

48TH ANNUAL DIAGNOSTIC SLIDE SESSION

CASE 2007-7

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Clinical history: This 72-year-old man first became ataxic at the age of 39 years though his disability was not serious enough to keep him out of the U.S. Air Force. He got married and had 7 children of whom no one has a similar disease. His parents were also not affected. At the time of examination, the patient was confined to a wheelchair. He could not transfer to a bed on his own, stand, or walk. His mental status inquiry revealed some lack of concentrating ability. He had prominent dysarthria and coarse nystagmus on right and left lateral gaze. Saccades were hypometric. Pupillary light reactions were normal, and the fundi were normal. He had some dysmetria of the arms and hands, with a distinct writhing component. His arms were strong but his legs were atrophic, spastic, and weak. A right hammer toe was present. Muscle stretch reflexes were lost but there was an intermittent right Babinski sign. A suprapubic catheter was in place. He had normal surface sensation everywhere. Vibratory sense was reduced but joint position sense in the toes was totally absent. Some examiners considered multiple system atrophy. His single cranial magnetic resonance imaging one year later showed cerebrotocerebellar atrophy. Gene testing for Friedreich's ataxia was negative. He developed visual failure, and optic atrophy became prominent. After admission to a nursing home, he began to refuse food, drink, and medications. He died after several weeks of progressive malnutrition. The general autopsy confirmed bronchopneumonia. The brain weighed 1,050 g.

Material submitted: One H&E-stained slide of the thalamus

Points for discussion:

- (1) Diagnosis
- (2) Pathogenesis