Parkinson’s Disease And GBA Gene Mutations: Clinical Correlations

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Glucocerebrosidase gene (GBA) mutations confer a 5-6 fold increased risk of developing Parkinson’s disease (PD) with particular phenotypic characteristics. The aim of this study is to determine the frequency of GBA mutations in central and southern Italy patients with PD and identify distinctive clinical features of the carriers.

We screened 194 PD patients from central and southern Italy, including 57 patients from Campania, evaluating frequency of GBA mutations and age at disease onset. In the patients from Campania we also assessed severity of motor involvement, frequency of cognitive impairment, psychiatric disorders, dysautonomia and pharmacological treatment.

GBA mutations occur in 13.4% of all our patients and in 17.54% of patients from Campania. This prevalence is much higher than previous estimates, both in international and Italian studies, probably due to an underestimation of the latter. GBA mutation carriers had earlier disease onset as compared to patients without mutations (53.2±9.2 vs 58.8±9.1, p=0.008). There is not significant difference in others clinical features, but patients with GBA mutations seem to have higher frequency of bradikynesia (80% vs 53.2%, p>0.05) and lower dopaminergic load (L-dopa Equivalent Daily Dose) as compared to patients without mutations (572.6±383.5 mg/die vs 724.1±593.2 mg/die, p>0.05).

This study demonstrates a strong association between GBA mutations and early onset Parkinson’s disease. These data also suggest a high prevalence of GBA mutation carriers in central and southern Italy, especially in Campania. Further studies are necessary to confirm these data and identify distinctive clinical features of the carriers.