Panel de esclerosis lateral amiotrófica (ELA)/demencia

Nuestro panel de esclerosis lateral amiotrófica (ELA)/demencia incluye genes que causan la enfermedad de Alzheimer, la demencia y la demencia frontotemporal, así como genes utilizados para el diagnóstico diferencial con superposición en cualquier punto de la historia natural de la enfermedad. Los genes dentro de este panel se han seleccionado cuidadosamente para aumentar el rendimiento del diagnóstico. Se incluyen enfermedades procesables que se superponen con el fenotipo (como la enfermedad de Wilson, la enfermedad de Niemann-Pick y la deficiencia de hexosaminidasa A). Este panel no detecta la enfermedad de Huntington.

Nº de genes:	105
Entrega:	25 días
Cobertura:	≥99.5% ≥20x Cobertura media con profundidad ≥150 x
Detalles:	Análisis de CNV incluido Análisis de Expansión de repetición: C9ORF72, PRNP

SÍNDROMES Y TRASTORNOS COMUNES CUBIERTOS

Enfermedad de alzheimer
Demencia
Demencia frontotemporal
Deficiencia de hexosaminidasa A
Enfermedad de Niemann-Pick
Enfermedad de Wilson

Resumen de genes y enfermedades asociadas (OMIM) incluidos en este panel:

Genes	OMIM (Genes)	Enfermedades asociadas (OMIM)	Herencia
ABCA7	605414	Alzheimer disease 9, susceptibility to	AD
ALS2	606352	Amyotrophic lateral sclerosis 2, juvenile;Spastic paralysis, infantile onset ascending;Primary lateral sclerosis, juvenile	AR
ANG	105850	Amyotrophic lateral sclerosis 9	

ANXA11	602572	Amytrophic lateral sclerosis 23	AD
APOE	107741	Sea-blue histiocyte disease;Lipoprotein glomerulopathy;?Alzheimer disease, protection against, due to APOE3-Christchurch;Hyperlipoproteinemia, type III;Coronary artery disease, severe, susceptibility to;?Macular degeneration, age-related;Alzheimer disease 2	AR, AD
APP	104760	Alzheimer disease 1, familial;Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants	AD
ARSA	607574	Metachromatic leukodystrophy	AR
ATL1	606439	Spastic paraplegia 3A, AD;Neuropathy, hereditary sensory, type ID	AD
ATP7B	606882	Wilson disease	AR
ATXN2	601517	Spinocerebellar ataxia 2;Parkinson disease, late-onset, susceptibility to;Amyotrophic lateral sclerosis, susceptibility to, 13	AD, AD, mi
BSCL2	606158	Lipodystrophy, congenital generalized, type 2;Encephalopathy, progressive, with or without lipodystrophy;Silver spastic paraplegia syndrome;Neuropathy, distal hereditary motor, type VC	AR, AD
C9orf72	614260	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1	AD
CCNF	600227	Frontotemporal dementia and/or amyotrophic lateral sclerosis 5	
CHCHD10	615903	?Myopathy, isolated Mi, AD;Spinal muscular atrophy, Jokela type;Frontotemporal dementia and/or amyotrophic lateral sclerosis 2	AD
CHMP2B	609512	Frontotemporal dementia and/or amytrophic lateral sclerosis 7	AD
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
CYLD	605018	Trichoepithelioma, multiple familial, 1;?Frontotemporal dementia and/or amytrophic lateral sclerosis 8;Cylindromatosis, familial;Brooke-Spiegler syndrome	AD

CYP27A1	606530	Cerebrotendinous xanthomatosis	AR
DCTN1	601143	Perry syndrome;Neuronopathy, distal hereditary motor, type VIIB;Amyotrophic lateral sclerosis, susceptibility to	AD, AD, AR
ERBB4	600543	Amyotrophic lateral sclerosis 19	AD
EWSR1	133450	Ewing sarcoma;Neuroepithelioma	
FIG4	609390	Amyotrophic lateral sclerosis 11;Yunis-Varon syndrome;Charcot-Marie-Tooth disease, type 4J;?Polymicrogyria, bilateral temporooccipital	AD, 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
GLE1	603371	Congenital arthrogryposis with anterior horn cell disease;Lethal congenital contracture syndrome 1	AR
GRN	138945	Ceroid lipofuscinosis, neuronal, 11;Frontotemporal lobar degeneration with ubiquitin-positive inclusions;Aphasia, primary progressive	AR, AD
HEXA	606869	Tay-Sachs disease;[Hex A pseudodeficiency];GM2-gangliosidosis, several forms	AR
HNRNPA1	164017	Amyotrophic lateral sclerosis 20;?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3	AD
HNRNPA2B1	600124	?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2	AD
HSPD1	118190	Leukodystrophy, hypomyelinating, 4;Spastic paraplegia 13, AD	AR, AD
ITM2B	603904	Dementia, familial British;?Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities;Dementia, familial Danish	AD
KIF5A	602821	Amyotrophic lateral sclerosis, susceptibility to, 25;Myoclonus, intractable, neonatal;Spastic paraplegia 10, AD	AD

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
MATR3	164015	Amyotrophic lateral sclerosis 21	AD
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			
NEFH	162230	Charcot-Marie-Tooth disease, axonal, type 2CC;?Amyotrophic lateral sclerosis, susceptibility to	AD, AD, AR
NEK1	604588	Short-rib thoracic dysplasia 6 with or without polydactyly;Amyotrophic lateral sclerosis, susceptibility to, 24	AR, DR, AD
NOTCH3	600276	Myofibromatosis, infantile 2;Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1;Lateral meningocele syndrome	AD
NPC1	607623	Niemann-Pick disease, type C1;Niemann-Pick disease, type D	AR
OPTN	602432	Glaucoma, normal tension, susceptibility to;Glaucoma 1, open angle, E;Amyotrophic lateral sclerosis 12 with or without frontotemporal dementia	AD
PANK2	606157	HARP syndrome;Neurodegeneration with brain iron accumulation 1	AR
PFN1	176610	Amyotrophic lateral sclerosis 18	
PRNP	176640	Insomnia, fatal familial;Creutzfeldt-Jakob disease;Gerstmann-Straussler disease;Prion disease with protracted course;Kuru, susceptibility to;Cerebral amyloid angiopathy, PRNP-related;Huntington disease-like 1	AD
PRPH	170710	Amyotrophic lateral sclerosis, susceptibility to	AD, 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
REEP1	609139	Spastic paraplegia 31, AD;?Neuronopathy, distal hereditary motor, type VB	AD
SETX	608465	Spinocerebellar ataxia, AR, with axonal neuropathy 2;Amyotrophic lateral sclerosis 4, juvenile	AR, AD
SIGMAR1	601978	?Amyotrophic lateral sclerosis 16, juvenile;?Spinal muscular atrophy, distal, AR, 2	AR

SLC52A3	613350	?Fazio-Londe disease;Brown-Vialetto-Van Laere syndrome 1	AR
SNCA	163890	Parkinson disease 1;Dementia, Lewy body;Parkinson disease 4	AD
SOD1	147450	Spastic tetraplegia and axial hypotonia, progressive;Amyotrophic lateral sclerosis 1	AR, AD, AR
SORL1	602005		
SPAST	604277	Spastic paraplegia 4, AD	AD
SPG11	610844	Spastic paraplegia 11, AR;Amyotrophic lateral sclerosis 5, juvenile;Charcot-Marie-Tooth disease, axonal, type 2X	AR
SQSTM1	601530	Paget disease of bone 3;Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset;Myopathy, distal, with rimmed vacuoles;Frontotemporal dementia and/or amyotrophic lateral sclerosis 3	AD, AR
TAF15	601574	Chondrosarcoma, extraskeletal myxoid	
TARDBP	605078	Frontotemporal lobar degeneration, TARDBP-related;Amyotrophic lateral sclerosis 10, with or without FTD	AD
TBK1	604834	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4;Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8	AD
TFG	602498	Hereditary motor and sensory neuropathy, Okinawa type;?Spastic paraplegia 57, AR	AD, AR
TREM2	605086	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2	AR
TUBA4A	191110	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia	AD
TYROBP	604142	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1	AR
UBE3A	601623	Angelman syndrome	AD
UBQLN2	300264	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia	XLD
VAPB	605704	Spinal muscular atrophy, late-onset, Finkel type;Amyotrophic lateral sclerosis 8	AD
VCP	601023	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1;Charcot-Marie-Tooth disease, type 2Y;Frontotemporal dementia and/or amyotrophic lateral sclerosis 6	AD

WASHC5	610657	Spastic paraplegia 8, AD;Ritscher-Schinzel syndrome 1	AD, AR
C9orf72	614260	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1	AD
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