

# A case with expanding white matter lesions

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A woman in her early 40s presented with the progression of spastic quadriparesis, dysarthria and dysphagia from 3 years ago.

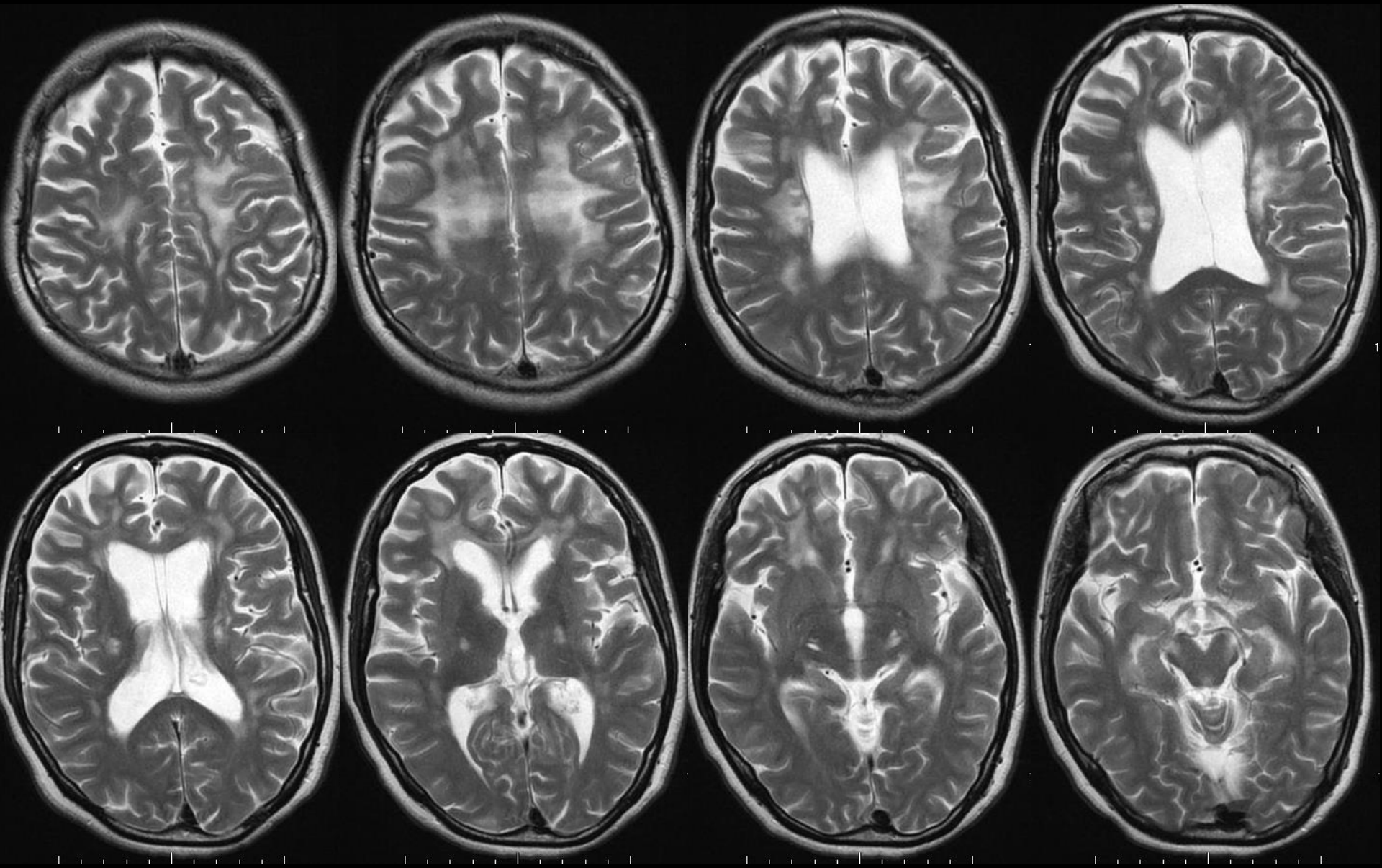
Her symptoms didn't improve after steroid therapy.

Results of the blood and CSF tests were normal.

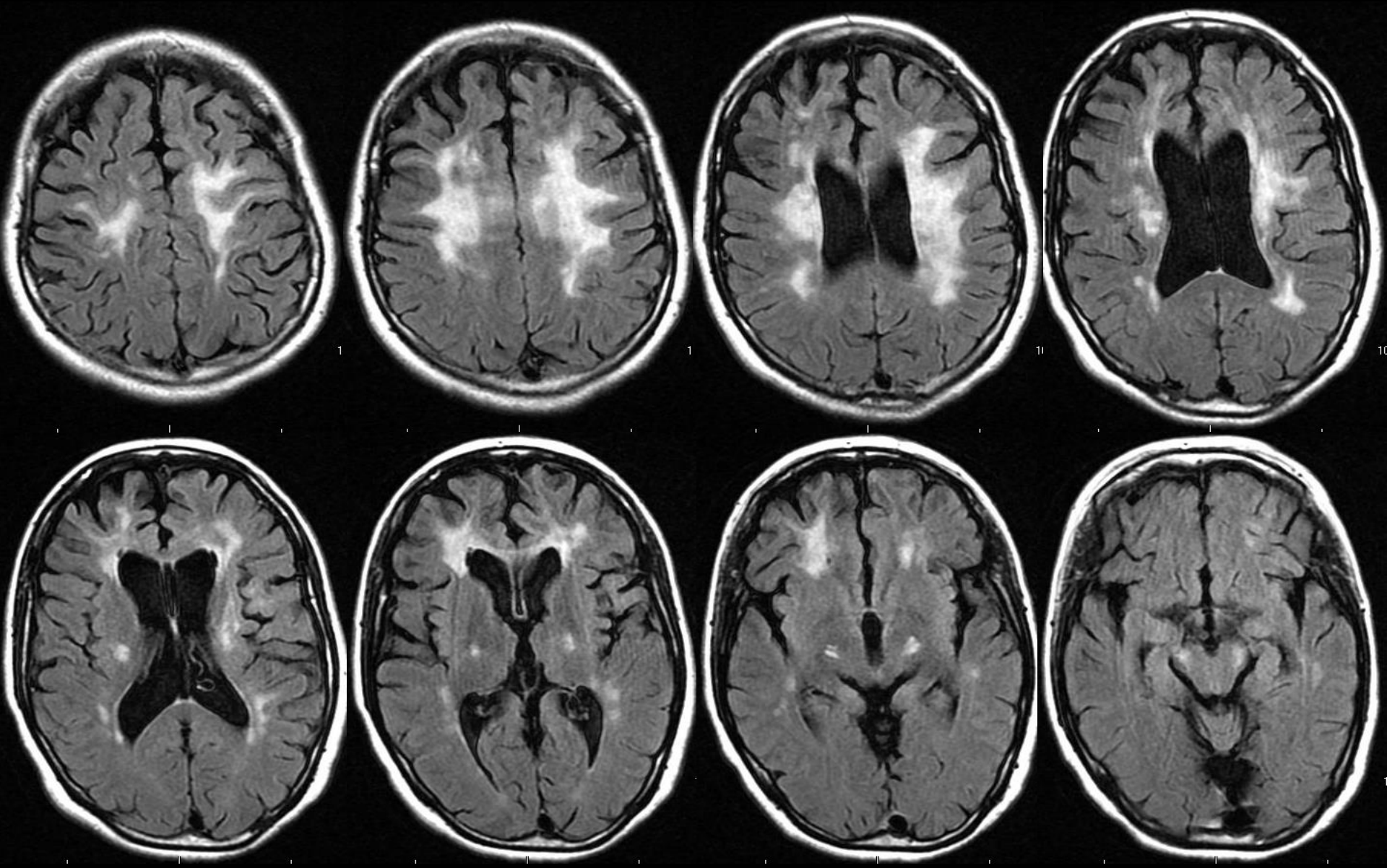
Past history: no significant.

Family history: no special findings.

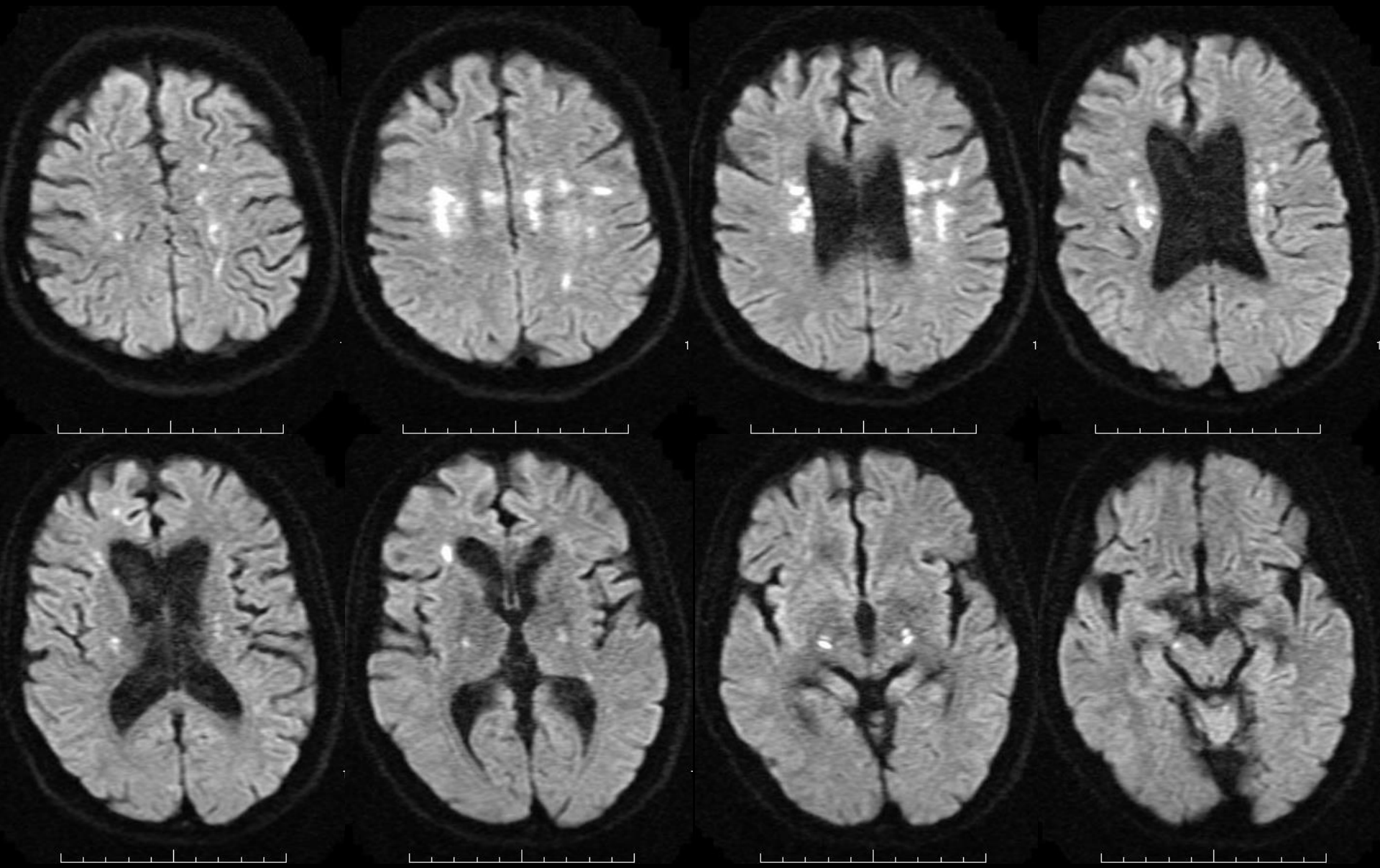
T2WI



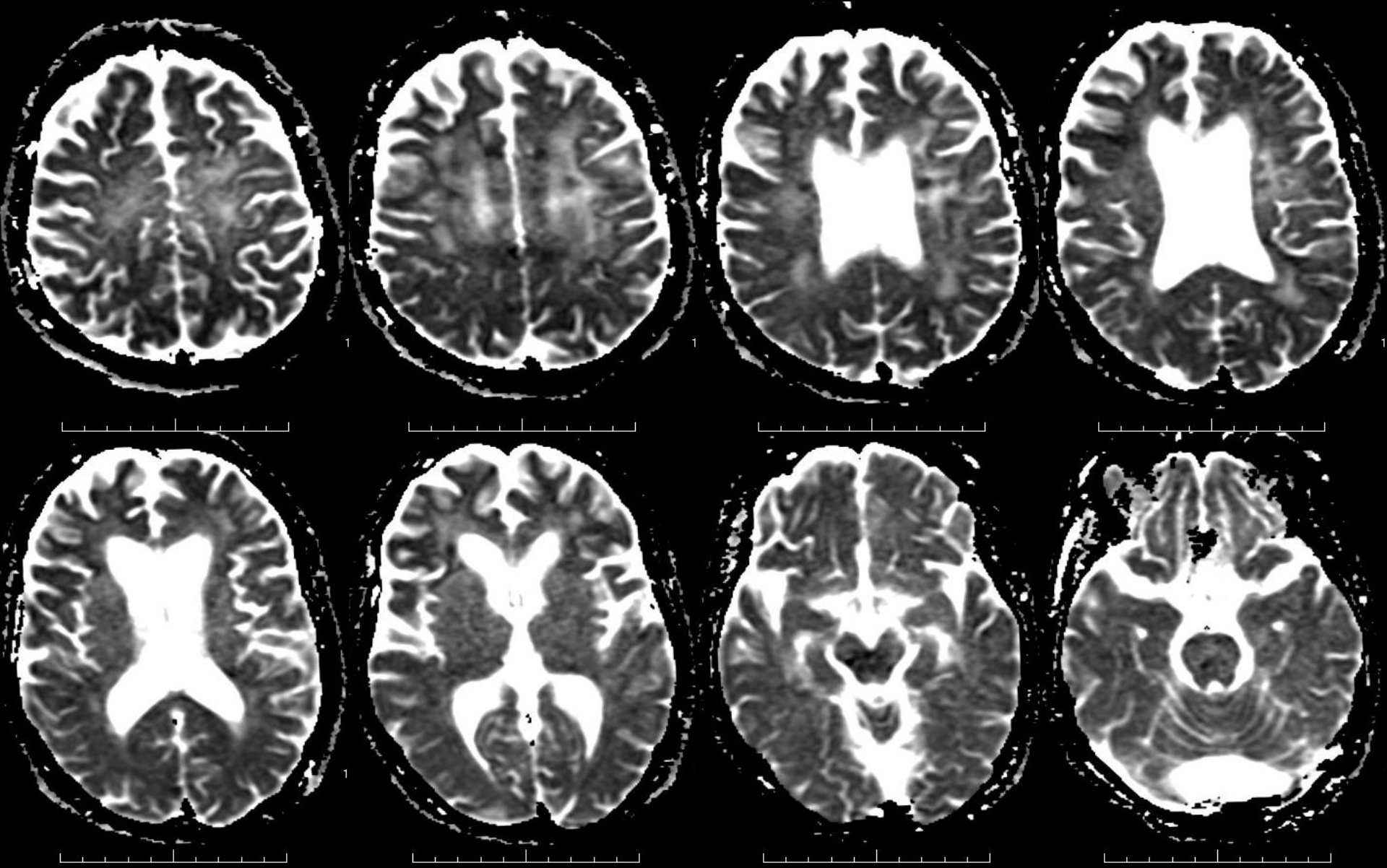
# FLAIR



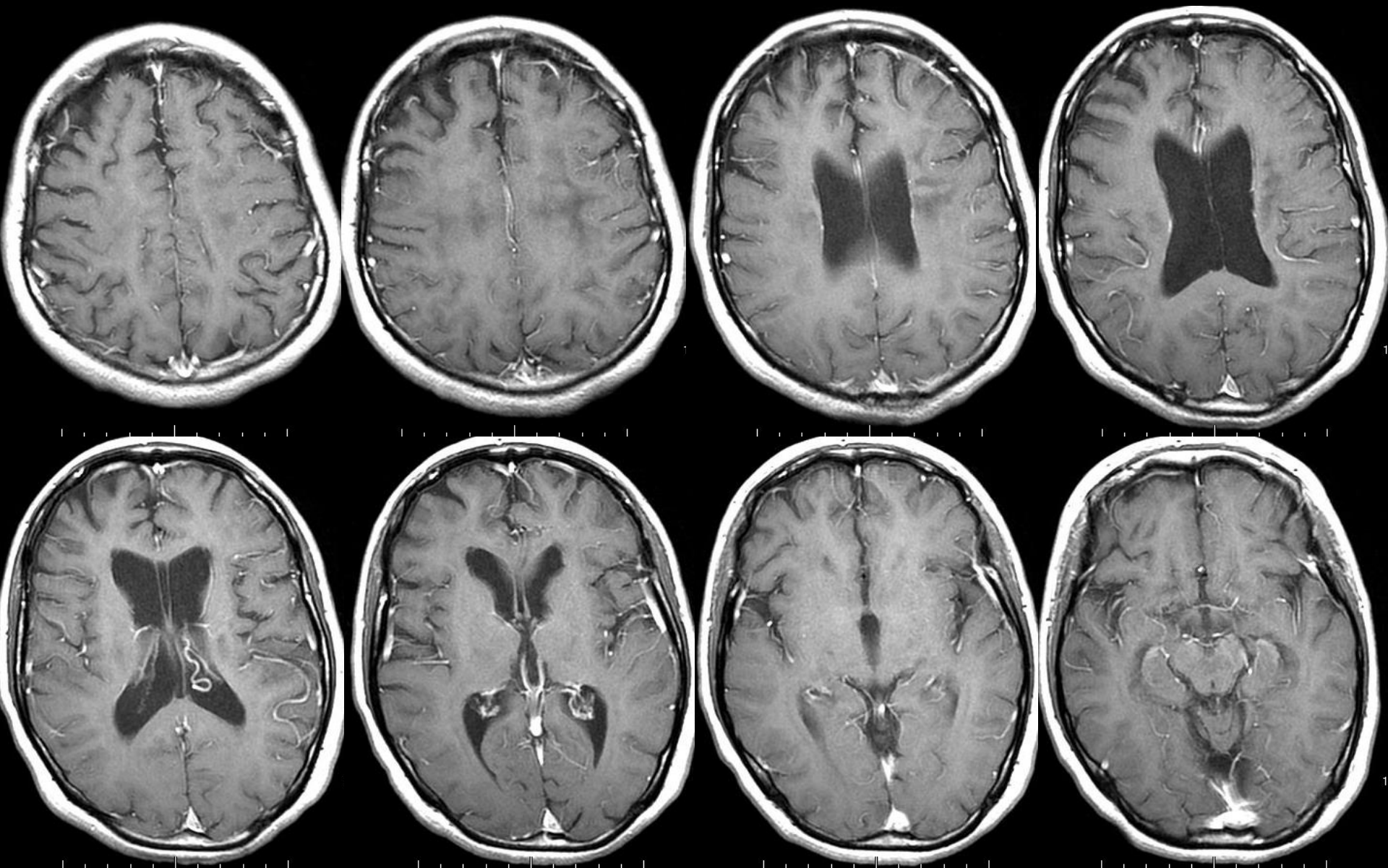
# DWI



ADC

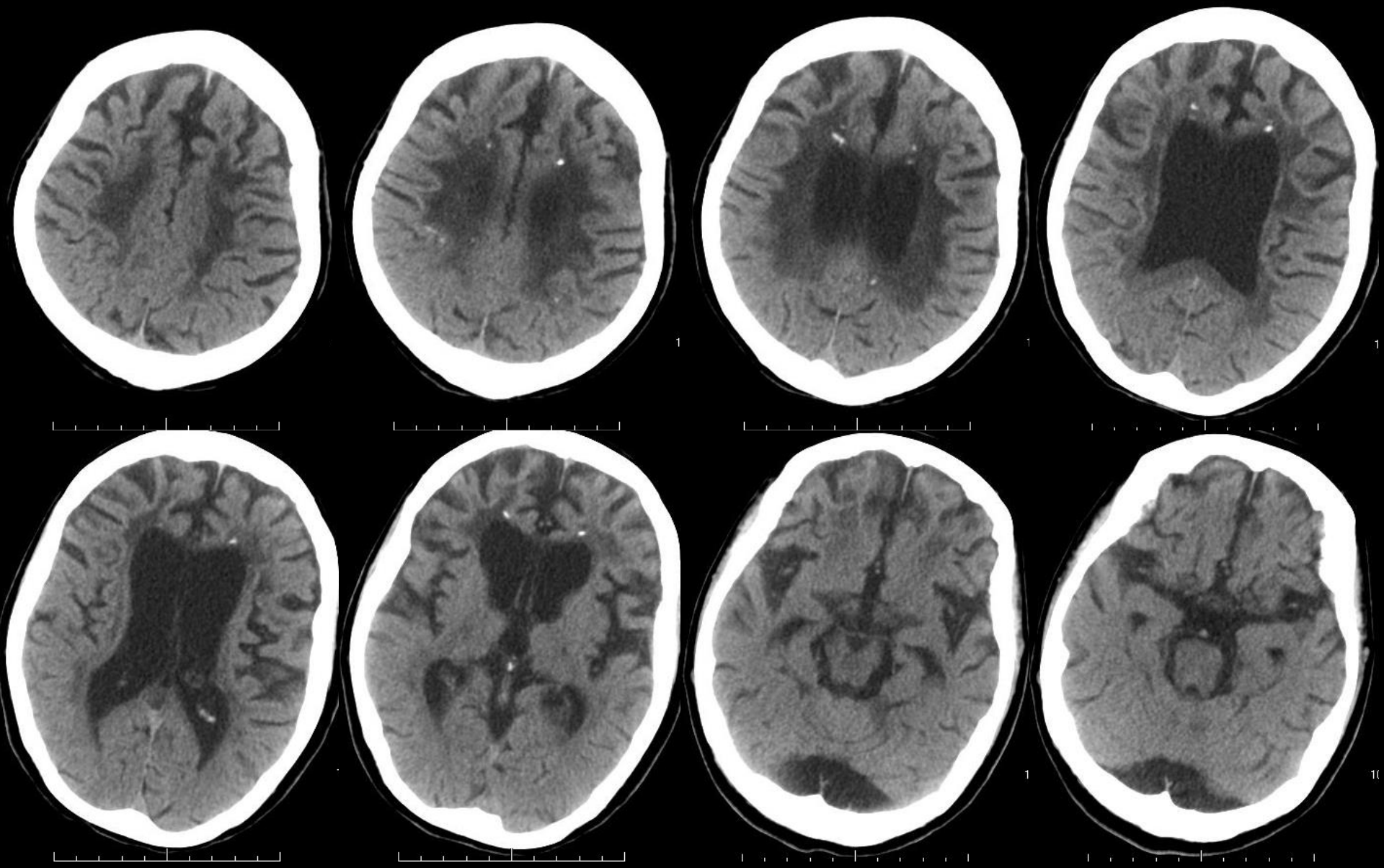


# Gd-T1WI



2 years later

CT



3  
years ago

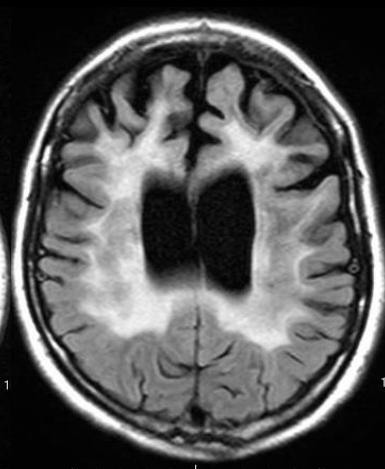
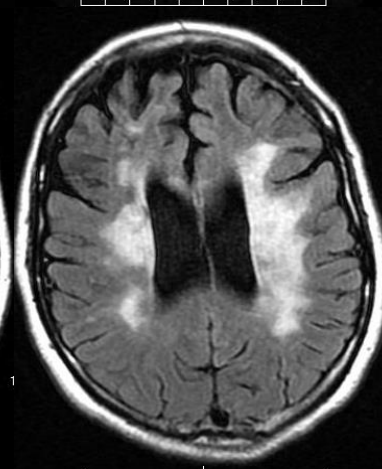
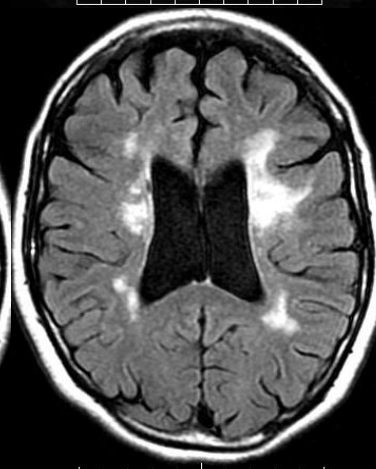
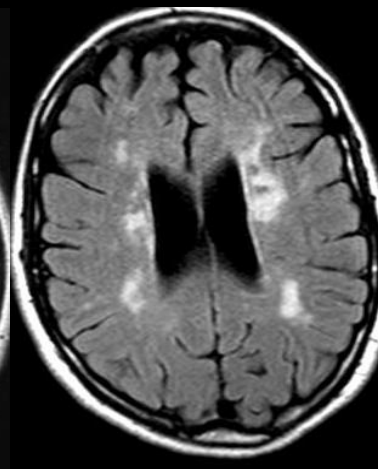
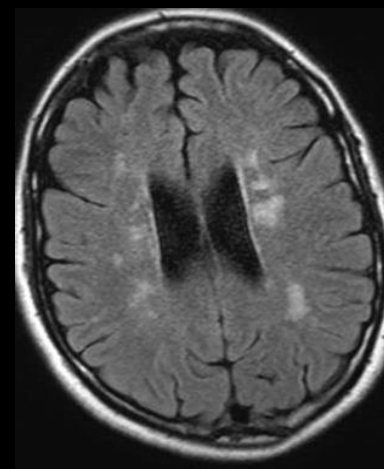
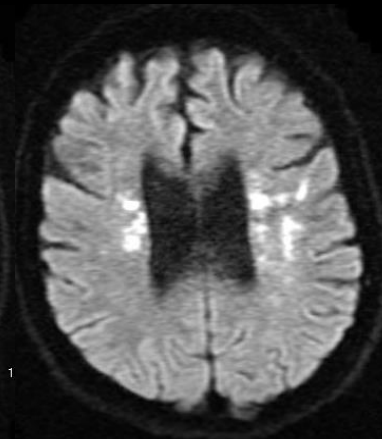
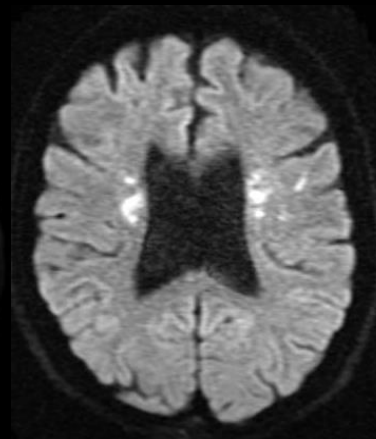
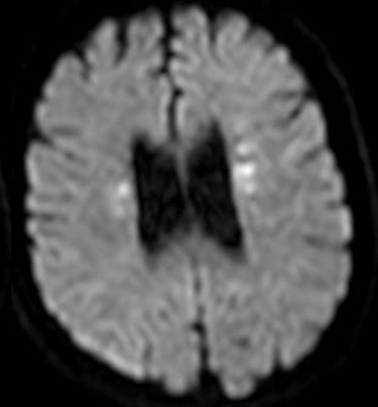
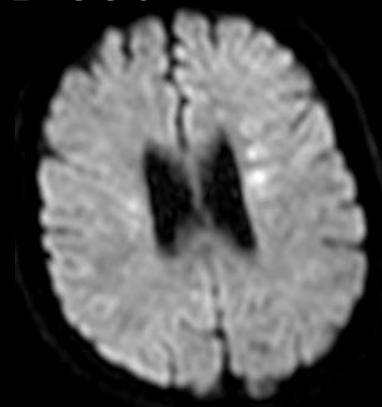
2  
years ago

1  
year ago

The large  
sample  
date

2  
years later

DWI

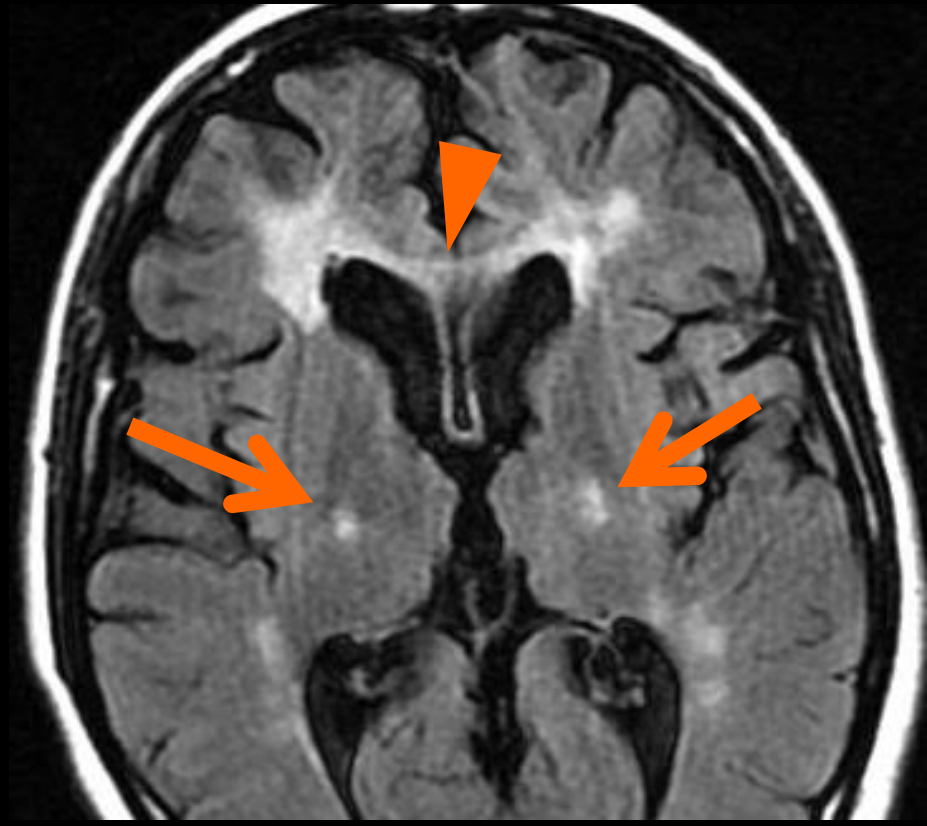
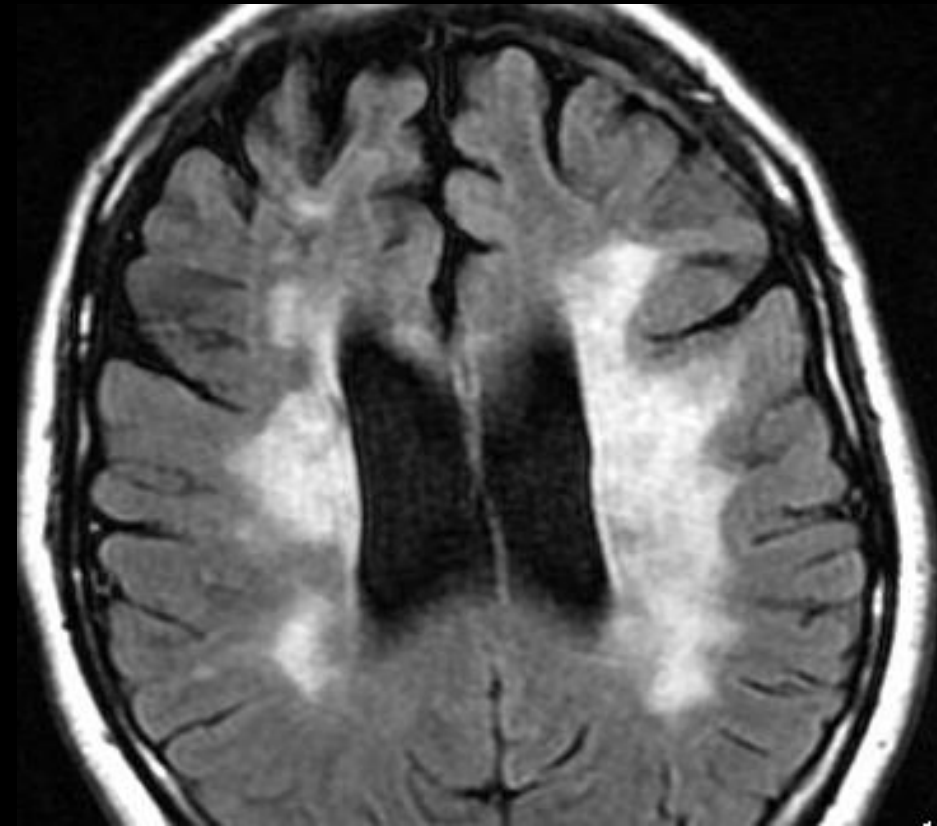


FLAIR



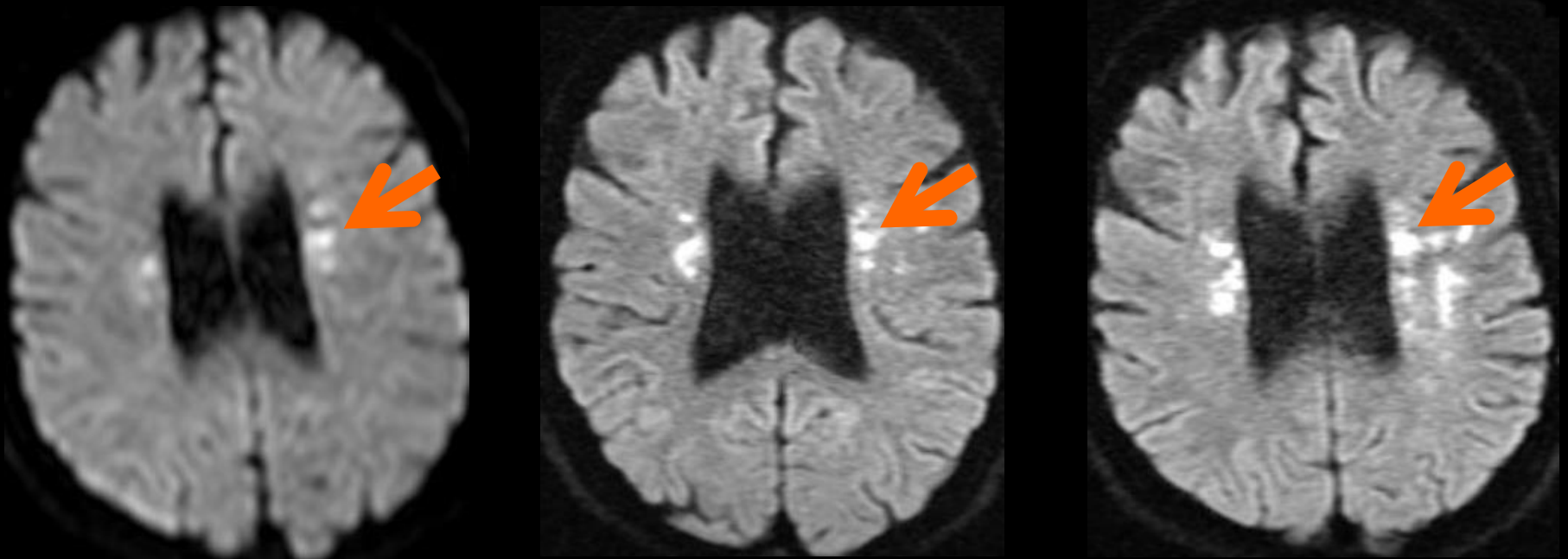
Deep white matter lesions  
sparing of U-fibers

Pyramidal tract lesions  
Callosal lesions



Frontal-predominant atrophy

Areas of bright DWI persisted and increased!



2 years

# Deep white matter + pyramidal tract lesions

MS; Multiple sclerosis

Vasculitis

MELAS; Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes

CADASIL/CARASIL; Cerebral autosomal dominant/recessive arteriopathy with subcortical infarcts and leukoencephalopathy

ALS-D; Amyotrophic lateral sclerosis with dementia

Adrenoleukodystrophy (adult-onset)

Metachromatic leukodystrophy (adult-onset)

Krabbe disease (adult-onset)

HDLS; Hereditary diffuse leukoencephalopathy with spheroids

A mutation in CSF1R was identified.

HDLS; Hereditary diffuse leukoencephalopathy  
with spheroids

She is now bedridden and mute.

# HDLS

- Autosomal dominant white matter disease (possibly sporadically)
- Mutations in **CSF1R** cause HDLS.  
(CSF1R= colony stimulating factor 1 receptor)
- 8 to 78 years (average: 38years)

## Symptoms:

- Preceding cognitive disturbance
- Spastic paresis, ataxia, dysarthria, epilepsy
- **Progressing rapidly** with death within several years

Rademakers R, et al. Nature Genetics. 2011;44:200-205

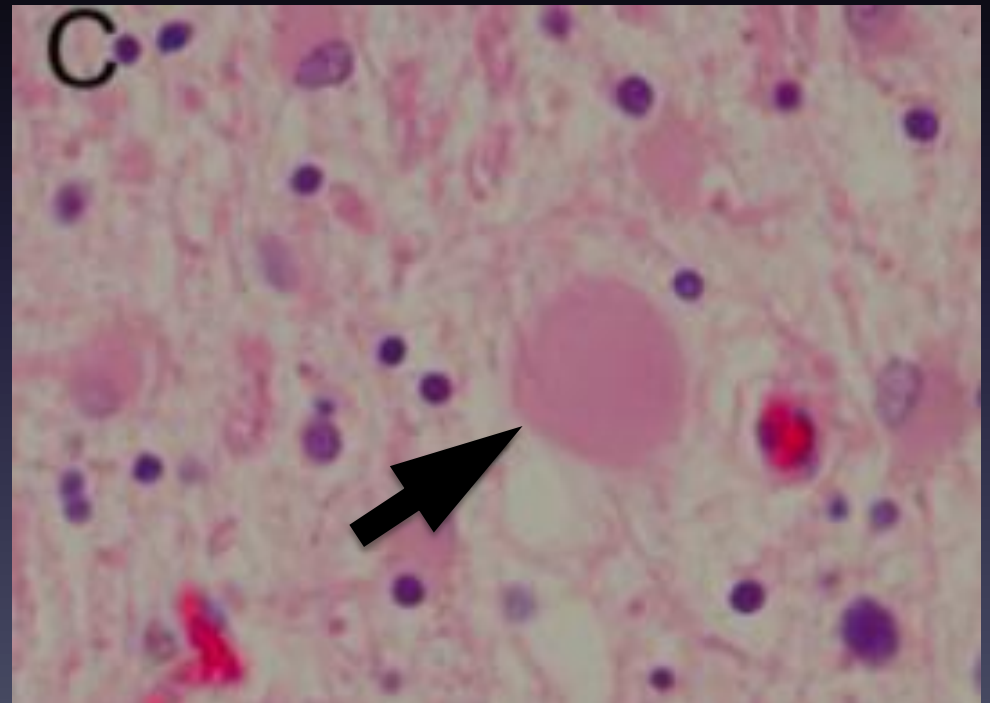
Axelsson R, et al. Acta Psychiatr Scand Suppl. 1984;314:1-65

# HDLS

## Pathological findings:

- Formation of axonal spheroids, loss of myelin sheaths, axonal destruction, gliosis; frontal predominance
- Sparing of U-fibers

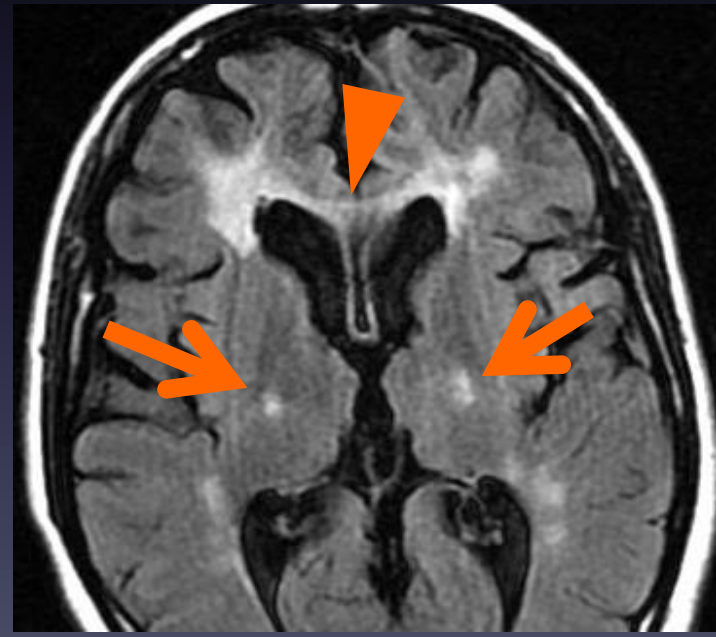
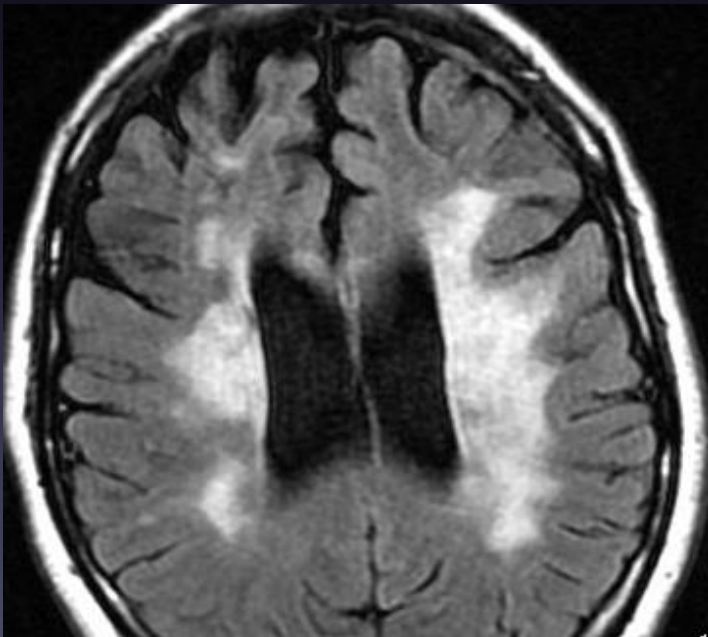
Axonal spheroids



# MRI findings in HDLS

## T2WI/FLAIR

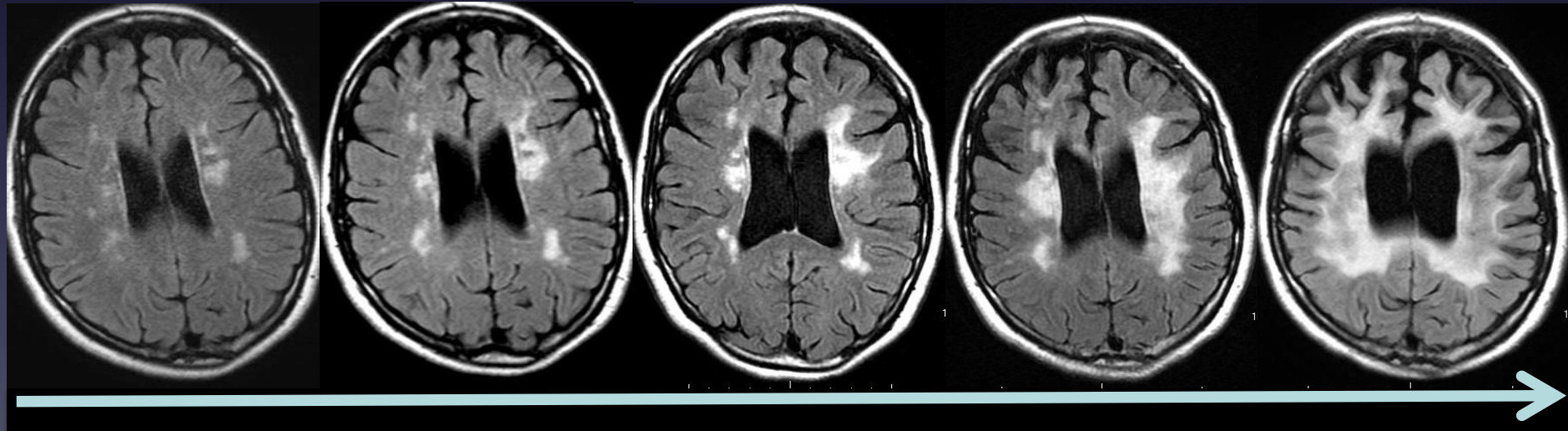
- Multifocal lesions in the **deep white matter** sparing of U-fibers; **frontal predominance**
- Involvement of **corpus callosum, pyramidal tracts**



# MRI findings in HDLS

## T2WI/FLAIR

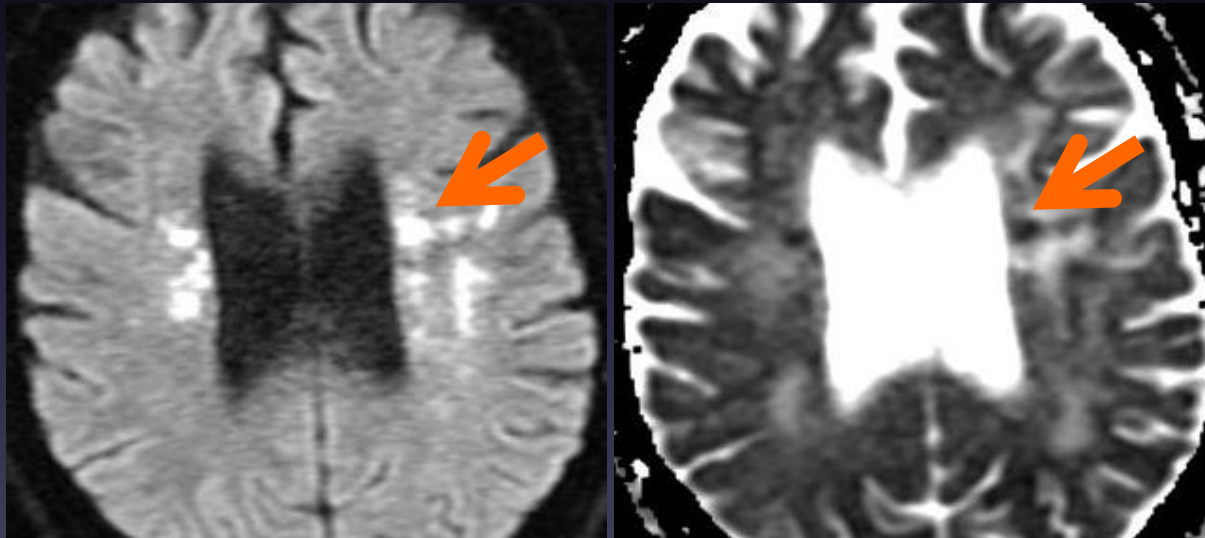
- Multifocal lesions in the **deep white matter** sparing of U-fibers; **frontal predominance**
- Involvement of **corpus callosum, pyramidal tracts**
- Confluent and diffuse with age
- Atrophy develops chronically



# MRI findings in HDLS

## DWI

- **Hyperintense foci** in the white matter lesions **persist and increase**
- Decreased ADC values in the acute phase



Mateen FJ, et al. J Neurol Neurosurg Psychiatry. 2010;81:619-622

Terasawa Y, et al. J Neurol Sci. 2013;335:213-215

吉田邦広 日磁医誌 2014;第34卷2号

# Conclusion

Persistent and increasing bright foci on DWI can be an indicator of HDLS.

