PARKINSON GENE PANEL DG 3.4.0 (36 genes)

Releasedate: 19-04-2022

Gene	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
ATP13A2	100,0%	100,0%	Spastic paraplegia 78, autosomal recessive, 617225 Kufor-Rakeb syndrome, 606693
ATP1A3	100,0%	100,0%	Alternating hemiplegia of childhood 2, 614820 Dystonia-12, 128235 CAPOS syndrome, 601338 Developmental and epileptic encephalopathy 99, 619606
C19orf12	100,0%	100,0%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
CHCHD2	100,0%	100,0%	Parkinson disease 22, autosomal dominant, 616710
CHMP2B	100,0%	100,0%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 7, 600795
CSF1R	100,0%	100,0%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids 1, 221820
DCTN1	100,0%	100,0%	Neuronopathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605
DNAJC6	100,0%	100,0%	Parkinson disease 19a, juvenile-onset, 615528 Parkinson disease 19b, early-onset, 615528
FBXO7	100,0%	100,0%	Parkinson disease 15, autosomal recessive, 260300
FTL	100,0%	100,0%	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
GBA	100,0%	100,0%	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
GCH1	100,0%	100,0%	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910
GRN	100,0%	100,0%	Aphasia, primary progressive, 607485 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706

LRRK2	100,0%	100,0%	No OMIM Disease ID
MAPT	100,0%	100,0%	Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540 Dementia, frontotemporal, with or without parkinsonism, 600274 Pick disease, 172700
MYORG	100,0%	100,0%	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317
PARK7	100,0%	100,0%	Parkinson disease 7, autosomal recessive early-onset, 606324
PDGFB	100,0%	100,0%	Meningioma, SIS-related, 607174 Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907
PDGFRB	100,0%	100,0%	Premature aging syndrome, Penttinen type, 601812 Kosaki overgrowth syndrome, 616592 Myofibromatosis, infantile, 1, 228550 Basal ganglia calcification, idiopathic, 4, 615007
PINK1	100,0%	100,0%	Parkinson disease 6, early onset, 605909
PLA2G6	92,3%	92,3%	Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217 Infantile neuroaxonal dystrophy 1, 256600
POLG	100,0%	100,0%	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	75,5%	75,3%	Adenocarcinoma of lung, somatic, 211980 Parkinson disease, juvenile, type 2, 600116 Ovarian cancer, somatic, 167000
PRKRA	100,0%	100,0%	Dystonia 16, 612067
PSEN1	100,0%	100,0%	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%	Basal ganglia calcification, idiopathic, 1, 213600
SLC30A10	100,0%	100,0%	Hypermanganesemia with dystonia 1, 613280
SLC39A14	93,6%	93,5%	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013

SLC6A3	100,0%	100,0%	Parkinsonism-dystonia, infantile, 1, 613135
SNCA	79,1%	79,1%	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543
TAF1	100,0%	100,0%	Intellectual developmental disorder, X-linked syndromic 33, 300966 Dystonia-Parkinsonism, X-linked, 314250
TH	100,0%	100,0%	Segawa syndrome, recessive, 605407
VPS13C	100,0%	100,0%	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS35	100,0%	100,0%	No OMIM Disease ID
WDR45	100,0%	100,0%	Neurodegeneration with brain iron accumulation 5, 300894
XPR1	100,0%	100,0%	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.
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TWIST is the chemistry used for WES analysis.
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 coverage denoting NC are non-protein coding genes.
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 : April 19th , 2022.
This list is accurate for panel version DG 3.4.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors