1995 Case 5

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The patient was a 62 year old man with a long history of a chronic progressive neurodegenerative disease who died at a local nursing home with the following history. From the age of 14 months, when the patient began to walk , he was never able to keep up with children his age and fell easily. At age five he was seen by a physician who noted him to be unsteady on his feet with his eyes closed. He had hyperactive knee jerks unsustained ankle clonus and positive Babinski's.. At age 12, he was seen at Yale where he was found to be generally weak, the legs more than the arms. The weakness was symmetrical with atrophy of the interossei of the hands and a suggestive atrophy of peroneal and anterior tibial muscles. Ankle jerks were absent, knee jerks were diminished with slightly hyperactive reflexes in the arms. Pyramidal, posterior column, and cerebellar signs were all negative. Sensation was intact except for loss of vibratory sense in the feet. Pes cavus was present. He had nystagmus on the extremes of lateral and upward gaze. There was marked unsteadiness in the upright posture and with support he walked with a widebased gait. His speech was high-pitched, monotonous and slow. Laboratory studies including a lumbar puncture were normal.

Family history revealed no other family members with any neurological disease. His sister was normal except for dextrocardia.

He was seen again at Yale at age 20.At that time his neurological exam was not much different than at age 12 although he had been using leg braces to help in walking. He also had atrophy of thenar and hypothenar eminence as well as of interossei muscles of the hand.. During the time between 12 and 20, the patient finished eighth grade at a special school. He also worked at a Defense plant as a grinder for nine months. He lived at home and did not work after the war was over.

At age 23, he was seen at Yale for vocational rehabilitation. At that time they noted that his IQ was dull-normal. Other findings had remained the same except his gait was even more unsteady. Adiodokokinesis and fine hand movements were performed adequately and he had no intention tremor. Speech was now slurred, high pitched, monotone and responses slow although the patient was cooperative. The patient reported that occasionally when he swallowed liquids they would come out his nose. He reported some difficulty in initiating the urinary stream but no urinary incontinence although he reported fecal incontinence.

At age 32 he was admitted to at a chronic care hospital
because of the death of his parents and his inability to care for himself. At that time he had both fecal and urinary incontinence, bilateral foot drop, weakness of all lower extremity muscles bilaterally as well as weakness of hip flexors. Both knee and ankle jerks were absent and vibratory and position sense was decreased in both lower extremities to the sacrum. He had no cerebellar deficit or fasciculations. A lumbar puncture showed increased protein.

At age 56 he was seen by another neurologist who noted several new findings including dysarthria, and intention tremor in his right arm. He now needed a walker to get around. Nerve conduction studies showed compressive neuropathy at the wrist and EMG findings were consistent with neuropathic disease rather than myopathic disease. He was seen several times at the MDA Clinic with further progression of his disease. The last note when he was age 60, indicated more progression of his disease. His voice was hypophonic with dysarthria. His lower extremities were markedly weak. His hands were clawed without strength although proximal strength was relatively preserved. He had a pronounced intention tremor. There were trace reflexes noted at the biceps but deep tendon reflexes were absent at other sites. Planter responses were silent and there was sensory loss distally in the lower extremities as well as in the hands. MRI showed cerebellar atrophy without pontine atrophy. Nerve conduction studies were consistent with a sensory neuropathy. Intellectual function was difficult to assess due to his problems with speaking, however, he was able to cooperate with the examiner.

He died at the age of 62 of acute and chronic aspiration pneumonia. At autopsy the brain showed mild cerebral and cerebellar atrophy.

Your slide is an LFB-PAS stained section of cortex and spinal cord.

Questions: 1. What is the diagnosis?
2. what is the etiology?