

# ALS GENE PANEL DG 2.7/DG 2.8

<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>
<i>ALS2</i>	184.7	99%	99%	Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225
<i>ANG</i>	164	100%	99%	Amyotrophic lateral sclerosis 9, 611895
<i>CHCHD10</i>	25.8	58%	38%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 Spinal muscular atrophy, Jokela type, 615048 ?Myopathy, isolated mitochondrial, autosomal dominant, 616209
<i>CHMP2B</i>	97.2	97%	89%	Amyotrophic lateral sclerosis 17, 614696 Dementia, familial, nonspecific, 600795
<i>ERBB4</i>	161.6	99%	98%	Amyotrophic lateral sclerosis 19, 615515
<i>FIG4</i>	187.1	99%	98%	Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691
<i>FUS</i>	159.4	99%	96%	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Tremor, hereditary essential, 4, 614782
<i>MATR3</i>	94	96%	89%	Amyotrophic lateral sclerosis 21, 606070
<i>OPTN</i>	120.8	99%	99%	Amyotrophic lateral sclerosis 12, 613435 Glaucoma 1, open angle, E, 137760 {Glaucoma, normal tension, susceptibility to}, 606657
<i>PFN1</i>	169.7	100%	100%	Amyotrophic lateral sclerosis 18, 614808
<i>SETX</i>	187.1	99%	99%	Amyotrophic lateral sclerosis 4, juvenile, 602433 Spinocerebellar ataxia, autosomal recessive 1, 606002
<i>SIGMAR1</i>	145.9	100%	99%	?Amyotrophic lateral sclerosis 16, juvenile, 614373 ?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726
<i>SOD1</i>	179.7	100%	99%	Amyotrophic lateral sclerosis 1, 105400
<i>SPG11</i>	146.9	98%	96%	Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360

<i>SQSTM1</i>	130.7	98%	94%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 Paget disease of bone 3, 167250
<i>TARDBP</i>	164.6	100%	99%	Amyotrophic lateral sclerosis 10, with or without FTD, 612069 Frontotemporal lobar degeneration, TARDBP-related, 612069
<i>TBK1</i>	120.7	96%	89%	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439
<i>TUBA4A</i>	221.1	100%	100%	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208
<i>UBQLN2</i>	149.2	99%	96%	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857
<i>VAPB</i>	128	98%	95%	Amyotrophic lateral sclerosis 8, 608627 Spinal muscular atrophy, late-onset, Finkel type, 182980
<i>VCP</i>	168.4	99%	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 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 : October 1st, 2016.

This list is accurate for panel versions DG 2.7 and DG 2.8 From DG 2.7 to DG 2.8 no changes were made to the content of the gene panels.

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

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