PARKINSON GENE PANEL DG 2.18 (36 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
ATP13A2	100%	99,50%	100%	100%	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225
ATP1A3	100%	99,90%	100%	100%	CAPOS syndrome, 601338 Alternating hemiplegia of childhood 2, 614820 Dystonia-12, 128235
C19orf12	100%	99,80%	100%	100%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
CHCHD2	98,40%	83,80%	100%	100%	Parkinson disease 22, autosomal dominant, 616710
CHMP2B	99,70%	96,70%	100%	100%	Amyotrophic lateral sclerosis 17, 614696 Dementia, familial, nonspecific, 600795
CSF1R	99,90%	99,30%	100%	100%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
DCTN1	100%	98,80%	100%	100%	Perry syndrome, 168605 Neuronopathy, distal hereditary motor, type VIIB, 607641
DNAJC6	100%	99,40%	100%	100%	Parkinson disease 19b, early-onset, 615528 Parkinson disease 19a, juvenile-onset, 615528
FBXO7	99,80%	97,90%	100%	100%	Parkinson disease 15, autosomal recessive, 260300
FTL	98,50%	89,40%	100%	100%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159 L-ferritin deficiency, dominant and recessive, 615604
GBA	100%	100%	100%	100%	Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 Gaucher disease, type I, 230800 Gaucher disease, perinatal lethal, 608013 Gaucher disease, type II, 230900
GCH1	99,90%	95,50%	100%	100%	Hyperphenylalaninemia, BH4-deficient, B, 233910 Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230
GRN	100%	100%	100%	100%	Ceroid lipofuscinosis, neuronal, 11, 614706 Aphasia, primary progressive, 607485 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485

Gene	Col2	Col3	Col4	Col5	Disease
KIAA1161	100%	100%	100%	100%	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317
LRRK2	99,70%	97,80%	100%	100%	No OMIM disease ID
MAPT	100%	99,50%	100%	100%	Pick disease, 172700 Dementia, frontotemporal, with or without parkinsonism, 600274 Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540
PARK7	100%	100%	100%	100%	Parkinson disease 7, autosomal recessive early-onset, 606324
PDGFB	100%	99,30%	100%	100%	Dermatofibrosarcoma protuberans, 607907 Basal ganglia calcification, idiopathic, 5, 615483 Meningioma, SIS-related, 607174
PDGFRB	99,20%	97,50%	100%	100%	Basal ganglia calcification, idiopathic, 4, 615007 Kosaki overgrowth syndrome, 616592 Myofibromatosis, infantile, 1, 228550 Premature aging syndrome, Penttinen type, 601812
TAF1	99,80%	97,70%	100%	100%	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
PINK1	90,70%	86,90%	99,90%	99,40%	Parkinson disease 6, early onset, 605909
PLA2G6	99,90%	98,30%	100%	100%	Infantile neuroaxonal dystrophy 1, 256600 Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217
POLG	100%	99,30%	100%	100%	Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
PRKN	79,80%	78,80%	89,80%	89,80%	Parkinson disease, juvenile, type 2, 600116 Ovarian cancer, somatic, 167000 Adenocarcinoma of lung, somatic, 211980
PRKRA	100%	99,40%	100%	100%	Dystonia 16, 612067
PSEN1	100%	100%	100%	100%	Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 ?Acne inversa, familial, 3, 613737 Alzheimer disease, type 3, 607822 Dementia, frontotemporal, 600274 Pick disease, 172700 Cardiomyopathy, dilated, 1U, 613694
SLC20A2	100%	99,20%	100%	100%	Basal ganglia calcification, idiopathic, 1, 213600
SLC30A10	100%	100%	100%	100%	Hypermanganesemia with dystonia 1, 613280

Gene	Covered 10x	Covered 20x			Phenotype
SLC39A14	100%	99,40%	93,50%	93,50%	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC6A3	100%	100%	100%	100%	Parkinsonism-dystonia, infantile, 1, 613135
SNCA	100%	100%	100%	100%	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543
TH	99,30%	96,10%	100%	100%	Segawa syndrome, recessive, 605407
VPS13C	99,40%	96,90%	100%	100%	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS35	97,30%	91,30%	100%	100%	No OMIM disease ID
WDR45	96,40%	89,70%	100%	100%	Neurodegeneration with brain iron accumulation 5, 300894
XPR1	100%	99,90%	100%	100%	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.

Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.

TWIST is the chemistry used for (in-house) TURBO/RAPID 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-DNA coding genes.

non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.

This list is accurate for panel version DG 2.18

Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors