ALS GENE PANEL DG 3.2.0 (27 genes)

Releasedate: 16-09-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype Description and OMIM disease ID*
ALS2	99,9	99,8	100	100	Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225 Amyotrophic lateral sclerosis 2, juvenile, 205100
ANG	100	99,7	100	100	Amyotrophic lateral sclerosis 9, 611895
ANXA11	99,8	97,6	100	100	Amyotrophic lateral sclerosis 23, 617839
CFAP410	100	99,6	100	100	Retinal dystrophy with macular staphyloma, 617547 Spondylometaphyseal dysplasia, axial, 602271
CHCHD10	57,8	42	100	100	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 Spinal muscular atrophy, Jokela type, 615048 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911
CHMP2B	99,6	98,5	100	100	Frontotemporal dementia and/or amyotrophic lateral sclerosis 7, 600795
ERBB4	99,9	99,5	100	100	Amyotrophic lateral sclerosis 19, 615515
FIG4	99,8	99,7	100	100	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228
FUS	98,4	95,3	100	100	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Essential tremor, hereditary, 4, 614782
GRN	100	100	100	100	Aphasia, primary progressive, 607485 Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 Ceroid lipofuscinosis, neuronal, 11, 614706
KIF5A	100	99,8	100	100	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187
MAPT	99,9	98,9	100	100	Supranuclear palsy, progressive, 601104 Supranuclear palsy, progressive atypical, 260540 Dementia, frontotemporal, with or without parkinsonism, 600274 Pick disease, 172700
MATR3	96,5	90,6	100	100	Amyotrophic lateral sclerosis 21, 606070

NEK1	99,5	98,2	100	99,9	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
OPTN	99,9	99,9	100	100	Glaucoma 1, open angle, E, 137760 Amyotrophic lateral sclerosis 12 with or without frontotemporal dementia, 613435
PFN1	100	100	100	100	Amyotrophic lateral sclerosis 18, 614808
SETX	99,8	99,6	100	100	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433
SIGMAR1	100	100	100	100	?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726 ?Amyotrophic lateral sclerosis 16, juvenile, 614373
SOD1	100	100	100	100	Spastic tetraplegia and axial hypotonia, progressive, 618598 Amyotrophic lateral sclerosis 1, 105400
SPG11	99,8	99	100	100	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SQSTM1	99,8	97,8	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	99,9	100	100	Frontotemporal lobar degeneration, TARDBP-related, 612069 Amyotrophic lateral sclerosis 10, with or without FTD, 612069
TBK1	99,3	97,5	100	99,8	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439
TUBA4A	100	100	100	100	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208
UBQLN2	99,8	98,7	100	100	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857
VAPB	99,8	99,4	100	99,9	Spinal muscular atrophy, late-onset, Finkel type, 182980 Amyotrophic lateral sclerosis 8, 608627
VCP	100	99,1	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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Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID 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 : September 16th , 2021.
This list is accurate for panel version DG 3.2.0
Ad 1. “No OMIM Disease ID” signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors