Supplemental data

Appendix e-1. Cognitive tasks associated with each cognitive domain

Cognitive domain	Cognitive tasks
Attention /8	Digit span[1] and numeric substitution[1]
Executive /7	Go/No-Go task [2] Graphic version of the Luria alternating sequence[3] Trail Making B [4] Rey complex figure test copy [5] Lexical verbal fluency (letter P) Abstract thinking (proverb interpretation)*
Visuospatial /12	Cube copy test [1] and Clock drawing [6]
Memory /5	Five-word free and cued recall [1]
Language /3	Naming[7] Comprehension[8] Repetition[1]

* local cognitive test (Clinique Interdisciplinaire de Mémoire, CHU de Québec, Québec, Canada, www.cliniquedememoire.ca).

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3. Luria AR. Human brain and psychological processes. New York,: Harper & Row, 1966.
4. Barncord SW, Wanlass RL. The symbol trail making test: test development and utility as a measure of cognitive impairment. Appl Neuropsychol 2001;8:99-103.
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Appendix e-2. Detailed cognitive evaluations of SCA34 patients

Case III-3
This patient was evaluated at the memory clinic at 80 years of age. He had achieved theology studies and worked as a pastor. He was independent for activities of daily living but lived in a supervised religious housing community. Upon examination, the patient showed euphoria. He insisted on meeting the secretary who scheduled his medical appointment. There were no language deficits. He scored 27/30 on the Mini-Mental State Examination (MMSE), losing one point on calculation and two points on memory recall. He scored 24/30 on the Montreal Cognitive Assessment (MoCA), losing 3 points for the trail making, cube copy, and clock drawing. He also lost 3 points on memory recall, which improved with cueing. He failed on the Luria test, alternating graphic sequence and Go/No-Go task. The Rey Complex Figure Test (RCFT) was significantly impaired, and he proceeded by juxtaposition of details. On praxis assessment tasks, the patient presented body-part-as-object (BPO) gesture.

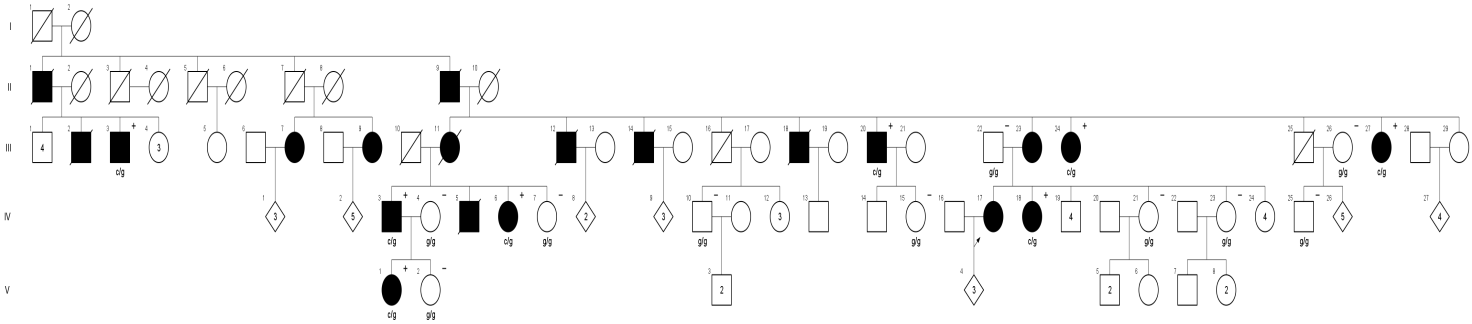
Case IV-3
This patient was evaluated at the memory clinic at 72 years of age; he was a male with a grade 11 education. He used to be an iron worker. He was mildly impaired on instrumental activities of daily living and lost his driver's licence following a road test examination. He scored 25/30 on the MMSE, losing 4 points on calculation and one point on memory recall, which improved with cueing. On follow-up, his MMSE dropped to 21. MoCA score was 22/30, he failed the trail making and cube copy. Clock drawing was mildly impaired. Working memory was altered, he failed backward digit span and months backward. Lexical verbal fluency was significantly decreased. Abstract thinking was slightly diminished. He correctly performed the Luria but failed the Go/No-Go task. The RCFT was impaired, and he proceeded by juxtaposition of details. Language was preserved. There was no apraxia.

Case IV-6
This patient was evaluated at the memory clinic at 70 years of age; she was a female with a grade 12 education. She worked as a secretary. She complained about attention deficits from an early age. She had trouble finding her way while driving over five years ago, but did not report other functional impairment. Her MMSE score was 23/30. She lost her points on calculation, pentagons drawing and memory recall, which improved with cueing. She scored 24/30 on the MoCA. She failed the cube copy and her lexical verbal fluency was impaired. She failed the Go/No-Go and Luria tests. She missed 4 of 16 words on naming task. Copying of the RCFT was performed with mild errors. She did well on praxis assessment.

Case IV-18
This patient was evaluated at the memory clinic at 64 years of age; she was a female with a grade 12 education. She worked as a transcriptionist but had been dismissed because of reduced performance. When questioned, she claimed that she had character incompatibility with her manager. On examination, she showed euphoric affects and lack of insight. She was talkative and disinhibited; she made inappropriate comments about physical appearance of physicians. She scored 27/30 on MMSE, losing one point on calculation and two points on memory recall. She scored 24/30 on the MoCA, she failed the trail making and cube copy. Her lexical verbal fluency was altered. She was unable to perform the Go/No-Go and Luria test. The RCFT was significantly impaired. Praxis assessment tasks were performed correctly.

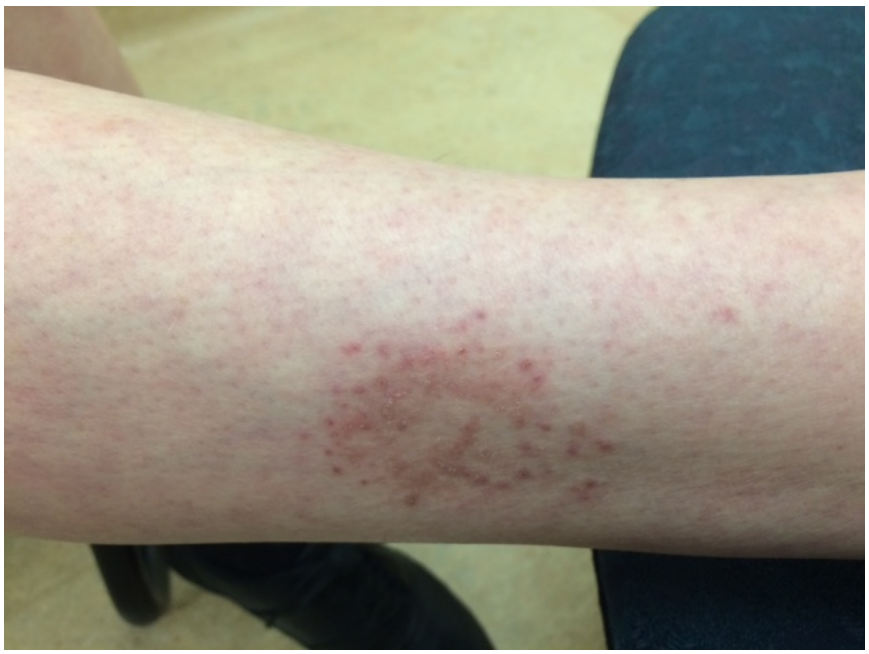
Case V-1
This patient was evaluated at the memory clinic at 42 years of age; she was a female with 14 years of education. She had had a diagnosis of attention deficit-hyperactivity disorder at the age of 30, which was treated with methylphenidate, and has also been diagnosed with panic disorder. She scored 28/30 on the MMSE, losing two points on memory recall, which were recovered by cueing. Her MoCA score was 29/30, she missed one point on the lexical verbal fluency. She showed impaired working memory and selective attention on detailed neuropsychological assessment. The RCFT was drawn perfectly. Subtle impulsiveness was noted.

Figure e-1. Pedigree of a French-Canadian Family with the c.504 G>C mutation in *ELOVL4*.



Symbols with a + sign in the right corner indicate that the patients is bearing the identified c.504 G>C mutation in the *ELOVL4* gene, and the genotype is indicated below the symbol. Symbols with a – sign indicate the absence of a mutated allele in *ELOVL4*.

Figure e-2. Dermatologic lesion in a SCA34 patient



Evaluation by a dermatologist revealed nummular dermatitis, but no sign of erythrokeratodermia variabilis.

Table e-1. Clinical and paraclinical characteristics of patients with mutations in *ELOVL4* in present and previous studies

Characteristic	Present Study	Giroux et al. 1972[1] Cadieux-Dion et al. 2014[2]	Ozaki et al. 2015[3]	Bourassa et al. 2015[4]	Bourque et al. 2018[5]
No of patients, n	9	19	9	1	1
Ethnic origin	French-Canadian	French-Canadian	Japanese	Brazil	English Canadian
Mutation	c.504G>C, p.L168F	c.504G>C, p.L168F	c.736T>G, p.W246G	c.539A>C, p.Q180P	c.698C>T, p.T233M
Mean age at onset, y	47	51	33.9	Mid 20s	15
Ataxia	Gait and limb ataxia with dysarthria (9/9)	Gait ataxia (12/19) limb ataxia (9/19) and dysarthria (6/19)	Gait and limb ataxia with dysarthria (9/9)	Gait and limb ataxia with dysarthria (1/1)	Gait ataxia (1/1), absence of limb ataxia or dysarthria
Oculomotor abnormalities	Nystagmus (7/8) hypometric saccades and saccadic pursuit (8/8)	Nystagmus (7/19) slow pursuit (5/19), slow saccades (3/19)	Nystagmus (7/9) supranuclear gaze palsy (3/9), altered smooth pursuit (5/9)	Mild bilateral ophtalmoplegia, diplopia, horizontal gaze-evoked nystagmus (1/1)	Square wave jerks, periodic alternating skew deviation, saccadic pursuit (1/1)
Motor neuron involvement	Decreased DTR (2/9)	Decreased DTR (7/19)	Increased DTR or Babinski (8/9)	None	Decreased DTR (1/1)
Dermatologic involvement	Nummular dermatitis (1/9) Absence of EKV (9/9)	Active or past EKV (14/19)	None	Past EKV	Active EK
Cognitive involvement	Alterations in executive, visuospatial attention (5/5), psychiatric features (3/5)	Cognition appeared normal	Not reported	Not reported	Not reported
Neuroimaging	Mild to severe cerebellar (5/5) and pontine (3/5) atrophy. Hot cross bun sign (2/5)	Cerebellar (6/9), pontine (5/9) and/or cerebral (4/9) atrophy, normal MRI (3/9)	Cerebellar and pontine atrophy (8/8), hot cross bun sign(4/6) or pontine linear hyperintensity (2/6)	Cerebellar and pontine atrophy (1/1)	Mild cerebellar and pontine atrophy (1/1)
Electrophysiolo-gical anomalies	Peripheral neuropathy (2/3), Normal (1/3)	Mild peripheral axonal neuropathy (4/8)	None	Not reported	Normal (1/1)

DTR deep tendon reflexes; EK(V) erythrokeratodermia (variabilis); MRI Magnetic resonance imaging; VLCFA very long chain fatty acids; VLC-SFA very long chain saturated fatty acids

1. Giroux JM, Barbeau A. Erythrokeratodermia with ataxia. Arch Dermatol 1972;106:183-188.
2. Cadieux-Dion M, Turcotte-Gauthier M, Noreau A, et al. Expanding the clinical phenotype associated with ELOVL4 mutation: study of a large French-Canadian family with autosomal dominant spinocerebellar ataxia and erythrokeratodermia. JAMA Neurol 2014;71:470-475.
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