PARKINSON GENE PANEL DG 2.16 (36 genes)

Releasedate: 07-06-2019

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
ATP13A2	134,1	99.9%	99.7%	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225
ATP1A3	159,8	100.0%	100.0%	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235
C19orf12	104,2	100.0%	99.8%	?Spastic paraplegia 43, autosomal recessive, 615043 Neurodegeneration with brain iron accumulation 4, 614298
CHCHD2	69,7	99.9%	93.7%	Parkinson disease 22, autosomal dominant, 616710
CHMP2B	90,2	99.5%	97.7%	Amyotrophic lateral sclerosis 17, 614696 Dementia, familial, nonspecific, 600795
CSF1R	113,3	99.9%	99.1%	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
DCTN1	112,6	99.9%	99.2%	Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605 {Amyotrophic lateral sclerosis, susceptibility to}, 105400
DNAJC6	126,5	99.9%	99.0%	Parkinson disease 19a, juvenile-onset, 615528 Parkinson disease 19b, early-onset, 615528
FBXO7	152,8	99.9%	99.6%	Parkinson disease 15, autosomal recessive, 260300
FTL	145,2	99.7%	96.7%	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
GBA	169,8	100.0%	100.0%	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
GCH1	84,8	100.0%	99.5%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GRN	174,1	100.0%	100.0%	Aphasia, primary progressive, 607485

				Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
KIAA1161	NC	NC	NC	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317
LRRK2	117,5	99.7%	97.2%	{Parkinson disease 8}, 607060
MAPT	151,6	99.9%	99.6%	Dementia, frontotemporal, with or without parkinsonism, 600274 Pick disease, 172700 Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540 {Parkinson disease, susceptibility to}, 168600
PARK7	83,5	100.0%	99.8%	Parkinson disease 7, autosomal recessive early-onset, 606324
PDGFB	115,4	100.0%	100.0%	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174
PDGFRB	126,6	99.7%	98.0%	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
PINK1	87,3	96.4%	90.7%	Parkinson disease 6, early onset, 605909
PLA2G6	111,9	99.8%	98.2%	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
POLG	113,9	100.0%	99.6%	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, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
PRKN	82,1	79.9%	78.1%	Adenocarcinoma of lung, somatic, 211980 Ovarian cancer, somatic, 167000 Parkinson disease, juvenile, type 2, 600116
PRKRA	190,7	100.0%	100.0%	Dystonia 16, 612067
PSEN1	131,5	100.0%	100.0%	?Acne inversa, familial, 3, 613737 Alzheimer disease, type 3, 607822 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Cardiomyopathy, dilated, 1U, 613694 Dementia, frontotemporal, 600274 Pick disease, 172700

SLC20A2	108,6	100.0%	98.5%	Basal ganglia calcification, idiopathic, 1, 213600
SLC30A10	176,1	100.0%	100.0%	Hypermanganesemia with dystonia 1, 613280
SLC39A14	95,4	99.9%	97.9%	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC6A3	133	100.0%	99.9%	Parkinsonism-dystonia, infantile, 1, 613135 {Nicotine dependence, protection against}, 188890
SNCA	105	100.0%	100.0%	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543
TAF1	86,8	99.1%	95.5%	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TH	96,3	100.0%	98.2%	Segawa syndrome, recessive, 605407
VPS13C	110,2	99.5%	97.0%	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS35	83,8	95.7%	88.0%	{Parkinson disease 17}, 614203
WDR45	68,7	96.8%	88.9%	Neurodegeneration with brain iron accumulation 5, 300894
XPR1	126	100.0%	99.6%	Basal ganglia calcification, idiopathic, 6, 616413

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
Median Coverage describes the average number of reads seen across 50 exomes.
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : May 8th, 2019.
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors