Whole Exome Sequencing
Gene package Parkinson, version 2.1, 25-2-2022

Technical information
DNA was enriched using the Agilent SureSelectXT Human All Exon V7 capture kit and paired-end sequenced on the Illumina platform (outsourced). Sequencing data are demultiplexed with bcl2fastq2 Conversion Software from Illumina. Illumina DRAGEN Bio-IT Platform is used for read mapping to the hg19 genome and sequence variant detection. The detected sequence variants are annotated and filtered with Alissa Interpret software and classified with Alamut Visual. Copy number 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 annotated and filtered 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). The sensitivity to detect variants using this technology is not 100%; pathogenic variants could be missed. 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).

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HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
ATP1A3	182350	100	100	99.45	94.42
ATP6AP2	300556	100	100	99.59	93.78
CHCHD2	616244	100	100	100	91.78
COMT	116790	100	100	100	100
CSF1R	164770	100	99.58	97.75	90.20
DCTN1	601143	100	99.35	98.33	91.75
DNAJC13	614334	100	100	99.96	98.65
DNAJC6	608375	100	100	100	97.28
EIF2AK2	176871	100	100	99.74	95.75
EIF4G1	600495	100	99.30	98.58	93.78
FBXO7	605648	100	98.28	91.87	91.87
FTL	134790	100	100	95.12	84.71
GBA	606463	100	100	100	99.29
GCH1	600225	94.02	94.02	94.02	92.99
GRN	138945	100	100	100	97.18
HTRA2	606441	100	100	100	99.54
JAM2	606870	100	96.95	90.99	89.86
LRP10	609921	100	99.32	96.80	89.43
LRRK2	609007	100	99.70	98.95	96.98
MAPT	157140	95.53	92.66	89.69	79.70
PARK7	602533	100	100	100	98.11
PDGFB	190040	100	100	100	96.14
PDGFRB	173410	100	100	99.40	94.76

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	% covered ≥10x	% covered ≥20x	% covered ≥30x	% covered ≥50x
PINK1	608309	98.53	95.03	90.78	77.59
PLA2G6	603604	100	100	99.56	94.14
POLG	174763	100	100	98.60	90.96
PRKN	602544	100	99.08	97.31	93.27
PSAP	176801	100	100	100	95.51
PTRHD1	617342	100	100	100	100
RAB29	603949	100	100	100	99.01
RAB39B	300774	100	100	100	100
SEMA6B	608873	99.36	89.71	80.97	70.89
SLC18A2	193001	100	100	100	98.13
SLC20A2	158378	100	100	100	94.67
SLC30A10	611146	100	100	100	100
SLC6A3	126455	100	99.86	98.69	92.78
SNCA	163890	100	100	100	98.27
SPR	182125	100	100	99.05	88.20
SYNJ1	604297	99.84	97.11	94.83	92.88
TAF1	313650	100	99.95	99.77	97.63
TH	191290	100	98.04	94.21	86.67
UCHL1	191342	100	97.35	82.45	77.62
UQCRC1	191328	100	100	100	98.21
VPS13C	608879	99.07	98.02	97.41	94.36
VPS35	601501	100	100	100	99.16

- OMIM release used: 23-9-2021
- The statistics above are based on a set of 104 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