A 40-year-old woman with delayed motor milestones and high arches since childhood was investigated for a progressive gait disorder from the age of 24 years. On clinical examination, a spastic and ataxic gait was present, with mild ataxia in upper limbs, dysarthria, and nystagmus. Nerve conduction study/EMG revealed a demyelinating neuropathy. MRI of the brain (figure 1) and fundus photography (figure 2) suggested autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) and the diagnosis was genetically confirmed. The combination of spinocerebellar ataxia with demyelinating neuropathy, superior vermis atrophy, and pontine linear hypointensities should prompt the diagnosis of ARSACS, which may be an underdiagnosed condition outside Quebec.

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