Robert H Brown

List of Publications by Year
in descending order

172	34,129	8181 76	4548 171
papers	citations	h-index	g-index

186	186	186	26792
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutations in Cu/Zn superoxide dismutase gene are associated with familial amyotrophic lateral sclerosis. Nature, 1993, 362, 59-62.	27.8	6,331
2	Decoding ALS: from genes to mechanism. Nature, 2016, 539, 197-206.	27.8	1,533
3	Amyotrophic Lateral Sclerosis. New England Journal of Medicine, 2017, 377, 162-172.	27.0	1,264
4	Motor neurons in Cu/Zn superoxide dismutase-deficient mice develop normally but exhibit enhanced cell death after axonal injury. Nature Genetics, 1996, 13, 43-47.	21.4	1,153
5	Molecular biology of amyotrophic lateral sclerosis: insights from genetics. Nature Reviews Neuroscience, 2006, 7, 710-723.	10.2	984
6	Mutant dynactin in motor neuron disease. Nature Genetics, 2003, 33, 455-456.	21.4	884
7	Dysferlin, a novel skeletal muscle gene, is mutated in Miyoshi myopathy and limb girdle muscular dystrophy. Nature Genetics, 1998, 20, 31-36.	21.4	857
8	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. Science, 2015, 347, 1436-1441.	12.6	823
9	Evidence of Increased Oxidative Damage in Both Sporadic and Familial Amyotrophic Lateral Sclerosis. Journal of Neurochemistry, 1997, 69, 2064-2074.	3.9	671
10	A gene encoding a putative GTPase regulator is mutated in familial amyotrophic lateral sclerosis 2. Nature Genetics, 2001, 29, 166-173.	21.4	635
11	ANG mutations segregate with familial and 'sporadic' amyotrophic lateral sclerosis. Nature Genetics, 2006, 38, 411-413.	21.4	617
12	Wild-type and mutant SOD1 share an aberrant conformation and a common pathogenic pathway in ALS. Nature Neuroscience, 2010, 13, 1396-1403.	14.8	600
13	Intrinsic Membrane Hyperexcitability of Amyotrophic Lateral Sclerosis Patient-Derived Motor Neurons. Cell Reports, 2014, 7, 1-11.	6.4	583
14	dSarm/Sarm1 Is Required for Activation of an Injury-Induced Axon Death Pathway. Science, 2012, 337, 481-484.	12.6	558
15	Mutations in the profilin 1 gene cause familial amyotrophic lateral sclerosis. Nature, 2012, 488, 499-503.	27.8	522
16	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	8.1	517
17	Variants of the elongator protein 3 (ELP3) gene are associated with motor neuron degeneration. Human Molecular Genetics, 2009, 18, 472-481.	2.9	512
18	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1043-1048.	21.4	494

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19	XBP-1 deficiency in the nervous system protects against amyotrophic lateral sclerosis by increasing autophagy. Genes and Development, 2009, 23, 2294-2306.	5.9	463
20	Mutant FUS proteins that cause amyotrophic lateral sclerosis incorporate into stress granules. Human Molecular Genetics, 2010, 19, 4160-4175.	2.9	447
21	Pathways Disrupted in Human ALS Motor Neurons Identified through Genetic Correction of Mutant SOD1. Cell Stem Cell, 2014, 14, 781-795.	11.1	392
22	Immediate and long-term consequences of COVID-19 infections for the development of neurological disease. Alzheimer's Research and Therapy, 2020, 12, 69.	6.2	367
23	A Met-to-Val mutation in the skeletal muscle Na+ channel Î±-subunit in hyperkalaemic periodic paralysis. Nature, 1991, 354, 387-389.	27.8	356
24	Genome-wide association study identifies 19p13.3 (UNC13A) and 9p21.2 as susceptibility loci for sporadic amyotrophic lateral sclerosis. Nature Genetics, 2009, 41, 1083-1087.	21.4	344
25	Dysferlin Interacts with Annexins A1 and A2 and Mediates Sarcolemmal Wound-healing. Journal of Biological Chemistry, 2003, 278, 50466-50473.	3.4	336
26	Sodium phenylbutyrate prolongs survival and regulates expression of antiâ€apoptotic genes in transgenic amyotrophic lateral sclerosis mice. Journal of Neurochemistry, 2005, 93, 1087-1098.	3.9	315
27	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. Neuron, 2014, 84, 324-331.	8.1	308
28	miRNA malfunction causes spinal motor neuron disease. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 13111-13116.	7.1	299
29	Inhibition of ICE slows ALS in mice. Nature, 1997, 388, 31-31.	27.8	298
30	Discovery of a Biomarker and Lead Small Molecules to Target r(GGGGCC)-Associated Defects in c9FTD/ALS. Neuron, 2014, 83, 1043-1050.	8.1	289
31	SPTLC1 is mutated in hereditary sensory neuropathy, type 1. Nature Genetics, 2001, 27, 261-262.	21.4	284
32	SOD1 mutants linked to amyotrophic lateral sclerosis selectively inactivate a glial glutamate transporter. Nature Neuroscience, 1999, 2, 427-433.	14.8	282
33	EPHA4 is a disease modifier of amyotrophic lateral sclerosis in animal models and in humans. Nature Medicine, 2012, 18, 1418-1422.	30.7	269
34	Emerging mechanisms of molecular pathology in ALS. Journal of Clinical Investigation, 2015, 125, 1767-1779.	8.2	244
35	Respiratory dysfunction and management in spinal cord injury. Respiratory Care, 2006, 51, 853-68;discussion 869-70.	1.6	240
36	Sixteen novel mutations in the Cu/Zn superoxide dismutase gene in amyotrophic lateral sclerosis: a decade of discoveries, defects and disputes. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders: Official Publication of the World Federation of Neurology, Research Group on Motor Neuron Diseases, 2003, 4, 62-73.	1.2	234

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37	Common and rare variant association analyses in amyotrophic lateral sclerosis identify 15 risk loci with distinct genetic architectures and neuron-specific biology. Nature Genetics, 2021, 53, 1636-1648.	21.4	223
38	Linkage of recessive familial amyotrophic lateral sclerosis to chromosome 2q33â€“q35. Nature Genetics, 1994, 7, 425-428.	21.4	221
39	Neonatal Neuronal Circuitry Shows Hyperexcitable Disturbance in a Mouse Model of the Adult-Onset Neurodegenerative Disease Amyotrophic Lateral Sclerosis. Journal of Neuroscience, 2008, 28, 10864-10874.	3.6	219
40	Human C9ORF72 Hexanucleotide Expansion Reproduces RNA Foci and Dipeptide Repeat Proteins but Not Neurodegeneration in BAC Transgenic Mice. Neuron, 2015, 88, 902-909.	8.1	219
41	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	21.4	218
42	Chromosome 9p21 in sporadic amyotrophic lateral sclerosis in the UK and seven other countries: a genome-wide association study. Lancet Neurology, The, 2010, 9, 986-994.	10.2	205
43	The C9ORF72 expansion mutation is a common cause of ALS+/â€”FTD in Europe and has a single founder. European Journal of Human Genetics, 2013, 21, 102-108.	2.8	201
44	Proteomic profiling of cerebrospinal fluid identifies biomarkers for amyotrophic lateral sclerosis. Journal of Neurochemistry, 2005, 95, 1461-1471.	3.9	191
45	A Yeast Model of FUS/TLS-Dependent Cytotoxicity. PLoS Biology, 2011, 9, e1001052.	5.6	191
46	Peripheral neuropathy in the acquired immunodeficiency syndrome. Annals of Neurology, 1988, 23, 485-492.	5.3	186
47	TDP-43 gains function due to perturbed autoregulation in a Tardbp knock-in mouse model of ALS-FTD. Nature Neuroscience, 2018, 21, 552-563.	14.8	181
48	Poly(GP) proteins are a useful pharmacodynamic marker for C9ORF72 -associated amyotrophic lateral sclerosis. Science Translational Medicine, 2017, 9, .	12.4	179
49	Reduced expression of the Kinesin-Associated Protein 3 (KIFAP3) gene increases survival in sporadic amyotrophic lateral sclerosis. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 9004-9009.	7.1	177
50	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. Nature Communications, 2016, 7, 11253.	12.8	174
51	Angiogenin variants in Parkinson disease and amyotrophic lateral sclerosis. Annals of Neurology, 2011, 70, 964-973.	5.3	168
52	Disruption of muscle membrane and phenotype divergence in two novel mouse models of dysferlin deficiency. Human Molecular Genetics, 2004, 13, 1999-2010.	2.9	166
53	Attenuated traumatic axonal injury and improved functional outcome after traumatic brain injury in mice lackingSarm1. Brain, 2016, 139, 1094-1105.	7.6	155
54	Genetics of Amyotrophic Lateral Sclerosis. Cold Spring Harbor Perspectives in Medicine, 2018, 8, a024125.	6.2	151

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55	SOD1Suppression with Adeno-Associated Virus and MicroRNA in Familial ALS. New England Journal of Medicine, 2020, 383, 151-158.	27.0	151
56	Rapid Communication: Cu/Zn Superoxide Dismutase Activity in Familial and Sporadic Amyotrophic Lateral Sclerosis. Journal of Neurochemistry, 1994, 62, 384-387.	3.9	143
57	Dysferlin in Membrane Trafficking and Patch Repair. Traffic, 2007, 8, 785-794.	2.7	138
58	Phase 2 study of sodium phenylbutyrate in ALS. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2009, 10, 99-106.	2.1	135
59	Mutations in the vesicular trafficking protein annexin A11 are associated with amyotrophic lateral sclerosis. Science Translational Medicine, 2017, 9, .	12.4	129
60	Mutations in the N-terminal Actin-Binding Domain of Filamin C Cause a Distal Myopathy. American Journal of Human Genetics, 2011, 88, 729-740.	6.2	124
61	Amyotrophic Lateral Sclerosis. New England Journal of Medicine, 2017, 377, 1602-1602.	27.0	118
62	Amyotrophic lateral sclerosis: Problems and prospects. Annals of Neurology, 2013, 74, 309-316.	5.3	117
63	Mutations in the glutamate transporter EAAT2 gene do not cause abnormal EAAT2 transcripts in amyotrophic lateral sclerosis. Annals of Neurology, 1998, 43, 645-653.	5.3	109
64	<scp>ALS</scp>â€linked protein disulfide isomerase variants cause motor dysfunction. EMBO Journal, 2016, 35, 845-865.	7.8	109
65	Superoxide Dismutase Concentration and Activity in Familial Amyotrophic Lateral Sclerosis. Journal of Neurochemistry, 1995, 64, 2366-2369.	3.9	101
66	Inhibition of Fast Axonal Transport by Pathogenic SOD1 Involves Activation of p38 MAP Kinase. PLoS ONE, 2013, 8, e65235.	2.5	100
67	Mutant SOD1-expressing astrocytes release toxic factors that trigger motoneuron death by inducing hyperexcitability. Journal of Neurophysiology, 2013, 109, 2803-2814.	1.8	99
68	A novel, blood-based diagnostic assay for limb girdle muscular dystrophy 2B and miyoshi myopathy. Annals of Neurology, 2002, 51, 129-133.	5.3	98
69	Childhood amyotrophic lateral sclerosis caused by excess sphingolipid synthesis. Nature Medicine, 2021, 27, 1197-1204.	30.7	96
70	NurOwn, phase 2, randomized, clinical trial in patients with ALS. Neurology, 2019, 93, e2294-e2305.	1.1	95
71	<scp>C9orf72</scp> and <scp>UNC13A</scp> are shared risk loci for amyotrophic lateral sclerosis and frontotemporal dementia: A genomeâ€wide metaâ€analysis. Annals of Neurology, 2014, 76, 120-133.	5.3	91
72	Endoplasmic reticulum stress leads to accumulation of wild-type SOD1 aggregates associated with sporadic amyotrophic lateral sclerosis. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 8209-8214.	7.1	88

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73	Novel mutations support a role for Profilin 1 in the pathogenesis of ALS. Neurobiology of Aging, 2015, 36, 1602.e17-1602.e27.	3.1	87
74	Mutant PFN1 causes ALS phenotypes and progressive motor neuron degeneration in mice by a gain of toxicity. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E6209-E6218.	7.1	85
75	Therapeutic rAAVrh10 MediatedSOD1Silencing in AdultSOD1^{G93A}Mice and Nonhuman Primates. Human Gene Therapy, 2016, 27, 19-31.	2.7	85
76	Randomized trial of <scp>l</scp> -serine in patients with hereditary sensory and autonomic neuropathy type 1. Neurology, 2019, 92, e359-e370.	1.1	83
77	Extracellular microRNAs in human circulation are associated with miRISC complexes that are accessible to anti-AGO2 antibody and can bind target mimic oligonucleotides. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 24213-24223.	7.1	83
78	Widespread spinal cord transduction by intrathecal injection of rAAV delivers efficacious RNAi therapy for amyotrophic lateral sclerosis. Human Molecular Genetics, 2014, 23, 668-681.	2.9	81
79	Apolipoprotein E ?4 allele is not associated with earlier age at onset in amyotrophic lateral sclerosis. Annals of Neurology, 1995, 38, 460-463.	5.3	80
80	Sequence variants in human neurofilament proteins: Absence of linkage to familial amyotrophic lateral sclerosis. Annals of Neurology, 1996, 40, 603-610.	5.3	78
81	Mitochondrial damage revealed by immunoselection for ALS-linked misfolded SOD1. Human Molecular Genetics, 2013, 22, 3947-3959.	2.9	78
82	The distinct genetic pattern of ALS in Turkey and novel mutations. Neurobiology of Aging, 2015, 36, 1764.e9-1764.e18.	3.1	78
83	Adenoâ€associated virusâ€“delivered artificial microRNA extends survival and delays paralysis in an amyotrophic lateral sclerosis mouse model. Annals of Neurology, 2016, 79, 687-700.	5.3	78
84	A randomized trial of mexiletine in ALS. Neurology, 2016, 86, 1474-1481.	1.1	72
85	Suppression of mutant C9orf72 expression by a potent mixed backbone antisense oligonucleotide. Nature Medicine, 2022, 28, 117-124.	30.7	72
86	Age-Dependent TDP-43-Mediated Motor Neuron Degeneration Requires GSK3, hat-trick, and xmas-2. Current Biology, 2015, 25, 2130-2136.	3.9	71
87	Identification of rare protein disulfide isomerase gene variants in amyotrophic lateral sclerosis patients. Gene, 2015, 566, 158-165.	2.2	70
88	Age of onset of amyotrophic lateral sclerosis is modulated by a locus on 1p34.1. Neurobiology of Aging, 2013, 34, 357.e7-357.e19.	3.1	69
89	Mutant Profilin1 transgenic mice recapitulate cardinal features of motor neuron disease. Human Molecular Genetics, 2017, 26, ddw429.	2.9	67
90	Toxic mutants in Charcot's sclerosis. Nature, 1995, 378, 342-343.	27.8	64

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91	Homologous Recombination Mediates Functional Recovery of Dysferlin Deficiency following AAV5 Gene Transfer. PLoS ONE, 2012, 7, e39233.	2.5	64
92	Longitudinal biomarkers in amyotrophic lateral sclerosis. Annals of Clinical and Translational Neurology, 2020, 7, 1103-1116.	3.7	62
93	Safe and effective superoxide dismutase 1 silencing using artificial microRNA in macaques. Science Translational Medicine, 2018, 10, .	12.4	59
94	ALS-associated missense and nonsense TBK1 mutations can both cause loss of kinase function. Neurobiology of Aging, 2018, 71, 266.e1-266.e10.	3.1	59
95	Association of a Locus in theCAMTA1Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. JAMA Neurology, 2016, 73, 812.	9.0	57
96	Amyotrophic Lateral Sclerosis: Review. Seminars in Neurology, 2015, 35, 469-476.	1.4	56
97	Silencing strategies for therapy of SOD1-mediated ALS. Neuroscience Letters, 2017, 636, 32-39.	2.1	55
98	SOD1 aggregates in ALS: Cause, correlate or consequence?. Nature Medicine, 1998, 4, 1362-1364.	30.7	54
99	Modulation of actin polymerization affects nucleocytoplasmic transport in multiple forms of amyotrophic lateral sclerosis. Nature Communications, 2019, 10, 3827.	12.8	54
100	Mapping of Gene Expression Reveals CYP27A1 as a Susceptibility Gene for Sporadic ALS. PLoS ONE, 2012, 7, e35333.	2.5	50
101	AAV gene therapy for Tay-Sachs disease. Nature Medicine, 2022, 28, 251-259.	30.7	49
102	Dystrophin-Associated Proteins and the Muscular Dystrophies: A Glossary. Brain Pathology, 1996, 6, 19-24.	4.1	46
103	Loss of Sarm1 does not suppress motor neuron degeneration in the SOD1G93A mouse model of amyotrophic lateral sclerosis. Human Molecular Genetics, 2018, 27, 3761-3771.	2.9	45
104	A randomized <scp>placeboâ€controlled</scp> phase 3 study of mesenchymal stem cells induced to secrete high levels of neurotrophic factors in amyotrophic lateral sclerosis. Muscle and Nerve, 2022, 65, 291-302.	2.2	41
105	Genetic ablation of NMDA receptor subunit NR3B in mouse reveals motoneuronal and nonmotoneuronal phenotypes. European Journal of Neuroscience, 2007, 26, 1407-1420.	2.6	40
106	The Role of Copy Number Variation in Susceptibility to Amyotrophic Lateral Sclerosis: Genome-Wide Association Study and Comparison with Published Loci. PLoS ONE, 2009, 4, e8175.	2.5	39
107	Self-Control and External Control of Mechanical Ventilation Give Equal Air Hunger Relief. American Journal of Respiratory and Critical Care Medicine, 1998, 157, 415-420.	5.6	37
108	A common haplotype within the PON1 promoter region is associated with sporadic ALS. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2008, 9, 306-314.	2.1	37

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109	Human genetics and neuropathology suggest a link between miR-218 and amyotrophic lateral sclerosis pathophysiology. Science Translational Medicine, 2019, 11, .	12.4	37
110	Cortical Spreading Depression Denotes Concussion Injury. Journal of Neurotrauma, 2019, 36, 1008-1017.	3.4	34
111	Calpainopathy and eosinophilic myositis. Annals of Neurology, 2006, 59, 875-877.	5.3	33
112	Dysferlin overexpression in skeletal muscle produces a progressive myopathy. Annals of Neurology, 2010, 67, 384-393.	5.3	33
113	Excessive release of inorganic polyphosphate by ALS/FTD astrocytes causes non-cell-autonomous toxicity to motoneurons. Neuron, 2022, 110, 1656-1670.e12.	8.1	33
114	Superoxide dismutase and familial amyotrophic lateral sclerosis: New insights into mechanisms and treatments. Annals of Neurology, 1996, 39, 145-146.	5.3	32
115	Evaluation of the Golgi trafficking protein VPS54 (wobbler) as a candidate for ALS. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2008, 9, 141-148.	2.1	31
116	Type 1 human poliovirus binds to human synaptosomes. Annals of Neurology, 1987, 21, 64-70.	5.3	30
117	50bp deletion in the promoter for superoxide dismutase 1 (SOD1) reduces SOD1 expression in vitro and may correlate with increased age of onset of sporadic amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2008, 9, 229-237.	2.1	29
118	Prospective natural history study of C9orf72 ALS clinical characteristics and biomarkers. Neurology, 2019, 93, e1605-e1617.	1.1	29
119	Poly(GR) and poly(GA) in cerebrospinal fluid as potential biomarkers for C9ORF72-ALS/FTD. Nature Communications, 2022, 13, 2799.	12.8	28
120	A novel dysferlin mutant pseudoexon bypassed with antisense oligonucleotides. Annals of Clinical and Translational Neurology, 2014, 1, 703-720.	3.7	24
121	Heteroplasmy in Chronic External Ophthalmoplegia: Clinical and Molecular Observations. Pediatric Research, 1990, 28, 542-548.	2.3	23
122	Tetanus toxin fragment C fusion facilitates protein delivery to CNS neurons from cerebrospinal fluid in mice. Journal of Neurochemistry, 2005, 95, 1118-1131.	3.9	23
123	Breathlessness in Spinal Cord Injury Depends on Injury Level. Journal of Spinal Cord Medicine, 1999, 22, 97-101.	1.4	22
124	Analysis of a genetic defect in the TATA box of the SOD1 gene in a patient with familial amyotrophic lateral sclerosis. Muscle and Nerve, 2007, 36, 704-707.	2.2	22
125	A C9ORF72 BAC mouse model recapitulates key epigenetic perturbations of ALS/FTD. Molecular Neurodegeneration, 2017, 12, 46.	10.8	22
126	A new approach for rare variation collapsing on functional protein domains implicates specific genic regions in ALS. Genome Research, 2019, 29, 809-818.	5.5	21

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127	An update on superoxide dismutase 1 in familial amyotrophic lateral sclerosis. Journal of the Neurological Sciences, 1996, 139, 10-15.	0.6	20
128	Medications and laboratory parameters as prognostic factors in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2008, 9, 369-374.	2.1	20
129	Association of UBQLN1 mutation with Brownâ€“Vialettoâ€“Van Laere syndrome but not typical ALS. Neurobiology of Disease, 2012, 48, 391-398.	4.4	20
130	Cross-sectional associations of pulmonary function with systemic inflammation and oxidative stress in individuals with chronic spinal cord injury. Journal of Spinal Cord Medicine, 2016, 39, 344-352.	1.4	20
131	Correction of pseudoexon splicing caused by a novel intronic dysferlin mutation. Annals of Clinical and Translational Neurology, 2019, 6, 642-654.	3.7	20
132	Risk factors for hepatocellular carcinoma (HCC) in the northeast of the United States: results of a caseâ€“control study. Cancer Causes and Control, 2020, 31, 321-332.	1.8	20
133	Low-level overexpression of wild type TDP-43 causes late-onset, progressive neurodegeneration and paralysis in mice. PLoS ONE, 2022, 17, e0255710.	2.5	20
134	A Reinnervating MicroRNA. Science, 2009, 326, 1494-1495.	12.6	18
135	Genetic ablation of phospholipase C delta 1 increases survival in SOD1G93A mice. Neurobiology of Disease, 2013, 60, 11-17.	4.4	18
136	Variants in candidate ALS modifier genes linked to Cu/Zn superoxide dismutase do not explain divergent survival phenotypes. Neuroscience Letters, 2006, 392, 52-57.	2.1	17
137	Intralingual and Intrapleural AAV Gene Therapy Prolongs Survival in a SOD1 ALS Mouse Model. Molecular Therapy - Methods and Clinical Development, 2020, 17, 246-257.	4.1	17
138	Irreversible reduction in potassium fluxes accompanies terminal differentiation of human myoblasts to myotubes. Journal of Cellular Physiology, 1987, 132, 57-64.	4.1	16
139	Nucleic Acid Therapeutics for Neurological Diseases. Neurotherapeutics, 2019, 16, 245-247.	4.4	16
140	Genetic diversity of axon degenerative mechanisms in models of Parkinson's disease. Neurobiology of Disease, 2021, 155, 105368.	4.4	16
141	Whole-genome sequencing reveals that variants in the Interleukin 18 Receptor Accessory Protein 3â€²UTR protect against ALS. Nature Neuroscience, 2022, 25, 433-445.	14.8	16
142	Prevalence of Huntington's disease gene CAG repeat alleles in sporadic amyotrophic lateral sclerosis patients. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 265-269.	2.1	15
143	Breathlessness and Exercise in Spinal Cord Injury. Journal of Spinal Cord Medicine, 1999, 22, 297-302.	1.4	13
144	Amyotrophic Lateral Sclerosis: An Emerging Era of Collaborative Gene Discovery. PLoS ONE, 2007, 2, e1254.	2.5	13

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145	Proteolytic fragment or new gene product?. Nature, 1988, 336, 210-210.	27.8	12
146	Molecular genetic analysis of dysferlin in Japanese patients with Miyoshi myopathy. Proceedings of the Japan Academy Series B: Physical and Biological Sciences, 1999, 75, 207-212.	3.8	12
147	Finding a Treatment for ALS â€" Will Gene Editing Cut It?. New England Journal of Medicine, 2018, 378, 1454-1456.	27.0	11
148	Mutation in protein disulfide isomerase A3 causes neurodevelopmental defects by disturbing endoplasmic reticulum proteostasis. EMBO Journal, 2022, 41, e105531.	7.8	11
149	Case 35-2006. New England Journal of Medicine, 2006, 355, 2132-2142.	27.0	10
150	No association ofDYNC1H1with sporadic ALS in a caseâ€control study of a northern European derived population: A tagging SNP approach. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2006, 7, 46-56.	2.1	9
151	Genetic susceptibility to hepatocellular carcinoma in chromosome 22q13.31, findings of a genomeâ€wide association study. JGH Open, 2021, 5, 1363-1372.	1.6	9
152	Amyotrophic Lateral Sclerosis â€" A New Role for Old Drugs. New England Journal of Medicine, 2005, 352, 1376-1378.	27.0	8
153	Endogenous retroviruses in ALS: A reawakening?. Science Translational Medicine, 2015, 7, 307fs40.	12.4	8
154	Mutant SOD1 protein increases Nav1.3 channel excitability. Journal of Biological Physics, 2016, 42, 351-370.	1.5	8
155	AAV-Mediated Gene Therapy for Glycosphingolipid Biosynthesis Deficiencies. Trends in Molecular Medicine, 2021, 27, 520-523.	6.7	8
156	Experience in Developing a Combined Activity and Verbal Group Therapy Program with Latency-Age Boys. International Journal of Group Psychotherapy, 1975, 25, 331-337.	0.6	7
157	Binding the Abdomen Can Improve Speech in Men With Phrenic Nerve Pacers. American Journal of Speech-Language Pathology, 2002, 11, 71-76.	1.8	7
158	DNA sequence analysis of the conserved region around the SOD1 gene locus in recessively inherited ALS. Neuroscience Letters, 2009, 463, 64-69.	2.1	6
159	Imaging Net Retrograde Axonal Transport In Vivo: A Physiological Biomarker. Annals of Neurology, 2022, 91, 716-729.	5.3	6
160	Neuron Research Leaps Ahead. Science, 2008, 321, 1169-1170.	12.6	5
161	Vanadium, aluminum, magnesium and manganese are not elevated in hair samples in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2010, 11, 492-493.	2.1	5
162	Defective daily temperature regulation in a mouse model of amyotrophic lateral sclerosis. Experimental Neurology, 2019, 311, 305-312.	4.1	5

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163	Lipids in Airway Mucus of Acute Quadriplegic Patients. Experimental Lung Research, 1990, 16, 369-385.	1.2	4
164	Further analysis of KIFAP3 gene in ALS patients from Switzerland and Sweden. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2017, 18, 302-304.	1.7	4
165	The Dawning of a New Era in the Molecular Biology of the Muscular Dystrophies. Brain Pathology, 1996, 6, 17-17.	4.1	3
166	International engagement by <scp>U</scp>nited <scp>S</scp>tates academic neurology departments: A national survey. Annals of Neurology, 2015, 78, 150-152.	5.3	3
167	Sodium phenylbutyrate prolongs survival and regulates expression of anti-apoptotic genes in transgenic amyotrophic lateral sclerosis mice. Journal of Neurochemistry, 2006, 96, 908-908.	3.9	2
168	Amyotrophic Lateral Sclerosis: Fuel for the Corticofugal Feud. Annals of Neurology, 2020, 88, 682-684.	5.3	2
169	The genetics of amyotrophic lateral sclerosis. , 2005, , 758-771.		0
170	Reply to comment on: A novel dysferlin mutant pseudoexon bypassed with antisense oligonucleotides. Annals of Clinical and Translational Neurology, 2015, 2, 785-785.	3.7	0
171	Juvenile Amyotrophic Lateral Sclerosis. , 2015, , 146-159.		0
172	Impact of Intralingual AAVrh10â€miRSOD1 Injection on Respiratory Function in SOD1 G93A Mice. FASEB Journal, 2018, 32, 743.7.	0.5	0