

Creutzfeldt-Jakob disease

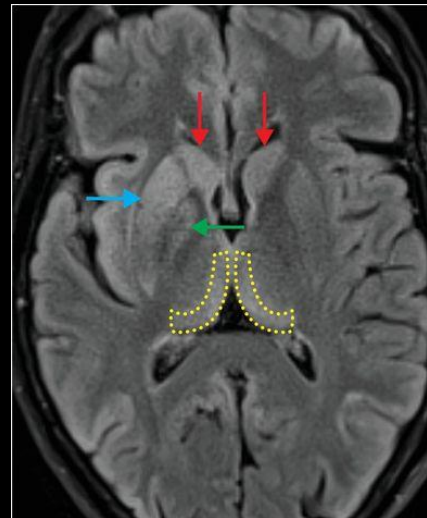
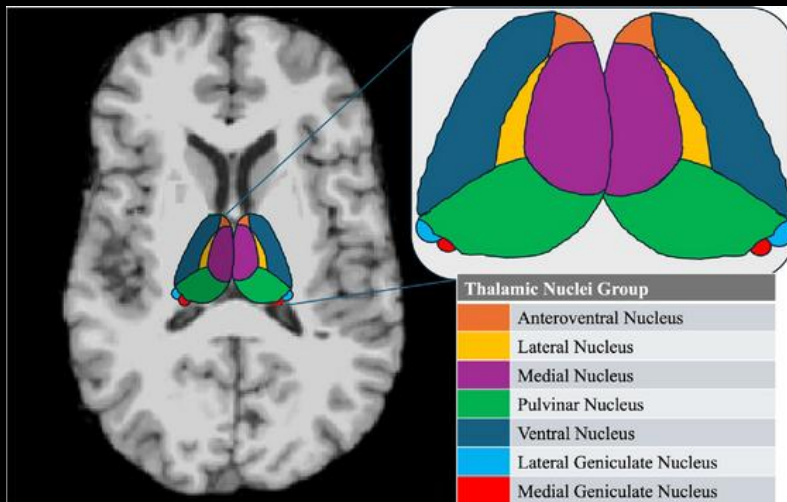
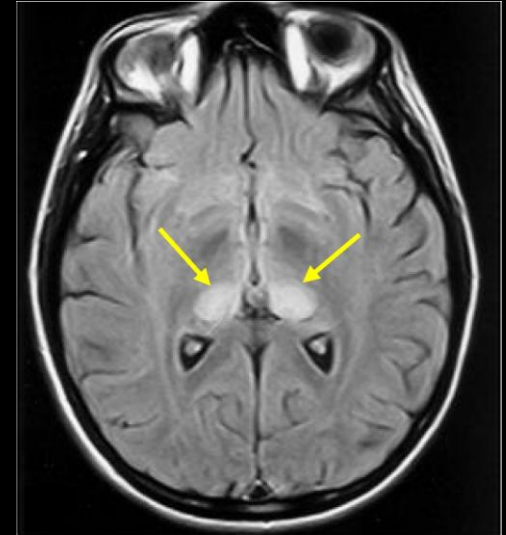
- Rapidly progressing, fatal, potentially transmissible dementia caused by prion.
- sCJD : Spontaneous PrP^c → PrP^{Sc} or somatic mutation
- Familial CJD (fCJD): Mutations in PRNP
- Iatrogenic CJD: Infection from prion-containing material
 - Surgical instruments, dura mater grafts
 - Cadaveric corneal transplants, human growth hormone preparations
- vCJD: Bovine spongiform encephalopathy in cattle is transmitted to humans through infected beef
 - Primarily present in UK
 - a.k.a. new variant CJD (nvCJD)

Imaging

- Best imaging clue: Progressive T2 hyperintensity of basal ganglia (BG), thalamus, and cerebral cortex
- Predominantly gray matter (GM): Caudate and putamen > globus pallidus (GP)
 - **Thalamus: Common in variant CJD (vCJD)**
 - Cerebral cortex: Frontal, parietal, and temporal
- Heidenhain variant: Occipital lobe
- 2 signs seen in 90% of vCJD but can also occur in sporadic CJD (sCJD)
 - "Pulvinar" sign: Symmetric T2 hyperintensity of pulvinar of thalamus
 - "Hockey stick" sign: Symmetric pulvinar and dorsomedial thalamic nuclear hyperintensity
- Best imaging tool: MR with DWI

Pulvinar sign radiology

- Variant Creutzfeldt-Jakob disease
- Fabry disease: the
 - Hyperintense signal is seen on T1 rather than T2
 - Can have late adult onset (< 30)
 - Normally kids
- Acute disseminated encephalomyelitis (ADEM)
- Status epilepticus
- Bilateral thalamic infarcts

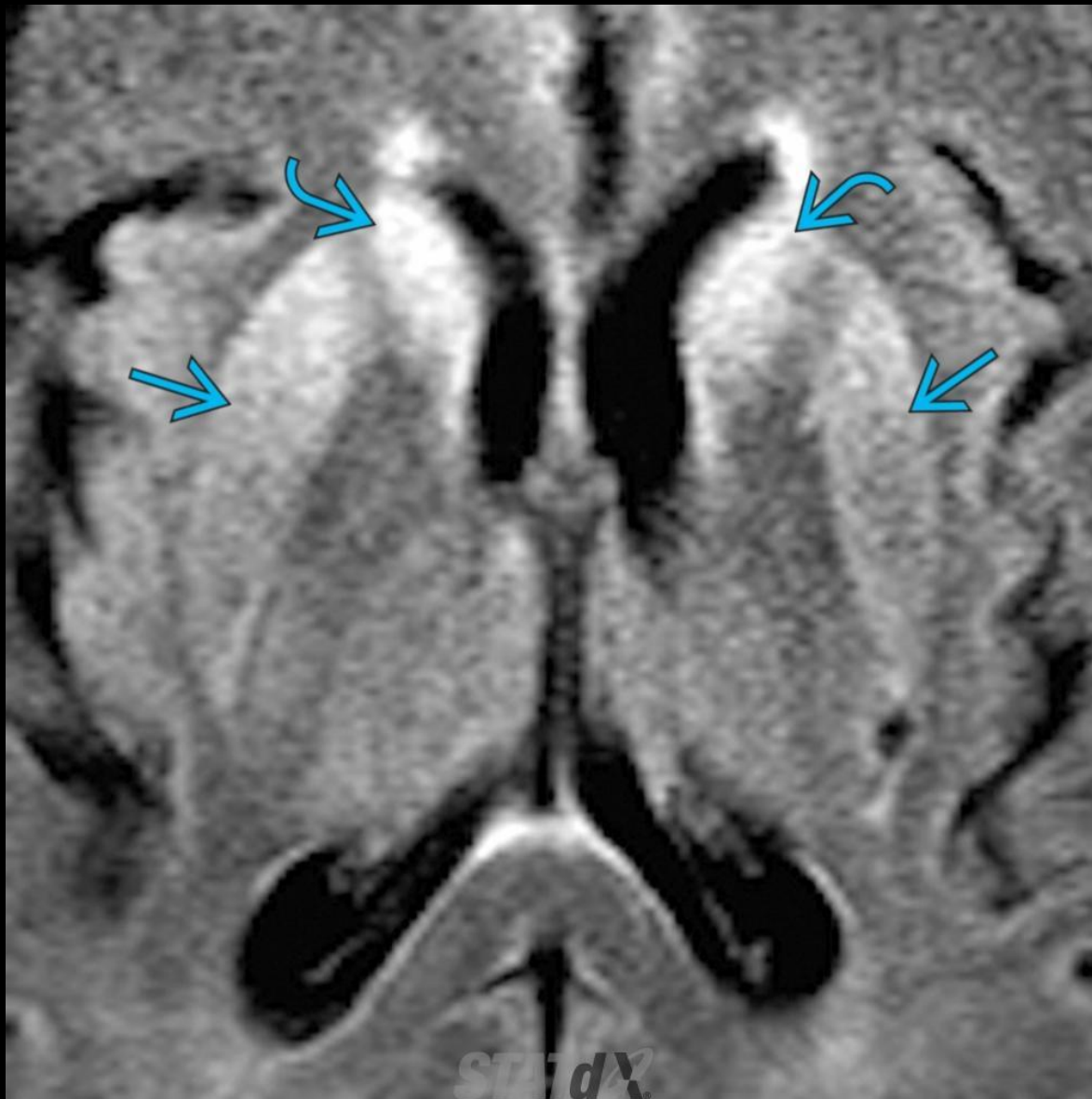


- Symmetrical dorsomedial thalamic and pulvinar hyperintense signal bilaterally, producing the “hockey-stick” sign (yellow)
- Hyperintense signal in the cortex, bilateral caudate (red arrows),
- Right putamen (blue arrow)
- Right globus pallidus (green arrow)

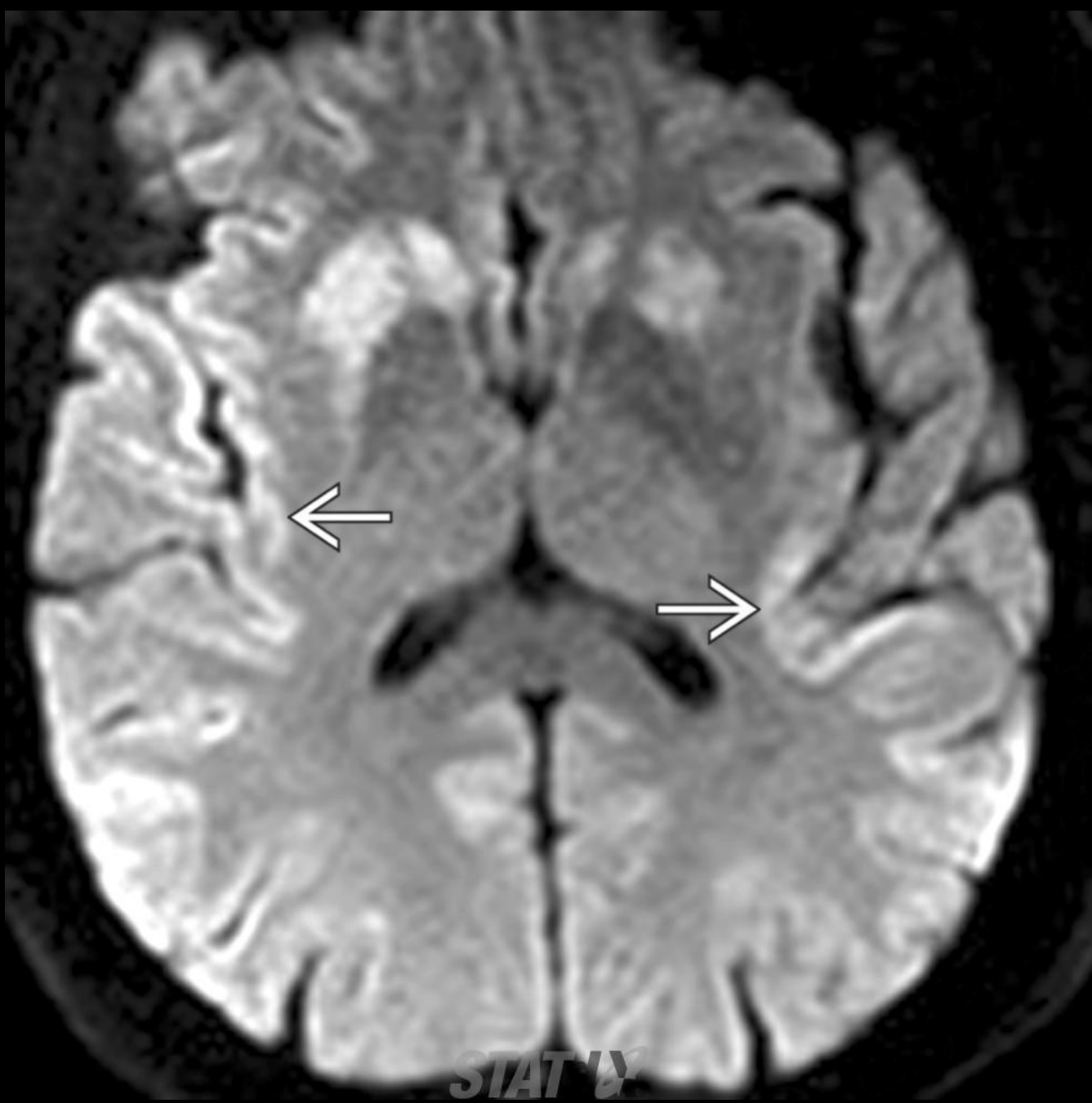
Variant CJD

CJD

- Classic CJD
 - Mostly sporadic, with smaller hereditary and iatrogenic subsets; it is not linked to food exposure in most cases.
- vCJD
 - Acquired form strongly linked to bovine spongiform encephalopathy (BSE, “mad cow disease”) via consumption of contaminated beef and, rarely, via blood transfusion
- Imaging, Pathology, and Tissue Involvement
 - **Classic CJD** - shows **cortical ribboning** and basal ganglia hyperintensity on MRI and has characteristic periodic sharp wave complexes on EEG; lymphoid tissue typically does not show abundant prion deposition.
 - **vCJD** characteristically shows the **pulvinar sign** (bilateral posterior thalamic hyperintensity) on MRI and demonstrates florid amyloid plaques with abundant prion deposition in brain and lymphoid tissues (tonsil, spleen).



Axial FLAIR MR in the same patient shows corresponding high signal in the caudate nuclei (cyan curved arrow) and putamina (cyan solid arrow). Creutzfeldt-Jakob disease (CJD) is a rapidly progressing, fatal, neurodegenerative disorder caused by a prion. MR with DWI is the imaging procedure of choice.



Axial DWI MR shows asymmetric diffusion restriction in the caudate nuclei and putamen. Involvement of the anterior more than the posterior putamen is typical of CJD. There is also asymmetric hyperintensity in the frontal and temporal lobe cortical ribbons (white solid arrow), typical of sCJD. (Courtesy, N. F. Ho, MD.)

Creutzfeldt-Jakob disease

“DDX: Acute infarct”

