Marka van Blitterswijk

List of Publications by Year
in descending order

77	7,596	66234	76769
papers	citations	42 h-index	74 g-index

81	81	81	8092
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Unconventional Translation of C9ORF72 GGGGCC Expansion Generates Insoluble Polypeptides Specific to c9FTD/ALS. Neuron, 2013, 77, 639-646.	3.8	962
2	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	3.8	517
3	Antisense transcripts of the expanded C9ORF72 hexanucleotide repeat form nuclear RNA foci and undergo repeat-associated non-ATG translation in c9FTD/ALS. Acta Neuropathologica, 2013, 126, 829-844.	3.9	506
4	Distinct brain transcriptome profiles in C9orf72-associated and sporadic ALS. Nature Neuroscience, 2015, 18, 1175-1182.	7.1	330
5	Evidence for an oligogenic basis of amyotrophic lateral sclerosis. Human Molecular Genetics, 2012, 21, 3776-3784.	1.4	307
6	Detection of long repeat expansions from PCR-free whole-genome sequence data. Genome Research, 2017, 27, 1895-1903.	2.4	277
7	Whole-genome sequencing reveals important role for TBK1 and OPTN mutations in frontotemporal lobar degeneration without motor neuron disease. Acta Neuropathologica, 2015, 130, 77-92.	3.9	267
8	Association between repeat sizes and clinical and pathological characteristics in carriers of C9ORF72 repeat expansions (Xpansize-72): a cross-sectional cohort study. Lancet Neurology, The, 2013, 12, 978-988.	4.9	232
9	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1037-1042.	9.4	218
10	Poly(GP) proteins are a useful pharmacodynamic marker for C9ORF72 -associated amyotrophic lateral sclerosis. Science Translational Medicine, 2017, 9, .	5.8	179
11	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. Nature Communications, 2016, 7, 11253.	5.8	174
12	How do C9ORF72 repeat expansions cause amyotrophic lateral sclerosis and frontotemporal dementia. Current Opinion in Neurology, 2012, 25, 689-700.	1.8	169
13	Angiogenin variants in Parkinson disease and amyotrophic lateral sclerosis. Annals of Neurology, 2011, 70, 964-973.	2.8	168
14	Expanded ATXN2 CAG repeat size in ALS identifies genetic overlap between ALS and SCA2. Neurology, 2011, 76, 2066-2072.	1.5	151
15	TMEM106B protects C9ORF72 expansion carriers against frontotemporal dementia. Acta Neuropathologica, 2014, 127, 397-406.	3.9	133
16	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.	1.5	118
17	Primary lateral sclerosis: consensus diagnostic criteria. Journal of Neurology, Neurosurgery and Psychiatry, 2020, 91, 373-377.	0.9	118
18	Truncated stathmin-2 is a marker of TDP-43 pathology in frontotemporal dementia. Journal of Clinical Investigation, 2020, 130, 6080-6092.	3.9	117

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19	Spt4 selectively regulates the expression of C9orf72 sense and antisense mutant transcripts. Science, 2016, 353, 708-712.	6.0	116
20	Long-read sequencing across the C9orf72 â€˜GGGGCCâ€™ repeat expansion: implications for clinical use and genetic discovery efforts in human disease. Molecular Neurodegeneration, 2018, 13, 46.	4.4	111
21	Novel clinical associations with specific C9ORF72 transcripts in patients with repeat expansions in C9ORF72. Acta Neuropathologica, 2015, 130, 863-876.	3.9	104
22	VCP mutations in familial and sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2012, 33, 837.e7-837.e13.	1.5	103
23	Repetitive element transcripts are elevated in the brain of C9orf72 ALS/FTLD patients. Human Molecular Genetics, 2017, 26, 3421-3431.	1.4	101
24	Hexanucleotide repeat expansions in C9ORF72 in the spectrum of motor neuron diseases. Neurology, 2012, 79, 878-882.	1.5	100
25	Potential genetic modifiers of disease risk and age at onset in patients with frontotemporal lobar degeneration and GRN mutations: a genome-wide association study. Lancet Neurology, The, 2018, 17, 548-558.	4.9	97
26	Genome-wide analyses as part of the international FTLD-TDP whole-genome sequencing consortium reveals novel disease risk factors and increases support for immune dysfunction in FTLD. Acta Neuropathologica, 2019, 137, 879-899.	3.9	90
27	Cerebellar c9RAN proteins associate with clinical and neuropathological characteristics of C9ORF72 repeat expansion carriers. Acta Neuropathologica, 2015, 130, 559-573.	3.9	89
28	C9ORF72 repeat expansions in cases with previously identified pathogenic mutations. Neurology, 2013, 81, 1332-1341.	1.5	84
29	In-depth clinico-pathological examination of RNA foci in a large cohort of C9ORF72 expansion carriers. Acta Neuropathologica, 2017, 134, 255-269.	3.9	76
30	Ataxin-2 as potential disease modifier in C9ORF72 expansion carriers. Neurobiology of Aging, 2014, 35, 2421.e13-2421.e17.	1.5	74
31	TYROBP genetic variants in early-onset Alzheimer's disease. Neurobiology of Aging, 2016, 48, 222.e9-222.e15.	1.5	69
32	Prosaposin is a regulator of progranulin levels and oligomerization. Nature Communications, 2016, 7, 11992.	5.8	68
33	Paraoxonase gene mutations in amyotrophic lateral sclerosis. Annals of Neurology, 2010, 68, 102-107.	2.8	67
34	TDP-43 plasma levels are higher in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2012, 13, 446-451.	2.3	66
35	Genetic modifiers in carriers of repeat expansions in the C9ORF72 gene. Molecular Neurodegeneration, 2014, 9, 38.	4.4	63
36	RNA processing pathways in amyotrophic lateral sclerosis. Neurogenetics, 2010, 11, 275-290.	0.7	61

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37	C9orf72 -derived arginine-containing dipeptide repeats associate with axonal transport machinery and impede microtubule-based motility. Science Advances, 2021, 7, .	4.7	57
38	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 448-460.e4.	3.8	56
39	Genetic Overlap between Apparently Sporadic Motor Neuron Diseases. PLoS ONE, 2012, 7, e48983.	1.1	55
40	Jump from Pre-mutation to Pathologic Expansion in C9orf72. American Journal of Human Genetics, 2015, 96, 962-970.	2.6	50
41	VAPB and C9orf72 mutations in 1 familial amyotrophic lateral sclerosis patient. Neurobiology of Aging, 2012, 33, 2950.e1-2950.e4.	1.5	47
42	Novel optineurin mutations in sporadic amyotrophic lateral sclerosis patients. Neurobiology of Aging, 2012, 33, 1016.e1-1016.e7.	1.5	46
43	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	4.5	46
44	Mutational analysis of TARDBP in neurodegenerative diseases. Neurobiology of Aging, 2011, 32, 2096-2099.	1.5	43
45	Genetic screening of a large series of North American sporadic and familial frontotemporal dementia cases. Alzheimer's and Dementia, 2020, 16, 118-130.	0.4	43
46	Neuroprotective Effect of Oligodendrocyte Precursor Cell Transplantation in a Long-Term Model of Periventricular Leukomalacia. American Journal of Pathology, 2009, 175, 2332-2342.	1.9	41
47	Identical twins with the C9orf72 repeat expansion are discordant for ALS. Neurology, 2014, 83, 1476-1478.	1.5	40
48	Extensive transcriptomic study emphasizes importance of vesicular transport in C9orf72 expansion carriers. Acta Neuropathologica Communications, 2019, 7, 150.	2.4	40
49	Elevated methylation levels, reduced expression levels, and frequent contractions in a clinical cohort of C9orf72 expansion carriers. Molecular Neurodegeneration, 2020, 15, 7.	4.4	34
50	Unaffected mosaic C9orf72 case. Neurology, 2018, 90, e323-e331.	1.5	33
51	Toward allele-specific targeting therapy and pharmacodynamic marker for spinocerebellar ataxia type 3. Science Translational Medicine, 2020, 12, .	5.8	32
52	Progressive amnestic dementia, hippocampal sclerosis, and mutation in C9ORF72. Acta Neuropathologica, 2013, 126, 545-554.	3.9	30
53	TMEM106B haplotypes have distinct gene expression patterns in aged brain. Molecular Neurodegeneration, 2018, 13, 35.	4.4	30
54	Novel causal genes and disease modifiers. Nature Reviews Neurology, 2013, 9, 63-64.	4.9	28

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55	ExpandedC9ORF72Hexanucleotide Repeat in Depressive Pseudodementia. JAMA Neurology, 2014, 71, 775.	4.5	28
56	Unravelling the clinical spectrum and the role of repeat length in C9ORF72 repeat expansions. Journal of Neurology, Neurosurgery and Psychiatry, 2021, 92, 502-509.	0.9	28
57	Anti-superoxide dismutase antibodies are associated with survival in patients with sporadic amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2011, 12, 430-438.	2.3	27
58	Profilin-1 mutations are rare in patients with amyotrophic lateral sclerosis and frontotemporal dementia. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2013, 14, 463-469.	1.1	25
59	Bromodomain inhibitors regulate the C9ORF72 locus in ALS. Experimental Neurology, 2015, 271, 241-250.	2.0	25
60	C9orf72 promoter hypermethylation is reduced while hydroxymethylation is acquired during reprogramming of ALS patient cells. Experimental Neurology, 2016, 277, 171-177.	2.0	21
61	Characterization of FUS Mutations in Amyotrophic Lateral Sclerosis Using RNA-Seq. PLoS ONE, 2013, 8, e60788.	1.1	21
62	Microglia in frontotemporal lobar degeneration with progranulin or C9ORF72 mutations. Annals of Clinical and Translational Neurology, 2019, 6, 1782-1796.	1.7	20
63	Excess of Rare Damaging TUBA4A Variants Suggests Cytoskeletal Defects in ALS. Neuron, 2014, 84, 241-243.	3.8	18
64	Long-read targeted sequencing uncovers clinicopathological associations for C9orf72-linked diseases. Brain, 2021, 144, 1082-1088.	3.7	17
65	C9orf72 repeats compromise nucleocytoplasmic transport. Nature Reviews Neurology, 2015, 11, 670-672.	4.9	12
66	Abnormal expression of homeobox genes and transthyretin in C9ORF72 expansion carriers. Neurology: Genetics, 2017, 3, e161.	0.9	12
67	Rare and common paraoxonase gene variants in amyotrophic lateral sclerosis patients. Neurobiology of Aging, 2012, 33, 1845.e1-1845.e3.	1.5	11
68	Revealing the Mutational Spectrum in Southern Africans With Amyotrophic Lateral Sclerosis. Neurology: Genetics, 2022, 8, e654.	0.9	10
69	UBQLN2 in familial amyotrophic lateral sclerosis in the Netherlands. Neurobiology of Aging, 2012, 33, 2233.e7-2233.e8.	1.5	9
70	Mutations in the TRPV4 Gene Are Not Associated With Sporadic Progressive Muscular Atrophy. Archives of Neurology, 2012, 69, 790-1.	4.9	6
71	Preface: promoting research in PLS: current knowledge and future challenges. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2020, 21, 1-2.	1.1	6
72	Shared brain transcriptomic signature in TDP-43 type A FTLD patients with or without GRN mutations. Brain, 2022, 145, 2472-2485.	3.7	6

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73	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. SSRN Electronic Journal, 0, , .	0.4	4
74	Mutational analysis of TARDBP in Parkinson's disease. Neurobiology of Aging, 2013, 34, 1517.e1-1517.e3.	1.5	3
75	Clinical variability and additional mutations in amyotrophic lateral sclerosis patients with p.<scp>N</scp>352<scp>S</scp> mutations in <scp>TARDBP</scp>. Neuropathology and Applied Neurobiology, 2014, 40, 356-358.	1.8	1
76	ALS-FTD Complex Disorder due to C9ORF72 Gene Mutation: Description of First Polish Family. European Neurology, 2014, 72, 64-71.	0.6	1
77	Repeat expansions in myoclonic epilepsy. Nature Genetics, 2018, 50, 477-478.	9.4	0