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CASE 2001-01

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Clinical History:

The proband, a male, had an episode of generalized seizures at the age of 24. Prior to that time, he had lived a healthy life working as an architectural draftsman; however, afterwards, he had numerous episodes of action myoclonus. His performance at work deteriorated as he began having difficulties with memory, occasionally being unable to write his own name. At the age of 27, seizures reappeared; they were myoclonic, complex partial, and tonic-clonic seizures. In spite of aggressive treatment, his seizures were difficult to control and there were several episodes of status epilepticus. A neurological examination revealed slow speech, diplopia, vertical and horizontal directional nystagmus, dysarthria and myoclonus in the extremities. The tendon reflexes of the extremities were increased, except for the Achilles tendon reflex, which could not be elicited. Sensory examination showed hypoalgesia in a glove and stocking distribution. Neuropsychological examinations revealed severe generalized impairment consistent with advanced dementia. Routine laboratory tests were normal. EEGs showed frequent spikes as well as spike and wave complexes in the central and temporal regions bilaterally. Cerebral atrophy was evident on CT and MRI scans. As the disease progressed, myoclonus of the face and extremities worsened as well as cerebellar ataxia. He died from aspiration pneumonia at age 43.

Family history:

The proband's mother, presented seizures at 25 years of age and was diagnosed as having progressive myoclonus epilepsy. She died at 37 years of age. The proband's brother was also diagnosed as having progressive myoclonus epilepsy. He died at age 43 and showed pathologic features identical to those of the proband.

Necropsy findings:

The fresh brain weighed 980 g and was diffusely atrophic. The most striking cytological finding was the presence of eosinophilic and PAS-positive bodies in the neuronal perikaryon and cell processes throughout most gray matter areas of the brain and spinal cord. In the peripheral nervous system, the eosinophilic and PAS-positive bodies were seen in the dorsal root ganglia.

At the cytological examination of peripheral organs, intracytoplasmic bodies comparable to those seen in neurons were not present. The lungs showed organized aspiration pneumonia and acute bronchopneumonia.

Material submitted:

One hematoxylin and eosin stained section of the frontal cortex.
One unstained section of the frontal cortex.

Points for discussion:

1. Differential diagnoses and final diagnosis
2. Etiology of disease