

ALS GENE PANEL DG 2.9/DG 2.10

<i>Gene</i>	<i>Median coverage</i>	<i>% covered > 10x</i>	<i>% covered > 20x</i>	<i>Associated Phenotype description and OMIM disease ID</i>
<i>ALS2</i>	192.8	99%	99%	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225
<i>ANG</i>	175.1	100%	99%	Amyotrophic lateral sclerosis 9, 611895
<i>CHCHD10</i>	27.8	65%	42%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 Spinal muscular atrophy, Jokela type, 615048 ?Myopathy, isolated mitochondrial, autosomal dominant, 616209
<i>CHMP2B</i>	112.3	99%	95%	Amyotrophic lateral sclerosis 17, 614696 Dementia, familial, nonspecific, 600795
<i>ERBB4</i>	172.4	99%	99%	Amyotrophic lateral sclerosis 19, 615515
<i>FIG4</i>	214.4	100%	99%	Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691
<i>FUS</i>	148.1	97%	94%	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Tremor, hereditary essential, 4, 614782
<i>MATR3</i>	107.4	97%	93%	Amyotrophic lateral sclerosis 21, 606070
<i>OPTN</i>	120	100%	99%	Amyotrophic lateral sclerosis 12, 613435 Glaucoma 1, open angle, E, 137760 {Glaucoma, normal tension, susceptibility to}, 606657
<i>PFN1</i>	177.4	100%	100%	Amyotrophic lateral sclerosis 18, 614808
<i>SETX</i>	197.6	100%	99%	Amyotrophic lateral sclerosis 4, juvenile, 602433 Spinocerebellar ataxia, autosomal recessive 1, 606002
<i>SIGMAR1</i>	144.5	100%	100%	?Amyotrophic lateral sclerosis 16, juvenile, 614373 ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726
<i>SOD1</i>	150.2	100%	99%	Amyotrophic lateral sclerosis 1, 105400
<i>SPG11</i>	145	99%	98%	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360

<i>SQSTM1</i>	142.3	99%	96%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Paget disease of bone 3, 167250
<i>TARDBP</i>	176.5	100%	100%	Amyotrophic lateral sclerosis 10, with or without FTD, 612069 Frontotemporal lobar degeneration, TARDBP-related, 612069
<i>TBK1</i>	119.5	99%	95%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439
<i>TUBA4A</i>	237.6	100%	100%	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208
<i>UBQLN2</i>	138.3	99%	98%	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857
<i>VAPB</i>	119.5	99%	98%	Amyotrophic lateral sclerosis 8, 608627 Spinal muscular atrophy, late-onset, Finkel type, 182980
<i>VCP</i>	155.7	100%	99%	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 HGNC 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 : April 14th, 2017.

This list is accurate for panel versions DG 2.9 and DG 2.10

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