M A Van Es

List of Publications by Year
in descending order

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102 papers	8,117 citations	66343 42 h-index	54911 84 g-index
106 all docs	106 docs citations	106 times ranked	10153 citing authors

#	Article	IF	Citations
1	Mortality in polytrauma patients with moderate to severe TBI on par with isolated TBI patients: TBI as last frontier in polytrauma patients. Injury, 2022, 53, 1443-1448.	1.7	12
2	Structural variation analysis of 6,500 whole genome sequences in amyotrophic lateral sclerosis. Npj Genomic Medicine, 2022, 7, 8.	3.8	23
3	Cortical and subcortical changes in resting-state neuronal activity and connectivity in early symptomatic ALS and advanced frontotemporal dementia. NeuroImage: Clinical, 2022, 34, 102965.	2.7	3
4	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
5	Clinical trials in pediatric ALS: a TRICALS feasibility study. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2022, 23, 481-488.	1.7	3
6	Facial Onset Sensory and Motor Neuronopathy. Neurology: Clinical Practice, 2021, 11, 147-157.	1.6	16
7	TDP-43 proteinopathies: a new wave of neurodegenerative diseases. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 86-95.	1.9	174
8	The Effect of <scp>SMN</scp> Gene Dosage on <scp>ALS</scp> Risk and Disease Severity. Annals of Neurology, 2021, 89, 686-697.	5.3	10
9	Distinctive pattern of temporal atrophy in patients with frontotemporal dementia and the I383V variant in TARDBP. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 787-789.	1.9	5
10	Genotype-phenotype correlations of KIF5A stalk domain variants. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2021, 22, 561-570.	1.7	9
11	Incidence, Prevalence, and Geographical Clustering of Motor Neuron Disease in the Netherlands. Neurology, 2021, 96, .	1.1	19
12	Epidemiology of paediatric moderate and severe traumatic brain injury in the Netherlands. European Journal of Paediatric Neurology, 2021, 35, 123-129.	1.6	4
13	Discussing Personalized Prognosis Empowers Patients with Amyotrophic Lateral Sclerosis to Regain Control over Their Future: A Qualitative Study. Brain Sciences, 2021, 11, 1597.	2.3	4
14	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
15	Pharmacogenetic interactions in amyotrophic lateral sclerosis: a step closer to a cure?. Pharmacogenomics Journal, 2020, 20, 220-226.	2.0	14
16	KIF1A variants are a frequent cause of autosomal dominant hereditary spastic paraplegia. European Journal of Human Genetics, 2020, 28, 40-49.	2.8	65
17	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
18	5â€²ValCAC tRNA fragment generated as part of a protective angiogenin response provides prognostic value in amyotrophic lateral sclerosis. Brain Communications, 2020, 2, fcaa138.	3.3	16

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19	TRICALS: creating a highway toward a cure. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2020, 21, 496-501.	1.7	20
20	Is it accurate to classify ALS as a neuromuscular disorder?. Expert Review of Neurotherapeutics, 2020, 20, 895-906.	2.8	12
21	Discussing personalized prognosis in amyotrophic lateral sclerosis: development of a communication guide. BMC Neurology, 2020, 20, 446.	1.8	12
22	Multimodal longitudinal study of structural brain involvement in amyotrophic lateral sclerosis. Neurology, 2020, 94, e2592-e2604.	1.1	46
23	Use of Multimodal Imaging and Clinical Biomarkers in Presymptomatic Carriers of C9orf72 Repeat Expansion. JAMA Neurology, 2020, 77, 1008.	9.0	45
24	An overview of screening instruments for cognition and behavior in patients with ALS: selecting the appropriate tool for clinical practice. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2020, 21, 324-336.	1.7	35
25	A placebo-controlled trial to investigate the safety and efficacy of Penicillin G/Hydrocortisone in patients with ALS (PHALS trial). Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2020, 21, 584-592.	1.7	4
26	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
27	Neuro-imaging in amyotrophic lateral sclerosis: Should we shift towards the periphery?. Clinical Neurophysiology, 2020, 131, 2286-2288.	1.5	1
28	The Distinct Traits of the UNC13A Polymorphism in Amyotrophic Lateral Sclerosis. Annals of Neurology, 2020, 88, 796-806.	5.3	23
29	ATXN1 repeat expansions confer risk for amyotrophic lateral sclerosis and contribute to TDP-43 mislocalization. Brain Communications, 2020, 2, fcaa064.	3.3	33
30	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
31	A case of ALS with posterior cortical atrophy. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2019, 20, 506-510.	1.7	2
32	Implications of spirometric reference values for amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2019, 20, 473-480.	1.7	4
33	Cross-sectional and longitudinal assessment of the upper cervical spinal cord in motor neuron disease. NeuroImage: Clinical, 2019, 24, 101984.	2.7	18
34	Prospective natural history study of C9orf72 ALS clinical characteristics and biomarkers. Neurology, 2019, 93, e1605-e1617.	1.1	29
35	A neuropsychological and behavioral study of PLS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2019, 20, 376-384.	1.7	19
36	Diagnostic value of emergency medical services provider judgement in the identification of head injuries among trauma patients. European Journal of Neurology, 2019, 26, 274-280.	3.3	7

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37	Refining eligibility criteria for amyotrophic lateral sclerosis clinical trials. Neurology, 2019, 92, .	1.1	66
38	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
39	Derivation of norms for the Dutch version of the Edinburgh cognitive and behavioral ALS screen. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2019, 20, 19-27.	1.7	17
40	â€œALS reversalsâ€• demographics, disease characteristics, treatments, and co-morbidities. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2018, 19, 495-499.	1.7	33
41	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
42	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	8.1	517
43	Reconsidering the causality of TIA1 mutations in ALS. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2018, 19, 1-3.	1.7	22
44	Outcome in Patients with Isolated Moderate to Severe Traumatic Brain Injury. Critical Care Research and Practice, 2018, 2018, 1-7.	1.1	27
45	Whole blood transcriptome analysis in amyotrophic lateral sclerosis: A biomarker study. PLoS ONE, 2018, 13, e0198874.	2.5	37
46	Senataxin mutations elicit motor neuron degeneration phenotypes and yield TDP-43 mislocalization in ALS4 mice and human patients. Acta Neuropathologica, 2018, 136, 425-443.	7.7	43
47	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
48	Amyotrophic lateral sclerosis. Lancet, The, 2017, 390, 2084-2098.	13.7	867
49	Diagnostic value of sonography in treatment-naive chronic inflammatory neuropathies. Neurology, 2017, 88, 143-151.	1.1	135
50	Meta-analysis of pharmacogenetic interactions in amyotrophic lateral sclerosis clinical trials. Neurology, 2017, 89, 1915-1922.	1.1	82
51	Detection of long repeat expansions from PCR-free whole-genome sequence data. Genome Research, 2017, 27, 1895-1903.	5.5	277
52	Susceptible genes and disease mechanisms identified in frontotemporal dementia and frontotemporal dementia with Amyotrophic Lateral Sclerosis by DNA-methylation and GWAS. Scientific Reports, 2017, 7, 8899.	3.3	30
53	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
54	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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55	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	21.4	218
56	Association of a Locus in theCAMTA1Gene With Survival in Patients With Sporadic Amyotrophic Lateral Sclerosis. JAMA Neurology, 2016, 73, 812.	9.0	57
57	Large-scale screening in sporadic amyotrophic lateral sclerosis identifies genetic modifiers in C9orf72 repeat carriers. Neurobiology of Aging, 2016, 39, 220.e9-220.e15.	3.1	20
58	Brain morphologic changes in asymptomatic C9orf72 repeat expansion carriers. Neurology, 2015, 85, 1780-1788.	1.1	66
59	Depolarized Inactivation Overcomes Impaired Activation to Produce DRG Neuron Hyperexcitability in a Na_v1.7 Mutation in a Patient with Distal Limb Pain. Journal of Neuroscience, 2014, 34, 12328-12340.	3.6	18
60	Analysis of the KIFAP3 gene in amyotrophic lateral sclerosis: a multicenter survival study. Neurobiology of Aging, 2014, 35, 2420.e13-2420.e14.	3.1	16
61	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
62	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
63	<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
64	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
65	Serum angiogenin levels are elevated in ALS, but not Parkinson's disease: TableÂ1. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 1439-1440.	1.9	11
66	Screening for rare variants in the coding region of ALS-associated genes at 9p21.2 and 19p13.3. Neurobiology of Aging, 2013, 34, 1518.e5-1518.e7.	3.1	16
67	H63D polymorphism in HFE is not associated with amyotrophic lateral sclerosis. Neurobiology of Aging, 2013, 34, 1517.e5-1517.e7.	3.1	19
68	Mutational analysis of TARDBP in Parkinson's disease. Neurobiology of Aging, 2013, 34, 1517.e1-1517.e3.	3.1	3
69	Gene expression profile of SOD1-G93A mouse spinal cord, blood and muscle. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 190-198.	1.7	18
70	Amyotrophic lateral sclerosis is not linked to multiple sclerosis in a population based study. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 940-941.	1.9	9
71	Evidence for an oligogenic basis of amyotrophic lateral sclerosis. Human Molecular Genetics, 2012, 21, 3776-3784.	2.9	307
72	Rare and common paraoxonase gene variants in amyotrophic lateral sclerosis patients. Neurobiology of Aging, 2012, 33, 1845.e1-1845.e3.	3.1	11

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73	Polymorphisms in the GluR2 gene are not associated with amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 418-420.	3.1	8
74	Novel optineurin mutations in sporadic amyotrophic lateral sclerosis patients. Neurobiology of Aging, 2012, 33, 1016.e1-1016.e7.	3.1	46
75	UNC13A is a modifier of survival in amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 630.e3-630.e8.	3.1	107
76	VAPB and C9orf72 mutations in 1 familial amyotrophic lateral sclerosis patient. Neurobiology of Aging, 2012, 33, 2950.e1-2950.e4.	3.1	47
77	Mapping of Gene Expression Reveals CYP27A1 as a Susceptibility Gene for Sporadic ALS. PLoS ONE, 2012, 7, e35333.	2.5	50
78	Angiogenin, a piece of the complex puzzle of neurodegeneration. Annals of Neurology, 2012, 71, 727-728.	5.3	1
79	Genetic Overlap between Apparently Sporadic Motor Neuron Diseases. PLoS ONE, 2012, 7, e48983.	2.5	55
80	Angiogenin variants in Parkinson disease and amyotrophic lateral sclerosis. Annals of Neurology, 2011, 70, 964-973.	5.3	168
81	A coâ€segregating microduplication of chromosome 15q11.2 pinpoints two risk genes for autism spectrum disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2010, 153B, 960-966.	1.7	76
82	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
83	A sequence variant at 4p16.3 confers susceptibility to urinary bladder cancer. Nature Genetics, 2010, 42, 415-419.	21.4	169
84	A large genome scan for rare CNVs in amyotrophic lateral sclerosis. Human Molecular Genetics, 2010, 19, 4091-4099.	2.9	51
85	FUS Mutations in Familial Amyotrophic Lateral Sclerosis in the Netherlands. Archives of Neurology, 2010, 67, 224-30.	4.5	66
86	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
87	Tau levels do not influence human ALS or motor neuron degeneration in the SOD1 ^{G93A} mouse. Neurology, 2010, 74, 1687-1693.	1.1	18
88	Gene-Network Analysis Identifies Susceptibility Genes Related to Glycobiology in Autism. PLoS ONE, 2009, 4, e5324.	2.5	119
89	Genome-wide association study in premature ovarian failure patients suggests ADAMTS19 as a possible candidate gene. Human Reproduction, 2009, 24, 2372-2378.	0.9	90
90	A CASE OF ALS-FTD IN A LARGE FALS PEDIGREE WITH A K17I ANG MUTATION. Neurology, 2009, 72, 287-288.	1.1	68

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91	DPP6 IS ASSOCIATED WITH SUSCEPTIBILITY TO PROGRESSIVE SPINAL MUSCULAR ATROPHY. Neurology, 2009, 72, 1184-1185.	1.1	11
92	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
93	A large-scale international meta-analysis of paraoxonase gene polymorphisms in sporadic ALS. Neurology, 2009, 73, 16-24.	1.1	66
94	Weighted gene co-expression network analysis of the peripheral blood from Amyotrophic Lateral Sclerosis patients. BMC Genomics, 2009, 10, 405.	2.8	156
95	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
96	Alzheimerâ€™s disease beyond APOE. Nature Genetics, 2009, 41, 1047-1048.	21.4	39
97	Analysis ofFGGYas a risk factor for sporadic amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2009, 10, 441-447.	2.1	15
98	Genetic variation in DPP6 is associated with susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2008, 40, 29-31.	21.4	205
99	Copy-number variation in sporadic amyotrophic lateral sclerosis: a genome-wide screen. Lancet Neurology, The, 2008, 7, 319-326.	10.2	85
100	Progranulin genetic variability contributes to amyotrophic lateral sclerosis. Neurology, 2008, 71, 253-259.	1.1	148
101	Analysis of genome-wide copy number variation in Irish and Dutch ALS populations. Human Molecular Genetics, 2008, 17, 3392-3398.	2.9	41
102	ITPR2 as a susceptibility gene in sporadic amyotrophic lateral sclerosis: a genome-wide association study. Lancet Neurology, The, 2007, 6, 869-877.	10.2	195