

Supplementary Material

The Degree of Cardiovascular Autonomic Dysfunction Is Not Different in *GBA*-Related and Idiopathic Parkinson's Disease Patients: A Case-Control Instrumental Evaluation

Supplementary Table 1. List of genes included in the targeted or virtual NGS panel.

Gene	Transcript ID	UniProt ID	RefSeq	Inheritance
<i>SNCA</i>	ENST00000394991.8	P37840	NM_000345.4	AD
<i>LRRK2</i>	ENST00000298910.12	Q5S007	NM_198578.4	AD
<i>VPS35</i>	ENST00000299138.12	Q96QK1	NM_018206.6	AD
<i>GBA</i>	ENST00000368373.8	P04062	NM_000157.4	AD, risk factor
<i>RAB39B</i>	ENST00000369454.4	Q96DA2	NM_171998.4	X-linked
<i>PRKN</i>	ENST00000366898.6	O60260	NM_004562.3	AR
<i>PINK1</i>	ENST00000321556.5	Q9BXM7	NM_032409.3	AR
<i>DJ-1</i>	ENST00000338639.10	Q99497	NM_007262.5	AR
<i>ATP13A2</i>	ENST00000326735.13	Q9NQ11	NM_022089.4	AR
<i>PLA2G6</i>	ENST00000332509.8	O60733	NM_003560.4	AR
<i>DNAJC6</i>	ENST00000371069.5	O75061	NM_001256864.2	AR
<i>SYNJ1</i>	ENST00000433931.7	J3KQV8	NM_003895.3	AR
<i>FBXO7</i>	ENST00000266087.12	Q9Y3I1	NM_012179.4	AR
<i>VPS13C</i>	ENST00000644861.2	Q709C8	NM_020821.3	AR
<i>PTRHD1</i>	ENST00000328379.6	Q6GMV3	NM_001013663.2	AR
<i>POLG</i>	ENST00000268124.11	E5KNU5 P54098	NM_002693.3	AD, AR
<i>TWINK</i>	ENST00000311916.8	Q96RR1-1	NM_021830.5	AD, AR
<i>OPAI</i>	ENST00000361510.8	O60313-10	NM_130837.3	AD, AR
<i>SLC25A46</i>	ENST00000355943.8	Q96AG3-1	NM_138773.4	AR

AD, autosomal dominant; AR, autosomal recessive

Supplementary Table 2. GBA variants identified in the study.

Patient	Nucleotide change	Amino acid change	class of variant
1	<i>c.1226A>G</i>	<i>p.Asn409Ser</i>	<i>mild</i>
2	<i>c.1271T>C</i>	<i>p.Leu424Pro</i>	<i>unknown</i>
3	<i>c.1448T>C</i>	<i>p.Leu483Pro</i>	<i>severe</i>
4	<i>c.1226A>G</i>	<i>p.Asn409Ser</i>	<i>mild</i>
5*	<i>c.1444G>A</i>	<i>p.Asp482Asn</i>	<i>unknown</i>
	<i>c.1200G>A</i>	<i>p.Met400Ile</i>	<i>unknown</i>
6	<i>c.1448T>C</i>	<i>p.Leu483Pro</i>	<i>severe</i>
7	<i>c.1093G>A</i>	<i>p.Glu365Lys</i>	<i>risk</i>
8*	<i>c.1223C>T</i>	<i>p.Thr408Met</i>	<i>risk</i>
	<i>c.1093G>A</i>	<i>p.Glu365Lys</i>	<i>risk</i>
9	<i>c.1093G>A</i>	<i>p.Glu365Lys</i>	<i>risk</i>
10	<i>c.1226A>G</i>	<i>p.Asn409Ser</i>	<i>mild</i>
11	<i>c.1226A>G</i>	<i>p.Asn409Ser</i>	<i>mild</i>
12	<i>c.1093G>A</i>	<i>p.Glu365Lys</i>	<i>risk</i>
13	<i>c.1093G>A</i>	<i>p.Glu365Lys</i>	<i>risk</i>
14	<i>c.1223C>T</i>	<i>p.Thr408Met</i>	<i>risk</i>
15	<i>c.509G>A</i>	<i>p.Arg170His</i>	<i>unknown</i>
16	<i>c.1223C>T</i>	<i>p.Thr408Met</i>	<i>risk</i>
17	<i>c.1448T>C</i>	<i>p.Leu483Pro</i>	<i>severe</i>
18	<i>c.1223C>T</i>	<i>p.Thr408Met</i>	<i>risk</i>
19	<i>c.1443C>A</i>	<i>p.Asn481Lys</i>	<i>unknown</i>
20	<i>c.1013del</i>	<i>p.Pro338Glnfs*26</i>	<i>severe</i>
21*	<i>c.1469A>G</i>	<i>p.His490Arg</i>	<i>mild</i>
	<i>c.1379G>A</i>	<i>p.Gly460Asp</i>	<i>unknown</i>
22	<i>c.508C>T</i>	<i>p.Arg170Cys</i>	<i>severe</i>
23*	<i>c.259C>T</i>	<i>p.Arg87Trp;</i>	<i>mild</i>
	<i>c.1226A>G</i>	<i>p.Asn409Ser</i>	<i>mild</i>

*variants were found in cis.

Supplementary Table 3. Neuropsychological profile of the study sample

	Total PD sample	<i>GBA</i> -mutated group	Non-mutated group	p
	69	23	46	
Years of education, <i>y</i>	11.8 ± 3.8	11.8 ± 3.2	11.8 ± 4.0	0.979
PD-MCI, <i>n</i> (%)	14 (16.9)	6 (26.1)	8 (17.4)	0.318
MMSEc	27.8 ± 2.7	28.1 ± 1.8	27.6 ± 3.3	0.682
Brief Mental Deterioration Battery	1.9 (1.3-2.5)	1.9 (1.4 – 2.3)	1.9 (1.2 – 2.5)	0.786
Rey's 15 Words:				
immediate recall	39.4 ± 8.9	39.3 ± 9.7	39.4 ± 9.6	0.968
delayed recall	7.3 ± 1.9	6.7 ± 2.1	7.5 ± 1.8	0.287
Immediate Visual Memory	19.9 (17.9-20.8)	20.2 (16.3-20.6)	19.9 (18.2-21.1)	0.410
Copy Design: simple	10.9 ± 2.4	12.3 ± 5.8	10.2 ± 1.5	0.099
Digit Span				
Forward	5.8 ± 1.0	5.9 ± 0.9	5.7 ± 1.1	0.660
Backward	4.5 ± 1.0	4.5 ± 0.4	4.4 ± 1.1	0.889
Barrage Test				
time	50.8 (41.5-69.0)	48.0 (39.0-70.0)	55.0 (42.0-68.0)	0.808
errors	0.0 (0.0-0.0)	0.0 (0.0-1.0)	0.0 (0.0-0.0)	0.432
score	11.0 (10.0-13.0)	11.0 (11.0-12.0)	12.0 (10.0-13.0)	0.874
final result	0.2 (-0.7-0.3)	0.2 (-0.7-1.1)	0.1 (-0.7-0.2)	0.440
Rey-Osterrieth Complex Figure Test				
direct copy	31.5 ± 3.8	31.0 ± 5.1	31.7 ± 3.3	0.791
delayed recall	15.9 ± 4.7	17.3 ± 5.1	15.1 ± 4.4	0.387
Simple Verbal Analogies Test	16.5 ± 2.7	16.6 ± 2.1	16.5 ± 2.9	0.839
Verbal Fluency				
Phonemic	31.6 (23.9-43.5)	31.8 (20.3-41.4)	30.8 (25.3-43.8)	0.679
Semantic	43.5 (36.0-51.0)	43.0 (40.0-50.0)	44.0 (35.0-52.0)	0.907
Stroop test				
time	18.8 (14.1-24.8)	21.8 (16.0-28.5)	17.8 (11.8-24.3)	0.083
errors	0.0 (-0.37 – 1.5)	0.2 (-0.2 – 1.6)	0.0 (-0.5 – 1.4)	0.461
Frontal Assessment Battery	16.5 (14.2-16.9)	14.7 (13.9-16.9)	16.5 (15.2-17.2)	0.397

Data are expressed as mean ± standard deviation or median (interquartile range)

Statistically significant p-values are denoted in bold (p value ≤ 0.05).

GBA, glucocerebrosidase gene; *n*, sample size; MMSEc, corrected Mini-Mental State Examination; PD-MCI, Mild Cognitive Impairment in Parkinson's Disease; PD, Parkinson's disease