

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.

Nº de genes:	115
Entrega:	25 días
Cobertura:	$\geq 99.5\% \geq 20x$ Cobertura media con profundidad $\geq 150x$
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

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

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			

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

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	Hyper manganeseemia with dystonia 1	AR

SLC39A14	608736	?Hyperostosis cranialis interna;Hyper-manganesemia 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