

SYNE1 Ataxia

SYNE1 gene mutations were identified as a cause of late-onset pure cerebellar syndrome. Non-cerebellar symptoms, including cognitive impairment, were already described in this condition. The aim of this study was to perform a detailed cognitive and psychiatric description of patients with SYNE1 gene mutations. We performed neuropsychological and psychiatric evaluations of six patients with SYNE1 ataxia and compared their performance with 18 normal controls paired for age and education level. SYNE1 ataxia patients present cognitive dysfunction, characterized by impairment in attention and processing speed domains. Otherwise, the psychiatric assessment reported low levels of overall behavioral symptoms with only some minor anxiety-related complaints. Although this is a small sample of patients, these results suggest that SYNE1 ataxia patients may represent a model to investigate effects of cerebellar degeneration in higher hierarchical cognitive functions. For further studies, abstract thinking impairment in schizophrenia may be related to dysfunction in cerebellum pathways ¹⁾.

¹⁾

Gama MTD, Braga-Neto P, Dutra LA, Alessi H, Maria LA, Gadelha AA, Ortiz BB, Kunii I, Correia-Silva SR, Dias da Silva MR, Dion PA, Rouleau GA, França MC Jr, Barsottini OGP, Pedroso JL. Cognitive and Psychiatric Evaluation in SYNE1 Ataxia. *Cerebellum*. 2019 May 2. doi: 10.1007/s12311-019-01033-5. [Epub ahead of print] PubMed PMID: 31049853.

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Last update: **2024/06/07 02:52**

