AMYOTROPHIC LATERAL SCLEROSIS, ALS PANEL DG-5.1.0 (27 GENES)

Gene	*Twist X2 covered 10x*	*Twist X2 covered 20x*	*srWGS covered 10x*	*srWGS covered 15x*	*srWGS covered 20x*	*Associated Phenotype description and OMIM disease ID*
ALS2	97%	96.3%	100%	100%	99.8%	Primary lateral sclerosis, juvenile, 606353;Spastic paralysis, infantile onset ascending, 607225;Amyotrophic lateral sclerosis 2, juvenile, 205100
ANG	100%	100%	100%	100%	99.1%	Amyotrophic lateral sclerosis 9, 611895
ANXA11	100%	98.9%	100%	100%	99.5%	Amyotrophic lateral sclerosis 23, 617839;Inclusion body myopathy and brain white matter abnormalities, 619733
CFAP410	100%	100%	100%	100%	99.3%	Retinal dystrophy with macular staphyloma, 617547;Spondylometaphyseal dysplasia, axial, 602271
CHCHD10	100%	100%	100%	100%	99.5%	?Myopathy, isolated mitochondrial, autosomal dominant, 616209;Spinal muscular atrophy, Jokela type, 615048;Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911
CHMP2B	90.8%	88.1%	100%	100%	99.7%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 7, 600795
ERBB4	100%	100%	100%	100%	99.9%	Amyotrophic lateral sclerosis 19, 615515

FIG4	98.4%	98.4%	100%	100%	99.9%	Yunis-Varon syndrome, 216340;?Polymicrogyria, bilateral temporooccipital, 612691;Amyotrophic lateral sclerosis 11, 612577;Charcot-Marie-Tooth disease, type 4J, 611228
FUS	100%	100%	100%	100%	99.2%	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030;Essential tremor, hereditary, 4, 614782
GRN	100%	100%	100%	100%	99.7%	Frontotemporal dementia 2, 607485;Aphasia, primary progressive, 607485;Ceroid lipofuscinosis, neuronal, 11, 614706
KIF5A	100%	100%	100%	100%	99.5%	Myoclonus, intractable, neonatal, 617235;{Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921;Spastic paraplegia 10, autosomal dominant, 604187
MAPT	95.4%	95.4%	100%	99.9%	99.2%	Supranuclear palsy, progressive, 601104;Frontotemporal dementia 1, with or without parkinsonism, 600274;Supranuclear palsy, progressive atypical, 260540;{Parkinson disease, susceptibility to}, 168600;Pick disease, 172700
MATR3	100%	100%	100%	100%	99.6%	Amyotrophic lateral sclerosis 21, 606070

NEK1	100%	100%	100%	100%	99.8%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520;?Orofaciodigital syndrome II, 252100;{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
OPTN	100%	100%	100%	100%	99.8%	Glaucoma 1, open angle, E, 137760;Amyotrophic lateral sclerosis 12 with or without frontotemporal dementia, 613435;{Glaucoma, normal tension, susceptibility to}, 606657
PFN1	100%	100%	100%	100%	99.5%	Amyotrophic lateral sclerosis 18, 614808
SETX	100%	100%	100%	100%	99.8%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002;Amyotrophic lateral sclerosis 4, juvenile, 602433
SIGMAR1	100%	100%	100%	100%	100%	?Neuronopathy, distal hereditary motor, autosomal recessive 2, 605726;?Amyotrophic lateral sclerosis 16, juvenile, 614373
SOD1	100%	100%	100%	100%	99.7%	Spastic tetraplegia and axial hypotonia, progressive, 618598;Amyotrophic lateral sclerosis 1, 105400
SPG11	99.6%	99.6%	100%	100%	99.7%	Amyotrophic lateral sclerosis 5, juvenile, 602099;Charcot-Marie-Tooth disease, axonal, type 2X, 616668;Spastic paraplegia 11, autosomal recessive, 604360

SQSTM1	100%	100%	100%	100%	98.9%	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145;Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437;Myopathy, distal, with rimmed vacuoles, 617158;Paget disease of bone 3, 167250
TARDBP	100%	100%	100%	100%	99.7%	Frontotemporal lobar degeneration, TARDBP-related, 612069;Amyotrophic lateral sclerosis 10, with or without FTD, 612069
TBK1	100%	100%	100%	100%	99.9%	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8}, 617900;Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439;Autoinflammation with arthritis and vasculitis, 620880
TUBA4A	100%	100%	100%	100%	99.6%	Oocyte/zygote/embryo maturation arrest 23, 621231;Spastic ataxia 11, autosomal dominant, 621226;Frontotemporal dementia and/or amyotrophic lateral sclerosis 9, 616208;Congenital myopathy 26, 621225
UBQLN2	100%	100%	98.3%	90.4%	72.6%	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857

VAPB	91.4%	91.4%	100%	100%	99.9%	Spinal muscular atrophy, late-onset, Finkel type, 182980;Amyotrophic lateral sclerosis 8, 608627
VCP	100%	99.6%	100%	100%	99.8%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954;Charcot-Marie-Tooth disease, type 2Y, 616687;Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320

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.

TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.
This list is accurate for panel version DG 5.1.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors