PARKINSON GENE PANEL DG 2.11 (35 genes)

Gene	*Median*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
ATP13A2	117.5	100	98	Kufor-Rakeb syndrome, 606693 ?Ceroid lipofuscinosis, neuronal, 12, 606693
ATP1A3	177.6	100	100	Alternating hemiplegia of childhood 2, 614820 CAPOS syndrome, 601338 Dystonia-12, 128235
C19orf12	94.3	100	99	Neurodegeneration with brain iron accumulation 4, 614298 ?Spastic paraplegia 43, autosomal recessive, 615043
CHCHD2	95.6	99	91	Parkinson disease 22, autosomal dominant, 616710
CHMP2B	91.9	98	91	Amyotrophic lateral sclerosis 17, 614696 Dementia, familial, nonspecific, 600795
CSF1R	140	99	98	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820
DCTN1	131.9	99	98	Neuropathy, distal hereditary motor, type VIIB, 607641 Perry syndrome, 168605 {Amyotrophic lateral sclerosis, susceptibility to}, 105400
DNAJC6	160.9	99	99	Parkinson disease 19, juvenile-onset, 615528
FBXO7	189.7	98	93	Parkinson disease 15, autosomal recessive, 260300
FTL	148.1	98	93	Hyperferritinemia-cataract syndrome, 600886 L-ferritin deficiency, dominant and recessive, 615604 Neurodegeneration with brain iron accumulation 3, 606159
GBA	240.7	100	100	Gaucher disease, perinatal lethal, 608013 Gaucher disease, type I, 230800 Gaucher disease, type II, 230900 Gaucher disease, type III, 231000 Gaucher disease, type IIIC, 231005 {Lewy body dementia, susceptibility to}, 127750 {Parkinson disease, late-onset, susceptibility to}, 168600
GCH1	74.4	97	86	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230 Hyperphenylalaninemia, BH4-deficient, B, 233910

GRN	184.8	100	100	Aphasia, primary progressive, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485
LRRK2	116.9	98	93	{Parkinson disease 8}, 607060
MAPT	134.4	100	98	Dementia, frontotemporal, with or without parkinsonism, 600274 Pick disease, 172700 Supranuclear palsy, progressive atypical, 260540 Supranuclear palsy, progressive, 601104 {Parkinson disease, susceptibility to}, 168600
PARK2	123.7	99	98	Adenocarcinoma of lung, somatic, 211980 Adenocarcinoma, ovarian, somatic, 167000 Parkinson disease, juvenile, type 2, 600116 {Leprosy, susceptibility to}, 607572
PARK7	86.9	100	99	Parkinson disease 7,autosomal recessive early-onset,606324
PDGFB	95.2	100	100	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174
PDGFRB	147.2	99	96	Basal ganglia calcification, idiopathic, 4, 615007 Kosaki overgrowth syndrome, 616592 Myeloproliferative disorder with eosinophilia, 131440 Myofibromatosis, infantile, 1, 228550 Premature aging syndrome, Penttinen type, 601812
PINK1	90.4	87	81	Parkinson disease 6, early onset, 605909
PLA2G6	117.6	99	98	Infantile neuroaxonal dystrophy 1, 256600 Neurodegeneration with brain iron accumulation 2B, 610217 Parkinson disease 14, autosomal recessive, 612953
POLG	114.4	100	99	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
PRKRA	178.7	99	98	Dystonia 16, 612067

PSEN1	153.1	99	98	Acne inversa, familial, 3, 613737 Alzheimer disease, type 3, 607822 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Cardiomyopathy, dilated, 1U, 613694 Dementia, frontotemporal, 600274 Pick disease, 172700
SLC20A2	119.1	99	97	Basal ganglia calcification, idiopathic, 1, 213600
SLC30A3	102.1	100	99	No OMIM phenotype ?Alzheimer disease, early-onset (Rovelet-Lecrux (2012) Eur J Hum Genet 20,613) ?Landau-Kleffner syndrome (Conroy (2014) Epilepsia 55,858)
SLC39A14	107.9	99	98	Hypermanganesemia with dystonia 2, 617013
SLC6A3	146	100	99	Parkinsonism-dystonia, infantile, 613135 {Nicotine dependence, protection against}, 188890
SNCA	129.8	100	100	Dementia, Lewy body, 127750 Parkinson disease 1, 168601 Parkinson disease 4, 605543
TAF1	113.3	99	96	Dystonia-Parkinsonism, X-linked, 314250 Mental retardation, X-linked, syndromic 33, 300966
TH	68.4	97	88	Segawa syndrome, recessive, 605407
VPS13C	106.5	96	90	Parkinson disease 23, autosomal recessive, early onset, 616840
VPS35	94	96	89	{Parkinson disease 17}, 614203
WDR45	75.6	97	90	Neurodegeneration with brain iron acculation 5, 300894
XPR1	131.6	100	99	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 : April 14th, 2017.
This list is accurate for panel version DG 2.11
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors