PARKINSON DISEASE PANEL DG-4.2.0 (36 GENES)

Gene	*Twist X2 covered 10x*	*Twist X2 covered 20x*	*srWGS covered 10x*	*srWGS covered 15x*	*srWGS covered 20x*	*Associated Phenotype description and OMIM disease ID*
ATP13A2	100%	100%	100%	99.9%	98.9%	Spastic paraplegia 78, autosomal recessive, 617225;Kufor-Rakeb syndrome, 606693
ATP1A3	100%	100%	100%	99.9%	98.3%	Alternating hemiplegia of childhood 2, 614820;Dystonia-12, 128235;CAPOS syndrome, 601338;Developmental and epileptic encephalopathy 99, 619606
C19orf12	100%	100%	100%	99.8%	97.5%	Neurodegeneration with brain iron accumulation 4, 614298;?Spastic paraplegia 43, autosomal recessive, 615043
CHCHD2	100%	100%	100%	100%	99.6%	Parkinson disease 22, autosomal dominant, 616710

CHMP2B	92.7%	88.2%	100%	99.9%	99%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 7, 600795
CSF1R	100%	100%	100%	100%	98.9%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476;Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820
DCTN1	100%	100%	100%	99.9%	98.7%	Perry syndrome, 168605;{Amyotrophic lateral sclerosis, susceptibility to}, 105400;Neuronopathy, distal hereditary motor, autosomal dominant 14, 607641
DNAJC6	100%	100%	100%	99.9%	99.4%	Parkinson disease 19a, juvenile-onset, 615528;Parkinson disease 19b, early-onset, 615528
FBXO7	100%	100%	100%	100%	99.5%	Parkinson disease 15, autosomal recessive, 260300

FTL	100%	100%	100%	99.9%	98.8%	Hyperferritinemia-cataract syndrome, 600886;L-ferritin deficiency, dominant and recessive, 615604;Neurodegeneration with brain iron accumulation 3, 606159
GBA1	100%	100%	100%	100%	99.3%	{Lewy body dementia, susceptibility to}, 127750;Gaucher disease, type II, 230900;Gaucher disease, type IIIC, 231005;Gaucher disease, type III, 231000;Gaucher disease, type I, 230800;Gaucher disease, perinatal lethal, 608013;{Parkinson disease, late-onset, susceptibility to}, 168600
GCH1	100%	100%	100%	100%	99.8%	Dystonia, DOPA-responsive, 128230;Hyperphenylalaninemia, BH4-deficient, B, 233910

GRN	100%	100%	100%	100%	99.6%	Frontotemporal dementia 2, 607485;Aphasia, primary progressive, 607485;Ceroid lipofuscinosis, neuronal, 11, 614706
LRRK2	100%	100%	100%	99.9%	99%	{Parkinson disease 8}, 607060
MAPT	95.4%	95.3%	100%	99.9%	98.5%	Supranuclear palsy, progressive, 601104;Frontotemporal dementia 1, with or without parkinsonism, 600274;Supranuclear palsy, progressive atypical, 260540;{Parkinson disease, susceptibility to}, 168600;Pick disease, 172700
MYORG	100%	100%	100%	100%	99.2%	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317
PARK7	100%	100%	100%	100%	99.7%	Parkinson disease 7, autosomal recessive early-onset, 606324

PDGFB	100%	100%	100%	100%	99.4%	Meningioma, SIS-related, 607174;Basal ganglia calcification, idiopathic, 5, 615483;Dermatofibrosarcoma protuberans, 607907
PDGFRB	100%	100%	100%	100%	98.5%	Premature aging syndrome, Penttinen type, 601812;Kosaki overgrowth syndrome, 616592;Myofibromatosis, infantile, 1, 228550;Basal ganglia calcification, idiopathic, 4, 615007;Myeloproliferative disorder with eosinophilia, 131440
PINK1	100%	100%	100%	99.9%	98.2%	Parkinson disease 6, early onset, 605909
PLA2G6	100%	99.9%	100%	100%	98.9%	Parkinson disease 14, autosomal recessive, 612953;Neurodegeneration with brain iron accumulation 2B, 610217;Infantile neuroaxonal dystrophy 1, 256600

POLG	100%	100%	100%	99.9%	99.2%	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459;Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662;Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700;Progressive external ophthalmoplegia, autosomal dominant 1, 157640;Progressive external ophthalmoplegia, autosomal recessive 1, 258450
PRKN	100%	100%	100%	99.9%	98.5%	Adenocarcinoma of lung, somatic, 211980;Parkinson disease, juvenile, type 2, 600116;Ovarian cancer, somatic, 167000
PRKRA	100%	100%	100%	100%	99.6%	Dystonia 16, 612067

PSEN1	100%	100%	100%	100%	99.6%	Pick disease, 172700;Dementia, frontotemporal, 600274;?Acne inversa, familial, 3, 613737;?Cardiomyopathy, dilated, 1U, 613694;Alzheimer disease, type 3, with or without spastic paraparesis, 607822
SLC20A2	100%	100%	100%	99.9%	98.8%	Basal ganglia calcification, idiopathic, 1, 213600
SLC30A10	100%	100%	100%	99.9%	98.6%	Hypermanganesemia with dystonia 1, 613280
SLC39A14	93.6%	93.6%	100%	100%	98.9%	?Hyperostosis cranalis interna, 144755;Hypermanganesemia with dystonia 2, 617013
SLC6A3	100%	100%	100%	99.9%	98.9%	Parkinsonism-dystonia, infantile, 1, 613135;{Nicotine dependence, protection against}, 188890
SNCA	100%	100%	100%	100%	99.4%	Dementia, Lewy body, 127750;Parkinson disease 1, 168601;Parkinson disease 4, 605543

TAF1	98.7%	98.5%	98.4%	86.5%	67.2%	Intellectual developmental disorder, X-linked syndromic 33, 300966;Dystonia-Parkinsonism, X-linked, 314250
TH	100%	100%	100%	100%	98.4%	Segawa syndrome, recessive, 605407
VPS13C	100%	100%	100%	100%	99.5%	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS35	100%	100%	100%	100%	99.7%	{Parkinson disease 17}, 614203
WDR45	100%	99.9%	98.6%	86.7%	66.5%	Neurodegeneration with brain iron accumulation 5, 300894
XPR1	100%	100%	100%	100%	99.4%	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.
TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
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srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.
This list is accurate for panel version DG 4.2.0

Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors