Molecular Genetics and Metabolism Reports 14 (2018) 43

Contents lists available at ScienceDirect

Molecular Genetics and Metabolism Reports

journal homepage: www.elsevier.com/locate/ymgmr

Correspondence

Motor involvement in Fabry disease

Dear Editor,

We have read with great interest the article published by Wise and colleagues on the last issue of Molecular Genetics and Metabolism Reports [1]. The Authors performed a survey study investigating the prevalence of Parkinson Disease (PD) in individuals affected by Fabry Disease (FD), and found that 2 patients out of 90 (2.2%) were diagnosed with PD. Although considering all the limitations of their study design, the Authors conclude that there might be an increased risk of developing PD in individuals with α-galactosidase A mutations.

The present work is not the first study exploring and suggesting a possible motor involvement in FD. Indeed, as reported in the manuscript, the presence of subtle motor symptoms in FD, with slower gait and transfer speed, poorer fine manual dexterity and lower hand speed, has been reported [2]. In line with these evidences, both electrophysiology and advanced MRI studies showed an involvement of the primary motor cortex in this condition. In particular, with transcranial magnetic stimulation an increase of excitatory neurotransmission in motor cortex circuits of FD patients was found [3], while a reduction of the functional connectivity between motor cortices and both basal ganglia and cerebellum was proved by a resting-state functional MRI study, resembling findings present in early phases of PD [4]. Finally, pathology studies demonstrated the presence of abnormal globotriaosylceramide accumulation in the substantia nigra of FD patients, a central hub of the basal ganglia motor circuit [5].

We think that all these evidences, together with the results of this study, further support the hypothesis of a possible extrapyramidal involvement in FD. However, only future prospective studies, conducted on large samples of FD patients with the integration of different modalities, as well as a direct comparisons between FD and PD patients, can further confirm or dismiss this pathophysiological hypothesis.

Study funding

No funding reported.

Disclosure

The author reports no disclosures relevant to the manuscript.

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https://doi.org/10.1016/j.ymgmr.2017.11.006
Received 27 November 2017; Accepted 28 November 2017