PARKINSON GENE PANEL DG 2.17 (36 genes)

Releasedate: 06-12-2019

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ATP13A2	149.6	100.0%	99.8%	Kufor-Rakeb syndrome, 606693 Spastic paraplegia 78, autosomal recessive, 617225
ATP1A3	173.9	100.0%	100.0%	CAPOS syndrome, 601338 Alternating hemiplegia of childhood 2, 614820 Dystonia-12, 128235
C19orf12	117.5	100.0%	99.9%	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
CHCHD2	75.2	100.0%	96.6%	Parkinson disease 22, autosomal dominant, 616710
CHMP2B	87.0	100.0%	96.8%	Amyotrophic lateral sclerosis 17, 614696 Dementia, familial, nonspecific, 600795
CSF1R	121.9	100.0%	99.4%	Brain abnormalities, neurodegeneration, and dysosteosclerosis, 618476 Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
DCTN1	121.1	100.0%	99.5%	Perry syndrome, 168605 Neuropathy, distal hereditary motor, type VIIB, 607641
DNAJC6	131.2	100.0%	99.4%	Parkinson disease 19b, early-onset, 615528 Parkinson disease 19a, juvenile-onset, 615528
FBXO7	157.7	100.0%	99.5%	Parkinson disease 15, autosomal recessive, 260300
FTL	164.3	100.0%	98.4%	Hyperferritinemia-cataract syndrome, 600886 Neurodegeneration with brain iron accumulation 3, 606159 L-ferritin deficiency, dominant and recessive, 615604
GBA	180.2	100.0%	100.0%	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	91.0	99.9%	99.4%	Hyperphenylalaninemia, BH4-deficient, B, 233910 Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230
GRN	193.7	100.0%	100.0%	Ceroid lipofuscinosis, neuronal, 11, 614706 Aphasia, primary progressive, 607485 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
KIAA1161	284.1	100.0%	100.0%	Basal ganglia calcification, idiopathic, 7, autosomal recessive, 618317

LRRK2	115.4	99.6%	96.5%	No OMIM Disease ID
MAPT	165.9	100.0%	99.6%	Pick disease, 172700 Dementia, frontotemporal, with or without parkinsonism, 600274 Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540
PARK7	88.7	100.0%	100.0%	Parkinson disease 7, autosomal recessive early-onset, 606324
PDGFB	127.3	100.0%	100.0%	Dermatofibrosarcoma protuberans, 607907 Basal ganglia calcification, idiopathic, 5, 615483 Meningioma, SIS-related, 607174
PDGFRB	138.0	99.8%	98.2%	Kosaki overgrowth syndrome, 616592 Basal ganglia calcification, idiopathic, 4, 615007 Premature aging syndrome, Penttinen type, 601812 Myofibromatosis, infantile, 1, 228550
PINK1	96.5	97.0%	92.1%	Parkinson disease 6, early onset, 605909
PLA2G6	121.0	99.9%	98.6%	Infantile neuroaxonal dystrophy 1, 256600 Parkinson disease 14, autosomal recessive, 612953 Neurodegeneration with brain iron accumulation 2B, 610217
POLG	124.4	100.0%	99.8%	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	86.2	80.1%	78.6%	Parkinson disease, juvenile, type 2, 600116 Ovarian cancer, somatic, 167000 Adenocarcinoma of lung, somatic, 211980
PRKRA	191.8	100.0%	99.9%	Dystonia 16, 612067
PSEN1	132.4	100.0%	100.0%	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	117.5	99.9%	98.5%	Basal ganglia calcification, idiopathic, 1, 213600
SLC30A10	200.7	100.0%	100.0%	Hypermanganesemia with dystonia 1, 613280
SLC39A14	101.7	99.9%	98.8%	?Hyperostosis cranalis interna, 144755 Hypermanganesemia with dystonia 2, 617013
SLC6A3	142.7	100.0%	99.9%	Parkinsonism-dystonia, infantile, 1, 613135

SNCA	102.9	100.0%	100.0%	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543
TAF1	89.0	99.4%	96.2%	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TH	106.8	100.0%	99.2%	Segawa syndrome, recessive, 605407
VPS13C	106.4	99.4%	95.8%	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS35	83.4	95.4%	87.2%	No OMIM disease ID
WDR45	74.7	97.1%	90.6%	Neurodegeneration with brain iron accumulation 5, 300894
XPR1	126.1	100.0%	99.9%	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.
Median Coverage describes the average number of reads seen across 50 exomes.
% 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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : December 11th , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors