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List of Publications by Year in descending order

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96 papers	8,655 citations	53794 45 h-index	51608 86 g-index
121 all docs	121 docs citations	121 times ranked	12345 citing authors

#	Article	IF	Citations
1	C9orf72 repeat expansions cause neurodegeneration in Drosophila through arginine-rich proteins. Science, 2014, 345, 1192-1194.	12.6	632
2	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	8.1	517
3	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
4	Neurofilament light chain. Neurology, 2015, 84, 2247-2257.	1.1	412
5	Mutations in the Matrin 3 gene cause familial amyotrophic lateral sclerosis. Nature Neuroscience, 2014, 17, 664-666.	14.8	398
6	Biallelic expansion of an intronic repeat in RFC1 is a common cause of late-onset ataxia. Nature Genetics, 2019, 51, 649-658.	21.4	338
7	Large C9orf72 Hexanucleotide Repeat Expansions Are Seen in Multiple Neurodegenerative Syndromes and Are More Frequent Than Expected in the UK Population. American Journal of Human Genetics, 2013, 92, 345-353.	6.2	297
8	C9orf72 frontotemporal lobar degeneration is characterised by frequent neuronal sense and antisense RNA foci. Acta Neuropathologica, 2013, 126, 845-857.	7.7	289
9	C9orf72 hexanucleotide repeat associated with amyotrophic lateral sclerosis and frontotemporal dementia forms RNA G-quadruplexes. Scientific Reports, 2012, 2, 1016.	3.3	275
10	C9orf72 expansions in frontotemporal dementia and amyotrophic lateral sclerosis. Lancet Neurology, The, 2015, 14, 291-301.	10.2	270
11	Is SOD1 loss of function involved in amyotrophic lateral sclerosis?. Brain, 2013, 136, 2342-2358.	7.6	237
12	TDP-43 loss and ALS-risk SNPs drive mis-splicing and depletion of UNC13A. Nature, 2022, 603, 131-137.	27.8	188
13	Gâ€quadruplexes: Emerging roles in neurodegenerative diseases and the nonâ€coding transcriptome. FEBS Letters, 2015, 589, 1653-1668.	2.8	185
14	Postmortem Cortex Samples Identify Distinct Molecular Subtypes of ALS: Retrotransposon Activation, Oxidative Stress, and Activated Glia. Cell Reports, 2019, 29, 1164-1177.e5.	6.4	184
15	Gâ€quadruplexâ€binding small molecules ameliorate C9orf72 <scp>FTD</scp> / <scp>ALS</scp> pathology inÂvitro and inÂvivo. EMBO Molecular Medicine, 2018, 10, 22-31.	6.9	178
16	Nuclear import impairment causes cytoplasmic trans-activation response DNA-binding protein accumulation and is associated with frontotemporal lobar degeneration. Brain, 2010, 133, 1763-1771.	7.6	165
17	The Overlapping Genetics of Amyotrophic Lateral Sclerosis and Frontotemporal Dementia. Frontiers in Neuroscience, 2020, 14, 42.	2.8	152
18	Different Intracellular Pathomechanisms Produce DiverseMyelin Protein ZeroNeuropathies in Transgenic Mice. Journal of Neuroscience, 2006, 26, 2358-2368.	3.6	144

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19	Systemic inflammatory response and neuromuscular involvement in amyotrophic lateral sclerosis. Neurology: Neuroimmunology and NeuroInflammation, 2016, 3, e244.	6.0	129
20	Mice with endogenous <scp>TDP</scp> â€43 mutations exhibit gain of splicing function and characteristics of amyotrophic lateral sclerosis. EMBO Journal, 2018, 37, .	7.8	129
21	Homozygosity for the C9orf72 GGGGCC repeat expansion in frontotemporal dementia. Acta Neuropathologica, 2013, 126, 401-409.	7.7	126
22	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. Annals of Neurology, 2019, 85, 470-481.	5.3	118
23	Truncated stathmin-2 is a marker of TDP-43 pathology in frontotemporal dementia. Journal of Clinical Investigation, 2020, 130, 6080-6092.	8.2	117
24	The C9orf72 repeat expansion itself is methylated in ALS and FTLD patients. Acta Neuropathologica, 2015, 129, 715-727.	7.7	114
25	SOD1 and TDP-43 animal models of amyotrophic lateral sclerosis: recent advances in understanding disease toward the development of clinical treatments. Mammalian Genome, 2011, 22, 420-448.	2.2	113
26	SOD1 Function and Its Implications for Amyotrophic Lateral Sclerosis Pathology. Neuroscientist, 2015, 21, 519-529.	3.5	113
27	Proteasome Inhibition and Aggresome Formation in Sporadic Inclusion-Body Myositis and in Amyloid-Î² Precursor Protein-Overexpressing Cultured Human Muscle Fibers. American Journal of Pathology, 2005, 167, 517-526.	3.8	105
28	Humanized mutant FUS drives progressive motor neuron degeneration without aggregation in â€˜FUSDelta14â€™ knockin mice. Brain, 2017, 140, 2797-2805.	7.6	95
29	C9orf72 arginine-rich dipeptide proteins interact with ribosomal proteins in vivo to induce a toxic translational arrest that is rescued by eIF1A. Acta Neuropathologica, 2019, 137, 487-500.	7.7	94
30	Plasma neurofilament heavy chain levels and disease progression in amyotrophic lateral sclerosis: insights from a longitudinal study. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 565-573.	1.9	91
31	ATXN2 trinucleotide repeat length correlates with risk of ALS. Neurobiology of Aging, 2017, 51, 178.e1-178.e9.	3.1	86
32	Quantitative analysis of cryptic splicing associated with TDP-43 depletion. BMC Medical Genomics, 2017, 10, 38.	1.5	81
33	Cytoplasmic functions of TDP-43 and FUS and their role in ALS. Seminars in Cell and Developmental Biology, 2020, 99, 193-201.	5.0	80
34	Correlation of clinical and molecular features in spinal bulbar muscular atrophy. Neurology, 2014, 82, 2077-2084.	1.1	76
35	Evaluation of methodologies for microRNA biomarker detection by next generation sequencing. RNA Biology, 2018, 15, 1133-1145.	3.1	74
36	Whole genome sequencing for the diagnosis of neurological repeat expansion disorders in the UK: a retrospective diagnostic accuracy and prospective clinical validation study. Lancet Neurology, The, 2022, 21, 234-245.	10.2	74

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37	A comprehensive analysis of rare genetic variation in amyotrophic lateral sclerosis in the UK. Brain, 2017, 140, 1611-1618.	7.6	71
38	FUS ALS-causative mutations impair FUS autoregulation and splicing factor networks through intron retention. Nucleic Acids Research, 2020, 48, 6889-6905.	14.5	70
39	Value of systematic genetic screening of patients with amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 510-518.	1.9	69
40	The role of hnRNPs in frontotemporal dementia and amyotrophic lateral sclerosis. Acta Neuropathologica, 2020, 140, 599-623.	7.7	62
41	Circulating miR-181 is a prognostic biomarker for amyotrophic lateral sclerosis. Nature Neuroscience, 2021, 24, 1534-1541.	14.8	57
42	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 448-460.e4.	8.1	56
43	Unexpected similarities between C9ORF72 and sporadic forms of ALS/FTD suggest a common disease mechanism. ELife, 2018, 7, .	6.0	53
44	A novel SOD1-ALS mutation separates central and peripheral effects of mutant SOD1 toxicity. Human Molecular Genetics, 2015, 24, 1883-1897.	2.9	52
45	Skeletal muscle MRI differentiates SBMA and ALS and correlates with disease severity. Neurology, 2019, 93, e895-e907.	1.1	51
46	Mice Carrying ALS Mutant TDP-43, but Not Mutant FUS, Display InÂVivo Defects in Axonal Transport of Signaling Endosomes. Cell Reports, 2020, 30, 3655-3662.e2.	6.4	51
47	Screening a UK amyotrophic lateral sclerosis cohort provides evidence of multiple origins of the C9orf72 expansion. Neurobiology of Aging, 2015, 36, 546.e1-546.e7.	3.1	48
48	Beyond motor neurons: expanding the clinical spectrum in Kennedyâ€™s disease. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 808-812.	1.9	48
49	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	9.0	46
50	Muscle and not neuronal biomarkers correlate with severity in spinal and bulbar muscular atrophy. Neurology, 2019, 92, e1205-e1211.	1.1	41
51	Novel CLN3 mutation causing autophagic vacuolar myopathy. Neurology, 2014, 82, 2072-2076.	1.1	37
52	Widespread RNA metabolism impairment in sporadic inclusion body myositis TDP43-proteinopathy. Neurobiology of Aging, 2014, 35, 1491-1498.	3.1	36
53	FUS-ALS mutants alter FMRP phase separation equilibrium and impair protein translation. Science Advances, 2021, 7, .	10.3	36
54	Investigation of next-generation sequencing technologies as a diagnostic tool for amyotrophic lateral sclerosis. Neurobiology of Aging, 2015, 36, 1600.e5-1600.e8.	3.1	32

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55	Multicentre appraisal of amyotrophic lateral sclerosis biofluid biomarkers shows primacy of blood neurofilament light chain. Brain Communications, 2022, 4, fcac029.	3.3	29
56	Neuregulin 1 type III improves peripheral nerve myelination in a mouse model of congenital hypomyelinating neuropathy. Human Molecular Genetics, 2019, 28, 1260-1273.	2.9	28
57	Disease mechanism, biomarker and therapeutics for spinal and bulbar muscular atrophy (SBMA). Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 1085-1091.	1.9	28
58	An integrated multi-omic analysis of iPSC-derived motor neurons from C9ORF72 ALS patients. IScience, 2021, 24, 103221.	4.1	27
59	HnRNP K mislocalisation is a novel protein pathology of frontotemporal lobar degeneration and ageing and leads to cryptic splicing. Acta Neuropathologica, 2021, 142, 609-627.	7.7	24
60	Novel mutations in human and mouse SCN4A implicate AMPK in myotonia and periodic paralysis. Brain, 2014, 137, 3171-3185.	7.6	23
61	Uses for humanised mouse models in precision medicine for neurodegenerative disease. Mammalian Genome, 2019, 30, 173-191.	2.2	22
62	ALS-related FUS mutations alter axon growth in motoneurons and affect HuD/ELAVL4 and FMRP activity. Communications Biology, 2021, 4, 1025.	4.4	21
63	Cell environment shapes TDP-43 function with implications in neuronal and muscle disease. Communications Biology, 2022, 5, 314.	4.4	21
64	Transcriptomic analysis of frontotemporal lobar degeneration with TDP-43 pathology reveals cellular alterations across multiple brain regions. Acta Neuropathologica, 2022, 143, 383-401.	7.7	20
65	Deficiency of the zinc finger protein ZFP106 causes motor and sensory neurodegeneration. Human Molecular Genetics, 2016, 25, 291-307.	2.9	19
66	A Nonsense Mutation in Mouse Tardbp Affects TDP43 Alternative Splicing Activity and Causes Limb-Clasping and Body Tone Defects. PLoS ONE, 2014, 9, e85962.	2.5	18
67	Profilin1 E117G is a moderate risk factor for amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 506-508.	1.9	17
68	Sequencing analysis of the spinal bulbar muscular atrophy CAG expansion reveals absence of repeat interruptions. Neurobiology of Aging, 2014, 35, 443.e1-443.e3.	3.1	16
69	Recent advances in bulbar syndromes. Current Opinion in Neurology, 2014, 27, 506-514.	3.6	15
70	Clinical Trials in Spinal and Bulbar Muscular Atrophyâ€"Past, Present, and Future. Journal of Molecular Neuroscience, 2016, 58, 379-387.	2.3	15
71	P0S63del impedes the arrival of wild-type P0 glycoprotein to myelin in CMT1B mice. Human Molecular Genetics, 2011, 20, 2081-2090.	2.9	14
72	The snowball effect of RNA binding protein dysfunction in amyotrophic lateral sclerosis. Brain, 2018, 141, 1236-1238.	7.6	14

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73	A nonsense mutation in myelin protein zero causes congenital hypomyelination neuropathy through altered P0 membrane targeting and gain of abnormal function. Human Molecular Genetics, 2019, 28, 124-132.	2.9	12
74	Travelling Together: A Unifying Pathomechanism forÂALS. Trends in Neurosciences, 2020, 43, 1-2.	8.6	12
75	NMJ-Analyser identifies subtle early changes in mouse models of neuromuscular disease. Scientific Reports, 2021, 11, 12251.	3.3	12
76	Plasma pNfH levels differentiate SBMA from ALS. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 215-217.	1.9	11
77	HnRNP K mislocalisation in neurons of the dentate nucleus is a novel neuropathological feature of neurodegenerative disease and ageing. Neuropathology and Applied Neurobiology, 2022, 48, .	3.2	11
78	Graphical modelling of molecular networks underlying sporadic inclusion body myositis. Molecular BioSystems, 2013, 9, 1736.	2.9	10
79	Towards a European Registry and Biorepository for Patients with Spinal and Bulbar Muscular Atrophy. Journal of Molecular Neuroscience, 2016, 58, 394-400.	2.3	10
80	No effect of <scp>AR</scp> polyG polymorphism on spinal and bulbar muscular atrophy phenotype. European Journal of Neurology, 2016, 23, 1134-1136.	3.3	8
81	Humoral response to neurofilaments and dipeptide repeats in ALS progression. Annals of Clinical and Translational Neurology, 2021, 8, 1831-1844.	3.7	8
82	CHCHD10Pro34Ser is not a highly penetrant pathogenic variant for amyotrophic lateral sclerosis and frontotemporal dementia. Brain, 2016, 139, e9-e9.	7.6	7
83	TDP-43 mutations increase HNRNP A1-7B through gain of splicing function. Brain, 2018, 141, e83-e83.	7.6	7
84	A Comparison of Low Read Depth QuantSeq 3â€² Sequencing to Total RNA-Seq in FUS Mutant Mice. Frontiers in Genetics, 2020, 11, 562445.	2.3	6
85	AR cooperates with SMAD4 to maintain skeletal muscle homeostasis. Acta Neuropathologica, 2022, 143, 713-731.	7.7	6
86	FUS is not dysregulated by the spinal bulbar muscular atrophy androgen receptor polyglutamine repeat expansion. Neurobiology of Aging, 2013, 34, 1516.e17-1516.e19.	3.1	5
87	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. SSRN Electronic Journal, 0, , .	0.4	4
88	Antisense makes sense for amyotrophic lateral sclerosis. Lancet Neurology, The, 2013, 12, 416-417.	10.2	3
89	Neuromuscular diseases: progress in gene discovery drives diagnostics and therapeutics. Lancet Neurology, The, 2015, 14, 13-14.	10.2	3
90	Musclesense: a Trained, Artificial Neural Network for the Anatomical Segmentation of Lower Limb Magnetic Resonance Images in Neuromuscular Diseases. Neuroinformatics, 2021, 19, 379-383.	2.8	2

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91	An unusual presentation for SOD1â€ALS: Isolated facial diplegia. Muscle and Nerve, 2013, 48, 994-995.	2.2	1
92	DNA Editing for Amyotrophic Lateral Sclerosis: Leading Off First Base. CRISPR Journal, 2020, 3, 75-77.	2.9	1
93	ALS Mice Carrying Pathological Mutant TDP-43, But Not Mutant FUS, Display Axonal Transport Defects <i>in vivo</i>. SSRN Electronic Journal, 0, , .	0.4	1
94	Post-transcriptional modifications caused by TDP-43 mutations in mouse and man. SpringerPlus, 2015, 4, L52.	1.2	0
95	Markers of cognitive resilience and a framework for investigating clinical heterogeneity in <scp>ALS</scp> â€. Journal of Pathology, 2022, , .	4.5	0
96	The role of the RNA binding protein HnRNP K in the pathogenesis of frontotemporal lobar degeneration.. Alzheimer's and Dementia, 2021, 17 Suppl 3, e052826.	0.8	0