Case 2010-12

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Clinical History
This 39-year-old woman had first symptoms of an ataxic disorder at the age of 10 (1978). She was unsteady on her feet and later developed a speech impediment. At the age of 25, a presumptive diagnosis of spinocerebellar ataxia (SCA) was made, but all genetic tests available at that time were negative (SCA 1, 2, 3, 6, and Friedreich ataxia). The family history included essential tremor in her mother and maternal grandfather. The patient married and had a normal son. A detailed neurological examination in October 2001 (age 33) revealed normal mental status; saccadic intrusions into ocular pursuit movements; ataxia; dysmetria; dysarthria; hearing loss; modest hyperreflexia; and a right Babinski sign. She faithfully made follow-up visits and received abundant support from her parents. The last neurological examination occurred in June 2007. She had progressed to intense rigidity of her extremities; dystonia; leg spasticity; and sustained ankle clonus. She was bed-bound and required a gastrostomy tube. Magnetic resonance imaging was unrevealing. After release to a long-term hospital, she contracted pneumonia and died.

Additional Information
The general autopsy revealed pulmonary congestion and an angiomyolipoma of the right kidney. The brain weighed 1321 g; the substantia nigra was pale

Material Submitted
H&E-stained section of cerebellar vermis
Virtual Slide (click here)

Points for Discussion
1. Diagnosis
2. Clinicoanatomical correlation