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List of Publications by Year
in descending order

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107 papers	5,443 citations	109321 35 h-index	98798 67 g-index
110 all docs	110 docs citations	110 times ranked	7837 citing authors

#	Article	IF	Citations
1	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	8.1	517
2	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
3	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. Neuron, 2014, 84, 324-331.	8.1	308
4	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
5	Disruption of skeletal muscle mitochondrial network genes and miRNAs in amyotrophic lateral sclerosis. Neurobiology of Disease, 2013, 49, 107-117.	4.4	194
6	Mutations of FUS gene in sporadic amyotrophic lateral sclerosis. Journal of Medical Genetics, 2010, 47, 190-194.	3.2	152
7	Human neural stem cell transplantation in ALS: initial results from a phase I trial. Journal of Translational Medicine, 2015, 13, 17.	4.4	151
8	Diagnostic and Prognostic Biomarkers in Amyotrophic Lateral Sclerosis. JAMA Neurology, 2017, 74, 525.	9.0	139
9	Spinal and bulbar muscular atrophy: Skeletal muscle pathology in male patients and heterozygous females. Journal of the Neurological Sciences, 2008, 264, 100-105.	0.6	133
10	Human skeletal muscle atrophy in amyotrophic lateral sclerosis reveals a reduction in Akt and an increase in atroginâ€1. FASEB Journal, 2006, 20, 583-585.	0.5	127
11	A genome-wide association meta-analysis identifies a novel locus at 17q11.2 associated with sporadic amyotrophic lateral sclerosis. Human Molecular Genetics, 2014, 23, 2220-2231.	2.9	123
12	Factors predicting survival in ALS: a multicenter Italian study. Journal of Neurology, 2017, 264, 54-63.	3.6	96
13	Rapamycin treatment for amyotrophic lateral sclerosis. Medicine (United States), 2018, 97, e11119.	1.0	96
14	Humanized mutant FUS drives progressive motor neuron degeneration without aggregation in â€˜FUSDelta14â€™ knockin mice. Brain, 2017, 140, 2797-2805.	7.6	95
15	ATXN2 trinucleotide repeat length correlates with risk of ALS. Neurobiology of Aging, 2017, 51, 178.e1-178.e9.	3.1	86
16	C9ORF72 repeat expansion in a large Italian ALS cohort: evidence of a founder effect. Neurobiology of Aging, 2012, 33, 2528.e7-2528.e14.	3.1	74
17	Ubiquilin 2mutations in Italian patients with amyotrophic lateral sclerosis and frontotemporal dementia. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 183-187.	1.9	74
18	Glycolytic-to-oxidative fiber-type switch and mTOR signaling activation are early-onset features of SBMA muscle modified by high-fat diet. Acta Neuropathologica, 2016, 132, 127-144.	7.7	74

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19	Results from Phase I Clinical Trial with Intraspinal Injection of Neural Stem Cells in Amyotrophic Lateral Sclerosis: A Long-Term Outcome. Stem Cells Translational Medicine, 2019, 8, 887-897.	3.3	71
20	FUS ALS-causative mutations impair FUS autoregulation and splicing factor networks through intron retention. Nucleic Acids Research, 2020, 48, 6889-6905.	14.5	70
21	Neurofilaments in motor neuron disorders: towards promising diagnostic and prognostic biomarkers. Molecular Neurodegeneration, 2020, 15, 58.	10.8	68
22	Safety and efficacy of nabiximols on spasticity symptoms in patients with motor neuron disease (CANALS): a multicentre, double-blind, randomised, placebo-controlled, phase 2 trial. Lancet Neurology, The, 2019, 18, 155-164.	10.2	63
23	Non-neural phenotype of spinal and bulbar muscular atrophy: results from a large cohort of Italian patients. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 810-816.	1.9	59
24	Genetic Modifiers of Duchenne Muscular Dystrophy and Dilated Cardiomyopathy. PLoS ONE, 2015, 10, e0141240.	2.5	58
25	Association of a Locus in theCAMTA1Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. JAMA Neurology, 2016, 73, 812.	9.0	57
26	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 448-460.e4.	8.1	56
27	Multimodal structural MRI in the diagnosis of motor neuron diseases. NeuroImage: Clinical, 2017, 16, 240-247.	2.7	55
28	Clinical and molecular characterization of limbâ€girdle muscular dystrophy due to LAMA2 mutations. Muscle and Nerve, 2011, 44, 703-709.	2.2	52
29	Beyond motor neurons: expanding the clinical spectrum in Kennedyâ€™s disease. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 808-812.	1.9	48
30	Pilot trial of clenbuterol in spinal and bulbar muscular atrophy. Neurology, 2013, 80, 2095-2098.	1.1	47
31	TUBA4A gene analysis in sporadic amyotrophic lateral sclerosis: identification of novel mutations. Journal of Neurology, 2015, 262, 1376-1378.	3.6	44
32	Proteostasis and ALS: protocol for a phase II, randomised, double-blind, placebo-controlled, multicentre clinical trial for colchicine in ALS (Co-ALS). BMJ Open, 2019, 9, e028486.	1.9	44
33	Androgen-dependent impairment of myogenesis in spinal and bulbar muscular atrophy. Acta Neuropathologica, 2013, 126, 109-121.	7.7	41
34	Muscle and not neuronal biomarkers correlate with severity in spinal and bulbar muscular atrophy. Neurology, 2019, 92, e1205-e1211.	1.1	41
35	Onset Manifestations of Spinal and Bulbar Muscular Atrophy (Kennedyâ€™s Disease). Journal of Molecular Neuroscience, 2016, 58, 321-329.	2.3	40
36	Clinical and molecular cross-sectional study of a cohort of adult type III spinal muscular atrophy patients: clues from a biomarker study. European Journal of Human Genetics, 2013, 21, 630-636.	2.8	39

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37	TBK1mutations in Italian patients with amyotrophic lateral sclerosis: genetic and functional characterisation. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 869-875.	1.9	38
38	Skeletal Muscle Satellite Cells in Amyotrophic Lateral Sclerosis. Ultrastructural Pathology, 2014, 38, 295-302.	0.9	37
39	Increased mitophagy in the skeletal muscle of spinal and bulbar muscular atrophy patients. Human Molecular Genetics, 2017, 26, ddx019.	2.9	37
40	The Italian multicenter experience with edaravone in amyotrophic lateral sclerosis. Journal of Neurology, 2020, 267, 3258-3267.	3.6	37
41	Functional changes in Becker muscular dystrophy: implications for clinical trials in dystrophinopathies. Scientific Reports, 2016, 6, 32439.	3.3	36
42	Screening of the PFN1 gene in sporadic amyotrophic lateral sclerosis and in frontotemporal dementia. Neurobiology of Aging, 2013, 34, 1517.e9-1517.e10.	3.1	35
43	Muscle MRI and functional outcome measures in Becker muscular dystrophy. Scientific Reports, 2017, 7, 16060.	3.3	35
44	Comparative Analysis of C9orf72 and Sporadic Disease in a Large Multicenter ALS Population: The Effect of Male Sex on Survival of C9orf72 Positive Patients. Frontiers in Neuroscience, 2019, 13, 485.	2.8	35
45	The unfolded protein response in amyotrophic later sclerosis: results of a phase 2 trial. Brain, 2021, 144, 2635-2647.	7.6	33
46	Erythropoietin in amyotrophic lateral sclerosis: a multicentre, randomised, double blind, placebo controlled, phase III study. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 879-886.	1.9	32
47	Cardiomyopathy in patients with POMT1-related congenital and limb-girdle muscular dystrophy. European Journal of Human Genetics, 2012, 20, 1234-1239.	2.8	31
48	Progress in enzyme replacement therapy in glycogen storage disease type II. Therapeutic Advances in Neurological Disorders, 2009, 2, 143-153.	3.5	30
49	Analysis of hnRNPA1, A2/B1, and A3 genes in patients with amyotrophic lateral sclerosis. Neurobiology of Aging, 2013, 34, 2695.e11-2695.e12.	3.1	30
50	Longitudinal evaluation of SMN levels as biomarker for spinal muscular atrophy: results of a phase IIb double-blind study of salbutamol. Journal of Medical Genetics, 2019, 56, 293-300.	3.2	30
51	Natural history of upper motor neuron-dominant ALS. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2010, 11, 424-429.	2.1	29
52	Hypnosis-based psychodynamic treatment in ALS: a longitudinal study on patients and their caregivers. Frontiers in Psychology, 2015, 6, 822.	2.1	29
53	Clinical and molecular study in a long-surviving patient with MLASA syndrome due to novel PUS1 mutations. Neurogenetics, 2016, 17, 65-70.	1.4	29
54	No association of DPP6 with amyotrophic lateral sclerosis in an Italian population. Neurobiology of Aging, 2011, 32, 966-967.	3.1	28

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55	Safety, tolerability, and preliminary efficacy of an IGF-1 mimetic in patients with spinal and bulbar muscular atrophy: a randomised, placebo-controlled trial. Lancet Neurology, The, 2018, 17, 1043-1052.	10.2	28
56	Female gender doubles executive dysfunction risk in ALS: a case-control study in 165 patients. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 574-579.	1.9	26
57	Beta-agonist stimulation ameliorates the phenotype of spinal and bulbar muscular atrophy mice and patient-derived myotubes. Scientific Reports, 2017, 7, 41046.	3.3	26
58	ALS risk but not phenotype is affected by ataxin-2 intermediate length polyglutamine expansion. Neurology, 2011, 76, 2030-2031.	1.1	25
59	Sorting Rare ALS Genetic Variants by Targeted Re-Sequencing Panel in Italian Patients: OPTN, VCP, and SQSTM1 Variants Account for 3% of Rare Genetic Forms. Journal of Clinical Medicine, 2020, 9, 412.	2.4	24
60	SPP1 genotype and glucocorticoid treatment modify osteopontin expression in Duchenne muscular dystrophy cells. Human Molecular Genetics, 2017, 26, 3342-3351.	2.9	23
61	MEF2 impairment underlies skeletal muscle atrophy in polyglutamine disease. Acta Neuropathologica, 2020, 140, 63-80.	7.7	23
62	Psychopathological features and suicidal ideation in amyotrophic lateral sclerosis patients. Neurological Sciences, 2010, 31, 735-740.	1.9	22
63	TGFBR2 but not SPP1 genotype modulates osteopontin expression in Duchenne muscular dystrophy muscle. Journal of Pathology, 2012, 228, 251-259.	4.5	22
64	Theory of mind, empathy and neuropsychological functioning in X-linked Spinal and Bulbar Muscular Atrophy: a controlled study of 20 patients. Journal of Neurology, 2015, 262, 394-401.	3.6	21
65	Intraspinal stem cell transplantation for amyotrophic lateral sclerosis: Ready for efficacy clinical trials?. Cytotherapy, 2016, 18, 1471-1475.	0.7	21
66	Polyglutamine-Expanded Androgen Receptor Alteration of Skeletal Muscle Homeostasis and Myonuclear Aggregation Are Affected by Sex, Age and Muscle Metabolism. Cells, 2020, 9, 325.	4.1	21
67	TDP-43 in skeletal muscle of patients affected with amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2010, 11, 240-243.	2.1	19
68	Efficacy of Hypnosis-Based Treatment in Amyotrophic Lateral Sclerosis: A Pilot Study. Frontiers in Psychology, 2012, 3, 465.	2.1	19
69	Comorbidity of dementia with amyotrophic lateral sclerosis (ALS): insights from a large multicenter Italian cohort. Journal of Neurology, 2017, 264, 2224-2231.	3.6	19
70	Decoding distinctive features of plasma extracellular vesicles in amyotrophic lateral sclerosis. Molecular Neurodegeneration, 2021, 16, 52.	10.8	19
71	Parkinson-like features in ALS with predominant upper motor neuron involvement. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 137-143.	2.1	18
72	Collagen XIX Alpha 1 Improves Prognosis in Amyotrophic Lateral Sclerosis. , 2019, 10, 278.		18

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73	Mutational analysis of VCP gene in familial amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 630.e1-630.e2.	3.1	17
74	210th ENMC International Workshop: Research and clinical management of patients with spinal and bulbar muscular atrophy, 27â€“29 March, 2015, Naarden, The Netherlands. Neuromuscular Disorders, 2015, 25, 802-812.	0.6	16
75	Impact on children of a parent with ALS: a case-control study. Frontiers in Psychology, 2015, 6, 288.	2.1	16
76	Safety and efficacy of edaravone compared to historical controls in patients with amyotrophic lateral sclerosis from North-Eastern Italy. Journal of the Neurological Sciences, 2019, 404, 47-51.	0.6	16
77	Transforming growth factor beta 1 signaling is altered in the spinal cord and muscle of amyotrophic lateral sclerosis mice and patients. Neurobiology of Aging, 2019, 82, 48-59.	3.1	15
78	A pilot trial with clenbuterol in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2006, 7, 252-254.	2.1	14
79	No evidence of cardiomyopathy in spinal and bulbar muscular atrophy. Acta Neurologica Scandinavica, 2013, 128, e30-e32.	2.1	14
80	Protein misfolding, amyotrophic lateral sclerosis and guanabenz: protocol for a phase II RCT with futility design (ProMISe trial). BMJ Open, 2017, 7, e015434.	1.9	14
81	New <scp>FIG</scp>4 gene mutations causing aggressive <scp>ALS</scp>. European Journal of Neurology, 2018, 25, e41-e42.	3.3	14
82	The clinical spectrum of CASQ1-related myopathy. Neurology, 2018, 91, e1629-e1641.	1.1	14
83	Clinical features and outcomes of the flail arm and flail leg and pure lower motor neuron MND variants: a multicentre Italian study. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 1001-1003.	1.9	14
84	Muscle histopathology in upper motor neuron-dominant amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2008, 9, 287-293.	2.1	13
85	Improving the knowledge of amyotrophic lateral sclerosis genetics: novel SOD1 and FUS variants. Neurobiology of Aging, 2014, 35, 1212.e7-1212.e10.	3.1	13
86	Brain MRI shows white matter sparing in Kennedy's disease and slowâ€progressing lower motor neuron disease. Human Brain Mapping, 2019, 40, 3102-3112.	3.6	12
87	Insights into the genetic epidemiology of spinal and bulbar muscular atrophy: prevalence estimation and multiple founder haplotypes in the Veneto Italian region. European Journal of Neurology, 2019, 26, 519-524.	3.3	12
88	Evaluation of peripherin in biofluids of patients with motor neuron diseases. Annals of Clinical and Translational Neurology, 2021, 8, 1750-1754.	3.7	11
89	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
90	Cardiac Function in Types II and III Spinal Muscular Atrophy: Should We Change Standards of Care?. Neuropediatrics, 2015, 46, 033-036.	0.6	9

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91	Validation of the Italian version of the SBMA Functional Rating Scale as outcome measure. Neurological Sciences, 2016, 37, 1815-1821.	1.9	9
92	Brainstem glucose hypermetabolism in ALS/FTD and shorten survival: a ¹⁸F-FDG PET/MR study. Journal of Nuclear Medicine, 2021, , jnumed.121.262232.	5.0	9
93	Specific numerical processing impairment in ALS patients. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 6-12.	1.7	8
94	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
95	Unimpaired Neuropsychological Performance and Enhanced Memory Recall in Patients with Sbma: A Large Sample Comparative Study. Scientific Reports, 2018, 8, 13627.	3.3	8
96	MetabolicÂalterations in spinal and bulbar muscular atrophy. Revue Neurologique, 2020, 176, 780-787.	1.5	7
97	Empathy-based supportive treatment in amyotrophic lateral sclerosis: A pragmatic study. American Journal of Clinical Hypnosis, 2021, 63, 202-216.	0.6	7
98	Enhanced Neural Empathic Responses in Patients with Spino-Bulbar Muscular Atrophy: An Electrophysiological Study. Brain Sciences, 2021, 11, 16.	2.3	7
99	The relevance of migraine in the clinical spectrum of mitochondrial disorders. Scientific Reports, 2022, 12, 4222.	3.3	7
100	Mitochondrial implications in bulbospinal muscular atrophy (Kennedy disease). Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2016, 17, 112-118.	1.7	6
101	Quality of life assessment in adult spinal muscular atrophy patients treated with nusinersen. Journal of Neurology, 2022, 269, 3264-3275.	3.6	6
102	AR cooperates with SMAD4 to maintain skeletal muscle homeostasis. Acta Neuropathologica, 2022, 143, 713-731.	7.7	6
103	CSF Heavy Neurofilament May Discriminate and Predict Motor Neuron Diseases with Upper Motor Neuron Involvement. Biomedicines, 2021, 9, 1623.	3.2	4
104	The Role of Motor System in Mental Rotation: New Insights from Myotonic Dystrophy Type 1. Journal of the International Neuropsychological Society, 2020, 26, 492-502.	1.8	3
105	The blurred scenario of motor neuron disorders linked toSpatacsinmutations: a case report. European Journal of Neurology, 2014, 21, e85-e86.	3.3	2
106	Preliminary design and validation of the â€œ6-K-scaleâ€•for bulbar symptoms evaluation in SBMA. Neurological Sciences, 2019, 40, 1393-1401.	1.9	2
107	Disease Propagation in Amyotrophic Lateral Sclerosis: Insights and Hopes. Journal of Neurology & Stroke, 2015, 2, .	0.1	0