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Saturday

Myotonic Dystrophy-1 as a model of frontotemporal degeneration in non-coding region microsatellite diseases with motor involvement

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State of the Art:
C9-ALS/FTD is a member of the sub-family of non-coding region microsatellite diseases driven by both RNA and protein mediated toxicity. It is difficult to characterize progressive changes in ALS/FTD personality due to the aggressive nature of the disease process, while case studies indicate delusions and paranoid schizophrenia (Zucchi et al. 2019). We hypothesized that Myotonic Dystrophy-1 (DM-1), due to commonalities that include being a non-coding region microsatellite disease with muscle involvement, with abnormalities of the hypothalamic–pituitary–adrenal axis (Takeshima et al., 2018; Dedeene et al., 2020) and cognitive deficits associated with the frontal and temporal brain regions (Winblad et al., 2016), could provide insight to progression of the ALS/FTD disease process, beyond that derived from genetically unrelated FTD without muscle disease.

Methodology:
Neuropsychological investigations were performed in 21 DM-1 patients of normal intelligence and 10 matched healthy controls (HC). Between group differences were analyzed by t-tests. For DM-1, within group differences for cataracts, as a biomarker of disease progression, were analyzed by Spearman rho correlations.

Results:
In comparison to HC, DM-1 evidenced significantly lower capacities for attention, semantic fluency, visuospatial processing, visual learning and recall, and letter fluency. Personality assessment found the highest frequency traits for Borderline, Avoidant, Passive Aggressive and Paranoid. The presence of cataracts was significantly correlated with Avoidant and Borderline traits.

Conclusion: Current findings evidence a concordance between cognitive and personality changes in ALS/FTD and DM-1. DM-1 may serve as a better model of ALS/FTD progression than genetically unrelated FTD without muscle disease.

Conflicts of interest

No Conflicts of Interest.