Initial Presentation of MELAS in an Adult Woman with Associated Cerebellar Atrophy

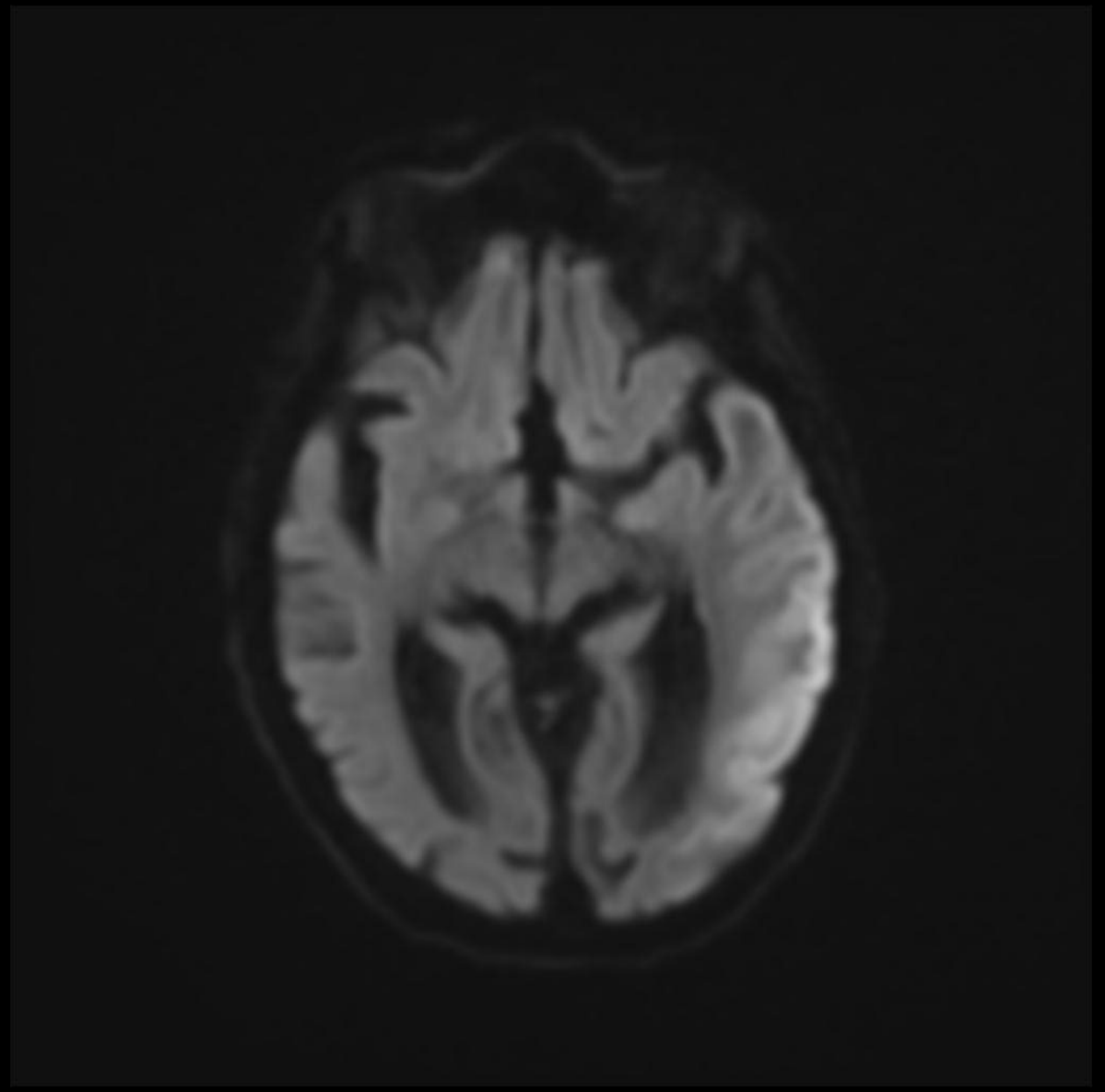
Hall Braud MS, Christopher Rouse MD, Meredith Rouse MD

Clinical Presentation

- 51-year-old female who presented to the ED with new onset seizure
- Past medical history:
  - Sensorineural hearing loss, T2DM, chronic debility, short stature, and recent cognitive decline
- Physical exam:
  - Vital signs within normal limits
  - Post-ictal appearance
  - Notable for aphasia and right sided hemiparesis

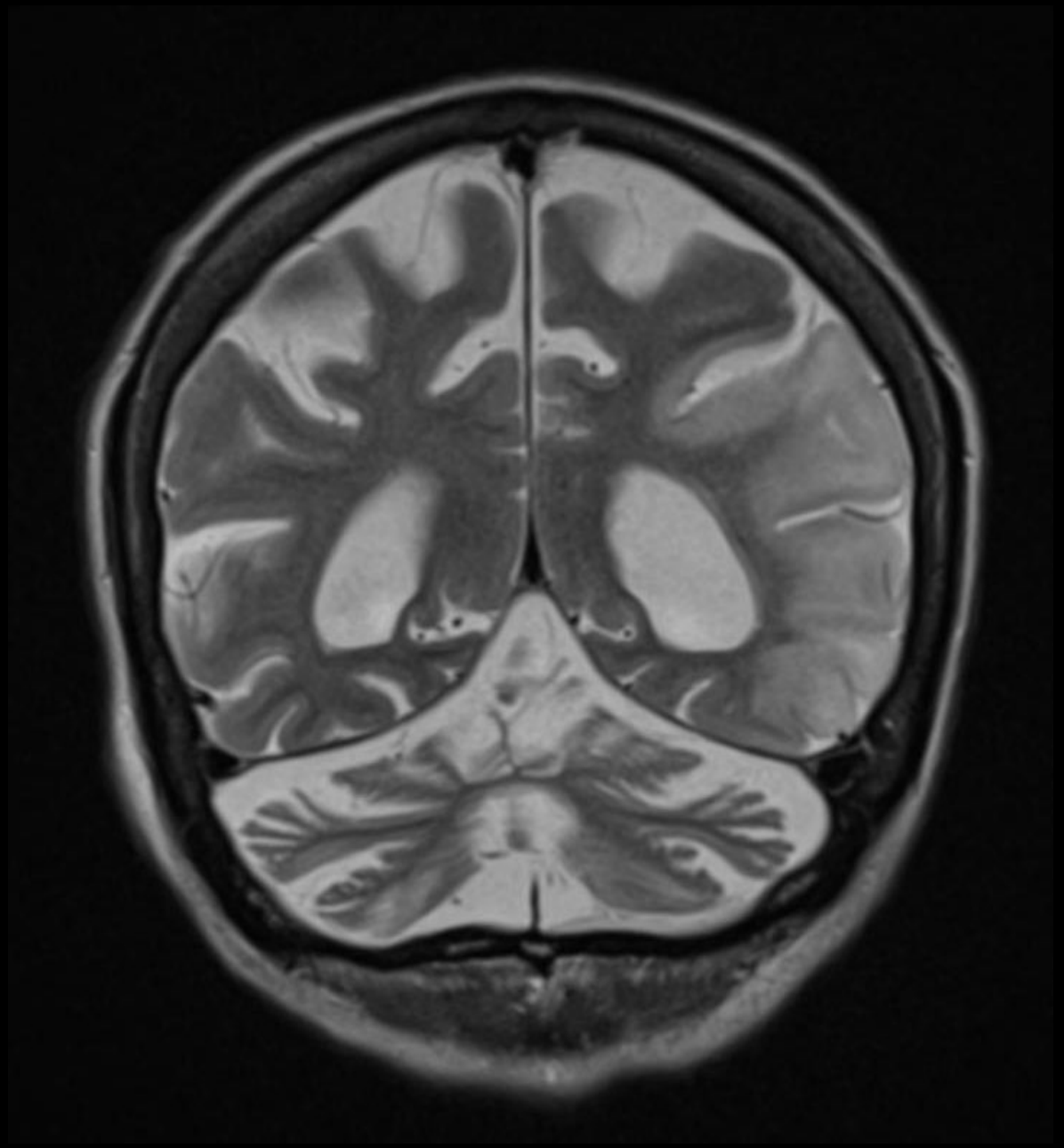
Imaging Discussion

- CT Head without contrast
  - Significant for indistinct hypodensity of left temporal lobe concerning for stroke-like lesion of the left MCA distribution
- MRI brain (T2 FLAIR, right)
  - Gyriform diffusion restriction and cortical edema of temporparietal and the left anterior occipital lobes



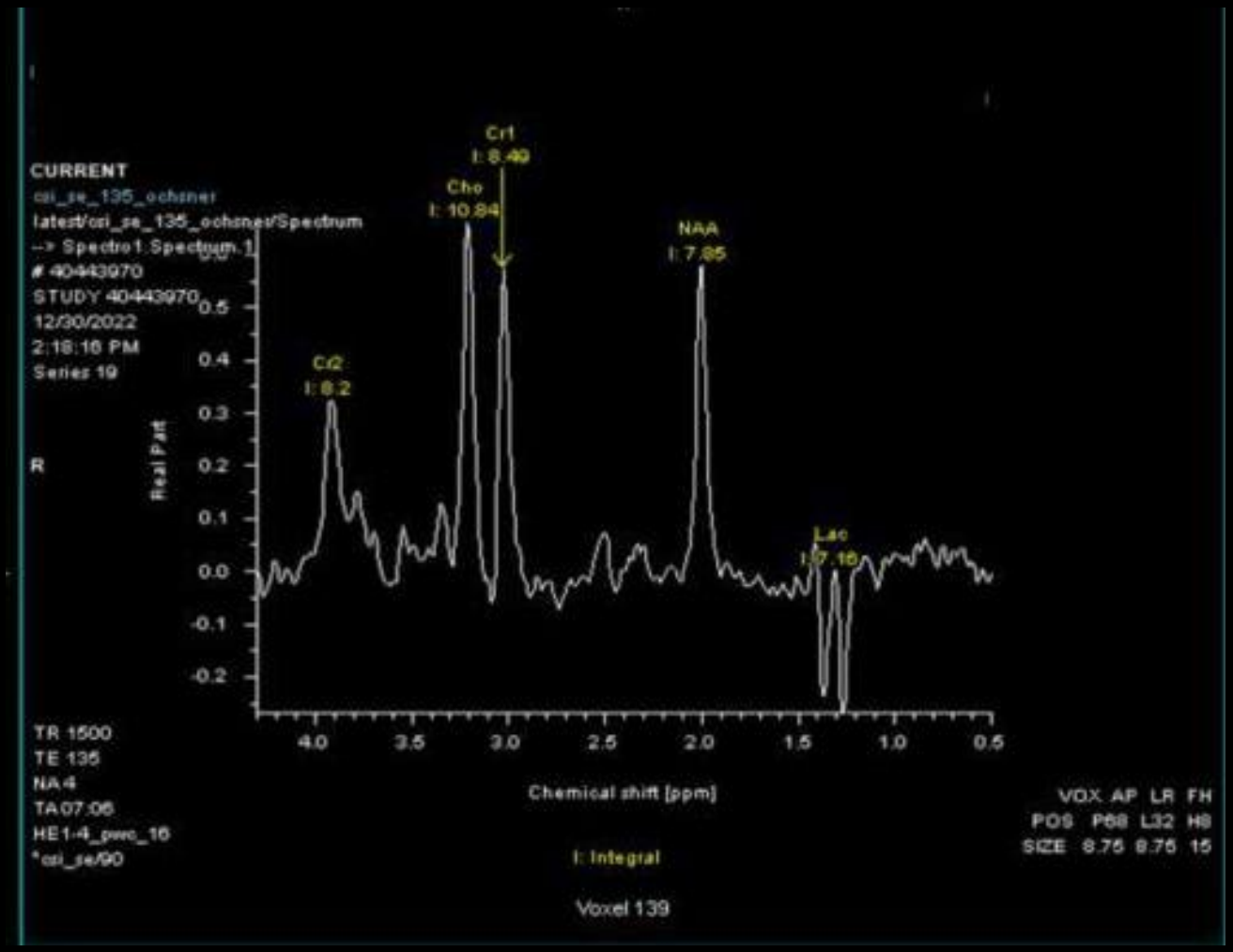
Imaging Discussion

- MRI brain (T2, right)
  - T2 hyperintensity of the left temporoparietal lobe
  - Note additional finding of cerebellar atrophy

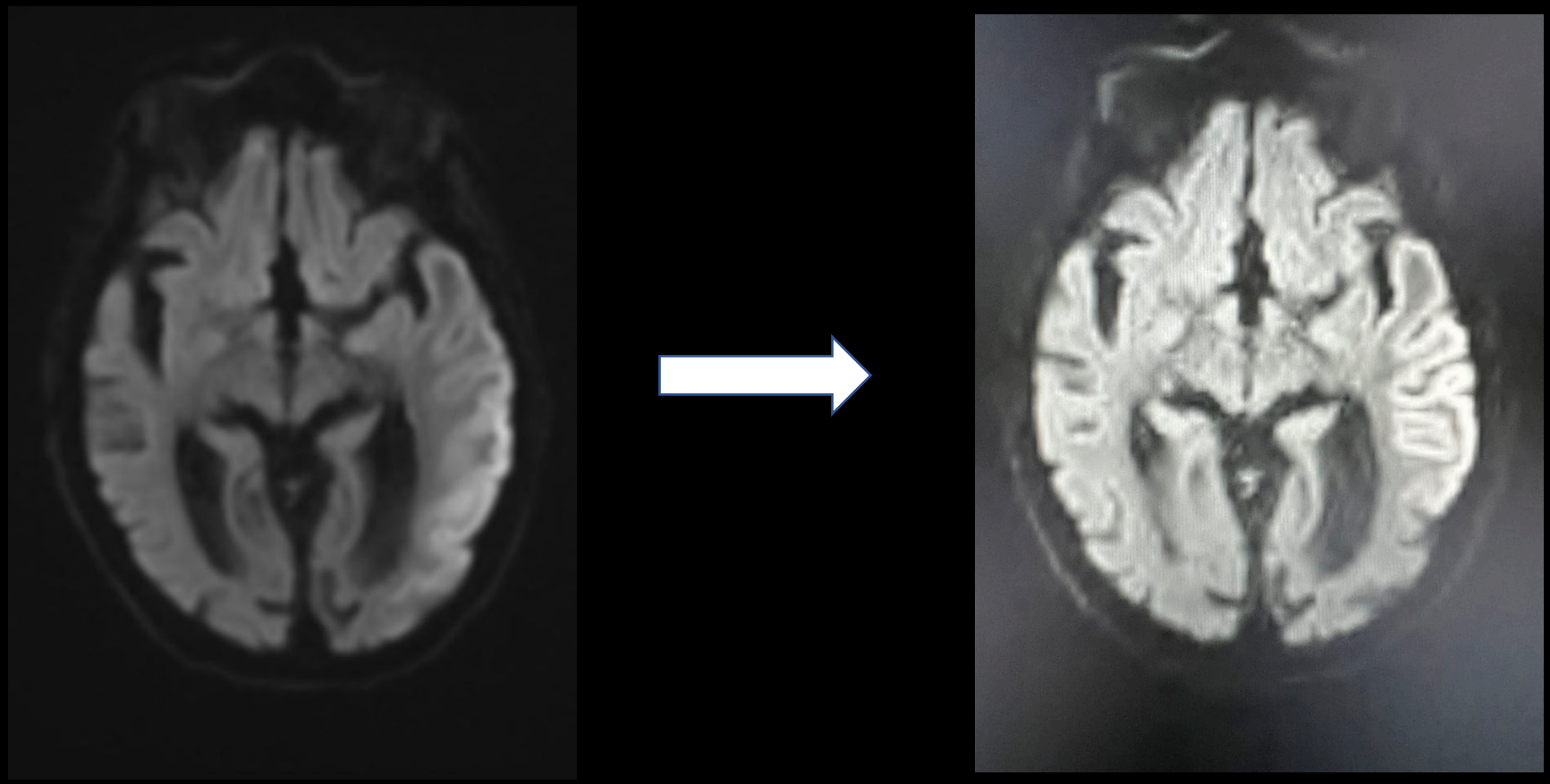


Imaging Discussion

- MR Spectroscopy
  - Focus of elevated lactate peak within the left parietal subcortical white matter concerning for anaerobic metabolism



Imaging Discussion



- Imaging taken in the weeks following the episode (right image) showed atypical progression of the lesions not consistent with ischemic stroke
  - Persistent cortical restricted diffusion and migration of lesion

Management

- Started on Keppra for seizure prophylaxis
- Genetic testing performed
  - Notable for multiple mitochondrial DNA abnormalities and in particular a pathogenic variant of the MT-TL1 gene (m.3424A>G) with 10% heteroplasmy
- This particular mutation is highly associated with mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes (MELAS)

Outcome

- Given the constellation of findings including past medical history of sensorineural hearing loss, stroke-like lesions and cerebellar atrophy on brain MRI, spectroscopy showing a lactate peak in the affected regions, and a definitive mitochondrial DNA mutation, the patient was diagnosed with MELAS

Take Home Points

- MELAS typically presents in individuals before age 20 following normal development but may present as first time or recurrent strokes in adult patients

- The stroke-like episodes seen with MELAS are distinct from traditional stroke and appear differently on brain MRI

Take Home Points

- Cerebellar atrophy is a finding associated with MELAS and can be used to distinguish the disorder from ischemic stroke.

- Elevated lactate peak on MR spectroscopy is another finding that can be used to distinguish MELAS from other causes of cortical restricted diffusion.