PARKINSON DISEASE PANEL DG-5.1.0 (34 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%	100%	99.5%	Spastic paraplegia 78, autosomal recessive, 617225;Kufor-Rakeb syndrome, 606693
ATP1A3	100%	100%	100%	100%	99.2%	Alternating hemiplegia of childhood 2, 614820;Dystonia-12, 128235;CAPOS syndrome, 601338;Developmental and epileptic encephalopathy 99, 619606
C19orf12	100%	100%	100%	99.9%	98.7%	Neurodegeneration with brain iron accumulation 4, 614298;?Spastic paraplegia 43, autosomal recessive, 615043
CHCHD2	90.1%	90.1%	100%	100%	99.8%	Parkinson disease 22, autosomal dominant, 616710
DCTN1	100%	100%	100%	100%	99%	Perry syndrome, 168605;{Amyotrophic lateral sclerosis, susceptibility to}, 105400;Neuronopathy, distal hereditary motor, autosomal dominant 14, 607641
DNAJC6	100%	100%	100%	99.9%	99.6%	Parkinson disease 19a, juvenile-onset, 615528;Parkinson disease 19b, early-onset, 615528
FBXO7	99.1%	98.1%	100%	100%	99.6%	Parkinson disease 15, autosomal recessive, 260300

FTL	100%	100%	100%	100%	99.3%	Hyperferritinemia-cataract syndrome, 600886;L-ferritin deficiency, dominant and recessive, 615604;Neurodegeneration with brain iron accumulation 3, 606159
GBA1	100%	98.3%	100%	100%	99.5%	{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%	99.5%	Dystonia, DOPA-responsive, 128230;Hyperphenylalaninemia, BH4-deficient, B, 233910
LRRK2	100%	100%	100%	100%	99.7%	{Parkinson disease 8}, 607060
MYORG	100%	100%	100%	99.9%	99.1%	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317
PANK2	100%	100%	100%	100%	99.6%	Neurodegeneration with brain iron accumulation 1, 234200
PARK7	97.5%	93.1%	100%	99.8%	99.4%	Parkinson disease 7, autosomal recessive early-onset, 606324

PDGFB	99.7%	94.7%	100%	100%	99.8%	Meningioma, SIS-related, 607174;Basal ganglia calcification, idiopathic, 5, 615483
PDGFRB	99.3%	97.4%	100%	100%	99.2%	Premature aging syndrome, Penttinen type, 601812;?Ocular pterygium-digital keloid dysplasia syndrome, 621091;Kosaki overgrowth syndrome, 616592;Myofibromatosis, infantile, 1, 228550;Basal ganglia calcification, idiopathic, 4, 615007
PINK1	96.9%	96.9%	100%	100%	99.2%	Parkinson disease 6, early onset, 605909
PLA2G6	100%	100%	100%	100%	99.4%	Parkinson disease 14, autosomal recessive, 612953;Neurodegeneration with brain iron accumulation 2B, 610217;Infantile neuroaxonal dystrophy 1, 256600
POLG	100%	100%	100%	100%	99.8%	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%	100%	99.8%	Adenocarcinoma of lung, somatic, 211980;Parkinson disease, juvenile, type 2, 600116;Ovarian cancer, somatic, 167000
PRKRA	100%	100%	100%	100%	99.8%	Dystonia 16, 612067
RAB32	100%	100%	100%	99.9%	99.3%	{Parkinson disease 26, autosomal dominant, susceptibility to}, 620923
SLC20A2	100%	99.5%	100%	100%	99.7%	Basal ganglia calcification, idiopathic, 1, 213600
SLC30A10	100%	100%	100%	100%	99.3%	Hypermanganesemia with dystonia 1, 613280
SLC39A14	94%	94%	100%	100%	99.5%	?Hyperostosis cranalis interna, 144755;Hypermanganesemia with dystonia 2, 617013
SLC6A3	93.9%	92.4%	100%	100%	99.6%	Parkinsonism-dystonia, infantile, 1, 613135;{Nicotine dependence, protection against}, 188890
SNCA	100%	100%	100%	100%	99.9%	Dementia, Lewy body, 127750;Parkinson disease 1, 168601;Parkinson disease 4, 605543
SYNJ1	100%	100%	100%	100%	99.8%	Parkinson disease 20, early-onset, 615530;Developmental and epileptic encephalopathy 53, 617389
TAF1	98.7%	98.7%	98.9%	89%	70.8%	Intellectual developmental disorder, X-linked syndromic 33, 300966;Dystonia-Parkinsonism, X-linked, 314250

TH	100%	99.8%	100%	100%	99.2%	Segawa syndrome, recessive, 605407
VPS13C	100%	100%	100%	100%	99.8%	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS35	100%	100%	100%	100%	99.9%	{Parkinson disease 17}, 614203
WDR45	98.4%	98%	98.4%	87.9%	68.6%	Neurodegeneration with brain iron accumulation 5, 300894
XPR1	100%	100%	100%	100%	99.7%	Basal ganglia calcification, idiopathic, 6, 616413

Gene symbols used follow HGNC 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 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 5.1.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors