Marka van Blitterswijk

List of Publications by Citations

79 papers	5,486 citations	39 h-index	74 g-index
81 ext. papers	6,681 ext. citations	10.9 avg, IF	4.83 L-index

#	Paper	IF	Citations
79	Unconventional translation of C9ORF72 GGGGCC expansion generates insoluble polypeptides specific to c9FTD/ALS. *Neuron*, **2013**, 77, 639-46	13.9	783
78	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, 829-44	14.3	392
77	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. *Neuron*, **2018**, 97, 1268-1283.e6	13.9	296
76	Evidence for an oligogenic basis of amyotrophic lateral sclerosis. *Human Molecular Genetics*, **2012**, 21, 3776-84	5.6	251
75	Distinct brain transcriptome profiles in C9orf72-associated and sporadic ALS. *Nature Neuroscience*, **2015**, 18, 1175-82	25.5	235
74	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	14.3	222
73	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, 978-88	24.1	200
72	Detection of long repeat expansions from PCR-free whole-genome sequence data. *Genome Research*, **2017**, 27, 1895-1903	9.7	159
71	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. *Nature Genetics*, **2016**, 48, 1037-42	36.3	149
70	Angiogenin variants in Parkinson disease and amyotrophic lateral sclerosis. *Annals of Neurology*, **2011**, 70, 964-73	9.4	144
69	How do C9ORF72 repeat expansions cause amyotrophic lateral sclerosis and frontotemporal dementia: can we learn from other noncoding repeat expansion disorders?. *Current Opinion in Neurology*, **2012**, 25, 689-700	7.1	136
68	Poly(GP) proteins are a useful pharmacodynamic marker for -associated amyotrophic lateral sclerosis. *Science Translational Medicine*, **2017**, 9,	17.5	128
67	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. *Nature Communications*, **2016**, 7, 11253	17.4	126
66	Expanded ATXN2 CAG repeat size in ALS identifies genetic overlap between ALS and SCA2. *Neurology*, **2011**, 76, 2066-72	6.5	126
65	TMEM106B protects C9ORF72 expansion carriers against frontotemporal dementia. *Acta Neuropathologica*, **2014**, 127, 397-406	14.3	108
64	A blinded international study on the reliability of genetic testing for GGGGCC-repeat expansions in C9orf72 reveals marked differences in results among 14 laboratories. *Journal of Medical Genetics*, **2014**, 51, 419-24	5.8	96
63	Spt4 selectively regulates the expression of C9orf72 sense and antisense mutant transcripts. *Science*, **2016**, 353, 708-12	33.3	92

62	VCP mutations in familial and sporadic amyotrophic lateral sclerosis. *Neurobiology of Aging*, **2012**, 33, 837.e7-13	5.6	84
61	Novel clinical associations with specific C9ORF72 transcripts in patients with repeat expansions in C9ORF72. *Acta Neuropathologica*, **2015**, 130, 863-76	14.3	81
60	Hexanucleotide repeat expansions in C9ORF72 in the spectrum of motor neuron diseases. *Neurology*, **2012**, 79, 878-82	6.5	81
59	C9ORF72 repeat expansions in cases with previously identified pathogenic mutations. *Neurology*, **2013**, 81, 1332-41	6.5	75
58	Cerebellar c9RAN proteins associate with clinical and neuropathological characteristics of C9ORF72 repeat expansion carriers. *Acta Neuropathologica*, **2015**, 130, 559-73	14.3	72
57	Long-read sequencing across the C9orf72 'GGGGCC' repeat expansion: implications for clinical use and genetic discovery efforts in human disease. *Molecular Neurodegeneration*, **2018**, 13, 46	19	66
56	Repetitive element transcripts are elevated in the brain of C9orf72 ALS/FTLD patients. *Human Molecular Genetics*, **2017**, 26, 3421-3431	5.6	63
55	Ataxin-2 as potential disease modifier in C9ORF72 expansion carriers. *Neurobiology of Aging*, **2014**, 35, 2421.e13-7	5.6	62
54	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	24.1	60
53	Primary lateral sclerosis: consensus diagnostic criteria. *Journal of Neurology, Neurosurgery and Psychiatry*, **2020**, 91, 373-377	5.5	59
52	In-depth clinico-pathological examination of RNA foci in a large cohort of C9ORF72 expansion carriers. *Acta Neuropathologica*, **2017**, 134, 255-269	14.3	57
51	Paraoxonase gene mutations in amyotrophic lateral sclerosis. *Annals of Neurology*, **2010**, 68, 102-7	9.4	56
50	RNA processing pathways in amyotrophic lateral sclerosis. *Neurogenetics*, **2010**, 11, 275-90	3	54
49	TDP-43 plasma levels are higher in amyotrophic lateral sclerosis. *Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders*, **2012**, 13, 446-51		53
48	Genetic modifiers in carriers of repeat expansions in the C9ORF72 gene. *Molecular Neurodegeneration*, **2014**, 9, 38	19	51
47	TYROBP genetic variants in early-onset Alzheimer's disease. *Neurobiology of Aging*, **2016**, 48, 222.e9-222.e15	5.6	51
46	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	14.3	50
45	Prosaposin is a regulator of progranulin levels and oligomerization. *Nature Communications*, **2016**, 7, 11992	17.4	49

44	Genetic overlap between apparently sporadic motor neuron diseases. *PLoS ONE*, **2012**, 7, e48983	3.7	48
43	Jump from pre-mutation to pathologic expansion in C9orf72. *American Journal of Human Genetics*, **2015**, 96, 962-70	11	41
42	VAPB and C9orf72 mutations in 1 familial amyotrophic lateral sclerosis patient. *Neurobiology of Aging*, **2012**, 33, 2950.e1-4	5.6	40
41	Identical twins with the C9orf72 repeat expansion are discordant for ALS. *Neurology*, **2014**, 83, 1476-8	6.5	39
40	Novel optineurin mutations in sporadic amyotrophic lateral sclerosis patients. *Neurobiology of Aging*, **2012**, 33, 1016.e1-7	5.6	37
39	Mutational analysis of TARDBP in neurodegenerative diseases. *Neurobiology of Aging*, **2011**, 32, 2096-9	5.6	36
38	Truncated stathmin-2 is a marker of TDP-43 pathology in frontotemporal dementia. *Journal of Clinical Investigation*, **2020**, 130, 6080-6092	15.9	34
37	Neuroprotective effect of oligodendrocyte precursor cell transplantation in a long-term model of periventricular leukomalacia. *American Journal of Pathology*, **2009**, 175, 2332-42	5.8	28
36	Genetic screening of a large series of North American sporadic and familial frontotemporal dementia cases. *Alzheimerss and Dementia*, **2020**, 16, 118-130	1.2	25
35	Unaffected mosaic case: RNA foci, dipeptide proteins, but upregulated C9orf72 expression. *Neurology*, **2018**, 90, e323-e331	6.5	24
34	Expanded C9ORF72 hexanucleotide repeat in depressive pseudodementia. *JAMA Neurology*, **2014**, 71, 775-81	17.2	24
33	Profilin-1 mutations are rare in patients with amyotrophic lateral sclerosis and frontotemporal dementia. *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, **2013**, 14, 463-9	3.6	23
32	Motor neuron disease in 2012: Novel causal genes and disease modifiers. *Nature Reviews Neurology* , **2013**, 9, 63-4	15	22
31	Progressive amnestic dementia, hippocampal sclerosis, and mutation in C9ORF72. *Acta Neuropathologica*, **2013**, 126, 545-54	14.3	22
30	Elevated methylation levels, reduced expression levels, and frequent contractions in a clinical cohort of C9orf72 expansion carriers. *Molecular Neurodegeneration*, **2020**, 15, 7	19	20
29	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. *Neuron*, **2021**, 109, 448-460.e4	13.9	20
28	Bromodomain inhibitors regulate the C9ORF72 locus in ALS. *Experimental Neurology*, **2015**, 271, 241-50	5.7	19
27	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, 430-8		19

26	Extensive transcriptomic study emphasizes importance of vesicular transport in C9orf72 expansion carriers. *Acta Neuropathologica Communications*, **2019**, 7, 150	7.3	18
25	-derived arginine-containing dipeptide repeats associate with axonal transport machinery and impede microtubule-based motility. *Science Advances*, **2021**, 7,	14.3	17
24	C9orf72 promoter hypermethylation is reduced while hydroxymethylation is acquired during reprogramming of ALS patient cells. *Experimental Neurology*, **2016**, 277, 171-177	5.7	16
23	Characterization of FUS mutations in amyotrophic lateral sclerosis using RNA-Seq. *PLoS ONE*, **2013**, 8, e60788	3.7	16
22	TMEM106B haplotypes have distinct gene expression patterns in aged brain. *Molecular Neurodegeneration*, **2018**, 13, 35	19	15
21	Toward allele-specific targeting therapy and pharmacodynamic marker for spinocerebellar ataxia type 3. *Science Translational Medicine*, **2020**, 12,	17.5	15
20	Excess of rare damaging TUBA4A variants suggests cytoskeletal defects in ALS. *Neuron*, **2014**, 84, 241-3	13.9	13
19	Neurodegenerative disease: C9orf72 repeats compromise nucleocytoplasmic transport. *Nature Reviews Neurology*, **2015**, 11, 670-2	15	11
18	Microglia in frontotemporal lobar degeneration with progranulin or C9ORF72 mutations. *Annals of Clinical and Translational Neurology*, **2019**, 6, 1782-1796	5.3	11
17	Abnormal expression of homeobox genes and transthyretin in expansion carriers. *Neurology: Genetics*, **2017**, 3, e161	3.8	9
16	UBQLN2 in familial amyotrophic lateral sclerosis in The Netherlands. *Neurobiology of Aging*, **2012**, 33, 2233.e7-2233.e8	5.6	9
15	Rare and common paraoxonase gene variants in amyotrophic lateral sclerosis patients. *Neurobiology of Aging*, **2012**, 33, 1845.e1-3	5.6	7
14	C9orf72-derived arginine-containing dipeptide repeats associate with axonal transport machinery and impede microtubule-based motility		7
13	Detection of long repeat expansions from PCR-free whole-genome sequence data		6
12	Unravelling the clinical spectrum and the role of repeat length in repeat expansions. *Journal of Neurology, Neurosurgery and Psychiatry*, **2021**, 92, 502-509	5.5	6
11	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. *JAMA Neurology*, **2021**, 78, 1236-1248	17.2	5
10	Mutations in the TRPV4 gene are not associated with sporadic progressive muscular atrophy. *Archives of Neurology*, **2012**, 69, 790-1		4
9	Mutational analysis of TARDBP in Parkinson's disease. *Neurobiology of Aging*, **2013**, 34, 1517.e1-3	5.6	2

8	Revealing the Mutational Spectrum in Southern Africans With Amyotrophic Lateral Sclerosis.. *Neurology: Genetics*, **2022**, 8, e654	3.8	2
7	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. *SSRN Electronic Journal*,	1	2
6	Long-read targeted sequencing uncovers clinicopathological associations for C9orf72-linked diseases. *Brain*, **2021**, 144, 1082-1088	11.2	2
5	Clinical variability and additional mutations in amyotrophic lateral sclerosis patients with p.N352S mutations in TARDBP. *Neuropathology and Applied Neurobiology*, **2014**, 40, 356-8	5.2	1
4	ALS-FTD complex disorder due to C9ORF72 gene mutation: description of first Polish family. *European Neurology*, **2014**, 72, 64-71	2.1	1
3	Long-read sequencing across the C9orf72 GGGGCC repeat expansion: implications for clinical use and genetic discovery efforts in human disease		1
2	Single-cell profiling of the human primary motor cortex in ALS and FTLD		1
1	Repeat expansions in myoclonic epilepsy. *Nature Genetics*, **2018**, 50, 477-478	36.3	