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CASE 2011-8

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Clinical History: The patient was a 21-year-old female with progressive neurologic deterioration. She initially presented at the age of 10 years with tremors and headaches. Her family history was remarkable for a paternal great grandfather with myasthenia gravis, a maternal great grandmother with tremors, (onset in the 7th decade), and a maternal grandfather with tremors (onset in the 6th decade). A maternal aunt was born with mental retardation. Notably, both parents reportedly had seizures in the past. At the age of 12 years, the patient developed dysarthric speech. A presumptive diagnosis of myasthenia gravis was excluded. A muscle biopsy performed in at age 18 revealed chronic neurogenic atrophy. The patient subsequently developed temperature instability, tongue fasciculations, worsening dysarthria, tremors, and cognitive decline. Three years later the patient underwent bilateral deep brain stimulator placement resulting in temporary improvement of her tremors. Her clinical condition worsened, progressing to disabling dyskinesia, severe dysphagia, and inability to walk or stand. Because of repeated complications and in agreement between the patient and her family, the gastric feeding tube was removed; death followed shortly thereafter.

Autopsy findings: The fixed brain weighed 1150 grams, not including the left frontal pole, which was removed at the referring institution prior to fixation. External examination of the brain demonstrated a normal gyral pattern. The cranial nerves and blood vessels showed no abnormalities. Coronal sections of the cerebrum demonstrate bifrontal tracks traversing the frontal neocortex, centrum semiovale, and basal ganglia, both terminating in the ipsilateral subthalamic nuclei. The cortical ribbon was uniform and of normal thickness. Cross sections of the brainstem and parasagittal sections of the cerebellum show tan discoloration of the substantia nigra, but are otherwise without significant gross abnormalities.

Material submitted: H&E section of cerebrum, unstained slide

Points for discussion: 1. Diagnosis, 2. Pathogenesis