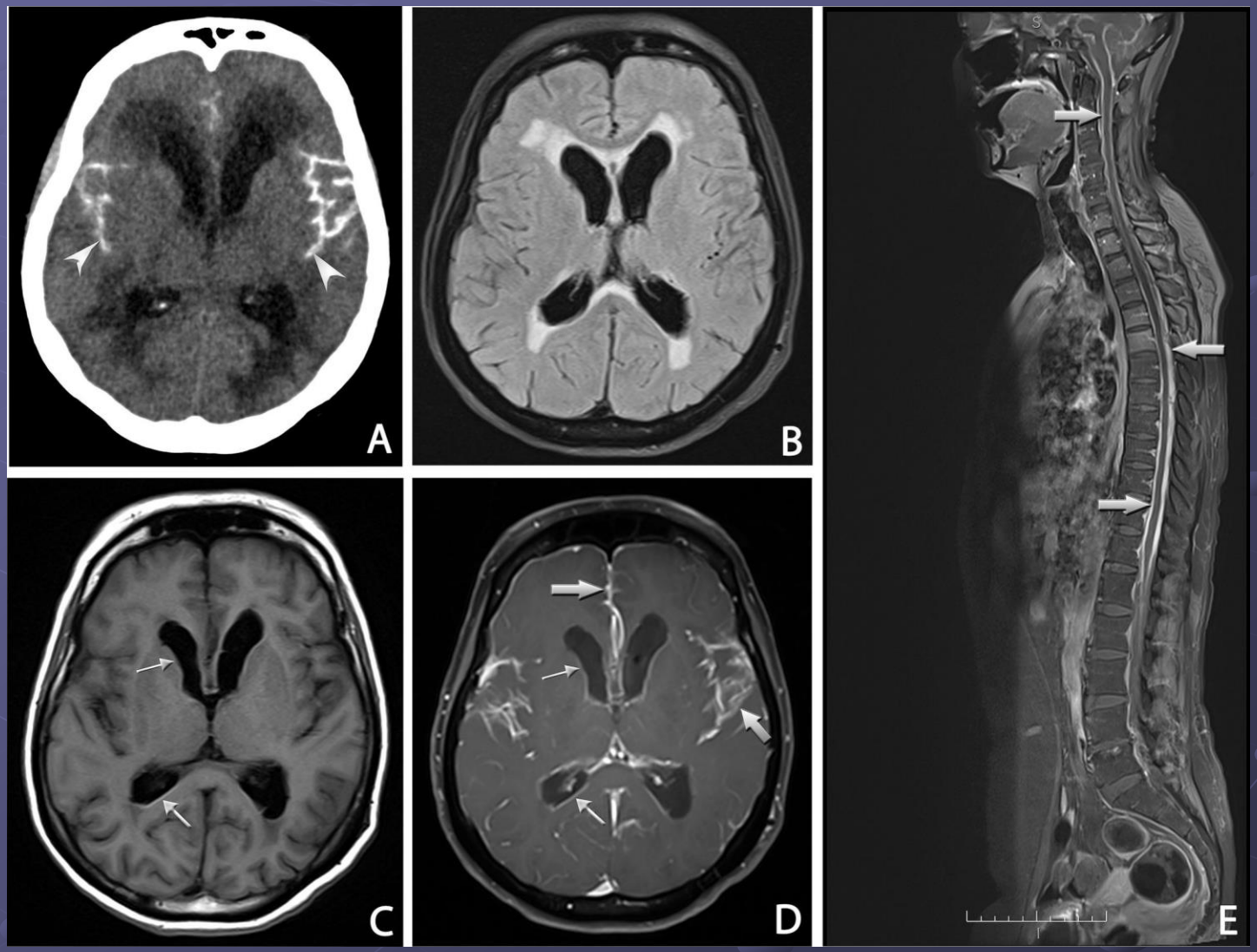
Hereditary transthyretin amyloidosis

- Transthyretin (ATTR) amyloidosis
- Rare autosomal dominant disorder in which mutations in TTR lead to the accumulation of misfolded amyloid fibrils in a variety of organs including peripheral nerves, the heart, the eye, the kidney, and the autonomic and central nervous system.
- The most common clinical manifestations include familial amyloid polyneuropathy and cardiomyopathy.
- Leptomeningeal amyloidosis represents the least common phenotype.
- It is characterized by the deposition of abnormal transthyretin amyloid aggregates in the pial and arachnoid membranes as well as the subarachnoid vessels.
- Clinical manifestations vary, though progressive neurological and cognitive decline are the most common symptoms.

TTR associated leptomeningeal amyloidosis



Leptomeningeal amyloidosis

- The imaging reveals diffuse sulcal calcifications and extensive leptomeningeal enhancement, alongside ventricular dilatation with periventricular hyperintensities. These distinctive findings strongly suggest leptomeningeal amyloidosis.

Leptomeningeal amyloidosis with SAH