

Leonard H Van Den Berg

List of Publications by Year in descending order

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Version: 2024-02-01

304
papers

21,601
citations

15503

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. <i>Nature Genetics</i> , 2013, 45, 1238-1243.	21.4	1,544
2	Amyotrophic lateral sclerosis. <i>Nature Reviews Disease Primers</i> , 2017, 3, 17071.	30.5	885
3	Amyotrophic lateral sclerosis. <i>Lancet, The</i> , 2017, 390, 2084-2098.	13.7	867
4	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. <i>Science</i> , 2015, 347, 1436-1441.	12.6	823
5	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , 2018, 97, 1268-1283.e6.	8.1	517
6	Clinical diagnosis and management of amyotrophic lateral sclerosis. <i>Nature Reviews Neurology</i> , 2011, 7, 639-649.	10.1	503
7	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1043-1048.	21.4	494
8	Haploinsufficiency leads to neurodegeneration in C9ORF72 ALS/FTD human induced motor neurons. <i>Nature Medicine</i> , 2018, 24, 313-325.	30.7	445
9	Microglia innately develop within cerebral organoids. <i>Nature Communications</i> , 2018, 9, 4167.	12.8	405
10	Disease variants alter transcription factor levels and methylation of their binding sites. <i>Nature Genetics</i> , 2017, 49, 131-138.	21.4	390
11	Identification of context-dependent expression quantitative trait loci in whole blood. <i>Nature Genetics</i> , 2017, 49, 139-145.	21.4	363
12	Prognosis for patients with amyotrophic lateral sclerosis: development and validation of a personalised prediction model. <i>Lancet Neurology, The</i> , 2018, 17, 423-433.	10.2	342
13	Exome-wide Rare Variant Analysis Identifies TUBA4A Mutations Associated with Familial ALS. <i>Neuron</i> , 2014, 84, 324-331.	8.1	308
14	Analysis of amyotrophic lateral sclerosis as a multistep process: a population-based modelling study. <i>Lancet Neurology, The</i> , 2014, 13, 1108-1113.	10.2	302
15	Amyotrophic lateral sclerosis: moving towards a new classification system. <i>Lancet Neurology, The</i> , 2016, 15, 1182-1194.	10.2	301
16	Detection of long repeat expansions from PCR-free whole-genome sequence data. <i>Genome Research</i> , 2017, 27, 1895-1903.	5.5	277
17	Multidisciplinary ALS care improves quality of life in patients with ALS. <i>Neurology</i> , 2005, 65, 1264-1267.	1.1	273
18	Population based epidemiology of amyotrophic lateral sclerosis using capture-recapture methodology. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2011, 82, 1165-1170.	1.9	273

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19	A proposal for new diagnostic criteria for ALS. <i>Clinical Neurophysiology</i> , 2020, 131, 1975-1978.	1.5	268
20	A randomized sequential trial of creatine in amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 2003, 53, 437-445.	5.3	260
21	Gene discovery in amyotrophic lateral sclerosis: implications for clinical management. <i>Nature Reviews Neurology</i> , 2017, 13, 96-104.	10.1	245
22	Population genetic differentiation of height and body mass index across Europe. <i>Nature Genetics</i> , 2015, 47, 1357-1362.	21.4	227
23	<scp>C</scp>9orf72 ablation in mice does not cause motor neuron degeneration or motor deficits. <i>Annals of Neurology</i> , 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. <i>Nature Genetics</i> , 2021, 53, 1636-1648.	21.4	223
25	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. <i>Nature Genetics</i> , 2016, 48, 1037-1042.	21.4	218
26	Dexramipexole versus placebo for patients with amyotrophic lateral sclerosis (EMPOWER): a randomised, double-blind, phase 3 trial. <i>Lancet Neurology</i> , The, 2013, 12, 1059-1067.	10.2	216
27	Hypermetabolism in ALS is associated with greater functional decline and shorter survival. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 1016-1023.	1.9	177
28	TDP-43 proteinopathies: a new wave of neurodegenerative diseases. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2021, 92, 86-95.	1.9	174
29	Multifocal motor neuropathy: Diagnostic criteria that predict the response to immunoglobulin treatment. <i>Annals of Neurology</i> , 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. <i>Nature Genetics</i> , 2020, 52, 1303-1313.	21.4	163
31	A natural history study of late onset spinal muscular atrophy types 3b and 4. <i>Journal of Neurology</i> , 2008, 255, 1400-1404.	3.6	158
32	Blood lipids influence DNA methylation in circulating cells. <i>Genome Biology</i> , 2016, 17, 138.	8.8	154
33	The role of <i>TREM2</i> R47H as a risk factor for Alzheimer's disease, frontotemporal lobar degeneration, amyotrophic lateral sclerosis, and Parkinson's disease. <i>Alzheimer's and Dementia</i> , 2015, 11, 1407-1416.	0.8	152
34	Improving clinical trial outcomes in amyotrophic lateral sclerosis. <i>Nature Reviews Neurology</i> , 2021, 17, 104-118.	10.1	152
35	Deciphering amyotrophic lateral sclerosis: What phenotype, neuropathology and genetics are telling us about pathogenesis. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2013, 14, 5-18.	1.7	142
36	Diagnostic value of sonography in treatment-naive chronic inflammatory neuropathies. <i>Neurology</i> , 2017, 88, 143-151.	1.1	135

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37	Deep learning predictions of survival based on MRI in amyotrophic lateral sclerosis. <i>NeuroImage: Clinical</i> , 2017, 13, 361-369.	2.7	135
38	Muscle strength and motor function throughout life in a cross-sectional cohort of 180 patients with spinal muscular atrophy types 1-4. <i>European Journal of Neurology</i> , 2018, 25, 512-518.	3.3	126
39	Age-related accrual of methylomic variability is linked to fundamental ageing mechanisms. <i>Genome Biology</i> , 2016, 17, 191.	8.8	120
40	A blinded international study on the reliability of genetic testing for GGGGCC-repeat expansions in <i>C9orf72</i> reveals marked differences in results among 14 laboratories. <i>Journal of Medical Genetics</i> , 2014, 51, 419-424.	3.2	118
41	Primary lateral sclerosis: consensus diagnostic criteria. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2020, 91, 373-377.	1.9	118
42	Cell Specific eQTL Analysis without Sorting Cells. <i>PLoS Genetics</i> , 2015, 11, e1005223.	3.5	115
43	Genetic correlation between amyotrophic lateral sclerosis and schizophrenia. <i>Nature Communications</i> , 2017, 8, 14774.	12.8	114
44	Comparative interactomics analysis of different ALS-associated proteins identifies converging molecular pathways. <i>Acta Neuropathologica</i> , 2016, 132, 175-196.	7.7	113
45	Homozygous deletion of the survival motor neuron 2 gene is a prognostic factor in sporadic ALS. <i>Neurology</i> , 2001, 56, 749-752.	1.1	111
46	Randomized sequential trial of valproic acid in amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 2009, 66, 227-234.	5.3	111
47	Revised Airlie House consensus guidelines for design and implementation of ALS clinical trials. <i>Neurology</i> , 2019, 92, e1610-e1623.	1.1	105
48	Full ablation of <i>C9orf72</i> in mice causes immune system-related pathology and neoplastic events but no motor neuron defects. <i>Acta Neuropathologica</i> , 2016, 132, 145-147.	7.7	104
49	Negative selection in humans and fruit flies involves synergistic epistasis. <i>Science</i> , 2017, 356, 539-542.	12.6	103
50	Genomic signals of migration and continuity in Britain before the Anglo-Saxons. <i>Nature Communications</i> , 2016, 7, 10326.	12.8	100
51	Incidence of polyneuropathy in Utrecht, the Netherlands. <i>Neurology</i> , 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. <i>Lancet Neurology</i> , The, 2017, 16, 513-522.	10.2	95
53	<i>C9orf72</i> genotypes producing less SMN protein increase susceptibility to and severity of sporadic ALS. <i>Neurology</i> , 2005, 65, 820-825.	1.1	94
54	Cortical thickness in ALS: towards a marker for upper motor neuron involvement. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 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. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2017, 88, 365-367.	1.9	94
56	Cross-ethnic meta-analysis identifies association of the GPX3-TNIP1 locus with amyotrophic lateral sclerosis. <i>Nature Communications</i> , 2017, 8, 611.	12.8	93
57	The ALS-FTD-Q. <i>Neurology</i> , 2012, 79, 1377-1383.	1.1	91
58	The changing picture of amyotrophic lateral sclerosis: lessons from European registers. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2017, 88, 557-563.	1.9	89
59	The Combined Assessment of Function and Survival (CAFS): A new endpoint for ALS clinical trials. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2013, 14, 162-168.	1.7	88
60	Effect of Presymptomatic Body Mass Index and Consumption of Fat and Alcohol on Amyotrophic Lateral Sclerosis. <i>JAMA Neurology</i> , 2015, 72, 1155.	9.0	87
61	ATXN2 trinucleotide repeat length correlates with risk of ALS. <i>Neurobiology of Aging</i> , 2017, 51, 178.e1-178.e9.	3.1	86
62	Associations of autozygosity with a broad range of human phenotypes. <i>Nature Communications</i> , 2019, 10, 4957.	12.8	84
63	Joint sequencing of human and pathogen genomes reveals the genetics of pneumococcal meningitis. <i>Nature Communications</i> , 2019, 10, 2176.	12.8	83
64	Transethnic Genome-Wide Association Study Provides Insights in the Genetic Architecture and Heritability of Long QT Syndrome. <i>Circulation</i> , 2020, 142, 324-338.	1.6	83
65	Meta-analysis of pharmacogenetic interactions in amyotrophic lateral sclerosis clinical trials. <i>Neurology</i> , 2017, 89, 1915-1922.	1.1	82
66	Caregiver burden in amyotrophic lateral sclerosis: A systematic review. <i>Palliative Medicine</i> , 2018, 32, 231-245.	3.1	82
67	Subcortical structures in amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , 2015, 36, 1075-1082.	3.1	78
68	Shared vulnerability for connectome alterations across psychiatric and neurological brain disorders. <i>Nature Human Behaviour</i> , 2019, 3, 988-998.	12.0	75
69	Genetic variants associated with longitudinal changes in brain structure across the lifespan. <i>Nature Neuroscience</i> , 2022, 25, 421-432.	14.8	75
70	Lithium lacks effect on survival in amyotrophic lateral sclerosis: a phase IIb randomised sequential trial. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 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. <i>NeuroImage</i> , 2016, 124, 762-769.	4.2	74
72	Widespread structural brain involvement in ALS is not limited to the C9orf72 repeat expansion. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, 1354-1360.	1.9	69

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73	Cardiac pathology in spinal muscular atrophy: a systematic review. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 67.	2.7	67
74	Brain morphologic changes in asymptomatic <i>C9orf72</i> repeat expansion carriers. <i>Neurology</i> , 2015, 85, 1780-1788.	1.1	66
75	Refining eligibility criteria for amyotrophic lateral sclerosis clinical trials. <i>Neurology</i> , 2019, 92, .	1.1	66
76	Kuramoto model simulation of neural hubs and dynamic synchrony in the human cerebral connectome. <i>BMC Neuroscience</i> , 2015, 16, 54.	1.9	65
77	KIF1A variants are a frequent cause of autosomal dominant hereditary spastic paraplegia. <i>European Journal of Human Genetics</i> , 2020, 28, 40-49.	2.8	65
78	Large-scale SOD1 mutation screening provides evidence for genetic heterogeneity in amyotrophic lateral sclerosis. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2010, 81, 562-566.	1.9	64
79	Safety and tolerability of Triumeq in amyotrophic lateral sclerosis: the Lighthouse trial. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 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. <i>Lancet Neurology</i> , The, 2017, 16, 208-216.	10.2	62
81	Monitoring disease progression with plasma creatinine in amyotrophic lateral sclerosis clinical trials. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 156-161.	1.9	62
82	Edaravone: a new treatment for ALS on the horizon?. <i>Lancet Neurology</i> , 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. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2019, 20, 432-440.	1.7	60
84	Addressing heterogeneity in amyotrophic lateral sclerosis CLINICAL TRIALS. <i>Muscle and Nerve</i> , 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. <i>Health and Quality of Life Outcomes</i> , 2016, 14, 107.	2.4	58
86	Association of a Locus in the <i>CAMTA1</i> Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. <i>JAMA Neurology</i> , 2016, 73, 812.	9.0	57
87	Natural history of lung function in spinal muscular atrophy. <i>Orphanet Journal of Rare Diseases</i> , 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. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 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. <i>Nature Genetics</i> , 2022, 54, 232-239.	21.4	55
90	<i>SMN1</i> gene duplications are associated with sporadic ALS. <i>Neurology</i> , 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. <i>Environmental Health Perspectives</i> , 2017, 125, 097023.	6.0	54
92	Population-based analysis of survival in spinal muscular atrophy. <i>Neurology</i> , 2020, 94, e1634-e1644.	1.1	54
93	Autoantibody pathogenicity in a multifocal motor neuropathy induced pluripotent stem cellâ€“derived model. <i>Annals of Neurology</i> , 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. <i>European Journal of Neurology</i> , 2017, 24, 1307-1313.	3.3	51
95	Connectomeâ€“Based Propagation Model in Amyotrophic Lateral Sclerosis. <i>Annals of Neurology</i> , 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. <i>Clinical Neurophysiology</i> , 2015, 126, 1413-1420.	1.5	49
97	Meta-analysis of genome-wide DNA methylation identifies shared associations across neurodegenerative disorders. <i>Genome Biology</i> , 2021, 22, 90.	8.8	49
98	A case-control study of hormonal exposures as etiologic factors for ALS in women. <i>Neurology</i> , 2017, 89, 1283-1290.	1.1	48
99	Multifocal motor neuropathy: controversies and priorities. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2020, 91, 140-148.	1.9	48
100	The frontotemporal syndrome of ALS is associated with poor survival. <i>Journal of Neurology</i> , 2016, 263, 2476-2483.	3.6	46
101	Multimodal longitudinal study of structural brain involvement in amyotrophic lateral sclerosis. <i>Neurology</i> , 2020, 94, e2592-e2604.	1.1	46
102	Prognostic value of weight loss in patients with amyotrophic lateral sclerosis: a population-based study. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2020, 91, 867-875.	1.9	46
103	Association of Variants in the <i>SPTLC1</i> Gene With Juvenile Amyotrophic Lateral Sclerosis. <i>JAMA Neurology</i> , 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. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2018, 89, 797-803.	1.9	45
105	Cognitive and behavioural changes in PLS and PMA:challenging the concept of restricted phenotypes. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2019, 90, 141-147.	1.9	45
106	Natural course of scoliosis and lifetime risk of scoliosis surgery in spinal muscular atrophy. <i>Neurology</i> , 2019, 93, e149-e158.	1.1	45
107	Use of Multimodal Imaging and Clinical Biomarkers in Presymptomatic Carriers of <i>C9orf72</i> Repeat Expansion. <i>JAMA Neurology</i> , 2020, 77, 1008.	9.0	45
108	Respiratory measures in amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 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. <i>Neurology</i> , 2020, 95, e1988-e1998.	1.1	44
110	Association of maternal prenatal smoking GFI1-locus and cardio-metabolic phenotypes in 18,212 adults. <i>EBioMedicine</i> , 2018, 38, 206-216.	6.1	43
111	A Phase 2, Double-Blind, Randomized, Dose-Ranging Trial Of <i>Reldesemtiv</i> In Patients With ALS. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 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. <i>Acta Neuropathologica</i> , 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. <i>Journal of Neurology</i> , 2017, 264, 1413-1420.	3.6	41
114	July 2017 ENCALS statement on edaravone. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 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. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2020, 91, 75-81.	1.9	41
116	Pharmacokinetics of intravenous immunoglobulin in multifocal motor neuropathy. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2014, 85, 1145-1148.	1.9	40
117	MRI of the brachial plexus in polyneuropathy associated with monoclonal gammopathy. <i>Muscle and Nerve</i> , 2001, 24, 1312-1318.	2.2	39
118	Accelerometry for remote monitoring of physical activity in amyotrophic lateral sclerosis: a longitudinal cohort study. <i>Journal of Neurology</i> , 2019, 266, 2387-2395.	3.6	39
119	Rasch-built Overall Disability Scale for Multifocal motor neuropathy (^{MMN} ^{RODS}). <i>Journal of the Peripheral Nervous System</i> , 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. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2019, 20, 497-505.	1.7	38
121	Nerve ultrasound improves detection of treatment-responsive chronic inflammatory neuropathies. <i>Neurology</i> , 2020, 94, e1470-e1479.	1.1	38
122	Genome-wide study of DNA methylation shows alterations in metabolic, inflammatory, and cholesterol pathways in ALS. <i>Science Translational Medicine</i> , 2022, 14, eabj0264.	12.4	38
123	Bulbar muscle MRI changes in patients with SMA with reduced mouth opening and dysphagia. <i>Neurology</i> , 2014, 83, 1060-1066.	1.1	37
124	Factors related to caregiver strain in ALS: a longitudinal study. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, 775-781.	1.9	37
125	MRI shows thickening and altered diffusion in the median and ulnar nerves in multifocal motor neuropathy. <i>European Radiology</i> , 2017, 27, 2216-2224.	4.5	37
126	Whole blood transcriptome analysis in amyotrophic lateral sclerosis: A biomarker study. <i>PLoS ONE</i> , 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. <i>Science Translational Medicine</i> , 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. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 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, Double-blind, Placebo-controlled, Crossover Trial. <i>Clinical Pharmacology and Therapeutics</i> , 2018, 104, 1136-1145.	4.7	36
130	Correlates of health related quality of life in adult patients with spinal muscular atrophy. <i>Muscle and Nerve</i> , 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. <i>Lancet Neurology</i> , The, 2021, 20, 373-384.	10.2	35
132	Patterns of symptom development in patients with motor neuron disease. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2018, 19, 21-28.	1.7	34
133	Taking a risk: a therapeutic focus on ataxin-2 in amyotrophic lateral sclerosis?. <i>Trends in Molecular Medicine</i> , 2014, 20, 25-35.	6.7	33
134	C9orf72 expansion differentially affects males with spinal onset amyotrophic lateral sclerosis. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2017, 88, 281.1-281.	1.9	33
135	ALS reversals: demographics, disease characteristics, treatments, and co-morbidities. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2018, 19, 495-499.	1.7	33
136	ATXN1 repeat expansions confer risk for amyotrophic lateral sclerosis and contribute to TDP-43 mislocalization. <i>Brain Communications</i> , 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. <i>PLoS ONE</i> , 2016, 11, e0167087.	2.5	32
138	Comparative study of peripheral nerve Mri and ultrasound in multifocal motor neuropathy and amyotrophic lateral sclerosis. <i>Muscle and Nerve</i> , 2016, 54, 1133-1135.	2.2	32
139	Nerve ultrasound. <i>Neurology</i> , 2019, 92, .	1.1	32
140	Nerve ultrasound for diagnosing chronic inflammatory neuropathy. <i>Neurology</i> , 2020, 95, e1745-e1753.	1.1	32
141	Intragenic and structural variation in the SMN locus and clinical variability in spinal muscular atrophy. <i>Brain Communications</i> , 2020, 2, fcaa075.	3.3	32
142	A case series of PLS patients with frontotemporal dementia and overview of the literature. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2017, 18, 534-548.	1.7	31
143	Understanding the use of NIV in ALS: results of an international ALS specialist survey. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 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 2-4 (SPACE trial). <i>BMJ Open</i> , 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. <i>European Journal of Epidemiology</i> , 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. <i>Journal of the Peripheral Nervous System</i> , 2015, 20, 289-295.	3.1	30
147	Nerve sonography to detect peripheral nerve involvement in vasculitis syndromes. <i>Neurology: Clinical Practice</i> , 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. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2021, 22, 300-307.	1.7	30
149	Telehealth as part of specialized ALS care: feasibility and user experiences with ‘ALS home-monitoring and coaching’. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , 2020, 21, 183-192.	1.7	30
150	Comparing methods to combine functional loss and mortality in clinical trials for amyotrophic lateral sclerosis. <i>Clinical Epidemiology</i> , 2018, Volume 10, 333-341.	3.0	29
151	Nerve ultrasound can identify treatment-responsive chronic neuropathies without electrodiagnostic features of demyelination. <i>Muscle and Nerve</i> , 2019, 60, 415-419.	2.2	29
152	Prospective natural history study of <i>C9orf72</i> ALS clinical characteristics and biomarkers. <i>Neurology</i> , 2019, 93, e1605-e1617.	1.1	29
153	Progression of cognitive and behavioural impairment in early amyotrophic lateral sclerosis. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2020, 91, 779-780.	1.9	29
154	Blood Metal Levels and Amyotrophic Lateral Sclerosis Risk: A Prospective Cohort. <i>Annals of Neurology</i> , 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. <i>BMC Neurology</i> , 2011, 11, 70.	1.8	28
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