ALS GENE PANEL DG 3.5.0 (27 genes)

Releasedate: 05-12-2022

Gene	*TWIST X2 covered >10x*	*TWIST X2 covered >20x*	*Associated Phenotype description and OMIM disease ID*
ALS2	100%	100%	Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225 Amyotrophic lateral sclerosis 2, juvenile, 205100
ANG	100%	100%	Amyotrophic lateral sclerosis 9, 611895
ANXA11	100%	100%	Amyotrophic lateral sclerosis 23, 617839 Inclusion body myopathy and brain white matter abnormalities, 619733
CFAP410	100%	100%	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271
CHCHD10	100%	100%	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 Spinal muscular atrophy, Jokela type, 615048 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911
CHMP2B	100%	100%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 7, 600795
ERBB4	100%	100%	Amyotrophic lateral sclerosis 19, 615515
FIG4	100%	100%	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228
FUS	100%	100%	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Essential tremor, hereditary, 4, 614782
GRN	100%	100%	Aphasia, primary progressive, 607485 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706
KIF5A	100%	100%	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187
MAPT	100%	100%	Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540 Dementia, frontotemporal, with or without parkinsonism, 600274 Pick disease, 172700
MATR3	100%	100%	Amyotrophic lateral sclerosis 21, 606070
NEK1	100%	100%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520

OPTN	100%	100%	Glaucoma 1, open angle, E, 137760 Amyotrophic lateral sclerosis 12 with or without frontotemporal dementia, 613435
PFN1	100%	100%	Amyotrophic lateral sclerosis 18, 614808
SETX	100%	100%	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433
SIGMAR1	100%	100%	?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726 ?Amyotrophic lateral sclerosis 16, juvenile, 614373
SOD1	100%	100%	Spastic tetraplegia and axial hypotonia, progressive, 618598 Amyotrophic lateral sclerosis 1, 105400
SPG11	100%	100%	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SQSTM1	100%	100%	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%	Frontotemporal lobar degeneration, TARDBP-related, 612069 Amyotrophic lateral sclerosis 10, with or without FTD, 612069
TBK1	100%	100%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439
TUBA4A	100%	100%	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208
UBQLN2	100%	100%	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857
VAPB	100%	100%	Spinal muscular atrophy, late-onset, Finkel type, 182980 Amyotrophic lateral sclerosis 8, 608627
VCP	100%	100%	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 HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
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TWIST X2 is the chemistry used for WES analysis.
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 coverage denoting NC are non-protein coding genes.
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 28th , 2022.
This list is accurate for panel version DG 3.5.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors