MRI pattern approach of adult-onset inherited leukoencephalopathies

Joel C. Morgenlander, MD: The importance of clinical review articles is sometimes minimized by those responsible for medical journals or those serving on Appointments, Promotion and Tenure committees. However, for those of us benefitting from reading an excellent review article and incorporating lessons learned into practice, they are a crucial source of education. A prime example is the article by Labauge et al. concerning patterns of white matter abnormalities in adult-onset leukoencephalopathies. I recommended this article within my practice.

A 72-year-old man had clinical findings including cognitive deficits, gaze-evoked nystagmus, cogwheel rigidity, distal sensory loss with diminished reflexes, intention tremor, dystaxia on heel to shin, and a shuffling, unbalanced gait. A 77-year-old man had an examination showing cognitive deficits, cogwheel rigidity, action tremor, and an ataxic gait. I had seen the patient 8 years earlier and could not make a diagnosis at that time. After reviewing both patients’ MRI scans; noticing the pattern of white matter changes, particularly in the middle cerebellar peduncle (see figure); and reviewing the article described, I sent genetic testing for fragile X-associated tremor/ataxia syndrome. Both patients tested positive for premutation repeats of 107 and 87 repeat lengths, respectively. This diagnosis was helpful in caring for the patients and had genetic counseling ramifications for their families.

Review articles of quality are a source of important education for students and clinicians. Those who take the effort to put together such articles are contributing greatly to clinical care and should be praised academically and professionally for their efforts.

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