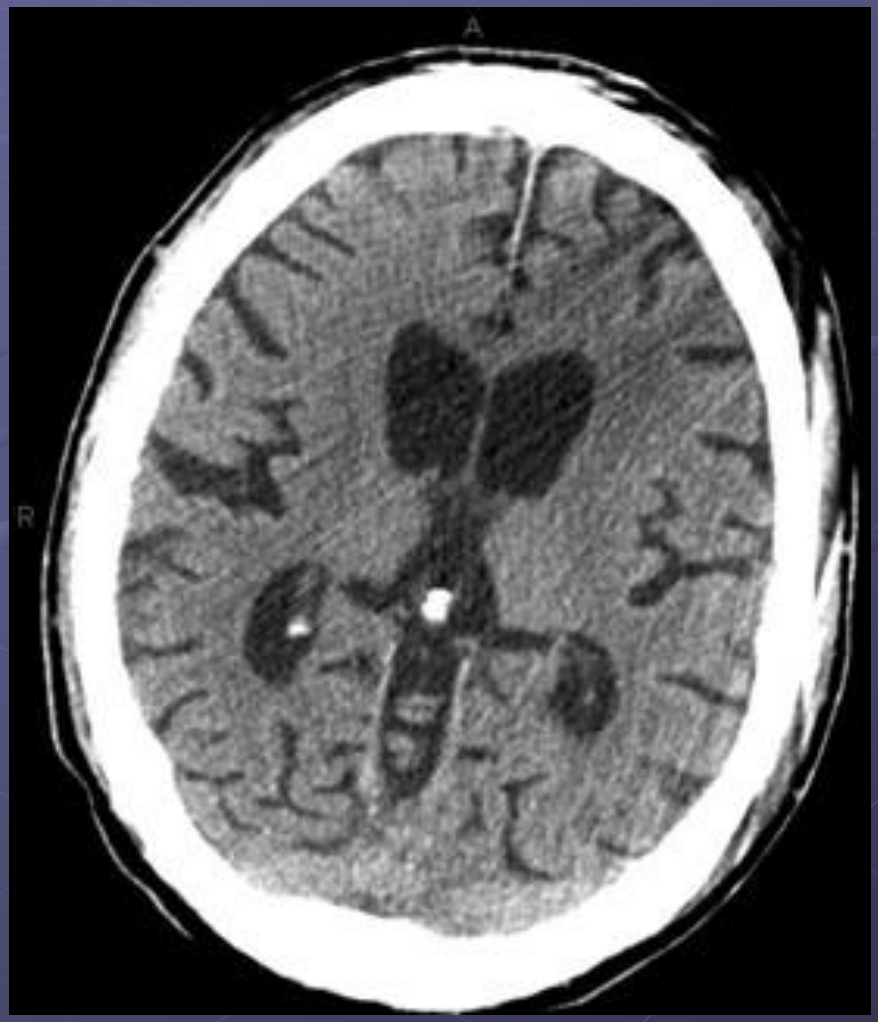
Huntington disease

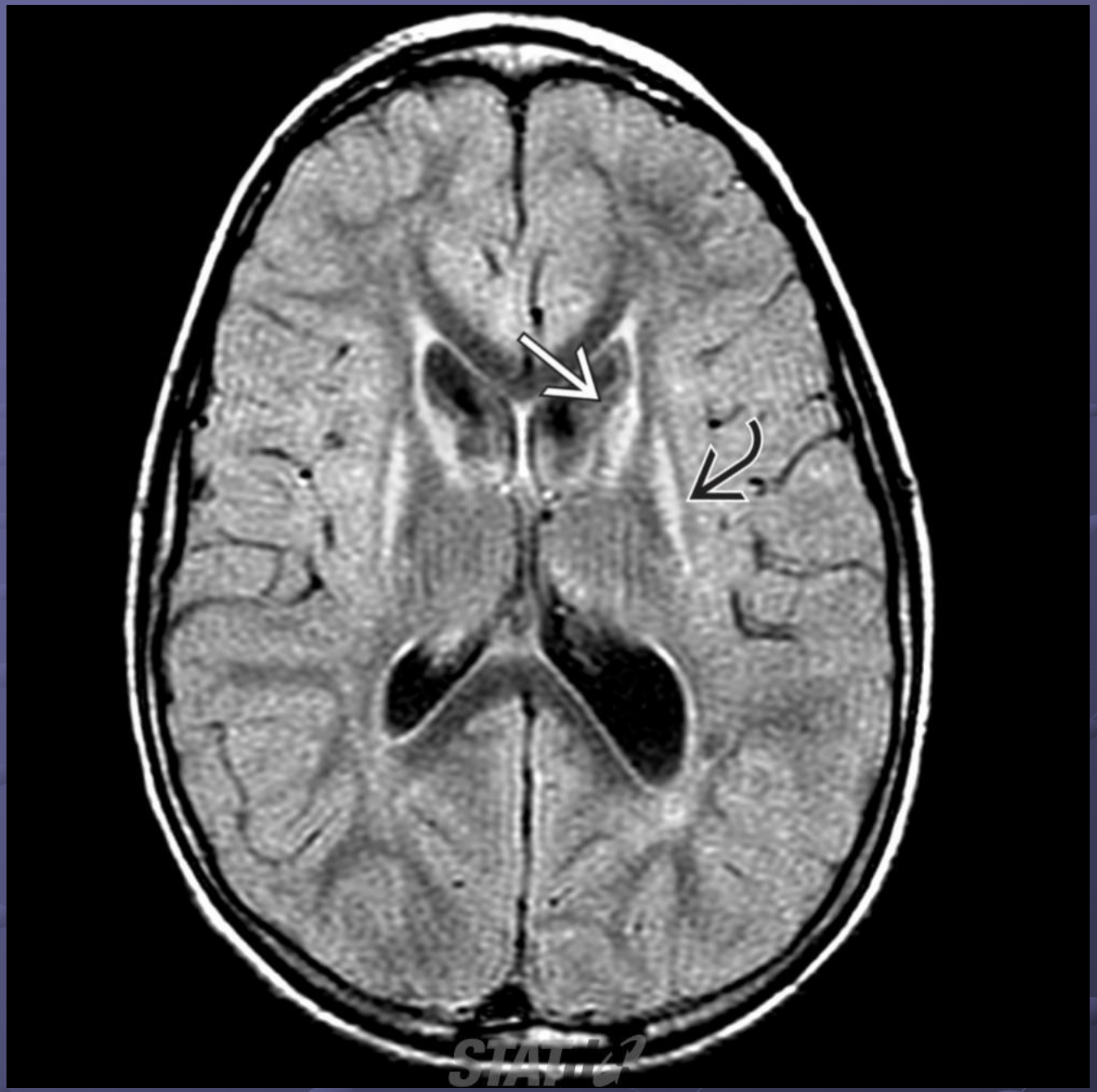
- Autosomal dominant with complete penetrance
- neurodegenerative disease
- Caused by a loss of GABAergic neurons of the basal ganglia, especially atrophy of the caudate nucleus and putamen.
- Characterized by progressive unintentional choreoathetoid movements, subcortical type dementia, behavioural changes, and psychosis which starts in midlife.
- Juvenile form

Imaging

- Diffuse cerebral atrophy
- Atrophy of caudate nucleus → frontal horns enlarged
- ↑ CC:IT ratio (bicaudate ratio)
  - Shrinkage of caudate nucleus (CN) and ↑ intercaudate distance (CC)
  - Increased intercaudate distance (CC) between medial aspects of CN
  - Most specific & sensitive measure for HD
- Hyperintense signal in CN, putamina in juvenile HD
- ↓ FDG uptake in BG before any detectable atrophy
- ± frontal lobe hypometabolism

Huntington disease





Axial FLAIR MR in an 8 year old with dysarthria, rigidity, and a family history of HD demonstrates a combination of volume loss and ↑ signal intensity of the caudate heads (white solid arrow) & putamina (black curved arrow).