Box 1. Design of the next-generation sequencing panel.

HGNC gene symbol	Inheritance	Disease onset	Role
PINK1	AR	Early-onset	Mitochondrial function/mitophagy
PRKN	AR	Early-onset	Mitochondrial function/mitophagy; ubiquitination; synaptic function
PARK7	AR	Early-onset	Infl ammation/immune system; mitochondrial function
SNCA	AD	Early-onset	Synaptic function; autophagy/lysosomal degradation; mitochondrial function
LRRK2	AD	Late-onset	Synaptic function; inflammation/immune system; autophagy/lysosomal degradation
VPS35	AD	Late-onset	Autophagy/lysosomal degradation; endocytosis
DCTN1	AD	NA	Cell division; axonal transport (including autophagosomes)
GCH1†	NA	NA	GTP binding; calcium ion binding; BH4 metabolism; metabolic pathways
GBA†	NA	NA	Inflammation/ immune system; autophagy/lysosomal degradation; metabolic pathways

Abbreviations: HGNC, HUGO Gene Nomenclature Committee; AR, autosomal recessive; AD, autosomal dominant; †, PD risk variant; NA, not applicable

Table 1. Characteristics of the PD patients.

Variable	PD (n=189)
Clinical subtype	
• Tremor-dominant (n)	43.6% (83)
• Hypokinetic-rigid dominant (n)	5.3% (10)
• PIGD (n)	50.8% (96)
No family history of PD (n)	83.6 % (158)
1st degree relative with PD (n)	11.6% (22)
2nd degree relative with PD (n)	4.8% (9)
MDS-UPDRS total score, mean (±SD)	72.8 (28.8)
HY, median (range)	3 (1-5)
SE-ADL, median (range)	80 (30-100)
BDI, mean (±SD)	14.8 (7.4)
MMSE, mean (±SD)	26.8 (3.5)

Abbrevations: BDI, Beck Depression Inventory; *HY*, Hoehn and Yahr stage; *MDS-UPDRS*, Movement Disorders Society Unified Parkinson´s Disease Rating Scale; *MMSE*, Mini Mental State Examination; *PD*, Parkinson´s disease; *PIGD*, Postural instability and gait disturbance dominant; *SE-ADL*, Schwab and England Activities of Daily Living Scale.

Table 2. Summary of genetic findings.

Gene	Chromosome	AA change	Interpretation	Count in PD	Count in controls
GBA	1	p.L396P (allele name L444P)	Pathogenic variant causing Gaucher disease	1	0
GBA	1	pT321M (allele name T369M)	Risk variant	10	3
GBA	1	p.E278K (allele name E326K)	Risk variant	6	3
GBA	1	p.L276I	Novel variant of unknown relevance	1	0
GBA	1	p.E10X	Novel pathogenic variant	1	0
GCH1	14	p.V204I	Likely pathogenic/Unknown relevance	2	0
GCH1	14	p.Q110E	Unknown relevance	1	0
LRRK2	12	p.E334K	Unknown relevance	2	1
LRRK2	12	p.R767H	Unknown relevance	1	0
LRRK2	12	p.R1514Q	Unknown relevance	4	3
PRKN	6	p.V109M	Unknown relevance	1	0
PRKN	6	p.R126W	Single heterozygous pathogenic variant	2	0
PRKN		p.A82E	Unknown relevance	0	3
PARK7	1	p.R98Q	Unknown relevance	4	0
PINK1	1	p.K186N	Unknown relevance	1	0
PINK1	1	p.G411S	Unknown relevance	1	2
PINK1	1	p.P209L	Unknown relevance	1	0
VPS35	16	p.G51S	Unknown relevance	1	1

Table 3. Summary of frequency of any rare variant found.

Gene	Prevalence in PD % (n)	Prevalence in controls % (n)	p-value
GBA	10.1 (19)	3.8 (6)	0.035*
GCH1	1.6 (3)	0 (0)	0.254
LRRK2	3.7 (7)	2.5 (4)	0.750
PRKN	1.6 (3)	1.9 (3)	1
PARK7	*2.1 (4)*	*0 (0)*	*0.128*
PINK1	1.6 (3)	1.3 (2)	1
VPS35	0.5 (1)	0.6 (1)	1

*Statistically significant difference