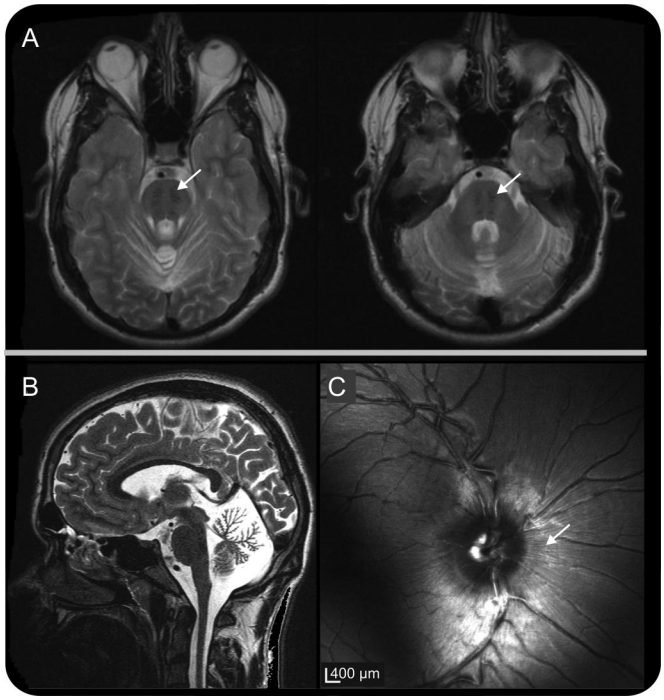
Characteristic MRI and funduscopic findings help diagnose ARSACS outside Quebec

Figure MRI



(A) T2-weighted MRI shows linear hypointensities near the pyramidal tract in the pons on transversal slices and (B) cerebellar atrophy, predominantly of the superior vermis on sagittal slices. (C) Funduscopy shows myelinated fibers radiating from the optic disk.

A 20-year-old German man had progressive ataxia, spasticity, and polyneuropathy from childhood. MRI revealed linear pontine hypointensities (figure, A) and cerebellar atrophy (figure, B). Funduscopy showed myelinated fibers radiating from the optic disc (figure, C). Autosomal recessive spastic ataxia Charlevoix-Saguenay (ARSACS) was diagnosed by identification of biallelic *SACS* gene mutations (chromosome 13q12). The gene product is sacsin, found in large neurons such as Purkinje cells, one of many ataxia-related proteins. ARSACS, first described in the French-Canadian founder population of Quebec, occurs outside Quebec,[1,2] suggesting that worldwide screening in early-onset cerebellar ataxia with characteristic pontine and funduscopic abnormalities may identify additional cases.

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