Whole Exome Sequencing
Gene package Parkinson, version 1.2, 30-9-2020

Technical information
DNA was enriched using Agilent SureSelect DNA + 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/). Variant detection is performed by the Genome Analysis Toolkit HaplotypeCaller (reference: http://www.broadinstitute.org/gatk/). The detected variants are filtered and annotated with Alissa Interpret software and classified with Alamut Visual. Additionally, MPLA analysis was performed for several (fragments of) Parkinson genes (SALSA P051/P052 Parkinson probemix). It is not excluded that pathogenic mutations 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).

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HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
ATP1A3	182350	221	100	100	100
ATP6AP2	300556	54	100	93	81
CHCHD2	616244	171	100	100	98
COMT	116790	373	100	100	100
CSF1R	164770	205	100	100	100
DCTN1	601143	162	100	100	99
DNAJC13	614334	58	99	91	78
DNAJC6	608375	125	100	100	96
EIF4G1	600495	178	100	100	99
FBXO7	605648	126	97	94	90
FTL	134790	188	100	100	100
GBA	606463	207	100	100	100
GCH1	600225	207	94	94	94
GIGYF2	612003	80	100	98	91
GRN	138945	336	100	100	100
HTRA2	606441	256	100	100	100
LRP10	609921	294	100	100	100
LRRK2	609007	65	98	92	84
MAPT	157140	170	100	100	99
PARK7	602533	76	100	99	85
PDGFB	190040	184	100	100	100
PDGFRB	173410	235	100	100	100
PINK1	608309	258	100	100	98

HGNC approved gene symbol	Phenotype description including OMIM phenotype ID(s)	median depth	% covered >10x	% covered >20x	% covered >30x
PLA2G6	603604	287	100	100	100
POLG	174763	188	100	100	100
PRKN	602544	138	100	100	99
PTRHD1	617342	264	100	100	100
RAB29	603949	102	100	100	100
RAB39B	300774	156	100	100	99
SLC18A2	193001	145	100	100	100
SLC20A2	158378	185	100	95	95
SLC30A10	611146	209	100	100	100
SLC6A3	126455	226	100	100	100
SNCA	163890	106	100	100	100
SPR	182125	142	100	100	100
SYNJ1	604297	83	98	95	90
TAF1	313650	71	100	99	95
TH	191290	266	100	100	100
UCHL1	191342	89	100	94	84
VPS13C	608879	63	96	87	77
VPS35	601501	82	100	97	91

- OMIM release used: 8-9-2019
- The statistics above are based on a set of 100 samples
- Median depth is the median of the mean sequence depth over the protein coding exons (±10bp flanking introns) of the longest transcript
- % Covered 10x , 20x and 30 x describes the percentage of a gene's coding sequence (±10bp flanking introns) that is covered at least 10x, 20x or 30x