# PARKINSON DISEASE PANEL DG 3.8.1 (36 GENES)

| *Gene* | *Twist X2 covered >10x* | *Twist X2 covered >20x* | *WGS covered >10x* | *WGS covered >20x* | *Associated Phenotype description and OMIM disease ID* |
|---|---|---|---|---|---|
| ATP13A2 | 100.0% | 100.0% | 100.0% | 99.8% | Spastic paraplegia 78, autosomal recessive, 617225;Kufor-Rakeb syndrome, 606693 |
| ATP1A3 | 100.0% | 100.0% | 100.0% | 99.5% | Alternating hemiplegia of childhood 2, 614820;Dystonia-12, 128235;CAPOS syndrome, 601338;Developmental and epileptic encephalopathy 99, 619606 |
| C19orf12 | 100.0% | 99.9% | 100.0% | 98.8% | Neurodegeneration with brain iron accumulation 4, 614298;?Spastic paraplegia 43, autosomal recessive, 615043 |
| CHCHD2 | 100.0% | 100.0% | 100.0% | 99.9% | Parkinson disease 22, autosomal dominant, 616710 |
| CHMP2B | 100.0% | 100.0% | 100.0% | 98.1% | Frontotemporal dementia and/or amyotrophic lateral sclerosis 7, 600795 |

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| CSF1R | 100.0% | 100.0% | 100.0% | 99.4% | Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476;Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820 |
| DCTN1 | 100.0% | 100.0% | 100.0% | 99.6% | Perry syndrome, 168605;{Amyotrophic lateral sclerosis, susceptibility to}, 105400;Neuronopathy, distal hereditary motor, autosomal dominant 14, 607641 |
| DNAJC6 | 100.0% | 100.0% | 100.0% | 99.3% | Parkinson disease 19a, juvenile-onset, 615528;Parkinson disease 19b, early-onset, 615528 |
| FBXO7 | 100.0% | 100.0% | 100.0% | 99.3% | Parkinson disease 15, autosomal recessive, 260300 |
| FTL | 100.0% | 100.0% | 100.0% | 99.2% | Hyperferritinemia-cataract syndrome, 600886;L-ferritin deficiency, dominant and recessive, 615604;Neurodegeneration with brain iron accumulation 3, 606159 |

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| GBA | 100.0% | 100.0% | 100.0% | 99.6% | {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 | 100.0% | 100.0% | 100.0% | 99.5% | Dystonia, DOPA-responsive, 128230;Hyperphenylalaninemia, BH4-deficient, B, 233910 |
| GRN | 100.0% | 100.0% | 100.0% | 99.8% | Aphasia, primary progressive, 607485;Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485;Ceroid lipofuscinosis, neuronal, 11, 614706 |
| LRRK2 | 100.0% | 100.0% | 100.0% | 99.1% | {Parkinson disease 8}, 607060 |

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| MAPT | 100.0% | 100.0% | 100.0% | 99.6% | Supranuclear palsy, progressive, 601104;Supranuclear palsy, progressive atypical, 260540;Dementia, frontotemporal, with or without parkinsonism, 600274;{Parkinson disease, susceptibility to}, 168600;Pick disease, 172700 |
| MYORG | 100.0% | 100.0% | 100.0% | 100.0% | Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317 |
| PARK7 | 100.0% | 100.0% | 100.0% | 99.5% | Parkinson disease 7, autosomal recessive early-onset, 606324 |
| PDGFB | 100.0% | 100.0% | 100.0% | 99.0% | Meningioma, SIS-related, 607174;Basal ganglia calcification, idiopathic, 5, 615483;Dermatofibrosarcoma protuberans, 607907 |
| PDGFRB | 100.0% | 100.0% | 100.0% | 99.8% | 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 |

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| PINK1 | 100.0% | 100.0% | 100.0% | 99.7% | Parkinson disease 6, early onset, 605909 |
| PLA2G6 | 100.0% | 99.9% | 100.0% | 99.5% | Parkinson disease 14, autosomal recessive, 612953;Neurodegeneration with brain iron accumulation 2B, 610217;Infantile neuroaxonal dystrophy 1, 256600 |
| POLG | 100.0% | 100.0% | 100.0% | 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 | 91.9% | 91.1% | 100.0% | 99.3% | Adenocarcinoma of lung, somatic, 211980;Parkinson disease, juvenile, type 2, 600116;Ovarian cancer, somatic, 167000 |
| PRKRA | 100.0% | 100.0% | 100.0% | 99.4% | Dystonia 16, 612067 |

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| PSEN1 | 100.0% | 100.0% | 100.0% | 99.6% | Pick disease, 172700;Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822;Dementia, frontotemporal, 600274;?Acne inversa, familial, 3, 613737;Cardiomyopathy, dilated, 1U, 613694;Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822;Alzheimer disease, type 3, 607822 |
| SLC20A2 | 100.0% | 100.0% | 100.0% | 99.6% | Basal ganglia calcification, idiopathic, 1, 213600 |
| SLC30A10 | 100.0% | 100.0% | 100.0% | 99.8% | Hypermanganesemia with dystonia 1, 613280 |
| SLC39A14 | 93.6% | 93.6% | 100.0% | 99.8% | ?Hyperostosis cranalis interna, 144755;Hypermanganesemia with dystonia 2, 617013 |
| SLC6A3 | 100.0% | 100.0% | 100.0% | 99.9% | Parkinsonism-dystonia, infantile, 1, 613135;{Nicotine dependence, protection against}, 188890 |
| SNCA | 100.0% | 100.0% | 100.0% | 99.5% | Dementia, Lewy body, 127750;Parkinson disease 1, 168601;Parkinson disease 4, 605543 |

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| TAF1 | 100.0% | 99.9% | 98.3% | 72.1% | Intellectual developmental disorder, X-linked syndromic 33, 300966;Dystonia-Parkinsonism, X-linked, 314250 |
| TH | 100.0% | 100.0% | 100.0% | 99.7% | Segawa syndrome, recessive, 605407 |
| VPS13C | 100.0% | 100.0% | 100.0% | 99.3% | Parkinson disease 23, autosomal recessive, early onset, 616840 |
| VPS35 | 100.0% | 100.0% | 100.0% | 99.3% | {Parkinson disease 17}, 614203 |
| WDR45 | 100.0% | 100.0% | 99.7% | 84.2% | Neurodegeneration with brain iron accumulation 5, 300894 |
| XPR1 | 100.0% | 100.0% | 100.0% | 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.*
*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.*
*srWGS GRCh38 Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.*
*srWGS GRCh38 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 : March 17th, 2023.*
*This list is accurate for panel version DG 3.8.1*