Case 6

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Clinical Abstract:

The patient was a 38 year-old woman who was well until age seven. At that time she began tripping and often could not keep up with her classmates at play. She was examined by a neurologist at age eleven who found bilateral pes cavus and bilateral Babinski responses. Sensory perception was normal. Romberg's sign was absent. At age twelve weakenss of legs was found. Cranial nerves were intact. There was neither nystagmus nor dysmetria. Pain, touch and position sense were intact. Knee jerks were hyperactive and bilateral Babinski responses were again present. Bilateral triple arthrodesis were performed to improve the patient's gait.

At age 15 a mild scoliosis was noted but she walked unaided until late in the second decade when she began to use crutches. In her early twenties she had to use a wheelchair. In her early thirties she could no longer care for herself and was admitted to a nursing home. All during this period she saw physicians only irregularly. By age thirty-four the patient was dysphagic and aphonic. At age thirty-five, after a pulmonary embolus and multiple lung abscesses due to aspiration pneumonia, a feeding gastrostomy was done. At that time she had a flaccid quadriparesis with severe muscle wasting but could grimace, smile and appeared alert. Only pectoralis reflexes were present. There was no nystagmus; optic discs and retinae were normal. She grimaced in response to painful stimuli everywhere. An EEG showed shifting bitemporal delta and theta activity without sharp waves or spikes.

She returned to the nursing home and died just after her thirty-eighth birthday. She had needed nearly total care for five years.

General autopsy showed pes cavus, muscle wasting, multiple pulmonary emboli and bilateral acute aspiration pneumonia. A bronchial carcinoid tumor was an incidental finding. The heart was normal and sections of skeletal muscle showed neurogenic atrophy.

Neuropathology: The brain was small with small pointed frontal lobes. The hemispheres were symmetrical and there was mild widening of the sulci and narrowing of the gyri in the frontal region. Coronal sections revealed slightly enlarged lateral ventricles. The white matter and corpus callosum appeared decreased in size. The basal ganglia and thalamus were grossly normal as was the cerebellum. Sectioning of the brainstem revealed a decrease in size of the pyramids. The cervical spinal cord was narrowed and the posterior columns of the spinal cord were chalky white.

Materal Submitted: Sections are from spinal cord and midbrain or corpus striatum.

Points of Discussion:
1. What is the diagnosis?
2. What are the intraneuronal inclusions?