ALS

Gene panel

Gene panel information

Gene panel	**ALS**
Version	4
Total genes	42
Activation date	Thursday 04 april 2024
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% coding region covered*	OMIM gene id	OMIM Phenotypes
ALS2	99.87 %	606352	Primary lateral sclerosis, juvenile, 606353 (3), Autosomal recessive; Spastic paralysis, infantile onset ascending, 607225 (3), Autosomal recessive; Amyotrophic lateral sclerosis 2, juvenile, 205100 (3), Autosomal recessive
ANG	100 %	105850	Amyotrophic lateral sclerosis 9, 611895 (3)
ANXA11	99.71 %	602572	Amyotrophic lateral sclerosis 23, 617839 (3), Autosomal dominant; Inclusion body myopathy and brain white matter abnormalities, 619733 (3), Autosomal dominant
CCNF	99.99 %	600227	Frontotemporal dementia and/or amyotrophic lateral sclerosis 5, 619141 (3), Autosomal dominant
CHCHD10	100 %	615903	?Myopathy, isolated mitochondrial, autosomal dominant, 616209 (3), Autosomal dominant; Spinal muscular atrophy, Jokela type, 615048 (3), Autosomal dominant; Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911 (3), Autosomal dominant
CHMP2B	99.8 %	609512	Frontotemporal dementia and/or amyotrophic lateral sclerosis 7, 600795 (3), Autosomal dominant
DCTN1	99.98 %	601143	Neuronopathy, distal hereditary motor, type VIIB, 607641 (3), Autosomal dominant; Perry syndrome, 168605 (3), Autosomal dominant; {Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal recessive, Autosomal dominant
DNAJC7	99.88 %	601964	*No OMIM phenotypes*
ERBB4	99.92 %	600543	Amyotrophic lateral sclerosis 19, 615515 (3), Autosomal dominant
FIG4	99.83 %	609390	Yunis-Varon syndrome, 216340 (3), Autosomal recessive; ?Polymicrogyria, bilateral temporooccipital, 612691 (3), Autosomal recessive; Amyotrophic lateral sclerosis 11, 612577 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 4J, 611228 (3), Autosomal recessive
FUS	99.93 %	137070	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030 (3); Essential tremor, hereditary, 4, 614782 (3), Autosomal dominant
GLE1	99.99 %	603371	Lethal congenital contracture syndrome 1, 253310 (3), Autosomal recessive; Congenital arthrogryposis with anterior horn cell disease, 611890 (3), Autosomal recessive
GRN	100 %	138945	Aphasia, primary progressive, 607485 (3), Autosomal dominant; Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485 (3), Autosomal dominant; Ceroid lipofuscinosis, neuronal, 11, 614706 (3), Autosomal recessive
HNRNPA1	62.92 %	164017	?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424 (3), Autosomal dominant; Amyotrophic lateral sclerosis 20, 615426 (3), Autosomal dominant
HNRNPA2B1	99.9 %	600124	?Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422 (3), Autosomal dominant

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KIF5A	99.91 %	602821	Myoclonus, intractable, neonatal, 617235 (3), Autosomal dominant; {Amyotrophic lateral sclerosis, susceptibility to, 25}, 617921 (3), Autosomal dominant; Spastic paraplegia 10, autosomal dominant, 604187 (3), Autosomal dominant
LYST	99.87 %	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
MATR3	99.86 %	164015	Amyotrophic lateral sclerosis 21, 606070 (3), Autosomal dominant
NEFH	100 %	162230	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924 (3), Autosomal dominant; {?Amyotrophic lateral sclerosis, susceptibility to}, 105400 (3), Autosomal recessive, Autosomal dominant
NEK1	99.83 %	604588	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 (3), Autosomal recessive, Digenic recessive; {Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 (3), Autosomal dominant
OPTN	99.98 %	602432	Glaucoma 1, open angle, E, 137760 (3), Autosomal dominant; Amyotrophic lateral sclerosis 12 with or without frontotemporal dementia, 613435 (3), Autosomal recessive, Autosomal dominant; {Glaucoma, normal tension, susceptibility to}, 606657 (3)
PFN1	74.59 %	176610	Amyotrophic lateral sclerosis 18, 614808 (3)
SETX	99.97 %	608465	Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy 2, 606002 (3), Autosomal recessive; Amyotrophic lateral sclerosis 4, juvenile, 602433 (3), Autosomal dominant
SIGMAR1	99.99 %	601978	?Spinal muscular atrophy, distal, autosomal recessive, 2, 605726 (3), Autosomal recessive; ?Amyotrophic lateral sclerosis 16, juvenile, 614373 (3), Autosomal recessive
SLC52A1	100 %	607883	Riboflavin deficiency, 615026 (3), Autosomal dominant
SLC52A2	100 %	607882	Brown-Vialetto-Van Laere syndrome 2, 614707 (3), Autosomal recessive
SLC52A3	99.94 %	613350	?Fazio-Londe disease, 211500 (3), Autosomal recessive; Brown-Vialetto-Van Laere syndrome 1, 211530 (3), Autosomal recessive
SOD1	99.97 %	147450	Spastic tetraplegia and axial hypotonia, progressive, 618598 (3), Autosomal recessive; Amyotrophic lateral sclerosis 1, 105400 (3), Autosomal recessive, Autosomal dominant
SORD	85.52 %	182500	Sorbitol dehydrogenase deficiency with peripheral neuropathy, 618912 (3), Autosomal recessive
SPAST	99.77 %	604277	Spastic paraplegia 4, autosomal dominant, 182601 (3), Autosomal dominant
SPG11	99.89 %	610844	Amyotrophic lateral sclerosis 5, juvenile, 602099 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2X, 616668 (3), Autosomal recessive; Spastic paraplegia 11, autosomal recessive, 604360 (3), Autosomal recessive
SPTLC1	99.74 %	605712	Amyotrophic lateral sclerosis 27, juvenile, 620285 (3), Autosomal dominant; Neuropathy, hereditary sensory and autonomic, type IA, 162400 (3), Autosomal dominant
SQSTM1	100 %	601530	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145 (3), Autosomal recessive; Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437 (3), Autosomal dominant; Myopathy, distal, with rimmed vacuoles, 617158 (3), Autosomal dominant; Paget disease of bone 3, 167250 (3), Autosomal dominant
TAF15	99.91 %	601574	Chondrosarcoma, extraskeletal myxoid, 612237 (1)
TARDBP	100 %	605078	Frontotemporal lobar degeneration, TARDBP-related, 612069 (3), Autosomal dominant; Amyotrophic lateral sclerosis 10, with or without FTD, 612069 (3), Autosomal dominant

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TBK1	99.07 %	604834	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8}, 617900 (3), Autosomal dominant; Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439 (3), Autosomal dominant
TIA1	99.72 %	603518	Welander distal myopathy, 604454 (3), Autosomal recessive, Autosomal dominant; Amyotrophic lateral sclerosis 26 with or without frontotemporal dementia, 619133 (3), Autosomal dominant
TUBA4A	100 %	191110	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia, 616208 (3), Autosomal dominant
UBQLN2	100 %	300264	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857 (3), X-linked dominant
UNC13A	99.99 %	609894	*No OMIM phenotypes*
VAPB	100 %	605704	Spinal muscular atrophy, late-onset, Finkel type, 182980 (3), Autosomal dominant; Amyotrophic lateral sclerosis 8, 608627 (3), Autosomal dominant
VCP	99.99 %	601023	Frontotemporal dementia and/or amyotrophic lateral sclerosis 6, 613954 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2Y, 616687 (3), Autosomal dominant; Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320 (3), Autosomal dominant

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2023-07-31**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

Possible phenotype mapping keys

(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known

(2) the disorder has been placed on the map by linkage; no mutation has been found

(3) the molecular basis for the disorder is known; a mutation has been found in the gene

(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).

Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).

A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.

* Exome panels: >=20x, HyperCap panels: >=30x