

CEREBELLAR COGNITIVE AFFECTIVE SYNDROME IN RFC1-RELATED DISORDER.

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Background: *RFC1* Expansions were recently described as the cause of the cerebellar ataxia, neuropathy and vestibular areflexia syndrome (CANVAS). So far, little is known about the frequency and profile of cognitive impairment in the disease. The Cerebellar Cognitive Affective Syndrome Scale (CCAS-S) is a scale specifically designed to capture neuropsychological changes in patients with cerebellar ataxia, but its use in *RFC1* patients has not yet been pursued.

Objective: to describe the prevalence and clinical profile of CCAS in patients with *RFC1*-related disorder.

Methods: This is an observational, multicentric study, including 10 patients with molecular confirmation of *RFC1* expansions enrolled between October/2022 and January/2023. The Brazilian Portuguese version of the CCAS-S was applied by the same rater and in the same physical space. A failure score of ≥ 3 cognitive domains indicate definite, ≥ 2 indicates probable and ≥ 1 possible CCAS.

Results: The mean age were 58.5 ± 6.7 years. The mean SARA score was 19.5 ± 6.68 . In our sample, 8 (80%) *RFC1* patients had definite CCAS and 2 (20%) had probable CCAS. Mean CCAS-S raw score was $69.6 (\pm 9.6)$ and the mean number of failed domains was $5 (\pm 2.3)$. The cognitive domains with the worst performance were respectively: abstract thinking (100% failure), visual-spatial (70%) and executive function (60%).

Conclusions: CCAS is frequent in *RFC1*-related disorder. CCAS-S is an interesting tool to assess cognitive decline in these patients.

Keywords: Cerebellar Cognitive Affective Syndrome, *RFC1*, CCAS, Cerebellar Cognitive Affective Syndrome Scale.