Leonard H Van Den Berg

List of Publications by Year
in descending order

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304 papers	21,601 citations	15504 65 h-index	14208 128 g-index
314 all docs	314 docs citations	314 times ranked	27702 citing authors

#	Article	IF	Citations
1	Systematic identification of trans eQTLs as putative drivers of known disease associations. Nature Genetics, 2013, 45, 1238-1243.	21.4	1,544
2	Amyotrophic lateral sclerosis. Nature Reviews Disease Primers, 2017, 3, 17071.	30.5	885
3	Amyotrophic lateral sclerosis. Lancet, The, 2017, 390, 2084-2098.	13.7	867
4	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. Science, 2015, 347, 1436-1441.	12.6	823
5	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	8.1	517
6	Clinical diagnosis and management of amyotrophic lateral sclerosis. Nature Reviews Neurology, 2011, 7, 639-649.	10.1	503
7	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
8	Haploinsufficiency leads to neurodegeneration in C9ORF72 ALS/FTD human induced motor neurons. Nature Medicine, 2018, 24, 313-325.	30.7	445
9	Microglia innately develop within cerebral organoids. Nature Communications, 2018, 9, 4167.	12.8	405
10	Disease variants alter transcription factor levels and methylation of their binding sites. Nature Genetics, 2017, 49, 131-138.	21.4	390
11	Identification of context-dependent expression quantitative trait loci in whole blood. Nature Genetics, 2017, 49, 139-145.	21.4	363
12	Prognosis for patients with amyotrophic lateral sclerosis: development and validation of a personalised prediction model. Lancet Neurology, The, 2018, 17, 423-433.	10.2	342
13	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. Neuron, 2014, 84, 324-331.	8.1	308
14	Analysis of amyotrophic lateral sclerosis as a multistep process: a population-based modelling study. Lancet Neurology, The, 2014, 13, 1108-1113.	10.2	302
15	Amyotrophic lateral sclerosis: moving towards a new classification system. Lancet Neurology, The, 2016, 15, 1182-1194.	10.2	301
16	Detection of long repeat expansions from PCR-free whole-genome sequence data. Genome Research, 2017, 27, 1895-1903.	5.5	277
17	Multidisciplinary ALS care improves quality of life in patients with ALS. Neurology, 2005, 65, 1264-1267.	1.1	273
18	Population based epidemiology of amyotrophic lateral sclerosis using capture-recapture methodology. Journal of Neurology, Neurosurgery and Psychiatry, 2011, 82, 1165-1170.	1.9	273

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19	A proposal for new diagnostic criteria for ALS. Clinical Neurophysiology, 2020, 131, 1975-1978.	1.5	268
20	A randomized sequential trial of creatine in amyotrophic lateral sclerosis. Annals of Neurology, 2003, 53, 437-445.	5.3	260
21	Gene discovery in amyotrophic lateral sclerosis: implications for clinical management. Nature Reviews Neurology, 2017, 13, 96-104.	10.1	245
22	Population genetic differentiation of height and body mass index across Europe. Nature Genetics, 2015, 47, 1357-1362.	21.4	227
23	<scp>C</scp>9orf72 ablation in mice does not cause motor neuron degeneration or motor deficits. Annals of Neurology, 2015, 78, 426-438.	5.3	225
24	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
25	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	21.4	218
26	Dexpramipexole versus placebo for patients with amyotrophic lateral sclerosis (EMPOWER): a randomised, double-blind, phase 3 trial. Lancet Neurology, The, 2013, 12, 1059-1067.	10.2	216
27	Hypermetabolism in ALS is associated with greater functional decline and shorter survival. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 1016-1023.	1.9	177
28	TDP-43 proteinopathies: a new wave of neurodegenerative diseases. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 86-95.	1.9	174
29	Multifocal motor neuropathy: Diagnostic criteria that predict the response to immunoglobulin treatment. Annals of Neurology, 2000, 48, 919-926.	5.3	164
30	Genome-wide association study of intracranial aneurysms identifies 17 risk loci and genetic overlap with clinical risk factors. Nature Genetics, 2020, 52, 1303-1313.	21.4	163
31	A natural history study of late onset spinal muscular atrophy types 3b and 4. Journal of Neurology, 2008, 255, 1400-1404.	3.6	158
32	Blood lipids influence DNA methylation in circulating cells. Genome Biology, 2016, 17, 138.	8.8	154
33	The role of TREM2 R47H as a risk factor for Alzheimer's disease, frontotemporal lobar degeneration, amyotrophic lateral sclerosis, and Parkinson's disease. Alzheimer's and Dementia, 2015, 11, 1407-1416.	0.8	152
34	Improving clinical trial outcomes in amyotrophic lateral sclerosis. Nature Reviews Neurology, 2021, 17, 104-118.	10.1	152
35	Deciphering amyotrophic lateral sclerosis: What phenotype, neuropathology and genetics are telling us about pathogenesis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 5-18.	1.7	142
36	Diagnostic value of sonography in treatment-naive chronic inflammatory neuropathies. Neurology, 2017, 88, 143-151.	1.1	135

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37	Deep learning predictions of survival based on MRI in amyotrophic lateral sclerosis. NeuroImage: Clinical, 2017, 13, 361-369.	2.7	135
38	Muscle strength and motor function throughout life in a crossâ€sectional cohort of 180 patients with spinal muscular atrophy types 1câ€“4. European Journal of Neurology, 2018, 25, 512-518.	3.3	126
39	Age-related accrual of methylomic variability is linked to fundamental ageing mechanisms. Genome Biology, 2016, 17, 191.	8.8	120
40	A blinded international study on the reliability of genetic testing for GGGGCC-repeat expansions inC9orf72reveals marked differences in results among 14 laboratories. Journal of Medical Genetics, 2014, 51, 419-424.	3.2	118
41	Primary lateral sclerosis: consensus diagnostic criteria. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 373-377.	1.9	118
42	Cell Specific eQTL Analysis without Sorting Cells. PLoS Genetics, 2015, 11, e1005223.	3.5	115
43	Genetic correlation between amyotrophic lateral sclerosis and schizophrenia. Nature Communications, 2017, 8, 14774.	12.8	114
44	Comparative interactomics analysis of different ALS-associated proteins identifies converging molecular pathways. Acta Neuropathologica, 2016, 132, 175-196.	7.7	113
45	Homozygous deletion of the survival motor neuron 2 gene is a prognostic factor in sporadic ALS. Neurology, 2001, 56, 749-752.	1.1	111
46	Randomized sequential trial of valproic acid in amyotrophic lateral sclerosis. Annals of Neurology, 2009, 66, 227-234.	5.3	111
47	Revised Airlie House consensus guidelines for design and implementation of ALS clinical trials. Neurology, 2019, 92, e1610-e1623.	1.1	105
48	Full ablation of C9orf72 in mice causes immune system-related pathology and neoplastic events but no motor neuron defects. Acta Neuropathologica, 2016, 132, 145-147.	7.7	104
49	Negative selection in humans and fruit flies involves synergistic epistasis. Science, 2017, 356, 539-542.	12.6	103
50	Genomic signals of migration and continuity in Britain before the Anglo-Saxons. Nature Communications, 2016, 7, 10326.	12.8	100
51	Incidence of polyneuropathy in Utrecht, the Netherlands. Neurology, 2015, 84, 259-264.	1.1	95
52	Safety and efficacy of olesoxime in patients with type 2 or non-ambulatory type 3 spinal muscular atrophy: a randomised, double-blind, placebo-controlled phase 2 trial. Lancet Neurology, The, 2017, 16, 513-522.	10.2	95
53	SMN genotypes producing less SMN protein increase susceptibility to and severity of sporadic ALS. Neurology, 2005, 65, 820-825.	1.1	94
54	Cortical thickness in ALS: towards a marker for upper motor neuron involvement. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 288-294.	1.9	94

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55	Association of motor milestones, SMN2 copy and outcome in spinal muscular atrophy types 0â€“4. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 365-367.	1.9	94
56	Cross-ethnic meta-analysis identifies association of the GPX3-TNIP1 locus with amyotrophic lateral sclerosis. Nature Communications, 2017, 8, 611.	12.8	93
57	The ALS-FTD-Q. Neurology, 2012, 79, 1377-1383.	1.1	91
58	The changing picture of amyotrophic lateral sclerosis: lessons from European registers. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 557-563.	1.9	89
59	The Combined Assessment of Function and Survival (CAFS): A new endpoint for ALS clinical trials. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 162-168.	1.7	88
60	Effect of Presymptomatic Body Mass Index and Consumption of Fat and Alcohol on Amyotrophic Lateral Sclerosis. JAMA Neurology, 2015, 72, 1155.	9.0	87
61	ATXN2 trinucleotide repeat length correlates with risk of ALS. Neurobiology of Aging, 2017, 51, 178.e1-178.e9.	3.1	86
62	Associations of autozygosity with a broad range of human phenotypes. Nature Communications, 2019, 10, 4957.	12.8	84
63	Joint sequencing of human and pathogen genomes reveals the genetics of pneumococcal meningitis. Nature Communications, 2019, 10, 2176.	12.8	83
64	Transethnic Genome-Wide Association Study Provides Insights in the Genetic Architecture and Heritability of Long QT Syndrome. Circulation, 2020, 142, 324-338.	1.6	83
65	Meta-analysis of pharmacogenetic interactions in amyotrophic lateral sclerosis clinical trials. Neurology, 2017, 89, 1915-1922.	1.1	82
66	Caregiver burden in amyotrophic lateral sclerosis: A systematic review. Palliative Medicine, 2018, 32, 231-245.	3.1	82
67	Subcortical structures in amyotrophic lateral sclerosis. Neurobiology of Aging, 2015, 36, 1075-1082.	3.1	78
68	Shared vulnerability for connectome alterations across psychiatric and neurological brain disorders. Nature Human Behaviour, 2019, 3, 988-998.	12.0	75
69	Genetic variants associated with longitudinal changes in brain structure across the lifespan. Nature Neuroscience, 2022, 25, 421-432.	14.8	75
70	Lithium lacks effect on survival in amyotrophic lateral sclerosis: a phase IIb randomised sequential trial. Journal of Neurology, Neurosurgery and Psychiatry, 2012, 83, 557-564.	1.9	74
71	Simulating disease propagation across white matter connectome reveals anatomical substrate for neuropathology staging in amyotrophic lateral sclerosis. NeuroImage, 2016, 124, 762-769.	4.2	74
72	Widespread structural brain involvement in ALS is not limited to theC9orf72repeat expansion. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 1354-1360.	1.9	69

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73	Cardiac pathology in spinal muscular atrophy: a systematic review. Orphanet Journal of Rare Diseases, 2017, 12, 67.	2.7	67
74	Brain morphologic changes in asymptomatic C9orf72 repeat expansion carriers. Neurology, 2015, 85, 1780-1788.	1.1	66
75	Refining eligibility criteria for amyotrophic lateral sclerosis clinical trials. Neurology, 2019, 92, .	1.1	66
76	Kuramoto model simulation of neural hubs and dynamic synchrony in the human cerebral connectome. BMC Neuroscience, 2015, 16, 54.	1.9	65
77	KIF1A variants are a frequent cause of autosomal dominant hereditary spastic paraplegia. European Journal of Human Genetics, 2020, 28, 40-49.	2.8	65
78	Large-scale SOD1 mutation screening provides evidence for genetic heterogeneity in amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, 562-566.	1.9	64
79	Safety and tolerability of Triumeq in amyotrophic lateral sclerosis: the Lighthouse trial. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2019, 20, 595-604.	1.7	63
80	Safety and efficacy of ozanezumab in patients with amyotrophic lateral sclerosis: a randomised, double-blind, placebo-controlled, phase 2 trial. Lancet Neurology, The, 2017, 16, 208-216.	10.2	62
81	Monitoring disease progression with plasma creatinine in amyotrophic lateral sclerosis clinical trials. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 156-161.	1.9	62
82	Edaravone: a new treatment for ALS on the horizon?. Lancet Neurology, The, 2017, 16, 490-491.	10.2	61
83	The project MinE databrowser: bringing large-scale whole-genome sequencing in ALS to researchers and the public. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2019, 20, 432-440.	1.7	60
84	Addressing heterogeneity in amyotrophic lateral sclerosis CLINICAL TRIALS. Muscle and Nerve, 2020, 62, 156-166.	2.2	60
85	Associations between psychological factors and health-related quality of life and global quality of life in patients with ALS: a systematic review. Health and Quality of Life Outcomes, 2016, 14, 107.	2.4	58
86	Association of a Locus in theCAMTA1Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. JAMA Neurology, 2016, 73, 812.	9.0	57
87	Natural history of lung function in spinal muscular atrophy. Orphanet Journal of Rare Diseases, 2020, 15, 88.	2.7	56
88	The current use of telehealth in ALS care and the barriers to and facilitators of implementation: a systematic review. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2020, 21, 167-182.	1.7	55
89	Genome-wide association analyses identify new Brugada syndrome risk loci and highlight a new mechanism of sodium channel regulation in disease susceptibility. Nature Genetics, 2022, 54, 232-239.	21.4	55
90	SMN1 gene duplications are associated with sporadic ALS. Neurology, 2012, 78, 776-780.	1.1	54

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91	Long-Term Air Pollution Exposure and Amyotrophic Lateral Sclerosis in Netherlands: A Population-based Caseâ€“control Study. Environmental Health Perspectives, 2017, 125, 097023.	6.0	54
92	Population-based analysis of survival in spinal muscular atrophy. Neurology, 2020, 94, e1634-e1644.	1.1	54
93	Autoantibody pathogenicity in a multifocal motor neuropathy induced pluripotent stem cellâ€“derived model. Annals of Neurology, 2016, 80, 71-88.	5.3	53
94	A comparative study of brachial plexus sonography and magnetic resonance imaging in chronic inflammatory demyelinating neuropathy and multifocal motor neuropathy. European Journal of Neurology, 2017, 24, 1307-1313.	3.3	51
95	Connectomeâ€Based Propagation Model in Amyotrophic Lateral Sclerosis. Annals of Neurology, 2020, 87, 725-738.	5.3	51
96	Distinctive patterns of sonographic nerve enlargement in Charcotâ€“Marieâ€“Tooth type 1A and hereditary neuropathy with pressure palsies. Clinical Neurophysiology, 2015, 126, 1413-1420.	1.5	49
97	Meta-analysis of genome-wide DNA methylation identifies shared associations across neurodegenerative disorders. Genome Biology, 2021, 22, 90.	8.8	49
98	A case-control study of hormonal exposures as etiologic factors for ALS in women. Neurology, 2017, 89, 1283-1290.	1.1	48
99	Multifocal motor neuropathy: controversies and priorities. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 140-148.	1.9	48
100	The frontotemporal syndrome of ALS is associated with poor survival. Journal of Neurology, 2016, 263, 2476-2483.	3.6	46
101	Multimodal longitudinal study of structural brain involvement in amyotrophic lateral sclerosis. Neurology, 2020, 94, e2592-e2604.	1.1	46
102	Prognostic value of weight loss in patients with amyotrophic lateral sclerosis: a population-based study. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 867-875.	1.9	46
103	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	9.0	46
104	Multicentre, cross-cultural, population-based, caseâ€“control study of physical activity as risk factor for amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2018, 89, 797-803.	1.9	45
105	Cognitive and behavioural changes in PLS and PMA:challenging the concept of restricted phenotypes. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 141-147.	1.9	45
106	Natural course of scoliosis and lifetime risk of scoliosis surgery in spinal muscular atrophy. Neurology, 2019, 93, e149-e158.	1.1	45
107	Use of Multimodal Imaging and Clinical Biomarkers in Presymptomatic Carriers of C9orf72 Repeat Expansion. JAMA Neurology, 2020, 77, 1008.	9.0	45
108	Respiratory measures in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2018, 19, 321-330.	1.7	44

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109	Muscle strength and motor function in adolescents and adults with spinal muscular atrophy. Neurology, 2020, 95, e1988-e1998.	1.1	44
110	Association of maternal prenatal smoking GFI1-locus and cardio-metabolic phenotypes in 18,212 adults. EBioMedicine, 2018, 38, 206-216.	6.1	43
111	A Phase 2, Double-Blind, Randomized, Dose-Ranging Trial Of Reldesemtiv In Patients With ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2021, 22, 287-299.	1.7	42
112	Serotonin 2B receptor slows disease progression and prevents degeneration of spinal cord mononuclear phagocytes in amyotrophic lateral sclerosis. Acta Neuropathologica, 2016, 131, 465-480.	7.7	41
113	Assessment of the factorial validity and reliability of the ALSFRS-R: a revision of its measurement model. Journal of Neurology, 2017, 264, 1413-1420.	3.6	41
114	July 2017 ENCALS statement on edaravone. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2017, 18, 471-474.	1.7	41
115	Development and assessment of the inter-rater and intra-rater reproducibility of a self-administration version of the ALSFRS-R. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 75-81.	1.9	41
116	Pharmacokinetics of intravenous immunoglobulin in multifocal motor neuropathy. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 1145-1148.	1.9	40
117	MRI of the brachial plexus in polyneuropathy associated with monoclonal gammopathy. Muscle and Nerve, 2001, 24, 1312-1318.	2.2	39
118	Accelerometry for remote monitoring of physical activity in amyotrophic lateral sclerosis: a longitudinal cohort study. Journal of Neurology, 2019, 266, 2387-2395.	3.6	39
119	Raschâ€built Overall Disability Scale for Multifocal motor neuropathy (<scp>MMNâ€RODS</scp>^{Â©}). Journal of the Peripheral Nervous System, 2015, 20, 296-305.	3.1	38
120	Loss of appetite is associated with a loss of weight and fat mass in patients with amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2019, 20, 497-505.	1.7	38
121	Nerve ultrasound improves detection of treatment-responsive chronic inflammatory neuropathies. Neurology, 2020, 94, e1470-e1479.	1.1	38
122	Genome-wide study of DNA methylation shows alterations in metabolic, inflammatory, and cholesterol pathways in ALS. Science Translational Medicine, 2022, 14, eabj0264.	12.4	38
123	Bulbar muscle MRI changes in patients with SMA with reduced mouth opening and dysphagia. Neurology, 2014, 83, 1060-1066.	1.1	37
124	Factors related to caregiver strain in ALS: a longitudinal study. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, 775-781.	1.9	37
125	MRI shows thickening and altered diffusion in the median and ulnar nerves in multifocal motor neuropathy. European Radiology, 2017, 27, 2216-2224.	4.5	37
126	Whole blood transcriptome analysis in amyotrophic lateral sclerosis: A biomarker study. PLoS ONE, 2018, 13, e0198874.	2.5	37

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127	Human genetics and neuropathology suggest a link between miR-218 and amyotrophic lateral sclerosis pathophysiology. Science Translational Medicine, 2019, 11, .	12.4	37
128	Cognitive behavioural therapy and quality of life in psychologically distressed patients with amyotrophic lateral sclerosis and their caregivers: Results of a prematurely stopped randomized controlled trial. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2015, 16, 309-315.	1.7	36
129	Acute Effects of Riluzole and Retigabine on Axonal Excitability in Patients With Amyotrophic Lateral Sclerosis: A Randomized, Doubleâ€Blind, Placeboâ€Controlled, Crossover Trial. Clinical Pharmacology and Therapeutics, 2018, 104, 1136-1145.	4.7	36
130	Correlates of health related quality of life in adult patients with spinal muscular atrophy. Muscle and Nerve, 2016, 54, 850-855.	2.2	35
131	Associations between lifestyle and amyotrophic lateral sclerosis stratified by C9orf72 genotype: a longitudinal, population-based, case-control study. Lancet Neurology, The, 2021, 20, 373-384.	10.2	35
132	Patterns of symptom development in patients with motor neuron disease. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2018, 19, 21-28.	1.7	34
133	Taking a risk: a therapeutic focus on ataxin-2 in amyotrophic lateral sclerosis?. Trends in Molecular Medicine, 2014, 20, 25-35.	6.7	33
134	C9orf72 expansion differentially affects males with spinal onset amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2017, 88, 281.1-281.	1.9	33
135	â€œALS reversalsâ€• demographics, disease characteristics, treatments, and co-morbidities. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2018, 19, 495-499.	1.7	33
136	ATXN1 repeat expansions confer risk for amyotrophic lateral sclerosis and contribute to TDP-43 mislocalization. Brain Communications, 2020, 2, fcaa064.	3.3	33
137	A Comparative Study of SMN Protein and mRNA in Blood and Fibroblasts in Patients with Spinal Muscular Atrophy and Healthy Controls. PLoS ONE, 2016, 11, e0167087.	2.5	32
138	Comparative study of peripheral nerve Mri and ultrasound in multifocal motor neuropathy and amyotrophic lateral sclerosis. Muscle and Nerve, 2016, 54, 1133-1135.	2.2	32
139	Nerve ultrasound. Neurology, 2019, 92, .	1.1	32
140	Nerve ultrasound for diagnosing chronic inflammatory neuropathy. Neurology, 2020, 95, e1745-e1753.	1.1	32
141	Intragenic and structural variation in the SMN locus and clinical variability in spinal muscular atrophy. Brain Communications, 2020, 2, fcaa075.	3.3	32
142	A case series of PLS patients with frontotemporal dementia and overview of the literature. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2017, 18, 534-548.	1.7	31
143	Understanding the use of NIV in ALS: results of an international ALS specialist survey. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2018, 19, 331-341.	1.7	31
144	Protocol for a phase II, monocentre, double-blind, placebo-controlled, cross-over trial to assess efficacy of pyridostigmine in patients with spinal muscular atrophy types 2â€“4 (SPACE trial). BMJ Open, 2018, 8, e019932.	1.9	31

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145	Validated inference of smoking habits from blood with a finite DNA methylation marker set. European Journal of Epidemiology, 2019, 34, 1055-1074.	5.7	31
146	Impairment measures versus inflammatory <scp>RODS</scp> in <scp>GBS</scp> and <scp>CIDP</scp>: a responsiveness comparison. Journal of the Peripheral Nervous System, 2015, 20, 289-295.	3.1	30
147	Nerve sonography to detect peripheral nerve involvement in vasculitis syndromes. Neurology: Clinical Practice, 2016, 6, 293-303.	1.6	30
148	An old friend who has overstayed their welcome: the ALSFRS-R total score as primary endpoint for ALS clinical trials. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2021, 22, 300-307.	1.7	30
149	Telehealth as part of specialized ALS care: feasibility and user experiences with â€œALS home-monitoring and coachingâ€. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2020, 21, 183-192.	1.7	30
150	Comparing methods to combine functional loss and mortality in clinical trials for amyotrophic lateral sclerosis. Clinical Epidemiology, 2018, Volume 10, 333-341.	3.0	29
151	Nerve ultrasound can identify treatmentâ€responsive chronic neuropathies without electrodiagnostic features of demyelination. Muscle and Nerve, 2019, 60, 415-419.	2.2	29
152	Prospective natural history study of C9orf72 ALS clinical characteristics and biomarkers. Neurology, 2019, 93, e1605-e1617.	1.1	29
153	Progression of cognitive and behavioural impairment in early amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 779-780.	1.9	29
154	Blood Metal Levels and Amyotrophic Lateral Sclerosis Risk: A Prospective Cohort. Annals of Neurology, 2021, 89, 125-133.	5.3	29
155	Effects of aerobic exercise therapy and cognitive behavioural therapy on functioning and quality of life in amyotrophic lateral sclerosis: protocol of the FACTS-2-ALS trial. BMC Neurology, 2011, 11, 70.	1.8	28
156	Comparing the <scp>NIS</scp> vs. <scp>MRC</scp> and <scp>INCAT</scp> sensory scale through Rasch analyses. Journal of the Peripheral Nervous System, 2015, 20, 277-288.	3.1	27
157	Genome-wide identification of genes regulating DNA methylation using genetic anchors for causal inference. Genome Biology, 2020, 21, 220.	8.8	27
158	Inhibition of HERV-K (HML-2) in amyotrophic lateral sclerosis patients on antiretroviral therapy. Journal of the Neurological Sciences, 2021, 423, 117358.	0.6	27
159	No mutations in hnRNPA1 and hnRNPA2B1 in Dutch patients with amyotrophic lateral sclerosis, frontotemporal dementia, and inclusion body myopathy. Neurobiology of Aging, 2014, 35, 1956.e9-1956.e11.	3.1	26
160	Association between alcohol exposure and the risk of amyotrophic lateral sclerosis in the Euro-MOTOR study. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 11-19.	1.9	26
161	Association of NIPA1 repeat expansions with amyotrophic lateral sclerosis in a large international cohort. Neurobiology of Aging, 2019, 74, 234.e9-234.e15.	3.1	26
162	Support needs of caregivers of patients with amyotrophic lateral sclerosis: A qualitative study. Palliative and Supportive Care, 2019, 17, 195-201.	1.0	26

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163	Drug treatment for spinal muscular atrophy types II and III. The Cochrane Library, 2020, 1, CD006282.	2.8	26
164	Blended psychosocial support for partners of patients with ALS and PMA: results of a randomized controlled trial. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2020, 21, 344-354.	1.7	26
165	Resting-state EEG reveals four subphenotypes of amyotrophic lateral sclerosis. Brain, 2022, 145, 621-631.	7.6	26
166	Complement activity is associated with disease severity in multifocal motor neuropathy. Neurology: Neuroimmunology and NeuroInflammation, 2015, 2, e119.	6.0	25
167	Proteomic profiling of the spinal cord in ALS: decreased ATP5D levels suggest synaptic dysfunction in ALS pathogenesis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2017, 18, 210-220.	1.7	25
168	Myasthenia gravis with muscle specific kinase antibodies mimicking amyotrophic lateral sclerosis. Neuromuscular Disorders, 2016, 26, 350-353.	0.6	24
169	No evidence for shared genetic basis of common variants in multiple sclerosis and amyotrophic lateral sclerosis. Human Molecular Genetics, 2014, 23, 1916-1922.	2.9	23
170	Evaluation of genetic risk loci for intracranial aneurysms in sporadic arteriovenous malformations of the brain. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, 524-529.	1.9	23
171	Bulbar Problems Self-Reported by Children and Adults with Spinal Muscular Atrophy. Journal of Neuromuscular Diseases, 2019, 6, 361-368.	2.6	23
172	The Distinct Traits of the UNC13A Polymorphism in Amyotrophic Lateral Sclerosis. Annals of Neurology, 2020, 88, 796-806.	5.3	23
173	Assessment of motor unit loss in patients with spinal muscular atrophy. Clinical Neurophysiology, 2020, 131, 1280-1286.	1.5	23
174	Structural variation analysis of 6,500 whole genome sequences in amyotrophic lateral sclerosis. Npj Genomic Medicine, 2022, 7, 8.	3.8	23
175	Rare genetic variation in UNC13A may modify survival in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2016, 17, 593-599.	1.7	22
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177	Amyotrophic lateral sclerosis as a multi-step process: an Australia population study. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2019, 20, 532-537.	1.7	22
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