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BRIEF COMMUNICATION

Cerebellum and neuropsychiatric disorders: insights from ARSACS

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Abstract Autosomal recessive spastic ataxia of Charlevoix–Saguenay (ARSACS) is a rare neurodegenerative disorder characterized by ataxia, spastic paraparesis, polyneuropathy, and evidence of superior cerebellar vermis atrophy at magnetic resonance imaging (MRI). Reports of atypical presentations and additional clinical or MRI findings have been recently published, but psychiatric disturbances have never been associated with ARSACS. We describe four ARSACS patients manifesting severe psychiatric symptoms including psychosis, panic disorder, and depression during the course of the disease. Our case reports further expand the ARSACS phenotype and add clinical data in favor of the hypothesized relationship between cerebellar dysfunction and psychiatric disorders.

Keywords ARSACS · Sacsin · Neuropsychiatric disorders · Cerebellum · Psychosis

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Introduction

Sacsin-related spastic ataxia (ARSACS, MIM 270550) is a neurodegenerative disorder originally described in Québec [1] and later documented worldwide upon a wider screening of *SACS*, the disease gene. The phenotype is characterized by the triad of early-onset ataxia, spastic paraparesis, and polyneuropathy. Brain magnetic resonance imaging (MRI) classically reveals atrophy of the superior cerebellar vermis and linear T2-hypointensities in the pons. Recently, the clinical and imaging spectrum of ARSACS has been expanded, and atypical presentations as well as additional findings have been observed [2–4].

Cognitive impairment is occasionally part of ARSACS and intelligence quotient (IQ) levels tend to be in the lower normal range, though most cases are able to cope well with daily activities. So far, only two ARSACS male sibs presenting deficits in cognitive and behavioral functioning consistent with the so-termed cerebellar cognitive affective syndrome (CCAS) have been reported [5]. Here, we describe four ARSACS patients with classical clinical phenotype manifesting severe psychiatric disturbances during the disease course.

Case reports

Case 1 is a 28-year-old woman with a history of mild delay in psychomotor development, gait unsteadiness, and urinary incontinence. Neurological examination at age 21 revealed spastic ataxia, dysarthria, pes cavus, areflexia except for brisk patellar reflexes, and Babinski sign. Full scale IQ scored 61 confirming mild intellectual disability. Nerve conduction study uncovered axonal sensory neuropathy. Brain MRI revealed upper vermis atrophy