Teaching Neuro*Images*: Diffuse posterior leukoencephalopathy in MELAS without stroke-like episodes

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A 46-year-old woman with a history of seizures, migraine, diabetes mellitus, and neurosensory hearing loss presented with progressive gait unsteadiness. Her mother had the same clinical features. Examination revealed short stature, cognitive deficit, and ataxia. Brain MRI showed diffuse atrophy and posterior leukoencephalopathy (figure). Lactate levels were slightly elevated in both serum and CSF. Mitochondrial DNA analysis revealed the m.3243A>G tRNALeu(UUR) mutation that causes mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes (MELAS). Brain MRI in MELAS classically shows lesions that predominantly affect the occipital cortex and do not conform to large-vessel territories. The posterior predominance of brain lesions in our patient, although nonspecific, suggested a MELAS-associated pathology. Diffuse symmetric leukoencephalopathy in patients with the m3243A>G mutation and lacking stroke-like episodes is extremely rare and their pathophysiology is unclear.[1,2] Diabetes mellitus might also play a role in these radiologic abnormalities.

REFERENCES

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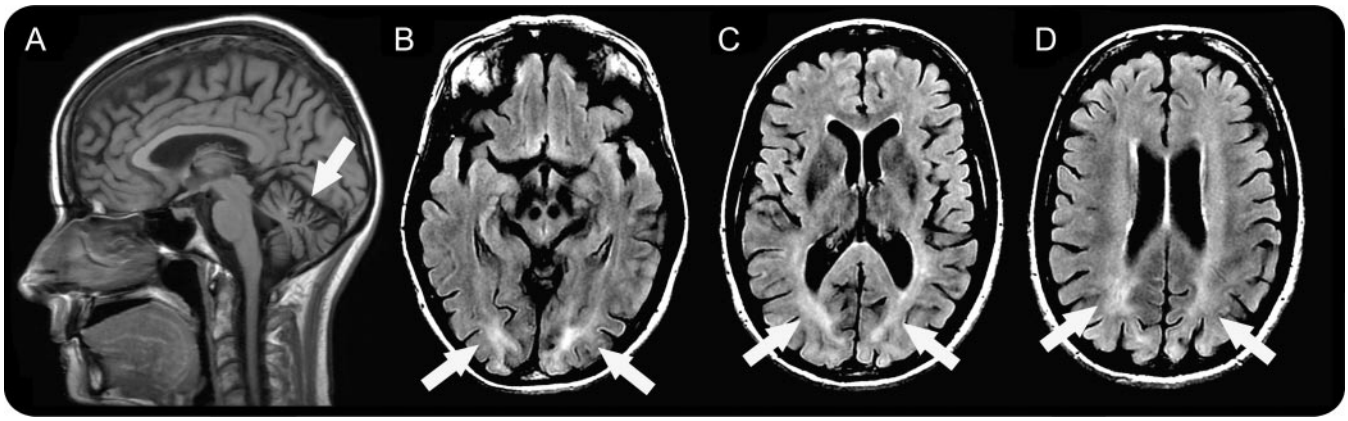
Figure Brain MRI



Brain 3-T MRI showing generalized brain atrophy with severe involvement of the cerebellar vermis on sagittal T1-weighted imaging (A) and diffuse symmetric posterior leukoencephalopathy on axial fluid-attenuated inversion recovery sequences (B–D). Diffusion-weighted imaging and apparent diffusion coefficient sequences were normal. Basal ganglia were normal and there were no intracerebral calcifications on MRI.

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