Vivianna M Van Deerlin

List of Publications by Year
in descending order

119 papers	17,676 citations	28190 55 h-index	20900 115 g-index
127 all docs	127 docs citations	127 times ranked	21677 citing authors

#	Article	IF	Citations
1	Genetic meta-analysis of diagnosed Alzheimerâ€™s disease identifies new risk loci and implicates AÎ², tau, immunity and lipid processing. Nature Genetics, 2019, 51, 414-430.	9.4	1,962
2	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	6.0	1,085
3	Frequency of the C9orf72 hexanucleotide repeat expansion in patients with amyotrophic lateral sclerosis and frontotemporal dementia: a cross-sectional study. Lancet Neurology, The, 2012, 11, 323-330.	4.9	1,039
4	Exome sequencing in amyotrophic lateral sclerosis identifies risk genes and pathways. Science, 2015, 347, 1436-1441.	6.0	823
5	Stages of pTDPâ€43 pathology in amyotrophic lateral sclerosis. Annals of Neurology, 2013, 74, 20-38.	2.8	820
6	Rare coding variants in PLCG2, ABI3, and TREM2 implicate microglial-mediated innate immunity in Alzheimer's disease. Nature Genetics, 2017, 49, 1373-1384.	9.4	783
7	TARDBP mutations in amyotrophic lateral sclerosis with TDP-43 neuropathology: a genetic and histopathological analysis. Lancet Neurology, The, 2008, 7, 409-416.	4.9	636
8	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. Neuron, 2018, 97, 1268-1283.e6.	3.8	517
9	Genome-wide association analyses identify new risk variants and the genetic architecture of amyotrophic lateral sclerosis. Nature Genetics, 2016, 48, 1043-1048.	9.4	494
10	Common variants at 7p21 are associated with frontotemporal lobar degeneration with TDP-43 inclusions. Nature Genetics, 2010, 42, 234-239.	9.4	479
11	Neurodegenerative disease concomitant proteinopathies are prevalent, age-related and APOE4-associated. Brain, 2018, 141, 2181-2193.	3.7	448
12	Neuropathological and genetic correlates of survival and dementia onset in synucleinopathies: a retrospective analysis. Lancet Neurology, The, 2017, 16, 55-65.	4.9	394
13	Trial of Transplantation of HCV-Infected Kidneys into Uninfected Recipients. New England Journal of Medicine, 2017, 376, 2394-2395.	13.9	315
14	Frontotemporal dementia and its subtypes: a genome-wide association study. Lancet Neurology, The, 2014, 13, 686-699.	4.9	302
15	Sequential distribution of pTDP-43 pathology in behavioral variant frontotemporal dementia (bvFTD). Acta Neuropathologica, 2014, 127, 423-439.	3.9	237
16	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.	9.4	223
17	Frontotemporal lobar degeneration: defining phenotypic diversity through personalized medicine. Acta Neuropathologica, 2015, 129, 469-491.	3.9	218
18	Genome-wide association study identifies four novel loci associated with Alzheimerâ€™s endophenotypes and disease modifiers. Acta Neuropathologica, 2017, 133, 839-856.	3.9	199

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19	Evidence for a role of the rare p.A152T variant in MAPT in increasing the risk for FTD-spectrum and Alzheimer's diseases. Human Molecular Genetics, 2012, 21, 3500-3512.	1.4	198
20	Genome sequencing analysis identifies new loci associated with Lewy body dementia and provides insights into its genetic architecture. Nature Genetics, 2021, 53, 294-303.	9.4	198
21	Investigating the genetic architecture of dementia with Lewy bodies: a two-stage genome-wide association study. Lancet Neurology, The, 2018, 17, 64-74.	4.9	195
22	Twelve-Month Outcomes After Transplant of Hepatitis Câ€“Infected Kidneys Into Uninfected Recipients. Annals of Internal Medicine, 2018, 169, 273-281.	2.0	193
23	CSF biomarkers associated with disease heterogeneity in early Parkinsonâ€™s disease: the Parkinsonâ€™s Progression Markers Initiative study. Acta Neuropathologica, 2016, 131, 935-949.	3.9	190
24	Association ofGBAMutations and the E326K Polymorphism With Motor and Cognitive Progression in Parkinson Disease. JAMA Neurology, 2016, 73, 1217.	4.5	185
25	Postmortem Cortex Samples Identify Distinct Molecular Subtypes of ALS: Retrotransposon Activation, Oxidative Stress, and Activated Glia. Cell Reports, 2019, 29, 1164-1177.e5.	2.9	184
26	Diagnosis of Parkinson's disease on the basis of clinical and genetic classification: a population-based modelling study. Lancet Neurology, The, 2015, 14, 1002-1009.	4.9	179
27	Age at symptom onset and death and disease duration in genetic frontotemporal dementia: an international retrospective cohort study. Lancet Neurology, The, 2020, 19, 145-156.	4.9	175
28	APOE,MAPT, andSNCAGenes and Cognitive Performance in Parkinson Disease. JAMA Neurology, 2014, 71, 1405.	4.5	172
29	Genome-wide association study of corticobasal degeneration identifies risk variants shared with progressive supranuclear palsy. Nature Communications, 2015, 6, 7247.	5.8	170
30	A platform for discovery: The University of Pennsylvania Integrated Neurodegenerative Disease Biobank. Alzheimer's and Dementia, 2014, 10, 477.	0.4	167
31	Effects of Multiple Genetic Loci on Age at Onset in Late-Onset Alzheimer Disease. JAMA Neurology, 2014, 71, 1394.	4.5	166
32	Expansion of the classification of FTLD-TDP: distinct pathology associated with rapidly progressive frontotemporal degeneration. Acta Neuropathologica, 2017, 134, 65-78.	3.9	163
33	Loss of brain tau defines novel sporadic and familial tauopathies with frontotemporal dementia. Annals of Neurology, 2001, 49, 165-175.	2.8	159
34	GBA Variants are associated with a distinct pattern of cognitive deficits in <scp>P</scp>arkinson's disease. Movement Disorders, 2016, 31, 95-102.	2.2	158
35	The role of TREM2 R47H as a risk factor for Alzheimer's disease, frontotemporal lobar degeneration, amyotrophic lateral sclerosis, and Parkinson's disease. Alzheimer's and Dementia, 2015, 11, 1407-1416.	0.4	152
36	Deep clinical and neuropathological phenotyping of <scp>P</scp>ick disease. Annals of Neurology, 2016, 79, 272-287.	2.8	146

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37	Cognitive decline and reduced survival inC9orf72expansion frontotemporal degeneration and amyotrophic lateral sclerosis. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 163-169.	0.9	141
38	Graft-Versus-Tumor Induction With Donor Leukocyte Infusions as Primary Therapy for Patients With Malignancies. Journal of Clinical Oncology, 1999, 17, 1234-1234.	0.8	124
39	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. Annals of Neurology, 2019, 85, 470-481.	2.8	118
40	APOE and TREM2 regulate amyloid-responsive microglia in Alzheimerâ€™s disease. Acta Neuropathologica, 2020, 140, 477-493.	3.9	117
41	Clinical marker for Alzheimer disease pathology in logopenic primary progressive aphasia. Neurology, 2017, 88, 2276-2284.	1.5	114
42	Identification of evolutionarily conserved gene networks mediating neurodegenerative dementia. Nature Medicine, 2019, 25, 152-164.	15.2	111
43	Development and Validation of Pedigree Classification Criteria for Frontotemporal Lobar Degeneration. JAMA Neurology, 2013, 70, 1411.	4.5	107
44	Risk genotypes at TMEM106B are associated with cognitive impairment in amyotrophic lateral sclerosis. Acta Neuropathologica, 2011, 121, 373-380.	3.9	102
45	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
46	<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.	2.8	91
47	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
48	Transplanting hepatitis C virusâ€“infected hearts into uninfected recipients: A single-arm trial. American Journal of Transplantation, 2019, 19, 2533-2542.	2.6	88
49	Sex-specific genetic predictors of Alzheimerâ€™s disease biomarkers. Acta Neuropathologica, 2018, 136, 857-872.	3.9	87
50	Autosomal dominant VCP hypomorph mutation impairs disaggregation of PHF-tau. Science, 2020, 370, .	6.0	85
51	C9ORF72 repeat expansions in cases with previously identified pathogenic mutations. Neurology, 2013, 81, 1332-1341.	1.5	84
52	Selective Genetic Overlap Between Amyotrophic Lateral Sclerosis and Diseases of the Frontotemporal Dementia Spectrum. JAMA Neurology, 2018, 75, 860.	4.5	79
53	The development and convergence of co-pathologies in Alzheimerâ€™s disease. Brain, 2021, 144, 953-962.	3.7	76
54	Semi-automated quantification of C9orf72 expansion size reveals inverse correlation between hexanucleotide repeat number and disease duration in frontotemporal degeneration. Acta Neuropathologica, 2015, 130, 363-372.	3.9	65

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55	TDP-43 Promotes Neurodegeneration by Impairing Chromatin Remodeling. Current Biology, 2017, 27, 3579-3590.e6.	1.8	63
56	Genome-wide, high-content siRNA screening identifies the Alzheimerâ€™s genetic risk factor FERMT2 as a major modulator of APP metabolism. Acta Neuropathologica, 2017, 133, 955-966.	3.9	60
57	Asymmetry of post-mortem neuropathology in behavioural-variant frontotemporal dementia. Brain, 2018, 141, 288-301.	3.7	56
58	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. Neuron, 2021, 109, 448-460.e4.	3.8	56
59	A 2-Step Cerebrospinal Algorithm for the Selection of Frontotemporal Lobar Degeneration Subtypes. JAMA Neurology, 2018, 75, 738.	4.5	54
60	Differences in the Presentation and Progression of Parkinson's Disease by Sex. Movement Disorders, 2021, 36, 106-117.	2.2	54
61	Clinical, Genetic, and Pathologic Characteristics of Patients With Frontotemporal Dementia and Progranulin Mutations. Archives of Neurology, 2007, 64, 1148.	4.9	52
62	Early Donor Chimerism Levels Predict Relapse and Survival after Allogeneic Stem Cell Transplantation with Reduced-Intensity Conditioning. Biology of Blood and Marrow Transplantation, 2014, 20, 1758-1766.	2.0	52
63	TMEM106B Effect on cognition in Parkinson disease and frontotemporal dementia. Annals of Neurology, 2019, 85, 801-811.	2.8	52
64	ALS-Plus syndrome: Non-pyramidal features in a large ALS cohort. Journal of the Neurological Sciences, 2014, 345, 118-124.	0.3	51
65	C9orf72 intermediate repeats are associated with corticobasal degeneration, increased C9orf72 expression and disruption of autophagy. Acta Neuropathologica, 2019, 138, 795-811.	3.9	50
66	PARK10 is a major locus for sporadic neuropathologically confirmed Parkinson disease. Neurology, 2015, 84, 972-980.	1.5	48
67	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. JAMA Neurology, 2021, 78, 1236.	4.5	46
68	Genetic and neuroanatomic associations in sporadic frontotemporal lobar degeneration. Neurobiology of Aging, 2014, 35, 1473-1482.	1.5	43
69	Alzheimerâ€™s Disease and Frontotemporal Dementia: The Current State of Genetics and Genetic Testing Since the Advent of Next-Generation Sequencing. Molecular Diagnosis and Therapy, 2018, 22, 505-513.	1.6	41
70	Cognitive reserve in frontotemporal degeneration. Neurology, 2016, 87, 1813-1819.	1.5	40
71	Aberrant activation of non-coding RNA targets of transcriptional elongation complexes contributes to TDP-43 toxicity. Nature Communications, 2018, 9, 4406.	5.8	40
72	Transcriptomic Changes Due to Cytoplasmic TDP-43 Expression Reveal Dysregulation of Histone Transcripts and Nuclear Chromatin. PLoS ONE, 2015, 10, e0141836.	1.1	40

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73	A C6orf10/LOC101929163 locus is associated with age of onset in C9orf72 carriers. Brain, 2018, 141, 2895-2907.	3.7	39
74	Lower plasma apolipoprotein A1 levels are found in Parkinson's disease and associate with apolipoprotein A1 genotype. Movement Disorders, 2015, 30, 805-812.	2.2	37
75	Large-scale exploratory genetic analysis of cognitive impairment in Parkinson's disease. Neurobiology of Aging, 2017, 56, 211.e1-211.e7.	1.5	37
76	APOE, thought disorder, and SPAREâ€AD predict cognitive decline in established Parkinson's disease. Movement Disorders, 2018, 33, 289-297.	2.2	35
77	Primary Tau Pathology, Not Copathology, Correlates With Clinical Symptoms in PSP and CBD. Journal of Neuropathology and Experimental Neurology, 2020, 79, 296-304.	0.9	35
78	Neuron loss and degeneration in the progression of TDP-43 in frontotemporal lobar degeneration. Acta Neuropathologica Communications, 2017, 5, 68.	2.4	34
79	Regional brain amyloid-Î² accumulation associates with domain-specific cognitive performance in Parkinson disease without dementia. PLoS ONE, 2017, 12, e0177924.	1.1	33
80	UNC13A polymorphism contributes to frontotemporal disease in sporadic amyotrophic lateral sclerosis. Neurobiology of Aging, 2019, 73, 190-199.	1.5	31
81	Genomewide Association Studies of <scp>LRRK2</scp> Modifiers of Parkinson's Disease. Annals of Neurology, 2021, 90, 76-88.	2.8	30
82	Heritability and genetic variance of dementia with Lewy bodies. Neurobiology of Disease, 2019, 127, 492-501.	2.1	29
83	Distinct characteristics of limbic-predominant age-related TDP-43 encephalopathy in Lewy body disease. Acta Neuropathologica, 2022, 143, 15-31.	3.9	29
84	Analysis of neurodegenerative disease-causing genes in dementia with Lewy bodies. Acta Neuropathologica Communications, 2020, 8, 5.	2.4	27
85	Myelin oligodendrocyte basic protein and prognosis in behavioral-variant frontotemporal dementia. Neurology, 2014, 83, 502-509.	1.5	26
86	Converging Patterns of Î±-Synuclein Pathology in Multiple System Atrophy. Journal of Neuropathology and Experimental Neurology, 2018, 77, 1005-1016.	0.9	26
87	TDP-43 pathology in a case of hereditary spastic paraplegia with a NIPA1/SPG6 mutation. Acta Neuropathologica, 2012, 124, 285-291.	3.9	24
88	Lack of evidence for Lrrk2 in Î±-synuclein pathological inclusions. Annals of Neurology, 2006, 60, 618-619.	2.8	23
89	Frontotemporal lobar degeneration proteinopathies have disparate microscopic patterns of white and grey matter pathology. Acta Neuropathologica Communications, 2021, 9, 30.	2.4	22
90	Tau immunotherapy is associated with glial responses in FTLD-tau. Acta Neuropathologica, 2021, 142, 243-257.	3.9	22

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91	Longitudinal structural gray matter and white matter MRI changes in presymptomatic progranulin mutation carriers. NeuroImage: Clinical, 2018, 19, 497-506.	1.4	21
92	Familial Frontotemporal Dementia: From Gene Discovery to Clinical Molecular Diagnostics. Clinical Chemistry, 2003, 49, 1717-1725.	1.5	20
93	Preemptive Treatment With Elbasvir and Grazoprevir for Hepatitis Câ€“Viremic Donor to Uninfected Recipient Kidney Transplantation. Kidney International Reports, 2020, 5, 459-467.	0.4	16
94	TMEM106B deficiency impairs cerebellar myelination and synaptic integrity with Purkinje cell loss. Acta Neuropathologica Communications, 2022, 10, 33.	2.4	16
95	Degeneration of the locus coeruleus is a common feature of tauopathies and distinct from TDP-43 proteinopathies in the frontotemporal lobar degeneration spectrum. Acta Neuropathologica, 2020, 140, 675-693.	3.9	15
96	TMEM106B modifies TDP-43 pathology in human ALS brain and cell-based models of TDP-43 proteinopathy. Acta Neuropathologica, 2021, 142, 629-642.	3.9	15
97	Genetic prediction of impulse control disorders in Parkinson's disease. Annals of Clinical and Translational Neurology, 2022, 9, 936-949.	1.7	15
98	Common neuropathological features underlie distinct clinical presentations in three siblings with hereditary diffuse leukoencephalopathy with spheroids caused by CSF1R p.Arg782His. Acta Neuropathologica Communications, 2015, 3, 42.	2.4	14
99	Validation of a Long-Read PCR Assay for Sensitive Detection and Sizing of C9orf72 Hexanucleotide Repeat Expansions. Journal of Molecular Diagnostics, 2018, 20, 871-882.	1.2	13
100	A comprehensive screening of copy number variability in dementia with Lewy bodies. Neurobiology of Aging, 2019, 75, 223.e1-223.e10.	1.5	13
101	Machine learning suggests polygenic risk for cognitive dysfunction in amyotrophic lateral sclerosis. EMBO Molecular Medicine, 2021, 13, e12595.	3.3	13
102	Biochemical and pathological characterization of frontotemporal dementia due to a Leu266Val mutation in microtubule-associated protein tau in an African American individual. Acta Neuropathologica, 2007, 113, 471-479.	3.9	12
103	The genetics and neuropathology of neurodegenerative disorders: perspectives and implications for research and clinical practice. Acta Neuropathologica, 2012, 124, 297-303.	3.9	12
104	Analysis of C9orf72 repeat expansions in a large international cohort of dementia with Lewy bodies. Neurobiology of Aging, 2017, 49, 214.e13-214.e15.	1.5	12
105	Signature laminar distributions of pathology in frontotemporal lobar degeneration. Acta Neuropathologica, 2022, 143, 363-382.	3.9	12
106	Genetic predictors of survival in behavioral variant frontotemporal degeneration. Neurology, 2019, 93, e1707-e1714.	1.5	11
107	Early emergence of anti-HCV antibody implicates donor origin in recipients of an HCV-infected organ. American Journal of Transplantation, 2019, 19, 2525-2532.	2.6	11
108	Hepatitis C virus genotyping of organ donor samples to aid in transplantation of <scp>HCV</scp>â€positive organs. Clinical Transplantation, 2018, 32, e13172.	0.8	9

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109	ADNC-RS, a clinical-genetic risk score, predicts Alzheimerâ€™s pathology in autopsy-confirmed Parkinsonâ€™s disease and Dementia with Lewy bodies. Acta Neuropathologica, 2020, 140, 449-461.	3.9	7
110	Plasma <scp>MIA</scp>, <scp>CRP</scp>, and Albumin Predict Cognitive Decline in Parkinson's Disease. Annals of Neurology, 2022, 92, 255-269.	2.8	7
111	Whole Clinic Research Enrollment in Parkinsonâ€™s Disease: The Molecular Integration in Neurological Diagnosis (MIND) Study. Journal of Parkinson's Disease, 2021, 11, 757-765.	1.5	5
112	Common genetic variation is associated with longitudinal decline and network features in behavioral variant frontotemporal degeneration. Neurobiology of Aging, 2021, 108, 16-23.	1.5	2
113	Saliva versus Upper Respiratory Swabs. Journal of Molecular Diagnostics, 2022, 24, 727-737.	1.2	2
114	Association of Mitochondrial DNA Genomic Variation With Risk of Pick Disease. Neurology, 2021, 96, e1755-e1760.	1.5	1
115	O2â€10â€05: Cerebrospinal Fluid Levels of Amyloid Beta and Tau as Endophenotypes Reveal Novel Variants Potentially Informative for Alzheimer's Disease. Alzheimer's and Dementia, 2016, 12, P252.	0.4	0
116	[P3â€“072]: MULTIâ€SITE EVALUATION OF THE AMPLIDEXÂ® PCR/CE TOMM40 KIT FOR RAPID AND ACCURATE GENOTYPING OF POLYâ€T LENGTH POLYMORPHISMS AT RS10524523 OF THE TOMM40 GENE. Alzheimer's and Dementia, 2017, 13, P959.	0.4	0
117	P1â€139: THE CONTRIBUTION OF SEXâ€SPECIFIC ASSOCIATIONS IN GENETIC STUDIES OF ALZHEIMER'S DISEASE PATHOLOGY. Alzheimer's and Dementia, 2018, 14, P327.	0.4	0
118	A Robust Xenotransplantation Model for Acute Myeloid Leukemia. Blood, 2008, 112, 2939-2939.	0.6	0
119	Mapping tau burden and neuronal loss in MAPT-associated frontotemporal lobar degeneration.. Alzheimer's and Dementia, 2021, 17 Suppl 3, e054141.	0.4	0