# PANEL DE LA ENFERMEDAD DE PARKINSON

Nuestro **panel de la enfermedad de Parkinson (EP)** identifica todas las variantes genéticas fisiopatológicamente relevantes para el desarrollo y el tratamiento de la EP. Los rasgos característicos de la EP incluyen la pérdida neuronal en áreas específicas de la sustancia negra y la acumulación generalizada de proteína intracelular α-sinucleína. La enfermedad se caracteriza por tres síntomas motores centrales: temblor, rigidez muscular y bradicinesia.

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| **Nº de genes:** | 115 |
| **Entrega:** | 25 días |
| **Cobertura:** | ≥99.5% ≥20x<br>Cobertura media con profundidad ≥150x |
| **Detalles:** | Análisis CNV incluido |

## SÍNDROMES Y TRASTORNOS COMUNES CUBIERTOS

**Enfermedad de Parkinson**

| Genes | OMIM (Gen) | Enfermedades asociadas (OMIM) | Herencia |
|---|---|---|---|
| APP | 104760 | Alzheimer disease 1, familial;Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants | AD |
| ATP13A2 | 610513 | Spastic paraplegia 78, AR;Kufor-Rakeb syndrome | AR |
| ATP1A3 | 182350 | Alternating hemiplegia of childhood 2;Dystonia-12;Developmental and epileptic encephalopathy 99;CAPOS syndrome | AD |
| ATP6AP2 | 300556 | Mental retardation, XL, syndromic, Hedera type;?Parkinsonism with spasticity, XL;Congenital disorder of glycosylation, type IIr | XLR |
| ATP7B | 606882 | Wilson disease | AR |
| C19orf12 | 614297 | Neurodegeneration with brain iron accumulation 4;Spastic paraplegia 43, AR | AD, AR, AR |

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| CHCHD10 | 615903 | Myopathy, isolated Mi, AD;Spinal muscular atrophy, Jokela type;Frontotemporal dementia and/or amyotrophic lateral sclerosis 2 | AD |
| CHCHD2 | 616244 | Parkinson disease 22, AD | AD |
| CLN3 | 607042 | Ceroid lipofuscinosis, neuronal, 3 | AR |
| COASY | 609855 | Neurodegeneration with brain iron accumulation 6;Pontocerebellar hypoplasia, type 12 | AR |
| CP | 117700 | Cerebellar ataxia;Hemosiderosis, systemic, due to aceruloplasminemia;[Hypoceruloplasminemia, hereditary] | AR |
| CSF1R | 164770 | Brain abnormalities, neurodegeneration, and dysosteosclerosis;Leukoencephalopathy, diffuse hereditary, with spheroids | AR, AD |
| CYP27A1 | 606530 | Cerebrotendinous xanthomatosis | AR |
| DCAF17 | 612515 | Woodhouse-Sakati syndrome | AR |
| DCTN1 | 601143 | Perry syndrome;Neuronopathy, distal hereditary motor, type VIIB;Amyotrophic lateral sclerosis, susceptibility to | AD, AD, AR |
| DNAJB2 | 604139 | Spinal muscular atrophy, distal, AR, 5 | AR |
| DNAJC12 | 606060 | Hyperphenylalaninemia, mild, non-BH4-deficient | AR |
| DNAJC13 | 614334 | | |
| DNAJC5 | 611203 | Ceroid lipofuscinosis, neuronal, 4, Parry type | AD |
| DNAJC6 | 608375 | Parkinson disease 19b, early-onset;Parkinson disease 19a, juvenile-onset | AR |
| EIF4G1 | 600495 | Parkinson disease 18 | AD |
| FBXO7 | 605648 | Parkinson disease 15, AR | AR |
| FTL | 134790 | Neurodegeneration with brain iron accumulation 3;Hyperferritinemia-cataract syndrome;L-ferritin deficiency, dominant and recessive | AD, AD, AR |
| FUS | 137070 | Essential tremor, hereditary, 4;Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia | AD |

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| GBA | 606463 | Lewy body dementia, susceptibility to;Gaucher disease, type IIIC;Parkinson disease, late-onset, susceptibility to;Gaucher disease, type II;Gaucher disease, type III;Gaucher disease, perinatal lethal;Gaucher disease, type I | AD, AR, AD, mi |
| GCH1 | 600225 | Dystonia, DOPA-responsive, with or without hyperphenylalaninemia;Hyperphenylalaninemia, BH4-deficient, B | AD, AR, AR |
| GIGYF2 | 612003 | Parkinson disease 11 | |
| GRN | 138945 | Ceroid lipofuscinosis, neuronal, 11;Frontotemporal lobar degeneration with ubiquitin-positive inclusions;Aphasia, primary progressive | AR, AD |
| HTRA2 | 606441 | Parkinson disease 13;3-methylglutaconic aciduria, type VIII | AR |
| JAM2 | 606870 | Basal ganglia calcification, idiopathic, 8, AR | AR |
| KIF5A | 602821 | Amyotrophic lateral sclerosis, susceptibility to, 25;Myoclonus, intractable, neonatal;Spastic paraplegia 10, AD | AD |
| LRP10 | 609921 | | |
| LRRK2 | 609007 | Parkinson disease 8 | AD |
| LYST | 606897 | Chediak-Higashi syndrome | AR |
| MAPT | 157140 | Parkinson disease, susceptibility to;Pick disease;Dementia, frontotemporal, with or without parkinsonism;Supranuclear palsy, progressive;Supranuclear palsy, progressive atypical | AD, mi, AD, AR |
| MT-ATP6 | | | |
| MT-ATP8 | | | |
| MT-CO1 | | | |
| MT-CO2 | | | |
| MT-CO3 | | | |
| MT-CYB | | | |
| MT-ND1 | | | |
| MT-ND2 | | | |
| MT-ND3 | | | |
| MT-ND4 | | | |
| MT-ND4L | | | |

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| MT-ND5 | | | |
| MT-ND6 | | | |
| MT-RNR1 | | | |
| MT-RNR2 | | | |
| MT-TA | | | |
| MT-TC | | | |
| MT-TD | | | |
| MT-TE | | | |
| MT-TF | | | |
| MT-TG | | | |
| MT-TH | | | |
| MT-TI | | | |
| MT-TK | | | |
| MT-TL1 | | | |
| MT-TL2 | | | |
| MT-TM | | | |
| MT-TN | | | |
| MT-TP | | | |
| MT-TQ | | | |
| MT-TR | | | |
| MT-TS1 | | | |
| MT-TS2 | | | |
| MT-TT | | | |
| MT-TV | | | |
| MT-TW | | | |
| MT-TY | | | |
| MYORG | 618255 | Basal ganglia calcification, idiopathic, 7, AR | AR |
| NPC1 | 607623 | Niemann-Pick disease, type C1;Niemann-Pick disease, type D | AR |
| NPC2 | 601015 | Niemann-pick disease, type C2 | AR |
| NUS1 | 610463 | ?Congenital disorder of glycosylation, type 1aa;Mental retardation, AD 55, with seizures | AR, AD |
| PANK2 | 606157 | HARP syndrome;Neurodegeneration with brain iron accumulation 1 | AR |
| PARK7 | 602533 | Parkinson disease 7, AR early-onset | AR |
| PDE10A | 610652 | Dyskinesia, limb and orofacial, infantile-onset;Striatal degeneration, AD | AR, AD |
| PDE8B | 603390 | Striatal degeneration, AD;Pigmented nodular adrenocortical disease, primary, 3 | AD |

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| PDGFB | 190040 | Meningioma, SIS-related;Basal ganglia calcification, idiopathic, 5;Dermatofibrosarcoma protuberans | AD |
| PDGFRB | 173410 | Myeloproliferative disorder with eosinophilia;Myofibromatosis, infantile, 1;Premature aging syndrome, Penttinen type;Basal ganglia calcification, idiopathic, 4;Kosaki overgrowth syndrome | AD |
| PINK1 | 608309 | Parkinson disease 6, early onset | AR |
| PLA2G6 | 603604 | Neurodegeneration with brain iron accumulation 2B;Parkinson disease 14, AR;Infantile neuroaxonal dystrophy 1 | AR |
| POLG | 174763 | Progressive external ophthalmoplegia, AR 1;Progressive external ophthalmoplegia, AD 1;Mi recessive ataxia syndrome (includes SANDO and SCAE);Mi DNA depletion syndrome 4B (MNGIE type);Mi DNA depletion syndrome 4A (Alpers type) | AR, AD |
| POLG2 | 604983 | Mi DNA depletion syndrome 16 (hepatic type);?Mi DNA depletion syndrome 16B (neuroophthalmic type);Progressive external ophthalmoplegia with Mi DNA deletions, AD 4 | AR, AD |
| PRKN | 602544 | Parkinson disease, juvenile, type 2;Ovarian cancer, somatic;Adenocarcinoma of lung, somatic | AR |
| PRKRA | 603424 | Dystonia 16 | AR |
| PSEN1 | 104311 | Cardiomyopathy, dilated, 1U;Pick disease;Alzheimer disease, type 3;Alzheimer disease, type 3, with spastic paraparesis and apraxia;?Acne inversa, familial, 3;Dementia, frontotemporal;Alzheimer disease, type 3, with spastic paraparesis and unusual plaques | AD |
| PSEN2 | 600759 | Alzheimer disease-4;Cardiomyopathy, dilated, 1V | AD |
| PTS | 612719 | Hyperphenylalaninemia, BH4-deficient, A | AR |
| RAB39B | 300774 | Mental retardation, XL 72;Waisman syndrome | XLR |
| SLC20A2 | 158378 | Basal ganglia calcification, idiopathic, 1 | AD |
| SLC30A10 | 611146 | Hypermanganesemia with dystonia 1 | AR |

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| SLC39A14 | 608736 | ?Hyperostosis cranalis interna;Hypermanganesemia with dystonia 2 | AD, AR |
| SLC6A3 | 126455 | Nicotine dependence, protection against;Parkinsonism-dystonia, infantile, 1 | AR |
| SMPD1 | 607608 | Niemann-Pick disease, type A;Niemann-Pick disease, type B | AR |
| SNCA | 163890 | Parkinson disease 1;Dementia, Lewy body;Parkinson disease 4 | AD |
| SNCB | 602569 | Dementia, Lewy body | AD |
| SPG11 | 610844 | Spastic paraplegia 11, AR;Amyotrophic lateral sclerosis 5, juvenile;Charcot-Marie-Tooth disease, axonal, type 2X | AR |
| SPR | 182125 | Dystonia, dopa-responsive, due to sepiapterin reductase deficiency | ?AD, AR |
| SYNJ1 | 604297 | Developmental and epileptic encephalopathy 53;Parkinson disease 20, early-onset | AR |
| TAF1 | 313650 | Mental retardation, XL, syndromic 33;Dystonia-Parkinsonism, XL | XLR |
| TARDBP | 605078 | Frontotemporal lobar degeneration, TARDBP-related;Amyotrophic lateral sclerosis 10, with or without FTD | AD |
| TENM4 | 610084 | Essential tremor, hereditary, 5 | AD |
| TH | 191290 | Segawa syndrome, recessive | AR |
| TMEM230 | 617019 | | |
| TWNK | 606075 | Progressive external ophthalmoplegia with Mi DNA deletions, AD 3;Perrault syndrome 5;Mi DNA depletion syndrome 7 (hepatocerebral type) | AD, AR |
| UCHL1 | 191342 | ?Parkinson disease 5, susceptibility to;Spastic paraplegia 79, AR | AD, AR |
| VPS13A | 605978 | Choreoacanthocytosis | AR |
| VPS13C | 608879 | Parkinson disease 23, AR, early onset | AR |
| VPS35 | 601501 | Parkinson disease 17 | AD |
| WDR45 | 300526 | Neurodegeneration with brain iron accumulation 5 | XLD |
| XPR1 | 605237 | Basal ganglia calcification, idiopathic, 6 | AD |