PERSPECTIVE

Cerebellar cognitive affective syndrome in friedreich ataxia: Causes and consequences

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ABSTRACT

With a pattern of impairment that falls under the cerebellar cognitive affective syndrome, people with Friedreich Ataxia (FRDA) exhibit significantly lower performances in many cognitive domains. Using multiple variable regression models, determine the primary predictor of the CCAS in a large cohort of people with FRDA. 39 FRDA patients participated in this monocentric observational study. The SARA was used to assess ataxic motor symptoms, and the CCAS-Scale was used to assess cognitive abilities (CCAS-S). Age, SARA, GAA1, Age of onset of Symptoms (ASO), Age, and Disease Duration (DD) were selected as covariates in a logistic regression model to predict CCAS-S failed items, a linear regression model using CCAS and covariates is used. Patients' median ages, SARA scores, ASO, DD, and GAA1 scores were 29.14, 22.10, 14.11, 15.9, and 712.238, respectively (4 point-mutations). The average CCAS-S raw score was 86/16, with an average of 2.9/1.6 failed items. There were 23 people with definite CCAS. This result justifies screening for CCAS, especially in patients with SARA>20, and supports the idea that cognitive and motor symptoms in FRDA share a common core cerebellar pathophysiology.

Key Words: *pathophysiology, screening, mutation*

INTRODUCTION

One of the most prevalent causes of hereditary cerebellar ataxia is Friedreich Ataxia (FA). Reduced levels of frataxin, a mitochondrial protein involved in the formation of iron-sulphur clusters and antioxidant defenses, are related to FA pathogenesis. In 98% of patients, lower frataxin levels are caused by homozygous enhanced expansion of an intronic GAA triplet repeat in the FXN gene, which inhibits the expression of frataxin through an epigenetic process. The 2% of instances that are still present are compound heterozygotes with a deletion or a point mutation in the FXN gene. Most of the remaining frataxin expression in patients with expansions comes from the shorter GAA repeat expansion (GAA1), whose length accounts for 30–50% of the variation in symptom start age and is a predictor of disease severity. Early atrophy of the spinal cords posterior columns and a gradual degeneration of the cerebellar dentate nuclei (DN) and their efferent fibers in the superior cerebellar pedunculi are two features of FA. Clinically, when cerebellar indications manifest, afflicted individuals exhibit symptoms. Then, throughout time, various abnormalities of the cerebellar, pyramidal, visual, and auditory systems lead to the development of the neurological disorder. Through modulation of neocortical activity via the dentato-thalamo-cortical circuits, the cerebellar posterior lobes contribute significantly to cognitive functioning. Known as the cerebellar cognitive affective syndrome, lesions in the cerebellar posterior lobes and/or disruption of dentato-thalamic pathways are linked to certain cognitive patterns (CCAS). Patients with FA are more likely to develop CCAS because to their substantial DN and efferent tracts involvement. However, there is less research on cognitive dysfunctions in FA compared to motor symptoms, despite mounting evidence of moderate but common cognitive dysfunctions. FA is also linked to neocortical changes that include structural atrophy, impaired metabolism, and functional connectivity, all of which may affect cognitive abilities. Clinicians, caregivers, and people with FA may be better able to recognize and treat cognitive issues if they have a better understanding of the factors that contribute to cognitive deficits in FA. Therefore, the purpose of this study is to evaluate the impact of the CCAS in a sizable cohort of people with FA and to determine the determinant of the CCAS using regression models.

CONCLUSION

The maintenance of memory T cell subsets in the BM may shift in response to BMI, as our study reports for the first time. In fact, when the markers CD28 and CD57 were taken into consideration, no relationships were found. So, in addition to age and CMV, BMI is another factor to take into account, especially when researching the phenotypic of effector/memory T cells. Planning metabolic therapies will help determine whether the scenario described in overweightpeople can be reversed, enhancing the ability of adaptive immune cells.

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