

# PARKINSON GENE PANEL DG 2.9 / DG 2.10

( 34 genes)

| Gene     | Median coverage | % coverage > 10x | % coverage > 20x | Associated Phenotype description and OMIM disease ID  |
|----------|-----------------|------------------|------------------|---|
| ATP13A2  | 129.3           | 100%             | 99%              | Kufor-Rakeb syndrome, 606693<br>?Ceroid lipofuscinosis, neuronal, 12, 606693  |
| ATP1A3   | 204.3           | 100%             | 100%             | Alternating hemiplegia of childhood 2, 614820<br>CAPOS syndrome, 601338<br>Dystonia-12, 128235  |
| C19orf12 | 110.3           | 100%             | 99%              | Neurodegeneration with brain iron accumulation 4, 614298<br>?Spastic paraplegia 43, autosomal recessive, 615043   |
| CHCHD2   | 93              | 97%              | 90%              | Parkinson disease 22, autosomal dominant, 616710  |
| CHMP2B   | 112.3           | 99%              | 95%              | Amyotrophic lateral sclerosis 17, 614696<br>Dementia, familial, nonspecific, 600795   |
| CSF1R    | 147.8           | 99%              | 98%              | Leukoencephalopathy, diffuse hereditary, with spheroids, 221820   |
| DCTN1    | 142.6           | 99%              | 98%              | Neuropathy, distal hereditary motor, type VIIB, 607641<br>Perry syndrome, 168605<br>{Amyotrophic lateral sclerosis, susceptibility to}, 105400  |
| DNAJC6   | 187             | 99%              | 98%              | Parkinson disease 19, juvenile-onset, 615528  |
| FBXO7    | 224.2           | 99%              | 98%              | Parkinson disease 15, autosomal recessive, 260300   |
| FTL      | 149.2           | 99%              | 93%              | Hyperferritinemia-cataract syndrome, 600886<br>L-ferritin deficiency, dominant and recessive, 615604<br>Neurodegeneration with brain iron accumulation 3, 606159                      |
| GCH1     | 92.5            | 97%              | 89%              | Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230<br>Hyperphenylalaninemia, BH4-deficient, B, 233910   |
| GRN      | 214.6           | 100%             | 100%             | Aphasia, primary progressive, 607485<br>Ceroid lipofuscinosis, neuronal, 11, 614706<br>Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485                   |
| LRRK2    | 140             | 99%              | 96%              | {Parkinson disease 8}, 607060   |
| MAPT     | 100             | 98%              | 93%              | Dementia, frontotemporal, with or without parkinsonism, 600274<br>Pick disease, 172700<br>Supranuclear palsy, progressive atypical, 260540<br>Supranuclear palsy, progressive, 601104 |

|         |       |      |      |   |
|---------|-------|------|------|---|
|         |       |      |      | {Parkinson disease, susceptibility to}, 168600  |
| PARK2   | 142.8 | 99%  | 99%  | Adenocarcinoma of lung, somatic, 211980<br>Adenocarcinoma, ovarian, somatic, 167000<br>Parkinson disease, juvenile, type 2, 600116<br>{Leprosy, susceptibility to}, 607572  |
| PARK7   | 88.1  | 99%  | 98%  | Parkinson disease 7,autosomal recessive early-onset,606324  |
| PDGFB   | 110.3 | 100% | 100% | Basal ganglia calcification, idiopathic, 5, 615483<br>Dermatofibrosarcoma protuberans, 607907<br>Meningioma, SIS-related, 607174  |
| PDGFRB  | 160.1 | 98%  | 95%  | Basal ganglia calcification, idiopathic, 4, 615007<br>Kosaki overgrowth syndrome, 616592<br>Myeloproliferative disorder with eosinophilia, 131440<br>Myofibromatosis, infantile, 1, 228550<br>Premature aging syndrome, Penttinen type, 601812  |
| PINK1   | 98    | 93%  | 89%  | Parkinson disease 6, early onset, 605909  |
| PLA2G6  | 135.5 | 99%  | 97%  | Infantile neuroaxonal dystrophy 1, 256600<br>Neurodegeneration with brain iron accumulation 2B, 610217<br>Parkinson disease 14, autosomal recessive, 612953   |
| POLG    | 128.1 | 100% | 99%  | Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700<br>Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662<br>Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459<br>Progressive external ophthalmoplegia, autosomal dominant 1, 157640<br>Progressive external ophthalmoplegia, autosomal recessive 1, 258450 |
| PRKRA   | 179.8 | 99%  | 99%  | Dystonia 16, 612067   |
| PSEN1   | 156.6 | 99%  | 97%  | Acne inversa, familial, 3, 613737<br>Alzheimer disease, type 3, 607822<br>Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822<br>Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822<br>Cardiomyopathy, dilated, 1U, 613694<br>Dementia, frontotemporal, 600274<br>Pick disease, 172700                 |
| SLC20A2 | 126.4 | 99%  | 97%  | Basal ganglia calcification, idiopathic, 1, 213600  |
| SLC30A3 | 100.1 | 99%  | 97%  | No OMIM phenotype<br>?Alzheimer disease, early-onset (Rovelet-Lecrux (2012) Eur J Hum Genet 20,613)<br>?Landau-Kleffner syndrome (Conroy (2014) Epilepsia 55,858)   |

|          |       |      |      |   |
|----------|-------|------|------|---|
| SLC39A14 | 120.5 | 99%  | 97%  | Hyper manganeseemia with dystonia 2, 617013   |
| SLC6A3   | 150.7 | 100% | 99%  | Parkinsonism-dystonia, infantile, 613135<br>{Nicotine dependence, protection against}, 188890 |
| SNCA     | 158.8 | 100% | 100% | Dementia, Lewy body, 127750<br>Parkinson disease 1, 168601<br>Parkinson disease 4, 605543     |
| TAF1     | 133.9 | 99%  | 97%  | Dystonia-Parkinsonism, X-linked, 314250<br>Mental retardation, X-linked, syndromic 33, 300966 |
| TH       | 87.6  | 97%  | 93%  | Segawa syndrome, recessive, 605407  |
| VPS13C   | 130.4 | 98%  | 94%  | Parkinson disease 23, autosomal recessive, early onset, 616840                                |
| VPS35    | 112.5 | 98%  | 92%  | {Parkinson disease 17}, 614203  |
| WDR45    | 85.2  | 95%  | 89%  | Neurodegeneration with brain iron accumulation 5, 300894                                      |
| XPR1     | 171.5 | 99%  | 99%  | Basal ganglia calcification, idiopathic, 6, 616413  |

Gene symbols used follow HGNC 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 14<sup>th</sup> 2017

This list is accurate for panel version DG 2.9 and DG 2.10

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

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