

49th ANNUAL DIAGNOSTIC SLIDE SESSION 2008

CASE 2008-3

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Clinical History:

- 16 year old white female presented in late 2005 with progressive right upper and lower extremity weakness, dysarthria, dysphagia and anorexia associated with 26 lb. weight loss.
- Medical History: In utero exposure to cocaine. Attention Deficit Hyperactivity Disorder (ADHD) since age 6 treated with Ritalin® and Concerta®.
- Family History: a similar neurologic disorder occurred in the patient's father (D.O.D. 43), half brother and half sister, the latter dying in their early 20's.
- Physical Exam: Weight: 42.5 Kg, below the third percentile. Mild scoliosis with deviation to the right. Weak gag reflex. Mild decrease of breath sounds. Weakness in neck flexors and extensors, as well as in shoulder shrug. Decreased strength proximally in the right upper and right lower extremities (3/5), right greater than left. Symmetric deep tendon reflexes (2+), except right biceps (1+). Negative Romberg sign. Difficulty balancing on her heels and walking on her heels. She could not squat. Sensation intact to touch, pain, temperature, vibration and position. Intelligible speech, but somewhat muffled.
- Neuroimaging: Normal cervical spine and brain MRI with and without contrast. Significant weakness in the oral and pharyngeal phases of swallowing on video fluoroscopy. Normal EEG. Normal motor and sensory nerve conduction studies. Abnormal EMG with a neuropathic pattern.
- Negative lumbar puncture.
- Muscle biopsy (right quadriceps) revealed severe denervation atrophy with associated reinnervation. Severe restrictive chest wall disease in pulmonary function tests.

Necropsy findings:

- External examination showed an estimated weight of 110 lbs, cachexia, atrophic lower extremities arranged in a frog-leg position. No gross abnormalities of the cerebrum, brainstem, cerebellum or spinal cord.

Material submitted: An H&E stained section of a spinal cord segment

Points for discussion: Diagnosis and differential diagnosis