

ALS GENE PANEL DG 2.13 (21 genes)

| Gene | Median coverage | % covered > 10x | % covered > 20x | Associated Phenotype description and OMIM disease ID |
|---------|-----------------|-----------------|-----------------|---|
| ALS2 | 170.2 | 99 | 99 | Amyotrophic lateral sclerosis 2, juvenile, 205100 Primary lateral sclerosis, juvenile, 606353 Spastic paralysis, infantile onset ascending, 607225 |
| ANG | 178 | 100 | 99 | Amyotrophic lateral sclerosis 9, 611895 |
| CHCHD10 | 20 | 43 | 35 | Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 Spinal muscular atrophy, Jokela type, 615048 ?Myopathy, isolated mitochondrial, autosomal dominant, 616209 |
| CHMP2B | 92 | 98 | 91 | Amyotrophic lateral sclerosis 17, 614696 Dementia, familial, nonspecific, 600795 |
| ERBB4 | 144.5 | 99 | 99 | Amyotrophic lateral sclerosis 19, 615515 |
| FIG4 | 154.9 | 99 | 98 | Amyotrophic lateral sclerosis 11, 612577 Charcot-Marie-Tooth disease, type 4J, 611228 Yunis-Varon syndrome, 216340 ?Polymicrogyria, bilateral temporooccipital, 612691 |
| FUS | 137.4 | 97 | 94 | Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 Tremor, hereditary essential, 4, 614782 |
| MATR3 | 92.5 | 95 | 88 | Amyotrophic lateral sclerosis 21, 606070 |
| OPTN | 113.8 | 100 | 99 | 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 | 99 | 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 | 96 | Amyotrophic lateral sclerosis 5, juvenile, 602099 Charcot-Marie-Tooth disease, axonal, type 2X, 616668 Spastic paraplegia 11, autosomal recessive, 604360 |

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|--------|-------|-----|-----|---|
| SQSTM1 | 109.1 | 98 | 94 | Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 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 | 90 | Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439 |
| TUBA4A | 220.6 | 100 | 100 | Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208 |
| UBQLN2 | 136.3 | 99 | 98 | Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857 |
| VAPB | 107.8 | 99 | 95 | Amyotrophic lateral sclerosis 8, 608627 Spinal muscular atrophy, late-onset, Finkel type, 182980 |
| VCP | 144.8 | 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 : April 18th, 2018.

This list is accurate for panel version DG 2.13

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