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Screening for Fabry's disease in a case series of Parkinson's disease patients from Southern Italy

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Introduction: Autophagy-lysosomal pathway dysfunction is involved in PD pathogenesis, causing abnormal accumulation of alpha-synuclein in Lewy bodies. Mutations in several genes encoding lysosomal enzymes have been associated with PD so far. Fabry disease (FD) is a X-linked lysosomal storage disease caused by alpha-galactosidase (α-GAL) deficiency, leading to deposition of globotriaosylceramide (Gb3) in nervous system and other organs. GLA gene variants and decreased α-GAL activity have been detected in PD patients in a few previous studies or case reports.

Objective: We aimed to assess α-GAL levels in a case series of PD patients from Campania, in Southern Italy.

Methods: One hundred forty-four unrelated subjects (88 males and 56 females) affected with PD were enrolled. Demographics data, medical and family history, and brain MRI findings were collected. α-GAL activity was measured using a fluorometric assay in males. GLA gene sequencing was performed in females and males with decreased α-GAL activity, who also underwent globotriaosylsphingosine (lyso-Gb3) levels analysis.

Results: The mean age ± SD was 67.3± 9.2 years, and mean age ± SD at onset was 58.4±10.2. The most common motor phenotype was tremor-dominant PD (n=75, 52%). Twenty-nine participants (20%) had a positive family history for PD, whereas 7 subjects (7%) reported at least one first family member suffering from cerebrovascular disease. Seventy-six patients (53%) had at least one cardiovascular risk factor and four (3%) had a past medical history of stroke or TIA. Brain MRI showed signs of cerebral small vessel disease in 58 cases (40%). α-GAL levels resulted lower than cut-off in 15 males (20.5±6.1 μmol/L/h), whereas lyso-Gb3 values were within reference range. GLA gene variants were not detected in any tested subjects.

Conclusion: Our results did not show any association between FD and PD. Further investigation is necessary to confirm these findings, including larger and more homogeneous cohorts.