Canadian Association of Neuropathologists
L'Association Canadienne des Neuropathologistes

CASE No 4

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This 83-year-old woman died from a progressive neurological illness that began at the age of 54 with unsteadiness of gait. Over the years, examiners described ataxia, dysarthria, dysmetria, dysdiadochokinesia, and intention tremor. The patient's father died at 79 but had no signs of ataxia. The mother died at 90, also without showing ataxia during life. The patient had 8 full brothers and 2 full sisters of whom at least 4 had (or have) ataxia. A maternal cousin has severe ataxia. In addition, the family history includes several members with multiple sclerosis or Parkinson's disease. A medical geneticist concluded that transmission was "dominant with incomplete penetrance". Testing for spinocerebellar ataxia types 1, 2, and 3; and for dentatorubropallidoluysin atrophy was negative. The autopsy was restricted to brain and spinal cord. Brain weight was 1068 g.

Material submitted: One hematoxylin & eosin-stained slide and one unstained slide of thoracic spinal cord

Point for discussion: Diagnosis