# ALS GENE PANEL DG 2.17 ( 22 genes)

Releasedate: 06-12-2019

| Gene | Median Coverage | % covered > 10x | % covered > 20x | Associated Phenotype description and OMIM disease ID |
|---|---|---|---|---|
| ALS2 | 145.2 | 100.0% | 99.9% | Primary lateral sclerosis, juvenile, 606353<br>Amyotrophic lateral sclerosis 2, juvenile, 205100<br>Spastic paralysis, infantile onset ascending, 607225 |
| ANG | 161.8 | 100.0% | 100.0% | Amyotrophic lateral sclerosis 9, 611895 |
| ANXA11 | 93.3 | 99.9% | 98.9% | Amytrophic lateral sclerosis 23, 617839 |
| CHCHD10 | 28.6 | 67.7% | 41.5% | Spinal muscular atrophy, Jokela type, 615048<br>Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911<br>?Myopathy, isolated mitochondrial, autosomal dominant, 616209 |
| CHMP2B | 87.0 | 100.0% | 96.8% | Amyotrophic lateral sclerosis 17, 614696<br>Dementia, familial, nonspecific, 600795 |
| ERBB4 | 127.4 | 100.0% | 99.3% | Amyotrophic lateral sclerosis 19, 615515 |
| FIG4 | 155.0 | 100.0% | 99.7% | Yunis-Varon syndrome, 216340<br>?Polymicrogyria, bilateral temporooccipital, 612691<br>Charcot-Marie-Tooth disease, type 4J, 611228<br>Amyotrophic lateral sclerosis 11, 612577 |
| FUS | 126.1 | 99.8% | 97.6% | Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030<br>Essential tremor, hereditary, 4, 614782 |
| MATR3 | 87.4 | 96.9% | 92.7% | Amyotrophic lateral sclerosis 21, 606070 |
| OPTN | 107.5 | 100.0% | 99.8% | Glaucoma 1, open angle, E, 137760<br>Amyotrophic lateral sclerosis 12, 613435 |
| PFN1 | 166.1 | 100.0% | 100.0% | Amyotrophic lateral sclerosis 18, 614808 |
| SETX | 153.0 | 100.0% | 99.7% | Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002<br>Amyotrophic lateral sclerosis 4, juvenile, 602433 |
| SIGMAR1 | 162.1 | 100.0% | 100.0% | ?Amyotrophic lateral sclerosis 16, juvenile, 614373<br>?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726 |
| SOD1 | 129.8 | 100.0% | 99.7% | Amyotrophic lateral sclerosis 1, 105400<br>Spastic tetraplegia and axial hypotonia, progressive, 618598 |
| SPG11 | 118.8 | 99.9% | 98.5% | Charcot-Marie-Tooth disease, axonal, type 2X, 616668<br>Spastic paraplegia 11, autosomal recessive, 604360<br>Amyotrophic lateral sclerosis 5, juvenile, 602099 |

| | | | | |
|---|---|---|---|---|
| SQSTM1 | 129.7 | 100.0% | 99.6% | Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437<br>Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145<br>Myopathy, distal, with rimmed vacuoles, 617158<br>Paget disease of bone 3, 167250 |
| TARDBP | 129.8 | 100.0% | 100.0% | Frontotemporal lobar degeneration, TARDBP-related, 612069<br>Amyotrophic lateral sclerosis 10, with or without FTD, 612069 |
| TBK1 | 98.9 | 99.8% | 97.4% | Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439 |
| TUBA4A | 173.6 | 100.0% | 100.0% | Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208 |
| UBQLN2 | 132.2 | 100.0% | 99.8% | Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857 |
| VAPB | 94.9 | 100.0% | 99.4% | Spinal muscular atrophy, late-onset, Finkel type, 182980<br>Amyotrophic lateral sclerosis 8, 608627 |
| VCP | 103.9 | 100.0% | 99.4% | Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320<br>Charcot-Marie-Tooth disease, type 2Y, 616687<br>Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 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.*
*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 11th , 2019.*
*This list is accurate for panel version DG 2.17*
*Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*