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CLINICAL PHENOMENA OF CHARLEVOIX-SAGUENAY ATAXIA IN TWO ADULT BROTHERS

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Introduction: Autosomal recessive ataxia of Charlevoix-Saguenay (ARSACS) is characterized by early onset cerebellar ataxia, lower limb spasticity, and sensorimotor axonal polyneuropathy. Early atrophy of the superior cerebellar vermis is always present. Molecular linkage analysis found that the ARSACS gene is located on chromosome 13q12. Cloning of the gene, SACS, demonstrated that it encodes the protein sacsin.

Objectives: Systematic evaluation of neurological, neuropsychiatric and neuropsychological features in two male siblings.

Aims: Investigation of the putative relationship between cerebellar dysfunction and affective symptoms.

Methods: Detailed neuropsychiatric and neuropsychological assessment.

Results: The first patient, aged 55, the disease started in early infancy and a severe progressive cerebellar syndrome with spasticity of the legs and axonal polyneuropathy developed. In his brother, aged 50, the debut of neurological symptoms was in preadolescence with a less severe deterioration over time. Cognitive functioning was only marginally impaired in the latter patient, whereas behavioural aberrations were present in the first patient only. Both patients showed a reduced cognitive and emotional responsivity to environmental events leading to impairments in several areas of daily life, such as lack of effort and strategic planning, as well as impulsivity and impoverished social interaction with emotional indifference. This symptom profile typically points towards the presence of an apathy syndrome.

Conclusions: In ARSACS, in addition to the motor impairments, it may be postulated that the cerebellar cognitive affective syndrome is present. Thus, this hereditary form of ataxia may be accompanied by a series of non-motor symptoms of which motivational and affective signs dominate.

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