

# Marka van Blitterswijk

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	Revealing the Mutational Spectrum in Southern Africans With Amyotrophic Lateral Sclerosis.. <i>Neurology: Genetics</i> , <b>2022</b> , 8, e654	3.8	2
78	-derived arginine-containing dipeptide repeats associate with axonal transport machinery and impede microtubule-based motility. <i>Science Advances</i> , <b>2021</b> , 7,	14.3	17
77	Long-read targeted sequencing uncovers clinicopathological associations for C9orf72-linked diseases. <i>Brain</i> , <b>2021</b> , 144, 1082-1088	11.2	2
76	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. <i>Neuron</i> , <b>2021</b> , 109, 448-460.e4	13.9	20
75	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. <i>JAMA Neurology</i> , <b>2021</b> , 78, 1236-1248	17.2	5
74	Unravelling the clinical spectrum and the role of repeat length in repeat expansions. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2021</b> , 92, 502-509	5.5	6
73	Genetic screening of a large series of North American sporadic and familial frontotemporal dementia cases. <i>Alzheimers and Dementia</i> , <b>2020</b> , 16, 118-130	1.2	25
72	Elevated methylation levels, reduced expression levels, and frequent contractions in a clinical cohort of C9orf72 expansion carriers. <i>Molecular Neurodegeneration</i> , <b>2020</b> , 15, 7	19	20
71	Primary lateral sclerosis: consensus diagnostic criteria. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , <b>2020</b> , 91, 373-377	5.5	59
70	Truncated stathmin-2 is a marker of TDP-43 pathology in frontotemporal dementia. <i>Journal of Clinical Investigation</i> , <b>2020</b> , 130, 6080-6092	15.9	34
69	Toward allele-specific targeting therapy and pharmacodynamic marker for spinocerebellar ataxia type 3. <i>Science Translational Medicine</i> , <b>2020</b> , 12,	17.5	15
68	Genome-wide analyses as part of the international FTLTDP whole-genome sequencing consortium reveals novel disease risk factors and increases support for immune dysfunction in FTLTDP. <i>Acta Neuropathologica</i> , <b>2019</b> , 137, 879-899	14.3	50
67	Extensive transcriptomic study emphasizes importance of vesicular transport in C9orf72 expansion carriers. <i>Acta Neuropathologica Communications</i> , <b>2019</b> , 7, 150	7.3	18
66	Microglia in frontotemporal lobar degeneration with progranulin or C9ORF72 mutations. <i>Annals of Clinical and Translational Neurology</i> , <b>2019</b> , 6, 1782-1796	5.3	11
65	Repeat expansions in myoclonic epilepsy. <i>Nature Genetics</i> , <b>2018</b> , 50, 477-478	36.3	
64	Unaffected mosaic case: RNA foci, dipeptide proteins, but upregulated C9orf72 expression. <i>Neurology</i> , <b>2018</b> , 90, e323-e331	6.5	24
63	Potential genetic modifiers of disease risk and age at onset in patients with frontotemporal lobar degeneration and GRN mutations: a genome-wide association study. <i>Lancet Neurology</i> , <b>2018</b> , 17, 548-558	24.1	60

62	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , <b>2018</b> , 97, 1268-1283.e6	13.9	296
61	TMEM106B haplotypes have distinct gene expression patterns in aged brain. <i>Molecular Neurodegeneration</i> , <b>2018</b> , 13, 35	19	15
60	Long-read sequencing across the C9orf72 'GGGGCC' repeat expansion: implications for clinical use and genetic discovery efforts in human disease. <i>Molecular Neurodegeneration</i> , <b>2018</b> , 13, 46	19	66
59	In-depth clinico-pathological examination of RNA foci in a large cohort of C9ORF72 expansion carriers. <i>Acta Neuropathologica</i> , <b>2017</b> , 134, 255-269	14.3	57
58	Poly(GP) proteins are a useful pharmacodynamic marker for -associated amyotrophic lateral sclerosis. <i>Science Translational Medicine</i> , <b>2017</b> , 9,	17.5	128
57	Detection of long repeat expansions from PCR-free whole-genome sequence data. <i>Genome Research</i> , <b>2017</b> , 27, 1895-1903	9.7	159
56	Repetitive element transcripts are elevated in the brain of C9orf72 ALS/FTLD patients. <i>Human Molecular Genetics</i> , <b>2017</b> , 26, 3421-3431	5.6	63
55	Abnormal expression of homeobox genes and transthyretin in expansion carriers. <i>Neurology: Genetics</i> , <b>2017</b> , 3, e161	3.8	9
54	Prosaposin is a regulator of progranulin levels and oligomerization. <i>Nature Communications</i> , <b>2016</b> , 7, 11992	17.4	49
53	NEK1 variants confer susceptibility to amyotrophic lateral sclerosis. <i>Nature Genetics</i> , <b>2016</b> , 48, 1037-42	36.3	149
52	CCNF mutations in amyotrophic lateral sclerosis and frontotemporal dementia. <i>Nature Communications</i> , <b>2016</b> , 7, 11253	17.4	126
51	C9orf72 promoter hypermethylation is reduced while hydroxymethylation is acquired during reprogramming of ALS patient cells. <i>Experimental Neurology</i> , <b>2016</b> , 277, 171-177	5.7	16
50	TYROBP genetic variants in early-onset Alzheimer's disease. <i>Neurobiology of Aging</i> , <b>2016</b> , 48, 222.e9-222.e15	36.5	51
49	Spt4 selectively regulates the expression of C9orf72 sense and antisense mutant transcripts. <i>Science</i> , <b>2016</b> , 353, 708-12	33.3	92
48	Bromodomain inhibitors regulate the C9ORF72 locus in ALS. <i>Experimental Neurology</i> , <b>2015</b> , 271, 241-50	5.7	19
47	Distinct brain transcriptome profiles in C9orf72-associated and sporadic ALS. <i>Nature Neuroscience</i> , <b>2015</b> , 18, 1175-82	25.5	235
46	Whole-genome sequencing reveals important role for TBK1 and OPTN mutations in frontotemporal lobar degeneration without motor neuron disease. <i>Acta Neuropathologica</i> , <b>2015</b> , 130, 77-92	14.3	222
45	Neurodegenerative disease: C9orf72 repeats compromise nucleocytoplasmic transport. <i>Nature Reviews Neurology</i> , <b>2015</b> , 11, 670-2	15	11

44	Novel clinical associations with specific C9ORF72 transcripts in patients with repeat expansions in C9ORF72. <i>Acta Neuropathologica</i> , <b>2015</b> , 130, 863-76	14.3	81
43	Cerebellar c9RAN proteins associate with clinical and neuropathological characteristics of C9ORF72 repeat expansion carriers. <i>Acta Neuropathologica</i> , <b>2015</b> , 130, 559-73	14.3	72
42	Jump from pre-mutation to pathologic expansion in C9orf72. <i>American Journal of Human Genetics</i> , <b>2015</b> , 96, 962-70	11	41
41	TMEM106B protects C9ORF72 expansion carriers against frontotemporal dementia. <i>Acta Neuropathologica</i> , <b>2014</b> , 127, 397-406	14.3	108
40	A blinded international study on the reliability of genetic testing for GGGGCC-repeat expansions in C9orf72 reveals marked differences in results among 14 laboratories. <i>Journal of Medical Genetics</i> , <b>2014</b> , 51, 419-24	5.8	96
39	Ataxin-2 as potential disease modifier in C9ORF72 expansion carriers. <i>Neurobiology of Aging</i> , <b>2014</b> , 35, 2421.e13-7	5.6	62
38	Excess of rare damaging TUBA4A variants suggests cytoskeletal defects in ALS. <i>Neuron</i> , <b>2014</b> , 84, 241-3	13.9	13
37	Identical twins with the C9orf72 repeat expansion are discordant for ALS. <i>Neurology</i> , <b>2014</b> , 83, 1476-8	6.5	39
36	Clinical variability and additional mutations in amyotrophic lateral sclerosis patients with p.N352S mutations in TARDBP. <i>Neuropathology and Applied Neurobiology</i> , <b>2014</b> , 40, 356-8	5.2	1
35	Genetic modifiers in carriers of repeat expansions in the C9ORF72 gene. <i>Molecular Neurodegeneration</i> , <b>2014</b> , 9, 38	19	51
34	Expanded C9ORF72 hexanucleotide repeat in depressive pseudodementia. <i>JAMA Neurology</i> , <b>2014</b> , 71, 775-81	17.2	24
33	ALS-FTD complex disorder due to C9ORF72 gene mutation: description of first Polish family. <i>European Neurology</i> , <b>2014</b> , 72, 64-71	2.1	1
32	Association between repeat sizes and clinical and pathological characteristics in carriers of C9ORF72 repeat expansions (Xpansize-72): a cross-sectional cohort study. <i>Lancet Neurology</i> , <b>2013</b> , 12, 978-88	24.1	200
31	Motor neuron disease in 2012: Novel causal genes and disease modifiers. <i>Nature Reviews Neurology</i> , <b>2013</b> , 9, 63-4	15	22
30	Unconventional translation of C9ORF72 GGGGCC expansion generates insoluble polypeptides specific to c9FTD/ALS. <i>Neuron</i> , <b>2013</b> , 77, 639-46	13.9	783
29	Mutational analysis of TARDBP in Parkinson's disease. <i>Neurobiology of Aging</i> , <b>2013</b> , 34, 1517.e1-3	5.6	2
28	Antisense transcripts of the expanded C9ORF72 hexanucleotide repeat form nuclear RNA foci and undergo repeat-associated non-ATG translation in c9FTD/ALS. <i>Acta Neuropathologica</i> , <b>2013</b> , 126, 829-44	14.3	392
27	Progressive amnesic dementia, hippocampal sclerosis, and mutation in C9ORF72. <i>Acta Neuropathologica</i> , <b>2013</b> , 126, 545-54	14.3	22

26	C9ORF72 repeat expansions in cases with previously identified pathogenic mutations. <i>Neurology</i> , <b>2013</b> , 81, 1332-41	6.5	75
25	Profilin-1 mutations are rare in patients with amyotrophic lateral sclerosis and frontotemporal dementia. <i>Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration</i> , <b>2013</b> , 14, 463-9	3.6	23
24	Characterization of FUS mutations in amyotrophic lateral sclerosis using RNA-Seq. <i>PLoS ONE</i> , <b>2013</b> , 8, e60788	3.7	16
23	Mutations in the TRPV4 gene are not associated with sporadic progressive muscular atrophy. <i>Archives of Neurology</i> , <b>2012</b> , 69, 790-1		4
22	Rare and common paraoxonase gene variants in amyotrophic lateral sclerosis patients. <i>Neurobiology of Aging</i> , <b>2012</b> , 33, 1845.e1-3	5.6	7
21	Novel optineurin mutations in sporadic amyotrophic lateral sclerosis patients. <i>Neurobiology of Aging</i> , <b>2012</b> , 33, 1016.e1-7	5.6	37
20	VCP mutations in familial and sporadic amyotrophic lateral sclerosis. <i>Neurobiology of Aging</i> , <b>2012</b> , 33, 837.e7-13	5.6	84
19	UBQLN2 in familial amyotrophic lateral sclerosis in The Netherlands. <i>Neurobiology of Aging</i> , <b>2012</b> , 33, 2233.e7-2233.e8	5.6	9
18	VAPB and C9orf72 mutations in 1 familial amyotrophic lateral sclerosis patient. <i>Neurobiology of Aging</i> , <b>2012</b> , 33, 2950.e1-4	5.6	40
17	TDP-43 plasma levels are higher in amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , <b>2012</b> , 13, 446-51		53
16	How do C9ORF72 repeat expansions cause amyotrophic lateral sclerosis and frontotemporal dementia: can we learn from other noncoding repeat expansion disorders?. <i>Current Opinion in Neurology</i> , <b>2012</b> , 25, 689-700	7.1	136
15	Evidence for an oligogenic basis of amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , <b>2012</b> , 21, 3776-84	5.6	251
14	Hexanucleotide repeat expansions in C9ORF72 in the spectrum of motor neuron diseases. <i>Neurology</i> , <b>2012</b> , 79, 878-82	6.5	81
13	Genetic overlap between apparently sporadic motor neuron diseases. <i>PLoS ONE</i> , <b>2012</b> , 7, e48983	3.7	48
12	Mutational analysis of TARDBP in neurodegenerative diseases. <i>Neurobiology of Aging</i> , <b>2011</b> , 32, 2096-9	5.6	36
11	Expanded ATXN2 CAG repeat size in ALS identifies genetic overlap between ALS and SCA2. <i>Neurology</i> , <b>2011</b> , 76, 2066-72	6.5	126
10	Angiogenin variants in Parkinson disease and amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , <b>2011</b> , 70, 964-73	9.4	144
9	Anti-superoxide dismutase antibodies are associated with survival in patients with sporadic amyotrophic lateral sclerosis. <i>Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders</i> , <b>2011</b> , 12, 430-8		19

8	RNA processing pathways in amyotrophic lateral sclerosis. <i>Neurogenetics</i> , <b>2010</b> , 11, 275-90	3	54
7	Paraoxonase gene mutations in amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , <b>2010</b> , 68, 102-7	9.4	56
6	Neuroprotective effect of oligodendrocyte precursor cell transplantation in a long-term model of periventricular leukomalacia. <i>American Journal of Pathology</i> , <b>2009</b> , 175, 2332-42	5.8	28
5	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. <i>SSRN Electronic Journal</i> ,	1	2
4	Long-read sequencing across the C9orf72 CAGGGCC repeat expansion: implications for clinical use and genetic discovery efforts in human disease		1
3	Detection of long repeat expansions from PCR-free whole-genome sequence data		6
2	C9orf72-derived arginine-containing dipeptide repeats associate with axonal transport machinery and impede microtubule-based motility		7
1	Single-cell profiling of the human primary motor cortex in ALS and FTLD		1