Whole Exome Sequencing
Gene package Parkinson, version 1, 30-9-2019

Technical information
DNA was enriched using Agilent SureSelect Clinical Research Exome V2 capture and paired-end sequenced on the Illumina platform (outsourced). The aim is to obtain 8.1 Giga base pairs per exome with a mapped fraction of 0.99. The average coverage of the exome is ~50x. Duplicate 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 Cartagenia 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)	OMIM gene ID	median depth	% covered >10x	% covered >20x	% covered >30x
ATP1A3	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235	182350	144	100	100	100
ATP6AP2	Mental retardation, syndromic, Hedera type, 300423 ?Parkinsonism with spasticity, 300911	300556	46	100	91	64
CHCHD2	Parkinson disease 22, 616710	616244	67	100	88	68
COMT	{Panic disorder, susceptibility to}, 167870 {Schizophrenia, susceptibility to}, 181500	116790	145	100	100	100
CSF1R	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids, 221820	164770	99	100	100	100
DCTN1	{Amyotrophic lateral sclerosis, susceptibility to}, 105400 Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605	601143	95	100	100	99
DNAJC13	No OMIM phenotype	614334	71	100	99	94
DNAJC6	Parkinson disease 19a, juvenile-onset, 615528 Parkinson disease 19b, early-onset, 615528	608375	84	100	100	98
EIF4G1	{Parkinson disease 18}, 614251	600495	100	100	100	100
FBXO7	Parkinson disease 15, 260300	605648	86	100	100	97

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FTL	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159	134790	144	100	100	100
GBA	Gaucher disease, perinatal lethal, 608013 Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 {Lewy body dementia, susceptibility to}, 127750 {Parkinson disease, late-onset, susceptibility to}, 168600	606463	172	100	100	100
GCH1	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910	600225	52	100	100	93
GIGYF2	{Parkinson disease 11}, 607688	612003	66	100	99	94
GRN	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485	138945	155	100	100	100
HTRA2	3-methylglutaconic aciduria, type VIII, 617248 {Parkinson disease 13}, 610297	606441	143	100	100	98
LRP10	No OMIM phenotype	609921	148	100	100	100
LRRK2	{Parkinson disease 8}, 607060	609007	78	100	100	96
MAPT	Dementia, frontotemporal, with or without parkinsonism, 600274 {Parkinson disease, susceptibility to}, 168600 Pick disease, 172700 Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540	157140	120	100	100	100
PARK7	Parkinson disease 7 early-onset, 606324	602533	68	100	99	87
PDGFB	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174	190040	101	100	100	100
PDGFRB	Basal ganglia calcification, idiopathic, 4, 615007 Kosaki overgrowth syndrome, 616592 Myeloproliferative disorder with eosinophilia, 131440 Myofibromatosis, infantile, 1, 228550 Premature aging syndrome, Penttinen type, 601812	173410	116	100	100	100
PINK1	Parkinson disease 6, early onset, 605909	608309	100	98	93	88
PLA2G6	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, 612953	603604	118	100	100	99

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POLG	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia 1, 157640 Progressive external ophthalmoplegia 1, 258450	174763	114	100	100	99
PRKN	Adenocarcinoma of lung, somatic, 211980 Ovarian cancer, somatic, 167000 Parkinson disease, juvenile, type 2, 600116	602544	96	100	100	99
PTRHD1	No OMIM phenotype	617342	119	100	100	100
RAB29	No OMIM phenotype	603949	68	100	100	99
RAB39B	Mental retardation 72, 300271 Waisman syndrome, 311510	300774	59	100	100	100
SLC18A2	?Parkinsonism-dystonia, infantile, 2, 618049	193001	74	100	100	97
SLC20A2	Basal ganglia calcification, idiopathic, 1, 213600	158378	99	100	100	95
SLC30A10	Hypermanganesemia with dystonia 1, 613280	611146	148	100	100	100
SLC6A3	{Nicotine dependence, protection against}, 188890 Parkinsonism-dystonia, infantile, 1, 613135	126455	102	100	100	100
SNCA	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543	163890	52	100	100	98
SPR	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716	182125	102	100	100	100
SYNJ1	Epileptic encephalopathy, early infantile, 53, 617389 Parkinson disease 20, early-onset, 615530	604297	62	100	99	92
TAF1	Dystonia-Parkinsonism, 314250 Mental retardation, syndromic 33, 300966	313650	54	100	97	87
TH	Segawa syndrome, recessive, 605407	191290	93	100	99	94
UCHL1	{?Parkinson disease 5, susceptibility to}, 613643 Spastic paraplegia 79, 615491	191342	79	100	100	99
VPS13C	Parkinson disease 23, early onset, 616840	608879	50	100	97	85
VPS35	{Parkinson disease 17}, 614203	601501	68	100	99	94

- Gene symbols according HGCN
- OMIM release used: 8-9-2019
- "No OMIM phenotypes" indicates a gene without a current OMIM association
- OMIM phenotypes between "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values
- OMIM phenotypes between "{}", indicate risk factors
- OMIM phenotypes with a question mark, "?", before the disease name indicates an unconfirmed or possibly spurious mapping
- 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

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