Whole Exome Sequencing
Gene package Parkinson, version 2, 30-9-2021

Technical information
DNA was enriched using Agilent SureSelect DNA + SureSelect OneSeq 300kb CNV Backbone + Human All Exon V7 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 10 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate and non-unique reads are excluded. Data are demultiplexed with bcl2fastq Conversion Software from Illumina. Reads are mapped to the genome using the BWA-MEM algorithm (reference: http://bio-bwa.sourceforge.net/). Sequence variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected sequence variants are filtered and annotated with Alissa Interpret software and classified with Alamut Visual. Copy variant detection is performed using the BAM multiscale reference method using depth of coverage analysis and dynamical bins in NexusClinical. The detected copy number variants are filtered and annotated with the NexusClinical software and classified using UCSC Genome Browser (NCBI37/hg19). Additionally, MPLA analysis was performed for several (fragments of) Parkinson genes (SALSA P051/P052 Parkinson probemix). It is not excluded that pathogenic variants are being missed using this technology. At this moment, there is not enough information about the sensitivity of this technique with respect to the detection of deletions and duplications of more than 5 nucleotides and of somatic mosaic mutations (all types of sequence changes).

Erasmus MC
University Medical Center Rotterdam

Dept. Clinical Genetics

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
ATP1A3	182350	100	100	99.43	95.12
ATP6AP2	300556	100	100	98.50	86.79
CHCHD2	616244	100	100	100	91.44
COMT	116790	100	100	100	100
CSF1R	164770	100	99.84	98.32	91.17
DCTN1	601143	100	99.91	98.94	93.34
DNAJC13	614334	100	100	99.87	98.28
DNAJC6	608375	100	100	100	97.10
EIF2AK2	176871	100	100	99.36	92.52
EIF4G1	600495	100	99.47	98.70	94.28
FBXO7	605648	100	98.48	91.87	91.87
FTL	134790	100	100	97.19	88.02
GBA	606463	100	100	100	100
GCH1	600225	94.02	94.02	94.02	91.38
GRN	138945	100	100	100	99.41
HTRA2	606441	100	100	100	99.38
JAM2	606870	100	91.30	90.99	85.82
LRP10	609921	100	99.96	98.33	91.38

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
LRRK2	609007	99.93	99.51	98.77	96.61
MAPT	157140	95.87	93.06	90.29	80.08
PARK7	602533	100	100	100	96.36
PDGFB	190040	100	100	100	97.03
PDGFRB	173410	100	100	99.59	94.94
PINK1	608309	98.32	93.98	88.89	76.09
PLA2G6	603604	100	100	99.74	96.69
POLG	174763	100	100	99.06	93.20
PRKN	602544	100	99.57	97.92	93.03
PSAP	176801	100	100	100	95.80
PTRHD1	617342	100	100	100	100
RAB29	603949	100	100	100	99.72
RAB39B	300774	100	100	100	100
SEMA6B	608873	99.43	88.02	80.97	71.05
SLC18A2	193001	100	100	100	98.56
SLC20A2	158378	100	100	99.58	94.67
SLC30A10	611146	100	100	100	99.38
SLC6A3	126455	100	100	99.32	95.75
SNCA	163890	100	100	100	100
SPR	182125	100	100	97.33	81.97
SYNJ1	604297	99.58	96.18	94.10	92.20
TAF1	313650	100	99.89	99.48	96.46
TH	191290	100	98.55	94.77	85.86
UCHL1	191342	100	93.64	82.45	78.15
UQCRC1	191328	100	100	100	98.62
VPS13C	608879	98.55	97.73	96.94	92.88
VPS35	601501	100	100	100	98.96

- OMIM release used: 23-9-2021
- The statistics above are based on a set of 150 samples
- % Covered 10x , 20x, 30x and 50x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x, 20x, 30x or 50x