**Supplemental methods**

**Whole exome sequencing and evaluation related to other genetic disorders presenting with ataxia.**

A DNA sample from the patient was analyzed by whole exome sequencing. Genomic DNA was captured by using SureSelect XT Human All Exon V6 (Agilent Technologies, Santa Clara, CA). Captured DNA was sequenced on a HiSeq 4000 (Illumina, San Diego, CA). The mean read depth against target regions was 85.0 reads with 96.0% of target regions being covered by 20 or more reads. The quality controlled reads were mapped to the human reference genome (UCSC hg19) by using BWA (Burrows-Wheeler Alignment Tool). After the removal of PCR duplications using Picard (v1.13), single nucleotide variants (SNVs) and short insertions and deletions (indels) were identified by using the Genome Analysis Toolkit (GATK; v3.4-0) and annotated by using SnpEff (v4.1g).

As a result, the number of missense variants and indels were 11,402 and 12,533, respectively. Among them, we focused on mutations in genes causing ataxia as previously reported1 (Table e-1). We used whole *exome* sequencing; therefore, *mitochondrial DNA* and *ATXN8OS* were not covered in the analysis. Our case did not have additional features such as deafness, diabetes, myoclonus, neuropathy, or spastic paraparesis, so therefore mitochondrial disease was unlikely2. Also, SCA8 was unlikely because the onset age of all reported SCA8 cases is below 73 years.

After filtering out the clearly benign mutations such as intron\_variant or synonymous\_variant, 1 inframe\_insertion in *ATXN2* which was already confirmed by Sanger sequencing (Figure D) and 33 missense\_variants were left in 14 genes causing ataxia (Table e-2). In the list, the non-reference allele frequency of existing variations in the 1000 Genomes combined East Asian population (phase 3) was relatively high for most mutations; therefore, these mutations were unlikely to cause ataxia. The lowest frequency for the mutation was 0.004 (0.4%) in the *GALC* gene; however, the evaluation by Variant Clinical Significance was "benign". In conclusion, we found no causative mutation among the candidate genes for ataxia except for the homozygous expansion of trinucleotide repeats in *ATXN2.*

**References**

1. Hadjivassiliou M, Martindale J, Shanmugarajah P, et al. Causes of progressive cerebellar ataxia: prospective evaluation of 1500 patients. J Neurol Neurosurg Psychiatry. 2017 Apr;88(4):301-309
2. Bargiela D, Shanmugarajah P, Lo C, et al. Mitochondrial pathology in progressive cerebellar ataxia. Cerebellum Ataxias. 2015 Dec 4;2:16