Autosomal recessive spastic ataxia of Charlevoix-Saguenay

Figure 1 Atrophy of the vermis, mainly of the upper part on a T2-weighted image (A, arrow), and linear hypointensities in the pons on fluid-attenuated inversion recovery image (B, arrow)

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)¹ and the diagnosis was genetically confirmed.² 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.

Figure 2 Color (A) and black and white (B) picture of fundus revealing hypermyelinated fibers in the retina, which are present in some cases of autosomal recessive spastic ataxia of Charlevoix-Saguenay

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