

# ALS GENE PANEL DG 2.15 (22 genes)

Releasedate: 31-01-2019

<i>Gene</i>	<i>Median coverage</i>	<i>% covered &gt; 10x</i>	<i>% covered &gt; 20x</i>	<i>Associated Phenotype Description and OMIM disease ID</i>
ALS2	170.2	99.9	99.2	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225
ANG	178	100	99.9	Amyotrophic lateral sclerosis 9, 611895
ANXA11	80.8	98.9	88.8	Amyotrophic lateral sclerosis 23, 617839
CHCHD10	20	43	35.2	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 Spinal muscular atrophy, Jokela type, 615048
CHMP2B	92	98.6	91.7	Amyotrophic lateral sclerosis 17, 614696 Dementia, familial, nonspecific, 600795
ERBB4	144.5	99.9	99.1	Amyotrophic lateral sclerosis 19, 615515
FIG4	154.9	99.8	98.4	?Polymicrogyria, bilateral temporooccipital, 612691 Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 Yunis-Varon syndrome, 216340
FUS	137.4	97.6	94.7	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Essential tremor, hereditary, 4, 614782
MATR3	92.5	95.7	88.8	Amyotrophic lateral sclerosis 21, 606070
OPTN	113.8	100	99.4	Amyotrophic lateral sclerosis 12, 613435 Glaucoma 1, open angle, E, 137760 {Glaucoma, normal tension, susceptibility to}, 606657
PFN1	152	100	100	Amyotrophic lateral sclerosis 18, 614808
SETX	163.2	99.9	99.1	Amyotrophic lateral sclerosis 4, juvenile, 602433 Spinocerebellar ataxia, autosomal recessive 1, 606002
SIGMAR1	148.5	100	100	?Amyotrophic lateral sclerosis 16, juvenile, 614373 ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726
SOD1	161.9	100	100	Amyotrophic lateral sclerosis 1, 105400

SPG11	129.2	99.2	96.9	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360
SQSTM1	109.1	98.6	94.5	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Myopathy, distal, with rimmed vacuoles, 617158 Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 Paget disease of bone 3, 167250
TARDBP	175.2	100	100	Amyotrophic lateral sclerosis 10, with or without FTD, 612069 Frontotemporal lobar degeneration, TARDBP-related, 612069
TBK1	102.5	97.8	90.7	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439 {Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8}, 617900
TUBA4A	220.6	100	100	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208
UBQLN2	136.3	99.7	98	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857
VAPB	107.8	99	95.9	Amyotrophic lateral sclerosis 8, 608627 Spinal muscular atrophy, late-onset, Finkel type, 182980
VCP	144.8	99.9	99.5	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 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.

Median Coverage describes the average number of reads seen across 50 exomes.

% 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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : December 31st, 2018.

This list is accurate for panel version DG 2.15

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors