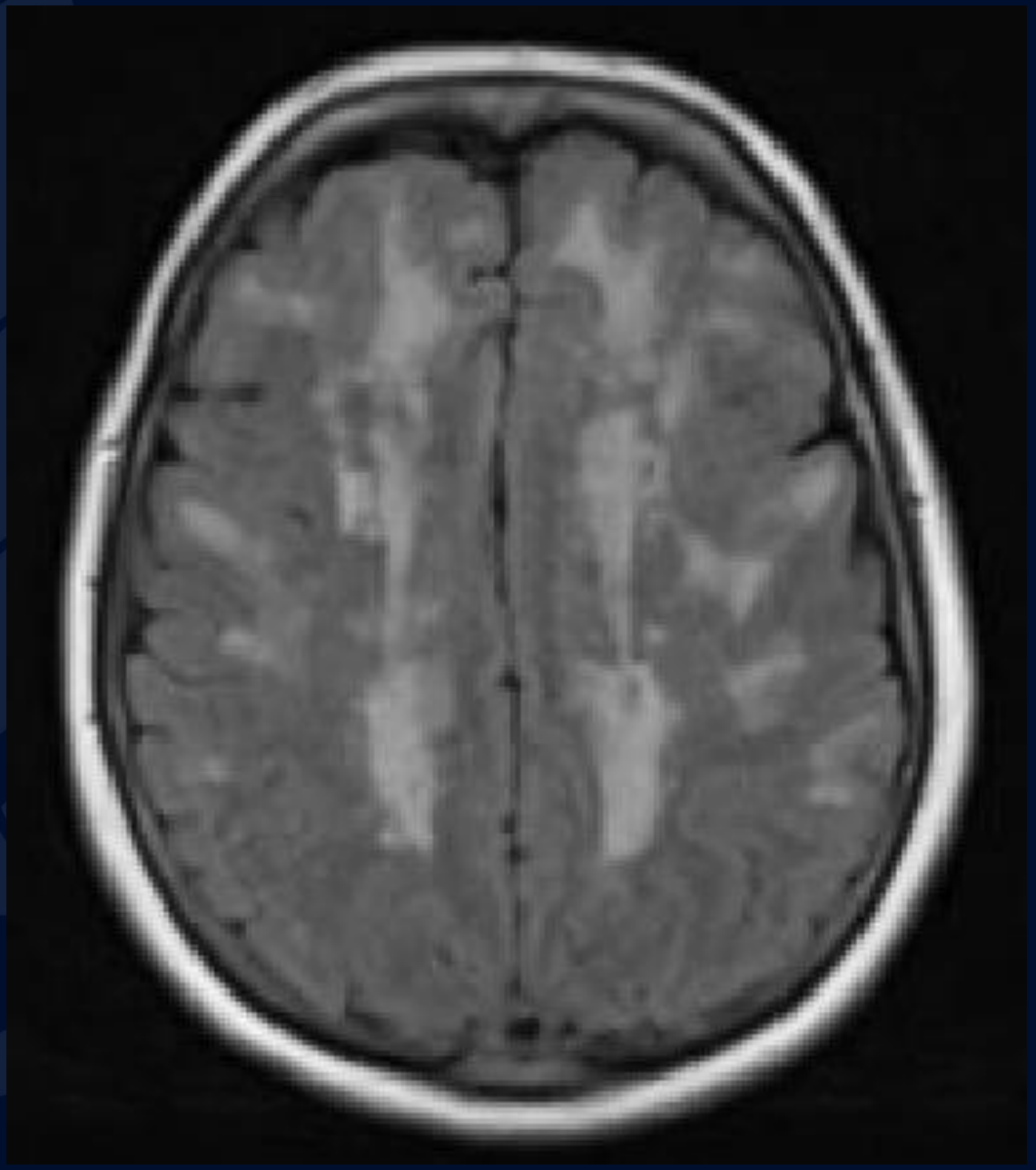

Axial Flair

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Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL)

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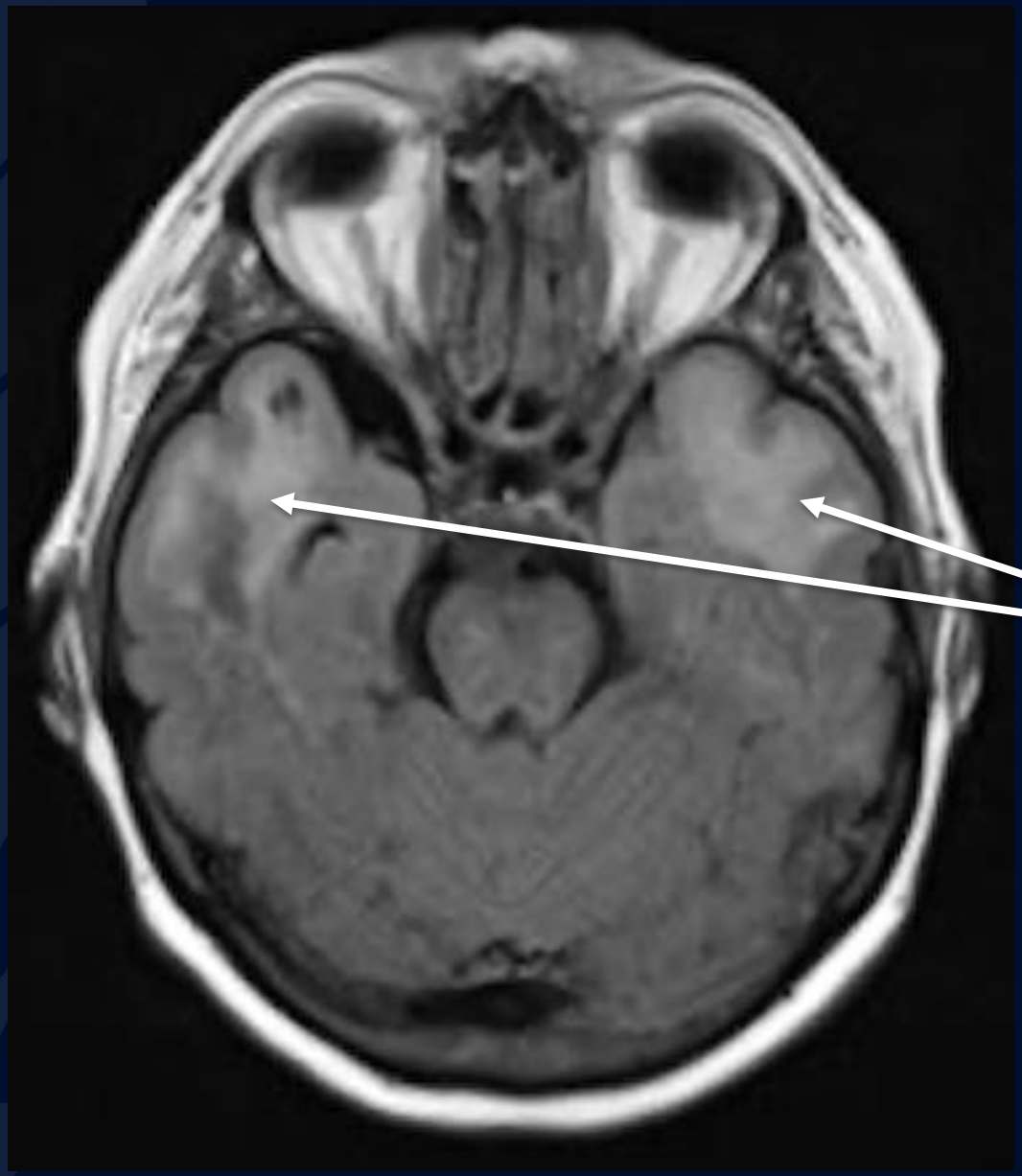

Axial Flair

Anterior temporal lobe involvement bilaterally

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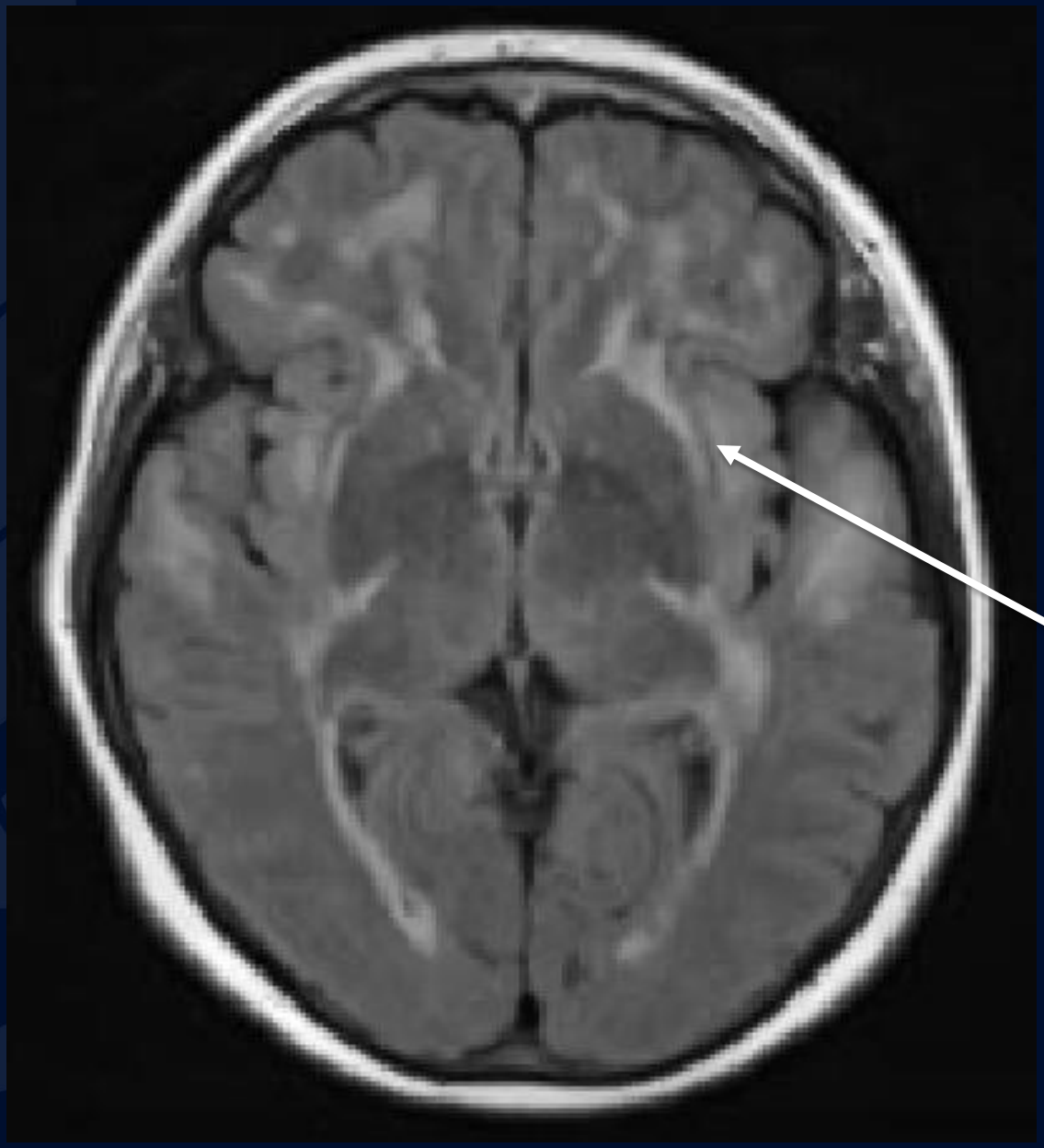

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External capsule involvement

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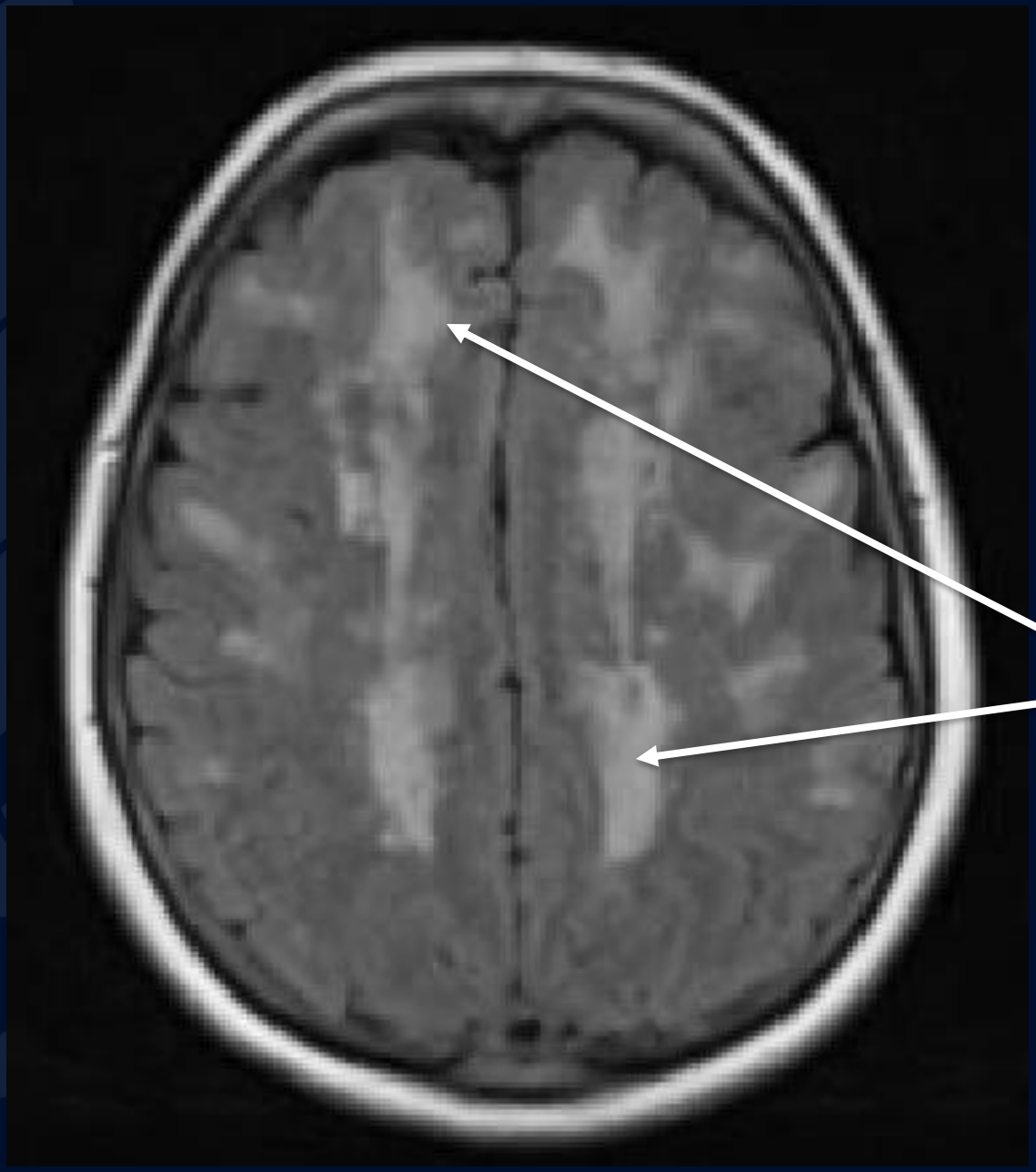

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Confluent white matter involvement

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CADASIL

- Autosomal dominant microvasculopathy characterized by recurrent lacunar and subcortical white matter ischemic strokes and vascular dementia in young and middle-age patients without known vascular risk factors. There is disproportionate cortical hypometabolism.
- An autosomal dominant trait, with patients typically becoming symptomatic in adulthood (30 to 50 years of age).
- Presentation is usually with recurrent TIAS or strokes in multiple vascular territories. Presenile dementia and migraines develop in the third-to-fourth decades of life. Clinically, CADASIL often has a similar presentation to migraines and may also have auras.
  - Depression, psychosis, pseudobulbar palsy and focal neurological defects as well as seizures are also seen.

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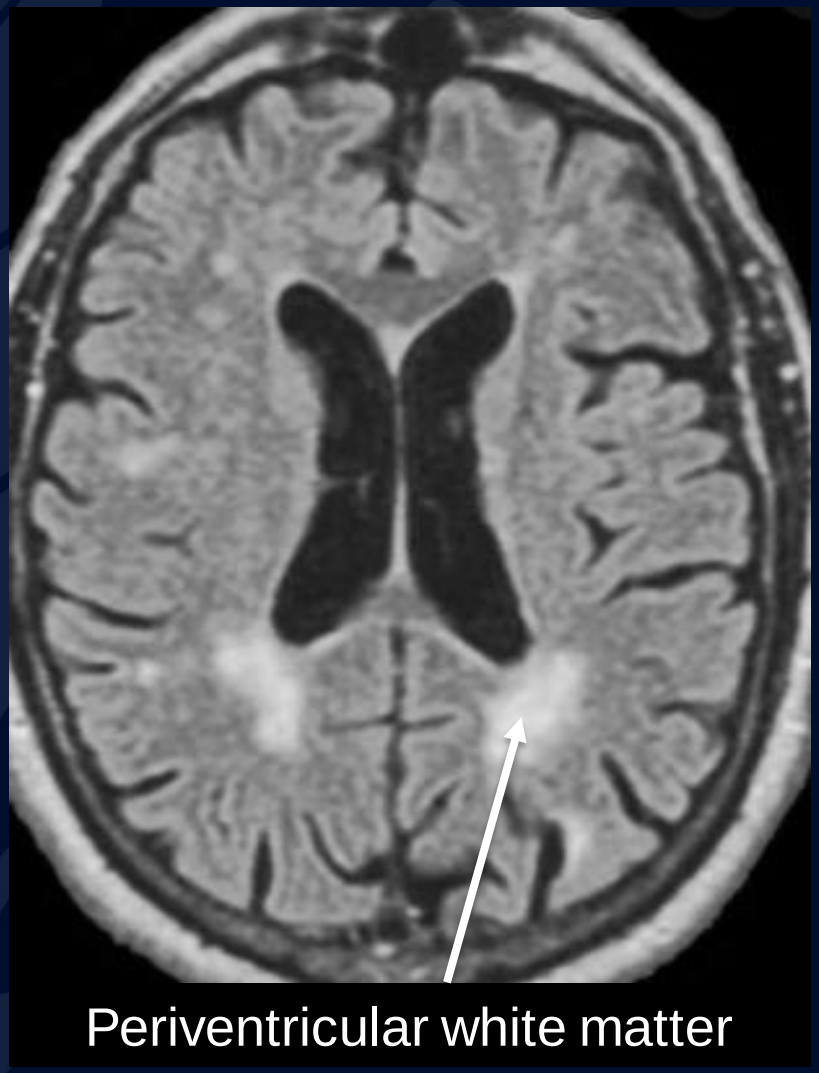
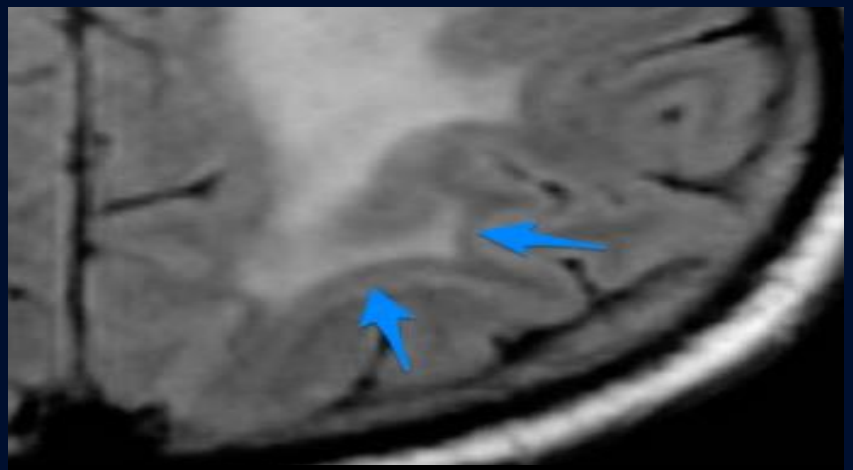
Pathology

- Results from a mutation on chromosome 19p13.12 involving the *NOTCH3* gene.
- Autosomal dominant trait.
- It results in small vessel and arteriole stenosis secondary to fibrotic thickening of the basement membrane of the vessels; pathological hallmark is the deposition of granular osmiophilic material in close relation to the vascular smooth muscle cells.

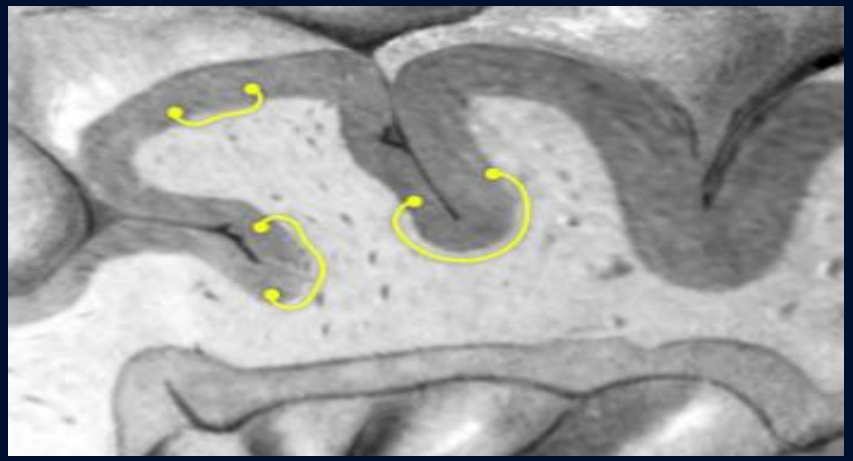
Anatomy



Periventricular white matter



Subcortical U-FIBERS



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Imaging Features

- MRI is the study of choice.
- Widespread confluent white matter hyperintensities.
- Subcortical white matter can be diffusely involved, in the initial course of the disease involvement of the anterior temporal lobe (86%) and external capsule (93%) are classical.
- Relative sparing of the occipital and orbitofrontal subcortical white matter , subcortical U fibers and cortex
- Cerebral microhemorrhages in 45% cases (25-70%) without characteristic distribution.
- Eventually, cerebral atrophy ensues, which correlated well with degree of cognitive decline.

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Treatment & Prognosis

- Variable but progressive course and leading to death between 50-70 years of age.
- Differential diagnosis: MELAS, SUSAC syndrome, CNS Vasculitis.
- Younger patient with small vessel ischemic white mater changes is the clue.
- Predilection of anterior temporal lobes white matter is a distinctive feature.
- Sparing of the cortex and subcortical U-fibers is typical.

References

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