

Canadian Association of Neuropathologists
L'Association Canadienne des Neuropathologistes

CASE No 9

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The patient was a 70-year-old male physical education teacher. Medical hx: asthma, hypertension. The patient was treated with gamma knife for a cavernous sinus meningioma in the year 2000. He was an ex-smoker and drank 15 alcoholic beverages a week. He was born in the Ukraine and had no family hx of relevance. In the summer of 2007 he began to experience tipping over while curling, slurring of speech, uncontrollable laughter and crying, weakness of upper and lower extremities, choking while taking fluids and muscle twitching in all limbs. Bladder and bowel function remained normal. This was followed by difficulties with fine hand movements such as combing, opening jars and using cutlery.

Physical examination showed left ptosis, enlarged and sluggish left pupil and restriction of extraocular movements of the left eye. There were brisk jaw jerks and flaccid and spastic dysarthria. Fasciculations in tongue and all extremities and trunk were noted. Muscle bulk was reduced in UE'S and clonus at both ankles was elicited. There was mild spasticity and reduced strength in all extremities with almost normal gait.

EMG study showed diffuse active and chronic motor neuron disease
The disease progressed rapidly and inexorably. No detailed clinical information is available. The patient died of respiratory failure on 11/25/2008. The duration of illness was about twenty months.

The autopsy was performed two hours after death. The right half of the brain was unremarkable on surface examination. In the cord the ventral gray exhibited a tan discoloration and was atrophic contrasting with pallor of corticospinal tracts.

Immunostaining of multiple regions and levels of CNS revealed:

Tau and amyloid beta: vestiges of positivity in entorhinal cortex;

Synuclein: -ve; TDP-43 protein: -ve ; and misfolded sod1: -ve

Molecular studies: negative for mutations in SOD1, fus and tdp-43 protein

Submitted: a) spinal cord: LFB/H&E
 b) Sensory motor area: H&E

Question: Diagnosis?