Introduction: A clinical association has been reported between Parkinson Disease (PD) and Gaucher’s disease.

Aim: The aim of this study was to ascertain the frequency of GBA mutations in a cohort of Spanish PD patients and healthy age-matched controls.

Methods: We screened a total 112 PD patients and 109 controls from Aragon, Spain, for GBA mutations by complete sequencing of all exons, as well as, intron/exon boundaries, of the GBA gene.

Results: We have found 7 patients (6.3%) heterozygous for GBA mutations (2 were heterozygous for L444P, 1 was heterozygous for E326K, and 4 were heterozygous for T369M) whereas only 4 control subjects (3.4%) carried GBA mutations (2 were heterozygous for N370S and 2 were heterozygous for T369M). We observed a higher heterozygote frequency for the mutations in GBA gene in patients with PD compared with control subjects. Patients with PD had greater odds of being carriers of Gaucher’s disease than did control subjects (odds ratio, 1.72; 95% CI, 0.51 - 5.77). Patients with PD carrying GBA mutations were younger than those not carriers (age at onset: 52.6±12.77 years vs. 60.2±10.20 years; p=0.065). In the present study, the clinical characteristics of PD patients carrying GBA mutation in heterozygosis were typical for PD.

Conclusions: This study suggests that GBA is a susceptibility gene for PD and GBA mutations are associated with earlier age at onset of disease in this cohort.