

Table 1

The 39 ALS-related genes examined in this study

Gene	Location	Name	Gene MIM number	NCBI Reference Sequence	Phenotype	Phenotype MIM number
<i>SOD1</i> , <i>ALS1</i>	21q22.11	Superoxide dismutase 1	147450	NM_000454.4	Amyotrophic lateral sclerosis 1	105400
<i>TARDBP</i> , <i>TDP43</i> , <i>ALS10</i>	1p36.22	TAR DNA-binding protein	605078	NM_007375.3	Amyotrophic lateral sclerosis 10, with or without frontotemporal dementia	612069
					Frontotemporal lobar degeneration, TARDBP-related	612069
<i>FUS</i> , <i>TLS</i> , <i>ALS6</i> , <i>ETM4</i>	16p11.2	Fusion, derived from 12-16 translocation, malignant liposarcom	137070	NM_004960.3	Essential tremor, hereditary, 4	614782
					Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia	608030
<i>ANG</i> , <i>RNASE5</i> , <i>ALS9</i>	14q11.2	Angiogenin	105850	NM_001145.4	Amyotrophic lateral sclerosis 9	611895
<i>CHCHD10</i> , <i>FTDALS2</i> , <i>SMAJ</i> , <i>IMMD</i>	22q11.23	Coiled-coil-helix-coiled-coil-helix domain-containing protein 10	615903	NM_001301339.1	?Myopathy, isolated mitochondrial, autosomal dominant	616209
					Frontotemporal dementia and/or amyotrophic lateral sclerosis 2	615911
					Spinal muscular atrophy, Jokela type	615048
<i>OPTN</i> , <i>GLC1E</i> , <i>FIP2</i> , <i>HYPL</i> , <i>NRP</i> , <i>ALS12</i>	10p13	Optineurin	602432	NM_001008211.1	{Glaucoma, normal tension, susceptibility to}	606657
					Glaucoma 1, open angle, E	137760
					Amyotrophic lateral sclerosis 12	613435

<i>ALS2,</i> <i>ALSJ,</i> <i>PLSJ,</i> <i>IAHSP</i>	2q33.1	Alsin	606352	NM_020919.3	Amyotrophic lateral sclerosis 2, juvenile	205100
					Primary lateral sclerosis, juvenile	606353
					Spastic paralysis, infantile onset ascending	607225
<i>TBK1,</i> <i>NAK,</i> <i>FTDALS4,</i> <i>IIAE8</i>	12q14.2	TANK-binding kinase 1	604834	NM_013254.3	{Encephalopathy, acute, infection-induced (herpes-specific), susceptibility to, 8}	617900
					Frontotemporal dementia and/or amyotrophic lateral sclerosis 4	616439
<i>SPG11,</i> <i>KIAA1840,</i> <i>FLJ21439,</i> <i>ALS5,</i> <i>CMT2X</i>	15q21.1	Spastascin	610844	NM_025137.3	Amyotrophic lateral sclerosis 5, juvenile	602099
					Charcot-Marie-Tooth disease, axonal, type 2X	616668
					Spastic paraplegia 11, autosomal recessive	604360
<i>PFN1,</i> <i>ALS18</i>	17p13.2	Profilin-1	176610	NM_005022.3	Amyotrophic lateral sclerosis 18	614808
<i>TUBA4A,</i> <i>TUBA1,</i> <i>ALS22</i>	2q35	Tubulin, alpha-4A	191110	NM_006000.2	Amyotrophic lateral sclerosis 22 with or without frontotemporal dementia	616208
<i>DAO,</i> <i>DAMOX</i>	12q24.11	D-amino-acid oxidase	124050	NM_001917.4	{Schizophrenia}	181500
<i>GRN,</i> <i>CLN11</i>	17q21.31	Granulin	138945	NM_002087.3	Aphasia, primary progressive	607485
					Ceroid lipofuscinosis, neuronal, 11	614706
					Frontotemporal lobar degeneration with ubiquitin-positive inclusions	607485

<i>SETX</i> , <i>SCAR1</i> , <i>AOA2</i> , <i>ALS4</i>	9q34.13	Senataxin	608465	NM_015046.6	Amyotrophic lateral sclerosis 4, juvenile	602433
					Spinocerebellar ataxia, autosomal recessive 1	606002
<i>TAF15</i> , <i>TAF2N</i> , <i>RBP56</i>	17q12	TAF15 RNA polymerase II, TATA box-binding protein- associated factor, 68kD	601574	NM_139215.2	Chondrosarcoma, extraskeletal myxoid	612237
<i>VCP</i> , <i>IBMPFD1</i> , <i>ALS14</i> , <i>CMT2Y</i>	9p13.3	Valosin-containing protein	601023	NM_007126.4	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1	167320
					Charcot-Marie-Tooth disease, type 2Y	616687
					Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia	613954
<i>UBQLN2</i> , <i>PLIC2</i> , <i>CHAP1</i> , <i>ALS15</i>	Xp11.21	Ubiquilin 2	300264	NM_013444.3	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia	300857
<i>SQSTM1</i> , <i>P62</i> , <i>PDB3</i> , <i>FTDALS3</i> , <i>NADGP</i> , <i>DMRV</i>	5q35.3	Sequestosome 1	601530	NM_003900.4	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3	616437
					Myopathy, distal, with rimmed vacuoles	617158
					Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset	617145
					Paget disease of bone 3	167250

<i>FIG4</i> , <i>KIAA0274</i> , <i>SAC3</i> , <i>ALS11</i> , <i>YVS</i> , <i>BTOP</i>	6q21	Fig4, <i>S. cerevisiae</i> , homolog of	609390	NM_014845.5	?Polymicrogyria, bilateral temporooccipital	612691
					Amyotrophic lateral sclerosis 11	612577
					Charcot-Marie-Tooth disease, type 4J	611228
					Yunis-Varon syndrome	216340
<i>ANO2</i> , <i>TMEM16B</i> , <i>C12orf3</i>	12p13.31	Anoctamin 2	610109	NM_001278596.1		
<i>APEX</i> , <i>APE</i>	14q11.2	APEX nuclease (multifunctional DNA repair enzyme)	107748	NM_001641.3		
<i>CEP112</i>	17q24.1	Centrosomal protein 112		XM_005257119.5		
<i>CHMP2B</i> , <i>DMT1</i> , <i>VPS2B</i> , <i>ALS17</i>	3p11.2	CHMP family, member 2B	609512	NM_014043.3	Amyotrophic lateral sclerosis 17	614696
					Dementia, familial, nonspecific	600795
<i>CNTF</i> , <i>HCNTF</i>	11q12.1	Ciliary neurotrophic factor	118945	NM_000614.3		
<i>DCTN1</i> , <i>HMN7B</i>	2p13.1	Dynactin 1 (p150, glued, <i>Drosophila</i> , homolog of)	601143	NM_004082.4	Neuropathy, distal hereditary motor, type VIIB	607641
					Perry syndrome	168605
					{ Amyotrophic lateral sclerosis, susceptibility to }	105400

<i>HFE,</i> <i>HLA-H,</i> <i>HFE1,</i> <i>MVCD7,</i> <i>TFQTL2</i>	6p22.2	Hemochromatosis gene	613609	NM_000410.3	Hemochromatosis	235200
					[Transferrin serum level QTL2]	614193
					{Alzheimer disease, susceptibility to}	104300
					{Microvascular complications of diabetes 7}	612635
					{Porphyria cutanea tarda, susceptibility to}	176100
					{Porphyria variegata, susceptibility to}	176200
<i>MATR3,</i> <i>MPD2,</i> <i>ALS21</i>	5q31.2	Matrin 3	164015	NM_199189.2	Amyotrophic lateral sclerosis 21	606070
<i>NEFH,</i> <i>CMT2CC</i>	22q12.2	Neurofilament, heavy polypeptide	162230	NM_021076.3	{Amyotrophic lateral sclerosis, susceptibility to}	105400
					Charcot-Marie-Tooth disease, axonal, type 2CC	616924
<i>PON1,</i> <i>PON,</i> <i>ESA,</i> <i>MVCD5</i>	7q21.3	Paraoxonase-1	168820	NM_000446.6	{Coronary artery disease, susceptibility to}	
					{Coronary artery spasm 2, susceptibility to}	
					{Microvascular complications of diabetes 5}	612633
					{Organophosphate poisoning, sensitivity to}	

<i>PON2</i>	7q21.3	Paraoxonase-2	602447	NM_000305.2	{Coronary artery disease, susceptibility to}	
<i>PRPH</i>	12q13.12	Peripherin	170710	NM_006262.3	{Amyotrophic lateral sclerosis, susceptibility to}	105400
<i>SMN1, SMA1, SMA2, SMA3, SMA4</i>	5q13.2	Survival of motor neuron 1, telomeric	600354	NM_001297715.1	Spinal muscular atrophy-1	253300
					Spinal muscular atrophy-2	253550
					Spinal muscular atrophy-3	253400
					Spinal muscular atrophy-4	271150
<i>SMN2</i>	5q13.2	Survival of motor neuron 2, centromeric	601627	NM_022875.2	{Spinal muscular atrophy, type III, modifier of}	253400
<i>VAPB, VAPC, ALS8</i>	20q13.32	Vesicle-associated membrane protein-associated protein B	605704	NM_004738.4	Amyotrophic lateral sclerosis 8	608627
					Spinal muscular atrophy, late-onset, Finkel type	182980
<i>VEGF, MVCD1</i>	6p21.1	Vascular endothelial growth factor	192240	NM_001171623.1	{Microvascular complications of diabetes 1}	603933
<i>CCNF, FBX1</i>	16p13.3	Cyclin F	600227	NM_001761.2		
<i>NEK1, SRTD6, SRPS2A, ALS24</i>	4q33	Never in mitosis gene A-related kinase 1	604588	NM_001199397.1	{Amyotrophic lateral sclerosis, susceptibility to, 24}	617892
					Short-rib thoracic dysplasia 6 with or without polydactyly	263520
<i>HNRNPA1, IBMPFD3, ALS20</i>	12q13.13	Heterogeneous nuclear ribonucleoprotein A1	164017	NM_002136.3	Amyotrophic lateral sclerosis 20	615426

					?Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3	615424
<i>ERBB4</i> , <i>HER4</i> , <i>ALS19</i>	2q34	Avian erythroblastic leukemia viral (v-erb-b2) oncogene homolog 4	600543	NM_005235.2	Amyotrophic lateral sclerosis 19	615515