ALS GENE PANEL DG 3.00 (23 genes)

Releasedate: 02-12-2020

Gene	Agilent V5 covered > 10x	Agilent V5 covered > 20x	TWIST covered > 10x	TWIST covered 20x	Associated Phenotype description and OMIM disease ID
ALS2	100	99,9	100	100	Primary lateral sclerosis, juvenile, 606353 Amyotrophic lateral sclerosis 2, juvenile, 205100 Spastic paralysis, infantile onset ascending, 607225
ANG	100	100	100	100	Amyotrophic lateral sclerosis 9, 611895
ANXA11	100	98,5	100	100	Amytrophic lateral sclerosis 23, 617839
CHCHD10	59,1	43,9	100	100	Spinal muscular atrophy, Jokela type, 615048 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 ?Myopathy, isolated mitochondrial, autosomal dominant, 616209
CHMP2B	99,7	96,7	100	100	Amyotrophic lateral sclerosis 17, 614696 Dementia, familial, nonspecific, 600795
ERBB4	100	99,5	100	100	Amyotrophic lateral sclerosis 19, 615515
FIG4	100	99,8	100	100	Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 Charcot-Marie-Tooth disease, type 4J, 611228 Amyotrophic lateral sclerosis 11, 612577
FUS	99,2	96,4	100	100	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Essential tremor, hereditary, 4, 614782
KIF5A	100	99,9	100	100	Myoclonus, intractable, neonatal, 617235 Spastic paraplegia 10, autosomal dominant, 604187 {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921
MATR3	97	93,4	100	100	Amyotrophic lateral sclerosis 21, 606070
OPTN	100	99,9	100	100	{Glaucoma, normal tension, susceptibility to}, 606657 Glaucoma 1, open angle, E, 137760 Amyotrophic lateral sclerosis 12, 613435
PFN1	100	100	100	100	Amyotrophic lateral sclerosis 18, 614808
SETX	100	99,8	100	100	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 Amyotrophic lateral sclerosis 4, juvenile, 602433

SIGMAR1	100	100	100	100	?Amyotrophic lateral sclerosis 16, juvenile, 614373 ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726
SOD1	100	99,9	100	100	Spastic tetraplegia and axial hypotonia, progressive, 618598 Amyotrophic lateral sclerosis 1, 105400
SPG11	100	99,3	100	100	Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360 Amyotrophic lateral sclerosis 5, juvenile, 602099
SQSTM1	98,8	95,5	100	100	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 Myopathy, distal, with rimmed vacuoles, 617158 Paget disease of bone 3, 167250
TARDBP	100	100	100	100	Frontotemporal lobar degeneration, TARDBP-related, 612069 Amyotrophic lateral sclerosis 10, with or without FTD, 612069
TBK1	99,7	97,2	100	100	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439 {Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8}, 617900
TUBA4A	100	100	100	100	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208
UBQLN2	100	99,4	100	100	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857
VAPB	100	99,9	100	100	Spinal muscular atrophy, late-onset, Finkel type, 182980 Amyotrophic lateral sclerosis 8, 608627
VCP	100	99,2	100	100	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 Charcot-Marie-Tooth disease, type 2Y, 616687 Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954

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.
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-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 20th , 2020.
This list is accurate for panel version DG 3.0.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors