

51st ANNUAL DIAGNOSTIC SLIDE SESSION 2101

CASE 2010-11

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Clinical history:

60-year-old, who twelve days prior to his death, had an episode of airway obstruction during a swallowing evaluation (June 2009), due to new onset choking episodes.

The patient had an engineering degree and managed his own business. He played golf and soccer. At age 56, he developed partial complex seizures. MRI showed slight atrophy of mesial temporal lobes and a cerebellar hemangioblastoma. A persistent and protracted neuropsychiatric symptomatology began to evolve including insomnia, suicidal ideation, panic attacks, tremors, unsteady gait, decreased social interaction, obsessions, weight loss attributed to compulsive exercising and loss of sex drive. Seizure activity was difficult to control. In 2008 he had to downsize his business, and developed forgetfulness and disorientation. He had "good days" when he read newspapers, played cards, and displayed a fluent speech. A vast number of hematological, biochemical, and radiological studies gave normal results. By May 2009, the patient had major memory problems. He had no abnormal movements or features of Parkinsonism. In June 2009, he began to experience choking episodes.

Family history: Mother died of ALS.

Autopsy findings:

Brain weighted 1550 grams, fresh and showed no significant atrophy. A cystic lesion with a mural nodule (3.5 x 3 cm) was identified in one of the cerebellar hemispheres, histologically consistent with a hemangioblastoma.

Microscopic examination revealed:

- Tau AT-8 immunostaining and Gallyas Ag impregnation: incipient argyrophilic grain disease.
- α -synuclein immunostaining: significant Lewy pathology in substantia nigra with trace involvement of amygdala.
- TDP-43 immunostain: incipient proteinopathy in hippocampus and entorhinal cortex.
- There was no positive staining for tau, α -synuclein or TDP-43 in neocortex.

Material submitted:

Any of sensory motor, frontal, temporal or parietal areas.

Points for discussion:

- A. What is the immunostain provided?
- B. What is the diagnosis?