Supplementary information

Supplement. Information about cases with *FGF14* GAA expansion between 200 and 250 repeats

Case 1.

A 78-year-old female presented with unsteady gait progressing over 3 years. On neurological examination, gaze-horizontal nystagmus, downbeat nystagmus, slurred speech, decrease of vibratory sense, poor heel-knee test, and wide-based gait were noted. Autonomic symptoms, including orthostatic hypotension, were not observed. None of the parents or siblings presented with cerebellar ataxia. Brain magnetic resonance imaging (MRI) showed atrophy of the cerebellum, and 3D - Stereotactic Surface Projections (SSP) analysis of single photon emission computed tomography with N-isopropyl-p-^{123}I-iodoamphetamine (^{123}I-IMP-SPECT) showed a small decrease in blood flow in the cerebellum. Seven years after onset, the patient is still able to walk unassisted. She had a pure GAA expansion of 225 repeats in the *FGF14* gene.

Case 2.

A 77-year-old female presented with unsteady gait and dysarthria progressing over 5 years. On neurological examination, gaze-horizontal nystagmus, scanning speech, poor finger-nose-finger test, poor heel-knee test, and wide-based gait were noted. Autonomic symptoms, including orthostatic hypotension, were not observed. None of the parents or siblings presented with cerebellar ataxia. Brain MRI showed mild atrophy of cerebellum, and 3D-SSP analysis of ^{123}I-IMP-SPECT showed no significant decrease of blood flow. Nine years after onset, the patient is still able to walk unassisted.

She had a pure GAA expansion of 206 repeats in the *FGF14* gene.